Arthroplasty in patients with rare conditions

Total knee arthroplasty in ochronosis

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ABSTRACT

Alkaptonuria is disorder of tyrosine metabolism due to deficiency of homogentisic oxidase characterized by excretion of homogentisic acid in urine, deposition of oxidized homogensitate pigments in connective tissues and articular cartilages (ochronosis). The result is dark pigmentation and weakening of the tissues resulting in chronic inflammation and osteoarthritis. Management of alkaptonuric ochronotic osteoarthritis is usually symptomatic and replacements have comparable outcomes to osteoarthritis in patients without ochronosis. I report a case of a patient with ochronosis of knee treated with total knee replacement and report operative pearls for surgery in this rare disorder.

Introduction

Alkaptonuria is an autosomal recessive disorder caused by the deficiency of homogentisate 1,2 dioxygenase activity (Fig. 1). It was first described by Virchow in 1866 [10]. Alkaptonuria is disorder of tyrosine metabolism due to deficiency of homogentisic oxidase characterized by excretion of homogentisic acid in urine, deposition of oxidized homogensitate pigments in connective tissues, the dermis, apocrine glands and articular cartilages (ochronosis). The pathogenesis of the disease is the polymerization of deposited HGA that discors and weakens the connective tissue, ultimately resulting in brittle tissue that is easily disrupted and leads to chronic inflammation, degeneration, and eventually osteoarthritis [1]. Patients with alkaptonuria are usually asymptomatic and arthropathy appears after the fourth decade [2].

Systemically, there is thickening and blue—black or gray—blue discoloration of ear cartilage. Other body locations include the eyelids, sclera, foreheads, cheeks, axillae, genitals, nail beds, buccal mucosa, larynx, tympanic eardrum and nasal tip. Simultaneously, this discoloration can occur in skin, tendons, ligaments, costochondral junctions, sclera, heart valves, the intima of blood vessels and cause lumbar intervertebral disc calcification and disc space narrowing [3–5].

The knee is the most common site of orthopedic abnormality. Other sites of involvement are hips, shoulders, sacroiliac joints and the pubic symphysis [6]. There is currently no definitive cure for alkaptonuric ochronosis. However, total joint replacement in published cases of ochronotic osteoarthritis report good results similar to osteoarthritic patients without ochronosis [7] I report the case of a 58-year-old female with a family history of ochronosis, who developed degenerative arthritis of the knee.

Case history

A 58-year old female patient was admitted with history of bilateral knee pain for 8 years. Pain was of gradual onset, interfering her day to day activities, more on right side and was not responding to conservative treatment. No cutaneous signs of ochronosis were noted at time of presentation. The patient had a family history of ochronosis in her mother and 2 sisters. The patient had history of bilateral knee pain for 8 years. Pain was of gradual onset, interfering her day to day activities, more on right side and was not responding to conservative treatment. No cutaneous signs of ochronosis were noted at time of presentation. The patient had a family history of ochronosis in her mother and 2 sisters. The patient had history of darkening of urine which was revealed by patient after surgery. On local examination of right knee, there was no swelling. On deep palpation, there was medial and lateral joint line tenderness. The range of motion of the right knee was 0°—95° and left knee was 0°—105° with significant pain. The patient walked with an antalgic gait. Clinical diagnosis of osteoarthritis was confirmed with anteroposterior and lateral radiographs demonstrating joint spaces narrowing.

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narrowing, peripheral new bone formation, subchondral bone sclerosis and arthritic changes of patella (Fig. 2). Cemented total knee replacement was offered after informed consent. During surgery, an anterior midline incision was used. The subcutaneous tissues revealed black nodules (Figs. 3 and 4). There was synovial hypertrophy. The joint capsule was contracted and partially black with a hard consistency. It was difficult to retract the patella as patellar tendon was stiff and there was a popping sound during exposure. No obvious extensor mechanism dissociation was found. On capsular incision, I observed no joint fluid, blackened articular cartilage of femur, tibia and patella (Fig. 5) along with blackened undersurface of patella (Fig. 6) and quadriceps tendons. After total synovectomy, I noted dark black stiff menisci inside the joint (Fig. 7). A 5 × 3 × 2 mm cavitary defect was found in tibia which was ultimately filled by cement. Multiple drill holes were made on the raw surface of femur and tibia before cementing. Standard cementing technique was performed without difficulty (Fig. 8).

There was excessive bleeding during surgery interfering the operative field even after tourniquet control (300 mm Hg). There were no other major complications occurred during the surgery. The patient had increased blood loss postoperatively, which was totaled 1150 ml. The drain was removed on 4th postoperative day, which was atypical, but left in place because of blood loss. Patient had mild postoperative pain. Her operated joint was mobilized on second post-operative day. Post-operative wound healing was uneventful. Patient was very happy and satisfied with improved R.O.M. (0–110) and painless knee. The knee society score was 84 on last follow up 18 months post-operatively.

Discussion

Alkaptonuria is disorder of tyrosine metabolism due to deficiency of homogentisic oxidase characterized by excretion of homogentisic acid in urine, deposition of oxidized homogensitate pigments in connective tissues and articular cartilages (ochronosis). There is no definitive cure for the disease but Nitisinone, an inhibitor of 4-hydroxyphenyl pyruvate dioxygenase has been shown to decrease urinary excretion of homogentisic acid [11]. The effectiveness of Nitisinone in treating ochronosis is unknown.
Patients presented with joint involvement have joint replacement as the final treatment option as replacement has a very good outcome as far as functional outcome is concerned [11]. Patients with alkaptonuria are usually asymptomatic and arthropathy appears after the fourth decade [2]. Ochronotic arthropathy is often diagnosed at surgery and may not be suspected until a blackened joint is found at surgery [8,9]. Early diagnosis of ochronosis is valuable to orthopedic surgeons to avoid tendon ruptures and preoperative cardiac clearance is also important because of the risk of valve calcification [11].

In the present case the diagnosis of ochronosis was not made until exploration of the joint and confirmed after taking complete family history retrospectively after surgery. So high suspicion is required for preoperative diagnosis. Ochronosis mainly affects the cartilage and does not affect the subchondral bone. In this case, the patellar tendon had mild black discoloration and while retracting the patella a popping sound was noted which can be a sign of tearing or detachment; fortunately I did not find such. Tendon can be friable and stiff due to ochronotic involvement and one should be careful while retracting patella. We need more such case presentations to prove real incidence of such involvement. Subchondral bone was normal in appearance and free of pigments but while taking cuts it was observed that it was of soft consistency (extra soft as compared to softness of osteoporotic bone as per tactile sensations while taking cuts). The reason behind altered consistency could not be found and it is not mentioned in prior case reports. As there is also lack of data suggesting choice between cemented or non-cemented prosthesis, we chose to drill holes in cut surfaces of the femur and tibia before cementing as a part or our routine procedure. Cement was used because of soft bone. Increased intraoperative and postoperative blood loss was found in earlier case reports. This may be due to synovectomy of the hypertrophied synovium which was more than what we usually observe in other common causes of arthritis. Our protocol is to remove drain on second postoperative day. But due to increased intra and early postoperative blood loss, drain was kept up to forth postoperative day. One could consider tranexamic acid to decrease blood loss.

Current controversies and future considerations

Future considerations include possible genetic interventions or the use of Nitisinone for treatment of this disorder. Blood management, use of bone cement, and spontaneous quadriceps rupture [12], are all possible considerations that might enhance surgical outcomes. Further research in these areas may be helpful.

Summary

Ochronosis is a musculoskeletal manifestation of alkaptonuria caused by the polymerization of deposited HGA, that discolors and weakens connective tissue [1]. Management of alkaptonuric ochronotic osteoarthritis is usually symptomatic and replacements have comparable outcomes to osteoarthritis in patients without
ochronosis. I report a case of a patient with ochronosis of knee treated with total knee replacement and report operative pearls for surgery in this rare disorder.

KEY POINTS

- A high suspicion and good history taking is required for pre-operative diagnosis. Cutaneous discoloration of the ear lobes is common.
- Consider preoperative cardiac clearance because of possibility of calcified cardiac valve disease [11].
- Be careful while retracting the patella, quadriceps rupture has been reported [12].
- Be careful while making cuts and consider use of cement because of softness of bones.
- Excess blood loss intra and postoperatively may occur. Consider tranexamic acid to decrease blood loss.

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