# PSEUDOXANTHOMA ELASTICUM

**http://www.aocd.org**

Pseudoxanthoma elasticum (PXE) is an inherited disorder of the connective tissue in the skin, eyes, gastrointestinal and cardiovascular system. Typically the skin lesions begin in childhood but are not noticed until adolescence because of there is a lack of symptoms in addition to the rash. It is important to recognize this disease early in order to minimize complications of the cardiovascular and gastrointestinal systems. There are several different subtypes of PXE.

A patient first presents to a physician with perfectly symmetric PXE skin lesions on the outer neck. These lesions are without symptoms but are of cosmetic concern. The manifestation of PXE on the skin is very distinctive. Small, yellow papules 1-5mm in diameter form in a linear pattern. The skin takes on a "plucked chicken" appearance. As the disease progresses other symptoms may occur like gastrointestinal bleeding, high blood pressure, chest pain, and blood in the urine.

The cause of PXE is the result of a mutation in a protein that is most abundant in the kidney and liver cells. It is not understood why this would lead to abnormalities in other organs.

Diagnosis is made on clinical findings and several lab tests. A physician may order a simple blood test, a urinalysis, and stool screen. A **biopsy** of the lesion in question may also be ordered.

Many of the changes that occur with PXE are irreversible but preventive measures may be taken to minimize the disease course. The sagging folds of skin that are seen late in the course of PXE can be easily corrected by surgical removal of redundant skin folds if the patient desires. Collagen and autologous fat injections may be options for mental (forehead) crease treatment.

Regular visits with an ophthalmologist are important to prevent eye damage. A consultation with a cardiologist or gastroenterologist may be necessary if heart or gastrointestinal problems exist.

**This information has been provided to you compliments of the American Osteopathic College of Dermatology and your physician.**