Alkaptonuria

Alkaptonuria is a rare inherited genetic disorder in which the body cannot process the amino acids phenylalanine and tyrosine, which occur in protein. It is caused by a mutation in the HGD gene for the enzyme homogentisate 1,2-dioxygenase (EC 1.13.11.5); if a person inherits abnormal copies from each parent (it is a recessive condition), the body accumulates an intermediate substance called homogentisic acid in the blood and tissues. Homogentisic acid and its oxidized form alkapton are excreted in the urine, giving it an unusually dark color. The accumulating homogentisic acid causes damage to cartilage (ochronosis, leading to osteoarthritis) and heart valves, as well as precipitating as kidney stones and stones in other organs.

Symptoms usually develop in people over 30 years old, although the dark discoloration of the urine is present from birth.