

BRUCK SYNDROME IN PREGNANCY

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INTRODUCTION:

Bruck syndrome is a rare, autosomal recessive condition associated with features of both arthrogryposis and osteogenesis imperfecta. It is characterised by congenital large joint contractures and bone fragility leading to fractures and deformities. There are fewer than 50 described cases of Bruck syndrome in the literature, with no reported cases in pregnancy. We describe a case of a successful pregnancy in a woman with Bruck syndrome.

Patient positioning



CASE REPORT:

A 27-year-old woman with osteogenesis imperfecta and significant lower limb contractures, presented in her first pregnancy. Her BMI was 15 kg/m² (30kg/130cm) and she was wheelchair-bound. Her past medical history included spinal-fusion surgery for scoliosis and iron deficiency anaemia which had not been fully investigated due to unsuccessful endoscopies. She required regular blood and iron infusions throughout pregnancy.

She was referred to a clinical geneticist, where a homozygous *FKBP10* gene mutation was identified, confirming **Bruck syndrome**. A Caesarean section was recommended due to her significant lower limb contractures and risks of an unplanned Caesarean section. A general anaesthetic was recommended due to the technical difficulties and unreliable spread of a regional analgesia. Pre-delivery lung function testing demonstrated a restrictive pattern. A simulated theatre trial was performed to address difficulties in positioning, access to the lower abdomen and delivering the baby. Occupational therapy recommended use of a weighted doll to identify difficulties with her home set-up.

At 36+4 weeks' gestation she presented with spontaneous contractions. A Caesarean section was performed under general anaesthesia via a high transverse uterine incision. Blood loss was 500ml which led to a haemoglobin drop from 96g/L to 64g/L, requiring 1-unit blood transfusion. Postoperative pain management was challenging but respiratory support was not required.

DISCUSSION:

This is the first reported case of pregnancy in a woman with Bruck syndrome. The main obstetric risks anticipated were preterm delivery and the risks of an unplanned, complicated Caesarean section. Early pre-delivery airway assessment and preparation for difficult intubation were essential to minimise neck and jaw manipulation and reducing risk of fractures/atlando-axial injury. Lung function testing indicated the possible requirement for post-operative respiratory support; however, it was predicted her lung function would improve once the impact of the gravid uterus was removed. Surgical challenges included positioning and access to the lower abdomen during Caesarean section. Care was taken to support the patient's contracted limbs without force to avoid dislocation or fracture. Blood loss was meticulously managed due to her anaemia and low weight.

CONCLUSION:

Pregnant women with Bruck syndrome require a multidisciplinary approach including input from obstetric and fetal medicine specialists, midwives, anaesthetists, geneticists, occupational therapists and physiotherapists.