INITIAL ULTRASOUND EXAMINATIONS

A 24 year old P1+0 attended for her routine anatomy examination at 20 weeks gestation. Severe brain abnormalities were identified. The posterior ventricle was enlarged and the cerebellum appeared abnormal. The patient was referred to the fetal medicine service.

Severe ventriculomegaly measuring 16.3mm

This examination showed severe bilateral ventriculomegaly (>15mm) and an absent cerebellar vermis.

Absent cerebellar vermis (1) and dilated fourth ventricle (2). This gave rise to an initial suspicion of Dandy-Walker Malformation.

FURTHER EXAMINATIONS AND DIAGNOSIS

An ultrasound exam at 27 weeks showed further deterioration with bilateral ventriculomegaly of 23mm

There was now dilatation of the anterior ventriciles, the third-ventricle and the cisterna magna.

NEUROSURGICAL REVIEW

The couple met with the fetal medicine consultant and a neurosurgeon at 29 weeks for further discussion and ultrasonographic examination.

Increasing ventriculomegaly of 26mm.

In the week interval, the ventriculomegaly had increased and the anterior ventriciles were no longer defined. There now appeared to be complete absence of the cerebellar vermis and further dilatation of the cisterna magna.

Agenesis of the cerebellar vermis and further dilatation of the cisterna magna.

MAGNETIC RESONANCE IMAGING

An MR examination was performed. It confirmed these findings and also discovered a z-shaped brainstem and cobblestone lissencephaly, in which the surface of the brain lacks normal folds and grooves and develops a bumpy, irregular appearance. The eye orbits were also found to be asymmetric.

Sagittal view: “Z-shaped” brainstem (1), small vermis (2) and occipital encephalocoele. Axial view: Asymmetric orbital globes.

The MR findings confirmed the previous ultrasound findings and now gave rise to a diagnosis of Walker-Warburg Syndrome.

WALKER-WARBURG SYNDROME

Walker-Warburg Syndrome (WWS) is a lethal, autosomal recessive genetic disorder effecting 1,300,000 live births. It is defined by hydrocephalus, smooth gyri, nearly absent sulci, retinal dysplasia and often encephalocoele. It is the most severe in a group of congenital muscular dystrophy conditions. Children that survive the fetal and neonatal period generally need full support and have a limited life expectancy.

Whilst this couple already had a healthy unaffected child, this syndrome has a high recurrence rate of 1:4 and therefore the couple need to be thoroughly counselled on the potential recurrence in future pregnancies. Rather than risk premature labour by performing an amniocentesis for prenatal karyotyping, the couple declined, deciding instead to await postnatal testing or a post-mortem.

Fetal/ Neonatal Outcome

A male infant was delivered. He initially was in poor condition and admitted to the neonatal intensive care unit for monitoring for 2 weeks. His condition improved enough to allow him home with his family for comfort/palliative measures.

Unfortunately the infant died at three months of age following seizure activity. At parental request, a post mortem was performed. This confirmed the prenatal ultrasound and MR findings and a diagnosis of Walker-Warburg was made.

CONCLUSION

Whilst the outcome of this situation could not be changed by the ultrasound findings, detection and proper diagnosis of abnormalities prenatally can at least somewhat prepare the parents for an adverse outcome. This case highlights the need for a multidisciplinary team approach in the unfortunate event of a poor prognosis.

REFERENCES


