

先天性脑穿通畸形囊肿 1 例报道

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脑穿通畸形囊肿(Canalization Cyst)是指大脑半球内有空洞或囊肿与脑室相通,其内充满脑脊液,可扩延至脑膜,但不进入蛛网膜下腔的一种疾病。其分为先天性脑穿通畸形囊肿(Porencephalic Cyst)和后天性脑穿通畸形囊肿。先天性脑穿通畸形囊肿多集中在早产儿、过期儿、难产儿,主要表现为智力低下、癫痫发作、肢体瘫痪等。在临床上成人以单纯部分性癫痫发作为首发症状就诊的患者却十分罕见,现将该病例报道如下。

1 临床资料

患者,男,30 岁。因“突发头晕、四肢僵硬后腰痛 4 h”为主诉于 2018 年 3 月 6 日入本院。患者 4 h 前睡眠中突发头晕,有明显天旋地转感,同时出现四肢僵硬,不能活动,具体表现为双上肢屈曲,双下肢伸直,自诉双手成“鸡爪”样,上述症状持续约 20 min 后缓解,随即出现双侧腰肌疼痛、活动加重,并伴有发热,体温最高达 38.5℃,于当地卫生院给予退热等对症治疗,腰痛无缓解,遂急诊入本院。患者发病过程中无意识障碍,无言语不清,无明显抽搐,无舌咬伤及大小便失禁。患者自诉身体健康,体力劳动不受限。否认外伤、手术史。患者父母身体健康,患者为独子,否认家族遗传病史。患者母亲否认早产、难产、产伤病史。但患者母亲诉患者出生时就有右手无力,活动欠灵活。从小患者用左手持筷、写字。生长发育与同龄人无明显差别。体育活动、体力劳动正常,学习成绩较差,小学毕业后辍学务工。患者 25 岁结婚,配偶健康,生育 1 子 1 女,均体健。患者妻子诉患者平素性格暴躁,易动怒、易激惹,经常与家人发生争吵;发病前 1 d 曾与家人激烈争吵,曾长时间熬夜玩手机;患者邻居诉患者自幼性格执拗、行为古怪,曾抓老鼠做烧烤吃。体格检查:身高 182 cm,体重 85 kg,体型健硕,营养状况好,皮肤黝黑,颅骨发育正常,未见局限性隆起;神清语利,双瞳孔等大正圆, D=3.5 mm,对光反应灵敏,双眼球向各方向运动充分;双侧额纹、鼻唇沟对称,双侧口角无歪斜,伸舌居中。鼓腮、龇牙、吹口哨动作完成好;颈软,无抵抗,右上肢远端肌力Ⅳ级,余肢体肌力Ⅴ级,右侧肱二头肌反射(+++),肱三头肌反射(+++),桡骨膜反射(+++),膝腱反射(+++),跟腱反射(+++),左侧腱反射正常;Babinski[R(+),L(-)],右侧深浅感觉障碍;右侧指鼻试验欠稳准,右侧轮替运动差,跟膝胫试验欠稳准。闭目难立征(+).入院头颅 CT 显示左

侧额叶脑穿通畸形囊肿(图 1)。

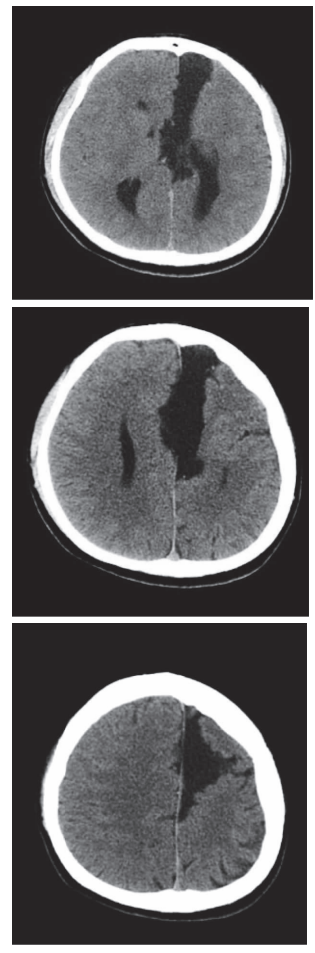


图 1 头颅 CT 显示左侧额叶脑穿通畸形囊肿

2 讨论

脑穿通畸形囊肿最早由 Heschl 于 1859 年提出,是一种罕见的疾病。其发病率极低,多由病例报道的形式呈现。其发病机制尚不明确,最新的研究指出脑穿通畸形可能与 COL4A1 基因突变有关。胎儿在母体妊娠 2~7 个月内大脑皮层出现原始畸形,局部脑组织长期存在血液循环障碍,导致脑组织缺氧、坏死、液化,最终导致脑内囊腔或空洞形成。囊肿与脑室相通,病变处缺乏脑组织支撑,局部受静水压大,加之囊壁本身有分泌功能,致使囊腔逐渐扩大,临近结构受

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(上接第 357 页)

压。故发病早期即出现相应症状与体征,并呈缓慢进行性加重趋势。本例患者虽出生后右手无力,但无进行加重的表现。后天性脑穿通畸形囊肿则常由于围产期产伤窒息或外伤脑出血、脑梗死、颅内感染继发脑组织软化坏死吸收而形成囊性液性暗区。本例患者无产伤史,无脑出血、脑梗死及外伤病史。考虑本例患者为先天性脑穿通畸形囊肿。

文献报道该病主要表现为三大主症如癫痫、脑瘫、低智。患者虽无典型的上述三主症,但仔细询问病史后却仍有迹可循。患者此次虽以腰痛为主诉就诊,但追问病史患者在出现腰痛前有过天旋地转的眩晕、全身僵硬,并伴有发热。考虑患者当时癫痫发作,肌张力明显增高,角弓反张,引起腰部肌肉拉伤。患者癫痫发作前首先出现眩晕,考虑为癫痫先兆。马顺昌等报道的 1 例成人先天性脑穿通畸形囊肿患者也出现了眩晕发作,继而出现癫痫发作。脑穿通畸形囊肿癫痫发作前眩晕是否具有特异性还有待证实。患者虽无明显肢体瘫痪,但自幼右手无力,不能从事精细劳动,持筷、写字均用左手。查体时右手肌力稍差,右侧腱反射亢进,巴宾斯基征

阳性。考虑患者右侧肢体存在轻偏瘫。患者正常交流可,但学习成绩较差,小学毕业后就辍学,不排除与智能减低有关。患者脾气暴躁、性格怪异,考虑与额叶病变有关。此外,国外病例报道的先天性脑穿通畸形囊肿,大部分病例在确诊前有过精神症状发作。该患者既往无明显的精神症状发作病史,也是该病例的特殊之处。

本病的治疗仍以对症治疗如抗癫痫治疗为主,也有人主张有症状者宜早期外科手术治疗。本例为成年男性患者,平常临床症状较轻微,不影响生活。本次发作为单纯部分性癫痫发作,持续时间较短,以休息、营养脑细胞对症治疗为主。但此类疾病,癫痫再发风险高,告知患者及家属,需定期随访;避免高空作业及开车等高风险工作,以免发生意外。该患者为成年男性,以单纯部分性癫痫发作为首发症状起病,脑穿通畸形囊肿的三大主症并不典型,在临床工作中极易被误诊、漏诊。提高对本病的认识,注意询问患者的生长发育史,及时完善颅脑 CT、核磁共振等相关检查,有利于尽早明确诊断,给予及时治疗,避免医疗资源浪费。

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