CYSTIC HYGROMA: MINI REVIEW FROM CASES REPORT

Alves C[1], Fragoso I[1], Moura M[1], Correia A[1], Lopes R[1], Carnide C[1], Correia P[1], Nunes S[1], Moreira A[1], Moutinho O[1]

1- Centro Hospitalar de Trás-os-Montes e Alto Douro, Vila Real, Portugal

1- Introduction/objectives

Cystic hygroma is a congenital malformation resulting from lymph accumulation in the jugular lymphatic sacs due to obstruction of the lymphatic system, most commonly in the fetal neck. In the first trimester, its overall incidence is approximately 1 in 100 fetuses. The diagnosis is based on ultrasound examination showing a single or multilocular fluid-filled structure in the nuchal region or extending along the entire length of the fetus. Multiple internal septae or trabeculae may be identified.

It is usually associated with an increased risk for fetal trisomy, especially trisomy 21, and structural malformations (primarily cardiac and skeletal), both of which increase the risk of miscarriage, fetal demise and neonatal death. Aneuploidy appears to be more frequent with septated than simple cystic hygromas.

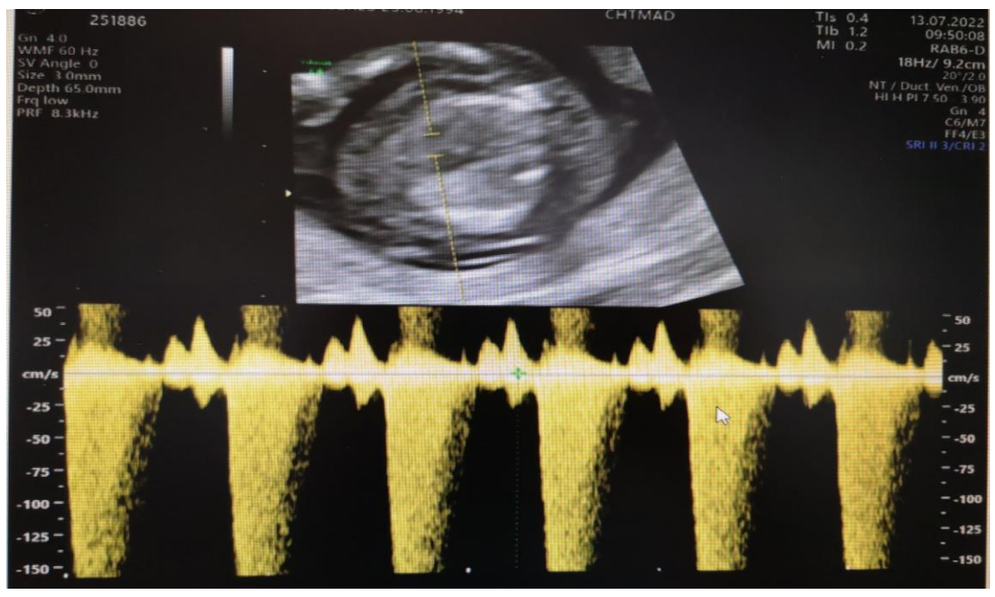
2- Methods

To present clinical cases of cystic hygromas detected at our prenatal diagnostic centre during 2022 with review of the literature.

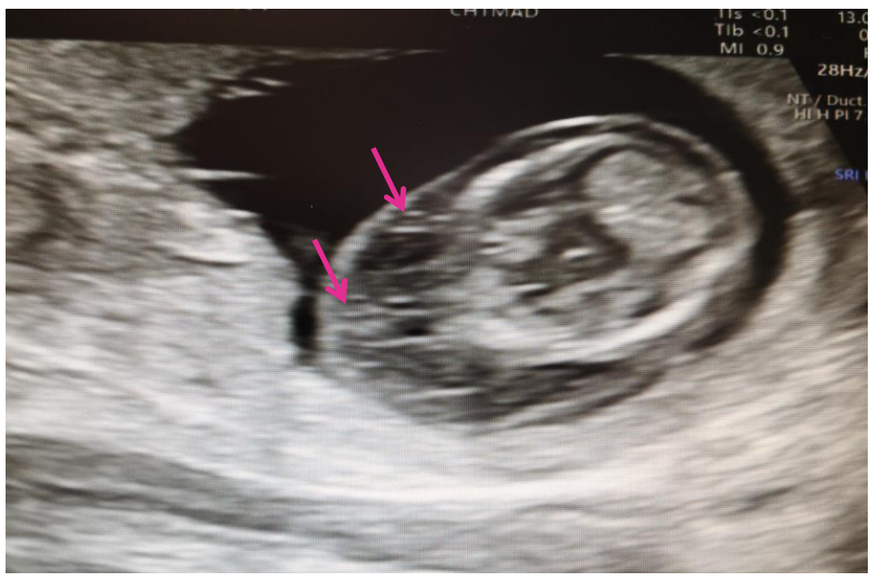
3- Results

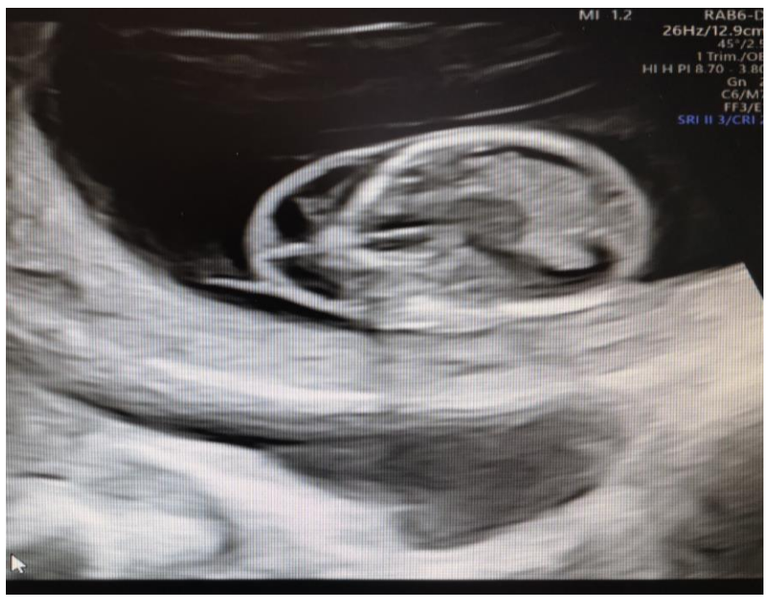
During 2022 we detected four cases of cystic hygromas, three of them associated with trisomy 21 (confirmed with karyotype obtained by invasive testing).

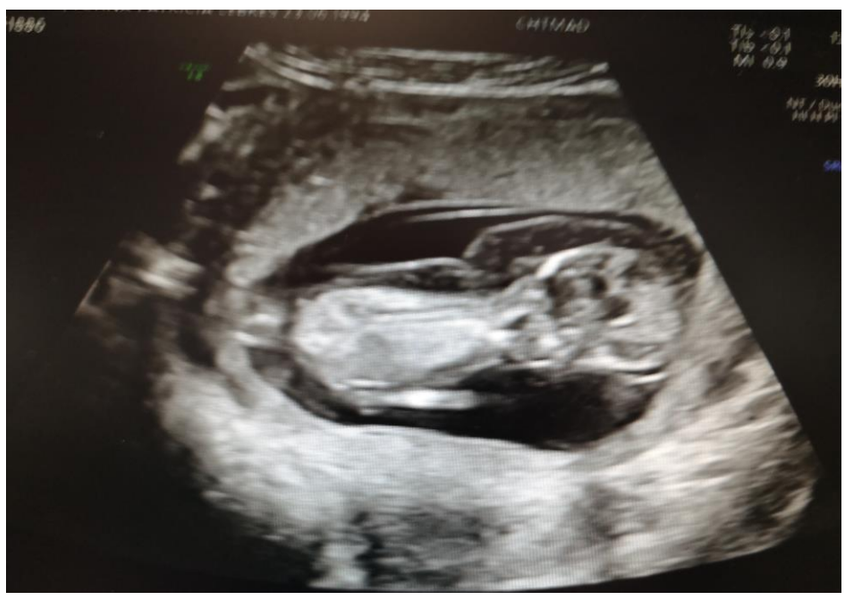
Regarding these three cases, the mean maternal age was 35 years and the mean gestational age at diagnosis was 13 weeks. The mean value of nuchal translucency was 3.70 mm. In two of the cases, there were also other sonographic findings associated with fetal aneuploidy, namely tricuspid regurgitation (Picture 1) and absent nasal bone. None of them showed other signs of structural anomalies. All these pregnancies were medically terminated by couple's option.



Picture 1. Tricuspid regurgitation.







Pictures 2-4. Ultrasound images of a first trimestre cystic hygroma (the arrows indicate the presence of multiple septations). Picture 4 reveals the extension of the lesion along the entire lenght of the fetus (which helps differentiate cystic hygroma from enlarged nuchal translucency).

We also obtained a fourth case of a simple cystic hygroma in a 31-year-old healthy woman (with a nuchal translucency of 4.20 mm and no other sonographic signs of aneuploidy). She underwent invasive testing which came back showing a 46,XY – normal chromosomal constitution. Three weeks after the diagnosis, the cystic hygroma eventually resolved and no other signs of fetal structural anomalies appeared during the rest of the pregnancy.

4- Conclusions

Most cystic hygromas are associated with an increased risk for fetal aneuploidy (particularly trisomy 21 during the first trimester and monosomy X during the second trimester) and structural malformations (mainly congenital heart defects). It should be differentiated from enlarged nuchal translucency (which is typically smaller and confined to the nuchal region), other neural tube defects, cystic teratomas (which have both cystic and solid components) or hemangiomas (classically have robust vascularity).

On the other hand, in euploid fetuses, the majority of simple cystic hygromas end up resolving during pregnancy and these neonates are usually phenotypically normal.