

## **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.

## **Why Study Testicular Microlithiasis?**

While skin and eyes are the most affected and well documented aspects of pseudoxanthoma elasticum (PXE), we know that other organs are sub-clinically affected. This means that there is mineralization in these organs, but they make no difference to the organ's function, or to the individual's health. We were contacted in 2004 by the parent of a boy who had testicular ultrasonography because he was experiencing pain. The radiologist found testicular microlithiasis. Microlithiasis are minute, tiny, pieces of mineral – usually calcium, but can be made of magnesium and other minerals as well. Testicular microlithiasis means small clusters of calcium have formed in the testicles. Because a number of studies show a relationship between testicular microlithiasis and testicular cancer, this was a concerning finding for the parent and the radiologist. Because we had heard many women complain of being diagnosed with calcifications in their breasts upon imaging, we wondered if we were seeing the same effect in men.

## The Study

We wanted to see if this discovery of testicular microlithiasis was common to men affected by PXE in general. We recruited and worked through a consenting process with eight men (ages 29 to 56 years old) and one boy (13 years old). The thirteen-year-old had an additional process in which he assented, and his parents consented to the study. Testicular ultrasonography (US) was performed on the nine individuals.

Two radiologists reviewed the US images by consensus for testicular microlithiasis, testicular masses, and additional testicular abnormalities. Testicular microlithiasis was judged to be classic when at least five microliths were seen on a single US image and to be limited when fewer than five microliths were seen on all obtained US images. Urologic physiologic examinations were also performed. A history and/or symptom of testicular disease also were recorded at the time of examination.

Shortly after conducting these ultrasounds, two additional men, aged 48 and 59 years, another 13-year-old boy offered to share their ultrasound images. We also obtained an autopsy donation at this time, and we performed histopathologic testicular analysis. This means we studied the tissue.

This brought our total to 12 participants. 11 (92%) had classic and one (8%) had limited testicular microlithiasis. None of the 12 participants had evidence of testicular cancer malignancy at US or on physical examination. The tissue (histopathologic) analysis at autopsy revealed intratubular microlithiasis without the calcification of elastic fibers in arterial walls that is characteristic of skin (cutaneous) PXE.

## Conclusion

These results suggested an association between PXE and testicular microlithiasis. It is likely that boys and men with PXE have testicular microlithiasis and likely do not have to worry about the microlithiasis being associated with cancer.

### Key Facts:

- Males who are affected by PXE likely have **microcalcifications in their testicles.**
- These are likely **not associated with cancer**, as they might be in men who do not have PXE.

### Original paper:

*Bercovitch RS, Januario JA, Terry SF, et al. Testicular microlithiasis in association with pseudoxanthoma elasticum. Radiology. 2005;237(2):550-554. doi:10.1148/radiol.2372041136*