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头颅核磁和肿瘤标志物在儿童颅内生殖细胞瘤诊治的价值

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Value of Brain Magnetic Resonance Imaging and Tumor Markers in the Diagnosis and Treatment of Intracranial Germinoma in Children

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摘要 目的 评价头颅核磁共振显像（MRI）和脑脊液及血清肿瘤标志物动态改变在儿童颅内生殖细胞瘤诊治的价值。方法"总结2009年1月至2010年12月诊治的5例中枢性尿崩症儿童患者（女性3例、男性2例），全部患者在初诊和随诊时进行头颅MRI增强扫描，并检测垂体前叶激素和脑脊液及血清肿瘤标志物人绒毛膜促性腺激素(hCG)和甲胎蛋白水平。**结果** 3例既往未经过检查和治疗，2例起病时病因未明，就诊于我院时病情加重。起病年龄8岁至12岁1个月，起病至就诊时间1至78个月。全部患者均以多尿、多饮症状起病，除1例以外，其余患者均有生长迟滞，第二性征未发育；1例患者在随访的2年内身高生长速度正常，已进入正常青春期发育，但是在颅内肿瘤显著增大后，5例患者均有垂体前叶功能减退，血浆泌乳素水平升高。3例分别在起病后18、24和78个月出现脑疝。3例起病时头颅MRI均表现为垂体柄增粗，在随访18~22个月表现为下丘脑-垂体区巨大占位，2例在起病后1和78个月首次就诊，MRI示颅内巨大占位；全部患者均有T1加权像垂体后叶高信号的消失。5例患者脑脊液hCG均升高，其中4例血清hCG也相应升高，并且随着肿瘤的增大而升高，放疗后随肿瘤的缩小而下降。只有1例脑脊液和血清甲胎蛋白显著升高。**结论** 诊断为“特发性中枢性尿崩症”的患者必须进行密切的随访来鉴别病因，尤其是合并有垂体前叶激素缺乏时。初诊时MRI表现为正常或单纯垂体柄增粗者，在随访过程中应连续观察头颅MRI增强扫描的变化，以尽早诊断出潜在的下丘脑垂体柄病变。推荐在初诊时评价脑脊液hCG水平，因为hCG升高可能早于MRI阳性表现。

关键词： 中枢性尿崩症 生殖细胞瘤

Abstract: Objective To evaluate the role of brain magnetic resonance imaging (MRI) and tumor markers in the cerebral spinal fluid (CSF) and serum in the diagnosis and treatment of intracranial germinoma in children. Methods Totally 5 children (3 girls and 2 boys) who were treated in our hospital between January 2009 and December 2010 due to central diabetes insipidus. All patients received contrast-enhanced brain MRI at presentation and during each follow-up: meanwhile, their anterior pituitary hormones and tumor markers including human chorionic gonadotropin (hCG) and alpha fetoprotein (AFP) were also determined. Results Three patients presented without prior evaluation, and two patients were referred to our hospital due to exaggerated disease of unknown cause. Their ages at presentation ranged from 8 years to 12 years 1 month, and the duration of symptoms at presentation was between 1 month to 78 months. All of them had polyuria and polydipsia at presentation. Except one child, the other 4 patients had growth retardation and failure in initiation of puberty. Although the growth rate and puberty development were normal during the 2-year follow-up for the excepted child, all child experienced anterior pituitary hypofunction and an increased concentration of plasma prolactin after the lesion became enlarged. Three patients had cerebral hernia, which presented in 18, 24, and 78 months, respectively. In three patients, brain MRI at presentation showed isolated pituitary stalk thickening, which further developed into massive tumor in the hypothalamus pituitary region 18-22 months later; in the remaining two patients, large brain tumor was found via MRI at their first presentations. In all five patients, the posterior pituitary gland (bright spot) disappeared on T1-weighted MRI images. CSF hCG elevated in all five patients, and serum hCG increased in four patients; the level of hCG varied with the mass size of tumor. Serum and CSF AFP increased in only one patient. Conclusions Patients with “idiopathic central diabetes insipidus” must be closely followed to identify the etiology, especially when anterior pituitary hormone deficiencies are detected. For patients with normal brain MRI results or simply isolated pituitary stalk thickening at presentation, the changes of serial contrast-enhanced brain MRI should be observed during follow-up to ensure the early detection of an evolving occult hypothalamic-stalk lesion. Determination of CSF hCG at the first presentation may be useful, because an increased CSF level of hCG precedes MRI abnormalities.

Keywords: central diabetes insipidus germinoma

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