WHOLE-GENOME SEQUENCING (WGS)

WGS is the most informative and detailed genetic test which includes nearly 100% of human DNA (over 6 billion DNA bases).

CellGenetics laboratory specialises in bioinformatic analysis of sequencing data generated by WGS and its interpretation in accordance with all internationally acclaimed standards and guidelines. We offer a wide portfolio of gene panels in:

- ✓Oncology
- ✓Cardiology
- ✓Gastroenterology
- ✓Neurology
- ✓Pulmonology
- ✓Nephrology
- ✓Endocrinology
- ✓Metabolism
- ✓Mitochondrial disorders
- ✓Hematology
- ✓Immunology
- ✓Skeletal disorders
- ✓Dermatology
- ✓ENT
- ✓Ophthalmology
- ✓Reproductive health
- ✓Prevention

What is Whole-genome Sequencing (WGS)?

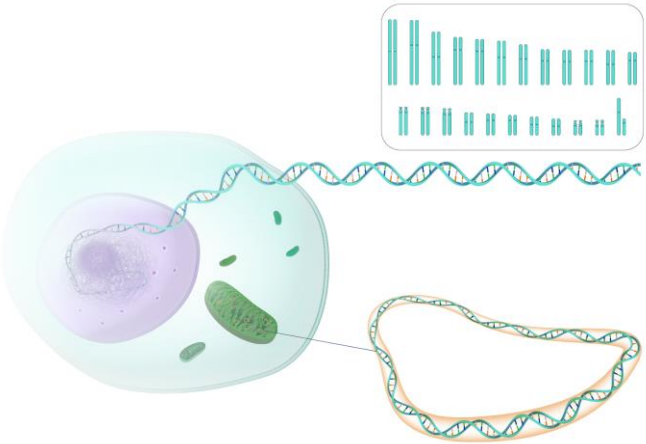
- The most informative and exhaustive genetic test
- Includes nearly 100% of human DNA (over 6 billion DNA bases)
- Applications in all fields of medicine
- Allows for targeted bioinformatic analysis of any gene or panel of genes associated with various diseases
- Includes the noncoding sequence of each gene as well as its regulatory elements—regions often overlooked by most routine genetic studies
- Higher diagnostic sensitivity than exome sequencing, as well as any other genetic test
- Shortens the time to genetic diagnosis for polygenic conditions, diseases with a complex clinical picture and unclear etiology
- Ability to detect copy number variants (CNV – deletions, duplications) as well as structural variations (SV)
- A profitable investment with the possibility of further analyses in the future

WGS can be beneficial for:

- patients with a genetic disease or suspicion of a certain clinical diagnosis
- people with a family history of (hereditary) diseases in order to determine predisposition
- partner couples planning a pregnancy who would like to minimize the risk of disease in their offspring
- people proactive about their health and would like to learn more about their body and physiology, optimizing their lifestyle
- people who would invest in their genome data to save in the long run from smaller laboratory genetic tests that would collectively cost more
- newborns in order to screen for rare early-onset diseases

The **human genome** is a complete set of nucleic acid sequences for humans, encoded as DNA within the 23 chromosome pairs in cell nuclei and in a small DNA molecule found within individual mitochondria. These are usually treated separately as the nuclear genome and the mitochondrial genome. Human genomes include both protein-coding DNA sequences and various types of DNA that does not encode proteins.

Whole genome sequencing (WGS), also known as full genome sequencing, complete genome sequencing, or entire genome sequencing, is the process of determining the entirety, or nearly the entirety, of the DNA sequence of an organism's genome at a single time. This entails sequencing all of an organism's chromosomal DNA as well as DNA contained in the mitochondria.
Whole genome sequencing has largely been used as a research tool, but was being introduced to clinics in 2014.In personalized medicine, whole genome sequence data presents an important tool to guide therapeutic intervention.



What is the difference between Whole-genome sequencing and Whole-exome sequencing?

Whole-genome sequencing (WGS) determines the order of all nucleotides in an individual's DNA and can uncover variation in any part of the human genome, including coding, noncoding, and mitochondrial DNA (mtDNA) regions.

Whole-exome sequencing (WES) focuses on the genomic protein-coding regions (exons). The exome is a relatively small proportion of the whole genome (approximately 2%).

In many cases, WGS is the better option than WES because DNA variations outside protein-coding regions can affect gene activity and protein production, potentially leading to genetic disorders. WGS includes the noncoding sequence of each gene as well as its regulatory elements—regions often overlooked by most routine genetic studies. WGS has higher diagnostic sensitivity than WES. WGS can also detect copy variants (CNV – deletions, duplications) as well as structural variations (SV) – options that WES cannot provide.

What is our workflow?

- Preliminary genetic consultation
- Blood draw and DNA extraction
- Whole-genome sequencing
- Bioinformatic analysis + genetic/ medical interpretation
- Detailed genetic report
- Genetic counseling

"We used to think that our fate was in our stars, but now we know that, in large measure, our fate is in our genes" **– James Watson**

"The genome is a book that wrote itself, continually adding, deleting and amending over four billion years." – **Matt Ridley**

Our portfolio includes but is not limited to:

Prevention	
Prevention /ACMG guidelines/	78 genes
Carrier screen	750 genes

Oncology	**190 genes**
BRCA1+BRCA2	2 genes
Hereditary breast and ovarian cancer syndrome	45 genes
Prostate cancer	30 genes
Colon cancer	30 genes
Fanconi anemia	24 genes
Neurofibromatosis	2 genes

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Cardiology	380 genes
Cardiomyopathy	220 genes
Arrhythmia	220 genes
Congenital heart defects (CHDs)	130 genes
Noonan syndrome	14 genes
Brugada syndrome	26 genes
Ehlers-Danlos syndrome	100 genes
Marfan syndrome	36 genes
Short and Long QT syndromes	20 genes
Familial hypercholesterolemia	24 genes

Neurology	2000 genes
Epilepsy	600 genes
Mental retardation	2000 genes
Autism	480 genes
Neuromuscular disorders	500 genes
Alzheimer's disease, Parkinson's disease and dementia	35 genes

Gastroenterology	150 genes
Colon cancer	30 genes
Pancreatic cancer	20 genes
Stomach cancer	40 genes
Familial adenomatous polyposis (FAP)	4 genes
Lynch syndrome	5 genes
Polycystic liver and kidney diseases	90 genes

Metabolic diseases	650 genes
Obesity	70 genes
Diabetes	100 genes
MODY (Maturity-onset diabetes of the young)	30 genes
Familial hypercholesterolemia	24 genes

Mitochondrial diseases	1800 genes
Mitochondrial genome	37 genes

Nephrology	600 genes
Polycystic kidney disease (PKD)	87 genes
Tubulopathy	40 genes
Nephrotic syndrome	100 genes
Alport syndrome	6 genes

Pulmonology	150 genes
Cystic fibrosis	1 gene
Chronic obstructive pulmonary disease	26 genes
Pulmonary arterial hypertension	23 genes

Immunology	650 genes
Autoimmunity	110 genes
Immunodeficiency	1000 genes
Congenital immunodeficiency	470 genes
Severe combined immunodeficiency (SCID)	80 genes

Hematology	250 genes
Aplastic anemia, spherocytosis, dyskeratosis	156 genes
Congenital hemolytic anemias	40 genes
Fanconi anemia	24 genes
Beta-thalassemia and sickle cell anemia	1 gene
Thrombophilia	25 genes

Ophthalmology	500 genes
Cataract	170 genes
Age-related macular degeneration	24 genes
Retinitis pigmentosa	190 genes
Glaucoma	37 genes
Aniridia	4 genes

Skeletal disorders	600 genes
Osteogenesis imperfecta and bone fragility	75 genes
Connective tissue disease	90 genes
Short stature and skeletal abnormalities	500 genes

Dermatology	250 genes
Familial malignant melanoma	20 genes
Neurofibromatosis	2 genes
Xeroderma pigmentosum	9 genes
Tuberous sclerosis	2 genes
Ichthyosis	60 genes
Epidermolysis bullosa	26 genes

ENT	
Deafness	400 genes
Syndromic deafness	200 genes
Non-syndromic deafness	100 genes
Congenital microtia/anotia	12 genes

Reproductive health	
Female infertility	330 genes
Premature ovarian failure	90 genes
Oocyte maturation defects	8 genes
Male infertility	220 genes
Disorders of spermatogenesis	45 genes
Invitro fertilization /for women/	50 genes