VIEWER'S CHOICE (LETTER TO EDITOR)

Alkaptonuria - A Rare Disorder Revisited

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Parents of a one month old girl sought consultation regarding brownish discolouration of their daughter's clothes, diapers and napkins noted since about 5th day of birth. The baby was the first issue born of non-consanguineous marriage. Medical history as well as family history was unremarkable. Physical examination revealed no abnormality. Single spot urine on collection appeared clear and yellow but it turned dark brown to black on prolonged exposure to atmosphere and also rapidly on addition of ten percent liquor ammonia or sodium or potassium hydroxide. The urine sample was sparingly acidic with specific gravity 1.010 and without any pus cell, red blood cell, hemoglobin or protein. The urine sample was subjected to the qualitative tests for homogentisic acid (HGA) (on the basis of its strong reducing property) as follows: -

Tests (urine sample)	Results
Boiling with Benedict's solution	Brownish precipitate
Mixing with 10 percent ferric chloride	greenish brown precipitate
Mixing with10 percent liquor ammonia and 3 percent silver nitrate solution	Black colour was produced from precipitate of silver

Urinary HGA was measured quantitatively by spectrophotometry or chromatography (1). Urinary creatinine level was measured by modified kinetic Jaffe's method. Urinary HGA to creatinine ratio was found 4.11 (in non-alkaptonuric person the ratio is < 0.01 mmol / millimoles of creatinine) (2). X-ray knee, hip and lumbar spine, ultrasonography of kidney, ureter and bladder and echocardiography were normal. Thus it was confirmed that the baby had alkaptonuria.

Alkaptonuria rarely reported from Indian subcontinent, is a rare autosomal recessive disorder of homogentisic acid oxidase deficiency characterized by homogentisic aciduria, ochronosis and arthritis resulting from accumulation of HGA and bezoquinone acetic acid in the body (3). Sir Garrod described an alkaptonuric baby, born of a consanguineous marriage, whose urine darkened in its napkin on standing and on exposure to air on 3rd day onwards after birth as because it contained HGA (4). The condition is very rare, affecting one in 250,000 to one million people worldwide (3). Male children are affected equally but sooner and more severely in comparison to females. Homogentisic aciduria is the only manifestation in childhood. Though alkaptonurics usually are born out of consanguineous marriages yet the present case is an exception. Early detection can pave the way for prevention or limitation in the development of complications. Restriction of protein, administration of ascorbic acid, nitisinone and avoiding unnecessary use of any other drugs can be helpful in delaying the progress of the disease. Finally, there is no definitive management available till today.

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