Ashy Ears

FIGURE 1. The (L) ear with blue-gray discoloration.

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A 56-year-old man was referred to our clinic because of the symmetric appearance of blue-gray discoloration on the helix cartilage of his ears (Figs. 1 and 2). The lesions presented a gradual deterioration during the last 10 years. The rest of his physical examination was unremarkable. According to his past medical history, the diagnosis of alkaptonuria was made at the age of 35 because of the appearance of low back pain and dark urine of approximately 3-years duration. At present, he has experienced arthritis of the spine.

Alkaptonuria is a rare, autosomal-recessive disease of tyrosine degradation resulting from accumulation of homogentisic acid (HGA) within the body due to deficiency of the hepatic enzyme homogentisate 1,2-dioxygenase[1]. *HGD* is the gene encoding homogentisate 1,2-dioxygenase and is the only gene known to be associated with alkaptonuria.

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FIGURE 2. The (R) ear with blue-gray discoloration.

HGA is oxidized to a pigment-like polymeric material responsible for the black color of urine that is deposited in the cartilage of the body, including ears[2]. Joint symptoms involving the spine are usually seen in the 3rd and 4th decade, while pigment depositions begin to appear approximately in the 4th decade of life[2,3].

A variety of therapeutic modalities have been investigated; however, no preventive or curative treatment has been yet approved for alkaptonuria[3]. Young individuals should be counseled for physical therapy to promote optimal muscle strength and flexibility, but they should avoid contact sports. Dietary decrease of phenylalanine and tyrosine intake has been proposed to reduce the production of HGA. High-dose vitamin C is usually recommended to decrease the deposition in cartilaginous tissues, but has no effect on HGA excretion[4]. Nitisinone has been proposed for the pharmacologic treatment of alkaptonuria[5]. It is a triketone herbicide that inhibits the enzyme that produces HGA and has been approved for the treatment of tyrosinemia type I. Further investigations to determine the potential benefit of nitisinone in alkaptonuria for slowing the progression of joint disease are currently in progress[3].

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