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Letter to Editor (Pages: 1857-1858)

Black Urine

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Dear editor-in-chief,

Alkaptonuria is a rare autosomal recessive metabolic disease, with a prevalence of 1: 250,000 to 1: 1,000,000 (1). Alkaptonuria occurs mostly in adults aged over thirty years (2). The disease is caused due to total inhibition of homogentisic acid oxidase enzyme, which catalyzes the conversion of homogentisic acid (HGA) to maleyl acetoacetate, fumaric acid and acetoacetic acid (3). It is usually deposited in outer ear cartilage, sclera, eyelid, cornea, conjunctiva, eyelids, mucous, skin, bone and internal cardiac structures (mitral valve, aorta and coronary arteries), as well as secreted in urine and sweat (2). The most common presentation in children is darkening of the urine exposed to air, while osteoarthritis followed by changes in eyes, ears, skin and cardiovascular is the most common symptom in adults (4). Alkaptonuria is a still frequently sub diagnosed and rare disease. We present here a child with the final diagnosis of alkaptonuria.

A two-year-old boy was born at term of healthy, non-consanguineous Iranian parents. His mother attended in the clinic with the history of sometimes discoloration of diapers after passing urine. She noticed that first at the age of one month with intensified in recent months. His physical examination and growth parameters were normal. His mother denied taking any medication (sorbitol, nitrofurantoin, metronidazole, methocarbamol, sena and methyldopa) (5). Qualitative urine examination showed dark black discoloration; by this history, alkaptonuria was the most clinical suspicious. In the following a 24-hour-urine sample was collected and sent for quantitative measurements. The urine sample was highly positive for homogentisic acid and negative for porphyrin metabolites. Examination of eyes, musculoskeletal systems, skin, and cardiovascular system was normal.

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There is no definite treatment of this disorder. Reduction of proteins, mainly phenylalanine and tyrosine has diminished homogentisic acid excretion. Whether a mild dietary restriction from early in life would avoid or minimize later complications is not known (2). Another recommendation is to prescription a high dose of vitamin C, which its antioxidant effect prohibits the oxidation of the homogentisic acid and deposition in cartilaginous tissues (2). Limited use of nitisinone, a potent inhibitor of the enzyme 4-Hydroxyphenylpyruvate dioxygenase (HPPD), which mediates formation and urinary excretion of homogentisic acid, has been reported (6). However, safety of prolonged use is still an open question (2). So far, we have reviewed the literature our case is the earliest age of presentation from Iran. The parents are explained about the disease and its complications and the need for further follow up. The follow-up with other specialties such as cardiologist, orthopedics, ophthalmologist and otorhinolaryngologist are needed (2).

In Conclusion, alkaptonuria is a still frequently sub diagnosed disease, of difficult management in spite of increasing multidisciplinary study and monitoring through complementary exams. Clinicians should be aware of multiple system involvement in this disorder, as early recognition and proper treatment may significantly improve the quality of life in these patients.

Key Words: Alkaptonuria, Black urine, Homogentisic acid.

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