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Case Report

An Enigmatic Color Change of Urine: Alkaptonuria

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ABSTRACT

Alkaptonuria is an exceedingly rare tyrosine metabolism disorder of autosomal recessive inheritance. Only a few instances of it have been observed in Bangladeshi children. Here, we talk about a 2-year-old boy who had dark urine and was later found to have alkaptonuria.

Keywords: Dark urine, Alkaptonuria

Introduction

Homogentisic acid accumulates in the blood and tissues as a result of alkaptonuria, a rare autosomal recessive disorder of tyrosine metabolism caused by a deficiency in the enzyme homogentisic acid oxidase (HGO). When urine is exposed to air, homogentisic acid and its oxidized analog alkapton exhibit a unique black tint [1].

The pigmentation known as ochronosis is caused by the accumulation of homogentisic acid and its metabolites in tissues. The sclera, conjunctiva, limbic cornea, heart valves-particularly the aortic valve-intervertebral disc, muscles, and other tissues all contain it [2]. Later in life, alkaptonuria can cause heart valve degeneration, arthritis, and joint destruction [3, 4]. We describe the situation surrounding a 2-year-old boy who had dark urine and was subsequently diagnosed with alkaptonuria

Case Report

Another mother complained of her 2-year-old son's dark urine at the renal clinic at Dr. MR Khan Shishu Hospital and intracerebral hemorrhage (ICH). She discovered it the next morning when the kid urinated on the bed overnight. The previous night's urine had stained the white bed cover a dark blackish tint. She also noticed the dark stain on the diaper in the morning while changing it. The mother was worried about her child's urine

A

B

Figure 1. Normal and dark urine

A. Urine immediately when passed in the morning. **B.** Urine turned to black when left standing.

containing blood. On request, she stored the child's pee in a pot; it was normal in color immediately after passing urine (Figure 1A), but when left standing, it turned black (Figure 1B). During his general assessment, nothing unusual was found. No signs of joint or cardiac problems confirmed by X-ray joints and echocardiography were observed, respectively. A routine urinalysis was negative for red blood cells and hemoglobin. A diagnosis of alkaptonuria was made when the amount of homogentisic acid in the urine was quantitatively measured and it was high at 1553 and the reference range is 1%. High-dose Vitamin C treatment was started at 1 gm/day. The parents received counseling regarding the illness, its long-standing complications, and the necessity for follow-up.

Discussion

Alkaptonuria is a very rare disorder in youngsters. Urine turns dark due to oxidation and polymerization of homogentisic acid, which is accelerated by an alkaline pH [5]. The primary symptom of this disorder is black urine on standing or stains the diaper, but most parents overlook or ignore this symptom.6 Only 21% of children with alkaptonuria are diagnosed before 1 year of age. The youngest case of alkaptonuria was reported at 7 days of age from Srilanka [6-8]. An adult patient with this disorder typically presents with arthritis, ocular, cutaneous, and cardiovascular ochronosis in their fourth decade [6]. In the case of our patient, his parents were unaware of it during her early infancy till he appeared at the age of 2 years.

Arthritis is the only incapacitating sign of this disease and affects nearly all patients as get older. It first appeared in large weight-bearing joints, such as the hips, spine, and knees. Patients with recurrent cardiac involvement have heart disease related to mitral and aortic valvulitis. Myocardial infarction caused by ischemic heart disease is one of the main causes of death. These clinical symptoms are not yet apparent due to our patient's incredibly early presentation, but we nonetheless urged them to follow up frequently to check for these concerns [8, 9].

Alkaptonuria has no particular treatment. Nitisinone has been proposed to be a potent inhibitor of the second enzyme para-hydroxyphenylpyruvic acid oxygenase in the tyrosine catabolic pathway; it can be used to prevent the formation of the offending homogentisic acid (HGA) molecule. Although it is currently only being tested on adults. In a few reported cases, they treated this with high-dose ascorbic acid, more commonly known as Vitamin C, an antioxidant believed to reduce HGA to benzoquinone acetate (BQA) via oxidation. However, the investigation revealed that although Vitamin C reduced the HGA to BQA conversion, it did not affect the urinary excretion of HGA [9, 10]. We also prescribed Vitamin C to the patient. 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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Conclusion

Despite its rarity, alkaptonuria ought to be considered as a cause of darkening urine in children. The patient must have a regular checkup to evaluate if there are any long-term effects.

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this research.

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Authors' contributions

All authors equally contributed to preparing this article.

Conflict of interest

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