CASE REPORT

A CASE REPORT OF MULTIPLE STONES IN RENAL, URINARY BLADDER & PROSTETIC URETHRA IN PATIENT OF ALKEPTONURIA
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ABSTRACT
A 45 y old Indian male patient developed multiple stone in renal pelvis and prostetic urethra due to alkeptonuria. X-ray and Ultrasoundogy suggest the stone in renal pelvis and cystoscopy suggest multiple stone in prostatic urethra. The stone may be due to deficiency of homogentisate oxidase and it leads to deposition of homogentisic acid in renal pelvis and prostetic urethra. The stone was removed by pyelolithotomy and cystoscopy with lithotripsy and patient was improved and discharged.

Keywords: Alkeptonuria, homogentisate oxidase, renal pelvis, prostaticurethra

INTRODUCTION
Alkaptonuria is a rare metabolic condition of autosomal recessive inheritance. It was the first disorder found in humans to attest the principles of Mendelian autosomal recessive inheritance 1. In this disorder, there is congenital deficiency of homogentisate oxidase 2. So homogentisate polymers are accumulated in various tissues causing black melanin pigmentation of connective tissues resulting in multiple joint arthritis, termed as ochronosis, cola color urine, eye and skin pigmentation, kidney, bladder, prostatic stones, cardiovascular disease like cardiac valve defects 3. Current incidence of alkaptonuria is 1 case in 250,000 to 1 million live births. Here I present a case report of multiple stones in renal pelvis, urinary bladder and prostatic urethra in alkaptonuric patient, successfully managed with surgical treatment

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A 45 years old male patient admitted in skin dept. with c/o skin lesion on both palm and cola coloured urine since 5 years.

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Multiple elevated pappules were found on both the ear lobes and dorsum of the wrist. Patient had hyperkeretosis on both the palm & soles, mild tendernes in right lumbar region, decresce hearing in both ear, shoulder joint &sacroiliac joint stiffness. His urine turned back to cola colour in sunlight.

INVESTIGATION
His Hb-13.5,Total count-7000, Platletcount-3.4laks, S.urea17, S.creatinine-0.5, S.Na-140meq/l, S.K-4.3meq/l,ESR-15, serum homogentisic acid->6.6microgram/ml, urine homogentisic acid->3.12 mmol/l. In X-ray K.U.B. show stone in Right renal pelvis ? stone in bladder, X-ray P.B.H. show opacity surrounding pubic symphysis. In ultrasoundography right side 12mm-staghorn calculi was present in renal pelvis. Patient was transferred to surgery dept. Surgeon was planed intera venuos pyelography (IVP). In IVP both kideny had normal excreting function. Surgeon had planned Rt. Pyelolithotomy. During operation surgeon could not catheterize the patient, then supra-pubic catheterization (S.P.C.) was done. Rt. Pyelolithotomy with 5 FR double lumen-J(D-J) stent was kept in operation. After 7 day all stitch was removed. After 15 days cystoscopy was planned for bladder stone. During cystoscopy found that multiple calculi present at prosthetic part of urethra. During cystoscopy prostetic calculi was removed. 16 French gauge Foly’s catheter was done & SPC in situ. Multiple stone in urinary bladder&D-J stent showed in digital X ray K.U.B.& PBH on next day. After 14 days cystoscopy was planned. On cystoscopy residual prostetic calculi & bladder stones&D-J stent were removed. After 2 days S.P.C. was removed.

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DISCUSSION

Alkaptonuria is a rare autosomal recessive inherited disease, caused by a mutation in the homogentisate 1, 2-dioxygenase (HGO) gene. Alkaptonuria arises in people who have inherited two abnormal HGD genes: one from each parent. This results in a deficiency of the enzyme homogentisic acid 1, 2-dioxygenase, responsible for the breakdown of homogentisic acid (HGA) in the liver and kidney, one of the end-products of the phenylalanine and tyrosine degradation pathway. Consequently, there is accumulation of HGA in the blood and several tissues, where it precipitates in the form of a pigment, the alkapton, giving them a dark ocre colouration; hence, this disease was termed ochronosis. Substance is also eliminated in large quantities in urine, where it is oxidated to benzoquinones by the action of light and alkaline pH, which in turn form melanin like polymers giving it the same dark colouration on standing. It has an incidence of 1:10,000,000, with male/female ratio of 2:1 and no race predominance. Alkaptonuria is associated with kidney and prostatic stones. Stones probably form because of the extremely high levels of urinary HGA excretion (renal clearance rates of 400 to 500 ml per minute). A unilateral ureteral obstruction by renal and prostatic ochronotic lithiasis is more common. Renal stone formation occurs, especially after the fifth decade. Prostate stones start to form at the same time. However, their development is not associated with the formation of kidney stones. In the prostatic alkaline pH, HGA polymerizes and precipitates, serving as a nidus for stone formation. The clinical features are due to the colouration and precipitation of HGA in the connective tissue, especially tendons, cartilages, endocardic and arterial subendothelial tissue and sclera, among others. Patients have a blue greyish colouration starting in sclera then extending to cornea, acquiring a more dark colour with time. Then it extends to the ear lobes and the back of the hands. Scrotum and axilary folds may also be affected. The pigment is eliminated by sweat and may be noticed on clothing. Ochronotic arthritis affects the spine and large joints, especially shoulders, hips and knees. It resembles ankylosing spondylitis. Diagnosis is based on its characteristic clinical features: dark urine, renal pelvis, bladder or prostatic stones, eye and skin colouration and joint pain, complemented by quantification of homogentisic acid in urine (5grm/24h) and tissues. The characteristic articular radiological findings and the more recently developed molecular analysis of the HGO gene mutations are also helpful. Different forms of treatment have been used in patients with ochronosis. Dietary restriction of phenylalanine and tyrosine reduces the excretion of HGA. Vitamin C has been used in high doses, producing a decrease in the urinary excretion of benzoquinone acetic acid (BQA), a toxic oxidation product of HGA. However, it has no effect in the elimination of homogentisic acid itself, and there are no reliable studies to prove vitamin C’s therapeutic effectiveness. Although it is a disease that does not change a patient’s overall survival, alkaptonuria can be very incapacitating, especially due to its osteoarticular sequelae, thus reducing one’s quality of life. Surveillance for renal, prostate and cardiac complications after the fourth decade of life and strict attention to pain control are advisable.

REFERENCES