Mammography

Summary of the paper:
Mammographic Findings in Pseudoxanthoma Elasticum**

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.

Mammograms detect mineralization
Over the years, there were isolated case reports of arterial and skin mineralization within the breasts of women affected by PXE, however no studies of mammography in PXE had been undertaken until PXE International decided this was important in 2003. The information here is still accurate and useful today.

PXE International gathered the mammograms of 51 women with confirmed PXE, defined as women having a positive skin biopsy and angioid streaks. We partnered with a radiologist at a hospital-based breast imaging center and compared the 51 mammograms to age-matched mammograms from 109 women who had been seen at the center. Each mammogram was examined by two radiologists who did not know whether they were looking at the mammogram of a woman with PXE or a woman without PXE. The radiologists noted characteristics such as breast density, calcifications, skin thickening, and vascular calcification. The results were statistically analyzed. In order to examine some more carefully, breast biopsy specimens from five patients with PXE were examined by a pathologist who did not participate in examining the mammograms, so that they were not influenced by the results of that analysis.

PO Box 189 • Damascus, MD 20872 • 202.362.9599
sterry@pxe.org • pxe.org

One in seven women affected by PXE had at least three of these signs: skin thickening, skin calcifications, breast microcalcification, and/or vascular calcification. There was no particular pattern of microcalcification that was either diagnostic or suggestive of PXE, although one third of women with PXE demonstrated both vascular and breast microcalcification, a combination that was not observed at all in the control group. Thickening of the skin can have several causes including leukemia, scarring, infection, or inflammation. However, if the woman does not have any of these conditions then skin thickening in the axillary (armpit) area appears to be the cause and that is a sign of PXE.

Breast Biopsies

Four of the breast biopsies showed fibrocystic disease, the presence of benign lumps in the breast, and two showed calcifications of the cells and layers associated with breast ducts and glands. Elastic tissue is present in small quantities around ducts within vessel walls in normal breast tissue, and in increased amounts around ducts in benign breast disease. It is possible that breast microcalcification seen around ducts may be the result of calcification of this elastic tissue, although this hypothesis could not be confirmed on any of the biopsies available for this study. One biopsy did show calcifications that were "typical" for PXE, affecting not only the skin and arteries right below it, but also the deep, soft tissue of the breast itself. The remaining cases showed calcification typical of fibrocystic disease, as mentioned earlier.

It appears that PXE is associated with a significantly increased incidence of breast microcalcifications, vascular calcification, axillary skin thickening and microcalcifications, and that although no particular mammographic finding is diagnostic of PXE, the finding of axillary skin abnormalities or any three of the above findings in mammography might suggest PXE in a distinctive diagnosis. The majority of breast calcifications in PXE have no effect on the breast, including breast feeding.

The bottom line: bring this Bulletin, or the original paper, to your radiologist. if your radiologist sees microcalcifications of the breast in a routine mammogram, it is highly likely that it is caused by PXE and no follow-up, including a biopsy or needless waiting and worrying, is needed.

**The original paper was published in the Journal of the American Academy of Dermatology, March 2003, Pages 359 – 366.

Authors:
Lionel Bercovitch, MD[1,4], Barbara Schepps, MD[2], Susan Koelliker, MD[2], Cynthia Magro, MD[3], Sharon Terry, MA[4], and Mark Lebwohl, MD[5]
Departments of Dermatology [1] and Diagnostic Imaging[2], Rhode Island Hospital and Brown University School of Medicine, and The Anne C. Pappas Center for Breast Imaging[2], Providence, Rhode Island, Department of Pathology[3], Harvard Medical School and Pathology Services, Inc.[3], Cambridge, Massachusetts, PXE International, Inc[4], Washington, DC, Department of Dermatology[5], Mount Sinai Hospital and Mount Sinai School of Medicine, New York, New York.