How a-CGH can help the diagnosis in the first trimester

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Objective

Case report.

Methods

Case report.

Results

First trimester screening was performed in a 38 years-old pregnant woman at 12 week (ultrasound and biochemical markers free-beta HGC and PAPP-A) elsewhere. The nuchal translucency was reported as 2, 4 mm, the nasal bone incert and the risk after calculation 1/200. Both prenatalist and patient came to our service for second opinion. The counsellor explained the risks of CVS or amnio and the couple decided for same-day CVS in our service. The result was 46, XY. Besides normal karyotype, they were advised not to miss an echocardiography and second trimester routine scan. At 18 week scan, mild ventriculomegaly, choroid plexus cyst, umbilical hernia, hypotelorism and abnormal right atrium were found. The couple was counselled again and an amniocentesis with a-CGH karyotype was proposed, they accepted and the result was Trisomy 18. The geneticist that performed aCGH is in contact with the laboratory of CVS to investigate if it is a trisomy restricted to fetus or a misdiagnosis in CVS.

Conclusion

The cases of uncertain diagnosis at first trimester merit a best approach including amniocentesis for a-CGH in all cases.