Ochronosis: A Rare Cause of Degenerative Arthropathy

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INTRODUCTION:

Ochronosis is a rare metabolic disorder associated with the accumulation of homogentisic acid due to alkaptonuria (a defect in the enzyme homogentisate 1,2-dioxygenase).

The pigment deposits in connective tissues, especially cartilage, causing them to become brittle and discolored (bluish-black), making it weak and prone to rupture, causing rapid joint degeneration.

It is established that joint compromise begins its manifestation after the fourth decade of life, beginning with pain and decreased mobility, with the knee becoming the most affected.

This relatively infrecuent autosomal recessive condition has a higher incidence in certain populations, like the Dominican Republic and Slovakia (1:19,000)

METHODS: case presentation

RESULTS:

1st Case

A 72-year old male patient, with a known morbid history of managed hypertension and DM II, who attended at the health center after presenting pain and functional limitation in the left knee of 5 years of evolution, without improvement of the condition.

2nd Case

A 67-year old female patient, with a known morbid history of managed hypertension, hysterectomy and ORIF left hip.

Attended the health center after presenting pain and functional limitation in the left knee of 2 years of evolution, without improvement of the condition.

Patient refers arthroscopic procedure, but no other details (other facility).

Physical examination reveals changes in skin coloration inboth lower limbs, without significant axial deviations in them.

DISCUSSION AND CONCLUSION:

Ochronotic arthropathy often mimics osteoarthritis but can be more aggressive. Diagnosisis often not made until surgery.

No curative medical treatment; symptomatic management and surgery are mainstays. A diet with protein restriction and rich in ascorbic acid is recommended to prevent oxidation.

Ultimately, the progression of the disease will lead to a joint replacement.