

CASE REPORT

OSTEOPETROSIS – A CHALLENGE IN RARE SITUATION.

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ABSTRACT: BACKGROUND: Osteopetrosis is a collective term used for a pathological condition with defective function of osteoclasts presented with range of sclerosing bone diseases along with skeletal, renal, haematological & neurological manifestation. It may be autosomal recessive (ARO), autosomal dominant (ADO) and X-linked. It presents with hydrocephalus, short stature and anaemia, involvement of ocular nerve or facial nerve. Hypocalcemia, tetany, seizures & secondary hypoparathyroidism is known to occur. **CASE REPORT:** Two patients of osteopetrosis were posted for orthopaedic surgery. Both patients were operated under combined spinal epidural anaesthesia. 2ml of 0.5%Bupivacaine+30mcg clonidine was given intrathecally & epidural supplementation was given with Bupivacaine 3 cc increments after two segment regression of sensory level. Postoperative analgesia was provided by epidural Bupivacaine 0.125% along with Inj. Tramadol 50 mg on patient's request. **CONCLUSION:** Even if administration of anesthesia is a challenge in patients of Osteopetrosis, regional anaesthesia can be given safely with proper preoperative preparation & intraoperative care.

KEY WORDS: osteopetrosis, femur fracture, combined spinal & epidural anaesthesia

INTRODUCTION: Osteopetrosis is a collective term used for a pathological condition with defective function of osteoclasts presented with range of sclerosing bone diseases along with skeletal, renal, haematological & neurological manifestation. It is classified as Infantile malignant or (ARO), Intermediate type or (ARO), Adult onset or (ADO)&X-linked Osteopetrosis.

ARO is a life threatening condition manifests in first few months with life span of 6 to 10 yrs present with seizures with normal Calcium levels, renal tubular acidosis, cerebral calcification, developmental delay, hypotonia, retinal atrophy & sensorineural deafness. ADO is type I and type II. Type I is associated with reduced number & size of osteoclasts & involvement of ocular nerve while type II is associated with proliferation of large & multinucleated osteoclasts, involvement of facial nerve, bony sclerosis, renal tubular acidosis & cerebral calcification. In X-linked Osteopetrosis severe immunodeficiency is observed with ectodermal changes.

The difficulties faced by Anaesthetists are difficult intubation due to facial deformities, head & mandibular involvement cervicomedullary stenosis may lead to cord trauma during intubation difficult spinal & epidural anaesthesia due to scoliosis and short stature. Leucoerythroblastic anaemia, pancytopenia, thrombocytopenia leading to excessive bleeding

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during airway instrumentation or surgery. Hepatosplenomegaly is seen due to secondary expansion of extramedullary haematopoiesis. Decreased myocardial contractility, renal tubular acidosis and mental retardation might be seen in these patients⁸. These patients are having high risk of developing hypocalcemia, tetany, seizures & secondary hypoparathyroidism. Bone marrow suppression with inhibition of medullary haematopoiesis resulting in pancytopenia. Here we want to discuss anaesthetic management of two patients of benign adult onset osteopetrosis posted for orthopaedic surgery.

CASE REPORT: Two patients of osteopetrosis were posted for open reduction & internal fixation of fracture of femur. A 22 yrs male had # left sided shaft femur while a 47 years male had right sided # subtrochantric femur. First patient had previous fracture of tibia whereas another 47yrs male had # subtrochantric femur right. Both patients were short statured height 139 cm & 135cm respectively. Both had frontal bossing, protruding eyes, facial puffiness, small sized head, short neck & inadequate mouth opening. Also both had Pigeon shaped chest, short hands & wide thumb.

Patients were investigated in detail. All blood investigations were normal except in first patient's haemoglobin was 8.2 gm% & borderline cardiomegaly on X-ray chest. Radiographs of long bones revealed diffuse sclerosis with obliterated modularly cavity. X-ray spine was suggestive of mild lumbar scoliosis. The fundus examination of right eye revealed partial optic atrophy with gradual diminution of vision since 6-7 yrs. Audiometry indicated left ear deafness. He had large tongue, high arched palate & irregular dentition. His mouth opening was 3.5 cm & mentohyoid distance was 5.5 cm. In second patient micrognathia, adequate mouth opening along with mentohyoid distance 5cm was observed. In second case family history of osteopetrosis was known with death of younger sister.

Both the patients were given oral antacids Tab Pantoprazol 40mg and inj. Taxim 1gm preoperatively. After obtaining written, valid consent patients were operated under combined spinal epidural anaesthesia. Epidural anaesthesia was given in L₂-L₃ space with 18 numbered needle trough which 18 numbered epidural catheter was put Lumbar puncture was done through same space, 2ml of 0.5% Bupivacaine+30mcg clonidine was given intrathecally. Epidural supplementation was given with Bupivacaine 0.5%, 3 cc increments after two segment regression of sensory level approximately after 95 mints of intrathecal drug administration. We got difficulty for identification of epidural space in first patient. T₈ level was achieved in first patient while T₁₀ level was achieved in second patient. Both patients were haemodynamically stable throughout the procedure & didn't require any vasopressors.

Eyes were protected with eye pads after instillation of natural tears & proper padding was done at pressure points. Postoperative analgesia was provided by epidural Bupivacaine 0.125% along with Inj. Tramadol 50 mg on patient's request. During surgery Ringer's lactate solution 1000ml along with 500 ml Hydroxyethyl starch & 500ml of 5% Dextrose was infused. Second patient required one unit of blood transfusion. The procedure was uneventful in both patients. There was no excessive bleeding intraoperatively but surgeon had difficulty in bone drilling & threading screws.

DISCUSSION: Osteopetrosis is a hereditary disorder involving skeleton and characterised by increased bone density on radiographs also known as Marbel Bone Disease or Osteosclerosis fragilis generalis at^{1,3}. It was diagnosed by German radiologist in 1904 hence named as Albers-Schonberg disease.¹ Multisystemic involvement like anaemia, pancytopenia,

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hepatosplenomegaly, renal tubular acidosis, compressive nerve palsies and mental retardation needs modified anaesthetic technique.^{1,2,3} Facial deformities like proptosis, high arched palate, broad facies, hypertelorism, mandibular hyper or hypoplasia², limited mandibular movements make oral intubation difficult while bony encroachment of nasal turbinates may preclude nasal intubation.^{2,4} Poor periodontal attachment leads to increased risk of intraoperative tooth loss⁴. Thrombocytopenia lead to profuse bleeding during intubation or surgery.² Hypocalcemia may lead to intraoperative seizures or tetany.⁴ Spinal or epidural anaesthesia is difficult due to bony thickening, scoliosis & short stature in these patients. Tendency of repeated fractures needs to take proper care while positioning during surgery & protruding eyes should be protected.

It was reported that incidence of difficult oral intubation is high along with air way obstruction & arterial desaturation⁵. Anesthetic management strategies should consider the factors that cause the high frequency of adverse airway events in this patient population⁶. Awake fibreoptic intubation is necessary in severe cases⁷. we preferred combined spinal epidural anaesthesia as in both patients. We did not have any difficulty in giving combined spinal epidural anaesthesia in one patient while another patient needed multiple pricks. Access of neuroaxial block may be technically difficult due to chronic compression fracture of lumbar or thoracic spine & potential for trauma to abnormal bone must be considered in these patients⁷. Besides these problems there is a possibility of interosseous injection with high serum levels of local anaesthetic solution⁷. So it is advisable to administer titrated doses of local anaesthetic drugs. Regional anaesthesia should be given after correction of thrombocytopenia if present. Peripheral nerve blocks can be given safely but for lower limb high doses of local anaesthetic drug may be needed.

Funnel like appearance known as Erlenmeyer flask deformity,¹ extremely radio dense vertebrae with alternate bands known as “rugger-jersey” sign are diagnostic of osteopetrosis³ which was seen in our patient. Bone within bone appearance “Endobone” is due to defect in metaphyseal bone remodelling resulting in greatly thickened cortices & obliteration of medullary space⁸.

Neurological effects of osteopetrosis results from restricted growth of foramina leading to Optic, Facial & Trigeminal neuropathies, strabismus, hearing loss, dysarthria, hydrocephalus, cerebral atrophy & developmental delay.⁹ Short stature might be due to impaired longitudinal growth. Both patients were belong to intermediate type osteopetrosis so there was no evidence of hepatosplenomegaly, anaemia, renal tubular acidosis & pancytopenia. There was evidence of involvement of Abducent nerve & Optic nerve in one patient without raised intracranial pressure in one patient.

Deficiency of carbonic unhydrase in osteoclasts causes defective hydrogen ion pumping & hence defective bone resorption results. Carbonic anhydrase II has activity in bone, kidney and brain. These Patients may present with slightly acidic blood with high chloride concentration. It is due to excessive leakage of bicarbonates from renal tubules.¹⁰

Infantile osteopetrosis is severe form of disease presents in early childhood. The life threatening complication is bone marrow failure leading to anaemia, hepatosplenomegaly, pancytopenia with increased susceptibility to infection^{1, 2,3} Benign type of osteopetrosis is diagnosed in late adolescent or adulthood. Bone marrow function is not compromised in these

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patients.³ Steroids, high-dose calcitriol, or interferon-gamma therapy may be beneficial, and bone marrow transplantation has been advocated.

CONCLUSION: For successful management of patient anaesthesiologist must be aware of etiopathogenesis of osteopetrosis & prepared for difficult intubation & regional block. Preoperative correction of anaemia & thrombocytopenia, proper positioning & protection of eyes is must. Regional anaesthesia can be given safely in milder osteopetrosis without difficulty.



Photograph showing abnormal facies



Photograph showing short hands

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Photograph showing abnormal Facies



Radiograph showing funnel shaped femur

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