

# Expanding the CHARGE geno-phenotype: a girl with novel *CHD7* deletion, hypogonadotropic hypogonadism, and agenesis of uterus and ovaries

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## Case presentation

A 18-year-old girl with cheilopalatoschisis, unilateral kidney agenesis, hearing problems (attributed to bilateral hypoplasia of auditory ossicles and cochlea), and aplasia of the semicircular canals presented with primary amenorrhea and subnormal breast development.

### *Clinical examination:*

- Mild facial dysmorphic features, mild hypoplastic ear cups
- Tanner A2 P2 M2

### *Technical examinations*

- GnRH-test: peak LH 0,6 IU/l, FSH 2,1 IU/l, oestradiol undetectable
- MRI of the pituitary gland: normal
- Bone age 13 years

### *Hypothesis: Hypogonadotropic hypogonadism*

*Treatment:* Oestrogen administration, gradually increased over 2 years

*Evolution:* Tanner score A2 P3 M5, persistent amenorrhea

### *Further examinations*

- Abdominal MRI: **uterus and ovaries not visualized**
- Genetic exploration: **c.3634\_3637 deletion in exon 15 of *CHD7***

## Conclusion

We describe a CHARGE girl with a hitherto unreported deletion in exon 15 of *CHD7*, with agenesis of uterus and ovaries, and with hypogonadotropic hypogonadism, thus expanding the phenotype of CHARGE syndrome.

In hypogonadotropic hypogonadism, proper treatment with sex-steroid hormones is indicated. In case of persistent primary amenorrhea evaluation of uterus and ovaries by ultrasound and MRI is advisable.