

A Case series on Core body syndrome in pregnancy

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INTRODUCTION

Congenital myopathies are a group of minimally progressive or non-progressive neuromuscular conditions which is present from birth. A classical type of congenital myopathy is called central core disease. This condition is often confused with muscular dystrophy. Central core disease can be associated with comorbidities which affect pregnancy and its management.

CASE DETAILS

This report consists of siblings with central core disease, their diagnosis and management in pregnancy.

Case 1: A 31-year-old primigravida was booked with a diagnosis of congenital myopathy. She had a strong family history of congenital myopathy and many family members were affected, but with variable penetrance. She was wheelchair bound and had an associated vitamin B12 deficiency. She had previous surgeries on her hip and knee at 9 and 11 years respectively. She declined fetal testing and presented with recurrent reduced fetal movements, which is one of the signs of fetal congenital myopathy. She went into preterm labour at 33 weeks and had a caesarean section in view of breech presentation. She delivered a baby who was unfortunately, subsequently diagnosed with the same condition and presented with hypotonia, slow respiration and developmental delay later.

Table 1
PRECONCEPTION
• Cardiac evaluation • Evaluation of lung function • Evaluation of osteopenia, skeletal deformities and other nutritional deficiencies • Preconceptionally folic acid and supplementation of deficient vitamins and nutrients • Genetic counselling explaining the mode of inheritance and estimating the risk to the offspring. This discussion should cover talks about preimplantation genetic diagnosis, gamete donation, adoption and post conceptional screening techniques.
Table 2
PREGNANCY
• Offer screening with chorionic villous sampling/ amniotic fluid testing • As the mobility may be restricted, a venous thromboembolism risk assessment must take place in the first trimester and a plan for thromboprophylaxis must be made. • The patient should be followed up in a consultant led unit with specialist midwife, occupational health therapist and social worker.
• Second trimester anomaly scan • Be aware of the possibility of recurrent reduced fetal movements • Anaesthetic evaluation, assess risk for malignant hyperthermia, kyphoscoliosis and to plan for pain relief and anaesthetic medications that can be used during labour and caesarean if necessary. • Aim for vaginal delivery, however positioning during labour must be discussed in a multidisciplinary meeting based on individual situation, considering risk factors like osteopenia and previous surgeries.
Table3
POSTPARTUM
• Active management of third stage of labour • Reassessment of venous thromboembolism scoring and thromboprophylaxis. • Neonatal assessment and plan for follow up • If the baby is affected, the mother will need additional help with feeding, mental health support, early acquaintance with suitable nursery and toddler groups. • Home visits by family doctor, paediatrician, specialist nurse and social worker may be arranged.

Case 2: A 32-year-old lady with previous two miscarriages presented in antenatal period with congenital myopathy. She had muscle weakness and could not walk long distances or climb stairs. She also had vitamin B12 deficiency along with severe kyphoscoliosis. She also declined fetal screening for congenital myopathy in early pregnancy and presented with recurrent reduced fetal movements. She had a didelphic uterus and had an elective caesarean at 38weeks in view of breech presentation. At birth the baby had meconium aspiration and needed CPAP for 2 days to support respiration. The baby also had feeding difficulties at birth.

DISCUSSION

Mothers with central core disease can have muscle weakness at varied levels. It is usually associated with kyphoscoliosis, osteopenia and vitamin deficiency. The condition can be associated with life threatening malignant hyperthermia. Occasionally, there could be associated cardiac condition and altered lung function. This is an autosomal dominant condition and neonates affected by this condition can have hypotonia, muscle weakness, weak cry and are likely to need respiratory support at birth. They usually have feeding difficulties and developmental delay. A management plan during pregnancy has been summarised in Tables 1,2 and 3.

CONCLUSION

Although there is no direct effect of this condition on pregnancy except possible reduced fetal movements, a multi-disciplinary team which involves a specialist nurse, Neurologist, Cardiologist, Physician, orthopaedician, Obstetrician, Anaesthetist, Paediatrician and social care is needed for management of such cases.