

Polyuria-Polydipsia—First Sign of a Rare Hematological Disease

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Introduction

Central diabetes insipidus (CDI) is a rare symptom in children [1]. The aetiology of CDI in childhood is heterogeneous. Most of the causes are idiopathic, while others can be heralded by rare disorders that infiltrate the pituitary stalk such as lymphocytic hypophysitis, germinoma, sarcoidosis or Langerhans cell histiocytosis (LCH) [2,3].

Case Presentation

We report a 12-year-old girl who presented in the Endocrinology Department for weight gain in the last 3 years and menstrual irregularities. From family history, we learned that the mother was diagnosed with Graves' disease and brother had hypothyroidism. On endocrine examination, she had abdominal obesity, goiter, polyuria (diuresis 10 L/24 h) and polydipsia (ingested liquids 10 L/24 h). The laboratory assessment revealed high cholesterol level, urinary density < 1005, autoimmune thyroiditis with euthyroid status, with no hormonal deficiencies. The fluid deprivation-vasopressin test demonstrated central diabetes insipidus (CDI) and desmopressin treatment was initiated. Imaging examination showed a pituitary microadenoma and thickening of the pituitary stalk. Thus, lymphocytic hypophysitis, sarcoidosis and granulation tissue lesions were considered in the differential diagnosis.

The diagnosis of LCH was suspected by bone X-ray showing multiple lytic bone lesions in the left femoral bone and skull. The disease was further confirmed by pathology of the biopsy specimen from the skull lesion. The cytostatic treatment was initiated, and after six doses, the 18F FDG PET CT revealed two lytic lesions in both femoral bones, and the cytostatic treatment continued to date.

Conclusions

LCH is a rare disease with reported incidence in 2.6–8.9 per million children who are <15 years of age each year [4]. Currently, children with organ involvement who do not have a good response to chemotherapy and have diabetes insipidus are the most difficult to manage [5].

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