

NONCLASSIC CYP21A2 DEFICIENCY: DIAGNOSIS AND CLINICAL EVOLUTION

C. Luzuriaga Tomás, C. Naranjo González, L. Bertholt Zuber, B. Ezquieta Zubicaray. Paediatric Endocrinology. Hospital Universitario Marqués de Valdecilla. Santander, Spain.

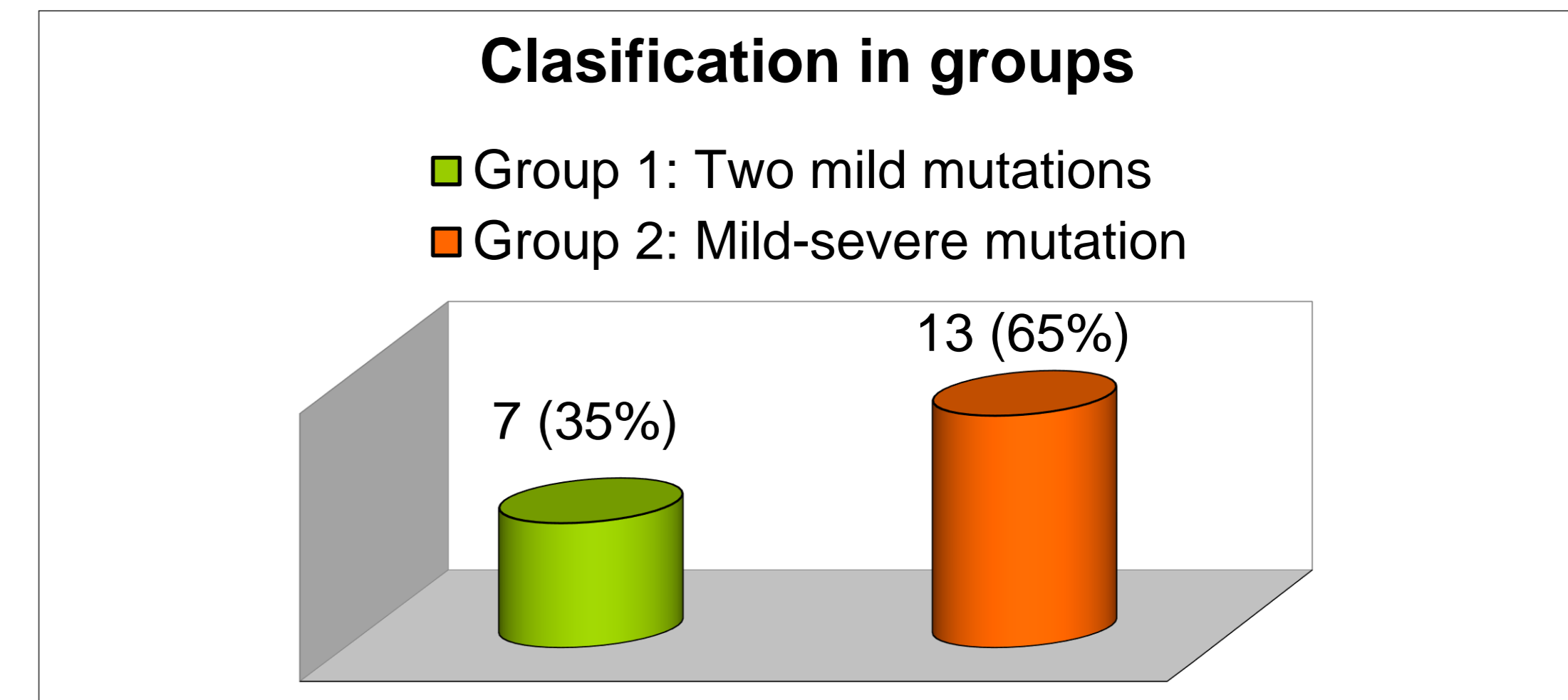
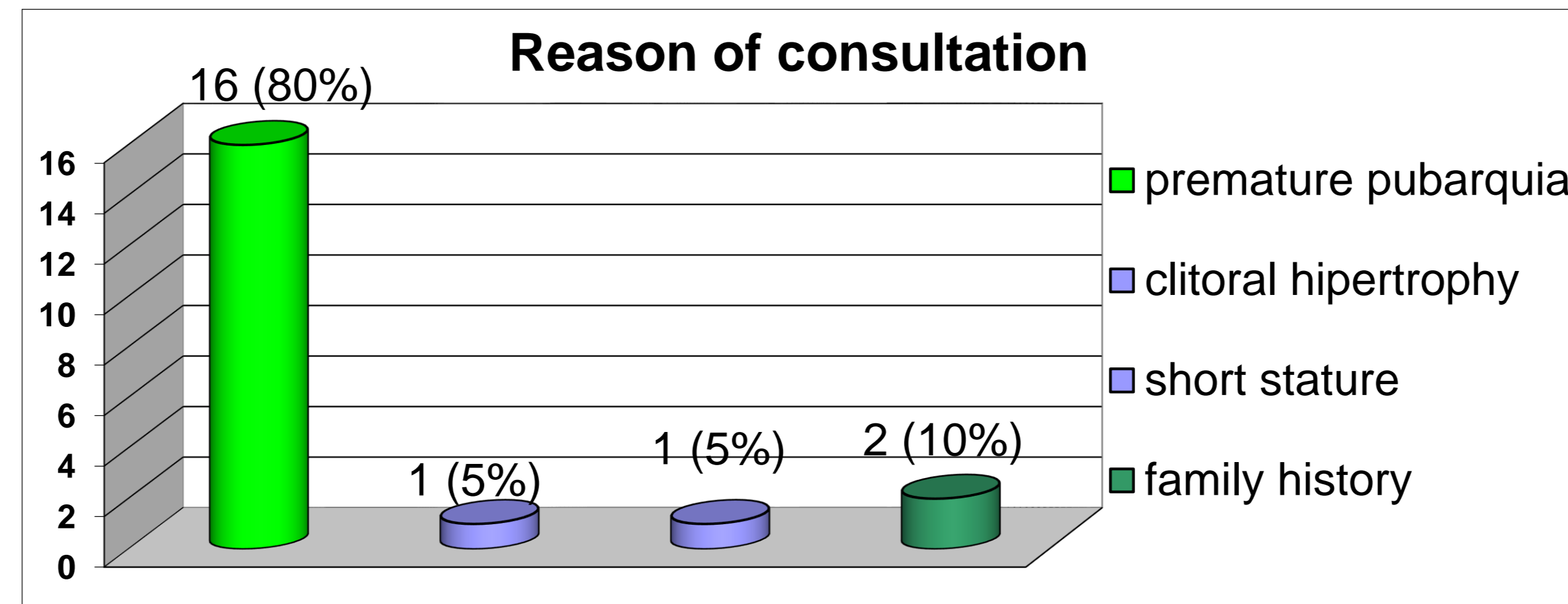
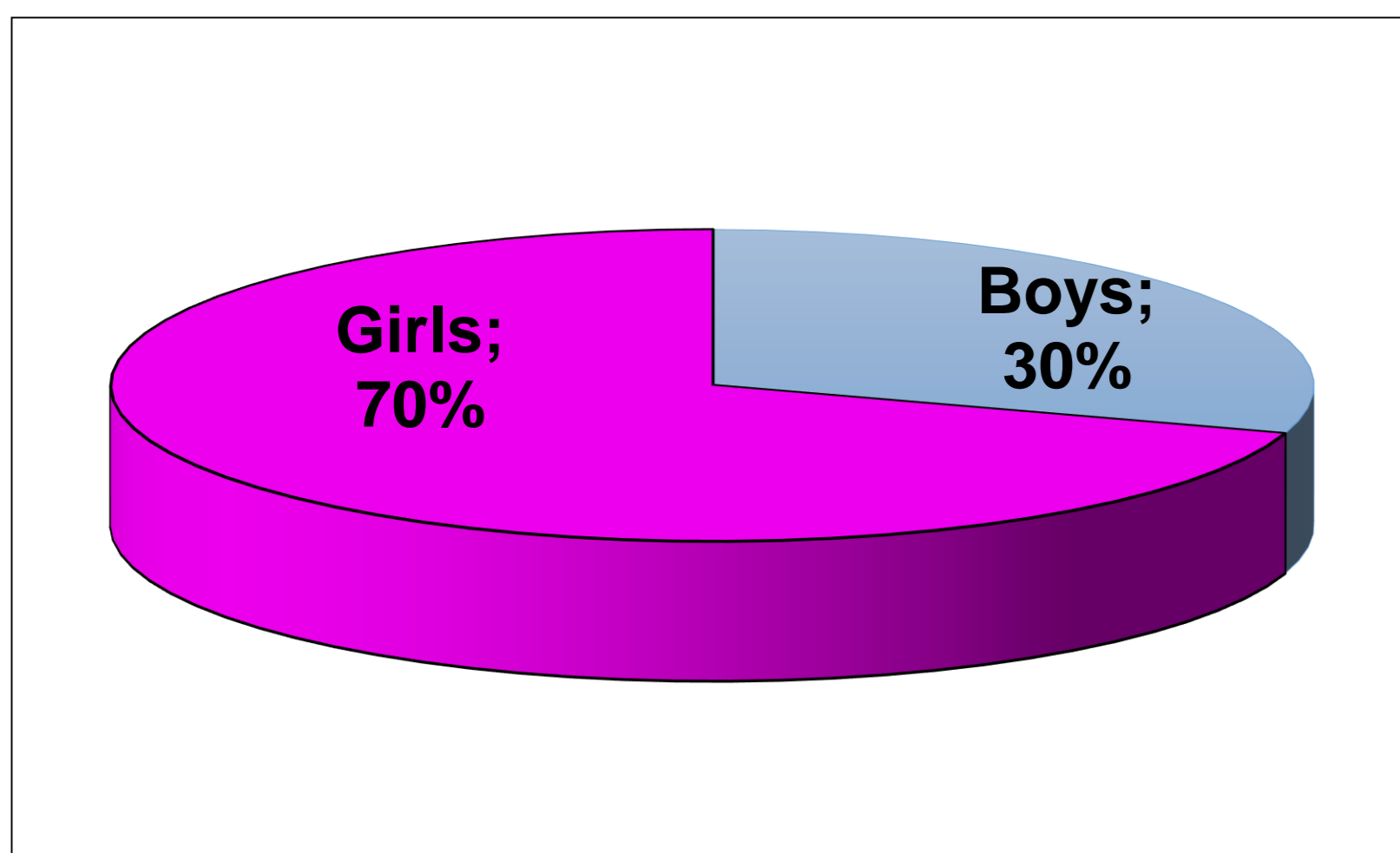
Background: The nonclassic congenital adrenal hyperplasia (CAH) is often presented as isolated premature pubarche in childhood. Definitive diagnosis is genetic.

Objective and hypothesis: To describe patients diagnosed in our hospital, clinical signs and laboratory results, that lead to genetic study. Analyze adult height and reproductive function.

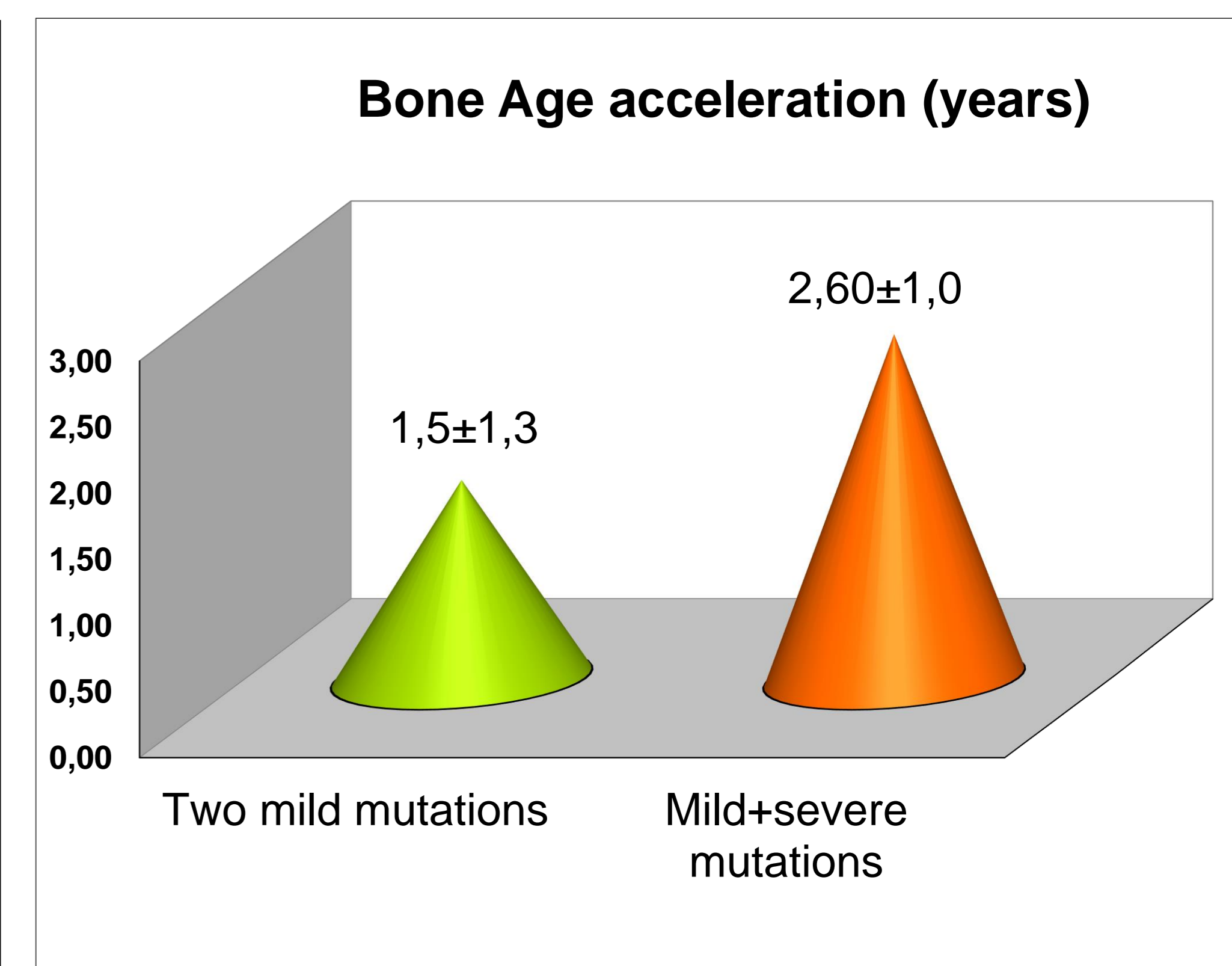
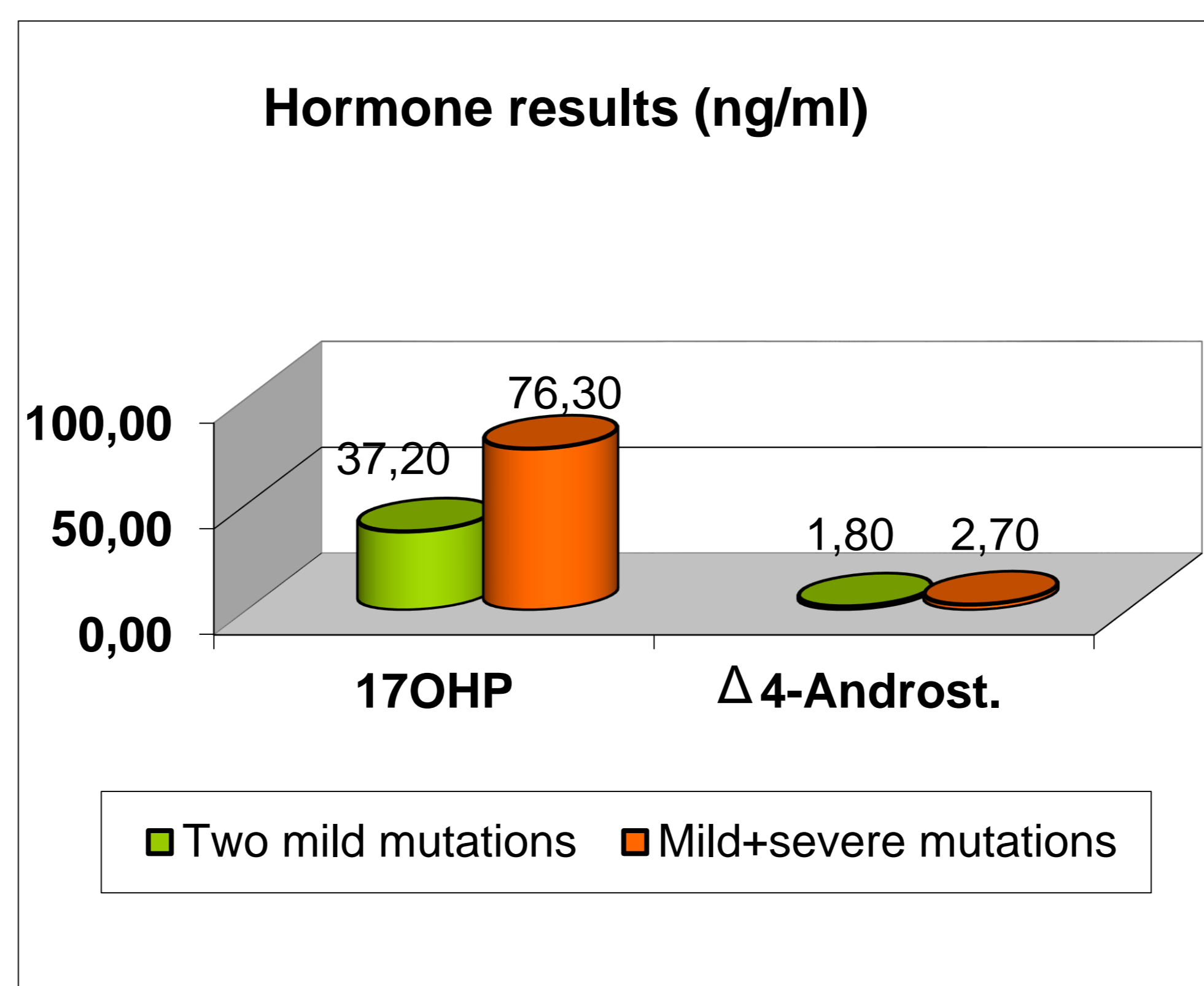
Method: Descriptive retrospective study of our population with genetic confirmation.

Results

We analyzed 20 patients: 14 females and 6 males. Age at diagnosis 7.54 ± 2.10 years (3 – 12.34 years).

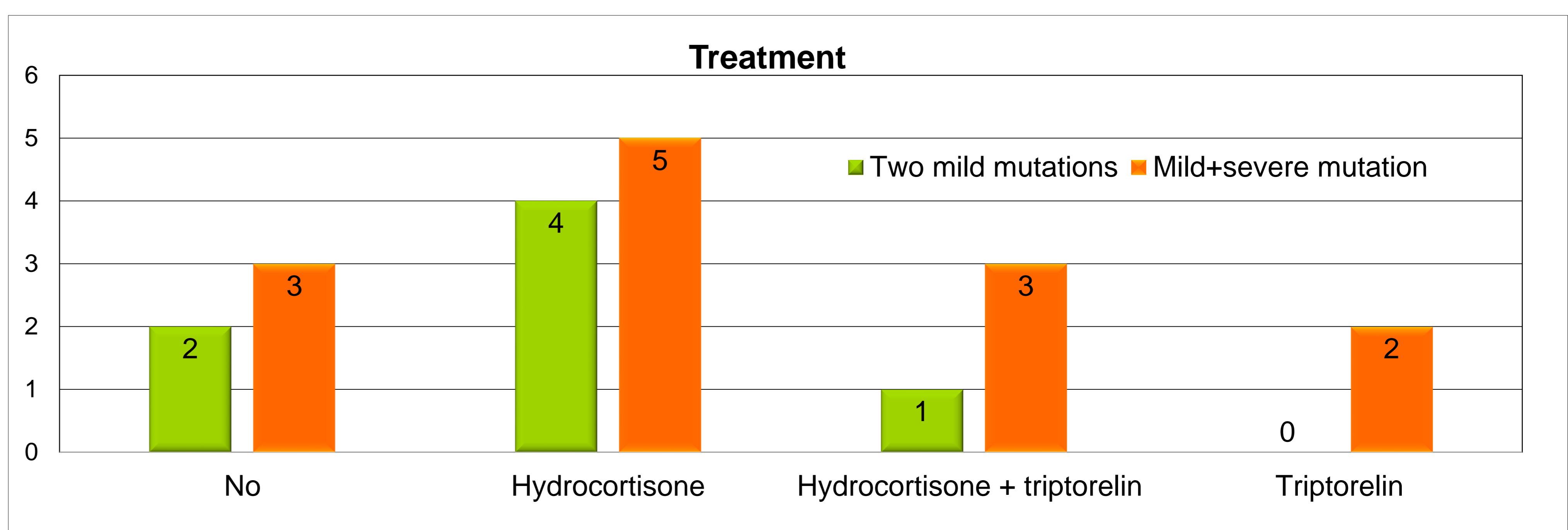


MUTATIONS	PATIENTS	TYPE	GROUP
p.Val282Leu + p.Val282Leu	6	Mild-mild	1
p.Val282Leu + p.Pro454Ser	1	Mild-mild	1
p.Val282Leu + c.1205_1206delAT	1	Mild-mild	1
p.Val282Leu + [c.293-13A/C>G; c.332-339del]	1	Mild-severe	2
p.Val282Leu + c.332-339del	2	Mild-severe	2
p.Val282Leu + p.Gln319Stop	1	Mild-severe	2
p.Val282Leu + [p.Gln319Stop; p.Ile173Asn]	1	Mild-severe	2
p.Val282Leu + [p.Gln319Stop; p.Arg357Trp]	3	Mild-severe	2
p.Val282Leu + gene conversion	3	Mild-severe	2
p.Val282Leu + c.293-13A/C>G	1	Mild-severe	2



Significant difference in hormone results.

Significant difference in bone age acceleration.



Age at diagnosis 7.54 ± 2.10 years (3 – 12.34 years).

- Group 1: 7.3 ± 2.9 years.
- Group 2: 7.6 ± 1.5 years.

Reproductive function:

- No primary amenorrhea.
- Functional ovarian hyperandrogenism (PCOS): 2 patients (Group 2).
- Azoospermia: 2 patients (Group 1: 1, Group 2: 1).

	GROUP 1	GROUP 2	TOTAL
FINAL HEIGHT	5	7	12
Exceed target height	1	1	2
Under target height	1 (untreated)	1 (treated)	2
Adult height $p < 3$ (Spanish population)	1	-	1

Conclusions

In our study predominate the presence of severe mutation, which must be taken into account for genetic counseling.

Premature pubarche and advanced bone age are the main clinical signs.

We got good results of adult height in both groups with hydrocortisone treatment, triptorelin or combined. Unable to ensure that no treatment deteriorate adult height. Larger studies are necessary.

In this type of CAH we recommend monitoring reproductive function.