Anatomy

Cleveland Clinic

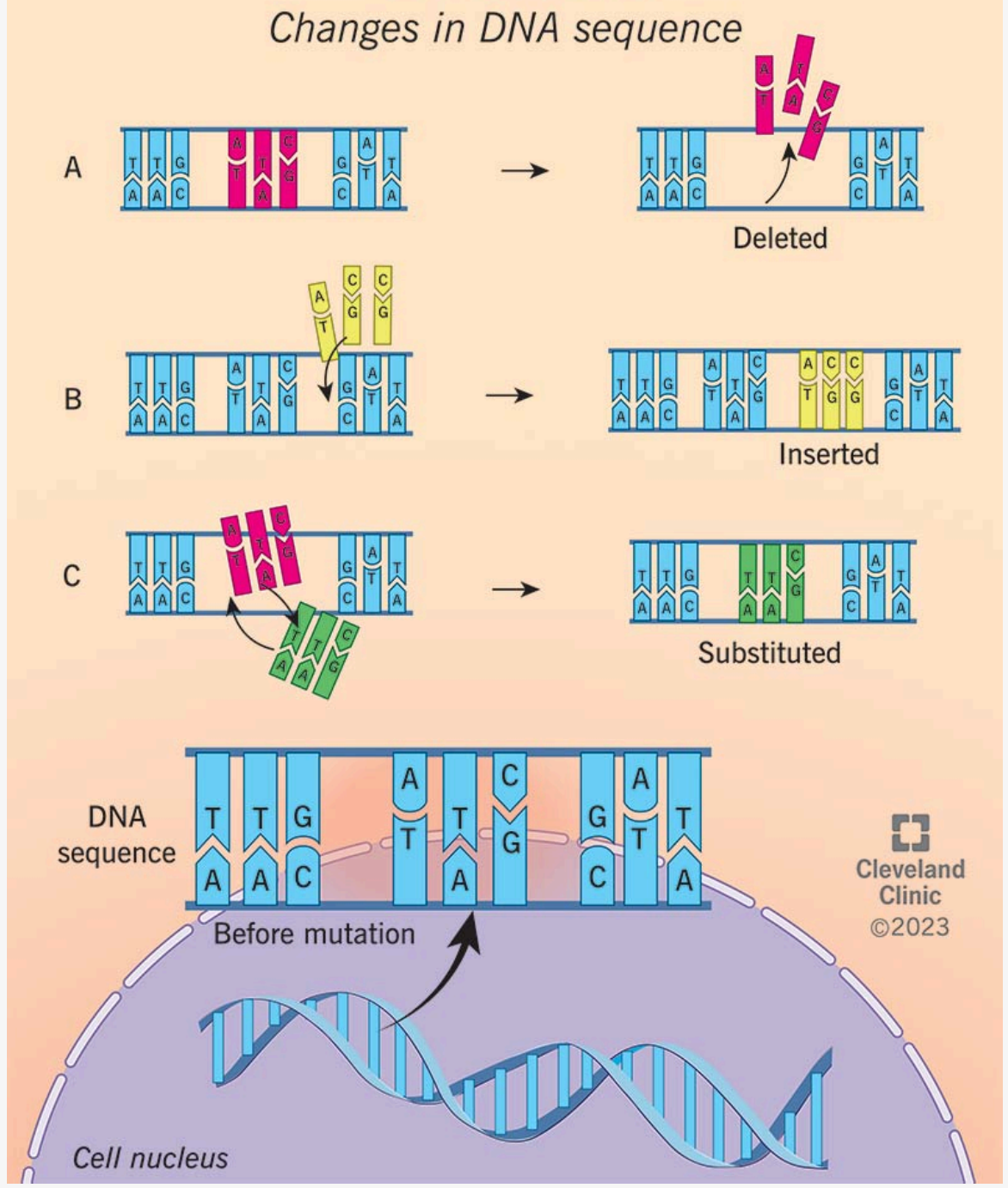
Genetic Mutations in Humans

Genetic mutations are changes to your DNA sequence that happen during cell division when your cells make copies of themselves. Your DNA tells your body how to form and function. Genetic mutations could lead to genetic conditions like cancer, or they could help humans better adapt to their environment over time.

Contents ↓

Overview Function Anatomy Conditions and Disorders Care

Overview



A genetic mutation is a change to your DNA sequence by removing, adding or replacing pieces of your DNA.

What are genetic mutations?

functions. If part of your DNA sequence is in the wrong place, isn't complete or is damaged, you might experience symptoms of a genetic condition.

When do genetic mutations happen?

Genetic mutations occur during cell division when your cells divide and replicate. There are two types of cell division:

- **Mitosis**: The process of making new cells for your body. During mitosis, your genes instruct your cells to split into two by making a copy of your chromosomes.
- **Meiosis**: The process of making egg and sperm cells for the next generation. During meiosis, chromosomes copy themselves with half the amount of chromosomes as the original (from 46 to 23). That's how you're able to get your genetic material equally from each parent.

How do genetic mutations happen?

Genetic mutations occur during cell division. When your cells divide, they handwrite your body's instruction manual by copying the original document word for word. There's a lot of room for error during cell division because your cells might substitute (replace), delete (remove) or insert (add) letters while they're copying. If you have an error (genetic mutation), your genetic instruction manual for your cells may not be readable by the cells, or may have missing parts or unnecessary parts added. All of this can mean that your cells can't function as they normally should.

Schedule Genetic Counseling

Find a Doctor and Specialists

Make an Appointment

Function

How do genetic mutations affect other organs?

A genetic mutation changes the information your cells need to form and function. Your genes are responsible for making proteins that tell your body what physical characteristics you should have. If you have a genetic mutation, you could experience symptoms of a genetic condition because your cells are doing a different job than they should be.

Symptoms of genetic conditions depend on which gene has a mutation. There are many different diseases and conditions caused by mutations. The signs and symptoms you experience could include:

- Physical characteristics like facial abnormalities, a cleft palate, webbed fingers and toes, or short stature
- Problems with cognitive (intellectual) function and developmental delays
- Vision or hearing loss

Are genetic mutations bad?

Not all genetic mutations lead to genetic disorders. Some genetic mutations don't have any effect on your health and well-being. This is because the change in the DNA sequence doesn't change how your cell functions.

Your body also has enzymes, which are a substance that creates chemical reactions in our body. These enzymes help your body protect itself from disease. Enzymes can repair a variety of genetic mutations before they affect how a cell functions.

Some genetic mutations even have a positive effect on humans. Changes in how cells work can sometimes improve the proteins that your cells produce and allow them to adapt to changes in your environment. An example of a positive genetic mutation is one that can protect a person from acquiring heart disease or diabetes, even with a history of smoking or being overweight.

How do genetic mutations lead to genetic variations?

A genetic mutation is a change to a gene's DNA sequence to produce something different. It creates a permanent change to that gene's DNA sequence.

Genetic variations are important for humans to evolve, which is the process of change over generations. A sporadic genetic mutation occurs in one

person's chance of survival, or freedom from disease, then it begins being passed through generations and spread through the population. As the mutation passes from generation to generation, it becomes a normal part of the human genome and evolves from a gene variant into a normal gene.

Anatomy

Where are genes in my body?

Genes reside on threadlike structures in your body called chromosomes. Chromosomes are in each cell in your body. There are trillions of cells in your body that make you who you are.

What are the different types of genetic mutations?

There are different types of genetic mutations based on where they form. Types of genetic mutations include:

- **Germline mutation**: A change in a gene that occurs in a parent's reproductive cells (egg or sperm) that affects the genetic makeup of their child (hereditary).
- **Somatic mutation**: A change in a gene that occurs after conception in the developing embryo that may become a baby. These occur in all cells in the developing body — except the sperm and egg. Somatic

Can I inherit genetic mutations?

Yes, you can inherit germline genetic mutations, while somatic mutations occur with no previous history of the mutation in your family. There are several patterns that genetic mutations can pass from the parent to a child (hereditary), like:

- **Autosomal dominant**. Only one parent needs to pass the genetic mutation onto their child for their child to inherit the mutation. Marfan syndrome is an example of a condition inherited in this pattern.
- **Autosomal recessive**. Both parents need to pass the same genetic mutation onto their child for their child to inherit the mutation. Sickle cell disease is an example of a condition inherited in this pattern.
- **X-linked dominant**. Babies have an X chromosome. Only one mutation on the X chromosome needs to pass from one parent to the child for the child to inherit the mutation. Fragile X syndrome is an example of a disorder inherited in this pattern.
- **X-linked recessive**. If only the father has the mutation, there's an 100% chance that female offspring will be carriers and no male offspring will be affected. If only the mother had the mutation, there's a 50% chance that female offspring will be carriers and a 50% chance male offspring will have the condition. If both parents have the mutation, 50% of male offspring will have the condition and 100% of female offspring will have the mutation. Color blindness is an example of a condition inherited in this pattern.

acquired the mutation from their parents. Thrombocytopenia is an example of a condition inherited in this pattern.

- **Y-linked:** Only male babies have a Y chromosome and can inherit this type. Only one mutation on the Y chromosome needs to pass to the child to inherit the mutation. Webbed toes are an example of a condition inherited in this pattern.
- **Codominant**. Each gene has two parts (one from the egg and one from the sperm). They usually work together to create a single trait. But sometimes, they each work separately to produce variations of the trait. Alpha-1 antitrypsin deficiency is an example of a condition inherited in this pattern.
- **Mitochondrial**. The mitochondrion is the part of a cell that creates energy. Only mitochondria (plural) from the egg survive fertilization, when the two cells come together. So, all maternal DNA in the embryo comes from the egg. This is why mitochondrial inheritance is also known as maternal inheritance. Leber hereditary optic neuropathy (sudden vision loss) is an example of a condition inherited in this pattern.

Conditions and Disorders

What are genetic disorders?

A genetic disorder is a condition caused by changes in your genome, or the genetic material present in a human. It includes your DNA, genes and

- Mutation of multiple genes (multifactorial inheritance)
- Mutation of one or more chromosomes
- Environmental factors (chemical exposure, UV rays) that change your genetic makeup

You can inherit the genetic condition from your parents (if it's germ cell DNA in the sperm or egg) or the genetic condition can happen randomly, without having a history of the genetic condition in your family.

What are common genetic disorders?

There are thousands of genetic conditions that exist. Some of the most common genetic conditions are:

- Alzheimer's disease
- Some cancers
- Cystic fibrosis
- Down syndrome
- Sickle cell disease

Is there a test that checks for genetic mutations?

If your healthcare provider suspects that you have a genetic condition or you're at risk of having a child with a genetic condition, they may offer a genetic test. There are many genetic tests that require a sample of your

chromosomes that cause genetic conditions. These tests can also let you know if you're at risk of having a child with a genetic condition, if you plan on fathering a child or becoming pregnant.

Care

How do I keep my genes healthy to prevent genetic mutations?

Some genetic mutations happen randomly and you can't prevent them from occurring. Other genetic mutations can be the result of changes in your environment. You can take steps to prevent some genetic mutations by:

- Not smoking
- Wearing sunscreen when out in the sun
- Avoiding chemical exposure (carcinogens) or exposure to radiation (X-ray exposure)
- Eating a nutritious, balanced diet and avoiding processed foods

A note from Cleveland Clinic

While some genetic mutations can lead to genetic conditions, most mutations don't cause symptoms in humans. It's difficult to prevent mutations from happening, especially as genetic mutations can occur randomly — some without being present in your family history. If you plan on having biological children and want to understand your risk of passing a

Care at Cleveland Clinic

Do certain health conditions seem to run in your family? Are you ready to find out if you're at risk? Cleveland Clinic's genetics team can help.

Schedule Genetic Counseling | Find a Doctor and Specialists | Make an Appointment

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References

Questions 216.444.2538

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