First trimester imaging in Dandy-Walker syndrome with vermian agenesis

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Introduction

Dandy walker is usually a 2nd trimester diagnosis. We report features that could give a hint at the first trimester scan.

Case

A 24 years old G1P0, with no consanguinity had a spontaneous pregnancy. First trimester ultrasound showed a normal NT at 1.29, normal nasal bone and adequate biometry. IT seemed blurry and larger than usual measuring 3mm. Second trimester biochemical screening was normal. Morphology scan at 21 weeks showed an abnormal posterior fossa with an enlarged cistern magna and a decreased vermian structure. Upper displacement of the cerebellar lobes was also noted. No other CNS or morphological abnormalities were seen.

The case was thoroughly discussed with the couple and they chose further investigation by amniocentesis. The latter revealed a normal caryotype and the absence of the most frequent microdeletions by microarrays. MRI was also performed and confirmed the diagnosis of vermian agenesis; the pediatrician highlighted the severity of the long term outcome and the couple elected then termination of pregnancy. Post mortem evaluation did not reveal additional abnormalities features. Upon reviewing the first trimester images there was a possible link between the sagittal images and the latter diagnosis. What would become the increased cistern magna would probably be the atypical intracerebral translucency noted then. Eventhough it is difficult to identify the cerebellum at 12 weeks, discrete anomalies of the posterior fossa in the first trimester could be a sign for vermian agenesis.

Conclusion

Dandy-Walker can be suspected at the first trimester scan, leading to an earlier diagnosis.