



Testicle Study Shows Additional Area of Mineralization in PXE Patients

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An article recently published in the medical journal *Radiology* confirmed the association of PXE and testicular microlithiasis. The article reported on a study conducted at Rhode Island Hospital in Providence, Rhode Island, USA. Lionel Bercovitch, M.D., was the lead researcher for the study, and Robert S. Bercovitch, M.D., was the lead author for the article. Others who worked on the study and were co-authors of the study were: Damian E. Dupuy, M.D., Alan D. Podis, M.D., Jennifer A. Januario, M.D., Sharon F. Terry, M.A., and Kim Boekelheide, M.D.

Testicular microlithiasis is a condition in which very small mineral deposits (microliths) are distributed randomly in the testicles. The condition is asymptomatic (causes no symptoms), so it is typically discovered during testing for other testicular anomalies.

PXE International became aware of this issue when a 13-year-old boy with PXE visited his physician, complaining of testicular pain. Ultimately, ultrasound images were taken of his testicles, and they revealed the presence of testicular microlithiasis. This discovery initially was of some concern because this finding is sometimes associated with testicular cancer (although there are ample published data showing this to be a normal finding in many males.) On the other hand, the presence of microliths in males with PXE as well as similar lesions found by ultrasound in other organs in both male and female PXE patients might be related to microscopic deposits of calcium and other minerals in elastic fibers found in PXE patients. Accordingly, PXE International and Rhode Island Hospital supported this study to explore the possibility of an association between PXE and testicular microlithiasis.

There was a total of 13 participants studied, all members of PXE International's research registry. Of this group, nine males were recruited to be studied prospectively, while three, having recently undergone testicular ultrasonography (ultrasound imagery), were studied retrospectively. The final participant was studied using tissue donated at the time of his autopsy. The ages of the participants varied from 13 to 56.

The prospectively studied participants were analyzed in three stages. First, the team reviewed each participant's medical history, particularly his urologic history. Then each participant underwent a physical examination of his testicles and the surrounding area. Finally, ultrasound images were taken of each participant's testicles and then analyzed.

The retrospective study of the three other participants (as well as the deceased participant) was limited to an analysis of earlier ultrasonography.

The results were clear: all 13 participants had testicular microlithiasis in varying degrees.

The meaning of the results is less clear. Notwithstanding the reported association of testicular microlithiasis and testicular cancer, it appears to occur normally (or in the absence of obvious testicular disease) in PXE and may actually be a hallmark of PXE. Therefore, testicular microlithiasis does not appear to be the risk factor for testicular cancer for males with PXE that it may be for the males in the general population. There is no known association between PXE and testicular cancer. Further, it may be that testicular microlithiasis merely represents another manifestation of PXE.

The microliths, the mineralized deposits, are similar to deposits found elsewhere in PXE patients—the kidneys, spleen, pancreas, skin, circulatory system and heart as well as in female breasts. Therefore, it is possible that the presence of testicular microlithiasis in males with PXE is related to the

mineralization of some elastic fibers found in individuals affected by PXE of both genders.

The authors recommend that males with PXE with confirmed testicular microlithiasis remain under long-term observation by ultrasonography and physical examination.