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Pseudoxanthoma elasticum

Pseudoxanthoma elasticum (PXE) is the name given to a group of connective tissue disorders that affects the elastic tissue of the skin, blood vessels, and the eyes. It is also known as Gronblad–Strandberg syndrome.

The basic fault in PXE appears to relate to genetic abnormalities on chromosome 16p13.1. The disease involves the transporter genes MRP6 or ABCC6 in at least some affected families. There is good evidence that, in affected individuals, collagen and ground substance are abnormal. Calcium then accumulates in abnormal elastic fibres in the skin, blood vessels, eyes and heart.

It affects about 1 in every 160,000 people. There are a number of subtypes of PXE, each with a slightly different clinical presentation and pattern of inheritance. Two are inherited as an autosomal dominant characteristic; this means half the children of an affected individual are affected. Three types of PXE are autosomal recessive (1 in 4 children affected).

Clinical features

Individuals with PXE have distinctive skin lesions, eye abnormalities, and changes of blood vessels but these may vary in distribution and severity

Skin

Pseudoxanthoma elasticum



The skin lesions of PXE are characteristic (but rather difficult to photograph). They consist of small, yellowish bumps in rows or a lacy pattern, which may join to make large patches. The skin is soft, lax and slightly wrinkled. The patches may be slightly pebbly in appearance, which has been described as cobblestoned.

The common sites affected are the sides of the neck, below the collar bones, the armpits, abdomen, groins, perineum and thighs.

These skin changes may develop in childhood but usually develop in early adulthood (before the age of 30). They may, in some cases, first appear in old age. Once they appear, they usually persist unchanged indefinitely.

Similar changes may occur in the soft palate, inside the lips, and lining of the stomach, rectum and vagina.

The diagnosis of PXE is usually straightforward as [skin biopsy](#) findings are characteristic.

Heart and blood vessels

The arteries of the heart and throughout the body may be affected. This may lead to pain in the legs on walking (claudication), stroke, heart pain (angina) and cardiomyopathy (heart muscle disease). Mitral valve prolapse occurs in about 5–8% of cases.

Eye

Vascular lesions known as angioid streaks affect the retina of most patients with PXE. They usually appear between 20 and 40 years of age. Angioid streaks may lead to progressive visual failure and occasionally blindness.

Obstetric risk

Patients with PXE have an increased risk of miscarriage in the first three months of pregnancy. Stretch marks develop during pregnancy in virtually all patients.

Treatment

- The most important aspect of treatment is to ensure that complications from blood vessel involvement are prevented or dealt with speedily by the appropriate specialist. Regular follow-up with a specialist vascular surgeon and/or cardiologist is recommended.
- There is currently no effective treatment for the skin lesions, but the appearance may be improved by plastic surgery.
- Restriction of dietary calcium has been tried with some benefit, but this is controversial.
- Laser photocoagulation may be helpful in preventing further bleeding at the back of the eye.
- As there is a risk of passing this condition on to the next generation, genetic counselling may be helpful.

Related Information

Other websites:

- [PXE International Inc.](#)
- [The PXE Support Group](#) – PIXiE (UK)
- [National Assn for Pseudoxanthoma Elasticum Inc.](#) (NAPE)
- [Pseudoxanthoma elasticum](#) – emedicine dermatology, the online textbook

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DermNet does not provide an on-line consultation service.
If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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