‘keyhole’ communication between the fourth ventricle and the cisterna magna at 20 weeks [4], the absence of the cerebellar vermis at 22 weeks [5] associated with a smooth cortex and ventriculomegaly. Due to the family history, we suspected JS based on MTS. This is the second case reported and it provides the earliest depiction of the MTS on sonography.

When a mutation is found by molecular diagnosis, preimplantation genetic diagnosis or prenatal invasive testing are offered to the family. However, in many cases, a specific mutation is not identified, and prenatal diagnosis of this severe disease may rely exclusively on imaging. Adding early prenatal sonography to the diagnostic tool kit for JS enhances management and counseling with regard to decisions for cases at risk.

References


Erratum

In the article by Gratacós E and Nicolaides K, entitled ‘Clinical perspective of cell-free DNA testing for fetal aneuploidies’ [Fetal Diagn Ther 2014;35:151–155, DOI: 10.1159/000362940], in the section Implications of a Low-Risk Result, the 1,000 in the first sentence should be replaced with 100 (‘The negative likelihood when cfDNA testing gives a low-risk result is about 100, 31 and 13 for trisomies 21, 18 and 13, respectively [5].’) and the 500,000 in the last sentence should be replaced with 50,000 (‘For example, if prior screening by the combined test had shown that the risk for trisomy 21 was 1 in 500 and cfDNA testing gives a low-risk result, the chance that the fetus is affected is 1 in 50,000; …’) and not as erroneously stated in a previous Erratum with 5,000 [Fetal Diagn Ther 2014;36:68].