Case Report of A Boy Presented with Brown Nappy

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Abstract

Background: Alkaptonuria is a rare tyrosine metabolism disorder due to a deficiency of the enzyme Homogentisic Acid Oxidase (HGO) and it carries autosomal recessive inheritance. We report this case, for increasing awareness among the doctor's society, so that early diagnosis of diseases from urine color.

Case Presentation: A 14-month-old boy, 2nd issue of a consanguineous couple brought to Outpatient Department of Paediatric Nephrology with symptoms of staining of nappy following urination on 1st March 2022. His urine chromatography report shows a high concentration of homogentisic acid and later diagnosed with a case of alkaptonuria.

Conclusion: Alkaptonuria has to be considered if any child presents with darkening of urine exposure to the atmosphere. Early identification of diseases would be beneficial for delaying potential complications.

Key words: Alkaptonuria; Brown nappy; Homogentisic acid.

Introduction

Certain metabolic disorders manifest as a particular color of urine. Urine that comes into contact with the atmosphere turns black when it has alkaptonuria.It is a hereditary autosomal recessive tyrosine metabolism condition brought on by a Homogentisic Acid Oxidase (HGO) deficiency. Homogentisic acid builds up in theheart, connective tissue, cartilage, blood, and weight-bearing joints and excreted in theurine, sweat, semen, etc. When urine comes into touch with air, too much homogentisic acid is eliminated, causing the urine to turn dark.[1]

The buildup of homogentisic acid and its metabolite causes a pigmentation known as ochronosis in the intervertebral disc, muscles, the limbic cornea, sclera, conjunctiva and heart valves, especially aortic valve and other tissues. Alkaptonuria can lead heart valve degeneration, joint destruction and arthritis later in life.[2] In clinics for children under one year old, just 21% of the youngsters are found to have the illness.[3] 1 in 250000 is the incidence. In addition to representing the UK, Germany, Lebanon, Sudan, Saudi Arabia, Turkey and other countries, it is exceptionally common in Slovakia (Incidence 1 in 19,000).[1]

We report a 14 month-old-kid who presented with dark urine and was later identified alkaptonuria, in order to facilitate early diagnosis of new instances and identification of potential problems.

Doctors from all specialities, including general practitioners, will be aware of this instance. They can identify a patient based only on the color of their urine and prevent further difficulties.

Case Presentation

On March1, 2022, Master 'A' a 14-month-old boy from Banshkhali, Chattogram was hospitalized to the Outpatient Department of Pediatric Nephrology at Chittagong Medical College Hospital with a history of staining of nappy after passing urine since birth. The baby was otherwise healthy. He is the second issue of a consanguineous parent and was exclusively breastfed for up to 6 months then started complementary feeding, not immunized and belongs to a low socio-economic status without significant drug history.

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Submitted on : 04.09.2023
Accepted on : 22.10.2023

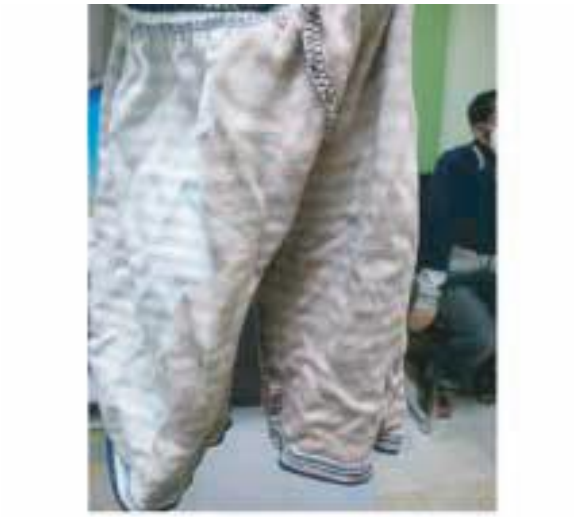

Image 1 Changes of color of pant after passing urine

Baby was active, well, alert, interested with surroundings, not pale, non icteric with normal vitals and age appropriate growth development. The locomotors system, precordium, abdomen, respiratory system and other systems findings were normal. Eye examination found normal.
Collected urine in a clear jar showed initially brown, then black discoloration after long standing exposures to atmosphere.

Image 2 Urine turns black after long standing exposure to atmosphere

The concentration of homogentisic acid in urine chromatography was found high.

In addition to being counseled about the illness, its implications and the need for follow up care, parents were encouraged to continue breastfeeding and provide complimentary feedings. At the 6 month check up, the infant is showing no sign of problems. Before commence the study necessary permission was taken from respective authorities and written informed consent was obtained from the patient's father for the publication of this case report and accompanying images.

Discussion

A rare autosomal recessive inborn error of mistake called alkaptonuria is brought on by a homogentisic acid oxidase deficit.[1]

The initial sign of alkaptonuria is a darkening of the urine upon exposure to air.[4-6, 2,3]

An alkaline pH accelerates the oxidation and polymerization of homogentisic acid,resulting in dark urine.[3]

Homogentisic acid builds up intracellularly and extracellularly, it is oxidized to benzoquinone acetate, which polymerizes to form a dark yellow or ochre pigment resembling melanin. Ochre buildup in the connective tissue, including the cartilage.[7]

There may be blue black staining of the palms, feet and ear cartilage as well as blackish discoloration of the eyes, knee painand stiffness in the metacarpal joints.[8]

The patient may exhibit some orthopedic problem, hyperchromic papules on the lateral border of the second fingers and a family history of the same ailment.[9]

The only debilitating symptom of this illness is arthritis, which almost all patient experience as they age and is one of the leading cause of death from valvular heart disease.[12]

Continued monitoring in conjunction with early disease recovery can prevent futue consequences.[6]

Patients with alkaptonuria might try a variety of trearment strategies. The majority of current care is palliative. Other treatment options are Ascorbic acid, low protein diet, lifestyle counseling, physiotherapy, regular surveillance for a treatable complication, pain management, organ replacement therapy, palliative surgery and potential disease modifying therapy-Nitisinone. Newer modalities of treatment are missing enzyme replacement or gene replacement.[10]
According to certain research, siblings, alkaptonurias differ phenotypically in terms of severity.[11]

Since the medication nitisonin,s buildup affects ochronosis, treatment usually centers on a metabolic pathway involving homogentisate.[12]

Limitation

We discussed only one case.

Conclusion

Alkaptonuria is a rare disorder, but not difficult to diagnose by seeing urine color. Consanguineous marriage can reduce the number of alkaptonuria cases.

Recommendation

To obtain experience in this subject, more cases are required.

Acknowledgement

We are very much grateful to patients and patients' attendants.

Contribution of authors

SA-Drafting, citing references & final approval.
MMQ-Design, citing references & final approval.
US-Design, critical revision & final approval.
AKMRK-Conception, drafting & final approval.

Disclosure

All the authors declared no conflict of interest.

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