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Osteogenesis Imperfecta : Anesthetic implications

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ABSTRACT

Osteogenesis imperfecta is an inherited connective tissue disorder with multi-system involvement.Commonly encountered complications include difficult airway, coagulation dysfunction , respiratory compromise due to skeletal deformity , congenital cardiac anomalies and these issues pose an anaesthetic challenge due to inherent complications associated with it. Proper pre-operative evaluation, preparation and vigilant intra-operative management will improve the outcome in these patients.

Osteogenesis Imperfecta is an inherited disorder of connective tissue caused by mutation of collagen 1 gene and usually transmitted as as an autosomal dominant trait.The pathophysiology of the disease revolves around the defects in collagen formation leading to easily fractured bones, hypermobile joints,blue or grey sclera,skeletal deformity and fragile skin.[1]

Osteogenesis Imperfecta is of 4 types with varying inheritance pattern and degree of severity.[2]

These patients also have a greater incidence of airway anomalies, thoracic anatomy abnormalities, coagulation dysfunction, hyperthyroidism, and an increased tendency to develop perioperative hyperthermia.[3]These isssues may be of concern during anesthetic management.

This article will provide a brief account of problems associated with osteogenesis imperfecta as well as measures to avoid complications intra-operatively.

Positioning

Due to propensity for these patients to develop fractures ,care must be taken when handling and transferring these patients along with careful padding of pressure points.[3]

Joint laxity can lead to dislocation,hence care should be taken to avoid over-extension during positioning for surgical exposure.[1]

Automated blood pressure cuffs may be hazardous as over inflation may may result in fracture.[4] During insertion of intravenous catheter , light pressure should be applied above the site of insertion or a loosely applied tourniquet may be used.[1]

Airway management

Airway management in an osteogenesis imperfecta patient coming for surgery is a challenge for anaesthetist. Due to abnormal skeletal growth , the risk of odontoaxial dislocation and hypermobile joints, a difficult airway must always be anticipated in these patients.[5]

Facial anomalies, risk of mandibular fracture, injury to teeth ,short neck,pigeon chest,predisposition to mucosal bleeding, associated basilar invagination with distorted airway anatomy,high chances of cervical vertebrae injury associated with osteogenesis imperfecta make direct laryngoscopy and intubation difficult.[6][7]

The optimal positioning can be achieved by carefully placing a blanket beneath the upper back to help restore anatomical relationships.[1]

Fibre-optic intubation would appear to be an ideal aid in securing the airway in these patients.[8] Other methods of maintaining cervical spine immobility during intubation include the use of an intubating laryngeal mask airway or in emergency cases the use of a stylet.[1]

Spinal deformity

Kyphoscoliosis and thoracic cage deformity is prevalent in osteogenesis imperfecta population.[1] Scoliosis results in reduced vital capacity , reduced FRC and restrictive pulmonary disease pattern charecterised by increased respiratory rate and decreased tidal volume.[9] More severe the thoracic curve (greater cobb's angle), more profound the disturbances in pulmonary function.[10] Significant pulmonary impairment may result in cor-pulmonale.[8] Also kyphoscoliosis and other thoracic skeletal deformities may predispose these patients to post-operative respiratory failure.[3]

Pulmonary compromise is the leading cause of death in these patients.[11]

Cardio-vascular system

Cardio-vascular involvement may occur in patients with osteogenesis imperfecta.Aortic insufficiency is the most common valvular abnormality followed by mitral regurgitation.[12]Other abnormalities like atrial or ventricular septal defect , patent ductus arteriosus , aortic dissection, left ventricular rupture have also been reported.[3]

Most of the abnormalities may prove insignificant and non-progressive requiring no surgical intervention.[1] Clinical examination and echo-cardiography may be helpful in detecting these abnormalities. Surgical procedures performed in these patients carry a higher risk of complications related to platelet dysfunction,friable tissues, impaired wound healing and musculoskeletal weakness and deformity.[13]Patients with mitral valve prolapse, valvular dysfunction or congenital heart disease should receive prophylactic antibiotics to prevent sub-acute bacterial endocarditis.[3]

Temperature dysregulation

There have been reports of increase in temperature in patients with osteogenesis imperfecta both with and without anaesthesia[8].It has been suggested that hyperthermia is the result of either abnormal central nervous system temperature regulation or abnormal cellular energy metabolism[1].Additionally , these patients have increased basal creatine k kinase and pyrophosphate concentrations , both associated with an increased risk of malignant hyperthermia[14]. Also thyroid storms can occur intraoperatively, as 50% of the patients with osteogenesis imperfecta have increased levels of thyroxine.[6]

It presents as fever, tachycardia, hypoxia, acidosis and elevated serum CPK levels.[15]

Routine use of anticholinergic medications like atropine and inhalational anesthetics (halothane,isoflurane) is to be avoided as

these agents can trigger hyperthermia in susceptible individuals.[3]

The use of depolarising muscle relaxant like succinyl choline should be avoided because of its potential to cause malignant hyperthermia and fractures due to fasciculations.[5] Pancuronium bromide or atracurium are the muscle relaxants of choice. [15] Pulse oximetry and end-tidal carbon dioxide monitoring may herald the warning of hypermetabolism intra-operatively.[8]

The presence of hyperthermia under anesthesia can be usually controlled with cooling,supplemental oxygen,sodium bicarbonate,cardiovascular stimulants and intravenous dantrolene.[1]

Haemorrhagic diathesis

A 10-30% in incidence of bleeding diathesis has been reported in patients with osteogenesis imperfecta. [16]These individuals have an increased risk of intraoperative bleeding despite having normal bleeding times and coagulation profile.[5] The coagulation defect appears to be due to the effect of the abnormal collagen on platelet-endothelial cell interactions and resulting in increased capillary fragility, defective contraction of small vessels after injury and defective platelet aggregation.[3] Other abnormalities noted include decreased platelet retention, decreased factor 8 antigen and ristocetin co-factor.[17]

Platelet works and Sonoclot evaluation helps in the correction of coagulopathy in these patients [12]Tourniquet test is a valuable screening test in patients with osteogenesis imperfecta.[17]

Patients with pre-operative platelet counts of < 10,000 to 20000 ×109/l should receive platelet transfusions before surgery.[1]

Coagulopathy with sudden development of widespread petechiae has also been reported.[18]Therefore due precautions regarding any unexpected bleeding should be taken in the form of availability of adequate blood, FFP, platelet concentrates, cryoprecipitates.[6]

Desmopressin, a synthetic analog of ADH may be useful for improving hemostasis and decreasing transfusion requirements in surgical patients with osteogenesis imperfecta.[16] Medications , most importantly NSAIDS which have deleterious effects on platelet function should be avoided [3].

Central nervous system

Central nervous system involvement is in the form of basilar impression or invagination syndrome, cranio-vertebral junction anomalies including atlanto-axial subluxation, hydrocephalus, early onset deafness and anatomical/developmental abnormalities.[3]

Basilar invagination is seen in 25% of subjects with osteogenesis imperfecta with highest frequency in type 4 B[19]. It occurs as the weight of the cranium deforms the soft skull base producing invagination of foramen magnum and translocation of the upper cervical vertebra into posterior cranial fossa causing severe neurologic disability, respiratory compromise and sudden deaths.[7]

The signs and symptoms include headache characteristically occipital,precipitated by coughing and laughter,trigeminal neuralgia,vertigo,weakness of arms and legs,horizontal,upbeat and convergence nystagmus,proprioceptive defect,sleep apnoea,seizures,papilledema in acute hydrocephalus.[20]

Screening of all patients of osteogenesis imperfecta particularly type 4 B with CT sagittal mid-line reconstruction of cranial base and hard palate is recommended.[19] Brain MRI can be done in selected cases.[20]

Excessive neck extension is to be avoided during intubation due to the risk of vertebral artery dissection and stroke.[21]

Anaesthetic management

Anesthetising a patient with osteogenesis imperfecta is a challenging event for most anaesthesiologist.

There have been several case reports of conductance of surgery under general as well as regional anesthesia in these patients.[5]

Regional anaesthesia is the method of choice.[6]Firstly , it avoids the need for tracheal intubation (laryngoscopy and tracheal intubation associated with risk of odonto-axial dislocation, fracture mandible and cervical vertebrae,injury to teeth) , makes development of hyperthermia less likely and facilitates detection of thyroid storm.[19]

But technical difficulty associated with epidural block placement , inability to tolerate supine position awake and a pre-existing coagulopathy from platelet abnormalities may preclude regional anesthesia in these patients.[2]

Caudal epidural is preferred to spinal anesthesia as it is difficult to perform lumbar puncture due to associated kyphoscoliosis and unpredictability of the level of block.[4]

When general anesthesia is considered, meticulous attention is required with the use of anticholinergic agents,inhalational anesthetics, neuro-muscular blocking agents, airway management, positioning of the patient and acute pain management.[6]

Fibre-optic intubation is the method of choice in these patients. [7]ILMA provides an acceptable control of airway for general anesthesia in these patients with minimal neck extension.[3]

Ketamine is the preferred anaesthetic agent but is contraindicated in the presence of hydrocephalus.[1]

Dexamethasone may be given pre-operatively to reduce airway edema.[8]

A Reduction in FRC (restrictive disease) and V/Q mismatch will lead to hypoxemia; Hence may require high FiO2 or PEEP.[1]

Delayed extubation should be anticipated in these patients and if extubation is allowed,supplemental oxygen should be administered t ill the patient is fully awake and recovered from anesthesia effects.[1]

To summarise ,osteogenesis imperfecta patients pose a significant challenge to anesthesiologists due to inherent complications associated .Among which difficult airway , problems with positioning , bleeding disorders ,cardiovascular anomalies are of prime importance. Proper pre-operative evaluation and intraoperative management will lessen the adverse outcomes in these patients.

REFERENCE

1. Oakley I, Reece LP. Anesthetic implications for the patient with osteogenesis imperfecta. AANA journal 2010;78:47–53. | 2. Vogel TM, Ratner EF, Thomas RC. Pregnancy Complicated By Severe Osteogenesis Imperfecta :A Report of two cases.Anesth Analg 2002;1315–7. | 3. Gregory AS, Joseph DT.Perioperative Care of the patient with osteogenesis imperfecta.Orthopaedics 2007:30 :1-9 | 4. Garg M, Jain M, Gupta A. Anaesthetic management of a case of osteogenesis imperfecta with urinary bladder stone - a case report. Indian J of anaesth 2009;53:68–70. | 5. Hosalli V, Hegde H V,Ambi U,Hulkund S. Anaesthetic Management of a Case of Osteogenesis Imperfecta with Bladder Outlet Obstruction : A Case Report. J of Clinical and Diagnostic Research 2011;5:640–2. | 6. Chandran R, Dave N , Padvi A , Garasia M.A rare presentation of a child with osteogenesis imperfecta and congenital laryngomalacia for herniotomy.Indian J of Anaesth 2011;55:534-6 | 7. Papneja C, Tandon MS ,Singh D , Ganjoo P.Anaesthetic management in a patient of Osteogenesis Imperfecta with basilar invagination.The Internet J of Anesthesiol 2006;10(2). | 8. Rodrigo C. Anesthesia for Maxillary and Mandibular Osteotomies in Osteogenesis Imperfecta.Anaesth Prog 1995;42 :17–20. | 9. Kulkarni HA, Ambareesha M.Scoliosis and anaesthetic considerations .Indian J of Anaesth 2007;51:486-95. | 10. Korula S, Saramma SP, Ipe S. Parturient with severe kyphoscoliosis: An anesthetic challenge. J of Obstet Anaesth Crit Care 2011;1:81-4. | 11. Widmann RF, Bitan FD, Laplaza FJ, Burke SW, DiMaio MF, Schneider R. Spinal deformity, pulmonary compromise, and quality of life in osteogenesis imperfecta. Spine 1999;24:1673–8. | 12. Huang J, Dinh M, Kuchle N, Zhou J .Anesthetic management for combined mitral valve replacement and aortic valve repair in a patient with osteogenesis imperfecta ..Ann Card Anaesth 2011;14:115-8. | 13. Wong RS, Follis FM, Shively BK , Wernly JA. Osteogenesis imperfecta and cardiovascular diseases. The Ann Thorac Surg 1995;60:1439-43 | 14. Ramton J ,KeelDA,ShanananEC,IngramGS.Occurence of malignant hyperpyrexia in a patient with osteogenesis imperfecta. Br J Anaesthesiol 1984;56:1443-45. | 15. Ryan CA,Al-Ghamdi AS,Gayle M , Finer NN. Osteogenesis Imperfecta and Hyperthermia.Anesth Analg 1989:68:811-4. | 16. Keegan MT , Whatcott BD,Harrison BA.Osteogenesis Imperfecta, Perioperative Bleeding, and Desmopressin. Anesthesiology 2002;97:1011-3 | 17. Evensen SA, Myhre L, Stormorken H. Haemostatic studies in osteogenesis imperfecta. Scand J of haematol 1984 2013 Jul 7];33(2):177–9 | 18. Bhandari G, Shahi K S, Bhadoria P, Bhalotra AR, Sandhya O D, Arya M Osteogenesis Imperfecta; No Place for Imperfect Anaesthesiologist Indian J Anaesth.2008;52:577 | 19. Sillence DO. Craniocervical abnormalities in osteogenesis imperfecta: genetic and molecular correlation. Pediatric radiology 1994;24(6):427–30. | 20. Hayes M, Parker G, Ell J, Sillence D. Basilar impression complicating osteogenesis imperfecta type IV: the clinical and neuroradiological findings in four cases. J neurol neurosurg psychiatry 1999;66:357-64. | 21. Zotter H,Zenz W, Gallisti S, Zoher B ,Lindbichler F.Stroke following appendectomy under general anaesthesia in a patient with basilar impression. Acta Anaesthesiol Scand 2000;44:1271-2.