



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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寄语专家介绍：李世緯，研究员，中国抗癫痫协会创会会长；国际抗癫痫联盟（ILAE）执行委员会公共卫生顾问，ILAE 全球宣传委员会成员（2017 - 2021）。曾任世界卫生组织助理总干事、驻南太平洋地区代表（1998 - 2003），国家卫生部国际合作司司长（1992 - 1997），北京市卫生局副局长（1987 - 1992），北京市神经外科研究所副所长（1987 - 1992），美国国立卫生研究院（NIH）作访问学者和访问科



学家（1983 - 1986）。致力于神经系统疾病流行病学研究、脑血管病社区人群综合性预防研究和癫痫社会控制的研究，发表学术论文 70 余篇，主编《神经系统疾病流行病学》《中国癫痫预防与控制绿皮书》等，获得国家和北京市科技进步奖十余项。2009 年获得国际抗癫痫联盟（ILAE）和国际癫痫病友会（IBE）“癫痫大使”奖（我国第一人）。2019 年获中国医师报该年度“推动行业前行的力量 - 十大医学杰出贡献专家”。2021 年获得“ILAE、IBE 终生成就奖”（我国第一人）。

CAAE 创会会长寄语青年委员

非常赞赏和喜欢中抗和青委：我们协会所有的成员事业心都非常强，而且大家非常和谐，不搞山头，互相之间的交流合作特别活跃，青委也是这样。所以我对青年委员们的活动非常赞赏。咱们的青委成员做为有一定成就的科技工作者和学者，现在都三、四十岁，正值当年，是一个学者的最佳年龄段，这个时期精力旺盛，事业初步有成，工作当中是骨干。同时也是负担最重的时期，不管是家庭还是工作方面。所以对这个最佳的年龄段一定要珍惜，一定要过好。

青年人都有成长阶段：以我为例，从 19 岁（1960 年）到现在学医 62 年，从一个西北穷山沟的卫生院里的大夫，到后来出任世界卫生组织的助理总干事、驻南太平洋地区的代表，这个过程也很有趣。我没有什么家庭背景，两岁时父亲去世，依靠母亲给别人织毛衣、洗衣服挣一点钱，供我上学长大。从一个农村医生到世界卫生组织的高级官员过程中，看似“过五关斩六将”，但是实际上，也有好几次的“夜走麦城”。

青年人应当不忘初心：我退休以后专注于癫痫，总得来说要有不忘初心、坚持向上的劲头。不久前我应邀给复旦大学附属华山医院的青年医师讲了一次课，题目叫《悬壶初心，济世全球》，我的前半生做为临床医生，治病救人做为“悬壶”；后来转到公共卫生领域，在做卫生行政工作。这不同于临床医生的工作，治疗一个个的病人，而是面向大众健康事业的，可谓“济世”。听过我的课后，有一个青年专家告诉我她的感受：“一定要保持正能量，秉承治病救人的初心，勇往直前”，这正符合我讲课的目的。我并非强调自己的成就，只是从我个人走的路，深感青年医生要有志向，记住自己的初心，就是治病救人。做为一名医务工作者，要面向大众，面向病人。

青年人应该有志向、理想和抱负：人们常说“某某人有野心”，野心这个词，听着是一个贬义词，但我理解为一个褒义词。因为心如果不野一点，怎么去做更大的事、更多的事？完全守着自己的小天地，没有野心，又怎能做出更大工作和贡献呢？所以我认为青年一代要有自己的志向和抱负，这其中包括两个方面：一方面如顾硕医生，是目前医院的党委书记，还有青委的副主委陈蕾医生，担任华西医院的副院长，在管理方面有所成就。另一方面，更多的人要在业务方面有自己的创造性成就。

学一学“科学心理学”：这门学科是研究科学创造活动中的心理规律和心理结构。其一，大家熟知的智力有几个要素：观察力、记忆力、思维力、想象力和操作力。首先要求有观察能力，要善于观察医疗实践中的具体现象，特别是一些奇怪的、解释不了的现象。在观察的基础上，经过思维、想象、操作，来达成新的成果。最简单的例子，大家都知道，牛顿是因为苹果掉在地上发现了重力，瓦特发现蒸汽吹起了茶壶盖，从而发明了蒸汽机。说明从观察到思维想象到操作，智力因素起了重要的作用。其二，还有非智力因素，包括兴趣、情绪、意志和性格，要对自己周围的世界感兴趣、有意志，才能做出科学创造。其三，灵感和机遇在科学创造活动当中的地位也是很重要的。我最近看到一些青年专家，如广州医学院的邓宇虹在研究脑肠轴，用益生菌治疗难治性癫痫，操德智医师报道了一例托吡酯治愈 West 综合征等等，对我们来说是一些很新的现象，如果我们能抓住这些现象，进行深入思考和研究，就有可能取得好的成果。希望大家能够学习科学创造中的心理规律，增强创造的自觉性，减少盲目性，对于攀登科学技术

高峰有重要的意义。

希望青委们要勇于打入国际：我几次强调“我们在国际抗癫痫联盟中的代表性不足”，我们有 1 万余名中国抗癫痫协会的会员，6000 多个注册会员。如此大的协会，在 ILAE 的领导机构里，我们的占比很小。我去年因年龄关系辞去了兼任的职务，但是谁能去代替我，是我现在经常考虑的问题。我的目光现在集中在青委里一些有实力、有成就的青年专家中，希望你们可以拿出自己的业务成绩，善于表现自己，勇于沟通，主动出击，争取在国际抗癫痫联盟和其它国际组织中有更多的中国专家加入其决策和专业领导机构中。

希望大家处理好与老专家、老师及下级医师的关系：起到承上启下的作用，要谦虚谨慎，要继承和突破，要青出于蓝而胜于蓝，我希望你们成为都能胜于蓝。

这几点就是我一个“老头”，一个过来人，对我们青年医师的希冀，供大家参考。

（根据李会长在 3 月 12 日青委会学术交流会讲话整理，并经审核、修订）

读后感：

李世焯创会会长在 2022 年 3 月 12 日第二届青委会的首次年会上致辞中，重点结合他个人成材和发展的过程，给年轻人提出了一些指导意见。虽然我知道李会长要讲的主题，但是当日听到这个讲话，还是非常震撼和感动，会后和几个青委的副主委商量，向李会长提出了把讲话内容整理后刊登的想法，得到了李会长热情的支持并亲自进行了审核、修订。

李会长首先以中抗为例告诉我们团结的重要，未来一定是多学科合作、多团队协作和多中心研究的时代。同时，教导我们要珍惜青春，要有志向、有理想抱负、要有国际视野。李会长还提醒青委们成材不是一蹴而就，要有不同的阶段，学会坚持，学会面对困难，敢于处理问题，特别是要依靠和尊重老师与老专家，处理好同事间的关系，形成合力。期间，李会长还向青委们推荐了研究科学创造活动中心理规律和心理结构的“科学心理学”，供大家学习，并特别提到了几位青委的工作。通过李会长简短、朴实而真诚的讲话，传递的是他几十年学习、生

活和工作（包括医生、研究员和行政领导）感受，更体现了他对癫痫事业和年轻癫痫医生的关心、爱护和期望。

青年人有激情、有想法，可以成为大江大河和奔腾的骏马，但如果不能很好把持，也可能成为泛滥的洪水或者脱缰的野马，需要前辈们的经验和指导。现在把这个讲话稿刊登出来，以便让更多的年轻医务人员学习，同时也可以在我们遇到困难和问题，再次学习，汲取能量，更好的健康成长。

（梁树立）



执信念之灯，照生命之境

CAAE 青年委员会粤桂琼闽大区

中山大学第一附属医院神经科 尹思静 龙定菊

“所信者目也，而目犹不可信；所恃者心也，而心犹不足恃。”

——孔子

病人以生命相托，我们便应该心无杂念全力以赴。生不能为相济世，亦当为医救人。所谓病情，发言容易，或持有据快论，而病源未必相符。病魔的伪装以及在医学岗位上的坚守，让我们不得不对自己种种苛求。

“医生，19 床病人已经到了。”伴着护士清脆的嗓音，我拿上笔记本和查体工具去到病房。那时她戴着口罩端正地坐在木椅上，看不清她脸上的表情，但眼里全是焦躁和不安。护士老师忙碌而熟练的给她安装脑电图电极，我在一旁开始了自己病史询问。她是一个 17 岁的女孩，因为“反复发作性意识不清伴四肢抽搐 7 年余”被姑姑带到我们医院诊治。在这 7 年间，她反复出现双眼上翻、肢体抽搐、牙关紧闭、面色发绀、呼之不应。姑姑也曾多次将她带至当地医院就诊，无一例外地被诊断为“癫痫”。患者自己也不规律的服用抗癫痫药物，尝试过各种民间“偏方”，但是治疗效果均不理想，发作频率也从最开始的一年几次逐渐发展为近期一晚数次。乍一看病史也并没有让我很惊讶，这不就是典型的癫痫，因为不遵医嘱服药而导致的症状控制不良吗？根据这些日子的临床经验，我不疑有他，迅速开好医嘱写完病历并交完班就下班了。

她如此不幸，因为长年被疾病困扰，却又是如此幸运，因为当天深夜“抽搐”又多次挑衅，视频脑电监测记录了她发作的全部过程。她发作开始时自觉胸闷心悸，随后面色苍白，逐渐意识不清，出现挣扎样动作。同步的心电监测可见“室扑”样改变，随后脑电全导慢活动。真相呼之欲出，再次追问了病史，我了解到她在每次抽搐前都有心慌、胸闷、呼吸困难等不适。我们立即联系心内科会诊转科。

随后的几天里，在 CCU 持续心电监护的过程中，她的心电记录提示“尖端扭

转型室速”，结合病史最后诊断为“长 QT 间期综合征”，后来便做了“双腔 ICD 植入术”，手术后便没有在出现过抽搐。那天我路过 CCU 去看她，她已经收拾好行李准备出院了，眉目里掩不住的喜悦，她说“我终于可以和其他正常女孩子一样了，回去后我想继续上学”。

这看似平常的抽搐，却令人细思极恐。回顾这个女孩的病史，在这 7 年之中，死神之手似乎已经扼住她的喉咙，只要稍加用力就能为这个年轻的生命划上死亡的句号。即使患者每一次都能从死神之手侥幸逃脱，但是这一次次看似短暂的发作，都可能是损伤患者的脑组织等脆弱而重要的脏器的利器，长此以往，往往会造成难以挽回的损伤，严重影响她本人及家人的生活质量。此外，患者长期以来被当做癫痫治疗，长期服用抗癫痫药物，不恰当的治疗不仅不能解决患者的病痛，而且还会带来或轻或重的药物不良反应。

她在这七年时间里，怀着期望最后失望地辗转于各个医院，也曾做过一些检查，但是却没有查到具体原因。身为管床医生的我也不由重新自我审视：作为一个刚出象牙塔便进入临床的年轻癫痫专科医生，接触了一些表现形式不同的“癫痫”，我自然而然地把见到的每一个以痫样发作为主的病人都归结于癫痫，给他们诊断为“癫痫”或者某种“癫痫综合征”。但是，眼见就一定为实吗？身为医生，我们必须用严谨的态度对待每一个患者，做好甄别，尽量给患者一个准确的诊断是我们的职责，也是我从这个女孩的经历中学到的教训。

在她身上，这 7 年来，我们与病魔交手为何屡战屡败？随着科技日新月异的发展，各种辅助检查技术更新迭代，临床的诊疗工作却因为更依赖这些冰冷的仪器，而弱化了本该熟练掌握的病史询问及体格检查。的确，“心源性晕厥”与“癫痫”从临床症状上非常难以鉴别，但她的病程中在意识丧失前反复出现的心悸、胸闷、呼吸困难等表现开始却也被我忽视，如果我有足够的临床基础累积，不把思维局限于专科本身的疾病，是否能早一些发现她有心脏病变？如果在查体时不单单把重点局限在神经系统，而是认真的完成系统的全身体格检查，我是否能从心脏听诊中发现异常，从而尽早为她找到病因？但是，恰当的辅助检查也不可或缺的，要是这次住院她发作的同时没有心电去监测到心电图的变化，那我是否会继续把她当做“癫痫”治疗？

我满怀热忱步入临床，以为自己可以如电视剧里的青年医生那样信手拈来轻

松解决患者的病痛，但现实的诊疗情况却山重水复，有不少的病人因为种种原因被漏诊或者误诊，他们用自己的健康甚至生命成为我们成长路上的教材，而我们要做的就是在漫漫医学之路不畏艰辛夯实基础，不畏浮云总结教训，不断地提高自身的能力，及时给患者做出最佳诊治方案。通过对这个病历的回顾和思考，我受益匪浅。William Osler 曾说：“如果你认真倾听你的病人，他们会告诉你正确的诊断”，临床工作固然繁忙琐碎，但详细了解病史是我们正确诊疗的开端。在学好自己专科相关知识和技能的同时亦需要兼顾其他各科知识的输入融合，拥有全面的知识储备，不仅是诊疗工作的基础，更是精准诊疗必备基石。

想起她出院时候的笑容，我知道一种叫做职业成就感的東西已在我心里悄然生长，让我以更加坚定的步伐迈向医学之路的远方。

医患一家，同心筑梦

CAAE 青年委员会川渝滇黔藏大区

四川大学华西二院神经科 甘靖

工作在医院，我们有幸能参与到很多人与疾病抗争的过程。以精湛的技术为矛，与患者并肩作战；以人文关怀为盾，悉心保护因疾病而煎熬的内心。我们与患者或许是初相逢，或许曾多次共抗病魔，理解、关爱与尊重从不缺席。用心付出的同时，患者的满意度、依从性悄然提升，医务人员内心的人文关怀之花也随之盛放。

陌生的医院环境、陌生的医务人员、各种不适的检查与治疗……都在加剧患儿的恐惧与不安，他们用哭闹与不配合抗争。我们心疼的同时也希望做点什么。2016 年四川大学华西第二医院团委与护理部共同发起人文关怀项目“Hi，患儿

儿童医疗辅导”，我们儿童神经科作为示范病房参与其中。第一次活动是“小丑医生”。当我们戴上小丑的红鼻子，穿上小丑的服装，小朋友们竟然愿意主动亲近我们了。那一刻，我们是为小朋友们治疗的医生、护士阿姨，也是让他们开怀大笑的好朋友。初次人文关怀触电，让我们充满惊喜，同时，也带给我们无限的想象力。我们期待以更多的人文关怀方式改善住院体验！

我们设置画展墙，为小朋友们的涂鸦设置了一处展示的天地。绘画是情绪的释放，也是治疗。给予我们走进小朋友内心的地图，让我们看到小朋友的无助，让我们知道怎么温暖和帮助他们。也让我们看到小朋友的梦想，惊叹于他们的乐观与坚强。也有很多小朋友将他们的感谢与梦想画在了纸上，让我们深切感受到温暖他人的同时也在被他人所温暖！也更有藏区小朋友为我们医务工作者谱写的诗词，让我们也感受到了力量！



还记得那个 4 岁多的小女孩，因为考虑额叶癫痫来到我们病区。频繁的发作、治疗效果的不佳以及沉重的家庭经济负担让患儿与家属疲惫不堪。医生们反复的讨论、研究患儿的病历，考量患儿的家庭经济条件，选择最适切的检查 ASL，出结果后积极联系外科，在最短的时间内解决患儿的问题。术后恢复良好的她回到病房，我们的画展墙上展出了她的作品，画作中的甘医生成为了她心目中的“超人”！

纪念尊敬的黄希顺教授

CAAE 青年委员会湘鄂豫大区

郑州大学第一附属医院神经科 张海峰

2021 年 11 月 23 日 11 时 58 分我省著名的癫痫病专家、领航人黄希顺教授因病逝世，享年 73 岁。黄老师一生投身医疗事业，孜孜以求，刻苦研究医疗理论，对技术精益求精，注重总结经验，勇于实践，不断探索。黄老师在郑州大学第一附属医院工作 40 年，其中 20 多年的时间在与病痛斗争，但依旧坚守岗位，战斗在抗癫痫事业的第一线。黄老师在 1981 年出版的《丙戊酸药物的临床及药理研究》，是国内第一个关于抗癫痫药物的中文版专著；1982 年开设河南省首个癫痫门诊，是我省癫痫治疗的创始人；1988 年获得国际抗癫痫联盟课题，随后申请到我省卫生系统及我院的第一个国家自然科学基金；1992 年，出版癫痫常识的科普读物《癫痫防治 200 问》，深受到海内外患者的好评，多次再版。黄老师桃李满天下，培养的许多研究生已成为我省医疗界的中坚力量。从上世纪 80 年代，黄老师就开始做癫痫病史登记，编码存档，以便于随时调取，分析研究，一做就是 20 年。黄老师的去世，不仅是我院的一大损失，也是我省抗癫痫事业的重大损失。他虽然离开了我们，但他高尚的品质和崇高的思想永远活在我们的心中。

初识黄老师是在 2008 年，记得那年我读研究生，导师是连亚军教授，和黄老师都是研究癫痫疾病的，同在一个科室对他也慢慢的熟悉起来。印象中：黄老师教学很严谨，对学生的要求很严格，再忙也会边看病边教导学生，发现学生出现错误马上就纠正；另外，黄老师神经内科专业功底很深厚，其实他不仅仅是癫痫看的好，按黄老师自己的话说是“因为看癫痫太出名所以门诊基本被慕名而来的癫痫病人包了，别的病慢慢就没有机会看了”，随着时间沉淀这种现象越来越明显。听过黄老师分析神经内科病例，觉得思路非常清晰、对神经解剖非常精通；还有，黄老师知识很渊博、很爱国、爱家乡，历史、地理、军事、民族等等几乎无所不知，无论说什么话题都是滔滔不绝。黄老师那代人经历过很多艰难困苦，常提毛主席理论（特别是实践论），总是说国家和家乡的好，对老乡也很好，当然对患者也很好。再就是一些记忆碎片了，比如：带着我们用利多卡因控制了一

个非常难治、近持续的局灶性癫痫发作；看到回族同胞给我们讲穆斯林的起源；把自己比作川军抗日英雄王铭章；给我们讲过去用电休克法治疗精神病（黄老师曾在驻马店精神病院干过几年）等等。直到最近这些年才知道黄老师居然已经受脊索瘤折磨了 20 多年，但当时我们都没有看出来，现在才明白黄老师每天忍着病痛奋斗在抗癫痫的第一线，直到最近几年因行走不便才逐渐减少门诊量（仍坚持坐轮椅上门诊），去年因必须住院治疗才彻底停止工作。

最近，黄老师的自传《玉汝河》出版了，科里的大夫们看到都舍不得放下了。黄老师的一生很精彩，从贫苦农民的孩子到读小学、初中、高中，再到当革委会副主任、大队干部、教师、工农兵大学生、驻马店精神病院医生、读华西的研究生、拿 ILAE 课题和河南医学界第一个国家自然科学基金、与假药贩子斗争...但也让我们感到难过，哪里知道黄老师从 97 年就开始不断的做手术，几乎一两年要做一次。随后大概 12 年以后，骶尾部上一一直有一个不能愈合的洞需要不断换药，期间黄老师坚持外出参加学术会议、讲课，每次都要带着好几个换药包，讲课前换上，讲完课换掉。最近这两年黄老师住在我们科，我也做过他几个月的主管医生，黄老师每次都是用笑容迎接我们查房，但我们却不忍看他那深可见骨的窦口。去年底，我要去廖卫平教授那里学习，他知道后就找我聊起他们当年的往事，说到兴起仿佛病痛都消失了一般...

黄老师遇到难题不断探索和钻研，遇到痛苦乐观而无畏，积极向上。黄老师走了，但他乐观、坚强、忘我、利他的精神，值得我学习一辈子。我们要继承他的遗愿，求真务实、努力拼搏；同时，我们也要学习连亚军主任等对黄老师的敬重和悉心照顾，做好一代代的传承，团结协作，为我省卫生事业的发展而努力奋斗。

黄老师一路走好，天堂没有病痛。

编号: XEY-2022-1-1

引用格式: Shi Y, Chen G, Sun D, Hu C, Liu Z, Shen D, Wang J, Song T, Zhang W, Li J, Ren X, Han T, Ding C, Wang Y, Fang F. Phenotypes and genotypes of mitochondrial diseases with mtDNA variations in Chinese children: A multi-center study. *Mitochondrion*. 2022;62:139-150. doi: 10.1016/j.mito.2021.11.006.

第一作者: 孙丹

Abstract

Mitochondrial DNA (mtDNA) associated mitochondrial diseases hold a crucial position but comprehensive and systematic studies are relatively rare. Among the 262 patients of four children's hospitals in China, 96%-point mutations (30 alleles in 11 genes encoding tRNA, rRNA, Complex I and V) and 4%-deletions (seven of ten had not been reported before) were identified as the cause of 14 phenotypes. MILS presented the highest genetic heterogeneity, while the m.3243A > G mutation was the only "hotspot" mutation with a wide range of phenotypes. The degrees of heteroplasmy in the leukocytes of MM were higher than MELAS. The heteroplasmy level of patients was higher than that in mild and carrier group, while we found low-level heteroplasmy pathogenic mutations as well. Some homoplasmic variations (e.g., m.9176 T > C mutation) are having high incomplete penetrance. For a suspected MELAS, m.3243A > G mutation was recommended to detect first; while for a suspected LS, trios-WES and mtDNA genome sequencing by NGS were recommended first in both blood and urine.

编号: XEY-2022-1-2

引用格式: Guang S, Mao L, Zhong L, Liu F, Pan Z, Yin F, Peng J. Hormonal Therapy for Infantile Spasms: A Systematic Review and Meta-Analysis. *Front Neurol*. 2022;13:772333. doi: 10.3389/fneur.2022.772333.

通讯作者：彭镜

Abstract

Objective: The limitations of adrenocorticotrophic hormone (ACTH) treatment for infantile spasms (ISs), such as high costs, limited availability, and adverse effects (AEs), make it necessary to explore whether corticosteroids are optimal alternatives. Many other compelling treatments have gone through trials due to the suboptimal effectiveness of hormonal therapy. A systematic review and meta-analysis were performed to evaluate the effectiveness and safety of hormonal therapy for patients with ISs. **Methods:** EMBASE, Ovid MEDLINE, Cochrane Central Register of Controlled Trials (CENTRAL), and online registers were searched through April 2021 for randomized controlled trials (RCTs). **Results:** A total of 19 RCTs (N = 1,279) were included. There was no significant difference in the effectiveness of oral corticosteroids and ACTH in electro-clinical response (risk ratio [RR] = 0.85, 95% CI 0.41–1.76). Low-dose ACTH had similar effectiveness in electro-clinical response compared to usual-dose group (RR = 0.94, 95% CI 0.60–1.47) but conferred a lower risk of AEs (RR = 1.71, 95% CI 1.08–2.71). ACTH was more beneficial in controlling spasms than vigabatrin (VGB) (RR = 1.31, 95% CI 1.05–1.64) for patients without tuberous sclerosis complex (TSC). All RCTs were connected through network meta-analysis, and we found that ketogenic diet (KD), zonisamide, methylprednisolone, or combined treatment of hormonal therapy with topiramate (TPM) or pyridoxine was not different in electro-clinical response compared to usual-dose ACTH. **Conclusion:** Our analysis showed that oral corticosteroids could be optional alternatives when ACTH is not applicable, and ACTH is more beneficial for patients without TSC. Moreover, low-dose ACTH is recommended due to comparative effectiveness but lower risk of AEs. However, due to the high heterogeneity of included patients and treatment protocols, these results must be interpreted with caution. RCTs with multicentric involvement and larger sample size are needed for solid evaluation of other alternative treatments.

编号: XEY-2022-1-3

引用格式: Peng P, Kessi M, Mao L, He F, Zhang C, Chen C, Pang N, Yin F, Pan Z, Peng J. Etiologic Classification of 541 Infantile Spasms Cases: A Cohort Study. *Front Pediatr.* 2022;10:774828. doi: 10.3389/fped.2022.774828.

通讯作者: 彭镜

Abstract

Objective: To explore the etiology of infantile spasms (IS) in a large Chinese cohort based on the United States National Infantile Spasms Consortium (NISC) classification. **Methods:** In the present study, we recruited IS patients diagnosed at a single center (Xiangya Hospital, Central South University) between Jan 2010 and Aug 2019. Thereafter, we collected their clinical and genetic information retrospectively. Their underlying etiologies were classified according to the NISC classification and then compared in different scenarios to understand their distribution. **Results:** A total of 541 patients with IS from 18 provinces were included in this study. The underlying etiology was identified in 53.2% of the cases: structural-acquired, 25.3%; genetic, 12.9%; genetic-structural, 7.2%; structural-congenital, 5.0%; metabolic, 2.4%; infections, 0.4% and immune, 0%. Whole-exome sequencing (WES) provided the highest diagnostic yield (26.9%). In structural-acquired IS, the proportion of hypoglycemic brain injuries was significant, second only to hypoxic-ischemic encephalopathy. There was no patient discovered to have Down syndrome. STXBP1, CDKL5, TSC2, KCNQ2, IRF2BPL, and TSC1 were the most frequently implicated genes. Genetic causes were found to be the most common cause of IS in the early onset group, while structural-acquired etiologies were common in males and preterm babies. Patients with pre-spasm seizures were associated with a higher proportion of identified causes than those without. Nonacquired structural etiologies were more common in patients without hypsarrhythmia than in those with hypsarrhythmia. **Significance:** The most prevalent cause of IS was structural acquired followed by genetic causes. When brain MRI fails to detect the etiology, we propose WES as the next step. Structural-

acquired IS and cases with genetic disorders are characteristic of the Chinese cohort, however, the etiology differs with the patient's age of onset, gestation age at birth, sex, and the presence/absence of both pre-spasm seizures, and hypsarrhythmia.

编号: XEY-2022-1-4

引用格式: Sun Y, Peng J, Liang D, Ye X, Xu N, Chen L, Yan D, Zhang H, Xiao B, Qiu W, Shen Y, Pang N, Liu Y, Liang C, Qin Z, Luo J, Chen F, Wang J, Zhang Z, Wei H, Du J, Yan H, Duan R, Wang J, Zhang Y, Liao S, Sun K, Wu L, Yu Y. Genome sequencing demonstrates high diagnostic yield in children with undiagnosed global developmental delay/intellectual disability: A prospective study. Hum Mutat. 2022. doi: 10.1002/humu.24347.

第一作者: 彭镜

Abstract

Genome sequencing (GS) has been used in the diagnosis of global developmental delay (GDD)/intellectual disability (ID). However, the performance of GS in patients with inconclusive results from chromosomal microarray analysis (CMA) and exome sequencing (ES) is unknown. We recruited 100 pediatric GDD/ID patients from multiple sites in China from February 2018 to August 2020 for GS. Patients have received at least one genomic diagnostic test before enrollment. Reanalysis of their CMA/ES data was performed. The yield of GS was calculated and explanations for missed diagnoses by CMA/ES were investigated. Clinical utility was assessed by interviewing the parents by phone. The overall diagnostic yield of GS was 21%. Seven cases could have been solved with reanalysis of ES data. Thirteen families were missed by previous CMA/ES due to improper methodology. Two remained unsolved after ES reanalysis due to complex variants missed by ES, and a CNV in untranslated regions. Follow-up of the diagnosed families revealed that nine families experienced changes in clinical management, including identification of targeted treatments, cessation of unnecessary treatment, and considerations for family planning. GS demonstrated high

diagnostic yield and clinical utility in this undiagnosed GDD/ID cohort, detecting a wide range of variant types of different sizes in a single workflow. **KEYWORDS** clinical utility, diagnostic yield, genome sequencing, global developmental delay, intellectual disability

编号: XEY-2022-1-5

引用格式: Hou J, Zhu H, Xiao L, Zhao CW, Liao G, Tang Y, Feng L. Alterations in Cortical-Subcortical Metabolism in Temporal Lobe Epilepsy With Impaired Awareness Seizures. *Front Aging Neurosci.* 2022 Mar 10;14:849774. doi: 10.3389/fnagi.2022.849774.

通讯作者: 冯莉

Abstract

Objective: The features of cerebral metabolism associated with loss of consciousness in patients with temporal lobe epilepsy (TLE) have not been fully elucidated. We aim to investigate the alterations in cortical-subcortical metabolism in temporal lobe epilepsy with impaired awareness seizures (IAS). **Methods:** Regional cerebral metabolism was measured using fluorine-18-fluorodeoxyglucose positron emission tomography (18F-FDG PET) in patients with TLE-IAS and healthy controls. All patients had a comprehensive evaluation to confirm their seizure origin and lateralization. Videos of all seizures were viewed and rated by at least two epileptologists to identify the state of consciousness when a seizure occurred. By synthesizing the seizure history, semeiology, and video EEG of all patients, as long as the patients had one seizure with impaired awareness, she/he will be included. 76 patients with TLE-IAS and 60 age-matched healthy controls were enrolled in this study. Regional cerebral metabolic patterns were analyzed for TLE-IAS and healthy control groups using statistical parametric mapping. Besides, we compared the MRI-negative patients and MRI-positive patients with healthy controls, respectively. **Results:** There were no significant differences in the age and sex of TLE-IAS patients and healthy

control. TLE-IAS patients showed extensive bilateral hypermetabolism in the frontoparietal regions, cingulate gyrus, corpus callosum, occipital lobes, basal ganglia, thalamus, brainstem, and cerebellum. The region of metabolic change was more extensive in right TLE-IAS than that of the left, including extensive hypometabolism in the ipsilateral temporal, frontal, parietal, and insular lobes. And contralateral temporal lobe, bilateral frontoparietal regions, occipital lobes, the anterior and posterior regions of the cingulate gyrus, bilateral thalamus, bilateral basal ganglia, brainstem, and bilateral cerebellum showed hypermetabolism. The TLE patients with impaired awareness seizure showed hypermetabolism in the cortical-subcortical network including the arousal system. Additionally, 48 MRI-positive and 28 MRI-negative TLE-IAS patients were included in our study. TLE-IAS patients with MRI-negative and MRI-positive were both showed hypermetabolism in the cingulate gyrus. Hypometabolism in the bilateral temporal lobe was showed in the TLE-IAS with MRI-positive. **Conclusion:** These findings suggested that the repetitive consciousness impairing ictal events may have an accumulative effect on brain metabolism, resulting in abnormal interictal cortical-subcortical metabolic disturbance in TLE patients with impaired awareness seizure. Understanding these metabolic mechanisms may guide future clinical treatments to prevent seizure-related awareness deficits and improve quality of life in people with TLE.

编号: XEY-2022-1-6

引用格式: Wu Y, Huang K, Wen S, Xiao B, Feng L. Validation of the Chinese Version of the Stigma Scale of Epilepsy. *Front Neurol.* 2022 Feb 7;13:796296. doi: 10.3389/fneur.2022.796296.

通讯作者: 冯莉

Abstract

Purpose: This study was carried out to test the validity and reliability of the Chinese version of the Stigma Scale of Epilepsy (SSE), with aim to better understand the public

stigmatizing attitudes of epilepsy in China and help elucidate stigma determinants for interventions. **Methods:** The SSE was translated into Simplified Chinese Mandarin. In this study, most of the participants were enrolled via convenience sampling by randomly distributing questionnaires on the streets and parts of the participants were recruited by an online platform named Wenjuanxing. We assessed the psychometric properties of the SSE in 310 Chinese native-speaker. Cronbach's alpha was tested for reliability. Index of Content Validity (CVI) was calculated. Exploratory and confirmatory analysis were used to explore the factor structure and verify the validity of SSE. **Results:** The Cronbach's alpha is 0.936 for the overall scale, and the CVI value is greater than 0.78. The exploratory factor analysis (EFA) extracted SSE six factors: the fear of seizure attacks (factor 1), sympathy for patients with epilepsy (PWEs) (factor 2); difficulties faced by PWEs (factor 3); speculation on PWEs' feeling (factor 4); discrimination against PWEs (factor 5); and knowledge about epilepsy (factor 6). The item 13 was proven to be problematic and has been eliminated. The confirmatory factor analysis (CFA) ensured the great construct validity ($\chi^2/SD = 1.725$, goodness of fit index (GFI) = 0.916, and root mean square error of approximation (RMSEA) = 0.048), convergent validity (the factor loads of each item corresponding to each latent variable >0.6 , average variance extracted (AVE) >0.5 , and composite reliability (CR) >0.7), and discrimination validity (all of the absolute value of correlation coefficient are <0.5 , and less than the square root of AVE) of the SSE. **Conclusions:** The Chinese version of the SSE scale was a valid and reliable tool to measure epilepsy-associated stigma in the Chinese society.

编号: XEY-2022-1-7

引用格式: Huang K, Wu Y, He Q, Yang H, Du Y, Xiao B, Feng L. Validity and reliability of the Chinese version of the epilepsy stigma scale. *Epilepsy Behav.* 2022;127:108531. doi: 10.1016/j.yebeh.2021.108531.

通讯作者: 冯莉

Abstract

Objective: This study was conducted to test the validity and reliability of the Chinese version of the epilepsy stigma scale (ESS), which aims to better understand the stigma of patients with epilepsy (PWEs), lays the foundation for future investigation and explores appropriate strategies to mitigate PWEs' stigma in Chinese culture. **Methods:** The scale was translated following standard procedures. For psychometric validation, the Chinese version of the ESS was administered to 214 PWEs above the age of 16 who were diagnosed with epilepsy by two trained epileptologists and were taking anti-seizure drugs for at least a month. All of the patients were recruited from Xiangya Hospital of Central South University of China from August 2021 to September 2021. **Results:** The Cronbach's alpha coefficient was 0.893 for the entire scale, 0.903 for felt stigma, and 0.688 for enacted stigma. Exploratory and confirmatory factor analyses were conducted and showed that the scale was grouped under two dimensions, and the results of confirmatory factor analysis support the structure. **Conclusion:** The Chinese version of the ESS is a valid and reliable tool to assess epilepsy-related stigma in Chinese culture.

编号: XEY-2022-1-8

引用格式: Chen Y, Chen X, Kang H. Case Report: Moving Tumor-Like Foci Behind Refractory Epilepsy-Cerebral Sparganosis Successfully Treated by Surgery After Failure of Praziquantel Treatment. *Front Neurol.* 2022 Feb 10;13:838849. doi: 10.3389/fneur.2022.838849.

通讯作者: 康慧聪

Abstract

Cerebral sparganosis is clinically nonspecific and easily misdiagnosed, exposing patients to the risk of severe brain damage and neurological dysfunction caused by actively migrating larvae. Diagnostic biomarkers from typical cases can help to establish an early diagnosis and proper treatment. We present a 25-year-old woman who

suffered from nine years of refractory epilepsy and was misdiagnosed with glioma and subjected to surgery. The postoperative pathology confirmed granuloma, and the tumor-like foci reappeared three months later. Along with the “tunnel sign” on MRI, cerebral sparganosis was suspected and confirmed by positive serum and cerebrospinal fluid antibodies against *Spirometra mansoni*. The patient visited us after a failure of four cycles of praziquantel treatment, recurrent seizures and hemiplegia with basal ganglia foci. Craniotomy was not carried out until the larva moved to the superficial lobe on follow-up MRIs, and pathology revealed sparganosis granuloma. The patient became seizure-free and recovered myodynamia but had long-lasting cognitive dysfunction due to severe brain damage. This case indicated the importance of tunnel signs and moving tumor-like foci on MRI as diagnostic clues of cerebral sparganosis. An early diagnosis is vitally important to avoid severe neural dysfunction by the long-living and moving larvae. Surgical removal of the larva is a critical remedy for cases failed by praziquantel treatment.

论文收录说明

- 以八大区为单位收集，本期因部分地区和青委受疫情影响严重，相关论文待下一期同步收集；
- 收集范围包括 SCI（SCIE、ESCI）收录的论文、《ACTA Epileptologica》、《Epilepsia Open》、《癫痫杂志》发表论文；
- 收录的为当期正式发表，未正式发表的论文在收集后将于下一期收录；
- 收录格式为引用格式+摘要。

编号: HJL-2022-1-1

引用格式: Wan HJ, Hu WH, Wang X, Zhang C, Wang SS, Zheng Z, Zhou F, Sang Xia S, Yu X, Song F, Sun B, Wang Y. McLeod syndrome with a novel XK frameshift mutation: A case report. *Medicine (Baltimore)*. 2022;101(10):e28996. doi: 10.1097/MD.00000000000028996.

通讯作者: 王莹

Abstract

Introduction: McLeod syndrome (MLS) is a rare X-linked neurohematologic disorder caused by loss of function mutation in the XK gene, however, variations in the XK gene await elucidation. Here we reported the clinical phenotype and genetic features of a patient with MLS caused by a novel frameshift mutation of the XK gene. **Patient concerns:** The 44-year-old male patient presented chorea, cognitive impairment, mental disorders and seizures, accompanied by peripheral neuropathy, hyperCKemia, and acanthocytosis. The proband's mother had mild chorea. One elder brother who died 10 years ago without a confirmed diagnosis showed symptoms of both chorea and mental disorders, the other elder brother also developed mild chorea. **Diagnosis:** The patient was diagnosed with MLS based on the family history, clinical manifestations, and accessory examinations. Whole-exome sequencing studies revealed a novel frameshift mutation resulted from a nucleotide variation in exon 2 (452delA) which leads to an amino acid residue conversion from Gln to Arg and early termination of XK protein (Gln151ArgfsTer2). The patient and one of his elder brothers are hemizygotes, and his mother is heterozygous. **Interventions:** The patient was treated with haloperidol to control chorea and levetiracetam to control seizure. **Outcomes:** Six months after treatment, the proband was seizure-free but with little improvement in chorea and cognitive dysfunction. **Conclusion:** We described a family with MLS, which was caused by a novel frameshift mutation in the XK gene. The causes of mild clinical presentation of the proband's mother need further investigation.

编号: YGQM-2022-1-1

引用格式: Wan HJ, Hu WH, Wang X, Zhang C, Wang SS, Zheng Z, Zhou F, Sang L, Zhang K, Zhang JG, Shao XQ. Interictal pattern on scalp electroencephalogram predicts excellent surgical outcome of epilepsy caused by focal cortical dysplasia. *Epilepsia Open*. 2022. DOI: 10.1002/epi4.12587.

第一作者: 万慧娟

Abstract

Objective: Focal cortical dysplasia (FCD) represents an essential cause of drug-resistant epilepsy with surgery as an effective treatment option. This study aimed to identify the important predictors of favorable surgical outcomes and the impact of the interictal scalp electroencephalogram (EEG) patterns in predicting postsurgical seizure outcomes. **Methods:** We retrospectively evaluated 210 consecutive patients between 2015 and 2019. They were diagnosed with FCD by pathology, underwent resection, and had at least one year of postsurgical follow-up. Predictors of seizure freedom were analyzed. **Results:** Based on the information at the latest follow-up, seizure outcome was classified as Engel Class I (seizure-free) in 81.4% and Engel Class II-IV (non-seizure-free) in 18.6% of patients. There were 43, 105, and 62 cases of FCD type I, type II, and type III, respectively. The interictal EEG showed a repetitive discharge pattern (REDP) in 87 (41.4%) patients, polyspike discharge pattern (PDP) in 41 (19.5%), and the coexistence of REDP and PDP in the same location in 32 (15.2%) patients. The analyzed patterns in order of frequency were repetitive discharges lasting 5 seconds or more (32.4%); polyspikes (16.7%); RED type 1 (11.4%); continuous epileptiform discharges occupying >80% of the recording (11.4%); RED type 2 (6.2%); brushes (3.3%); focal, fast, continuous spikes (2.4%); focal fast rhythmic epileptiform discharges (1.43%); and frequent rhythmic bursting epileptiform activity (1.4%). The coexistence of REDP and PDP in the same location on scalp EEG and complete resection of the assumed epileptogenic zone (EZ) was independently associated with favorable postsurgical prognosis. **Significance:** Resective epilepsy surgery for

intractable epilepsy caused by FCD has favorable outcomes. Interictal scalp EEG patterns were revealed to be predictive of excellent surgical outcomes and may help clinical decision-making and enable better presurgical evaluation.

编号: YGQM-2022-1-2

引用格式: Zhang L, Zhou H, Zhang W, Ling X, Zeng C, Tang Y, Gan J, Tan Q, Hu X, Li H, Cheng B, Xu H, Guo Q. Electroclinical and Multimodality Neuroimaging Characteristics and Predictors of Post-Surgical Outcome in Focal Cortical Dysplasia Type IIIa. Front Bioeng Biotechnol. 2022;9:810897. DOI: 10.3389/fbioe.2021.810897.

通讯作者: 郭强

Abstract

Focal cortical dysplasia (FCD) type IIIa is an easily ignored cause of intractable temporal lobe epilepsy. This study aimed to analyze the clinical, electrophysiological, and imaging characteristics in FCD type IIIa and to search for predictors associated with postoperative outcome in order to identify potential candidates for epilepsy surgery. We performed a retrospective review including sixty-six patients with FCD type IIIa who underwent resection for drug-resistant epilepsy. We evaluated the clinical, electrophysiological, and neuroimaging features for potential association with seizure outcome. Univariate and multivariate analyses were conducted to explore their predictive role on the seizure outcome. We demonstrated that thirty-nine (59.1%) patients had seizure freedom outcomes (Engel class Ia) with a median postsurgical follow-up lasting 29.5 months. By univariate analysis, duration of epilepsy (less than 12 years) ($p = 0.044$), absence of contralateral insular lobe hypometabolism on PET/MRI (p Log-rank = 0.025), and complete resection of epileptogenic area (p Log-rank = 0.004) were associated with seizure outcome. The incomplete resection of the epileptogenic area (hazard ratio = 2.977, 95% CI 1.218-7.277, $p = 0.017$) was the only independent predictor for seizure recurrence after surgery by multivariate analysis. The

results of past history, semiology, electrophysiological, and MRI were not associated with seizure outcomes. Carefully included patients with FCD type IIIa through a comprehensive evaluation of their clinical, electrophysiological, and neuroimaging characteristics can be good candidates for resection. Several preoperative factors appear to be predictive of the postoperative outcome and may help in optimizing the selection of ideal candidates to benefit from epilepsy surgery.

编号: YGQM-2022-1-3

引用格式: Guo Y, Miao Q, Zhang Y, Wang C, Liang M, Li X, Qiu W, Shi G, Zhai Q, Chen Z. A novel missense creatine mutant of CaBP4, c.464G>A (p.G155D), associated with autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE), reduces the expression of CaBP4. *Transl Pediatr.* 2022;11(3):396-402. DOI: 10.21037/tp-22-54.

通讯作者: 陈志红

Abstract

Background: CaBP4 encodes Ca²⁺-binding protein 4, a neuronal Ca²⁺-binding protein that participates in many cellular processes by regulating the concentration of free Ca²⁺ ions. De novo CaBP4 variants have been identified as a cause of congenital stationary night blindness (CSNB). However, we recently reported a 4-generation pedigree with 11 individuals diagnosed with autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE) that were validated with only one novel missense mutation, c.464G>A (p.G155D), in CaBP4. De novo CaBP4 variants have never been reported to be related with ADNFLE. This study aimed to identify whether c.464G>A (p.G155D) in CaBP4 reduced the expression of CaBP4. **Methods:** In vitro experiments using recombinant protein expressed in human neuron cells were utilized in this study. Real-time polymerase chain reaction (RT-PCR) was performed to evaluate the effect of c.464G>A on CaBP4 mRNA expression. Western blot was performed to assess the effect of c.464G>A on CaBP4 protein expression. **Results:** According to the RT-PCR

and Western blot results, c.464G>A (p.G155D) was associated with an increased expression of CaBP4 mRNA and a reduced expression of CaBP4 protein. **Conclusions:** These results reveal that c.464G>A (p.G155D) in CaBP4 reduced the expression of CaBP4 by reducing the stability of the CaBP4 protein. Mutations in the CaBP4 gene may be associated with ADNFLE.

编号: YGQM-2022-1-4

引用格式: Ye Y, Hu Z, Mai J, Chen L, Cao D, Liao J, Duan J. A de novo PUM1 Variant in a Girl With a Dravet-Like Syndrome: Case Report and Literature Review. *Front Pediatr.* 2022;10:759889. DOI: 10.3389/fped.2022.759889.

第一作者: 叶园珍

Abstract

In the recent 3 years, subjects with Pumilio1-associated developmental disability, ataxia, and seizure syndrome have been identified as harboring Pumilio homolog 1 (PUM1) mutations. However, the characteristics of the seizure phenotype remain to be elucidated. We herein described a 3-year-old female proband who was diagnosed with developmental and epileptic encephalopathy presenting with some features suggestive of a Dravet-like syndrome. For genetic analyses, trio-based whole-exome sequencing and array comparative genomic hybridization were performed. Consequently, a de novo heterozygous missense variant was identified in exon 22 of the PUM1 gene: NM_001020658: c.3439C > T (p.Arg1147Trp). Upon thoroughly reviewing the existing literature, nine cases of PUM1 mutation-related epilepsy were identified, and their clinical features were summarized. A relationship between PUM1 mutation and clinical manifestations characteristic of a Dravet-like syndrome was proposed. To our knowledge, this is the first report of a patient with PUM1 mutation presenting with a Dravet-like syndrome.

编号: YGQM-2022-1-5

引用格式: Lin S, Liao J, Zhao X, Hu Y, Chen L, Chen Y, Liu G, Yao Y, Su Q, Scheffer IE, Wen F. Focal Epilepsy in Children With Tuberous Sclerosis Complex: Does Vigabatrin Control Focal Seizures? J Child Neurol. 2022 Mar 23;8830738211048326. DOI: 10.1177/08830738211048326.

第一作者: 林素芳

Abstract

We evaluated the efficacy and safety of vigabatrin in focal epilepsy associated with tuberous sclerosis complex by retrospectively reviewing patients with focal epilepsy and tuberous sclerosis complex treated with vigabatrin at a pediatric epilepsy center over an 8-year period. Of 85 patients, 20 (23.5%) were seizure-free for >12 months, 45 (52.9%) were responders ($\geq 50\%$ seizure reduction), and 20 (23.5%) were nonresponders. The median age (in months) at seizure onset in the seizure-free group (median, 15; interquartile range [IQR], 6-23.3) was higher than that of responders (median, 5; IQR, 3-14) and nonresponders (median, 6; IQR, 2-12). Fewer patients in the seizure-free group had calcification in their largest tubers, but the presence of tuber calcification did not differ among groups. Vigabatrin is more likely to result in seizure freedom in children with tuberous sclerosis complex who have later infantile onset of focal seizures and no calcification in their largest tuber.

编号: YGQM-2022-1-6

引用格式: Huang D, Wen X, Lu C, Zhang B, Fu Z, Huang Y, Niu K, Yang F. Investigating the molecular mechanism of Compound Danshen Dropping Pills for the treatment of epilepsy by utilizing network pharmacology and molecular docking technology. Ann Transl Med. 2022;10(4):216. DOI: 10.21037/atm-22-195.

第一作者: 黄丹

Abstract

Background: Compound Danshen Dropping Pills (CDDP) is widely used in clinical treatment of epilepsy. But the underlying active ingredients and molecular mechanisms are unclear. Our study aims to investigate the active components and functional mechanisms of CDDP in treating epilepsy using a network pharmacology approach. **Methods:** Candidate constituents and targets of CDDP were searched on the Traditional Chinese Medicine Systems Pharmacology database. NCBI and Genecards were used to establish a database of epilepsy targets. Next, used Cytoscape software, the interactive network diagram of "drug-active component-target" was drawn. Based on the STRING database we constructed protein-protein interaction network and analyzed protein-protein interaction relationships. Gene ontology analysis and Kyoto Encyclopedia of Genes and Genomes pathway enrichment analysis were performed for the common targets. Molecular docking provided an evaluation tool for verifying the combination of components and targets, which was performed using Auto-dock. **Results:** Sixty bioactive components, corresponding to 79 therapeutic targets for epilepsy, were successfully identified. Functional enrichment analysis showed that CDDP plays a pharmacological role in the treatment of epilepsy by regulating serotonergic synapses, morphine addiction, nicotine addiction and other pathways, as well as the NF- κ B signaling pathway. Molecular docking analysis showed that representative components may be closely bound to key targets. **Conclusions:** This network pharmacology study revealed the synergistic effects of multiple components, targets, and pathways of CDDP in the treatment of epilepsy, which will deepen our understanding of the underlying molecular mechanisms of CDDP in the treatment of epilepsy and lay the foundation for further experimental studies.

编号: YGQM-2022-1-7

引用格式: Hu J, Chen YH, Fang X, Zhou Y, Chen F. Clinical manifestations and prenatal diagnosis of Ullrich congenital muscular dystrophy: A case report. *World J Clin Cases*. 2022 Jan 7;10(1):338-344. DOI: 10.12998/wjcc.v10.i1.338.

第一作者：胡君

Abstract

Background: Ullrich congenital muscular dystrophy (UCMD) is one of the collagen-VI-related myopathies caused by mutations of COL6A1, COL6A2, and COL6A3 genes. Affected individuals are characterized by muscle weakness, proximal joint contracture, distal joint hyperlaxity, and progressive respiratory failure. There is currently no cure for UCMD. Here, we report the clinical manifestations and prenatal diagnosis of compound heterozygous mutations of the COL6A2 gene in a Chinese family with UCMD. **Case summary:** A 3-year-old boy, his 4-year-old brother, their parents, and a 20-wk-old fetus in the mother's womb were included in the study. The brothers had the typical manifestations of the early-severe subtype: A delayed motor milestone (never walking independently), torticollis, scoliosis, proximal joint contracture, distal joint hyperextension, right hip joint dislocation, and calcaneal protuberance. Both brothers were found by whole-exome sequencing and Sanger sequencing to carry two mutations of the COL6A2 gene (c.1353_c.1354insC, p.Arg453ProfsTer42/c.2105G>A, p.Trp702Ter). The absence of collagen VI staining in the younger brother's muscle was identified accurately. Genetic counseling and prenatal diagnosis were crucial for the family, as the autosomal recessive genetic disease affected a quarter of the patient's siblings. The fetus of the mother's third child underwent prenatal diagnosis and carried the same two mutations of COL6A2, confirmed in the amniotic fluid by multiplex ligation-dependent probe amplification and short tandem repeats. After a painful psychological struggle, the parents finally decided to terminate the pregnancy. **Conclusion:** We report a Chinese family suffering from UCMD. By clarifying the COL6A2 mutations in the probands, the parents had the opportunity to opt for voluntary interruption of the third UCMD pregnancy.

编号: ZWG-2022-1-1

引用格式: Xu ZY, Shen CH, Cai MT, Zhang GF, Ding MP, Guo Y. Managing depression and anxiety in patients with epilepsy in eastern China: A survey of epilepsy health professionals in Zhejiang Province. *Epilepsy Behav.* 2022 Feb;127:108516. doi: 10.1016/j.yebeh.2021.108516.

通讯作者: 郭谊

Abstract

Objective: We aimed to evaluate the assessment and management of epilepsy with anxiety and depression, and their clinical practice based on a survey. **Methods:** A cross-sectional survey of epilepsy health professionals was undertaken in Zhejiang Province using the modified International League Against Epilepsy (ILAE) Psychology Task Force questionnaire. We recorded the characteristics of participants and the practice of screening, referral, and treatment for depression and anxiety disorders. A total of 146 participants joined in the survey, of which 76.0% were neurologists, and 69 participants were the member of the Zhejiang Association Against Epilepsy (ZAAE). **Results:** This survey revealed that almost all participants (87.7%) agreed that screening for depression and anxiety in patients with epilepsy (PWEs) was very important; however, the frequency of screening was very low (41.1% of participants screened less than 10% of patients, and 34.2% participants screened between 10% and 30% of patients). A higher frequency of screening was reported in the member group and compared with that in the non-member group ($P = 0.025$). The main barrier to screening was the lack of time during clinic visits: 81.5% participants included screening questions as part of their clinical review. When anxiety/depression was diagnosed, the next step should be to refer patients to a psychiatrist (78.1%). No standardized procedures and lack of mental health specialists trained to assess and/or manage PWEs, were the main barriers to follow-up assessment and management. Lack of appropriately trained mental health specialists was also the main barrier to psychological treatment for depression and anxiety. **Conclusion:** This survey highlighted that epilepsy healthcare professionals in

Zhejiang province agreed on the importance of screening for psychiatric comorbidities in PWEs; however, the screening and management were actually insufficient. Certain barriers to screening, referral, and treatment were presented and improvements were recommended.

编号: ZWG-2022-1-2

引用格式: Guo Y, Xu ZY, Cai MT, Gong WX, Shen CH. **Epilepsy With Suicide: A Bibliometrics Study and Visualization Analysis via CiteSpace. Front Neurol 2021;12:823474. DOI: 10.3389/fneur.2021.823474**

第一作者: 郭谊

Abstract

Objective: The purpose of this study was to analyze the research status of epilepsy with suicide and to determine the hotspots and frontiers via CiteSpace. **Method:** We searched the Web of Science Core Collection (WoSCC) for studies related to epilepsy and suicide from inception to September 30, 2021. We used CiteSpace to generate online maps of collaboration between countries, institutions, and authors, and revealed hot spots and frontiers in epilepsy with suicide. **Results:** A total of 631 publications related to epilepsy with suicide were retrieved from theWoSCC. AndresM. Kanner was the most published author (25 papers). The USA and Columbia University were the leading country and institution in this field, with 275 and 25 papers, respectively. There were active cooperation between institutions, countries, and authors. Hot topics focused on depression, antiseizure medications, pediatric epilepsy, and risk factors of suicide in patients with epilepsy (PWEs). **Conclusions:** Based on the CiteSpace findings, this study detected active collaboration among countries, institutions and authors. The main current research trends include suicide caused by depression, suicide caused by the use of antiseizure medications, suicide in children with epilepsy, and risk factors for suicide in PWEs. Thus, more attention should be paid to the psychiatric comorbidity of PWEs

(especially pediatric epilepsy), the suicidal tendency of PWEs, and the rational use of antiseizure medications in the future.

编号: ZWG-2022-1-3

引用格式: Zheng Y, Ming WJ, Zheng Z, Jiang HJ, Chen C, Wu H, Wang ZJ, Xu SS, Zhu JM, Ding MP, Wang S, Ding Y. Pearls & Oysters: Parietal Lobe Epilepsy in Disguise: Motor Attacks Induced by Proprioceptive Triggers. Neurology 2022;98:509-513. DOI: 10.1212/WNL.0000000000200050

通讯作者 丁瑶

Abstract

Pearls: Parietal lobe epilepsy (PLE) is characterized by frequent auras and diverse semiologies, largely due to the elaborate connections of the parietal lobe to other regions. Proprioceptive-induced seizures are a rare but unique expression of PLE. They are characterized by motor seizure attacks precipitated by proprioceptive stimuli of the extremities. Focal cortical dysplasia (FCD) is a common etiology of drug-refractory PLE. Bottom-of-sulcus dysplasia (BOSD) represents a distinctive subtype notable for its highly localized and subtle lesions, as well as an excellent surgical outcome. **Oysters:** PLE should be part of the differential in patients with paroxysmal movement disorders. Thorough history taking, especially of the triggers and motor attacks, is essential for a correct diagnosis. The presence of localized rhythmic epileptic discharges (REDs) on electroencephalography serves as an important indicator of the easily misdiagnosed FCD. A thorough history taking, careful review of MRI, and application of multimodal imaging postprocessing further facilitate subtle lesion detection. A transient response to antiseizure medications is common in FCD-associated epilepsy. However, early referral to surgery evaluation should be considered because drug refractoriness is likely to develop with time.

编号: ZWG-2022-1-4

引用格式: 郭崇伦, 慕洁 审. 肌阵挛性失张力癫痫的表型与遗传谱. 癫痫杂志, 2022, 8(1): 74-81. doi: 10.7507/2096-0247.20220018

译者: 郭崇伦

Abstract

文章旨在描述一个大样本量的癫痫伴肌阵挛性失张力发作 (MAE) 患者神经发育损害程度, 并确定其遗传学病因。采用标准化的神经心理学仪器对 MAE 患者的癫痫特征、智力残疾、自闭症谱系障碍和注意缺陷/多动障碍进行深入的表型分析。我们对癫痫和神经精神疾病的基因集进行外显子分析(全外显子测序), 以确定遗传学病因。研究共分析了 101 例 MAE 患者 (70% 为男性)。发作年龄中位数为 34 月龄 (范围 6~72 月龄)。主要发作类型为肌阵挛性失张力发作或失张力发作 100%、全身强直阵挛性发作 72%、肌阵挛性发作 69%、失神性发作 60%、强直性发作 19%。研究观察到 62% 的患者有智力障碍, 69% 的患者适应行为评分极低。此外, 24% 表现出自闭症症状, 37% 表现出注意力缺陷/多动症状。85 例患者中的 12 例 (14%) 发现了致病性变异, 包括 5 例先前发表的患者。这些基因是 SYNGAP1 (n=3)、KIAA2022 (n=2)、SLC6A1 (n=2) 以及 KCNNA2、SCN2A、STX1B、KCNB1 和 MECP2 (各 n=1)。此外, 研究还分别在 3 例患者中分别鉴定了 1 个新的候选基因 —ASH1L、CHD4 和 SMARCA2。研究发现, MAE 与明显的神经发育障碍有关。MAE 具有遗传异质性, 通过外显子分析在 14% 的队列中确定了致病的遗传病因。这些发现表明 MAE 是几种病因的表现, 而不是一个离散的综合征实体。

欢迎大家引用上述论文

编号: CYDQZ-2022-1-1

引用格式: Li W , Lai W , Peng A , Chen L. Two cases of anesthetics-induced epileptic seizures: a case report and literature review. Acta Epileptologica, 2022, 4(1).

通讯作者: 陈蕾

Abstract

Background: Anesthetics like propofol have been reported to be capable of controlling status epilepticus. However, we have observed during daily clinical work that some anesthetics can induce epileptic seizures. Therefore, this study aims to explore the relationships between anesthetics and epilepsy. **Case Presentation:** We collect and report two cases of anesthetics-induced epileptic seizures, in order to arouse attention towards this critical phenomenon. We also summarize the current research progress on this topic, analyze associations between anesthetics and epilepsy, and discuss the mechanisms underlying the associations. Two females, seizure-free for more than 3years, presented with anesthetics-induced epileptic seizures and controlled by administration of Midazolam and Diazepam. By literature review, we included six studies which found that the occurrence of epileptic seizures has no relationship with the age/sex of patients, the surgery procedure, nor the type of anesthesia. It is closely related to the decreased blood concentration of AEDs and the inadequate preoperative preparation. **Conclusions:** Unnecessary surgery should be avoided for patients with epilepsy, and anesthetists should make careful selections on anesthetics and prepare adequately for surgery.

编号: CYDQZ-2022-1-2

引用格式: Peng A, Qiu X, Ji S, Hu D, Dong B, Song T, Huang C, Chen L. The impact of childhood parental loss on risk for depression and anxiety in adulthood:

A community-based study in Southwest China. J Affect Disord. 2022;298(Pt A):104-109. doi: 10.1016/j.jad.2021.10.093.

通讯作者: 陈蕾

Abstract

Purpose: Lack of parental raising is an important reason for parental loss in China due to urbanization. We aimed to explore the association between parental loss (including parental death, divorce, alcoholism, crime, drug addiction, bedridden, and lack of parental raising) before 17 years old and mental health disorders in adulthood in Southwest China. **Methods:** Childhood parental loss, age, gender, socioeconomic status, smoking, drinking, and medical history were self-reported. Patient Health Questionnaire 9 (PHQ-9) and Generalized Anxiety Disorder 7 (GAD-7) were used for identifying depression and anxiety, respectively. We use logistic regression with inverse probability weighting according to the propensity score to assess the risk of childhood parental loss on mental health disorders in adulthood. **Results:** A total of 8014 adults were enrolled in this study. Childhood parental loss increased the risk of adulthood depression (OR, 1.60, 95%CI, 1.30–1.98) but not anxiety ($p=0.07$) after adjustment of all covariates. Sensitivity analyses including logistic regression with original data, nearest neighbor matching, full matching, and propensity score as the only covariate all yielded similar results. **Limitations:** Recall bias could not be fully eliminated due to the retrospective nature of study design. Our study had been conducted in a less-prosperous area in Southwest China and the results may not be representative at nation level. **Conclusions:** Childhood parental loss was associated with depression in adulthood in Southwest China, highlighting the need for early intervention in children with parental loss, to prevent long-term negative effects on their mental health.

编号: CYDQZ-2022-1-3

引用格式: Tang Y, Peng A, Peng B, He S, Zhao X, Zhu Y, Lai W, Song T, Chen L. Association between patent foramen ovale and migraine without aura: a

community-based cross-sectional study in China. *BMJ Open*. 2022 Mar 31;12(3):e056937. doi: 10.1136/bmjopen-2021-056937.

通讯作者: 陈蕾

Abstract

Objectives To assess the influence of patent foramen ovale (PFO) on the prevalence of migraine without aura based on propensity score-matched samples in Southwest China. Design Propensity-matched cross-sectional study. Participants Residents over 20 years of age were recruited from 15 communities of Western China from July 2020 to October 2020. A total of 3741 residents having accepted to undergo contrast-transthoracic echocardiography and a standard structured questionnaire was assessed for the relationship between PFO and migraine without aura. Primary and secondary outcome measures The primary outcome measures were the prevalence of migraine without aura across different degrees of right–left shunts. Results A total of 3741 participants were included. Among them, 881 participants were diagnosed with PFO. The prevalence of migraine without aura in the PFO group was 12.83%, significantly higher than the other group (7.83%, $p < 0.001$; OR=1.71, 95% CI 1.19 to 2.47). Conclusion This community-based cross-sectional study pointed to a strong association between PFO and migraine without aura, especially when the shunt is large

编号: CYDQZ-2022-1-4

引用格式: Peng A, Lai W, He S, Li W, Song T, Ji S, Zhao X and Chen L. Association Between Early Parental Death and Loneliness in Adulthood: A Community-Based Study in Southwest China. *Front. Psychiatry*. 2022, 13:784000. doi: 10.3389/fpsy.2022.784000

通讯作者: 陈蕾

Abstract

Loneliness is a growing public health problem that threatens physical and mental health

to a large extent. Compelling evidence has shown that premature parental death is strongly associated with many mental health disorders in adulthood, but whether it increases the risk of loneliness remains unclear. In this large communitybased study, we included 32,682 adult participants (20–93 years old) from Southwest China and used the three-item short version of University of California, Los Angeles, Loneliness Scale to identify participants with loneliness. A total of 1,975 participants reported loneliness, which resulted in a loneliness prevalence of 6.0% in Southwest China. Logistic regression was used to evaluate the association between early parental death and loneliness after adjusting for age, gender, education level, marital status, smoking and drinking status, living status, and chronic diseases. We found that early parental death was significantly associated with loneliness [odds ratio (OR) = 1.21, 95% confidence interval (CI), 1.03–1.42]. A Sensitivity analysis excluding those with mental health disorders (796 participants) yielded similar results (OR = 1.26, 95% CI = 1.06–1.49). We also found that being younger, single, divorced, or widowed, and more educated; living alone; and having chronic disorders were associated with loneliness. We conclude that childhood parental death is associated with loneliness in adulthood, suggesting the need for early intervention in affected children to prevent long-term adverse neuropsychiatric effects.

编号: CYDQZ-2022-1-5

引用格式: Kang L, Duan Y, Chen C, Li S, Li M, Chen L and Wen Z. Structure Activity Relationship (SAR) Model for Predicting Teratogenic Risk of Antiseizure Medications in Pregnancy by Using Support Vector Machine. *Front. Pharmacol.* 2022, 13:747935. doi: 10.3389/fphar.2022.747935

通讯作者: 陈蕾

Abstract

Teratogenicity is one of the main concerns in clinical medications of pregnant women. Prescription of antiseizure medications (ASMs) in women with epilepsy during

pregnancy may cause teratogenic effects on the fetus. Although large scale epilepsy pregnancy registries played an important role in evaluating the teratogenic risk of ASMs, for most ASMs, especially the newly approved ones, the potential teratogenic risk cannot be effectively assessed due to the lack of evidence. In this study, the analyses are performed on any medication, with a focus on ASMs. We curated a list containing the drugs with potential teratogenicity based on the US Food and Drug Administration (FDA)-approved drug labeling, and established a support vector machine (SVM) model for detecting drugs with high teratogenic risk. The model was validated by using the post-marketing surveillance data from US FDA Spontaneous Adverse Events Reporting System (FAERS) and applied to the prediction of potential teratogenic risk of ASMs. Our results showed that our proposed model outperformed the state-of-art approaches, including logistic regression (LR), random forest (RF) and extreme gradient boosting (XGBoost), when detecting the high teratogenic risk of drugs (MCC and recall rate were 0.312 and 0.851, respectively). Among 196 drugs with teratogenic potential reported by FAERS, 136 (69.4%) drugs were correctly predicted. For the eight commonly used ASMs, 4 of them were predicted as high teratogenic risk drugs, including topiramate, phenobarbital, valproate and phenytoin (predicted probabilities of teratogenic risk were 0.69, 0.60 0.59, and 0.56, respectively), which were consistent with the statement in FDA-approved drug labeling and the high reported prevalence of teratogenicity in epilepsy pregnancy registries. In addition, the structural alerts in ASMs that related to the genotoxic carcinogenicity and mutagenicity, idiosyncratic adverse reaction, potential electrophilic agents and endocrine disruption were identified and discussed. Our findings can be a good complementary for the teratogenic risk assessment in drug development and facilitate the determination of pharmacological therapies during pregnancy.

编号: CYDQZ-2022-1-6

引用格式: Shen KF, Yue J, Wu ZF, Wu KF, Zhu G, Yang XL, Wang ZK, Wang J,

Liu SY, Yang H, Zhang CQ. Fibroblast growth factor 13 is involved in the pathogenesis of temporal lobe epilepsy. *Cereb Cortex*. 2022 Feb 23:bhac012. doi: 10.1093/cercor/bhac012.

通讯作者：张春青

Abstract

Background: Temporal lobe epilepsy (TLE) is the most common drug-resistant epilepsy in adults, with pathological mechanisms remaining to be fully elucidated. Fibroblast Growth Factor 13 (FGF13) encodes an intracellular protein involved in microtubule stabilization and regulation of voltage-gated sodium channels (VGSCs) function. FGF13 mutation has been identified in patients with inherent seizure, suggesting a potential association between FGF13 and the etiology of TLE. Here, we set to explore the pathological role of FGF13 in the etiology of TLE. **Results:** We found that the expression of FGF13 was increased in the cortical lesions and CA1 region of sclerotic hippocampus and correlated with the seizure frequency in TLE patients. Also, Fgf13 expression was increased in the hippocampus of chronic TLE mice generated by kainic acid (KA) injection. Furthermore, Fgf13 knockdown or overexpression was respectively found to attenuate or potentiate the effects of KA on axonal length, somatic area and the VGSCs-mediated current in the hippocampal neurons. **Conclusions:** Taken together, these findings suggest that FGF13 is involved in the pathogenesis of TLE by modulating microtubule activity and neuronal excitability.

编号：CYDQZ-2022-1-7

引用格式： Shen KF, Duan QT, Duan W, Xu SL, An N, Ke YY, Wang LT, Liu SY, Yang H, Zhang CQ. Vascular endothelial growth factor-C modulates cortical NMDA receptor activity in cortical lesions of young patients and rat model with focal cortical dysplasia. *Brain Pathol*. 2022:e13065. doi: 10.1111/bpa.13065.

通讯作者：张春青

Abstract

Emergence of dysmorphic neurons is the primary pathology in focal cortical dysplasia (FCD) associated pediatric intractable epilepsy; however, the etiologies related to the development and function of dysmorphic neurons are not fully understood. Our previous studies revealed that the expression of vascular endothelial growth factor-C (VEGF-C) and corresponding receptors VEGFR-2, VEGFR-3 was increased in the epileptic lesions of patients with tuberous sclerosis complex or mesial temporal lobe epilepsy. Here, we showed that the expression of VEGF-C, VEGFR-2, and VEGFR-3 was increased at both mRNA and protein levels in patients with cortical lesions of type I, IIa, and IIb FCD. The immunoreactivity of VEGF-C, VEGFR-2 and VEGFR-3 was located in the micro-columnar neurons in FCD type I lesions, dysplastic neurons (DNs) in FCD type IIa lesions, balloon cells (BCs) and astrocytes in FCD type IIb lesions. Additionally, the amplitude of evoked-EPSCs (eEPSC) mediated by NMDA receptor, the ratio of NMDA receptor- and AMPA receptor-mediated eEPSC were increased in the dysmorphic neurons of FCD rats established by prenatal X-ray radiation. Furthermore, NMDA receptor mediated current in dysmorphic neurons was further potentiated by exogenous administration of VEGF-C, however, could be antagonized by ki8751, the blocker of VEGFR-2. These results suggest that VEGF-C system participate in the pathogenesis of cortical lesions in patients with FCD in association with modulating NMDA receptor-mediated currents.

编号: CYDQZ-2022-1-8

引用格式: Dong L, Zheng Q, Cheng Y, Zhou M, Wang M, Xu J, Xu Z, Wu G, Yu Y, Ye L, Feng Z. Gut Microbial Characteristics of Adult Patients With Epilepsy. *Front Neurosci.* 2022 Feb 16;16:803538. doi: 10.3389/fnins.2022.803538. eCollection 2022.

通讯作者: 冯占辉

Abstract

Objective: To characterize the intestinal flora of patients with epilepsy and its correlation with epilepsy. **Methods:** Patients with ages > 18 years were consecutively enrolled from the outpatient department, Affiliated Hospital of Guizhou Medical University from January 2018 to December 2019. A total of 71 subjects were recruited, including epilepsy patients (n = 41) as an observation group and patient family members (n = 30) as a control group. Fresh stool specimens of all the subjects were collected. The 16S ribosomal RNA sequencing was analyzed to determine changes in intestinal flora composition and its correlation with epilepsy. Subgroup analysis was then conducted. All patients with epilepsy were divided into an urban group (n = 21) and a rural group (n = 20) according to the region, and bioinformatics analyses were repeated between subgroups. **Results:** LEfSe analysis showed that *Fusobacterium*, *Megasphaera*, *Alloprevotella*, and *Sutterella* had relatively increased abundance in the epilepsy group at the genus level. Correlation analysis suggested that *Fusobacterium* sp. ($r = 0.584, P < 0.01$), *Fusobacterium mortiferum* ($r = 0.560, P < 0.01$), *Ruminococcus gnavus* ($r = 0.541, P < 0.01$), and *Bacteroides fragilis* ($r = 0.506, P < 0.01$) were significantly positively correlated with the occurrence of epilepsy ($r \geq 0.5, P < 0.05$). PICRUST function prediction analysis showed that there were significant differences in 16 pathways between the groups at level 3. Comparing the rural group with the urban group, *Proteobacteria* increased at the phylum level and *Escherichia coli*, *Fusobacterium varium*, *Prevotella stercorea*, and *Prevotellaceae* bacterium DJF VR15 increased at the species level in the rural group. **Conclusion:** There were significant differences in the composition and functional pathways of gut flora between epilepsy patients and patient family members. The *Fusobacterium* may become a potential biomarker for the diagnosis of epilepsy.

编号: CYDQZ-2022-1-9

引用格式: Mao Q, Wang C, Wen W, Zhou M, Tang J, Chen C, Cheng Y, Wu Q, Zhang X, Feng Z, Wang M. A meta-analysis of the association between calprotectin

and the severity of COVID-19. *J Infect.* 2022;84(3):e31-e33. doi: 10.1016/j.jinf.2022.01.022.

通讯作者: 冯占辉

No abstract available

编号: CYDQZ-2022-1-10

引用格式: Wang M, Wen W, Zhou M, Wang C, Feng ZH. Meta-Analysis of Risk of Myocarditis After Messenger RNA COVID-19 Vaccine. *Am J Cardiol.* 2022;167:155-157. doi: 10.1016/j.amjcard.2021.12.007.

通讯作者: 冯占辉

No abstract available

编号: CYDQZ-2022-1-11

引用格式: Chen D, Peng X, Zhan Y, Wu P, Jiang L, Hu Y. Efficacy and safety of intravenous high-dose immunoglobulin in treatment of the severe form of Japanese encephalitis. *Neurol Sci.* 2022. doi: 10.1007/s10072-022-05891-6.

通讯作者: 胡越

Abstract

Purpose: To explore the efficacy and safety of intravenous immunoglobulin (IVIG) in the treatment of severe Japanese encephalitis (JE). **Methods:** A retrospective study of 124 children diagnosed with the severe or very severe form of JE was undertaken. There were 62 cases in the IVIG group and control group. The efficacy, safety, and tolerability of IVIG were evaluated 3 days, as well as 1, 2, and 3 weeks after IVIG, respectively, and the prognosis was assessed at 6 months. **Results:** Cox regression survival analysis suggested that the IVIG group reached the criteria for efficacious treatment faster than

that in the control group. The duration of unconsciousness and the number of days of seizures, a dull response to light, the pyramidal sign, and meningeal-irritation sign in the IVIG group were shorter than those in the control group ($p < 0.05$). The number of complications occurring in the IVIG group (including gastrointestinal bleeding and pneumonia) was less than that in the control group ($p < 0.05$). Cox regression survival analysis suggested that age ($p = 0.003$) and imaging abnormalities ($p = 0.042$) had an effect on the efficacy of IVIG treatment. The Liverpool Outcome Score at 6 months showed that the prognosis of the IVIG group was better than that of the control group ($p < 0.05$). IVIG treatment was safe and tolerable. **Conclusion:** IVIG showed good efficacy, safety, and tolerance for treatment of the severe form of JE. The age and imaging abnormalities of patients affect the efficacy of IVIG treatment.

编号: CYDQZ-2022-1-12

引用格式: Ding X, Zhou J, Zhao L, Chen M, Wang S, Zhang M, Zhang X, Jiang G. Intestinal Flora Composition Determines Microglia Activation and Improves Epileptic Episode Progress. *Front Cell Infect Microbiol.* 2022;12:835217. doi: 10.3389/fcimb.2022.835217.

通讯作者: 蒋国会

Abstract

In response to environmental stimuli, immune memory mediates the plasticity of myeloid cells. Immune training and immune tolerance are two aspects of plasticity. Microglia that are immunologically trained or immunologically tolerant are endowed with a tendency to differentiate into alternative dominant phenotypes (M1/M2). Male C57BL/6 mice (immune training group, immune-tolerant group, and control group) were used to establish the kainic acid epilepsy model. The seizure grade, duration, latency, hippocampal potential, and energy density were used to evaluate seizures, and the changes in the polarization of microglia were detected by western blot. 16S rDNA sequencing showed that the abundance of Ruminococcus in the immune-tolerant group

was the dominant flora. Our research connections Intestinal microorganisms, brain immune status, and epilepsy behavior together. Pro-inflammatory M1 phenotype and anti-inflammatory M2 phenotype mediate and enhance and suppress subsequent inflammation, respectively. We conclude that intestinal microorganisms influence the occurrence and development of epilepsy by regulating the polarization of microglia.

中国科学院预警期刊参考目录 (2022 年 1 月更新, 医学期刊)

序号	杂志名称	ISSN 号	预警等级
1	Aging-US	1945-4589	中
2	American Journal of Cancer Research	2156-6976	中
3	American Journal of Translational Research	1943-8141	中
4	BIOMEDICINE & PHARMACOTHERAPY	0753-3322	中
5	Brazilian Journal of Medical and Biological Research	0100-879X	中
6	Cancer Biomarkers	1574-0153	中
7	EXperimental and Molecular Pathology	0014-4800	中
8	Experimental and Therapeutic Medicine	1792-0981	中
9	International Journal of Immunopathology and Pharmacology	2058-7384	中
10	International Journal of Molecular Medicine	1107-3765	中
11	Journal of Biomaterials and Tissue Engineering	2157-9083	中
12	Journal of International Medical Research	0300-0605	中
13	Medical Science Monitor	1643-3750	中
14	Molecular Medicine Reports	1791-2997	中
15	Oncology Letters	1792-1074	中
16	Oncology Research	0965-0407	中
17	Oncotargets and Therapy	1178-6930	中
18	Cancer Cell International	1475-2867	中
19	Cancer Management and Research	1179-1322	中
20	Journal of Cancer	1837-9664	中
21	Journal of Cellular and Molecular Medicine	1582-1838	中
22	World Journal of Clinical Cases	2307-8960	中

编号: JJJJM-2022-1-1

引用格式: Wang S, Pan J, Zhao M, Wang X, Zhang C, Li T, Wang M, Wang J, Zhou J, Liu C, Sun Y, Zhu M, Qi X, Luan G, Guan Y. Characteristics, surgical outcomes, and influential factors of epilepsy in Sturge-Weber syndrome. *Brain*. 2022:awab470. doi: 10.1093/brain/awab470.

通讯作者: 关宇光

Abstract

Few studies have reported the clinical presentation, surgical treatment, outcomes, and influential factors for patients with epilepsy and Sturge-Weber syndrome. This large-scale retrospective study continuously enrolled 132 patients with Sturge-Weber syndrome and epilepsy from January 2008 to December 2018 at our hospital to analyze their characteristics. Among these patients, 90 underwent epilepsy surgery, and their postoperative 2-year follow-up seizure, cognitive, and motor functional outcomes were assessed and analyzed. Univariable and multivariable logistic analyses were conducted to explore the influential factors. Among the Sturge-Weber syndrome patients for whom characteristics were analyzed ($n = 132$), 76.52% of patients had their first epileptic seizures within their first year of life. The risk factors for cognitive decline were seizure history ≥ 2 years (adjusted odds ratio [aOR] = 3.829, 95% confidence interval [CI]: 1.810-9.021, $p = 0.008$), bilateral leptomeningeal angiomas (aOR = 3.173, 95% CI: 1.970-48.194, $p = 0.013$), age at onset < 1 year (aOR = 2.903, 95% CI: 1.230-6.514, $p = 0.013$), brain calcification (aOR = 2.375, 95% CI: 1.396-5.201, $p = 0.021$) and left leptomeningeal angiomas (aOR = 2.228, 95% CI: 1.351-32.571, $p = 0.030$). Of the patients who underwent epilepsy surgery ($n = 90$), 44 were subject to focal resection, and 46 underwent hemisphere surgery (19 anatomical hemispherectomies and 27 modified hemispherotomies). A postoperative seizure-free status, favorable cognitive outcomes, and favorable motor outcomes were achieved in 83.33%, 44.44%, and 43.33% of surgical patients, respectively. The modified hemispherotomy group had similar surgical outcomes, less intraoperative blood loss and shorter postoperative hospital

stays than the anatomical hemispherectomy group. Regarding seizure outcomes, full resection (aOR = 11.115, 95% CI: 1.260-98.067, $p = 0.020$) and age at surgery < 2 years (aOR = 6.040, 95% CI: 1.444-73.367, $p = 0.031$) were positive influential factors for focal resection. Age at surgery < 2 years (aOR = 15.053, 95% CI: 1.050-215.899, $p = 0.036$) and infrequent seizures (aOR = 8.426, 95% CI: 1.086-87.442, $p = 0.042$; monthly vs. weekly) were positive influential factors for hemisphere surgery. In conclusion, epilepsy surgery resulted in a good postoperative seizure-free rate and favorable cognitive and motor functional outcomes and showed acceptable safety for patients with epilepsy and Sturge-Weber syndrome. Modified hemispherotomy is a less invasive and safer type of hemisphere surgery than traditional anatomic hemispherectomy with similar surgical outcomes. Early surgery may be helpful to achieve better seizure outcomes and cognitive protection, while the risk of surgery for young children should also be considered.

编号: JJJJM-2022-1-2

引用格式: Yang B, Mo J, Zhang C, Wang X, Sang L, Zheng Z, Gao D, Zhao X, Wang Y, Liu C, Zhao B, Guo Z, Shao X, Zhang J, Zhang K, Hu W. Clinical features of automatisms and correlation with the seizure onset zones: A cluster analysis of 74 surgically-treated cases. *Seizure.* 2022;94:82-89. doi: 10.1016/j.seizure.2021.11.015

通讯作者: 张凯

Abstract

OBJECTIVES: To identify semiologic features of automatisms correlating to different seizure onset zones (SOZ). **METHODS:** In total, 204 seizures from 74 patients with either oral or manual automatisms were assessed. Patients were divided into four groups depending on the SOZ into frontal, posterior, neocortical temporal, and mesial temporal cortex groups. A k-means analysis was applied on 11 semiologic features on a multi-criteria scale. Then, the resulting clinical patterns were correlated with the SOZs

determined by presurgical anatomy-electroclinical data (25 cases with stereo-EEG).

RESULTS: Four clinical patterns of automatisms with different accompanying symptoms were identified. The clinical features of clusters 1 and 4 were mostly found in temporal epilepsy whereas clusters 2 and 3 were more frequent in extratemporal epilepsy. Cluster 1 was significantly correlated with mesial temporal lobe epilepsy ($p = .017$) and was characterized by aura, postictal confusion, short automatisms delay. Cluster 3 included 1/3 patients with frontal lobe epilepsy and was characterised by emotionality. Cluster 4 was related to neocortical temporal lobe epilepsy and characterised by dystonia and short automatism delay ($p = .011$). **CONCLUSION:** The distinct semiologic patterns of automatisms may provide information which may allow clinicians to define the SOZs. These findings could improve diagnostic accuracy and surgical outcome.

编号: JJJJM-2022-1-3

引用格式: Liu T, Ding J, Zhang S, Wang Y, Xu J, Yuan L, et al. Independent temporal lobe epilepsy in patients with tuberous sclerosis complex. *Pediatr Investig.* 2022;6:23–28. DOI :10.1002/ped4.1231

第一作者: 刘婷红 丁晶 **通讯作者:** 梁树立

Abstract

Tuberous sclerosis complex (TSC) is a rare disease that involves multiple organs, including the brain; approximately 80%–90% of TSC patients exhibit TSC-associated epilepsy. Independent temporal lobe epilepsy (TLE), TSC-unrelated epilepsy, is particularly rare in patients with TSC. Here, we describe three patients with TSC with independent TLEs that were confirmed by stereo-electroencephalography (EEG), postoperative pathological findings, and seizure outcome at follow-up. The patients were retrospectively enrolled at two centers; their ictal epileptiform discharge onsets were determined using electrode contacts in the hippocampus during stereo-EEG. The three patients underwent anterior temporal lobectomies and remained seizure-free at 1–

5 years after surgery. Postoperative pathological examinations confirmed hippocampal sclerosis in all three patients. Furthermore, postoperative intelligence quotient improvement was evident in one patient, while the quality of life was improved in two patients at 12 months after surgery.

编号: JJJJM-2022-1-4

引用格式: 于国静, 季涛云, 车圆圆, 王文, 刘通, 王宏杰, 王若凡, 王爽, 刘庆祝, 蔡立新. 儿童 Sturge-Weber 综合征继发癫痫的临床脑电图特征及手术治疗.

癫痫杂志. 2022, 8(1): 41-47. doi: 10.7507/2096-0247.20220011

通讯作者: 季涛云

Abstract

目的 总结经手术治疗儿童 Sturge-Weber 综合征 (Sturge-Weber syndrome, SWS) 继发癫痫的癫痫发作特点、脑电图特征、手术方式及术后疗效, 提高对此类疾病的认识。**方法** 回顾性分析我院儿童癫痫中心自 2015 年 5 月—2020 年 5 月经手术治疗 7 例 SWS 的患儿资料。收集 7 例患儿人口学特征、癫痫发作形式、脑电图、头颅影像学、手术方式及预后等。**结果** 共收集患儿 7 例, 其中男 1 例、女 6 例。癫痫起病年龄 2 月龄~1 岁 6 月龄, 平均 (6.43±5.26) 月龄。7 例患儿均以局灶性发作起病, 2 例病程中出现癫痫持续状态 (1 例为持续性部分性癫痫), 1 例有痉挛发作, 1 例表现为丛集性发作的特点。发作间期脑电图特点: ① 随着病程延长, 脑电图逐渐加重, 表现为患侧慢波逐渐增多 (7/7), 波幅逐渐变低 (7/7), 生理波消失 (4/7); ② 发作间期脑电图常表现为无癫痫样放电/偶见或少量癫痫样放电。4 例行半球离术, 1 例颞顶枕离断术, 2 例行病灶切除术。术后随访时间 6 个月~5 年, 平均随访时间 (79.29±24.50) 个月。6 例术后定期随访均 Engel IIa, 1 例术后 3 个月 Engel II 级, 术后 6 个月及 1 年 Engel III 级。**结论** SWS 继发癫痫发作形式多样, 脑电图具有特征性改变。若为药物难治性癫痫, 通过详尽的术前评估, 采取合理手术方式, 可取的较好的治疗效果。

编号: SGQNX-2022-1-1

引用格式: Wang X, Mu P, Zhang W and Liu Y. Case Report: Not All Neurological Symptoms Respond Well to Penicillin in Patients With Neurosyphilis. *Front. Neurol.* 2022, 12:813829. doi: 10.3389/fneur.2021.813829

通讯作者: 刘永红

Abstract

Patients with neurosyphilis present with a wide range of neurological symptoms, the response of which to penicillin is not well known. In this paper, we analyzed the clinical video-electroencephalogram (EEG) features of neurosyphilis in a 54-year-old man who exhibited with rhythmic orofacial involuntary movements, Argyll Robertson pupil, frequent paroxysmal oral-automatism seizures, periodic lateralized discharges (PLEDs) with triphasic waves, behavioral changes, and memory decline. After treatment with penicillin, PLED and seizures disappeared and behavioral changes and memory decline were significantly improved, but rhythmic orofacial involuntary movements and Argyll Robertson pupil persisted, which indicates an irreversible characteristic of late stage neurosyphilis syndromes.

编号: SGQNX-2022-1-2

引用格式: Dou X, Li D, Wu F, Wang Z, Niu M, Wu Y, Deng T, Wang D, Li X. The clinical features, treatment and outcomes of 33 children from Northwestern China with Anti-N-methyl-D-aspartate receptor encephalitis. *Neurol Res.* 2022 May;44(5):429-438. doi: 10.1080/01616412.2021.2000824.

通讯作者: 李霞

Abstract

Objectives: We analyzed the clinical features and outcomes of children with anti-N-methyl-D-aspartate receptor (anti-NMDAR) encephalitis in Northwestern China.

Methods: We retrospectively recruited 33 pediatric patients with anti-NMDAR encephalitis in Northwestern China from December 2013 to April 2020. The demographics, clinical features, treatments, and outcomes were reviewed. **Results:** 33 patients with anti-NMDAR encephalitis were enrolled in this study (a median age of 6.8 years, 20 females and 13 males). The initial symptoms included seizures (42.4%), psychiatric symptoms (39.4%), speech dysfunction (12.1%), and paralysis (6.1%). During the course of the disease, 31 patients (93.9%) presented with psychiatric symptoms, 29 patients (87.9%) presented with speech dysfunction, 25 patients (75.8%) presented with movement disorders and 24 patients (72.7%) presented with sleep disorders followed by seizures, consciousness disturbance, autonomic nervous dysfunction, paralysis, and hypoventilation. 12 patients (36.4%) had abnormal cerebrospinal fluid (CSF) findings, 10 patients (30.3%) exhibited abnormal brain magnetic resonance imaging (MRI) results, and 29 patients (87.9%) showed abnormal Electroencephalography (EEG) findings. None of the patients had tumors. All patients received first-line immunotherapy and 8 patients both received first and second-line immunotherapy. 30 of the 33 patients achieved good outcomes (score on the modified Rankin Scale [mRS] of 0–2), whilst the other 3 patients had poor outcomes (mRS score of 3–6). **Conclusions:** Patients with higher CSF anti-NMDAR body titer were more likely to develop sleep disorders, consciousness disturbances and more severe disease states.

欢迎大家引用上述论文

中国抗癫痫协会青年委员会 2021 年度学术报告及交流大会

中国抗癫痫协会青年委员会 2021 年度学术报告及交流大会于 2022 年 3 月 12-13 日在海南省海口市隆重召开。今年的主题是“新时代创新争先 新征程踏浪筑梦”。由于新冠疫情的原因，这是第二年度青年委员会的学术年会由次年的 1 月



调整了 3 月。本次会议之前也是各地出现了疫情抬头的趋势，在洪震会长、张慧秘书长、段力副秘书长的精心筹划和海南医学院附属医院、海南省抗癫痫协会、李其富青委团队的严密组织下，青年委员会年会得到平安、顺利召开。除了青年委员会外，CAAE 洪震会长、周东、王学峰、周列民副会长、秘书处张慧秘书长、段力副秘长和杨天明、李晓裔、王淑荣、李文玲、李岩等省市抗癫痫协会会长现场参会，李世绰创会会长、廖卫平、孙伟、陈述花教授等常青委和第一届青委们线上参加了会议并致辞或学术讲座。

本次会议继承了既往学术年会的活动，在开幕式中洪震会长和李世绰创会会长分别致辞，洪会长肯定了新一届青委会 2021 年的工作，并对 2022 年的工作提出了新的要求和期望。李会长重点以谈心的形式和青委们进行了交流。海南省卫生健康委周长强书记兼主任就海南卫生健康事业和产业的发展政策与规划做了详细的介绍，邀请青年学者到海南健康岛发展创来。海南医学院第一附属医院顾硕书记代表东道主做欢迎致辞，他表示全力支持癫痫专业的发展，同时以自己到海南的发展经历诚挚邀请青年医学专家到海南工作。最后，梁树立主委汇报青委会 2021 年工作和 2022 年工作计划。开幕式由李其富教授主持。癫痫热点争鸣、年度最佳研究评比依然精彩纷呈。八大区总结评比是在原有 NEW 项目八大区评比基础上首次进行八大区工作的全面总结，负责人全面协调，各个青委参与其中，虽然过程有些紧张和辛苦，但回顾工作，更多

的是感动和动，同时通过对比，取长补短，大家都摩拳擦掌，努力做好 2022 年工作。人文环节今天分为上下半场，上半场由海南省卫生健康委医政医管局高新谱副局长作了“开放、创新-海南医疗区”专题讲座，就海南大健康领域的规划和进展，特别是乐城的定位和前景等进行了全面的讲解，并与顾硕书记及青委们进行了讨论，下半场是以“种下希望、耕耘梦想”为主题的团建活动，当日正值第 44 个全民义务植树节，全体参会人员赶赴海口市五源河国家湿地公园，分中抗秘书处、海南抗癫痫协会、青委八大区等十组，共同种下了 20 棵英雄树—木棉树和四季各异的黄花风铃木树，目前这些树长势良好。青年联谊会简单而热烈，同时充分体现了海南的特色，既有海南医学院学生们组织的海南民歌《久久不见久久见》、黎族特色竹竿舞，也有曾经响彻湖北武汉国际会展中心方舱医院的儋州调声《嘱姑九点半》。另外，海南特色植物的有奖竞猜（奖品是洪会长等人主译的专业书籍）也让大家收获人文知识和专业知识。

另外这次会议上特别增加了多中心研究展示与招募环节，增加这部分内容主要是为了推动以共同兴趣为导向的多中心研究的开展。共有 11 个多中心主研究项目公开招募，其中大会报告 5 项，海报展示 6 项，会后这些多中心研究项目都有青委单位报名参加。





2021年度八大区工作评比

卓越奖

川渝黔滇藏

杰出奖

京津冀晋蒙

优秀奖

粤桂琼闽

优胜奖

豫鄂湘、申苏鲁、陕甘宁青新

浙皖赣、黑吉辽



最佳研究获奖名单

年度最佳研究论文一等奖

张春青 陆军军医大学新桥医院

年度最佳研究论文二等奖

李其富 海南医学院第一附属医院

张捷 北京大学第一医院

年度最佳研究论文三等奖

王秀 首都医科大学附属北京天坛医院

关宇光 首都医科大学三博脑科医院

郝勇 上海交通大学医学院附属仁济医院

年度最佳研究论文优秀奖

彭炳蔚 广州市妇女儿童医疗中心

叶园珍 深圳市儿童医院

林婉挥 福建医科大学附属协和医院

最佳人气奖

李其富 海南医学院第一附属医院



青年委员会多中心项目展示（大会汇报项目）

单位	地区	科室	联系人	项目名称	合作单位标准	招募患者要求
首都医科大学附属北京儿童医院等	北京	功能神经外科	刘婷红	中国癫痫数据银行项目	1. 癫痫专科门诊； 2. 省市级以上医院重点从事癫痫工作人员 3. 各级癫痫中心单位	否
四川大学华西医院	四川省	神经内科	陈蕾	女性癫痫队列研究项目参研中心招募	1、二级甲等及以上医院；2、神经内科、神经外科或独立癫痫中心；3、具有专业的癫痫医师，具备开展癫痫专科研究的条件。	18-45岁的育龄期女性癫痫患者。
北京大学第一医院	北京	儿科	季涛云	儿童癫痫术后抗癫痫发作药物减/停药时间点与癫痫发作预后的关系	中国抗癫痫协会批准的三级癫痫中心	纳入标准；排除标准（见详情）
首都医科大学三博脑医院	北京	功能神经外科	关宇光	中国脑面血管瘤病多中心协作研究计划书	1、参加医院应是三级甲等医院或三级专科医院及中国抗癫痫协会二级以上癫痫中心； 2、参加人员应具备副主任医师以上职称； 3、完成 SWS 切除性手术二例及以上并完成随访。	1、符合脑面血管瘤病临床诊断，即影像学证实软脑膜血管瘤，常伴脑萎缩、钙化灶等典型影像学表现，伴或不伴面部血管痣、青光眼、认知发育异常、运动功能障碍等典型临床表现，完成切除/离断手术并术后病理学辅助进一步确诊； 2、就诊及手术完成时间需在 2000 年 1 月至 2020 年 12 月之间。
空军军医大学西京医院	陕西省	神经内科	刘永红	《脑电监测期间的 Near-SUDEP》多中心临床研究申报	脑电监测中心	脑电监测期间发生 Near-SUDEP 的癫痫患者

欢迎各位青委、同行积极参加多中心研究项目

青年委员会多中心项目展示（海报展示项目）

单位	地区	科室	联系人	项目名称	合作单位标准	招募患者要求
深圳市儿童医院	广东省	癫痫中心	操德智	特殊益生菌 BF839 添加治疗儿童难治性癫痫多中心合作项目	二级及以上癫痫中心，月平均诊治难治性癫痫儿童 20 例以上。	1、诊断为药物难治性癫痫的患者；2、每月至少有一次癫痫发作；3、现阶段服药规律，两种及以上药物联合治疗；4、年龄<18岁
吉林大学第一医院	吉林省	儿科	姜慧轶	伴临床下样放电的初诊抽动障碍患儿首选药物治疗后执行功能分析	三级甲等医院，有小儿神经科或发育行为科。	选择 2022 年 1 月 ~ 2022 年 12 月 吉大一院二部儿科伴临床下样放电的初诊抽动障碍患儿。
空军军医大学西京医院	陕西省	神经内科	张歆博	“自身免疫性脑炎急性症状性病性发作”多中心临床研究申报	脑电监测中心	符合 AE 诊断标准；AE 发病期间（后）出现癫痫发作。
上海交通大学医学院附属瑞金医院	上海	神经外科	刘强强	双侧丘脑中央中核电刺激治疗药物难治性癫痫的多中心、单盲、随机对照临床研究	1、具有 DBS 手术条件的外科中心，预计入组数量超过 5 例，可以成为多中心（预计多中心数量为 4-6 个）。 2、暂不具备 DBS 手术条件的单位，可以推荐患者至多中心手术，入组病例记为推荐单位。	1、年龄 10-60 岁。 2、符合药物难治性癫痫的诊断标准； 3、发作类型：全面性发作为主； 4、同意参加本研究，签署知情同意书； 5、愿遵守相关试验方案条例，按规定期限前来复查并行相关检查。
首都医科大学附属北京儿童医院	北京	功能神经外科	袁柳	TSC 相关癫痫 VNS 治疗的多中心研究	中国抗癫痫协会批准的三级、二级癫痫中心	纳入标准；排除标准（见详情）
首都医科大学附属北京儿童医院	北京	神经科	邓劼	生酮饮食治疗单基因变异所致发育性癫痫性脑病的临床研究	1、具有儿童癫痫的诊断和治疗能力，开展门诊及病房诊疗，拥有脑电图仪等检查设备； 2、拥有经验丰富的小儿神经专业医师和生酮营养师，具备遗传学检测判读能力，对本研究的患儿进行专人管理及随访； 3、至少曾成功开展 1 例儿童癫痫的 KD 治疗。	纳入标准；排除标准（见详情）

欢迎各位青委、同行积极参加多中心研究项目

领读学术

领读学术是一个传统的项目，2022 年我们做了一些改变，全年将开展 10 场，首场和年终场为全国性会议，中间 8 场由各大区分别组织主办。另外最主要的是在既往文献解读+研究和论文发表经验分享为主的基础上，融合入研究全过程的指导和讨论，包括如何发现问题、凝练问题、开展研究、统计分析、论文撰写和投稿发表等。

领读学术
— 开启您学术新篇章 —

直播时间：2022年1月22日 09:00-11:30

会议日程

时间	内容	讲者
大会时间：2022年1月22日（星期六）09:00-11:30 大会主席：姜玉武 梁树立		
09:00-09:10	开场视频	
09:10-09:15	开场寄语	姜玉武 梁树立
09:15-09:45	"Publish or perish" - How to publish	Selim Ramin Benbadis
09:45-10:15	互动讨论	吴海欣 陈蕾 陈子怡 周健 彭镜 刘永红 梁建民 王爽
10:15-10:35	GSK卫星会： 2021NICE指南解读-局灶性癫痫	陈子怡
10:35-11:05	Response to antiseizure medications in epileptic patients with malformation of cortical development	王爽
11:05-11:25	互动讨论	吴海欣 陈蕾 陈子怡 周健 梁建民 王爽
11:25-11:30	会议总结	姜玉武 梁树立

会议将全程在线直播
并开通在线讨论
敬请扫描二维码观看精彩内容

会议截图显示多位专家在线参与讨论。

2022 年的首场领读学术于 1 月 22 日线上召开。由青委会第一届主任委员姜玉武教授和第二届主任委员梁树立教授共同主持。Selim Ramin 教授连线介绍了文章发表体会；王爽教授介绍了自己文章发表经验；陈子怡教授进行了指南解读。

超 700 人次加入直播间，10 家医院线上集体参会，人员覆盖北京、浙江、陕西、广东、上海、重庆、辽宁、江苏、吉林等全国 32 个省市自治区。

领读学术
— 开启智慧学术新篇章 —

直播时间: 2022年3月20日 09:30-12:00

• 会议日程 •

时间	内容	讲者
🕒 大会时间: 2022年3月20日(星期日) 09:30-12:00		
👤 大会主席: 陈子怡		
09:30-09:40	GSK开场视频	
09:40-09:45	开场寄语	陈子怡
09:45-10:15	Recessive PKD1 mutations are associated with FS/EFS+ and the genotype-phenotype correlation	何娜
10:15-10:35	互动讨论	余璐 李其富 林燕舞
10:35-10:55	GSK卫星会-2021NICE指南解读-局灶性癫痫篇	郭燕舞
10:55-11:35	能力提升-控制混杂因素的统计方法	张璐
11:35-11:55	互动讨论	余璐 李其富 林燕舞
11:55-12:00	会议总结	陈子怡

会议将全程在线直播
并开通在线讨论
敬请扫描二维码观看精彩内容

2022年的首场大区承领读学术活动, 由粤桂琼闽大区于3月20日线上召开。

会议由陈子怡副主委主持, 何娜教授连线介绍了自己文章发表心路, 梅斯医学科学部的张璐老师讲解了统计中如何控制混杂因素, 郭燕舞教授进行了指南解读。

超400人加入直播间, 6家医院线上集体参会, 人员覆盖广东、北京、四川、上海、重庆、广西、浙江、海南、黑龙江、陕西等29个省市自治区。

领读学术
— 开启智慧学术新篇章 —

临床科研培训系列课程

为了提升癫痫和电生理相关临床研究水平,《癫痫与神经电生理学杂志》编辑部和中国抗癫痫协会青年委员会针对各位杂志编委的研究生和 CAAE 青年委员等年轻科技工作者开展相关科研培训工作。既往的科研培训多倾向于理论学习,但是学生往往听不进去,真正面对数据时无从下手,对自己数据的整体分析没有信心,不知道自己做出来的结果有什么优势与不足。本次临床科研培训从实战出发,帮助学员尽快掌握科研设计、数据收集、数据清理、数据分析、书写文章的能力。采用软件实操培训的模式,把理论融入实操中,从而学会临床科研设计、数据收集模式、数据清理、数据呈现模式、掌握数据的缺陷与不足。同时,课程中也会解决各位编委在投稿中遇到的实际问题。

课程讲座每周1次,时间为周三上午11:00-12:00(腾讯会议)每节课45分钟。所有课程均由青年委员、杂志常务主编冯占辉博士授课。

主要内容包括:

- [1] Epidata 应用:解决问基于单中心和多中心数据的收集、数据清理、数据输出。
- [2] SPSS 应用:解决如何统计数据,做出初步分析,把握统计基本方法。
- [3] Prism 应用:解决如何做出完美的数据分析图,提供参考数据和全程实操,在实际操作中,学会做出有美感的图
- [4] PASS 应用:解决如何估算样本量、把握度。
- [5] Review manager 应用:解决同类型文章数据整合问题(meta分析)。
- [6] TCGA 数据库应用:解决如何下载数据和利用数据库二次分析问题。

癫痫与神经电生理学
杂志编辑部
中国抗癫痫协会
青年委员会

数据文件质控(软件实操)

CHK文件建立和数据间的比对

冯占辉

癫痫与神经电生理学杂志常务主编



讲师介绍
博士,博士生导师,贵州省抗癫痫协会常务理事,长期从事癫痫临床诊治工作,近5年发表SCI文章48篇,中文文章68篇。

直播时间
2022年4月27日(周三)11:00

腾讯会议号:473 297 780



扫码看直播

统筹策划

张 慧 段立嵘 梁树立

刊头题字

李世焯

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THINKINGS AND ACTIONS

