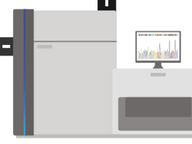
SAMPLE PREPARATION

DNA is extracted and prepared for whole genome sequencing

SEQUENCING



DNA from the whole genome is sequenced using next generation sequencing methods

BIOINFORMATICS

Computers and software tools are used to filter large quantities of data. The filters used depend on the clinical question and sample from the genome that has been sequenced.

Rare disease/cancer

The new DNA sequence is quality assured to ensure the bases are correctly identified.

The genome sequence is 'reassembled'. This is called **sequence alignment**.

The newly sequenced genome is compared against a reference genome to identify variants. This is known as **variant calling**.

This produces a file known as a **variant call file.**

This details all the variants that have been discovered in the newly sequenced genome. Some will detail many hundreds of thousands; some only a few.

Infectious disease

The newly sequenced genomes (from different samples) are quality assured to ensure the bases are correctly identified.

The sequenced genomes are compared to a reference genome and each other.

This enables identification of the exact pathogen.

Which determines any differences between the pathogen genomes from different people.

INTERPRETATION/ANALYSIS

A team of scientists and clinical professionals will work together to review and interrogate the information in order to provide diagnosis, aid treatment and management and identify the source of an outbreak.

They will do this using a variety of tools and resources, depending on the clinical question.

Research and genetic databases

Research and literature

Statistics and modeling

Information about the patient (phenotypic and/or epidemiological data)

HOW RESULTS ARE ANALYSED

Rare disease

Variations in the sample are analysed to:

- Make a diagnosis
- Identify possible treatments

Cancer

Specific variants are identified to:

- Provide targeted treatments
- Ensure patient has access to any relevant clincial trials

Infectious disease

The pathogen sequence is compared to other known outbreaks in order to:

- Identify specific type of pathogen
- Track disease spread and evolution
- Identify source
- And identify effective treatment

REPORT IS GENERATED

Rare disease and cancer

A report detailing the findings of whole genome sequencing is produced.

This report will be sent to the clinical team and used to inform the ongoing management of the patient.

Infectious disease

A report detailing the findings of whole genome sequencing is produced.

This information will be used for infectious control measures and, in some cases, patient management.

REMEMBER !

We don't always find an answer.

This is usually because there is not enough information about a specific variant, or not enough cases to track an outbreak.