

先天性无运铁蛋白血症一例

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【摘要】 目的 对 1 例以重度贫血为主诉入院的疑难病例进行确诊。方法 根据患儿的症状、体征、特殊检查(骨髓活检、肝活检、运铁蛋白浓度测定)患儿父母血液检查结果及文献资料进行分析确诊。结果 患儿以严重贫血、肝脾肿大为主要表现,血浆 β 球蛋白量为 5% (正常 7% ~ 13%),运铁蛋白浓度为 0.0147 g/L (正常 2.52 ~ 4.29 g/L)。肝活检显示肝内大量含铁血黄素沉着(肝细胞和枯否细胞)和轻微纤维化。符合先天性无运铁蛋白血症。患者父亲的运铁蛋白浓度为 1.29 g/L,患者母亲的运铁蛋白浓度为 1.4 g/L,仅为正常人的一半。结论 确诊患儿为先天性无运铁蛋白血症,此症为常染色体隐性遗传,极其少见,应提高认识,以防漏诊。

【关键词】 贫血; 转铁蛋白; 含铁血黄素沉着症

Congenital atransferrinemia: a case report LIN Guozhen, CHEN Le, WANG Wei, LI Yunzhu, CHEN Shunian. Ruijin Hospital, Shanghai Second Medical University, Shanghai, 200025 China

【Abstract】 Objective Congenital atransferrinemia (CAT) is an extremely rarely, recessively inherited disorder. To date, only eight cases have been reported in the published literatures of the world, and there is no report in China. The authors diagnosed a difficult and complicated case suffering from severe anemia for 3 years. **Methods** A thirteen-year-old boy was transferred to the Pediatric Department, Ruijin Hospital for recurrent anemia with significant hepatosplenomegaly three years. The child had required transfusion of whole blood and iron many times, but there was no benefit to the anemia. The child denied a family history of anemia, and his parents are normal. Diagnosis was confirmed according to the symptoms, signs, special laboratory examinations (liver biopsy, bone marrow biopsy, serum TRF, etc.) and the previously published papers of other authors. **Results** The patient manifested as severe anemia, hepatomegaly and splenectasis. The beta-globulin fraction was 5% (normal value: 7% ~ 13%) on serum protein electrophoresis, and assays for TRF showed 0.0147g/L (normal value: 2.52 ~ 4.29g/L). The liver biopsy revealed the significant deposition of hemosiderin within the hepatocytes and kupffer cells. The parent of the boy showed a low level of TRF. His father's TRF was 1.29g/L, and his mother's TRF was 1.4 g/L. CAT is an autosomal recessive disease. **Conclusion** The diagnosis of congenital atransferrinemia was established, which remind that doctors must pay a great attention to this rare disease.

【Key words】 Anemia; Transferrin; Hemosiderosis

先天性无运铁蛋白血症 (congenital atransferrinemia, CAT) 是一种罕见的隐性遗传性疾病,到目前为止,世界文献只有 8 例报道,国内尚未有报道。我科最近收治一例以重度贫血为主诉入院的患儿,经检查确诊为先天性无运铁蛋白血症,现报告如下。

病例资料

患儿,13 岁,安徽籍,因发现贫血 3 年入院。否认有家族史,外院诊断缺铁性贫血给予多次输血、铁剂治疗均无效转至我院。

入院体检:明显贫血貌,无明显黄疸,生长发育欠佳,心率 98 次/min,心前区有 II 收缩期杂音,肝

肋下 2.5 cm,脾肋下 2 cm,其余阴性。

入院后化验及检查: Hb: 20 ~ 70 g/L,红细胞平均容积: 68 ~ 74 fL,红细胞平均血红蛋白浓度: 276 ~ 280 g/L,红细胞平均血红蛋白量: 20 ~ 21 pg,网织红细胞: 0.04 ~ 0.06, WBC: $(2.9 \sim 5.2) \times 10^9/L$, PLT: $(180 \sim 270) \times 10^9/L$ 。红细胞形态: 明显大小不一,部分呈泪滴形,可见破裂红细胞、裂片红细胞、椭圆形红细胞。大部分红细胞中央淡染区明显扩大。溶血系列检查: 胎儿型血红蛋白: 0.065 (正常 < 0.02),血清结合珠蛋白: 210 mg/L (正常 981 ~ 2740 mg/L),其余阴性。骨髓穿刺: 增生性贫血,粒系伴成熟障碍,骨髓活检: 骨髓造血细胞和脂肪细胞比例 0.8: 0.2,粒红比例倒置,红系增生,以中晚幼红细胞为主。粒系减少,巨核细胞可见。甲、乙、丙、戊型肝炎

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