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A Rare Disease, Alkaptonuria Cause for Low Backache

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Abstract: A rare case of Alkaptonuria (AKU) with Low backache (LBA) is being reported. AKU is a rare metabolic disorder inherited as an autosomal recessive mode that can cause premature degenerative arthritis of the spine and peripheral large joints. Homogentisic Acid (HGA) is an immediate degradation product in the metabolism of tyrosine and phenylalanine. It is characterized by darkening of the urine on standing and ascorbic acid pays an important role in the metabolism of tyrosine and phenylalanine. Incidence of this disease is 1:2,50,000 people. Hereby present a case of middle-aged man who developed low back pain secondary to AKU.
Keywords: Alkaptonuria, low backache, homogentisic acid, ascorbic acid.

INTRODUCTION

Alkaptonuria is a rare inherited genetic disorder in which the body cannot process the amino acids phenylalanine and tyrosine, which occur in protein and in which urine coloration is black. People with alkaptonuria typically develop arthritis, particularly in the spine and large joints, beginning in early adulthood. The worldwide prevalence of Alkaptonuria (AKU) is 1 case in 2,50,000-10,00,000 live births [1]. So far, 950 AKU sufferers have been identified in 40 countries [2, 3].

CASE PRESENTATION

- A 49-year-old male working in Central Govt at Hindupur, Andhra Pradesh and presented with a history of low backache since 10 years.
- He had difficulty in restriction in back movements, bending and scoliosis.
- He had reported the occurrence of urine turning black after some time.
- He was also experiencing early morning stiffness, which improved with activity, and pain on both the knee joints without swelling.
- The patient had a family history of similar complaints and the affected family members also demonstrated back pain.
- His wife is NACO +ve recently detected, but, he is negative.

EXAMINATION

- Physical examination demonstrated that the blackish discoloration of ears as well as the pigmentation of sclera.
- Examination of the spine showed kyphoscoliosis with normal spinal mobility.
- Anterio-posterior X-ray of the spine showed inter-vertebral disc calcification with normal sacroiliac joint.

DISCUSSION AND CONCLUSION

AKU, a rare defect with autosomal recessive inheritance, is caused by the abnormal tyrosine catabolism due to the deficiency in homogentisic acid oxidase (HGAO) [4]. The enzyme catalyzes the catabolism of 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

1. Phornphutkul C, Introne WJ, Perry MB, Bernardini I, Murphey MD, Fitzpatrick DL, Anderson PD, Huizing M, Anikster Y, Gerber LH, Gahl WA; Natural history of alkaptonuria. New England journal of medicine, 2002; 347(26):2111-21.
2. Grant Mitchell A, Rezvani I; Defects in metabolism of amino acids: Tyrosine. In Nelson Textbook of Pediatrics Volume 1. 18th edition. Edited by: Kleigman, Behrman, Jenson, Stanton. Elsevier Publication: Indian reprint; 2008; 534.
3. Ranganath L, Taylor AM, Shenkin A, Fraser WD, Jarvis J, Gallagher JA, Sireau N; Identification of alkaptonuria in the general population: a United Kingdom experience describing the challenges, possible solutions and persistent barriers. J Inherit Metab Dis., 2011; 34:723-30.
4. Develop AKUre. 950 AKU patients [Internet]. Cambridge (UK): The AKU Society and the Develop AKUre Consortium: c2014 [cited 15 May 2013]. Available from: http://www.developakure.eu/950-aku-patients/
5. Schimizu I, Hamad T, Khaprey Z, Miyanishi K. Hara T; Ochronotic arthropathy pathological evidence of acute destruction of the hip joint. Clin. Rheumatoid., 2007; 26: 1189:91.