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SHORT COMMUNICATION

# DUCHENNE MUSCULAR DYSTROPHY ASSOCIATED WITH FETAL PLEURAL EFFUSION AND POLYHYDRAMNIOS

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## SUMMARY

A case of fetal pleural effusion in a fetus affected with Duchenne muscular dystrophy (DMD) is reported. This case is discussed in the context of the previous observation of frequent stillbirths among male fetuses in DMD families.

KEY WORDS Duchenne muscular dystrophy Fetal pleural effusion

## INTRODUCTION

Fetal pleural effusions are rare, occurring in 1 in 15 000 pregnancies (reviewed in Weber and Philipson, 1992). They are associated with a high rate of fetal and neonatal mortality. An underlying abnormality is frequently not found, and the known differential diagnosis includes a variety of non-specific causes. Here we report on a fetus presenting at 28 weeks with unilateral fetal hydrothorax who has Duchenne muscular dystrophy (DMD). We suggest that fetal pleural effusions could explain the high rate of stillbirths and neonatal mortality in DMD.

## CASE REPORT

A 28-year-old gravida 6 para 2034 (two sets of twins) black female was referred for ultrasound at 28 weeks for confirmation of dates. A family history of DMD was ascertained after fetal abnormalities were noted on ultrasound. The patient's first pregnancy produced twin boys who we examined at age 10 years and found to have proximal muscle weakness, positive Gower sign, pseudohypertrophy of the calves, and elevation of serum creatine phosphokinase to >4000 IU/l consistent with a diagnosis of DMD. The patient's brother had DMD diagnosed at the age of 12. He died from complications of DMD at the age of 20. At 28 weeks' gestation an ultrasound for confirmation of dates revealed a unilateral fetal pleural effusion measuring 5·4 × 1·9 cm and increased amniotic fluid (Figure 1). The rest of the ultrasound examination was normal. Sonography repeated 2 weeks later showed appropriate interval growth, a decrease in the size of the effusion to 5·0 × 1·2 cm, and marked polyhydramnios.

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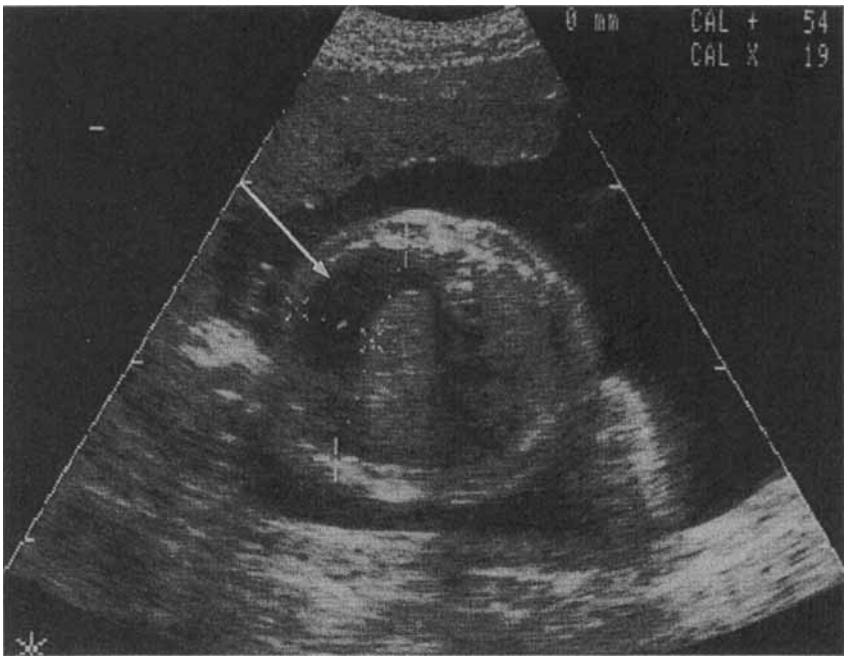


Figure 1. Sonogram of the thorax at 28 weeks of gestation. Note the large, right pleural effusion (arrow)

The patient refused prenatal diagnosis to rule out aneuploidy as the cause of the effusion. The fetal echocardiogram was normal. Fetal ultrasounds were repeated biweekly. The effusion and polyhydramnios resolved at 36 weeks and the patient had a spontaneous vaginal delivery at term. Physical examination was unremarkable. Muscles had normal consistency. Serum creatine phosphokinase was 80 000 IU/l on day 1 of life. There was no myoglobinuria. Subsequent serum creatine phosphokinase was >3000 IU/l at 6 weeks of age. Chest X-ray showed resolution of the effusion, but fluid could be seen in the interlobular fissure of the right lung. There was no evidence of lung hypoplasia. The child had an otherwise unremarkable neonatal course in the well child nursery and was discharged at 3 days of life. Polymerase chain reaction of dystrophin DNA from the affected infant and his affected sibs yielded normal-sized fragments of the dystrophin gene. This study cannot exclude a point mutation or small deletion and is normal in approximately 35 per cent of DMD patients. The patient refused muscle biopsy for confirmation of the diagnosis.

## DISCUSSION

The finding of large pleural effusions is rare and reports in the literature have been recently reviewed (Weber and Philipson, 1992). Most pleural effusions noted prenatally persist throughout pregnancy and may result in pulmonary hypoplasia. There is a large likelihood of poor outcome. Pleural effusions without hydrops fetalis, as here, are more frequently associated with a good outcome. Frequently,

no cause can be found for the pleural effusions. We are unaware of previous reports of pleural effusions in fetuses affected with DMD and their association may not be causal. Although infants affected with DMD are generally normal in the neonatal period, high serum levels of creatine phosphokinase are found, even to the extreme level reported here (Breningstall *et al.*, 1988). The clinical course of children with DMD includes motor delay and ultimately progressive loss of motor function resulting in death due to respiratory failure. Cardiomyopathy is frequent in patients with DMD. Induced fetal cardiac dysfunction in experimental animals rapidly leads to fetal pleural effusions that can progress to include polyhydramnios and hydrops fetalis (Gest *et al.*, 1990). We speculate that the effusion that appeared in this fetus could be attributable to cardiac dysfunction related to abnormalities of dystrophin. Fetal pleural effusions occur in fetuses affected with other myopathies, as in congenital myotonic dystrophy (Curry *et al.*, 1987), although their pathogenesis is not understood. Population studies have shown an unexplained increased rate of male stillbirths and neonatal deaths in families with DMD (Danieli *et al.*, 1980; Lane *et al.*, 1983). We do not know if pleural effusions occur in other fetuses with DMD, but speculate that pulmonary hypoplasia secondary to pleural effusions, or hydrops fetalis secondary to their progression, could lead to the observed stillbirths and neonatal deaths. Although spontaneous resolution of fetal pleural effusions is associated with neonatal survival, this case illustrates that infants affected *in utero* with pleural effusions may have an underlying disorder which may only be identified by long-term follow-up.

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