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CLINICAL

Osteopetrosis

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ABSTRACT

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INTRODUCTION

Osteopetrosis:

Osteopetrosis or marble Bone Disease is a bone disease that makes bones abnormally dense and prone to fracture.

The different types of osteopetrosis which are usually distinguished by their pattern of

inheritance are-

1. Autosomal Dominant
2. Autosomal Recessive
3. X linked.

Autosomal Dominant Osteopetrosis also known as Albers-Schonberg disease is typically mildest type. signs & symptoms includes multiple bone

fracture, scoliosis, arthritis of hip, osteomyelitis. problems are detected in early childhood or late adolescent. Autosomal Recessive is a more severe form of disorder that becomes apparent in early infancy. features of autosomal recessive includes slow growth, short stature, dental abnormalities, hepatosplenomegaly, anaemia, recurrent infections. depending on the genetic changes involved, people with severe osteopetrosis can also have brain abnormalities, intellectual disability or epilepsy. Common findings in osteopetrosis are cranial nerve

compressions manifesting visual & hearing loss, nutritional anaemia, thrombocytopenia, brittle and osteomyelitic bones, hypocalcemic seizures with secondary hyperparathyroidism & muscular hypotonia. judicious use of transfusions is warranted as significant immunosuppression is a feature of this disease. A few individuals have been diagnosed with Intermediate autosomal osteopetrosis which has features of both. some affected individuals have abnormal calcium deposits in brain and a form of kidney disease called renal tubular acidosis. AD osteoporosis affects

about 1 in 20,000 people and AR Osteoporosis affects 1 in 2,50,000 people.

[FIGURE 1]

CASE REPORT:

18 yr old male, known case of osteopetrosis with # Rt. Shaft femur p/f ORIF with DHS .

On examination patient had short stature , short neck, pectus carinatum ,bifid palate,double teeth lining, Obstructive sleep apnea, MPC grading III, on auscultation bilateral crepts were present.

On investigations labs were within normal limits. Radiological investigations, CXR-normal, x-ray spine showed marble bone

appearance/ window within window appearance.PFT within normal limits, 2D echo was normal.Patient was previously operated for #Rt tibia under general anaesthesia at peripheral institute.

Patient was accepted for elective surgery with high risk consent and ICU backup. In operating room all standard monitors were attached. 2 wide bore IV access secured.Regional anaesthesia procedure was explained to patient. Sitting position given,Under all aseptic precautions painting and draping done.

Local anaesthetic given at L3-L4 with Inj. Lignocaine 2% 2CC. Epidural needle inserted

,LOR at 3cm,catheter fixed at 7cm. Spinal anaesthesia given with 25G Quincke™ s needle,Inj Bupivacaine 3cc with Inj Clonidine 15mcg.supine Position given & level achieved till T10 dermatome.operative procedure began after adequate spinal anaesthesia achieved.

[FIGURE 2]

DISCUSSION:

We were able to successfully place central neuraxial block in our case,however this may not be the case always.complications related to regional anaesthesia may occur due anatomical & structural differences of the bone, also

inadverent intraosseous injection of local anaesthetic may occur during regional anaesthesia.

The osteopetrotic bones are difficult to drill and surgical time can escalate.due to brittle bones chances of fractures increases intraop also during anaesthesia or surgical positioning.

This patient also had significant risk of difficult airway due to high arched narrow hard palate,obliterated nasal passage due to congestion,wide lower hyperplastic jaw,restricted TM joint movement and cervical spine fracture also hypognathism and narrowing of

oropharynx may result in upper airway collapse and negative pressure pulmonary edema following extubation.

CONCLUSION:

Osteopetrosis(Marble Bone Disease) is associated with various

challenging anatomical and physiological alterations but with proper knowledge,preoperative identification and planning of anaesthesia complexities we can manage the cases successfully.

Acknowledgement

Conflict of Interest

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