Prenatal Diagnosis of Walker-Warburg Syndrome

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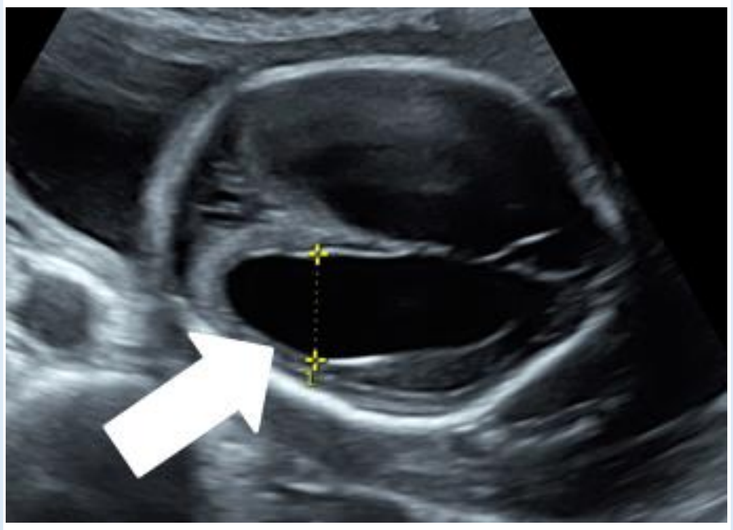
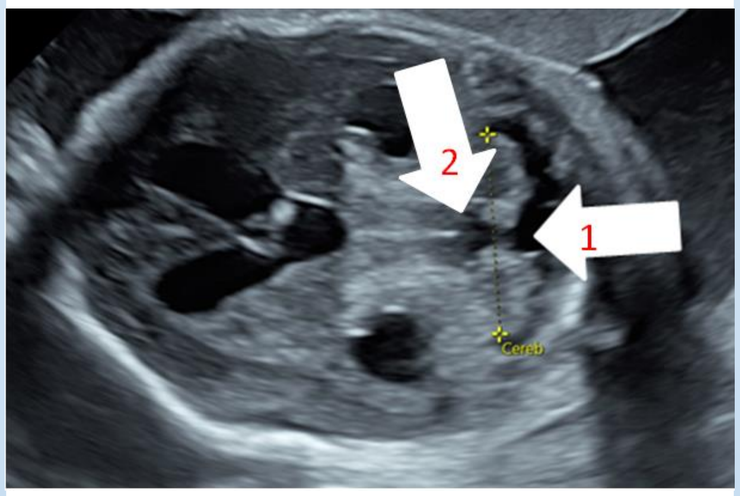
INITIAL ULTRASOUND EXAMINATIONS

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

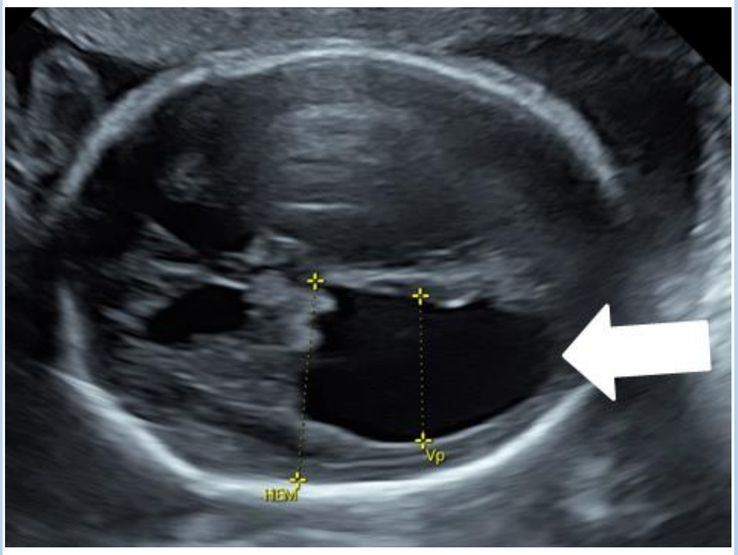
This examination showed **severe bilateral ventriculomegaly** (>15mm) and an **absent cerebellar vermis.**



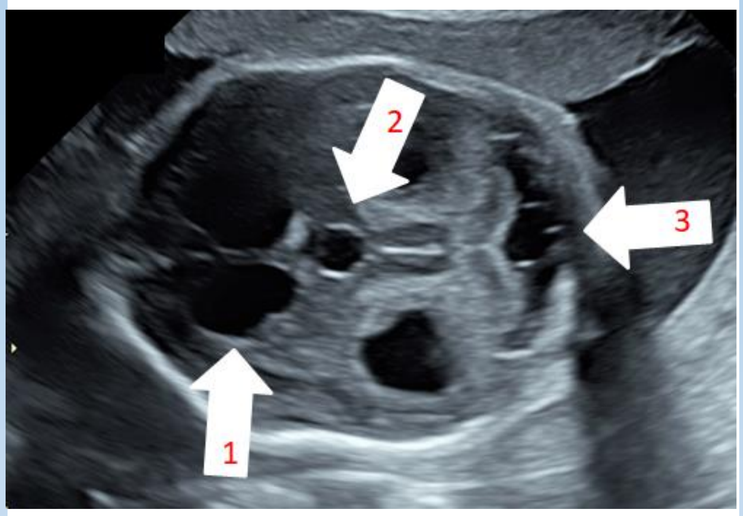
Absent cerebellar vermis (1) and dilated fourth ventricle (2).
This gave rise to an initial suspicion of **Dandy-Walker Malformation**.

FURTHER EXAMINATIONS AND DIAGNOSIS

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



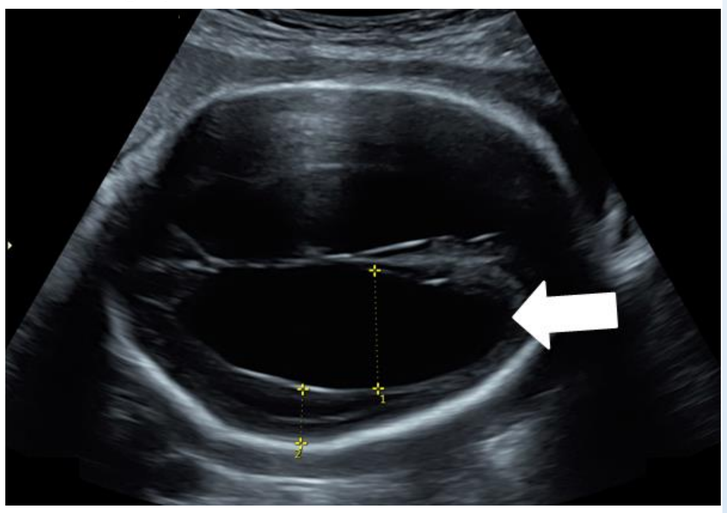
There was now dilatation of the anterior ventricles, the third-ventricle and the cisterna magna .



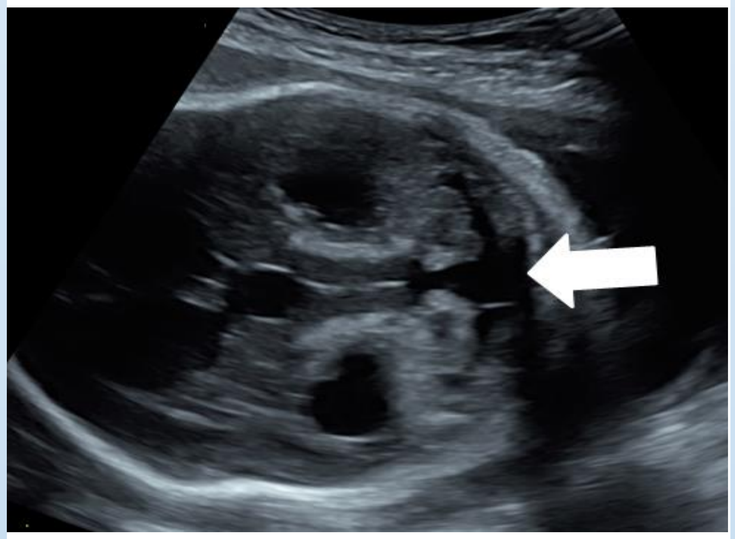
Dilatation of the anterior ventricles (1), the third ventricle (2) and the cisterna magna (3).

NEUROSURGICAL REVIEW

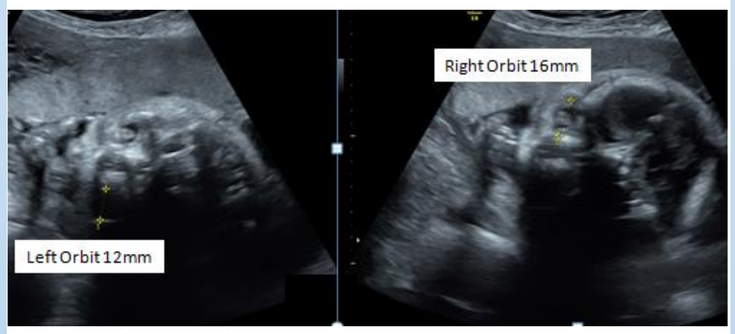
The couple met with the fetal medicine consultant and a neurosurgeon at 28 weeks for further discussion and ultrasonographic examination.



Increasing ventriculomegaly of 26mm.
In the week interval, the ventriculomegaly had increased and the anterior ventricles 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.



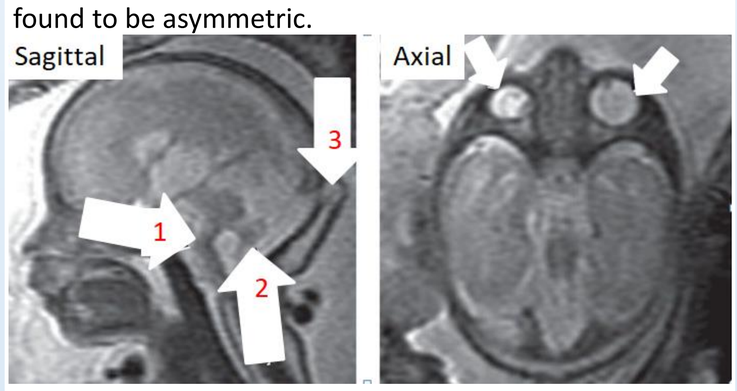
Asymmetric eye orbits.
The asymmetric eye orbits were more apparant and measurements suggested micro-ophthalmia and hypotelorism. This further indicated muscle-eye-brain disease on the spectrum on Walker-Warburg Syndrome.
The rest of the fetal anatomy was re-examined and still found to be normal.
The fetal medicine specialist and neurosurgeon discussed the findings with the couple. Whilst there would be an initial, thorough neonatal examination, based on the most recent ultrasound examination findings it would be unlikely that surgical interventions such as shunting would be beneficial and instead palliative/comfort measures may be of the best interest.

WALKER-WARBURG SYNDROME

Walker-Walburg Syndrome (WWS) is a **lethal**, autosomal recessive **genetic disorder** effecting **1:100,000** live births.
It is defined by **hydrocephalus, smooth gyri, nearly absent sulci, retinal dysplasia** and often **encephalocele**. It is the most severe in a group of congenital muscular dystrophy conditions.
Children that survive the fetal and neo-natal 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 thouroughly 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.

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.

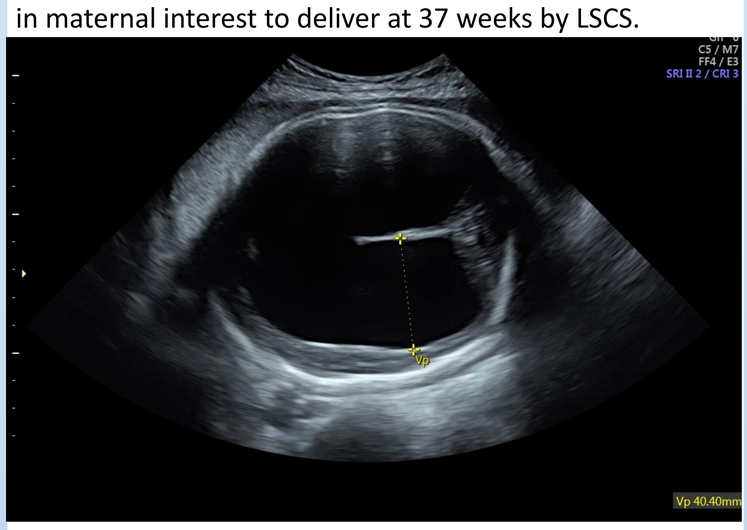


MRI. Sagittal view- “Z-shaped” brainstem (1), small vermis (2) and occipital encephalocele. Axial view- Asymetric orbital globes.

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

Fetal/ Neonatal Outcome

A further examination at 36 weeks confirmed worsening ventriculomegaly of 40mm and a head circumference of 41 weeks. Due to a previous LSCS delivery, it was decided in maternal interest to deliver at 37 weeks by LSCS.



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 MRI 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.

EQUIPMENT USED

The G.E. Voluson E8 was used for the examinations.
A 4-8MHz curvilinear prob was used for the anatomy scan and a 1-5MHz probe for the later gestation examinations.

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