同卵双胞胎迟发型鸟氨酸氨甲酰基转移酶缺乏症病例分析并文献复习

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摘要:

目的：探讨迟发型鸟氨酸氨甲酰转移酶缺乏症（OTCD）的临床特点、诊治经验，提高对该病的认识。方法：回顾性分析我院确诊的一对同卵双胞胎兄弟OTCD的临床诊疗经过和实验室检查，并结合国内外资料进行文献复习。结果：同胞弟弟，14岁4月，病程1年余，1年前曾出现呕吐，此次仍以呕吐起病，后出现频繁抽搐、昏迷。血氨增高，尿气相色谱-质谱分析提示尿嘧啶和乳清酸增高。同胞哥哥，无明显临床表现，血氨正常，尿气相色谱-质谱分析提示尿嘧啶和乳清酸正常。遗传病医学外显子组基因测序明确其致病位点为鸟氨酸氨甲酰基转移酶（OTC）基因c.119G>A（p.Arg40His），患儿母亲携带c.119G>A（p.Arg40His）突变。结论：OTCD临床表现缺乏特异性，易误诊，对于原因不明的神经、精神症状或消化道症状患儿应尽早完善血氨、血尿质谱分析，从而有利于早期诊断，确诊则需要基因检测。治疗上以降低血氨，避免高氨血症对神经系统的损伤为原则，早期干预可改善预后。

关键词: 鸟氨酸氨甲酰基转移酶缺乏症 高氨血症 串联质谱法 气相色谱-质谱法

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Report of Identical Twins with Late-Onset Ornithine Transcarbamlase Deficiency and Literatures Report

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Abstract:

Objective: To discuss the clinical characteristic,diagnosis experiences and deepen understanding of late-onset ornithine transcarbamlase deficiency (OTCD). Methods: Data of the clinical diagnosis ,treatment and lab results of the twins with OTCD were analyzed ,and the domestic and international literature was reviewed. Results: The twin younger brother,14 years and 4 months old,course over 1 year, he vomited 1 year ago and the initial symptom this time is vomiting followed by frequent convulsions,coma,hyperammonemia. The urine gas chromatography (GC) showed uracil and saratin increased. The twin elder brother, 14 years and 4 months old, is asymtomatic. Blood ammonia is normal, urine gas GC also. The pathogenicity site confirmed by genetic disease exons gene sequencing is ornithine transcarbamlase gene c.119G>A (p.Arg40His), which the mother carried. Conclusion: Patients of OTCD lack idiosyncracy and easy to be misdiagnosed. Blood ammonia, blood and urine GC should be completed as soon as possible for early diagnosis when patients with agnogenic neurological, mental or gastrointestinal symptoms. OTCD can be confirmed by gene detection. The mainstays of treatment is lowering the plasma ammonia level in case of the risk of neurological damage.Early intervention can improve the prognosis.

Key words: ornithine transcarbamylase deficiency hyperammonemia tandem massspectrometry gaschromatography- massspectrometry