

What Is PXE?

Pseudoxanthoma elasticum, (PXE), is an inherited disorder that causes some tissue in the body to become mineralized, that is, calcium and other minerals are deposited in the tissue. This can result in changes in the skin, eyes, cardiovascular system and gastrointestinal system. PXE was recognized over a hundred years ago. A number of significant advances have been made in the past few decades.

What Are the Effects of PXE?

PXE results in a variety of signs and symptoms. The number, type, and severity of signs of PXE are different for each person. Certain effects of PXE can cause serious medical problems while others have less impact. The effects of PXE may include: skin changes; changes in the retina of the eye that may result in significant loss of central vision; changes in the cardiovascular system that may involve calcification of arteries and decreased blood flow in the arms and legs; changes in the gastrointestinal system that may lead to bleeding in the stomach or intestines.

Genetics

The gene associated with PXE is called *ABCC6*. Every living person has the gene called *ABCC6*. It is not true that people affected by PXE have the “PXE gene”; instead they have a change (mutation) in the gene associated with PXE, the *ABCC6* gene.

All of our genes come in pairs, so we all have two copies of every gene. Some genes are dominant, meaning that a change (mutation) in just one copy of the gene will cause a sign or a symptom. Some genes are recessive, meaning that both genes must have a change (mutation) in order to cause a sign or symptom.

PXE is a recessive trait, so an individual will only have PXE if they have changes (mutations) in both copies of *ABCC6*. A person is a carrier of PXE if they have a change (mutation) in only one copy of *ABCC6*.

Inheritance

In the examples below, ‘a’ means there is a change in *ABCC6*, while ‘A’ means there is no change or mutation. By drawing a small chart, called a Punnett square, we can see all the possible combinations of gene pairs.

Symbols Used

A = ABCC6 gene with no change or mutation
a = ABCC6 gene with a change or mutation

AA = not a carrier, doesn't have PXE

Aa = carrier

aa = has PXE

Possible combinations

Note that these are the odds each and every pregnancy. That means you can have one of these combinations occur more than the odds would suggest. Just like you have 50% odds of getting heads or tails, but you can flip a coin and get heads 10 times in a row.

Example 1:

Neither parent has PXE.

Parent 1 is a carrier and is symbolized with Aa

Parent 2 doesn't have PXE and is not a carrier and is symbolized with AA

	A	a
A	AA	Aa
A	AA	Aa

Each of the four boxes represents the odds for each pregnancy so you can see that this hypothetical couple has a 50% chance that a child will not be a carrier (AA) and a 50% chance that a child will be a carrier (Aa)

Example 2:

Neither parent has PXE.

Parent 1 is a carrier and is symbolized with Aa

Parent 2 is also a carrier and is symbolized with Aa

This is the most common scenario by far, and likely why you have PXE.

	A	a
A	AA	Aa
a	aA	aa

Each of the four boxes represents the odds for each pregnancy so you can see that this hypothetical couple has a:

25% (one out of four boxes) chance that a child will not have PXE and not be a carrier (AA);

50% chance that a child will be a carrier (Aa); or

25% chance that a child will be affected by PXE (aa).

Example 3:

One parent has PXE.

Parent 1 is a carrier and is symbolized with Aa

Parent 2 has PXE and is symbolized with aa

This is the reason some families have two generations with PXE, we have never found one with three generations.

	A	a
a	aA	aa
a	aA	aa

Each of the four boxes represents the odds for each pregnancy so you can see that this hypothetical couple has a:

50% (two out of four boxes) chance that a child be a carrier (aA); and

50% chance that a child will be affected by PXE (aa).

Example 4:

Parent 1 has PXE and is symbolized with aa

Parent 2 has PXE and is symbolized with aa

There is no question here, all of the children will have PXE.

	a	a
a	aa	aa
a	aa	aa

Each of the four boxes represents the odds for each pregnancy so you can see that this hypothetical couple has a:

100% (four out of four boxes) chance that a child will be affected by PXE (aa).