Sir,
A 44-year-old female born of nonconsanguineous marriage, presented with asymptomatic dark raised lesions on hands and face for 2 years. She additionally had severe backache and hip joint pain for 3 years and reduced vision for 1 year. She denied history of prior application of topical creams. She did not notice any staining of clothing or dark-colored urine on prolonged standing. The family history was not contributory.

Her general and systemic examination findings were unremarkable. Cutaneous examination showed multiple bluish-black ill-defined plaques on the malar area, nose, both hands, and right ear lobe [Figure 1a-c], [Figure 2a and b]. The nose additionally had caviar-like hyperpigmented papules. Thickening and reduced flexibility of helices were noted. Both eyes showed bluish-brown pigmentation over the sclera [Figure 3a and b]. Skeletal examination revealed painful internal rotation with decreased range of movements over bilateral hip joints.

Blood urea and serum creatinine were marginally raised. Urine turned dark on prolonged standing [Figure 4]. Screening for homogentisic acid in urine was negative. Chest X-ray, ECG, and 2D ECHO detected no abnormality. Ultrasonography of the abdomen and pelvis showed increased cortical echogenicity of the right kidney. Radiological examination of the cervical and lumbar spine showed diffuse disc bulges and desiccation at various levels. MRI of hip joint revealed acetabulum protrusion and bone marrow edema. Skin biopsy showed sharply defined ochre-colored banana-shaped fibers in the dermis. [Figure 5a-c].

A diagnosis of endogenous ochronosis with chronic renal disease was made. Tablet vitamin C daily and a low-protein diet were advised and regular follow-up with an orthopedician and a nephrologist was suggested.

Endogenous ochronosis is described as pigmentary changes developing due to alkaptonuria, a rare autosomal recessive disorder, due to deficiency of homogentisic acid oxidase in kidney and liver leading to accumulation of homogentisic acid. It leads to the characteristic triad of darkening of urine (one of the early signs), ochronosis, and arthropathy.

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Figure 1: (a) Multiple symmetrical bluish-black ill-defined plaques on the malar area and nose with caviar-like hyperpigmented papules on the nose; (b and c) Hyperpigmented plaques on hypothenar and thenar eminences, lateral border, and tips of fingers along with ill-defined hyperpigmentation on dorsomedial aspect of hand

Figure 2: (a) Bluish-black pigmentation of right ear lobule; (b) left lobule of ear with no pigmentation
sun-exposed sites, ears and nose cartilages, areas of high eccrine sweat gland density becomes obvious in the 4th decade.[2]

The only disabling effect, ochronotic arthropathy, typically affecting large joints, and spine occurs in almost all patients with advancing age. An increased risk of myocardial infarction with calcification and stenosis of aortic valves and coronaries have also been identified. Pigment deposits can form stones in the prostate, urethra, and kidneys due to high urinary homogentisic acid excretion.

The diagnosis is made with the presence of the clinical triad and confirmed by screening of urine for homogentisic acid.[3]

No effective treatment exists. A low-protein diet and vitamin C supplements help in reducing connective tissue damage.[4] Nitisinone, a triketone herbicide, has also been tried.[5] Active surveillance for cardiac, renal, and prostate complications should be done after the 4th decade.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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