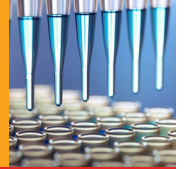


Next Generation Sequencing

Information for health professionals



NGS OVERVIEW

Next Generation Sequencing (NGS) is a cutting-edge technology for genetic testing.

This technology provides a breakthrough in diagnostic yield: in a single test, a gene, several genes (panel) or the whole exome are analyzed at a fraction of the time and cost than before.

NGS APPLICATION

GENE PANELS FOR ALL MEDICAL AREAS

CGC Genetics's NGS panels consist on the simultaneous sequencing of multiple genes associated with a particular disease or phenotype.

We provide over 200 panels for all medical specialties. All NGS panels available at CGC Genetics are custom designed by our team and regularly updated to include additional genes, leading to a high diagnostic yield.

Please find a comprehensive list of all our panels and the latest version of each, at www.cgccgenetics.com

DISEASE EXOME BY CGC GENETICS

The human genome contains 20.000 genes, of which only a fraction are known to be related to disease or clinical phenotype. Disease exome is the largest sequencing panel currently available, including >6 000 clinically relevant genes.

This diagnostic tool is available to physicians and for patients with an uncertain, unspecific or complex phenotype. Disease exome has an average depth of coverage of >100x with >95% of the target regions covered.

WHOLE EXOME SEQUENCING – WES

Whole Exome Sequencing (WES) entails the sequencing of all genomic coding regions, i.e., the exons.

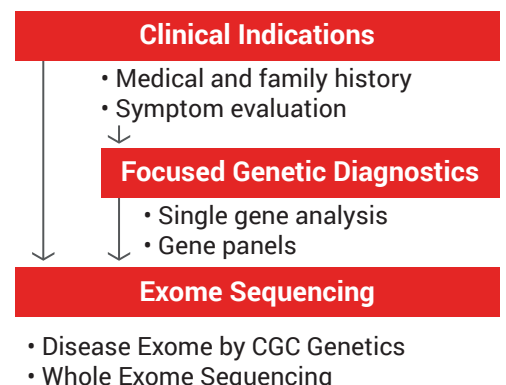
WES purpose is to search for genetic variants throughout the 20 000 genes that make up the exome.

Variants may be either detected in genes related with defined clinical conditions or in genes whose association with disease has not been yet described.

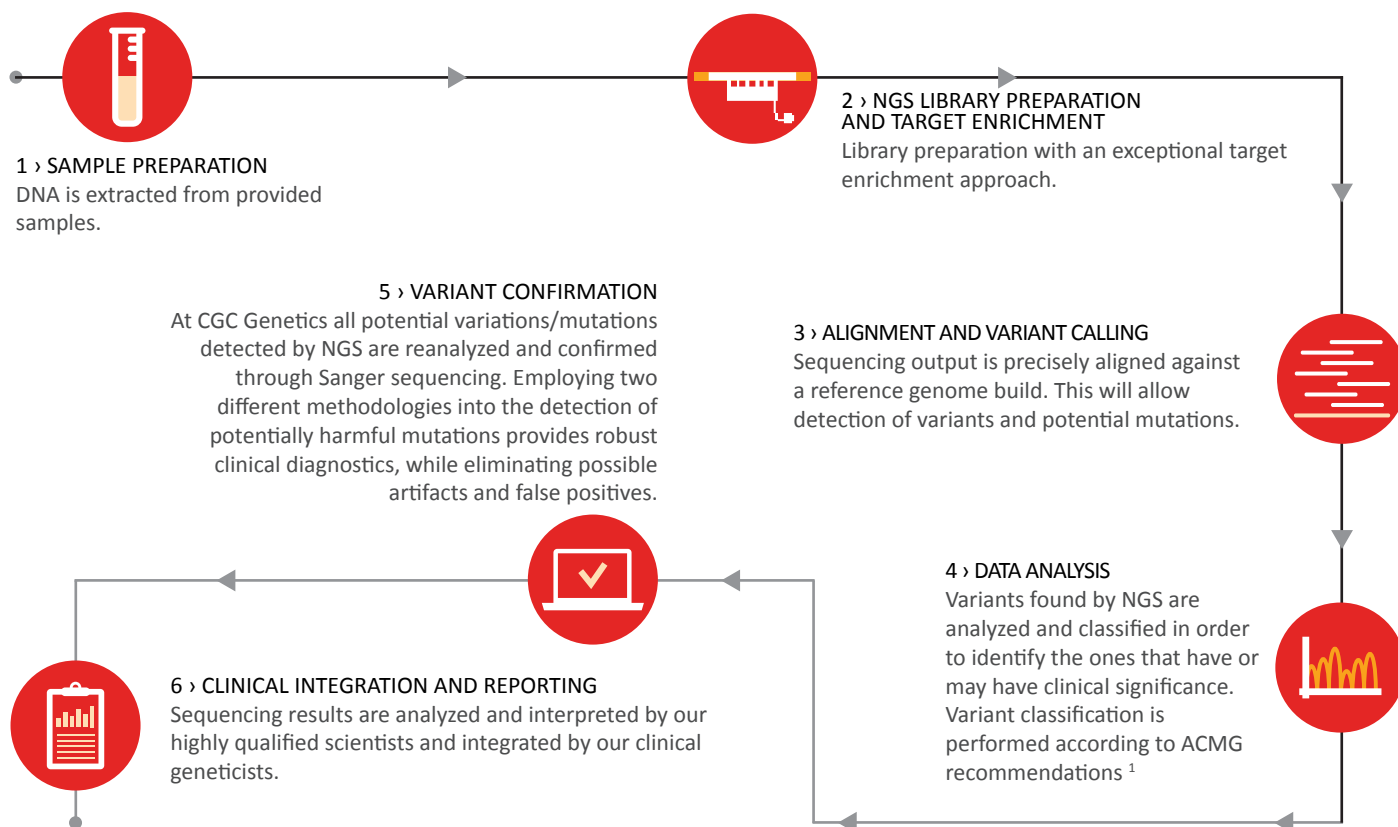
For a more efficient variant filtering, CGC Genetics recommends WES TRIO, i.e., performing WES in both the patient and the parents. With this approach, it is possible that both parents serve as a reference for filtering benign variants, or ascertain the parent-of-origin for each variant, making WES an effective diagnostic tool. WES has an average depth of coverage of 100x, with more than 90% coverage of coding regions.

MITOCHONDRIAL DNA TEST

Exome sequencing only analyzes nuclear DNA, excluding mitochondrial DNA. Mitochondrial DNA contains 37 genes involved in several mitochondrial diseases. CGC Genetics offers a panel for detection of mitochondrial diseases with complete sequencing of the mitochondrial genome.



METHODOLOGY PROCESS



¹ Richards S et al., Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine. 2015, 17(5):405-24.

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