Genetics & Hereditary Cancer

Pim Suwannarat, MD

CANCER SURVIVOR DAY

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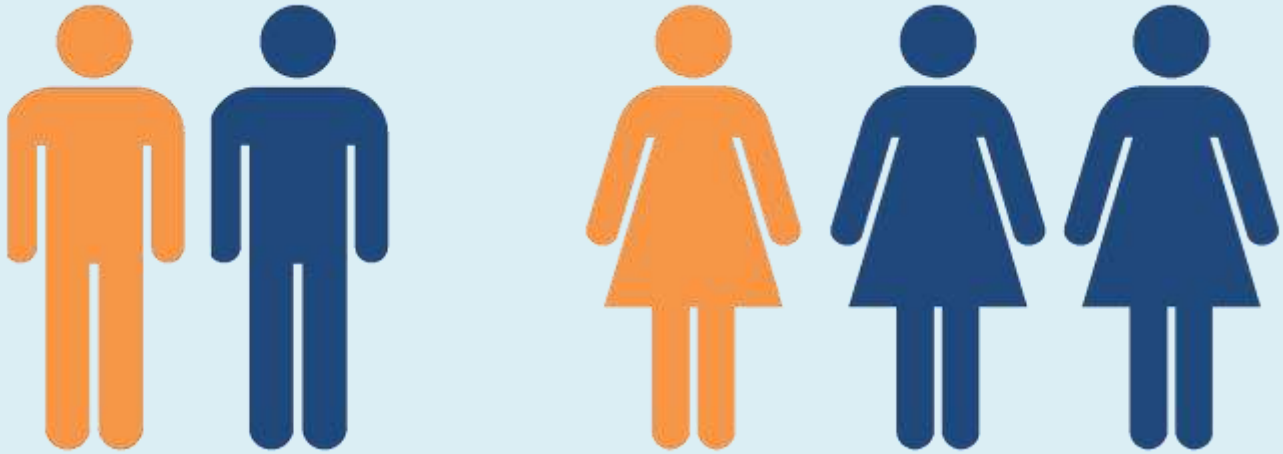
Cancer is Common



1 in 2 men and 1 in 3 women will be diagnosed with cancer

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You cannot inherit cancer.

You can only inherit a SUSCEPTIBILITY to cancer.

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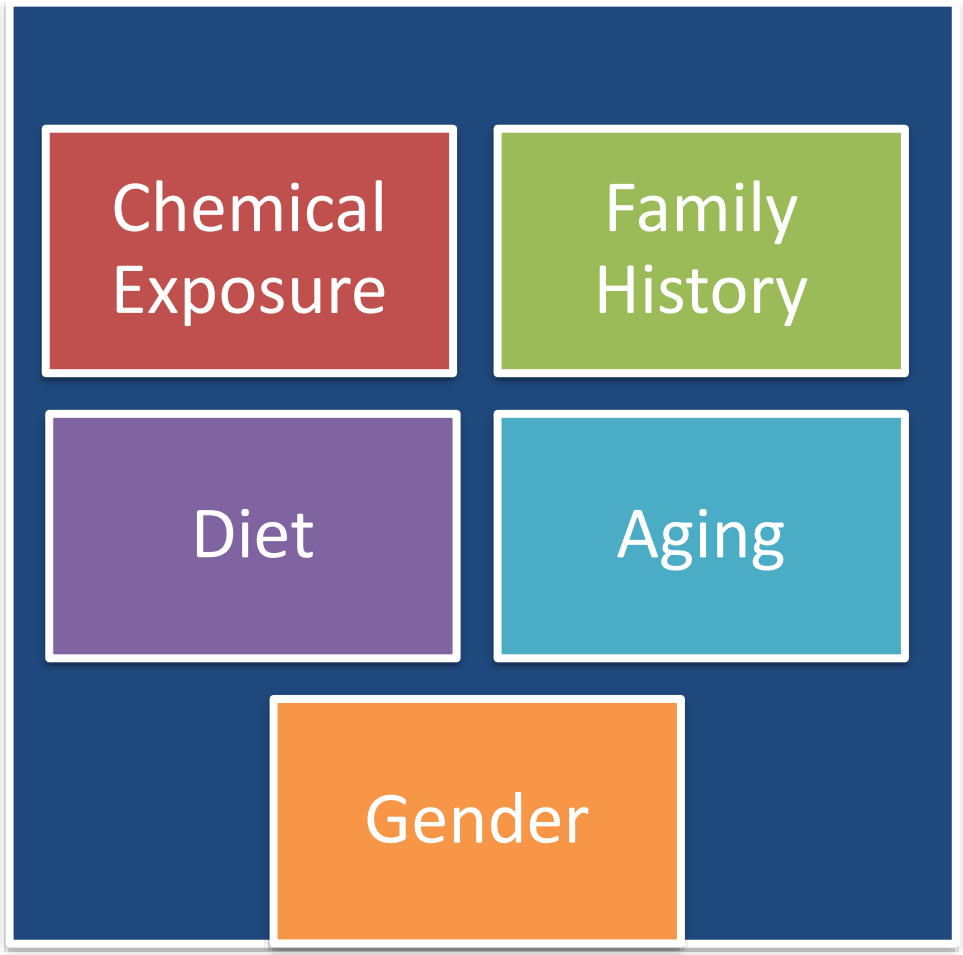
Cancer Risk Factors



- You may have no risk factors and have cancer
- You may have all the risk factors and not have cancer

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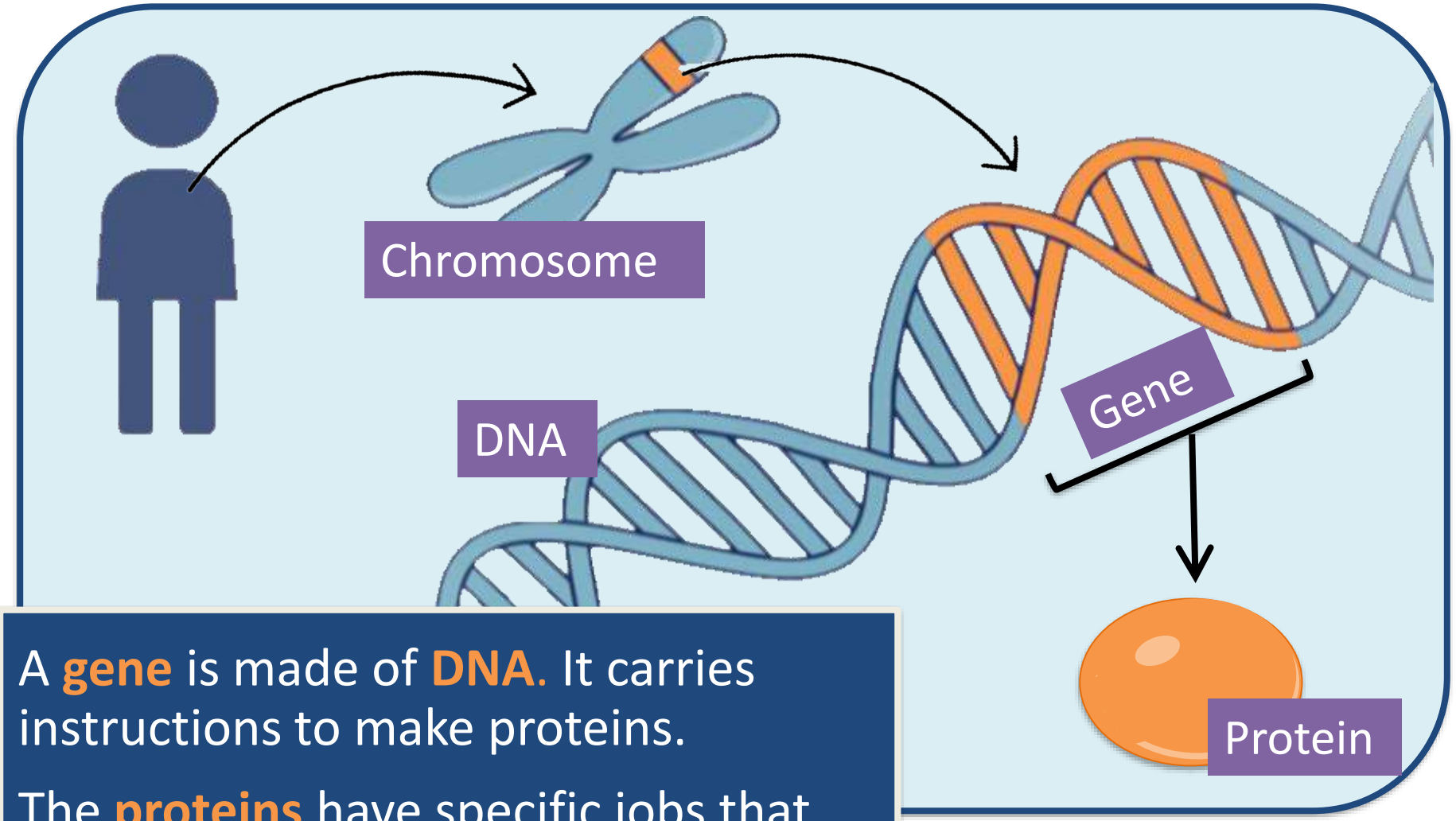
What is a Gene?



A **gene** is made of **DNA.** It carries instructions to make proteins.

The **proteins** have specific jobs that help your body work normally.

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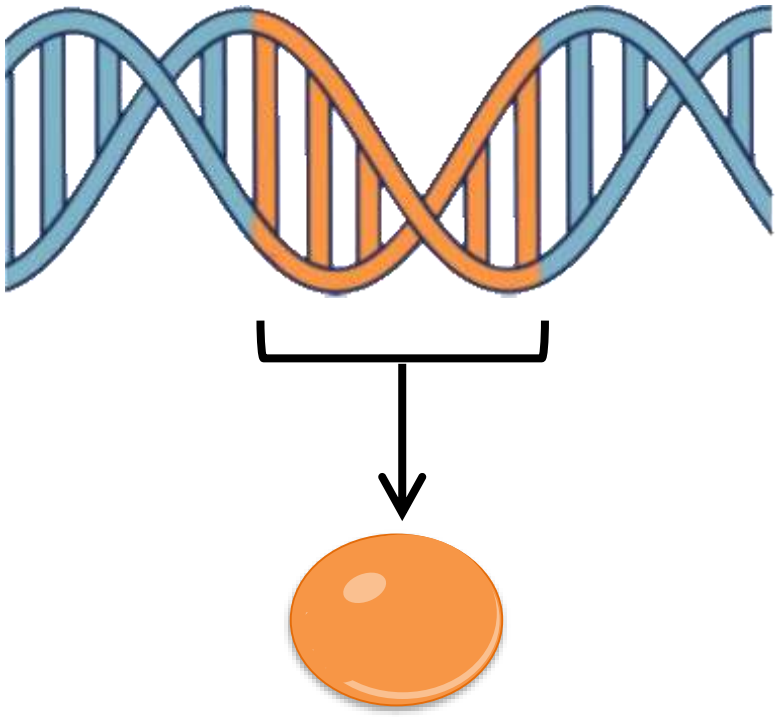
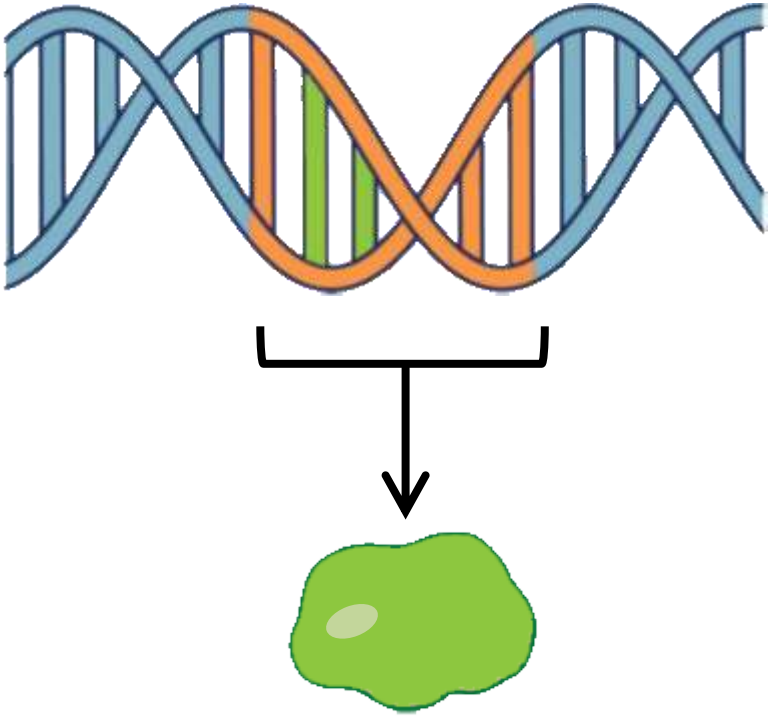
What is a Genetic Mutation?

Normal Gene



Healthy Protein

Mutated Gene



Damaged Protein

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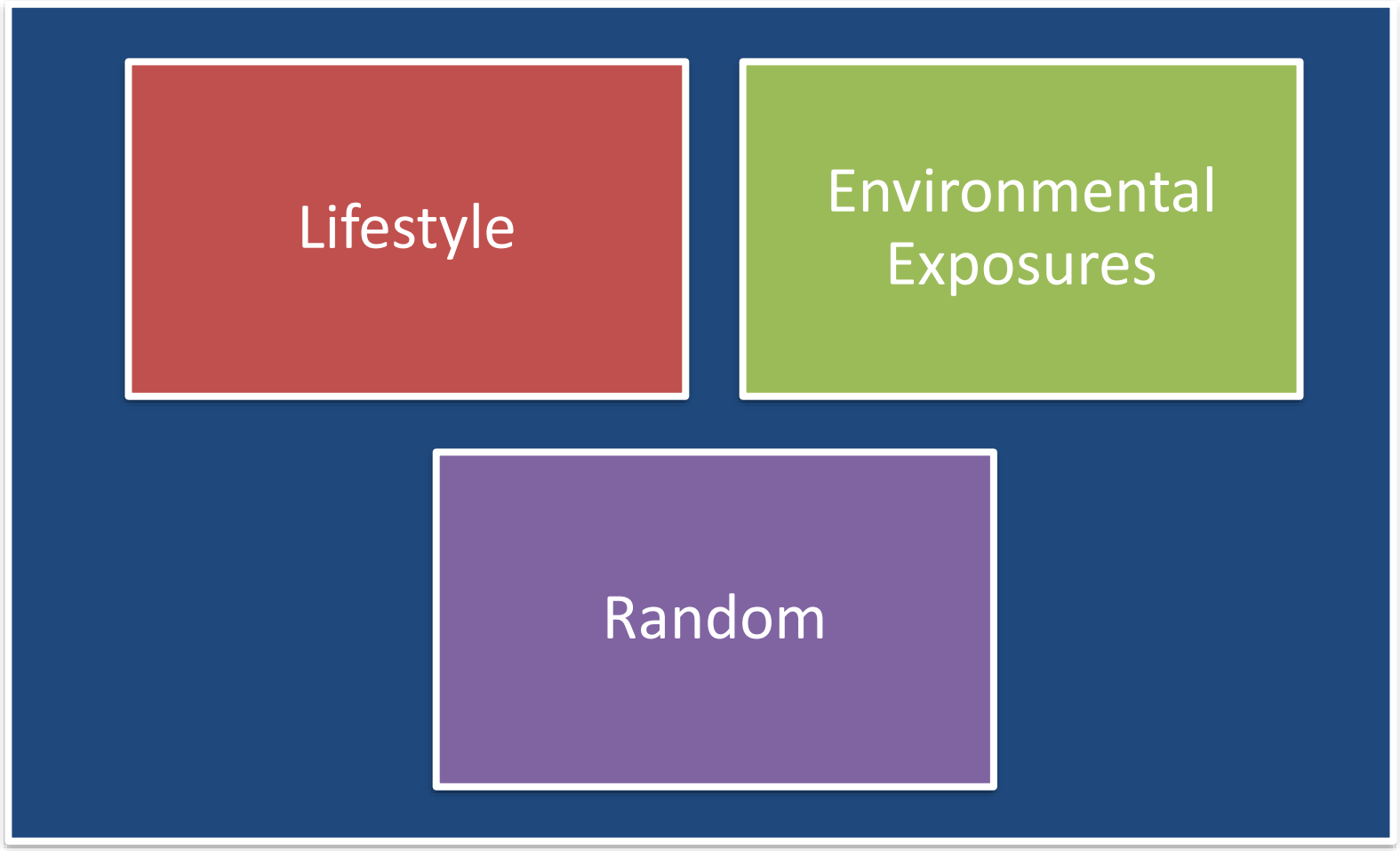
How Do Mutations Happen?



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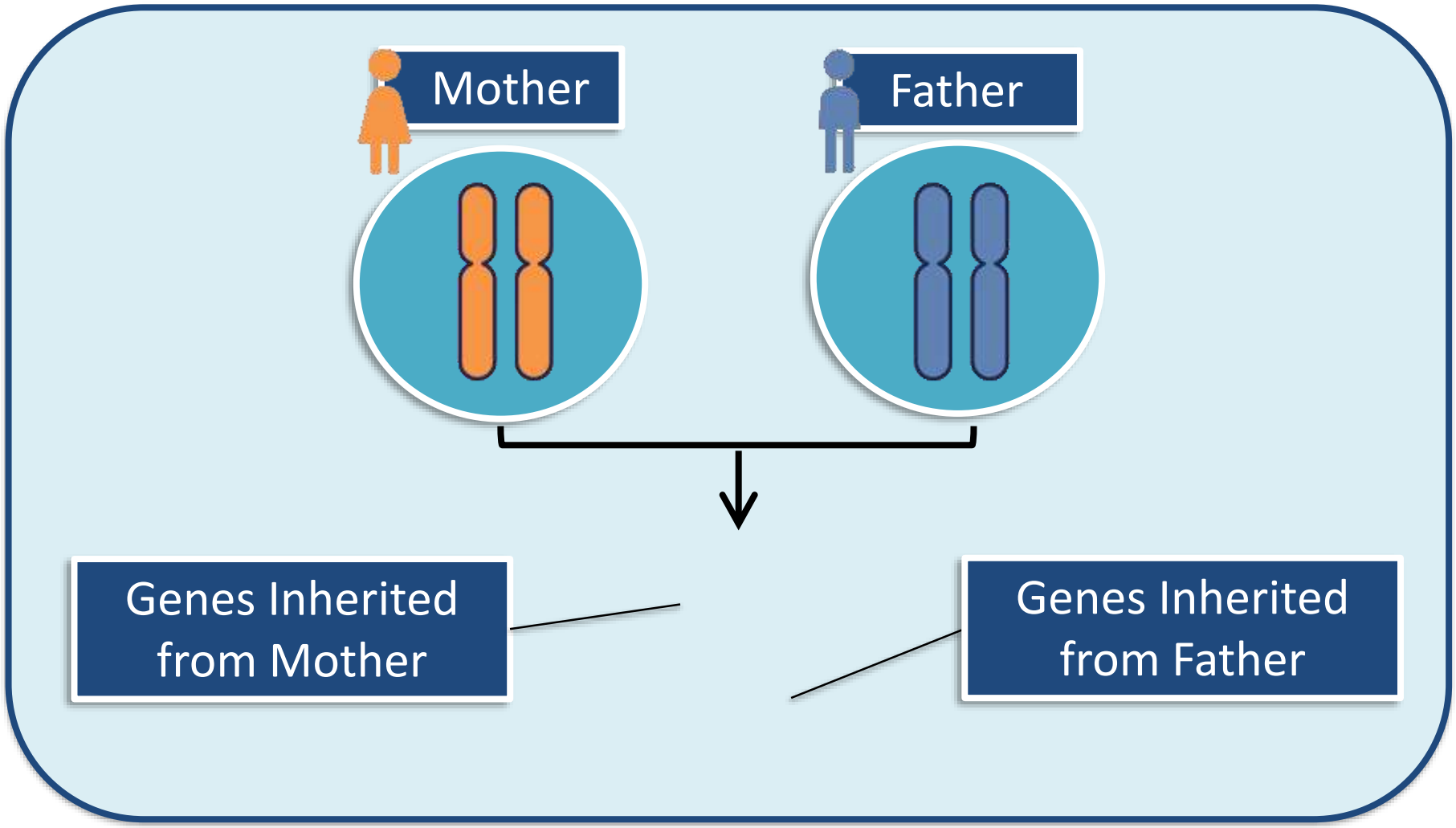
How Does Cancer Start?

- Mutations can lead to loss of control over cell growth
- With uncontrolled cell growth, a mass of excess tissue may form (tumor)
- A tumor becomes cancerous when it has the ability to spread to other areas of the body.

Inherited Cancer Risk Genes

- Everyone has two copies of each gene
- Most genes that are linked with increased risk for cancer are “Tumor suppressor genes”
- Hundreds of different mutations, but usually just one mutation in any family

How Are Genes Inherited?



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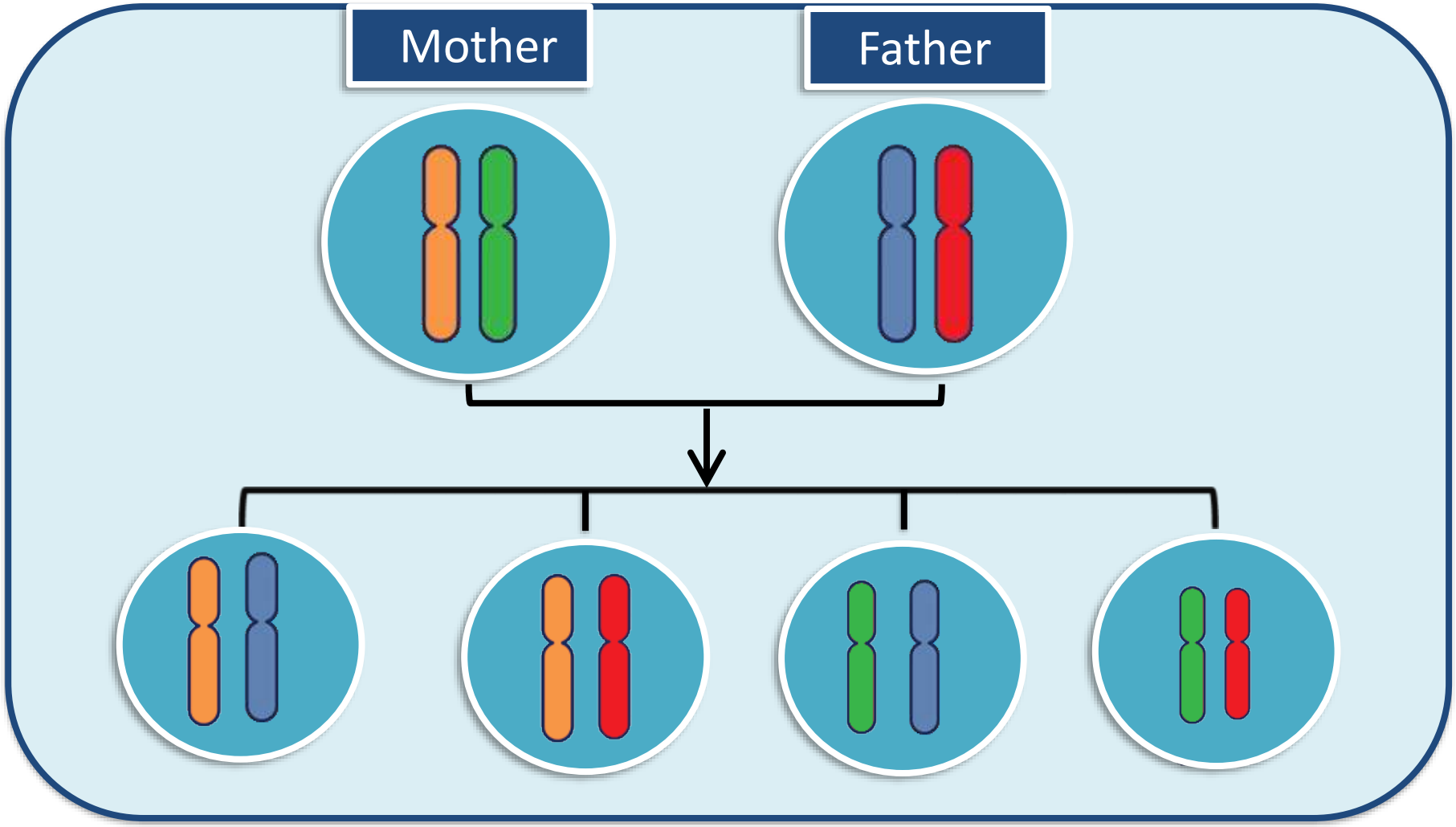
How Are Genes Inherited?



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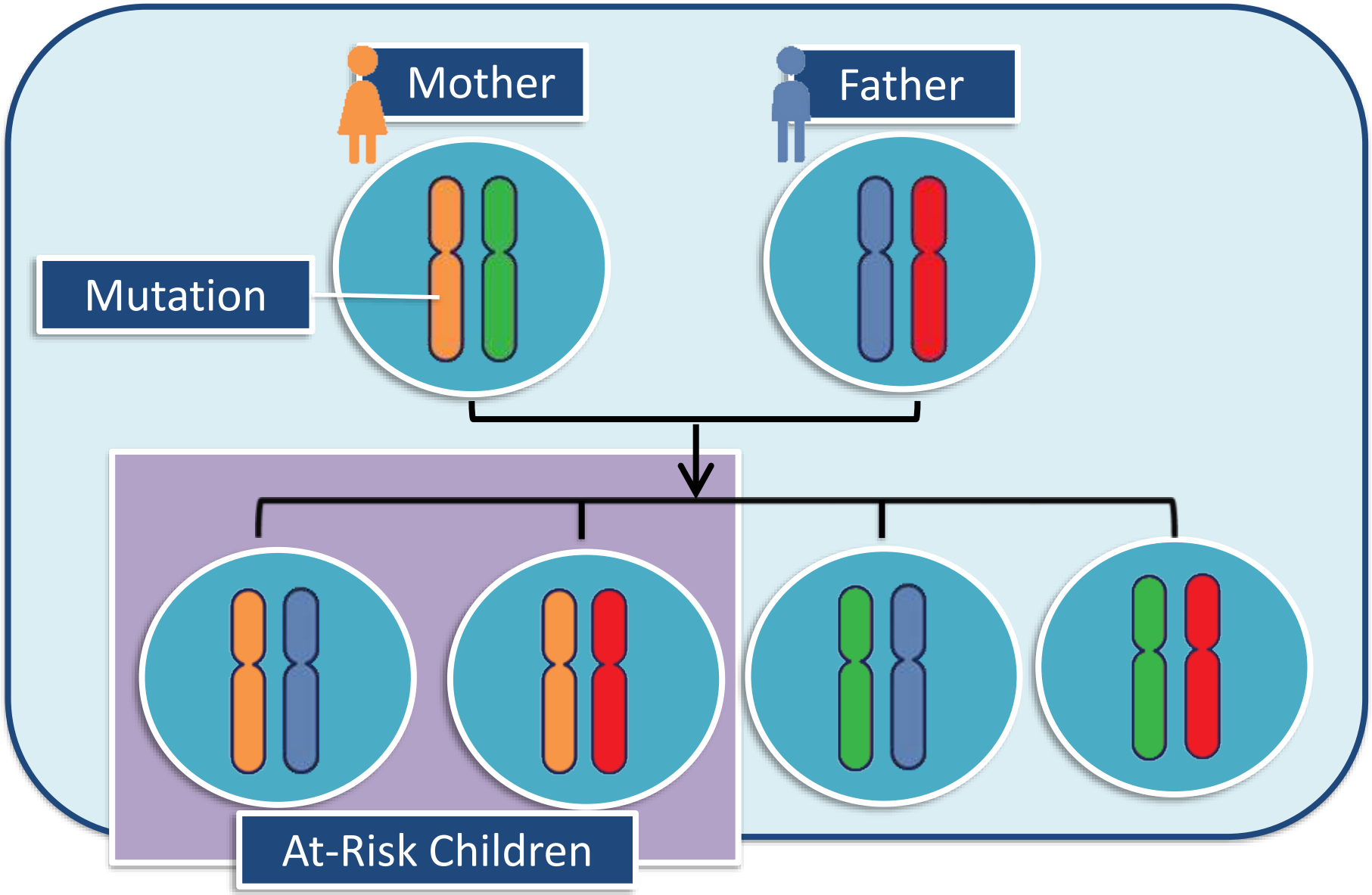
Autosomal Dominant Inheritance



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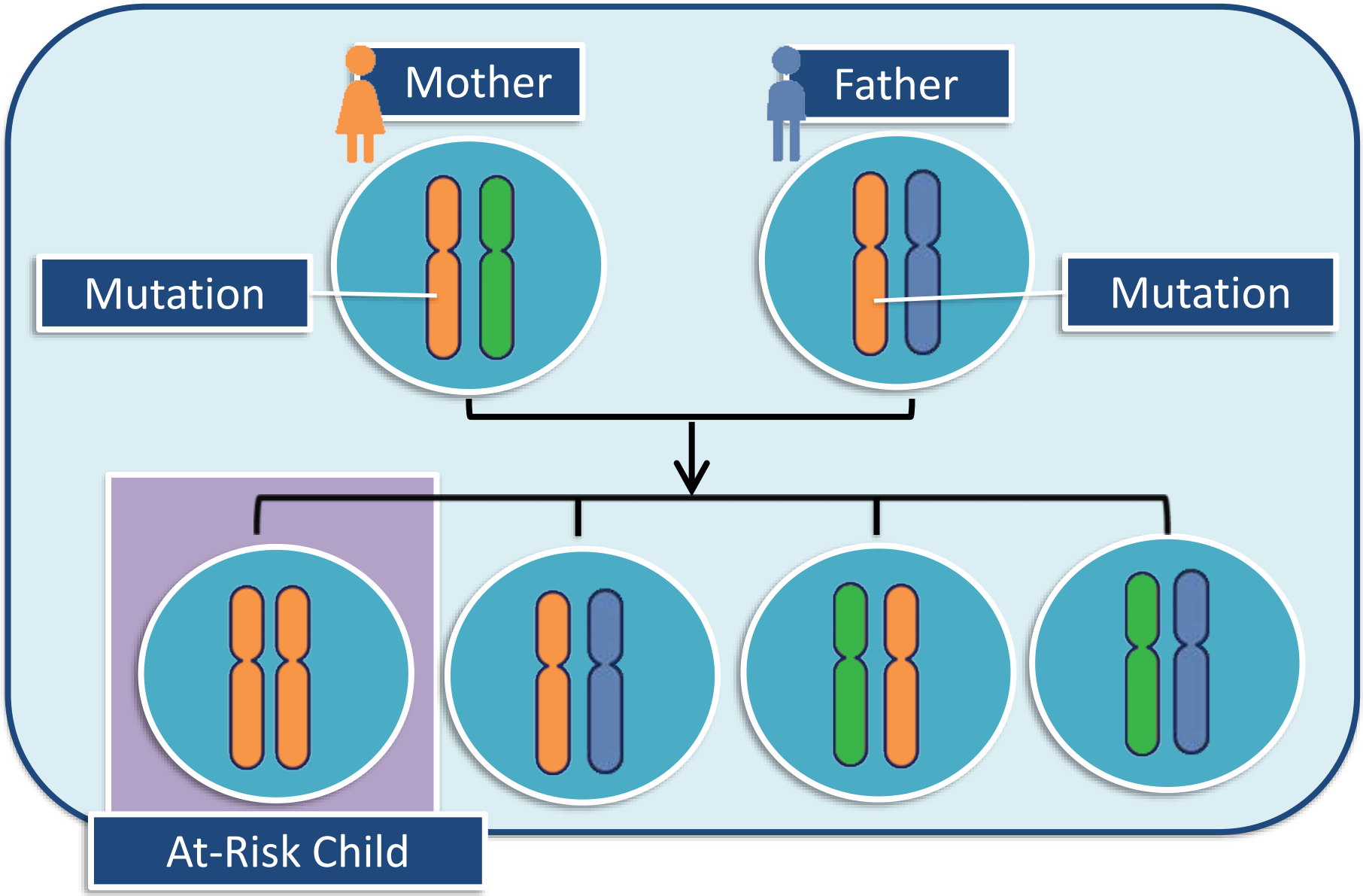
Autosomal Recessive Inheritance



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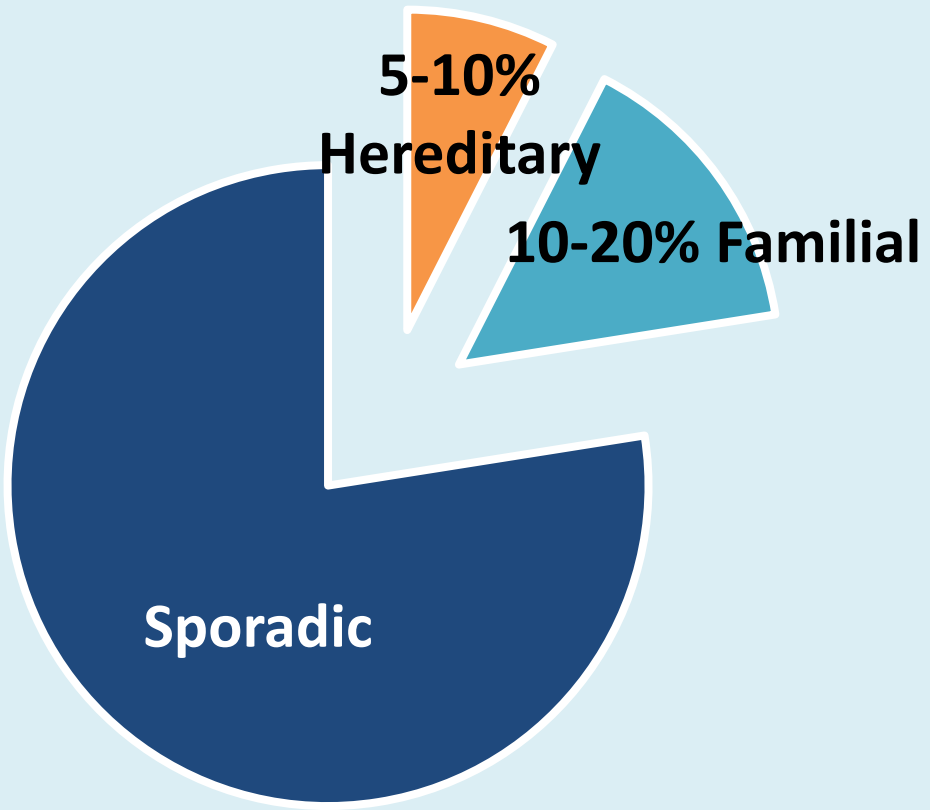
Categories of Cancer Risk



Hereditary: Inherited gene mutation. Significantly increased cancer risk

Familial: Multiple genes & environmental factors involved. Some increased cancer risk

Sporadic: Cancer occurs by chance or relative to environmental factors. General population cancer risk

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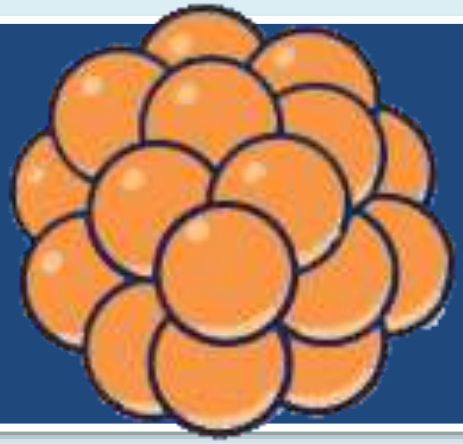
Somatic vs Germline Mutation

Germline Mutation

Change in the gene that is inherited



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Somatic Mutation

Change in the gene that arose in the tumor and is confined to the tumor

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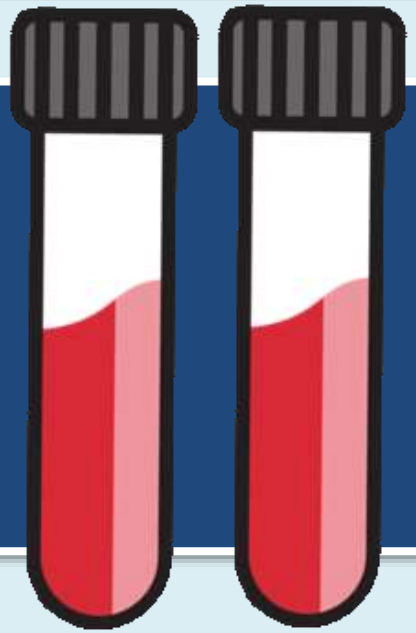
Tumor Testing vs Germline Testing

Tumor Testing

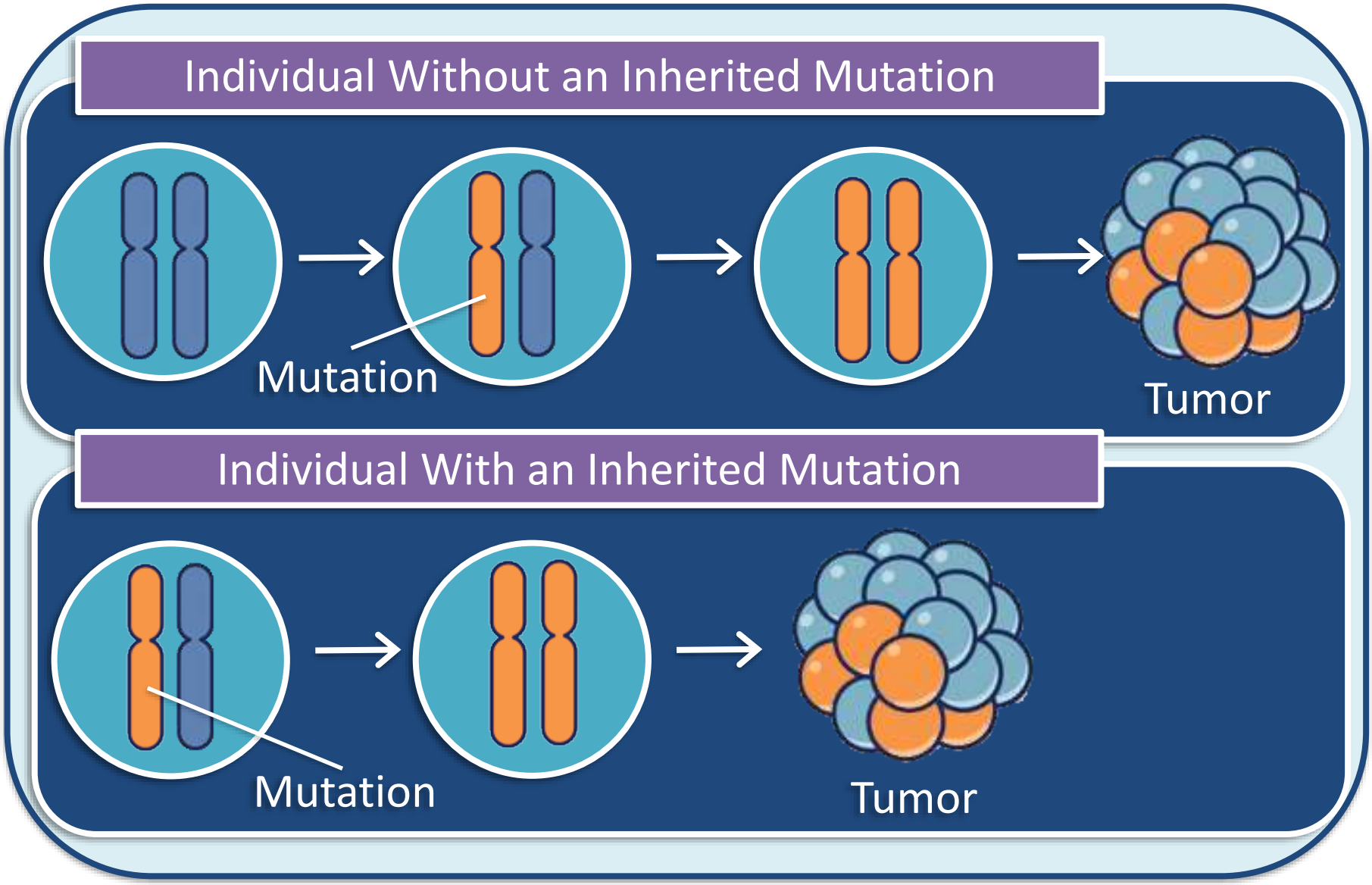
Can help guide treatment options (i.e., chemotherapy)



Germline Testing

Can help determine if a mutation was inherited and help guide treatment and risk management options

The Two-Hit Hypothesis



Family History Suggestive of Hereditary Cancer

- Cancer in more than one generation
- Several close relatives with the same or related types of cancer
- Multiple cancers
- Certain rare cancers
- Cancer at an early age

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Taking your own family history

- List all your relatives
- Identify anyone with cancer
- Include the person's age at cancer diagnosis
- Gather details about each cancer

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Genetic Counseling

- A genetic counselor can help estimate the risk for cancer
- Many people over estimate their risk for cancer
- Your cancer risk considers your personal medical history and the history of cancer in your family

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Genetic Testing

- Process to identify a mutation
- May better define your cancer risk
- May let you personalize your cancer risk management
- Can give you information to share with relatives

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Limitations of Genetic Testing

- Not all mutations are detectable
- Not all cancer genes are known
- Results may not clarify your cancer risk
- Positive results do not determine if or when you will develop cancer

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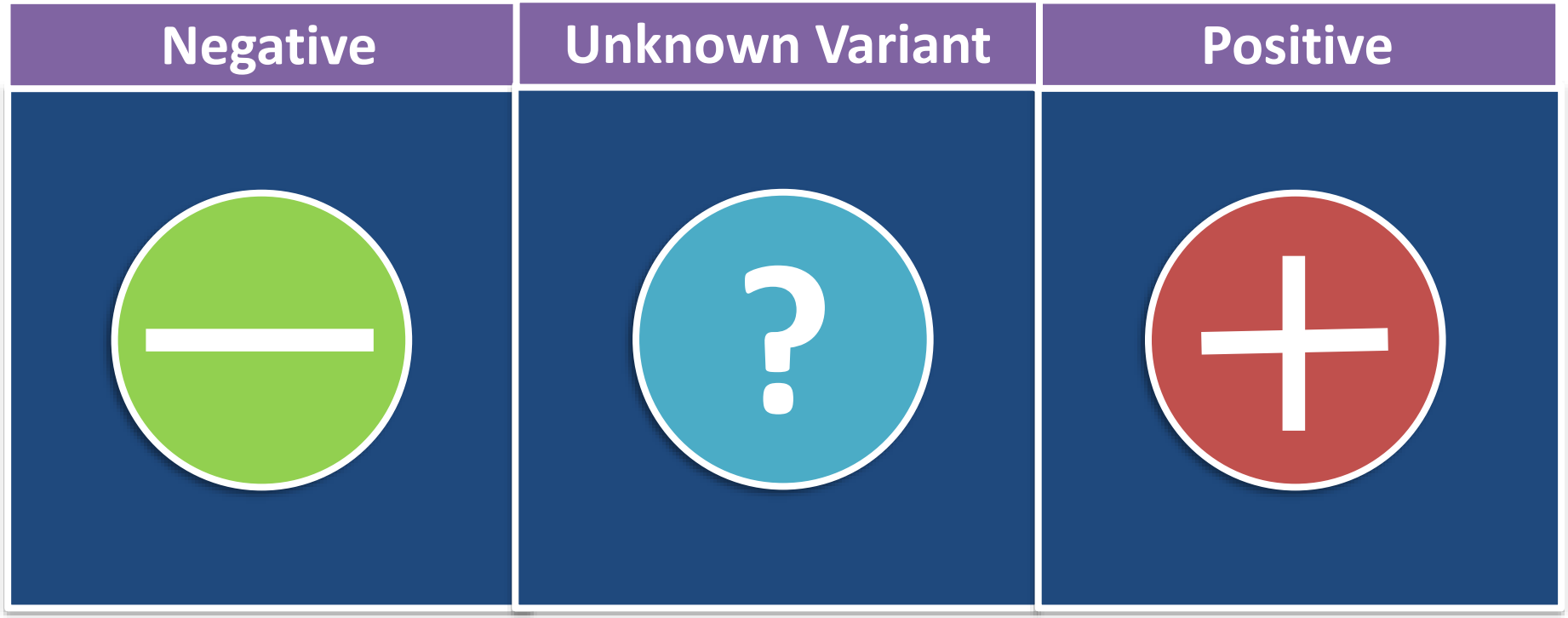
Possible Results of Genetic Testing



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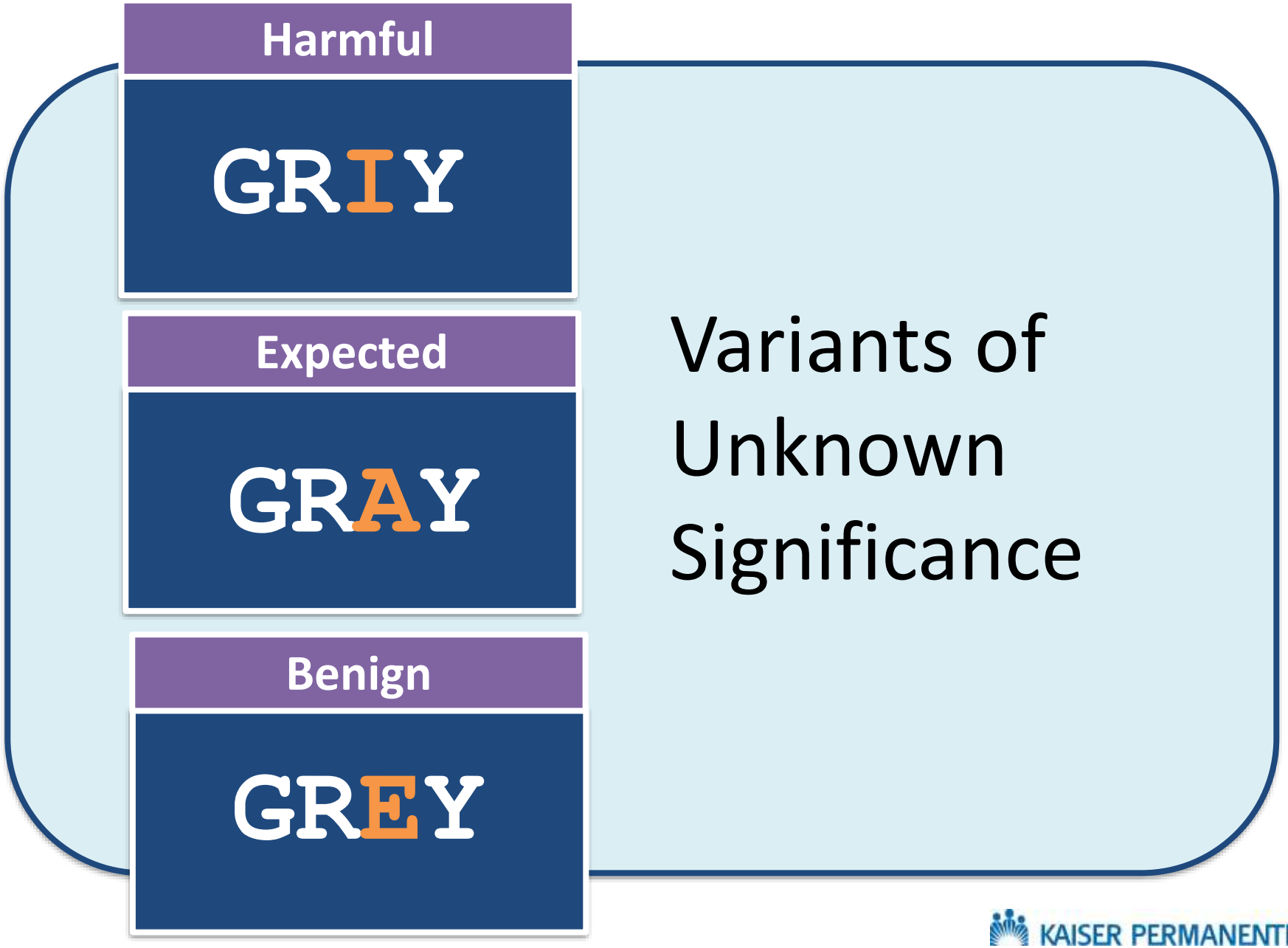

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How Are Testing Results Used?

Medical care based on personal and family history of cancer

Medical care based on cancer risks specific to gene mutation

Makes testing available to relatives

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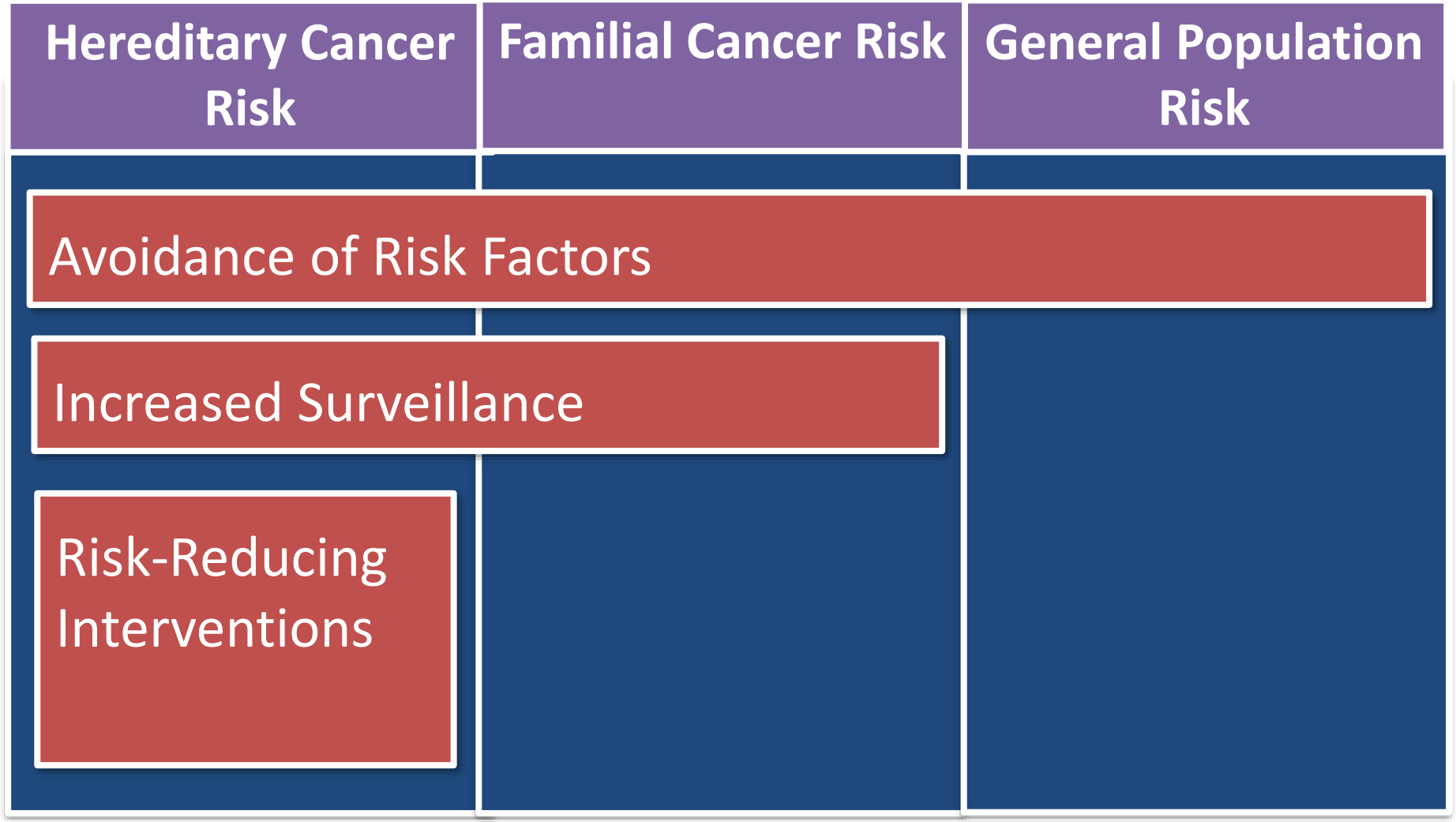
Management Options



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Genetic Testing & Family Members

Genetic Testing for Known Mutation in Family

Possible Increased Screening

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Health Insurance

Disability Insurance
Life Insurance

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Common Misconception #1

I already had cancer, I don’t need genetic testing

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Common Misconception #2

Several of my aunts and my grandmother on my father's side had breast cancer, but no one on my mother side, so I'm not at higher risk for breast cancer.

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Common Misconception #3

Nearly everyone in my family had cancer, I'm guaranteed to have cancer

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Take Home Points

- Cancer is the result of multiple genetic changes
- Most cancer is not hereditary
- Genetic testing is not offered to low risk families
- Cancer testing is best initiated in the person with cancer

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Take Home Points Continued

- Not everyone with a known cancer susceptibility mutation will develop cancer
- Everyone can benefit from recommended cancer screenings such as mammography and colonoscopy.

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THANK YOU

Questions?

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