Monogenic Inheritance Patterns

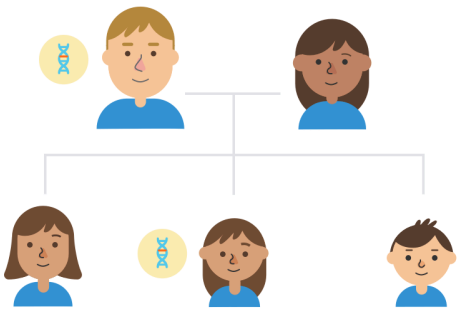
Some health conditions are caused by single genetic variants with a big impact. These are called **monogenic conditions**. One unique aspect of monogenic conditions is that they tend to follow predictable inheritance patterns.

The inheritance pattern of a monogenic condition depends on two main questions:

- **Which chromosome is involved?** If the genetic variant that causes the disease is located on a numbered chromosome (1-22), the inheritance pattern is called "autosomal" (chromosomes 1-22 are called autosomes). If the variant is on the X chromosome, the inheritance pattern is X-linked.
- **How many copies are needed?** If having one copy of a genetic variant is enough to cause disease or increase risk, the condition is considered "dominant." If two copies are needed (one variant from each parent), the condition is considered "recessive."

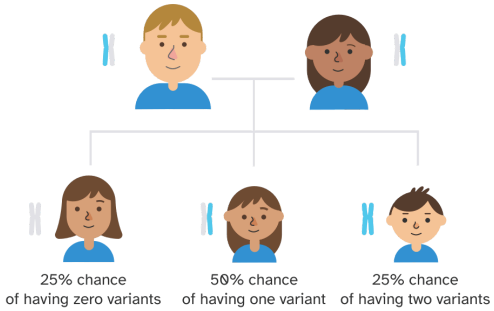
Here are three examples of common inheritance patterns.

Autosomal dominant



- Inheriting a **single genetic variant** is sufficient to cause the condition
- Variants can be passed down from **either parent**
- Each biological child of a parent with the condition has a **50% chance** of having the same condition
- **Examples:** BRCA1/2-related cancer risks, familial hypercholesterolemia

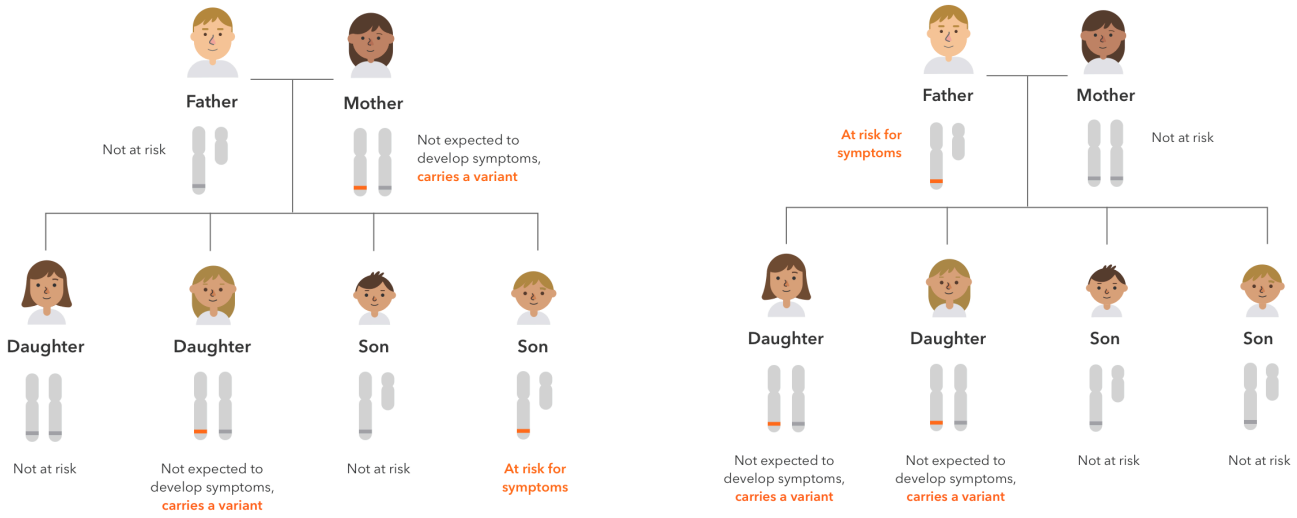
Autosomal recessive



- Must inherit **two genetic variants** (one from each parent) to have the condition
- People with just one variant are called **carriers** and usually don't have symptoms
- If both parents are carriers, each biological child has a **25% chance** of having the condition
- **Examples:** sickle cell anemia, cystic fibrosis

More info on the next page ↓

X-linked



- In the diagram above, the larger chromosome is X and the smaller chromosome is Y.
    - Females typically have two X chromosomes. They pass one of those X chromosomes on to each of their children (both male and female).
    - Males typically have one X chromosome and one Y chromosome. They pass along an X chromosome to each of their daughters and a Y chromosome to each of their sons.
- For **females**, the impact of an X-linked variant depends on how many copies are inherited. If they inherit just one variant, they still have a working copy of the gene on their other X chromosome. As a result, they may have no symptoms or mild symptoms. On the other hand, if they inherit two variants (one on each X chromosome), then they're likely to have the condition.
- For **males**, a single genetic variant is sufficient to cause an X-linked condition, because there's no "backup" copy of the gene. As a result, X-linked conditions tend to be more common in males than females.
- The two family trees above show what happens when a mother (left) or father (right) has a genetic variant that causes an X-linked condition. Try to trace the variant through the family tree and spot the differences in how that variant is inherited.
- **Examples:** G6PD deficiency, Duchenne muscular dystrophy

There are other types of monogenic inheritance, too — including Y-linked and mitochondrial inheritance — plus variations like incomplete penetrance and codominance. You can learn more about these patterns at MedlinePlus.

Note: In the diagram above, we use the labels mother, father, daughter, and son as familiar, shorthand ways to describe family relationships in X-linked inheritance. These terms are meant to reflect how X and Y chromosomes are typically passed down, not how people identify or what roles they hold in their families. We also use the terms female and male to describe birth sex as it relates to chromosomes (XX or XY), because X-linked conditions depend on which sex chromosomes a person has. We recognize that birth sex, gender identity, and family roles are not the same thing, and that not everyone with two X chromosomes identifies as female or as a mother. These labels are used only to describe patterns of chromosome inheritance, not to define people's identities.