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Images in Medicine

# A case of 'blue skin' and 'dark urine'

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ABSTRACT

A 60-year-old female presented with a 20-year history of progressive dark bluish discoloration of skin and passage of dark colored urine, painful arthritis and a recent history of invasive ductal carcinoma of right breast. Skin biopsy revealed hyaline material which was Periodic-Acid-Schiff stain positive and Congo-red stain negative, urine analysis revealed dark urine with presence reducing substance and radio-imaging showed intervertebral ossification and joint ankyloses. The patient was managed symptomatically with physiotherapy and acetaminophen on as required basis. This image is reported as a classic case of Alkaptonuria with clinical, histopathological and radio-imaging findings and the presence of invasive ductal breast carcinoma in the same patient.



## Introduction

A 60-year-old female presented with a 20-year history of progressive discoloration of face, hands and feet, passage of dark colored urine with history of painful hip and knee joint, total hip replacement of right side and a recent history of invasive ductal carcinoma of right breast.

## Clinical and imaging findings

Skin examination revealed dark bluish discoloration of forehead, malar areas, nose, external ears, sclera, dorsum of hands and feet (Fig. 1). Urine analysis revealed dark discoloration of urine with presence of reducing substance (Fig. 2). Skin biopsy showed epidermal thinning with clefted hyaline like material in superficial dermis, breakdown of dermal collagen with occasional crescentic and vermiform shaped ochre colored fibers. The hyaline material was Periodic-Acid-Schiff stain positive and Congo-red stain negative.

Radio imaging with Plain radiograph and magnetic resonance imaging of spine and pelvis revealed ossification of intervertebral discs, ankyloses of antero-inferior part of right sacroiliac joint with partial ankyloses of lumbar facetal joints and a post hip replacement right side status (Fig. 3). Renal function tests and echocardiogram were normal. Based on the clinical presentation, urinalysis, skin biopsy and radio imaging she was diagnosed as a case of alkaptonuria. Quantitative measurements of homogentisic acid in urine were not done.

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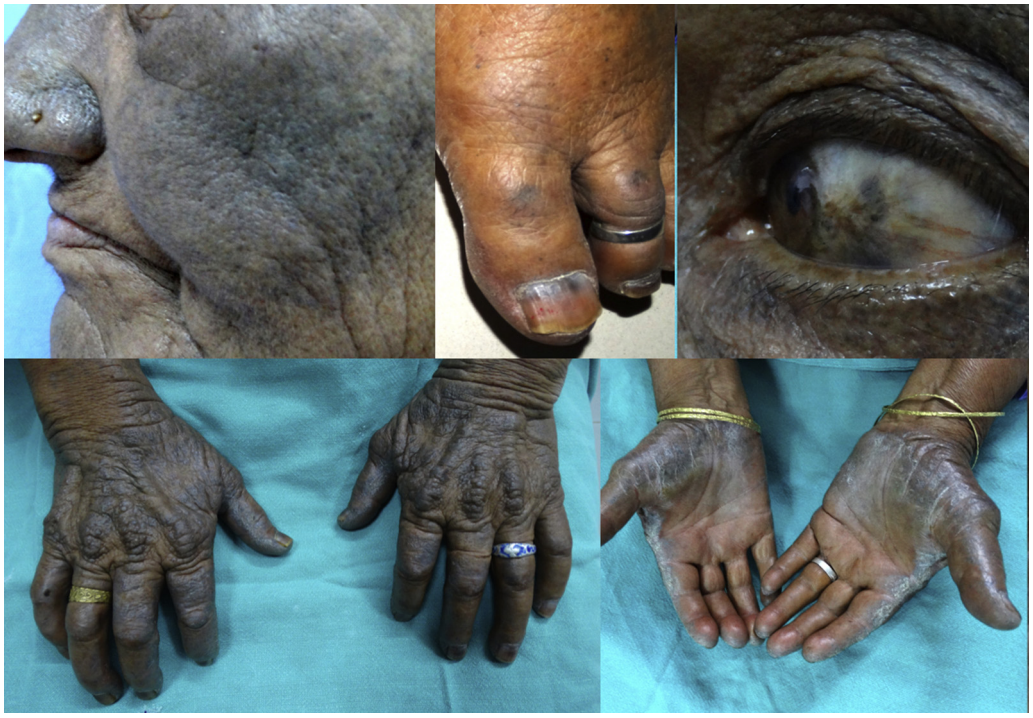

**Fig. 1 – (a) Face pigmentation on malar areas and nose. (b) Nail pigmentation. (c) Scleral pigmentation. (d) Ochronosis with waxy papules and plaques on dorsum and palmar aspects of hands.**

## Discussion

Alkaptonuria is a rare, autosomal recessive, metabolic disorder affecting 1 in 250,000 to 1 million people worldwide, while certain communities of Slovakia have a much higher incidence.[1,2] First reported in 1584 as "black urine disease",[3] it is caused by a deficiency in homogentisic acid oxidase which results in accumulation and deposition of oxidation and polymerization product of homogentisic acid in cartilage, eyelids, forehead, cheeks, axillae, genital regions, nail beds, buccal mucosa, larynx, tympanic membrane, and tendons.

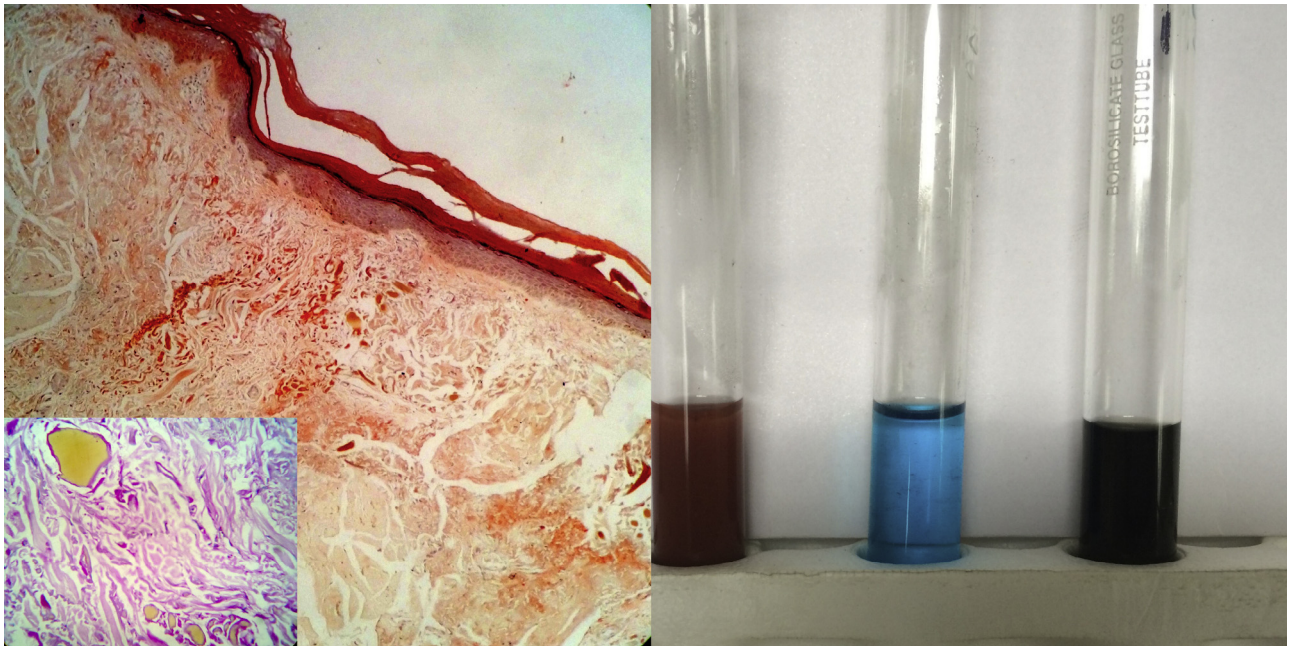


**Fig. 2 – Ochronotic pigment in dermis with evidence of dermal collagen breakdown and periodic acid Schiff stain positive material, PAS 10× with H&E 40× as inset (left). Urinalysis showing positive control in left tube, negative control in middle tube, and test sample showing black discoloration with dark green pigmentation after Benedict's test (right).**

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