

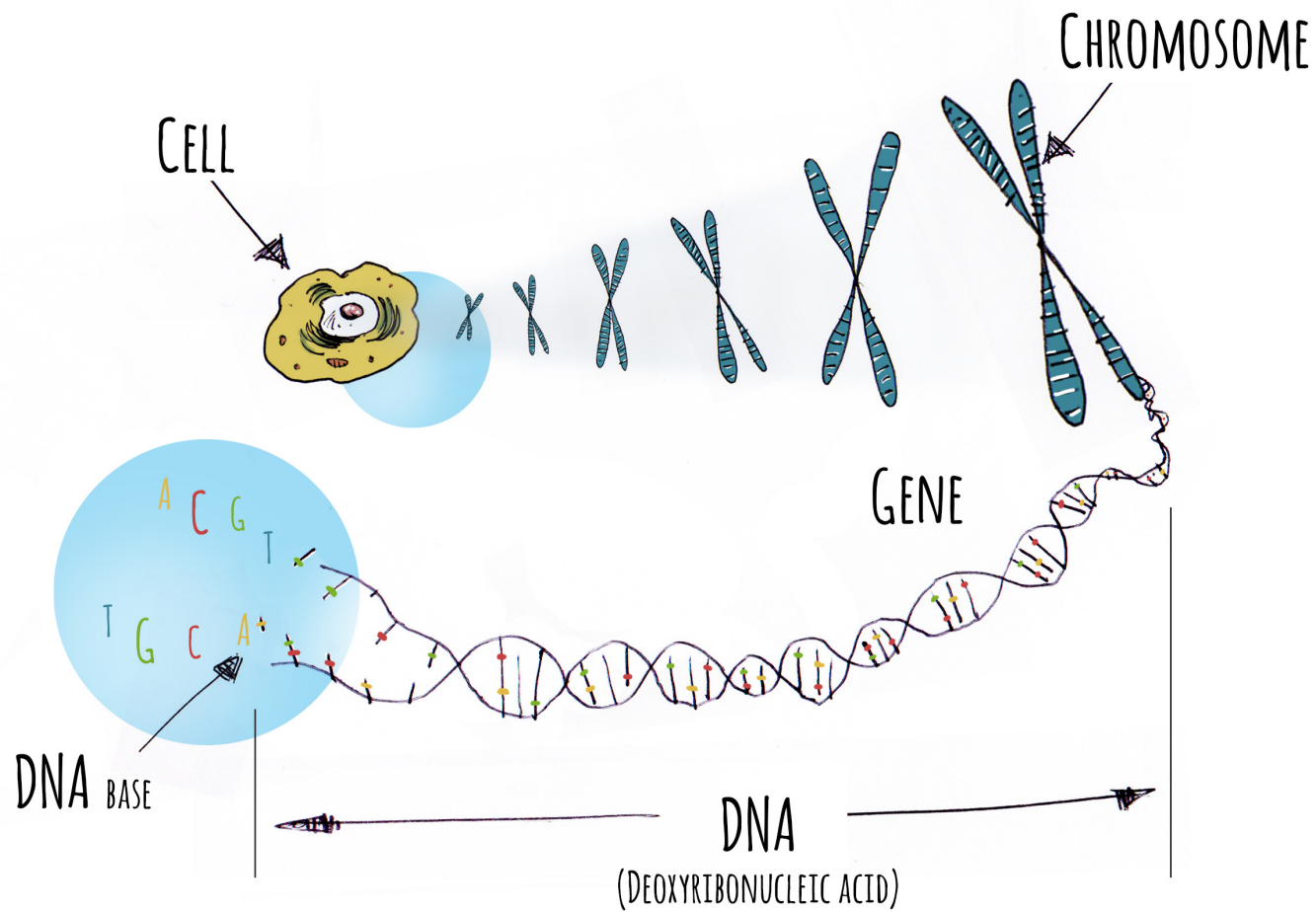
# ExomeNext

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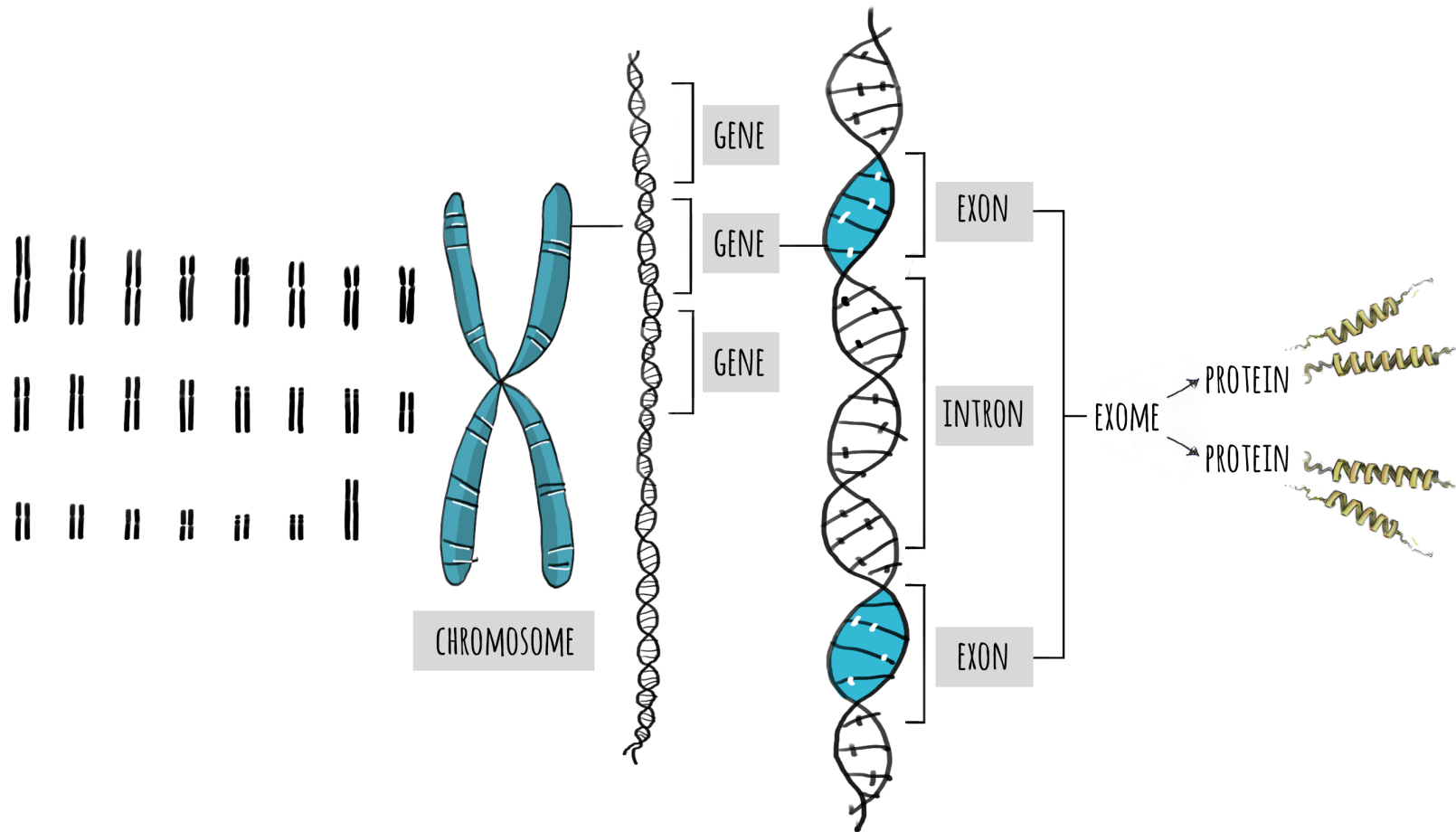
A GUIDE TO EXOME SEQUENCING



# What is DNA?

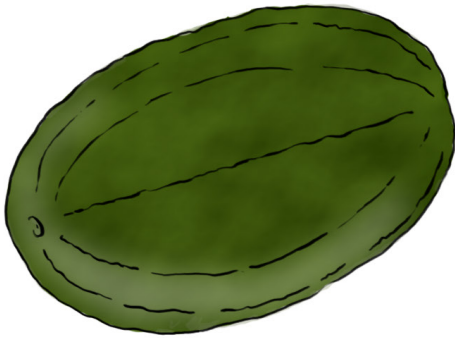


# What is the Exome?

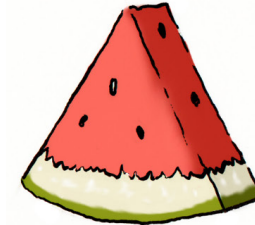


# Why is the Exome Important?

WATERMELON = GENOME



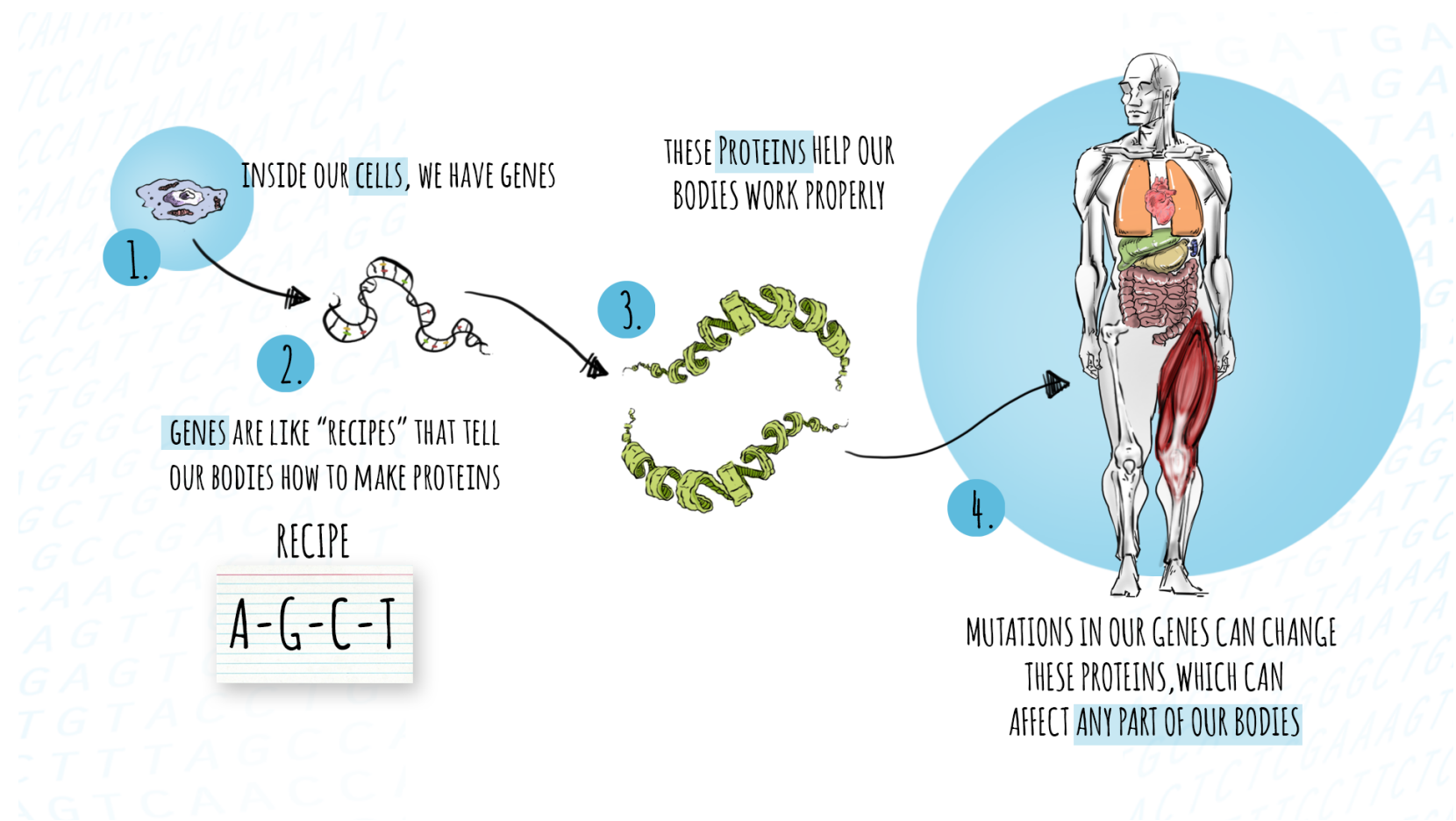
SLICE = EXOME 1-2%



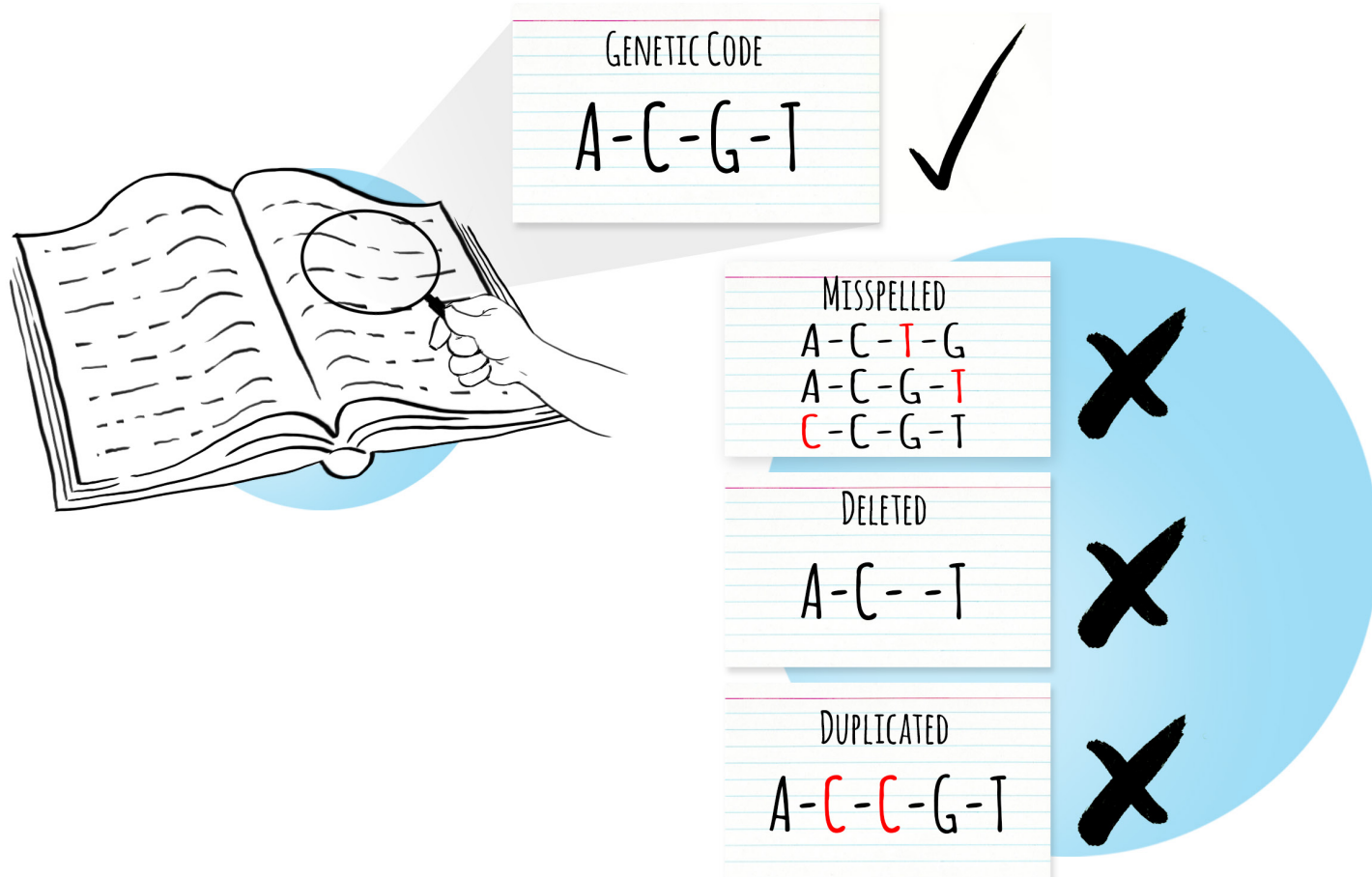
SEEDS = GENES



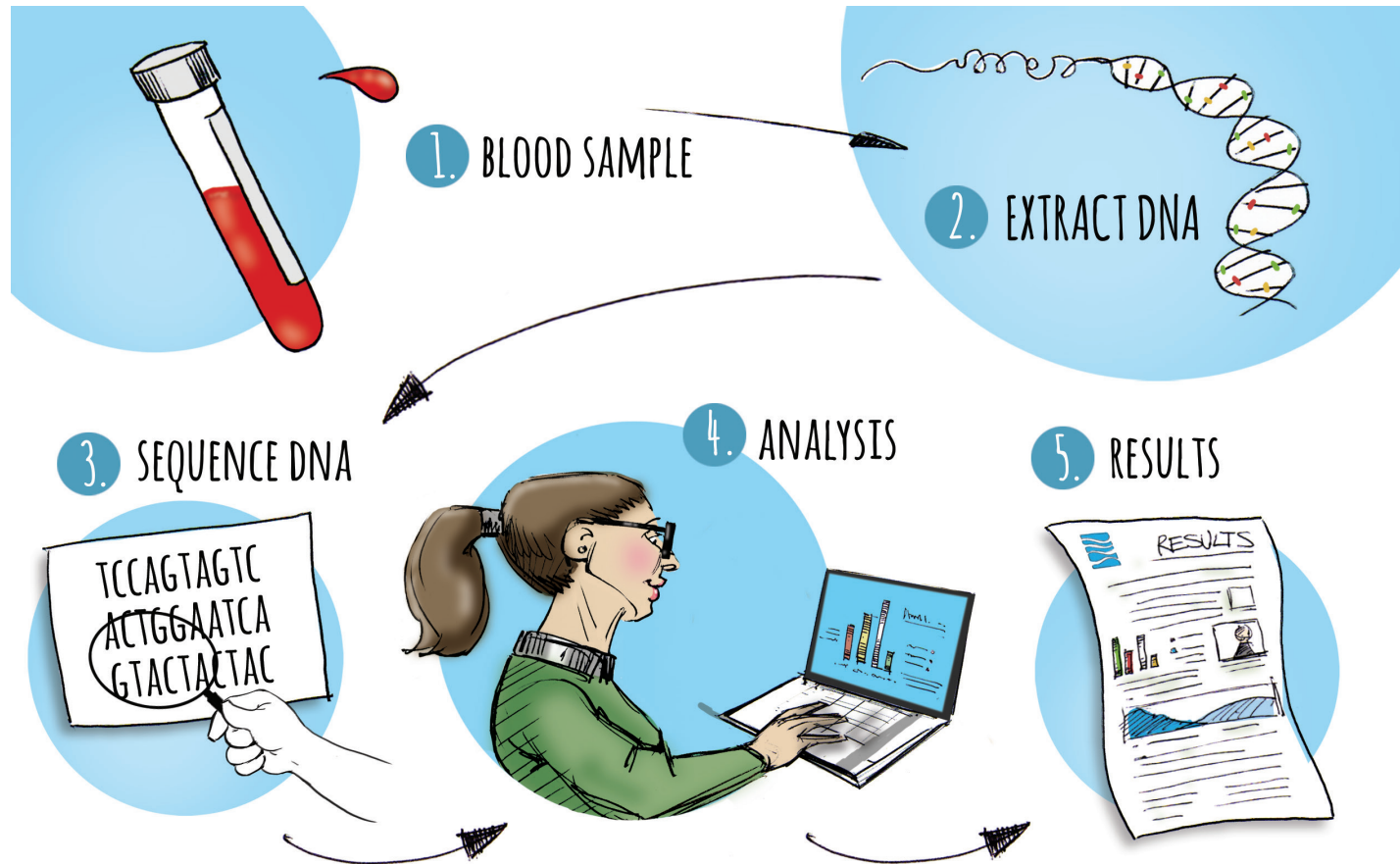
# What Do Genes Do?



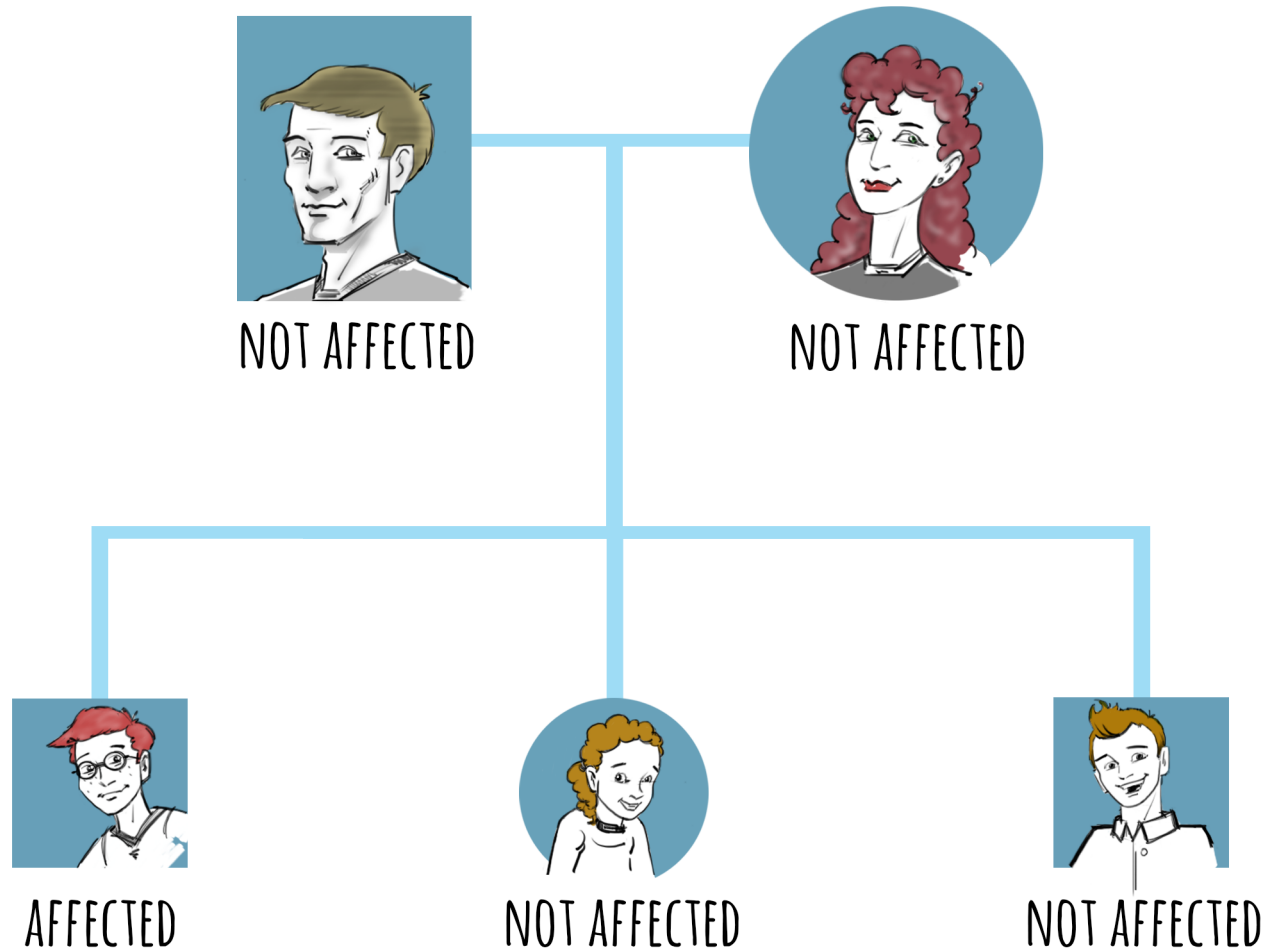
# What is a Mutation?



# How is ExomeNext Done?



# Testing Family Members



## Is ExomeNext Right For You?

ExomeNext might be something to consider if your or your family member's medical concern could be due to an underlying genetic change, but:

- Your previous genetic testing did not find an answer (or diagnosis)
- There is no specific genetic testing available for the genetic condition you may have
- Your medical concerns may be caused by changes in more than one gene

## What Are the Benefits of ExomeNext?

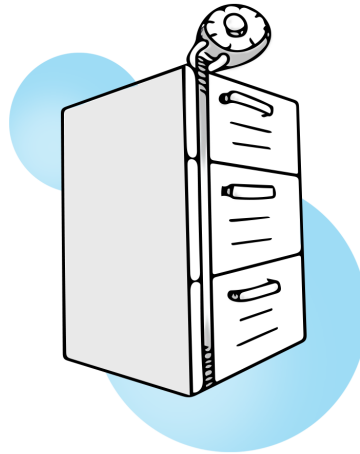
- Quicker and more cost-effective since it studies multiple genes at once
- Can suggest changes to treatment or medical care
- Removes feelings of uncertainty related to having an undiagnosed disease
- Provides awareness about health concerns to your other family members

# Things to Consider Before Testing

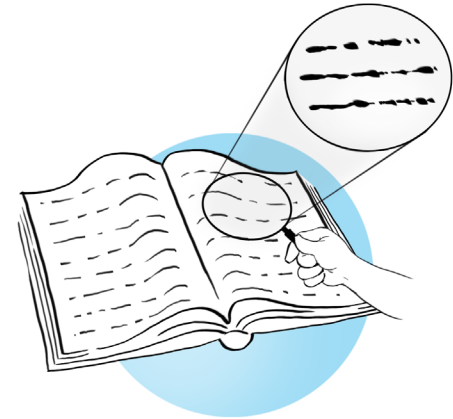
## LIMITATIONS



## PRIVACY



## SECONDARY FINDINGS



## Supplementary Information

RESULT TYPE	OVERALL RESULT CATEGORY		RESULT MEANING
Primary Result	Characterized Genetic Etiologies	Positive	A mutation was found in a gene known to be associated with your medical concerns
		Likely Positive	A mutation was found in a gene that most likely causes your medical concerns
		Uncertain	A genetic change was found, but it is unclear if it is the cause of your medical concerns
		Negative	The underlying cause of your medical concerns has not been found among genes associated with genetic conditions
	Candidate Genetic Etiologies	Uncertain	A genetic change was found, but based on information we know today, it is unclear this is the cause of your medical concerns
		Negative	The underlying genetic cause of your medical concerns has not been found
Secondary Findings Result		Positive	Mutation(s) was found in a gene that is not associated with your medical concerns
		Negative	No mutation(s) was found among a list of genes that are not associated with your medical concerns

## Characterized Genetic Etiologies: Positive

### Result

Mutation(s) was found in a gene known to be associated with your medical concerns

### Medical Management

Your doctor or genetic counselor will discuss if or how this result changes your or your family's medical care

### Genetic Testing for Family Members

Other family members may consider testing for the specific mutation(s)

### Future Testing

Usually not recommended/necessary

## Characterized Genetic Etiologies: Likely Positive

### Result

A mutation was found in a gene that most likely causes your medical concerns

### Medical Management

Your doctor or genetic counselor will discuss if or how this result changes your or your family's medical care

### Genetic Testing for Family Members

Other family members may consider testing for the specific mutation(s)

### Future Testing

Usually not recommended/necessary

## Characterized Genetic Etiologies: **Uncertain**

### Result

A genetic change was found, but it is unclear if it is the cause of your medical concerns

### Medical Management

Your doctor or genetic counselor will discuss if or how this result changes your or your family's medical care

### Genetic Testing for Family Members

In some cases, testing other family members for this specific gene change(s) may help to better understand your results

### Future Testing

Studying your exome data again in the future may be helpful

## Characterized Genetic Etiologies: Negative

### Result

The underlying cause of your medical concerns has not been found in genes associated with genetic conditions

### Medical Management

Your doctor or genetic counselor will discuss if or how this result changes your or your family's medical care

### Genetic Testing for Family Members

Testing other family members is not recommended at this time

### Future Testing

Studying your exome data again in the future may be helpful

## Candidate Genetic Etiologies: **Uncertain**

### Result

A change(s) in a gene was found, but based on the information we know today, it is not clear if this is the cause of your medical concerns

### Medical Management

Your doctor or genetic counselor will discuss if or how this result changes your or your family's medical care

### Genetic Testing for Family Members

In some cases, testing other family members for this specific gene change(s) may help to better understand your results

### Future Testing

Studying your exome data again in the future may be helpful

## Candidate Genetic Etiologies: Negative

### Result

No relevant mutation(s) was identified among genes that could cause a genetic condition. The underlying cause of your medical concerns has not been found.

### Medical Management

No relevant mutation(s) was identified among genes that could cause a genetic condition. The underlying cause of your medical concerns has not been found.

### Genetic Testing for Family Members

Testing other family members is not recommended at this time

### Future Testing

Studying your exome data again in the future may be helpful

