

A Rare Case of Central Diabetes Insipidus in a Pediatric Patient

Um Caso Raro de Diabetes Insípida Central em Idade Pediátrica

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Palavras-chave: Criança; Diabetes Insípida Neurogénica/diagnóstico; Hipófise/diagnóstico; Hipofisite Autoimune/diagnóstico

Central diabetes insipidus (CDI) in children is rare^{1,2} and pituitary stalk thickening (PST) is present in approximately one-third of cases.¹ While histopathological analysis is usually required to reach a definitive diagnosis, a presumptive diagnosis of lymphocytic infundibulo-neurohypophysitis (LINH) can be made in cases with self-limited PST and decreasing anterior pituitary size, even without histological confirmation.¹ The optimal management strategy for children with LINH remains unclear.

A 3-year-old white girl was admitted to our hospital with a one-month history of polyuria and polydipsia. There was no history of head trauma, febrile illness, skin rash, bone pain, or chronic cough. Blood chemistry tests, including glycemia, urea, creatinine, and electrolytes, were all within normal range. Urinalysis showed hypotonic urine and no glycosuria. A water deprivation test was inconclusive, showing only a partial increase in urine osmolality. A brain magnetic resonance imaging (MRI) showed diffuse thickening of the pituitary stalk and absence of neurohypophysis bright spot on T1-weighted images (Fig. 1A). Central diabetes insipidus secondary to a pituitary lesion was suspected, and desmopressin was initiated. Anterior pituitary function was normal. Further investigation excluded Langerhans cell histiocytosis and germ-cell tumors (β -human chorionic gonad-

otropin and α -fetoprotein were negative both in blood and in cerebrospinal fluid). No abnormalities were found on whole-body radiographs or on abdominal, renal, and cervical ultrasound. Whole body 18-fluoro-2-deoxy-D-glucose positron emission tomography/computed tomography (FDG-PET/CT) scan showed no abnormal tracer uptake. Over time, serial MRI showed spontaneous reduction of the PST (Fig. 1B), and a presumptive diagnosis of LINH was made. After careful consideration, the medical team decided not to perform a stalk biopsy and not start immune-modulating therapy, while maintaining close clinical and neuroimaging monitoring. During follow-up, the patient developed growth hormone deficiency and central hypothyroidism. Presently, six years after the initial presentation, she remains clinically stable, without evidence of other pituitary hormonal deficits or other abnormalities.

Childhood-onset LINH is extremely rare²; its main clinical presentation is CDI,^{1,2} and, usually, anterior pituitary function is unaffected at diagnosis.^{1,3} However, lymphocytic infiltration may extend to the adenohypophysis,² just like in our patient. Typical neuroimaging findings include diffuse PST, and absence of neurohypophysis hyperintensity.⁴ Retrospective studies describe spontaneous regression or even a complete normalization of the PST in most patients with LINH.^{1,2}

While histopathology remains the gold-standard for diagnosing hypophysitis,^{1,2,3} the risk of stalk biopsy must be weighed. In our case, biopsy was not performed, due to the risk of complete stalk sectioning. Therefore, diagnosis was assumed in the absence of histological findings, and close monitoring ensued.

To date, there is scarce information to guide optimal treatment.^{1,5} Both clinical surveillance and corticosteroids

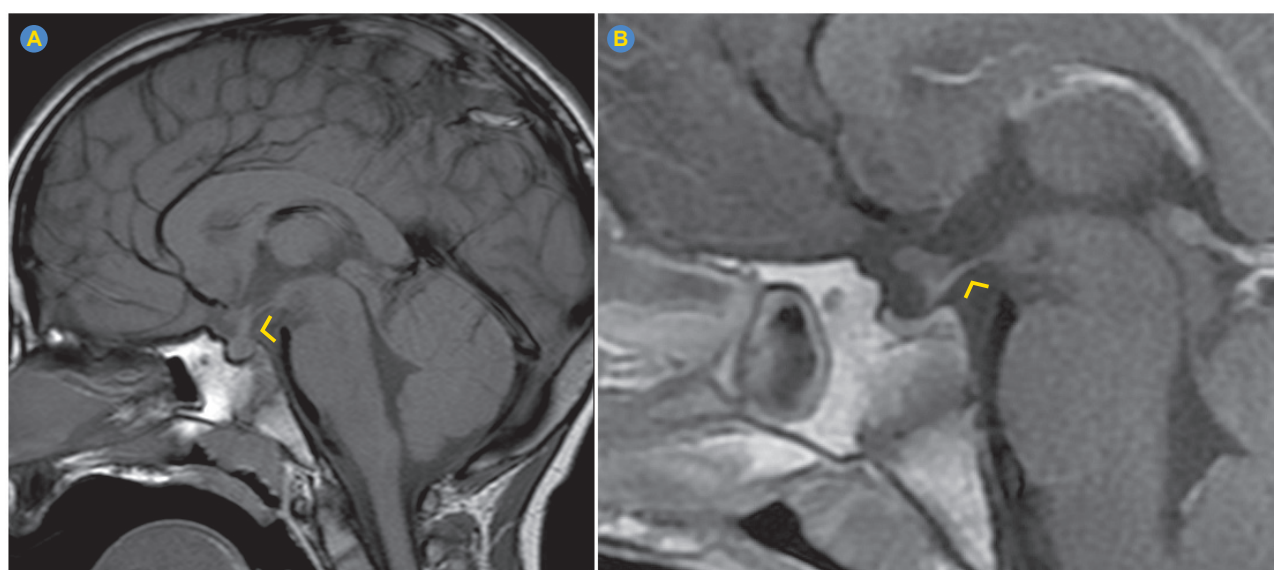


Figure 1 – Head MRI at diagnosis with sagittal T1-weighted turbo-spin echo (TSE) sequences. There is a thickening of the pituitary stalk (yellow arrowhead) and an absence of the neurohypophysis bright spot (A). Follow-up head MRI at 4-years, with sagittal T1-weighted TSE sequences, after intravenous gadolinium injection. Normal thickness of the pituitary stalk, with a small adenohypophysis (height = 3mm) (B).

have been reported as successful management strategies.⁵ Recently, conflicting clinical outcomes of high-dose corticosteroids have been reported, namely referring to pituitary axis recovery.⁵ Furthermore, given that the inflammatory process is mostly self-limiting,¹ spontaneous recovery in patients using a conservative approach can also be observed, and therefore the risks and benefits of corticosteroids should be taken into consideration when deciding on a treatment approach.⁵ Initial clinical surveillance has been deemed appropriate for patients presenting with mild symptoms and/or signs.⁵ The decision in our patient was not initiating immune-modulating treatment, and a spontaneous regression of the PST was observed.

In conclusion, the differential diagnosis in children presenting with CDI and PST is challenging; although rare, childhood-onset LINH must be considered, namely in cases of CDI. While definitive diagnosis requires pituitary biopsy, a presumptive diagnosis can be made based on clinical, biochemical, and neuroimaging assessment. Although there is no consensus about the optimal management strategy, clinical surveillance may be appropriate in selected patients. Close follow-up with serial neuroimaging, hormonal assessment, and clinical evaluation is mandatory. Prospective studies are needed in order to develop evidence-based guidelines for its management.

AUTHOR CONTRIBUTIONS

ARH: Literature search, study design, writing of the

manuscript.

MML: Literature search, writing of the manuscript.

BR, MLS: Critical review of the manuscript.

CP: Study design and critical review of the manuscript.

All authors approved the final version to be published.

PROTECTION OF HUMANS AND ANIMALS

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the Helsinki Declaration of the World Medical Association updated in 2013.

DATA CONFIDENTIALITY

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

PATIENT CONSENT

Obtained.

COMPETING INTERESTS

The authors have declared that no competing interests exist.

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