CAAE·青年思行

CAAE Youth Thinkings and Actions

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中国抗癫痫协会青年委员会

CHINA ASSOCIATION AGAINST EPILEPSY YOUTH COMMITTEE

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爱的回响艺术展

——医学人文让病房变得更加温暖

CAAE 青年委员会鄂豫湘大区

武汉儿童医院神经内科　胡玲　张瑾瑾

医本仁术，医学是一门以心灵温暖心灵的科学。这张照片相信大家都很熟悉，它来自 100 多年前的杭州，两位主人公一老一小，小的看上去四五岁，长衫马甲，长者一身西式装扮。这是当时的广济医院（现“浙大二院”）首任院长梅藤更先生与小患者互相鞠躬致敬。

2021 年 6 月,国务院办公厅印发《关于推动公立医院高质量发展的意见》明确：“建设特色鲜明的医院文化。唱响大医精诚、医者仁心主旋律，以充满人文关怀的医疗服务赢得患者、社会的信任和尊重。”医者人文素养与公立医院高质量发展密不可分，新时代呼唤医学人文的回归、要让人文主导医学、要让人文照亮医学。评判医务人员的人文标准应该是要拿起人文的听诊器，注重每个服务环节的人文质量管理。

作为人文关怀示范病区，我们神经内科一直秉承人文关怀理念，着力推进以患者需求为导向，以解决专科问题为目标的人文行动，不仅竭尽所能帮助患儿解除身体上的病痛，更关注患儿和家长的内心，帮助他们疏解内心焦虑、恐惧和忧愁。治愈过程也许很漫长，而我们一直都在努力；帮助很平常，而我们却从未停止过脚步；而安慰，是我们能够给予最平凡却最有效的能量。

小玉，15 岁 7 月，因“童年情绪障碍，多发性硬化，癫痫”于 2024 年 5 月 25 日收治神经内科。她也是神经内科的“老”病人，3 岁发病，每年来院复查，与神经内科的医护们结下了深厚的“革命友谊”。有感于孙丹主任长期以来的辛苦付出，住院次日她亲自给孙丹主任颁发了荣誉证书，为孙丹主任点赞。通过观察，孙丹主任发现患儿喜欢绘画，提议趁六一儿童节之际，举办小

玉画展。护士长迅速联系社工部专职社工策划活动方案，方案不断完善，以期通过此次活动，达到心灵绘画的力量，探索内心世界的疗愈之旅。

方案一经确定，护士长盛情邀请小玉参与画展的布置，并在画展后能进行“绘画讲座”，指导住院的小朋友们画画。欣然兴奋接受任务的“小画家”，在短短四天时间里，通过画笔绘出一幅幅心中所想、心之所向的二十四幅画作，并珍重交付护士长，表示捐献给科室。

2024 年 5 月 31 日通过小玉的亲自布置，这场特别的画展在神经内科“儿童活动室”如期召开。活动现场，小玉为病房的其他小朋友们特别绘画了儿童节主题的画作，参加活动的小朋友们也纷纷拿起了画笔，用作品去追逐时间的脚印，把孩子的纯真留在萌动的瞬间。

画展上，小玉为大家讲解每一幅画的创作初心与灵感来源，孩子们都很受触动，同为癫痫治疗的患儿，原来住院生活也可以变得多姿多彩。画展后小玉拿起画笔，教小孩子们画小老虎，希望孩子们像小老虎一样勇敢、强壮，大家都能拥有战胜困难的勇气和战胜病魔的信心。其实，这就是神经内科经常开展的慢病“同伴教育”中的一幕。

武汉儿童医院神经内科在在科室原有的慢病管理模式下，透过医护人员、医务社工、社会力量的跨专业团队协作方式，创新慢病患儿医务服务模式，对于患儿入院到出院有一个全流程的关注和支持。为服务对象提供多维度的支持，协作患儿家庭缓解经济、心理、康复照顾、教育、社会融入等问题，提升其疾病接受度及处理因病衍生的问题的能力，促进家庭自助互助，更好平衡康复和生活，更好的回归家庭、校园和社会。

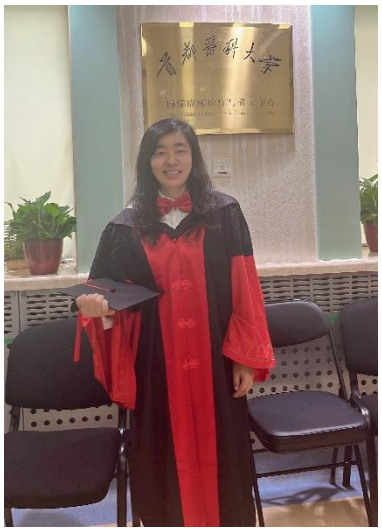
对患儿有一种好奇心或侦探般的态度往往会将病例转化为我们能够同情的真实人物。运用儿童游戏工具包、改造住院环境、拓展童心诊所、未来医生观察团等儿童视角的品牌活动，体现武汉儿童医院人文服务特色。正如爱德华·利文斯顿·特鲁多的哲学所说，“有时，去治愈，常常，去帮助，总是，去安慰”。医院也是人文关怀的萌芽地，医学人文实践十分重要。在孩子成长的每个阶段多少都会遇到来自外界的“声音”，尤其要注重对孩子们内心的关注，帮助他们建立起强大信心，生活中“让我们受苦的都不是事情本身，而是我们对事情的看法”。期望孩子们都能茁壮成长，健康快乐。愿医患间的尊重与关爱代代传承！

首都医科大学三博脑科医院 刘钊 人物（栾国明教授）绘画作品

我与癫痫的故事

CAAE 青年委员会京津冀晋蒙大区

首都医科大学附属北京儿童医院神经内科 李华



与癫痫结缘，始于 21 年前的春天，我从山东的一座小城只身来到了首都医科大学附属北京儿童医院神康中心脑电图室工作（现在为“神经中心脑电图室”）。往事一幕幕，恍若在眼前：抽搐的孩子常常让我不知所措，曲曲折折的脑电波形让我无所适从。这时候，神康中心主任邹丽萍教授似乎看穿了我窘迫的心态，把我叫到了身边，拍了拍我的肩膀，语重心长地说：“你可以的……”邹教授的一席话，让我备受鼓舞，不仅给了我动力和信心，更让我感受到了前辈对年轻人的提携和信任。于是我跑遍了整个北京城，买来了市面上关于脑电图学的四本书，开始了我在癫痫领域的学习和探索。

东晋时期的书法家王羲之练字的故事让我深受启发：他在练字时，常常将自己的墨池放在一个隐秘的角落，因为只有他自己知道墨池的存在，他深知书法练习需要长时间的专注与沉浸，墨池中的墨水也需要长时间的磨练与积累……王羲之练字的专注与执着，让深刻理解了精益求精的内涵。

从此以后，白天工作时，我在神康中心脑电图室跟着老师学习贴电极，练习脑电图操作技术；下班后去菜市场买来冬瓜，在冬瓜上练习电极安放位置；晚上，认真阅读脑电图书。有时候我会主动要求上夜班，因为我有自己的小心思：可以利用夜间时间练习阅图。每每夜班时，我把白天监测的患者脑电图重新阅过无数次，不懂的问题抽时间向老师请教，或者从书本上找答案。几个月过去了，当我再次面对一个孩子抽搐发作时，我不再是那样慌乱了，面对癫痫患者的脑电图也变得淡定了许多。

令我印象最深的一件事是对于一个重症脑炎患儿的诊断和治疗。那是 2012 年 3 月 9 日，急诊科紧急转送来一名 15 岁男孩，当班医生发现患儿发热、言语

错乱、抽搐发作，随后转变为意识模糊、呼吸急促等危重症状。但是接下来的一系列检查中，除脑电图提示异常外，其他检查均正常。这时候的我们迷茫了，孩子到底怎么了？我们冷静下来，重新查阅了患儿的临床及脑电图资料，意外发现这份脑电图异常不仅显示出弥漫性或局灶性慢波、癫痫样放电外，还出现了一种类似于δ刷的波形。经过查阅文献，神经内科团队果断地指向了一个可能的诊断——抗 NMDA 受体脑炎。这种脑炎是一种新近认识的自身免疫性脑炎，临床表现非常复杂多样，容易漏诊、误诊，如果不及时诊断和治疗，患者病死率和病残率很高。抗 NMDA 受体脑炎自 2007 年被人类认识以来，日渐受到医学界的重视。神经内科团队 2012 年率先报道了我国儿童这种脑炎，并相继开展了一系列深入研究。作为团队成员之一，我也在 2012 年完成了自己第一篇关于抗 NMDA 受体脑炎脑电图的论文。我们深刻地明白：我们不仅要治好患儿的病，更要安抚好家长焦虑的心。

十几年过去了，当时的患儿如今已组建了幸福美满的家庭，并有了一个可爱的儿子，每年，他都会到儿童医院来看我们，每年春节我们都会收到他的新年祝福！这个患儿的诊断、治疗经历，让我深深懂得：一名医者的冷静、果断来自于过硬的专业基本功！于是，2011 年，我踏入了首都医科大学的校门，开始了自己的临床基本功学习之旅——攻读儿内科学医学硕士学位。

2014 年 6 月，儿童医院成立了癫痫中心，我非常幸运地被安排在病房脑电图室，参与术前评估脑电监测工作。记得一个难忘的上午，我跟着同事到手术室观摩、学习难治性癫痫患者术中脑电监测，进入手术室的一瞬间，我看到一个难治性癫痫患者被麻醉后安静地躺在手术台上，我被触动了，再一次深深体会了生死时速的紧迫感与责任感。那次手术中，我不禁感慨医学的高科技化，主刀医生操作熟练、果断但不失谨慎，让我心中顿生敬佩之意，更深刻地体会到，我要正确认识自己的专业水平，需要进行刻苦钻研，重新出发。

那段时间，我重新拾起了大学时期学过的《断层解剖学》，与我的同事们一起承担难治性癫痫患者术前头皮脑电监测工作，我还自告奋勇承担起难治性癫痫患者立体定向脑电监测工作。由于第一次接触颅内脑电图，一份图我常常需要从早上八点开始，连续看到晚上，尽管身体非常疲惫，但是我与同事都坚持下来了，艰辛和兴奋，彼此支持和鼓励，团队合作的精神让我深受感动。然而，近几年的

工作经历，让我感受到了内科思维与外科思维的差距，也激发了我对立体定向脑电的兴趣。生命和健康是一个人最宝贵的东西，患儿家长将孩子的生命与健康托付给我们，我们责任重大。我深知只有站在巨人的肩膀上，才能走得更远。2020 年 9 月，我再一次踏入首都医科大学的校门，攻读博士学位课程，三年的博士学位学习之旅，让我铭记老师循循善诱的教导，学会了面对问题、解决问题的思维。尽管此时的我，依然还有很多短板，但心结已打开，接受自己的不完美，再次与以往告别！

癫痫，是一种常见的神经系统疾病，从脑电图的视角看，癫痫具有复杂性。脑电图是评估癫痫的重要方法，癫痫与脑电图有不解之缘！我热爱脑电图学这个专业，”匠心独运、精益求精”成为我的座右铭。我与癫痫的缘分，也许就是一个不断学习的历程，让我不断去追寻、攀登更高的山峰！

《CAAE 青年思行》论文摘要收录及作者标注说明

- 以八大区为单位收集，可以存在收集不全的情况；
- 收集范围包括 SCI（SCIE、ESCI）收录的论文、《ACTA Epileptologica》和《癫痫杂志》发表论文；
- 针对市县级医院的基层青年委员，还将收录在中华系列杂志和中国高质量期刊发表的中文论文；
- 收录的论文为 2024 年 1-5 月发表的论文；
- 收录格式为引用格式+摘要；
- 作者中将列出全部作者名字的英文缩写或中文，但仅重点标注第一作者（含共同）和通讯作者的中文名字，以便青委们之间更好直接交流和合作，我们深知任何工作都是一个团队完成，特别是一些老师或专家也是文章通讯作者或共同通讯作者、合作作者或者没有署名，青年人的工作都在他（她）们的指导和帮助下完成。

我与癫痫患者的故事

——走进您心，伴您同行

CAAE 青年委员会川渝滇黔藏大区

重庆医科大学附属第二医院癫痫中心 张瑾

癫痫患者梵高的作品《星空》举世闻名，他用夸张的手法，生动地描绘了充满运动和变化的星空，天空中飞卷的星云，好像鲜花一样怒放，那柔韧的枝条虽然力量微薄，却无畏地抽向天空。而直上云端的巨大柏树，形如一团黑色的火舌，不屈地奋力伸展着枝叶，纵使生活艰难，依然不向命运低头。

这是梵高通过绘画作品传递给我们的情绪和态度，下面我想讲一讲我和癫痫患者绘画作品中的故事。

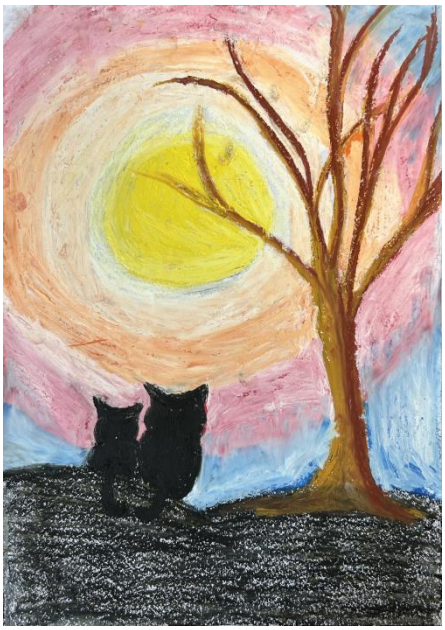
一. 微光

这幅作品的创作者叫小华（化名），女性，16 岁。癫痫病史 10 余年的小华早早就辍学了，花样的年纪却辗转在各个医院中虚度，小华的脸上很少看到有笑容。

我问道：“小华，你为什么不开心呢？”

小华回答：“姐姐，生病十几年来，看了好多医院。我没有朋友，也没有爱好，我感觉我的生活已经没有希望了。”

接下来的日子，我给外地来的小华做重庆旅游攻略，鼓励她闲暇之余出去看看；和她分享癫痫患者回归正常生活的案例；和她讨论当下热门的话题；给她推荐好看的电影和好听的音乐。



出院回家后，小华给我发来短信，她说：“这个世界好像也没有那么糟糕，谢谢你让我重新看到了生活的美好！”

作者：小华（化名）

作品名称：微光

作者心语：穿透黑暗，去寻找黎明前的光

二、海底童趣

作者：小思（化名）

作品名称：海底童趣

作者心语：

坐着章鱼时光机，遇见童心、勇气和希望

这幅作品的创作者叫小思（化名），男性，23 岁。还记得第一次见到小思的样子，高高帅帅，白白净净，一双机灵的圆眼睛骨碌碌转，嘴巴叽叽喳喳说个不停。那时候的小思 18 岁，准备入伍当兵时体检查出颅内静脉窦血栓，从此与我们结了缘。6 年的疾病折磨，反复的癫痫样发作以及蛛网膜下腔出血，当初那个高大帅气的少年早已不是当初阳光的样子，身体日渐佝偻，眼神逐渐涣散。

我问他：“小思，还想去当兵吗？”

小思回答：“嗯.....。”这时候的小思说话已经不成句了,当听到“当兵”两个字,我看到他的眼神迅速闪过一丝欣喜,继而又暗淡下来。

接下来的日子，分享网络上的军人故事和部队趣事成了我们俩的聊天日常，“两岁幼童被卡，退伍军人徒手爬至五楼施救”、“最美光背兵哥哥勇救溺水儿童”.....故事很小，力量却很大。出院时小思创作了绘画作品“海底童趣”，他傻笑着告诉我说：“小叮当有任意门，我有时光机，还是可以穿越回最美好的 18 岁。”是的，满怀希望就会所向披靡！

三、蝶变

作者：小琴（化名）

作品名称：蝶变

作者心语：

突破瓶颈和未知，破茧成蝶，迎风飞翔

这副作品的创作者叫小琴（化名），傣族，患病以前是位能歌善舞的美丽大姐。熟识以后，大姐得意的拿出生病以前的跳舞视频给我看，视频中的她长发飘逸，裙袂飞扬，像美丽的蝴蝶翩翩舞动。我拿∂出女儿的跳舞视频要和她“远程 PK”，大姐摸了摸因为手术被剃光头发的大脑门，不服气的说道：“给我一点点长头发的时间，我还是广场坝子上最闪亮的星！”就像小琴留下的作品，翩翩起舞在花丛间的蝴蝶，每一片翅膀都闪耀着绚丽的光彩，给我们的生活带来无比的生机与活力。

以上是我和癫痫患者的一些小故事，与其说是我在帮助癫痫患者，倒不如说是这些患者在点滴中治愈我,让我在困顿迷茫时有了向前的勇气。

最后致患者的话：同样的画笔，有人画山，有人画海，都是风光；同样的人生，有人顺利，有人波折，都是精彩！您的病痛我虽无法“身受”，却在尽力“感同”，疾病抗争之路，我们伴您同行！

圆 28 岁女性癫痫患者一个幸福母亲梦

CAAE 青年委员会陕西咸阳市

咸阳市第一人民医院 王沛 张永锋

“王医生，给您报个喜，我生了!”今年 3 月，收到娇娇（化名）的这条微信，我的内心生出了极大的喜悦，激动的心情更是无以言表，我想这是为她诞下新生命的祝福，更是为她的“新生”。此刻，我们都是世界上最幸福的人。

28 岁的娇娇是个因为反复癫痫发作而导致不能生育的药物难治性癫痫的女性，在第一次怀孕过程中，因为反复癫痫发作导致流产，对患者本人产生了极大的精神刺激，导致患者的婚姻破裂，不能进行正常的工作。可以说，疾病就像被推倒的多米诺骨牌，彻底改变了她的命运，不仅因为治疗为其父母造成了极大的经济压力，也让她的心理一次又一次遭受“毁灭”式打击，导致她多次有轻生的念头。

第一次在神经内科癫痫中心接诊娇娇时，她是 70 多岁的父母陪同下来就诊的，我对她进行了详细的病史询问，药物服用情况、癫痫发作情况及癫痫共患病的全面的评估，对患者癫痫的病因进行了详细的查找，最终确定为药物难治性癫痫。得知在此之前，她有长达 10 余年的癫痫病史，曾就诊外院同时给予口服“丙戊酸钠片、卡马西平片、丙戊酸镁缓释片、苯妥英钠片、苯巴比妥钠片”等多种抗癫痫发作药物治疗，服药期间怀孕 1 次，但因癫痫发作导致流产，深知对于这样的患者，无论是身心都非常脆弱。

“癫痫是可以控制的，也是可以生育的，并且癫痫不会对胎儿造成严重的损害，可以孕育健康的宝宝!”当患者听到“可控”、“可以孕育健康宝宝”，黯淡的眼神突然盯着我，好像需要这样可预期的结果支撑她做一些改变!在之后长达 10 时间的就诊、随访过程中，我中心的医护人员都熟知了这样一位患者，她渴望一丝丝信心去面对生活，渴望有心理安慰、深入交流及关心爱护，渴望有自己的健康宝宝。全方位的关怀就像往内心深处已近干涸的土壤上浇灌的一丝清泉，能给心灵注入希望。我科使用了对患者妊娠及胎儿相对安全的新型抗癫痫药物，奥卡西平片、左乙拉西坦片、拉莫三嗪片，逐渐在“可控”的病情中她重拾生活的勇

气，我也鼓励她在癫痫控制后可以重新工作及步入婚姻，重新组建幸福的家庭。

庆幸的是，随诊 1 年后，娇娇未有癫痫发作，同时脑电图复查 3 次均未见脑电图癫痫样放电，癫痫控制后患者再次迎来了婚姻，同时在 1 年后成功怀孕并产下一名健康的小宝宝.....

娇娇的来信仿佛是她幸福生活的缩影，也让我平凡的工作充满自豪感、成就感。医学是生命至上的科学，是人世间最神圣的事业，没有任何一种事业的价值会高于人的生命。作为一名医务工作者，最大的幸福就是患者的康复，患者离开医院时脸上灿烂的微笑。那将是我们持续进行医学工作永不枯竭的动力。

女性癫痫一直是癫痫患者的重点关注问题，许许多多的女性癫痫患者因为癫痫导致流产、失业、离婚等，对其人生造成了种种困扰，我院癫痫中心致力于详细诊治、精心呵护，帮助患者解除了癫痫带来的种种困扰及伤害，为她们的人生开创崭新的、幸福的、美满的人生篇章。我们多希望像娇娇这样的动人故事更多的在人世间发生，多希望癫痫患者可以过上“无痫生活”，拥有一个“无癫痫、抱健康”的幸福人生，多希望这样的人间大爱可以继续传递.....

我与中抗共成长

CAAE 青年委员会陕甘青宁新大区

空军军医大学西京医院神经内科　刘佳　杨蕾

站在护理事业的光辉舞台上，我心潮澎湃，无比自豪能与尊贵的中国抗癫痫协会（CAAE）并肩同行，在提升癫痫患者生命质量的宏伟征途中，我不仅见证了

自我专业的蜕变，更深刻领悟到护理之光如何温暖社会的每一个角落。

我的护理旅程，是始于梦想，兴于奉献的故事。CAAE 的每一次培训与研讨，都是我专业版图上不可或缺的里程碑。在那里，我与同仁们的思想碰撞出智慧的火花，从专家的睿智中汲取宝贵知识，最新治疗理念与护理技术的灌溉，让我的专业之树更加茁壮。这份成长，让我在细微处见真章，于急难中显从容，学会了以细腻入微的关怀，编织个性化护理的经纬，为患者点亮希望的灯塔。

在这条充满温情与挑战的成长轨迹上，我邂逅过一个家庭，他们的故事如同夜空中最亮的星，照亮了护理之路的深远意义。孩子患有难治性癫痫，经济困难重重。父亲孤军奋战，支撑全家，孩子的医疗需求紧迫而沉重。我们科室毅然伸出援手，开辟医疗绿径，确保其治疗之路无阻，更在细节中展现温情，如亲人般关怀，解决交通与药物难题，疫情期间也不曾让爱隔离。

随着时间推移，政策的春风和我们不懈的努力带来了转机：费用减免、低保支持，孩子的病情好转，家庭重现生机。这些正面变化如同希望之光，不仅照亮了这个家庭，也坚定了我们作为医护人员的信念——我们在编织爱、希望与重生的图谱，护理工作超越了职业本身，成为对生命的深情礼赞。

护理，对我而言，远超乎职业定义，它是灵魂深处的呼唤，是对生命的最高敬仰。在与患者的紧密相连中，我成为了他们痛苦中的一抹温柔，是携手并进的同路人。那些共度风雨的日子，让我确信，我们不仅是治愈身体的护航者，更是心灵慰藉的引路人，用爱与专业，共同绘制生命中最美的风景线。

回望与 CAAE 同行的时光，我满载感激，内心更加坚定了作为护士的骄傲与责任。我学会了用心聆听每一个心跳背后的故事，理解每一次呼吸中的期待，支持每一段艰难却坚韧的旅程。

未来的日子里，我将不忘初心，继续在这条既充满挑战又满怀收获的道路上坚定前行，不懈提升自我，以更高水准的护理技艺，为癫痫患者带去更多的温暖、希望与光明。因为，我知道，在这条爱的征途上，我们共同守护的，不仅仅是健康，更是生命的尊严与奇迹。

医疗的意义

CAAE 青年委员会京津冀晋蒙大区

首都医科大学宣武医院功能神经外科 乔梁

几年前的一个下午，我如常完成一台神经外科手术，来到病房查房。多年的临床工作已经使我在大脑中习惯性的将患者分为几类：对于术前患者，需要确认（偶有也会推翻）他们在门诊得到的初步诊断，完善相关检查化验，着重评估手术适应证或禁忌证。如果适合手术，就来安排术前谈话，向患者及家属介绍手术目的、流程、预期效果，当然还有可能的并发症。通常，还要安抚患者的术前紧张情绪。

对于术后患者，首要关注的是手术效果（虽然有些手术未必即刻见效）以及是否有并发症，如果一切顺利，浅浅的成就感之余（多年临床历练后早已不会为一台手术的成功就欣喜若狂）更要叮嘱好患者各项术后恢复注意事项。

对于临近出院的患者，我会检查他们的切口愈合状况。宣武医院功能神经外科的患者大多从全国各地慕名而来，许多在交通、时间和花费上不方便定期复诊，所以做好出院宣教（包括介绍互联网问诊）尤为重要。

走入病房，我看到当天刚入院的一位小伙子：小杨（化名），28 岁。翻看他的门诊病历，上面写着简要病史和初步诊断：难治性癫痫。癫痫是一种神经系统的慢性疾病，常表现为发作性意识丧失伴肢体抽搐（也可有不同形式）。功能神经外科收治的癫痫患者大多病情重、病史长、用药多但效果不佳、并常伴有不同程度的记忆减退、认知障碍、情绪问题等。再加之发作之不可预期性，癫痫就如“不定时炸弹”般严重影响患者的生长发育、就学就业，甚至可能威胁生命。

眼前的小杨坐在病床上，戴着眼镜，斯斯文文。交谈中得知他顺利完成了大学本科和研究生学业。考虑上文说过的那些因素，这真是值得祝贺的成就，我心想。

“你现在从事什么工作呢？“我问道。

”我目前在##工作，担任程序员。”小杨回答。

我眼前一亮：##是国内鼎鼎有名的互联网企业。在近年来严峻的就业形势下，

聘入互联网“大厂”要经过层层筛选。对健康的大学生尚不简单，小杨“带病”走到今天实属不易。他自食其力，甚至赚钱为自己看病，我对他多了一份感佩。

“那你现在发作多吗，对生活工作影响大吗？“我关心道。

“是的，乔大夫。我隔三差五就会有愣神发作，好在没肢体抽搐，仍坚持上班。但不久前有一次大发作在办公室跌倒，把我同事都吓坏了，紧急送我去医院。我现在服药量不小，但效果不好，所以来寻求是否能手术治疗。我特别担心疾病会继续发展到影响我的工作。”小杨回答时露出求助的眼神。

我仿佛看到眼前的这位年轻人正在背着沉重包袱，艰难的奋力前行。很多时候，也许正是这种减轻患者疾病重负的天然使命感驱策着医务人员不断付出心智体力，精进技术，传递温暖。

“作为程序员，你需要夜间加班多吗？”我继续问道。

“是的，我这次住院还带来了电脑，不想把工作落下太多”。小杨指指床头柜上的苹果电脑。

我想到前一段媒体常讨论的“996”现象，“我理解你的工作需求。但记得尽量不熬夜，要规律作息，这样有助于减少发作。特别是不要依靠咖啡、浓茶提神。”

“谢谢乔大夫提醒。我得病以来也查询过不少资料，对酒精、咖啡、浓茶都戒掉了。我会尽量避免熬夜加班，但有时候项目来了……”。

……

走出病房，我仍被小杨的正能量感动，体会到医患互动不只是单向施援。

近年来，癫痫学领域已经取得了巨大发展，产生了许多新的治疗方案，但具体到小杨这样的病例，什么是最适合的？

接下来的数天，我们为小杨进行了脑电图、磁共振等相关检查，确认了“难治性癫痫”的诊断。脑电图提示在小杨即使没有发作的时间段，脑内也存在异常癫痫样放电。看来在药物治疗效果有限的基础上确实应该考虑手术干预。

我向小杨进一步解释手术方案，“癫痫外科有不同术式。致痫灶切除术虽然术后无发作比例较高，但手术切除是不可逆的，其风险（包括对于认知功能的影响）值得重视。多学科专家会诊认为，基于你的具体病情和病灶分布，应该考虑神经调控治疗。”

小杨认真的聆听。

我继续介绍，“这种神经调控方法叫迷走神经电刺激。它通过植入体内的设备刺激迷走神经，从而逐步降低大脑的兴奋性，达到减少减轻发作的目的。它的优势在于不会对你目前良好的认知功能产生负面影响，甚至还可能改善情绪。这是一种微创、可逆和可调节的技术手段。”

“所以，这种方法听起来像中医的调理？它不是通过破坏性的切除脑组织，而是调整大脑兴奋性？”小杨问。

“对，可以这样理解。神经调控的效果也许不如切除术那样立竿见影，更需要耐心和时间。但是它最大的优势在于对患者功能的保护，而且保留脑组织的完整也为未来可能应用新的治疗方法预留了机会。”我解释道。

小杨重重的点了点头，我感到也感谢他的信任。想起国内知名的儿童神经科专家邹丽萍教授曾将医疗过程比喻为医生背着患者过河，互相协作才可能共赢，此刻我深以为然。

几天后，小杨成功的接受了手术，术后恢复良好，没有任何并发症。我从内心为他高兴。记得出院当天，小杨等待办理手续时又坐在医院楼梯间噼噼啪啪的敲起了电脑，大概又接到新的项目吧。看得出工作对于他的重要意义。

之后的几年，我和同事们共同经历了抗击新冠疫情以及疫情后时代。接诊的患者一批又一批，完成的手术一台又一台，繁忙的工作中我几乎淡忘了小杨。直到不久前，我路过某互联网公司。正值下班，大厦里涌出熙熙攘攘、活泼朝气的年轻面孔。我突然想起小杨，他大概就是这些年轻人之一吧。他现在身体好吗，工作顺利吗？

这些年轻人，如同各行各业的广大劳动者一样。他们正在通过勤奋打拼实现个人价值、同时实实在在的建设国家、奉献社会。医疗服务的大多数对象正是这样的普通人。医疗行业正是通过保障每一位劳动者、每一位平凡公民的健康，而为国家和社会发展提供助力。

“健康中国”的大战略并不虚无缥缈，它就落实在每一个公民的健康保障上，它关乎国家的综合实力强盛和社会整体文明程度。促进每个公民健康，促进每个劳动者健康，才能建设起健康强大的国家。这就是我理解的医疗的意义。

编号：JJJM-2024-1-1

引用格式：Ma H, Zhu L, Yang X, Ao M, Zhang S, Guo M, Dai X, Ma X, Zhang X. Genetic and phenotypic analysis of 225 Chinese children with developmental delay and/or intellectual disability using whole-exome sequencing. BMC Genomics. 2024;25(1):391.

通信作者：马秀伟

Abstract

Background: Developmental delay (DD), or intellectual disability (ID) is a very large group of early onset disorders that affects 1–2% of children worldwide, which have diverse genetic causes that should be identified. Genetic studies can elucidate the pathogenesis underlying DD/ID. **Methods:** In this study, whole-exome sequencing (WES) was performed on 225 Chinese DD/ID children (208 cases were sequenced as proband-parent trio) who were classified into seven phenotype subgroups. The phenotype and genomic data of patients with DD/ID were further retrospectively analyzed. There were 96/225 (42.67%; 95% confidence interval [CI] 36.15–49.18%) patients were found to have causative single nucleotide variants (SNVs) and small insertions/deletions (Indels) associated with DD/ID based on WES data. The diagnostic yields among the seven subgroups ranged from 31.25 to 71.43%. Three specific clinical features, hearing loss, visual loss, and facial dysmorphism, can significantly increase the diagnostic yield of WES in patients with DD/ID (*P*=0.005, *P*=0.005, and *P*=0.039, respectively). Of note, hearing loss (odds ratio [OR]=1.86%; 95% CI=1.00-3.46, *P*=0.046) or abnormal brainstem auditory evoked potential (BAEP) (OR=1.91, 95% CI=1.02–3.50, *P*=0.042) was independently associated with causative genetic variants in DD/ID children. **Conclusions：** Our findings enrich the variation spectrums of SNVs/Indels associated with DD/ID, highlight the value genetic testing for DD/ID children, stress the importance of BAEP screen in DD/ID children, and help to facilitate early diagnose, clinical management and reproductive decisions, improve therapeutic response to medical treatment.

编号：JJJM-2024-1-2

引用格式：Sun T, Wu S, Liu X, Tao JX, Wang Q. Impact of intracranial subclinical seizures on seizure outcomes after SLAH in patients with mesial temporal lobe epilepsy. Clin Neurophysiol. 2024;160:121-129.

第一作者：孙太欣

Abstract

Objective: To investigate the association between subclinical seizures detected on intracranial electroencephalographic (i-SCSs) recordings and mesial temporal sclerosis (MTS), as well as their impact on surgical outcomes of stereotactic laser amygdalohippocampotomy (SLAH). **Methods:** A retrospective review was conducted on 27 patients with drug-resistant mesial temporal lobe epilepsy (MTLE) who underwent SLAH. The number of seizures detected on scalp EEG and iEEG was assessed. Patients were followed for a minimum of 3 years after SLAH. **Results:** Of the 1715 seizures recorded from mesial temporal regions, 1640 were identified as i-SCSs. Patients with MTS were associated with favorable short- and long-term surgical outcomes. Patients with MTS had a higher number of i-SCSs compared to patients without MTS. The numbers of i-SCSs were higher in patients with Engel I-II outcomes, but no significant statistical difference was found. However, it was observed that patients with MTS who achieved Engel I-II classification had higher numbers of i-SCSs than patients without MTS ($P < 0.05$). **Conclusion:** Patients with MTS exhibited favorable short-term and long-term surgical outcome after SLAH. A higher number of i-SCSs was significantly associated with MTS in patients with MTLE. The number of i-SCSs tended to be higher in patients with Engel I-II surgical outcomes. **Significance:** The association between i-SCSs, MTS, and surgical outcomes in MTLE patients undergoing SLAH has significant implications for understanding the underlying mechanisms and identifying potential therapeutic targets to enhance surgical outcomes.

编号：JJJM-2024-1-3

引用格式：Wang W, Huang Q, Zhou Q, Han J, Zhang X, Li L, Lin Y, Wang Y. Multimodal noninvasive evaluation in MRI-negative epilepsy patients. Epilepsia Open. 2024;9(2):765-775.

通讯作者：林一聪

Abstract

Background: Presurgical evaluation is still challenging for MRI-negative epilepsy patients. As noninvasive modalities are the easiest acceptable and economic methods in determining the epileptogenic zone, we analyzed the localization value of common noninvasive methods in MRI-negative epilepsy patients. **Methods**: In this study, we included epilepsy patients undergoing presurgical evaluation with presurgical negative MRI. MRI postprocessing was performed using a Morphometric Analysis Program (MAP) on T1-weighted volumetric MRI. The relationship between MAP, magnetoencephalography (MEG), scalp electroencephalogram (EEG) and seizure outcomes were analyzed to figure out the localization value of different noninvasive methods. **Results**: Eight-six patients were included in this study. Complete resection of the MAP-positive regions or the MEG-positive regions was positively associated with seizure freedom ($p = 0.028$ and 0.007, respectively). When an area is co-localized by MAP and MEG，the resection of the area was significantly associated with seizure freedom ($p = 0.006$). However, neither the EEG lateralization nor the EEG localization showed statistical association with the surgical outcome ($p = 0.683$ and 0.505, respectively). **Conclusions**: Scalp EEG had limited role in presurgical localization and predicting seizure outcome, combining MAP and MEG results can significantly improve the localization of epileptogenic lesions and have positive association with seizure-free outcome.

编号：JJJM-2024-1-4

引用格式：Yan Z, Yang Y, Wang J, Deng Q, Zhang L, Wang M, Zhou J, Guan Y, Luan G, Wang M. Posterior cingulate epilepsy: Seizure semiology and intracranial electrical stimulation using SEEG. Seizure. 2024;119:28-35.

通讯作者 王梦阳

Abstract

Purpose: This study aimed to explore seizure semiology and the effects of intracerebral electrical stimulation on the human posterior cingulate cortex (PCC) using Stereoelectroencephalography (SEEG) to deepen our comprehension of posterior cingulate epilepsy (PCE). **Methods:** This study examined the characteristics of seizures through video documentation, by assessing the outcomes of intracranial electrical stimulation (iES) during SEEG. We further identified the connection between the observed semiology and precise anatomical locations within the PCC subregions where seizure onset zones (SOZ) were identified. **Results:** Analysis was conducted on 59 seizures from 15 patients recorded via SEEG. Behavioural arrest emerged as the predominant manifestation across the PCC subregions. Where ictal activity extended to both the mesial and lateral temporal cortex, automatism was predominantly observed in seizures originating from the ventral PCC (vPCC). The retrosplenial cortex (RSC) is associated with complex motor behaviour, with seizure discharges spreading to the temporal lobe. Seizures originating from the PCC include axial tonic and autonomic seizures. Only one case of positive clinical seizures was documented. High frequencies of iES within the PCC induced various clinical responses, categorised as vestibular, visual, psychological, and autonomic, with vestibular reactions primarily occurring in the dorsal PCC (dPCC) and RSC, visual responses in the left RSC, and autonomic reactions in the vPCC and RSC. **Conclusion:** The manifestations of seizures in PCE vary according to the SOZ and the patterns of seizure propagation. The occurrence of seizures induced by iES is exceedingly rare, indicating that mapping of the PCC could pinpoint the primary sector of PCC

编号：JJJM-2024-1-5

引用格式：Yang Y, Zhang C, Cao C, Su W, Zhao N, Yue W. Clinical Features of Patients with Myelin Oligodendrocyte Glycoprotein Antibody-Associated Disease and Isolated Seizure Symptoms. Neuropsychiatr Dis Treat. 2024;20:61-67. doi: 10.2147/NDT.S444853.

通讯作者 岳伟

Abstract

Background: Myelin oligodendrocyte glycoprotein (MOG) antibody–associated encephalitis is a new clinical phenotype of inflammatory demyelinating diseases. Some MOG antibody–positive patients with central nervous system demyelinating events present with isolated seizures. However, there are gaps in the epidemiological knowledge regarding seizures with MOG antibody–associated encephalitis in adults. This study characterized the clinical features and treatment of MOG antibody–positive patients with isolated seizures. **Methods:** We reviewed all the patients admitted to Tianjin Huanhu Hospital between Jan. 1st 2017 and Jan. 1st 2022, to screen the MOG antibody–positive patients with isolated seizures, and collected the concerned patients' information regarding epidemiology, clinical presentations, laboratory and radiological characteristics, electroencephalogram (EEG), treatments, and prognoses. **Results:** We collected six MOG antibody–positive adult patients who had isolated symptomatic seizures. The mean age of the patients was 33 years (range, 29–40 years), and five (83.3%) were men. All patients presented with motor seizures, five (83.3%) had cognitive dysfunction, and only one (16.7%) had status epilepticus. Five (83.3%) patients had a good response to immunotherapy and antiseizure medications; only one had a sequela. The cerebrospinal fluid or serum anti-MOG antibody test turned negative over time. **Conclusions:** The most common seizure type in patients with MOG antibody–associated encephalitis with isolated seizures was focal to bilateral tonic-clonic seizures, and most patients had a good prognosis. Adding antiseizure medications were beneficial for MOG antibody–positive patients with seizures.

Relapses and sequelae were associated with low-dose, short-time, or delayed therapy, and wide-range demyelinating brain damage.

编号：JJJM-2024-1-6

引用格式：Li W, Meng J, Lei J, Li C, Yue W. Carbon monoxide poisoning with hippocampi lesions on MRI: cases report and literature review. BMC Neurol. 2024;24(1):159.

通讯作者 岳伟

Abstract

Background: Carbon monoxide (CO) poisoning is now one of the leading causes of poisoning-related mortality worldwide. The central nervous system is the most vulnerable structure in acute CO poisoning. MRI is of great significance in the diagnosis and prognosis of CO toxic encephalopathy. The imaging features of CO poisoning are diverse. We report atypical hippocampal lesions observed on MRI in four patients after acute CO exposure. **Case presentations:** We report four patients who presented to the emergency department with loss of consciousness. The diagnosis of CO poisoning was confirmed on the basis of their detailed history, physical examination and laboratory tests. Brain MRI in all of these patients revealed abnormal signal intensity in hippocampi bilaterally. They all received hyperbaric oxygen therapy. The prognosis of all four patients was poor. **Conclusions:** Hippocampi, as a relatively rare lesion on MRI of CO poisoning, is of important significance both in the early and delayed stages of acute CO poisoning. In this article, we summarize the case reports of hippocampal lesions on MRI in patients with CO poisoning in recent years, in order to provide reference for the diagnosis and prognosis of CO poisoning.

编号：JJJM-2024-1-7

引用格式：Zhang J, Ling L, Xiang L, Li W, Bao P, Yue W. Clinical features of neuronal intranuclear inclusion disease with seizures: a systematic literature review. Front Neurol. 2024;15:1387399.

通讯作者 岳伟

Abstract

Background: Infant, junior, and adult patients with neuronal intranuclear inclusion disease (NIID) present with various types of seizures. We aimed to conduct a systematic literature review on the clinical characteristics of NIID with seizures to provide novel insight for early diagnosis and treatment and to improve prognosis of these patients. **Methods:** We used keywords to screen articles related to NIID and seizures, and data concerning the clinical characteristics of patients, including demographic features, disease characteristics of the seizures, treatment responses, imaging examinations, and other auxiliary examination results were extracted. **Results:** The included studies comprised 21 patients with NIID with seizures. The most common clinical phenotypes were cognitive impairment (76.20%) and impaired consciousness (57.14%), and generalized onset motor seizures (46.15%) represented the most common type. Compared with infantile and juvenile cases, the use of antiepileptic drugs in adults led to significant seizure control and symptom improvement, in addition to providing a better prognosis. The number of GGC sequence repeats in the NOTCH2NLC gene in six NIID patients with seizures who underwent genetic testing ranged 72–134. **Conclusions:** The most common clinical phenotypes in patients with NIID with seizures were cognitive impairment and consciousness disorders. Patients with NIID presented with various types of seizures, with the most common being generalized onset motor seizures. Adult patients had a better prognosis and were relatively stable. The early diagnosis of NIID with seizures is of great significance for treatment and to improve prognosis.

编号：JJJM-2024-1-8

引用格式：Chen G, Wang A, Zhang X, Li Y, Xia X, Tian X, Li J, Miao Z, Yue W. Systemic Immune-Inflammation Response is Associated with Futile Recanalization After Endovascular Treatment. Neurocrit Care. 2024. doi: 10.1007/s12028-023-01930-y.

通讯作者 岳伟

Abstract

Background: Frequent incidence of futile recanalization decreases the benefit of endovascular treatment (EVT) in acute ischemic stroke. We hypothesized that the inflammation and immune response after ischemic are associated with futile recanalization. We aimed to investigate the correlation of admission systemic immune-inflammation index (SII) with futile recanalization post EVT. **Methods:** Patients with successful recanalization (modified Thrombolysis in Cerebral Ischemia angiographic score 2b-3) and maintained artery recanalized after 24 h of EVT were chosen from a prospective nationwide registry study. Futile recanalization was defined as a poor functional outcome (modified Rankin Scale score 3-6) at 90 days, irrespective of a successful recanalization. At admission, SII was calculated as (platelet count × neutrophil count)/lymphocyte count/100. Logistic regression analysis helped to test the relationship of SII with futile recanalization. **Results:** Among the 1,002 patients included, futile recanalization occurred in 508 (50.70%). No matter whether tested as quartiles or continuous variables, SII was significantly associated with futile recanalization ($P < 0.05$), and for every one standard deviation increase of SII, the risk of futile recanalization elevated by 22.3% (odds ratio 1.223, 95% confidence interval 1.053-1.444, $P = 0.0093$). Moreover, no significant interactions could be observed between SII or SII quartiles and age, baseline National Institutes of Health Stroke Scale scores, onset-to-recanalization time, and modified Thrombolysis in Cerebral Ischemia angiographic scores (all P for interaction > 0.05). **Conclusions:** Early SII elevation was associated with an increased risk of futile recanalization among patients with EVT. Our

results indicated that therapeutic drug targeting hyperreactive immune-inflammation response might be helpful for reducing the incidence of futile recanalization.

编号：JJJM-2024-1-9

引用格式：Zhang J, Ling L, Xiang L, Li W, Bao P, Yue W. Role of the gut microbiota in complications after ischemic stroke. Front Cell Infect Microbiol. 2024;14:1334581.

通讯作者 岳伟

Abstract

Ischemic stroke (IS) is a serious central nervous system disease. Post-IS complications, such as post-stroke cognitive impairment (PSCI), post-stroke depression (PSD), hemorrhagic transformation (HT), gastrointestinal dysfunction, cardiovascular events, and post-stroke infection (PSI), result in neurological deficits. The microbiota-gut-brain axis (MGBA) facilitates bidirectional signal transduction and communication between the intestines and the brain. Recent studies have reported alterations in gut microbiota diversity post-IS, suggesting the involvement of gut microbiota in post-IS complications through various mechanisms such as bacterial translocation, immune regulation, and production of gut bacterial metabolites, thereby affecting disease prognosis. In this review, to provide insights into the prevention and treatment of post-IS complications and improvement of the long-term prognosis of IS, we summarize the interaction between the gut microbiota and IS, along with the effects of the gut microbiota on post-IS complications.

编号：JJJM-2024-1-10

引用格式：Du P, Zhang X, Lian X, Hölscher C, Xue G. O-GlcNAcylation and Its

Roles in Neurodegenerative Diseases. J Alzheimers Dis. 2024;97(3):1051-1068.

通讯作者：薛国芳

Abstract

As a non-classical post-translational modification, O-linked β-N-acetylglucosamine (O-GlcNAc) modification (O-GlcNAcylation) is widely found in human organ systems, particularly in our brains, and is indispensable for healthy cell biology. With the increasing age of the global population, the incidence of neurodegenerative diseases is increasing, too. The common characteristic of these disorders is the aggregation of abnormal proteins in the brain. Current research has found that O-GlcNAcylation dysregulation is involved in misfolding or aggregation of these abnormal proteins to mediate disease progression, but the specific mechanism has not been defined. This paper reviews recent studies on O-GlcNAcylation's roles in several neurodegenerative disorders such as Alzheimer's disease, Parkinson's disease, amyotrophic lateral sclerosis, Huntington's disease, Machado-Joseph's disease, and giant axonal neuropathy, and shows that O-GlcNAcylation, as glucose metabolism sensor, mediating synaptic function, participating in oxidative stress response and signaling pathway conduction, directly or indirectly regulates characteristic pathological protein toxicity and affects disease progression. The existing results suggest that targeting O-GlcNAcylation will provide new ideas for clinical diagnosis, prevention, and treatment of neurodegenerative diseases.

编号：JJJM-2024-1-11

引用格式：Adamu A, Li S, Gao F, Xue G. The role of neuroinflammation in neurodegenerative diseases: current understanding and future therapeutic targets. Front Aging Neurosci. 2024:16:1347987.

通讯作者：薛国芳

Abstract

Neuroinflammation refers to a highly complicated reaction of the central nervous system (CNS) to certain stimuli such as trauma, infection, and neurodegenerative diseases. This is a cellular immune response whereby glial cells are activated, inflammatory mediators are liberated and reactive oxygen and nitrogen species are synthesized. Neuroinflammation is a key process that helps protect the brain from pathogens, but inappropriate, or protracted inflammation yields pathological states such as Parkinson's disease, Alzheimer's, Multiple Sclerosis, and other neurodegenerative disorders that showcase various pathways of neurodegeneration distributed in various parts of the CNS. This review reveals the major neuroinflammatory signaling pathways associated with neurodegeneration. Additionally, it explores promising therapeutic avenues, such as stem cell therapy, genetic intervention, and nanoparticles, aiming to regulate neuroinflammation and potentially impede or decelerate the advancement of these conditions. A comprehensive understanding of the intricate connection between neuroinflammation and these diseases is pivotal for the development of future treatment strategies that can alleviate the burden imposed by these devastating disorders.

编号：JJJM -2024-1-12

引用格式：Kuang S, Zhang S, Cui Z, Ge M, Yuan L, Wang J, Wei Z, Xu J, Zhai F, Liang S. Clinical characteristics and surgical outcomes of low-grade epilepsy-associated brain tumors. Ther Adv Neurol Disord. 2024;17:17562864241237851.

通信作者：梁树立

Abstract

Background: Low-grade epilepsy-associated brain tumors (LEATs) are found to be the second most common lesion-related epilepsy. Malignant potential of LEATs is very low and the overall survival is good, so the focus of treatment is focused more on seizure outcome rather than oncological prognosis. **Objectives:** This study was conducted to evaluate the risk factors of seizure outcomes after resection in patients with LEATs. **Design:** A retrospective study. **Methods:** A retrospective analysis of

patients with LEATs who underwent resective surgery in our three epilepsy centers between October 2010 and April 2023 with a minimum follow-up of 1 year. Demography, clinical characters, neurophysiology, and molecular neuropathology were assessed for association with postoperative seizure outcomes at 1-, 2-, and 5-year follow-up. Synthetic minority oversampling technique (SMOTE) algorithm model was performed to handle the imbalance of data distribution. Gaussian Naïve Bayes (GNB) algorithms were created as a basis for classifying outcomes according to observation indicators. **Results:** A total of 111 patients were enrolled in the cohort. The most common pathology was ganglioglioma (n = 37, 33.3%). The percentage of patients with seizure freedom was 91.0% (101/111) at 1-year follow-up, 87.5% (77/88) at 2-year follow-up, and 79.1% (53/67) at 5-year follow-up. Partial resection had a significantly poor seizure outcome compared to total resection and supratotal resection ($p < 0.05$). The epileptiform discharge on post-resective intraoperative electrocorticography (ECoG) or postoperative scalp electroencephalography (EEG) were negative factors on postoperative seizure freedom at 1-, 2-, or 5-year follow-ups ($p < 0.05$). The area under the receiver-operating characteristic curve value of the GNB-SMOTE model was 0.95 (95% CI, 0.876-1.000), 0.892 (95% CI, 0.656-0.934), and 0.786 (95% CI, 0.491-0.937) at 1-, 2-, and 5-year follow-up, respectively. **Conclusion:** The partial resection, post-resective intraoperative ECoG, and postoperative scalp EEG were valuable indicators of poor seizure outcomes. The utilization of post-resective intraoperative ECoG is beneficial to improve seizure outcomes. Based on the data diversity and completeness of three medical centers, a multivariate correlation analysis model was established based on GNB algorithm.

编号：JJJM -2024-1-13

引用格式：Zhang A, Liu T, Xu J, Zhao Q, Wang X, Jiang Z, Liang S, Cui Y, Li Y. Efficacy of deep brain stimulation for Tourette syndrome and its comorbidities: A meta-analysis. Neurotherapeutics. 2024: e00360.

通信作者：梁树立

Abstract

Tourette Syndrome (TS) is a neurodevelopmental disorder characterized by multiple motor and vocal tics, often accompanied by comorbid disorders. Optional treatments for patients with TS include behavioral therapy, pharmacotherapy, and neurostimulation techniques. Deep brain stimulation (DBS) has been considered a therapeutic approach for refractory TS and its comorbid symptoms. However, systematic comparison is necessary to understand the therapeutic effect of DBS among patients with TS with various comorbid symptoms, demographic characteristics, or stimulation targets. Consequently, our research aimed to assess the clinical efficacy of DBS in alleviating the symptoms of TS and its comorbidities. A systematic literature search was conducted across five databases: PubMed, Web of Science, MEDLINE, Embase, and PsycINFO. The primary outcome was the mean change in the global score of the Yale Global Tic Severity Scale (YGTSS), which assesses the severity of tics. The secondary outcomes included mean improvement of comorbid symptoms, such as obsessive-compulsive behaviors (OCB), depression symptoms and anxiety symptoms. In total, 51 studies with 673 participants were included in this meta-analysis. Overall, the DBS led to a significant improvement in tic symptoms ($p < 0.001$), as well as the comorbid obsessive-compulsive, depression, and anxiety symptoms with effect sizes of 1.88, 0.88, 1.04, and 0.76 accordingly. In the subgroup analysis, we found that striatum stimulation led to a more significant improvement in OCB in patients with TS compared to that observed with thalamic stimulation ($p = 0.017$). The relationship between sex, age, and target with the improvement of tics, depression, and anxiety was not statistically significant ($p = 0.923$, 0.438, 0.591 for different male proportions; $p =$ 0.463, 0.425, 0.105 for different age groups; $p = 0.619$, 0.113, 0.053 for different targets). In conclusion, DBS is an efficient treatment option for TS, as well as the comorbid OCB, depression symptoms, and anxiety symptoms. It is important to highlight that stimulating the striatum is more effective in managing obsessive-compulsive symptoms compared to stimulating the thalamus.

编号：JJJM -2024-1-14

引用格式： **Feng WX, Wang XF, Wu Y, Li XM, Chen SH, Wang XH, Wang ZH,Fang F, Chen CH. Clinical analysis of PAFAH1B1 gene variants in pediatric patients with epilepsy. Seizure. 2024:117:98-104.**

第一作者/通讯作者 冯卫星

Abstract

Purpose: PAFAH1B1, also known as LIS1, is associated with type I lissencephaly in humans, which is a severe developmental brain disorder believed to result from abnormal neuronal migration. Our objective was to characterize the genotypes and phenotypes of PAFAH1B1-related epilepsy.**Methods:**We conducted a comprehensive analysis of the medical histories, magnetic resonance imaging findings, and video-electroencephalogram recordings of 11 patients with PAFAH1B1 variants at the Neurology Department of Beijing Children's Hospital from June 2017 to November 2022.**Results:**The age of onset of epilepsy ranged from 2 months to 4 years, with a median onset age of 5 months. Among these 11 patients (comprising 6 boys and 5 girls), all were diagnosed with lissencephaly type 1. Predominantly, generalized tonic-clonic and spasm seizures characterized PAFAH1B1-related epilepsy. Additionally, 10 out of the 11 patients exhibited severe developmental disorders. All patients exhibited de novo variants, with three individuals displaying 17p13.3 deletions linked to haploinsufficiency of PAFAH1B1. Four variants were previously unreported. Notably, three patients with 17p13.3 deletions displayed developmental delay and drug resistant epilepsy, whereas the single patient with mild developmental delay, Intelligence Quotient (IQ) 57 and well-controlled seizures had a splicing-site variant.**Conclusion:**The severity of the phenotype in patients with PAFAH1B1 variants ranged from drug-responsive seizures to severe epileptic encephalopathy. These observations underscore the clinical heterogeneity of PAFAH1B1-related disorders, with most patients exhibiting developmental disorders. Moreover, the severity of epilepsy appears to be linked to genetic variations.

编号：JJJM -2024-1-15

引用格式：Yao Y, Wang X, Zhao B, Mo J, Guo Z, Yang B, Li Z, Fan X, Cai D, Sang L, Zheng Z, Shao X, Ai L, Hu W, Zhang C, Zhang K. Hypometabolic patterns are related to post-surgical seizure outcomes in focal cortical dysplasia: A semi-quantitative study. Epilepsia Open. 2024;9(2):653-664 .

通信作者：张凯 张弨 胡文瀚

Abstract

Objective: Fluorine-18-fluorodeoxyglucose-positron emission tomography (FDG-PET) is routinely used for presurgical evaluation in many epilepsy centers. Hypometabolic characteristics have been extensively examined in prior studies, but the metabolic patterns associated with specific pathological types of drug-resistant epilepsy remain to be fully defined. This study was developed to explore the relationship between metabolic patterns or characteristics and surgical outcomes in type I and II focal cortical dysplasia (FCD) patients based on results from a large cohort. **Methods:** Data from individuals who underwent epilepsy surgery from 2014 to 2019 with a follow-up duration of over 3 years and a pathological classification of type I or II FCD in our hospital were retrospectively analyzed. Hypometabolic patterns were quantitatively identified via statistical parametric mapping (SPM) and qualitatively analyzed via visual examination of PET-MRI co-registration images. Univariate analyses were used to explore the relationship between metabolic patterns and surgical outcomes. **Results:** In total, this study included data from 210 patients. Following SPM calculations, four hypometabolic patterns were defined including unilobar, multi-lobar, and remote patterns as well as cases where no pattern was evident. In type II FCD patients, the unilobar pattern was associated with the best surgical outcomes ($p = 0.014$). In visual analysis, single gyrus ($p = 0.032$) and Clear-cut hypometabolism edge ($p = 0.040$) patterns exhibited better surgery outcomes in the type II FCD group. **Conclusions:** PET metabolic patterns are well-correlated with the prognosis of type II FCD patients. However, similar correlations were not observed in type I FCD,

potentially owing to the complex distribution of the epileptogenic region. **Plain language summary:** In this study, we demonstrated that FDG-PET was a crucial examination for patients with FCD, which was a common cause of epilepsy. We compared the surgical prognosis for patients with different hypometabolism distribution patterns and found that clear and focal abnormal region in PET was correlated with good surgical outcome in type II FCD patients.

编号：JJJM -2024-1-16

引用格式：Zhao X, Hu X, Guo Z, Hu W, Zhang C, Mo J, Zhang K. Deep Learning Approaches for Imaging-Based Automated Segmentation of Tuberous Sclerosis Complex. J Clin Med. 2024;13(3):680.

通信作者：张凯

Abstract

The present study presents a novel approach for identifying epileptogenic tubers in patients with tuberous sclerosis complex (TSC) and automating tuber segmentation using a three-dimensional convolutional neural network (3D CNN). The study retrospectively included 31 TSC patients whose lesions were manually annotated from multiparametric neuroimaging data. Epileptogenic tubers were determined via presurgical evaluation and stereoelectroencephalography recording. Neuroimaging metrics were extracted and compared between epileptogenic and non-epileptogenic tubers. Additionally, five datasets with different preprocessing strategies were used to construct and train 3D CNNs for automated tuber segmentation. The normalized positron emission tomography (PET) metabolic value was significantly lower in epileptogenic tubers defined via presurgical evaluation ($p = 0.001$). The CNNs showed high performance for localizing tubers, with an accuracy between 0.992 and 0.994 across the five datasets. The automated segmentations were highly correlated with clinician-based features. The neuroimaging characteristics for epileptogenic tubers were demonstrated, increasing surgical confidence in clinical practice. The validated

deep learning detection algorithm yielded a high performance in determining tubers with an excellent agreement with reference clinician-based segmentation. Collectively, when coupled with our investigation of minimal input requirements, the approach outlined in this study represents a clinically invaluable tool for the management of TSC.

编号：JJJM -2024-1-17

引用格式：Yang B, Zhao B, Li C, Mo J, Guo Z, Li Z, Yao Y, Fan X, Cai D, Sang L, Zheng Z, Gao D, Zhao X, Wang X, Zhang C, Hu W, Shao X, Zhang J, Zhang K. Localizing seizure onset zone by a cortico-cortical evoked potentials-based machine learning approach in focal epilepsy. Clin Neurophysiol. 2024;158:103-113.

通信作者：张凯

Abstract

Objective: We aimed to develop a new approach for identifying the localization of the seizure onset zone (SOZ) based on corticocortical evoked potentials (CCEPs) and to compare the connectivity patterns in patients with different clinical phenotypes. **Methods:** Fifty patients who underwent stereoelectroencephalography and CCEP procedures were included. Logistic regression was used in the model, and six CCEP metrics were input as features: root mean square of the first peak (N1RMS) and second peak (N2RMS), peak latency, onset latency, width duration, and area. **Results:** The area under the curve (AUC) for localizing the SOZ ranged from 0.88 to 0.93. The N1RMS values in the hippocampus sclerosis (HS) group were greater than that of the focal cortical dysplasia (FCD) IIa group ($p < 0.001$), independent of the distance between the recorded and stimulated sites. The sensitivity of localization was higher in the seizure-free group than in the non-seizure-free group ($p = 0.036$). **Conclusions:** This new method can be used to predict the SOZ localization in various focal epilepsy phenotypes. Significance: This study proposed a machine-learning approach for localizing the SOZ. Moreover, we examined how clinical phenotypes impact large-scale abnormality of the epileptogenic networks.

编号：JJJM -2024-1-18

引用格式：Li Z, Zhao B, Hu W, Zhang C, Wang X, Liu C, Mo J, Guo Z, Yang B, Yao Y, Shao X, Zhang J, Zhang K. Practical measurements distinguishing physiological and pathological stereoelectroencephalography channels based on high-frequency oscillations in the human brain. Epilepsia Open. 2024 May 29. doi: 10.1002/epi4.12950.

通信作者：张凯

Abstract

Objective: The present study aimed to identify various distinguishing features for use in the accurate classification of stereoelectroencephalography (SEEG) channels based on high-frequency oscillations (HFOs) inside and outside the epileptogenic zone (EZ). **Methods:** HFOs were detected in patients with focal epilepsy who underwent SEEG. Subsequently, HFOs within the seizure-onset and early spread zones were defined as pathological HFOs, whereas others were defined as physiological. Three features of HFOs were identified at the channel level, namely, morphological repetition, rhythmicity, and phase-amplitude coupling (PAC). A machine-learning (ML) classifier was then built to distinguish two HFO types at the channel level by application of the above-mentioned features, and the contributions were quantified. Further verification of the characteristics and classifier performance was performed in relation to various conscious states, imaging results, EZ location, and surgical outcomes. **Results:** Thirty-five patients were included in this study, from whom 166 104 pathological HFOs in 255 channels and 53 374 physiological HFOs in 282 channels were entered into the analysis pipeline. The results revealed that the morphological repetitions of pathological HFOs were markedly higher than those of the physiological HFOs; this was also observed for rhythmicity and PAC. The classifier exhibited high accuracy in differentiating between the two forms of HFOs, as indicated by an area under the curve (AUC) of 0.89. Both PAC and rhythmicity contributed significantly to this distinction. The subgroup analyses supported these findings. **Significance:** The suggested HFO features can accurately distinguish between pathological and physiological channels substantially

improving its usefulness in clinical localization. **Plain language summary:** In this study, we computed three quantitative features associated with HFOs in each SEEG channel and then constructed a machine learning-based classifier for the classification of pathological and physiological channels. The classifier performed well in distinguishing the two channel types under different levels of consciousness as well as in terms of imaging results, EZ location, and patient surgical outcomes.

编号：JJJM -2024-1-19

引用格式：Wu Z, Ren Z, Gao R, Sun K, Sun F, Liu T, Zheng S, Wang W, Zhang G. Impact of subthalamic nucleus deep brain stimulation at different frequencies on neurogenesis in a rat model of Parkinson's disease. Heliyon. 2024;10(10):e30730.

通信作者：翟锋

Abstract

Neurogenesis, play a vital role in neuronal plasticity of adult mammalian brains, and its dysregulation is present in the pathophysiology of Parkinson's disease (PD). While subthalamic nucleus deep brain stimulation (STN-DBS) at various frequencies has been proven effective in alleviating PD symptoms, its influence on neurogenesis remains unclear. This study aimed to investigate the effects of 1-week electrical stimulation at frequencies of 60Hz, 130Hz, and 180Hz on neurogenesis in the subventricular zone (SVZ) of PD rats. A hemiparkinsonian rat model was established using 6-hydroxydopamine and categorized into six groups: control, PD, sham stimulation, 60Hz stimulation, 130Hz stimulation, and 180Hz stimulation. Motor function was assessed using the open field test and rotarod test after one week of STN-DBS at different frequencies. Tyrosine hydroxylase (TH) expression in brain tissue was analyzed via Western blot and immunohistochemistry. Immunofluorescence analysis was conducted to evaluate the expression of BrdU/Sox2, BrdU/GFAP, Ki67/GFAP, and BrdU/DCX in bilateral SVZ and the rostral migratory stream (RMS). Our findings revealed that high-

frequency STN-DBS improved motor function. Specifically, stimulation at 130Hz increased dopaminergic neuron survival in the PD rat model, while significantly enhancing the proliferation of neural stem cells (NSCs) and neuroblasts in bilateral SVZ. Moreover, this stimulation effectively facilitated the generation of new NSCs in the ipsilateral RMS and triggered the emergence of fresh neuroblasts in bilateral RMS, with notable presence within the lesioned striatum. Conversely, electrical stimulation at 60Hz and 180Hz did not exhibit comparable effects. The observed promotion of neurogenesis in PD rats following STN-DBS provides valuable insights into the mechanistic basis of this therapeutic approach for PD.

编号：JJJM -2024-1-20

引用格式：Huang Q, Xie P, Zhou J, Ding H, Liu Z, Li T, Guan Y, Wang M, Wang J, Teng P, Zhu M, Ma K, Wu H, Luan G, Zhai F. Predictors of seizure outcomes in stereo-electroencephalography-guided radio-frequency thermocoagulation for MRI-negative epilepsy. Ther Adv Chronic Dis. 2024;15:20406223241236258.

通信作者：翟锋

Abstract

Background: One-third of intractable epilepsy patients have no visually identifiable focus for neurosurgery based on imaging tests [magnetic resonance imaging (MRI)-negative cases]. Stereo-electroencephalography-guided radio-frequency thermocoagulation (SEEG-guided RF-TC) is utilized in the clinical treatment of epilepsy to lower the incidence of complications post-open surgery. **Objective:** This study aimed to identify prognostic factors and long-term seizure outcomes in SEEG-guided RF-TC for patients with MRI-negative epilepsy. **Design:** This was a single-center retrospective cohort study. **Methods:** We included 30 patients who had undergone SEEG-guided RF-TC at Sanbo Brain Hospital, Capital Medical University, from April 2015 to December 2019. The probability of remaining seizure-free and the plotted survival curves were analyzed. Prognostic factors were analyzed using log-rank

tests in univariate analysis and the Cox regression model in multivariate analysis. **Results:** With a mean time of 31.07 ± 2.64 months (median 30.00, interquartile range: 18.00-40.00 months), 11 out of 30 patients (36.7%) were classified as International League Against Epilepsy class 1 in the last follow-up. The mean time of remaining seizure-free was 21.33 ± 4.55 months [95% confidence interval (CI) 12.41-30.25], and the median time was 3.00 ± 0.54 months (95% CI 1.94-4.06). Despite falling in the initial year, the probability of remaining seizure-free gradually stabilizes in the subsequent years. The patients were more likely to obtain seizure freedom when the epileptogenic zone was located in the insular lobe or with one focus on the limbic system (p = 0.034, hazard ratio 5.019, 95% CI 1.125-22.387). **Conclusion:** Our findings may be applied to guide individualized surgical interventions and help clinicians make better decisions.

编号：JJJM-2023-1-21

引用格式：Chen W, Liu G, Cui L, Tian F, Zhang J, Zhao J, Lv Y, Du J, Huan X, Wu Y, Zhang Y. Evaluation of metagenomic and pathogen-targeted next-generation sequencing for diagnosis of meningitis and encephalitis in adults: A multicenter prospective observational cohort study in China. J Infect. 2024;88(5):106143.

第一作者 陈卫碧

Abstract

Background Next-generation sequencing (NGS) might aid in identification of causal pathogens. However, the optimal approaches applied to cerebrospinal fluid (CSF) for detection are unclear, and studies evaluating application of different NGS workflows for diagnosis of intracranial infections are limited. **Methods** In this multicenter, prospective observational cohort study, we described the diagnostic efficacy of pathogen-targeted NGS (ptNGS) and metagenomic NGS (mNGS) compared to that of composite microbiologic assays, for infectious meningitis/encephalitis (M/E). **Results**

In total, 152 patients diagnosed with clinically suspected M/E at four tertiary hospitals were enrolled; ptNGS and mNGS were used in parallel for pathogen detection in CSF. Among the 89 patients who were diagnosed with definite infectious M/E, 57 and 39 patients had causal microbial detection via ptNGS and mNGS, respectively. The overall accuracy of ptNGS was 65.1%, with a positive percent agreement (PPA) of 64% and a negative percent agreement (NPA) of 66.7%; and the overall accuracy of mNGS was 47.4%, with a PPA of 43.8% and an NPA of 52.4% after discrepancy analysis. There was a significant difference in the detection efficiency between these two methods both for PPA (sensitivity) and overall accuracy for pathogen detection ($P<0.05$). **Conclusions** NGS testings have provided new information in addition to conventional microbiologic tests. ptNGS seems to have superior performance over mNGS for common causative pathogen detection in CSF for infectious M/E.

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第一作者 梁树立

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临床问题 1：病变相关癫痫中，难治性癫痫的比例是否存在差异？

推荐意见 1：病变性质不同，其相关癫痫中难治性癫痫的比例也不同，约 30%～80%为药物难治性癫痫（推荐比例 93.5%，反对比例 0%）。

临床问题 2：病变相关癫痫中，药物治疗无发作的患者是否考虑手术治疗？

推荐意见 2：对于非功能区、单个、局灶性病变相关的癫痫，即使 1～2 种抗癫痫发作药物可以控制，也建议考虑手术治疗。此外也需结合患者的起病年龄、身体状况、手术意愿及对手术预后的期望值综合评估（推荐比例 90.3%，反对比例 6.5%）。

临床问题 3：病变相关癫痫中，不同病变的致痫区如何界定？

推荐意见 3：不同的病变依据其病理学及组织 学特征，各自存在特殊的致痫性及其与致痫区的关系。发育性神经肿瘤的致痫区与结构性病变较为 一致，而脑血管性病变、脑软化的致痫区主要在结 构性病变临近区域，脑胶质瘤中则结构性病变本身及周围区域可能为致痫区（推荐比例 100%，反对比例 0%）。

临床问题 4：对于病变相关癫痫而言，病变区 全切除如何定义？

推荐意见 4：病变区的全切除范围，通常以 MRI-FLAIR 序列显示范围为标准，血管性病变则以 SWI 或 DWI 序列显示范围为标准。术后切除范围的验证应当在 24～72h 内进行 MRI 检查，通过术前术后 MRI 的融合来显示病变区是否完全切除 （推荐比例 87.1%，反对比例 6.5%）。

临床问题 5：病变相关癫痫中，哪些手段可以实现病变全区切除？

推荐意见 5：采用术中导航及超声等技术可提高病变区全切除的概率。根据病变大小及性质的不同，可以采用整块切除和分块切除等不同的手术策略，以提高病变区全切除效果（推荐比例 93.5%，反对比例 3.2%）。

临床问题 6：对于病变相关癫痫而言，病变区超全切除如何定义？

推荐意见 6：癫痫病变区的超全切除应当包括病变及其临近皮质和整个致痫区（推荐比例 87.1%， 反对比例 0%）。

临床问题 7：病变相关癫痫中，哪些手段可以实现病变区超全切除？

推荐意见 7：病变区位于非功能区的患者，病变扩大切除是超全切除的基础，原则下切除范围要到达脑沟。术中皮质脑电图等可以帮助对致痫病变的边界进行确认。颞叶病灶没有明确累及颞叶内侧结构时，应综合结构及功能影像、症状学、神经心理结果来考虑是否对颞叶内侧结构予以切除，必要时需进行颅内电极脑电图监测（推荐比例 96.8%，反对比例 3.2%）。

临床问题 8：开颅癫痫手术中，哪些情况被认为是严重并发症？

推荐意见 8：开颅手术后需非计划二次手术治疗和永久影响脑功能的并发症主要是脑出血、脑梗死、功能区切除相关的功能障碍、症状性脑积水及颅内感染等（推荐比例 100%，反对比例 0%）。

临床问题 9-1：癫痫开颅手术中，如何避免术后感染的发生？一旦发生如何处理？

推荐意见 9：严格消毒和无菌操作，减少开放脑室，严密缝合硬脑膜可以减少术后感染的发生。 可疑感染时早期腰椎穿刺检查明确感染及其病源菌，合理使用抗生素（推荐比例 100%，反对比例 0%）。

临床问题 9-2：癫痫开颅手术中，如何避免颅 内出血的发生？一旦发生如何处理？

推荐意见 10：严格硬膜悬吊、严密止血、减少 术中对脑组织不必要牵拉、严密硬脑膜缝合可以减 少颅内出血的发生。术后出现颅内出血可以通过药物止血、腰椎穿刺治疗；如果血肿量较大，必要时积极行血肿清除和/或去骨瓣减压（推荐比例 100%，反对比例 0%）。

临床问题 9-3：癫痫开颅手术中，如何避免脑梗死的发生？一旦发生如何处理

推荐意见 11：严格显微镜下操作，尽可能保留病变周边的动静脉血管。如果考虑是动脉性脑梗死时，可以应用药物治疗。如果出现大范围脑梗死、严重颅内高压、脑疝等症状时，应当积极进行 去大骨瓣减压（推荐比例 100%，反对比例 0%）。

临床问题 9-4：癫痫开颅手术中，如何避免脑积水的发生？一旦发生如何处理？

推荐意见 12：严密止血，减少开放脑室，反复冲洗术区，减少止血材料应用、严密缝合硬脑膜。术后积极进行腰椎穿刺，释放血性脑脊液，必要时也可以进行腰大池置管引流，减少脑积水可能性。一旦诊断脑积水，早期脱水治疗，存在明显颅高 症状，应当进行脑室-腹腔分流手术（推荐比例 96.8%，反对比例 0%）。

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审稿专家：王小强、毛之奇、田宏、冯卫星、任杰、刘婷红、刘翔宇、杨岸超、吴洵昳、张希、张恒、张春青、陈心、季涛云、孟强、徐硕、徐成伟、高薇、郭燕舞、梁树立、彭伟锋、樊星、操德智

临床问题 1：不同 MCD 相关癫痫中难治性癫痫的比例一样吗？

推荐意见 1：不同 MCD 相关癫痫的分子病理学机制不同，抗癫痫发作药物的选择各异，各自难治性癫痫的比例有所差别，但其中绝大部分都是药物难治的（推荐比例 96.8%，反对比例 0.0%）。

临床问题 2：癫痫患者发现 MCD，一定是致痫灶吗？

推荐意见 2：MCD 是癫痫的重要病因，但影像学发现 MCD 的患者不一定都伴随癫痫发作（推荐比例 100%，反对比例 0.0%）。

临床问题 3-1：FCD 的影像病理灶与致痫灶一致吗？

推荐意见 3：多数 FCD 的影像病理灶与致痫灶一致，其致痫性及范围根据 FCD 亚型的不同而有所差异（推荐比例 96.8%，反对比例 3.2%）。

临床问题 3-2：灰质异位的影像病理灶与致痫灶一致吗？

推荐意见 4：GMH 的致痫起源有单独异位结节起始、单独相关皮质起始及同时起始三种模式，且存在多个异位结节同时受累的情况（推荐比例 96.8%，反对比例 0.0%）。

临床问题 3-3：HH 的影像病理灶与致痫灶一致吗？

推荐意见 5：强迫性不自主发笑发作确认来自于 HH，其他类型发作可能来自于 HH 内或 HH 外，甚至因为继发致痫性成为 HH 外独立起源（推荐比例 100% 反对比例 0.0%）。

临床问题 3-4：其它 MCD 的影像病理灶与致痫灶一致吗？

推荐意见 6：PMG、结节性硬化、脑裂畸形的致痫灶较影像病理灶更为复杂（推荐比例 00%，反对比例 0.0%）。

临床问题 4：对于 FCD 而言，全切除如何定义？

推荐意见 7：全切程度的判断并非以 FCD 病变本身全切为标准，而是评估手术实际切除范围是否涵盖了术前预定的脑区结构（推荐比例 96.8%， 反对比例 0.0%）。

临床问题 6：通过哪些手段能够实现 FCD 的全切除？

推荐意见 9：FCD 全切除的概念至少包含两个层面，即在理论层面上正确界定切除的范围，以及 在技术层面上实现目标区域的完整切除（推荐比例 96.8%，反对比例 0.0%）。

临床问题 7：HH 患者的 SEEG 设计的要点有哪些？

推荐意见 10：电极置入的策略与致痫区范围有关，其核心策略是使 HH 内尽可能多地分布电极触点，并确保电极触点覆盖瘤蒂周围，同时还应考虑是否存在下丘脑以外的致痫区（推荐比例 96.8%，反对比例 0.0%）。

临床问题 8：PNH 患者行 SEEG 时其设计的要点有哪些？

推荐意见 11：成功的 PNH-SEEG 方案是将电极置入假定的致痫结节、周围皮质和直接传播区域，单纯针对结节的置入方案需谨慎（推荐比例 100%，反对比例 0.0%）。

临床问题 10：通过哪些手段可以实现 GMH 全切除？

推荐意见 13：采用电生理监测结合神经导航技术可以提高病理灶全切除的概率，可通过术中对痫性放电的监测指导切除范围的界定，若合并其他脑部异常应当考虑扩大切除范围（推荐比例 93.5%，反对比例 0.0%）。

临床问题 11：通过哪些手段可以实现 MCD 全切除？

推荐意见 14：不同 MCD 的解剖位置和与其 功能联系的皮质范围有所不同，电生理监测及神经导航技术的应用，有助于采取扩大切除的方式以完全切除致痫灶（推荐比例 93.5%，反对比例 0.0%）。

临床问题 12：哪些因素可以影响癫痫术后早期癫痫发作的发生？

推荐意见 15：癫痫患者术后早期癫痫发作主要与起病年龄、病程、发作类型、术前发作频率、手术切除部位、手术方式等有关。围手术期停用抗癫痫发作药物引起血药浓度下降，手术本身对大脑皮层的刺激，围手术期的感染、低血钠、代谢性酸中毒等并发症也可引起术后早期癫痫发作（推荐比例 100%，反对比例 0.0%）。

临床问题 13：如何预防癫痫术后早期癫痫发作？

推荐意见 16：为预防术后早期癫痫的发生，需要避免各种可能诱发因素的发生，及时处理各种并发症，如出血、感染、皮下积液等，预防性使用抗癫痫发作药物（推荐比例 93.5%，反对比例 0.0%）。

临床问题 14：出现癫痫术后早期癫痫发作如何处理？

推荐意见 17：癫痫术后早期发作应积极寻找病因，选择合适的抗癫痫发作药物进行治疗，防止继发性脑损伤（推荐比例 100%，反对比例 0.0%）。

立志而圣则圣矣，立志而贤则贤矣。

——习近平总在五四运动 100 周年上的讲话

典故：王守仁《教条示龙场诸生》

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引用格式：Duan H, Pan C, Wu T, Peng J, Yang L. MT-TN mutations lead to progressive mitochondrial encephalopathy and promotes mitophagy. Biochim Biophys Acta Mol Basis Dis. 2024;1870(4):167043.

通信作者：彭镜

Abstract

Mitochondrial encephalopathy is a neurological disorder caused by impaired mitochondrial function and energy production. One of the genetic causes of this condition is the mutation of MT-TN, a gene that encodes the mitochondrial transfer RNA (tRNA) for asparagine. MT-TN mutations affect the stability and structure of the tRNA, resulting in reduced protein synthesis and complex enzymatic deficiency of the mitochondrial respiratory chain. Our patient cohort manifests with epileptic encephalopathy, ataxia, hypotonia, and bilateral basal ganglia calcification, which differs from previously reported cases. MT-TN mutation deficiency leads to decreased basal and maximal oxygen consumption rates, disrupted spare respiratory capacity, declined mitochondrial membrane potential, and impaired ATP production. Moreover, MT-TN mutations promote mitophagy, a process of selective degradation of damaged mitochondria by autophagy. Excessive mitophagy further leads to mitochondrial biogensis as a compensatory mechanism. In this study, we provided evidence of pathogenicity for two MT-TN mutations, m.5688 T > C and m.G5691A, explored the molecular mechanisms, and summarized the clinical manifestations of MT-TN mutations. Our study expanded the genotype and phenotypic spectrum and provided new insight into mt-tRNA (Asn)-associated mitochondrial encephalopathy.

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引用格式：He H, Li X, Guzman GA, Bungert-Plümke S, Franzen A, Lin X, Zhu

H, Peng G, Zhang H, Yu Y, Sun S, Huang Z, Zhai Q, Chen Z, Peng J, Guzman RE. Expanding the genetic and phenotypic relevance of CLCN4 variants in neurodevelopmental condition: 13 new patients. J Neurol. 2024 May 17.

通讯作者 彭镜

Abstract

Objectives: CLCN4 variations have recently been identified as a genetic cause of X-linked neurodevelopmental disorders. This study aims to broaden the phenotypic spectrum of CLCN4-related condition and correlate it with functional consequences of CLCN4 variants. **Methods**: We described 13 individuals with CLCN4-related neurodevelopmental disorder. We analyzed the functional consequence of the unreported variants using heterologous expression, biochemistry, confocal fluorescent microscopy, patch-clamp electrophysiology, and minigene splicing assay. **Results**: We identified five novel (p.R41W, p.L348V, p.G480R, p.R603W, c.1576 + 5G > A) and three known (p.T203I, p.V275M, p.A555V) pathogenic CLCN4 variants in 13 Chinese patients. The p.V275M variant is found at high frequency and seen in four unrelated individuals. All had global developmental delay (GDD)/intellectual disability (ID). Seizures were present in eight individuals, and 62.5% of them developed refractory epilepsy. Five individuals without seizures showed moderate to severe GDD/ID. Developmental delay precedes seizure onset in most patients. The variants p.R41W, p.L348V, and p.R603W compromise the anion/exchange function of ClC-4. p.R41W partially impairs ClC-3/ClC-4 association. p.G480R reduces ClC-4 expression levels and impairs the heterodimerization with ClC-3. The c.1576 + 5G > A variant causes 22 bp deletion of exon 10. **Conclusions**: We further define and broaden the clinical and mutational spectrum of CLCN4-related neurodevelopmental conditions. The p.V275M variant may be a potential hotspot CLCN4 variant in Chinese patients. The five novel variants cause loss of function of ClC-4. Transport dysfunction, protein instability, intracellular trafficking defect, or failure of ClC-4 to oligomerize may contribute to the pathophysiological events leading to CLCN4-related neurodevelopmental disorder.

编号：XEY-2024-1-3

引用格式：Xie C, Kessi M, Yin F, Peng J. Roles of KCNA2 in Neurological Diseases: from Physiology to Pathology. Mol Neurobiol. 2024. doi: 10.1007/s12035-024-04120-9

通讯作者 彭镜

Abstract

Potassium voltage-gated channel subfamily a member 2 (Kv1.2, encoded by KCNA2) is highly expressed in the central and peripheral nervous systems. Based on the patch clamp studies, gain-of function (GOF), loss-of-function (LOF), and a mixed type (GOF/LOF) variants can cause different conditions/disorders. KCNA2-related neurological diseases include epilepsy, intellectual disability (ID), attention deficit/hyperactive disorder (ADHD), autism spectrum disorder (ASD), pain as well as autoimmune and movement disorders. Currently, the molecular mechanisms for the reported variants in causing diverse disorders are unknown. Consequently, this review brings up to date the related information regarding the structure and function of Kv1.2 channel, expression patterns, neuronal localizations, and tetramerization as well as important cell and animal models. In addition, it provides updates on human genetic variants, genotype-phenotype correlations especially highlighting the deep insight into clinical prognosis of KCNA2-related developmental and epileptic encephalopathy, mechanisms, and the potential treatment targets for all KCNA2-related neurological disorders.

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引用格式：Liao D, Zhong L, Yang L, He F, Deng X, Yin F, Peng J. Clinical and radiological features, treatment responses and prognosis in pediatric patients with co-existing anti-N-methyl-D-aspartate receptor and myelin oligodendrocyte glycoprotein antibody-associated encephalitis: A single center study. Mult Scler

Relat Disord. 2024;81:105133.

通讯作者 彭镜

Abstract

Objectives: To characterize the clinical and radiological features, treatment responses and outcomes of children with co-existing anti-N-methyl-D-aspartate receptor (NMDAR) and myelin oligodendrocyte glycoprotein (MOG) antibody-associated encephalitis. **Methods**: Clinical manifestations, imaging features, effectiveness of treatment and outcomes of patients who were cerebral spinal fluid(CSF)-positive for NMDAR-antibody(NMDAR-ab) and seropositive for MOG-antibody(MOG-ab) were analyzed. **Results**: Twelve patients including 8 females and 4 males were enrolled. The median onset age was 9 years, ranging from 2.2 to 12.8 years. Behavioral changes and/or psychiatric symptoms (n = 8/12), seizures (n = 8/12), encephalopathy (n = 7/12) were 3 of the most common symptoms. Brain magnetic resonance imaging(MRI) of all the patients showed T2/fluid attenuation inversion recovery(FLAIR) abnormal signal in the cerebral white matter at least once in the courses of disease, 2 of whom developed new brain lesions which were asymptomatic. All of the patients had supratentorial lesions. Spinal cord MRI was performed in 7 patients. Only 1 patient showed related abnormalities with increased T2 signal in the spinal cord C1-5. Nine patients underwent optic nerve MRI; 5 patients demonstrated abnormal results, among whom 4 exhibited T2 abnormal signal (2 were symptom-free) and 1 showed a little effusion in bilateral optic nerve sheats. Intravenous immunoglobulin (IVIG) and intravenous methylprednisolone (IVMP) were the most common used therapies in those patients. Nine patients were treated with second-line therapy to prevent relapses. For total 29 clinical attacks, the median modified Rankin Scale (mRS) before treatment and after therapy of acute stage was 1 and 0, respectively. Seven of 12 patients(58.3 %) experienced clinical relapses. In terms of outcome, all of the patients' mRS of last follow-up (≥6 months) was ≤2. **Conclusions**: Behavioral changes and/or psychiatric symptoms, seizures and encephalopathy were common in children with co-existing anti-NMDAR and MOG antibody-associated encephalitis. A minority of subjects may

develop asymptomatic lesions on brain and optic nerve MRI. The relapse rate of this disease is relatively high. The majority of patients responded well to the immunotherapies and had a good outcome(mRS of last follow-up≤2).

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引用格式：Pang N, Li K, Tan S, Chen M, He F, Chen C, Yang L, Zhang C, Deng X, Yang L, Mao L, Wang G, Duan H, Wang X, Zhang W, Guo H, Peng J, Yin F, Xia K. Targeted sequencing identifies risk variants in 202 candidate genes for neurodevelopmental disorders. Gene. 2024;897:148071.

通讯作者 彭镜

Abstract

With the continuous deepening of genetic research on neurodevelopmental disorders (NDDs), more patients have been identified the causal or candidate genes. However, it is still urgent needed to increase the sample size to confirm the associations between variants and clinical manifestations. We previously performed molecular inversion probe sequencing of autism spectrum disorder (ASD) candidate genes in 1543 ASD patients. In this study, we used the same method to detect de novo variants (DNVs) in 665 NDD patients with intellectual disability (ID) and/or epilepsy (EP) for genetic analysis and diagnosis. We compared findings from ID/EP and ASD patients to improve our understanding of different subgroups of NDDs. We identified 72 novel variants and 39 DNVs. A totally of 5.71 % (38/665) of the patients were genetically diagnosed by this sequencing strategy. ID/EP patients demonstrated a higher prevalence of likely gene disruptive DNVs in ASD genes than the healthy population. Regarding high-risk genes, SCN1A and CKDL5 were more frequently mutated in ID/EP patients than in ASD patients. Our data provide an overview of the mutation burden in ID/EP patients from the perspective of high risk ASD genes, indicating the differences and association of NDDs subgroups.

编号：XEY-2024-1-6

引用格式：Zhou C, Xie F, Wang D, Huang X, Guo D, Du Y, Xiao L, Liu D, Xiao B, Yang Z, Feng L. Preoperative structural-functional coupling at the default mode network predicts surgical outcomes of temporal lobe epilepsy. Epilepsia. 2024;65(4):1115-1127.

通讯作者 冯莉

Abstract

Objective: Structural-functional coupling (SFC) has shown great promise in predicting postsurgical seizure recurrence in patients with temporal lobe epilepsy (TLE). In this study, we aimed to clarify the global alterations in SFC in TLE patients and predict their surgical outcomes using SFC features. **Methods:** This study analyzed presurgical diffusion and functional magnetic resonance imaging data from 71 TLE patients and 48 healthy controls (HCs). TLE patients were categorized into seizure-free (SF) and non-seizure-free (nSF) groups based on postsurgical recurrence. Individual functional connectivity (FC), structural connectivity (SC), and SFC were quantified at the regional and modular levels. The data were compared between the TLE and HC groups as well as among the TLE, SF, and nSF groups. The features of SFC, SC, and FC were categorized into three datasets: the modular SFC dataset, regional SFC dataset, and SC/FC dataset. Each dataset was independently integrated into a cross-validated machine learning model to classify surgical outcomes. **Results:** Compared with HCs, the visual and subcortical modules exhibited decoupling in TLE patients ($p < .05$). Multiple default mode network (DMN)-related SFCs were significantly higher in the nSF group than in the SF group ($p < .05$). Models trained using the modular SFC dataset demonstrated the highest predictive performance. The final prediction model achieved an area under the receiver operating characteristic curve of .893 with an overall accuracy of 0.887. **Significance:** Presurgical hyper-SFC in the DMN was strongly associated with postoperative seizure recurrence. Furthermore, our results introduce a novel SFC-based machine learning model to precisely classify the surgical outcomes of TLE.

编号：XEY-2024-1-7

引用格式：Xiao L, Yang J, Zhu H, Zhou M, Li J, Liu D, Tang Y, Feng L*, Hu S*. [18F]SynVesT-1 and [18F]FDG quantitative PET imaging in the presurgical evaluation of MRI-negative children with focal cortical dysplasia type II. Eur J Nucl Med Mol Imaging. 2024 May;51(6):1651-1661.

通讯作者 冯莉

Abstract

Purpose: MRI-negative children with focal cortical dysplasia type II (FCD II) are one of the most challenging cases in surgical epilepsy management. We aimed to utilize quantitative positron emission tomography (QPET) analysis to complement [18F]SynVesT-1 and [18F]FDG PET imaging and facilitate the localization of epileptogenic foci in pediatric MRI-negative FCD II patients. **Methods:** We prospectively enrolled 17 MRI-negative children with FCD II who underwent [18F]SynVesT-1 and [18F]FDG PET before surgical resection. The QPET scans were analyzed using statistical parametric mapping (SPM) with respect to healthy controls. The sensitivity, specificity, positive predictive value (PPV), negative predictive value (NPV), and area under the curve (AUC) of [18F]SynVesT-1 PET, [18F]FDG PET, [18F]SynVesT-1 QPET, and [18F]FDG QPET in the localization of epileptogenic foci were assessed. Additionally, we developed a multivariate prediction model based on dual trace PET/QPET assessment. **Results:** The AUC values of [18F]FDG PET and [18F]SynVesT-1 PET were 0.861 (sensitivity = 94.1%, specificity = 78.2%, PPV = 38.1%, NPV = 98.9%) and 0.908 (sensitivity = 82.4%, specificity = 99.2%, PPV = 93.3%, NPV = 97.5%), respectively. [18F]FDG QPET showed lower sensitivity (76.5%) and NPV (96.6%) but higher specificity (95.0%) and PPV (68.4%) than visual assessment, while [18F]SynVesT-1 QPET exhibited higher sensitivity (94.1%) and NPV (99.1%) but lower specificity (97.5%) and PPV (84.2%). The multivariate prediction model had the highest AUC value (AUC = 0.996, sensitivity = 100.0%, specificity = 96.6%, PPV = 81.0%, NPV = 100%). **Conclusions:** The multivariate

prediction model based on [18F]SynVesT-1 and [18F]FDG PET/QPET assessments holds promise in noninvasively identifying epileptogenic regions in MRI-negative children with FCD II. Furthermore, the combination of visual assessment and QPET may improve the sensitivity and specificity of diagnostic tests in localizing epileptogenic foci and achieving a preferable surgical outcome in MRI-negative FCD II.

编号：XEY-2024-1-8

引用格式：Tang Y, Xiao L, Deng C, Zhu H, Gao X, Li J, Yang Z, Liu D, Feng L, Hu S. [^{F}]FDG PET metabolic patterns in mesial temporal lobe epilepsy with different pathological types. Eur Radiol. 2024;34(2):887-898.

通讯作者 冯莉

Abstract

Objectives: To investigate [18F]FDG PET patterns of mesial temporal lobe epilepsy (MTLE) patients with distinct pathologic types and provide possible guidance for predicting long-term prognoses of patients undergoing epilepsy surgery. **Methods:** This was a retrospective review of MTLE patients who underwent anterior temporal lobectomy between 2016 and 2021. Patients were classified as having chronic inflammation and gliosis (gliosis, n = 44), hippocampal sclerosis (HS, n = 43), or focal cortical dysplasia plus HS (FCD-HS, n = 13) based on the postoperative pathological diagnosis. Metabolic patterns and the severity of metabolic abnormalities were investigated among MTLE patients and healthy controls (HCs). The standardized uptake value (SUV), SUV ratio (SUVr), and asymmetry index (AI) of regions of interest were applied to evaluate the severity of metabolic abnormalities. Imaging processing was performed with statistical parametric mapping (SPM12). **Results:** With a mean follow-up of 2.8 years, the seizure freedom (Engel class IA) rates of gliosis, HS, and FCD-HS were 54.55%, 62.79%, and 69.23%, respectively. The patients in the gliosis group presented a metabolic pattern with a larger involvement of extratemporal areas,

including the ipsilateral insula. SUV, SUVr, and AI in ROIs were decreased for patients in all three MTLE groups compared with those of HCs, but the differences among all three MTLE groups were not significant. **Conclusions:** MTLE patients with isolated gliosis had the worst prognosis and hypometabolism in the insula, but the degree of metabolic decrease did not differ from the other two groups. Hypometabolic regions should be prioritized for [18F]FDG PET presurgical evaluation rather than [18F]FDG uptake values. **Clinical relevance statement:** This study proposes guidance for optimizing the operation scheme in patients with refractory MTLE and emphasizes the potential of molecular neuroimaging with PET using selected tracers to predict the postsurgical histology of patients with refractory MTLE epilepsy.

编号：XEY-2024-1-9

引用格式：Huang K, Tian Z, Zhang Q, Yang H, Wen S, Feng J, Tang W, Wang Q, Feng L. Reduced eye gaze fixation during emotion recognition among patients with temporal lobe epilepsy. J Neurol. 2024;271(5):2560-2572.

通讯作者 冯莉

Abstract

Objectives: To investigate the facial scan patterns during emotion recognition (ER) through the dynamic facial expression task and the awareness of social interference test (TASIT) using eye tracking (ET) technology, and to find some ET indicators that can accurately depict the ER process, which is a beneficial supplement to existing ER assessment tools. **Method:** Ninety-six patients with TLE and 88 healthy controls (HCs) were recruited. All participants watched the dynamic facial expression task and TASIT including a synchronized eye movement recording and recognized the emotion (anger, disgust, happiness, or sadness). The accuracy of ER was recorded. The first fixation time, first fixation duration, dwell time, and fixation count were selected and analyzed. **Results:** TLE patients exhibited ER impairment especially for disgust ($Z = -3.391$; $p = 0.001$) and sadness ($Z = -3.145$; $p = 0.002$). TLE patients fixated less on the face, as

evidenced by the reduced fixation count (Z = - 2.549; p = 0.011) of the face and a significant decrease in the fixation count rate (Z = - 1.993; p = 0.046). During the dynamic facial expression task, TLE patients focused less on the eyes, as evidenced by the decreased first fixation duration (Z = - 4.322; p = 0.000), dwell time (Z = - 4.083; p = 0.000), and fixation count (Z = - 3.699; p = 0.000) of the eyes. **Conclusion:** TLE patients had ER impairment, especially regarding negative emotions, which may be attributable to their reduced fixation on the eyes during ER, and the increased fixation on the mouth could be a compensatory effect to improve ER performance. Eye-tracking technology could provide the process indicators of ER, and is a valuable supplement to traditional ER assessment tasks.

编号：XEY-2024-1-10

引用格式：Wu Z, Li X, Huang Y, Huang K, Xiao B, Chi Y, Feng L*, Yang H*. Effects of a Nurse-Led Cognitive Behavioral Intervention for Parents of Children With Epilepsy. Pediatr Neurol. 2024;154:70-78.

通讯作者 冯莉

Abstract

Background: This study aimed to evaluate the effects of a nurse-led cognitive behavioral intervention for parents of children with epilepsy (CWE). **Methods:** The study recruited 238 CWE from the neurology ward of Xiangya Hospital from March 2019 to August 2022. According to the interventions after discharge, the children and their parents were randomly divided into 117 parent-child dyads in the intervention group and 121 parent-child dyads in the control group. The seizure severity and treatment compliance in CWE as well as the parents' psychological states and satisfaction with the care provided by nurses were compared before and after intervention. **Results:** The follow-up six months after discharge showed that the seizure frequency among CWE in the intervention group was significantly less than the controls (P = 0.048). Compared with the controls, the intervention group also reported fewer

symptoms of anxiety and depression, better sleep quality, and more positive attitudes toward epilepsy, as well as higher nursing satisfaction ($P < 0.001$). The correlation analysis indicated the correlation of CWE's seizure severity was correlated with the compliance, parents' psychological states, and parents' satisfaction with the care provided by nurses. **Conclusions:** The adoption of the nurse-led cognitive behavioral intervention on parents of CWE can improve the parents' mental health status and their satisfaction with the nurses, which can have a positive association with the seizure severity of CWE. In light of this information, this nursing intervention may be a new method for the long-term disease management of CWE.

编号：XEY-2024-1-11

引用格式：Liu Y, Yang H, Gan S, He L, Zeng R, Xiao T, Wu L. A novel mutation of DNA2 regulates neuronal cell membrane potential and epileptogenesis. Cell Death Discov. 2024;10(1):259.

通讯作者 吴丽文

Abstract

Mesial temporal lobe epilepsy (MTLE) is one of the most intractable epilepsies. Previously, we reported that mitochondrial DNA deletions were associated with epileptogenesis. While the underlying mechanism of mitochondrial DNA deletions during epileptogenesis remain unknown. In this study, a novel somatic mutation of DNA2 gene was identified in the hippocampal tissue of two MTLE patients carrying mitochondrial DNA deletions, and this mutation decreased the full-length expression of DNA2 protein significantly, aborting its normal functions. Then, we knocked down the DNA2 protein in zebrafish, and we demonstrated that zebrafish with DNA2 deficiency showed decreased expression of mitochondrial complex II-IV, and exhibited hallmarks of epileptic seizures, including abnormal development of the zebrafish and epileptiform discharge signals in brain, compared to the Cas9-control group. Moreover, our cell-based assays showed that DNA2 deletion resulted in accumulated

mitochondrial DNA damage, abnormal oxidative phosphorylation and decreased ATP production in cells. Inadequate ATP generation in cells lead to declined Na+, K+-ATPase activity and change of cell membrane potential. Together, these disorders caused by DNA2 depletion increased cell apoptosis and inhibited the differentiation of SH-SY5Y into branched neuronal phenotype. In conclusion, DNA2 deficiency regulated the cell membrane potential via affecting ATP production by mitochondria and Na+, K+-ATPase activity, and also affected neuronal cell growth and differentiation. These disorders caused by DNA2 dysfunction are important causes of epilepsy. In summary, we are the first to report the pathogenic somatic mutation of DNA2 gene in the patients with MTLE disease, and we uncovered the mechanism of DNA2 regulating the epilepsy. This study provides new insight into the pathogenesis of epilepsy and underscore the value of DNA2 in epilepsy.

编号：XEY-2024-1-12

引用格式：Haiyan Yang, Hongmei Liao, Siyi Gan, Ting Xiao, Liwen Wu. A novel splicing variant in MICAL-1 gene is associated with epilepsy. Eur J Med Genetic. 2024;104946

通讯作者 吴丽文

Abstract

Germline MICAL1 defects have been rarely reported in patients with epilepsy and the genotype-phenotype association remains unclear. In this study, the patient was a 4.6 years old girl who presented with onset of recurrent focal seizures with onset at age 3.4 years. EEG showed abnormal δ-wave activity in the right central and middle temporal lobe. Trio WES showed a novel heterozygous variant c.-43-1G > A in the MICAL1 gene in the patient and her normal mother. Minigene verified two abnormal transcripts due to the mutation, which was predicted to interrupt 5'UTR structures of MICAL1. The patient was clinically diagnosed with benign childhood epilepsy with centrotemporal spike (BECTS). As far as we know, this is the first BECTS case with

documented MICAL1 mutation. Novel MICAL1 variant c.-43-1G > A putatively interrupted MICAL1 translation by changing 5'UTR structures and, however, further functioning study is needed.

编号：XEY-2024-1-13

引用格式：Zhao S, Lian R, Jin L, Li M, Jia T, Xu F, Du K, Wang L, Guo Q, Dong Y. Clinical and genetic analysis of infants with pontocerebellar hypoplasia type 6 caused by RARS2 variations. Epilepsia Open. 2024;9(1):250-257.

通讯作者 董燕

Abstract

Objective: Defects in RARS2 cause cerebellopontine hypoplasia type 6 (pontocerebellar hypoplasia type 6, PCH6, OMIM: #611523), a rare autosomal recessive inherited mitochondrial disease. Here**,** we report two male patients and their respective family histories. **Methods:** We describe the clinical presentation and magnetic resonance imaging (MRI) findings of these patients. Whole-exome sequencing was used to identify the genetic mutations. **Results:** One patient showed hypoglycemia, high lactic acid levels (fluctuating from 6.7 to 14.1 mmol/L), and frequent seizures after birth, with progressive atrophy of the cerebrum, cerebellum, and pons. The other patient presented with early infantile developmental and epileptic encephalopathies (EIDEEs) with an initial developmental delay followed by infantile epileptic spasm syndrome (IESS) at 5 months old, with no imaging changes. Whole-exome sequencing identified compound heterozygous RARS2 variants c.25A>G (p.I9V) with c.1261C>T (p.Q421*) and c.1A>G (p.M1V) with c.122A>G (p.D41G) in these two patients. Of these loci, c.1261C>T and c.122A>G have not been previously reported. **Significance:** Our findings have expanded the RARS2 gene variant spectrum and present EIDEEs and IESS as phenotypes which deepened the association between PCH6 and RARS2. **Plain language summary:** Defects in RARS2 cause cerebellopontine hypoplasia type 6, a rare autosomal recessive inherited mitochondrial

disease. Two patients with RARS2 variants were reported in this article. One patient showed hypoglycemia, high lactic acid levels, and frequent seizures after birth, with progressive atrophy of the cerebrum, cerebellum, and Page 3 of 21 Epilepsia OpenFor Review Only pons. The other patient presented with an initial developmental delay followed by refractory epilepsy at 5 months old, with no imaging changes. Our findings deepened the association between PCH6 and RARS2.

编号：XEY-2024-1-14

引用格式：Zhang K, Yao H, Yang J, Jia T, Shan Q, Li D, Li M, Gan L, Wang X, Dong Y. Analysis of clinical characteristics and histopathological transcription in 40 patients afflicted by epilepsy stemming from focal cortical dysplasia. Epilepsia Open. 2024 Jun;9(3):981-995. doi: 10.1002/epi4.12928.

通讯作者 董燕

Abstract

Objective: This study aims to comprehensively analyze the clinical characteristics and identify the differentially expressed genes associated with drug-resistant epilepsy (DRE) in patients with focal cortical dysplasia (FCD). **Methods:** A retrospective investigation was conducted from July 2019 to June 2022, involving 40 pediatric cases of DRE linked to FCD. Subsequent follow-ups were done to assess post-surgical outcomes. Transcriptomic sequencing and quantitative reverse transcription polymerase chain reaction (qRT-PCR) were used to examine differential gene expression between the FCD and control groups. **Results:** Among the 40 patients included in the study, focal to bilateral tonic-clonic seizures (13/40, 32.50%) and epileptic spasms (9/40, 22.50%) were the predominant seizure types. Magnetic resonance imaging (MRI) showed frequent involvement of the frontal (22/40, 55%) and temporal lobes (12/40, 30%). In cases with negative MRI results (13/13, 100%), positron emission tomography/computed tomography (PET-CT) scans revealed hypometabolic lesions. Fused MRI/PET-CT images demonstrated lesion reduction in 40.74% (11/27) of cases

compared with PET-CT alone, while 59.26% (16/27) yielded results consistent with PET-CT findings. FCD type II was identified in 26 cases, and FCD type I in 13 cases. At the last follow-up, 38 patients were prescribed an average of 1.27 ± 1.05 anti-seizure medications (ASMs), with two patients discontinuing treatment. After a postoperative follow-up period of 23.50 months, 75% (30/40) of patients achieved Engel class I outcome. Transcriptomic sequencing and qRT-PCR analysis identified several genes primarily associated with cilia, including CFAP47, CFAP126, JHY, RSPH4A, and SPAG1. **Significance:** This study highlights focal to bilateral tonic-clonic seizures as the most common seizure type in patients with DRE due to FCD. Surgical intervention primarily targeted lesions in the frontal and temporal lobes. Patients with FCD-related DRE showed a promising prognosis for seizure control post-surgery. The identified genes, including CFAP47, CFAP126, JHY, RSPH4A, and SPAG1, could serve as potential biomarkers for FCD. **Plain language summary:** This study aimed to comprehensively evaluate the clinical data of individuals affected by focal cortical dysplasia and analyze transcriptomic data from brain tissues. We found that focal to bilateral tonic-clonic seizures were the most prevalent seizure type in patients with drug-resistant epilepsy. In cases treated surgically, the frontal and temporal lobes were the primary sites of the lesions. Moreover, patients with focal cortical dysplasia-induced drug-resistant epilepsy exhibited a favorable prognosis for seizure control after surgery. CFAP47, CFAP126, JHY, RSPH4A, and SPAG1 have emerged as potential pathogenic genes for the development of focal cortical dysplasia.

编号：XEY-2024-1-14

引用格式：Zhao T, Zhang X, Cui X, Chen Y, Wang N, Bin Wang, Ren Z, Sun L, Zhao P, Xu J, Han X. Awareness, attitudes and first aid knowledge of epilepsy among university students - A cross-sectional study in Henan Province, China. Epilepsy Res. 2024;201:107315.

第一作者 赵婷

Abstract

Purpose: Epilepsy is a debilitating disease that can lead to series of social and psychological issues, impairing the quality of life of people with epilepsy (PWE). This survey aimed to investigate the awareness, attitudes, and firstaid knowledge of epilepsy in university students. **Method:** This cross-sectional study was conducted in Henan Province, China between January 1 and April 30, 2022. Students majored in education, medicine, science and engineering from 8 universities attended the study. The survey questionnaire comprised 28 questions covering 4 sections: demographic characteristics, awareness of epilepsy, attitudes toward PWE and knowledge of first aid for seizures. **Results**: A total of 2376 university students completed the questionnaire. 94.7% heard of epilepsy. In the first aid knowledge section, individual question was correctly answered by at least 50% students, 9.3% students correctly answered all questions. Attitude toward PWE was independently ($R2 = 0.108$, $F = 73.227$, $p < 0.001$) associated with both awareness of epilepsy ($B = 0.411$, $p < 0.001$) and first aid knowledge of epilepsy ($B = 0.047$, $p = 0.001$). Among the three majors, medical students had more positive attitudes toward PWE than students majored in education, science and engineering ($p < 0.05$). However, medical students performed worse among the groups when answering the first aid knowledge questions. **Conclusion**: This survey showed that university students in Central China had a good awareness of epilepsy. For medical students, improvements are necessary for the awareness of the first aid knowledge for seizure.

编号：XEY-2024-1-16

引用格式：Zhao T, Zhang X, Cui X, Su S, Li L, Chen Y, Wang N, Sun L, Zhao J, Zhang J, Han X, Cao J. Inhibiting the IRAK4/NF-κB/NLRP3 signaling pathway can reduce pyroptosis in hippocampal neurons and seizure episodes in epilepsy. Exp Neurol. 2024;377:114794.

第一作者 赵婷

Abstract

Background: Interleukin-1 receptor-associated kinase 4 (IRAK4) plays an important role in immune modulation in various central nervous system disorders. However, IRAK4 has not been reported in epilepsy models in animal and clinical studies, nor has its involvement in regulating pyroptosis in epilepsy. **Method:** First, we performed transcriptome sequencing, quantitative real-time polymerase chain reaction, and western blot analysis on the hippocampal tissues of refractory epilepsy patients to measure the mRNA and protein levels of IRAK4 and pyroptosis-related proteins. Second, we successfully established a pentylenetetrazol (PTZ)-induced seizure mouse model. We conducted behavioral tests, electroencephalography, virus injection, and molecular biology experiments to investigate the role of IRAK4 in seizure activity regulation. **Results:** IRAK4 is upregulated in the hippocampus of epilepsy patients and PTZ-induced seizure model mice. IRAK4 expression is observed in the hilar neurons of PTZ-induced mice. Knocking down IRAK4 in PTZ-induced mice downregulated pyroptosis-related protein expression and alleviated seizure activity. Overexpressing IRAK4 in naive mice upregulated pyroptosis-related protein expression and increased PTZ-induced abnormal neuronal discharges. IRAK4 and NF-κB were found to bind to each other in patient hippocampal tissue samples. Pyrrolidine dithiocarbamate reversed the pyroptosis-related protein expression increase caused by PTZ. PF-06650833 alleviated seizure activity and inhibited pyroptosis in PTZ-induced seizure mice. **Conclusion:** IRAK4 plays a key role in the pathological process of epilepsy, and its potential mechanism may be related to pyroptosis mediated by the NF-κB/NLRP3 signaling pathway. PF-06650833 has potential as a therapeutic agent for alleviating epilepsy.

编号：XEY-2024-1-17

引用格式：Zhao T, Zhang X, Cui X, Su S, Li L, Chen Y, Wang N, Sun L, Zhao J,

Zhang J, Han X, Cao J. Oridonin exerts anticonvulsant profile and neuroprotective activity in epileptic mice by inhibiting NLRP3-mediated pyroptosis. Int Immunopharmacol. 2024;134:112247.

第一作者 赵婷

Abstract

Background: Epilepsy is a chronic disabling disease poorly controlled by available antiseizure medications. Oridonin, a bioactive alkaloid with anti-inflammatory properties and neuroprotective effects, can inhibit the increased excitability of neurons caused by glutamate accumulation at the cellular level. However, whether oridonin affects neuronal excitability and whether it has antiepileptic potential has not been reported in animal models or clinical studies. **Method:** Pentylenetetrazol was injected into mice to create a model of chronic epilepsy. Seizure severity was assessed using the Racine scale, and the duration and latency of seizures were observed. Abnormal neuronal discharge was detected using electroencephalography, and neuronal excitability was assessed using calcium imaging. Damage to hippocampal neurons was evaluated using Hematoxylin-Eosin and Nissl staining. The expression of the NOD-like receptor thermal protein domain associated protein 3 (NLRP3) inflammasome and other pyroptosis-related proteins was determined using western blotting and immunofluorescence. A neuronal pyroptosis model was established using the supernatant of BV2 cells treated with lipopolysaccharide and adenosine triphosphate to stimulate hippocampal neurons. **Results:** Oridonin (1 and 5 mg/kg) reduced neuronal damage, increased the latency of seizures, and shortened the duration of fully kindled seizures in chronic epilepsy model mice. Oridonin decreased abnormal discharge during epileptic episodes and suppressed increased neuronal excitability. In vitro experiments showed that oridonin alleviated pyroptosis in hippocampal HT22 neurons. **Conclusion***:* Oridonin exerts neuroprotective effects by inhibiting pyroptosis through the NLRP3/caspase-1 pathway in chronic epilepsy model mice. It also reduces pyroptosis in hippocampal neurons in vitro, suggesting its potential as a therapy for epilepsy.

编号：XEY-2024-1-18

引用格式：Li S, Yi J, Tuo Y, Nie G, Wang J, Wang Y, Sun D, Liu Z. Population pharmacokinetics and dosing optimization of perampanel in children with epilepsy: A real-world study. Epilepsia. 2024;65(6):1687-1697.

通讯作者 孙丹

Abstract

Objective: The purposes of this study were to explore the pharmacokinetics of perampanel (PER) in children with epilepsy, identify factors that contribute to pharmacokinetic variations among subjects, evaluate the connection between PER exposure and clinical outcome, and establish an evidence-based approach for tailoring individualized antiepileptic treatment in this specific population. **Methods:** In this prospective study, PER plasma concentrations and genetic information on metabolic enzymes were obtained from 194 patients younger than 18 years. The disposition kinetics of PER in pediatric patients following oral dosing were characterized using nonlinear mixed effect models. The effective range for the plasma concentration of PER was determined by assessing the efficacy and safety of PER treatment and analyzing the relationship between drug exposure and clinical response. Monte Carlo simulations were then performed to evaluate and optimize the current dosing regimens. **Results:** The pharmacokinetic profile of PER was adequately described by a onecompartment model with first-order absorption and elimination. Body weight, total bilirubin level, and concomitant oxcarbazepine were found to have significant influences on PER pharmacokinetics. Model estimates of apparent clearance and volume of distribution were .016±.009L/h/kg and 1.47±.78L/kg, respectively. The effective range predicted from plasma concentration data in responders was 215–862μg/L. Dosing scenarios stratified according to essential covariates were proposed through simulation analysis.

编号：XEY-2024-1-19

引用格式：Huang S, Hu C, Zhong M, Li Q, Dai Y, Ma J, Qin J, Sun D. Clinical phenotypes of developmental and epileptic encephalopathy-related recurrent KCNH5 missense variant p.R327H in Chinese children. Epilepsy Behav Rep. 2024;26:100671.

通讯作者 孙丹

Abstract

KCNH5 gene encodes for the voltage-gated potassium channel protein Kv10.2. Here, we investigated the clinical features of developmental and epileptic encephalopathy (DEE) in five Chinese pediatric patients with a missense mutation (p.R327H) in KCNH5 gene. These patients had undergone video EEG to evaluate background features and epileptiform activity, as well as 3.0 T MRI scans for structural analysis and intelligence assessments using the Gesell Developmental Observation or Wechsler Intelligence Scale for Children. Seizure onset occurs between 4 and 10 months of age, with focal and generalized tonic-clonic seizures being common. Initial EEG findings showed multiple multifocal sharp waves, sharp slow waves or spike slow waves, and spike waves. Brain MRI revealed widened extracerebral space in only one patient. Mechanistically, the KCNH5 mutation disrupts the two hydrogen bonds between Arg327 and Asp304 residues, potentially altering the protein's structural stability and function. Almost 80 % of patients receiving add-on valproic acid (VPA) therapy experienced a reduction in epileptic seizure frequency. Altogether, this study presents the first Chinese cohort of pediatric DEE patients with the KCNH5 p.R327H mutation, highlighting focal seizures as the predominant seizure type and incomplete mutation penetrance. Add-on VPA therapy was likely effective in the early stages of DEE pathogenesis.

编号：XEY-2024-1-20

引用格式：Wang J, Mei Y, Liang S, Li SC, Chen C, Nie G, Tuo YL, Sun D, Wang Y. How to handle a missed or delayed dose of lacosamide in pediatric patients with epilepsy? a mode-informed individual dosing. Epilepsy Behav. 2024;151:109601.

通讯作者 孙丹

Abstract

This study aims to investigate the effects on the pharmacokinetic (PK) of lacosamide (LCM), and to guide the individual dosing regimens for children and ones with poor medication adherence. Population PK research was performed based on 164 plasma samples of 113 pediatric patients aged from 1.75 to 14.42 years old. The PK characteristic of LCM was developed by a one-compartment model with first-order elimination. The typical value of apparent clearance (CL) and apparent volume of distribution (Vd) was 1.91 L·h− 1 and 56.53 L respectively. In the final model, the variability of CL was significantly associated with the body surface area (BSA) and elevated uric acid (UA) level. In contrast, the impact of some prevalent anti-seizure medicines, such as valproic acid, levetiracetam, oxcarbazepine, lamotrigine, and perampanel, and gene polymorphisms of Cytochrome P450 (CYP)2C19, ATP-binding cassette (ABC)B1, and ABCC2 had no clinical significance on the PK parameters of LCM. BSA-based dosing regimen of LCM was provided according to Monte Carlo simulation approach; while the dosage should reduce half in patients with an UA level of more than 400 μmol·L-1 comparing with an UA level of 100 μmol·L-1. Individualize remedial doses of about 0.5- to 1.5-fold of regular doses were recommended in six common scenarios of missed or delayed doses, that depended on the delayed time. In current study, the population PK model of LCM in children with epilepsy was developed successfully. The BSA-based dosing regimen and individualized remedial strategy were recommended to guarantee the precise administration of LCM.

编号：XEY-2024-1-21

引用格式：Jiao L, Kang H, Geng Y, Liu X, Wang M, Shu K. The role of the nucleus basalis of Meynert in neuromodulation therapy: a systematic review from the perspective of neural network oscillations. Front Aging Neurosci. 2024;16:1376764.

第一作者 康慧聪

Abstract

As a crucial component of the cerebral cholinergic system and the Papez circuit in the basal forebrain, dysfunction of the nucleus basalis of Meynert (NBM) is associated with various neurodegenerative disorders. However, no drugs, including existing cholinesterase inhibitors, have been shown to reverse this dysfunction. Due to advancements in neuromodulation technology, researchers are exploring the use of deep brain stimulation (DBS) therapy targeting the NBM (NBM-DBS) to treat mental and neurological disorders as well as the related mechanisms. Herein, we provided an update on the research progress on cognition-related neural network oscillations and complex anatomical and projective relationships between the NBM and other cognitive structures and circuits. Furthermore, we reviewed previous animal studies of NBM lesions, NBM-DBS models, and clinical case studies to summarize the important functions of the NBM in neuromodulation. In addition to elucidating the mechanism of the NBM neural network, future research should focus on to other types of neurons in the NBM, despite the fact that cholinergic neurons are still the key target for cell type-specific activation by DBS.

编号：XEY-2024-1-22

引用格式：Zhou Q, Zhang N, Wang M, Zhao Q, Zhu S, Kang H. Adenosine kinase gene modified mesenchymal stem cell transplantation retards seizure severity and associated cognitive impairment in a temporal lobe epilepsy rat model. Epilepsy Res. 2024;200:107303.

通讯作者 康慧聪

Abstract

Purpose: Temporal lobe epilepsy (TLE) has a high risk of developing drug resistant and cognitive comorbidities. Adenosine has potential anticonvulsant effects as an inhibitory neurotransmitter, but drugs targeting its receptors and metabolic enzyme has inevitable side effects. Therefore, we investigated adenosine augmentation therapy for seizure control and cognitive comorbidities in TLE animals. Methods: Using lentiviral vectors coexpressing miRNA inhibiting the expression of adenosine kinase (ADK), we produced ADK$^-$-rMSC (ADK knockdown rat mesenchymal stem cell). ADK$^-$-rMSC and LV-con-rMSC (rMSC transduced by randomized scrambled control sequence) were transplanted into the hippocampus of TLE rat respectively. ADK$^-$+DPCPX group was transplanted with ADK$^-$-rMSC and intraperitoneally injected with DPCPX (adenosine A1 receptor antagonist). Seizure behavior, EEG, CA1 pyramidal neuron apoptosis, and behavior in Morris water maze and novel object recognition test were studied. **Results:** Adenosine concentration in the supernatants of 105 ADK$^-$-rMSCs was 13.8 ng/ml but not detectable in LV-con-rMSCs. ADK$^-$-rMSC ($n = 11$) transplantation decreased spontaneous recurrent seizure (SRS) duration compared to LV-con-rMSC ($n = 11$, $P < 0.05$). CA1 neuron apoptosis was decreased in ADK$^-$-rMSC ($n = 3$, $P < 0.05$). ADK--rMSC ($n = 11$) improved the Morris water maze performance of TLE rats compared to LV-con-rMSC ($n = 11$, escape latency, $P < 0.01$; entries in target quadrant, $P < 0.05$). The effect of ADK$^-$-rMSC on neuron apoptosis and spatial memory were counteracted by DPCPX. However, ADK$^-$-rMSC didn't improve the performance in novel object recognition test. **Conclusion:** Adenosine augmentation-based ADK$^-$-rMSC transplantation is a promising therapeutic candidate for TLE and related cognitive comorbidities.

编号：XEY-2024-1-23

引用格式：Wu X, Shi M, Chen Y, Lian Y, Fang S, Zhang H. Effect and Mechanism

of LIN28 on Ferroptosis in Mg2+-free Rat Hippocampal Neuron Model of Epilepsy. Neurochem Res. 2024;49(7):1655-1664.

通讯作者 张海峰

Abstract

Studies have demonstrated that LIN28 is expressed in the CNS and may exert protective effects on neurons. However, it remains unknown whether LIN28 regulates ferroptosis in the context of epilepsy. In this study, we established an epilepsy model by culturing hippocampal neurons from rats in a magnesium-free (Mg2+-free) medium. In Mg2+-depleted conditions, hippocampal neurons exhibited reduced LIN28 expression, heightened miR-142-5p expression, decreased glutathione peroxidase (GPX) activity and expression, elevated levels of reactive oxygen species (ROS) and malondialdehyde (MDA), resulting in a significant decline in cell viability and an increase in ferroptosis. Conversely, overexpression of LIN28 reversed these trends in the mentioned indices. Altogether, this study reveals that LIN28 may exert neuroprotective effects by inhibiting the miR-142-5p expression and suppressing ferroptosis in hippocampal neurons induced by Mg2+-free via increasing GPX4 expression.

编号：XEY-2024-1-24

引用格式：Wu X, Zhang H, Shi M, Fang S. Clinical features in antiglycine receptor antibody-related disease: a case report and update literature review. Front. Immunol. 2023; 15:1387591.

通讯作者 张海峰

Abstract

Background and objectives: Antiglycine receptor (anti-GlyR) antibody mediates multiple immune-related diseases. This study aimed to summarize the clinical features to enhance our understanding of anti-GlyR antibody-related disease. **Methods:** By collecting clinical information from admitted patients positive for glycine receptor (GlyR) antibody, the clinical characteristics of a new patient positive for GlyR antibody

were reported in this study. To obtain additional information regarding anti-GlyR antibody-linked illness, clinical data and findings on both newly reported instances in this study and previously published cases were merged and analyzed. **Results:** A new case of anti-GlyR antibody-related progressive encephalomyelitis with rigidity and myoclonus (PERM) was identified in this study. A 20-year-old man with only positive cerebrospinal fluid anti-GlyR antibody had a good prognosis with first-line immunotherapy. The literature review indicated that the common clinical manifestations of anti-GlyR antibody-related disease included PERM or stiff-person syndrome (SPS) (n = 179, 50.1%), epileptic seizure (n = 94, 26.3%), and other neurological disorders (n = 84, 24.5%). Other neurological issues included demyelination, inflammation, cerebellar ataxia and movement disorders, encephalitis, acute psychosis, cognitive impairment or dementia, celiac disease, Parkinson’s disease, neuropathic pain and allodynia, steroid-responsive deafness, hemiballism/tics, laryngeal dystonia, and generalized weakness included respiratory muscles. The group of PERM/SPS exhibited a better response to immunotherapy than others. **Conclusions:** The findings suggest the presence of multiple clinical phenotypes in anti-GlyR antibody-related disease. Common clinical phenotypes include PERM, SPS, epileptic seizure, and paraneoplastic disease. Patients with PERM/SPS respond well to immunotherapy.

道阻且长，行则将至

——习近平在中国北京世界园艺博览会开幕式上的讲话

典故：《诗经·蒹葭》

编号：CYDQZ-2024-1-1

引用格式：Xia Y, Lai W, Sha L, Duan Y, Chen L. Causal link between oxidative stress and epilepsy: A two-sample Mendelian randomization study. Brain Behav. 2024;14(6):e3549.

通讯作者 陈蕾

Abstract

Background: Although a growing body of research has indicated a strong link between oxidative stress and epilepsy, the exact nature of their interaction remains elusive. To elucidate this intricate relationship, we conducted a bidirectional Mendelian randomization (MR) analysis employing two independent datasets. **Methods:** A two-sample MR analysis was performed using instrumental variables derived from genome-wide association study summary statistics of oxidative stress injury biomarkers (OSIB) and epilepsy. The OSIBs were selected from eight primary metabolic pathways associated with oxidative stress. Additionally, seven distinct epilepsy phenotypes were considered, which encompassed all epilepsy, generalized epilepsy, generalized tonic-clonic seizures, focal epilepsy, focal epilepsy with hippocampal sclerosis (focal HS), focal epilepsy with lesions other than HS (focal NHS), and lesion-negative focal epilepsy. Causal estimates were computed using the inverse-variance weighted method or the Wald ratio method, and the robustness of causality was assessed through sensitivity analyses. **Results:** For OSIB and epilepsy, 520 and 23 genetic variants, respectively, were selectively extracted as instrumental variants. Genetically predicted higher kynurenine level was associated with a decreased risk of focal epilepsy (odds ratio [OR] 1.950, 95% CI 1.373-2.528, p = .023) and focal NHS (OR 1.276, 95% CI 1.100-1.453, p = .006). For reverse analysis, there was a suggestive effect of focal NHS on urate (OR 1.19 × 1015, 95% CI 11.19 × 1015 to 1.19 × 1015, p = .0000746) and total bilirubin (Tb) (OR 4.98, 95% CI 3.423-6.543, p = .044). In addition, genetic predisposition to focal HS was associated with higher Tb levels (OR 9.83, 95% CI 7.77-11.888, p = .034). **Conclusion:** This MR study provides compelling evidence of a

robust association between oxidative stress and epilepsy, with a notable emphasis on a causal relationship between oxidative stress and focal epilepsy. Additional research is warranted to confirm the connection between oxidative stress and the risk of epilepsy and to unravel the underlying mechanisms.

编号：CYDQZ-2024-1-2

引用格式：Ji S, Dong B, Tang Y, Li H, Lai W, Li Y, Chen Y, Peng A, Chen L. Therapeutic value of patent foramen ovale closure for drug-resistant epilepsy: A case series report. Epilepsia Open. 2024; doi: 10.1002/epi4.12960.

通讯作者 陈蕾

Abstract

Background: Closure surgery of patent foramen ovale (PFO) has been found to effectively control cryptogenic stroke and migraine, but it is uncertain whether PFO closure could also alleviate epileptic seizures. This study aims to observe the therapeutic effect of PFO closure on epileptic seizures. **Methods:** Since July 11th, 2017, in the neurology department of West China Hospital, Sichuan University, Chengdu, we have been regularly monitoring patients with epilepsy who have undergone PFO closure. The patient's clinical information, such as frequency, duration, and severity of seizures, before and after surgery was recorded in detail as well as postoperative safety events. **Results:** Of the 31 epilepsy patients who confirmed PFO observed (27 cases were drug-resistant epilepsy, 87.10%), average age of surgery was 23.74 years, and 12 cases were female (38.71%). After one-year follow-up, 26 patients (83.87%) achieved remission of seizure frequency, and 22 of whom (70.97%) experienced a remission of more than 50%. Additionally, compared to before surgery, 22 cases (70.97%) reported a decrease in the average seizure duration, and 20 cases (64.52%) reported a reduction in seizure severity. In the seizure indicators of frequency, average duration and severity, significant differences were identified between preoperative and postoperative comparisons with all test p values were <0.05. Furthermore, no serious safety events

were reported except for one patient who briefly reported chest pain, and all patients expressed effective PFO closure. **Significance:** The PFO closure has been shown for the first time to result in a significant reduction in the frequency, duration, and severity of seizures. Patients with drug-resistant epilepsy and PFO with a large shunt are ideal candidates for undergoing PFO closure. **Plain language summary:** Since PFO closure was found to have a good therapeutic effect on cryptogenic stroke and migraine, it has become a credible complementary therapy for the treatment of neurological diseases, and drug-resistant epilepsy with PFO is expected to become the next target disease that PFO closure could significantly improve.

编号：CYDQZ-2024-1-3

引用格式：Xia Y, Duan Y, Sha L, Lai W, Zhang Z, Hou J, Chen L. Whole-cycle management of women with epilepsy of child-bearing age: ontology construction and application. BMC Med Inform Decis Mak. 2024;24(1):101.

通讯作者 陈蕾

Abstract

Background: The effective management of epilepsy in women of child-bearing age necessitates a concerted effort from multidisciplinary teams. Nevertheless, there exists an inadequacy in the seamless exchange of knowledge among healthcare providers within this context. Consequently, it is imperative to enhance the availability of informatics resources and the development of decision support tools to address this issue comprehensively. **Materials and methods:** The development of the Women with Epilepsy of Child-Bearing Age Ontology (WWECA) adhered to established ontology construction principles. The ontology's scope and universal terminology were initially established by the development team and subsequently subjected to external evaluation through a rapid Delphi consensus exercise involving domain experts. Additional entities and attribute annotation data were sourced from authoritative guideline documents and specialized terminology databases within the respective field.

Furthermore, the ontology has played a pivotal role in steering the creation of an online question-and-answer system, which is actively employed and assessed by a diverse group of multidisciplinary healthcare providers. **Results:** WWECA successfully integrated a total of 609 entities encompassing various facets related to the diagnosis and medication for women of child-bearing age afflicted with epilepsy. The ontology exhibited a maximum depth of 8 within its hierarchical structure. Each of these entities featured three fundamental attributes, namely Chinese labels, definitions, and synonyms. The evaluation of WWECA involved 35 experts from 10 different hospitals across China, resulting in a favorable consensus among the experts. Furthermore, the ontology-driven online question and answer system underwent evaluation by a panel of 10 experts, including neurologists, obstetricians, and gynecologists. This evaluation yielded an average rating of 4.2, signifying a positive reception and endorsement of the system's utility and effectiveness. **Conclusions:** Our ontology and the associated online question and answer system hold the potential to serve as a scalable assistant for healthcare providers engaged in the management of women with epilepsy (WWE). In the future, this developmental framework has the potential for broader application in the context of long-term management of more intricate chronic health conditions.

编号：CYDQZ-2024-1-4

引用格式：Duan Y, Yang X, Zhang M, Qi X, Jin Y, Wang Z, Chen L. Adaptive Dosage Strategy of Levetiracetam in Chinese Epileptic Patients: Focus on Pregnant Women. J Pharm Sci. 2024;113(5):1385-1394.

通讯作者 陈蕾

Abstract

There is presently no efficient dose individualization strategy for the use of antiseizure medications in epileptic pregnant patients. This study aimed to develop a population pharmacokinetics model for levetiracetam and propose a tailored adaptive

individualized dosage strategy for epileptic pregnant patients. A total of 322 levetiracetam plasma concentrations from 238 patients with epilepsy were included, including 216 women with epilepsy (20.83% of whom were pregnant). The levetiracetam plasma concentration was measured using a validated ultra-performance liquid chromatography-tandem mass spectrometry assay, and the data were modeled using a nonlinear mixed-effects model. The resultant model served as the basis for simulating the dosage adjustment strategy. A one-compartment model with first-order elimination best described the pharmacokinetic data of levetiracetam. The apparent clearance (CL/F) was 3.43 L/h (95% CI 3.30-3.56) and the apparent volume of distribution was 43.7 L (95% CI 40.4-47.0) for a typical individual of 57.2 kg. Pregnancy and body weight were found to be significant covariates of CL/F of levetiracetam. The recommended regimen of levetiracetam could be predicted by the population pharmacokinetic model based on body weight, gestational age, and the daily dose of levetiracetam taken before pregnancy.

编号：CYDQZ-2024-1-5

引用格式：Cao Y, Li H, Chen M, Wang P, Shi F, Zhu X, Peng A, Li S, Chen L. Evaluation and systematic review of guidance documents for status epilepticus. Epilepsy Behav. 2024;150:109555.

通讯作者 陈蕾

Abstract

Guidance documents play a pivotal role in shaping the management of status epilepticus (SE). However, the methodological quality of these documents remains uncertain. In this systematic review, we comprehensively searched 12 literature and guideline databases to assess the quality of clinical practice guidelines and consensus statements related to SE management using the AGREE II methodology. Additionally, we summarized the associated recommendations. We identified a total of 14 clinical practice guidelines and 11 consensus statements spanning the period from 1993 to 2022.

The median score for clarity of presentation was 71.8% (ranging from 15.3% to 91.7%), indicating generally good clarity. However, the aspect of editorial independence received poor ratings, with a median score of 32.1% (ranging from 0% to 83.3%). Notably, the 2016 guideline published by the American Epilepsy Society in Epilepsy (AES) received the highest overall scores. Across these guidance documents, there was consistency in the definition and diagnosis of SE. However, significant variability was observed in therapeutic recommendations, particularly in terms of the timing for adding or changing medications. The methodological approaches used in most SE guidance documents require improvement, and the disparities in recommendations highlight existing gaps in evidence. Enhanced methodological rigor results in increased standardization of the guideline, consequently augmenting its reference value. Given the urgency of SE as an emergency condition, it is imperative that these documents also address relevant management strategies before admission.

编号：CYDQZ-2024-1-6

引用格式：Lai W, Wu Y, Sha L, Lai Q, Yang X, Ai F, Zhang Q, Bu F, He S, Zhu X, Chen L. Identifying Genetic Factors of Polycystic Ovary Syndrome in Women with Epilepsy: A Whole-Genome Sequencing Study. Neuroendocrinology. 2024;114(3):223-233.

通讯作者 陈蕾

Abstract

Background: Women with epilepsy (WWE) are more likely to develop reproductive endocrine disorders, especially polycystic ovary syndrome (PCOS). This study aimed to explore the genetic factors of PCOS in WWE in hope of improving individual precision diagnosis and treatment. **Methods:** WWE registered at West China Hospital between January 2022 and October 2022 were enrolled in this study. Demographic and epilepsy-related characteristics were recorded, and blood samples were collected for hormones, glucose metabolism testing, and whole-genome sequencing. **Results:** After

sample sequencing, quality control, and variants selection, association analyses were performed. Pathway analysis was performed to identify involved biological pathways. The overall and PCOS "burden score" of each individual were calculated to count the deleterious variants. A total of 95 WWE were included in this study and 19 patients were diagnosed with PCOS. WWE with PCOS showed a significantly different hormone profiles and a tendency of impaired glucose metabolism. The most commonly associated genes were ZFYVE28, COL19A1, SIK3, ANKK1, PPIG, and REPIN1. The top 3 canonical pathways are adipogenesis pathway, epoxysqualene biosynthesis signaling, and glutamate degradation signaling. The most significant common variant was rs11914038 located in gene CELSR1 and rs651748 located in gene ZBTB16. In human gene connectome prioritizations, ITGA9, PNPLA2, and DAB2 are the top 3 genes having the shortest distance to known PCOS genes. **Conclusion:** Genetic factors involved in the abnormal regulation of glucose and insulin metabolism are likely to be associated with the comorbidity of PCOS in WWE. Interventions targeting these processes should be given more priority in clinical practice.

编号：CYDQZ-2024-1-7

引用格式：Dai L, Huang J, Shen KF, Yang XL, Zhu G, Zhang L, Wang ZK, Liu SY, Liao X, Xu SL, Yang H, Li XY, Zhang CQ. Altered expression of the Plexin-B2 system in tuberous sclerosis complex and focal cortical dysplasia IIb lesions. Histol Histopathol. 2024 Jan 10:18707.

通讯作者 张春青

Abstract

Tuberous sclerosis complex (TSC) and focal cortical dysplasia (FCD) type IIb are the predominant causes of drug-refractory epilepsy in children. Dysmorphic neurons (DNs), giant cells (GCs), and balloon cells (BCs) are the most typical pathogenic profiles in cortical lesions of TSC and FCD IIb patients. However, mechanisms underlying the pathological processes of TSC and FCD IIb remain obscure. The Plexin-B2-Sema4C

signalling pathway plays critical roles in neuronal morphogenesis and corticogenesis during the development of the central nervous system. However, the role of the Plexin-B2 system in the pathogenic process of TSC and FCD IIb has not been identified. In the present study, we investigated the expression and cell distribution characteristics of Plexin-B2 and Sema4C in TSC and FCD IIb lesions with molecular technologies. Our results showed that the mRNA and protein levels of Plexin-B2 expression were significantly increased both in TSC and FCD IIb lesions versus that in the control cortex. Notably, Plexin-B2 was also predominantly observed in GCs in TSC epileptic lesions and BCs in FCD IIb lesions. In contrast, the expression of Sema4C, the ligand of Plexin-B2, was significantly decreased in DNs, GCs, and BCs in TSC and FCD IIb epileptic lesions. Additionally, Plexin-B2 and Sema4C were expressed in astrocytes and microglia cells in TSC and FCD IIb lesions. Furthermore, the expression of Plexin-B2 was positively correlated with seizure frequency in TSC and FCD IIb patients. In conclusion, our results showed the Plexin-B2-Sema4C system was abnormally expressed in cortical lesions of TSC and FCD IIb patients, signifying that the Plexin-B2-Sema4C system may play a role in the pathogenic development of TSC and FCD IIb.

编号：CYDQZ-2024-1-8

引用格式：Qin N, Cao Q, Li F, Wang W, Peng X, Wang L. A nomogram based on quantitative EEG to predict the prognosis of nontraumatic coma patients in the neuro-intensive care unit. Intensive Crit Care Nurs. 2024;83:103618.

通讯作者 彭希

Abstract

Objective: We aimed to establish a quantitative electroencephalography-based prognostic prediction model specifically tailored for nontraumatic coma patients to guide clinical work. **Methods:** This retrospective study included 126 patients with nontraumatic coma admitted to the First Affiliated Hospital of Chongqing Medical

University from December 2020 to December 2022. Six in-hospital deaths were excluded. The Glasgow Outcome Scale assessed the prognosis at 3 months after discharge. The least absolute shrinkage and selection operator regression analysis and stepwise regression method were applied to select the most relevant predictors. We developed a predictive model using binary logistic regression and then presented it as a nomogram. We assessed the predictive effectiveness and clinical utility of the model. **Results:** After excluding six deaths that occurred within the hospital, a total of 120 patients were included in this study. Three predictor variables were identified, including APACHE II score [39.129 (1.4244-1074.9000)], sleep cycle [OR: 0.006 (0.0002-0.1808)], and RAV [0.068 (0.0049-0.9500)]. The prognostic prediction model showed exceptional discriminative ability, with an AUC of 0.939 (95 % CI: 0.899-0.979).Conclusion: A lack of sleep cycles, smaller relative alpha variants, and higher APACHE II scores were associated with a poor prognosis of nontraumatic coma patients in the neurointensive care unit at 3 months after discharge. **Clinical implication:** This study presents a novel methodology for the prognostic assessment of nontraumatic coma patients and is anticipated to play a significant role in clinical practice.

编号：CYDQZ-2024-1-9

引用格式：Wu X, Qin N, Peng X, Wang L. Exploring odontogenic brain abscesses: a comprehensive review. Acta Neurol Belg. 2024. doi: 10.1007/s13760-024-02569-y.

通讯作者 彭希

Abstract

Introduction: Whether in neurology or dentistry, odontogenic brain abscess stands as an ailment demanding undivided attention. The onset of this disease is insidious, with a relatively low incidence rate but a markedly high fatality rate. Moreover, its symptoms lack specificity, easily leading to misdiagnosis, oversight, and treatment delays. Hence,

clinicians should maintain heightened vigilance when faced with pathogenic bacteria of dental origin in patients. **Areas covered:** This paper encapsulates the latest research findings on the clinical manifestations and essential treatment points of odontogenic brain abscess. It may offer a crucial reference for prompt diagnosis and improved therapeutic approaches. **Expert opinions:** Odontogenic brain abscess, an infection of the cerebral parenchyma, usually appears in immunocompromised patients with dental ailments or postdental surgeries. The main pathogenic microorganisms include Streptococcus intermedius, Fusobacterium nucleatum, Streptococcus anginosus, and Millerella. Given the undetectable and nonspecific symptoms in patients, the diagnostic process relies on microbiological methods. Therefore, clinicians should actively investigate and identify the pathogenic microorganisms of odontogenic brain abscess for early detection and selection of appropriate treatment regimens to avoid disease management delays.

编号：CYDQZ-2024-1-10

引用格式：Li R, Xiong Y, Pan S, Lei W, Shu X, Shi X, Tian M. Role of TRAK1 variants in epilepsy: genotype-phenotype analysis in a pediatric case of epilepsy with developmental disorder. Front Mol Neurosci, 2024;17, doi:10.3389/fnmol.2024.1342371

通讯作者 田茂强

Abstract

Purpose: The TRAK1 gene is mapped to chromosome 3p22.1 and encodes trafficking protein kinesin binding 1. The aim of this study was to investigate the genotype–phenotype of TRAK1-associated epilepsy. **Methods:** Trio-based whole-exome sequencing was performed on a cohort of 98 patients with epilepsy of unknown etiologies. Protein modeling and the VarCards database were used to predict the damaging effects of the variants. Detailed neurological phenotypes of all patients with epilepsy having TRAK1 variants were analyzed to assess the genotype–phenotype

correlations. **Results:** A novel TRAK1 compound heterozygous variant comprising variant c.835C > T, p.Arg279Cys and variant c.2560A > C, p.Lys854Gln was identified in one pediatric patient. Protein modeling and VarCards database analyses revealed that the variants were damaging. The patient received a diagnosis of early infantile epileptic spasms with a developmental disorder; he became seizure free through valproate and adrenocorticotropic hormone treatment. Further results for six variants in 12 patients with epilepsy indicated that biallelic TRAK1 variants (including homozygous or compound heterozygous variants) were associated with epilepsy with developmental disorders. Among these patients, eight (67%) had epileptic spasms and seven (58%) were intractable to anti-seizure medicines. Moreover, eight patients experienced refractory status epilepticus, of which seven (88%) died in early life. To our knowledge, this is the first reported case of epilepsy caused by TRAK1 compound heterozygous variants. **Conclusion:** Biallelic TRAK1 variants can cause epilepsy and developmental disorders. In these patients, seizures progress to status epilepticus, suggesting a high risk for poor outcomes and the requirement of early treatment.

编号：CYDQZ-2024-1-11

引用格式：Zhou X, Yu Yang, Zhenzhen Tai, Haiqing Zhang, Juan Yang, Zhong Luo, Zucai Xu. The mechanism of mitochondrial autophagy regulating Clathrin-mediated endocytosis in epilepsy. Epilepsia Open. 2024; doi: 10.1002/epi4.12945

通讯作者 徐祖才

Abstract

Objective: To determine whether inhibition of mitophagy affects seizures through Clathrin-mediated endocytosis (CME). **Methods:** Pentylenetetrazol (PTZ) was intraperitoneally injected daily to establish a chronic PTZ-kindled seizure. The Western blot (WB) was employed to compare the differences in Parkin protein expression between the epilepsy group and the control group. Immunofluorescence was employed to detect the expression of MitoTracker and LysoTracker. Transferrin-

Alexa488 (Tf-A488) was injected into the hippocampus of mice. We evaluated the effect of 3-methyladenine (3-MA) on epilepsy behavior through observation in PTZ-kindled models. **Results:** The methylated derivative of adenine, known as 3-MA, has been extensively utilized in the field of autophagy research. The transferrin protein is internalized from the extracellular environment into the intracellular space via the CME pathway. Tf-A488 employs a fluorescent marker to track CME. Western blot showed that the expression of Parkin was significantly increased in the PTZ-kindled model ($P < 0.05$), while 3-MA could reduce the expression ($P < 0.05$). The fluorescence uptake of MitoTracker and LysoTracker was increased in the primary cultured neurons induced by magnesium-free extracellular fluid ($P < 0.05$); the fluorescence uptake of Tf-A488 was significantly decreased in the 3-MA group compared with the control group ($P < 0.05$). Following hippocampal injection of Tf-A488, both the epilepsy group and the 3-MA group exhibited decreased fluorescence uptake, with a more pronounced effect observed in the 3-MA group. Inhibition of mitophagy by 3-MA from d 3 to d 9 progressively exacerbated seizure severity and shortened latency.

编号：CYDQZ-2024-1-12

引用格式：Li C, Wang Z, Ren M, Ren S, Wu G, Wang L. Synaptic vesicle protein 2A mitigates parthanatos via apoptosis-inducing factor in a rat model of pharmacoresistant epilepsy. CNS Neurosci Ther. 2024;30(5):e14778.

通讯作者：王丽琨

Abstract

Aims: Synaptic vesicle protein 2A (SV2A) is a unique therapeutic target for pharma1 coresistant epilepsy (PRE). As seizure-induced neuronal programmed death, part1 hanatos was rarely reported in PRE. Apoptosis-inducing factor (AIF), which has been implicated in parthanatos, shares a common cytoprotective function with SV2A. We aimed to investigate whether parthanatos participates in PRE and is mitigated by SV2A

via AIF. **Methods:** An intraperitoneal injection of lithium chloride-pilocarpine was used to establish an epileptic rat model, and phenytoin and phenobarbital sodium were utilized to select PRE and pharmacosensitive rats. The expression of SV2A was manipulated via lentivirus delivery into the hippocampus. Video surveillance was used to assess epileptic ethology. Biochemical tests were employed to test hippocampal tissues following a successful SV2A infection. Molecular dynamic calculations were used to simulate the interaction between SV2A and AIF. **Results:** Parthanatos core index, PARP1, PAR, nuclear AIF and MIF, γ-H2AX, and TUNEL staining were all increased in PRE. SV2A is bound to AIF to form a stable complex, successfully inhibiting AIF and MIF nuclear translocation and parthanatos and consequently mitigating spontaneous recurrent seizures in PRE. Moreover, part 1 hanatos deteriorated after the SV2A reduction. Significance: SV2A protected hippocampal neurons and mitigated epileptic seizures by inhibiting parthanatos via binding to AIF in PRE.

编号：CYDQZ-2024-1-13

引用格式：Zheng Q, Cheng YR, Wang M, Ma X, Ye L, Xu Z, Feng Z. COVID-19 vaccinations for patients with epilepsy in Guizhou Province, China: A cross-sectional study. Heliyon. 2024;10(7):e29354.

通讯作者 冯占辉

Abstract

Several COVID-19 vaccines have been approved for emergency use according to China's immunization programs. These vaccines has created hope for patients with epilepsy, because the vaccines can help to reduce their risk of becoming infected with the severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2). The aim of this study was to investigate the COVID-19 vaccine safety in patients with epilepsy. Here, we assessed the time of symptom control and the features of adverse events of seizure patients following their COVID-19 vaccinations. The results showed that adverse

events of COVID-19 vaccinations for epilepsy patients included local pain at the injection site, dizziness and headache, epileptic attack, somnolence, limb weakness, limb pain, allergy, and fever. In addition, the average recovery time of the adverse events was approximately 42 h. More importantly, our study showed that it was relatively safe to vaccinate epilepsy patients who did not experience seizures for approximately 12 months prior to the immunization date.

编号：CYDQZ-2024-1-14

引用格式：Zeng Q, Xia XQ, Jiang L, Chen J, Liu YH, Hu Y. Efficacy and safety of adjunctive perampanel treatment in pediatric patients with epilepsy aged 4−12 years: a real-world study. J Neurol. 2024; doi：10.1007/s00415-024-12416-y.

通讯作者 胡越

Abstract

Objective: To determine the efficacy and safety of perampanel (PER) as an adjunctive therapy in children aged 4–12 years with epilepsy. **Methods:** We performed a non-randomized, open-label, placebo-uncontrolled, real-world self-controlled study that included 216 young children (aged 4–12 years) with epilepsy who received PER as adjunctive therapy at the children's hospital affiliated with Chongqing Medical University from July 4, 2020, to September 20, 2023. **Results:** (1) The efficacy rates of adjunctive PER therapy at 3, 6, 9, and 12 months were 62.8%, 67.8%, 65.3%, and 61.2%, respectively. PER showed efficacy in alleviating focal seizures, generalized tonic–clonic seizures, myoclonic seizures, and absence seizures. The efficacy rates for variants of self-limited epilepsy with centrotemporal spikes (SeLECTS) and Lennox-Gastaut syndrome (LGS) were 89.5% and 66.7%, respectively. (2) Focal non-motor onset seizures with or without impaired awareness, focal to bilateral tonic–clonic seizures (FBTCS), LGS, variants of SeLECTS, the number of concomitant antiseizure medications (ASMs), a family history of epilepsy, and focal lesions on cranial magnetic

resonance imaging were independent factors affecting efficacy. The order of PER addition did not affect efficacy. The retention rates at 3, 6, 9, and 12 months were 90.7%, 84.7%, 74.7%, 64.9%, respectively. (3) Adverse reactions occurred in 45 patients (45/216, 20.8%), with irritability/aggressive behavior (18/216, 8.3%) and somnolence (14/216, 6.5%) being the most common. Twelve patients (12/216, 5.6%) withdrew from the study because of adverse reactions. **Conclusion:** In young Chinese children with epilepsy, PER is effective, safe, and well-tolerated as an adjunctive therapy, making it a viable option for use with broad-spectrum ASM

编号：CYDQZ-2024-1-15

引用格式：Yi YJ, Zhang SM, Dai JL, Zheng H, Peng XL, Cheng L, Chen HS, Hu Y. MiR-23b-3p Improves Brain Damage after Status Epilepticus by Reducing the Formation of Pathological High-Frequency Oscillations via Inhibition of cx43 in Rat Hippocampus. ACS Chemical Neuroscience. 2024; doi:10.1021/acschemneuro.4c00112.

通讯作者 胡越

Abstract

In order to investigate the effectiveness and safety of miR-23b-3p in anti-seizure activity and to elucidate the regulatory relationship between miR-23b-3p and Cx43 in the nervous system, we have established a lithium chloride-pilocarpine (PILO) status epilepticus (SE) model. Rats were randomly divided into the following groups: seizure control (PILO), valproate sodium (VPA+PILO), recombinant miR-23b-3p overexpression (miR+PILO), miR-23b-3p sponges (Sponges+PILO), and scramble sequence negative control (Scramble+PILO) (n = 6/group). After experiments, we got the following results. In the acute phase, the time required for rats to reach stage IV after PILO injection was significantly longer in VPA+PILO and miR+PILO. In the chronic phase after SE, the frequency of spontaneous recurrent seizures (SRSs) in VPA+PILO and miR+PILO was significantly reduced. At 10 min before seizure

cessation, the average energy expression of fast ripples (FRs) in VPA+PILO and miR+PILO was significantly lower than in PILO. After 28 days of seizure, Cx43 expression in PILO was significantly increased, and Beclin1expression in all groups was significantly increased. After 28 days of SE, the numbers of hippocampal necrotic cells and synaptic structures in the hippocampal CA3 region in VPA+PILO and miR+PILO were significantly lower and higher than in PILO,respectively. There were no significant differences in biochemical indicators among the experimental group rats 28 days after SE compared to the seizure control group. Based on the previous facts, we can reach the conclusion that MiR-23b-3p targets and blocks the expression of hippocampal Cx43 which can reduce the formation of pathological FRs, thereby alleviating the severity of seizures, improving seizure-induced brain damage.

编号：CYDQZ-2024-1-16

引用格式：Liu J, Lin H, Wang D, Chen N, Li T. Distinct contribution of monocarboxylate transporter 2 to infantile epileptic spasms syndrome. Med Hypoth. 2024;188: 111359.

通讯作者：李听松

Abstract

Infantile epileptic spasms syndrome (IESS) is the most common refractory epileptic encephalopathy in early brain development, its pathogenesis remains elusive. In the adult brain, glucose serves as the predominant metabolic fuel. However, ketone bodies and lactate are more important in neural energy metabolism during early development. This has been further provided by the effectiveness of ketogenic diet (KD) therapy in IESS. When the circulatory pool of ketone bodies is increased by KD, the brain utilizes ketone bodies preferentially to meet the high energy demand of neurons which is beneficial to mitigate seizure activity and promote prognosis for children with IESS. Neuronal monocarboxylate transporter 2 (MCT2) is crucial in this process, transporting ketone bodies and lactate to mitochondria for adenosine triphosphate (ATP) generation

for cellular energy. The inhibition of MCT2 has been linked to mitochondrial dysfunction and its reduction has been found in adult animal epilepsy models. The mitochondrial energy metabolism disorder is a recognized central pathological aspect of epileptogenesis. Therefore, we hypothesize that neuronal MCT2, an essential gatekeeper of energy metabolism, may play a critical role in the genesis and propagation of spasms in IESS by markedly affecting brain metabolic homeostasis and mitochondrial function. We propose to conduct animal studies and clinical studies to investigate the relationship among MCT2, severity of IESS, susceptibility of IESS, and mitochondrial dysfunction from a metabolic perspective.

编号：CYDQZ-2024-1-17

引用格式：Yang J, Chen C, Chen N, Zheng H, Chen Y, Li X, Jia Q, Li T. Clinical characteristics and rehabilitation potential in children with cerebral palsy based on MRI classification system. Front Pediatr. 2024;12:1382172.

通讯作者：李听松

Abstract

Background: The correlation of clinical characteristics of cerebral palsy (CP) and the magnetic resonance imaging classification system (MRICS) for (CP) is inconsistent. Specifically, the variance in rehabilitation potential across MRICS remains underexplored. **Aims:** To investigate the clinical characteristics and potential for rehabilitation in children with CP based on MRICS. Materials and methods: Children with CP admitted to the Department of Rehabilitation, Children's Hospital of Chongqing Medical University between 2017 and 2021 were included in the study. Qualified cases underwent a follow-up period of at least one year. The clinical characteristics of CP among different MRICS were analyzed, then the rehabilitation potential was explored by a retrospective cohort study. **Results:** Among the 384 initially enrolled children, the male-to-female ratio was 2.3:1, and the median age of

diagnosis was 6.5 months (interquartile range: 4-12). The most prevalent MRICS categorization was predominant white matter injury (40.6%), followed by miscellaneous (29.2%) and predominant gray matter injury (15.6%). For the predominant white matter injury and miscellaneous categories, spastic diplegia emerged as the leading subtype of CP, with incidences of 59.6% and 36.6%, respectively, while mixed CP (36.7%) was the most common type in children with predominant gray matter. Notably, 76.4% of children with predominant white matter injury were classified as levels I-III on the gross motor function classification system (GMFCS), indicating significantly less severity than other groups ($\chi 2 = 12.438$, $p = 0.013$). No significant difference across MRICS categories was observed for the manual ability classification system (MACS) ($H = 8.176$, $p = 0.085$). Rehabilitation potential regarding fine motor function and adaptability based on Gesell assessment was dependent on MRICS over the follow-up period. Children with normal MRI scans exhibited superior rehabilitation outcomes. Commencing rehabilitation at an earlier stage produced consistent and beneficial results in terms of fine motor function and adaptability across all MRICS categories. Moreover, participants below 2 years of age demonstrated enhanced rehabilitation potential regarding fine motor outcomes and adaptability within the MRICS framework. **Conclusion:** MRICS displayed a significant association with clinical characteristics and rehabilitation efficacy in children with CP.

编号：CYDQZ-2024-1-18

引用格式：Yang Y, Shangguan Y, Wang X, Liu R, Shen Z, Tang M and Jiang G. The efffcacy and safety of third-generation antiseizure medications and non-invasive brain stimulation to treat refractory epilepsy: a systematic review and network meta-analysis study. Front. Neurol. 2024: 14:1307296.

第一作者 上官亚菲　　通讯作者 蒋国会

Abstract

Background: The new antiseizure medications (ASMs) and non-invasive brain stimulation (NIBS) are controversial in controlling seizures. So, this network meta-analysis aimed to evaluate the efficacy and safety of five third-generation ASMs and two NIBS therapies for the treatment of refractory epilepsy. **Methods:** We searched PubMed, EMBASE, Cochrane Library and Web of Science databases. Brivaracetam (BRV), cenobamate (CNB), eslicarbazepine acetate (ESL), lacosamide (LCM), perampanel (PER), repetitive transcranial magnetic stimulation (rTMS), and transcranial direct current stimulation (tDCS) were selected as additional treatments for refractory epilepsy in randomized controlled studies and other cohort studies. Randomized, double-blind, placebo-controlled, add-on studies that evaluated the efficacy or safety of medication and non-invasive brain stimulation and included patients with seizures were uncontrolled by one or more concomitant ASMs were identified. A random effects model was used to incorporate possible heterogeneity. The primary outcome was the change in seizure frequency from baseline, and secondary outcomes included the proportion of patients with ≥50% reduction in seizure frequency, and the rate of treatment-emergent adverse events. **Results:** Forty-five studies were analyzed. The five ASMs and two NIBS decreased seizure frequency from baseline compared with placebo. The 50% responder rates of the five antiseizure drugs were significantly higher than that of placebo, and the ASMs were associated with fewer adverse events than placebo ($p < 0.05$). The surface under the cumulative ranking analysis revealed that ESL was most effective in decreasing the seizure frequency from baseline, whereas CNB provided the best 50% responder rate. BRV was the best tolerated. No significant publication bias was identified for each outcome index. **Conclusion:** The five third-generation ASMs were more effective in controlling seizures than placebo, among which CNB, ESL, and LCM were most effective, and BRV exhibited better safety. Although rTMS and tDCS did not reduce seizure frequency as effectively as the five drugs, their safety was confirmed.

编号：HJL-2024-1-1

引用格式：Guo X, Yu J, Quan C, Xiao J, Wang J, Zhang B, Hao X, Wu X, Liang J. The effect of N-methyl-D-aspartate receptor antagonists on the mismatch negativity of event-related potentials and its regulatory factors: A systematic review and meta-analysis. J Psychiatr Res. 2024;172:210-220.

通讯作者 梁建民

Abstract

This study investigates the influence of N-methyl-D-aspartate receptor (NMDAR) antagonists on the mismatch negativity (MMN) components of event-related potentials (ERPs) in healthy subjects and explores whether NMDAR antagonists have different effects on MMN components under different types of antagonists, drug dosages, and deviant stimuli. We conducted a comprehensive literature search of PubMed, EMBASE, and the Cochrane Library from inception to August 1, 2023 for studies comparing the MMN components between the NMDAR antagonist intervention group and the control group (or baseline). All statistical analyses were performed using Stata version 12.0 software. Sixteen articles were included in the systematic review: 13 articles were included in the meta-analysis of MMN amplitudes, and seven articles were included in the meta-analysis of MMN latencies. The pooled analysis showed that NMDAR antagonists reduced MMN amplitudes [SMD (95% CI) = 0.32 (0.16, 0.47), P < 0.01, I2 = 47.3%, p < 0.01] and prolonged MMN latencies [SMD (95% CI) = 0.31 (0.13, 0.49), P = 0.16, I2 = 28.3%, p < 0.01]. The type of antagonist drug regulates the effect of NMDAR antagonists on MMN amplitudes. Different antagonists, doses of antagonists, and types of deviant stimuli can also have different effects on MMN. These findings indicate a correlation between NMDAR and MMN, which may provide a foundation for the application of ERP-MMN in the early identification of NMDAR encephalitis.

编号：HJL-2024-1-2

引用格式：Yu J, Zhang Y, Cai L, Sun Q, Li W, Zhou J, Liang J, Wang Z. The Changed Nocturnal Sleep Structure and Higher Anxiety, Depression, and Fatigue in Patients with Narcolepsy Type 1. Nat Sci Sleep. 2024;16:725-735.

通讯作者 梁建民

Abstract

Purpose: This study aimed to evaluate nocturnal sleep structure and anxiety, depression, and fatigue in patients with narcolepsy type 1 (NT1). **Methods:** Thirty NT1 patients and thirty-five healthy controls were enrolled and evaluated using the Epworth sleepiness scale (ESS), Generalized Anxiety Disorder-7, Patient Health Questionnaire-9, Fatigue Severity Scale (FSS), polysomnography, multiple sleep latency test, and brain function state monitoring. Statistical analyses were performed using SPSS Statistics for Windows, version 23.0. Benjamini-Hochberg correction was performed to control the false discovery rate. **Results:** Apart from typical clinical manifestations, patients with NT1 are prone to comorbidities such as nocturnal sleep disorders, anxiety, depression, and fatigue. Compared with the control group, patients with NT1 exhibited abnormal sleep structure, including increased total sleep time (P adj=0.007), decreased sleep efficiency (P adj=0.002), shortening of sleep onset latency (P adj<0.001), elevated wake after sleep onset (P adj=0.002), increased N1% (P adj=0.006), and reduced N2%, N3%, and REM% (P adj=0.007, P adj<0.001, P adj=0.013). Thirty-seven percent of patients had moderate to severe obstructive sleep apnea-hypopnea syndrome. And sixty percent of patients were complicated with REM sleep without atonia. Patients with NT1 displayed increased anxiety propensity (P adj<0.001), and increased brain fatigue (P adj=0.020) in brain function state monitoring. FSS scores were positively correlated with brain fatigue (P adj<0.001) and mean sleep latency was inversely correlated with FSS scores and brain fatigue (P adj=0.013, P adj=0.029). Additionally, ESS scores and brain fatigue decreased after 3 months of therapy (P=0.012, P=0.030). **Conclusion:** NT1 patients had abnormal nocturnal sleep structures, who showed increased anxiety, depression, and fatigue. Excessive daytime sleepiness and fatigue improved after 3

months of treatment with methylphenidate hydrochloride prolonged-release tablets in combination with venlafaxine.

编号：HJL-2024-1-3

引用格式：Yang N, Chen L, Zhang Y, Wu X, Hao Y, Yang F, Yang Z, Liang J. Novel NARS2 variants in a patient with early-onset status epilepticus: case study and literature review. BMC Pediatr. 2024;24(1):96.

通讯作者 梁建民

Abstract

Background: NARS2 as a member of aminoacyl-tRNA synthetases was necessary to covalently join a specific tRNA to its cognate amino acid. Biallelic variants in NARS2 were reported with disorders such as Leigh syndrome, deafness, epilepsy, and severe myopathy. **Case presentation:** Detailed clinical phenotypes were collected and the NARS2 variants were discovered by whole exome sequencing and verified by Sanger sequencing. Additionally, 3D protein structure visualization was performed by UCSF Chimera. The proband in our study had early-onset status epilepticus with abnormal EEG and MRI results. She also performed global developmental delay (GDD) and myocardial dysfunction. Next-generation sequencing (NGS) and Sanger sequencing revealed compound heterozygous missense variants [NM_024678.6:exon14: c.1352G > A(p.Arg451His); c.707T > C(p.Phe236Ser)] of the NARS2 gene. The proband develops refractory epilepsy with GDD and hyperlactatemia. Unfortunately, she finally died for status seizures two months later. **Conclusion:** We discovered two novel missense variants of NARS2 in a patient with early-onset status epilepticus and myocardial dysfunction. The NGS enables the patient to be clearly diagnosed as combined oxidative phosphorylation deficiency 24 (COXPD24, OMIM:616,239), and our findings expands the spectrum of gene variants in COXPD24.

编号：HJL-2024-1-4

引用格式：Li X, Quan P, Si Y, Liu F, Fan Y, Ding F, Sun L, Liu H, Huang S, Sun L, Yang F, Yao L. The microRNA-211-5p/P2RX7/ERK/GPX4 axis regulates epilepsy-associated neuronal ferroptosis and oxidative stress. J Neuroinflam. 2024;21(1):13.

通讯作者 孙林琳

Abstract

Ferroptosis is an iron-dependent cell death mechanism involving the accumulation of lipid peroxides. As a critical regulator, glutathione peroxidase 4 (GPX4) has been demonstrated to be downregulated in epilepsy. However, the mechanism of ferroptosis in epilepsy remains unclear. In this study, bioinformatics analysis, analysis of epilepsy patient blood samples and cell and mouse experiments revealed strong associations among epilepsy, ferroptosis, microRNA-211-5p and purinergic receptor P2X 7 (P2RX7). P2RX7 is a nonselective ligand-gated homotrimeric cation channel, and its activation mainly increases neuronal activity during epileptic seizures. In our study, the upregulation of P2RX7 in epilepsy was attributed to the downregulation of microRNA (miR)-211-5p. Furthermore, P2RX7 has been found to regulate GPX4/HO-1 by alleviating lipid peroxidation induced by suppression of the MAPK/ERK signaling pathway in murine models. The dynamic decrease in miR-211-5p expression induces hypersynchronization and both nonconvulsive and convulsive seizures, and forebrain miR-211-5p suppression exacerbates long-lasting pentylenetetrazole-induced seizures. Additionally, in this study, induction of miR-211-5p expression or genetic-silencing of P2RX7 significantly reduced the seizure score and duration in murine models through the abovementioned pathways. These results suggest that the miR-211-5p/P2RX7 axis is a novel target for suppressing both ferroptosis and epilepsy.

编号：HJL-2024-1-5

引用格式：仲文强，陈雪莲，朱延梅. 事件相关电位在癫痫疾病中的应用现状研究[J].癫痫杂志,2024,10(02):133-140.

通讯作者 朱延梅

摘要

事件相关电位（Event related potentials，ERPs）是一种从脑电中提取的与一定的刺激相关联的电位 活动，具有客观、易操作、实时反映大脑认知处理过程的优势，广泛用于研究阿尔兹海默病（Alzheimer's disease， AD）、帕金森病（Parkinson's disease，PD）、卒中、精神分裂症等疾病的相关病理生理机制 。癫痫作为神经科常见 疾病之一，ERPs 可从神经电生理层面探究癫痫患者出现认知障碍、焦虑抑郁情绪的原因并对其做出客观评估。 现就 ERPs 在癫痫患者中的应用现状进行综述。

SCI收录的癫痫领域部分杂志

杂志名	IF	分区
Epilepsia	6.6	Q1
Epilepsy Currents	5.8	Q1
Epilepsia Open	2.8	Q2
Epilepsy & Behavior	2.3	Q2
Epilepsy Research	2.0	Q3
Epileptic Disorders	1.9	Q3
Acta Epileptologica	1.2	Q4

编号：YGQM-2024-1-1

引用格式：Huang WY, Zhang HY, Li X, Zhang JM, Chen JJ, Chen ZY, Ni GZ. Prognostic factors underlying the development of drug-resistant epilepsy in patients with autoimmune encephalitis: a retrospective cohort study. J Neurol. 2024; doi: 10.1007/s00415-024-12432-y.

通讯作者：陈子怡 倪冠中

Abstract

Objective: The aim of our study was to analyze the characteristics of patients with autoimmune encephalitis (AE) to identify prognostic factors associated with the development of drug-resistant epilepsy (DRE). **Methods:** In this retrospective observational cohort study, we enrolled adult patients with AE between January 2016 and December 2022. The patients were categorized into two groups based on the presence or absence of DRE at the last follow-up. The predictors of the development of DRE were investigated using logistic regression analysis. **Results:** Among 121 AE patients, 75.2% (n = 91) experienced acute symptomatic seizures, and 29.8% (n = 36) developed DRE at the last follow-up. On multivariate regression analysis, the factors associated with DRE were antibody negativity (OR 3.628, 95% CI 1.092-12.050, p = 0.035), focal seizure (OR 6.431, 95% CI 1.838-22.508, p = 0.004), refractory status epilepticus (OR 8.802, 95% CI 2.445-31.689, p = 0.001), interictal epileptiform discharges on EEG (OR 6.773, 95% CI 2.206-20.790, p = 0.001), and T2/FLAIR hyperintensity in the limbic system (OR 3.286, 95% CI 1.060-10.183, p = 0.039). **Conclusions:** In this study, the risk of developing DRE was mainly observed among AE patients who were negative for antibodies or had focal seizures, refractory status epilepticus, interictal epileptiform discharges on EEG, and T2/FLAIR hyperintensity in the limbic system.

编号：YGQM-2024-1-2

引用格式：Wang F, Ren J, Cui W, Zhou Y, Yao P, Lai X, Pang Y, Chen Z, Lin Y, Liu H. Verbal memory network mapping in individual patients predicts postoperative functional impairments[J]. Hum Brain Mapp. 2024;45(7):e26691.

第一作者：王丰

Abstract

Verbal memory decline is a significant concern following temporal lobe surgeries in patients with epilepsy, emphasizing the need for precision presurgical verbal memory mapping to optimize functional outcomes. However, the inter-individual variability in functional networks and brain function-structural dissociations pose challenges when relying solely on group-level atlases or anatomical landmarks for surgical guidance. Here, we aimed to develop and validate a personalized functional mapping technique for verbal memory using precision resting-state functional MRI (rs-fMRI) and neurosurgery. A total of 38 patients with refractory epilepsy scheduled for surgical interventions were enrolled and 28 patients were analyzed in the study. Baseline 30-min rs-fMRI scanning, verbal memory and language assessments were collected for each patient before surgery. Personalized verbal memory networks (PVMN) were delineated based on preoperative rs-fMRI data for each patient. The accuracy of PVMN was assessed by comparing post-operative functional impairments and the overlapping extent between PVMN and surgical lesions. A total of 14 out of 28 patients experienced clinically meaningful declines in verbal memory after surgery. The personalized network and the group-level atlas exhibited 100% and 75.0% accuracy in predicting postoperative verbal memory declines, respectively. Moreover, six patients with extra-temporal lesions that overlapped with PVMN showed selective impairments in verbal memory. Furthermore, the lesioned ratio of the personalized network rather than the group-level atlas was significantly correlated with postoperative declines in verbal memory (personalized networks: r = -0.39, p = .038; group-level atlas: r = -0.19, p = .332). In conclusion, our personalized functional mapping technique, using precision

rs-fMRI, offers valuable insights into individual variability in the verbal memory network and holds promise in precision verbal memory network mapping in individuals.

编号：YGQM-2024-1-3

引用格式：Wang F, Hong ST, Zhang Y, Xing Z, Lin YX. ^{18}F-FDG-PET/CT for Localizing the Epileptogenic Focus in Patients with Different Types of Focal Cortical Dysplasia. Neuropsychiatr Dis Treat. 2024;20:211-220.

第一作者 王丰

Abstract

Purpose: To determine the diagnostic and localization value of 18F-fluorodeoxyglucose-positron emission tomography (PET)/computed tomography (CT) in patients with focal cortical dysplasia (FCD) who underwent epilepsy surgery. **Methods:** One hundred and eight patients with pathologically proven FCD who underwent surgery for refractory epilepsy were retrospectively analyzed. All patients underwent magnetic resonance imaging (MRI), 18F-FDG-PET/CT, and video electroencephalography. An MRI diagnosis of FCD was defined as MRI+. A PET/CT diagnosis of FCD was defined as PET/CT+. **Results:** MRI and PET/CT detected FCD in 20.37% and 93.52% of patients, respectively. The difference was significant. Twenty-one patients were MRI+/PET+, 80 were MRI-/PET+, six were MRI-/PET-, and one was MRI+/PET-. The MRI positivity rate was lowest in patients with FCD type IIIa (5.6%, $P < 0.05$). Prevalence of MRI-/PET+ was highest in patients with FCD type IIIa (88.89%, $P < 0.05$). **Conclusion:** PET/CT is superior to MRI in detecting FCD. FCD type IIIa was more likely than other types to show MRI-/PET+. This suggests that PET/CT has particular diagnostic value for FCD type IIIa patients with negative MRI findings.

编号：YGQM-2024-1-4

引用格式：Hu X, Fang Z, Wang F, Mei Z, Huang X, Lin Y, Lin Z. A causal relationship between gut microbiota and subcortical brain structures contributes to the microbiota-gut-brain axis: a Mendelian randomization study. Cerebral Cortex. 2024;34(2):bhae056.

第一作者：王丰

Abstract

A correlation between gut microbiota and brain structure, referring to as a component of the gut-brain axis, has been observed in observational studies. However, the causality of this relationship and its specific bacterial taxa remains uncertain. To reveal the causal effects of gut microbiota on subcortical brain volume, we applied Mendelian randomization (MR) studies in this study. Genome-wide association study data were obtained from the MiBioGen Consortium (n = 18,340) and the Enhancing Neuro Imaging Genetics through Meta-Analysis Consortium (n = 13,170). The primary estimate was obtained utilizing the inverse-variance weighted, while heterogeneity and pleiotropy were assessed using the Cochrane Q statistic, MR Pleiotropy RESidual Sum and Outlier, and MR-Egger intercept. Our findings provide strong evidence that a higher abundance of the genus Parasutterella is causally correlated with a decrease in intracranial volume (β = -30,921.33, 95% CI -46,671.78 to -15,170.88, P = 1.19 × 10-4), and the genus FamilyXIIIUCG001 is associated with a decrease in thalamus volume (β = -141.96, 95% CI: -214.81 to -69.12, P = 1.0× 10-4). This MR study offers novel perspectives on the intricate interplay between the gut microbiota and subcortical brain volume, thereby lending some support to the existence of the microbiota-gut-brain axis.

编号：YGQM-2024-1-5

引用格式：Li H, Wang Y, Guo J, Zhang PQ, Xu Z, Peng K, Dong XL, Zhao LM.

Effcacy and safety of modifed medium-chain triglyceride ketogenic diet in patients with drug-resistant epilepsy. Acta Epileptologica. 2024:7(1):44-52.

第一作者 李花

Abstract

Background Medium-chain triglyceride ketogenic diet (MCTKD) is previously less commonly used in China. This study was aimed to assess the efcacy and safety of the modifed MCTKD in the treatment of drug-resistant epilepsy in Chinese patients. **Methods** Patients with drug-resistant epilepsy were enrolled to receive treatment with modifed MCTKD in Guangdong Sanjiu Brain Hospital during December 2020 and September 2022. The modifed MCTKD contained fat that provided 50–70% of the total energy, as well as proteins and carbohydrates that provided 20–30% and 20% of energy, respectively. The fat component was composed of 20–30% medium-chain triglycerides (MCTs) and 30–40% longchain triglycerides. The efcacy and safety of the diet were assessed at 1, 3 and 6 months. **Results** A total of 123 patients aged 2.5 to 65 years, were included in this study. The response rates at 1, 3 and 6 months were 49.6%, 43.1%, and 30.9%, respectively. The seizure freedom rates at 1, 3 and 6 months were 12.2%, 10.6%, and 6.5%, respectively. The retention rates **at** 1, 3 and 6 months were 98.4%, 65.0% and 33.3% respectively. Side efects occurred in 21.14% of patients, which were predominantly gastrointestinal symptoms such as abdominal pain, diarrhea, vomiting, and constipation, and most of them resolved after dietary adjustments. A total of 82 patients (66.7%) discontinued the treatment with the reason of refusing to eat (8.1%), poor efcacy (35.0%), poor compliance (4.9%), and inability to follow-up (9.8%). Only 4 patients (3.3%) withdrew the diet due to side effects. **Conclusions** The modifed MCTKD with MCTs providing 20–30% of energy has a good safety in patients with drugresistant epilepsy, but its efectiveness needs to be enhanced. Further modifcations of MCTKD with an optimal energy ratio are required to achieve a better efcacy and safety.

编号：YGQM-2024-1-6

引用格式：Mai JH, Li H, He HY, Huang TS, Lin CM, Lan S, Xiao XH, He SL, Lu XG, Chen L, Li B, Luo XF, Wang H, Liao JX, Cao DZ. Efficacy and safety of perampanel as the first add-on therapy for children with epilepsy: A real-world multicenter prospective observational study. Seizure .2024;117:44-49.

第一作者：李花　通讯作者 操德智

Abstract

Objective: Perampanel (PER) is a new anti-seizure medication (ASM) with a novel mechanism of action. This study aimed to determine the efficacy and safety of PER when added to monotherapy in children and adolescents (age, 4–18 years) with epilepsy. **Method:** A multicenter prospective observational study was performed on children and adolescents (age, 4–18 years) with epilepsy who did not respond to ASM monotherapy between July 2021 and October 2022. PER was used as the first add-on therapy for the enrolled patients. Seizure-free rate, response rate, inefficacy rate, and drug retention rate were the main observation indicators during the 6 months of treatment. The patients were grouped based on treatment efficacy, and factors affecting efficacy were statistically analyzed. Adverse reactions were also recorded. **Results:** In this study, 93 patients with epilepsy were enrolled; among them, 9 patients were lost to follow-up (attrition rate, 9.7 %), and 84 were included in the analysis. Five patients with unknown efficacy discontinued taking PER early due to intolerable adverse reactions, and 79 patients (48 males, 31 females; mean age, 11.0 ± 3.9 years) finally remained. Genetic epilepsy and structural epilepsy were found in 22 patients and 36 patients, respectively. The mean duration of epilepsy history at the time of PER initiation was 4.0 ± 3.8 years, and the mean maintenance dosage of add-on PER was 4.5 ± 1.8 mg/day (equivalent to 0.14 ± 0.07 mg/kg/day). Among the 79 patients, 28 patients were diagnosed with epilepsy syndrome, including 13 patients having self limited epilepsy with centrotemporal spikes, among whom 9 patients were seizure-free after adding PER

during the 6-month follow-up (seizure-free rate, 69.2 %). For these 79 patients, the seizure-free, response, and retention rates at the end of follow-up were 45.6 %, 74.7 %, and 82.1 %, respectively. Among the 84 patients included in the analyses, adverse reactions occurred in 20 patients, mainly dizziness (8 patients), somnolence (6 patients), and irritability (4 patients), and 4 patients developed two adverse reactions simultaneously. Univariate analyses revealed statistically significant differences in efficacy between groups with structural and non-structural epilepsy and between groups with different baseline concomitant ASMs, suggesting that these factors affected the efficacy of PER as the first add-on therapy. **Conclusion:** The overall response rate of PER as the first add-on therapy for children and adolescents with epilepsy who were followed up for 6 months was 74.7 %, indicating a relatively favorable safety and tolerability profile. The group of the baseline concomitant ASM administered and the etiological classification of epilepsy as either structural or non-structural were the factors influencing the efficacy of PER as the first add-on therapy.

编号：YGQM-2024-1-7

引用格式：Wu H, Liao K, Tan Z, Zeng C, Wu B, Zhou Z, Zhou H, Tang Y, Gong J, Ye W, Ling X, Guo Q, Xu H. A PET-based radiomics nomogram for individualized predictions of seizure outcomes after temporal lobe epilepsy surgery. Seizure. 2024;119:17-27.

通讯作者 郭强

Abstract

Purpose: To establish and validate a novel nomogram based on clinical characteristics and [18F]FDG PET radiomics for the prediction of postsurgical seizure freedom in patients with temporal lobe epilepsy (TLE). **Patients and methods:** 234 patients with drug-refractory TLE patients were included with a median follow-up time of 24 months after surgery. The correlation coefficient redundancy analysis and LASSO Cox regression were used to characterize risk factors. The Cox model was conducted to

develop a Clinic-PET nomogram to predict the relapse status in the training set (n = 171). The nomogram's performance was estimated through discrimination, calibration, and clinical utility. The prognostic prediction model was validated in the test set (n = 63). **Results:** Eight radiomics features were selected to assess the radiomics score (radscore) of the operation side (Lat_radscore) and the asymmetric index (AI) of the radiomics score (AI_radscore). AI_radscor, Lat_radscor, secondarily generalized seizures (SGS), and duration between seizure onset and surgery (Durmon) were significant predictors of seizure-free outcomes. The final model had a C-index of 0.68 (95 %CI: 0.59-0.77) for complete freedom from seizures and time-dependent AUROC was 0.65 at 12 months, 0.65 at 36 months, and 0.59 at 60 months in the test set. A web application derived from the primary predictive model was displayed for economic and efficient use. **Conclusions:** A PET-based radiomics nomogram is clinically promising for predicting seizure outcomes after temporal lobe epilepsy surgery.

编号：YGQM-2024-1-8

引用格式：Liao K, Wu H, Jiang Y, Dong C, Zhou H, Wu B, Tang Y, Gong J, Ye W, Hu Y, Guo Q, Xu H. Machine learning techniques based on 18F-FDG PET radiomics features of temporal regions for the classification of temporal lobe epilepsy patients from healthy controls. Front Neurol. 2024;15:1377538.

通讯作者 郭强

Abstract

Background: This study aimed to investigate the clinical application of 18F-FDG PET radiomics features for temporal lobe epilepsy and to create PET radiomics-based machine learning models for differentiating temporal lobe epilepsy (TLE) patients from healthy controls. **Methods:** A total of 347 subjects who underwent 18F-FDG PET scans from March 2014 to January 2020 (234 TLE patients: 25.50 ± 8.89 years, 141 male patients and 93 female patients; and 113 controls: 27.59 ± 6.94 years, 48 male

individuals and 65 female individuals) were allocated to the training (n = 248) and test (n = 99) sets. All 3D PET images were registered to the Montreal Neurological Institute template. PyRadiomics was used to extract radiomics features from the temporal regions segmented according to the Automated Anatomical Labeling (AAL) atlas. The least absolute shrinkage and selection operator (LASSO) and Boruta algorithms were applied to select the radiomics features significantly associated with TLE. Eleven machine-learning algorithms were used to establish models and to select the best model in the training set. **Results:** The final radiomics features (n = 7) used for model training were selected through the combinations of the LASSO and the Boruta algorithms with cross-validation. All data were randomly divided into a training set (n = 248) and a testing set (n = 99). Among 11 machine-learning algorithms, the logistic regression (AUC 0.984, F1-Score 0.959) model performed the best in the training set. Then, we deployed the corresponding online website version showing the details of the LR model for convenience. The AUCs of the tuned logistic regression model in the training and test sets were 0.981 and 0.957, respectively. Furthermore, the calibration curves demonstrated satisfactory alignment (visually assessed) for identifying the TLE patients. **Conclusion:** The radiomics model from temporal regions can be a potential method for distinguishing TLE. Machine learning-based diagnosis of TLE from preoperative FDG PET images could serve as a useful preoperative diagnostic tool.

编号：YGQM-2024-1-9

引用格式：Jin L, Li Y, Luo S, Peng Q, Zhai QX, Zhai JX, Gao LD, Guo JJ, Song W, Yi YH, He N, Chen YJ. Reprint of: Recessive APC2 missense variants associated with epilepsies without neurodevelopmental disorders. Seizure. 2024;116:87-92.

通讯作者：何娜

Abstract

Objectives: The APC2 gene, encoding adenomatous polyposis coli protein-2, is involved in cytoskeletal regulation in neurons responding to endogenous extracellular signals and plays an important role in brain development. Previously, the APC2 variants have been reported to be associated with cortical dysplasia and intellectual disability. This study aims to explore the association between APC2 variants and epilepsy. **Methods:** Whole-exome sequencing (WES) was performed in cases (trios) with epilepsies of unknown causes. The damaging effects of variants were predicted by protein modeling and in silico tools. Previously reported APC2 variants were reviewed to analyze the genotype-phenotype correlations. **Results:** Four pairs of compound heterozygous missense variants were identified in four unrelated patients with epilepsy without brain malformation/intellectual disability. All variants presented no or low allele frequencies in the controls. The missense variants were predicted to be damaging by silico tools, and affect hydrogen bonding with surrounding amino acids or decreased protein stability. Patients with variants that resulted in significant changes in protein stability exhibited more severe and intractable epilepsy, whereas patients with variants that had minor effect on protein stability exhibited relatively mild phenotypes. The previously reported APC2 variants in patients with complex cortical dysplasia with other brain malformations-10 (CDCBM10; MIM: 618677) were all truncating variants; in contrast, the variants identified in epilepsy in this study were all missense variants, suggesting a potential genotype-phenotype correlation. **Significance:** This study suggests that APC2 is potentially associated with epilepsy without brain malformation/intellectual disability. The genotype-phenotype correlation helps to understand the underlying mechanisms of phenotypic heterogeneity.

编号：YGQM-2024-1-10

引用格式：Cao B, Peng B, Tian Y, Wang X, Li X, Zhu H, Shen H, Chen W. Clinical and genetic analysis of 18 patients with KCNQ2 mutations from South China. Turk J Pediatr. 2024;66(2):191-204.

通讯作者：彭炳蔚

Abstract

Background: We aimed to delineate the genotype and phenotype of patients with KCNQ2 mutations from South China. **Methods:** Clinical manifestations and characteristics of KCNQ2 mutations of patients from South China were analyzed. Previous patients with mutations detected in this study were reviewed. **Results:** Eighteen epilepsy patients with KCNQ2 mutations, including seven self-limited neonatal epilepsy (SeLNE), two self-limited infantile epilepsy (SeLIE) and nine developmental and epileptic encephalopathy (DEE) were enrolled. The age of onset (p=0.006), mutation types (p=0.029), hypertonia (p=0.000), and seizure offset (p=0.029) were different in self-limited epilepsy (SeLE) and DEE. De novo mutations were mainly detected in DEE patients (p=0.026). The mutation position, EEG or the age of onset were not predictive for the seizure or ID/DD outcome in DEE, while the development of patients free of seizures was better than that of patients with seizures (p=0.008). Sodium channel blockers were the most effective anti-seizure medication, while the age of starting sodium channel blockers did not affect the seizure or development offset. We first discovered the seizure recurrence ratio in SeLNE/SeLIE was 23.1% in South China. Four novel mutations (c.790T>C, c.355_363delGAGAAGAG, c.296+2T>G, 20q13.33del) were discovered. Each of eight mutations (c.1918delC, c.1678C>T, c.683A>G, c.833T>C, c.868G>A, c.638G>A, c.997C>T, c.830C>T) only resulted in SeLE or DEE, while heterogeneity was also found. Six patients in this study have enriched the known phenotype caused by the mutations (c.365C>T, c.1A>G, c.683A>G, c.833T>C, c.830C>T, c.1678C>T). **Conclusion:** This research has expanded known phenotype and genotype of KCNQ2-related epilepsy, and the different clinical features of SeLE and DEE from South China.

编号：YGQM-2024-1-11

引用格式：Song Y, Gao M, Wei B, Huang X, Yang Z, Zou J, Guo Y.

Mitochondrial ferritin alleviates ferroptosis in a kainic acid-induced mouse epilepsy model by regulating iron homeostasis: Involvement of nuclear factor erythroid 2-related factor 2. CNS Neurosci Ther. 2024;30(3):e14663.

通讯作者 郭燕舞

Abstract

Background: Epilepsy is a widespread and chronic disease of the central nervous system caused by a variety of factors. Mitochondrial ferritin (FtMt) refers to ferritin located within the mitochondria that may protect neurons against oxidative stress by binding excess free iron ions in the cytoplasm. However, the potential role of FtMt in epilepsy remains unclear. We aimed to investigate whether FtMt and its related mechanisms can regulate epilepsy by modulating ferroptosis. **Methods:** Three weeks after injection of adeno-associated virus (AAV) in the skull of adult male C57BL/6 mice, kainic acid (KA) was injected into the hippocampus to induce seizures. Primary hippocampal neurons were transfected with siRNA using a glutamate-mediated epilepsy model. After specific treatments, Western blot analysis, immunofluorescence, EEG recording, transmission electron microscopy, iron staining, silver staining, and Nissl staining were performed. **Results:** At different time points after KA injection, the expression of FtMt protein in the hippocampus of mice showed varying degrees of increase. Knockdown of the FtMt gene by AAV resulted in an increase in intracellular free iron levels and a decrease in the function of iron transport-related proteins, promoting neuronal ferroptosis and exacerbating epileptic brain activity in the hippocampus of seizure mice. Additionally, increasing the expression level of FtMt protein was achieved by AAV-mediated upregulation of nuclear factor erythroid 2-related factor 2 (Nrf2) gene in the hippocampus of seizure mice. **Conclusions:** In epilepsy, Nrf2 modulates ferroptosis by involving the expression of FtMt and may be a potential therapeutic mechanism of neuronal injury after epilepsy. Targeting this relevant process for treatment may be a therapeutic strategy to prevent epilepsy.

编号：YGQM-2024-1-12

引用格式 Hu Z, Tang L, Xu X, Zhan Y. Association between physical activity and psychosocial status in adults with epilepsy: Results from the 2022 National health Interview survey. Epilepsy Behav. 2024 ;156:109836.

通讯作者：许晓伟

Abstract

Objective: The study aimed to investigate the association between physical activity and the four dimensions of psychosocial status in adults with epilepsy. **Methods:** The data of individuals with epilepsy utilized in this cross-sectional study were derived from the 2022 National Health Interview Survey(NHIS). Physical activity was analyzed based on walking, moderate or vigorous intensity physical activity and the 2018 Physical Activity Guidelines (PAG) for Americans. The psychosocial status of the participants was assessed using self-report questionnaires that evaluated life satisfaction, symptoms of depression and anxiety, and social functioning. A multivariate ordinal regression model was employed to estimate odds ratios (ORs) and corresponding 95% confidence intervals (CIs) following adjustment for potential confounding factors. **Results:** In total of 424 individuals with epilepsy(mean age:48.0 years; male: 40.6 %) were included in this study. About 39.9 % of the participants met the 2018 PAG for aerobic activity. After controlling for potential confounding factors, individuals who adhered to the 2018 PAG for aerobic activity were found to have a higher likelihood of reporting increased life satisfaction (OR, 0.39; 95 % CI: 0.21, 0.71), decreased symptoms of depression (OR, 0.53; 95 % CI: 0.30, 0.94), and improved social functioning (OR, 0.42; 95 % CI: 0.24, 0.74). However, no significant association was observed between physical activity and anxiety symptoms among individuals with epilepsy. **Conclusions:** This study emphasizes that moderate to vigorous physical activity enhances psychosocial health in individuals with epilepsy. Nevertheless, it is important to note that a causal relationship cannot be inferred from these findings, and

further verification through randomized controlled trials is necessary.

编号：YGQM-2024-1-13

引用格式：Zheng Y, She Y, Su Z, Huang K, Chen S, Zhou L. A novel pathogenic variant in TDP2 causes spinocerebellar ataxia autosomal recessive 23 accompanied by pituitary tumor and hyperhidrosis: a case report. Neurol Sci. 2024;45(6):2881-2885

通讯作者 陈树达

Abstract

DP2 gene encodes tyrosyl DNA phosphodiesterase 2, an enzyme required for effective repair of the DNA double-strand breaks (DSBs). Spinocerebellar ataxia autosomal recessive 23 (SCAR23) is a rare disease caused by the pathogenic mutation of TDP2 gene and characterized by intellectual disability, progressive ataxia and refractory epilepsy. Thus far, merely nine patients harboring five different variants (c.425 + 1G > A; c.413_414delinsAA, p. Ser138*; c.400C > T, p. Arg134*; c.636 + 3_ 636 + 6 del; c.4G > T, p. Glu2*) in TDP2 gene have been reported. Here, we describe the tenth patient with a novel variant (c.650del, p. Gly217GlufsTer7) and new phenotype (pituitary tumor and hyperhidrosis).

编号：YGQM-2024-1-14

引用格式: Zhu C, Li J, Wei D, Wu L, Zhang Y, Huang H, Lin W. Intrinsic brain activity differences in perampanel-responsive and non-responsive drug-resistant epilepsy patients: an EEG microstate analysis. Ther Adv Neurol Disord. 2024 Jan 30;17:17562864241227293.

通讯作者：林婉挥

Abstract

Background: Drug-resistant epilepsy (DRE) patients exhibit aberrant large-scale brain networks. Perampanel may be a therapeutic option for controlling seizures in these patients. **Objective:** We aim to explore the differences of resting-state electroencephalogram (EEG) microstate in perampanel-responsive and non-responsive DRE patients. **Design:** Retrospective study. **Methods:** Clinical data were collected from DRE patients who received perampanel treatment at the Fujian Medical University Union Hospital from June 2020 to September 2021, with a minimum follow-up of 6 months. Patients were classified into three groups based on the extent of reduction in seizure frequency: non-responsive (seizure reduction <50%), responsive (seizure reduction >50% but not seizure-free), and seizure-free. Resting-state EEG data sets of all participants were subjected to EEG microstate analysis. The study comprehensively compared the mean duration, frequency per second, and temporal coverage of each microstate among the three groups. **Results:** A total of 76 perampanel-treated DRE patients were categorized into three groups based on their response to treatment: non-responsive (n = 20), responsive (n = 36), and seizure-free (n = 20), according to the degree of seizure frequency reduction. The results of EEG microstate analysis revealed no statistically significant distinctions in frequency, duration, and coverage of microstate D in these DRE patients. However, the seizure-free group showed significantly increased duration and coverage of microstate A, frequency and coverage of microstate B, and significantly decreased duration, frequency, and coverage of microstate C when compared with the other groups. **Conclusion:** Microstate A, B, and D is associated with the sensorimotor network, visual network, salience network, and attention network, respectively. This study demonstrates statistically significant differences in the sensorimotor, visual, and salience networks, but not in the attention network, between perampanel-responsive and non-responsive DRE patients.

编号：YGQM-2024-1-15

引用格式: Fang S, Zhu C, Zhang J, Wu L, Zhang Y, Huang H, Lin W. EEG microstates in epilepsy with and without cognitive dysfunction: Alteration in intrinsic brain activity. Epilepsy Behav. 2024;154:109729.

通讯作者 林婉挥

Abstract

Objective: This study aims to investigate the difference between epilepsy comorbid with and without cognitive dysfunction. **Method:** Participants were classified into patients with epilepsy comorbid cognitive dysfunction (PCCD) and patients with epilepsy without comorbid cognitive dysfunction (nPCCD). Microstate analysis was applied based on 20-channel electroencephalography (EEG) to detect the dynamic changes in the whole brain. The coverage, occurrence per second, duration, and transition probability were calculated. **Result:** The occurrence per second and the coverage of microstate B in the PCCD group were higher than that of the nPCCD group. Coverage in microstate D was lower in the PCCD group than in the nPCCD group. In addition, the PCCD group has a higher probability of A to B and B to A transitions and a lower probability of A to D and D to A transitions. **Conclusion:** Our research scrutinizes the disparities observed within EEG microstates among epilepsy patients both with and without comorbid cognitive dysfunction. **Significance:** EEG microstate analysis offers a novel metric for assessing neuropsychiatric disorders and supplies evidence for investigating the mechanisms and the dynamic change of epilepsy comorbid cognitive dysfunction.

编号：YGQM-2024-1-16

引用格式: Wang WL, Ren YH, Hou WL, Zhang XB, Yang CL, An WM, Xu F, and Wang FP, Identification of hub genes significantly linked to tuberous sclerosis

related-epilepsy and lipid metabolism via bioinformatics analysis. Front Neurol. 2024;15:1354062.

通讯作者：王逢鹏

Abstract

Background: Tuberous sclerosis complex (TSC) is one of the most common genetic causes of epilepsy. Identifying differentially expressed lipid metabolism related genes (DELMRGs) is crucial for guiding treatment decisions. **Methods:** We acquired tuberous sclerosis related epilepsy (TSE) datasets, GSE16969 and GSE62019. Differential expression analysis identified 1,421 differentially expressed genes (DEGs). Intersecting these with lipid metabolism related genes (LMRGs) yielded 103 DELMRGs. DELMRGs underwent enrichment analyses, biomarker selection, disease classification modeling, immune infiltration analysis, weighted gene co-expression network analysis (WGCNA) and AUCell analysis. **Results:** In TSE datasets, 103 DELMRGs were identified. Four diagnostic biomarkers (ALOX12B, CBS, CPT1C, and DAGLB) showed high accuracy for epilepsy diagnosis, with an AUC value of 0.9592. Significant differences ($p < 0.05$) in Plasma cells, T cells regulatory (Tregs), and Macrophages M2 were observed between diagnostic groups. Microglia cells were highly correlated with lipid metabolism functions. **Conclusions:** Our research unveiled potential DELMRGs (ALOX12B, CBS, CPT1C and DAGLB) in TSE, which may provide new ideas for studying the pathogenesis of epilepsy.

欢迎大家引用上述文献

编号：SGQNX-2024-1-1

引用格式: Li D, Sun N, Guo Y, Huang S, Yin C, Xiao Y, Ma W. Investigating the Effects of Perampanel on Autophagy-mediated Regulation of GluA2 and PSD95 in Epilepsy. Mol Neurobiol. 2024; doi: 10.1007/s12035-024-04136-1.

第一作者 李丹

Abstract

Epilepsy is a chronic neurological disorder characterized by recurrent seizures. Despite various treatment approaches, a significant number of patients continue to experience uncontrolled seizures, leading to refractory epilepsy. The emergence of novel anti-epileptic drugs, such as perampanel (PER), has provided promising options for effective epilepsy treatment. However, the specific mechanisms underlying the therapeutic effects of PER remain unclear. This study aimed to investi gate the intrinsic molecular regulatory mechanisms involved in the downregulation of GluA2, a key subunit of α-amino-3 hydroxy-5-methyl-4-isoxazolepropionic acid receptors, following epileptic seizures. Primary mouse hippocampal neurons were cultured and subjected to an epilepsy cell model. The expression levels of GluA2 and autophagy-related proteins were assessed using Western blotting and real-time fluorescent quantitative PCR. Immunofluorescence and immunohistochemistry techniques were employed to investigate the nuclear translocation of CREB-regulated transcriptional coactivator 1 (CRTC1). Additionally, status epilepticus animal models were established to further validate the findings. The epilepsy cell model exhibited a significant decrease in GluA2 expression, accompanied by elevated levels of autophagy-related proteins. Immu nofluorescence analysis revealed the nuclear translocation of CRTC1, which correlated with the expression of autophagy related genes. Treatment with an autophagy inhibitor reversed the decreased expression of GluA2 in the epilepsy cell model. Furthermore, the calcium/calmodulin-dependent protein phosphatase inhibitor FK506 and CaN overexpression affected the dephosphorylation and nuclear translocation of CRTC1, consequently influencing GluA2 expression. Animal model results further supported the involvement of these molecular mechanisms in epilepsy. Our findings suggest that

the downregulation of GluA2 following epileptic seizures involves the activation of autophagy and the regulation of CRTC1 nuclear transloca tion. These intrinsic molecular regulatory mechanisms provide potential targets for developing novel therapeutic strategies to alleviate refractory epilepsy and preserve cognitive functions in patients.

编号：SGQNX-2024-1-2

引用格式：Li D, Sun N, Xiang L, Liu J, Wang X, Yang L, Huang S. Neurophysiological Characteristics in Type II and Type III 5q Spinal Muscular Atrophy Patients: Impact of Nusinersen Treatment. Drug Des Devel Ther. 2024;18:953-965.

第一作者 李丹

Abstract

Objective: This study aimed to observe the neurophysiological characteristics of type II and type III 5q spinal muscular atrophy (SMA) patients and the changes in peripheral motor nerve electrophysiology after Nusinersen treatment, as well as the influencing factors. **Methods:** This single-center retrospective case-control study collected clinical data and peripheral motor nerve CMAP parameters from 42 5qSMA patients and 42 healthy controls at the Second Affiliated Hospital of Xi'an Jiaotong University (January 2021 to December 2022). It evaluated changes in motor function and CMAP amplitude before and after Nusinersen treatment. **Results:** Our investigation encompassed all symptomatic and genetically confirmed SMA patients, consisting of 32 type II and 10 type III cases, with a median age of 57 months (29.5 to 96 months). Comparative analysis with healthy controls revealed substantial reductions in CMAP amplitudes across various nerves in both type II and type III patients. Despite the administration of Nusinersen treatment for 6 or 14 months to the entire cohort, discernible alterations in motor nerve amplitudes were not observed, except for a significant improvement in

younger patients (≤36 months) at the 14-month mark. Further scrutiny within the type II subgroup unveiled that individuals with a disease duration ≤12 months experienced a noteworthy upswing in femoral nerve amplitude, a statistically significant difference when compared to those with >12 months of disease duration. **Conclusion:** Motor nerve amplitudes were significantly decreased in type II and type III 5q SMA patients compared to healthy controls. Nusinersen treatment showed better improvement in motor nerve amplitudes in younger age groups and those with shorter disease duration, indicating a treatment-time dependence.

编号：SGQNX-2024-1-3

引用格式: Lu M, Wang X, Sun N, Huang S, Yang L, Li D. Metabolomics of cerebrospinal fluid reveals candidate diagnostic biomarkers to distinguish between spinal muscular atrophy type II and type III. CNS Neurosci Ther. 2024;30(4):e14718.

通讯作者 李丹

Abstract

Aims: Classification of spinal muscular atrophy (SMA) is associated with the clinical prognosis; however, objective classification markers are scarce. This study aimed to identify metabolic markers in the cerebrospinal fluid (CSF) of children with SMA types II and III. **Methods:** CSF samples were collected from 40 patients with SMA (27 with type II and 13 with type III) and analyzed for metabolites. **Results:** We identified 135 metabolites associated with SMA types II and III. These were associated with lysine degradation and arginine, proline, and tyrosine metabolism. We identified seven metabolites associated with the Hammersmith Functional Motor Scale: 4-chlorophenylacetic acid, adb-chminaca,(+/-)-, dodecyl benzenesulfonic acid, norethindrone acetate, 4-(undecan-5-yl) benzene-1-sulfonic acid, dihydromaleimide beta-d-glucoside, and cinobufagin. Potential typing biomarkers, N-

cyclohexylformamide, cinobufagin, cotinine glucuronide, N-myristoyl arginine, 4-chlorophenylacetic acid, geranic acid, 4-(undecan-5-yl) benzene, and 7,8-diamino pelargonate, showed good predictive performance. Among these, N-myristoyl arginine was unaffected by the gene phenotype. **Conclusion:** This study identified metabolic markers are promising candidate prognostic factors for SMA. We also identified the metabolic pathways associated with the severity of SMA. These assessments can help predict the outcomes of screening SMA classification biomarkers.

编号：SGQNX-2024-1-4

引用格式: Wang X, Yuan N, Zhu J, Wang B, Zhang W, Liu Y. Fever-induced acute sleep terrors in children and adolescents following SARS-CoV-2 infection. Sleep Breath. 2024; doi: 10.1007/s11325-024-03038-9.

通讯作者 刘永红

Abstract

Objective: This study aims to provide physicians with insights into the clinical manifestations and outcomes of children and young adolescents experiencing sleep terrors following SARS-CoV-2 infection. **Methods:** We enrolled patients who developed new onset sleep terrors after SARS-CoV-2infection fromDecember2022to April 2023 in the Xijing hospital, Xi'an, China. **Results:** We enrolled six patients who experienced sleep terrors following SARS-CoV-2 infection. Out of these patients, five were children and only one was an adolescent, with a mean age of 9 years. Neuroimaging results were negative for all cases. Sleep terrors occurred during both the active course of COVID-19 illness and the recovery period in all patients. Symptoms included crying or screaming in terror, hyperactivity, inappropriate behavior and periods of mental confusion during sleep. These episodes typically occurred 40 min to 1 h after falling asleep. EEG monitoring confirmed two patients' episodes occurred during non-rapid eye movement (NREM) stage 3 sleep. The duration of sleep terrors

ranged from 3mines to30 mines, with each patient experiencing 3-4 to 30-40 instances. Initially, the frequency of episodes was highest at 3-4 times per night, gradually decreasing to once a night, then once a week, until complete disappearance. No medical intervention was required. Clinical follow-up ranged from 6 to 12 months, with spontaneous remission occurring within 1 week to 2 months for different patients. **Conclusion:** SARS-CoV-2 infection may precipitate acute sleep terrors in children and adolescents. The course of these sleep terrors is generally benign, with all patients achieving spontaneous complete remission over time.

编号：SGQNX-2024-1-5

引用格式: Pan Y, Zhao D, Zhang X, Yuan N, Yang L, Jia Y, Guo Y, Chen Z, Wang Z, Qu S, Bao J, Liu Y. Machine learning-Based model for prediction of Narcolepsy Type 1 in Patients with Obstructive Sleep Apnea with Excessive Daytime Sleepiness. Nat Sci Sleep. 2024;16:639-652.

通讯作者 刘永红

Abstract

Background: Excessive daytime sleepiness (EDS) forms a prevalent symptom of obstructive sleep apnea (OSA) and narcolepsy type 1 (NT1), while the latter might always be overlooked. Machine learning (ML) models can enable the early detection of these conditions, which has never been applied for diagnosis of NT1. **Objective:** The study aimed to develop ML prediction models to help non-sleep specialist clinicians identify high probability of comorbid NT1 in patients with OSA early. **Methods:** Totally, clinical features of 246 patients with OSA in three sleep centers were collected and analyzed for the development of nine ML models. LASSO regression was used for feature selection. Various metrics such as the area under the receiver operating curve (AUC), calibration curve, and decision curve analysis (DCA) were employed to evaluate and compare the performance of these ML models. Model interpretability was

demonstrated by Shapley Additive explanations (SHAP). **Results:** Based on the analysis of AUC, DCA, and calibration curves, the Gradient Boosting Machine (GBM) model demonstrated superior performance compared to other machine learning (ML) models. The top five features used in the GBM model, ranked by feature importance, were age of onset, total limb movements index, sleep latency, non-REM (Rapid Eye Movement) sleep stage 2 and severity of OSA. **Conclusion:** The study yielded a simple and feasible screening ML-based model for the early identification of NT1 in patients with OSA, which warrants further verification in more extensive clinical practices.

编号：SGQNX-2024-1-6

引用格式: Hu G, Pan Y, Yuan N, Yang Z, Shi X, Ma S, Li S, Hou X, Liu F, Li D, Bao J, Liu Y. Tongue Biting Event in Patients with Sleep-Related Facial Mandibular Myoclonus: A Case Series Study. Nat Sci Sleep. 2024:16:207-215.

通讯作者 刘永红

Abstract

Background: Sleep-related facial mandibular myoclonus (SRFMM) remains rare in clinical practice. The aim of this study was to provide a comprehensive understanding of the electroclinical manner, therapeutic regimen, and prognosis of SRFMM. **Methods:** Twenty-three patients who were diagnosed with SRFMM by clinical manifestation, video-electroencephalography (EEG) and electromyography over bilateral masseter and temporalis muscles were enrolled. Clinical and electrophysiological evaluation as well as follow-up information were recorded and analyzed. **Results:** The cohort involved 4 infants and 19 adults with a mean onset age of 43.5 years for SRFMM, among whom 19 were male. Twenty-one patients complained of tongue injuries and disturbed night-time sleep. SRFMM in 4 patients were ascribed to oral aripiprazole, brainstem ischemia and brain trauma. In 62 SRFMM episodes, 93.5% occurred in NREM sleep and 6.5% in REM sleep, and all events were

associated with EEG arousals. In 13 patients with or without clonazepam, the motor events gradually disappeared, and the rest turned to be sporadic. **Conclusion:** SRFMM is a characteristic parasomnia manifested by tongue biting and accompanying facial mandibular myoclonus, leading to disrupted sleep. Besides adults, infants can also experience SRFMM with spontaneous remission. Most patients respond well to clonazepam, eventually with favorable prognosis.

编号：SGQNX-2024-1-7

引用格式：Wang X, Pan Y, Marcuse L, Yuan N, Liu Y. Clinical and video-polysomnographic characterization of restless sleep disorder in adult patients. Sleep Bio Rhyth. 2024; doi.org/10.1007/s41105-024-00524-1.

通讯作者 刘永红

Abstract

Adults with restless sleep disorder (RSD) have never been studied clinically and polysomnographically. This study aimed to describe the clinical manifestation, duration, and distribution of sleep-related movements in adult patients with restless sleep disorder. Patients who had performed VPSG from Jan 2021 to Jan 2022 and met the diagnosis criteria of RSD were enrolled in the study. Patients' bed partners were also interviewed or telephoned in identifying this disorder. Scoring of movements during sleep was according to the diagnosis criteria of RSD and scoring of large muscle group movements during sleep proposed by the International RLS Study Group in 2020 and 2021, respectively. The clinical manifestation, the distribution of sleep stage as well as the types and duration of the movements were carefully recorded and analyzed. We included ten patients in the study with a mean age of 27.6 years (range 22–38). There was a male prevalence in adults with RSD. The study highlighted the findings from video-polysomnography, which indicated frequent sleep-related movements occurring throughout the Night. These movements were most prominent during N1 and N2 sleep stage, followed by REM sleep, while fewer movements were observed during N3 sleep.

Adults with RSD experienced significant daytime functioning impairments, including non-refreshing sleep, daytime fatigue/sleepiness, and mood disturbance. Two of the patients in the study were diagnosed with anxiety and depression, further underscoring the impact of RSD on mental health. Adult patients also suffer from severe RSD, and the RSD that originates in childhood tends to persist into adulthood. In these cases, longer duration of the disease and poor sleep quality may be associated with an increased risk of developing psychiatric comorbidities. Our cases represent an objectively documented type of RSD in younger adult patients.

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杂志名	IF	分区
Lancet Neurology	46.5	Q1
JAMA Neurology	20.4	Q1
Brain	10.6	Q1
Acta Neuropathologica	9.3	Q1
Journal of Neurology Neurosurgery and Psychiatry	8.7	Q1
Annals of Neurology	8.1	Q1
Neurology-Neuroimmunology & Neuroinflammation	7.8	Q1
Neurology	7.7	Q1
Brain Stimulation	7.6	Q1
Neurotherapeutics	5.6	Q1
Journal of Neurology	4.8	Q1
Therapeutic Advances in Neurological Disorders	4.7	Q1
European journal of Neurology	4.5	Q1
Neurosurgery	3.9	Q1
Journal of Neurosurgery	3.5	Q1
Pediatric Neurology	3.2	Q2
Neuromodulation	3.2	Q2

编号：SSL-2024-1-1

引用格式: Xu Y, Wang Q, Zhang Y, Chen Y, Xu L, Zhu G, Ma C, Wu X. Long-term treatment with Perampanel of Chinese patients with focal-onset seizures, especially in sleep-related epilepsy: a prospective real-world observational study. Front Neurol. 2024;15:1364295.

通讯作者 吴洵轶

Abstract

Background: There is currently a lack of studies examining the long-term therapeutic effectiveness of the third-generation anti-sezure medication, perampanel (PER), for focal-onset seizures (FOS), particularly in Chinese patients with sleep-related epilepsy (SRE). Additionally, the appropriate dosage, plasma concentration, and the relationship between dose and plasma concentration of PER in Chinese patients are still uncertain. **Methods:** A prospective, single-center, 24-month observational study was conducted in patients diagnosed with FOS, with a focus on patients with SRE. Changes in seizure frequency from baseline, adverse events, and retention rates were analyzed at 12 and 24 months following the start of the treatment. Tolerability was evaluated based on adverse events and discontinuation profiles. PER plasma concentrations were used to assess dose-concentration-response relationships. **Results:** A total of 175 patients were included (median age: 25 years; range: 4–72 years; 53. 1% males and 46.9% females), with the SRE population accounting for 49. 1% (n = 86). The patients diagnosed with SRE showed considerably higher response rates than those who did not have this diagnosis ($p = 0.025$, odds ratio = 3.8). Additionally, the SRE group adhered better to PER treatment ($r = 0.0009$). Patients with a shorter duration of epilepsy (median: 3 years; range:2–7 years) demonstrated a more favorable therapeutic response to PER ($p = 0.032$). Throughout the administration of maintenance doses, among the entire FOS population, the concentration of PER (C0) ranged between 101.5 and 917.4 ng/mL (median, 232.0 ng/mL), and the mean plasma concentration of PER in the responders was 292.8 ng/mL. We revealed a linear relationship between PER dose and plasma

concentration, regardless of whether PER was used as monotherapy or add-on therapy. The retention rates were 77.7% and 65. 1% at 12 and 24 months, respectively. Drug-related adverse events occurred in 45.0% of the patients and were mostly manageable. **Conclusions:** PER effectively reduced seizure frequency in Chinese patients with FOS, particularly in those with SRE, over a 24-month period. The treatment was well-tolerated and had a clear linear dose-plasma concentration relationship.

编号：SSL-2024-1-2

引用格式: Lu R, Wang M, Zhang Y, Li H, Zhou Y, Wang Y, Zhao R. Safety, Accuracy, and Efficacy of Robot-Assisted Stereo Electroencephalography in Children of Different Ages. Neurosurgery. 2024;95(1):137–45.

通讯作者 赵瑞

Abstract

Background: Aimed to investigate the safety, accuracy, and efficacy of stereo electroencephalography (SEEG) in children of various ages, with particular emphasis on those younger than 3 years. There is limited guidance regarding whether SEEG can conducted on very young children. **Methods:** This retrospective study was conducted between July 2018 and August 2022. It involved 88 patients who underwent 99 robot-assisted SEEG procedures at our center. The patients were categorized into 3 groups based on their age at the time of the robot-assisted SEEG procedures: group 1 (3 years and younger, n = 28), group 2 (age 3-6 years, n = 27), and group 3 (older than 6 years, n = 44). Clinical data, SEEG demographics, complications, and seizure outcomes were analyzed. **Results:** A total of 675 electrodes were implanted, with an average of 6.82 ± 3.47 (2.00-16.00) electrodes per patient (P = .052). The average target point error for the 675 electrodes was 1.93 ± 1.11 mm, and the average entry point error was 1.30 ± 0.97 mm (P = .536 and P = .549, respectively). The overall percentage of complications was 6.06% (P = .879). No severe or long-term neurologic impairment was observed. Of the total 99 procedures included in this study, 78 were admitted for epilepsy surgery for the

first time, while 9 patients were treated twice and 1 patient was treated 3 times. There were 21 radiofrequency thermocoagulation and 78 second-stage resective procedures performed after SEEG. There was no statistically significant difference in Engel class I outcomes among the patients who underwent SEEG in the 3 age groups (P = .621). **Conclusions:** Robot-assisted SEEG were demonstrated to be safe, accurate, and efficient across different age groups of children. This technique is suitable for children younger than 3 years who have indications for SEEG placement.

编号：SSL-2024-1-3

引用格式: Xu Y, Lu R, Li H, Feng W, Zhao R. A spectrum of AKT3 activating mutations cause focal malformations of cortical development (FMCDs) in cortical organoids. Biochim Biophys Acta Mol Basis Dis. 2024 ;1870(6):167232.

通讯作者 赵瑞

Abstract

Background: Focal malformations of cortical development (FMCDs) are brain disorders mainly caused by hyperactive mTOR signaling due to both inactivating and activating mutations of genes in the PI3K-AKT-mTOR pathway. Among them, mosaic and somatic activating mutations of the mTOR pathway activators are more frequently linked to severe form of FMCDs. A human stem cell-based FMCDs model to study these activating mutations is still lacking. **Methods:** we genetically engineer human embryonic stem cell lines carrying these activating mutations to generate cortical organoids. **Results:** Mosaic and somatic expression of AKT3 activating mutations in cortical organoids mimicking the disease presentation with overproliferation and the formation of dysmorphic neurons. In parallel comparison of various AKT3 activating mutations reveals that stronger mutation is associated with more severe neuronal migratory and overgrowth defects. **Conclusions:** we have established a feasible human stem cell-based model for FMCDs that could help to better understand

pathogenic mechanism and develop novel therapeutic strategy.

编号：SSL-2024-1-4

引用格式:Su S, Zhao F, Zhang H, Liu Y, Li Z, Zhang H, Wang Y, Fang F, Liu Y. Establishment of a transgene-free iPS cell line (SDCHi003-A) from a young patient bearing a NPRL2 mutation and suffering from Epilepsy. Stem Cell Res. 2024,76:103366.

第一作者 张洪伟

Abstract

Background: Epilepsy affects ~ 65 million people worldwide. Status epilepticus can lead to life-threatening if untreated. **Methods:** In this study, peripheral blood mononuclear cells were isolated from a young patient patient bearing a Nitrogen Perntease Regulator Like 2 Protein (NPRL2) mutation and suffering from Epilepsy verified by clinical and genetic diagnosis. Induced pluripotent stem cells (iPSCs) were established by a non-integrative method, using plasmids carrying OCT4, SOX2, KLF4, BCL-XL and C-MYC. **Results:**The established iPSCs presented typical pluripotent cells morphology, normal karyotype, and potential to differentiate into three germ layers. **Conclusions:** Our approach offers a useful model to explore pathogenesis and therapy of Epilepsy.

编号：SSL-2024-1-5

引用格式: Wang Y, Geng G, Hu W, Zhang H, Liu Y, Gao Z, Zhang H, Shi J. Epileptic seizures as an initial symptom for Sturge-Weber syndrome type III: A report of two cases. Exp Ther Med. 2024 28(1):299.

通讯作者 张洪伟

Abstract

Background: Sturge-Weber syndrome (SWS) type III, a rare neurocutaneous disorder, presents diagnostic challenges due to its variable clinical manifestations. **Methods:** The present study focuses on enhancing the understanding of this syndrome by conducting a detailed analysis of two pediatric cases and providing a comprehensive review of the existing literature. **Results:** The cases, managed at the Children's Hospital Affiliated to Shandong University (Jinan, China), highlight the diverse clinical presentations and successful management strategies for SWS type III. In the first case, a 4-year-old male patient exhibited paroxysmal hemiplegia, epileptic seizures and cerebral angiographic findings indicative of left pia mater and venous malformation. The second case involved a 2.5-year-old male patient presenting with recurrent seizures and angiographic findings on the right side. Both cases underscore the importance of considering epileptic seizures, acquired and transient hemiplegia and cognitive impairments in the diagnosis of SWS type III. **Conclusions:** The present study provides insights into the effective use of both pharmacological and surgical interventions, drawing from the positive outcomes observed in these cases. The findings emphasize the need for heightened awareness and a meticulous approach in diagnosing and treating SWS type III, contributing to the better management and prognosis of this condition.

编号：SSL-2024-1-6

引用格式: Zhang, H, Deng, J, Gao, Z, Wang, Y, Zhao, F, Zhao, H, Fang, F. Clinical phenotype and genotype of NPRL2-related epilepsy: Four cases reports and literature review. Seizure. 2024;116, 100-106.

第一作者 张洪伟

Abstract

Background: NPRL2-related epilepsy, caused by pathogenic germline variants of the NPRL2 gene, is a newly discovered childhood epilepsy linked to enhanced mTORC1 signalling. However, the phenotype and genotype of NPRL2 variants are still poorly

understood. Here, we summarize the association between the phenotype and genotype of NPRL2-related epilepsy. **Methods:** A retrospective analysis was conducted for four Chinese children with epilepsy due to likely pathogenic NPRL2 variants identified through whole-exome sequencing (WES). Previous reports of patients with NPRL2-related epilepsy were reviewed systematically. **Results:** One of our patients presented focal epilepsy involving the central region, which should be distinguished from self-limited epilepsy with centrotemporal spikes (SeLECTS). The four novel likely pathogenic NPRL2 variants consisted of two nonsense variants, one frameshift variant, and one copy number variant (CNV). Bioinformatics analysis revealed the two nonsense variants to be highly conserved and cause alterations in protein structure. Including our four cases, a total of 33 patients with NPRL2-related epilepsy have been identified to date. The most common presentation is focal epilepsy (70%), including sleep-related hypermotor epilepsy (SHE), temporal lobe epilepsy (TLE), and frontal lobe epilepsy (FLE). Infantile epileptic spasms syndrome (IESS) is also a notable feature of NPRL2-related epilepsy. Malformations of cortical development (MCD, 8/20), especially focal cortical dysplasia (FCD, 6/20), are common neuroimaging abnormalities. Two-thirds of the NPRL2 variants reported are loss of function (LoF) (14/21). Among these mutations, c.100C>T (p.Arg34*) and c.314T>C (p.Leu105Pro) have been detected in two families (likely due to a founder effect). **Conclusion:** NPRL2-related epilepsy shows high phenotypic and genotypic heterogeneity. Our study expands the genotype spectrum of NPRL2-related epilepsy, and the phenotype of focal epilepsy involving the central region should be clearly distinguished with SeLECTS, with reference value for clinical diagnosis.

编号：SSL-2024-1-7

引用格式: Li X, Pei Y, Ge Y, Xu L, Zhang Y, Zheng L, Ding D, Hong Z, PeiminYu. Epilepsy and driving: A preliminary survey of people with epilepsy at an epilepsy clinic in China. Epilepsy Behav. 2024;153:109668.

通讯作者：虞培敏

Abstract

Background: Driving is an important part of the daily life for most adults, and restrictions on driving can significantly affect the quality of life for people with epilepsy. This study aimed to investigate the current driving status of patients at an epilepsy clinic in China. **Method:** Study participants were administered a survey by a questionnaire including the demographic and clinical characteristics of seizure, driving-related questions and attitudes to driving. **Results:** A total of 101 patients responded the survey. Among 33(32.7%) who hold the driving license, 20 (60.6%) still drive, 3 had seizures while driving, and the rate of traffic accidents was 0. There was no significant difference in seizure frequency and type of medication between patients with and without the driving license, but compliance with medication was significantly better for those who held the driving license. **Conclusions:** One-third of people with epilepsy hold the driving license and good drug compliance is a favorable factor for driving. Standardizing different levels of restriction on driving for people with epilepsy is urgently needed.

编号：SSL-2024-1-8

引用格式: Liu Q, Lin Z, Shen Y, Zhu J, Song J, Zhang C, Lu Y, Xu J. Use of Compressed Sensing Accelerated, Low-Velocity Encoded, Isotropic Resolution, Phase Contrast Magnetic Resonance Angiography for SEEG Electrode Implantation. World Neurosurg. 2024;181:e18-e28.

第一作者 刘强强

Abstract

Background: We assessed the feasibility of using compressed sensing accelerated, low-velocity encoded, isotropic resolution phase contrast (CLIP) magnetic resonance angiography (MRA) for avascular trajectory planning of stereoelectroencephalography.

Method: Ten healthy subjects (1 woman and 9 men; age, 33.6 ± 9.0 years) and 20 consecutive patients (12 female patients; age, 22 ± 13.6 years) were enrolled in the present study. The healthy subjects underwent CLIP-MRA, and 3 other phase contrast MRA protocols with conventional parallel imaging (PI) acceleration, including low resolution with twofold PI (PI2), high resolution (HR) with fivefold PI (PI5), and HR-PI2. The patients underwent CLIP-MRA and computed tomography angiography (CTA). The image qualities were evaluated. The numbers and locations of trajectory–vessel conflict detected using CLIP-MRA were noted. **Results:** With similar scan durations, CLIP-MRA achieved higher spatial resolution compared with low resolution with PI2 and detected significantly more branches compared with HR-PI5. With the same spatial resolution, the signal/noise and contrast/noise ratios of CLIP-MRA were higher than those with HR-PI2 with a shorter scan duration. For the 12 adult patients (10 female patients; 28.8 ± 12.7 years), CLIP-MRA had better signal/noise and contrast/noise ratios than CTA. The trajectory had required modification for 14 of the 20 patients (70%), with a proportion of trajectory modification of 10.7% (23 of 215 electrodes). The middle meningeal artery, cortical vessel, and skull vessel were the main vessels with conflict ($n = 11$, $n = 7$, and $n = 5$, respectively). **Conclusions:** In the present study, CLIP-MRA provided a clear cortical vascular display noninvasively without intravascular contrast and radiation. The middle meningeal artery and diploic and emissary veins were the main conflict vessels and could be clearly displayed using CLIP-MRA but not CTA.

欢迎大家引用上述文献

编号：ZWG-2024-1-1

引用格式: Ding Y, Cen Z, Zheng Y, Qiu X, Ye Y, Chen X, Hu L, Wang B, Wang Z, Yin H, Shen C, Ming W, Ge Y, Xie F, Yang D, Ouyang Z, Wang H, Wu S, Ding M, Wang S, Luo W. Seizures and electrophysiological features in familial cortical myoclonic tremor with epilepsy 1. Clin Transl Neurol. 2024;11(2):414-423. doi: 10.1002/acn3.51961.Epub 2023 Dec 7.

第一作者 丁瑶　　通讯作者 王爽

Abstract

Objectives: To investigate and characterize epileptic seizures and electrophysiological features of familial cortical myoclonic tremor with epilepsy (FCMTE) type 1 patients in a large Chinese cohort. **Methods:** We systematically evaluated 125 FCMTEtype 1 patients carrying the pentanucleotide (TTTCA) repeat expansion in the SAMD12 gene in China. **Results:** Among the 28 probands, epileptic seizures (96.4%, 27/28) were the most common reason for an initial clinic visit. Ninety-seven (77.6%, 97/125) patients had experienced seizures. The seizures onset age was 36.5 ± 9.0 years, which was 6.9 years later than cortical tremors. The seizures were largely rare (<1/year, 58.8%) and occasional (1-6/year, 37.1%). Prolonged prodromes were reported in 57.7% (56/97). Thirty-one patients (24.8%, 31/125) reported photosensitivity history, and 79.5% (31/39) had a photoparoxysmal response. Interictal epileptiform discharges (IEDs) were recorded in 69.1% (56/81) of patients. Thirty-three patients showed generalized IEDs and 72.7% (24/33) were occipitally dominant, while 23 patients presented with focal IEDs with 65.2% (15/23) taking place over the occipital lobe. Overnight EEG of FCMTE patients displayed paradoxical sleep-wake fluctuation, with a higher average IED index of 0.82 ± 0.88/min during wakefulness and a lower IED index of 0.04 ± 0.06/min during non-rapid eye movement sleep stages I-II. **Interpretation:** FCMTE type 1 has a benign course of epilepsy and distinct clinical and electrophysiological features. In addition to a positive family history and cortical myoclonus tremor, the seizure prodromes, specific seizure triggers, photosensitivity, distribution of IEDs, and

unique fluctuations during sleep-wake cycle are cues for proper genetic testing and an early diagnosis of FCMTE.

编号：ZWG-2024-1-2

引用格式: Ye H, Ye L, Hu L, Yang Y, Ge Y, Chen R, Wang S, Jin B, Ming W, Wang Z, Xu S, Xu C, Wang Y, Ding Y, Zhu J, Ding M, Chen Z, Wang S, Chen C. Widespread slow oscillations support interictal epileptiform discharge networks in focal epilepsy. Neurobiol Dis. 2024;191:106409.

通讯作者 王爽

Abstract

Background: Interictal epileptiform discharges (IEDs) often co-occur across spatially-separated cortical regions, forming IED networks. However, the factors prompting IED propagation remain unelucidated. We hypothesized that slow oscillations (SOs) might facilitate IED propagation. **Methods:** The amplitude and phase synchronization of SOs preceding propagating and non-propagating IEDs were compared in 22 patients with focal epilepsy undergoing intracranial electroencephalography (EEG) evaluation. Intracranial channels were categorized into the irritative zone (IZ) and normal zone (NOZ) regarding the presence of IEDs. **Results:** During wakefulness, we found that pre-IED SOs within the IZ exhibited higher amplitudes for propagating IEDs than non-propagating IEDs (delta band: $p = 0.001$, theta band: $p < 0.001$). This increase in SOs was also concurrently observed in the NOZ (delta band: $p = 0.04$). Similarly, the inter-channel phase synchronization of SOs prior to propagating IEDs was higher than those preceding non-propagating IEDs in the IZ (delta band: $p = 0.04$). Through sliding window analysis, we observed that SOs preceding propagating IEDs progressively increased in amplitude and phase synchronization, while those preceding non-propagating IEDs remained relatively stable. Significant differences in amplitude occurred approximately 1150 ms before IEDs. During non-rapid eye movement (NREM) sleep, SOs on scalp recordings also showed higher amplitudes before

intracranial propagating IEDs than before non-propagating IEDs (delta band: $p = 0.006$). Furthermore, the analysis of IED density around sleep SOs revealed that only high-amplitude sleep SOs demonstrated correlation with IED propagation. **Conclusions:** This study highlights that transient but widely distributed SOs are associated with IED propagation as well as generation in focal epilepsy during sleep and wakefulness, providing new insight into the EEG substrate supporting IED networks.

编号：ZWG-2024-1-3

引用格式: Li Q, Cao Y, Zhang J, Fu Y, Shen B, Wang S, Fang J. Pregnancy-related knowledge in women with epilepsy in childbearing age: A pilot questionnaire survey from China. Brain Behav, 2024;14(2): e3400.

通讯作者 方嘉佳

Abstract

Purpose: We aim to understand the knowledge of and attitudes toward pregnancy issues among women with epilepsy (WWE) and their caregivers and analyze the answers from the questionnaire to expose topics that require educational activities; thus, WWE experiences pregnancy better. **Methods:** WWE at their childbearing age and/or their caregivers who entered the Fourth Affiliated Hospital of Zhejiang University for treatment of their condition were invited to fill out a questionnaire between March 1 and November 31, 2022.**Results:** A combined total of 205 WWE and 142 caregivers completed the questionnaires. Among the surveyed WWE, a majority (63.74%) reported experiencing at least one miscarriage or induced abortion. However, a significant proportion (84.62%) of these WWE were still able to successfully give birth to at least one child. Furthermore, the offspring of these WWE showed no significant differences compared to the offspring of women without epilepsy, as reported by 93.51% of the participants. The participants' knowledge regarding the impact of epilepsy on pregnancy was found to be comparable, with average scores of 7.74 and 7.84, respectively. The participants exhibited a limited comprehension of antiseizure

medications (ASMs)-related knowledge, specifically pertaining to ASMs adjustment during pregnancy (17.56% vs. 16.90%) and offspring outcomes (30.24% vs. 26.06%). Statistical analysis revealed significant correlations between the overall score and education level ($p < .001$), as well as epilepsy duration ($p = .008$). Regarding the source of knowledge, participants acknowledged primarily relying on neurologists, who remained their preferred choice for consultation. **Conclusions:** In our study, the understanding of pregnancy-related knowledge did not differ from WWE and their caregivers, both are far from satisfactory in certain areas. It is urgent for WWE and their caregivers to improve their pregnancy-related knowledge of epilepsy. As their primary access is from knowledgeable health care professionals like neurologists, well-trained neurologists in epilepsy management during pregnancy are in need.

编号：ZWG-2024-1-4

引用格式：Li Q, Zhang Z, Fang J. Hormonal Changes in Women with Epilepsy. Neuropsychiatr Dis Treat, 2024:20 373-388.

通讯作者 方嘉佳

Abstract

Epilepsy is a prevalent neurological disorder among women globally, often requiring long-term treatment. Hormonal fluctuations in women with epilepsy (WWE) can have reciprocal effects on epilepsy and antiseizure medications (ASMs), posing significant challenges for WWE. Notably, WWE commonly experience endocrine alterations such as thyroid dysfunctions, low bone metabolism, and reproductive hormone irregularities. On the one hand, the presence of hormones in women with epilepsy affects their susceptibility to epilepsy as well as the metabolism of antiseizure medications in various ways. On the other hand, epilepsy itself and the use of antiseizure medications impact the production, secretion, and metabolism of hormones, resulting in low fertility, increased risk of pregnancy complications, negative offspring outcomes, and so on. In order to develop more precise treatment strategies in the future, it is necessary to

comprehend the explicit relationships between hormones, epilepsy, and antiseizure medications, as well as to elucidate the currently known mechanisms underlying these interactions.

编号：ZWG-2024-1-5

引用格式: Cao Y, Jiang L, Zhang J, Fu Y, Li Q, Fu W, Zhu J, Xiang X., Zhao G, Kong D, Chen X, Fang J. A fast and non-invasive artificial intelligence olfactory-like system that aids diagnosis of Parkinson's disease. Eur J Neurol. 2024;31(3): e16167.

通讯作者 方嘉佳

Abstract

Background: Several previous studies have shown that skin sebum analysis can be used to diagnose Parkinson's disease (PD). The aim of this study was to develop a portable artificial intelligence olfactory-like (AIO) system based on gas chromatographic analysis of the volatile organic compounds (VOCs) in patient sebum and explore its application value in the diagnosis of PD. **Methods:** The skin VOCs from 121 PD patients and 129 healthy controls were analyzed using the AIO system and three classic machine learning models were established, including the gradient boosting decision tree (GBDT), random forest and extreme gradient boosting, to assist the diagnosis of PD and predict its severity. **Results:** A 20-s time series of AIO system data were collected from each participant. The VOC peaks at a large number of time points roughly concentrated around 5-12 s were significantly higher in PD subjects. The gradient boosting decision tree model showed the best ability to differentiate PD from healthy controls, yielding a sensitivity of 83.33% and a specificity of 84.00%. However, the system failed to predict PD progression scored by Hoehn-Yahr stage. **Conclusions:** This study provides a fast, low-cost and non-invasive method to distinguish PD patients from healthy controls. Furthermore, our study also indicates abnormal sebaceous gland secretion in PD patients, providing new evidence for exploring the pathogenesis of PD.

编号：ZWG-2024-1-6

引用格式: Zhang J, Zuo H, Fu Y, Cao Y, Li Q, Zhang Q, Zheng Y, Wang Y, Wu D., Chen W, Fang J. Intranasal delivery of phenytoin loaded layered double hydroxide nanoparticles improves therapeutic effect on epileptic seizures. J Nanobiotechnology. 2024, 22(1): 144.

通讯作者 方嘉佳

Abstract

Improving the efficiency of antiseizure medication entering the brain is the key to reducing its peripheral toxicity. A combination of intranasal administration and nanomedicine presents a practical approach for treating epileptic seizures via bypassing the blood-brain barrier. In this study, phenytoin (PHT) loaded layered double hydroxide nanoparticles (BSA-LDHs-PHT) were fabricated via a coprecipitation − hydrothermal method for epileptic seizure control. In this study, we expound on the preparation method and characterization of BSA-LDHs-PHT. In-vitro drug release experiment shows both rapid and continuous drug release from BSA-LDHs-PHT, which is crucial for acute seizure control and chronic epilepsy therapy. In-vivo biodistribution assays after intranasal administration indicate excellent brain targeting ability of BSA-LDHs. Compared to BSA-Cyanine5.5, BSA-LDHs-Cyanine5.5 were associated with a higher brain/peripheral ratio across all tested time points. Following intranasal delivery with small doses of BSA-LDHs-PHT, the latency of seizures in the pentylenetetrazole-induced mouse models was effectively improved. Collectively, the present study successfully designed and applied BSA-LDHs-PHT as a promising strategy for treating epileptic seizures with an enhanced therapeutic effect.

编号：ZWG-2024-1-7

引用格式: Fu Y, Zhang J, Cao Y, Ye L, Zheng R, Li Q, Shen B, Shi Y, Cao J,

Fang J. Recognition memory deficits detected through eye-tracking in well-controlled children with self-limited epilepsy with centrotemporal spikes. Epilepsia. 2024;doi: 10.1111/epi.17902

通讯作者 方嘉佳

Abstract

Objective: Children with self-limited epilepsy characterized by centrotemporal spikes (SeLECTS) exhibit cognitive deficits in memory during the active phase, but there is currently a lack of studies and techniques to assess their memory development after well-controlled seizures. In this study, we employed eye-tracking techniques to investigate visual memory and its association with clinical factors and global intellectual ability, aiming to identify potential risk factors by examining encoding and recognition processes. **Methods:** A total of 26 recruited patients diagnosed with SeLECTS who had been seizure-free for at least 2 years, along with 24 control subjects, underwent Wechsler cognitive assessment and an eye-movement-based memory task while video-electroencephalographic (EEG) data were recorded. Fixation and pupil data related to eye movements were utilized to detect distinct memory processes and subsequently to compare the cognitive performance of patients exhibiting different regression patterns on EEG. **Results:** The findings revealed persistent impairments in visual memory among children with SeLECTS after being well controlled, primarily observed in the recognition stage rather than the encoding phase. Furthermore, the age at onset, frequency of seizures, and interictal epileptiform discharges exhibited significant correlations with eye movement data. **Significance:** Children with SeLECTS exhibit persistent recognition memory impairment after being well controlled for the disease. Controlling the frequency of seizures and reducing prolonged epileptiform activity may improve memory cognitive development. The application of the eye-tracking technique may provide novel insights into exploring memory cognition as well as underlying mechanisms associated with pediatric epilepsy.

2024 年 CAAE 青年委员会年度学术报告与交流大会

2024 年 3 月 23-24 日中国抗癫痫协会青年委员会在 CAAE 秘书处的指导下，CAAE 青年委员会年度学术报告与交流大会在在汉文化发源地徐州召开。中国抗癫痫协会李世绰创会会长、洪震名誉会长、周东会长、周列民、邓艳春、林卫红、张国君、丁玎副会长，廖卫平监事长，江苏省抗癫痫协会杨天明创会会长、李岩会长，张华、杨卫东、李晓裔、徐淑军等省协会领导、杨小枫、姚一、钟建民等常务理事和 300 余位青委与青年癫痫工作者参加了本次学术会议。

本次大会以“彭城万里，南秀北雄”为主题，秉承“创新与争鸣”的主旋律，迸发出癫痫学界青年人的激情和活力。大会开幕式由徐州市儿童医院神经内科主任刘晓鸣教授担任主持。中国抗癫痫协会会长周东教授、徐州市政府张晨秘书长、徐州市儿童医院张洪永院长、江苏省抗癫痫协会李岩会长等领导先后致词。周会长首先肯定了过去的一年，青委会各项工作取得的优异成效，特别是 “西部行”活动不仅使青年医生得到锻炼和成长，也使得医疗资源下沉，造福了广大的患者。同时周会长也希望在新的一年里，青委们能够务实笃行，做好临床工作；勇毅创新，做好科学研究，特别是在人工智能、脑机接口、大数据、基因精准治疗方面要取得新进展、新突破；在癫痫诊疗与研究这个长赛道上，埋头苦干、奋发进取、合作共享，共创中国抗癫痫事业的美好未来。

中国抗癫痫协会青年委员会主任委员梁树立教授致辞并作 2023 年度工作总结及 2024 年度工作计划汇报。在大家的共同努力下，青委会圆满完成了 2023 年各项既定任务。在立体化的西部行活动、菁 Young 计划、癫痫数据银行与多中心研究项目，在领读学术、CAAE 青年思行、NEW 项目、青委学术会议、建立癫痫儿科登记系统等工作方面，均取得了不俗的成绩。根据工作需要，经量化考核、常务理事会讨论通过，本次特别增补 15 位新委员加入。他希望在新的一年，青委们更多参与到青委会各项工作中来，建立以兴趣为导向的多种协作方式，培养研究型人才，推动合作共享，开展成果转化。此外 2024 年青委会计划建立委员考评机制，继续加强国内外学者互动交流合作，全面加强科普宣传工作和人文建设。

本次会议，特增设“大师论道”环节，有幸邀请到中国工程院院士、首都医科大学附属北京天坛医院神经外科学中心主任江涛教授和中国抗癫痫协会创会会长李世绰教授。江院士在作了题为“神经肿瘤新药研发范式的建立与应用”的演讲。江涛院士诚挚希望各位同道携手合作，共同推动神经肿瘤药物研发，为中国和全世界的患者带来希望和健康。中国抗癫痫协会李世绰创会会长为大家带来了“科学心理学”的讲座，表示青年人是协会发展的中坚力量、是协会的未来，青委们要认真钻研、立志做有理想、敢担当、能吃苦、肯奋斗的新时代好青年。随后在大师与青委面对面环节青委与两位大师展开了激烈的讨论。

“青委年度最佳研究评比”、 “2023 癫痫领域热点问题争鸣与研讨” 和“多中心科研协作项目展示与招募”等单元中，青委们全程用流畅的英语或中文进行演讲和讨论，展示出事业发展“后浪”辈出，相信在未来国际癫痫学术舞台上，他们将越来越多地发出中国好声音。在最佳研究评比中孟祥红获得一等奖，王中科、邓劼获得二等奖，姜慧轶、于昊、杨雅男获得三等奖；王中科依靠创新性的研究成果、流利的英文表达和阳光的形象斩获最佳人气奖。此外，青委八大区还进行了 2023 年工作情况的亮点汇报，每个大区都在学术发展、人才培养、服务患者等方向快速发展，同时也有鲜明的区域特色和人文建设亮点，通过交流大区间可以互相借鉴，互相促进。

创新与争鸣仍是青委学术会议永恒的主题。本次会议的研究热点围绕癫痫病、癫痫的亚分子机制、癫痫的神经调控的新靶点、癫痫诊疗可穿戴设备、MOGHE、NCSE 等进行了演讲。参与会议讨论的不仅有青年学者、中年专家，更廖卫平、张国君教授等资深专家，现在激烈的讨论和思想碰撞让大家更加深入了解会议的内容与主题。

合作是青委会突出特点，今年有 11 个多中心合作项目进行了展示，而且 3 个项目为前期开展项目的进展汇报，青年学者会对多中心项目积极性非常高，而且部分项目已经形成初步成果，相信 2024 年的学术年会上会有优秀成果进行报告。也希望越来越多的青年专家持续合作，不断产生有较大影响力的研究结果，发出更多中国青年癫痫医生的声音。

人文环节是青委会年会的传统和特色项目，本次人文活动的主题是“智能驱动未来，创新成就卓越”，活动的主场有着“国之重器”之称的徐工集团。本次分为三个阶段，第一个阶段是由徐工集团的金牌讲解员在会场进行了徐工发展历程、先进智造和徐工精神的介绍，3 月 24 日下午 4 点驱车到达徐工生产车间，让大家第一次近距离领略工程机械之美，感知新时代、筑梦新时代。医学与工程机械制造业虽有着行业之隔，但在强国建设与民族复兴中承担着同样的重任，既需要工匠精神和担当，也需要自主创新与执着坚持。随后，开展了人文知识竞赛检验学习收获。

青年癫痫学者联谊茶话会是展示学术之外青年人青春活力和多才多艺的舞台，也是一次放飞自我为年轻充电、展示真诚增进友谊的过程。今年联谊会的主题是“青春筑梦，汉韵彭城”。首先是徐州癫痫工作者联合表演的节目“德润古彭、晁采新徐”，带我们穿越古今，领略汉文化之美和徐州发展。此后八大区分

别组织了地方特点明显的舞蹈、歌伴舞、豫剧、合唱、诗朗诵等节日，最后秘书处和爱心企业一起表演和小合唱。今年首次以茶话会的形式举办，大家观看节目更专心，联谊会的效果也更显著。

本次会议的成功举办是在中国抗癫痫协会秘书处张慧老师和段立嵘秘书长等人员的精心策划和细心准备下完成。江苏省抗癫痫协会杨天明创会会长、李岩会长、徐州市儿童医院领导、刘晓鸣主任、徐州医科大学附属医院樊红彬主任带领的团队为会议举办做了全面的支持和准备工作。三位会长、多位中抗副会长、省协会会长和常青委们专程参会讲话和指导，青委会和八大区的负责人员专门召开了全体会议讨论相关学术和人文工作，并建立了多个小组分工负责，全体青委热情饱满地全程参加了会议，部分不能到会青委也按要求履行了请假手续。每一次会议的成功举办都有许多台前幕后人员的默默付出和支持。

2025，相聚武汉！

菁 YOUNG 计划 2023 年病例总决赛

菁 YOUNG 计划旨在提升青年医生在癫痫疾病领域的诊疗水平，推动精准医疗的发展。2023 年菁 YOUNG 计划以病例比赛为主要形式，2024 年 3 月 24 日历经海选、半决赛，共 9 位选手闯进在徐州举办的决赛。会议专门邀请了邓艳春副会长、李岩教授、孙美珍教授、徐淑军教授和青委会副主委等进行点评和作为评委。

最终，经过激烈的竞争和评委们的严格评审，耿磊钰医生凭借其对“反反复复中追寻——隐匿的变异”病例的深入分析和精彩汇报，荣获大赛一等奖。黄晓利医生和李光健医生分别获得二等奖和三等奖，其他选手（马洁卉、张洪伟、徐达、张歆博、沈春红、于杰罡）也以其优异的表现获得优秀奖。本次赛事展示出青年医生的专业素养和丰富的临床经验，更呈现出 CAAE 青年医师积极思考、勇于争先的精神风貌。

菁 YOUNG 计划 2024 年北区辩论赛

2024 年菁 YOUNG 计划围绕基因诊断的意义和合理应用等将开展四场辩论赛，由八大区两两捉对开辩。为更好的辩出水平，青委会专门组织的相关赛前培训，由周列民副会长为八大区负责人、辩论赛领队和骨干人员进行了辩论赛形式和技巧的讲解。为了更好的促进两支队伍对主题的全面深入了解，今年辩论赛采用不提前设立正反方形式，仅在开赛前 24 小时才抽签决定正反方队伍。每支队伍设领队 1 名， 辩手 5-6 名（含替补队员 1-2 名）。

首场辩论赛于 2024 年 5 月 26 日在天津举办，辩题是“基因检查对指导癫痫临床治疗是否有重要意义”，李文玲教授带领的京津冀晋蒙大区代表队持正方观点，而梁建民教授带领的黑吉辽大区代表队持反方观点。会议专门邀请了秘书处张慧老师、天津抗癫痫协会杨卫东会长、山西省抗癫痫协会会长孙美珍教授、第一届青委副主任委员张月华教授和孙伟教授，以及杨志仙教授到会指导。陶哲教授担任辩论主持，经过陈述、攻辩、自由辩论和总结陈词四个阶段的精彩辩论，最终京津冀晋蒙大区代表队以 1 分险胜。

首场辩论赛精彩纷呈，体现了青年委员们深厚的知识沉淀、精心的策划准备、成熟的辩论技巧和阳光的形象心态。通过辩论大家更加深入认识到合理基因检测和准确结果分析的重要意义，必将会使更多的癫痫患者早期得到合理诊治，提高癫痫控制水平和生活质量。期待后期更加激烈的三场辩论赛。

CHINA ASSOCIATION AGAINST EPILEPSY YOUTH COMMITTEE

THINKINGS AND ACTIONS

统筹策划	张　慧　段立嵘　梁树立
刊头题字	李世绰
资料来源	全体青年委员　CAAE网站
资料收集	丁　瑶　关宇光　何　芳 李光建　梁树立　刘晓英 倪冠中　闫志强　张春青
资料整理	刘婷红
排版美工	刘婷红　王雅婷