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Surgical treatment of subconal ventricular septal defect

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Abstract Objective: To study the clinical characteristics and therapeutic efficiency of subconal ventricular septal defect. Methods: The ventricular septal defect was closed with patch repair in 191 cases and with direct sutures in 18 cases. The coexisted aortic valvular insufficiency was treated with plication and hanging of the aortic valve. Results: 4 patients died and all the others recovered satisfactorily. No residual fistula or new occurrence of aortic insufficiency was found. Conclusion: It is important to treat the subconal ventricular septal defect with patch repair and aortic valvular insufficiency with plication of the aortic valve.

Key words subconal ventricular septal defect; aortic valve; valvular insufficiency; surgical treatment

V型糖原沉积病1例报告

A case report of McArdle's disease

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1 临床资料

患者,女,20岁。因发作性双下肢疼痛17年入院。患者自3岁时不明原因出现双大腿发作性疼痛,每次持续数小时至十余小时不等,均为持续性酸胀痛,不放射,左侧较右侧明显,劳累及激烈活动诱发或加剧,应用"去痛片"等止痛药物可缓解,间歇期活动正常。无其它不适。自小身体及智力发育可,家族中患者母亲有相似病史,在当地医院按"风湿性关节炎"治疗效差。

入院查体:发育及营养状况好。心肺无异常发现。神经系统检查:四肢肌张力低,双侧肢体腱反射减低,余无异常。辅助检查:血常规,肝、肾功能,血沉,脑脊液均正常;抗核抗体及类风湿因子阴性;尿常规:WBC 25/μL, UBG 10 mg/L, BIL 10 mg/L, Pro+,一周后复查:WBC 20/μL, UBG 25 mg/L, BIL 18 mg/L, Pro+,三周后复查正常;肌酸激酶同功酶32.1 IU/L(正常为<25 IU/L);乳酸运动试验:运动前1.38 mmol/L,运动后3,5,10 min分别为1.12、1.27、1.22 mmol/L;EMG示神经传导速度正常,多相电位增多;ECG正常。肌肉活检:肌核增多,肌纤维大小不等,部分横纹消失,有不同程度变性、坏死,可见再生纤维和炎性细胞浸润,间质血管无明显改变。高碘酸希夫反应(PAS)见肌纤维中阳性物质沉积;电镜下部分线粒体嵴变平,粗面内质网脱颗粒,肌膜下、肌丝间有微小颗粒状物质沉积。病理诊断为糖原沉积病V型(McArdle病)。

2 讨论

McArdle病为糖原沉积病之一,系常染色体隐性遗传使体内尤其是肌肉组织中磷酸化酶缺乏,葡萄糖氧化磷酸化受抑制[1],导致运动时能量供应障碍而出现肌肉无力和痉挛。临床表现为:运动后肌肉无力和痉挛,伴剧烈疼痛,多发于下肢,休息后可好转;部分病人可有继减现象。少数病人可伴发急性肾功能不全、慢性咀嚼的无力和痉挛等[2]。实验室检查可见肌酶谱轻度升高,前臂缺血运动试验血乳酸水平不增或仅轻度升高,伴ECG和EMG改变等,但必须依靠肌活检方可确诊。近来文献报道约90%患者可通过分离白细胞的DNA进行诊断而避免肌肉活检[3]。患者母亲有相似病史,其家族性有待进一步研究。该病的遗传学改变比较复杂,且与人种密切相关。在白色人种和德国人中,R49X的反义突变约占85%,而日本人群中以第17号外显子中708、709码的单码缺失最常见,占约73%[4]。我国目前无该方面资料。

本病例有下列特点:①口服止痛药物有效,可能与运动后局部肌肉中释放致痛物质有关;②无肌肉萎缩,光镜下无肌纤维萎缩及脂质化,可能与病程进展缓慢有关;③尿常规轻度异常是否与肾功能受损有关需要进一步观察。

关键词 糖原沉积病V型

中图法分类号 R589.1

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