

One of the primary body systems affected by pseudoxanthoma elasticum (PXE) is the eye. Just about all people affected by PXE have some sign or symptom in their eyes. The first sign, called "peau d'orange", appears to be present before the second and more noticed sign, called angioid streaks. 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. In the periphery of the body, elastic tissue can become mineralized. Sometimes these areas break or crack. Breaks in Bruch's membrane are called angioid streaks. Sometimes these breaks are described as looking like "mud cracks" or "cracks in porcelain".

The presence of angioid streaks does not affect vision. You should begin to see an

Important Terms

Peau d'orange: a mottled, "skin of an orange" look on the retina
Angioid streaks: irregular, red to brown to gray lines that radiate mostly from the area around the optic nerve
Bruch's membrane: layer underneath the retina
Choroid: the vascular layer of the eye
Macula: part of the retina where central vision is focused

ophthalmologist annually once you have been diagnosed with PXE. They will monitor any changes in your eye. And, you are your own best detective when it comes to whether or not you are having a retinal bleed.

A retinal bleed happens when blood vessels from the underlying choroid (a blood vessel rich layer) grow through the cracks. It is possible for these tiny blood vessels to leak and bleed. If this happens, you can usually detect it early by looking at an Amsler grid, like the one on the next page.

If you catch your bleed early, you should get an appointment with your retinologist immediately. Your retinologist will likely suggest injecting the area of the bleed with an anti-angiogenesis medication. This helps most people to retain their vision. In rare cases, people lose enough central vision however to become legally blind, unable to see except through peripheral vision. This used to be the norm, and now most people keep their vision because of better and better treatments.

Long ago, and for many older PXEers, laser surgery was used if the leaking was a way from the central retina area. This was not a useful treatment, it led to damage of the retina and did not stop the blood vessels from growing the way the anti-angiogenesis medication does.

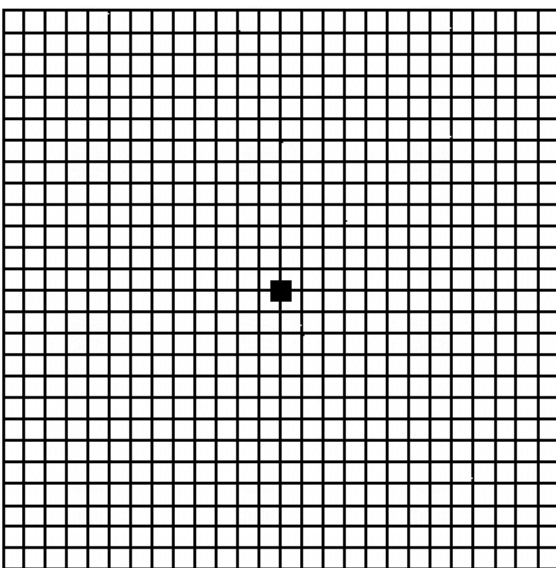
It is important that you have an annual, thorough ophthalmologic examination. This must include dilating your pupil so that the ophthalmologist can look for angioid streaks. Sometimes a retinal specialist will also suggest that photographs of the retina be taken for documentation and for comparison in the future. Some tests, like a fluorescein angiogram or optical coherence tomography are used to give the retinologist a better sense of what is happening beneath your retina. These are not routine tests but do help when you think you see a bleed.

Bottom line – use the Amsler grid daily. This grid may assist in identifying an area of visual change, or distortion, that might be caused by a leak or bleed. If you see an area of distortion, you should immediately report this to your retinologist.

It has been suggested that people with PXE not engage in activities that might cause direct eye injury without wearing eye protection. This would include football, boxing, and basketball. In addition, performing the valsalva maneuver – holding your breath and straining – such as heavy weightlifting could cause an eye bleed. Aerobic exercise is recommended for overall health, however.

If you lose your central vision, contacting a low vision clinic may be helpful. These clinics can help you overcome some of your disability and learn to accommodate what you cannot overcome, improving your mobility and your use of assistive devices.

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. We have other bulletins available to you. Contact us at info@pxe.org or 202.362.9599.



Amsler Grid

1. Tape the Amsler grid at eye level where light is consistent and without glare.
2. If you have glasses, put them on, and cover one eye.
3. Fix your gaze on the center black dot.
4. Keeping your gaze fixed, try to see if any lines are distorted or missing.
5. Mark the defect on the chart.
6. TEST EACH EYE SEPARATELY.
7. If the distortion is new or has worsened, arrange to see your eye doctor at once.
8. Always keep the Amsler grid the same distance from your eyes each time you test.