

Anaesthetic implications for an obstetric presentation of Mucopolysaccharidosis type 1 (Hurler-Scheie syndrome)

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Case

A 30 year old woman recently diagnosed with Mucopolysaccharidosis type 1, (Hurler-Scheie syndrome) was reviewed in the obstetric anaesthesia high risk clinic.

- She had multi-system manifestations of the condition, the key areas of concern with regards to her anaesthesia were:
- Restrictive spirometry pattern: FEV₁ 46% FVC 45%
 - Moderate obstructive sleep apnoea
 - Micrognathia (had undergone jaw surgery to try and improve) and macroglossia.
 - Significant short stature, a booking weight of 34kg
 - Atlanto occipital instability; MRI spine illustrated cervical stenosis but no compression (with no contraindications to neuraxial anaesthesia)
 - Severe visual impairment
 - Coagulation abnormality with low levels of factor XI
 - Bicuspid aortic valve with mixed aortic (mild regurgitation and moderate stenosis) and mitral disease (mild stenosis, mild-moderate regurgitation) but normal LV systolic function.

She had previously undergone maxillofacial surgery for micrognathia, for which awake fibreoptic intubation had been challenging. Airway assessment revealed a class 4 mallampati score, adequate mouth opening although macroglossia, and adequate neck movements.

Her English was limited and she was extremely worried at the prospect of having a general anaesthetic and not being able to be with her husband. This was a major deciding factor in her anaesthetic management during surgery.

Written consent by the patient was gained for presentation and publication.

Mucopolysaccharidosis type 1: Hurler-Scheie Syndrome

Mucopolysaccharidosis type 1 (MPS1) is one of a group of inherited metabolic conditions, the mucopolysaccharidoses [1]. These are a subset of lysosomal storage disorders, which result in incomplete degradation of glycosaminoglycans (previously known as mucopolysaccharides). The undigested glycosaminoglycans accumulate in various tissues causing multisystem disease.

There are three forms of MPS1: Hurlers syndrome (the most severe form), Scheie syndrome (an attenuated form) and Hurler-Scheie syndrome (an intermediate form). The attenuated forms (Hurler-Scheie and Scheie syndromes) have a slower disease progression and as such are usually diagnosed later in life. There is a wide spectrum of disease severity across the attenuated forms. Inheritance in autosomal recessive and incidence of the attenuated form is approximately 1:500,000 live births.

Multiprofessional Working

During her assessment prior to surgery, she met several specialists. Any updates of her care were discussed at the weekly high-risk meeting, including input from obstetricians, midwives, maternal-medicine physicians and anaesthetists. There was additional input from cardiologists, haematologists, geneticists, respiratory medicine and intensive care teams. The availability of video conferencing made this process very straightforward.

The couple lived 45 minutes from the tertiary centre where the bulk of their care took place. This involved ongoing inter-hospital communication for results and regular travelling for the couple. Steps were made to incorporate multiple assessments into one day to minimise the number of trips undertaken. As English was not her first language, steps were made to optimise understanding during visits, including use of translation services.

Reference

1. Mucopolysaccharidosis Type I - Symptoms, Causes, Treatment, National Organization for Rare Disorders (NORD). Available from: <https://rarediseases.org/rare-diseases/mucopolysaccharidosis-type-i/> (accessed 22/4/2024)



Pre-operative assessment

She was seen in anaesthetic cardiac clinic where the information already gleaned was consolidated and a discussion took place between her and her partner about the anaesthetic options and associated risks and benefits.

- The key issues were:
- known difficult airway
 - potentially difficult regional anaesthesia
 - low height body weight, concerns re: bleeding and high block
 - valvular abnormalities causing concern with rapid changes in SVR and avoidance of some uterotonics
 - mild coagulation abnormality (factor XI deficiency)
 - Significant deconditioning in pregnancy, almost wheelchair bound prior to CS
 - language barrier with patient

It was made clear that the safest plan was likely to be a planned general anaesthetic with an awake fibreoptic intubation, however the patient was very clear that she wanted to be awake with her husband present unless it absolutely could not be avoided. After long discussions with the couple, an anaesthetic management plan was agreed on.

Planning

A planned caesarean section was arranged to occur at 32 weeks gestation. A significant proportion of the operating list was reserved, to enable all staff to focus attention and to mitigate for potential complications. Two consultant anaesthetists and consultant surgeons managed the case, with support from cell salvage and neonatal teams.

- As the maternity site is distant from other theatre suites in the hospital, equipment and teams needed to be arranged in advance to enable them to be immediately available in case of need. There were several areas that required specific planning:
- Small sized (paediatric) equipment needed to be available for both regional and general anaesthetic procedures, with spares and alternative sizes.
 - With coagulation abnormalities and low body weight, a factor replacement (FFP pre-op) and reduced major obstetric haemorrhage threshold was derived, with planned use of cell salvage.
 - Familiarity with paediatric difficult airway and patients with cardiac abnormalities was prepared for through anaesthetic consultant cover on the case.
 - In addition the paediatric airway team were on standby with local paediatric ENT surgeons on site

A briefing was undertaken on the morning of surgery with all teams present.

Regional Anaesthesia

Prior to anaesthesia, two large bore intravenous cannulae were inserted and an arterial cannula for invasive monitoring. According to the patient’s preference she underwent a combined spinal epidural approach for regional anaesthesia. Whilst low dose spinal anaesthesia was sited with ease, epidural siting was complicated by blood in the catheter and inability to thread the catheter. The third attempt was successful enabling cautious epidural top up. Once appropriate block height had been confirmed (assessed by both consultants and agreed by patient) surgery commenced.

Unfortunately, prior to uterine incision the patient complained of pain and so induction of general anaesthesia was required.

General Anaesthesia

Due to her significant deconditioning in pregnancy, high flow nasal oxygen was commenced from the start and following placement of neuraxial anaesthesia, attention was paid to optimise a ramped position.

As expected, intubation was difficult with a very anterior position to her larynx making use of bougie and stylet difficult prior to desaturation. Initial attempts undertaken with Glidescope® and D blade were unsuccessful. With two handed bag mask ventilation and an oropharyngeal airway, oxygenation was manageable allowing successful asleep nasal fibreoptic intubation.

The case was complicated by major obstetric haemorrhage (1.4L) requiring packed red cells in addition to cell salvage blood and IV crystalloid fluid. Unfortunately, this likely contributed to pulmonary oedema, requiring diuresis.

Post-operative Care

The patient went to intensive care post-operatively as planned. She was extubated the next day and had a three day stay here prior to discharge back to the post-natal ward and discharge from hospital five days after surgery.

What have we learnt?

Multidisciplinary collaboration and extensive planning enabled the patient to reach 32 weeks gestation. Her theatre journey was complex, requiring enactment of each level of her anaesthetic plan. We attempted to respect her very strongly expressed wish to avoid a GA by attempting regional anaesthesia; unfortunately, this was difficult to site and inadequate for surgery. Despite this and a major haemorrhage both her and her baby did well.

Regular tertiary reviews and counselling were necessary due to her complexity and physical deterioration, however for the couple this was a noted burden. Thankfully, planning and preparation ensured that appropriate equipment and clinically experienced staff were available to ensure a successful outcome.