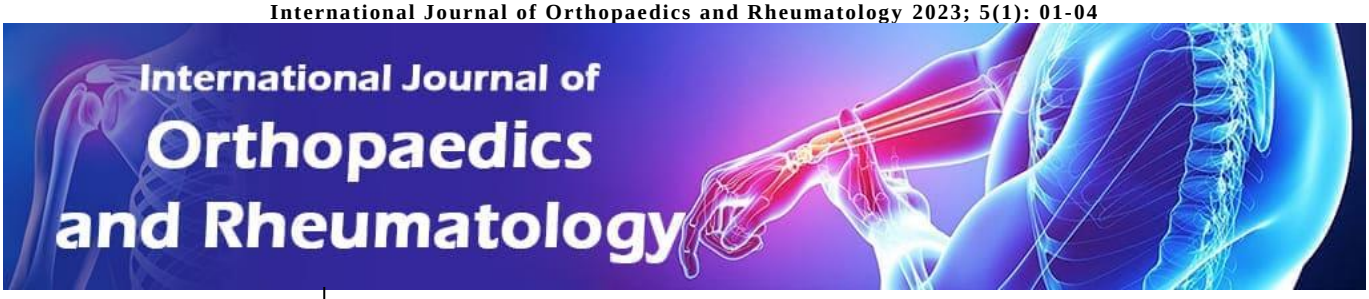

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Alkaptonuria presenting as osteoarthritis in elderly female: A case report

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Abstract
Background: Alkaptonuria is an autosomal recessive trait that deals with dysfunctional catabolism of tyrosine. It is a very rare disorder presenting in early childhood but ignored and progresses later in adulthood to present similar to any arthritis. One specific feature of alkaptonuria is the accumulation homogentisic acid in connective tissue of the body. It is diagnosed as discoloration of urine in childhood and as osteoarthritis complicated by ochronosis in adulthood. The only identified treatment is symptomatic relief along with supplements. The main stay of treatment is to prevent complications.
Case: This case report represents an elderly female who was diagnosed with osteoarthritis at 40 years of age. The patient had multiple complications of osteoarthritis leading to the multiple replacements of her joints over the years. Patient was found with ochronosis on one of the previous surgeries but diagnosis was not changed. Later, prominent ochronosis was found on her right knee, on ear pinna, and on her sclera. Patient was put on symptomatic relief medication and referred to a rheumatologist. All in all, it is important to go through proper investigations and further look into the diagnosis done carefully.
Conclusion: Alkaptonuria is a rare disease that is autosomally inherited in recessive manner and is hardly distinguishable from osteoarthritis. It is a combination between orthopedic and rheumatologist identification. Mainly charectarized by accumulation of the acid that causes bluish discoloration, alkaptonuria can be only treated through symptomatic relief. As a nutshell, it should be highly considered for proper diagnosis on presentation to prevent harmful complications.

Keywords: Alkaptonuria, ochronosis, Osteoarthritis, rheumatology, case report

Introduction
Alkaptonuria is an autosomal recessive trait on chromosome 3q21-23 caused by the deficiency in homogentisic acid 1, 2 dioxygenases (HGD), an important enzyme in the pathway of catabolism of tyrosine [1]. The prevalence of the disease is 1:250,000-1,000,000 cases. Only 950 cases were reported throughout 40 countries. It is characterized by an increase in the deposition of homogentisic acid in the connective tissues of the body resulting in ochronosis; a bluish-black discoloration of the connective tissues, especially in the articular surfaces of big joints. Moreover, the excess homogentisic acid in the urine causes it to turn dark in color [1, 2].
In pediatric age group the diagnosis is missed. By the third decade, patients present with symptoms mimicking osteoarthritis, and ankylosing spondylitis which later progress to be complicated with ochronosis [1, 3]. Clinical presentation of Alkaptonuria is sufficient for diagnosis. Homogentisic acid is increased in the accumulated fluids and tissue [4].
The only identified treatment up till now remains to be symptom relief. The approach is the main stay to prevent further complications and multiple surgeries. Vitamin supplementation could be added to along with vitamin D to improve the quality of the bone damaged and preserved [5]. This case report explains a case of a female who had multiple surgeries for osteoarthritis to finally be diagnosed with alkaptonuria.

Case report
A 72-year-old female nonsmoker, known to have osteoarthritis, osteoporosis and rheumatoid arthritis admitted to our hospital for elective right total knee replacement for severe knee osteoarthritis.

The history goes back to 30 years ago when the patient was diagnosed with rheumatoid arthritis and severe osteoporosis at an early age (40 years). Following the diagnosis, the patient was maintained on anti-inflammatory and vitamin D with Calcium supplements only.

At the age of 52, the patient underwent left total hip replacement for severe left hip osteoarthritis. Following her surgery 3 years later, the patient was readmitted to our hospital for right total hip replacement secondary to severe right hip osteoarthritis. At that time, the patient was also suffering from intermittent bilateral knee pain, and lower back pain. During the investigations, radiological report showed severe degenerative osteoarthritis of the right hip, narrowing of the joint space, and severe L3, 4, 5-disc space narrowing. (Fig. 1)

Subsequent to her right total hip replacement, the operative note with the gross and the histological pathology report revealed femoral head deformity and edge discoloration but the diagnosis remained as severe osteoarthritis.

In 2019, the patient was readmitted to the ward for elective left total knee replacement for severe knee osteoarthritis diagnosed by X-ray. During the surgery, we were surprised by the bluish-black discoloration of the quadriceps tendon and the patellar tendon along with the articular surfaces of the femur, tibia and patellae, and the surrounding soft tissue. The patellar tendon was very fragile causing worrisome about a possible avulsion. (Fig. 2)

Following the operation, the patient was examined physically and found to have a bluish black discoloration of the sclera and the cartilage of the ear. (Fig. 3 and 4)

The patient was diagnosed with Alkaptonuria based on her past medical history and present complaint and findings intra-operatively. She was referred to a rheumatologist and prescribed symptomatic treatment to prevent further complication.

Discussion

Although osteoarthritis typically presents with stiffness, instability and radiographic changes, it can also present with symptoms that are non-specific [6]. It is previously mentioned that Alkaptonuria can be mistaken for osteoarthritis if no ochronosis was found [3]. The most common degenerative disease affecting more than 25% of the population is osteoarthritis, whereas alkaptonuria is a rare genetic disorder that has been reported in less than 1000 cases till now [1, 6]. Our 72-year-old patient is presented with a previous diagnosis of osteoarthritis at the age of 40.

The knee is most frequently affected by articular surface disease as osteoarthritis and ochronosis due to the repetitive stress it is exposed to and the load it carries [8]. The patient of our case had previous history of repair surgeries of the hip and knee and was currently presenting for right knee replacement due to osteoarthritis.

Aromatic acids as Tyrosine and Phenylalanine, contained in foods and metabolized by our body, are important in the synthesis of multiple hormones. Any defect in specific step of the catabolism of tyrosine will result in specific genetic disorders [1]. The metabolic enzyme system of tyrosine of the cartilage is inhibited in Alkaptonuria by the deposition of the pigments that acts like a chemical irritant. This chemical irritation results in fragility and disintegration of the structure affected [9], thus leading to similar clinical symptoms with other diseases. Ochronosis is a bluish-black discoloration of the connective tissues, especially in the articular surfaces of big joints such as the hips and knees resulting from the accumulation of homogentisic acid [9]. The patient of our case was diagnosed for osteoarthritis due to the recurrent presentation of diffuse pain in joints (knee and hip) with restricted movement and stiffness. During her hip replacement, incidental pigmentation on the joint was discovered with irregularity of femoral head surface indicating to onchronosis but no further investigations were done. Within her current surgery the report along with the macroscopic appearance clearly showed bluish-black pigmentation on the edges of the cartilage.

Deposition in other areas of the body might be involved such as the skin, ears, eyes and heart [5]. Noting that after the surgery a complete proper physical exam was done revealing pigmentation on other sites. Bluish sclera and black ear pigmentation were significant to find. The scleral deposition may be one of the 4 different types [10].

Clinical presentation of alkaptonuria is sufficient for the diagnosis in addition to the finding of homogentisic acid accumulated in urine. To confirm the disease gas liquid chromatography on urine is done [4]. In the present case, no further urinary analysis or tests were done to confirm the diagnosis based on lab with our patient.

The patient was then referred to a rheumatologist for further investigations. She was also put on non-steroidal analgesic drug's as analgesics and other symptomatic treatment to prevent any upcoming complication. Research found that alkaptonuria is a disease to be managed symptomatically [5]. Additional supplements might help increase bone density and maintain adequate well-being for the patient. Ascorbic acid was found to have an antioxidant effect that helps in decreasing the deposition in cartilaginous tissue [10].

Makassed General Hospital
Makassed General Hospital
[redacted] Department

In-Patient Discharge Summary
Results Report

Date 29/12/2004 Service جنوبي اول
Patient ID 221874 1 Room 156 A
Name
Physician د.زياد الحاج

Date	Code	Examination Requested
26/12/04	X05103	HIP AP.LAT.2 FILMS

RIGHT HIP:

Severe degenerative osteoarthritis of the right hip with severe narrowing of the joint space.

Left total hip prosthesis.

Severe L3-4-5 disk space narrowing.

X ray no 37521.

KT/dm Kh.Tayyarah,M.D.

Fig 1: X-ray result of previous surgery

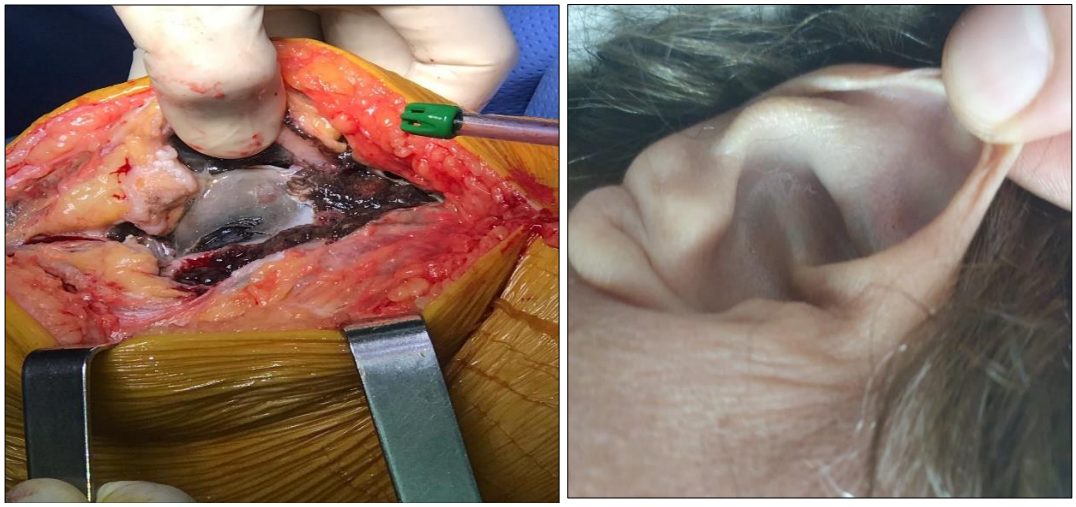

Fig 2: Knee and ear pinna cartilage with discoloration

Conclusion

Alkaptunoria is a rare disease rheumatological in origin. Presence of alkaptonuria is mainly misdiagnosed by osteoarthritis. The proper clinical evaluation should be done earlier for better outcomes. Such case report opens doors for trials and series to dig and know more about this disease.

Abbreviations

1. Homogentisic acid 1, 2 dioxygenases (HGD).

Ethics approval and consent to participate

The participant has agreed to participate verbally and no IRB approval was needed. The case is totally anonymous from any data containing patient's details and all data were kept locked in the authors computer.

Consenting

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal

Availability of data

Availability of all supported data are present

Conflict of interest

No conflict of interests found

Funding's

No funding's were given

Author's contributions

All others of the previous case report contributed equally in collecting information, literature review, writing, and following up this case report

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