# Pituitary Duplication – A rare and heterogenous spectrum

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## Background

Pituitary gland duplication is a very rare developmental abnormality. It is often associated with other midline anomalies including cleft palate, spinal cord and corpus callosum defects, termed duplication of the pituitary gland-plus syndrome. Duplication of the pituitary gland may arise from blastogenesis defects, with splitting of the rostral end of the notochord, though the exact mechanism is still unclear. Thus far no causative genetic mutations have been found.

## Objective

- Collaborate to describe the largest case series reported in the literature
- Characterise the different clinical phenotypes associated with duplication of the pituitary gland

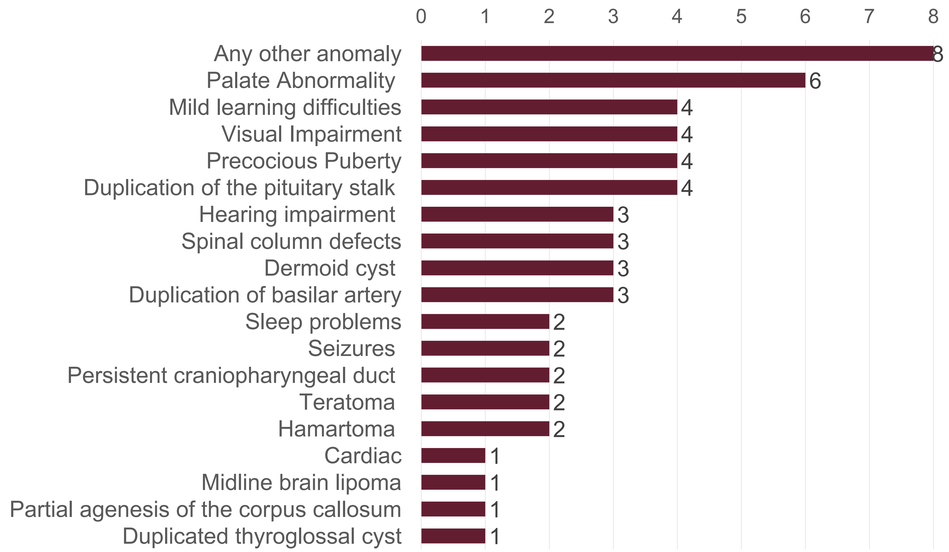
## Methods

Retrospective review of patient records of patients with pituitary duplication from 4 centres in Europe.

## Results

**Cohort:** 8 patients with pituitary duplication – 4 female, 4 male

**Clinical features:**




| Clinical feature | Further detail |
|---|---|
| Palate Abnormality | Cleft n=5<br>High arched n=1 |
| Precocious Puberty | 3 female, 1 male |
| Dermoid cyst | Nasal n=3<br>Palate n=1 |
| Hamartoma | Third ventricle n=1<br>Soft palate n=1 |
| Teratoma | Nasopharynx n=1<br>Palatine n=1 |
| Spinal column defects | Cervical cleft n=1<br>Arnold Chiari malformation and myelomeningocele n=1 |
| Hearing impairment | Chronic ear infections |
| Visual Impairment | Optic nerve hypoplasia n=2<br>Possible small optic discs n=1 |
| Cardiac | Patent ductus arteriosus and patent foramen ovale |

**Prognosis**: There was one death in this cohort in early childhood, and the eldest are now in their third decade.
**Genetic Investigation:** Six of the 8 children underwent whole exome sequencing (with trio analysis in 5 patients), but no relevant genetic variants were identified.

## Key Message

- This series highlights the wide phenotypic spectrum associated with duplication of the pituitary gland
- This helps to inform the patient, their family, and the clinicians to highlight the possible associated pathology, and to direct their management
- Future research
  - Ongoing collaboration to further characterise this syndrome, and to discover possible aetiological factors

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