Rare disease

Management of a pregnancy complicated by type III spinal muscular atrophy

L Howarth, T Glanville

Department of Obstetrics and Gynaecology, St James's University Hospital, Leeds, West Yorkshire, UK

Correspondence to Dr L Howarth, louisehowarth@doctors.org.uk

Summary

The authors report the successful management of a pregnancy in a patient with spinal muscular atrophy (SMA) type III. It is a genetically inherited condition causing increasing weakness of the skeletal muscle. The patient in our case was confined to a wheelchair due to marked weakness in her lower limbs. A review of the available literature identified potential risk factors for the antenatal, intrapartum and postpartum period. These include increased risk of thromboembolism, urinary tract infections, intrauterine growth restriction and preterm delivery, anaesthetic problems and increased risk of uterine atony with subsequent postpartum haemorrhage. The authors report the management of these risk factors and the reasons for delivery by Caesarean section at 32 weeks. Apart from a decline in muscle function postdelivery requiring physiotherapy, there were no adverse outcomes for mother or baby. In both this case and on review of the literature, it is proven that a successful pregnancy is possible with SMA.

BACKGROUND

Spinal muscular atrophy (SMA) is a genetically transmitted neuromuscular disorder characterised by progressive degeneration of the anterior horn cell leading to a lower motor neuron lesion. Increasing weakness of skeletal muscle leads to an inability to move limbs and respiratory dysfunction.¹ There is a vast diversity in severity and speed of progression. Types I–III are autosomal recessive but there is also an autosomal dominant and an X-linked recessive form.² Type I is the most severe form usually resulting in death in infancy. Type II usually presents in infancy with death before adulthood.³ Type III has an extended duration and is considered mild.⁴ At present, there is no cure but supportive treatment is available with physiotherapy and surgery.² Preconception genetic testing is available to patients affected by SMA and their partners. Unfortunately, there is still little information or guidelines in the medical literature about the impact of pregnancy on clinical condition in SMA.⁵ Management of a pregnancy complicated by type III SMA is described here and highlights the advantage of multidisciplinary care to achieve a successful outcome for the mother and baby.⁶

CASE PRESENTATION

A 29-year-old nulliparous woman presented with type III SMA. There was marked weakness of the proximal lower limb musculature with a fixed flexion deformity of the hips and she was confined to a wheelchair. She had previous operations on her spine to correct progressive thoracic and lumbar scoliosis, which left her with metal rods and pins in her back.

She was interested in starting a family and sought genetic advice and her partner was tested and found not to be a carrier for SMA. In addition, she sought pre-conceptual counselling from a consultant in fetal and maternal medicine regarding management. She was advised about her risk of urinary tract infections and preterm delivery

(table 1). It was advised that she would need regular mid-stream urine microscopy and culture and cervical screening. If there was evidence of cervical shortening, consideration could be to inserting a cervical stitch or progesterone therapy. She also had an increased risk of thromboembolism with her reduced mobility. She had no personal or family history of thromboembolism, so it was decided that antenatal heparin could be avoided but 6 weeks of post-natal heparin was advised. She was counselled about the mode of delivery and while vaginal deliveries with SMA have been reported, in view of her stature and anticipated cephalopelvic disproportion it was advised that it would be unlikely she would reach full term and the aim would be for delivery by Caesarean section.

Following removal of the Mirena coil, she conceived spontaneously. A scan at 9 weeks demonstrated a viable intrauterine pregnancy. Booking body mass index was 20, weight 46 kg and height 152 cm. She was seen in the early pregnancy unit at 12 weeks with vaginal spotting but no cause for bleeding was identified. A scan at 13 weeks showed a nuchal translucency of 1.70 mm. Combined first trimester biochemistry and nuchal translucency gave a low-risk assessment for chromosomal abnormalities. Routine booking blood were taken. A plan for regular mid-stream urine microscopy and culture, cervical screening at 17 and 22 weeks, anaesthetic referral, compression stockings and follow-up anatomy scan with the consultant at 20 weeks was made. Anaesthetic review was initially at 15 weeks and the decision for a spinal catheter was made; the option of general anaesthesia was also discussed. Cervical screening at 17 and 22 weeks showed no evidence of funneling. Anatomy scan at 20 weeks was normal with normal uterine artery dopplers. Scans at 25, 27 and 29 weeks showed normal growth, normal amniotic fluid volumes and biophysical profiles. A plan was made for elective Caesarean section at approximately 34 weeks. She was reviewed by the orthopaedic spinal surgery team at 30 weeks because

Table 1 Summary of identified risk factors and management

Risk factor	Management
Baby affected by SMA	Preconception genetic counselling Partner tested for SMA found not to be a carrier
Urinary tract infections	Regular mid-stream urine microscopy and culture
Thromboembolism	Postnatal heparin Compression stockings worn throughout pregnancy and in postnatal period Antenatal heparin not advised as although mobility decreased compared with average patient there was no personal or family history
Preterm delivery	Cervical screening at 17 and 22 weeks To optimise chances of successful outcome with a cervical stitch if there was any evidence of shortening or progesterone therapy
Anaesthetic problems	Early referral to anaesthetic team for baseline assessment at 15 weeks followed by a second review the week prior to delivery date
Poor uterine capacity leading to increased risk of IUGR	Scanned at 20, 25, 27 and 29 weeks to check growth, amniotic fluid and biophysical profile
Immobility	See by risk assessment midwife and physiotherapists
Uterine atony and PPH	Syntocinon infusion given

IUGR, intrauterine growth restriction; PPH, postpartum haemorrhage; SMA, spinal muscular atrophy.

of episodes of increasing back pain. Apart from an admission with abdominal discomfort and back pain, she did not have any symptoms requiring inpatient assessment. However, after 30 weeks, there was increasing abdominal and back pain, as a result of the gravid uterus and small stature. This was managed with oral analgesia. Admission was declined.

Antenatal review prior to planned admission allowed multidisciplinary assessment. She was also reviewed by the risk assessment midwife who formulated a moving and handling plan in addition to deciding what equipment would be needed to optimise her care. Caesarean section was performed at 32 weeks following steroid cover. The gestation was chosen as a balance between anticipated premature delivery, fetal respiratory prematurity and maternal discomfort. By this gestation, the patient was struggling to mobilise due to increasing back and abdominal pain. Spinal catheter was successfully used for regional anaesthesia. Due to her tilted pelvis and inability to straighten her legs, positioning on the operating table was difficult. A Pfannenstiel incision was performed. The uterus was noted to be rotated to the right and the lower segment was poorly formed, however, a lower uterine incision was possible. A syntocinon infusion was given postdelivery as the patient was at risk of uterine atony. A live male infant weighing 1840 g with an Apgar score of 10 at 5 min was delivered; arterial pH was 7.31. She was transferred to the postnatal ward the same day where she received daily input from the physiotherapists due to deterioration in muscular condition. Heparin was prescribed for 6 weeks postpartum. She was discharged 7 days postdelivery and her baby remained for observation at the neonatal unit.

INVESTIGATIONS

- Cervical screening at 17 and 22 weeks.
- Scanning at 20, 25, 27 and 29 weeks to check growth, amniotic fluid and biophysical profile.

OUTCOME AND FOLLOW-UP

- Delivery by Caesarean section at 32 weeks gestation.

DISCUSSION

A MEDLINE search from 1950 to November 2009 using the key words pregnancy and SMA was undertaken.

Complications included premature labour, presumed to be due to muscle weakness and kyphoscoliosis impeding a successful gestation to be obtained,^{7 8} prolonged labour,⁹ delayed postpartum recovery and muscle wasting in the second trimester of pregnancy. Muscle weakness was found to be worse because of the effects of SMA on pregnancy and involuntary inactivity as a result of being pregnant.⁸ Neither age of onset nor course of disease was found to be useful in predicting worsening SMA in pregnancy.⁸ Worsening respiratory function is also reported in pregnancy complicated by SMA. This may be a result of wheelchair dependency as well as respiratory muscle involvement. Pressure exerted by the diaphragm may exacerbate these problems.⁵ Increased rate of urinary tract infections also complicates SMA pregnancies.¹⁰

Mode of delivery needs to be planned carefully and Caesarean section is more common in SMA pregnancies although vaginal delivery may be possible even in patients with type II SMA.^{8 10} Musculoskeletal deformities such as scoliosis and contractures of the legs not only have an impact on the mode of delivery but also on the method of analgesia. Musculoskeletal deformities lead to failure of regional techniques and therefore a general anaesthetic may have to be used.⁵ Contractures of legs, fixed flexion deformity of the hips and scoliosis also cause difficulty when it comes to gaining access to the lower abdomen as was encountered in this case. Delayed postpartum recovery in wheelchair users meant that assistance in caring for the baby was necessary in this case. Women with SMA planning for a pregnancy should be informed that they may experience a worsening of muscle weakness secondary to the pregnancy and delivery.⁸

Learning points

- Management of delivery is a balance between neonatal versus maternal factors.^{6 11}
- •In both this case and on review of the available literature, it is proven that a successful pregnancy is possible with SMA.
- •Thorough pre-pregnancy counselling and a multidisciplinary team approach facilitate best results.

Competing interests None.

Patient consent Obtained.

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