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## OBJECTIVES

To present a unique case of *PROP1* mutation and XYY syndrome in one patient. There are none other reported cases of this combination.

## METHODS

We describe the clinical case of 7-year-old boy with short stature and hypothyroidism.

## RESULTS



The boy was born at term from unrelated healthy parents. His birth length and weight were 53 cm and 3950 g respectively. The patient complained about short stature at two years old (SDS= -2.75).

Chromosome test performed at 5 y.o.: **47, XYY**.

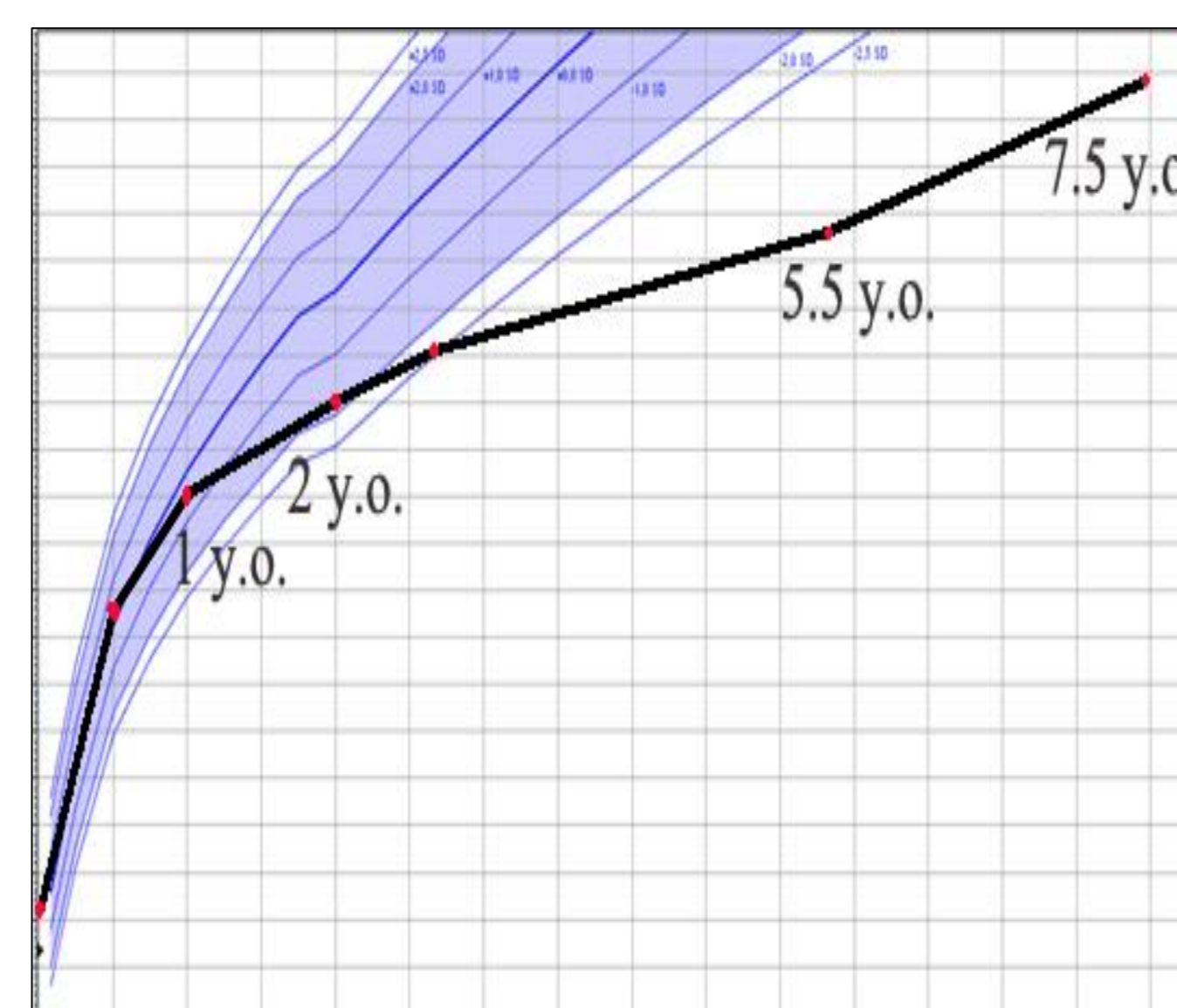
Physical examination at 7 y.o.: short stature (SDS -5.2), overweight (SDS +1.3), Tanner 1, volume testes D=S=1 ml, dry skin and pastosity.

Laboratory results: low levels of free T4 (7.0 pmol/l), IGF-1 (3 ng/ml), normal levels of TSH (1.1 mIU/L), cortisol (537 nmol/l) and prolactin (307 mIU/L).

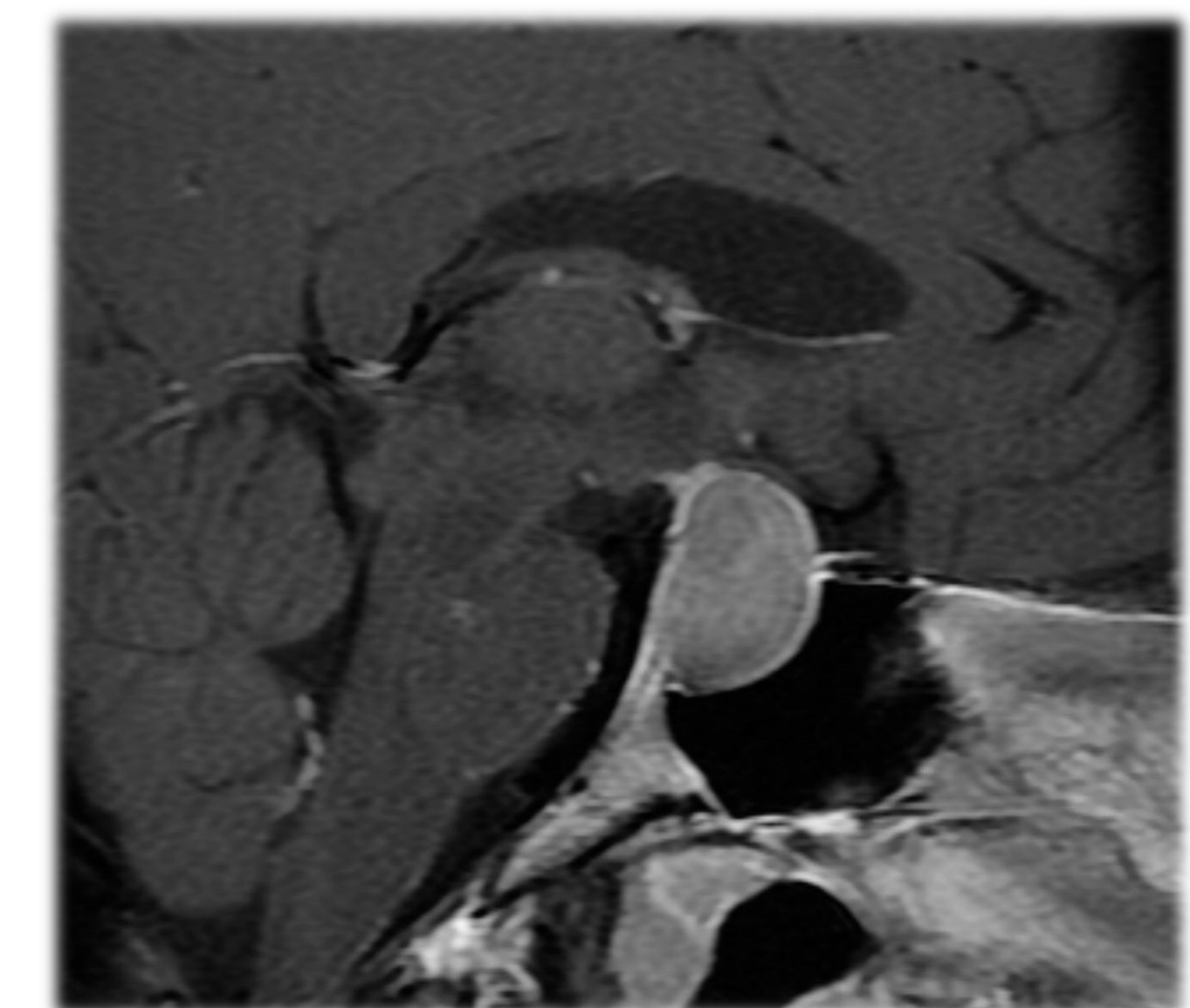
Brain MRI: anterior pituitary hyperplasia.

Genetic analysis: **compound heterozygous mutation of *PROP1* gene** (c.150delA and c.301\_302delAG).

Growth hormone and levothyroxin therapy was started. No clinical signs of XYY syndrome were found.



Growth chart



Brain MRI

## CONCLUSIONS

We presented a unique clinical case of mutations in *PROP1* gene in combination with 47, XYY karyotype. Patient monitoring is required in order to detect possible abnormalities.

## References

1. Castinetti F, Reynaud R, Saveanu A et al. MECHANISMS IN ENDOCRINOLOGY: An update in the genetic aetiologies of combined pituitary hormone deficiency. Eur J Endocrinol. 2016 Jun;174(6):P 239-47.
2. Kim IW, Khadilkar AC, Ko EY, Sabanegh ES Jr. 47,XYY Syndrome and Male Infertility. Rev Urol. 2013;15(4):188-96.

Authors have nothing to disclose

