Open Peer Review on Qeios

Alkaptonuria

National Cancer Institute

Source

National Cancer Institute. Alkaptonuria. NCI Thesaurus. Code C84546.

A rare autosomal recessive disorder characterized by abnormalities in the metabolism of phenylalanine and tyrosine. It results in the accumulation in the blood of homogentisic acid which is excreted in the urine. The presence of homogentisic acid in the urine causes its color to turn black. The excessive amount of homogentisic acid in the blood may cause damage to cartilage and heart valves, and may result in the formation of kidney stones.