

### Osteopetrosis a significant bone disease – A Case Study

#### Abstract

**Background:** Osteopetrosis is a genetic condition that affects just a few people. illness characterised by osteosclerosis, Due to inadequate osteoclast resorption, there is destruction of the medullary canal, calcified cartilage, and hard bone. Patients with osteopetrosis are more likely to develop osteoarthritis. Only a few studies have documented a patient who underwent both a complete Arthroplasty of the hip and knee (TKA). In this article, a patient with osteopetrosis describes a case of left and right hip osteoarthritis.

**Clinical Findings:** - walking problems, Calcium deficiency, the optic nerve in the brain is under pressure. Hearing loss, as well as distress, can lead to neurological issues.

**Diagnostic Evaluation:** - CBC Investigation, blood test, Hb-12.9%, Total RBC count-4.84million/cumm, RDW-13.2%, HCT-36.7%, Total WBC Count-26200 cu mm, Monocytes-01%, Granulocytes- 30%, Lymphocytes- 65%, Total platelet count-3.5 Lacs/cu mm, ALT(SGPT)-17 U/L, AST (SGOT)-44 U/L.

**Peripheral Smear:** - RBCs-Normocytic Normochromic Platelets-Adequate on smear, No Hemiparasite seen, WBCs-Neutrophilic leucocytosis.

**Therapeutic Intervention:** - Calcium 1200mg, D3 8000IU, vitamin D supplements calcitriol 0.25mcg/ml, hormone replacement therapy, Bone marrow transplantation (BMT), Alendronate70mg, (Fosamax), Risedronate35mg OD (Actonel), Inj Boniva 3mg OD, Inj Zoledronic acid 4mg BD (Recast), various medications, and/or surgery.

**Outcome:** - After therapy, the child's behaviour improves. Osteoporosis is a condition in which excessive bone resorption causes decreased bone mass and skeletal fragility, with the most serious clinical consequence being increased fracture risk.

**Conclusion:** - The patient was hospitalised to AVBRH with a known case of Osteopetrosis illness and complained of difficulties awakening, discomfort, and neurological issues. After receiving appropriate therapy and surgery, his condition improved. According to this case study, in patients with osteopetrosis, total joint arthroplasty is an effective therapy for painful hip and knee osteoarthritis.

**Keywords:** -Total hip arthroplasty (THA), Total knee arthroplasty (TKA), Osteopetrosis.

#### Introduction: -

It is a rare hereditary illness characterised by osteosclerosis, Insufficient osteoclast resorption causes medullary canal obliteration, petrified cartilage, and stiff bone. People with osteopetrosis are more likely to develop osteoarthritis. For individuals suffering from Total joint arthroplasty is the therapy of choice for pain and dysfunction, then it is fraught with intraoperative and postoperative problems. Only a few studies have documented a patient who underwent both a complete hip and A total knee replacement is a surgical procedure that replaces the whole knee

joint (TKA). In this article, an osteopetrosis patient presents a situation of left hip and right knee osteoarthritis.<sup>1</sup> THA and TKA were used on a Osteopetrosis patient, 59 years old, in the left hip and right knee is excruciating.

Osteopetrosis is a infrequent genetic sickness originally designated in 1904 by German radiologist Albert Schonberg. Reduced osteoclast activity causes osteosclerosis, medullary cavity obliteration, calcified cartilage, and brittle bone are all symptoms of osteosclerosis. There are three primary kinds of osteopetrosis: There are three types of malignancies in children: malignant, intermediate, and benign.<sup>2</sup> The intermediate autosomal recessive type has an adult life expectancy. Because of the reduced bone vascularity and insufficient white blood cells, this kind has the greatest risk of jaw osteomyelitis. It has a long-life hope disdain an enlarged risk of cracks and other musculoskeletal disorders such as hip and knee osteoarthritis. THA and TKA are still viable options for osteoporotic individuals suffering from painful osteoarthritis. Few research on arthroplasty in individuals with osteopetrosis have been published too far. This is the first time THA and TKA have been described in a patient with osteopetrosis that we are aware of. Patient demonstrated great clinical performance and was happy through the medical product after more than a year of follow-up.<sup>3</sup>

**Case presentation:** For previous 13 years, this 59-year-old female with osteopetrosis has been suffering left hip discomfort and bilateral, right greater-than-left, knee pain, as well as increasing activity limitation. She had never had a prior fracture. The x-rays indicated severe osteoarthritis in the left hip, as well as acetabular erosion, proximal femoral head displacement, and decreased joint interstellar. X-rays of the right knee revealed comparable osteoarthritic changes. THA in the left hip and TKA in the right knee were agreed upon by the patient. Preoperative planning and preparation were meticulous. In the first instance, a total left hip arthroplasty was done. A posterolateral approach was used to execute the THA procedure. There were significant degenerative alterations in the femoral leg, but no indications of downfall or avascular caput necrosis. Before placing the femoral component, the acetabulum was visible and prepped (Zimmer, USA).

**Patient Identification:** - A 59-year-old woman from Burtipura, Gondia, was brought to AVBRH on June 29, 2021, with a recognised case of Osteopetrosis illness. Her weight is 50 kilogrammes and her height is 159 centimetres.

**Present Medical History:** - His parents admitted a 59-year-old woman to AVBRH on June 29, 2021, with a complaint of difficulty walking. Calcium shortage, as well as strain on the brain's optic nerve There's a relationship between hearing loss, pain, and neurological problems. I was weak and inactive when I was admitted.

**Past medical history:** - In My patient was diagnosed with Osteopetrosis and was admitted to the hospital because she was in pain. Her left leg was X-rayed because she had no previous medical history.

**Present Surgical History:** - Patient do not have any present medical surgical history.

**Past Intervention and Outcome:** - My patient was diagnosed with Osteopetrosis and presented to the hospital with left leg pain, difficulties walking, and an X-ray of the left leg. He was diagnosed with Osteopetrosis Disease after that. Then, from time to time, it was proved to be helpful for future sickness treatment because the patient did not develop problems until then.

**Clinical Finding:** - The optic nerve in the brain is under strain due to calcium shortage, which is a stumbling block. Hearing loss, along with pain, can cause neurological problems.

**Ethology:** - Osteoporosis is a bone disease that develops when the body loses an excessive amount of bone or does not generate enough bone. As a result, bones become brittle and can fracture as a result of a in severe situations, a tumble, sneezing, or small bumps may occur. "Osteoporosis" means "porous bone."

**Physical Examination:** - There aren't many abnormalities discovered in a head-to-toe examination, except for abdominal distention. The youngster is then and has a dull appearance, he's weak and uncooperative, and he's restless and weary.

**Diagnosis Assessment:** - CBC Invesgation, Blood test, Hb -12gm%, Total RBC Count - 4.84million /cu mm, RDW-13.2%, HCT-36.7%, Total WBC Cont-26200 /cu mm, Monocytes-01%, Granulocytes-30%, Lymphocytes-65 %, AST (SGOT)-44U/L, ALT (SGPT)-17, Alkaline phosphate-239 U/L, Total protein-6.7 g/dl, Albumin-3.7 g/dl, Globulin-3.0gm/dl, Total bilirubin-0.3 mg / dl, Bilirubin Conjugated-0.1 mg / dl, Bilirubin Unconjugated-0.2 mg /dl, Platelet Count 3.5 micro litter, MCV -75.8%, MCH -26.7 %, MCHC -35.1%. Eosinophil -01%, Basophil 0%, KFT-Urea 18%, Creatinine – 03 %, Sodium – 142%, Potassium-4.8%,

**Peripheral smear:** - RBCs-Normocytic Normochromic Platelets-Adequate on smear, No Hemiparasite seen, WBCs-Neutrophilic leucocytosis.

**Therapeutic Intervention:** - Calcium 1200mg, D3 8000IU, vitamin D supplements calcitriol 0.25mcg/ml, hormone replacement therapy, Bone marrow transplantation (BMT), Alendronate70mg, (Fosamax), Risedronate35mg OD (Actonel), Inj Boniva 3mg OD, Inj Zoledronic acid 4mg BD (Recast), various medications, and/or surgery.

**Radiological Investigation:** - Dual energy x-ray absorptiometry (hip and spine) is a quantitative approach for verifying osteoporosis diagnosis. Metabolic bone disorders impact bone as a tissue; histologically, all bones are implicated, although radiologic characteristics are not always present.

**X-ray:** - When an X-ray of my patient's left leg is taken, there aren't many abnormalities discovered. Leg anomalies are seen. When an X-ray of my patient's left leg is taken, there aren't many abnormalities discovered. Leg anomalies are seen.

**Discussion:** -

For the time being, there is no cure for benign osteopetrosis. Treatment is primarily focused on symptomatic treatment of problems and is hence supportive. Nonsteroidal anti-inflammatory medications can be used to treat symptomatic joint osteoarthritis in this patient population, However, in some situations, complete joint replacement is required. Preoperative planning and preparation are critical for effective THA and TKA in osteopetrosis patients. The small and occasionally absent Intramedullary reaming can be difficult due to the medullary canal, and iatrogenic fractures are prevalent with this procedure. Due to tiny surfaces and dense bone, component size may be problematic, and cement-bone interdigitation may be difficult.<sup>3</sup> Finally, because of the inadequate Because of the concentrated body fluid stream to the bone and qualified neutropenia, these individuals are at a greater risk of osteomyelitis. The greater trochanter was injured in our case when constructing the proximal femur's very hard bone has a medullary canal. This fracture also wanted to be repaired, and the patient was recommended to uniform a knee brace for a week. During surgery, overheating of the bone was also an issue, which we handled by carefully drilling at a slow speed while constantly injecting cold saline to the bone.<sup>4</sup> Osteoporosis can be avoided by eating a healthy diet as a kid and avoiding medicines that accelerate bone loss. A healthy diet, exercise, and fall avoidance are all helpful in preventing fractured bones in people with osteoporosis Changing one's lifestyle, such as quitting smoking and abstaining from alcohol,

may be beneficial. Bisphosphonate medicines can help those who have previously fractured bones due to osteoporosis from breaking them again. They are less effective in those who have osteoporosis but have never had a bone fracture. They don't seem to have an effect on the chances of dying. Osteoporosis is becoming more common as individuals get older. Around 15% of Caucasians in their 50s and 70% of those over the age of 80 suffer from the disease. Women are more prone than men to be affected by this disease. Depending on the technique of diagnosis, 2% to 8% of men and 9% to 38% of women in the industrialised world are affected.<sup>5-6</sup> Few of the concerned studies and articles were reviewed<sup>7-16</sup>.

### **Conclusions: -**

According to our case study, for osteopetrosis-related hip and knee osteoarthritis, Total joint arthroplasty is a viable option for therapy. This situation demonstrates the practical challenges and problems related in osteopetrosis patients; complete joint arthroplasty is used. Sclerotic bone, a damaged medullary canal, and perioperative fractures all contributed to the patient's discomfort. The operating time is considerably increased. To get good results, careful planning, the proper tools, and expertise are required.

### **COMPETING INTERESTS DISCLAIMER:**

Authors have declared that no competing interests exist. The products used for this research are commonly and predominantly use products in our area of research and country. There is absolutely no conflict of interest between the authors and producers of the products because we do not intend to use these products as an avenue for any litigation but for the advancement of knowledge. Also, the research was not funded by the producing company rather it was funded by personal efforts of the authors.

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