

What Is PXE?

Pseudoxanthoma elasticum, (PXE), is an inherited disorder that causes some tissue in the body to become mineralized, that is, calcium and other minerals are deposited in the tissue. This can result in changes in the skin, eyes, cardiovascular system and gastrointestinal system. PXE was recognized over a hundred years ago. 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.

Reconstructive Surgery

“I had always said that plastic surgery was something I would never have done. Never! But now I really love the results,” said Isobel of Pennsylvania, USA. “However, there was a short time immediately after the surgery, when I would have said that I shouldn’t have had it done.”

Isobel, who has PXE, had reconstructive (plastic) surgery done on her neck in December 1996. Though her primary care physician and the plastic surgeon had both tried to prepare her for the immediate after-effects of this major surgical procedure, she was aghast, nevertheless. After the 3 1/2-hour procedure, she felt terrible, and the swelling and bruising seemed worse than she expected. Besides, the hospital discharged her after only one night. That initial recovery period was rough. However, by the time she took a vacation trip six weeks later, Isobel was completely recovered and delighted with the outcome of the surgery. For Isobel, insurance coverage was approved without undue difficulty.

Plastic surgeons effectively perform their usual procedures with individuals who have PXE, with typical outcomes. Consulting a number of surgeons over the past two decades, we have garnered information about the indications for surgery, pre-operative considerations, and the risks and uncertainties associated with plastic surgery. We have also consulted on the topic of issue of insurance coverage.

The wrinkles, folds, and looseness of the skin that may result from PXE, is called “laxity and skin redundancy”. This excess skin can be removed from the neck, armpits, thighs, breasts, and abdomen by means of plastic surgery. However, while plastic surgery is effective in reducing excess skin, it is not helpful in changing the “cobblestone” appearance of the skin. When PXE causes the mineralization of mid-dermis, the middle layer of the skin, the skin loses its ability to spring back. The skin sags and stretches. Elasticity cannot be restored to the skin, not even by means of plastic surgery. Therefore, the individual affected by PXE may want to make use of a procedure called “direct excision”. The folds and loose skin can be, essentially, smoothed out by pulling it all in one direction, somewhat as one might do when making a bed. After smoothing out the folds, extra skin remains. In bed-making, the extra sheet is turned down over the blankets. In direct excision, the extra skin is cut away and the incision is closed.

One problem with direct excision is that the incision is not always easy to hide, and it may leave an obvious surgical scar. When done to reduce the loose skin around the armpit, the surgeon can hide the scar under the arm. However even this scar might be visible if you were to wear a sleeveless shirt. Direct excision is the only option for some procedures. In these cases, you should weigh the possibility of scarring and the location of the scar against the expected results.

For individuals with a history of scarring, especially with obvious or thickened scars, discussion about where the surgical scar would be located should be undertaken, and whether the scar would be preferable to the loose skin. Individuals affected by PXE do not scar any differently than the general population, but the general population has a wide variation in how well people heal. A “good healer” is more likely to have good results from direct excision. The surgeon can assess this for you. Moreover, a surgeon can plan to locate the scar in a less conspicuous spot.

Procedures other than direct excision are more commonly done for people with PXE. A “face lift”, “neck lift” or tightening of the skin of the breasts or abdomen is more like a “redraping” of the skin. Many times, the surgeon tightens underlying muscle and removes fat. They make incisions at strategic and less conspicuous spots. For example, in a face-lift or a combination face lift and neck lift, incisions may be located at the scalp line or behind the ears. For these types of surgery, the term “redraping” may be as clear an analogy of what is done as is possible.

In abdominoplasty, plastic surgery to the abdomen, the surgeon rearranges the skin in several directions, then replaces it in a slightly different position. He or she makes an incision, with possibility of scarring, around the navel so that it ends up in its original

position. For the breasts, during a mastoplasty, skin may be re-shaped around the nipples, with the possibility of incisions around the nipples. Again, if you are considering plastic surgery you should find out exactly where the incisions will be and understand that permanent scarring is a possibility.

In plastic surgery, as in every kind of major surgery, there are always risks and the possibility of complications. Before deciding upon surgery, you will have a very thorough pre-operative cardiac workup, as well as an evaluation of all other systems. Since many procedures require general anesthesia, you will have a good, extensive pre-operative evaluation with a reputable surgeon. A cardiac workup will likely include, at the very least, a cardiogram, chest x-ray, and ultrasound of the heart. You should not smoke for several weeks before the surgery. Of course, smoking is never advisable for those with PXE.

It appears that individuals affected by PXE heal at the same rate as the general population, which again is highly variable and individual. Certain individuals who scar more extensively might develop keloids or hypertrophic scars such as occur in the general population.

Potential Complications

Among the complications that infrequently occur in the general population, are the risk of infections, reaction to anesthesia, and the risk of hematoma (bleeding under the skin). Certain complications are specific to the procedure done, or to the part of the body that is being treated. An infrequent but possible risk associated with a face-lift is injury to nerves of the face, more likely the loss of sensory nerves than those that affect movement.

Mastoplasty, the procedure done to reshape the breast, carries a risk of asymmetry or decreased sensitivity of the nipples, and difficulties in breast-feeding. Women would do best to wait until their childbearing is finished. Plastic surgery does not halt the process of aging, loose skin, or folded skin due to PXE. Nor do its results last if a patient's skin stretches due to childbearing or weight gain. It is important to be realistic about what plastic surgery can accomplish and to be aware of possible complications.

Insurance Coverage

A big frustration that individuals affected by PXE deal with in deciding whether to undergo plastic surgery is the issue of insurance coverage. It is often initially denied because those who determine coverage do not understand that this need is the result of a "congenital defect", technical language for a deviation from "normal" because of a disease one was born with. However, potential patients who are willing to appeal the decision and to fight for coverage have been successful, while some others have had no trouble in obtaining insurance coverage from first request.

The key to the decision often rests in how your surgeon details your need and is congruent with the definition of plastic surgery as reconstructive surgery. According to the definition

of The American Society of Plastic and Reconstructive Surgeons (ASPRS) (www.plasticsurgery.org), reconstructive surgery corrects abnormal structures caused by congenital defects, or by trauma or disease. Reconstructive surgery is commonly indicated if the surgery corrects the function of the body. On the other hand, if the surgeon calls it cosmetic, the ASPRS defines cosmetic surgery as surgery to normal structures of the body to improve appearance and self-esteem. Many insurance carriers approve payment for reconstructive surgery and reject it for cosmetic surgery. The same procedure might be considered cosmetic by one insurer and reconstructive by another. PXE International can provide a letter for you to accompany your request for coverage. In this letter, PXE International clearly states that only surgery can mitigate the skin effects of this congenital disorder. People seeking plastic surgery for PXE are seeking to correct an abnormal structure, affected by disease.

Because PXE is a congenital condition, corrective procedures should be defined as reconstructive surgery. However, authorities sometimes reject claims at first because the loose skin of a person with PXE does continue to carry out its function. Careful appeals and, when necessary, involvement of one's state insurance commissioner, have worked to gain insurance coverage for those with PXE who have been willing to keep fighting.

The bottom line is that PXE International is able to help you get coverage.

Key Facts

- **PXE is a congenital**, or inherited disease, and often causes loose, lax, folds of skin on the neck, under the arms, and in the groin.
- **Reconstructive surgery can help** make the skin folds less uncomfortable and prevent infection.
- These surgeries are often **major surgery** so you should consider them carefully.
- These surgeries can be covered by insurance, and PXE International is able to help by **writing appeal letters** if you are denied.