Alkaptonuria

Alkaptonuria, also known as ochronosis, is an autosomal recessive metabolic disorder caused by a defect in the enzyme homogentisic acid oxidase. This enzyme participates in the degradation of tyrosine to fumarate. As a result of the enzyme deficiency, a toxic tyrosine byproduct called homogentisic acid accumulates in the blood and is excreted in large amounts in the urine. Excessive amounts of homogentisic acid is harmful to cartilage leading to severe and debilitating arthritis. Alkaptonuria can also cause darkening of connective tissue in sun exposed areas and around sweat glands. Urine may also turn black if collected and left exposed to open air for a long period of time.

PLAY PICMONIC

Autosomal Recessive

Recessive-chocolate

This metabolic disorder is inherited in an autosomal recessive fashion.

Deficiency of homogentisic acid oxidase

Home-gentleman Acidic-lemon Ox-daisies

Homogentisic acid oxidase is an enzyme that metabolizes homogentisic acid into 4-maleylacetoacetate.

In degradative pathway of tyrosine to fumarate

Tire turning into Fuming-tire

This enzyme participates in the metabolism of tyrosine to fumarate.

Homogentisic acid harmful to cartilage

Damage to Cartilage-cart

Excessive amounts of homogentisic acid is harmful to cartilage, leading to severe and debilitating arthritis.

Arthritis

King-Arthur

Excessive amounts of homogentisic acid is harmful to cartilage, leading to severe and debilitating arthritis.

Dark Connective Tissue

Dark Ears and Nose

Alkaptonuria can cause darkening of connective tissue in sun exposed areas and around sweat glands. This is because the accumulated homogentisic acid causes pigmented deposits in connective tissues throughout the body.

Urine turns black on standing

Black Urine

Urine can turn black if collected and left exposed to open air for a long period of time in alkaptonuria.