

## **Congenital Myotonic Dystrophy: A Rare Cause of Polyhydramnios**

Congenital myotonic dystrophy is an autosomal dominant condition with estimated incidence of 13/100000(1). We describe here a case of congenital myotonic dystrophy suspected antenatally due to maternal polyhydramnios on antenatal ultrasound scan.

A twenty-seven-year-old primigravida had an anomaly scan at 20 weeks which was normal, but a repeat scan at 30 weeks revealed polyhydramnios. The fetus showed talipes, dysfunctional fetal swallowing and decreased movements. This along with maternal features of mild myopathic facies, weakness and slow release of hand grip, suggested a possibility of congenital myotonic dystrophy in the baby. Molecular genetic analysis of the mother confirmed CTG expansion.

The baby, delivered at 35 week gestation, was floppy, cyanosed and made no respiratory effort at birth needing ventilation. The baby had facial dysmorphism (inverted V shaped upper lip, floppy lower lip, hollowing of jaw and temples, ptosis), hypotonia and talipes. Reflexes were difficult to elicit. The cranial ultrasonography showed dilated ventricles and a generous amount of fluid around brain without any signs of raised intracranial pressure. Initial feeding difficulties resolved by the time of discharge at around 3 weeks. At follow-up he is failing to thrive and having recurrent chest infections.

Polyhydramnios is a relatively common obstetric complication. Major causes include maternal diabetes mellitus, chromosomal disorders, iso-immunogenic disease, congenital abnormalities, multiple gestations, with a significant percentage being idiopathic.

Esplin, *et al.*(2) described myotonic

dystrophy as a significant cause of idiopathic polyhydramnios. They suggested that all women having polyhydramnios with either a family history of myotonic dystrophy or ultrasonographic evidence of fetal hypotonia, including positional abnormalities of extremities should be offered DNA testing. It appears that prenatal ventriculomegaly may also be an important ultrasonographic finding suggesting congenital myotonic dystrophy. If idiopathic hydramnios is identified in a mother and dysfunctional fetal swallowing is seen, three fetal neuromuscular disorders should be ruled out –X-linked myotubular myopathy, congenital myotonic dystrophy and congenital nemaline myopathy(3).

The myotonic dystrophy gene is located on the long arm of chromosome 19, band 13q. This gene contains an unstable trineucleotide - CTG (cytosine-thymine-guanine) and usually passed on from mother. Myotonic dystrophy can have variable severity according to the age of onset and CTG repeat number. Our mother had lower CTG repeats than the baby who had 3100 repeats. This shows classical anticipation and correlates well with the severity of disease in the baby and the late detection of the mother. If the mother is affected the risk of recurrence is 1:8 and if the first child is affected the risk rises to 1:3 in the second pregnancy.

Connolly, *et al.*(4) showed that good prognosis depends on gestational age(>35 weeks) and length of mechanical ventilation (<21days). Data suggests a 25% chance of mortality within 18 months of life and 50% chance of survival into the mid thirties.

The diagnosis of congenital form is difficult in those cases where the mother has not been diagnosed. Delest, *et al.*(5) showed that family and personal history may give hints. In our case the finding of poly-hydramnios, club feet, and decreased fetal movement suggested the

possibility of fetal neuromuscular disease which prompted clinical examination followed by genetic testing of the mother which confirmed the myotonic dystrophy gene expansion. Prenatal diagnosis in fetus was not attempted because of the late presentational and the genetic confirmation in mother.

**Sandeep Jayawant,  
Rajiv Sinha,**

*John Radcliffe Hospital, Oxford.*

*Correspondence to:*

**Rajiv Sinha,**

*Flat 95, Ivy Lane, Headington,  
Oxford OX3 9 DY.*

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## Laparoscopy in Suspected Meckel's Diverticulum: Negative Nuclear Scan Notwithstanding

A one-year-old boy was admitted with painless lower gastrointestinal bleeding since one week. Investigations done at another hospital showed a hemoglobin of 6.2 g/dL for which blood transfusion had been given. The child had also undergone upper and lower gastrointestinal endoscopy and a technetium<sup>99m</sup> pertechnetate scan, which were all reported as normal. At admission to this hospital, physical examination revealed no abnormality. The clinical possibility of Meckel's diverticulum was discussed with the parents and a diagnostic laparoscopy was offered. Laparoscopy using a 5 mm umbilical port revealed a Meckel's diverticulum. Using two 3 mm secondary ports the diverticulum

was delivered out of the abdomen through the umbilical incision. A wedge resection of the diverticulum with intestinal anastomosis was done.

Meckel's diverticulum is the most common congenital anomaly of the gastrointestinal tract involving the small bowel(1). In infants and younger children, painless lower gastrointestinal bleed is the commonest manifestation. The bleeding may be brisk and blood transfusion is often required. A pre-operative diagnosis of a Meckel's diverticulum is often difficult to make.

Routine evaluation of these children would include a hemogram, endoscopic evaluation of the gastrointestinal tract and a radioisotope scan. Abdominal ultrasonography is commonly performed but rarely helps in diagnosis. Barium studies have little utility. The most useful method to detect a