

Alkaptonuria is an inherited condition that causes urine to turn black when exposed to air. Ochronosis, a buildup of dark pigment in connective tissues such as cartilage and skin, is also characteristic of the disorder. Excess homogentisic acid and related compounds are deposited in connective tissues, which causes cartilage and skin to darken. People with alkaptonuria typically develop arthritis, particularly in the spine and large joints, beginning in early adulthood. This enzyme helps break down the amino acids phenylalanine and tyrosine, which are important building blocks of proteins. Mutations in the HGD gene cause alkaptonuria.