PRENATAL DIAGNOSIS: TURNER'S SYNDROME

Marta Luísa Rodrigues¹; Prescillia Marques¹; Rute Branco¹; Florência Sepúlveda¹; Ana Cristina Costa¹; Antónia Nazaré¹.

1 - Obstetrics and Gynecology, Prof. Doctor Fernando Fonseca Hospital, Lisbon, Portugal

PROBLEM STATEMENT

Turner syndrome (TS) is caused by the absence of the X chromosome (45XO) or by the presence of an abnormal or dysfunctional X chromosome. It is the most common genetic alteration in women, affecting about 1 in every 2000-5000 female newborns, although only 1% of fetuses with TS conclude gestation. It is the only chromosomal monosomy compatible with life.

The diagnosis is made during adolescence, but may occur in the prenatal period as a result of an amniocentesis or to suggestive fetal ultrasound alterations: increased nuchal translucency, cervical cystic septum, fetal hydrops, shortening of the femur and cardiac malformations. Note that TS is not related to advanced maternal age.

METHODS

Presentation of a clinical case and review of the literature regarding changes associated with TS in the prenatal period.

RESULTS

34 years old, caucasian pregnant, Gravida-3 Para-2 Abortus-0, with no personal or family history of relief.

Pregnancy monitored from the first trimester, in which there were no analytical or ultrasound alterations (skull-caudal length of 46.1 mm, nuchal translucency of 1.40 mm, nose bones present). The combined biochemical screening of the first trimester was negative, with the premise that if the nuchal translucency was not considered, it would have been positive for the trisomy 21. In this context, it performed the integrated screening whose result was negative.

On morphological ultrasound, at 20 weeks gestation, there was edema at the nuchal, prefrontal and lower limbs (pictures 1-3), without other changes. In view of the ultrasound alterations visualized, she performed blood tests, fetal echocardiography and amniocentesis.

- **Analytically:** there were no parameters of recent viral infection.
- **Fetal echocardiogram:** initially, revealed three small ventricular septal defects and small pericardial effusion and, subsequently, only a predominance of the right cavities.
- **Amniocentesis:** 45 XO - Turner Syndrome.

The pregnant chose to keep the pregnancy.

On ultrasound of the third trimester, at 30 weeks, the fetus maintained pre-frontal edema and in the lower limbs, although slight, and did not present other alterations.

Eutocic delivery at 38 weeks - newborn with 2735g, Apgar Index 10/10/10, no apparent malformations or intercurrents.

CONCLUSION

We describe a clinical case of TS diagnosed in the prenatal period in the context of ultrasound alterations. This case allows us to verify that although there is a wide spectrum of ultrasound manifestations associated with this syndrome, these can be more or less severe and, consequently, it can be translated either at the level of the abortion rate (only 1% concludes gestation) or at the prognosis level after birth. The prenatal diagnosis of TS is important in that it allows to anticipate and guide all the clinic and psychosocial problems associated with it.

Picture 1: Nuchal edema, 2nd T.

Picture 2: Prefrontal edema, 2nd T.

Picture 3: Lower limb edema, 2nd T.

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