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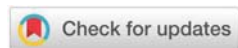
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Case Report

Large Prostatic Calculus in an Alkaptonuria patient – Caught unaware

Abstract

Alkaptonuria, a metabolic disorder with urological manifestation goes unnoticed till we are aware of it. Here we present an alkaptonuric patient with difficult catheterisation was diagnosed with a large prostatic calculus and his management.

Introduction

Alkaptonuria is a rare inherited metabolic disorder involving the serum homogentisic acid levels. Most of the people affected have varied presentations. Very rarely genitourinary system is involved. Prostatic calculi and black coloured urine are the urological presentations of this condition.

Case Report

55 years old male a known case of Alkaptonuria, recently underwent left hip replacement surgery. He developed urinary retention just after surgery. Per urethral catheterisation was attempted but could not be passed and hence cystoscopy was done. To our surprise cystoscopy showed a calculus in prostatic fossa that could not be pushed back into bladder and suprapubic catheterisation (SPC) was done. Xray Pelvis revealed speckled calcification behind pubic symphysis. Cystolitholapaxy was planned electively (Figure 1). Cystoscopy after 3 weeks revealed large prostatic urethral calculus of size of 2.5cms. As the stone could not be pushed into urinary bladder, it was fragmented with laser energy in prostatic fossa into small fragments. After 120 minutes of fragmentation, the fragments were pushed into bladder and washed out. Patient had a stable post operative period. Stone fragments were analysed which showed bilirubin and cholesterol. His urine was positive for alkaptonuria. Xray spine revealed multilevel intervertebral disc calcification with severe narrowing of the intervertebral disc spaces and associated anterior osteophytes along the lumbar vertebrae. His catheters were removed on POD 5 and was voiding with Qmax of 15ml/sec.



Figure 1: A. Xray Pelvis, B. Stone fragmentation with laser, C. Stone fragments, D. Post operative cysto-urethrogram.

Discussion

Alkaptonuria is a rare metabolic disorder first described by Sir Archibald Edward Garrod; who first used the expression, ‘inborn errors of metabolism’, to describe four rare disorders - alkaptonuria, albinism, cystinuria and pentosuria. He discovered that such disorders resulted from enzymatic defects in the catabolic pathways of aminoacids and sugars [1,2]. Autosomal recessive mutations of the homogentisic acid (HGA)oxidase gene, located on human chromosome 3q 21-q 23, results in a defect in the metabolism of homogentisic acid [2]. There is accumulation of homogentisic acid in patients with alkaptonuria. Oxidation of this acid leads to formation of a blackish melanin like pigment which is selectively deposited