

Inheritance and PXE

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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.

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 Punet 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

Possible Combinations

AA = not a carrier, doesn’t have PXE

Aa = carrier

aa = has PXE

EXAMPLES

Example 1:

Parent 1 = **Aa** = carrier

Parent 2 = **AA** = not a carrier, doesn't have PXE

| | | |
|----------|-----------|-----------|
| | A | a |
| A | AA | Aa |
| A | AA | Aa |

Result 1:

50% chance that a child will not have PXE or be a carrier (**AA**)

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

Example 2:

Parent 1 = **Aa** = carrier

Parent 2 = **Aa** = carrier

| | | |
|----------|-----------|-----------|
| | A | a |
| A | AA | Aa |
| a | Aa | aa |

Result 2:

25% chance that a child will not have PXE or be a carrier (**AA**)

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

25% chance that a child will have PXE (**aa**)

Example 3:

Parent 1 = **aa** = has PXE

Parent 2 = **Aa** = carrier

| | | |
|----------|-----------|-----------|
| | a | a |
| A | Aa | Aa |
| a | aa | aa |

Result 3:

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

50% chance that a child will have PXE (**aa**)

Example 4:

Parent 1 = **aa** = has PXE

Parent 2 = **AA** = not a carrier, doesn't have PXE

| | | |
|----------|-----------|-----------|
| | a | a |
| A | Aa | Aa |
| A | Aa | Aa |

Result 4:

100% chance that a child will be a carrier (**Aa**)

Example 5:

Parent 1 = **aa** = has PXE

Parent 2 = **aa** = has PXE

| | | |
|----------|-----------|-----------|
| | a | a |
| a | aa | aa |
| a | aa | aa |

Result 5:

100% chance that a child will have PXE (**aa**)