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Introduction

46 XY sex development disorders are a group of rare congenital conditions in which chromosomal, gonadal or anatomic sex is atypical. Less than 20% of cases have a precise genetic diagnosis.

We report here the case of a patient suffering from a 46 XY sexual development disorder secondary to the MAMLD 1 gene mutation.



Before surgery



After a cure of hypospadias , bifurcated scrotum, and several courses of medical micropenis therapy

Observation

The patient is a nine month infant who was admitted for abnormalities of the external genital organs.

The physical examination, revealed a 1.5 cm micropenis with posterior hypospadias, and normal positioned gonads.

Blood karyotype showed 46 XY chromosome formula.

Exocrine testicular function was found to be normal with an AMH level of 236,9 ng/ml, while endocrine function assessments are planned.

The genetic study revealed a new mutation of the MAMLD 1 gene (c.G 2217 A:p.W739X). The patient has benefited from a cure of hypospadias and bifurcated scrotum, as well as several courses of medical micropenis therapy (cutaneous dihydrotestosterone treatment).

Discussion and conclusion

The MAMLD 1 gene is located at the position 28 of the long arm of the X chromosome. (1)

This gene's mutation is responsible for the fetal Leydig cells function alteration during the critical period of sexual development. At birth, it leads to a 46 XY sexual development disorder.

(2)

Therefore, testicular function is most often conserved during infancy, but it needs surveillance as it may deteriorate in long term.

(2)

Not conflict of interest

References:

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