Neural tube defects as markers for trisomy 18

Sutmöller ATC, Dürks PT, Barth MB, Poli JHZ, Vogt GN, Feltes IC, Sampaio VO, Sa DC, Betat RS, Rosa RFM
Hospital Materno Infantil Presidente Vargas, Porto Alegre, Brazil

Objective

To report the aditional prenatal findings of a trisomy 18 case with myelomeningocele.

Methods

Case report.

Results

We report the case of a G6P5 36 year-old woman. She was referred to our centre due to polyhydramnios and myelomeningocele at 25 weeks of pregnancy. She had no first trimester screening. Morphological ultrasound at 28 weeks' gestation showed polyhydramnios (AFI = 29 mm); lemon sign; dilatation of the lateral and third ventricles with apparent cranial displacement suggestive of agenesis of the corpus callosum; the cerebellum was not identified and there was a lumbosacral spina bifida covered by a thin membrane. At this time, an amniocentesis for fetal karyotyping was performed. MRI confirmed the agenesis of the corpus callosus and showed, in addition to the findings observed in the ultrasound, altered cerebral gyri and disclosed signs of herniation of the cerebellar tonsils into the cervical canal, compatible with Arnold-Chiari type II. Fetal echocardiography performed soon after showed a ventricular septal defect of moderate to large size. Karyotype confirmed a full trisomy of chromosome 18 or Edwards syndrome.

Conclusion

Chromosomal abnormalities, mainly trisomy 18, are observed in 7-16% of fetuses with neural tube defects. This association becomes stronger when there are any other associated abnormalities. Neural tube defects have been observed in about 6-12% of cases of trisomy 18, mainly myelomeningocele as in our case. The association of trisomy 18 with other neural tube defects, as anencephaly or encephalocele, exists but is considered rare.