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个案与短篇

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MRI诊断Lhermitte-Duclos病1例

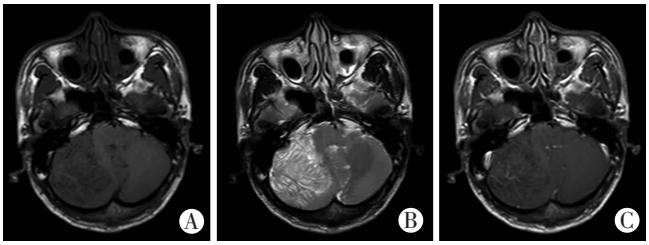
谢珊珊,程敬亮,张 勇 (450052 郑州,郑州大学第一附属医院磁共振科)

[**关键词**] Lhermitte Duclos病;小脑;磁共振成像
[**中图法分类号**] R745.7 [**文献标志码**] B

Lhermitte-Duclos病(lhermitte-duclos disease,LDD),又称小脑发育不良性神经节细胞瘤,是一种罕见的起源于小脑皮层的缓慢进展性占位性病变,最早由Lhermitte和Duclos报道并命名[1]。LDD在MRI上常有典型的影像学表现。本院收治1例,经MRI诊断和病理证实,现报告如下。

1 临床资料

患者,男性,因头部不适在当地医院查头颅MRI发现右侧小脑占位,为求进一步治疗来我院就诊,以"右侧小脑占位"于2013年10月15日收入院。体格检查:神志清,查体合作,四肢活动可,肌力Ⅴ级,病理征阴性。头颅MRI示:右侧小脑半球及桥小脑结合臂片状长T_1混杂长T_2信号,其内弥漫性夹杂不规则线条样更长T_1短T_2信号影,病变整体呈斑纹状(图1A、B),DWI高b上呈等信号;相邻脑干、四脑室受压变形,局部中线结构稍向左侧偏移。小脑扁桃体变尖下移,超出枕骨大孔水平约7 mm;幕上脑室系统稍扩大。增强扫描示上述病灶内少量不规则线条样轻度强化影(图1C),MRI诊断为小脑发育不良性神经节细胞瘤。术中见灰黄色肿瘤组织,质软,类脑组织,呈洋葱皮样分层分叶状,边界尚清,无包膜。术后病理检查示:小脑皮质结构紊乱,形态异常,发育不良的神经节细胞散在分布,肥大的神经节细胞胞质丰富,呈圆形或卵圆形,核仁明显(图2),确诊为:小脑发育不良性神经节细胞瘤。



A:T_1 WI轴位;B:T_2 WI轴位;C:Gd-增强扫描
图1 Lhermitte-Duclos病患者的MRI表现

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[**通信作者**] 程敬亮,E-mail:chengjl-2008@163.com
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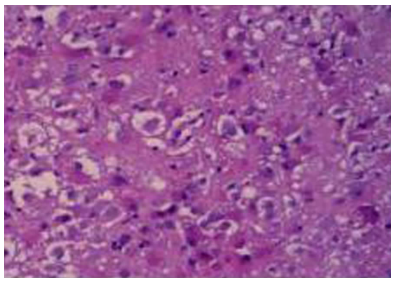


图 2 Lhermitte-Duclos 病脑组织病理学变化 (HE ×400)

2 讨论

LDD 自 1920 年首次报道至今,其性质、发病机制及遗传学改变颇具争议,是发育畸形、错构瘤,还是肿瘤,众说纷纭。2007 年 WHO 将其纳入“神经元和混合神经元-神经胶质肿瘤”,属 WHO Ⅰ级[2]。LDD 可散发,也可合并 Cowden 病,后者为一种罕见的常染色体显性遗传病,以来源于 3 个胚层的多发错构瘤为特征,且患者患恶性肿瘤的风险较高,多为乳腺癌、甲状腺癌和子宫内膜癌[3]。自 1991 年以来,LDD 被认为是 Cowden 病的一部分,且为 Cowden 病的特异性诊断标准[4]。由于 Cowden 病患者全身肿瘤的高发病率,当 LDD 确诊时,对患者进行全身肿瘤筛查和干预十分必要。

正常的小脑皮质由内颗粒细胞层、外分子层及中间的浦肯野细胞层构成。在 LDD 患者,正常的小脑皮质板层结构消失,表现为局限性或弥漫性小脑皮质颗粒细胞层肥大增生和外分子层过度髓鞘化[5],浦肯野细胞层结构消失、白质萎缩,前者往往形成占位效应。LDD 的这些组织学改变似乎兼具先天发育畸形和肿瘤的特点,但 LDD 无浸润性生长和灶周水肿。基于其组织学基础,LDD 在 MRI 上特征性的表现为条纹状改变,即增大小脑叶片皮质的外层部分与正常脑组织相比 T_1WI 呈稍低信号,T_2WI 上呈稍高信号,再加上脑沟内脑脊液的信号特点共同构成 T_1WI 等、低信号,T_2WI 等、高信号交替改变的条纹征。本例即符合此典型表现。

另外,在病变脑叶皮质板层周围有扩大的静脉血管走行,磁敏感加权成像可以清晰显示[6],这是除条纹征外的又一典型 MRI 表现。同时,这些静脉血管也是增强扫描病灶内呈现线条样强化的原因[6-7],本例患者增强扫描即可见此强化征象。静脉血管内含有大量脱氧血红蛋白,后者为顺磁性物质,因 MRI 扫描仪对顺磁性物质的敏感性和磁场强度呈正相关,所以高场强 MRI 扫描仪可以更好地显示这些静脉血管,从而明确病变边界[4],指导手术彻底切除,改善患者预后(因该病复发常与病变切除不彻底有关[8])。本例患者未行 SWI 检查,无法证实 SWI 血管表现。

LDD 虽较为罕见,但 MRI 表现典型,可明确诊断,并明确病变位置、范围及周围组织受压情况,为临床诊疗提供更多信息。另外,MR 在患者远期随访中也起着重要作用。

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