Search Histiocytosis X facing insipid diabetes with thickened pituitary stalk

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INTRODUCTION

Insipid diabetes is a rare disease in pediatric endocrinology. Facing a thickened pituitary stalk on MRI pituitary, the main diagnosis to mention are: dysgerminoma, histiocytosis, sarcoidosis, autoimmune hypophysitis [1]. Central Diabetes Insipidus can be the first manifestation of Langerhans Cell Histiocytosis [2]. Histiocytosis is a rare and often underdiagnosed cause.

CASE REPORT

We report the case of a teenage girl who presented polyuria - polydipsia syndrome at the age of 14 years consequent full central insipid diabetes confirmed by water restriction test.

MRI showed pituitary stalk thickened measured at 2.9 mm with a loss signal of post-pituitary.

The initial analysis were negative (blood and cerebrospinal fluid markers of dysgerminoma, body skeletal radiographies, bone scintigraphy, ear scan, lung scan, auto-immune research).

The patient was treated with Desmopressin.

Later, the pituitary stalk has grown to 5 mm (Figure 1 and 2) with apparition of an asthenia, a weight gain, and several anterior pituitary deficits in gonadal and somatotropic axis. The Growth Hormone Deficit (GHD) could not be supplemented until the diagnosis of dysgerminoma had not been dismissed.

A stalk biopsy was performed at the age of 15 ½ showing nonspecific inflammatory tissue and CD1a + marker in favour of Histiocytosis X. GH was started after the diagnosis confirmation.

Complementary analysis showed bone defects (Figure 3) and asymptomatic atypical lung nodules which were not present during the initial radiographies.

After two years, the last MRI showed a pituitary stalk increased to 9 mm with compression on the left side of optic chiasm (Figure 4), posing an indication of systemic treatment for Histiocytosis.

CONCLUSION

Histiocytosis X is a difficult and late diagnosis. Annual repetitions skeletal radiographies in search of bone’s lesion, skin biopsy, lung scanner [2] and repeat pituitary MRI before placing the indication of tige biopsy must be part of the key elements of diagnosis.

References