Your Child and PXE

A PXE International Guide

Introduction

Learning that your child has pseudoxanthoma elasticum (PXE) can be overwhelming. Parents often feel shocked, confused, sad, and uncertain about what the future will hold. PXE International understands these feelings deeply—we were founded by families who have walked this same path.

This guide offers clear, current medical understanding and a message of reassurance. PXE affects each individual differently. Most people with PXE live long, full lives. Medical knowledge has advanced dramatically over the past three decades since PXE International's founding, and effective ways now exist to monitor, prevent, and treat many complications once thought inevitable.

What is PXE?

Pseudoxanthoma elasticum (PXE) is a rare inherited condition in which certain elastic tissues in the body become abnormally mineralized. It most often affects the skin, eyes, and mid-sized arteries, and less commonly the gastrointestinal system. PXE usually results from pathogenic variants (changes) in both copies of the gene *ABCC6*. This gene helps the body produce inorganic pyrophosphate, a substance carried in the blood that helps prevent unwanted mineralization (tiny calcium deposits) in elastic tissues. When an individual lacks a properly functioning copy of the gene, mineralization can occur in specific areas of the body. This is not the case for carriers, who have one working copy and one damaged copy.

Individuals with PXE are born with it, but its course is typically slow and highly variable, so it is not always diagnosed. Some people develop only mild skin findings, while others may experience changes in vision or arteries later in life.

PXE affects both males and females and occurs worldwide, in all populations. It is estimated to occur in approximately 1 in 25,000 to 1 in 50,000 people—though many remain individuals remain undiagnosed.

Skin Findings

Skin changes are often the first visible sign of PXE and can appear in childhood. While these changes usually do not cause pain or health issues, they can indicate the presence of the condition.
Typical features include:

- Small yellow-orange bumps (1–5 mm) called papules that may merge (called plaques) and eventually cause wrinkled areas.
- Commonly appear on the sides of the neck, underarms, inside elbows, behind knees, or in the groin. Sometimes they appear around the navel.
- The skin may gradually become loose or wrinkled in these areas over time.

Dermatologists may confirm the diagnosis of PXE with a small skin biopsy. Although these changes are mostly cosmetic, they can be emotionally difficult. No medical treatment is needed. Some people may opt for reconstructive surgery to remove the lax skin. It appears that reconstruction lasts about 20 years.

Eye (Ophthalmologic) Findings

The eyes are the most medically important area to monitor in PXE. The retina—the light-sensitive layer at the back of the eye—contains delicate elastic tissue that can show early signs of PXE long before vision is affected. Most children have some signs of PXE in the eye, but will have no symptoms for decades (or ever, if we are successful in establishing treatments for PXE).
Common findings include:

- Peau d’orange: a subtle 'orange-peel' pattern seen by an eye doctor, sometimes appearing in childhood. (Note this does not refer to skin in PXE.)
- Angioid streaks: fine lines that radiate from the optic nerve area beneath the retina. These are common in PXE and do not, by themselves, cause vision loss.
- Retinal bleeding (choroidal neovascularization): in some people, new blood vessels can grow through the weakened retinal layer and leak, causing blurring or distortion in central vision.

The good news: Today, most retinal bleeding can be effectively treated with medications known as anti-VEGF injections (for example, Eylea®, Lucentis®, or Avastin®). These treatments often stabilize or even improve vision.

Eye care recommendations:

- Schedule a baseline exam with a retina specialist soon after diagnosis.
- Follow up yearly, or more often if advised.
- Report any sudden changes in vision immediately; early treatment makes a big difference.

- Encourage protective eyewear to reduce risk of direct eye trauma (e.g., boxing, baseball, or contact sports).

Since the advent of antiangiogenesis injections, most people with PXE retain useful vision throughout life and do not lose peripheral (side) vision.

Vascular Findings

PXE can affect medium-sized arteries by causing tiny mineral deposits in the elastic part of the artery wall. This can sometimes reduce blood flow later in life, especially to the arms and legs. Serious circulation problems are uncommon in children, but good lifelong preventive care is important.

Recommended care:

- Periodic blood pressure checks and lipid (cholesterol) testing – in children only as often as the pediatrician recommends, no need to add extra tests because of PXE.
- Encourage regular physical activity such as walking, biking with a helmet, swimming, or any way your child wants to move and play. Disregard any older references to restricting activities.
- Maintain a heart-healthy diet: plenty of fruits, vegetables, and whole foods, low in saturated fat and added sugars. Do not avoid calcium, despite old recommendations to do so.
- Avoid smoking and secondhand smoke.

Adults with PXE may be monitored for hypertension, peripheral artery disease, or early signs of coronary artery disease. But children do not need to be monitored unless they show symptoms. With good preventive care, many adults avoid significant vascular problems. When we have a treatment for PXE, though it will not reverse any damage, it will stop the progression of the disease. Therefore children will likely never have these symptoms.

Gastrointestinal Findings

Rarely, PXE can cause bleeding in the stomach or intestines. This is uncommon in anyone with PXE but important to recognize.

- If your child experiences vomiting of blood, black stools, or unexplained anemia, notify your doctor immediately.
- Avoid medications that irritate the stomach or increase bleeding risk, such as aspirin, ibuprofen (Motrin®, Advil®), or naproxen (Aleve®), unless specifically prescribed.
- Acetaminophen (Tylenol®) is generally the safest medication for pain or fever.

Genetics and Inheritance

PXE is inherited in an autosomal recessive pattern. This means a person must receive two altered copies of the *ABCC6* gene—one from each parent—to show signs of PXE. Parents who carry one altered copy are carriers and do not have symptoms.
For each pregnancy between two carriers:

- 25% chance the child will have PXE (two altered genes)
- 50% chance the child will be a carrier (one altered gene)
- 25% chance the child will have two typical copies of the gene

Most families have no previous history of PXE, because two carriers have not had a child together. PXE rarely appears in more than one generation.

Genetic testing for *ABCC6* is now widely available. PXE International offers support for families seeking testing, counseling, and inclusion in the PXE Research Registry. Learn more: www.pxe.org.

In some cases, infants and children are found to possess two pathogenic variants of the ABCC6 gene, leading a laboratory or pediatrician to diagnose them with pseudoxanthoma elasticum (PXE). However, without clinical signs, a definitive diagnosis of PXE cannot be made. Additionally, pathogenic variants in *ABCC6* may be associated with other medical conditions.

After the Diagnosis

Receiving a diagnosis can be upsetting and adjusting takes time. PXE International recommends these steps to support your child's health and peace of mind:

- Take a breath. Take several breaths. This is a very slowly progressing condition, and by the time your child has any significant symptoms, there will be treatments available.
- Encourage balanced meals, daily movement, and emotional well-being. But don't make PXE the issue; these are the right things to encourage for any child.
- Encourage sports and activities, and ask them to wear eye protection when playing games where a ball or stick could hit the eye and be detrimental.

Emotional support: It's normal to grieve and worry about the future. Connecting with other PXE families can provide strength, understanding, and practical advice. PXE International offers online forums, family meetings, a Facebook group for parents, another for adult PXEers, a WhatsApp group for young women (18 to 40 years old), and educational webinars.

Medical follow-up:

- Normal annual visits to a pediatrician.
- Annual visits with an ophthalmologist.
- Immediate attention to any new vision symptoms or unusual bleeding.

- Discuss any medications or supplements that would encourage bleeding (blood thinners) with your doctor before use.

Outlook: Children with PXE can expect a normal lifespan. With regular monitoring and current and future treatments, complications can often be prevented or managed. Research is progressing rapidly toward therapies that address the underlying cause of PXE. We have two that we are sure will be effective, and we just need to raise the necessary funding for the clinical trial.

PXE International

PXE International is a nonprofit organization founded by families affected by pseudoxanthoma elasticum. Our mission is to discover treatments and cures for PXE while ensuring that every affected person and family has access to the most current information and compassionate community support.

We invite you to connect, learn, and participate in research that will help build a better future for all people with PXE.

Contact us:
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