Alkaptonuria - Severe (11)

Alkaptonuria is a rare inherited metabolic disorder caused by a deficiency of the enzyme homogentisate 1,2-dioxygenase. This leads to the accumulation of homogentisic acid in the body, resulting in darkened urine, pigmentation of connective tissues (ochronosis), and progressive joint and spine problems. Severe cases can significantly impact mobility and quality of life.