What Is PXE?
Pseudoxanthoma elasticum (PXE) is an inherited disorder that causes mineralization in some tissue in the body. This means calcium and other minerals are sometimes deposited in skin, eyes, the cardiovascular system and the gastrointestinal system. PXE was recognized over a hundred years ago and a number of significant advances have been made in the past few decades.

What Are the Effects of PXE?
PXE results in a variety of signs and symptoms. The number, type, and severity of signs of PXE are different for each person. Certain effects of PXE can cause serious medical problems while others have less impact. The effects of PXE may include: skin changes; changes in the retina of the eye that may result in significant loss of central vision; changes in the cardiovascular system that may involve calcification of arteries and decreased blood flow in the arms and legs; changes in the gastrointestinal system that may lead to bleeding in the stomach or intestines. Not everyone gets these symptoms.

The Skin
Skin changes are often the first indication that a person has pseudoxanthoma elasticum (PXE). Typically, these changes appear on the sides of the neck and progress to other specific parts of the body.

The affected individual might notice small bumps on the skin. These are called lesions, or “papules” (see Figure 1). They are usually yellow or yellow-orange in color and irregular or rhomboid in shape. The lesions may group together to form a larger area of affected skin, resembling a rash or having a "cobblestone" appearance. These lesions can progress slowly and unpredictably, from the neck downward. With time, the lesions can come together to form plaques and loose and wrinkly skin (see Figure 2).

The lesions do not cause any problems other than their effect on appearance. They are asymptomatic. They are usually distributed symmetrically, equally on both the right and left sides of the body. They tend to progress downward, affecting the underarm (axillae), the inside of the elbow (antecubital...
fossa), groin, and the back of the knee (popliteal fossa). Sometimes the navel (periumbilical area) and inner lip (oral mucosa) are affected.

Skin signs of PXE can occur in young children, but the age of onset and the age of detection vary greatly from one individual to another. The areas of the body that are most affected are those which bend and flex. The neck, the underarms, the skin on the inside of the elbows, the groin, and the skin behind the knees may be progressively affected, leading to loose folds in these areas. The extent of skin involvement also varies greatly from individual to individual. In some individuals, there is little apparent skin involvement. In others, a great deal of skin is involved.

There is not presently any way to determine how much skin will be affected, at what rate the lesions will progress, or what affect the environment or diet may have on the progression. Generally, progression is slow. In late stages of the disease, the skin may have loose and saggy folds. In cases where this is unsightly, cosmetic surgery can be performed to tighten the skin. The outcome is usually good, though there have been some reported cases of stretched scars.

The definitive diagnostic tool at this time is a skin biopsy found to be positive for pseudoxanthoma elasticum (PXE). A dermatologist will take a small biopsy. The skin will show clumped, fragmented elastic fibers that are caused by small amounts of minerals not normally found in this tissue. It is possible to have PXE and not have any apparent skin lesions. In some individuals, careful examination of the skin by a dermatologist does not reveal any visible lesions, but a positive skin biopsy indicates the diagnosis of PXE.

Once the diagnosis is made, the affected individual should consider all the ramifications of the disease. The dermatologist should take a detailed history and refer you to an ophthalmologist and perhaps a cardiologist. PXE can cause eye, cardiac, vascular and gastrointestinal difficulties. It is recommended that affected individuals not take aspirin or non-steroidal anti-inflammatory medications. These may encourage gastric problems such as bleeding. For more information, please see our General Bulletin.

Some researchers have suggested that altering one's diet to include only the minimum daily requirement of calcium may slow the calcification of connective tissue throughout the body. There is no evidence to support this suggestion. There is no treatment for PXE or for the skin manifestations. Plastic surgery is sometimes used to correct lax skin. See our Plastic Surgery Bulletin for more information.

Fortunately, the effects of PXE on the various organ systems of the body are similar to those produced by much more common conditions – so you need to find a doctor who is caring and listens and takes the time to become educated about PXE. If necessary, they can read our medical bulletins and learn how to care for you. They can also consult us (we have gathered data on more than 4000 affected individuals), if necessary.