

Case report

Alkaptonuria in a 5-year-old boy in Iraq

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Introduction

Alkaptonuria is a rare inherited genetic disorder of tyrosine metabolism that causes the urine to turn black on contact with the air when homogentisic acid is oxidized to form a pigment-like polymer material [1,2]. The earliest sign of the disorder is the tendency for babies' diapers to stain black; later on in childhood and early adulthood, there is an asymptomatic, progressive deposition of the polymer into collagenous tissues [2-4]. One report suggests an incidence as high as 1 in every 25 000 live births, although worldwide it is certainly far lower [1,4-6]. Our objective in reporting this case study is to highlight the importance of early detection of the disease by paediatricians, to raise awareness and to prevent late complications by simple treatment.

Case report

Our patient was a 5-year-old boy, from a first-cousin consanguineous couple. The mother had a full-term normal vaginal delivery at hospital, with routine labour and a healthy postnatal period. After the first few months of the baby's life, the mother noticed a dark black colour on the diapers, some time after urination or a change of urine colour on ferric material when he urinated outside the bathroom; otherwise, the child was normal. The couple consulted our subspecialty clinic in Al Kadhimia hospital

for genetic counselling because they had suffered secondary infertility with 2 abortions. They became increasingly anxious about the dark-coloured urine when the new baby was 6 months of age. They became concerned that he might have the same problem, so they finally consulted our clinic for investigation and counselling.

Clinical examination recorded a boy, 5 years old, active, alert, with no pallor and no jaundice. Development was normal. All growth parameters on the chart were within the normal range for age. Chest, heart and abdominal examinations were normal. Examination of joints was normal, so too was the neurological examination.

Investigations were performed as follows. On testing urine for reducing substance, Benedict's reagent gave a strong positive. For urine and serum chromatography, the homogentisic acid level was very high. Other items were normal. After urination on a diaper, we observed the colour change to black.

We talked to the parents and informed them that the diagnosis was alkaptonuria. We referred the child to the ophthalmologist for baseline examination, which was normal, showing no depositions. Echocardiography results were also normal.

Treatment was started with vitamin C (1 g/day). This is a large amount since if there is a delay in diagnosis and treatment of this disease it can result in arthritis and ochronosis (darkening of the tissues) due

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to the slow accumulation of the dark polymer of homogentisic acid in cartilage and other mesenchymal tissue; this leads to dark blackened spots in the sclera, cornea, ear cartilage and arthritis with advancing age especially in men, with a high incidence of heart disease [4,6].

Discussion

Alkaptonuria is a rare autosomal recessive disorder of inborn error of metabolism as a result of deficiency of the homogentisic acid oxidase enzyme [7]. A literature review for the last 3 years revealed that the reported cases are few and often present late; for example, Yoshikai et al. reported

a 65-year-old male with aortic valve regurgitation and inferior myocardial infarction [8,9]. Another study presented 5 cases with ochronosis and joint pain [10], while Kazancioglu presented a 33-year-old male with end-stage renal failure [11]. Others like Chevez present pigmented conjunctival lesions [12]. Our patient was also male, only 5 years old, and with early detection we can protect him from all the above-mentioned complications of ochronosis by giving him high doses of vitamin C. Early detection is better than treating late complications. It is recommended that families investigate suspected cases in new babies to ensure early detection.

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