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CASE REPORT

Anaesthetic management during labour of a manifesting carrier of Duchenne muscular dystrophy

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SUMMARY. We describe the peripartum anaesthetic management of a 36-year-old woman who was a manifesting carrier of Duchenne muscular dystrophy. Duchenne muscular dystrophy is an X-linked recessive disorder affecting young males associated with severe complications during anaesthesia if depolarising neuromuscular blocking drugs and volatile agents are used. A manifesting carrier is a heterozygous female who demonstrates the disease in a milder form than in males. This probably occurs because of skewed X-inactivation. We planned to establish regional anaesthesia should an operation be necessary during labour or delivery and to use propofol total intravenous anaesthesia and rocuronium if general anaesthesia became unavoidable. At 37 weeks, the woman went into spontaneous labour, but fetal distress necessitated caesarean section for which combined spinal-epidural anaesthesia was used.

Keywords: Genetic diseases; X-linked, muscular dystrophy; Duchenne; Anaesthesia, obstetric

INTRODUCTION

Duchenne muscular dystrophy (DMD) is an X-linked disease of skeletal and cardiac muscle usually affecting males. General anaesthesia in affected individuals is associated with life-threatening peri-operative complications including cardiac arrhythmias, cardiac arrest and a malignant hyperpyrexia-like syndrome.[1] It is recommended that suxamethonium and inhaled volatile agents should be avoided in this population. Females who carry the abnormal X chromosome are usually asymptomatic but, rarely, can be manifesting carriers of the disease with symptoms and signs of the disease process. We consider issues surrounding the anaesthetic management during delivery of a manifesting carrier of DMD.

CASE HISTORY

The case concerns a 36-year-old woman who was a manifesting carrier of DMD. Since early childhood her calf muscles had been bulky, she could never run fast and performed badly on the school sports ground. From the age of 25, muscle weakness was pronounced and she began to have difficulty climbing stairs and rising from sitting to standing, indicating marked proximal weakness. Investigations showed a raised creatine kinase at 1920 units/L (normal: 25-170) suggesting muscular degeneration. Muscle biopsy confirmed DMD; there was a mosaic of dystrophin-positive and -negative muscle fibres indicating partial expression of the faulty gene. Her parents, sister and relatives had no history of the disease, which suggests a new mutation of the X chromosome. Molecular genetic studies were performed to determine the precise genetic abnormality and to direct screening of her family and their children. However, investigations using quantitative polymerase chain reaction, protein truncation tests and direct gene sequencing failed to show which X chromosome carried the mutant gene. It was therefore impossible to tell if the abnormality was maternal or paternal in origin.

Her first pregnancy had been nine years previously. During that pregnancy genetic analysis of a chorionic villus sample (CVS) at 12 weeks showed a female karyotype. The fetus was therefore at low risk of having DMD but had a 50% chance of being a carrier of the disease. After genetic counselling she continued with the pregnancy. An anaesthetic review at the time commented on the dangers of suxamethonium and inhaled volatile agents and that a regional block would

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be more suitable if caesarean section was required, but a detailed anaesthetic plan was not formulated. At term she went into spontaneous labour and required a ventouse delivery due to failure to progress. A pudendal nerve block was used for analgesia. She had a healthy baby girl.

In this, her second pregnancy, genetic analysis of the CVS taken at 12 weeks showed a male karyotype. The fetus had a 50% chance of having DMD. Fetal muscle biopsy at 22 weeks was normal, indicating that the fetus had inherited the normal X chromosome. She was referred to the obstetric anaesthetic service at 31 weeks' gestation. She was 1.65 m in height and weighed 60.3 kg at booking. Airway examination showed a Mallampati class 2 score.[2] A plan for her anaesthetic management was discussed. She was advised that we would try to avoid general anaesthesia if possible. We suggested starting epidural analgesia early in labour. If an epidural catheter was not in place and urgent caesarean section was required, standard spinal anaesthesia would be used but a combined spinal-epidural (CSE) would be preferred if time allowed. If general anaesthesia was unavoidable, she would be given rocuronium 0.6 mg/kg instead of suxamethonium at rapid sequence induction and total intravenous anaesthesia with propofol for maintenance of anaesthesia. A new breathing circuit would be used and the anaesthetic machine flushed with fresh gas for 15 min if she was admitted to the delivery suite in labour. The patient consented to this plan.

At 37 weeks she went into spontaneous labour. She progressed rapidly and her cervix was fully dilated on arrival to the delivery suite. She became fatigued and early decelerations were seen on the cardiotocograph. She was taken to theatre for assisted vaginal delivery. Blood pressure, oxygen saturation and electrocardiogram were monitored. In view of the possibility that a caesarean section might be required, a CSE was performed with epidural catheter insertion at L2/3 and spinal at L3/4. Clear cerebrospinal fluid (CSF) could be aspirated freely at the beginning, middle and end of injection. A total of 2.5 mL of 0.5% hyperbaric bupivacaine with diamorphine 300 μg was injected. This is the standard dose in our institution. Fifteen minutes after insertion, the block extended from C5 to L3 assessed by absence of cold sensation to ethyl chloride spray. Motor block was complete and 18 mg of ephedrine was required to maintain the blood pressure. The patient had no subjective respiratory compromise. Analgesia for the trial of ventouse was satisfactory but the attempt failed and preparations were made for caesarean section. An epidural top-up was not used to extend the block distally because of concern over increasing the block height, but over the next 20 min the block of cold sensation reached S1. The caesarean section was started and the baby boy delivered. He required suction and ventilatory support with bag and mask in the first minute. Apgar scores were 3, 8 and 10 at 1, 5 and 10 min respectively. The umbilical venous pH was 7.17. He was monitored on the special care baby unit for three days. He required no respiratory or cardiovascular support, was bottle fed and discharged home on day seven. The patient was comfortable during the procedure and her recovery thereafter was uncomplicated.

DISCUSSION

DMD is an X-linked recessive genetic condition affecting 30 per 100 000 live male births. It is characterised by progressive muscle weakness caused by the lack of dystrophin in muscle cells. Typically it affects the muscles of the shoulders and pelvis first, leading to difficulty climbing stairs and getting up from sitting on the floor (Gower's sign). It is usually diagnosed between the ages of 2 and 6 years and the disease progresses with death in early adulthood secondary to respiratory insufficiency. Cardiac muscle is also affected and can lead to cardiac failure. The genetic abnormality can be a new mutation of the X chromosome or, more commonly, the abnormality is inherited and there is a family history of affected males. To find the exact genetic mutation quantitative polymerase chain reaction, protein truncation tests and direct gene sequencing are used. Our patient was extensively investigated but the results were inconclusive. Her daughter has not been investigated because if she has inherited the same mutation the results would have a high probability of, again, being inconclusive. As her second child is a male without DMD, his X chromosome is normal.

Paediatric anaesthetists have most experience of perioperative complications and how to avoid them during their care of boys with DMD undergoing surgery. Complications include dysrythmias and cardiac arrest.[1,3,4] Rhabdomyolysis, raised creatine kinase, myoglobinuria, metabolic acidosis and acute renal failure are reported, suggesting a malignant hyperpyrexia-like syndrome.[1,3–6] The triggers for the above are depolarising neuromuscular blocking drugs and inhaled volatile agents. In planning an anaesthetic for a male patient with DMD, depolarising neuromuscular blocking drugs and inhaled volatile agents should be avoided. Monitoring and prompt treatment for malignant hyperpyrexia are vital. Regional anaesthesia is recommended where possible.[1]

Females are usually asymptomatic carriers of the abnormal gene. However, 2.5% of female carriers have symptoms of the disease.[6] These manifesting carriers have a prevalence 1 per 100 000 of live female births. Normally, carriers are unaffected because they have two X chromosomes, one normal and one affected. At