Congenital Duplication of the Pituitary in Two patients with Learning Difficulties, Midline Defects and Nasopharyngeal Tumours, Presenting with Central Precocious/Early Puberty

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Background

• Sophie presented with central precocious puberty aged 6.8y. She therefore had cranial imaging and was found to have duplication of the pituitary gland (DPG).

• Subsequently we identified a further case, Michael, who also had DPG, midline defects and a nasal dermoid, with severe learning difficulties and advanced puberty.

• This is a rare malformation with fewer than 40 cases described since 1880.[1]

Case 1: Sophie

• Sophie presented with a pharyngeal teratoma, excised at 39 days of age.

• She was further noted to have a cleft palate and thyroglossal cyst.

• She had multiple additional primary teeth. Moderate learning difficulties were evident by 4y.

• Subsequently, she presented at age 6.8y with precocious puberty (B2 A1 P1).

• Bone age was 8.8y. Pituitary Function: FSH 1.6mIU/L, LH <0.2mIU/L, Oestradiol 50pmol/L, otherwise normal.

• An MRI scan revealed DPG and multiple midline cervical spine abnormalities, figure 1.

Case 2: Michael

• Michael presented at 1.8y with a nasal dermoid.

• He was further noted to have a lingual hamartoma and cleft palate.

• He went on to develop speech and language delay with social and communication difficulties and severe learning difficulties.

• MRI was performed at 10.9y because of advanced pubertal status and deteriorating behaviour, and showed complete DPG, figure 2.

• There were also multiple midline cervical spine abnormalities, figure 3.

• Limited pituitary testing was performed, and was normal (Prolactin 500mIU/L FT3 5.4 pmol/L (3.8-6.0) free thyroxine 15 pmol/L (8-21).

Learning Point

• Of note, a previous MRI in each case, had failed to include pituitary/hypothalamic sequences, and therefore the DPG was missed.

Discussion

• The association of DPG with cleft palate, learning difficulties and benign nasopharyngeal tumours is recognised as DPG Plus Syndrome.[1]

• DPG was previously thought to be a very rare condition, arising during blastogenesis. The aetiology is unknown, and no genetic cause has been identified.[1]

• Although, rare, findings in our two cases were characteristic.

• Cranial imaging, including the pituitary and hypothalamus is diagnostic and should be performed in cases of benign nasopharyngeal tumours with cleft palate.

• DPG may be more common than previously recognised in view of the previous cranial imaging in both cases not detecting DPG, and these two children live in the same part of East Anglia.

Figure 1. T1 weighted MRI showing a W-shaped floor of the third ventricle with two separate infundibula

Figure 2. Two pituitary stalks are visible below the optic chiasm. The posterior pituitary bright spot of the right gland and the anterior pituitary of the left both lie adjacent to the respective cavernous sinuses. No midline stalk or pituitary fossa is visible.

Figure 3. Bifid odontoid process, wide separation of the lateral masses of C1 and partial duplication of the C2 synchondrosis, upper cervical discs and vertebral bodies.