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

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BACKGROUND

• Abnormal posterior pituitary gland development can be associated with a migration defect or can be due to neurodegeneration 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

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

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

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 GHD deficiency (1 case)
• CHARGE syndrome (1 case)
• motor developmental delay (1 case)
• septo-optic dysplasia (1 case)

Table 1: Summary of clinical and laboratory findings

<table>
<thead>
<tr>
<th>Age</th>
<th>Case 1</th>
<th>Case 2</th>
<th>Case 3</th>
<th>Case 4</th>
<th>Case 5</th>
<th>Case 6</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical Presentation</td>
<td>short stature</td>
<td>short stature</td>
<td>poor growth developmental delay</td>
<td>choanal atresia</td>
<td>short stature</td>
<td>asymptomatic bilateral optic nerve swelling</td>
</tr>
<tr>
<td>Dysmorphic Features</td>
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<td>none</td>
<td>none</td>
<td>none</td>
<td>none</td>
<td>none</td>
</tr>
<tr>
<td>TSH (mU/L)</td>
<td>2.19/12.5</td>
<td>3.7</td>
<td>1.87/15.9</td>
<td>678/745.1</td>
<td>519/7.6</td>
<td></td>
</tr>
<tr>
<td>Cortisol level (nmol/L)</td>
<td>319 random</td>
<td>-</td>
<td>522 random</td>
<td>751 random</td>
<td>7.15/7.11</td>
<td></td>
</tr>
<tr>
<td>GH peak on stimulation test (mg/L)</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>6.10/mg/L</td>
<td></td>
</tr>
<tr>
<td>Bone age*</td>
<td>CA: 4y6m</td>
<td>CA: 3.12/m</td>
<td>CA: 2.1/m</td>
<td>CA: 1.8/m</td>
<td>CA: 4y6/m</td>
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</tr>
<tr>
<td>GHD</td>
<td>none</td>
<td>none</td>
<td>none</td>
<td>none</td>
<td>none</td>
<td></td>
</tr>
</tbody>
</table>

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

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

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

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

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 vermillon hypoplasia; D: septo agenesis and perisylvian polymicrogyria; E: quadrigeminal plate arachnoid cyst and hydrocephalus

The authors declare no conflict of interest

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Pituitary, neuroendocrinology and puberty

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