Sir,

Congenital nephrogenic diabetes insipidus (CNDI) is a rare disease characterized by the inability of the kidney to respond to arginine vasopressin (AVP). The absence of the neurohypophyseal ‘bright signal’ on T1 sequence magnetic resonance imaging (MRI) is considered as an argument in favour of the diagnosis of central diabetes insipidus (CDI). This observation is challenged as we hereby present a case of a child diagnosed with CNDI and who did not present MRI pituitary bright signal.

A 6-month-old male presented with failure to thrive, polyuria and polydypsia. Family history revealed that the mother, 35 years of age, had been presenting polydypsia and polyuria, and she was investigated at the age of 6 years with no concluding diagnosis. The patient’s physical exam showed a weight of 5215 g (−3 DS) and clinical signs of dehydration. The patient’s plasma sodium level was 155 mmol/L, osmolality 305 mOsm/kg and urine osmolality 150 mOsm/kg. Brain MRI showed in T1 sequences the absence of the posterior pituitary bright signal suggesting the diagnosis of CDI (Figure 1). The child was treated with synthetic AVP analogue 1-desamino-8-D-arginine vasopressin (DDAVP) without improvement, which led to the consideration of CNDI. The diagnosis was confirmed by an elevated serum level of AVP of 214 pmol/L (reference value ≤4.34 pmol/L) and by genetic analysis demonstrating a T106C mutation in the V2R (X-linked CNDI). The child was treated with thiazide diuretic and increased fluids with restricted sodium intake. This resulted in catch-up growth and improved neurological development. A follow-up MRI was performed 6 months after the start

Fig. 1. Sagittal T1 image shows absence of the posterior pituitary bright signal.
of therapy with the same technique. At that time, the child’s weight had improved to 9310 g (−1.5 DS) corresponding to a gain of 22 g per day, and he did not present any clinical signs of dehydration and had a normal plasma level of sodium (140 mmol/L). MRI showed that the bright signal was still absent.

Brain MRI was also performed on the child’s mother presenting the same gene mutation (R106C). As expected in CNDI, the posterior pituitary bright signal was present on T1 images (Figure 2).

Discussion

MRI represents the first choice imaging modality in CDI. It displays a characteristic bright signal of the posterior lobe of the pituitary gland on T1-weighted images and can help to evaluate the functional status of the neurohypophyseal system [1]. The origin of this bright signal is still a matter of debate, and multiple theories have been evoked.

Mark et al. reported that the bright signal could be due to the fat pad in the sella [2]. However, Nishimura et al. insisted that the bright signal is caused by neurosecretory granules containing vasopressin [3]. On the other hand, Kucharczyk et al. reported that the bright signal represents the AVP content in the neurosecretory granules and/or the intracellular lipid droplets in the glial cell processes of the posterior lobe [4]. This bright signal is physiologically absent in ∼10% of healthy individuals [5], and the signal intensity depends on the production and secretion of AVP [6]. A literature review concludes that the absence of an MRI posterior bright signal in patients with DI is usually associated with hypothalamic neurohypophyseal axis lesions and correlates closely with undetectable plasma AVP levels.

Little is known about posterior pituitary bright signal and nephrogenic diabetes insipidus. Five cases in only two papers were found [1,7]. Three of them did not have the bright signal on MR T1W imaging. The number of cases found was thus limited, and the description of these cases was neither complete nor detailed as authors have not focused on the correlation between these cases and the presence or absence of the signal.

In our case report of CNDI, the bright signal was undetectable on MR T1W imaging, and this remained unchanged, even after good rehydration. This observation raised the question of a possible correlation between the absence of the bright signal and the type of gene mutation. For this reason, we performed an MRI on the child’s mother, a carrier of the same gene mutation (R106C), and discovered that she has a bright signal, indicating that the absence of signal was not related to the R106C mutation. We conclude that our patient could be classified within the percentage of normal patients described in the literature with idiopathic absence of posterior pituitary hyperintensity. Thus, the hypothesis that the bright signal is due to the presence of AVP in the neurosecretory granules should be further investigated.

The interest of this case report is to raise the pediatric nephrologists’ attention to the fact that the diagnosis of DI is based on clinical presentation associated with abnormal blood and urine osmolality, and AVP concentration. MRI does not necessarily reflect the origin (central or nephrogenic) of diabetes insipidus and should only be performed in diagnosed cases of CDI.

Conflict of interest statement. We have had no involvements that could be perceived as prejudicing the impartiality of the article reported.

Children’s Hospital of Geneva. Pediatric Nephrology, 6 Rue Willy-Donzé, 1211 Geneva 14, Switzerland E-mail: hassib.chehade@hcuge.ch

1. Maghnie M, Villa A, Arico M et al. Correlation between MRI of posterior pituitary and neurohypophyseal function in children with diabetes insipidus. J Clin Endocrinol Metab 1992; 14: 795–800
2. Mark L, Peck P, Daniels D et al. The pituitary fossa: a correlative anatomic and MR study. Radiology 1984; 153: 453–457
3. Nishimura K, Fujisawa I, Togashi K et al. Posterior lobe of the pituitary: identification by lack of chemical shift artefact in MR imaging. J Comput Assist Tomogr 1986; 10: 899–902
4. Kucharczyk J, Kucharczyk W, Berry I et al. Histochemical characterization and functional significance of the hyperintense signal on MR images of the posterior pituitary. AJR Am J Roentgenol 1989; 152: 153–157
5. Colombo N, Berry I, Kucharczyk J. Posterior pituitary gland appearance on MR images in normal and pathologic states. Radiology 1987; 165: 481–485
6. Kurokawa H, Fujisawa I, Nakano Y. Posterior lobe of the pituitary gland: correlation between signal intensity on T1-weighted MR images and vasopressin concentration. Radiology 1998; 207: 79–83
7. Moses AM, Clayton B, Hochhauser L. Use of T1-weighted MR imaging to differentiate between primary polydipsia and central diabetes insipidus. AJNR Am J Neuroradiol 1992; 13: 1273–1277

doi: 10.1093/ndtplus/sfq120