MAMLD 1 gene mutation and 46 XY sex development disorder: a case report

B. Bousyf (Dr1), Y. Lazreg (Dr1), M. Tajir (Pr)2, H Latrech* (Pr)1.

1. Endocrinology-Diabetology-Nutrition Department, Mohammed VI University Hospital Center, Mohammed I University, Oujda, Morocco.
2. Laboratory of epidemiology clinical research and public health, Faculty of Medicine and Pharmacy, Mohammed I University, Oujda, Morocco.

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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.

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:


(2) Yasuko Fujisawa and al. Long-term clinical course in three patients with MAMLD1 mutations. Endocrine Journal 2016, 63 (9), 835-839