Gonadal Mosaicism - A Case Report

Rodrigues, Joana[1]; Martins, Ana Rita[1]; Coutinho, Ana Luísa[1]; Hauwaert, Roxane[1] ; Mourinha, Vera[1]; Almeida, Diana[1]; Ferreira, Ângela[1]

[1]Centro Hospitalar Universitário do Algarve - Unidade de Faro, Portugal

Objetives

Gonadal mosaicism has an important role in disorders of sexual development. There are a wide range of clinical manifestations, varying from sexual ambiguity, postnatal virilization or a normal male or female phenotype. We describe the evolution of a pregnancy with prenatal diagnosis of mosaicism 45 X0 / 46 XY and the postnatal period.

Methods

Healthy pregnant women, multiparous, was referred at 21 weeks and 2 days for suspected ambiguous genitalia in the second trimester ultrasound (*Figure 1*). Pregnant women chose to collect blood for fetal DNA. After the result, amniocentesis was performed and QF-PCR and karyotype analysis were requested. Subsequent management included fetal echocardiogram and ultrasound reassesments.

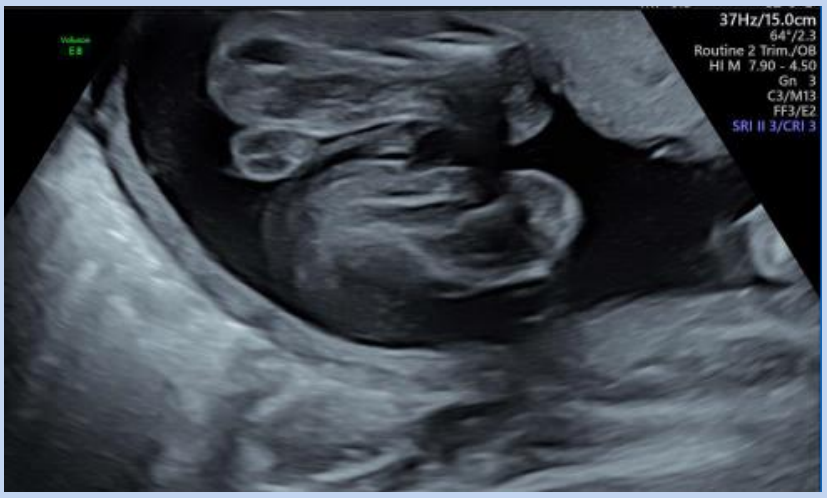

Figure 1. Image from the second trimester ultrasound where ambiguous genitalia was suspected.

Results

Fetal DNA showed an increased risk of monosomy X. Initially QF-PCR revealed a normal, XY fetus, but karyotype showed a 45 X0 / 46 XY mosaicism. In fetal echocardiogram, aortic coarctation was suspected. Subsequent ultrasound reassessments did not reveal other malformations and on third trimester ultrasound one of the testis was identified. Delivery happened at 38 weeks. The newborn had an Apgar score of 9/10/10 and has a male phenotype. Newborn needed surveillance in the Intensive Care Unit due to aortic coarctation suspicious, that was not confirmed. In the physical exam, newborn showed left cryptorchidism. Referred to Genetics, where the karyotype (from peripheral blood) revealed a mosaicism 47 XYY (as the main lineage) and 45 X0.

CONCLUSIONS: Sexual chromosomes mosaicisms as 45, X0/46, XY and 45, X0/47, XYY are important causes of ambiguous genitalia. Phenotypic expression can be broad. The main mechanisms responsible for the phenotype of mosaic patient is not well understood and despite prenatal diagnosis, postnatal karyotyped should always be performed.

References: Khan MR, Bukhari I, Junxiang T, Hui L, Muhammad N, Fan C, Zhu J, Wu M. A Novel Sex Chromosome Mosaicism 45,X/45,Y/46,XY/46,YY/47,XYY Causing Ambiguous Genitalia. Ann Clin Lab Sci. 2017 Nov;47(6):761-764. PMID: 29263053.