OCHRONOSIS

http://www.aocd.org

Ochronosis is a bluish-black discoloration of the skin and cartilage. It is generally caused by a rare genetic disease called alkaptonuria. People with this disease have a deficiency in an enzyme called homogentisic acid oxidase which allows the build-up of certain substances that eventually deposit in connective tissue found throughout the body. Several medications have also been reported to cause ochronosis.

This disease entity may be first noticed in newborns as their urine may be darker than normal. People are often without symptoms until later in life when the skin, sclera (white part of the eye), ears, and nose may develop pigmentary changes. Ear wax of a person with ochronosis will also be dark in color. A build-up of certain substances in the joints may cause degenerative changes and arthritis in these patients. There also seems to be an increased risk of cardiac disease because of deposits within the blood vessel walls.

A localized type of ochronosis can occur in the skin over areas where topical hydroquinone cream is applied. This is mainly seen when high concentration, 6 to 8%, of hydroquinone is used over a long period of time.

Your doctor can perform blood and urine tests to check for this condition. At the present time, there are no treatments for ochronosis. Certain elimination diets may be beneficial. A referral to a specialist may be needed to discuss potential complications such as joint and heart disease.