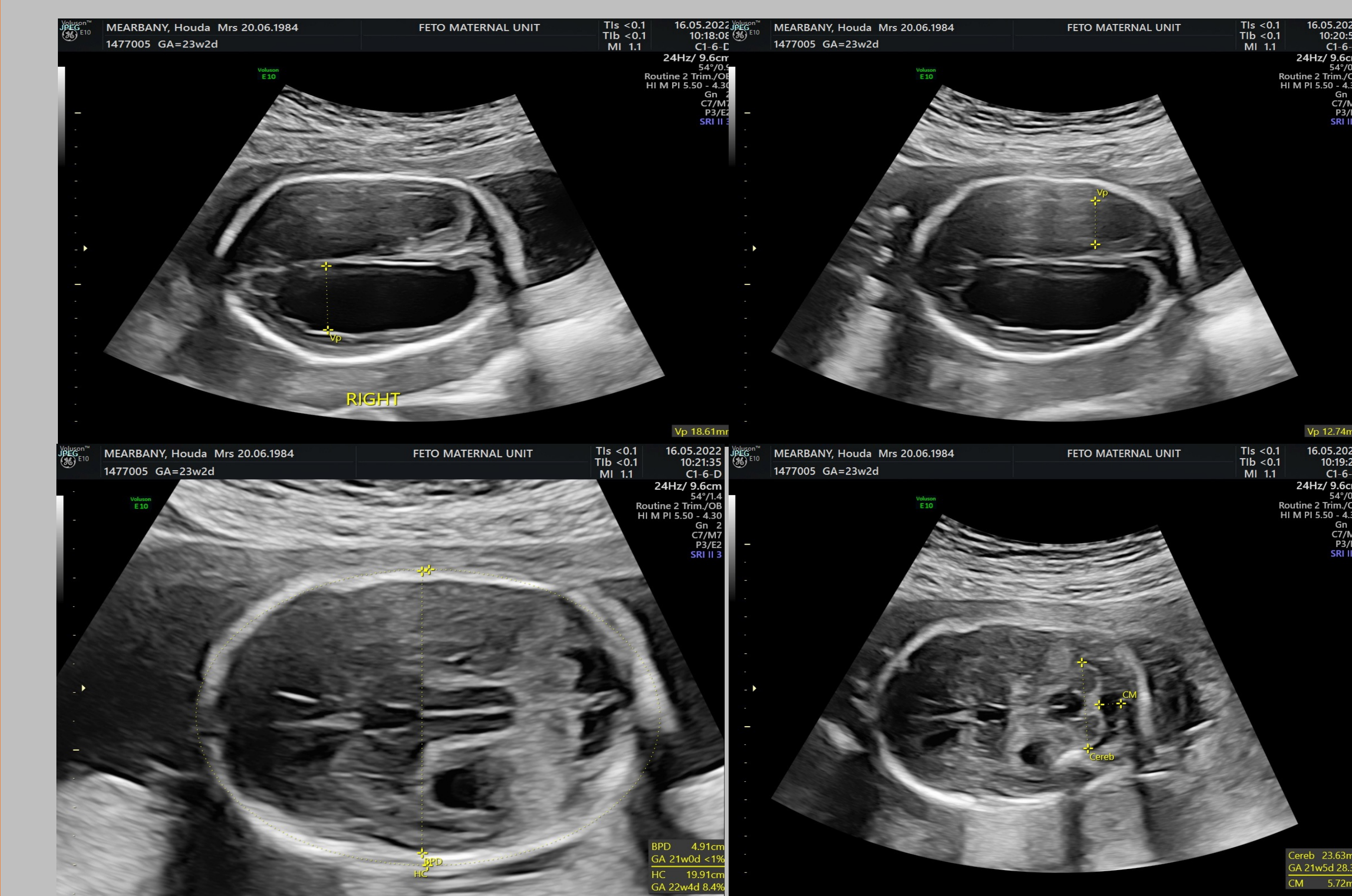


WWS: Walker Warburg Syndrome

Objective:

Walker-Warburg Syndrome (WWS) has been also referred as “Muscular dystrophy-dystroglycanopathy type A” (MDDGA) or “Cerebro-ocular dysplasia-muscular dystrophy” (COD-MD) or “hydrocephalus, agyria and retinal dysplasia” (HARD) syndromes, is a rare form of congenital muscular dystrophy that is also characterised by ocular manifestations, cerebral malformations, developmental delay and sometimes other organ manifestations. WWS can be suspected via routine ultrasound and/or fetal MRI during pregnancy and confirmed at or shortly after birth. The prognosis is poor and most of the cases don’t live past infancy.



Case Report:

We present a case of a 38 years old woman, G9P7-1, on a consanguineous relationship. Her first 2 pregnancies were uncomplicated and she delivered 2 term healthy babies. The third pregnancy was complicated by the diagnosis of progressive severe ventriculomegaly and the baby died 40 days after birth – the MRI was consistent with WWS. She had another pregnancy that was terminated at 13 weeks as the

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fetus was diagnosed early (on first trimester) with ventriculomegaly –a postnatal microarray was performed and no abnormalities of clinical significance were demonstrated. She had 3 other uncomplicated pregnancies.

In current pregnancy, the fetus was diagnosed with severe ventriculomegaly at 21 weeks (morphology USS). She was seen in The Feto maternal unit and the findings were confirmed. Low risk NIPT has been reported as a negative infections screening. Invasive genetic testing was declined.

A MRI was requested and performed at 24 weeks showed “Ventricular atrial measurements are left 15mm and right 20mm. Third ventricle 5mm. There is abnormal fissuration, sulcation and opercularisation. Only very shallow sylvian fissure are seen which could be secondary to hydrocephalous. Pontine bulge is absent and the brainstem appears kinked. The vermis is hypoplastic and deformed. Cerebellar hemisphere appears hypoplastic. Cisterna magna appears prominent size 7mm”

Paediatric Neurology team has been consulted and discussed that the findings were highly suspicious of recurrence of the WWS. After discussion the findings and prognosis with both parents, decision was made for comfort care pathway for the baby after delivery.

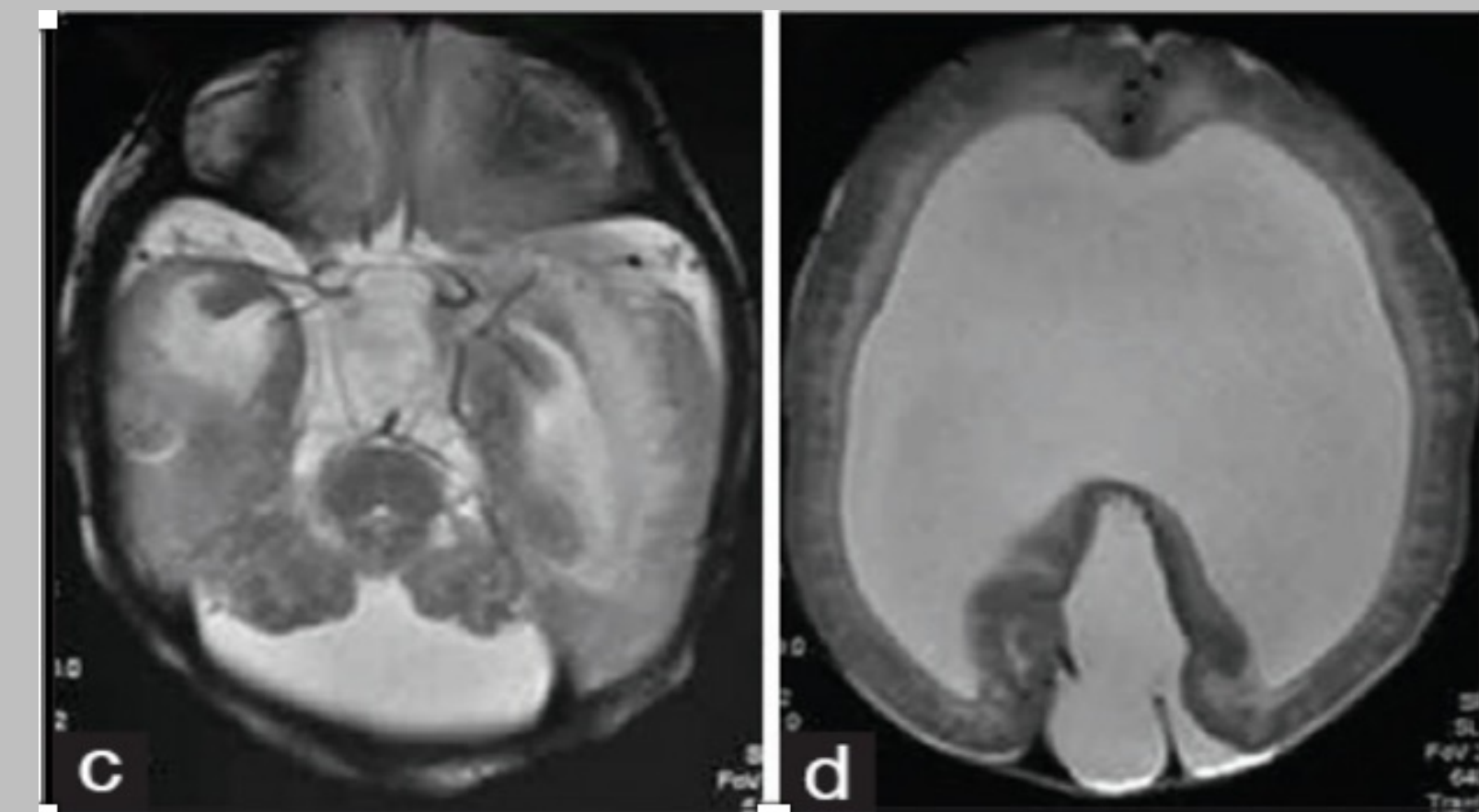
Induction of labour was started at 38+4 as there was progressive ventriculomegaly with increasing head circumference and the patient delivered vaginally a female, with birthweight 2760g and Apgar scores 6 and 7 at 1 and 5 minutes.

The mother was discharged home on D2 postpartum with follow up plan on D3.

Baby was discharged home on D3 with comfort care plan.

Discussion

WWS is inherited as an autosomal recessive pattern and 14 implicated genes have been described (POMT...) These mutations cause Defective glucosylation of alpha dystrogly can complex, which destabilises muscle fibres and affects migration of neurons in early development. However, genetic testing does not always identify a genetic cause. If a mutation is identified, it may be useful for counselling and management of future pregnancies. In our case, the genetic testing (microarray) was only performed on the pregnancy that was terminated as the parents declined any test.



The management of this condition may be challenging as there is no cure and the prognosis is poor, but the discussion should involve pregnancy management options and postnatal care planning, which may include treatments for seizures, hydrocephaly management and other supportive treatments.

Conclusion:

WWS has very poor prognosis and most cases do not live past infancy. However; multidisciplinary team involvement is essential to achieve the best possible outcome for the child and parents. The counselling about future pregnancies should include discussion of the risk of recurrence (~25%) and a plan for management (genetic testing if a gene was identified or early anatomy USS followed by serial USS)

References

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