

PXE and Skin

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

The affected individual might notice small bumps on the skin. These are called lesions. 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. Some people have referred to this as "cobblestone" appearance. Others have said that the neck has an unwashed appearance.

The lesions do not cause any problems in and of themselves. 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.

The age of onset and the age of detection vary greatly from one individual to another. The extent of skin involvement also varies greatly from individual to individual. In some individuals, there is no 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 effect 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 show small amounts of minerals not normally found in this tissue.

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.

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 a cardiologist. PXE can cause eye, cardiac, vascular and gastro-intestinal difficulties. There may be special issues for women and pediatric patients. It is recommended that affected individuals not take aspirin or non-steroidal anti-inflammatory medications. These may encourage gastric problems such as bleeding.

The bulletins and newsletters of PXE International, Inc. are available on cassette tape.