CASE REPORT

Pseudoxanthoma elasticum

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Case Presentation

A 57-year-old male presents to primary care with sub-acute vision changes. The vision changes involving central vision had been noted for several weeks before presenting to the clinic. He denies trauma or eye pain. Review of systems is also negative for neurological symptoms including facial weakness and headaches. The patient has no significant past medical or surgical history. He reported taking only an over the counter non-steroidal anti-inflammatory drug as needed for chronic back pain. He has no known allergies, and family history is noncontributory.

His vital signs are within normal limits. The physical exam including neurologic is unremarkable. Visual acuity was 20/80 in the right eye and 20/20 in the left. Nondialated fundoscopic exam was limited and he was urgently referred to ophthalmology. Ophthalmologic exam noted bilateral angioid streaks in the right eye with choroidal neovascularization along with macular degeneration. He was started on Aflibercept injections in the right eye. Based on the eye findings the ophthalmologist suspects the diagnosis of pseudoxanthoma elasticum.

Brain and neck MRI/MRA demonstrates multiple small foci of T2 and FLAIR hyperintensity, primarily in the subcortical cerebral white matter. Similar findings are reported in patients with mixed connective tissue disorders such as pseudoxanthoma elasticum.

The patient was also referred to dermatology. Several characteristic skin lesions are noted and a punch biopsy of a left neck lesion demonstrated basophilic, irregular and granular elastic fibers in the dermis, consistent with diagnosis of pseudoxanthoma elasticum.

Discussion

Pseudoxanthoma elasticum (PXE) is an autosomal recessive disease that results in abnormal elastic tissue and calcifications. Although typically familial, sporadic mutations can occur.[1] The causative defect is a mutation in the ABCC6 gene on chromosome 16 that encodes an ATP binding cassette transporter.[2-4] The prevalence of PXE is around 1 in 25,000 people.

Onset of the disease varies from childhood to early adulthood although most affected patients will display subtle cutaneous findings during childhood. The main organ systems involved include the skin, eyes, and vascular system.[5-7]

The classic cutaneous findings are collections of small yellow-orange papules in flexural areas, such as the sides of the neck, antecubital fossae, axillae, and groin. They also commonly occur on the inner lower lip and periumbilical regions. The papules often coalesce into plaques. The papules are asymptomatic and are usually identified incidentally. Skin biopsy confirms the diagnosis. Skin findings occur in 80 percent of individuals before the age of 20.

The main eye finding is angioid streaks, which are tears in the Bruch's membrane. Angioid streaks appear as dark asymmetrical lines stemming from the optic disk. This is seen in more than 90% of patients with PXE. Additional complaints include macular degeneration and retinal deformities, including retinal hemorrhage, that can contribute to central vision loss.[8]

Common vascular findings are accelerated atherosclerosis due to calcifications. This can lead to cerebrovascular disease, renovascular disease, and myocardial infarction. Lastly, approximately 10-15% of patients with PXE develop gastrointestinal hemorrhaging.[8]

There is no fully effective treatment for PXE. Patient with PXE should avoid the use of tobacco and smoking due to vascular dysfunction. Because of the increased risk of retinal hemorrhage, contact sports should be avoided. Aspirin and nonsteroidal anti-inflammatory agents are not recommended due to increased risk of retinal and GI hemorrhages. Statins are often prescribed and if started early may prevent abnormal vascular calcifications. All patients with PXE and their families should be referred for genetic counseling.[7-8]

Conclusion

Pseudoxanthoma elasticum is an autosomal recessive disease that results in abnormal elastic tissue. Main manifestations of the disease include skin, vascular, and ocular findings. Patients with PXE should have close ophthalmologic monitoring due to a high risk of vision loss.

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