EP04.25 - Antenatal diagnosis of a 16q- syndrome in a fetus with bilateral cleft lip and palate, cerebellar hypoplasia and club feet - a case report

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Introduction - Case report

We present a case of a 39 year old primigravid patient, who asked for a second opinion in 23/3 weeks of pregnancy because of multiple fetal anomalies seen on specialized ultrasound scan and amniocentesis with a normal karyotype, further genetic testing was declined by the parents at this stage. We saw a SGA-fet (10th centile) presenting with multiple anomalies shown in fig. 1. The combination of anomalies suggested a syndromal cause, but prognostic evaluation at this stage remained difficult with the dilemma of termination was no option for us without a concise diagnosis and the isolated anomalies being assumingly well treatable. After intensive counselling, the parents eventually agreed to further investigation.

16q- syndrome

Using Array-CGH an extremely rare (prevalence < 1:1000000) de novo-deletion at chromosome 16q22 was detected (46XX, del(16)(q22q23), 6,06 mb large), going along with a 16q- syndrome. There are only a few case reports in the literature. Typical symptoms are SGA, microcephaly, moderate to severe neurological, motorial and intellectual handicaps, spastic dysplegia, dysmorphic face, large fontanelles and wide skull sutures. Furthermore, 16q- is associated with cleft lip and palate as well as congenital cardiac disease. Genetic workup delivered the diagnosis to asses the developmental prognosis.

Conclusion

The parents fed back content with the chosen management of the situation, refraining from active fetocide, labour induction, care in the delivery room and psychological support. For some parents this could be a good alternative to fetocide to handle this crucial situation.