A RARE CASE OF DIABETES MELLITUS TYPE 1 IN A CHILD WITH NEUROFIBROMATOSIS TYPE 1

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Introduction

Neurofibromatosis type 1 (NF1) is an autosomal dominant multisystemic neurocutaneous disorder characterized by an increased risk of benign and malignant tumor formation (neurofibromas, glioma and gastrointestinal stromal tumor). The incidence has been described to be around 1 in 2,500–3,500 live births, and the estimated prevalence is 1 in 4,000–5,000. Although it was first described by von Recklinghausen in 1882, the formal diagnostic criteria were published a century later, in 1987. The diagnosis can be reached based on the positive family history as well as the following clinical features: a) pigmentary abnormalities such as café-au-lait macules which usually consist the earliest manifestation, axillary or inguinal freckling and Lisch nodules, b) neurofibromas, c) distinctive osseous lesion and d) optic pathway glioma. Diabetes mellitus is rarely seen in association with NF1, as to our knowledge, there have only been reported 3 cases of NF1 and diabetes mellitus.

Case description

A 12-year-old boy, diagnosed with NF1 at the age of 4, and no other relevant personal or family history, was admitted to the hospital presenting polydipsia, polyuria, and enuresis nocturna for the last ten days. On his physical examination, he was haemodynamically stable, with multiple café-au-lait spots (>20) of 1–5 cm in diameter and one plexiform neurofibroma on his neck. Multiple subcutaneous neurofibromas were found on his abdomen palpation. Laboratory tests revealed a blood glucose level of 488mg/dL (27.1mmol/L), a glycated haemoglobin level of 9.6% and traces of ketone bodies while liver and renal function was normal; urinalysis revealed glucosuria (3+) and ketonuria (4+). Acidosis was not detected on his blood gases analysis (pH=7.34). Insulin level was 5.4μIU/L and C-peptide was 1ng/mL. His thyroid and celiac antibodies tests were negative. Insulin treatment was initiated, and normoglycemia was maintained. His course was uncomplicated. Later, the patient underwent further investigation (abdominal ultrasonography and MRI) to search for somatostatinoma, on the one hand as possible manifestation of NF1 and on the other as likely cause of diabetes mellitus.

Conclusion

Studying the current literature, an ongoing association between NF1 and autoimmune diseases, such as juvenile arthritis, multiple sclerosis etc., is revealed. As the number of reports on the coexistence of NF1 and autoimmune diseases increases, including diabetes mellitus type 1, an association rather than a coincidence becomes more likely, but further investigation needs to be done.

References

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