Mammographic Findings in Pseudoxanthoma Elasticum

The original paper was published in the Journal of the American Academy of Dermatology, March 2003, Pages 359 – 366. Reprints are available from PXE International. This summary was written by Holly Briggs for the Spring Summer 2003 MemberGram.

Authors:
Lionel Bercovitch, MD1,4, Barbara Schepps, MD2, Susan Koelliker, MD2, Cynthia Magro, MD3, Sharon Terry, MA4, and Mark Lebwohl, MD5

Departments of Dermatology1 and Diagnostic Imaging2, Rhode Island Hospital and Brown University School of Medicine, and The Anne C. Pappas Center for Breast Imaging, Providence, Rhode Island2, Department of Pathology, Harvard Medical School and Pathology Services, Inc., Cambridge, Massachusetts3, PXE International, Inc., Washington, DC4, Department of Dermatology, Mount Sinai Hospital and Mount Sinai School of Medicine, New York, New York2, 3 Dr. Magro is currently with the Department of Pathology, Ohio State University College of Medicine, Columbus, OH.

The mineralization of elastic tissues within the body, particularly that of the skin, middle layer of the arteries, and the membrane behind the eye, called the Bruch’s membrane, is characteristic of pseudoxanthoma elasticum (PXE). As a result, PXE sometimes results in skin changes, arterial problems, and loss of central vision. There have been 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 this study. Mammography uses radio waves to detect changes in the soft tissue of the breast. This study was undertaken because women affected by PXE repeatedly reported to PXE International that their clinicians had observed calcifications in their mammograms. Mineralization is associated with PXE and there has been no analysis of a large number of mammograms.

These researchers compared the mammograms of 51 women with confirmed PXE (‘confirmed’ is defined as having a positive skin biopsy and angioid streaks) to the blindly selected mammograms of 109 women from a hospital-based breast imaging center. Each mammogram was examined by two radiologists who noted characteristics such as breast density and calcification, skin thickening, and vascular calcification. Then 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, and who was therefore not influenced by the results of that analysis.

Nearly 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 in the control group. Thickening of the skin can have several causes including leukemia, scarring, infection, inflammation, and skin lesions. However, if the woman does not have any of these conditions then skin thickening in the axillary (armpit) area appears to be a sign of PXE.
Four of the breast biopsies showed fibrocystic disease, the presence of benign lumps in the breast, and two showed calcification 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 can be suggested that breast microcalcification seen here 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 are benign. It is hoped that a further study of breast biopsy material might be helpful in understanding the basis of PXE mammographic findings reported in this study.

ACKNOWLEDGMENTS
The authors acknowledge the invaluable assistance of: The office staff of PXE International, Inc. in recruiting volunteers and tracking down mammograms; James Campbell, MD, MS, who provided invaluable statistical advice; Lisa Goldstein, MD and H. Louis McCombs, MD, for providing breast biopsy material; and Leslie Robinson-Bostom, MD and Rohini Sahuja, MD for assistance with photomicrography.