

Pregnancy and Charcot-Marie-Tooth Disease: Case Report

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Charcot-Marie-Tooth disease (CMTD) is a rare demyelinating disease of sensory and motor neurons which is transmitted in an autosomal dominant manner. It is associated with increased complications during pregnancy and emergency interventions during birth. The authors report a patient with CMTD who had an uncomplicated vaginal delivery.

HISTORY AND PRESENTATION

A 34-year-old Caucasian primigravida with known CMTD (type 1) presented at 10 weeks of pregnancy. Her CMTD, diagnosed in her 20s, caused fatigue and muscle weakness in her legs that were of typical champagne bottle appearance. She had no scoliosis, or respiratory or speech difficulties. Her gait was normal. She had not had any difficulties conceiving. After genetic counselling, she decided not to have prenatal invasive testing. She also had counselling from the paediatrician, and subsequently decided not to have neonatal diagnosis as the disease has a potential for very late presentation, sometimes as late as 50 years. She felt it may be more ethical to wait until the child would be capable of making decisions himself.

The patient's antenatal period was uneventful. Her 20-week anomaly scan was unremarkable. Clinically, as the growth of the fetus was satisfactory, she did not require any further scans. She had a discussion with

the anaesthesiologist regarding pain relief in labour, and was advised that fatigue during labour might interfere with maternal effort once the second stage of labour was reached. Epidural analgesia was recommended such that she might conserve muscle power until the head of the fetus was low in the pelvis. It was explained that spinal and epidural anaesthesia were not contraindicated. However, if she needed general anaesthesia, it was decided to avoid succinylcholine as it might have a prolonged effect on her.

Her labour began spontaneously at 41 weeks. She received epidural anaesthesia with bupivacaine and fentanyl for pain relief. She progressed well, did not need any augmentation, and in 10 hours spontaneously delivered a male baby weighing 3.26 kg. The baby was born in good condition and did not need any resuscitation. The Apgar score and neonatal examination were normal. The blood loss during delivery was 250 mL. The patient had an uneventful postpartum period and went home the next day.

DISCUSSION

CMTD affects one in 2,500 people. Symptoms begin most often in adolescence or early adulthood, but may be delayed until mid adulthood. It manifests as fatigue, weakness, scoliosis and, rarely, swallowing and speech difficulties and respiratory insufficiency. In addition,

the lower legs may take an "inverted champagne bottle" appearance due to peroneal muscle atrophy. As yet there is no known cure or treatment to arrest the progression.

CMTD may be associated with an entirely normal pregnancy in early or mild disease. However, a potential for significant morbidity exists if the disease is advanced and in later pregnancy. Advanced disease is also associated with older maternal age. Exacerbation of CMTD during pregnancy is known to occur. Patients notice increasing weakness which may be temporary or persistent. Exacerbations are usually recurrent in subsequent pregnancies. Among patients with subjective disabilities from childhood or youth, the risk of a noticeable exacerbation in at least one pregnancy is 50%, whereas there is no influence in pregnancy in patients with adult-onset CMTD.¹

The treatment of these patients requires a multidisciplinary team approach involving an obstetrician, a neurologist, a geneticist, an anaesthesiologist and a paediatrician. There is a 50% chance of inheritance, and genetic counselling is advised. Prenatal diagnosis is possible using amniocentesis or chorionic villus sampling. Fluorescent in situ hybridization is a reliable means for diagnosing the duplication of 17p12, which is found in 98% of patients with type 1 CMTD.²

Instrumental deliveries are more common among patients with CMTD because

of muscle fatigue and maternal exhaustion. Postpartum haemorrhage and presentation anomalies were shown to be more common in those affected.³ There is no evidence that CMTD is associated with any deleterious fetal outcome such as intrauterine growth restriction.

Referral to an anaesthesiologist is useful to plan general anaesthesia, if required, and

to allow discussion regarding intrapartum analgesia. Muscle relaxants can have a prolonged effect. However, it has been shown that succinylcholine can be used safely in patients with CMTD.⁴ Muscle relaxants such as atracurium and mivacurium are not shown to have a prolonged effect in these patients.⁵

Effective analgesia in the form of an epidural may be beneficial in avoiding muscle fatigue

in the first stage of labour, thus facilitating an unassisted second stage.

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