

Can eye complications of PXE be reversed?

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Vision loss is a common symptom of pseudoxanthoma elasticum (PXE) primarily as a result of changes in the eye, particularly peau d'orange, and angioid streaks. These are found in almost all people affected by PXE. These changes begin at an early age. The Bruch's membrane (BM), an elastin-rich layer separating the retinal pigment epithelium from the choroid layer, becomes thickened and calcified as people get older.

These changes to the eye occur as breaks in Bruch's membrane develop due to calcification. The break in Bruch's membrane itself has no effect on visual function. As PXE progresses, blood vessels grow through this membrane. This is called choroidal neovascularizations (CNV). Fluid & blood may leak when these blood vessels rupture. These are commonly referred to as "bleeds" and they cause visual distortion.

As these hemorrhages beneath the retina (bleeds) heal, scarring develops. If this is in the macula, the center of the retina, then it can cause central vision loss.

PXE is similar to the much more common age- related macular degeneration (AMD). With the growing number of people affected by AMD, more research and subsequent treatment options have become available to the benefit of PXErs. However, the concern for PXE affected individuals is that the changes to the retina occur at a much younger age than in AMD, requiring treatment earlier in life, with many more treatments and over a longer period of time, often several decades more than with AMD.

Current Treatment Options

CNVs occur before the development of a scar. They are the only PXE complication that can be treated at this time. Currently, it is treated by injections of vascular endothelial growth factor (VEGF). These are considered the most effective in preventing or limiting scar formation and the resulting vision loss. Recent studies with both AMD and PXE affected individuals have shown the VEGF intravitreal injections, Lucentis (ranibizumab), Avastin (bevacizumab) & Eyelea (aflibercept) appear to be comparable in terms of safety and effectiveness.

Previously used treatments include laser photocoagulation (hot laser) and photodynamic laser (cold laser) therapy with verteporfin. Due to scarring and a high rate of reoccurrence, these treatments have largely been replaced by the VEGF inhibitors.

Future Treatment Options

The Macular Degeneration Association has recently reported that a topical treatment for neovascular AMD is “within sight”. Intravitreal injections might be replaced with a topical application, such as eye drops, in the future.

Can eye complications from PXE be reversed?

The VEGF inhibitors, Lucentis, Avastin, and Eyelea, have been reported to be successful in vision improvement/restoring vision loss in PXE when people receive the injection in a timely manner. There is no evidence of reversal of retinal damage in humans at this point in time.

There is some research in animals to see if retinal cells can regenerate. Currently there are no therapies that can reverse the eye damage from PXE.

Areas of Research Interest

Gene therapy is an area of interest for some PXE affected individuals and their families. While there is no research being conducted at this time involving PXE, some ongoing studies offer insight into future research possibilities.