أكاديمية كاوست
KAUST ACADEMY

Introduction to Bioinformatics

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Day 3: Sequence Analysis
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Introduction to Bioinformatics

Application: diagnosing human genetic disease

Part 1: Personalized Medicine

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Personalized Medicine

Personalized Medicine

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- Tailoring medical treatment to individual characteristics of each patient.

- Role of Bioinformatics: Analyzing genetic and clinical data to customize treatment plans.

Bioinformatics applications in Biomedical Field

- Drug Discovery and Development
- Genomic and Genetic Studies
- Disease Prediction and Prevention
- Protein Structure Prediction
- Diagnosing human genetic disease

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Part 2: Exome Analysis

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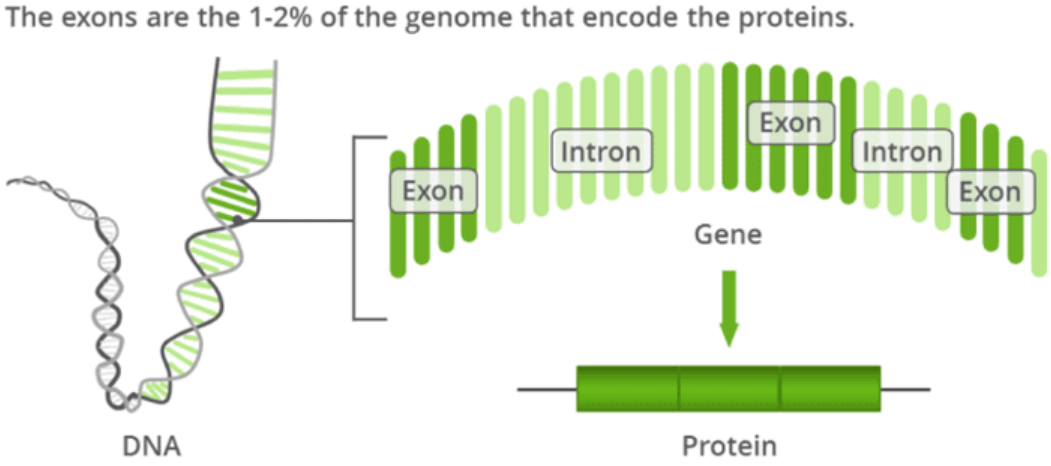
What is Exome?

An exome is the sequence of all the exons in a genome. Exons are the protein coding segments of a genome, plus their associated 5'- and 3'-untranslated regions.

The term "exon" comes from "EXpressed regiON". Exons are the regions that get translated, or expressed as proteins, as opposed to the intron, or “INTRagenic regiON” which is not represented in the final protein.



Check Galaxy Slides ☺

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Galaxy PROJECT

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Part 3: Trio Analysis

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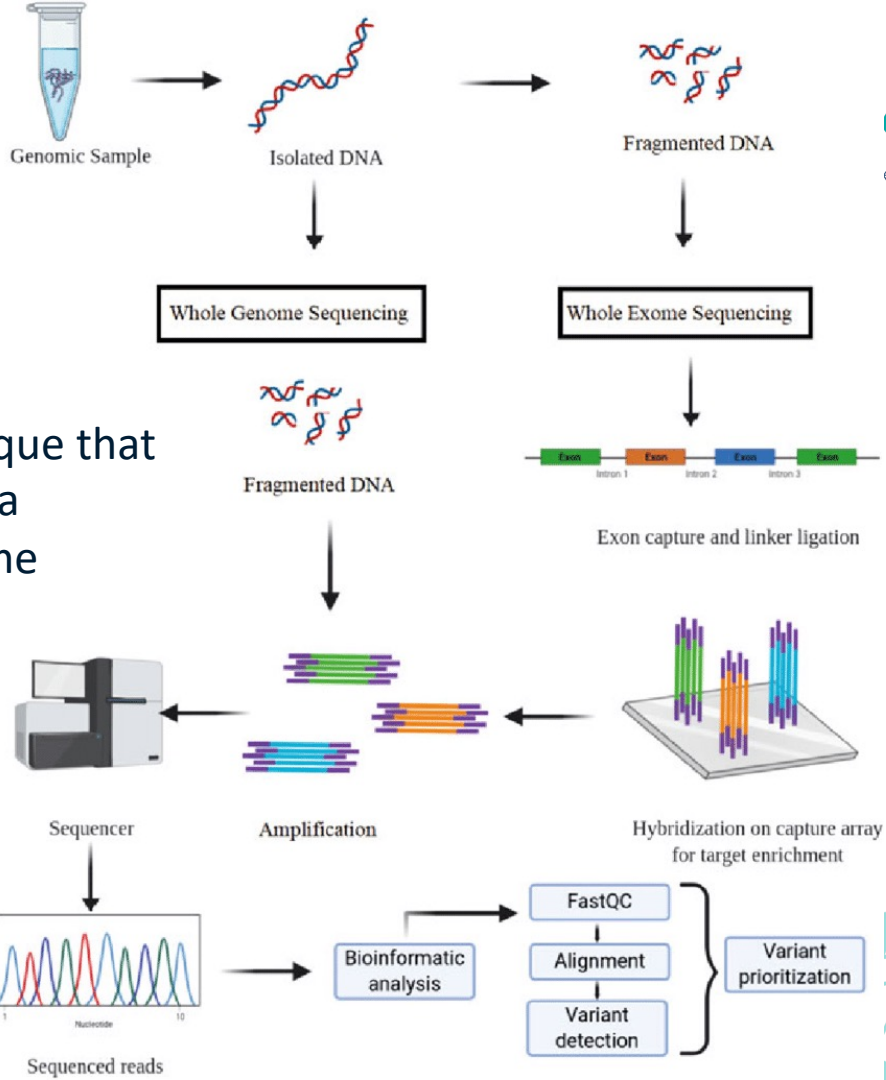
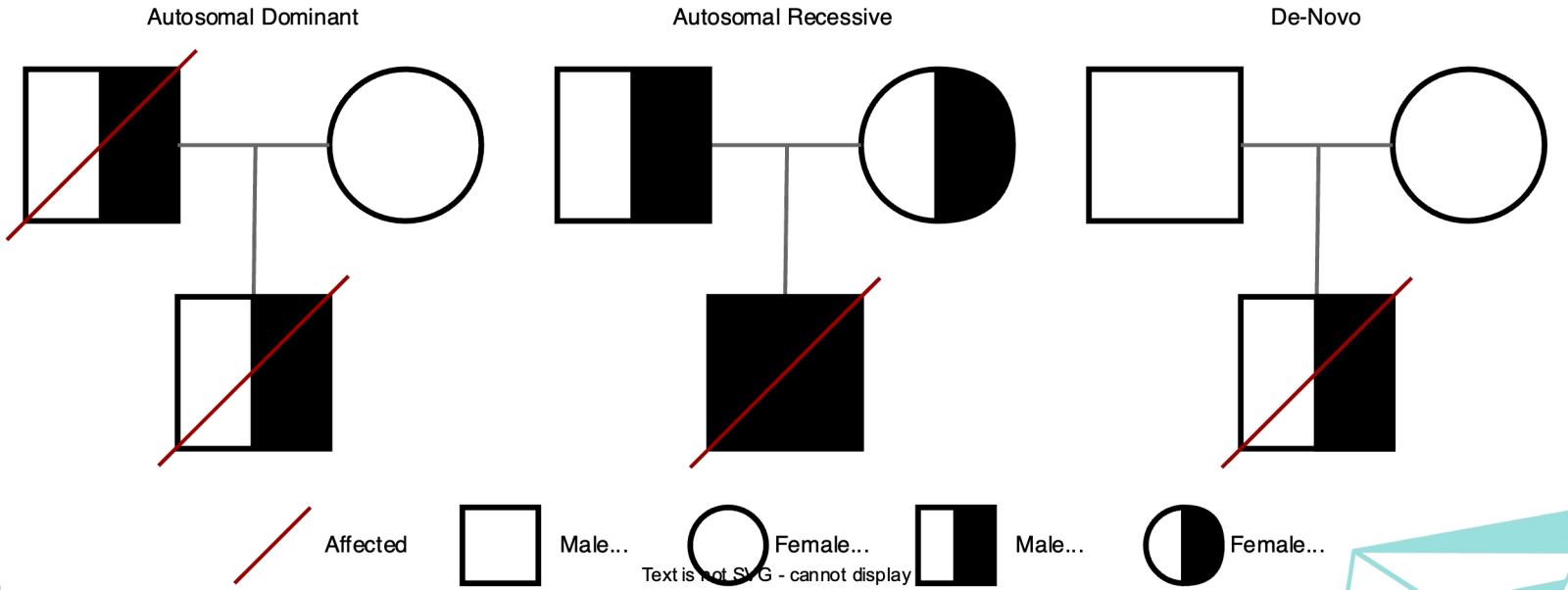
What is Exome sequencing?

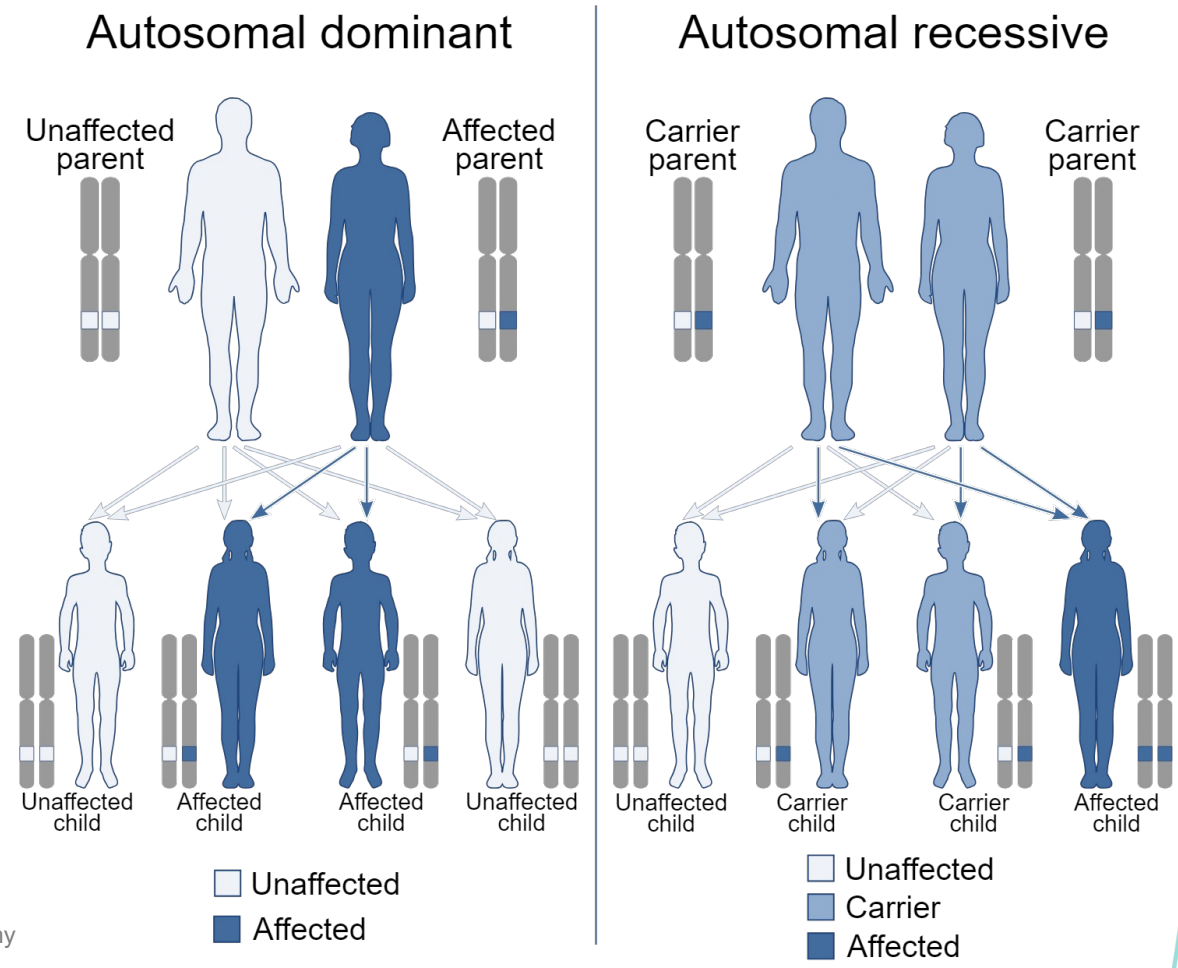
Exome sequencing is a genomic technique that analyzes the protein-coding regions of a genome. It's also known as whole exome sequencing.



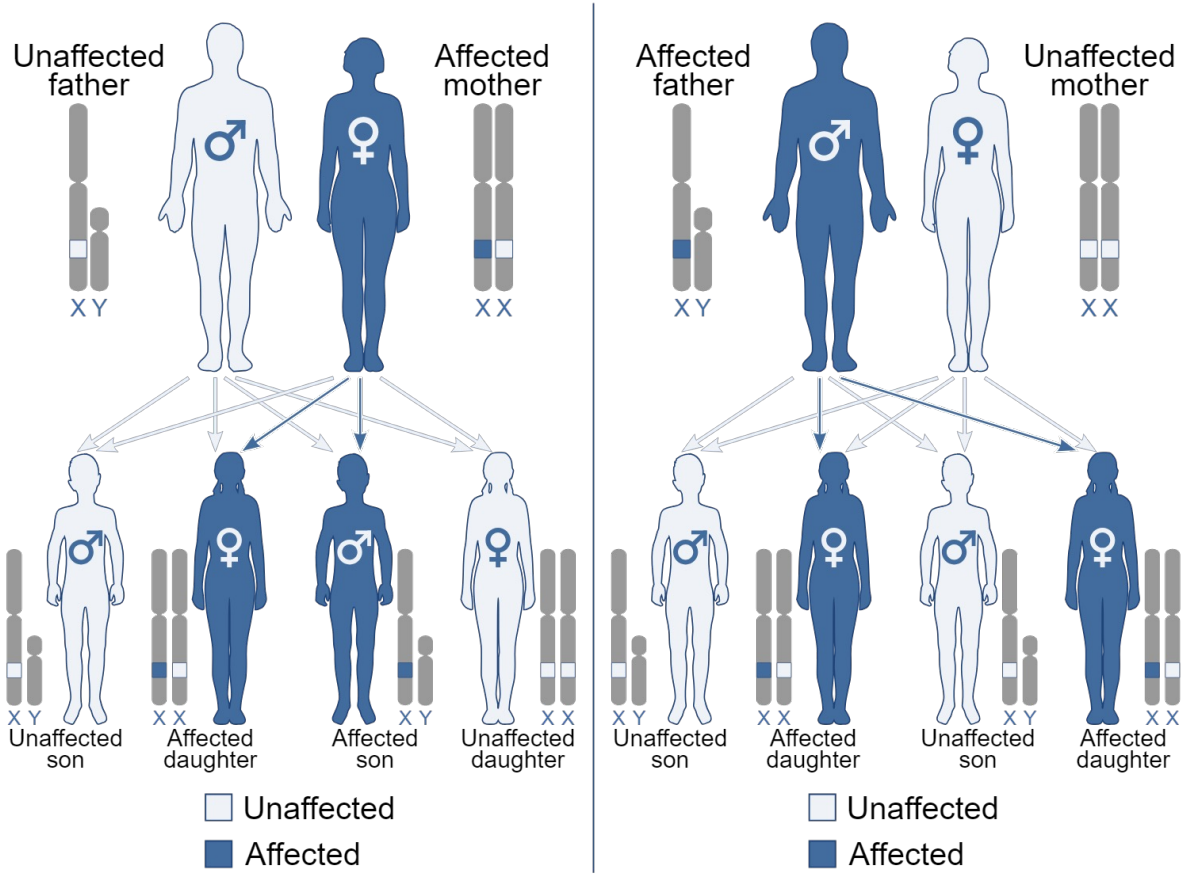
Trio Analysis

- DNA is sequenced of both the patient and parents.
- Discover causal mutations of inherited diseases




Autosomal dominant
Unaffected parent
Affected parent
Unaffected child
Affected child
Affected child
Unaffected child
Unaffected
Affected
Autosomal recessive
Carrier parent
Carrier parent
Unaffected child
Carrier child
Carrier child
Affected child
Unaffected
Carrier
Affected

X-linked dominant



Note: some X-linked dominant disorders are embryonic lethal in males, and most affect females less severely.

Hands-on and Practical Part

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GitHub

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Done with Day 3, Heyyyyy!

Thank You !

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