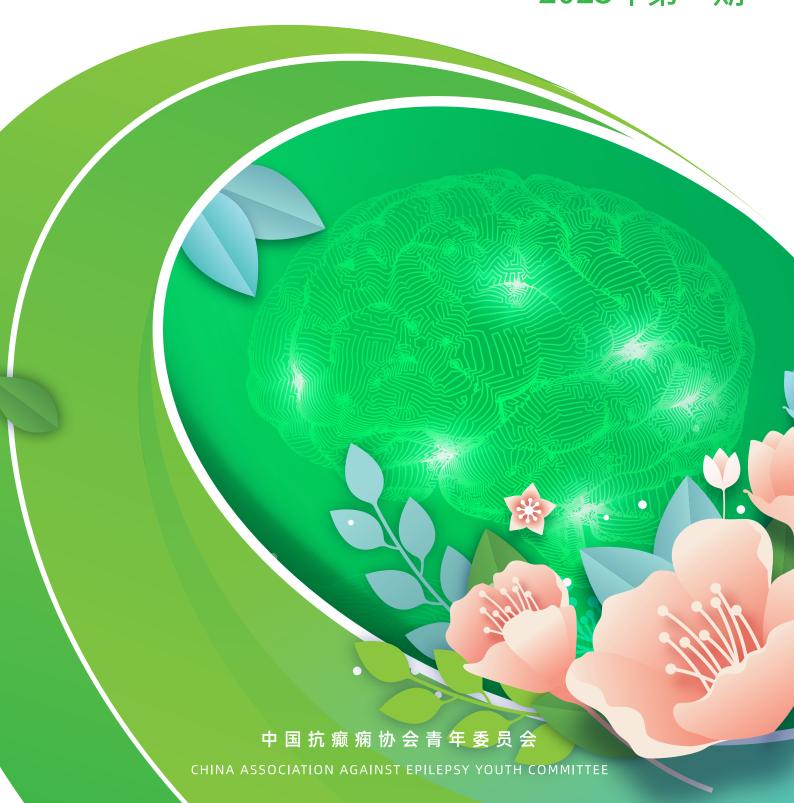


# CAAE·青年思行

CAAE Youth Thinkings and Actions 2023年第一期



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寄语专家简介:洪震,教授、博士生导师,现任复旦大学附属华山医院神经病学研究所所长、复旦大学癫痫诊治中心主任、中国抗癫痫协会会长。从事神经病学的临床工作已有四十余年,在神经内科临床,尤其是癫痫的诊治和临床药理工作中积累了一些经验。先后承担了国家"九五、十五、十一五、十二五"攻关课题、国家自然科学基金、上海市科委重大课题,并担任多个新药Ⅲ期临床



试验牵头单位负责人,多个国际多中心临床试验中心负责人。获卫生部科技进步奖一项、教育部科学技术进步奖、中华医学科技奖三项、上海市科技进步奖两项、上海市医学科技奖两项。先后发表论文 250 多篇,其中 SCI 论文近百篇。主编及参与编写《神经病学》、《现代癫痫学》、《现代神经流行病学》、《神经系统疾病基础与临床》、《精神病学》、《实用内科学》、《实用神经病学》等多部专著与教材。曾任《中华神经科杂志》副主编、《癫痫杂志》、《中国临床神经科学杂志》主编和多本专业杂志编委。已培养博士生 31 名,硕士生 37 名。

# CAAE 会长寄语青年委员

# 洪震教授在 2022 年青委年度学术报告与交流大会上的讲话

非常高兴参加 "CAAE 青年委员会年度学术报告及交流大会",本次会议是疫情全面放开之后协会召开的第一次成规模的线下会议,拉开了协会全年学术会议交流的序幕,本次参会人数突破了 300 人,这也预示着癫痫领域在后疫情时代蓬勃发展的强劲势头。

3月份的全国"两会"是全面贯彻落实党的二十大会议精神,踏上实现第二个百年奋斗目标新征程的重要会议,绘就的宏伟蓝图鼓舞人心,吹响的时代号角催人奋进。青年人是这个时代的晴雨表,时代的责任赋予青年人,时代的荣誉属于青年人。习近平总书记在中国共产党第二十次全国代表大会上强调:"青年强,则国家强。当代中国青年生逢其时,施展才干的舞台无比广阔,实现梦想的前景无比光明。"我们广大青年医生也要做到不忘初心,立志做有理想、敢担当、能吃苦、肯奋斗的新时代好青年。

青年委员会作为协会的一个综合性分支机构,开展了一系列卓有成效的工作,"西部行"、"领读学术"、"菁 YOUNG 计划"等项目不仅促进了癫痫学术交流,也培养和锻炼了一批青年骨干,为我国卫生健康事业持续、高质量发展锻造了坚强后备力量。希望大家在南昌这座英雄的城市,通过丰富的学术交流提升攻克医学难关的勇气,在意义深远的人文活动中汲取健康强国的不竭动力。

(以上内容根据现场录音整理, 并经过洪震会长删减、审核)

#### 读后感:

洪老师是我的研究生导师,他也是中国抗癫痫协会的会长。洪震教授平时对学生甚少直接批评、但他在骨子里对学生要求很高,行事果断、具有很强的原则性,他的为人和他在癫痫领域的造诣,一直是我们学习的榜样!我和诸多师弟师妹们也一直在努力追随恩师的脚步。

洪老师用习近平总书记在中国共产党第二十次全国代表大会上的讲话激励 我们新一届青委,凡事要做到不忘初心,立志成为有理想、敢担当、能吃苦、肯 奋斗的新时代好青年。他还希望我们能学习英雄、汲取英雄的力量,助力健康强 国。我相信我们广大青委必将谨遵前辈的教诲,团结奋进、继续做好各方面工作, 不负众望、争取在未来获得更多的创新发展!

(申苏鲁大区负责人:吴洵昳)

# 《CAAE 青年思行》论文摘要收录说明

- 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 2023 年 1-3 月发表的论文
- 收录格式为引用格式+摘要。

寄语专家简介:李世绰,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

# 李世绰教授在 2022 年青委年度学术报告与交流大会上的讲话

最近由于身体情况,暂停了一段时间工作。但是青委会是中国抗癫痫协会的未来和明天,是非常重要的一个委员会,中国抗癫痫协会未来的发展就是要寄希望于你们身上。所以,青委会的会我一定要参加并讲话。洪震会长、傅超书记、张明会长和树立主委的讲话都很全面。傅超书记充满革命的激情和诗意的讲话,令人印象深刻,相信是我们江西省抗癫痫事业有力支持者之一。

我今天的讲话,没有打稿子,按照心中所想到的,说几点意见:

首先,我们真是应该珍惜我们的青春,我今天看到一些同事,几年不见,已 头顶一顶花白头发,眼看着进入壮年的阶段了,盛年不重来,一日难再晨,岁月 不饶人啊。所以我觉得我们青年委员们,一定要珍惜自己的青春!

其次,当前网络上简直是什么千奇百怪的论调都有,有正面的,也有好多是以 正面的面孔出来的谬论,各种自媒体上的,五花八门。在复杂的国内外的大环境 中,希望年轻人提高政治的敏感和嗅觉,要站在大局的和历史的角度去看待问题,跟着党中央的步伐,不受各种错误思想的干扰,要认真领会、贯彻党的二十大和两会精神。

第三,将中国抗癫痫协会进一步融入国际学术界是刻不容缓的任务。最近,WHO 领导层发生了一些不合常规的变动,我国选派的助理总干事解除合同回国了,他原来对咱们抗癫痫事业做过很大支持。另外,丁玎教授目前是 IBE 的副主席,是两大癫痫国际组织当中的唯一的中国代表。ILAE 执委会,上届我国有两个成员,现在一个都没有,作为会员人数最多的 ILAE 成员国,这是不应当的。如何改变这一局面,争取在国际组织中占据应有的位置,获得更多的话语权,是我们要认真解决的问题。我希望包括丁玎教授在内的各位专家群策群力,为我们协会进一步融入国际组织,争取更多中国人的话语权做出成绩来。。

我们必须拿出真正的实力,拿出创新的成果,才能使中国抗癫痫协会在国际癫痫界的地位更加巩固和提高。一方面,加强对新技术的推广,虽然存在部分国外输入的新技术,但是中国病例多,可用数量和质量来获得成就;另一方面,中国有200多个癫痫中心,怎样能够发挥癫痫中心联合体的作用?青委会做过一些多中心研究的尝试,我觉得你们应当再好好设计一下,根据国际癫痫学界的进展情况,找出哪一方面我们有优势的领域,充分发挥自身优势,组织多中心研究,产出高质量科研论文,取得创新性成果。总之,创新的思路对于争取更高的国际学术地位非常重要。

第四,关于英文。虽然现在各国有个学中文的热潮,但是在可预期的未来,没有英文在世界上是走不通的。我知道好多青委的英文水平很高,比我们这一代人英文要棒得多。我希望所有青委们的英语一定要过关,不管是参加国际论文讨论、演讲还是辩论,都要有相当高的英语水平。在英文学习上绝对不能够放松,一定要把英语学好!英语学好了,你才能在国际上畅通无阻!

最后,希望中国抗癫痫协会继续保持建立十几年以来的良好风气,在今年协会换届工作中,不出现影响团结的事情和乱象,充分展现团结协作以及不计个人名利的优秀传统。有人担心如果个别资深专家,在选举当中没能获得理想的结果,会不会就把精力转到别的组织去?我认为即使你把精力转到别的组织去,只要你在认真发展抗癫痫事业,在为癫痫患者解除痛苦,那也没什么不好。咱们提倡的

就是,从事与癫痫防控相关的各个组织,我中有你,你中有我,大家一起推进抗癫痫事业。当然,我也不相信我们的老专家会因为个人得失,就把协会的事情放下不管了。同时,协会不可避免地要更新换代,年龄一代换一代。我已经八十多了,当然已经退休好久了,下一届我们洪震会长,期满以后也要退下来。下一个会长总要接上来,再下一个也要准备着,这一代一代的更新是个自然规律,在这更新换代的过程当中,我希望我们青委们,以积极的正当的姿态,参与到更新换代当中,我们会逐渐的建立越来越年轻的一代代领导集体,这样才能使我们协会永远保持活力!

以上五点意见,仅供大家参考,如果有错误不当的地方,请批评指正。

(以上内容根据现场录音整理, 并经过李世绰创会会长删减、审核)

## 读后感:

李世绰创会会长是中国抗癫痫协会青年委员会的共同倡议和创建者,对青委会的建设和每个青年委员的成长都特别关心。因为知道李会长处于身体康复之初,当我请示李会长是否可以参与青委年会致辞时还是有些犹豫和顾虑,虽然非常希望能听到李会长的教导和方向引领,但也怕影响了李会长的康复,也怕耽误更多更重要的协会工作。李会长收到邀请后,当即表示一定会参会,还对不能现场见到年轻的青委会表达了遗憾。

李会长在自己从事研究、管理、国际组织任职和协会建立发展的经验与思考基础上,结合国内外局势与学术领域现状,从珍惜青春、坚定政治定力与明辨是非、增强实力与融入国际行业发展、学好英文与加强国际交流、团结协作与传承发展等五个方面,用肺腑之言对我国青年癫痫工作者提出了明确要求、表达了殷切希望、指明了发展方向。会后我与许多青委交流,大家都非常感动,而感动的不仅仅是李会长康复之初就参会致辞,更重要的是感受到李会长对协会青委们的关爱与希望,对癫痫事业发展的高屋建瓴和深思熟虑。大家都倍感责任与动力,青委会一致表示,一定以时不我待的激情、走偏千山万水提高癫痫诊疗水平、想尽千方百计进入国际学术组织,说尽千言万语争取癫痫医患权益,吃尽千辛万苦开展创新研究,助力中国抗癫痫事业的高质量发展。

(青委会负责人: 梁树立)

# 从"山海情"到"京宁情" CAAE 青年委员会京津冀晋蒙大区 北京大学第一医院 谢涵 季涛云

宁夏西海固地区,地处偏远,干旱贫瘠。1996年国家确立了"福建对口帮扶宁夏"的政策,经过20余年的努力,曾经的"干沙滩"变成了如今的"金沙滩",也谱写出了2021年热播剧《山海情》中的一个个鲜活动人的扶贫故事。

通过经济帮扶,宁夏人民富起来了。然而,宁夏地区的医疗条件依然薄弱。 2018年响应国家的号召,北京大学第一医院(北大医院)儿科与宁夏妇幼保健院/宁夏儿童医院签署了紧密型合作协议,每年派遣北大医院儿科专家团队来宁夏,对宁夏儿童医院儿科各项业务进行精准帮扶。2022年8月随着国家区域医疗中心的建立,医院正式更名为北京大学第一医院宁夏妇女儿童医院,合作进入了新的篇章。一个个新的"京宁情"的故事正在发生。

宁夏儿童医学发展落后于国内发达地区。自 2021 年 1 月开,北大医院儿科先后派出多名儿童神经专业的医生支援其建设。派出儿童神经专业常杏芝主任医师和季涛云主任医师担任北大医院宁夏儿童医院副院长,派出儿童神经专业谢涵主治医师(中国抗癫痫协会青年委员)负责儿童神经病房的工作。调研发现宁夏地区癫痫儿童并不少见,但接受规范化抗癫痫治疗的比例比较低。造成这一现象的原因包括宁夏地区多数医疗机构未开设儿童神经专科,儿童神经专科医师和脑电图专业技师匮乏等。在这诸多原因之中,癫痫儿童不能获得高质量的脑电图监测,是阻碍宁夏地区癫痫儿童获得规范化诊治的突出问题之一。

在北大医院儿科团队的帮扶下,北大医院宁夏儿童医院脑电图室自 2021 年 2 月正式开展工作,2022 年 8 月开展了长程视频脑电图监测。2021 年 12 月在宁夏儿童医院成功建立了与北大医院儿科脑电图室的网络远程会诊平台 (图 1),实现了脑电图的远程实时会诊,极大提升了北大医院宁夏儿童医院的脑电图诊断水平。

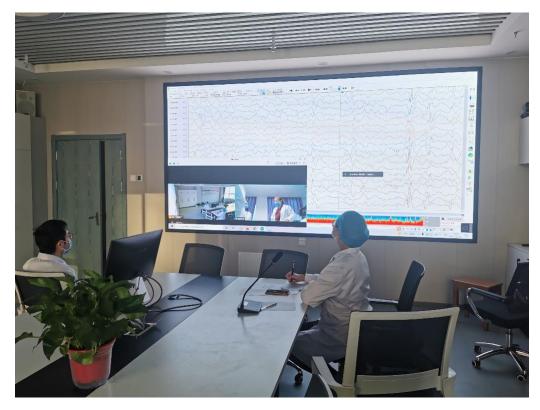


图 1: 宁夏儿童医院脑电图远程会诊平台(正在与北大医院儿科脑电图室网络会诊中)

除了深化宁夏儿童医院的脑电图室工作,三位医生每周出儿童神经专业门诊,服务于宁夏及周边地区的患儿,并一对一带教培养宁夏儿童医院的小儿神经专业医师,积极推动宁夏地区基层医疗机构开展脑电图监测等,积极构建以北大医院宁夏妇儿医院为中心,向上联系北大医院儿科,向下辐射宁夏区域内各县市级医疗机构的三级儿童癫痫规范化诊治体系。宁夏经济帮扶的"山海情",已是20余载,硕果累累;宁夏医疗帮扶的"京宁情",已5个年头,方兴未艾。目前,在宁夏地区儿童癫痫的规范化诊治的道路上仍充满着挑战,如宁夏地区大多数医疗机构的儿童癫痫诊治能力还比较薄弱,癫痫儿童及其家属对疾病的认识还比较匮乏等。北大医院儿科神经专业的同事们将不忘初心,继续砥砺前行。同时呼吁中国抗癫痫协会的各位专家同仁们能积极加入我们,能多来宁夏指导,延续"京宁情",让宁夏这片医疗的"干沙滩"早日变成"金沙滩"。

# 心怀仁爱之心,绽放儿科人文之花 CAAE 青年委员会鄂豫湘大区 中南大学湘雅医院儿童医学中心小儿神经专科 周霞

医学,是关爱生命的事业;人文,是医学的本质属性。古今往来,医学与人文相伴而生,医学最能够体现以人为本。关怀是链接医护和患者的桥梁,医疗工作因融入了人文关怀才丰富、深刻而有温度。病房每天上演着不同的故事,而有些暖心的温情时常发生,总有一些是我们不能忘怀的。其实,生活中打动人心的并不是并不一定是你为他做了多少轰轰烈烈的大事儿,而是与患者相处的许多细小的瞬间,也就是一句亲切的问候,一个温暖的眼神,一双无助时握住的手,甚至不经意间自然流露出如亲似友般的关怀,都会成为维护医患关系的纽带。

## 因为懂得, 所以慈悲

6岁的轩轩是个因癫痫发作经治疗控制不住的难治性癫痫的男孩,癫痫这种疾病发生在儿童身上,对家庭来说无疑是毁灭性的打击。为了治愈孩子的病,父母带着轩轩四处求医,曾吃过中、西药物,做过康复治疗,但抽搐仍然控制不住。其父母为了治好孩子的病,让这个本就不富裕的家庭更加窘迫。通过病友群的推荐,父母决定带轩轩到湘雅医院找彭镜教授看看,但因为轩轩已经生病了两年,家中积蓄不多,不得已,轩轩父母让爷爷奶奶带着孩子从云南出发,自己选择了继续工作为轩轩赚取医疗费。均已近70岁的轩轩爷爷奶奶带着轩轩千里迢迢、展转来到湘雅医院,因为挂不上彭教授号,让两老满脸愁容,看到孩子受尽病痛折磨,爷爷奶奶百般无奈之下冲到彭教授诊室说明了情况,彭教师立即为轩轩加了号,做病情评估,建议轩轩住院完善检查和治疗,并帮助其联系了小儿神经专科43病室的床位,将轩轩快速安置在病房,对此,爷爷奶奶非常感恩。

轩轩爷爷奶奶一直生活在农村,不会讲普通话,因为语言文化差异,爷爷奶奶的方言医护人员听不懂,很多疾病信息无法采集清楚,于是医生通过电话和父母沟通病情,医护人员在工作电脑上寻找,发现另外还有一个籍贯为云南的家属,似乎像看到救命稻草,请来了家属帮忙衔接沟通,看到有热心家属的加入使沟通顺畅,医生们在热心家属的同意下,将俩个人安置在同一病房,解决了沟通障碍

的问题。看到轩轩小儿神经专科的医护人员心如刀绞,他脸上、手臂、大腿多处地方被烫得发黑,部分地方已经化脓,原来是轩轩父母通过各种渠道听到治疗癫痫的偏方"火灸",因为病急乱投医导致了轩轩被伤害。处理好轩轩伤口后,医护人员语重心长的对轩轩的爷爷奶奶做好治疗癫痫的疾病科学宣教,希望以后不要再做这类"火灸"治疗,护士仍然不放心,通过电话联系到轩轩父母强调了"火灸"对于轩轩的弊端,确保轩轩全家人都知道此偏方的不可取。护士长周霞老师知道病房里来了"特殊"的轩轩后,在晨会上号召大家积极帮助轩轩爷爷奶奶解决生活和医疗上的困难,交代外出检查的陪检人员一定要将轩轩家人送至检查室,做完检查安全送回病房;同时和送餐人员沟通,请送餐员一定要按时将饭菜送至轩轩爷爷奶奶的床旁,确保他们不会饿肚子。



轩轩的癫痫入院时发作,一天发作三到四次,具体表现为突然惊叫后意识丧失,两眼向右上方斜视,嘴角向右侧抽动,肢体强直,喉间痰鸣,面唇青紫,小便失禁,每次发作时间约 2-3 分钟,发作过后意识逐渐恢复。这让本应阳光开朗的轩轩因癫痫发作让他脑部神经受到严重损伤,轩轩也变得沉默寡言失去了往日的活泼开朗。也让担心轩轩的奶奶夜不能寐,只能暗暗留下无奈和辛酸的眼泪,夜班护士发现年迈的奶奶伤心难过,把她带到护士站,听完奶奶的哭诉,安抚着奶奶,在我们病房有非常多类型的孩子,他们的治疗效果有的很好,控制住发作

后还可以继续上学,希望奶奶先好好休息保重自己身体,保存体力才能更好的照顾轩轩。第二天请了医院社工部的社工给轩轩一家做陪伴,并带着轩轩在我们的"向阳花"爱心小屋医学护理人文关怀基地专门为患儿提供常规性的医学人文关怀,让他结识了许多活泼的同龄小病友,鼓励小病友们多多参与社工部的活动、阅读我们爱心阅览车上的书本,邀请轩轩和爷爷奶奶到我们科室"畅谈室",进行沟通畅谈,给予心理支持,包括但不限于倾听、共情、安慰与开导、解释、建议和指导等方法,帮助缓解焦虑紧张情绪,促进病情恢复。主治医生张慈柳对患儿进行详细的体查、病史收集、既往治疗和检查以后,综合分析,给患者更换了治疗药物,通过几天观察,轩轩的抽搐情况明显好转,这让长吁短叹的爷爷奶奶充满的生机活力。

"因为懂得,所以慈悲"这是著名作家张爱玲的一句话。因为懂得疾病本身的复杂多变、崎岖坎坷以及无可奈何,所以懂得患者的艰难困苦,当医护人员懂得病人的时候,慈悲油然而生,便有了和谐。医疗是最富有人性色彩的服务。病人来医院就医,正处于生命中脆弱的时刻,这时病人最渴求的就是人性的温暖,而最能赢得病人心的就是人文关怀。在这时一句简单的问候都可能发挥疗愈人心的作用,用最简单的话语、最柔和的语气,给患者最大的勇气,支撑他们对抗疾病的勇气和信心,握握患者的手、拍拍他们的肩膀、拥抱他们。

### 爱让夏天的风变得柔和

7月的长沙骄阳似火,人潮涌动。家长来来往往的身影,医护人员匆匆的步伐,还有小孩子阵发性的啼哭,都让病房的温度显得格外高。而在中南大学湘雅 医院儿童医学中心小儿神经专科 43 病室脑电图检查室里,这里的患儿更是哭声 不断。

在临床诊疗中,我们常常会接诊癫痫的孩子,其实大多数就是民间俗称"羊癫疯"的患儿,为了观察病情的进展,我们常常需要做脑电图检查,有些患儿更是需要反复多次来进行脑电图检查,来回往返医院的这条路就成了家长们熟知的复查之路,这条路虽然漫长,但家长们都不辞辛苦,因为这是他们心中一条带着希望的路。

"医生!护士!辛苦你们帮帮忙吧!我大老远从外省赶过来的,现在没有床

做检查,这可怎么办呀?""阿姨您先别急,我们现在马上调整一下,给您孩子 腾一个床位出来!"

这个场景发生在脑电图检查室里,当时一位焦急万分的家长抱着孩子在检查室门口来回踱步。当时这位家长背着一个大包,双手抱着意识模糊的孩子,坐了几个小时的大巴车赶到这里,就是希望医生能看看孩子的情况。她不停在病房来回踱步,眼神中满是担忧。如果按照之前的检查流程,本来这位家长给孩子约上的检查已经排到了一个月之后,但考虑到疫情期间,这位家长一个人带着孩子十分辛苦,在科主任及护士长的调配下,小儿神经专科的医护人员们以患儿为中心,体患儿之苦,察家长之情,立马帮助这位家长办理入院手续流程,这个孩子安排了床位,并安抚好了家属的情绪,详细解答了家属的问题。"这里的医护人员用温柔、和蔼的话语给我一步步指导,若没有你们,真不知道怎么办了!真的十分感谢!"当我们给孩子戴上用于检查的帽子,评估好孩子的病情,指导完癫痫用药之后,家长悬着的心才放下来,他的一番简单话语就是对我们的高度评价,简单的话语,暖心的语句,都是我们最好的情感加温器。

## "向阳花爱心小屋"医学人文关怀暖人心

"这个孩子能够活下来,这不是单纯的偶然,它是一个由医生、家属、社会慈善,也包括孩子自身的求生力量共同来造就的奇迹!它有必然的因素在里面。" 也正是带着这样的哲学观与感恩心,博博妈妈每每与朋友交往聊天中,总是会由衷地讲述这个故事。

4岁的博博发作性肢体抽搐,每间隔 20 天左右就会发作,一天内发作数次。常年服用药物控制症状,但仍控制不住发作,常年服用药物控制症状,但仍控制不住发作。此外,博博的生长发育较同龄人缓慢,发育商相当于 10 个月大婴儿,不会说话、不理解他人言语。每当癫痫发作,都令他更加痛苦。辗转于国内多家医院,来我院就诊,被诊断为"结节性硬化、孤独症",因为就诊时间长,家庭条件差,父母每天唉声叹气,愁容满面,科室联系医务社工为孩子寻找医疗救助平台,为患者筹集善款 4.5 万元,并组织多学科讨论,为孩子实施了致痫结节切除术,结合药物治疗,术后未再出现癫痫发作,预后良好。

科室在2016年2月2日携手医务社工部共建首个医学人文关怀基地,驻地

病房医务社工部的工作人员联合志愿者为患儿提供常规性的系列医学人文关怀活动。开展文艺课堂:拼图、绘画、折纸、音乐等;爱心义剪、节假日送温暖、提供救助;有心理咨询师证的专业人员为有情绪障碍的患儿及家属进行心理疏导。科室采取多种形式、多种渠道、多种方式来给患者提供个性化关怀服务,提高患者就医感受。共同推动科室文化建设,创造人文氛围,培养人文理念,传承人文精神,塑造人文灵魂。







孩子不仅是祖国的花朵,同样也是祖国的未来,关爱孩子们的身心健康,特别是那些与疾病斗争的孩子尤其重要,我们不仅要帮助解决孩子们身体上的病痛,同时也要给孩子们一定的心灵慰藉,从而更好促进孩子们的身心发展,正如特鲁

多医生所言: "有时去治愈,常常去帮助,总是去安慰"。在不断强化临床专业技能的同时,也不断提升人文关怀与医疗服务水平,将人性化服务融入每个工作细节之中,不断进取,以满怀一颗热忱之心,守护万千孩子的生命健康。

"健康所系,性命相托"是我们牢记在心的誓言,"救死扶伤,无私奉献"是我们的人生信条。我们时刻将责任感外化于行,内化于心,真正带给别人温暖的医护人员既是患者的健康守门人,也是患儿心中的英雄。我们用爱心、细心、耐心,用诚意和正义,为孩子们撑起生命的保护伞,治愈疾病,减轻痛苦,延长生命,让患儿快乐地、有质量地生活。一切为了孩子,孩子们能健康成长,那我们这份守护的力量也就绽放了光芒。













编号: JJJM-2023-1-1

引用格式: Zhang L, Wang J, Yang Y, Deng Q, Yan Z, Liu X, Zhang S, Zhou J, Guan Y, Liu C, Luan G, Wang M. Semiology characteristics and location of epileptogenic zone in motor seizures of axial and shoulder girdle muscles assessed by video stereoelectroencephalography study in 65 patients. Epileptic Disord. 2023. doi: 10.1002/epd2.20054.

## 通信作者: 王梦阳

#### **Abstract**

Objective: To study the semiology characteristics of motor seizures of axial and shoulder girdle muscles (ASMs) by stereoelectroencephalography (SEEG) and its value in determining location of epileptogenic zone. Methods: A total of 598 patients underwent SEEG assessment in Sanbo Brain Hospital were reviewed; 65 patients with ASMs selected. Thirteen semiology feature items were extracted according to the location and symmetry of involved axial muscles, direction of movement, etc. Seizures were grouped with items, and the k-means was used to analyze association between ASMs semiology characteristics and seizure-onset zone (SOZ). Results: The SOZs of ASMs involved 23 combinations of seven different brain regions: 31 patients (47.7%) had one brain region, 19 (29.2%) had two, 14 (21.5%) had three, and one patient (1.5%) had four. One hundred and fifteen brain regions were analyzed. Seven brain regions accounted for a significant difference in chi-square test,  $\chi 2 = 62.79$ , p < 0.0001, with the highest proportion of insular and perisylvian. The k-means method identified two clusters: cluster 1 had a high degree of agreement with temporal lobe epilepsy (12/15), characterized by less shrug-like movement, later involvement of axial shoulder girdle muscles, longer duration, and lower seizure frequency; cluster 2 had a high degree of agreement with posterior cortex epilepsy (14/18), characterized by earlier involvement of axial shoulder girdle muscles, shorter duration, and higher seizure frequency. In frontal lobe, insular and perisylvian, anterior and middle cingulate gyrus, are the two categories accounted for similar proportion. Seizure-onset lateralized at the

contralateral of unilateral cervical tonic, with rate of seizure-free was 73.7%. **Significance:** The incidence of ASMs is high in insular and perisylvian. Unilateral cervical tonic seizures have good lateralizing value. Based on semiology characteristics, ASMs can be roughly clustered into two categories, which can only effectively distinguish the origins of temporal lobe and posterior cortex, with low discrimination for the seizure-onset of other lobes.

编号: JJJM-2023-1-2

引用格式: Xie H, Ma J, Ji T, Liu Q, Cai L, Wu Y. Efficacy of vagus nerve stimulation in 95 children of drug-resistant epilepsy with structural etiology. Epilepsy Behav. 2023;140:109107. doi: 10.1016/j.yebeh.2023.109107.

第一作者: 谢涵

#### **Abstract**

Vagus nerve stimulation (VNS) is one of the treatment options for drug-resistant epilepsy (DRE). To analyze the efficacy of VNS in children of DRE with structural etiology, we conducted a cohort study including 95 patients of DRE with structural etiology who underwent VNS treatment. Patients were followed up every 3 months at the outpatient department or via a remote programming platform. The median follow-up period was 2.6 years (range 1.0-4.6 years). The respective responder rates at 6, 12, 18, and 24 months of follow-up were 40.0% (38/95), 52.6% (50/95), 56.0% (47/84), and 59.7% (37/62). The respective seizure-free rates at 12, 18, and 24 months of follow-up were 8.4% (8/95), 9.5% (8/84), and 9.7% (6/62). The patients were divided into four groups based on etiologies: malformations of cortical development (n = 26), post-encephalitic lesions (n = 36), perinatal brain injury lesions (n = 31), and hippocampal sclerosis (n = 2). The respective responder rates at 12 months of follow-up in these groups were 53.8% (14/26), 52.8% (19/36), 51.6% (16/31), and 50.0% (1/2). There were no significant differences in gender, age at onset, age at stimulator implantation, epilepsy duration prior to VNS implantation, number of anti-

seizure medications ever tried before VNS treatment, pulse amplitude of VNS, specific structuraletiologies, lobe distribution or hemispheric side of structural lesions between responders and non-responders. Of the 95 patients, 8 (8.4%) underwent lesion surgery or hemispherectomy before VNS implantation, and 6/8 (75%) of these patients had a >50% reduction in seizure frequency. One patient who had a corpus callosotomy before VNS implantation had no response to VNS treatment. In conclusion, VNS is an effective treatment in children of DRE with structural etiology. There was no significant difference in VNSefficacy in patients with different structural etiologies. Vagus nerve stimulation treatment may also control seizures well in some patients with poor outcomes after lesion resection or hemispherectomy before VNS implantation.

编号: JJJM -2023-1-3

引用格式: Kuang S, Wang J, Wei Z, Zhai F, Liang S. The regulatory function of lncRNA and constructed network in epilepsy. Neurol Sci. 2023;44(5):1543-1554. doi: 10.1007/s10072-023-06648-5.

通信作者: 梁树立

#### Abstract

**Background:** Epilepsy is a neurological disease characterized by neural network dysfunction. Although most reports indicate that the pathological process of epilepsy is related to inflammation, synaptic plasticity, cell apoptosis, and ion channel dysfunction, the underlying molecular mechanisms of epilepsy are not fully understood. **Methods:** This review summarizes the latest literature on the roles and characteristics of long noncoding RNAs (lncRNAs) in the pathogenesis of epilepsy. **Results:** lncRNAs are a class of long transcripts without protein-coding functions that perform important regulatory functions in various biological processes. lncRNAs are involved in the regulation of the pathological process of epilepsy and are abnormally expressed in both patients and animal models. This review provides an overview of research progress in

epilepsy, the multifunctional features of lncRNAs, the lncRNA expression pattern related to epileptogenesis and status epilepticus, and the potential mechanisms for the two interactions contributing to epileptogenesis and progression. **Conclusion:** lncRNAs can serve as new diagnostic markers and therapeutic targets for epilepsy in the future.

编号: JJJM -2023-1-4

引用格式: Gao F, Liu C, Zhang L, Liu T, Wang Z, Song Z, Cai H, Fang Z, Chen J, Wang J, Han M, Wang J, Lin K, Wang R, Li M, Mei Q, Ma X, Liang S, Gou G, Xue N. Wearable and flexible electrochemical sensors for sweat analysis: a review. Microsyst Nanoeng. 2023;9:1. doi: 10.1038/s41378-022-00443-6.

通信作者:梁树立

#### **Abstract**

Flexible wearable sweat sensors allow continuous, real-time, noninvasive detection of sweat analytes, provide insight into human physiology at the molecular level, and have received significant attention for their promising applications in personalized health monitoring. Electrochemical sensors are the best choice for wearable sweat sensors due to their high performance, low cost, miniaturization, and wide applicability. Recent developments in soft microfluidics, multiplexed biosensing, energy harvesting devices, and materials have advanced the compatibility of wearable electrochemical sweat-sensing platforms. In this review, we summarize the potential of sweat for medical detection and methods for sweat stimulation and collection. This paper provides an overview of the components of wearable sweat sensors and recent developments in materials and power supply technologies and highlights some typical sensing platforms for different types of analytes. Finally, the paper ends with a discussion of the challenges and a view of the prospective development of this exciting field.

编号: JJJM -2023-1-5

引用格式: Xu J, Wang J, Liu T, Wang Y, Chen F, Yuan L, Zhai F, Ge M, Liang S. Factors that Influence Subdural Hemorrhage Secondary to Intracranial Arachnoid Cysts in Children. World Neurosurg. 2023:S1878-8750(23)00324-8. doi: 10.1016/j.wneu.2023.03.029.

## 通信作者: 梁树立

#### **Abstract**

**Objective:** This study aimed to investigate factors that influence subdural haemorrhage (SDH) secondary to intracranial arachnoid cysts (IACs) in children. Methods: Data of children with unruptured IACs (IAC group) and those with SDH secondary to IACs (IAC-SDH group) were analyzed. Nine factors, sex, age, birth type (vaginal or caesarean), symptoms, side (left, right, or midline), location (temporal or nontemporal), image type (I, II, or III), volume, and maximal diameter, were selected. IACs were classified as types I, II, and III according to their morphological changes observed on computed tomography images. Results: There were 117 boys (74.5%) and 40 girls (25.5%); 144 (91.7%) patients comprised the IAC group and 13 (8.3%) comprised the IAC-SDH group. There were 85 (53.8%) IACs on the left side, 53 (33.5%) on the right side, 20 (12.7%) in the midline region, and 91 (58.0%) in the temporal region. The univariate analysis showed significant differences in age, birth type, symptoms, cyst location, cyst volume, and cyst maximal diameter (P < 0.05) between the 2 groups. Logistic regression using the synthetic minority oversampling technique model showed that image type III and birth type were independent factors that influenced SDH secondary to IACs ( $\beta 0 = 4.143$ ;  $\beta$  for image type = -3.979;  $\beta$  for birth type = -2.542) and that the representative area under the receiver-operating characteristic curve value was 0.948 (95% confidence interval, 0.898-0.997). Conclusions: IACs are more common in boys than in girls. They can be divided into 3 groups according to their morphological changes on computed tomography images. Image type III and caesarean delivery were independent factors that influenced SDH secondary to IACs.

编号: JJJM-2023-1-6

引用格式: 李安, Adamu Alhamdu, 薛国芳. 新型冠状病毒感染期间对癫痫患者的疾病管理建议. 癫痫杂志. 2023;9(2):173-177. doi: 10.7507/2096-0247.202212005.

通信作者: 薛国芳

#### **Abstract**

新型冠状病毒感染已持续三年之久,随着国务院感染防控二十条和新十条政策的 出台,全国上下 防控政策由全面防控调整至精确防控,国内感染防控模式进入 新阶段。在新型冠状病毒感染期间,癫痫患者在就 医、用药等方面受到很大影响。在新型冠状病毒感染防控的新阶段,癫痫患者的疾病管理仍受到不同程度的 影响。但由于缺乏及时更新的指导建议,该患者群体广泛出现焦虑、抑郁等心 理健康问题,对疾病控制极为不利。 为了帮助癫痫患者管理病情、控制癫痫发 作,从感染防控、日常生活、用药、就医等方面提出指导性建议供癫痫患者及家 人参考。

编号: JJJM-2023-1-7

引用格式: 胡文瀚, 柏建军, 周文静, 张建国, 张凯. 磁共振引导下激光间质热疗术治疗药物难治性癫痫的操作规范. 癫痫杂志. 2023;9(1): 1-4. doi: 10.7507/2096-0247.202211008.

第一作者: 胡文瀚 通信作者: 张凯

激光间质热疗术(Laser interstitial thermal therapy,LITT)由 Bown 于 1983 年首次描述, 1990 年 Sugiyama 首次使用该术式治疗脑部病变,美国食品药品监督管理局(Food and Drug Administration,FDA)于 2007 年批准了 LITT 治疗颅内疾病,Curry 等于 2012 年报道了磁共振引导下激光间质热疗术(MRI-guided laser

interstitial thermal therapy,MgLITT)治疗癫痫。此后,MgLITT治疗癫痫得到了 迅速的发展。来自论文的数据统计显示,世界范围内共有 1800 多例癫痫患者接 受了 MgLITT 手术,涉及的病理包括下丘脑错构瘤(Hypothalamic hamartoma, HH)、颞叶内侧硬化、局灶性皮质发育不良(Focal cortical dysplasia,FCD)、各 种发育性肿瘤、海绵状血管畸形等。依据诸多文献报道的结果,MgLITT 治疗多 种病理所致癫痫的疗效不劣于传统的开颅切除性手术,并以安全、准确、微创的 优势,在未来可能改变癫痫外科的手术模式。目前已上市的 MgLITT 设备包括美 国 Medtronic 公司的 Visualase 系统和加拿大 Monteris 公司的 NeuroBlate 系统, 我国的 Sinovation 公司于 2020 年研制出国产 MgLITT 系统,同年完成动物实验 并开始了上市前的临床试验。2020年8月12日北京天坛医院率先开展了MgLITT 治疗一例颞叶内侧癫痫,截止到 2022 年 5 月,已完成 230 例 MgLITT 治疗不同 病理类型癫痫的手术,初步积累了相关的经验。清华大学玉泉医院于2020年11 月开始开展 MgLITT 治疗难治性癫痫患者,证实在非术中磁共振条件下也可安 全有效的开展 MgLITT 治疗。中国抗癫痫协会谭启富基金管理委员会委托北京 天坛医院和清华大学玉泉医院相关人员撰写 MgLITT 手术操作规范,从术前患 者筛选、手术流程和术后管理等方面分别介绍,旨在助力于这一技术未来在中国 的规范开展。

# 青春虚度无所成,白首衔悲亦何及。

——《放歌行》

# 纸上得来终觉浅, 绝知此事要躬行

——《冬夜读书示子聿》

编号: HJL-2023-1-1

引用格式: Huang C, You Z, He Y, Li J, Liu Y, Peng C, Liu Z, Liu X and Sun J. Combined transcriptomics and proteomics forecast analysis for potential biomarker in the acute phase of temporal lobe epilepsy. Front Neurosci, 2023; 17:1145805. doi: 10.3389/fnins.2023.1145805...

通信作者:孙家行

#### **Abstract**

**Background:** Temporal lobe epilepsy (TLE) is a common chronic episodic illness of the nervous system. However, the precise mechanisms of dysfunction and diagnostic biomarkers in the acute phase of TLE are uncertain and hard to diagnose. Thus, we intended to qualify potential biomarkers in the acute phase of TLE for clinical diagnostics and therapeutic purposes. Methods: An intra-hippocampal injection of kainic acid was used to induce an epileptic model in mice. First, with a TMT/iTRAQ quantitative labeling proteomics approach, we screened for differentially expressed proteins (DEPs) in the acute phase of TLE. Then, differentially expressed genes (DEGs) in the acute phase of TLE were identified by linear modeling on microarray data (limma) and weighted gene co-expression network analysis (WGCNA) using the publicly available microarray dataset GSE88992. Co-expressed genes (proteins) in the acute phase of TLE were identified by overlap analysis of DEPs and DEGs. The least absolute shrinkage and selection operator (LASSO) regression and support vector machine recursive feature elimination (SVM-RFE) algorithms were used to screen Hub genes in the acute phase of TLE, and logistic regression algorithms were applied to develop a novel diagnostic model for the acute phase of TLE, and the sensitivity of the diagnostic model was validated using receiver operating characteristic (ROC) curves. Results: We screened a total of 10 co-expressed genes (proteins) from TLE-associated DEGs and DEPs utilizing proteomic and transcriptome analysis. LASSO and SVM-RFE algorithms for machine learning were applied to identify three Hub genes: Ctla2a, Hapln2, and Pecam1. A logistic regression algorithm was applied to establish and

validate a novel diagnostic model for the acute phase of TLE based on three Hub genes in the publicly accessible datasets GSE88992, GSE49030, and GSE79129. **Conclusion:** Our study establishes a reliable model for screening and diagnosing the acute phase of TLE that provides a theoretical basis for adding diagnostic biomarkers for TLE acute phase genes.

编号: HJL-2023-1-2

引用格式: Zhang W, Xin M, Song G, Liang J. Childhood absence epilepsy patients with cognitive impairment have decreased sleep spindle density. Sleep Med. 2023;103:89-97.

## 通信作者: 梁建民

#### **Abstract**

Objective: To explore the differences in sleep spindle (SS) characteristics during stage N2 sleep between children with childhood absence epilepsy and healthy controls, and between children with childhood absence epilepsy with or without cognitive impairment. Methods: We recruited 29 children (14 females, 15 males, mean age: 8 (2.5) years) with childhood absence epilepsy who did not undergone antiseizure treatments previously and 30 age-matched controls (14 females, 16 males, mean age: 9 (3.0) years). For all patients, data on medical history were collected. Each child was monitored overnight by long-term video electroencephalography and was evaluated by the Wechsler Intelligence Scale for Children-Fourth Edition. Next, we compared anterior SS characteristics, including density, frequency, cycle length, duration, amplitude, and percentage of sleep stages. Results: The childhood absence epilepsy group exhibited lower spindle density and duration in the first 37.5 min of stage N2 sleep than the control group (P < 0.01). A decrease in spindle density could be observed in the childhood absence epilepsy group with aggravated cognition impairment. The spindle density was substantially lower in the cognitively impaired group than in the cognitively unimpaired group (P < 0.01). No significant differences were observed in SS amplitude, SS frequency, SS cycle length, and the distribution of sleep stages. **Conclusions:** Reduction in spindle density and duration is associated with the mechanisms underlying childhood absence epilepsy. The deficit in SS density is related with impaired cognition. This deficiency in SSs may be a useful predictive indicator of cognitive impairment in children with absence epilepsy, indicating that SSs may become a useful biomarker and potential adjuvant anti-seizure target for cognitive impairment caused by childhood absence epilepsy.

编号: HJL-2023-1-3

引用格式: Zhang B, Chen T, Hao X, Xin M, Liang J. Electroclinical characteristics of photosensitive epilepsy: A retrospective study of 31 Chinese children and literature review. Front Pediatr. 2023;11:994817.

通信作者: 梁建民

#### **Abstract**

**Objective:** The objective of this study was to better understand the clinical features of photosensitive epilepsy (PSE) in Chinese children. **Methods:** Thirty-one children with PSE were screened out of 398 children with epilepsy who were consecutively diagnosed by the video-electroencephalogram (VEEG) monitoring method and by using an intermittent photic stimulation (IPS) test. Their EEGs and clinical features were retrospectively analyzed, and their treatment outcomes were followed up. **Results:** PSE accounted for 7.79% (31/398) of children with epilepsy during the observation period in our single epilepsy center. The male to female ratio of PSE was 1:3.43, and the average seizure onset age was  $7.8 \pm 3.28$  years. The highest range of frequency sensitivity of the IPS test for the induction of EEG epileptic discharge or electroclinical seizures was within 10-20 Hz. Electroclinical seizures were induced in 41.94% (13/31) of PSE patients by using the IPS test, while EEG discharge without clinical seizures was induced in 58.06% (18/31) of PSE patients. Among all PSE patients, an IPS-positive reaction in the eye-closure state was induced in 83.87% of patients, and this

rate was significantly higher than that in the eye-opened state (41.94%) or eye-closed state (35.48%). (Eye-closure IPS stimulation means: make the subjects close their eyes at the beginning of each stimulation, open their eyes at the end of the stimulation, and close their eyes again at the beginning of the next stimulation, and so on. While Eye-closed IPS stimulation means the stimulation is started after 5 s of eye closure, and the subjects are kept closed throughout the whole process.) The common and effective drugs used for single or combined therapy in PSE children were valproic acid and levetiracetam. **Conclusion:** This study provides some useful information about electroclinical characteristics in a cohort of 31 PSE children. It may be beneficial for pediatric neurologists in terms of paying more attention to PSE and correctly dealing with it.



2022年3月12日中抗协会领导和青委们在海口市种下的木棉树和 黄花风铃木在海南省抗癫痫协会和李其富青委团队的看护下全部存 活并茁壮成长

编号: ZWG-2023-1-1

引用格式: Zhu Y, Guo C. A hand motion capture method based on infrared thermography for measuring fine motor skills in biomedicine. Artif Intell Med. 2023; 135:102474. doi: 10.1016/j.artmed.2022.102474.

通信作者: 郭崇伦

#### **Abstract**

Many biomedical applications require fine motor skill assessments; however, real-time and contactless fine motor skill assessments are not typically implemented. In this study, we followed the 2D-to-3D pipeline principle and proposed a transformer-based spatialtemporal network to accurately regress 3D hand joint locations by inputting infrared thermal video for eliminating need of multiple cameras or RGB-D devices. We also developed a dataset composed of infrared thermal videos and ground truth annotations for training. The label represents a set of 3D joint locations from infrared optical trackers, which is considered the gold standard for clinical applications. To demonstrate their potential, the proposed method was used to measure the finger motion angle, and we investigated its accuracy by comparing the proposal with the Azure Kinect system and Leap Motion system. On the proposed dataset, the proposed method achieved a 3D hand pose mean error of less than 14 mm and outperforms the other deep learning methods. When the error thresholds were larger than approximately 35 mm, our method first to achieved excellent performance (>80%) in terms of the fraction of good frames. For the finger motion angle calculation task, the proposed and commercial systems had comparable inter-system reliability (ICC2,1 ranging from 0.81 to 0.83) and excellent validity (Pearson's r-values ranging from 0.82 to 0.86). We believe that the proposed approaches can capture hand motion and measure finger motion angles and can be used in different biomedicine scenarios as an effective evaluation tool for fine motor skills.

编号: ZWG -2023-1-2

引用格式: Chen C, Wang Y, Ye L, Xu J, Ming W, Liu X, Hu L, Ye H, Xu C, Wang Y, Wang Z, Ding Y, Zhu J, Ding M, Chen Z, Wang S. A region-specific modulation of sleep slow waves on interictal epilepsy markers in focal epilepsy. Epilepsia. 2023; 64(4):973-985. doi: 10.1111/epi.17518.

## 通信作者: 王爽

#### **Abstract**

Objective: Sleep strongly activates interictal epileptic activity through an unclear mechanism. We investigated how scalp sleep slow waves (SSWs), whose positive and negative half-waves reflect the fluctuation of neuronal excitability between the up and down states, respectively, modulate interictal epileptic events in focal epilepsy. Methods: Simultaneous polysomnography was performed in 45 patients with drugresistant focal epilepsy during intracranial electroencephalographic recording. ScalpSSWs and intracranial spikes and ripples (80–250Hz) were detected; ripples were classified as type I (co-occurring with spikes) or type II (occurring alone). The Hilbert transform was used to analyze the distributions of spikes and ripples in the phases of SSWs. Results: Thirty patients with discrete seizure-onset zone (SOZ) and discernable sleep architecture were included. Intracranial spikes and ripples accumulated around the negative peaks of SSWs and increased with SSW amplitude. Phase analysis revealed that spikes and both ripple subtypes in SOZ were similarly facilitated by SSWs exclusively during down state. In exclusively irritative zones outside SOZ (EIZ), SSWs facilitated spikes and type I ripples across a wider range of phases and to a greater extent than those in SOZ. The type II and type I ripples in EIZ were modulated by SSWs in different patterns. Ripples in normal zones decreased specifically during the up-todown transition and then increased after the negative peak of SSW, with a characteristically high post-/pre-negative peak ratio. Significance: SSWs modulate interictal events in an amplitude-dependent and region-specific pattern. Pathological ripples and spikes were facilitated predominantly during the cortical down state.

Coupling analysis of SSWs could improve the discrimination of pathological and physiological ripples and facilitate seizure localization.

编号: ZWG-2023-1-3

引用格式: Xu S, Xu J, Chen C, Ye L, Wang S, Miao P, Zhu J. Multifocal mild malformation of cortical development with oligodendroglial hyperplasia (MOGHE) associated with SLC35A2 brain mosaicism. Clin Neurophysiol. 2023;145:22-25. doi: 10.1016/j.clinph.2022.11.001

通信作者: 王爽

Abstract

Mild malformation of cortical development with oligodendroglial hyperplasia (MOGHE) is a novel pathological entity proposed in 2017, which is associated with drug-resistant focal epilepsy (Schurr et al., 2017). The clinical features and pathogenesis of MOGHE have not yet been well characterized. Recently, the mosaic brain SLC35A2 variant was reported to be a genetic marker of MOGHE (Bonduelle et al., 2021). Here, we present a unique patient with two separate MOGHE lesions and SLC35A2 brain mosaicism, who achieved seizure freedom after comprehensive presurgical evaluation and tailored lobular resection

编号: ZWG -2023-1-4

引用格式: Hu L, Xiong K, Ye L, Yang Y, Chen C, Wang S, Ding Y, Wang Z, Ming W, Zheng Z, Jiang H, Li H, Zhu J, Xu C, Wang Y, Ding M, Chen Z, Wu Y, Wang S. Ictal EEG desynchronization during low-voltage fast activity for prediction of surgical outcomes in focal epilepsy. J Neurosurg. 2022 Dec 16:1-10. doi: 10.3171/2022.11.JNS221469.

通信作者:王爽

27

#### Abstract

OBJECTIVE: The authors investigated alterations in functional connectivity (FC) and EEG power during ictal onset patterns of low-voltage fast activity (LVFA) in drug-resistant focal epilepsy. They hypothesized that such changes would be useful to classify epilepsy surgical outcomes.

METHODS: In a cohort of 79 patients with drug-resistant focal epilepsy who underwent stereoelectroencephalography (SEEG) evaluation as well as resective surgery, FC changes during the peri-LVFA period were measured using nonlinear regression (h2) and power spectral properties within/between three regions: the seizure onset zone (SOZ), early propagation zone (PZ), and noninvolved zone (NIZ). Desynchronization and power desynchronization h2 indices were calculated to assess the degree of EEG desynchronization during LVFA. Multivariate logistic regression was employed to control for confounding factors. Finally, receiver operating characteristic curves were generated to evaluate the performance of desynchronization indices in predicting surgical outcome.

RESULTS: Fifty-three patients showed ictal LVFA and distinct zones of the SOZ, PZ, and NIZ. Among them, 39 patients (73.6%) achieved seizure freedom by the final follow-up. EEG desynchronization, measured by h2 analysis, was found in the seizure-free group during LVFA: FC decreased within the SOZ and between regions compared with the pre-LVFA and post-LVFA periods. In contrast, the non–seizure-free group showed no prominent EEG desynchronization. The h2 desynchronization index, but not the power desynchronization index, enabled classification of seizure-free versus non–seizure-free patients after resective surgery.

CONCLUSIONS: EEG desynchronization during the peri-LVFA period, measured by within-zone and between-zone h2 analysis, may be helpful for identifying patients with favorable postsurgical outcomes and also may potentially improve epileptogenic zone identification in the future.

编号: ZWG-2023-1-5

引用格式: Zhang Z, Li Q, Jiang T, Fang J. Reflex seizures induced by micturition:

a case report. Acta Epileptologica. 2023;5:4. doi:10.1186/s42494-022-00107-y.

通信作者: 方嘉佳

Abstract

Background: Reflex seizures (RS) induced by micturition are extremely rare, and the

clinical and electroencephalogram features of RS are not widely known among

clinicians. In particular, the origin of the epileptic area is still unclear. Case

presentation: An 8-year-old girl who had generalized tonic-clonic seizures was

diagnosed with RS induced by micturition based on the clinical manifestation and EEG

recordings. We also reviewed the clinical and EEG characteristics of RS induced by

micturition in literature by searching the databases of PubMed and MEDLINE using

keywords "micturition reflex seizure", "reflex seizure induced by micturition", and

"micturition induced seizure" by January 2022. We speculate that the mechanism of

micturition-induced RS may involve excessive neuronal excitation in regions that

participate in micturition. Conclusions: The RS in this patient was considered to be

induced by micturition. Awareness should be raised to this rare form of RS among

practitioners.

编号: ZWG -2023-1-6

引用格式:Lin R, Lin J, Xu Y, Yu Y, Foster E, Lin M, Xu H, Li X, Ye J, Yao F, Xu

H, Chen M, Chen J, Zheng H, Li J, Zheng M, Huang S, Zhu S, Yang Y, Wang X.

Development and validation of a novel radiomics-clinical model for predicting

post-stroke epilepsy after first-ever intracerebral haemorrhage. Eur Radiol. 2023

Feb 3. doi: 10.1007/s00330-023-09429-y.

通信作者:王新施

29

#### **Abstract**

Background: Post-stroke epilepsy (PSE) is associated with increased morbidity and mortality. This study aimed to develop and validate a novel prediction model combining clinical factors and radiomics features to accurately identify patients at high risk of developing PSE after intracerebral haemorrhage (ICH). Methods: Researchers performed a retrospective medical chart review to extract derivation and validation cohorts of patients with first-ever ICH that attended two tertiary hospitals in China between 2010-2020. Clinical data were extracted from electronic medical records and supplemented by tele-interview. Predictive clinical variables were selected by multivariable logistic regression to build the clinical model. Predictive radiomics features were identified and a Rad-score was calculated according to the coefficient of the selected feature. Both clinical variables and radiomic features were combined to build the radiomics-clinical model. Performance of the clinical, Rad-score, and combined models were compared. Results: 1571 patients were included in the analysis. Cortical involvement, early seizures within 7 days of ICH, NIHSS score, and ICH volume were included in the clinical model. Rad-score, instead of ICH volume, was included in the combined model. The combined model exhibited better discrimination ability and achieved an overall better benefit against threshold probability than the clinical model in the decision curve analysis (DCA). Conclusion: The combined radiomics-clinical model was better able to predict ICH-associated PSE compared to the clinical model. This can help clinicians better predict individual patient's risk of PSE following a first-ever ICH and facilitate earlier PSE diagnosis and treatment.

# 欢迎大家引用上述论文

编号: XEY-2023-1-1

引用格式: Wang X, You B, Yin F, Chen C, He H, Liu F, Pan Z, Ni X, Pang N, Peng J. A presumed missense variant in the U2AF2 gene causes exon skipping in neurodevelopmental diseases. J Hum Genet. 2023. doi: 10.1038/s10038-023-01128-2.

## 通信作者: 彭镜

#### Abstract

U2 small nuclear RNA auxiliary factor 2 (U2AF2) is an indispensable pre-mRNA splicing factor in the early process of splicing. Recently, U2AF2 was reported as a novel candidate gene associated with neurodevelopmental disorders. Herein, we report a patient with a novel presumed heterozygous missense variant in the U2AF2 gene (c.603G>T), who has a similar clinical phenotype as the patient reported before, including epilepsy, intellectual disability, language delay, microcephaly, and hypoplastic corpus callosum. We reviewed the phenotypic and genetic spectrum of patients with U2AF2-related neurological diseases, both newly diagnosed and previously reported. To investigate the possible pathogenesis, EBV-immortalized lymphoblastoid cells were derived from the peripheral blood obtained from the patient and control groups. Furthermore, according to the results of WB, RT-PCR, Q-PCR, and cDNA sequencing of RT-PCR products, the presumed missense variant c.603G>T caused exon 6 skipping in the U2AF2 mRNA transcript and led to a truncated protein (p.E163 E201del). Cell Counting Kit-8 (CCK-8) and cell cycle detection demonstrated that the variant c.603G>T inhibited the proliferation of patient lymphocyte cells compared with the control group. This study is aimed at expanding the phenotypic and genetic spectrum of U2AF2-related neurodevelopmental diseases and investigating the potential effects. This is the first report of the possible pathogenesis of a U2AF2 gene pathogenic variant in a patient with neurodevelopmental diseases and shows that a novel presumed missense variant in the U2AF2 gene causes exon skipping.

编号: XEY 2023-1-2

引用格式: Wu T, Zhang C, He F, Yang L, Yin F, Peng J. Large heterozygous deletion and uniparental disomy masquerading as homozygosity in CHKB gene. Mol Genet Genomic Med. 2023:e2162. doi: 10.1002/mgg3.2162.

通信作者: 彭镜

#### **Abstract**

Background: CHKB mutations have been described in 49 patients with megaconial congenital muscular dystrophy, which is a rare autosomal recessive disorder, of which 40 patients showed homozygosity. Methods: Peripheral blood genomic DNA samples were extracted from patients and their parents and were tested by whole exome sequencing. Quantitative PCR was performed to detect deletion. Single nucleotide polymorphism analysis was performed to identify uniparental disomy. Quantitative PCR and western blot were used to measure the expression level of CHKB in patient 1- derived immortalized lymphocytes. Mitochondria were observed in lymphocytes by electron microscopy. Results: Two unrelated cases born to non- consanguineous parents were diagnosed with megaconial congenital muscular dystrophy due to apparently homozygous mutations (patient 1: c.225- 2A>T; patient 2: c.701C>T) in the CHKB gene using whole exome sequencing. Quantitative PCR revealed that patient 1 had a large deletion encompassing the CHKB gene, inherited from the mother. Single nucleotide polymorphism analysis revealed patient 2 had paternal uniparental isodisomy containing the CHKB gene. In the immortalized lymphocytes from patient 1, decreased expression of CHKB was revealed by quantitative PCR and western blot, and giant mitochondria were observed using electron microscopy. Conclusion: We provide a possibility to detect giant mitochondria in othercells when muscle was not available. Moreover, clinicians should be aware that homozygous variants can be masqueraded by uniparental disomy or large deletions in offspring of nonconsanguineous parents, and excessive homozygositymay be misdiagnosed.

编号: XEY-2023-1-3

引用格式: Li C, Cai HB, Zhou Q, Zhang HQ, Wang M, Kang HC. Sleep disorders in the acute phase of coronavirus disease 2019: an overview and risk factor study. Ann Gen Psychiatry. 2023;22(1):3. doi: 10.1186/s12991-023-00431-8.

通信作者: 康慧聪

#### **Abstract**

Background: Sleep disorders are common during the outbreak of pandemic diseases, and similar disorders are noted in hospitalized COVID-19 patients. It is valuable to explore the clinical manifestations and risk factors for sleep disorders in COVID-19 patients. Methods: Inpatients with COVID-19 were enrolled. Detailed clinical information was collected, and sleep quality was assessed by PSQI. Patients were divided into a sleep disorder group and a normal group based on a PSQI  $\geq 7$ , and the clinical features were compared between the groups. Results: Fifty-three patients were enrolled, and 47.2% presented sleep disorders. Sleep disorders were associated with older age (> 50), anemia and carbon dioxide retention. Furthermore, factors associated with abnormal component scores of the PSQI were: (1) patients with older age were more likely to have decreased sleep quality, prolonged sleep latency, decreased sleep efficiency, sleep disturbances, and daytime dysfunction; (2) decreased sleep quality and prolonged sleep latency were associated with dyspnea, whereas carbon dioxide retention and more lobes involved in chest CT were associated with prolonged sleep latency; (3) decreased sleep efficiency was more prevalent in patients with anemia. Conclusions: Sleep disorders were prevalent in patients during the acute phase of COVID-19, and many risk factors (older age, anemia, carbon dioxide retention, the number of lobes involved in chest CT, and dyspnea) were identified. It is important to assess the presence of sleep disorders in patients to provide early intervention.

编号: XEY-2023-1-4

引用格式: Cai C, Kang H, Hashemi A, Chen D, Diwakar M, Haufe S, Sekihara K, Wu W, Nagarajan SS. Bayesian Algorithms for Joint Estimation of Brain Activity and Noise in Electromagnetic Imaging. IEEE Trans Med Imaging. 2023;42(3):762-773. doi: 10.1109/TMI.2022.3218074.

第一作者: 康慧聪

#### **Abstract**

Simultaneously estimating brain source activity and noise has long been a challenging task in electromagnetic brain imaging using magneto- and electroencephalography. The problem is challenging not only in terms of solving the NP-hard inverse problem of reconstructing unknown brain activity across thousands of voxels from a limited number of sensors, but also for the need to simultaneously estimate the noise and interference. We present a generative model with an augmented leadfield matrix to simultaneously estimate brain source activity and sensor noise statistics in electromagnetic brain imaging (EBI). We then derive three Bayesian inference algorithms for this generative model (expectation-maximization (EBI-EM), convex bounding (EBI-Convex) and fixed-point (EBI-Mackay)) to simultaneously estimate the hyperparameters of the prior distribution for brain source activity and sensor noise. A comprehensive performance evaluation for these three algorithms is performed. Simulations consistently show that the performance of EBI-Convex and EBI-Mackay updates is superior to that of EBI-EM. In contrast to the EBI-EM algorithm, both EBI-Convex and EBI-Mackay updates are quite robust to initialization, and are computationally efficient with fast convergence in the presence of both Gaussian and real brain noise. We also demonstrate that EBI-Convex and EBI-Mackay update algorithms can reconstruct complex brain activity with only a few trials of sensor data, and for resting-state data, achieving significant improvement in source reconstruction and noise learning for electromagnetic brain imaging.

编号: XEY-2023-1-5

引用格式: Zhu HY, Tang YX, Xiao L, Wen SR, Wu YX, Yang ZQ, Zhou L, Xiao B, Feng L, Hu S. Metabolic profiles and correlation with surgical outcomes in mesial versus neocortical temporal lobe epilepsy. CNS Neurosci Ther. 2023. doi: 10.1111/cns.14209.

#### 通信作者: 冯莉

#### **Abstract**

Aims: Differentiating mesial temporal lobe epilepsy (MTLE) and neocortical temporal lobe epilepsy (NTLE) remains challenging. Our study characterized the metabolic profiles between MTLE and NTLE and their correlation with surgical prognosis using 18 F-FDG-PET. Methods: A total of 137 patients with intractable temporal lobe epilepsy (TLE) and 40 age-matched healthy controls were recruited. Patients were divided into the MTLE group (N = 91) and the NTLE group (N = 46). 18 F-FDG-PET was used to measure the metabolism of regional cerebra, which was analyzed using statistical parametric mapping. The volume of abnormal metabolism in cerebral regions and their relationship with surgical prognosis were calculated for each surgical patient. **Results:** The cerebral hypometabolism of MTLE was limited to the ipsilateral temporal and insular lobes (p < 0.001, uncorrected). The NTLE patients showed hypometabolism in the ipsilateral temporal, frontal, and parietal lobes (p < 0.001, uncorrected). The MTLE patients showed extensive hypermetabolism in cerebral regions (p < 0.001, uncorrected). Hypermetabolism in NTLE was limited to the contralateral temporal lobe and cerebellum, ipsilateral frontal lobe, occipital lobe, and bilateral thalamus (p < 0.001, uncorrected). Among patients who underwent resection of epileptic lesions, 51 (67.1%) patients in the MTLE group and 10 (43.5%) in the NTLE group achieved Engel class IA outcome (p = 0.041). The volumes of metabolic increase for the frontal lobe or thalamus in the MTLE group were larger in non-Engel class IA patients than Engel class IA patients (p < 0.05). Conclusions: The spatial metabolic profile discriminated NTLE from MTLE. Hypermetabolism of the thalamus and frontal lobe in MTLE may facilitate preoperative counseling and surgical planning.

编号: XEY-2023-1-6

引用格式: Wang Y, Kong H, Wang Y, Jin P, Ding J, Li H, Wang H, Zhuo Z. A Retrospective Study on Clinical Features of Childhood Moyamoya Disease. Pediatr Neurol. 2023;138:17-24. doi: 10.1016/j.pediatrneurol.2022.08.007.

通信作者: 禚志红

#### **Abstract**

**Background:** Childhood moyamoya disease (MMD) can lead to progressive and irreversible neurological impairment. Early age at onset is likely associated with a worst prognosis of the disease. The study aims to summarize the clinical characteristics of childhood MMD for supporting the diagnosis and treatment of early MMD. **Methods:** A retrospective study was conducted on children aged 0-16 years who were diagnosed with MMD in the department of Neurology and neurosurgery of our hospital from October 2016 to April 2020. The clinical characteristics of children with MMD were summarized for analysis, and the distribution of sex and initial attacks type among different age groups were determined by data comparison. Results: The study surveyed 114 children (the male to female sex ratio of 1:1.07) with MMD and 6.1% of them had family history. The mean age of onset was 7.15±3.30 years old and the peak age of onset was 5 to 8 years old. The most commonly initial attacks type was transient ischemic attack (TIA) (62 cases, 54.4%) with limb weakness. The incidence of the initial attacks type in the three age groups was varied (P < 0.05). The result of overall prognosis was good in 86 cases (89.6%). Conclusions: In this study, MMD cases were mainly ischemic type and TIA was the most commonly initial attacks type. Infant group was more prone to have cerebral infarction, while preschool and school-age groups tended to have TIA. The treatments and prognosis of the studied MMD cases were achieved with good outcomes.

编号: CYDQZ-2023-1-1

引用格式: Wu Y, Bayrak CS, Dong B, He S, Stenson PD, Cooper DN, Itan Y, Chen L. Identifying shared genetic factors underlying epilepsy and congenital heart disease in Europeans. Hum Genet. 2023;142(2):275-288. doi: 10.1007/s00439-022-02502-4.

# 通讯作者: 陈蕾

#### **Abstract**

Epilepsy (EP) and congenital heart disease (CHD) are two apparently unrelated diseases that nevertheless display substantial mutual comorbidity. Thus, while congenital heart defects are associated with an elevated risk of developing epilepsy, the incidence of epilepsy in CHD patients correlates with CHD severity. Although genetic determinants have been postulated to underlie the comorbidity of EP and CHD, the precise genetic etiology is unknown. We performed variant and gene association analyses on EP and CHD patients separately, using whole exomes of genetically identified Europeans from the UK Biobank and Mount Sinai BioMe Biobank. We prioritized biologically plausible candidate genes and investigated the enriched pathways and other identified comorbidities by biological proximity calculation, pathway analyses, and gene-level phenome-wide association studies. Our variant- and gene-level results point to the Voltage-Gated Calcium Channels (VGCC) pathway as being a unifying framework for EP and CHD comorbidity. Additionally, pathway-level analyses indicated that the functions of disease-associated genes partially overlap between the two disease entities. Finally, phenome-wide association analyses of prioritized candidate genes revealed that cerebral blood flow and ulcerative colitis constitute the two main traits associated with both EP and CHD.

编号:CYDQZ-2023-1-2

引用格式: Li X, Peng A, Li L, Chen L. Association between walking and square dancing-oriented leisure-time physical activity and cognitive function among middle-aged and elderly people in Southwest China. BMC Geriatr. 2023;23(1):28. doi: 10.1186/s12877-023-03737-0. PMID: 36646990.

通讯作者: 陈蕾

#### Abstract

**Background:** Southwest China is facing a serious aging problem across the country, but the status of cognitive function in middle-aged and elderly people in this region is superior to the national average. This study intends to reveal the leisure-time physical activity (LTPA) pattern in this region and explore whether this pattern is beneficial for cognitive function. Methods: The data came from the 2019-2021 baseline survey on cognitive function of a natural population cohort conducted by West China Hospital of Sichuan University. A structured questionnaire was used to investigate the LTPA status of the participants, and the Mini-Mental State Examination was used to evaluate their cognitive function. Then, we used multiple linear regression to analyze the association between LTPA and cognitive level, and further subgroup analysis was carried out according to sex, age and waist-to-hip ratio. Results: A total of 2697 participants were enrolled, with an average age of 66.19  $\pm$  6.68 years. The average cognitive function score was 27.23  $\pm$  2.72, of which 8.60% indicated mild cognitive impairment. Their median LTPA level was 24.50 MET-hours per week, of which 70.37% reached the activity level recommended by WHO, with the main types being walking (1340 cases, 49.68%), square dancing (270 cases, 10.01%), or walking + square dancing (172 cases, 6.38%). Multiple linear regression showed that cognitive function increased with the amount of LTPA from 11.25 MET-hours/week to 36.40 MET-hours/week ( \beta 0.09 for  $11.25 \sim 24.50$  MET-hours/week,  $\beta$  0.38 for  $24.50 \sim 36.40$  MET-hours/week) but stabilized at more ( $\beta$  0.39 for  $\geq$ 36.40 MET-hours/week). The positive association persisted even for those who only walked ( $\beta$  0.37 for 24.50 ~ 36.40 MET-hours/week,

P = 0.008). Conclusions: Middle-aged and elderly people in Southwest China hold a relatively high level of LTPA status, and walking and square dancing-oriented LTPA are positively correlated with cognitive function.

编号: CYDQZ-2023-1-3

引用格式: Cao X, Shen Z, Wang X, Zhao J, Liu W, Jiang G. A Meta-analysis of Randomized Controlled Trials Comparing the Efficacy and Safety of Pregabalin and Gabapentin in the Treatment of Postherpetic Neuralgia. Pain Ther. 2023;12(1):1-18. doi: 10.1007/s40122-022-00451-4.

# 通讯作者: 蒋国会

#### **Abstract**

Objective: To systematically evaluate the clinical efficacy of pregabalin and gabapentin in the treatment of postherpetic neuralgia (PHN), including the difference in pain control and occurrence of adverse reactions. Methods: PubMed, MEDLINE, EMBASE, Cochrane Library, and Web of Science databases were searched for randomized controlled trials (RCTs) comparing the efficacy of pregabalin and gabapentin in patients with PHN. Data from studies meeting the inclusion criteria were extracted and the Cochrane Risk of Bias risk assessment tool was used to evaluate the quality of the included studies. Revman 5.3 and Stata17 was used to perform the metaanalysis and to detect publication bias. Results: A total of 14 RCTs with 3545 patients were included in this study, including 926 in the pregabalin treatment group, 1256 in the gabapentin treatment group, and 1363 in the placebo control group. Pregabalin was better than gabapentin in alleviating pain and improving the global perception of change in pain and sleep (P\0.05). Gabapentin was associated with a lower incidence of adverse events than pregabalin (P\0.05). Funnel plot and Begg s and Egger s tests showed no significant publication bias. Conclusion: Pregabalin appears to have a better overall therapeutic effect than gabapentin for patients with PHN, but gabapentin has a lower incidence of adverse reactions and a better safety profile. Clinicians should comprehensively consider patient factors and fully evaluate the advantages and disadvantages of each treatment option to select the most suitable drugs for patient use. Considering the limited quantity and quality of the existing literature, high-quality RCTs are need to confirm the advantages of pregabalin over gabapentin in the treatment of PHN and guide clinical decision-making.

编号: CYDQZ-2023-1-4

引用格式: Shen Z, Pu S, Cao X, Tang M, Wang S, Bai D, Jiang G. Bioinformatics and network pharmacology analysis of drug targets and mechanisms related to the comorbidity of epilepsy and migraine. Epilepsy Res. 2023;189:107066. doi: 10.1016/j.eplepsyres.2022.107066.

# 通讯作者: 蒋国会

#### **Abstract**

Objective: The present study aimed to explore the mechanisms underlying the comorbidity of epilepsy and migraine, identify potential common targets for drug intervention, and provide insight into new avenues for disease prevention and treatment using an integrated bioinformatic and network pharmacology approach. Methods: Disease targets in epilepsy and migraine were screened using the DisGeNET database to identify intersecting gene targets. Gene Ontology (GO) and Kyoto Encyclopedia of Genes and Genomes (KEEG) enrichment analyses were then performed using the WebGestalt database. Furthermore, the STRING database was used to construct a protein-protein interaction (PPI) network, and Cytoscape software was used to analyze the protein molecular signals at the intersection of epilepsy and migraine. The Drugbank database was used to identify common targets for antiepileptic drugs in epilepsy and migraine to further analyze the disease-gene-target-drug interaction network. Finally, molecular docking simulations were performed to verify the hypothesis that migraine and epilepsy share common diseases and drug targets. Results: A total of 178 common targets for epilepsy and migraine were identified using the

DisGeNET database, and the 24 genes most related to the disease were screened using the Score\_gda gene scoring system. GO enrichment analysis indicated that common targets were mainly enriched in biological processes and molecular functions, including membrane potential regulation, inorganic ion transmembrane transport, axonal signaling, and ion channel activity. KEGG pathway enrichment analysis indicated that the mechanism of action might be related to neuroactive ligand receptors, AGE-RAGE, cAMP, and VEGF signaling pathways. The PPI network construction and analysis results show that the PPI grid had 23 central nodes and 24 connected edges, with an average node degree of 2.09 and an average clustering coefficient of 0.384. The 10 genes with potentially important roles in epilepsy and migraine were CACNA1A, KCNQ2, KCNA1, SCN1A, PRRT2, SCN8A, KCNQ3, SCN2A, GRIN2A, and GABRG2. Drugbank database results indicated that antiepileptic drugs, including lamotrigine, topiramate, valproic acid, carbamazepine, gabapentin, and perampanel, also had common targets with migraine. The three most important targets exhibited strong binding affinity with drugs in the molecular docking simulations. Conclusion: Our systematic and comprehensive analyses of disease-gene-target-drug interaction networks identified several biological processes and molecular functions common to migraine and epilepsy, most of which were related to neuroactive ligand-receptor interactions. These data provide a new theoretical basis and reference for the clinical treatment of comorbid epilepsy and migraine and may aid in the development of novel pharmacological strategies.

编号: CYDQZ-2023-1-5

引用格式: Wang S, He X, Bao N, Chen M, Ding X. Zhang M, Zhao L, Wang S, Jiang G. Potentials of mir-9-5p in promoting epileptic seizure and improving survival of glioma patients. Acta Epileptologica. 2022; 4, 33. doi.org/10.1186/s42494-022-00097-x

通讯作者: 蒋国会

青年委员会 2023 年 1-3 月部分发表论文展示

Abstract

MicroRNAs (miRNAs) are a class of non-coding small RNAs with about 22

nucleotides in eukaryotes. They regulate gene expression at the post-transcriptional

level and play a key role in physiological and pathological processes. As one of the

most abundant miRNAs in the human brain, miRNA-9 (miR-9) has attracted extensive

attention due to its important role in the maintenance of normal function of the nervous

system and the occurrence and development of nervous system diseases. Hence, we

reviewed the neuroprotective effect of miR-9 in neurological diseases. MiR-9 may be

a potential target of nervous system diseases.

编号: CYDQZ-2023-1-6

引用格式: Wang S, Jiang G, Wang S. Neuroprotective Role of MiRNA-9 in

Neurological Diseases: A Mini Review. Curr Mol Med. 2023;23. doi:

10.2174/1566524023666221025123132.

通讯作者: 蒋国会

**Abstract** 

MicroRNAs (miRNAs) are a class of non-coding small RNAs with about 22

nucleotides in eukaryotes. It regulates gene expression at the post-transcriptional level

and plays a key role in physiological and pathological processes. As one of the most

abundant miRNAs in the human brain, miRNA-9 (miR-9) has attracted extensive

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system and the occurrence and development of nervous system diseases. Hence, we

reviewed the neuroprotective effect of miR-9 in neurological diseases. MiR-9 may be

a potential target of nervous system diseases.

编号: CYDQZ-2023-1-7

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引用格式: Tian MQ, Li RK, Yang F, Shu XM, Li J, Chen J, Peng LY, Yu XH, Yang CJ. Phenotypic expansion of KCNH1-associated disorders to include isolated epilepsy and its associations with genotypes and molecular sub-regional locations. CNS Neurosci Ther. 2023;29(1):270-281. doi: 10.1111/cns.14001.

## 第一作者及通讯作者: 田茂强

#### **Abstract**

**Purpose:** Genotype-phenotypic correlation of *KCNH1* variant remains elusive. This study aimed to expand the phenotypic spectrum of *KCNH1* and explore the correlations between epilepsy and molecular sub-regional locations. **Methods:** We performed whole-exome sequencing in a cohort of 98 patients with familiar febrile seizure (FS) or epilepsy with unexplained etiologies. The damaging effects of variants was predicted by protein modeling and multiple in silico tools. All reported patients with *KCNH1* pathogenic variants with detailed neurological phenotypes were analyzed to evaluate the genotype-phenotype correlation.

Results: Two novel *KCNH1* variants were identified in three cases, including two patients with FS with inherited variant (p.Ile113Thr) and one boy with epilepsy with de novo variant (p.Arg357Trp). Variant Ile113Thr was located within the eag domain, and variant p.Arg357Trp was located in transmembrane domain 4 of KCNH1, respectively. Two patients experienced refractory status epilepticus (SE), of which one patient died of acute encephalopathy induced by SE. Further analysis of 30 variants in 51 patients demonstrated that de novo variants associated with epileptic encephalopathy, while mosaic/somatic or germline variants cause isolated epilepsy/FS. All hotspot variants associated with epileptic encephalopathy clustered in transmembrane domain (S4 and S6), while those with isolated epilepsy/seizures or TBS/ZLS without epilepsy were scattered in the *KCNH1*. Conclusions: We found two novel missense variants of *KCNH1* in three individuals with isolated FS/epilepsy. Variants in the *KCNH1* cause a spectrum of epileptic disorders ranging from a benign form of genetic isolated epilepsy/FS to intractable form of epileptic encephalopathy. The genotypes and variant locations help explaining the phenotypic variation of patients with *KCNH1* variant.

编号: CYDQZ-2023-1-8

引用格式: Tian MQ, Liu XR, Lin SM, Wang J, Luo S, Gao LD, Chen XB, Liang XY, Liu ZG, He N, Yi YH, Liao WP; China Epilepsy Gene 1.0 Project. Variants in BRWD3 associated with X-linked partial epilepsy without intellectual disability. CNS Neurosci Ther. 2023;29(2):727-735. doi: 10.1111/cns.14057.

第一作者: 田茂强

#### **Abstract**

Aims: Etiology of the majority patients with idiopathic partial epilepsy (IPE) remains elusive. We thus screened the potential disease-associated variants in the patients with IPE. Methods: Trios-based whole exome sequencing was performed in a cohort of 320 patients with IPE. Frequency and molecular effects of variants were predicted. Results: Three novel BRWD3 variants were identified in five unrelated cases with IPE, which were four male cases and one female case. The variants included two recurrent missense variants (c.836C>T/p.Thr279Ile and c.4234A>C/p.Ile1412Leu) and one intronic variant close to splice site (c.2475+6A>G). The two missense variants were located in WD40 repeat domain and bromodomain, respectively. They were predicted to be damaging by silico tools and change hydrogen bonds with surrounding amino acids. The frequency of mutant alleles in this cohort was significantly higher than that in the controls of East Asian and all population of gnomAD. All these variants were inherited from the asymptomatic mothers. Four male cases presented frequent seizures at onset, while the female case only had two fever-triggered seizures. They showed good responses to valproate and lamotrigine, then finally became seizure free. All the cases had no intellectual disability. Further analysis demonstrated that all previously reported destructive variants of BRWD3 caused intellectual disability, while missense variants located in WD40 repeat domains and bromodomains of BRWD3 were associated with epilepsy. Conclusion: BRWD3 gene is potentially associated with X-linked partial epilepsy without intellectual disability. The genotypes and locations of BRWD3 variants may explain for their phenotypic variation.

编号: CYDQZ-2023-1-9

引用格式: Zheng Q, Ma P, Feng Z. Safety of COVID-19 vaccine in patients with epilepsy: a meta-analysis. Neurol Sci. 2023;44(1):13-17. doi: 10.1007/s10072-022-06453-6.

通讯作者: 冯占辉

#### **Abstract**

Coronavirus Disease 19 (COVID-19), caused by severe acute respiratory syndrome-coronavirus-2 (SARS-CoV-2), has been associated with greater than two million deaths worldwide. The safety of the COVID-19 vaccine for patients with epilepsy has been examined. Many observational trials of COVID-19 vaccine in patients with epilepsy have been analyzed using a systematic search of common databases between January 1, 2020, and August 20, 2022. Our research showed that a small percentage of vaccinated patients with epilepsy had a transient short-term increase of seizure frequency. The current COVID-19 vaccine for patients with epilepsy was safe. We found that the mRNA vaccine was relatively safer than the vector-based vaccine for patients with epilepsy. Future studies should determine adverse events of each vaccine type and critically identify mechanisms of severe adverse events following vaccination.

# 人生万事须自为,跬步江山即寥廓

——《王氏能远楼》

# 后生可畏,焉知来者之不如今也

----《论语》

编号: SSL-2023-1-1

引用格式: Chen ZP, Wang SJ, Zhao XS, Fang W, Wang ZG, Ye HJ, Wang MJ, Ke L, Huang TF, Lv P, Jiang XH, Zhang QP, Li L, Xie ST, Zhu JN, Hang CH, Chen DJ, Liu XY, Yan C. Lipid-accumulated reactive astrocytes promote disease progression in epilepsy. Nature Neuroscience, 2023;26:542–554. DOI https://doi.org/10.1038/s41593-023-01288-6.

通讯作者: 刘翔宇

#### **Abstract**

Reactive astrocytes play an important role in neurological diseases, but their molecular and functional phenotypes in epilepsy are unclear. Here, we show that in patients with temporal lobe epilepsy (TLE) and mouse models of epilepsy, excessive lipid accumulation in astrocytes leads to the formation of lipid-accumulated reactive astrocytes (LARAs), a new reactive astrocyte subtype characterized by elevated APOE expression. Genetic knockout of APOE inhibited LARA formation and seizure activities in epileptic mice. Single-nucleus RNA sequencing in TLE patients confirmed the existence of a LARA subpopulation with a distinct molecular signature. Functional studies in epilepsy mouse models and human brain slices showed that LARAs promote neuronal hyperactivity and disease progression. Targeting LARAs by intervention with lipid transport and metabolism could thus provide new therapeutic options for drug-resistant TLE.

编号: SSL-2023-1-2

引用格式: Zhang H, Deng J, Wang X, Chen C, Chen S, Dai L, Fang F. Clinical phenotypic and genotypic characterization of NPRL3-related epilepsy. Front Neurol. 2023; 14:1113747. doi: 10.3389/fneur.2023.1113747.

第一作者 张洪伟

#### Abstract

Background: As one of the assembly factors of the GATOR1 protein complex in the mechanism of rapamycin pathway, NPRL3 plays an important role in the pathogenesis of epilepsy. However, the correlation between genotype and clinical phenotype in patients with NPRL3-related epilepsy has not been clarified. Methods: A total of 11 Chinese children with NPRL3-related epilepsy were identified through whole-exome sequencing (WES). The data from the clinical presentation, laboratory data, brain imaging findings, genetic results, and treatment methods were collected. All previously reported cases with NPRL3-related epilepsy were collected and reviewed through PubMed search. Results: Among the 11 children, eight have not been reported, and two of them presented infantile spasms (ISs) as a new phenotype of NPRL3-related epilepsy. In addition, WES identified five frameshift mutations, three nonsense mutations, two missense mutations, and one exon deletion. Based on bioinformatics analysis, it was found that two missense mutation sites were highly conserved, and the c.400G>A mutation site of the NPRL3 gene caused the alteration of the protein structure. To date, 88 patients have been reported with NPRL3-related defects, including our 11 cases. The most common presentations were sleep-related hypermotor epilepsy (SHE), frontal lobe epilepsy (FLE), and temporal lobe epilepsy. A majority of patients (70%) presented normal neuroimaging results, and focal cortical dysplasia was the most common neuroimaging abnormality (62.5%). Among the NPRL3 gene mutations, loss of function (nonsense mutations, frameshift mutations, and exons deletion) was the most common genetic variation (75%). For 73% of patients with NPRL3-related epilepsy, monotherapy of sodium channel blockers was effective. Surgery was effective for 75% of children with neuroimaging abnormalities. Two cases unresponsive to surgery or anti-seizure medications were treated with ketogenic diets (KD), which were effective. One case was treated with rapamycin at an early stage of epilepsy, which was effective as well. Conclusion: NPRL3-related epilepsy has high clinical and genetic heterogeneity. SHE and FLE are the most common clinical presentations. Furthermore, ISs are the new phenotypes of NPRL3-related epilepsy, while the variants c.275G>A, c.745G>A, and c.1270C>T may be the most common NPRL3 gene mutations. Sodium

channel blockers, surgery, KD, and rapamycin may be the potential treatments for these patients. Our study expanded the clinical and genetic spectrum of NPRL3-related epilepsy and provided important information for the precise treatment of patients.

编号: SSL-2023-1-3

引用格式: 张洪伟, 张童, 金瑞峰, 高在芬, 刘勇. 岛叶癫痫的研究进展. 癫痫杂

志, 2023, 9(1): 43-47. doi: 10.7507/2096-0247.202210002

## 第一作者 张洪伟

## 摘要

随着立体定向脑电图(Stereoelectroencephalography,SEEG)技术的广泛使用,岛叶癫痫逐渐被认识,其症状学也逐渐被描述清楚。在岛叶癫痫的研究中,主观症状(如上腹部感觉、听觉和躯体感觉等)对于识别岛叶癫痫至关重要,客观的运动成分也比较突出。岛叶癫痫症状学的多样性与其特殊的皮层构筑特点、广泛的功能连接和致痫网络有关。由于岛叶特殊的位置关系,切除手术有一定的风险,SEEG 引导下立体定向脑电图引导下射频热凝毁(Radio frequency thermocoagulation,RF-TC)、激光间质热疗法(MRI-guided laser interstitial thermal therapy,LITT)具有接近于切除性手术的有效性,同时具有更高的安全性。这种精准、微创的治疗方法有可能会部分取代传统的切除性外科手术,使更多的癫痫患者获益,但目前来说该技术仍然只是一种尝试。

编号: SSL-2023-1-4

引用格式: 张洪伟, 耿贵富, 高在芬, 陈传美, 孟尧, 胡万冬, 张欢, 史建国, 刘勇. 儿童岛叶癫痫的特点与立体定向脑电图引导下射频热凝毁损的有效性及安全性分析. 癫痫杂志, 2023, 9(2): 91-95. doi: 10.7507/2096-0247.202211009

## 第一作者 张洪伟

## 摘要

的 探讨儿童岛叶癫痫的特点和立体定向脑电图(Stereotacticelectroencephalogram, SEEG) 引导射频热凝毁损技术 (Radiofrequency thermocoagulation, RF-TC) 在儿童药物难治性岛叶癫痫治疗中的有效性和安全 性。方法 回顾性分析2021年1月—2022年5月山东大学附属儿童医院癫痫中心收 治的7例经SEEG证实的岛叶癫痫患儿的临床资料,其中男3例、女4例,平均年龄 (6.6±3.5) 岁。所有患儿均接受I期无创术前评估,并SEEG电极置入并进行视频 脑电监测。根据SEEG结果和影像结果确定毁损触点,利用电极触点进行热凝毁 损。分别于术后3、6、12、18个月对患儿进行门诊复查或电话随访。采用 Engel 分级标准评估临床疗效,同时记录并发症情况。结果 6例(6/7)以夜间癫痫发 作为主,4例(4/7)表现为过度运动或复杂运动发作;3例(3/7)表现为局灶性 强直发作:仅1例患儿有先兆。7例患儿头皮脑电图间期均提示与手术侧别一致: 6例为围外侧裂区分布,1例局限于颞区。4例磁共振成像阴性,2例灰白质交界不 清;1例岛叶皮质增厚。7例患儿完成了电极置入,均完成半年以上随访,随访时 间为6.0~22.0(12.3±5.3)个月。至末次随访,7例患儿中,5例癫痫发作完全消 失(Engel la 级),2例术后仍有发作,术后未出现远期并发症。**结论** 儿童岛叶 癫痫先兆少见,运动症状突出,头皮脑电图以围侧裂区分布为主,SEEG引导下 RF-TC治疗儿童药物难治性岛叶癫痫的安全性和有效性均较好。

编号: SSL-2023-1-5

引用格式: 陆叶婷,来峰,李秉宜,朱翔毅,潘圣洁,徐敏,方琪,胡小伟. 棘波自动检测在颞叶癫痫患者长程脑电图中应用价值的初步研究. 癫痫杂志; 2023:9(1): 5-10. doi: 10.7507/2096-0247,202209011

通讯作者: 胡小伟

摘要

目的 探究 Persyst 自动检测技术在成人颞叶癫痫棘波自动检测中的应用价值。方法 连续选定苏州大学附属第一医院癫痫单元 2019 年 1 月 1 日—2019 年 12 月 31 日期间脑电图数据库中符合纳排标准的患者脑电图记录,由两名高年资、由中国抗癫痫协会认证脑电图技术专家阅读和标记长程脑电图中的发作间期癫痫样放电,将两名专家一致的结果作为棘波判定的"金标准",与 Persyst11、13、14 三个版本在 0.5~ 0.9 之间不同感知值下的棘波自动检测结果进行回顾性研究,计算敏感性、假阳性率、减少的工作量。 结果 共纳入 7 例颞叶癫痫患者,每例记录时间为 24~ 25 h,总记录时间为 169 h。两名阅图者总体一致率为 43.09%。自动检测棘波敏感性最高出现于感知值 0.5 时,分别为 62.26%、77.0% 和 67.28%,假阳性率最低出现于感知值为 0.9 时,分别为 62.26%、77.0% 和 67.28%,假阳性率最低出现于感知值为 0.9 时,分别为 0.37 个/min、0.85 个/min、0.46 个/min。借助自动分析最多可减少 14.59%~ 37.05% 的阅图量。 结论 Persyst 自动检测棘波的敏感性较高,假阳性也较高,不同版本间存在区别,与人工阅图进一步结合,减少一定工作量。通过合理的调整 Perception值可以既减少工作量又保证一定准确性。

# 为者常成,行者常至

——《晏子春秋》

看似寻常最奇崛,成如容易却艰辛。

——《题张司业诗》

编号: YGQM-2023-1-1

引用格式: Lin ZJ, Li B, Lin PX, Song W, Yan LM, Meng H, He N. Clinical

application of trio-based whole-exome sequencing in idiopathic generalized

epilepsy. Seizure. 2023:S1059-1311(23)00050-X. doi:

10.1016/j.seizure.2023.02.011.

通讯作者: 孟珩 何娜

Abstract

Purpose: Idiopathic generalized epilepsies (IGEs) are a common group of genetic

generalized epilepsies with high genetic heterogeneity and complex inheritance.

However, the genetic basis is still largely unknown. This study aimed to explore the

genetic etiologies in IGEs.

Methods: Trio-based whole-exome sequencing was performed in 60 cases with IGEs.

The pathogenicity of candidate genetic variants was evaluated by the criteria of the

American College of Medical Genetics and Genomics (ACMG), and the clinical

causality was assessed by concordance between the observed phenotype and the

reported phenotype.

Significance: Considering the genetic heterogeneity and complex inheritance of IGEs,

a comprehensive evaluation combined the ACMG scoring and assessment of clinical

concordance is suggested for the pathogenicity analysis of variants identified in clinical

screening. GABRB1 is probably a novel causative gene for IGE, which warrants further

studies.

编号: YGOM-2023-1-2

引用格式: Zou DF, Li XY, Lu XG, Wang HL, Song W, Zhang MW, Liu XR, Li

BM, Liao JX, Zhong JM, Meng H, Li B. Association of FAT1 with focal epilepsy

and correlation between seizure relapse and gene expression stage. Seizure.

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2023:S1059-1311(23)00063-8. doi: 10.1016/j.seizure.2023.03.003.

## 通讯作者: 孟珩

#### **Abstract**

Purpose: The FAT1 gene encodes FAT atypical cadherin 1, which is essential for foetal development, including brain development. This study aimed to investigate the relationship between FAT1 variants and epilepsy. Methods: Trio-based whole-exome sequencing was performed on a cohort of 313 patients with epilepsy. Additional cases with FAT1 variants were collected from the China Epilepsy Gene V.1.0 Matching Platform. Results: Four pairs of compound heterozygous missense FAT1 variants were identified in four unrelated patients with partial (focal) epilepsy and/or febrile seizures, but without intellectual disability/developmental abnormalities. These variants presented no/very low frequencies in the gnomAD database, and the aggregate frequencies in this cohort were significantly higher than those in controls. Two additional compound heterozygous missense variants were identified in two unrelated cases using the gene-matching platform. All patients experienced infrequent (yearly/monthly) complex partial seizures or secondary generalised tonic-clonic seizures. They responded well toantiseizure medication, but seizures relapsed in three cases when antiseizure medication were decreased or withdrawn after being seizurefree for three to six years, which correlated with the expression stage of FAT1. Genotype-phenotype analysis showed that epilepsy-associated FAT1 variants were missense, whereas non-epilepsy-associated variants were mainly truncated. The relationship between FAT1 and epilepsy was evaluated to be "Strong" by the Clinical Validity Framework of ClinGen. Conclusions: FAT1 is a potential causative gene of partial epilepsy and febrile seizures. Gene expression stage was suggested to be one of the considerations in determining the duration of antiseizure medication. Genotypephenotype correlation helps to explain the mechanisms underlying phenotypic variation..

编号: YGQM-2023-1-3

引用格式: Yang JH, Liu ZG, Liu CL, Zhang MR, Jia YL, Zhai QX, He MF, He N, Qiao JD. MED12 variants associated with X-linked recessive partial epilepsy without intellectual disability. Seizure. 2023:S1059-1311(23)00057-2. doi: 10.1016/j.seizure.2023.02.018.2.

#### 通讯作者: 何娜

#### Abstract

Objectives: The MED12 gene encodes mediator complex subunit 12, which is a component of the mediator complex involved in the transcriptional regulation of nearly all RNA polymerase II-dependent genes. MED12 variants have previously been associated with developmental disorders with or without nonspecific intellectual disability. This study aims to explore the association between MED12 variants and epilepsy. Materials and methods: Trios-based whole-exome sequencing was performed in a cohort of 349 unrelated cases with partial (focal) epilepsy without acquired causes. The genotype-phenotype correlations of MED12 variants were analyzed. Results: Five hemizygous missense MED12 variants, including c.958A>G/p.Ile320Val, c.1757G>A/p.Ser586Asn, c.2138C>T/p.Pro713Leu, c.3379T>C/p.Ser1127Pro, and c.4219A>C/p.Met1407Leu were identified in five unrelated males with partial epilepsy. All patients showed infrequent focal seizures and achieved seizure free without developmental abnormalities or intellectual disability. All the hemizygous variants were inherited from asymptomatic mothers (consistent with the X-linked recessive inheritance pattern) and were absent in the general population. The two variants with damaging hydrogen bonds were associated with early-onset seizures. Further genotype-phenotype analysis revealed that congenital anomaly disorder (Hardikar syndrome) was associated with (de novo) destructive variants in an X-linked dominant inheritance pattern, whereas epilepsy was associated with missense variants in an X-linked recessive inheritance pattern. Phenotypic features of intellectual disability appeared as the intermediate phenotype in terms of both genotype and inheritance. Epilepsy-related variants were located at the MED12-LCEWAV domain and the regions between MED12-LCEWAV and MED12-POL. **Conclusion:** MED12 is a potentially causative gene for X-linked recessive partial epilepsy without developmental or intellectual abnormalities. The genotype-phenotype correlation of MED12 variants explains the phenotypic variations and can help the genetic diagnosis.

编号: YGQM-2023-1-4

引用格式: Liu J, Zhang P, Zou Q, Liang J, Chen Y, Cai Y, Li S, Li J, Su J, Li Q. Status of epilepsy in the tropics: An overlooked perspective. Epilepsia Open. 2023;8(1):32-45. doi: 10.1002/epi4.12686.

通讯作者: 李其富

#### **Abstract**

Epilepsy is one of the most common serious chronic neurological diseases affecting people of all ages globally. It is characterized by recurrent seizures. About 50 million people worldwide have epilepsy. Indubitably, people with epilepsy (PWE) may be without access to appropriate treatment. Many studies have examined the molecular mechanisms and clinical aspects of epilepsy; nonetheless, the treatment gap exists in some special areas. In the tropics, the specific geographical and ecological conditions and a lack of medical resources result in neglect or delay of diagnosis for PWE. Herein, we summarized the epidemiology of epilepsy in the tropics and discussed the disease burden and existing problems, aiming to offer a medical environment for patients in need and highlight the importance of reducing the epileptic disease burden in tropical countries.

编号: YGQM-2023-1-5

引用格式: Wan H, Chen H, Zhang M, Feng T, Wang Y. Cerebral microbleeds is associated with dementia in Parkinson's disease. Acta Neurol Belg.

2023;123(2):407-413. doi: 10.1007/s13760-022-01918-z.

#### 第一作者 万慧娟

#### **Abstract**

**Introduction:** Emerging evidence suggests that cerebral small vessel disease (CSVD) may worsen cognitive functions in Parkinson's disease (PD). However, the efect of microbleeds on cognitive function in patients with PD remains unknown. This study explored the association between the presence, number and location of microbleeds with dementia in PD patients. Methods: This cross-sectional study included 431 patients with PD from Beijing Tiantan Hospital from May 2016 to August 2019. Cognition assessments (MMSE, MoCA) were performed for these patients. MRI imaging sequences were obtained and reviewed independently by two well-trained readers who were blind to all clinical data. Spearman's correlation analysis and logistic regression model analysis were further used for the assessments. Results: An association between cerebral microbleeds with cognitive ability and dementia in PD patients was revealed. A significance was observed between the total number of microbleeds and two widely used scores of cognitive assessments (Spearman R=-0.120 to MMSE with a p=0.016, and -0.117 to MoCA with a p=0.020). In detail, infratentorial microbleeds were associated with the level of cognition in PD patients (Spearman R=-0.099 to MMSE with a p=0.049, and -0.116 to MoCA with a p=0.021). Furthermore, logistic regression analysis results also confirmed such correlations between the number of microbleeds and cognitive ability after adjusting for age, cholesterol level, Hamilton Anxiety Scale, Hamilton Depression Scale, and white matter hyperintensity Fazekas score (OR 3.28, p=0.035, 95% CI 1.090-9.892). **Conclusions:** The occurrence of microbleeds, especially in the infratentorial locations, may worsen the cognitive function of PD patients and result in dementia. Management of cerebral vascular disease could be benefcial to patients with PD.

# 欢迎大家引用上述论文

# 领读学术

领读学术作为青委会的传统学术活动,一直得到了葛兰素公司的大力支持, 2023 年我们将继续开展了 1+8 场活动,其中第一场全国性活动于 2023 年 2 月 26 日胜利召开。



首场活动由梁树立青委担任主席。彭镜教授结合当前人才评价体系变化讲述了高质量论文的撰写和 SCI 收录论文的发表经验,系统的介绍了前期的研究基础积累过程。陈子怡青委以党的二十大对青年人才的要求等出发,系统讲解了临床资料的有效收集和临床研究设计。刘永红青委介绍了"拉莫三嗪—妇性癫痫治疗的甄选"。吴洵昳、陈蕾、周健、梁建民、王爽教授结合自身经验进行了讨论和点评,三位专家讲课,非常生动和有可比性,对大家感触颇深。超 800 人加入直播间,人员覆盖 28 个省市自治区的 40 多家医院。





# 青年委员会多中心项目展示

# 《自身免疫性脑炎相关精神症状》多中心临床研究

# 刘永红 张歆博 空军军医大学西京医院神经内科

**摘要:** 神经精神症状是自身免疫性脑炎(autoimmune encephalitis, AE)急性期(初始症状或非首发症状)最常见的症状之一,先前的研究报道,约 72%-95%的 AE 患者有精神病学临床表现,一些患者有长期孤立的精神病学表现。特征包括激动和攻击性、听觉和/或视觉幻觉、偏执和妄想。但是,不同的精神症状在不同类型 AE 中的好发特点、诊疗期间治疗方案、急性发作时处理方法尚不清楚。

**目的:** 通过对"自身免疫性脑炎相关精神症状"的多中心观察性随访研究,总结不同抗体类型脑炎的精神症状转归特点;通过精细的精神行为异常分类,以及是否合并其他神经系统症状,辅助我们更准确识别 AE 及其亚型。研究病程中精神类药物、ASMs 使用情况(包括精神症状急性发作、癫痫持续状态的紧急处理),探寻发作性症状的有效处理方式。

意义:通过本研究结果形成"早期评估病情发生发展、选择优效治疗方案"的成果;精确抗体筛选可为患者减少多类型抗体检测的经济负担,同时也使更多的"疑似"AE 患者进行抗体筛选、早期治疗、防止遗漏。

可行性:近年来,本单位逐步搭建自免脑病例库,目前已有十余家单位参与, 完成 300 多例患者资料录入。因此,我们希望能够与其他单位共同合作、数据共 享,继续完善自免脑多中心数据库建立,为自免脑相关研究建立大样本基础。

方法: 多中心观察性随访研究。收集 AE 患者出现精神症状、癫痫发作、睡眠障碍等相关数据,并对所有患者的其他临床数据进行收集,如发病年龄、起病症状、辅助检查(MRI、EEG、评估量表等)、AE 类型、用药方案等并进行随访。

(具体内容可联系负责人获取 Excel 表格)

合作单位要求: 具有自免脑诊治能力的医疗单位。

招募患者要求: 符合"确定的 AE"诊断标准(发现明确的阳性抗体)。

负责人及联系方式: 刘永红 13991236602; 张歆博 18069824332

# SUDEP/Near-SUDEP 的多中心临床研究

刘永红 王则直 空军军医大学西京医院神经内科

**摘要:** 癫痫性猝死(sudden unexpected death in epilepsy,SUDEP)是癫痫患 者突然、意外、有或无目击者、非创伤性和非溺水死亡,有或没有癫痫发作证据, 不包括记录的癫痫持续状态,尸检没有发现毒理学或解剖学死亡原因。SUDEP事 件占癫痫患者所有死亡人数的 18%, 也是继中风之后的神经系统疾病的第二大死 亡原因。越来越多的学者关注这一灾难性事件,给家庭和社会带来沉重的负担。 因此,尽早明确 SUDEP 的发病机制和危险因素,并提出预防策略和干预措施, 最终减少 SUDEP 的发生。根据 Nashef (2012) 关于 SUDEP 的新定义及分类标 准, Near-SUDEP 是指癫痫患者不存在明确原因下出现心跳、呼吸骤停后复苏存 活超过 1h。视频脑电监测在国内广泛开展,上述情况在许多脑电监测中心均有 发生。而根据之前国内研究显示,我国 SUDEP 的发生率为 2.34 1000 人/每年。 通过相关文献检索,国内鲜有开展 SUDEP 的临床研究。因此,我们计划回顾性 分析脑电监测中心既往数据,收集监测期间发生的 SUDEP/Near-SUDEP 的临床 电生理演变过程,对围癫痫发作期脑电图、心电图及呼吸改变的研究,从而为揭 示 SUDEP/Near-SUDEP 的发生机制提供依据,并分析其危险因素,如癫痫起病 年龄、性别、癫痫发作类型、基因检测结果、功能核磁共振等,为减少癫痫性猝 死提供预防基础。

合作单位标准: 国内多家脑电监测中心

Near-SUDEP 招募患者要求: (1) 患者符合 2014 年国际抗癫痫联盟(ILAE)癫痫的诊断标准; (2) 任何形式的癫痫发作,发作期或发作后出现的心律失常 (房颤、室颤、心脏停搏等),给予心肺复苏或心肺功能自行恢复且存活大于 1 小时; (3) 任何形式的癫痫发作,发作期或发作后出现的呼吸抑制(指脉氧下降、胸廓运动消失),给予心肺复苏或心肺功能自行恢复且存活大于 1 小时; (4) 任何形式的癫痫发作,发作后发生 PGES,期间心肺功能障碍(PGES 是指癫痫发作后 30s 内出现的全脑脑波波幅小于或等于 10μV,且持续时间大于 2s),给予心肺复苏或心肺功能自行恢复且存活大于 1 小时。

**SUDEP 招募患者要求:** (1) 明确诊断为癫痫; (2) 排除溺水、创伤或癫痫持续状态或其他已知原因导致的死亡; (3) 死亡时有或无癫痫发作证据; (4) 死亡发生在正常的活动和环境中(如床上、床边、家里、工作); (5) 尸检或医学未发现明确死因。

**排除标准:** 突发心肺功能抑制是由于已经确定的心脏疾病(如心律失常/心肌梗死等)、肺部疾病本身所致。

负责人及联系方式: 刘永红 13991236602

# 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 <sub>季涛云</sub>

## 北京大学第一医院儿童癫痫中心

癫痫手术是治疗药物难治性癫痫的有效方法。术后癫痫无发作率 50%~80%。但术后无发作患者的抗癫痫发作药物(anti-seizure medications,ASM)的管理仍然是目前最困难、未解决的治疗难题。目前尚无系统的前瞻性随机对照研究。国内外也缺乏关于术后减药、停药的标准化指南。Andermann 等人建议不要在术后一年内更换 ASM,并考虑在手术后两到三年停药。Kuzniecky 等将接受手术治疗颞叶癫痫病人(共 40 例)分为两组,一组术后 4~6 个月内继续维持术前多种 ASM治疗,另一组术后立即将手术前的多种 ASM 转换为单药(卡马西平)治疗。评估术后第一年内各方案的疗效和安全性,在癫痫发作复发率、复发类型和复发时间方面,两组间没有显着差异。在加拿大癫痫病学家中进行的一项调查显示,约50%的医生术后一年不调整 ASM,10%的医生会至少两年无发作才开始减量ASM。目前仍然缺少科学、合理的研究指导术后无发作患者的 ASM 的管理。考虑到长期应用 ASM 不良反应、ASM 对认知副作用、经济成本、长期口服 ASM的心理负担等,癫痫术后无癫痫发作的患者是否可以较早减/停 ASM 是目前亟须解决的问题。

本中心于 2022 年牵头开展了此项国内多中心研究,纳入癫痫术后无发作的 患者开展前瞻性随机对照研究,以建立科学的、有依据术后 ASM 减/停时间节点,并确定与 ASM 减/停时间节点相关的癫痫发作预后影响因素。目前已纳入患者 48 例。

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# 一项前瞻性、多中心研究:中链脂肪酸在成年难治性癫痫及共患病添加治疗的疗效、安全性及机制探讨

#### 林婉挥

#### 福建医科大学附属协和医院神经内科

**背景及目的:**最近的数据表明,添加中链甘油三酯(MCT)可提供中链脂肪酸辛酸和癸酸以及酮体作为辅助脑能量通过多种途径对啮齿动物癫痫发作模型以及人类癫痫有益。目前迫切需要在成年难治性癫痫及共患病进行更大规模的临床试验,以找到 MCT 的理想成分和剂量以及反应最佳的癫痫类型。

方法: 前瞻性、多中心、开放性设计,入组周期2年,随访期至少半年,符合纳入和排除标准。

MCT 方案设计: (1) 第一阶段 (第 1-4 周: MCT 适应期): 以增加三餐点心的形式,每餐 1 勺为起点 (9g),同时辅助牛奶 (不能耐受者舒化奶 100ml 左右),每周每餐增加 1 勺,4 周后的目标剂量。(2) 第二阶段(第 5-12 周:MAD 适应期): 改变饮食结构,逐渐减少碳水化合物这类主食。(3) 第三阶段 (第 13-24 周: 维持期):耐受者采用 MCT+MAD;不能耐受者仅采用 MCT; (4) 第四阶段 (6 个月-2 年: 拓展期): 有效果愿意继续采用生酮饮食治疗的继续维持至 2 年。

观察的指标: 1.发作情况 2.脑电情况 3.共患病情况 4.不良反应 5.血酮、尿酮、血脂、血糖、肝功能。依托生酮饮食管理平台。

**机制的探讨:**检测生酮饮食前后(至少半年)血液中脂肪酸代谢质谱、酮池的改变。

**患者标准:** 1.在签署知情同意书时,必须为年满 14-50 周岁(含)的男女受试者; 2. 体重至少 45 kg; 3. 符合 2017 年国际抗癫痫联盟 ILAE 难治性癫痫诊断标准: 规律服用两种抗癫痫药物且达有效剂量至少 1 年每月仍有发作,伴或不伴认知、情感障碍或头痛。4. 在过去 5 年内做过计算机断层扫描(CT)或核磁共振成像(MRI)扫描排除了癫痫的进行性原因。5. 育龄女性受试者必须采用可接受的避孕措施。排除标准: 1.进行血、尿代谢筛查,排除脂肪酸代谢障碍和生物氧化异常的相关疾病: 2.肠道不耐受: 如腹泻、胰腺炎、腹膜炎、肠道切除术后患者; 3.不能配合。

**合作单位要求:**有意愿,能配合完成随诊、完成皮层脑电图检查及血液检查。 **负责人及联系方式:** 林婉挥,13328200591

# 结节性硬化症相关癫痫 VNS 治疗的临床研究

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研究背景:结节性硬化症(tuberous sclerosis complex, TSC)患者 90%合并癫痫发作,其中至少 70%为药物难治性癫痫。癫痫外科是治疗难治性癫痫重要的方法,其中以切除性手术效果最好。181 例儿童 TSC 相关癫痫手术治疗的荟萃分析显示,56.3%患者术后无发作。2018 年申请人牵头完成的中国 TSC 相关癫痫手术治疗的回顾性分析显示术后 4 年的无发作率达到 60.8%。然而仍有许多癫痫患者不能确定致痫结节或者切除性手术后仍存在癫痫发作,就需要新的治疗方法。同时,迷走神经刺激术(vagus nerve stimulation, VNS)已经成为药物难治性癫痫的重要神经调控治疗癫痫,主要适用于不能进行切除性手术、不能确定致痫结节、开颅手术失败或者家属不同意进行开颅手术的结节性硬化症患者。

研究目的: 主要目的: 通过前瞻性对照研究明确结节性硬化症相关癫痫 VNS 治疗的临床癫痫控制、认知变化的有效性; 次要目的: 通过前瞻性对照研究明确结节性硬化症相关癫痫 VNS 治疗的安全性。

研究方法: 开放、前瞻性、非随机对照研究; 本研究是对临床对 TSC 相关癫痫常规治疗中的观察研究,不额外附加有创性检查或治疗; 分为 2 组。药物 V组: 初步评估有头皮脑电图、症状学均无局灶性特征,且无显著结节,但患者家属不同意 VNS 治疗,希望继续药物治疗; VNS 组: 初步评估有头皮脑电图、症状学均无局灶性特征,且无显著结节,患者家属同意 VNS 治疗,并最终完成 VNS治疗。

**患者入组标准:** ①临床诊断符合 2021 年版 TSC 国际诊断标准; ②癫痫诊断明确,病程 1 年以上,目前应用 2 种或以上抗癫痫发作药物,近 3 月癫痫发作>12次,曾应用 3 种以上抗癫痫发作药物; ③曾应用或未应用过雷帕霉素或依维莫司,应用或未应用过生酮饮食治疗; ④年龄 3-20岁,性别不限,有或无 TSC 家族史; ⑤经过初步评估(MRI、脑电图、发作症状、癫痫病史、神经科体征),符合《结节性硬化症相关癫痫外科治疗中国专家共识》标准中认定的,无法进行切除性手术或不适合颅内电极脑电图检查; ⑥家属同意入组并签署同意书。

**收集例数:** 120 例, VNS 和药物治疗组各 60 例(需要完成入组后 1 年随访)。

评估检查: 入组前和入组后 1 年、5 年均需要记录癫痫发作频率和类型、抗癫痫发作药物应用情况、mTOR 抑制剂应用情况、血常规和生化检查、IQ/DQ、QOL 神经科查体、心肾超声、胸片、发作间期 EEG,入组前还需要检查发作期 EEG、癫痫序列 MRI、基因检测,必要时 PET 检查。入组后需要记录治疗后药物副作用和/或手术并发症。

**合作单位要求:**可收集 VNS 组或药物治疗组病例,并完成 CRF 表格填写和相关术前术后评估检查。

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# 欢迎所有单位和个人参与上述多中心研究

# 2022 年 CAAE 青年委员会年度学术报告与交流大会

2023年3月25-26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人杰地灵的豫章故郡,洪都新府南昌召开。会议共300余人线下参会,除了青年委员我,廖卫平教授、周东、周列民、姜玉武、邓艳春副会长,张慧秘书长,段立嵘、丁玎副秘书长和江西抗癫痫协会张明会长、钟建民副会长,吉林省抗癫痫协会林卫红会长,江苏抗癫痫协会杨天明创会会长、李岩会长,天津抗癫痫协会杨卫东会长、陕西抗癫痫协会张华会长、河北抗癫痫协会李文玲会长、湖南抗癫痫协会彭镜会长等协会领导也莅临指导。阳春三月千里逢迎,高朋满座,共话中国青年癫痫发展事业,共叙CAAE青委的合作与友谊。





开幕式由江西省儿童医院信息中心主任、青委委员虞雄鹰主持。洪震会长在 讲话中对青委过去一年的工作给予了肯定,同时用习近平总书记在中国共产党第 二十次全国代表大会上讲话勉励青委们奋发有为,为我国卫生健康事业持续、高 质量发展锻造了坚强后备力量。江西省儿童医院傅超书记致辞中热情欢迎中抗专 家和青委会到英雄城南昌开展学术活动,高度概括了江西的历史和革命文化古迹, 全面介绍了医院在服务癫痫患者和开展创新发展方面的工作和未来规划。江西省 抗癫痫协会张明会长代表当地抗癫痫协会和工作者对会议在南昌召开表现欢迎 和感谢,简要介绍了省抗癫痫协会以提高当地癫痫诊疗能力、推动患者对癫痫的 正确认知等领域的工作,同时要求本省的参会人员认真学习讨论,促进江西抗癫 痫事业的发展。中国抗癫痫协会创会会长李世绰教授在致辞从珍惜青春、坚定政 治定力与明辨是非、增强实力与融入国际行业发展、学好英文与加强国际交流、团结协作与传承发展等五个方面,用肺腑之言对我国青年癫痫工作者提出了明确要求、表达了殷切希望、指明了发展方向。梁树立主任委员致欢迎辞并从立体化的抗癫痫西部行活动、菁 YOUNG 计划、领读学术、癫痫数据银行与多中心研究、CAAE 青年思行和指南工作、学术会议等 6 个方面对 2022 年青委进行了全面总结梳理,同时,也对 2023 年工作进行了部署。最后,提议青委们在新的一年既要务实笃行做好临床工作,更要勇毅创新搞好科学研究。

今年的学术内容包括 3 个单元:

癫痫热点问题争鸣与研讨是青委年会的特色栏目之一,今年由周健、季涛云、 王爽教授负责本部分的组稿工作,会议由他们和吴洵昳教授、陈子怡教授、孙丹 教授共同主持。本次会议首次特邀专家讲座,首都医科大学附属北京天坛医院王 群教授作题为"2022年癫痫领域 10 大进展"。中南大学湘雅医院彭镜教授进行 了"遗传性癫痫的基因治疗"的讲座,从基因治疗的概念、进展、未来方面等, 并结合自己相关研究进行了全面的介绍,廖卫平教授进行了点评和讨论。此外, 苏州市立医院王媚瑕医生、首都医科大学宣武医院乔梁教授、遂川县人民医院郭 崇伦教授、深圳市儿童医院操德智教授从癫痫共患精神疾病、Lennox-Gastaut 综 合征的多手段外科治疗、当癫痫遇上人工智能:进展与展望、儿童癫痫患者停药 时机选择等主题进行了新进展的介绍,周列民副会长、丁玎教授等专家先后参与 讨论。









青委年度最佳研究评比单元由刘永红、张凯、梁建民教授负责,并担任主持。 在 28 篇投稿中选择了 7 篇进行了汇报展示。经评委点评和现场观众投票,最终 中南大学湘雅医院冯莉医生获得最佳人气奖;首都医科大学附属北京天坛医院张 弨医生获得一等奖,中南大学湘雅医院冯莉、深圳市儿童医院李霖、浙江大学医 学院附属第二医院丁瑶三位医生获得二等奖;遵义医科大学附属医院田茂强、北 京大学第一医院于昊、武汉儿童医院王素梅三位医生获得三等奖。通过优秀论文 的演讲体现了青委们在 2022 年度的突出研究成果,也显示出在英语表达和理解方面存在一定差异,青委们需要进一步加强英文学习。

多中心研究环节由陈蕾、彭镜、李文玲教授负责并担任主行。两项关于饮食治疗的研究项目,分为是福建医科大学附属协和医院林婉挥医生汇报的"中链脂肪酸在成年难治性癫痫及共患病添加治疗的疗效、安全性及机制探讨"、首都医科大学附属北京儿童医院邓劼医生汇报的"生酮饮食治疗单基因变异所致婴儿期起病发育性癫痫性脑病疗效的多中心临床研究"项目;中南大学湘雅医院彭镜教授团队汇报了2项队列研究:贺海兰汇报的"电压门控氯离子通道和转运体相关癫痫/智力障碍队列研究"和潘邹汇报的"婴幼儿癫痫性痉挛综合征全国多中心长期队列研究"项目;空军军医大学西京医院刘永红教授团队有2项临床研究项目:张歆博汇报的"自身免疫性脑炎相关精神症状多中心临床研究"和王则直汇报的"SUDEP/Near-SUDEP 的多中心临床研究"项目。

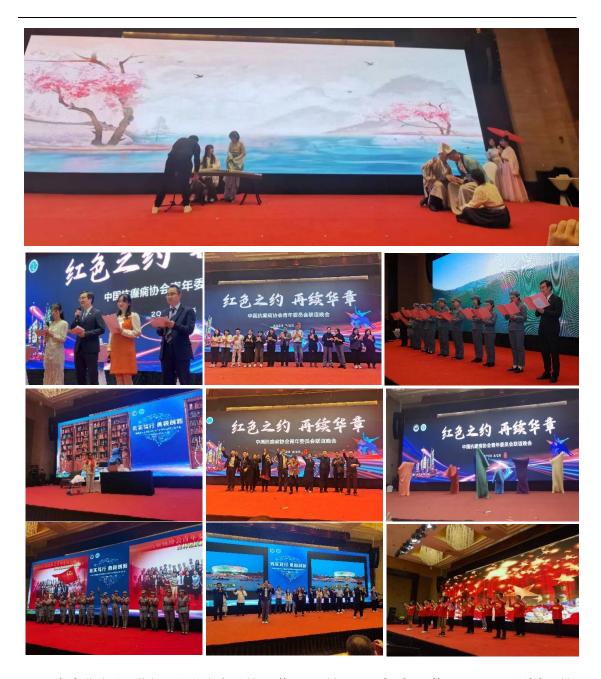


今年的人文环节包括 2 个单元:

人文建设可以帮助青年人更好地理解和认识世界、辨别是非,人文建设可以 提高青年人的文化修养,丰富精神财富,人文建设可以提高青年的道德水平和综 合素质。当然人文建设不可能一蹴而就,所以人文建设也是青年委员会长期坚持 的项目。今年的人文建设项目由孙丹和虞雄鹰教授负责。前期专门由张慧秘书长 牵头,成立了由段立嵘副秘书长、梁树立、孙丹和虞雄鹰教授参加的策划组。南昌的青委们进行了精心的准备和筹划,最终选择了以英雄雕像广场为出发点、南昌舰和江西省博物馆为参观学习场所,知识问答为检验形式的方案。在"追随前辈壮勇足迹,践行医学强国梦想"的口号声中,大家跟随导游的讲解认真参观,并不时发问深入交流,在晚上的相关知识问答中正确率超过90%,体现了青委们对人文学习的重视和丰厚的人文知识储备,最终黑吉辽大区依靠严密的团队合作、准确的知识记忆和领先抢答技巧获得第一名的好成绩。



青年癫痫学者联谊晚会是一个体现青委活力、团队协作和多才多艺的重要窗口,今年是后疫情时代的第一个线下联谊晚会,各大区的非常重视,节目也都经过精神筹划和排练,将文艺与癫痫有机结合在一起,真正做成了癫痫学者联谊会。中国抗癫痫协会秘书处的穿越情景剧和江西省儿童医院的舞台剧更是精彩纷呈。在大家的欢声笑语中,看到了青委们辛勤工作之余的放松、团聚后的喜悦和对未来发展的信心。联谊晚会由吴洵昳、陈子怡和郭强教授负责。虞雄鹰、郭谊、王剑虹和徐瑜欣担任晚会主持。



晚宴期间还进行了两项重要的工作,一是 2023 年度"菁 YOUNG 计划"的的启动仪式,在去年成功举办了以理论学习为主的"菁 YOUNG 计划"之后,今年更多集中在临床应用和病例展示,将理论和实验进行结合开展活动。启动会后将有开始 200 例比赛病例收集、初赛、复赛和决赛等一系列活动,体现青委们在精准诊断、精准治疗相关的临床与研究成果。最后,还进行了 2023 年度中国抗癫痫协会青年委员会学术报告和交流大会举办地的竞选,江苏省无锡市、徐州市和湖北省武汉市的青委们分别做了当地文化、美食、会议环境和癫痫事业发展等相关情况的汇报,最终经过现场票选,徐州成为明年会议的举办地。



本次会议的成功举办是在中国抗癫痫协会秘书处张慧秘书长和段立嵘副秘书长等人员的精心策划和细心准备下完成。江西省抗癫痫协会张明会长、江西省儿童医院领导、钟建民主任、吴华平副院长和虞雄鹰主任带领的团队为会议举办做了全面的支持和准备工作,特别是虞雄鹰主任从会场选择、人文路线挑选、晚会筹划等每个细节进行了推演安排,并担任了开幕式和晚会主持。两位会长、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。

2024, 相聚徐州!



统筹策划 张 慧 段立嵘 梁树立 **EPILEPSY YOUTH COMMITTEE** CHINA ASSOCIATION AGAINST 刊头题字 李世绰 资料来源 全体青年委员 CAAE网站 资料收集 梁建民 陈子怡 王 爽 陈 蕾 刘永红 吴洵昳 李文玲 张 凯 季涛云 刘婷红 资料整理 刘婷红 孙 丹 陈子怡 刘婷红 王雅婷 排版美工 THINKINGS AND ACTIONS