CAAE·青年思行

CAAE Youth Thinkings and Actions

2022年第三期

中国抗癫痫协会青年委员会

CHINA ASSOCIATION AGAINST EPILEPSY YOUTH COMMITTEE

目 录

"治愈"虽难求，“安慰”总常在

CAAE 青年委员会浙皖赣大区

浙江大学医学院附属第二医院 神经内科 癫痫中心 丁瑶

To Cure Sometimes, To Relieve Often, To Comfort Always------ E.L.Trudeau

第一部分

（该部分由一名癫痫患者的丈夫主要撰写）

“别看电脑了，早点休息吧，烧才刚退要多休息”。

“准备明天去上班，整理一下邮件，马上就好了，你先陪儿子睡吧。”

“检查报告都还没有出来呢，要不再休息两天，等检查报告出来再去上班吧。”

“烧都退了，没事了，马上就好了。”

那是 2020 年愚人节的前夜，当时的对话，我依旧记忆犹新，从那一刻开始命运对我们这个幸福和谐的小家庭开了一个难以承受的玩笑。

次日清晨，我被一阵床的震动摇晃吵醒，我心里正抱怨着孩子他妈起床动静怎么那么大，睁开眼发现她双眼上翻、全身抽搐、不省人事地躺在床上。手足无措的我，平生第一次拨打了 120 电话，急救、转院、急救……在反复几天的抢救中，她一直没有醒过来；在一次又一次的镇静药物治疗后，她依旧间断抽搐，无奈下我们终于辗转转院到了浙大二院的神经内科的病房。本以为可以松一口气了，没曾想，她的发作依旧反复并且持续出现，医生评估病情严重，当晚就转进了 ICU。看着医护人员将她推进 ICU，我彻底崩溃了，瘫坐在 ICU 门口的地上，那一刻我的心里充满了恐惧，在 ICU 的电梯间当众哭得声嘶力竭，我呼喊着医生，一定要救救她，拯救我们这个不幸的小家庭。

很快，我收到了病危通知书，医生说她得的是重症脑炎，在 ICU 里依旧是深昏迷状态，抽搐依旧持续，只能靠呼吸机维持呼吸，镇静药物都很难压制住抽搐发作；医生说由于颅压很高，压迫脑干，十分凶险，很大概率是无法醒过来的，即便醒过来以后估计也是植物人状态。

“念天地之悠悠 独怆然而涕下”，面对着突如其来的变故，想到家里年迈的父母和年幼的孩子，我表面坚定地说她一定能够醒过来的，然而内心却无比地无助和忐忑。

一天又一天，或许只有经历过的人才能体会在 ICU 门口等待的日子是何等的煎熬，无法用言语表述。阿昔洛韦，丙种球蛋白，甲强龙冲击，高颅压……这些原本陌生的专业名词，我反复从医生口中听到，于是我天天上网搜索、恶补医学知识，希望自己也能成为医学专家，拯救妻子。时值 2020 年的 4-5 月，正值武汉新冠疫情暴发不久，全国都谈疫色变，医院规定 ICU 的病人不能探视也不能陪护。家属每天唯一能做的就是在 ICU 门口等待医生的消息。我和我的家人常常为了等消息，在 ICU 门口一站就是一整天，甚至卷着铺盖在 ICU 门口守夜，然而等到的总是“还没有醒”……

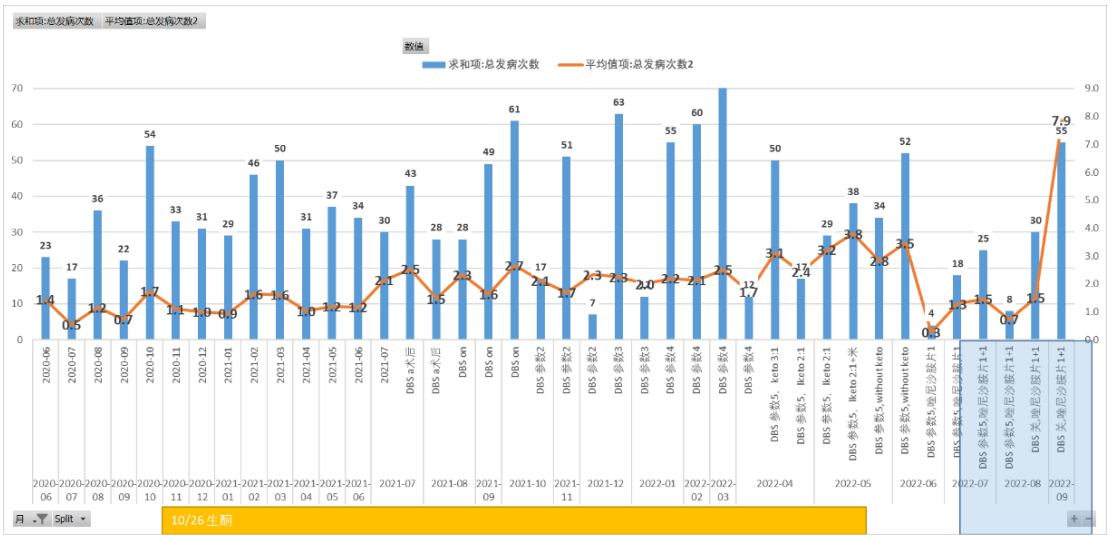
在近一个月无奈的等待，一次次的翘首期盼和一次次的悲伤失望之后，感谢上天眷顾，终于有一天，医生面容轻松地走出 ICU 的门口，告诉我，我的妻子有转醒的迹象了。那天，我坐在 ICU 的门口幻想着，应该会像电视剧里常见的剧情一样吧，妻子醒来后，能从 ICU 笑容满面地走出来至少是坐着轮椅被推出来……

终于有一天，医生通知我，妻子可以从 ICU 转出了，事与愿违，被推出来的与我之前日夜相守的妻子判若两人，虽然没有持续的抽搐、身上的大部分管子已经被拔除，她依旧处于意识模糊的状态。之后我们转入了康复科，经过数月的康复、高压氧等治疗之后，妻子的意识状态逐渐恢复，但始终无法记起过去的事情，也无法形成新的记忆。曾经的亲戚、朋友、同事来探望，她也不认识了；过去事情任何事情她都不记得了；刚吃过的东西、做过的事、说过的话、见过的人转身就忘，并且说话经常重复喃喃自语。医生说这是后遗症：认知障碍，我不知道人失去记忆是怎样一种体验，但我想她的内心一定充满着恐惧和无助。

除此之外，更可怕的是她每天都会发生癫痫性抽搐，一不小心就直挺挺地摔在地上，摔得头破血流，头上缝了一次又一次，身上的皮蹭破了一块又了块。看着她痛苦的样子，家里人也时刻神经紧绷。认知障碍又加癫痫，无论对于病人还是家属，简直就是地狱般的生活体验：给她买了轻便的安全帽，希望能够保护她头部，好不容易说服她带上，但她很快就忘记并脱掉了，头部依旧不停地受伤；我在房间铺好了软地板，让她待在房间里，但只要家人一不注意，她就从房间走

了出来，反复摔在瓷砖上，还造成了骨折……

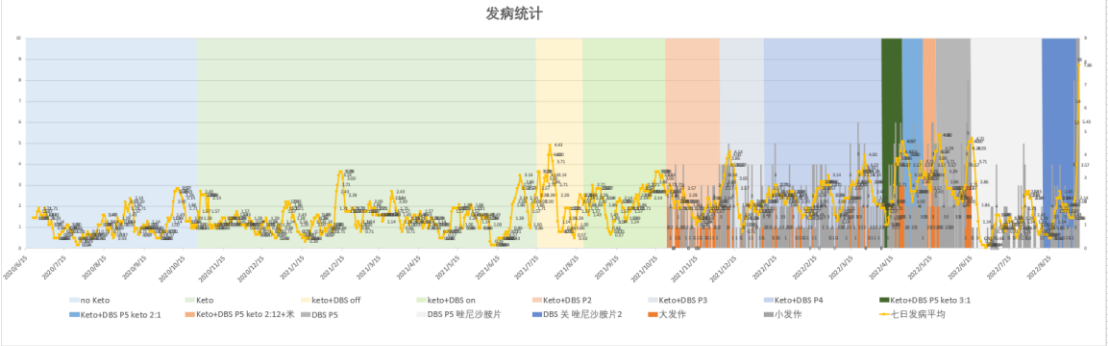
从出院开始，我每天都坚持做详细的癫痫发作记录（图 1-2），图表画了一张又一张，我希望尽我最大努力，好好记录，监督她用药，争取能减少发病频率，让她减少痛苦。在药物治疗方面，我们尝试了市面上能买到的各种各样的抗癫痫药物：奥卡西平、左乙拉西坦、丙戊酸钠、拉莫三嗪、吡仑帕奈……各种组合、各种尝试，均无明显效果。该发生的抽搐一次都没有减少，并且发病后短短五个月，她的体重就从最初的 100 斤不到迅速增加到 150 斤。通过医生的建议，2020 年 11 月我们启动了生酮治疗法，在生酮的初期，发作有所减少，体重从 150 斤回落到 100 斤上下，并且人的反应较前有明显的改善，似乎变得“聪明”了一些，我欣喜若狂，觉得看到了希望。然而好景不长，生酮 3 个月后除了体重和人的精神状态、认知有很明显的改善外，发作频率似乎又回到了基线状态。后来听说 DBS 手术对癫痫发作有改善作用，有 50%发作减少的概率，于是我们于 2021 年 7 月我们做 DBS 手术，DBS 手术参数调整后初期，发作似乎也有减少，然而随着时间的延长，发作频率似乎又回到了基线状态。



图一 患者丈夫描绘的发作次数随时间变化情况

很感激陪伴我和妻子在抗击病魔过程中一路走来的浙大二院的癫痫专科医生们，一路过关斩将，治疗过程中的各种艰难险阻唯有医生无私地帮我们共同面对。虽然，妻子目前的癫痫情况控制的还是不尽如人意，不过我们还在做各种尝试和努力，有专业医生的支持，我想随着科学技术的进步，总有癫痫控制的一天。

我经常思索，人生这道题，无论怎么选，都会有遗憾；无论你怎么精心策划，都抵不过命运的安排。认真做好每一天的事，积极面对，也许最终的结果，在冥冥之中自有安排。



图二 患者丈夫描绘的治疗方法与发作次数情况

第二部分

与患者以及家属接触的两年来，我被患者丈夫的诚挚所感动，每次门诊就诊都能见到患者拿着认真详细的治疗和发作记录图表。至今，这个病例的结局并不完美，让我想到长眠于纽约东北部撒拉湖畔特鲁多医生的墓志铭“To Cure Sometimes, To Relieve Often, To Comfort Always”------“偶尔去治愈，常常去帮助，总是去安慰”。这个病例和克鲁多的墓志铭使我陷入沉思，帮助和安慰是我们医生对于患者必要的人文关怀，而希望随着医学的不断进步，我想“治愈”的前缀今后并不是“偶尔”。

最近 20 年来，我国的癫痫领域变化日新月异；20 年前，我们与国际先进水平差距很大，而目前总体而言，中国的癫痫诊治水平已处于亚洲国家前列，接近欧美发达地区水平了，各种新型抗癫痫药物、手术、生酮饮食、神经调控治疗等手段日新月异。然而还是有一部分患者处于难治性癫痫的状态，特别是对于本文的这类重症脑炎后的癫痫患者。每次想到这些难治性癫痫的患者，就会敦促我们努力学习更多的专业知识、积极与国内外大咖交流、多阅读国外文献、多多做临床相关的诊治探索，唯有如此，治愈才能多一些、再多一些。我想这也成为我在抗癫痫领域不断前行和不懈努力的动力。因为，有太多的“妻子”、太多的“孩子”、太多的家庭等待着我们去治愈，与此同时，这些患者以及家庭更需要我们医生去陪伴、关怀和安慰。

交心式沟通

——封控时期癫痫患者及家属的“心境稳定剂”

CAAE 青年委员会粤桂琼闽大区

广东三九脑科医院癫痫中心 郭强

“憋了一整天都不让出病房门，我不管了，无论如何我就要出来透透气!”

“我都说了不能吃辣的，你们怎么给我配的餐？又不让我自己下去买！”

“配送口服药的怎么还没有送过来？我病情加重了谁负责？既然封控了就要保证物资药品配送准时呀!”

“我在隔离酒店里癫痫发作怎么办？我的安全谁来保障？”

“等着做手术，天天这么耗着，这耽误的住院天数、多出来的床位费怎么算？”

“车还在停车场呢？这么多天停车费贵得要死，医院给报销吗？”

“好不容易跟单位请假来治病，现在假期消耗完了，病还没有治!”

……

2022 年 7 月 10 日，“广州 710 疫情”，有三个阳性病例与广东三九脑科医院癫痫中心存在交集，癫痫中心所在的六楼病区无疑成为封控重点：全体 65 名医护人员成为密接被运送至酒店隔离，20 名轻症癫痫患者被办理出院另行隔离，6 区剩余 91 名癫痫患者和 106 位家属被就地封控隔离。癫痫中心医护集体突发“失能”，医院紧急增派三十余名神经内外科医护增援接管病区，维持基本诊疗工作和防控工作。

按“7+3”原则，癫痫中心的解封要在 10 天以后，而在这 10 天里，没有癫痫专科医生在场，所有患者都难以进行实质性的专科诊疗，而且封控期间对他们的种种限制约束，病房生活模式瞬间发生改变，有不少患者在病房度日如年。恐慌、紧张、焦虑、懊恼、愤怒、迷茫……各种情绪在病房蔓延。封控第二天，各种抱怨声接踵而至。

身处隔离酒店，一边完成疫情流调的各项统计任务，一边还要远程协助保障这 111 名患者的病情平稳及情绪平稳，癫痫中心团队面临着前所未有的挑战。我们在卫健委及院领导的指导下，最终平稳地应对了这次前所未有的挑战。后期复

盘，各种教训、经验不胜枚举。而今梳理出个中点滴，有关癫痫患者家属的心理疏导，也许能在行业内朋友们遇到类似情况作为借鉴与参考。

图 1：云查房：颅内电极置入术后患儿遥遥（化名）最近迷上了《侏罗纪世界 3》，那么……五只霸王龙前来保护你！图中上排左：护士长伍新颜，中：业务组长谭红平，右：运营助理周洋，下排左：主治医师陈淼彬，右：科主任郭强。

一、云查房

封控期间，患者每日面对着陌生的、非本专业的、全副武装的支援医护人员，而自己平时亲切的主治医生、主管护士不见所踪，这是患者恐慌、焦虑的重要原因。为了让患者心神安定，癫痫中心在隔离酒店第一日便开启了视频云查房。

中心作出整体部署：

以各业务小组为单位，业务小组长及主管医师每日至少两次云查房，逐一解决患者的疾病诊治、隔离起居等问题。

向全体医护强调，由于不能亲临现场，云查房容易遗漏患者重要信息，所以要求医护耐心询问每一个细节，不可流于形式地寒暄，并要求与现场的支援医生密切沟通形成联动。

主诊医生是患者在封控无助之时最信赖的人，自然也是稳控患者情绪的首席心理辅导者。云查房过程中应尤其注重与患者充分互动，讲究共情，设身处地地体验患者的处境，积极作出心理疏导。

通过十天的云查房举措，在封控期间最大程度地保障了医疗质量和医疗安全，极大的稳定了患者的不安情绪，患者普遍感到新颖、有趣和暖心。

二、患者的核心诉求是什么？

被封控在院的患者吐槽五花八门，问题解决难易程度有高有低，各位医生谈话方式不同，传递的信息也不完全一致。如何让团队所有成员都能够实施高效沟通，有效解决患者痛点？我们收集所有问题反馈，总结患者的抱怨与疑问不外乎三点：

我的病怎么办？

我的损失怎么办？（时间、金钱）

何时解封？

厘清了患者的核心诉求，癫痫中心便可针对上诉问题制定统一的回复，让医生们以一致、可信的口径实施有效沟通，避免躲闪其词。

对于病情，主管医生通过云查房，紧密跟进患者的病情动态，并与病房支援医生形成联动，用自己的专科知识为患者答疑解惑，并保障重要的诊治措施继续有序实施。

对于封控造成患者的部分财务和时间损失，中心积极的向医院申请，减免封控期间的床位费，承诺下一次脑电图复查优惠，并为患者赋上“癫痫中心 VIP”标识，承诺未来其就诊将优先及关照等，用实质性措施来弥补患者的损失。同时，医护人员积极的宣教：疫情防控大局的胜利，有赖于广大人民群众的支持，你们的鼎力配合、你们的各项损失癫痫中心都将铭记于心，未来，我们将以更优质的服务来汇报你们的付出！当话说到这个份上时，多数患者尚能释怀。

对于何时解封的问题，可以按照“7+3”原则初步向患者估算，让患者翘首可盼，但切记，医护人员决不可将话说得太满。因为疫情瞬息万变，核酸、健康码及流调数据不断在更新，政策也随时可能相应调整！

三、“给我个面子”

医院的减免、优惠依旧无法弥补患者的损失怎么办？

有些政策待遇无法做到人人绝对公平，患者之间互通有无之后，觉得自己吃亏了怎么办？

患者的诉求过于离谱，无法完全满足怎么办？

“给我个面子。”作为崇尚科学、正直儒雅的脑科医生，很少会抛出这么一句颇具江湖气息的话。但能说出这句话的医生，其所营建的医患关系一定不简单。

医学是有温度的，医患关系不应仅仅建立在医学科学的基础之上。患者有血有肉，医生也是如此。通过这次疫情风暴的洗礼，我们惊奇地发现，高水准的医疗服务不仅仅要求对患者机体施加科学的医学诊治措施，或简单加以微笑服务，或无限度满足其合理、不合理的诉求，更要求医护人员与患者交心、共情。有时候患者并不想做上帝，他更想与医生们做朋友！

试问，如果没有发自内心的关爱，没有与患者患难与共的真情，他们为什么要“给你面子”？因此，简单一个“面子”，是建立在医生与患者交心服务的基础上的；简单一个“面子”，折射出医患关系之外的兄弟、朋友情谊；简单一句“给我个面子”，也是对患者作出未来更优质服务的承诺。

沟通有两种方式：一种是目的式沟通，一种是交心式沟通。前者着重于向患者传递医学信息，履行告知义务；而交心式沟通则可以让双方的感情有了更深入的可能。事实胜于雄辩，在与患者交心沟通之后，沟通效率提升、效果明显。在不少患者，疫情带来的诸多委屈与不公，相较于与医生朋友的情分而言，“那都不算事”！

四、化干戈为玉帛

“不是说好周一解封吗？你们怎么出尔反尔，又延到周二了？”

“周三才能出院？周三下午孩子考试怎么赶得及？！”

“我周三无论如何一定要上班！”

“叫你们书记出来，解释不清我就打电话给卫健委！”

在同一个病房里的三位孩子家长，得知封控结束时间由周一调整到周二，自己的行程计划又被全部打乱之后，再也坐不住了，集体抗议。一直以来的压抑与不满，在封控到第 8 天时终于爆发出来。他们情绪激动，在病房大声呵斥医护人员，拒绝配合核酸检测。医护前几波沟通均以失败告终，眼看其他病患家属也即将引起共鸣，形势严峻！

我立即联系在病房忙于安抚工作的医生，让其召集三位意见最大的爸妈到一个单独房间，由我与他们视频沟通。

先做自我介绍：莲莲妈，曦曦妈，明明爸（均化名），你们辛苦了！我是癫痫中心主任郭强医生。

接下来逐一、耐心的听他们倾诉苦水，将他们抱怨的痛点做笔记，并快速想好如何回应。

随后，我逐一地回答他们，解封日延期到周二的具体原因是什么，周三出院能否保证、是否还会再次生变，出院时我们将如何快速办理手续以保障家长上班与孩子考试……

最后，我说了以下这番话：

“莲莲妈，曦曦妈，明明爸，首先我代表癫痫中心全体向你们抱歉，很愧疚因为疫情造成病房封控，给你们带来不好的就医体验。”

“第二，我想说，在全员红码隔离、全院封控一周多的时间里，我们保持着院内零感染。我们能够扛到今天，很大程度上有赖于你们过去 8 天的理解和付出。对于你们的支持与配合，我们心存感激，尽在不言中！未来的诊治路上，莲莲、曦曦、明明，还有病房的其他孩子，他们就是三九脑科医院癫痫中心的 VVVIP。若有任何需求，请随时跟我提出，我会想方设法让你们满意，用最优质的的服务来回报你们的付出。”

“相信这次封控，在每个人的人生中都是难以忘却的一段历程。不管苦过也好，委屈也好，我们是一起并肩走过的，这就像我们在癫痫诊治之路上并肩作战那样，我尽心尽力，您信任配合。我们非常珍惜这样一段特殊的缘分，珍爱这份来之不易的情谊。我们还希望，在以后的日子里，我们也能同舟共济，守望相助，不惧任何困难！”

……

随后，我与家长们互加了微信，成了患难与共的朋友。

这次沟通十分有效。三位家长也还是明事理之人，深知眼前的诸多不便多为疫情形势所逼，深刻理解了医护的不易，在剩下的两天里，他们遇事不再火冒三丈，非常配合理解。

五、后记

终于熬到 7 月 20 日，癫痫中心正式复工，各位被困多日的家长也顺利带孩子出院。到晚上，我逐一发信息关心他们的返程情况。家长们纷纷回信，回到家后，他们当天就立即投入到紧张的工作中去了。对我们十天来的关心、协调和照顾，他们都非常感谢。

交心式沟通作为患者家属心境稳定剂，运用在封控时期这样极端的情境下，还是卓有成效的。

多学科大爱伸出援手 紧要关头共同承担

CAAE 青年委员会粤桂琼闽大区

深圳市儿童医院癫痫中心　　操德智 李艺廉

2022 年 3 月，广东深圳，新冠肺炎疫情再起波澜。2022 年 3 月 11 日下午 14:30，我正在外面会诊一名不明原因发热的孩子。突然接到科里护士长的电话，说我们早上混采的一管核酸结果可疑阳性，所有参加混采的医护人员全部被隔离到我们医院的临时小平房里，进行单人单管的复核。

“啊！“我大叫一声。要知道那十个人中除了一名护士，其他全是我们医院癫痫外科的医生和脑电图技师。我自己因为早上采样时快了一步，放在了另外一管，而“幸免于难”！虽然我知道这种检测出现假阳性的可能性很小，但心里心存一丝侥幸，等复查结果出来再说。我迅速回到科里，护士长和剩下的护士、护工们正在进行环境采样和封控，眼看，科里的医生就剩了我一个“光杆司令”。接下来，我们在漫长的等待结果当中，做着最坏的打算！

很快，到下午 5 点多，复查结果出来了，是我们的一名技师不幸“中招”，确诊阳性了！而她前一天的核酸检测结果还是阴性，之前也没有明显的呼吸道症状，只是早上喉咙有点干涩的感觉。她自己前 1 周的行程也极其简单，严格的两点一线上班，路途中坐过地铁和公交，也都有严格戴口罩，没有与之前报道的阳性患者有过任何交集。这让我们所有人都感觉到病毒的狡猾和诡异！

很快，我们所有科室工作人员都被定为“核心密接”，而所有住院患者也都被

定为“密接”，所有家属都被定为“次密接”，同时还有前几天跟我们科医护人员，特别是我们那位技师有接触的，都被定为“密接”或“次密接”。我们医院瞬间面临了整个疫情期间以来最大的防控压力。还好，我们领导的反应及时，平时的预演到位。很快各职能部门各司其职，给我们进行了全方位的指导和安排。我们科的相关工作人员很快被转移到隔离酒店隔离。我们留在科里的几名工作人员当天也坚持在岗位继续做好病人的安抚和善后工作。

第二天，我们癫痫外科除一名在休假的护士外，其余所有员工均被拉到酒店隔离，同时我们的家属也被要求居家隔离或酒店隔离 7 天。

但疫情无情人有情，我们日常的工作并没有丝毫懈怠。

此时的病房中，有年龄未满 3 月整，前一天刚做完一场大脑半球次全离断的小宝宝；有术后不久正在观察期的小姑娘；有刚做颅内电极植入术，等待脑电监测记录及治疗的孩子，还有正在术前评估中或已评估完即将出院的孩子……

很快，在医务科的统一领导下，我们及时抽调了神经内科、神经外科、脑电图室的 “高手”前来帮忙，他们分别是：神经内科林素芳主任，神经外科刘明刚

主任，神经内科丁鑫医生，神经电生理童宛凌技师，神经内科田小琴护士，放射科尹少琼护士，血液肿瘤科赖碧芬护士，神经外科胡梦悦护士，中医科宋雨阳护士，供应室黄楚璇护士，癫痫外科李慧萍护士。他（她）们接到通知一点拒绝都没有，就准备好衣服，做好在病房进行 14 天封闭式工作的准备。

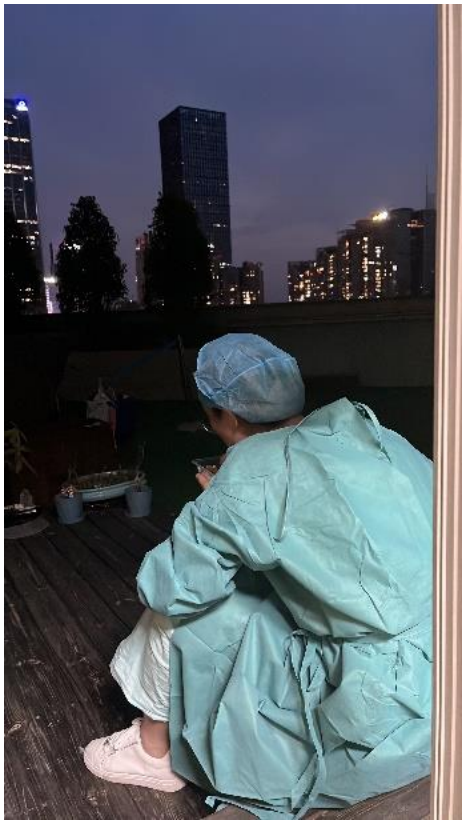
每日清晨，在操德智主任的主持下，所有人会在线上开晨会，分享当日查房情况，讨论病情，商议下一步做法。操德智主任，朱凤军主任，陈彦主任及各位管床医生、各位电生理医师技师做在线指导。

晨会结束后，癫痫外科的医生们也没闲着，各位都开通线上平台，帮助家长们线上咨询与购药。由于城市“慢行”，有些家长或处于“三区”（封控区、管控区或部分防范区）内无法外出，或正在隔离，或因交通管制无法到达医院。他们会因药快吃光了买不到而焦虑，会因孩子发作而忧心。我们的医生一对一的解答、安抚，帮他们尽量解决药物问题。操德智主任还设计了调查问卷，收集家长们对小朋友病情的担忧，并且每一个都亲自回复。

那段特殊时期，虽然我们不在你们身边，但是心里时刻挂念。终于在大家的努力下，病房中的孩子们都顺利出院了。

随着 3 月疫情平稳，这一段时光即将过去，生活也将回到正轨。癫痫外科全体医护将继续保持严谨态度，以人为本，为广大电宝宝排忧解难。

在这里要再次感谢每位在疫情袭来是逆行的英雄们!感谢你们辛苦的坚持与辛勤的付出，感谢你们每日穿着防护服在狭小的病区走出上万的步数，感谢你们在艰难时刻的共同承担。



谁说站在光里才是英雄

微笑-开启信任的大门

CAAE 青年委员会陕甘青宁新大区

延安大学咸阳医院 癫痫中心 年艳艳

“有时去治愈，常常去帮助，总是去安慰”。借用特鲁多医生的墓志铭来形容医护人员与癫痫患者们，我想是最合适不过的了。癫痫为可治疗性疾病，大多数病人预后较好。但不同类型的癫痫预后差异很大，有自发缓解、治疗后痊愈、长期服药控制和发展为难治性癫痫等几种预后形式。对于医护人员来说，面对癫痫患者，不光要有精湛的诊疗技术和精心的护理服务，还应具有在面对癫痫患者时足够的耐心和温暖的微笑。开启与癫痫患者信任大门的钥匙就是——微笑。

在临床上，和患者沟通所用最多的肢体语言就是微笑，前一段时间，我护理的一位癫痫患者，我就感同身受。是一位四十多岁的阿姨，确诊为癫痫，刚住院期间，她一句话也不说，脸上的表情都很少，查看她的病例，智能稍有障碍，但是不影响日常的生活能力和沟通，只记得前几天吐得厉害，进食差，每日在做完护理操作和相应的治疗后，我总是习惯性的会问一句：阿姨，今天吃饭了没？吐了有多少？同时，会交代在一旁的家属：虽然吐了，但是还是要吃饭。就这样过了几天，阿姨呕吐的症状较前缓解，到病房和阿姨的沟通也多了起来，渐渐地，阿姨也用微笑回馈我。每天还没等我开口，阿姨就先微笑着问我：吃饭了没？有时甚至在中午比较忙的时候，阿姨还会关心的说：“累了就歇一歇”。临床上还有许许多多像这位阿姨一样的癫痫患者，因为反复发作引起的智能减退，在面对这样的患者时，微笑更胜千言万语，他们会感受到作为医护人员的真诚。

还有一对母女，女儿在科室住了好几次，母亲已经六十多岁了，女儿四十多岁，但是由于癫痫反复的发作，女儿的衣食住行离不开人，每次来住院的时候，阿姨总是一脸疲惫，但是在女儿面前表现的依旧是那个无所不能的妈妈。因为经常住院的缘故，阿姨把科室的医护人员当成她最好的倾听者，背过女儿，在最信任的医护面前，把她的委屈和心酸释放出来。在她近期出院的那天早上，阿姨对我说，因为看病，把家里的房子也卖了，但是她不想接受别人的捐赠，靠着她的退休工资，把日子就这样朝前过着吧。阿姨抹完眼泪，依旧笑着对我说：没事，

阿姨能想得通，只要闺女病情稳定，我也就放心了。一时间，我也想不出更好的安慰语言，报以微笑给阿姨。这里的微笑不光代表着信任，更是一种鼓舞的力量。因病致贫，因病返贫的案例还有许多，这也更加坚定了我们规范诊治，精心护理癫痫患者的决心，为他们减轻身心痛苦，帮助癫痫患者家庭树立希望。

今年“国际癫痫关爱日”的主题是“癫痫关爱在社区”，通过举办不同形式的宣教活动，大力宣传在社区范围内癫痫关爱和管理的必要性和重要性，呼吁政府相关部门，社区广大民众，正确认识癫痫疾病、规范癫痫诊疗，给予综合关爱、创建和谐社会，对于我们医护人员来说，构建与癫痫患者之间信任的桥梁就是真诚的微笑，使他们在就诊的过程中，消除紧张感和焦虑感，从而产生战胜疾病的信心，能够与我们共同享受生命的精彩。

去伪存真，辨“症”假

CAAE 青年委员会湘鄂豫大区

湖北十堰太和医院　惠钰萍　赵峰

夫医学之要，莫先于明理，其次则在辩证，其次则在用药。理不明，证于何辨？证不辨，药于何用？

我常想，人在遇到危难时，别人问一声都是好的；有人病了，给他一碗白开水，人家心里也是暖烘烘的。救死扶伤，解除病人痛苦，维护病人健康，是医务工作者的神圣职责。医务工作者除了要有过硬的业务技术外，更要有一颗全心全意为人民服务的心，这是基本的、必备的条件。

临床临床，就要亲临病床，亲手掌握第一手资料，才能做出正确的判断。说着这句话脑海里就浮现了一位 6 岁小姑娘的身影，那是一个皮肤白皙，脸圆圆可爱的小女孩，在半年前上学一个月后开始出现每天夜间睡眠中突然惊醒伴大喊大叫，惊恐表情，不认得亲人，伴肢体挥舞动作，无明显的抽搐每次持续约十几秒，之后清醒，过程有印象，但描述不清，有时一晚上会出现几次，病后家长曾用偏

方（头部及上肢扎银针、推拿等）治疗，但效果欠佳，家长说也不知道是不是有精神病方面的遗传，孩子父亲的奶奶、姑姑、姑姑的女儿、外甥女都有精神类疾病病史，而且孩子在两年前被一个男人大声对她吼了一声之后，才突然出现睡眠中突然惊醒尖叫的情况。孩子自幼感觉也不合群，喜欢自己玩，好动、坐不住，易激惹，一不满足要求就哭闹不止，很难哄住。现在孩子上一年级了，老师经常反应孩子上课坐不住，扰乱秩序，作为家长的我们也确实有点着急，怕耽误学习和课堂，近期对她发脾气比较多，不听话就吵她，前两天在课堂上老师反应孩子突然出现了大喊大叫的情况，以前只有在睡觉的时候出现，我们在考虑是不是因为我们吵她太多了，所以害怕我们了，也让她的情况变糟糕了……

话语间听出了家长的自责与懊悔，家长说我们也带孩子去当地医院看了，做了头颅磁共振未见明显异常；心脏彩超:左房稍大，心功能正常；多动症测试 22 分，属于明显多动范畴；做了瑞文测试中度；血常规、肝、肾功能、电解质、甲状腺功能 5 项、生化全套均无异常。当时就推荐我们去看了心理行为门诊，做完心理测试后，医生告知我们孩子确实存在一些心理问题，包括行为和情绪方面，孩子也存在幻听的情况，经常会听到自己的好朋友或者陌生人的声音在自己的耳边对自己说话，声音还非常大，也会经常听到猫或狗的叫声，在入睡前或睡眠状态下听到，经常会被吓醒，会感到很害怕，心理医生就建议需要做心理治疗。

做了一个疗程的心理治疗后，行为和情绪上有好的转变，但是还是会有大声尖叫和惊恐的情况出现，孩子做心理治疗同期，心理医生也给我们家长提供了相应的心理疏导和家庭指导，所以一直到现在我们对待孩子已经很注意了，可为什么还是这样呢？我们也不知道怎么办了，现在就到你们医院来就诊再看看，希望能搞清楚到底怎么回事？

听完家长的话我先安抚道：“家长你们先别着急，我们其实很能理解你们的心情，我们的初衷和你们的愿景是一样的，都是希望孩子好。”说完就将他的病史全部梳理了一遍，汇报给上级医师，发现虽然她的心理问题突出，又有家族精神疾病病史，但小姑娘仍存在大声尖叫、惊恐表情的表现并未得到改善，即使这个问题看起来好像也能因为心理问题这个标签而顺理成章变得合理化，但从神经学考虑应该首选要先排除癫痫可能。之后就建议家长完善 24 小时视频脑电图，刚好在做脑电图的期间捕捉到了一次局灶性发作，临床表现就是一直存在的大声

尖、惊恐表情伴肢体挥舞动作，根据发作频率、发作持续时间、发作后的意识状态、发作症状以及发作间期放电和发作期的脑电图，临床结合脑电图结果，考虑为额叶癫痫。

在确诊后，跟家长进行沟通，告知这是额叶癫痫发作的一种临床表现形式，根据检查结果与临床表现，我们建议口服 xxx 抗癫痫药物。那么对于这个 6 岁的小姑娘来说到底是先用抗癫痫药物控制癫痫发作还是先用精神类药物治疗心理问题呢？因此我们要先理清病情的主次，对于癫痫患者伴发的精神障碍治疗，首先应合理运用抗癫痫药物治疗，对于原发疾病的治疗最为重要，积极控制癫痫发作的频率。因调节精神药物的作用可能有限，甚至某些抗精神病药物可以降低癫痫发作的阈值，诱发癫痫发作，所以在抗精神病药的使用过程中应该谨慎。如果没有搞清楚病因就先用精神类药物，那样就会加重病情。所以医学中常说“医学贵精，不精则害人匪细”。

这个病例对于低年资的我来说算是一次非常好的警醒，更加意识到询问病史的重要性，如果病史问诊不够详细，很容易就漏掉了重要信息，所以必须要亲手掌握第一手资料，才能帮助后期做出正确的判断。

在吃药三个月后，家长说孩子发作减少了，家长很开心，同时也在做心理疏导，行为和情绪也好转很多，我们也很欣慰。但其实当时家长得知是一种癫痫发作后他们是难以置信的，就问我们:“孩子不是心理问题吗？怎么会是癫痫了呢？”是呀，这两个问题对我们医务人员来说也是一种很好的反思，它是一种癫痫发作，但是我们却没有及时发现端倪，虽然现在的专病专诊我们已经做得很好了，但是面对复杂的病情，我们需要拓宽我们的思维模式，累积丰富的经验，全方位的把握病情，不能以偏概全。而且我们可以利用现有的资源，积极发挥各院癫痫 MDT 的作用，同时也能从各个专业角度出发探讨病情，给出专业化、合理化的建议，实现于患者病情及个人利益的最大化，也能提升医务人员的专业水平，实现共赢。

医学之路漫漫，要做一名好医生，首先一点要研究人，全心全意为人民服务，这就是医德，医德不光是愿望，更是一种行动，这个行动要贯穿医疗的全过程，贯穿医生的整个行医生涯。大约心细则眼明，而理名则心细，多读书辨证则理明识广。

编号：XEY-2022-4-1

引用格式： Tang Y, Yu J, Zhou J, Chen J, Hu S. ^{18}F-SynVesT-1 PET in focal cortical dysplasia type II with thickening cortex. Clin Nucl Med. 2022 47(8):741-743. doi: 10.1097/RLU.0000000000004214.

通信作者：陈晨

Abstract

A case of 2-year-old girl with intractable seizures underwent 18F-FDG PET and MRI for seizure focus localization. MRI demonstrated cortical thickening and blurring of the gray-white matter interface in the right postcentral gyrus with focal hypometabolism in 18F-FDG PET. The patient subsequently was enrolled in clinical trial of 18F-SynVesT-1 PET study in epilepsy; a more restricted area of reduced 18F-SynVesT-1 uptake was noted in the thickened postcentral gyrus. The surgical limits of resection were defined based on ictal semiology, electroencephalography, and imagings. The patient is seizure-free after epilepsy surgery, with histopathology of focal cortical dysplasia type IIb.

编号：XEY-2022-4-2

引用格式：Xie C, Liu F, He H, He F, Mao L, Wang X, Yin F, Peng J. Novel HCN1 Mutations Associated With Epilepsy and Impacts on Neuronal Excitability. Front Mol Neurosci. 2022;15:870182. doi: 10.3389/fnmol.2022.870182.

通信作者：彭镜

Abstract

Hyperpolarization-activated cyclic nucleotide-gated (HCN) channel plays a critical role in regulating the resting membrane potential and integrating synaptic transmission. Variants of HCN1 have been recognized as causes of epilepsy, and mutant HCN1 channels could act with loss-of-function (LOF), loss- and gain-of-function (LOF and GOF) and gain-of-function (GOF) mechanisms. However, phenotypes and

pathogenesis of HCN1-related epilepsy are still poorly understood. This study enrolled five epileptic cases carrying five different HCN1 variants: two pathogenic variants (I380F and S710Rfs*71), two likely pathogenic variants (E240G and A395G), and a paternally inherited variant (V572A). Four variants were novel. Electrophysiological experiments revealed impaired biophysical properties of the identified mutants, including current densities and activation/deactivation kinetics. Moreover, three variants exerted effects on the biophysical properties of wild-type HCN1 channels in heterozygous conditions. Immunofluorescence experiments showed that two variants reduced the protein expression of HCN1channels in neurons. Neurons expressing E240G (GOF) variant showed increased input resistance. However, the variant of I380F (LOF) increased the neuronal firing rate, thus leading to neuronal hyperexcitability. In conclusion, the present study expands the genotypic and phenotypic spectrum of patients with HCN1-related epilepsy and clarifies the underlying mechanisms. We reported five new cases including four unreported likely/pathogenic variants. We provided assessments of biophysical function for each variant, which could help patients to receive individual therapy in the future. We confirmed that HCN1 variants contributed to neuronal hyperexcitability by regulating input resistance and the action potential firing rate, and we have shown that they can affect protein expression in neurons for the first time.

编号：XEY-2022-4-3

引用格式：Xiao H, He H, Wu T, Ni X, Liu F, Yin F, Peng J. Functional Investigation of TUBB4A Variants Associated with Different Clinical Phenotypes. Mol Neurobiol. 2022;59(8):5056-5069. doi: 10.1007/s12035-022-02900-9.

通信作者：彭镜

Abstract

Dominant TUBB4A variants result in different phenotypes, including hypomyelination with atrophy of the basal ganglia and cerebellum (H-ABC), dystonia type 4 (DYT4),

and isolated hypomyelination. Here, we report four new patients with a novel TUBB4A variant (p.K324T) and three new patients with previously reported variants (p.Q292K, p.V255I, p.E410K). The individual carrying the novel p.K324T variant exhibits epilepsy of infancy with migrating focal seizures (EIMFS), while the other three have isolated hypomyelination phenotype. We also present a study of the cellular effects of TUBB4A variants responsible for H-ABC (p.D249N), DYT4 (p.R2G), a severe combined phenotype with combination of hypomyelination and EIMFS (p.K324T), and isolated hypomyelination (p.Q292K and p.E410K) on microtubule stability and dynamics, neurite outgrowth, dendritic spine development, and kinesin binding. Cellular-based assays reveal that all variants except p.R2G increase microtubule stability, decrease microtubule polymerization rates, reduce axonal outgrowth, and alter the density and shape of dendritic spines. We also find that the p.K324T and p.E410K variants perturb the binding of TUBB4A to KIF1A, a neuron-specific kinesin required for transport of synaptic vesicle precursors. Taken together, our data suggest that impaired microtubule stability and dynamics, defected axonal growth, and dendritic spine development form the common molecular basis of TUBB4A-related leukodystrophy. Impairment of TUBB4A binding to KIF1A is more likely to be involved in the isolated hypomyelination phenotype, which suggests that alterations in kinesin binding may cause different phenotypes. In conclusion, our study extends the spectrum of TUBB4A mutations and related phenotypes and provides insight into why different TUBB4A variants cause distinct clinical phenotypes.

编号：XEY-2022-4-4

引用格式：Wu T, He F, Xiao N, Han Y, Yang L, Peng J. Phenotype-Genotype Analysis Based on Molecular Classification in 135 Children With Mitochondrial Disease. Pediatr Neurol. 2022;132:11-18. doi: 10.1016/j.pediatrneurol.2022.04.013.

通信作者：彭镜

Abstract

Objectives: Over the past decades, mitochondrial disease classification has been mainly based on molecular defects. We aim to analyze phenotype-genotype correlation of mitochondrial disorders according to molecular classification. **Methods:** In this cohort study, we identified 135 individuals diagnosed with mitochondrial disorders, and all patients were divided into four subgroups based on molecular functions: the Respiratory Chain group (including subunits and assembly proteins in the respiratory chain), the Protein Synthesis group (including mitochondrial RNA metabolism, mitochondrial translation), the mitcohindrial DNA (mtDNA) Replication group, and the Others group (including cofactors, homeostasis, substrates, and inhibitors). **Results:** We found that in China, patients with the mtDNA variant constituted a large percentage of mitochondrial disease and were associated with a male preponderance in the Respiratory Chain group, whereas those in the Protein Synthesis group showed a relatively later onset and higher serum lactate level. In contrast, patients with nuclear DNA variants were younger at onset, with no specific lactate or cranial imaging features, especially in the others group, which contained several mitochondrial diseases with corresponding treatment. **Conclusion:** The mtDNA was recommended to detect first in patients with typical lactate and cranial imaging features. A broader consideration and detection are necessary for a better prognosis in an atypical patient.

编号：XEY-2022-4-5

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;43(5):568-581. 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.

编号：XEY-2022-4-6

引用格式：Ma J, Qian Q, Yan S, Dou H, Li C, Sun D. Child-Onset Cerebellar Ataxia Caused by Two Compound Heterozygous Variants in ADPRS Gene: A Case Report. Front Genet. 2022;12:788702. doi: 10.3389/fgene.2021.788702.

通信作者：孙丹

Abstract

Background: Gene variants of ADP-ribosylserine hydrosylase, also known as ADP-ribosylhydrolase-like 2 (ADPRS or ADPRLH2; OMIM: 610624), can cause stress-induced childhood-onset neurodegeneration with variable ataxia and seizures(CONDSIAS, OMIM: 618170), an ultra-rare neurodegenerative autosomal

recessive disorder. ADPRS encodes ADP-ribosylhydrolase 3, which removes poly(ADP-ribose) polymers, whose posttranslational addition occurs under stressful conditions. **Case Presentation:** After a respiratory tract infection, a 30-month-old male patient presented with unsteady gait that rendered walking impossible without external help. Neurological examination revealed acute cerebellar ataxia, electroencephalogram results were abnormal, and brain magnetic resonance imaging revealed slightly widened cerebellar sulci. Laboratory tests showed decreased levels of thyroid-stimulating hormone, and increased levels of plasma lactic acid and serum cardiac enzymes. The cerebrospinal fluid glucose test was positive. Four months after onset, the patient died of sudden convulsions. Using whole exome sequencing, we identified two novel compound heterozygous ADPRS variants: NM_017825.3:c.580C>T (p.Gln194Ter) and NM_017825.3:c.803-1G>A. RNA sequencing indicated that the former mutation might cause nonsense-mediated mRNA decay. The c.803-1G>A variant was found to be a splice-site mutation that leads to the transcriptional retention of intron 5. According to the guidelines of the American College of Medical Genetics and Genomics, the two variants were classified as pathogenic. **Conclusion:** We present the first report of the existence of two compound heterozygous
variants of ADPRS, which leads to CONDSIAS.

编号：XEY-2022-4-7

引用格式：Wang Y, Sun D, Mei Y, Wu S, Li X, Li S, Wang J, Gao L, Xu H, Tuo Y. Population Pharmacokinetics and Dosing Regimen Optimization of Latamoxef in Chinese Children. Pharmaceutics. 2022;14(5):1033.

第一作者：孙丹

Abstract

The present study aimed to establish population pharmacokinetic models of latamoxef, as well as its R- and S-epimers, and generate findings to guide the individualized administration of latamoxef in pediatric patients. A total of 145 in-hospital children

aged 0.08–10.58 years old were included in this study. Three population pharmacokinetic models of latamoxef and its R- and S-epimers were established. The stability and predictive ability of the final models were evaluated by utilizing goodness-of-fit plots, nonparametric bootstrapping, and normalized prediction distribution errors. The final model of total latamoxef was considered as a basis for the dosing regimen. A two-compartment model with first-order elimination best described the pharmacokinetics of total latamoxef. The population typical values of total latamoxef were as follows: central compartment distribution volume (V1) of 4.84 L, peripheral compartment distribution volume (V2) of 16.18 L, clearance (CL) of 1.00 L/h, and inter-compartmental clearance (Q) of 0.97 L/h. Moreover, R-epimer has a higher apparent volume of distribution and lower clearance than S-epimer. Body surface area (BSA) was identified as the most significant covariate to V, CL, and Q. Specific recommendations are given for dosage adjustment in pediatric patients based on BSA. This study highlights that a BSA-normalized dose of latamoxef was required when treating different bacteria to reach the therapeutic target more effectively.

编号：XEY-2022-4-8

引用格式：Xu L, Zhou Y, Ren X, Xu C, Ren R, Yan X, Li X, Yang H, Xu X, Guo X, Sheng G, Hua Y, Yuan Z, Wang S, Gu W, Sun D, Gao F. Expanding the Phenotypic and Genotypic Spectrum of ARFGEF1-Related Neurodevelopmental Disorder. Front Mol Neurosci. 2022;15:862096. doi: 10.3389/fnmol.2022.862096.

通信作者：孙丹

Abstract

Mono-allelic loss-of-function variants in ARFGEF1 have recently caused a developmental delay, intellectual disability, and epilepsy, with varying clinical expressivity. However, given the clinical heterogeneity and low-penetrance mutations of ARFGEF1-related neurodevelopmental disorder, the robustness of the gene-disease association requires additional evidence. In this study, five novel heterozygous

ARFGEF1 variants were identified in five unrelated pediatric patients with neurodevelopmental disorders, including one missense change (c.3539T>G), two canonical splice site variants (c.917-1G>T,c.2850+2T>A), and two frameshift (c.2923_c.2924delCT, c.4951delG) mutations resulting in truncation of ARFGEF1. The pathogenic/likely pathogenic variants presented here will be highly beneficial to patients undergoing genetic testing in the future by providing an expanded reference list of disease-causing variants.

编号：XEY-2022-4-9

引用格式：Huang L, Li H, Zhong J, Yang L, Chen G, Wang D, Zheng G, Han H, Han X, Long Y, Wang X, Liang J, Yu M, Shen X, Fan M, Fang F, Liao J, Sun D. Efficacy and Safety of the Ketogenic Diet for Mitochondrial Disease With Epilepsy: A Prospective, Open-labeled, Controlled Study. Front Neurol. 2022; 13:880944. doi: 10.3389/fneur.2022.880944.

通信作者：孙丹

Abstract

Background: The ketogenic diet (KD) is increasingly used to treat drug-resistant epilepsy because of its favorable effect on seizure reduction. Patients with mitochondrial diseases tend to experience seizures. Therefore, this study aimed to test the efficacy of the KD on participants with mitochondrial diseases in a controlled trial. **Methods:** Participants from fourteen clinical centers who were diagnosed with mitochondrial disease were semi-randomized to either the intervention (KD) or control group. The KD group followed a 3-month KD intervention, while the control group received a 1-month normal diet initially and then a 3-month KD intervention. The primary outcome measure was seizure reduction. Biomarker changes, cognitive impairments, and side effects were also recorded, if available. **Result:** A total of 33 participants were assigned to the KD (n = 22) and control groups (n = 11). In the KD group, 31.8% (7/22) of participants achieved ≥50% seizure reduction after 1 month of

diet intervention, which increased to 40.9%(9/22) at 3 months. In the control group, only 18.2% (2/11) of the participants had ≥50% seizure reduction during the normal diet period. After the control group was transferred to the KD, 63.6% (7/11) of participants had >50% seizure reduction, and this rate increased to 72.7% (8/11) at 3 months. The KD also showed high efficacy in participants with mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) or pathogenic variants in mitochondrial DNA (mtDNA) (90% and 93.3% response rates, respectively). The most frequent side effects reported at the 3-month review were vomiting, cold, hyperlipidemia, and bloating. **Conclusion:** The KD is a safe and effective therapy for seizure control in mitochondrial diseases, especially MELAS and pathogenic variants of mtDNA. KD intervention can be considered in the management of these patients.

编号：XEY-2022-4-10

引用格式：Wang S, Yu Y, Wang X, Deng X, Ma J, Liu Z, Gu W, Sun D. Emerging evidence of genotype-phenotype associations of developmental and epileptic encephalopathy due to KCNC2 mutation: Identification of novel R405G. Front Mol Neurosci. 2022;15:950255. doi: 10.3389/fnmol.2022.950255.

通信作者：孙丹

Abstract

Developmental and epileptic encephalopathies (DEEs) have high genetic heterogeneity, and DEE due to the potassium voltage-gated channel subfamily C member 2 (KCNC2) variant remains poorly understood, given the scarcity of related case studies. We report on two unrelated Chinese patients, an 11-year-old boy and a 5-year-old girl, diagnosed with global developmental delay (GDD), intellectual disability (ID), and focal impaired awareness seizure characterized by generalized spike and wave complexes on electroencephalogram (EEG) in the absence of significant brain lesions. Whole-exome sequencing (WES) and electrophysiological analysis were performed to detect genetic variants and evaluate functional changes of the mutant KCNC2, respectively.

Importantly, we identified a novel gain-of-function KCNC2 variant, R405G, in both patients. Previously reported variants, V471L, R351K, T437A, and T437N, and novel R405G were found in multiple unrelated patients with DEE, showing consistent genotype–phenotype associations. These findings emphasize that the KCNC2 gene is causative for DEE and facilitates treatment and prognosis in patients with DEE due to KCNC2 mutations.

编号：XEY-2022-4-11

引用格式：Yang H, Liao H, Gan S, Xiao T, Wu L. ARHGEF9 gene variant leads to developmental and epileptic encephalopathy: Genotypic phenotype analysis and treatment exploration. Mol Genet Genomic Med. 2022;10(7):e1967. doi: 10.1002/mgg3.1967.

通信作者：吴丽文

Abstract

Background: The ARHGEF9 gene variants have phenotypic heterogeneity, the number of reported clinical cases are limited and the genotype-phenotype relationship is still unpredictable. **Methods:** Clinical data of the patients and their family members were gathered in a retrospective study. The exome sequencing that was performed on peripheral blood samples was applied for genetic analysis. We used the ARHGEF9 gene as a key word to search the PubMed database for cases of ARHGEF9 gene variants that have previously been reported and summarized the reported ARHGEF9 gene variant sites, their corresponding clinical phenotypes, and effective treatment. **Results:** We described five patients with developmental and epileptic encephalopathy caused by ARHGEF9 gene variants. Among them, the antiepileptic treatment of valproic acid and levetiracetam was effective in two cases individually. The exome sequencing results showed five children with point mutations in the ARHGEF9 gene: p.R365H, p.M388V, p.D213E, and p.R63H. So far, a total of 40 children with ARHGEF9 gene variants have been reported. Their main clinical phenotypes include developmental delay, epilepsy,

epileptic encephalopathy, and autism spectrum disorders. The variants reported in the literature, including 22 de novo variants, nine maternal variants, and one unknown variant. There were 20 variants associated with epileptic phenotypes, of which six variants are effective for valproic acid treatment. **Conclusion:** The genotypes and phenotypes of ARHGEF9 gene variants represent a wide spectrum, and the clinical phenotype of epilepsy is often refractory and the prognosis is poor. The p.R365H, p.M388V, p.D213E, and p.R63H variants have not been reported in the current literature, and our stu

编号：XEY-2022-4-12

引用格式：Feng L, Jiao X, Zeng C, Zhao CW, Li R, Zhang L, Yang Z, Liao W, Liu D, Xiao B, Yang Z. Migration characteristics as a prognostic factor in cerebral sparganosis. Int J Infect Dis. 2022;117:28-36. doi: 10.1016/j.ijid.2022.01.005.

第一作者：冯莉

Abstract

Objective: To differentiate diagnostic and prognostic factors from the clinical material of patients with cerebral sparganosis in central South China. **Methods:** Consecutive patients with cerebral sparganosis from our hospital between 2010 and 2018 were retrospectively enrolled. The clinical manifestations, radiographic features, treatment, and outcomes of these patients were analyzed. **Results:** Thirty patients with cerebral sparganosis were included, and foci migration on magnetic resonance imaging was detected in 22 patients, from whom we observed 4 migration modes: interlobar migration (50.0%, 11/22); transmidline migration (27.3%, 6/22); transventricular migration (13.6%, 3/22); and cerebellum-brainstem migration (9.1%, 2/22). The percentage of good outcomes was higher in patients with live worm capture than in those without live worm capture (75.0%, 12/16 vs 33.3%, 2/6). Exposure to preoperative antiparasitic medication was associated with worm migration toward the cortical surface, which led to a higher probability of live worm capture. **Conclusions:**

We propose 4 modes of sparganosis migration that are correlated with worm capture and neurologic prognosis. We found that exposure to antiparasitic medication was associated with worm migration toward the cortical surface, leading to a higher probability of live worm capture. These observations suggest a novel significance for preoperative medication of cerebral sparganosis.

欢迎大家引用上述论文

论文收录说明

- 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；
- 收录的论文为 4-8 月正式发表，未正式发表的论文在收集后将于下一期收录。

编号：HJL-2022-4-1

引用格式：Cheng Z, Su J, Zhang K, Jiang H, Li B. Epigenetic Mechanism of Early Life Stress-Induced Depression: Focus on the Neurotransmitter Systems. Front Cell Dev Biol. 2022 Jul 5;10:929732. doi: 10.3389/fcell.2022.929732.

通讯作者：姜慧轶

Abstract

Depression has an alarmingly high prevalence worldwide. A growing body of evidence indicates that environmental factors signifificantly affect the neural development and function of the central nervous system and then induce psychiatric disorders. Early life stress (ELS) affects brain development and has been identifified as a major cause of depression. It could promote susceptibility to stress in adulthood. Recent studies have found that ELS induces epigenetic changes that subsequently affect transcriptional rates of differentially expressed genes. The epigenetic modififications involved in ELS include histone modififications, DNA methylation, and non-coding RNA. Understanding of these genetic modififications may identify mechanisms that may lead to new interventions for the treatment of depression. Many reports indicate that different types of ELS induce epigenetic modififications of genes involved in the neurotransmitter systems, such as the dopaminergic system, the serotonergic system, the gamma-aminobutyric acid (GABA)-ergic system, and the glutamatergic system, which further regulate gene expression and ultimately induce depression-like behaviors. In this article, we review the effects of epigenetic modififications on the neurotransmitter systems in depression-like outcomes produced by different types of ELS in recent years, aiming to provide new therapeutic targets for patients who suffer from depression.

编号：HJL-2022-4-1

引用格式：Liang J, Wang L, Hao X, Wang G, Wu X. Risk factors and prognosis of spinal cord injury without radiological abnormality in children in China. BMC Musculoskelet Disord. 2022;23(1):428. doi: 10.1186/s12891-022-05393-8.

第一作者：梁建民

Abstract

Background Compared to adults, spinal cord injury without radiographic abnormality (SCIWORA) is more common in children due to the congenital spinal soft tissue elasticity and immature vertebral bodies. In this study, we aimed to investigate the risk factors and prognosis associated with SCIWORA in China. **Method** We retrospectively examined patient records at the First Hospital of Jilin University from January 2007 to December 2020. Patients diagnosed with SCIWORA were included in the study group (n=16). The age, gender, history of trauma, symptoms, injury level of the spinal cord, the American Spinal Injury Association (ASIA) impairment score according to the International Standards for Neurological Classification of Spinal Cord Injury (ISNCSCI), as well as laboratory and imaging findings were analyzed. **Result** The study group included 16 patients with SCIWORA with a mean age of 6.69±2.51 y. The ISNCSCI impairment scale was significantly different between the pre-school age patients (≤7 years old) and school age patients (>7 years old) before (P=0.044) and after therapy (P=0.002). Similarly, magnetic resonance imaging demonstrated a significant difference in the spinal injury level between pre-school age and school age patients (P=0.041). Further, the study group was subdivided into three subgroups according to the cause of trauma: Dance, Taekwondo, or Falls. Magnetic resonance imaging revealed significant differences among the three subgroups (P=0.041). **Conclusion** Compared to school-age patients, pre-school-age patients were more vulnerable to SCIWORA with more severe ISNCSCI scores. Dance and Taekwondo are among the risk factors associated with SCIWORA in Chinese children.

编号：HJL-2022-4-1

引用格式：Kang J, Tian Z, Wei J, Mu Z, Liang J, Li M. Association between obstructive sleep apnea and Alzheimer's disease-related blood and cerebrospinal fluid biomarkers: A meta-analysis. J Clin Neurosci. 2022;102:87-94. doi: 10.1016/j.jocn.2022.06.004.

通讯作者：梁建民

Abstract

Introduction: Recent studies indicate that Alzheimer's disease- (AD) related biomarkers, including amyloid β (Aβ40 and Aβ42) and tau proteins (P-tau and T-tau), in blood and cerebrospinal fluid (CSF) are associated with obstructive sleep apnea (OSA). However, the results have been inconsistent. Therefore, the primary purpose of this meta-analysis was to determine the relationship between blood and CSF AD-related biomarkers and OSA. Methods: We searched the Embase, PubMed, Scopus, and Cochrane Library databases for relevant articles till February 2022. Results: Eight articles were finally included after the literature screening, including 446 patients with OSA and 286 controls. Pooled analysis showed that CSF Aβ42 (SMD = -0.220, P = 0.136), T-tau (SMD = 0.012, P = 0.89), and P-tau (SMD = 0.099, P = 0.274) levels were not different between patients with OSA and controls. In patients with moderate to severe OSA, CSF Aβ42 (SMD = -0.482, P = 0.031) were significantly lower than in controls. Blood T-tau (SMD = 0.560, P = 0.026), P-tau (SMD = 0.621, P < 0.001), and Aβ40 (SMD = 0.656, P < 0.001) levels were significantly higher in patients with OSA than in controls. Blood Aβ42 (SMD = 0.241, P = 0.232) were not different between patients with OSA and controls. Conclusion: OSA is associated with changes in AD-related markers. Higher OSA severity may be associated with the development of AD. AD-related biomarkers, especially in the blood, are clinically efficient, less invasively assessed and monitored, and may be useful for detecting OSA and related cognitive impairments. Further studies are needed to confirm these results.

编号：HJL-2022-4-1

引用格式：Fan Y, Chen D, Wang H, Pan Y, Peng X, Liu X, Liu Y. Automatic BASED scoring on scalp EEG in children with infantile spasms using convolutional neural network. Front Mol Biosci. 2022;9:931688. doi: 10.3389/fmolb.2022.931688

第一作者：范玉颖

Abstract

In recent years, the Burden of Amplitudes and Epileptiform Discharges (BASED) score has been used as a reliable, accurate, and feasible electroencephalogram (EEG) grading scale for infantile spasms. However, manual EEG annotation is, in general, very time-consuming, and BASED scoring is no exception. Convolutional neural networks (CNNs) have proven their great potential in many EEG classification problems. However, very few research studies have focused on the use of CNNs for BASED scoring, a challenging but vital task in the diagnosis and treatment of infantile spasms. This study proposes an automatic BASED scoring framework using EEG and a deep CNN. The feasibility of using CNN for automatic BASED scoring was investigated in 36 patients with infantile spasms by annotating their long-term EEG data with four levels of the BASED score (scores 5, 4, 3, and ≤2). In the validation set, the accuracy was 96.9% by applying a multi-layer CNN to classify the EEG data as a 4-label problem. The extensive experiments have demonstrated that our proposed approach offers high accuracy and, hence, is an important step toward an automatic BASED scoring algorithm. To the best of our knowledge, this is the first attempt to use a CNN to construct a BASED-based scoring model.

欢迎大家引用上述论文

编号：ZWG-2022-4-1

引用格式：Hua Y, Cui D, Han L, Xu L, Mao S, Yang C, Gao F, Yuan Z. A novel SCN9A gene variant identified in a Chinese girl with paroxysmal extreme pain disorder (PEPD): a rare case report. BMC Med Genomics. 2022;15(1):159. doi: 10.1186/s12920-022-01302-z.

通讯作者 袁哲锋

Abstract

Background: Paroxysmal extreme pain disorder (PEPD) is a rare autosomal dominant hereditary disease, characterized by paroxysmal burning pain in the rectum, eyes or mandible and autonomic nervous symptoms, including skin redness and bradycardia. PEPD is a sodium channel dysfunctional disorder caused by SCN9A gene variants. It occurs mainly in Caucasians and only one case has been reported in the Chinese population. Here, we report the second PEPD case in a Chinese indivisual. **Case presentation:** A 2 years and 6 months old girl initially presented with non-epileptic tonic seizures at 7 days after birth. Her clinical symptoms in order of presentation were non-epileptic tonic seizures, harlequin color change and pain. Genetic analysis showed the patient carried a heterozygous variant c.4384T>A (p.F1462I) in the SCN9A gene, which was speculated to cause PEPD symptoms. After administrating carbamazepine, the symptoms were relieved and the patient's condition improved. However, the patient's mother, who carries the same SCN9A variant as her daughter, only showed bradycardia and sinus arrest but no PEPD-related pain. **Conclusions:** This is the second PEPD case reported in the Chinese population. With the discovery of a novel variant in SCN9A, we expanded the genotype spectrum of PEPD. This is the first case suggesting that the clinical presentations of SCN9A-associated PEPD may show inter familial phenotypic diversity. In the future of clinical diagnosis, patients with triggered non-epileptic tonic seizures or pain and harlequin color change should be considered for PEPD and proper and prompt treatment should be given.

编号：ZWG-2022-4-2

引用格式：Zhang GF, Gong WX, Xu ZYR, Guo Y. Alzheimer's disease and epilepsy: The top 100 cited papers. Front. Aging Neurosci. 2022;14:926982. doi: 10.3389/fnagi.2022.926982

通讯作者：郭谊

Abstract

Background: Alzheimer's disease (AD) is one of the common neurodegenerative diseases, which often coexists with epilepsy. It is very significant to study the treatment options and the relationship between AD and epilepsy. **Aims:** The purpose of this study was to analyze the top 100 cited papers about AD and epilepsy using bibliometrics, and to describe the current situation and predict research hot spots. **Methods:** Top 100 papers were obtained from the Web of Science Core Collection (WoSCC). The WoSCC was used to analyze the author, institution, country, title, keywords, abstract, citation, subject category, publication year, impact factor (IF), and other functions. SPSS25 software was used for statistical analysis and CiteSpace V.5.7.R2 was used to visualize the information through collaborative networks. **Results:** The number of publications gradually increased from 2000 to 2021. The total citation count for the top 100 papers ranged from 15 to 433(mean = 67.43). The largest number of papers were published in 2016 (n = 11). Meanwhile, USA (centrality: 0.93) and Columbia University (centrality: 0.06) were the most influential research country and institutions, respectively. The top contributing journals was Journal of Alzheimer's Disease (8%). The IF for journals ranged from 1.819 to 53.44. A network analysis of the author's keywords showed that "beta" (centrality: 0.39), "amyloid beta" (centrality: 0.29), " hyperexcitability" (centrality: 0.29) and "disease" (centrality: 0.29) had a high degree of centrality. **Conclusion:** AD and epilepsy have been intensively studied in the past few years. The relationships, mechanisms and treatment of AD and epilepsy will be subjects of active research hotpots in future. These findings provide valuable information for clinicians

and scientists to identify new perspectives with potential collaborators and cooperative countries.

编号：ZWG-2022-4-3

引用格式：He F, Qiu J, Li H, Guo H, Wang S, Ding Y, Xu S, Wang Z, Feng J, Zhang P, Ding M, Wang S. Efficacy of the ketogenic diet in Chinese adults versus children with drug-resistant epilepsy: A pilot study. Epilepsy Behav. 2022;134:108820. doi: 10.1016/j.yebeh.2022.108820.

通讯作者：王爽

Abstract

Objective: We compared the efficacy and safety of ketogenic diet (KD) therapy as a treatment for Chinese adults versus children with drug-resistant epilepsy. **Methods:** The classic KD was initiated in 19 adults and 29 children with drug-resistant epilepsy. The KD ratio and the dosage of antiseizure medication (ASM) were delicately modulated by the ketogenic team. **Results:** At 12 months after diet initiation, 11 adults (8 on a KD ratio of 3:1 and 3 on a ratio of 2:1) and 20 children (9 on a ketogenic diet ratio of 3:1 and 11 on a ratio of 2:1) remained on the diet. The retention rate for adult KD therapy recipients was 79.0% at 6 months and 57.9% at 12 months after diet initiation, which was not significantly different from the retention rate for children (82.8% at 6 months and 68.9% at 12 months; $P > 0.05$). The efficacy rate of KD therapy (seizure freedom or 50% reduction in seizure frequency) did not significantly differ between adults (63.2%) and children (75.8%, $P = 0.517$). Alleviation of seizure severity was observed in 68.4% of adults and 63.6% of children who were not seizure free on KD therapy. Antiseizure medication was reduced in 34 out of all 48 individuals at the final follow-up. **Conclusion:** Our study demonstrated that KD therapy is a safe and effective treatment for Chinese adults as well as children with drug-resistant epilepsy.

编号：JJJJM-2022-4-1

引用格式：Yu H, Lv Q, Liu Q, Wang S, Ji T, Wang D, Wang W, Wang D, Jiang Y, LiuX, Cai L. Surgical treatment of pediatric intractable frontal lobe epilepsy due to malformation of cortical development. Acta Epileptologica. 20022;4, 23. doi. 10.1186/s42494-022-00090-4

第一作者：于昊

Abstract

Background: Malformation of cortical development (MCD) is a common cause of intractable epilepsy in children. In this study, the effectiveness of frontal lobe epilepsy (FLE) surgery in children with intractable epilepsy due to MCD was assessed and its prognostic factors were studied. **Methods:** Seventy-six patients with intractable FLE who received epilepsy surgery between January 2016 and March 2018 in Peking University First Hospital were recruited in this study. All the resected brain tissues were demonstrated to be MCD. All patients were followed up for at least 3 years. The clinical data and prognosis were analyzed retrospectively. Univariate and multivariate analyses were performed to investigate the correlations between clinical variables and prognostic outcome (Engel classification). **Results:** Sixty (78.9%) patients had Engel class I postoperative outcome. The mean age at surgery was 6.00 ± 4.24 years. Sixty-six patients (86.8%) had daily seizures, 40.2% of the patients had epileptic spasm, and 33% of the patients had extensive interictal EEG abnormalities, which, however, could not provide any helpful information for localizing epileptogenic zones. About 29% of the patients had normal MRI findings even by experienced radiologists, and 26% of the patients had epileptogenic lesion involving adjacent lobes. There was a significant correlation between acute postoperative seizure (APOS) and prognosis ($P < 0.05$): APOS predicted poor prognosis. There was a significant correlation between pathology and prognosis ($P < 0.05$): FCD IA and FCD IIB were correlated with a good outcome. Both variables with a significance level of $P < 0.05$ during univariate analysis, including pathology and APOS, were included in multivariate analysis, which were significant

independent predictors of prognosis. **Conclusions:** The clinical manifestations of pediatric intractable FLE due to MCD are more complicated than those in adults. Multidisplinary presurgical evaluation in pediatric epilepsy is mandatory. The surgical outcome of pediatric FLE due to MCD could reach a seizure-free rate of 78.9% with the follow-up of at least 3 years. The post-operative pathology and APOS may be related to the prognosis of surgery in this group of pediatric patients.

编号：JJJJM-2022-4-2

引用格式：Li T, Luan G, Zhou J. Correlation Between Ictal Signs and Anatomical Subgroups in Temporal Lobe Seizures: A Stereoelectroencephalography Study. Front Neurol. 2022;13:917079. doi: 10.3389/fneur.2022.917079.

通讯作者 周健

Abstract

Objective: Ictal semiology is a fundamental part of the presurgical evaluation of patients with temporal lobe epilepsy. We aimed to identify different anatomical and semiologic subgroups in temporal lobe seizures, and investigate the correlation between them. **Methods:** We enrolled 93 patients for whom stereoelectroencephalography exploration indicated that the seizure-onset zone was within the temporal lobe. Ictal signs and concomitant stereoelectroencephalography changes were carefully reviewed and quantified, and then cluster analysis and the Kendall correlation test were used to associate ictal signs with the temporal structures of patients. **Results:** Clustering analysis identified two main groups of temporal structures. Group 1 consisted of the medial temporal lobe structures and the temporal pole, which were divided into two subgroups. Group 1A included the hippocampal head, hippocampal body, and amygdala, and this subgroup correlated significantly with oroalimentary automatisms, feeling of fear, and epigastric auras. Group 1B included the hippocampal tail, temporal pole, and parahippocampal gyrus, and this subgroup correlated significantly with manual and oroalimentary automatisms. Group 2 consisted of the cortical structures of the temporal

lobe and was also divided into two subgroups. Group 2A included the superior and middle temporal gyrus, correlated significantly with bilateral rictus/facial contraction, generalized tonic-clonic seizure, and manual automatisms. Group 2B included Heschl's gyrus, the inferior temporal gyrus, and the fusiform gyrus, and this subgroup correlated significantly with auditory auras, focal hypokinetics, unilateral upper and lower limbs tonic posture/clonic signs, head/eye deviation, unilateral versive signs, and generalized tonic-clonic seizure. **Significance:** The temporal structures can be categorized according to the level at which each structure participates in seizures, and different anatomical subgroups can be correlated with different ictal signs. Identifying specific semiologic features can help us localize the epileptogenic zone and thus develop stereoelectroencephalography electrode implantation and surgical resection protocols for patients with temporal lobe epilepsy.

编号：JJJJM-2022-4-3

引用格式：Mo J, Wang Y, Zhang J, Cai L, Liu Q, Hu W, Sang L, Zhang C, Wang X, Shao X, Zhang K. Metabolic phenotyping of hand automatisms in mesial temporal lobe epilepsy. EJNMMI Res. 2022;12(1):32. doi: 10.1186/s13550-022-00902-1.

通讯作者 张凯

Abstract

Purpose: Hand automatisms (HA) are common clinical manifestations in mesial temporal lobe epilepsy. However, the location of the symptomatogenic zone (EZ) in HA as well as the networks involved, are still unclear. To have a better understanding of HA underlying mechanisms, we analyzed images from interictal [^{18}F] fluorodeoxyglucose-positron emission tomography (FDG-PET) in patients with mesial temporal lobe epilepsy (mTLE). **Methods:** We retrospectively recruited 79 mTLE patients and 18 healthy people that substituted the control group for the analysis. All patients underwent anterior temporal lobectomy and were seizure-free. Based on the semiology of the HA occurrence, the patients were divided into three subgroups:

patients with unilateral HA (Uni-HA), with bilateral HA (Bil-HA) and without HA (None-HA). We performed the intergroup comparison analysis of the interictal FDG-PET images and compared the functional connectivity within metabolic communities. **Results:** Our analysis showed that the metabolic patterns varied among the different groups. The Uni-HA subgroup had significant differences in the extratemporal lobe brain areas, mostly in the ipsilateral supplementary motor area (SMA) and middle cingulate cortex (MCC) when compared to the healthy control group. The Bil-HA subgroup demonstrated that the bilateral SMA and MCC areas were differentially affected, whereas in the None-HA subgroup the differences were evident in limited brain areas. The metabolic network involving HA showed a constrained network embedding the SMA and MCC brain regions. Furthermore, the increased metabolic synchronization between SMA and MCC was significantly correlated with HA. **Conclusion:** The metabolic pattern of HA was most conspicuous in SMA and MCC brain regions. Increased metabolic synchronization within SMA and MCC was considered as the major EZ of HA.

编号：JJJJM-2022-4-4

引用格式：Mo J, Zhang J, Hu W, Shao X, Sang L, Zheng Z, Zhang C, Wang Y, Wang X, Liu C, Zhao B, Zhang K. Neuroimaging gradient alterations and epileptogenic prediction in focal cortical dysplasia IIIa. J Neural Eng. 2022;19(2). doi: 10.1088/1741-2552/ac6628.

通讯作者 张凯

Abstract

Objective: Focal cortical dysplasia type IIIa (FCD IIIa) is a highly prevalent temporal lobe epilepsy but the seizure outcomes are not satisfactory after epilepsy surgery. Hence, quantitative neuroimaging, epileptogenic alterations, as well as their values in guiding surgery are worth exploring. **Approach**: We examined 69 patients with pathologically verified FCD IIIa using multimodal neuroimaging and stereoelectroencephalography

(SEEG). Among them, 18 received postoperative imaging which showed the extent of surgical resection and 9 underwent SEEG implantation. We also explored neuroimaging gradient alterations along with the distance to the temporal pole. Subsequently, the machine learning regression model was employed to predict whole-brain epileptogenicity. Lastly, the correlation between neuroimaging or epileptogenicity and surgical cavities was assessed. **Main results:** FCD IIIa displayed neuroimaging gradient alterations on the temporal neocortex, morphology-signal intensity decoupling, low similarity of intra-morphological features and high similarity of intra-signal intensity features. The support vector regression model was successfully applied at the whole-brain level to calculate the continuous epileptogenic value at each vertex (mean-squared error = 13.8 ± 9.8). **Significance:** Our study investigated the neuroimaging gradient alterations and epileptogenicity of FCD IIIa, along with their potential values in guiding suitable resection range and in predicting postoperative seizure outcomes. The conclusions from this study may facilitate an accurate presurgical examination of FCD IIIa. However, further investigation including a larger cohort is necessary to confirm the results.

编号：JJJJM-2022-4-5

引用格式：胡文瀚, 姚远, 莫嘉杰, 张弼, 王秀, 刘畅, 赵宝田, 张建国, 张凯. 磁共振引导下激光间质热疗治疗药物难治性癫痫的疗效分析. 癫痫杂志, 2022, 8(3): 202-206. doi: 10.7507/2096-0247.202202003

第一作者 胡文瀚　　通讯作者 张凯

摘要

目的 探究磁共振引导下激光间质热疗（Magnetic resonance-guided laser interstitial thermal therapy，MRgLITT）对药物难治性癫痫患者的治疗效果。**方法** 回顾性分析 2020 年 8 月—2021 年 2 月首都医科大学附属北京天坛医院神经外科收治的经 MRgLITT 的药物难治性癫痫患者的临床资料，包括患者的病变类型、术后并发

症及术后 1 年的 Engel 分级。**结果** 共纳入 55 例患者，其中男 27 例、女 28 例，平均（21.7 ± 14.1）岁，全部患者均顺利完成手术且均进行了术后 1 年的随访。纳入包括颅内肿瘤、下丘脑错构瘤、局灶性皮质发育不良、海绵状血管畸形，颞叶癫痫以及行胼胝体切开的患者，术后达到无发作（Engel I）的患者占 59.6%（31/52），姑息性手术平均缓解率为 68.6%。术后并发症主要包括神经功能缺损 6 例（10.9%）、出血 4 例（7.3%，1 例为症状性出血）和非感染性发热 2 例（3.6%），无严重和长期并发症。术后平均住院（4.7±1.6）天，术后恢复快。**结论** MRgLITT 技术逐渐成熟，具有广泛的适应症，对于恰当选择的病例，疗效可与切除性手术接近。该技术为药物难治性癫痫患者提供了一种安全及有效的治疗方式。

编号：JJJJM-2022-4-6

引用格式：Liang S, Fan X, Chen F, Liu Y, Qiu B, Zhang K, Qi S, Zhang G, Liu J, Zhang J, Wang J, Wang X, Song Z, Luan G, Yang X, Jiang R, Zhang H, Wang L, You Y, Shu K, Lu X, Gao G, Zhang B, Zhou J, Jin H, Han K, Li Y, Wei J, Yang K, You G, Ji H, Jiang Y, Wang Y, Lin Z, Li Y, Liu X, Hu J, Zhu J, Li W, Wang Y, Kang D, Feng H, Liu T, Chen X, Pan Y, Liu Z, Li G, Li Y, Ge M, Fu X, Wang Y, Zhou D, Li S, Jiang T, Hou L, Hong Z. Chinese guideline on the application of anti-seizure medications in the perioperative period of supratentorial craniocerebral surgery. Ther Adv Neurol Disord. 2022 Aug 16;15:17562864221114357. doi: 10.1177/17562864221114357.

第一作者：梁树立　樊星　刘永红 张凯　通讯作者 梁树立　李世绰　洪震

Abstract

Seizures are a common symptom of craniocerebral diseases, and epilepsy is one of the comorbidities of craniocerebral diseases. However, how to rationally use anti-seizure medications (ASMs) in the perioperative period of craniocerebral surgery to control or avoid seizures and reduce their associated harm is a problem. The China Association Against Epilepsy (CAAE) united with the Trauma Group of the Chinese Neurosurgery

Society, Glioma Professional Committee of the Chinese Anti-Cancer Association, Neuro-Oncology Branch of the Chinese Neuroscience Society, and Neurotraumatic Group of Chinese Trauma Society, and selected experts for consultancy regarding outcomes from evidence-based medicine in domestic and foreign literature. These experts referred to the existing research evidence, drug characteristics, Chinese FDA-approved indications, and expert experience, and finished the current guideline on the application of ASMs during the perioperative period of craniocerebral surgery, aiming to guide relevant clinical practice. This guideline consists of six sections: application scope of guideline, concepts of craniocerebral surgery-related seizures and epilepsy, postoperative application of ASMs in patients without seizures before surgery, application of ASMs in patients with seizures associated with lesions before surgery, emergency treatment of postoperative seizures, and 16 recommendations.

编号：JJJJM-2022-4-7

引用格式：Zhang S, Chen F, Zhai F, Liang S. Role of HMGB1/TLR4 and IL-1β/IL-1R1 Signaling Pathways in Epilepsy. Front Neurol. 2022;13:904225. doi: 10.3389/fneur.2022.904225.

通讯作者 梁树立

Abstract

Epilepsy is a chronic disorder of the nervous system characterized by recurrent seizures. Inflammation is one of the six major causes of epilepsy, and its role in the pathogenesis of epilepsy is gaining increasing attention. Two signaling pathways, the high mobility group box-1 (HMGB1)/toll-like receptor 4 (TLR4) and interleukin-1β (IL-1β)/interleukin-1 receptor 1 (IL-1R1) pathways, have become the focus of research in recent years. These two signaling pathways have potential as biomarkers in the prediction, prognosis, and targeted therapy of epilepsy. This review focuses on the association between epilepsy and the neuroinflammatory responses mediated by these two signaling pathways. We hope to contribute further in-depth studies on the role of

HMGB1/TLR4 and IL-1β/IL-1R1 signaling in epileptogenesis and provide insights into the development of specific agents targeting these two pathways.

编号：JJJJM-2022-4-8

引用格式： Yuan L, Wang Y, Cheng S, Zhang J, Zhang S, Liu T, Zhang G, Liang S. Interictal Discharge Pattern in Preschool-Aged Children With Tuberous Sclerosis Complex Before and After Resective Epilepsy Surgery. Front Neurol. 2022 May 31;13:868633. doi: 10.3389/fneur.2022.868633.

通讯作者 梁树立

Abstract

Objective: To analyze the interictal discharge (IID) patterns on pre-operative scalp electroencephalogram (EEG) and compare the changes in IID patterns after removal of epileptogenic tubers in preschool children with tuberous sclerosis complex (TSC)-related epilepsy. **Methods:** Thirty-five preschool children who underwent resective surgery for TSC-related epilepsy were enrolled retrospectively, and their EEG data collected before surgery to 3 years after surgery were analyzed. **Results:** Twenty-three (65.7%) patients were seizure-free post-operatively at 1-year follow-up, and 37-40% of post-operative patients rendered non-IID on scalp EEGs, and patients with focal IIDs or generalized IID patterns on pre-operative EEG presented a high percentage of normal post-operative scalp EEGs. IID patterns on pre-operative scalp EEGs did not influence the outcomes of post-operative seizure controls, while patients with non-IID and focal IID on post-operative EEGs were likely to achieve post-operative seizure freedom. Patients with new focal IIDs presented a significantly lower percentage of seizure freedom than those without new focal IIDs on post-operative EEGs at 3-year follow-up. **Conclusion:** Over 1/3 children with TSC presented normal scalp EEGs after resective epileptsy surgery. Patients with post-operative seizure freedom were more likely to have non-IIDs on post-operative EEGs. New focal IIDs were negative factors for seizure freedom at the 3-year follow-up.

编号：JJJJM-2022-4-9

引用格式： **Wang Y, Xu J, Liu T, Chen F, Chen S, Yuan L, Zhai F, Liang S. Diagnostic value of high-frequency oscillations for the epileptogenic zone: A systematic review and meta-analysis. Seizure. 2022;99:82-90. doi: 10.1016/j.seizure.2022.05.003.**

通讯作者 梁树立

Abstract

Background: Delineation of the epileptogenic zone (EZ) is crucial during resective surgery in patients with epilepsy. In recent years, high-frequency oscillations, including fast ripples and ripples, have been considered promising biomarkers of the EZ. The objective of this study was to perform a systematic review and meta-analysis to evaluate the diagnostic accuracy of fast ripples and ripples in identifying EZs. **Methods:** The PubMed, Embase, and Cochrane databases were searched systematically. The review process followed the Preferred Reporting Items for Systematic Reviews and Meta-analyses guidelines. A bivariate mixed-effects regression approach was used to obtain summary estimates of the sensitivity and specificity of fast ripples and ripples. **Results:** Thirteen studies were included in this review (12 for fast ripples and eight for ripples). Fast ripples had a pooled sensitivity of 0.80 (95% confidence interval [CI], 0.62-0.91), a specificity of 0.72 (95% CI, 0.55-0.85), and a summary area under the curve of 0.82 (95% CI, 0.79-0.86). The threshold effect was detected for ripples, and the summary area under the curve was 0.75 (95% CI, 0.71-0.79), with a sensitivity of 0.38 (95% CI, 0.18-0.64) and specificity of 0.90 (95% CI, 0.70-0.97). **Conclusions:** Fast ripples are a biomarker of the EZ with moderate diagnostic accuracy; in contrast, ripples are not as effective.

编号：JJJJM-2022-4-10

引用格式： **Chen F, Zhang S, Liu T, Yuan L, Wang Y, Zhang G, Liang S.**

Preliminary study on pathogenic mechanism of first Chinese family with PNKD. Transl Neurosci. 2022;13(1):125-133. doi: 10.1515/tnsci-2022-0222.

通讯作者 梁树立

Abstract

Background: The first Chinese family with paroxysmal non-kinesigenic dystonia (PNKD) was confirmed to harbour a *PNKD* mutation. However, the pathogenic mechanism of the PNKD-causing gene mutation was unclear. **Methods:** Wild-type and mutant *PNKD-L* plasmids were prepared and transfected into the C6 cell line to study the distribution and stability of PNKD protein in C6 cells and its effect on the glutathione content. The blood and cerebrospinal fluid (CSF) of 3 PNKD patients and 3 healthy controls were collected. The differentially expressed proteins were identified using isobaric tags for relative and absolute quantitation. Furthermore, Gene Ontology (GO) and Kyoto Encyclopaedia of Genes and Genomes (KEGG) enrichment analyses were performed, and the protein-protein interaction network was constructed. **Results:** Wild-type PNKD protein was mainly distributed in the membranes, whereas mutant PNKD protein was distributed throughout the C6 cells. After transfection with mutant *PNKD-L* plasmid, the glutathione content decreased significantly in C6 cells; the stability of the mutant PNKD protein was significantly low. There were 172 and 163 differentially expressed proteins in CSF and plasma, respectively, of PNKD patients and healthy controls. For these proteins, blood microparticle and complex activation (classical pathway) were the common GO enrichment term, and complex and coordination cascade pathway were the common KEGG enrichment pathway. Recombinant mothers against decapentaplegic homolog 4 (SMAD4) was one of the differentially expressed proteins; it exhibited a relationship with the aforementioned enrichment GO terms and KEGG pathway. **Conclusion:** PNKD protein was mainly distributed in cell membranes. *PNKD-L* mutation affected subcellular localisation, PNKD protein stability, and glutathione content. SMAD4 was found to be a potential biomarker for PNKD diagnosis.

编号：JJJJM-2022-4-11

引用格式：季涛云，王若凡，刘庆祝，王爽，于昊，刘畅，孙宇，王文，于国静，王东明，吴晔，姜玉武，刘晓燕，蔡立新. 21 例 Rasmussen 脑炎的临床特征、治疗及预后. 癫痫杂志, 2022, 8(5): 400-406. doi: 10.7507/2096-0247.202205012

第一作者：季涛云

摘要

目的 总结经手术治疗的 Rasmussen 脑炎（Rasmussen encephalitis，RE）的临床表型、电生理特点、影像学特点、治疗及预后，提高对此疾病的认识。

方法 回顾性分析 2014 年 10 月—2019 年 10 月于北京大学第一医院儿童癫痫中心行半球离断术的 21 例 RE 患者资料。收集人口学数据、起病年龄、起病方式、癫痫发作形式、脑电图特点、头颅影像学特点、手术及预后。**结果** 共收集 21 例患者，其中男 8 例、女 13 例，起病年龄（5.0±2.0）岁，手术时年龄（6.9±2.7）岁，手术时病程（1.7±1.3）年。20 例（20/21，95.2%）患者以局灶运动性发作起病，10 例（10/21，47.6%）患者有 2~3 种表现形式的局灶运动性发作。15 例（15/21，71.4%）患者有部分性癫痫持续状态（Epilepsia partialis continua，EPC），出现于病程（0.7±0.6）年。所有患者均有偏瘫，偏瘫出现于（0.9±0.6）年。脑电图显示所有患者受累半球节律减慢，其中有 4 例患者健侧半球随着病情的进展节律亦减慢。所有患者发作间期均有受累半球侧癫痫样放电，其中 6 例患者健侧亦有独立的癫痫样放电。21 例患者均行半球离断术。术后随访时间 2～7 年，所有患者均达到 Engel Ⅰ级。神经功能损害均较术前有不同程度恢复。**结论** RE 多发生于学龄期前后。局灶运动性发作是首发且最为常见的症状。随着病情的进展，发作形式呈现多样化，出现 EPC、偏瘫及认知功能下降。脑电图以患侧半球慢波及放电为主要表现，虽然部分患者亦可双侧受累，但明显不对称。通过手术治疗，患者在癫痫发作及发育方面均获得了良好的疗效。

编号：YGQM-2022-4-1

引用格式：Wang J, Wang J, Lu X, Song W, Luo S, Zou D, Hua L, Peng Q, Tian Y, Gao L, Liao W, He N. Recessive PKD1 Mutations Are Associated With Febrile Seizures and Epilepsy With Antecedent Febrile Seizures and the Genotype-Phenotype Correlation. 2022; 10;15:861159. doi: 10.3389/fnmol.2022.861159.

通讯作者 何娜

Abstract

Objective The *PKD1* encodes polycystin-1, a large transmembrane protein that plays important roles in cell proliferation, apoptosis, and cation transport. Previous studies have identified *PKD1* mutations in autosomal dominant polycystic kidney disease (ADPKD). However, the expression of *PKD1* in the brain is much higher than that in the kidney. This study aimed to explore the association between *PKD1* and epilepsy. **Methods** Trios-based whole-exome sequencing was performed in a cohort of 314 patients with febrile seizures or epilepsy with antecedent febrile seizures. The damaging effects of variants was predicted by protein modeling and multiple *in silico* tools. The genotype-phenotype association of *PKD1* mutations was systematically reviewed and analyzed. **Results** Eight pairs of compound heterozygous missense variants in *PKD1* were identified in eight unrelated patients. All patients suffered from febrile seizures or epilepsy with antecedent febrile seizures with favorable prognosis. All of the 16 heterozygous variants presented no or low allele frequencies in the gnomAD database, and presented statistically higher frequency in the case-cohort than that in controls. These missense variants were predicted to be damaging and/or affect hydrogen bonding or free energy stability of amino acids. Five patients showed generalized tonic-clonic seizures (GTCS), who all had one of the paired missense mutations located in the PKD repeat domain, suggesting that mutations in the PKD domains were possibly associated with GTCS. Further analysis demonstrated that monoallelic mutations with haploinsufficiency of *PKD1* potentially caused kidney disease, compound heterozygotes with superimposed effects of two missense mutations were associated

with epilepsy, whereas the homozygotes with complete loss of *PKD1* would be embryonically lethal. **Conclusions** *PKD1* gene was potentially a novel causative gene of epilepsy. The genotype-phenotype relationship of *PKD1* mutations suggested a quantitative correlation between genetic impairment and phenotypic variation, which will facilitate the genetic diagnosis and management in patients with *PKD1* mutations.

编号：YGQM-2022-4-2

引用格式： Wang Y, Li, C, XiongZ, Chen N, Wang X, Xu J, Wang Y, Liu L, Wu H, Huang C, Huang A, Tan J, Li Y, Li Q. Up-and-coming anti-epileptic effect of aloesone in Aloe vera: Evidenced by integrating network pharmacological analysis, in vitro, and in vivo models. Front Pharmacol.2022;13:962223. doi:10.3389/fphar.2022.962223

通讯作者 李其富

Abstract

Background: Aloe vera is a medically valuable plant with anti-epileptic activity; however, its mechanism of action remains unknown. In this study, network pharmacological, in vitro, and in vivo experiments were carried out to explore the potential anti-epileptic components and targets of Aloe vera. **Methods:** The main active components of Aloe vera were identified by searching the Traditional Chinese Medicine System Pharmacology database. Targets of Aloe vera were predicted using SwissTargetPrediction, whereas information about the epilepsy disease targets was obtained from Gene Cards. The protein-protein interaction network and core targets were screened according to the topological structure and CytoNCA plugin. The glutamate-induced HT22 cell line and pentylenetetrazol-induced seizure rats were used to confirm the effect of aloesone by detecting reactive oxygen species (ROS) and apoptosis, and predicting the targets. **Results:** A total of 14 core active components were selected based on the screening criteria of oral bioavailability ≥30% and drug-likeness ≥ 0.10. Four compounds, namely linoleic acid, aloesone, isoeleutherol

glucosiden qt, and anthranol, demonstrated the potential ability of crossing the blood-brain barrier. A total of 153 targets associated with epilepsy were predicted for the four compounds. Moreover, after network analysis with CytoNCA, 10 targets, namely, MAPK1, SRC, MARK3, EGFR, ESR1, PTGS2, PTPN11, JAK2, PPKCA, and FYN, were selected as the core genes, and SRC, which has been predicted to be the target of aloesone and anthranol, exhibited the highest subgraph centrality value. In vitro experiments confirmed that aloesone treatment significantly inhibited the glutamate-induced neuronal injury by reducing the intracellular ROS content and the early phase of apoptosis. Additionally, treatment with 50 mg/kg aloesone resulted in anti-seizure effects by reducing the seizure score and prolonging the latent period in acute and chronic rats. Furthermore, aloesone treatment increased the phosphorylation of c-SRC at Y418 and reduced the phosphorylation at Y529, simultaneously activating c-SRC. **Conclusion:** Integrating network pharmacology with in vitro and in vivo experiments demonstrated that aloesone, which inhibited seizure by activating c-SRC, is a potential anti-seizure compound present in Aloe vera.

编号：YGQM-2022-4-3

引用格式： Tong J, Zou Q, Wang S, Liu J, Chen R, Chen Y, Zhao W, Ma L, Li Q. Human herpesvirus 6B infection in mesial temporal lobe epilepsy: a meta-analysis. Acta Epileptologica.2022;4:9. doi.org/10.1186/s42494-022-00083-3.

通讯作者 李其富

Abstract

Background: Whether human herpesvirus 6B (HHV-6B) can affect mesial temporal lobe epilepsy (MTLE) remains controversial. The present meta-analysis was aimed to evaluate whether HHV-6B is significantly associated with MTLE. **Methods:** Six studies were included in this meta-analysis, comprising 183 MTLE patients and 75 controls. In these studies, HHV-6B infection in astrocytes and brain samples of MTLE patients and controls was investigated by polymerase chain reaction and

immunofluorescence. **Results:** The frequency of HHV-6B infection detection is significantly higher in the MTLE group than in the control group (OR = 9.42, 95%CI: 3.66 – 24.25), P < 0.00001). Although febrile convulsion is strongly associated with MTLE, the formation of febrile convulsion leading to MTLE is not associated with HHV-6B infection (OR = 2.68, 95%CI: 0.93 – 7.73), P = 0.07). Moreover, the HHV-6B-specific antigen is co-localized to cells positive for GFAP that morphologically resemble astrocytes. HHV-6B mainly infects astrocytes, oligodendrocytes and microglia, and could damage the vascular endothelial cells of the central nervous system. Conclusions: There is an association between HHV-6B infection and MTLE. Future large-scale, multi-center, controlled, prospective studies are required to confirm these findings. In addition, the exact mechanism underlying the effects of HHV-6B infection on MTLE needs to be further investigated.

编号：YGQM-2022-4-4

引用格式：Fan X, Guo Q, Zhang X, Fei L, He S, Weng X. Top-down modulation and cortical-AMG/HPC interaction in familiar face processing. Cerebral Cortex, 2022,;15(1):159. doi: 10.1093/cercor/bhac371.

第一作者：郭强

Abstract

Humans can accurately recognize familiar faces in only a few hundred milliseconds, but the underlying neural mechanism remains unclear. Here, we recorded intracranial electrophysiological signals from ventral temporal cortex (VTC), superior/middle temporal cortex (STC/MTC), medial parietal cortex (MPC), and amygdala/hippocampus (AMG/HPC) in 20 epilepsy patients while they viewed faces of famous people and strangers as well as common objects. In posterior VTC and MPC, familiarity-sensitive responses emerged significantly later than initial face-selective responses, suggesting that familiarity enhances face representations after they are first being extracted. Moreover, viewing famous faces increased the coupling between

cortical areas and AMG/HPC in multiple frequency bands. These findings advance our understanding of the neural basis of familiar face perception by identifying the top-down modulation in local face-selective response and interactions between cortical face areas and AMG/HPC.

编号：YGQM-2022-4-5

引用格式： Lin S, Long W, Wen J, Su Q, Liao J, Hu Z. Myelin oligodendrocyte glycoprotein antibody-associated aseptic meningitis without neurological parenchymal lesions: A novel phenotype. Mult Scler Relat Disord. 2022 23;68:104126. doi: 10.1016/j.msard.2022.104126.

第一作者/通讯作者：林素芳

Abstract

Background: Myelin oligodendrocyte glycoprotein (MOG) antibodies mediate inflammatory demyelinating dis- eases of the central nervous system. This study aimed to understand the clinical characteristics of MOG antibody- associated aseptic meningitis (MOGAM).

Methods: Here, we report the cases of two children with MOGAM. A systematic literature review was conducted and included patients who had MOGAM only, without neurological parenchymal lesions. The clinical charac- teristics that may have affected the outcome were statistically analyzed.

Results: We reviewed 12 cases of MOGAM; male: female = 9: 3. Prolonged fever lasting over 7 days (11/12) was the most frequent symptom, followed by headache (10/12), vomiting (5/12), and seizures (4/12). None of the patients had focal neurological manifestations or parenchymal lesions on imaging. Cerebrospinal fluid (CSF) leukocytosis was observed in all patients (12/12), and blood leukocytosis and elevated CSF pressure was observed in all patients who had corresponding results (9/9 and 4/4, respectively). Seizures occurrence was lower than that of MOG antibody-associated cortical encephalitis. Seven cases progressed to other MOG antibody-

associated diseases (MOGADs) in the later phase of MOGAM. Patients who did not progress to other MOGADs had a shorter disease duration from onset to the initiation of intravenous methylprednisolone than those who did. All the patients achieved full recovery after steroid treatment. One patient had relapses. Conclusions: MOGAM without inflammatory demyelination is a rare but distinct phenotype of MOGAD, with fewer clinical manifestations mimicking bacterial or viral meningitis/encephalomeningitis. Delayed diagnosis and treatment may induce the progression to other severe MOGADs. Early recognition of this unique autoim- mune aseptic meningitis may contribute to early diagnosis, treatment, and better outcomes.

编号：YGQM-2022-4-6

引用格式： Li L, Lin S, Tan Z, Chen L, Zeng Q, Sun Y, Li C, Liu Z, Lin C, Ren X, Zhang T, Li Y, Su Q, Li Y, Cao D, Liao J, Zhu F, Chen Y. Resective epilepsy surgery for West syndrome: The Hypsarrhythmic Asymmetric Scoring Scheme is a determining predictor of seizure outcome. Seizure. 2022;101:205-210. doi: 10.1016/j.seizure.2022.08.011.

第一作者：林素芳

Abstract

Objective: It has been suggested that asymmetric hypsarrhythmia is associated with structural etiology. We devised the Hypsarrhythmic Asymmetric Scoring Scheme (HASS) to quantify the degree of hypsarrhythmic asymmetry in a retrospective series of patients who underwent surgical treatment at our center. The present study aimed to investigate the role of HASS in predicting the postsurgical seizure outcomes. **Methods:** We retrospectively analyzed the records of 46 children with hypsarrhythmia who underwent resective epilepsy surgery between 2018 and 2020 and were followed up for at least 1 year after surgery. Hypsarrhythmia severity in each hemisphere was quantified and scored. The HASS score was calculated as the difference between the two hemispheres. Univariate results were submitted to logistic regression models to identify

independent predictors for favorable surgical outcomes. **Results:** Of the 46 patients who underwent resective surgery, Engel's class I-II outcomes were achieved in 34 (73.9%). The Engel I-II group had a significantly higher HASS score than the Engel III-IV group ($p<0.001$). Multivariate analysis showed that the HASS score was the only significant predictor of good outcomes ($p = 0.011$). Further receiver operating characteristic analysis showed that a threshold of 7 yielded a better seizure outcome with a sensitivity of 97.06% and specificity of 83.33%. **Significance:** As the first hypsarrhythmia scoring system specially designed for presurgical evaluation, the HASS score may contribute to predicting the postsurgical seizure outcome from the electroencephalography perspective.

《医疗机构工作人员廉洁从业九项准则》

1. **合法按劳取酬，不接受商业提成；**
2. **严守诚信原则，不参与欺诈骗保；**
3. **依据规范行医，不实施过度诊疗；**
4. **遵守工作规程，不违规接受捐赠；**
5. **恪守保密准则，不泄露患者隐私；**
6. **服从诊疗需要，不牟利转介患者；**
7. **维护诊疗秩序，不破坏就医公平；**
8. **共建和谐关系，不收受患方红包；**
9. **恪守交往底线，不收受企业回扣。**

编号：CYDQZ-2022-4-1

引用格式： Li S, Cai X, Yao C, Wang Y, Xiao X, Yang H, Yao Y, Chen L. Case Report: Late-Onset Lennox-Gastaut Syndrome Treated With Stereotactic Electroencephalography-Guided Radiofrequency Thermocoagulation Before Craniotomy. Front Neurol. 2022;13:857767. doi: 10.3389/fneur.2022.857767.

通讯作者 陈蕾

Abstract

The onset of Lennox-Gastaut syndrome (LGS), a severe epilepsy syndrome, is typically before 8 years of age. Late-onset LGS (with onset in adolescence and adulthood) is relatively rare clinically and has some differences from classical LGS. Herein, we describe the case of a patient with late-onset LGS and provide a literature review of such cases. The patient had focal epilepsy onset at 8 years of age. After a 9-year evolution, he suffered seizures of different types and had a diagnosis of late-onset LGS. Drug treatment was ineffective. Nothing was found on stereotactic electroencephalography (SEEG) and magnetic resonance imaging (MRI) during the course of the disease. After the second presurgical evaluation, we found a suspicious focus on high-resolution structural MRI which was verified by SEEG at last. After SEEG-guided radiofrequency thermocoagulation (RFTC), his seizures were controlled, and his cognitive function and quality of living clearly improved. However, his seizures recurred 2 years later, and he underwent left occipital resection. Thereafter, his seizures have been controlled until now. This case emphasizes the importance of high-resolution structural MRI in the treatment of LGS. Furthermore, it suggests that late-onset LGS may be caused by focal lesions and evolve from focal epilepsy. Thus, characterizing the clinical symptoms and performing individualized electroencephalographic follow-up are both very important. Additionally, the clinical outcome in this case implies the value and limitations of RFTC in patients with epilepsy and a clear focal lesion. Moreover, this case further supports differences between late-onset and classical LGS in terms of clinical manifestation, cognitive changes, prognosis, and treatment.

编号：CYDQZ-2022-4-2

引用格式：Lai W, Du D, Chen L. Metabolomics Provides Novel Insights into Epilepsy Diagnosis and Treatment: A Review. Neurochem Res. 2022;47(4):844-859. doi: 10.1007/s11064-021-03510-y.

通讯作者：陈蕾

Abstract

Epilepsy is one of the most common diseases of the central nervous system. The diagnosis of epilepsy mainly depends on electroencephalograms and symptomatology, while diagnostic biofuid markers are still lacking. In addition, approximately 30% of patients with epilepsy (PWE) show a poor response to the currently available anti-seizure medicines. An increasing number of studies have reported alterations in the blood, brain tissue, cerebrospinal fuid and urine metabolome in PWE and animal models of epilepsy. The aim of this review was to identify potential metabolic biomarkers and pathways that might facilitate diagnostic, therapeutic and prognostic determination in PWE and the understanding of the pathogenesis of the disease. The PubMed and Embase databases were searched for metabolomic studies of PWE and epileptic models published before December 2020. The study objectives, types of models and reported diferentially altered metabolites were examined and compared. Pathway analyses were performed using MetaboAnalyst 5.0 online software. Thirty-fve studies were included in this review. Metabolites such as glutamate, lactate and citrate were disturbed in both PWE and epileptic models, which might be potential biomarkers of epilepsy. Metabolic pathways including alanine, aspartate and glutamate metabolism; glycine, serine and threonine metabolism; glycerophospholipid metabolism; glyoxylate and dicarboxylate metabolism; and arginine and proline metabolism were involved in epilepsy. These pathways might play important roles in the pathogenesis of the disease. This review summarizes metabolites and metabolic pathways related to epilepsy.

编号：CYDQZ-2022-4-3

引用格式：Chen Q, Wang T, Kang D, Chen L. Protective effect of apolipoprotein E epsilon 3 on sporadic Alzheimer's disease in the Chinese population: a meta-analysis. Sci Rep. 2022;12(1):13620. doi: 10.1038/s41598-022-18033-x.

通讯作者：陈蕾

Abstract

Alzheimer's disease (AD) is fast becoming one of the most expensive, deadly and burdensome diseases in this century. It has the fastest-growing disease burden in China. Apolipoprotein E (APOE) polymorphic alleles are generally considered to be the primary genetic determinant of AD risk: individuals with the E4 allele are at increased risk of AD compared with individuals with the more common E3 allele. Since the intensity of the association varies among different ethnic groups, a separate meta-analysis of the Chinese population is needed. We searched Chinese and English databases to sift through literature over the past 20 years. Data on the APOE genotype and AD were collected for correlation analysis. OR was calculated according to APOE allele and genotype. A publication bias analysis and sensitivity analysis were performed, and the main results were further verified by subgroup analysis. The 116 eligible studies enrolled 23,396 patients with AD and 25,568 healthy controls. The study subjects covered at least 30 of the 34 provincial-level administrative regions (including Taiwan). The partial sex ratio was as follows: AD male/female; 10,291/11,240; control male/female, 11,304/12,428, [Formula: see text] = 0.122, P = 0.727. The results of the meta-analysis of alleles showed that I2 > 50% and Q statistics were significant for all genotypes; therefore, the random effect model was selected. The frequency of the ApoE ε4 allele in AD was higher than that in healthy controls, and the difference was statistically significant (OR 2.847, 95% CI [2.611-3.101], P < 0.001). The frequencies of ApoE ε3 and ε2 in AD were lower than those in healthy controls, and the differences were statistically significant (ε3: OR 0.539, 95% CI [0.504-0.576], P < 0.001; ε2: OR 0.771, 95% CI [0.705-0.843], P < 0.001). The results of the meta-analysis of AD

genotype showed that ApoE ε2/ε4 (OR 1.521, 95% CI [1.270-1.823], P < 0.001), ε3/ε4 (OR 2.491, 95% CI [2.267-2.738], P < 0.001) and ε4/ε4 (OR 5.481, 95% CI [4.801-6.257], P < 0.001) allele genotype frequencies were higher than those of the healthy controls. The differences were all statistically significant. Moreover, the ApoE ε2/ε2 (OR 0.612, 95% CI [0.504-0.743], P < 0.001), ε2/ε3 (OR 0.649, 95% CI [0.585-0.714], P < 0.001) and ε3/ε3 (OR 0.508, 95% CI [0.468-0.551], P < 0.001) genotypes were less frequent in patients with AD than in healthy controls, and the differences were statistically significant. The results of the sensitivity analysis and subgroup analysis were consistent with those of the whole model. These results provide support for the protective effect of the ApoE ε3/ε3 genotype against the development of AD. This research is the most comprehensive meta-analysis of the correlation between APOE and AD in the Chinese population by analysing the distribution of the APOE gene in patients with AD reported in the last 20 years. It was concluded that the APOE ε3 allele had a protective effect against sporadic AD in the Chinese population, with great significance, and that its protective effect was stronger than that of the ε2 allele.

编号：CYDQZ-2022-4-4

引用格式： Liu Y, Li Y, Wang Y, Lin C, Zhang D, Chen J, Ouyang L, Wu F, Zhang J, Chen L. Recent progress on vascular endothelial growth factor receptor inhibitors with dual targeting capabilities for tumor therapy. J Hematol Oncol. 2022;15(1):89. doi: 10.1186/s13045-022-01310-7.

通讯作者 陈蕾

Abstract

Vascular endothelial growth factor receptors (VEGFRs) are a family of receptor protein tyrosine kinases that play an important role in the regulation of tumor-induced angiogenesis. Currently, VEGFR inhibitors have been widely used in the treatment of various tumors. However, current VEGFR inhibitors are limited to a certain extent due to limited clinical efficacy and potential toxicity, which hinder their clinical application.

Thus, the development of new strategies to improve the clinical outcomes and minimize the toxic effects of VEGFR inhibitors is required. Given the synergistic effect of VEGFR and other therapies in tumor development and progression, VEGFR dual-target inhibitors are becoming an attractive approach due to their favorable pharmacodynamics, low toxicity, and anti-resistant effects. This perspective provides an overview of the development of VEGFR dual-target inhibitors from multiple aspects, including rational target combinations, drug discovery strategies, structure-activity relationships and future directions.

编号：CYDQZ-2022-4-5

引用格式： Tian M, Chen J, Li J, Pan H, Lei W, Shu X. Damaging novel mutations in PIGN cause developmental epileptic-dyskinetic encephalopathy: a case report. BMC Pediatr. 2022;22(1):222. doi: 10.1186/s12887-022-03246-w.

第一作者 田茂强

Abstract

Background: Mutations in PIGN, resulting in a glycosylphosphatidylinositol (GPI) anchor deficiency, typically leads to multiple congenital anomalies-hypotonia-seizures syndrome. However, the link between PIGN and epilepsy or paroxysmal non-kinesigenic dyskinesia (PNKD) is not well-described. This study reported a patient with PIGN mutation leading to developmental and epileptic encephalopathy and PNKD, to expand upon the genotype-phenotype correlation of PIGN. **Case presentation:** During the first 10 days of life, a girl exhibited paroxysmal staring episodes with durations that ranged from several minutes to hours. These episodes occurred 2-5 times daily and always occurred during wakefulness. Ictal electroencephalography revealed no abnormalities, and PNKD was diagnosed. The patient also exhibited severely delayed psychomotor development and generalized seizures at the age of 4 months. Results of brain magnetic resonance imaging and metabolic screenings were normal, but trio-based whole-exome sequencing identified two novel compound heterozygous PIGN

mutations (NM_176787; c.163C > T [p.R55 > X] and c.283C > T [p.R95W]). Flow cytometry analysis of the patient's granulocytes revealed dramatically reduced expression of GPI-anchored proteins. This indicated that the mutations compromised GPI functions. The patient got seizure-free for 1 year, and her dyskinesia episodes reduced significantly (1-2 times/month) after treatment with levetiracetam (600 mg/day) and clonazepam (1.5 mg/day). No progress was observed with respect to psychomotor development; however, no craniofacial dysmorphic features, cleft lip/palate, brachytelephalangy with nail hypoplasia, and internal malformations have been observed until now (6 years of age). **Conclusion:** This is the first study to document developmental and epileptic encephalopathy with PNKD in a human with PIGN mutations. This report expanded our understanding of the genotype-phenotype correlation of PIGN, and PIGN may be considered a potentially relevant gene when investigating cases of epilepsy or PNKD.

编号：CYDQZ-2022-4-6

引用格式： Hu Y, Cai P, Zhang H, Adilijiang A, Peng J, Li Y, Che S, Lan F, Liu C. A Comparation Between Frame-Based and Robot-Assisted in Stereotactic Biopsy. Front Neurol. 2022;13:928070. doi: 10.3389/fneur.2022.928070.

通讯作者 刘长青

Abstract

Introduction: Frame-based stereotactic biopsy is well-established to play an essential role in neurosurgery. In recent years, different robotic devices have been introduced in neurosurgery centers. This study aimed to compare the SINO surgical robot-assisted frameless brain biopsy with standard frame-based stereotactic biopsy in terms of efficacy, accuracy and complications. **Methods:** A retrospective analysis was performed on 151 consecutive patients who underwent stereotactic biopsy at Chongqing Sanbo Jiangling Hospital between August 2017 and December 2021. All

patients were divided into the frame-based group (n = 47) and the SINO surgical robot-assisted group (n = 104). The data collected included clinical characteristics, diagnostic yield, operation times, accuracy, and postoperative complications. **Results:** There was no significant difference in diagnostic yield between the frame-based group and the SINO surgical robot-assisted group (95.74 vs. 98.08%, $p > 0.05$). The mean operation time in the SINO surgical robot-assisted group was significantly shorter than in the frame-based group (29.36 ± 13.64 vs. 50.57 ± 41.08 min). The entry point error in the frame-based group was significantly higher than in the robot-assisted group [1.33 ± 0.40 mm (0.47-2.30) vs. 0.92 ± 0.27 mm (0.35-1.65), $P < 0.001$]. The target point error in the frame-based group was also significantly higher than in the robot-assisted group [1.63 ± 0.41 mm (0.74-2.65) vs. 1.10 ± 0.30 mm (0.69-2.03), $P < 0.001$]. Finally, there was no significant difference in postoperative complications between the two groups. **Conclusion:** Robot-assisted brain biopsy becomes an increasingly mainstream tool in the neurosurgical procedure. The SINO surgical robot-assisted platform is as efficient, accurate and safe as standard frame-based stereotactic biopsy and provides a reasonable alternative to stereotactic biopsy in neurosurgery.

编号：CYDQZ-2022-4-7

引用格式： Hu Y, Zhang H, Adilijiang A, Zhou J, Guan Y, Qi X, Wang M, Wang J, Wang X, Liu C, Luan G. Seizure outcomes and prognostic factors in patients with gangliogliomas associated with epilepsy. Front Surg. 2022;9:946201. doi: 10.3389/fsurg.2022.946201.

通讯作者 刘长青

Abstract

Introduction: Ganglioglioma (GG) patients often present with seizures. Although most patients can be seizure-free after tumor resection, some still experience seizures. The present study aimed to analyze a group of GGs patients associated with epilepsy and evaluate the seizure outcomes and prognostic factors. **Methods:** This retrospective

study involved clinical data collected from medical records of patients diagnosed with GG pathologically and underwent surgical resection in Sanbo Brain Hospital, Capital Medical University. The seizure outcomes were evaluated based on the International League Against Epilepsy (ILAE) seizure outcome classification. The prognostic factors were identified according to univariate and multivariate analysis.**Results:** A total of 222 patients were included, with a mean age at surgery of 19.19 ± 10.93 years. All patients were followed up at least for one year with a mean follow-up duration of 6.28 ± 3.17 years. At the final follow-up, 174 (78.4%) patients achieved ILAE Class 1 or 2. Univariate and multivariate analyses revealed that the short duration of seizures and gross total resection were significant positive factors for seizure-free. Bilateral interictal or ictal epileptiform discharges in preoperative video-electroencephalogram (VEEG) were related to poor seizure outcomes. **Conclusion:** Surgical resection is an effective treatment for patients with epilepsy associated with GGs. The analysis of predictive factors could effectively guide clinical practice and evaluate the prognosis of epilepsy with GG.

编号：CYDQZ-2022-4-8

引用格式： Zhao B, LiaoS, Zhong X, Luo Y, Hong S, Cheng M, Zhang J, Li J, Jiang L. Effectiveness and Safety of Oxcarbazepine vs. Levetiracetam as Monotherapy for Infantile Focal Epilepsy: A Longitudinal Cohort Study. Front Neurol. 2022;13:909191. doi: 10.3389/fneur.2022.909191.

通讯作者 李听松

Abstract

Objective: This study aimed to compare the effectiveness and safety of oxcarbazepine (OXC) vs. levetiracetam (LEV) for treating infantile focal epilepsy in a longitudinal cohort study. **Methods:** We enrolled 187 consecutive patients aged 2-24 months who received OXC or LEV as initial monotherapy; 161 patients completed the study. The longitudinal analysis involved anti-seizure medication (ASM) responsiveness, safety,

the establishment of epilepsy syndrome, and etiology over a median follow-up of 2 years (interquartile range [IQR] 1.6-2.4). The relative efficacy and retention rates of OXC vs. LEV were evaluated using generalized linear regression models and the Cox proportional hazards model. **Results:** The 161 patients who completed the study had comparable baseline demographics and clinical variables between the OXC group (n = 83) and LEV group (n = 78). Overall, the mean age at onset was 6 months (IQR 4.3-9). The most common epilepsy syndrome was self-limited familial/non-familial infantile epilepsy (54.7%). Epilepsy was related to genetic and unknown causes in 34.2 and 52.2% of the patients, respectively. OXC achieved significantly higher responses than LEV for seizure freedom (risk ratio [RR] = 1.71, 95% confidence interval [CI] = 1.28-2.73, P < 0.001) and 12-month retention rate after onset (hazard ratio [HR] = 1.84, 95% CI = 1.15-2.95, P = 0.007). Moreover, OXC showed more obvious effects for patients aged < 1 year diagnosed with self-limited familial/non-familial infantile epilepsy and non-syndromic epilepsy with genetic or unknown causes. The adverse events related to both OXC and LEV were well-tolerated. **Significance:** OXC could be an alternative to LEV for treating infantile focal epilepsy. OXC monotherapy can be considered first-line treatment for patients aged <12 months and those with epilepsy without developmental and epileptic encephalopathy.

编号：CYDQZ-2022-4-9

引用格式： Jiang Y, Zou N, Luo Y, Cheng M, Liao S, Hong S, Liang X, Zhong M, Li T, Jiang L. Cohort study of infantile epileptic spasms syndrome: etiological analysis and treatment of corticosteroids. Seizure. 2022;101:120-126. doi: 10.1016/j.seizure.2022.07.019.

通讯作者 李听松

Abstract

Background: Infantile epileptic spasms syndrome (IESS) is the most common type of severe epilepsy in infants. However, etiological frequency and optimized therapy,

particularly corticosteroid regimen and dose, remain unknown. **Methods:** An ambispective study of an IESS-diagnosed cohort was conducted. Etiologies were evaluated based on the 2017 International League Against Epilepsy classification system. Patients received intravenous dexamethasone or methylprednisolone for 3-5 consecutive days, followed by usual-dose (2 mg/kg/d) oral prednisone for 60-90 days with tapering doses for 1-2 months or high-dose (4 mg/kg/d) oral prednisone for 9-11 days with tapering doses for 2-4 weeks. Treatment responses were compared between the usual and high-dose prednisone groups after propensity score matching. Correlation analysis between treatment responses and underlying etiology was performed. **Results:** Of the 441 included participants, 218 (49.4%) cases had proven etiologies. The most common etiology of IESS was acquired-structural (23.6%), followed by genetic (15.4%) and congenital-structural (7.0%). Hypoxic-ischemic encephalopathy (55, 52.8%) was the most common acquired-structural etiology. Among the 242 patients administered corticosteroids, 95 received usual-dose oral prednisone and 147 received high-dose oral prednisone. After propensity score matching, 54 patients were included in the usual-dose and high-dose groups, respectively, and treatment effectiveness was compared. There were no significant differences in seizure freedom at days 13-14 (55.6% vs. 51.9%, $p = 0.700$) and continued seizure freedom between days 14-42 (29.6% vs. 38.9%, $p = 0.311$) post corticosteroid administration between the usual- and high-dose prednisone groups. The proportion of children achieving seizure cessation at days 13-14 ($\chi2 = 1.470$, $p = 0.698$) and days 14-42 ($\chi2 = 0.928$, $p = 0.836$) was similar in the different etiological subgroups. Unknown etiological group showed significantly higher resolution of hypsarrhythmia than other etiological groups ($\chi2 = 10.761$, $p = 0.009$). Both usual-dose and high-dose group showed tolerance to full-dose corticosteroids and similar adverse events over the observation period.**Conclusion:** IESS etiology was primarily related to structural causes. Clinical response in short-term follow-up was independent of prednisone dosage and underlying etiology. Better EEG responses may occur in patients with unknown etiology.

编号：CYDQZ-2022-4-10

引用格式： Shen K, Duan Q, Duan W, Xu S, An N, Ke Y, Wang L, Liu S, Yang H, Zhang C. 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;32(5):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：SSL-2022-4-1

引用格式： Chen Y, Wang Q, Xu Y, Wu D, Xu L, Zhu G, Wu X. Comparison of Lamotrigine and Oxcarbazepine Monotherapy Among Chinese Adult Patients With Newly-Diagnosed Focal-Onset Epilepsy: A Prospective Observational Study. Front. Neurol. 2022;13:855498. doi: 10.3389/fneur.2022.855498

通讯作者：吴洵昳

Abstract

Objective: We performed a prospective cohort study to compare the efficacy, safety, effect on mood, and quality of life between lamotrigine (LTG) and oxcarbazepine (OXC) monotherapy among Chinese adult patients with newly-diagnosed focal-onset epilepsy (FOE) with or without secondarily generalized tonic-clonic seizures. **Methods:** We enrolled 106 adult patients with new-onset FOE, of whom 56 were in the OXC group and 50 in the LTG group. Their clinical characteristics were detailly recorded especially basic seizure frequency, seizure types, and drug-related adverse events. Efficacy was evaluated as seizure-free (no seizure for 6 months), effective (seizure reduction by more than 50%), and ineffective (seizure reduction by less than 50%). Both intention-to-treat and per-protocol analyses were performed. We also assessed their mood state with the Zung Self-rating Scale for anxiety (Z-SAS) and Zung Self-rating Scale for Depression (Z-SDS) and quality of life (QOL) with Quality of Life in Epilepsy (QOLIE-31) at their baseline visit, 3-month visits, and 6-month visit. Intra-group comparisons in each group and inter-group comparisons between the two groups were made. Correlation analysis and multiple regression analysis were also conducted. **Results:** Except for gender, the two groups were well matched in any other characteristics such as primary seizure frequency and seizure types. In terms of efficacy, 33 patients in the OXC group were evaluated as seizure-free and 15 as effective, while in the LTG group, 31 were seizure-free, and nine were effective. No significant differences could be observed in efficacy between the two groups (P = 0.429). Through multiple logistic regression analysis, we found that OXC monotherapy was more likely

to predict a seizure-free state (OR = 1.76) than LTG, but the difference didn't reach statistical significance (P = 0.322) after correcting for other clinical variables. Both groups had adverse events such as fatigue, drowsiness, dizziness, rash, and gastrointestinal discomfort, most of which were mild and transient. In the OXC group, the scores of SAS (P = 0.067) and SDS (P = 0.004) reduced at the 6-month visit, while the score of Chen et al. Lamotrigine and Oxcarbazepine Monotherapy Comparison QOLIE-31 significantly increased (P = 0.001). In the LTG group, a significant decrease in SAS and SDS scores and an increase in QOLIE-31 scores could be witnessed (All P < 0.001). The inter-group comparison showed that improvement of SAS and SDS in the LTG group was more evident than that in the OXC group, which was of statistical significance. Correlational analysis indicated that the improvement of mood and life quality scales in both groups was independent of baseline seizure frequency and treatment efficacy. Multiple linear regression analysis indicated that LTG monotherapy was the only independent factor that could predict a better SAS (P = 0.01) and SDS (P = 0.019) outcome. **Conclusions:** OXC and LTG are effective as monotherapy and can be considered first-line selection among adult patients with new-onset FOE. Most adverse events are mild, transient, and tolerable. The two drugs improve the mood state of patients, though LTG is superior to OXC in this respect. OXC and LTG have great power in enhancing patients' quality of life. The positive effect on the psychosocial well-being of epilepsy patients may be one of the intrinsic pharmacological properties of LTG and OXC.

编号：SSL-2022-4-2

引用格式： L, Jun JW, Chang QW, Fang W, Chen CZ , Hong JW, Xiao Lye , Ji WX. FreeSurfer and 3D Slicer-Assisted SEEG Implantation for Drug-Resistant Epilepsy. Front. Neurorobot. 16:848746. doi: 10.3389/fnbot.2022.848746

第一作者：刘强强

Abstract

Objective: Our study aimed to develop an approach to improve the speed and resolution of cerebral-hemisphere and lesion modeling and evaluate the advantages and disadvantages of robot-assisted surgical planning software. Methods: We applied both conventional robot planning software (method 1) and open-source auxiliary software (FreeSurfer and 3D Slicer; method 2) to model the brain and lesions in 19 patients with drug-resistant epilepsy. The patients' mean age at implantation was 21.4 years (range, 6–52 years). Each patient received an average of 12 electrodes (range, 9–16) between May and November 2021. The electrode-implantation plan was designed based on the models established using the two methods. We statistically analyzed and compared the duration of designing the models and planning the implantation using these two methods and performed the surgeries with the implantation plan designed using the auxiliary software. **Results:** A significantly longer time was needed to reconstruct a cerebral-hemisphere model using method 1 (mean, 206 s) than using method 2 (mean, 20 s) ($p < 0.05$). Both methods identified a mean of 1.4 lesions (range, 1–5) in each patient. Overall, using method 1 required longer (mean, 130 s; range, 48–436) than using method 2 (mean, 68.1 s; range, 50–104; $p < 0.05$). In addition, the clarity of the model based on method 1 was lower than that based on method 2. To devise an electrode-implantation plan, it took 9.1–25.5 min (mean, 16) and 6.6–14.8 min (mean, 10.2) based on methods 1 and 2, respectively ($p < 0.05$). The average target point error of 231 electrodes amounted to 1.90 mm ± 0.37 mm (range, 0.33–3.61 mm). The average entry point error was 0.89 ± 0.26 mm (range, 0.17–1.67 mm). None of the patients presented with intracranial hemorrhage or infection, and no other serious complications were observed. **Conclusions:** FreeSurfer and 3D Slicer-assisted SEEG implantation is an excellent approach to enhance modeling speed and resolution, shorten the electrode-implantation planning time, and boost the efficiency of clinical work. These well-known, trusted open-source programs do not have explicitly restricted licenses. These tools, therefore, seem well suited for clinical-research applications under the premise of approval by an ethics committee, informed consent of the patient, and clinical judgment of the surgeon.

编号：SSL-2022-4-3

引用格式：Wei Z, Ye X, Wang C, Xu J, Zhang P, Liu Q, Zhao J. Case Report: Stereoelectroencephalography and Stereoelectroencephalography-Guided Radiofrequency Thermocoagulation in Familial Lateral Temporal Lobe Epilepsy. Front. Neurol. 13:864070. doi: 10.3389/fneur.2022.864070

通讯作者：刘强强

Abstract

Familial lateral temporal lobe epilepsy (FLTLE) is genetic focal epilepsy usually characterised by auditory symptoms. Most FLTLE cases can be controlled by anti-seizure medications, and to our best knowledge, there are no previous reports about stereoelectroencephalography (SEEG) used for patients with FLTLE. In this report, we present two patients with FLTLE in one family and their SEEG performances, together with 18F-flfluorodeoxyglucose (18F-FDG) PET and MRI results. In case 1, fast activities originated from the right superior temporal gyrus and spread rapidly to the right anterior insular lobe and hippocampus. In case 2, there were two seizure patterns: (1) The fast activities or sharp slow waves were identifified at the left superior temporal gyrus, then, sharp waves and spike waves spread in the left superior temporal gyrus; (2) There were fast activities and slow-wave oscillation originated in the left superior temporal gyrus, then, the fast activities spread in the left superior temporal gyrus and fifinally spread to the other sites. An SEEG-guided radiofrequency thermocoagulation was performed for both patients and one of them underwent resection surgery. Seizures are well-controlled and the patients are very satisfified with the therapeutic effects.

欢迎大家引用上述论文

编号：SGQNX-2022-4-1

引用格式：Liu Y, Ling T, Wang H, Yang Y, Song W, Wang T. Generation of an integration-free induced pluripotent stem cell line (LZUSHI001-A) from an epileptic patient with DGKG mutation. Stem Cell Res. 2022;61:102768. doi: 10.1016/j.scr.2022.102768. E

第一作者 刘亚青

Abstract

Epilepsy is a common chronic neurological disorder related to genetic factors. Base on the non-integrating episomal vector technique, a human induced pluripotent stem cell (iPSC) line, termed as LZUSHI001-A, was generated from peripheral blood mononuclear cells (PBMCs) of a 11-year-old male patient with Epilepsy, who had a heterozygous (c.2042G>A, p.R681Q) mutation in the DGKG gene. LZUSHI001-A offers a useful resource to investigate pathogenic mechanisms in epilepsy, as well as a cell-based model for drug development to treat epilepsy.

欢迎大家引用上述论文

菁 YOUNG 计划

菁 YOUNG 计划是中国抗癫痫协会青年委员会 2022 年度新开展的一项学术活动。2022 年 3 月 12 日项目在海南省海口市正式启动，全年计划完成 8 场学术活动，分别是八大区各承担，其中 4-8 月份实际了四场正式活动，得到了协会领导和广大青委们的认可。

自 2015 年国家提出启动精准医疗计划以来，依托高能量序和生物信息数据分析的技术进步等，加速了基因组、蛋白质组学技术等向临床应用的转化。特别是 CAAE 精准医学和药物不良反应监测专业委员会成立后，在廖卫平教授的带领下，我国癫痫领域在基因诊断等方面取得了突出成果，基因监测在癫痫领域提速发展。

青年委员们作为新一年的抗癫痫工作者，更应当积极投身为精准诊断与治疗的研究与应用。精准诊疗既不同于单纯依靠个案积累的经验，也不同于以书本知识和临床研究的循证医学，而应当是一种依托循证医学证据和个体化信息相结合的科学的个体化的诊疗活动。首先，癫痫的分层诊断，包括癫痫发作、癫痫、癫痫综合征和病因学的精准分类与癫痫共患病的准确诊断等对癫痫的治疗和预后判断非常重要；其次，癫痫作为一种有明显遗传异质性的疾病，同时，遗传也是癫痫的六大病因之一，准确掌握癫痫的染色体和基因相关信息至关重要；第三，无论是立体脑电图电极的置入、脑深部刺激电极的植入、激光间质热凝疗还是手术的切除都需要对解剖区域的准确定位和手术的精准实施。为此，在江苏思华药业股份在限公司的支持下，青委会制定了“菁 YOUNG 计划”。

会议日程

时 间	内 容	讲 者	主 席
09:00–09:05	中国抗癫痫协会青委会主委致辞	梁树立 教授	周 健 教授
09:05–09:10	中国抗癫痫协会领导致辞	张 慧 秘书长	
09:10–09:25	上海恩元检测产品介绍	邵 婕 经理	
09:25–09:50	神经发育障碍的诊治与遗传咨询	季涛云 教授	
09:50–10:15	儿童癫痫的病因筛查及精准治疗	石秀玉 教授	
10:15–10:20	全体休息		
10:20–10:45	胶质瘤相关癫痫的精准诊疗研究进展	樊 星 教授	梁树立 教授
10:45–11:10	癫痫基因检测在外科中的应用	王梦阳 教授	
11:10–11:35	基因检测报告解读原则与病例分享	冯卫星 教授	
11:35–11:50	癫痫精准医学展望及讨论	张 凯 教授 李文玲 教授	
11:50–12:00	会议总结	梁树立 教授	

会议日程

时 间	内 容	讲 者	主 席
09:00–09:05	大会主席致辞	陈子怡 教授	陈子怡 教授
09:05–09:10	中国抗癫痫协会领导致辞	张 慧 秘书长	
09:10–09:25	上海恩元检测产品介绍	邵 婕 经理	
09:25–09:50	药物基因组学在癫痫个体化治疗中的临床应用	倪冠中 教授	
09:50–10:15	Recessive PKD1 mutations are associated with FS/EFS+ & the genotype-phenotype correlation	何 娜 教授	
10:15–10:25	全体休息		
10:25–10:50	TBC1D24基因相关婴儿癫痫伴游走性局灶性发作的诊治	胡 君 教授	
10:50–11:15	一例癫痫伴脑病的病因探讨	彭炳蔚 教授	
11:15–11:40	如何提高基因检测技术在癫痫精准医学中的应用水平	郭 强 教授 李其富 教授 曾 涛 教授	
11:40–11:50	会议总结	陈子怡 教授	

5 月 8 日首场“菁 YOUNG 计划”学术活动由京津冀晋蒙大区周健副主任委员牵头，围绕癫痫病因学、遗传咨询、基因报告的解读、胶质瘤相关癫痫及癫痫外科的精准诊疗开展了学术报告和激烈的讨论。6 月 12 日陈子怡副主任委员牵头、粤桂琼闽大区完成的第二场学术活动则突出药物基因组学、癫痫基因相关研究和学术论文发表以及两个病例精准诊疗的实际实施进行了报告，同时就如何提高基因检测技术在癫痫精准医学中的应用水平为题开展的讨论。浙皖赣大区承办的第三场“菁 YOUNG 计划”学术活动由王爽教授担任主席，会议内容包括了指南解读、病例分析和维生素 B6 依赖性癫痫与 PCDH19 相关癫痫进行了讲解，会议还特别邀请了刘晓蓉教授对结节遗传学病因的抗癫痫发作药物选择进行了专题报告，同时回答了青委们在基因在癫痫诊疗中应用的不少困惑。

张慧秘书长对“菁 YOUNG 计划”特别重视，参加每所有场次的活动。青委们对精准诊疗有了更深入的理解，也一定会在实际工作中加强应用，提高临床水平，但跨区参与度有待提高。

会议日程

时 间	内 容	讲 者	主 席
08:30-08:35	大会主席致辞	王 爽 教授	王 爽 教授
08:35-08:40	中国抗癫痫协会青委会主委致辞	梁树立 教授	
08:40-08:45	中国抗癫痫协会领导致辞	张 慧 秘书长	
08:45-09:00	上海恩元检测产品介绍	刘聪敏 经理	
09:00-09:25	结合遗传学病因的抗癫痫药物选择	刘晓蓉 教授	
09:25-09:50	维生素B6依赖性癫痫的诊治	虞雄鹰 教授	
09:50-09:55	全体休息		
09:55-10:20	从ACNS2021术语看NCSE	王 康 教授	虞雄鹰 教授
10:20-10:45	PCDH19相关癫痫	袁哲峰 教授	
10:45-11:10	病例分享：容易“受惊吓”的女孩	沈春红 教授	
11:10-11:30	会议讨论与总结	王 爽 教授 虞雄鹰 教授	

领读学术

领读学术 2022 年我们全年将开展 10 场活动，首场全国会于 1 月 22 日线上召开、粤桂琼闽大区于 3 月 20 日线上召开首场区域性活动。

浙皖赣大区会议领读学术活动于 5 月 21 日线上召开，由王爽教授担任主席。蒋玉宝青委以 SCI 成形记介绍了文章写作经验、郭谊青委对局灶性癫痫药物优化治疗进行了文献解读；林上闸老师讲解了数据统计分析的思路和方法选择。虞雄鹰、齐印宝、王新施等青委共同讨论。覆盖全国 25 个省市，观看医院超 40 家，总观看人次超 300 人。

2022 年京津冀晋蒙大区会议领读学术活动于 6 月 18 日线上召开，由周健教授担任主席。李天富教授进行了临床和基础研究 SCI 发表的经验浅谈，王梦阳青委对结合 SANDA-II 研究介绍的经典药物的强势回归；梅斯医学科学部的林上闸老师讲解了文章选刊和修回的处理方法。孙伟、梁树立、季涛云等青委共同讨论。覆盖全国 27 个省市，线上加入观看医院超 35 家，总观看人次超 300 人。

陕甘青宁新大区领读学术于 6 月 25 日线上召开。刘永红教授担任主席。刘亚青青委以抗癫痫药物单药治疗与卒中后癫痫死亡率的关系为题介绍了相关研究；马磊青委对 NICE 指南局灶性癫痫篇进行了解读；梅斯医学科学部林上闹老师讲解了 META 分析的流程和统计。孙美珍教授、孙岩、王兰桂、王超等青委参加讨论。超 500 人加入直播间，人员覆盖等 30 个省市自治区，45 家医院。

陕甘青宁新大区领读学术于 8 月 28 日线上召开。刘永红教授担任主席。李听松青委以轻度胃肠炎伴婴幼儿良性惊厥为题介绍了 SCI 发表经验；吴欣桐青委对 SANDA-II 研究进行了解读；梅斯医学科学部的林上闹老师讲解了 SCI 论文写作的结构规范上半部分。孙美珍教授、孙岩、王兰桂、王超等青委参加讨论。超 600 人加入直播间，人员覆盖等 30 个省市自治区，45 家医院。

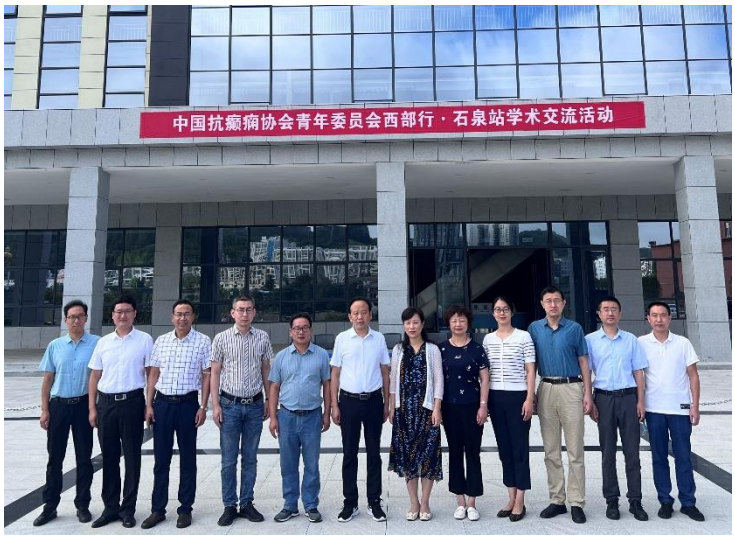
2022 年 CAAE 青年委员会线上抗癫痫●西部行

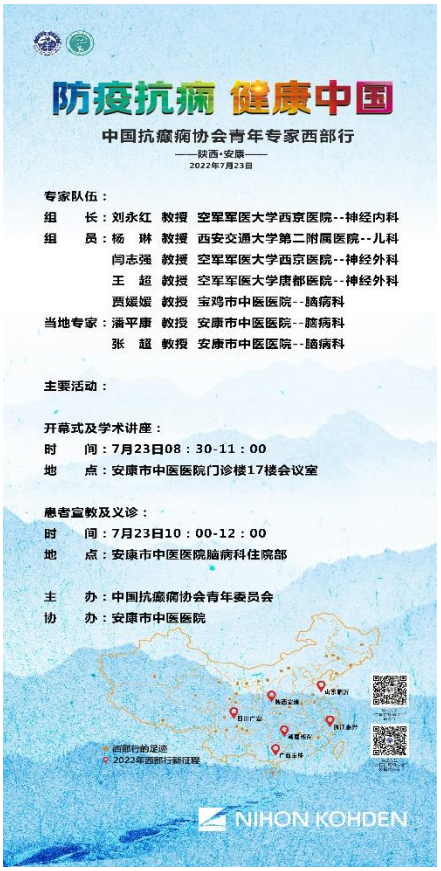
2021 年新开展的 CAAE 青年委员会线上西部行共完成了 10 场活动，覆盖 9 个省和自治区，得到了协会领导和秘书处领导的大力支持和肯定。今年将继续开展 8 场活动，主要是以八大区为主分别开展 1 场。

2022 年首场线上西部行活动由王爽教授牵头由浙皖赣青委承办的线上西部行活动—浙江海盐站顺利完成。这也是西部行活动首次走进浙江。海盐站与继往的线上西部行站点不同，没有承办过西部行活动，但浙皖赣大区负责人王爽教授在海盐代职支援 1 年，并至今持续帮扶，并且海盐也已经列入 2021 年西部行站点（因疫情未成行）。会上海盐人民医院神经内科介绍了近两年科室的工作，特别是癫痫领域的进展情况，王新施和丁晶青委进行了专题讲座，然后针对当地医院提供的疑难病例进行了讨论分析。张慧秘书长进行了致辞，对海盐当地的工作，特别是王爽教授帮扶下取得了成绩给予高度肯定，并表示疫情允许时尽快完成线下西部行活动。会议取得圆满成功。

2022 年 CAAE 青年委员会抗癫痫●西部行活动

由于疫情的原因，原计划 2021 年开展的六场线下西部行，仅完成了广西柳州场活动。今年青年委员们牢记使命在肩，在积极防疫的同时，继续开展抗癫痫西部行活动，助健康中国。为保证活动的顺利进行，今年仍然以各大区，特别是本省区青委专家为主组成活动小组的特点。2022 年 7 月 22-23 日湘鄂余大区在彭镜副主任委员的带领下，冯莉、卢军、陈晨等青年委员一共参与了第 71 场西部行活动，完成了湖南湘潭（125 站）和韶山（第 126 站）两站的系列活动。几乎同时，7 月 23-24 日陕甘青宁新大区闫志强、王超、贾媛媛和第一届青年委员杨琳等专家一行由刘永红副主任委员带队赴陕西安康（127 站）和石泉（128 站）完成了第 72 场 CAAE 抗癫痫西部行活动。





尽管新冠疫情仍在动态清零过程中，防疫的压力仍然不减。但这两场四站的西部行活动仍然得到了当地医院、抗癫痫医务人员和患者的热烈欢迎，医疗主要领导、相关科室主任及工作人员合计 500 人次参加了学术讲座活动，患者教育 400 人次，义诊和会诊、查房 300 人次。无论在时间和活动内容、流程都得到了很好的保持，张慧秘书长也特别进行了视频致辞，对相关给予大力支持。这两场活动分别得到了光电公司和卫材公司的大力支持，也是协会、医生、医药企业共同践行社会责任的共同体现。

统筹策划	张　慧　段立嵘　梁树立
刊头题字	李世绰
资料来源	全体青年委员　CAAE网站
资料收集	丁　瑶　刘亚青　张春青　梁建民 陈子怡　王　爽　陈　蕾　周　健 吴洵昳　刘永红　彭　镜　孙　丹 李文玲　张　凯　季涛云　郭　强
资料整理	梁树立　刘婷红
排版美工	刘婷红　王雅婷

CHINA ASSOCIATION AGAINST EPILEPSY YOUTH COMMITTEE

THINKINGS AND ACTIONS