Mutations in \textit{PROP1} gene in combination with 47, XYY karyotype: case report

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**OBJECTIVES**

To present a unique case of \textit{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.: \textbf{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: \textbf{compound heterozygous mutation of \textit{PROP1} gene} (c.150delA and c.301–302delAG).

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

**CONCLUSIONS**

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

Authors have nothing to disclose

**References**
