Anaesthetic considerations of obstetric patient with Stickler Syndrome: A Case Report

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Introduction

Stickler syndrome (hereditary progressive arthro-ophthalmopathy) is a rare genetic connective tissue disorder arising from mutations in genes encoding collagen types II, IX and XI.(1) The prevalence at birth is approximately 1/7,500 to 1/9,000 with the varying phenotypical expression.(2) Most common is autosomal-dominant inheritance, although autosomal-recessive forms of Stickler syndrome do occur. Stickler syndrome can affect several different systems and is characterised by ophthalmic, auditory, orofacial and skeletal manifestations. Orofacial features include Pierre Robin sequence (PRS), cleft palate and midfacial hypoplasia. Musculoskeletal problems include spinal abnormalities, hypermobility, epiphyseal dysplasia and premature osteoarthritis.(3) The combination of orofacial and skeletal manifestations presents significant anaesthetic challenges in obstetric patients with Stickler syndrome.

Case

A 36-year-old female, G2P1, ASA III, with Stickler syndrome presented for elective caesarean section at 39 weeks gestation. She had a previous caesarean section under combined spinal-epidural (CSE) anaesthesia without issue. She had a diagnosis of Stickler syndrome as a teenager and subsequent retinal detachment repair, Achilles' tendon surgery and several palate/cleft palate repairs. Airway assessment demonstrated a potentially difficult airway. There was evident moderate scoliosis of the lumbar spine on clinical exam. No lumbar spine imaging was available. Ultrasound examination of the lumbar spine revealed an access window on the convex angle at the L3-4 interspace. Placement of a CSE was difficult requiring ultrasound to guide placement. An 18G Touhy needle achieved loss of resistance at 7cm with a midline approach angled in the transverse plane toward the convex angle of the scoliosis. Needle-through-needle technique with 25G Whitacre needle revealed clear CSF and spinal anaesthetic was administered. An epidural catheter was sited with 5cm left in the epidural space. Sensory block to T6 was established bilaterally and an epidural top up was not needed during the case. The caesarean section was uncomplicated. Antibiotics, analgaesia, antiemetics and uterotonics given as standard. The epidural was removed at the end of the procedure. The patient was followed up with no issues or concerns prior to discharge.

Discussion

Stickler syndrome in the obstetric patient presents a unique combination of challenges. Craniofacial abnormalities paired with the physiological characteristics of the airway in pregnancy result in a potentially difficult airway and regional anaesthesia is recommended.(4) In this condition spinal abnormalities can also present a significant challenge in placement of successful neuraxial anaesthesia, especially in time critical obstetric situations and may increase the risk of complications from neuraxial. (5) Radiological imaging can provide vital information in patients with significant scoliosis, most used are x-ray and computed tomogram (CT). The Cobb angle can be calculated to give an indication of severity, vertebral body rotation. Research has shown ultrasound to be as affective as CT to guide neuraxial anaesthesia. The benefits of using ultrasound over CT are that it is portable, relatively inexpensive, free of ionizing radiation and provides images to the operator in real-time.(5)

In this case, neuraxial ultrasound provided anatomical information including the depth of spaces, identification of the midline and intervertebral level and the interspinous and interlaminar spaces, along with the angle of vertebral body rotation. Our patient had no previous imaging of her thoraco-lumbar spine. We noted an increase in the difficulty of the neuraxial anaesthesia compared with previous. Early anaesthetic assessment in this case would likely have revealed a change in the degree of scoliosis in this progressive condition and provided an opportunity to appropriately counsel the patient on their anaesthetic management and potential difficulties.

Given this we are advocating for pre-operative anaesthetic review well in advance of delivery for each pregnancy the patient has. In some severe cases this discussion may be best had prior to conception. Early pre-operative review allows for a thorough evaluation of the patient, their anaesthetic history, and any imaging they have. It also allows for clinical examination and to utilise preprocedural ultrasound examination in this setting. By performing preprocedural ultrasonography of the lumbar spine in the pre-operative setting we can identify and alert to a potentially difficult or impossible procedure. We can also document relevant data from the images obtained such as identification of the midline, vertebral body rotation and depth to ligamentum flavum. These will be of use to the anaesthesiologist on the day of delivery, particularly in a time sensitive scenario.

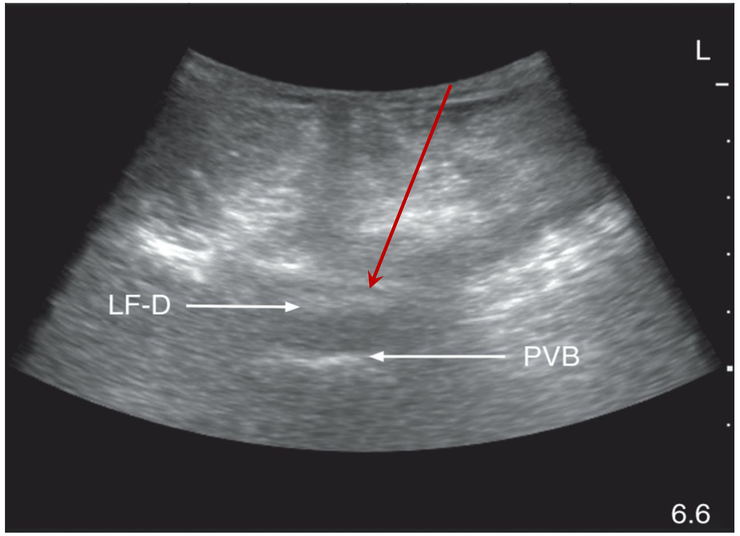


Figure 1: Example of Transverse Midline Ultrasound image showing rotation of vertebral body. Red arrow direction towards the convex angle of the scoliosis. LF-D, ligamentum flavum-dura mater complex; PVB, posterior vertebral body (9)

Conclusion

Our aim with this case is to notify our colleagues about the peri-operative considerations specific to those with craniofacial and spinal abnormalities in the obstetric setting and to emphasise the value of early pre-operative assessment. This case also serves to highlight the role of ultrasound in ensuring safe and correct placement of regional anaesthesia. The patient has consented to the presentation of their case and its management, considering it valuable to increase awareness of the condition.

References

1. Robin NH, Moran RT, Ala-kokko L. Stickler Syndrome Summary Genetic counseling Suggestive Findings. 2021;1–19.
2. Orphanet: Stickler Syndrome [Internet]. [cited 2021 Nov 13]. Available from: https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=828
3. Stickler GB, Hughes W, Houchin P. Clinical features of hereditary progressive arthroophthalmopathy (Stickler syndrome): A survey. Genet Med. 2001;3(3):192–6.
8. Porter D. Anaesthesia recommendations for patients suffering from Stickler Syndrome. Orphan Anesth. 2017;
9. Bowens C, Dobie KH, Devin CJ, Corey JM. An approach to neuraxial anaesthesia for the severely scoliotic spine. Br J Anaesth. 2013;111(5):807–11.

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