

Case Report

Antenatal

A 22 year old, primiparous female with known type 1 myotonic dystrophy presented to our unit at 29/40 pregnant. The patient received joint care with a tertiary obstetric unit, two multidisciplinary team (MDT) meetings were conducted including paediatric, obstetric and anaesthetic input. Video MDTs were utilized including fetal medical and genetic specialists from the tertiary unit who also confirmed the baby had congenital myotonic dystrophy by amniocentesis.

At 29/40 USS revealed polyhydramnios, EFW was within normal limits. A subsequent TORCH screen was negative. USS performed at 32 & 34/40 confirmed the persistence of polyhydramnios and identified unilateral talipes.

At 32/40 echocardiogram and spirometry were performed and concluded to be within normal limits. Respiratory clinic input was offered. The cardiac and respiratory MDT highlighted concerns about the risk of sleep apnoea. Postnatal CPAP was recommended.

At 30/40 the patient was reviewed for tightenings where a fetal fibronectin was positive. At 32+5/40 the patient was treated for a UTI and at 33/40 had a preterm, pre-labour, spontaneous rupture of membranes confirmed by amniure. The patient experienced reduced fetal movements from 32+5/40 and had 6 subsequent episodes. CTGs and PROM monitoring were normal.

Induction of labour was commenced at 36+4/40. An epidural was sited at 6cm. Syntocinon was commenced for delayed progress. Electrolytes were taken 6 hourly

A rotational kiwi in the room was successfully performed for CTG concerns and maternal exhaustion. The patient had manual removal of placenta in theatre. Total estimated blood loss was 2 litres.

MYOTONIC DYSTROPHY

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What is myotonic dystrophy?

Myotonic dystrophy is a rare (approx. 1 in 8000) multisystem disorder which can result in varied problems including muscle weakness and myotonia and cardiac conduction abnormalities.

Myotonic dystrophy is a genetic condition with autosomal dominant inheritance. There are two types of myotonic dystrophy. DM1 arises from an expansion of a cytosine-thymine-guanine (CTG) trinucleotide repeat in the 3'-untranslated region of the DM1 protein kinase (*DMPK*) gene. Genetic testing for the CTG repeat in the *DMPK* gene is considered the 'gold standard' test.

Case Report

Neonatal

The baby had low APGARs requiring early intubation and ventilatory support; transferred later to a tertiary centre (with failed extubation twice). Subsequently the baby developed a pneumothorax and multi resistant klebsiella colonisations requiring multiple antibiotics. The baby remains ventilator dependent ready for extubation to long term ventilation with extensive neurodisability input.

References

Sax TW, Rosenbaum RB. Neuromuscular disorders in pregnancy. Muscle Nerve. 2006 Nov;34(5):559-71. doi: 10.1002/mus.20661. PMID: 16969835.
•Harper PS. *Myotonic Dystrophy* 3rd edn. London, UK: W.B. Saunders; 2001 [[Google Scholar](#)]

How does myotonic dystrophy impact pregnancy?

Symptomatically, myotonic dystrophy generally has a benign course throughout pregnancy. A small proportion of women may experience an increase in weakness during pregnancy but this usually resolves postnatally.

Women with myotonic dystrophy have increased risks of fetal loss, prematurity and polyhydramnios. These patients may also experience recurrent reduced fetal movements. Generally these increased risks are attributed to the risk of inherited disease in the fetus. These patients will frequently also experience anxiety and depression antenatally and postnatally.

Pregnancies in patients with congenital myotonic dystrophy require additional antenatal multidisciplinary care and monitoring

Common risks include polyhydramnios, reduced fetal movements and preterm delivery

Important Aspects of Antenatal Care

- Pre-pregnancy counselling is advised
- Additional scans/fetal monitoring
- Investigation for alternate causes of polyhydramnios may be needed
- Recurrent reduced fetal movements need additional surveillance
- Early fetal medicine input is crucial for early counselling/planning with regards to exploring the risk of inheritance
- Patients should be aware of risks/presentation of preterm labour
- Early anaesthetic input is important- avoidance of general anaesthesia in type 1 MD is advisable where possible
- Multidisciplinary team management is key, including cardiology, physiotherapy, fetal medicine, medical physicians, neurologists and neonatologists