

PXE and the Eye

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One of the body systems affected by pseudoxanthoma elasticum (PXE) is the eye. It is thought that most affected individuals have angioid streaks. Other changes in the eye, called "peau d'orange", appear to be present before angioid streaks, so that younger members of a family can be screened for minor eye changes before angioid streaks appear. Angioid streaks are irregular, red to brown to gray lines that radiate mostly from the area around the optic nerve. Behind or underneath the retina is the retinal epithelium, which is attached to the lining of the eye by a thin layer called "Bruch's membrane" (named after the German anatomist who discovered it). This membrane is an elastin rich layer of connective tissue. Elastic tissue in the body of individuals affected with PXE can become mineralized. Sometimes these areas break or crack. Breaks in Bruch's membrane correspond to the angioid streaks. Some have described these breaks as looking like "mud cracks" or "cracks in porcelain".

The presence of angioid streaks does not affect vision. However, sometimes blood vessels from the underlying choroid (a blood vessel rich layer) can grow through the cracks. It is possible for these tiny blood vessels to leak and bleed. If this hemorrhage is in the area of the macula, the place where central vision is focused, there can be permanent central vision loss.

A person with PXE rarely, if ever, becomes blind. They can lose enough central vision however to become legally blind, unable to see except through peripheral vision.

Sometimes laser surgery is suggested, if the leaking occurs a secure distance from the central retina area. The small blood vessels are photocoagulated (sealed) using the laser. Even with the best methods it is successful in about 50% to 60% of cases. Reoccurrence is common. There is, therefore, some controversy about the use of laser surgery for PXE patients.

It is important that people affected with PXE have a diligent, annual, complete ophthalmologic examination. This will necessarily include dilating the pupil so that the ophthalmologist can look for angioid streaks. Sometimes a retinal specialist will also suggest that photographs of the retina be taken for documentation and for comparison in the future. Rarely, a fluorescein angiogram may be necessary, if the retinal specialist suspects that abnormal blood vessels are growing underneath the retina or if there is unexplained leakage of fluid or blood under the retina. Fluorescein angiograms are not part of a routine or complete medical examination of the eyes and should not be done 'annually' or 'routinely'. Every affected individual should have an annual examination with an ophthalmologist.

If an affected individual is at risk for retinal hemorrhage, she or he may want to use an Amsler Grid. This is a grid that may assist in identifying an area of visual distortion that might be caused by a new leak or bleed. If any new area of bleeding is discovered, the person should immediately report this to her or his ophthalmologist. Instructions for using the

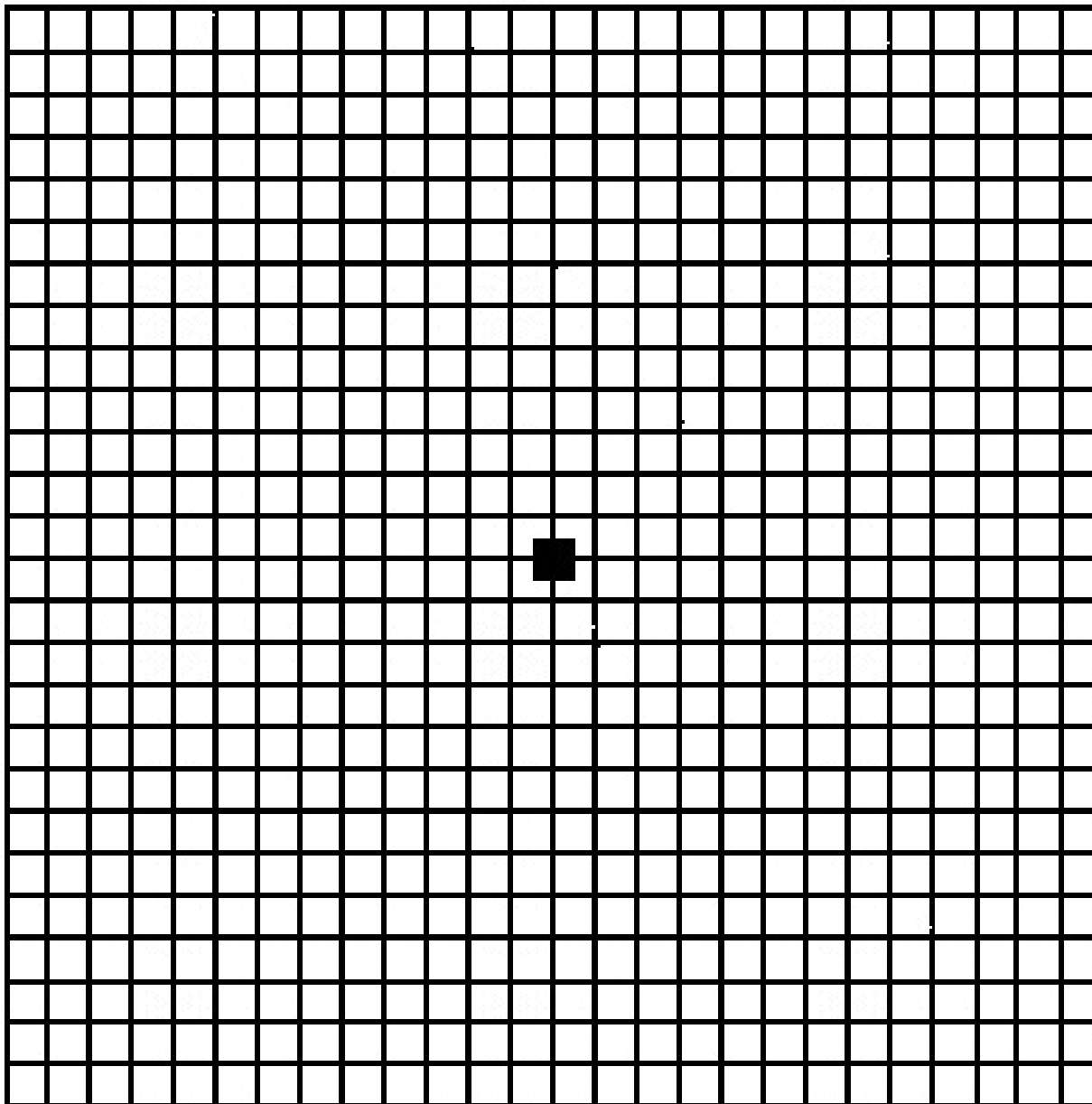
Amsler Grid can be found on the web at:
www.vrmny.com/amsler.htm.

It has been suggested that people with PXE not engage in activities that might cause direct eye injury. This would include football, boxing, heavy weight lifting or deep sea diving. However, good aerobic exercise is recommended.

If a person loses central vision, contacting a low vision clinic may be helpful. These clinics can help the affected individual overcome some of

their disability and learn to accommodate what they cannot overcome, both for mobility and for use of magnifying devices and reading aids.

PXE affects the skin, heart, vascular and gastrointestinal systems in some cases. There may be special issues for women and pediatric patients. It is recommended that individuals educate their physician. PXE International can help you do this.



Amsler Grid

The bulletins and newsletters of PXE International, Inc. are also available on audiocassette .