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.

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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.

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