Coexistence of multiple defects of pituitary hormones, short neck, hyposomatotropism and bilateral sensorineural hearing loss make diagnosis of **CONGENITAL MULTIPLE PANHYPOPITUITARISM (CMPHD)** associated with LHX3 GENE MUTATION.

Functional studies demonstrated that this variation determines the inability of the synthesized protein to bind to DNA altering activity of the normal protein.

**PRENATAL HISTORY**
- Prenatal US: **IUGR, rhizomelia, polihidramnios**
- Amniocentesis: **karyotype 46, XY**

**AT BIRTH,** at 39 GW by emergency CS:
- weight and length <3rd percentile
- head circumference < 10-25th percentile
- Physical examination: short neck, cryptorchidism and microphallus

**POSTNATAL PERIOD:**
- **early** episode of **hypoglycemia** (20 mg/dl - 1.11 mmol/l) \(\rightarrow\) infusion of D10% through umbilical vein catheter followed by naso-gastric feeding
- **recurrent** episodes of **hypoglycemia** from 8th-14th day of life \(\rightarrow\) corticosteroids
- jaundice with indirect bilirubin in 1\textsuperscript{st} week \(\rightarrow\) several cycles of **phototherapy**

**Laboratory tests**
- TSH, fT4, fT3 ↓
- PRL ↓
- FSH ↓
- LH ↓
- GH ↓
- ACTH, cortisol ↓

**INVESTIGATION in the 1\textsuperscript{st} month of life**
- **CNS US:** normal
- **Echocardiography:** normal
- **Thorax, abdomen, full skeleton X ray:** no evident abnormalities
- **Abdomen US:** mild renal pelvis dilatation, no other abnormalities
- **CNS MRI:** no morphologic alterations of pituitary gland and stalk

Recalled at hypothyroidism screening: pathological ABR and diagnosis of bilateral sensorineural hearing loss.

**Coexistence of multiple defect of pituitary hormones, short neck, hyposomatotropism and bilateral sensorineural hearing loss make diagnosis of **CONGENITAL MULTIPLE PANHYPOPITUITARISM (CMPHD)** associated with LHX3 GENE MUTATION.**

Functional studies demonstrated that this variation determines the inability of the synthesized protein to bind to DNA altering activity of the normal protein.

**Because of the same mutation was detected in his mother, asymptomatic, we excluded that this mutation alone is the cause of baby G. panhypopituitarism. SNP’s array excluded hemizygosity: this phenotype may be correlated with a digenic form.**

Now baby G. is treated with L-tiroxina, hydrocortisone and rhGH and underwent bilateral orchidopexy.
