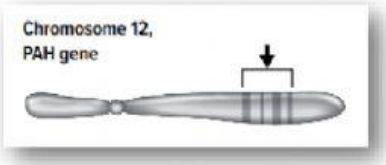
# Genetic disorders

Drag and drop

## Genetic disorders

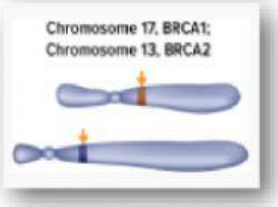
| Defective gene or chromosome | Disorder | Description |
|---|---|---|
|   | | |
| | Breast cancer and ovarian cancer | People with defective BRCA1 and/or BRCA2 genes have an increased risk of developing breast cancer and ovarian cancer. |
|   | | |
| | Phenylketonuria (PKU). | People with defective PAH genes cannot break down the amino acid phenylalanine. If phenylalanine builds up in the blood, it poisons nerve cells. |

LIVEWORKSHEETS




Cystic fibrosis

People with Williams Syndrome are missing part of chromosome 7, including the elastin gene. The protein made from the elastin gene makes blood vessels strong and stretchy.




Williams Syndrome

with defective CFTR genes, salt cannot move in and out of cells normally. Mucus builds up outside cells. The mucus can block airways in lungs and affect digestion.

LIVEWORKSHEETS

**Questions**

What causes cystic fibrosis?

Why do some women get tested to find out if they have a mutation in the *BRCA1* and *BRCA2* genes?

An individual cannot break down phenylalanine. What mutation does the person have?

Is this mutation beneficial, neutral, or harmful, and why?

**Answers**

harmful because nerve damage can result if the disorder isn't treated.

a mutation in the *PAH* gene

a mutation in the *CFTR* gene

Women with a mutation in *BCRA1* or *BCRA2* have a greater chance of developing breast and/or ovarian cancer than other women. Some women want to know if they have the increased risk and may choose to take preventative measures if they do.

LIVEWORKSHEETS