Clinical and Endocrinological Manifestations of Partial Ectopic Posterior Pituitary: a new imaging entity

McGill University Health Centre

Marina Ybarra[1,2], Rawan Hafiz[3],Julia E von Oettingen[3], Marie-Ève Robinson[3], Helen Bui[3], Christine Saint-Martin[3]

[1]INRS- Institute Armand Frappier , Laval [2]CHU - Sainte Justine Research Center, Montreal [3]McGill University Health Center, Montreal - Canada;

BACKGROUND

- Abnormal posterior pituitary gland development can be associated with a migration defect or can be due to neuro-degeneration of the hypothalamic nuclei
- Developmental abnormality of the posterior pituitary can lead to an ectopic posterior pituitary (EPP) at the median eminence or along the pituitary stalk, with partial or complete pituitary stalk agenesis
- EPP can be associated with endocrine manifestations. To our knowledge, partial ectopic posterior pituitary (PEPP) has never been reported before; the endocrinological consequences are, therefore, unknown

OBJECTIVE

To describe six cases of possible partial ectopic posterior pituitary gland (PEPP) seen on head magnetic resonance imaging (MRI) and their associated clinical and endocrinological manifestations

METHODS

Design: Single-centre case series from a tertiary Public Pediatric University Health Centre in Montreal, Canada

Participants: 2 boys and 4 girls between 8-day- and 14-year-old, with possible PEPP on head MRI

Data Collection Methods: Cases of children with possible PEPP were selected prospectively from 2005 to 2017, based on head MRI findings. History, exam findings and hormonal evaluation were extracted from the medical record, and images were reviewed and interpreted by an experienced pediatric neuro-radiologist

The authors declare no conflict of interest

RESULTS

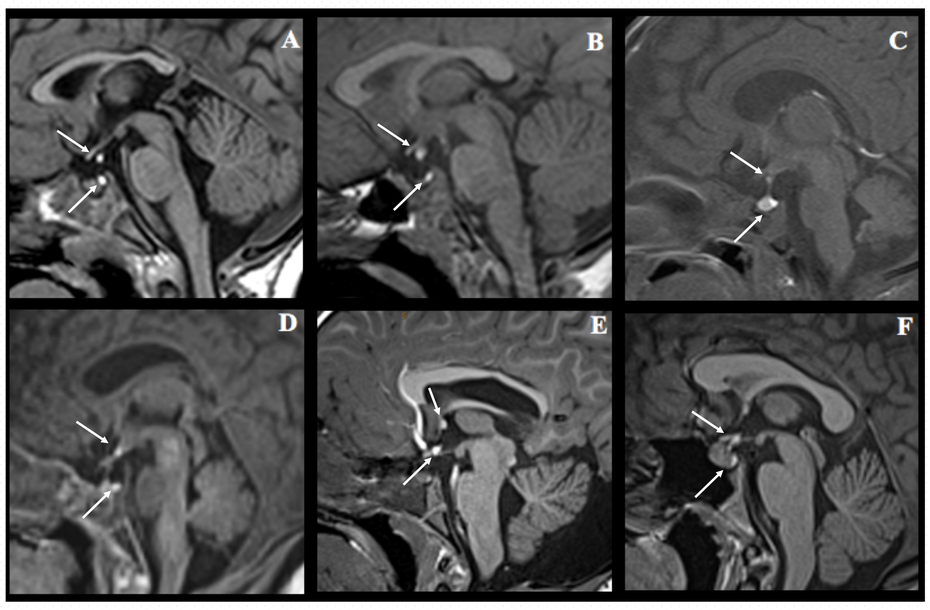
All the cases : **presence of two midline bright spots** on the thin focused T1 weighted sequences obtained with fat suppression technique:

- **one bright spot was located at the normal expected site** of the neurohypophysis in the posterior sella
- **another was in the midline median eminence or along the normal appearing pituitary stalk above the sella**, most likely corresponding to a partial presentation of an ectopic posterior pituitary gland

The possible PEPP was **associated with** different clinical phenotypes:

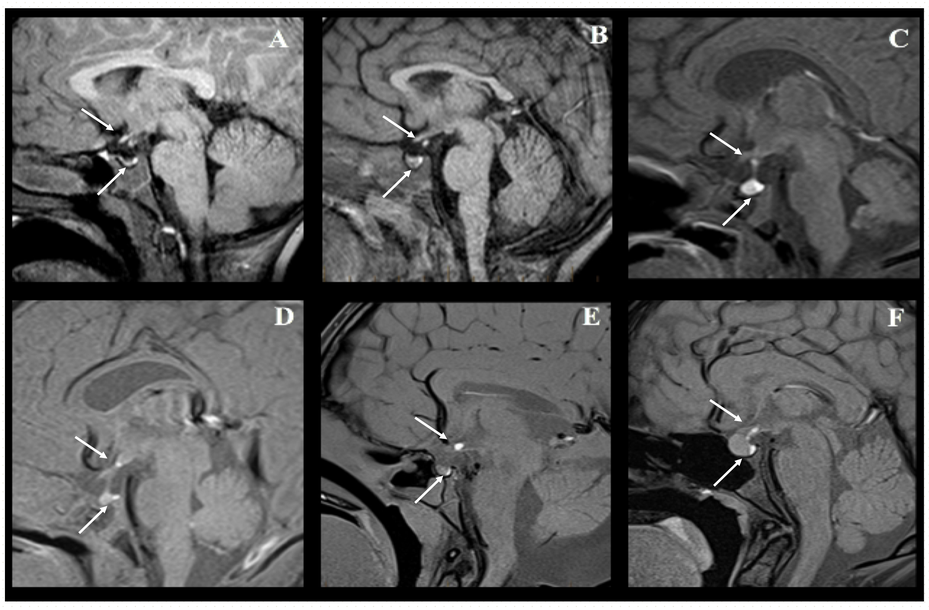
- isolated GH deficiency (1 case)
- combined TSH and GH deficiency (1 case)
- CHARGE syndrome (1 case)
- motor developmental delay (1 case)
- septo-optic dysplasia (1 case)

Figure 1: Head MRI - T1-weighted with no fat suppression



Legend : A second and ectopic T1 bright spot is noted on midline above the sella either at the floor of the V3 or at the infundibulum level.

Figure 2: Head MRI - T1-weighted with fat suppression



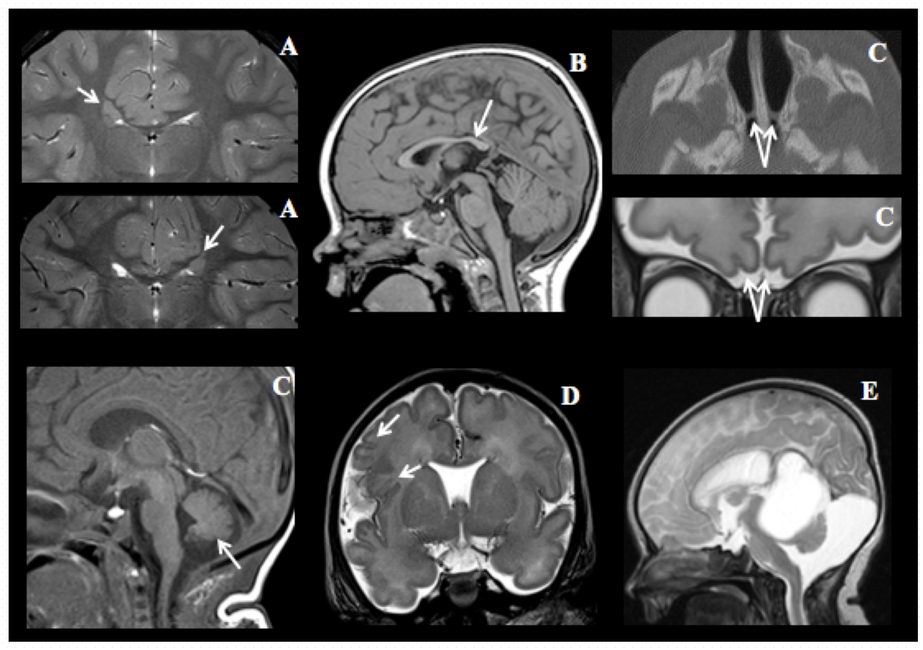
Legend: A second and ectopic T1 bright spot persists and is unchanged, noted on midline above the sella either at the floor of the third ventricle or at the infundibulum level.

Table 1: Summary of clinical and laboratory findings

	Case 1	Case 2	Case 3	Case 4	Case 5	Case 6
Age	4y6m	2y3m	8 d	19 d	7y4m	14y10m
Clinical Presentation	short stature	poor growth develop-mental delay strabismus	CHARGE Sd.	SOD	short stature Autism Arachnoid cyst	asympto-matic bilateral optic nerve swelling
Dysmorphic Features	none	short nose prominent ears small chin large forehead shorter right leg	small ears small jaw small nose hyperte-lorism	none	none	none
TSH / f-T4 (mU/L / pmol/L)	2.19/12.5	2.6/11.4	4.36 / -	1.18/7.6	5.1/7.7	1.75/11.1
Cortisol level (nmol/L)	319 random	-	678 random	522 random	741.5 peak	519 random
GH peak on Stimulation Test - ug/L	3.7	-	-	-	1.8	-
Bone age*	3y CA: 4y6m	3-12m CA: 2y3m	-	-	6y10m CA: 8y	-
Endocrine manifestations	GHD	none	none	none	GH and TSH deficiency	none

Legend: SOD: septo-optic-dysplasia; GHD: growth hormone deficiency; CA: chronological age * by Greulich and Pyle standards

Figure 3: Associated imaging features of the head MRI A, B, C, D, and E of cases 1, 2, 3, 4 & 5, respectively



Legend:. A: bilateral periventricular nodular heterotopia of grey matter, B: dysmorphic splenium of the corpus callosum C: choanal atresia, absence of olfactory bulbs and vermian hypoplasia; D: septum agenesis and perisylvian polymicrogytia; E: quadrigeminal plate arachnoid cyst and hydrocephalus

CONCLUSIONS

- To our knowledge, PEPP has never previously been described
- 2/6 of our cases of PEPP are associated with pituitary hormone deficiencies
- Long-term follow-up may reveal further endocrine manifestations
- We hypothesize that PEPP developed as a result of partial reversal of the process that initially interrupted the normal migration of the posterior pituitary gland, perhaps by micro- or macro-environmental factors