Gonadal dysgenesis, 46 XY about 5 familial cases

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Introduction

The 46 XY DSD sexual disorders are responsible for a range of phenotypic impairments, ranging from an ambiguous phenotype to a complete female phenotype. This is often a sporadic disease.
In this context, we report 5 cases of gonadal dysgenesis46 XY belonging to the same family and of particular phenotypic expression thus posing the problem of etiopathogenic link with genes involved in sexual differentiation

Results

We report 5 patients belonging to the same family of 10 members from a consanguineous marriage, presenting for a micropenis with bilateral cryptorchidism and gynecomastia. The mean age at diagnosis was 30.5 years (range: 17-37 years).
On examination, the phenotype was of ambiguous type with a large average size of 182 cm without dysmorphic syndrome in all cases. The size of the penis was between 1 and 2 cm (<- 3DS); a gynecomastia (S2-S5) was present in 4 cases, with gonads absent in 2 cases, ectopic at the inguinal level in 2 cases and in place but hypoplastic in the fifth brother.

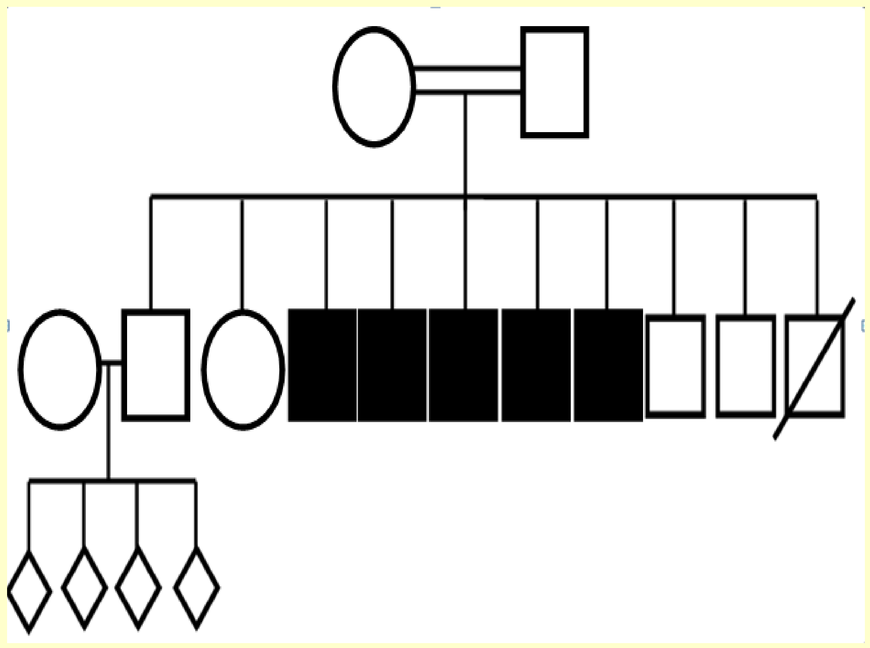

Fig:1 Family pedigree

Ultrasonography and pelvic CT had confirmed the presence of two gonads in the inguinal position in two cases.
The karyotype performed in 3 cases showed a homogeneous chromosomal formula compatible with a male genetic sex, type 46 XY (14 mitoses).
Hormonal exploration showed testosterone levels that collapsed to an average of 0.2 ng / ml, contrasting with high levels of gonadotropins, especially FSH, averaging 81 mIU / ml (range: 55-110); the average LH level was 29 mIU / ml (range: 9-47).
The AMH level was undetectable in the index case whereas it was 21.17 ng / ml (1.5-11.8) in a control from the family (Mr B).
The diagnosis of a familial gonadal dysgenesis is evoked and makes use of the biomolecular study of genes involved in testicular differentiation.

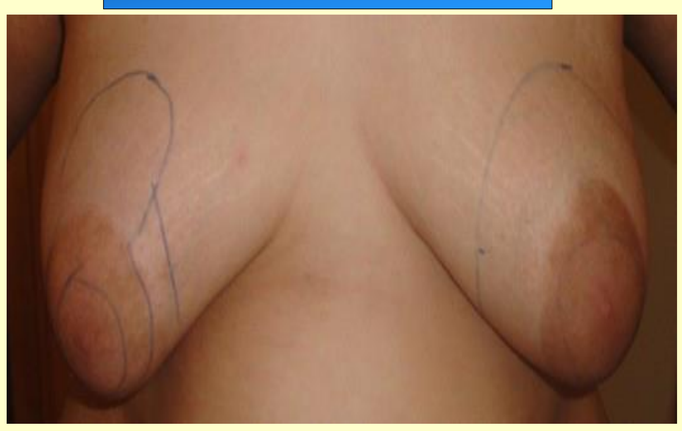

Fig:2: Gynecomastia

Fig:3: Micropenis

CONCLUSION

The abnormalities of sexual differentiation cover a wide spectrum of phenotypic and genotypic abnormalities and pose a real etiological problem; our cohort being particular by the occurrence of a table of gonadal dysgenesis in 5 members of the same family, and certainly involving one or more genes involved in the cascade of sexual differentiation. Certainly the progress of molecular biology will be of great help for understanding the phenotypic variability as well as the atypical aspects of gonadal dysgenesis.