Alkaptonuria is a rare inherited genetic disorder in which the body cannot process the amino acids phenylalanine and tyrosine, which occur in proteins and in which urine coloration is black in people with alkaptonuria. This typically develops arthritis, particularly in the spine and large joints, beginning in early adulthood. Homogentisic acid (HGA) is an immediate degradation product in the metabolism of tyrosine and phenylalanine, which can cause pigment deposition in the connective tissue of eye, ear, cartilage, and cardiac valves. Gomogentisic acid oxidase (HGAO) converts HGA to homogentisic acid (HGA) to molecules that can be used in Krebs cycle. Deposition of HGA produces gray to blue pigment in the tissues. It can cause pigment deposition in the connective tissue of eye, ear, cartilage and cardiac valves. Generally, the disease will be asymptomatic, but may appear in the middle age due to early degenerative arthritis involving lower spine, and shoulder and hip joints requiring early joint replacement.

AKU should be suspected in patients with the following clinical triad:
1. Degenerative arthritis (premature) i.e. in young patients of <45 yrs.
2. Urine turning blue-black on standing (due to alkalinization).

3. Abnormal pigmentation of connective tissues in eye, ear, and cardiac valves.

Since HGA is absent in the blood plasma and urine of healthy subjects, both of them can be used for the disease diagnosis. In AKU patients, the average levels of HGA noted in the plasma and urine are 6.6 micrograms/ml and 3.12 mmol/ mmol of creatinine respectively [1].

Confirmatory tests for diagnosis are paper chromatography and thin layer chromatography. Characteristic radiological findings include vertebral disc calcification, chondrocalcinosis, and osteoarthritis of multiple joints (especially spine) at young age.

Since most of the young patients with AKU present with low back pain, it can mimic ankylosing spondylitis. Generally, the disease could be suspected in young patients with low back pain and X-ray showing inter-vertebral disc calcification with normal sacroiliac joint. They can also have premature osteoarthritis of knee or other major joints [5].

A detailed history and examination may reveal black staining of the undergarment, change in the urine color on longstanding, pigmentation of sclera, and grayish hue discoloration of the ears. First-degree relatives may have symptomatic scleral pigmentation and ear discoloration before they develop any joint manifestation (as many are unaware of the urine color change or staining of clothes).

No treatment modality has been unequivocally proven to reduce the complications of AKU. Commonly advocated treatment strategies include large doses of ascorbic acid (vitamin C) and dietary restriction of tyrosine and phenylalanine. Although, dietary restriction may be effective in children, it has not shown any benefits in adults [1].

REFERENCES