Alkaptonuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Alkaptonuria. ORPHA:56

Alkaptonuria is a metabolic disease characterized by the accumulation of homogentisic acid (HGA) and its oxidized product, benzoquinone acetic acid (BQA), in various tissues (e.g. cartilage, connective tissue) and body fluids (urine, sweat), causing urine to darken when exposed to air as well as grey-blue coloration of the sclera and ear helix (ochronosis), and a disabling joint disease involving both the axial and peripheral joints (ochronotic arthropathy).