Anaesthetic Implications for a Parturient with Noonan's Syndrome

Dr. Shilpi Sethi , Dr. Samantha James, University Hospital Coventry and Warwickshire

NHS
University Hospitals Coventry and Warwickshire
NHS Trust

Introduction

Noonan syndrome (NS) is an autosomal dominant genetic disorder

Incidence of 1 in 1000 to 2500 live births

Associated with distinct systemic features and linked with key genetic mutations [1]

Peripartum management presents unique anaesthetic challenge

Case

28-year-old primigravida
37 + 3 weeks' gestation
Induction of labour due to comorbidities

Confirmed diagnosis of Noonan's syndrome, with genetic testing revealing a pathogenic mutation in the PTPN11 gene

Physical features: webbed neck, short stature (153 cm, 46 kg), ocular albinism, and optic disc hypoplasia.
Airway: Mallampati class II, temporomandibular joint pain
Spine: mild kyphoscoliosis

Echocardiogram: thickened pulmonary valve, mild pulmonary valve stenosis and, good LV systolic function, aneurysmal interatrial septum

Peripartum management

Detailed antenatal anaesthetic assessment prior to admission

Options for labour analgesia

Preprocedural neuraxial ultrasound assessment

Anaesthetic provision for LSCS – Neuraxial vs General anaesthetic

Failed induction → Category 2 LSCS

Coagulation studies normal

Successful USG spinal anaesthesia – paramedian approach, low dose spinal with opiod

Uterotonics – carbetocin

EBL 450ml

Uneventful recovery

Discussion

No consensus on the optimal anaesthetic technique

Although neuraxial anaesthesia can be technically challenging, it remains advantageous when feasible due to the risks associated with general anaesthesia, including airway difficulty and the potential for adverse cardiopulmonary responses

PTPN11 encodes cell-signaling mediator SHP-2, a non-transmembrane protein tyrosine phosphatase which modulates platelet responses, reduced platelet aggregation, low levels of specific clotting factors [2]

Preoperative ultrasound facilitates identification of intervertebral spaces, estimates the depth of the epidural space, and improves first-attempt success rates

Conclusion

Thorough preoperative evaluation

Pre-procedural imaging[3]

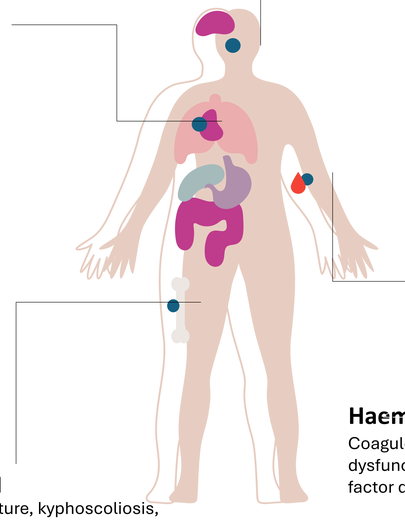
Multidisciplinary planning

Customised anaesthetic approach

Systemic presentation

Cardiovascular
Congenital heart disease, pulmonary stenosis, atrial septal defects, hypertrophic cardiomyopathy

Craniofacial
Short broad nose, ocular hypertelorism, micrognathia, short neck and excess skin folds



Haematological
Coagulopathy, platelet dysfunction, clotting factor deficiencies

Skeletal
Short stature, kyphoscoliosis, chest wall deformities

References

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[3] Karmakar MK, Li X, Ho AM, Kwok WH, Chui PT. Real–time ultrasound–guided paramedian epidural access: evaluation of a novel in–plane technique. Br J Anaesth. 2009; 102:845–854.