Ochronotic Arthropathy: Total Joint Arthroplasty in Alkaptonuria – A Case Study

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First described by Sir Achibald Garrod in 1908, alkaptonuria is an inherited genetic disorder of metabolism which is characterized by the absence of the enzyme homogentisic acid oxidase resulting in the accumulation of homogentisic acid. It is a rare autosomal recessive disorder resulting from two copies of the defective HGD gene seen more commonly in certain populations of Slovakia and the Dominican Republic. Ochronosis is the dark pigmentation of connective tissues due to the accumulation of homogentisic acid. Joint abnormalities occur due to chronic inflammation, degenerative changes and eventually early osteoarthritis with severe cartilage destruction causing significant morbidity.

We present a case study of a 67 years old Malaysian male with alkaptonuria who had presented with a history of longstanding neck pain, bilateral shoulder pain, bilateral hip and knee pain. Over the course of the patient’s follow up with us for 4 years, cemen ted bilateral hip and knee replacements were performed separately at different timings. Findings of black stained articular surfaces, adjacent tendons and ligaments were observed intraoperatively. Post operatively in the recovery period patient presented with blisters over sites of plaster adhesions otherwise had an unremarkable recovery. In the post-operative follow up period patient is ambulating pain free with good motion of the hip and knee joints.

In view of current therapeutic approaches to alkaptonuria has been relatively unsuccessful and are palliative, the early presentation and severity of arthritides in patients with ochronotic arthropathy sometimes necessitates arthroplasty. This report describes a case of ochronotic bilateral hip and knee treated with total joint arthroplasty with excellent outcomes.

REFERENCES: