myDNAmap
neurology

myDNAmap
the power of your genetics

myDNAmap is a medical genome company that was established with the aim of helping the general public to access all of the information included in their DNA, so they can better care for their health and that of their family.

myDNAmap offers a unique and comprehensive service in the field of preventative medicine. We sequence the whole genome containing all of the genes associated with a person's health and wellbeing. We provide pre- and post-sequencing advice, which will answer any questions and explain the results. We have developed **myDNAapp,** a mobile app where you can consult your results, contact health and science professionals and enter information regarding your health, which helps to personalise your genetic report. Furthermore, **myDNAmap** offers annual updates, which include the most recent discoveries in the field of genetics.

We know that every one of us is unique and individual just like our DNA.

Knowing your genetic profile will help you to take care of your health and that of your family. Today, tomorrow and always.

Always check **mydnamap.com** for the latest updates of our services.

myDNAmap neurology

Knowing your genetic predisposition to neurological diseases enables you to:

- establish preventative treatment strategies.
- modify habits that contribute to the development of symptoms.
- identify the best tools for a personalised treatment..
- detect family history that must be borne in mind and have genetic counselling to make adequate decisions.

In the **myDNAmap Neurology Panel** we analyse more than **450 genes** associated with monogenetic hereditary neurological diseases. This allows you to be able to act effectively in the prevention, prognosis and treatment of both the most frequent diseases. as well as diseases considered to be more rare or low frequency.

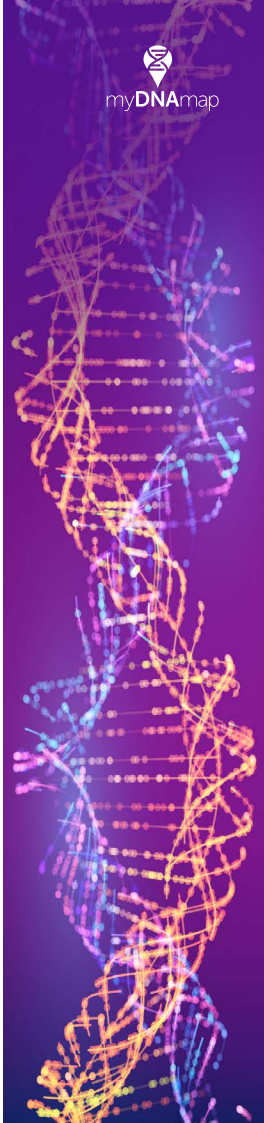
Movement disorders
- Dystonia hereditary
- Parkinson's disease and parkinsonism

Neurodegenerative disorders
- Dementia
- Amyotrophic lateral sclerosis (ALS)
- Hereditary Alzheimer's disease
- Hereditary Parkinson's disease and parkinsonism
- Prion diseases

Neuromuscular disorders
- Comprehensive neurodegenerative disorders panel
- Congenital myasthenic syndrome
- Malignant hyperthermia
- Muscular dystrophy
- Dystroglycanopathy
- Emery-Dreifuss muscular dystrophy
- Limb-girdle muscular dystrophy

Related neuropathy and disorders
- Neuropathies
- Charcot-Marie-Tooth disease
- Hereditary spastic paraplegia

- **Musculoskeletal disorders associated with cardiomyopathy**
- **Epilepsy and disorders due to abnormal brain development**



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Genetic factors play a pivotal role in the development of many diseases, including neurological ones. Knowing our genetic information early allows us to make decisions regarding our health. We are thus able to actively contribute to the prevention, delay in the onset of a disease or alleviate its symptoms.

Neurology is one of the specialisms that is greatly benefiting from the progression of genetics and sequencing technologies, like whole genome sequencing. Genome testing can be used to find out the predisposition to serious neurological symptoms. It is important to know that having a genetic predisposition to a disease does not necessarily mean that this disease will develop. Environmental factors influence its manifestation and it is possible to take control of them preventively.

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Some data regarding neurological diseases

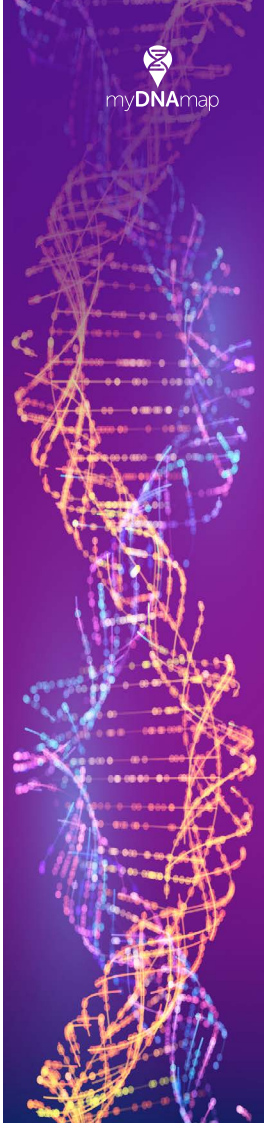
The genetics of neurological diseases is complex as, on occasions, the variants in a gene are determined for the development of the disease, but in other cases, those same genetic variants may be modulated by environmental factors like diet or drug consumption. For this reason, some individuals with different lifestyle habits and the same associated gene can develop the disease, whilst others may not. For this reason, at **myDNAmap app** we gather all information associated with lifestyle habits by means of a questionnaire created by our health professionals. It includes all relevant information for a sound good risk assessment.

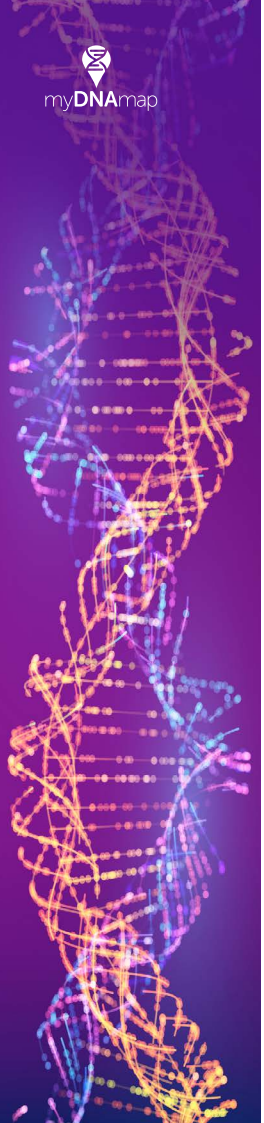
Parkinson's disease

Parkinson's is a neurodegenerative disorder that affects the neurons, which are responsible for controlling movement. These affected neurons do not produce a sufficient quantity of dopamine, the substance that is responsible for controlling a person's voluntary movements. This causes symptoms like trembling, balance and coordination problems, limb stiffness and slowness of movement. In this genetic test we analyse genes known to be most frequently associated with Parkinson's disease.

Alzheimer's disease and other types of dementia

Dementia causes a progressive neurodegenerative process. Different genetic and environmental factors trigger dementia, as well as symptoms. Some of these types of dementia are frontotemporal or vascular. Among them is Alzheimer's disease, which is the most common and represents 60 to 70% of cases.
Alzheimer's disease is a neurodegenerative disorder that causes cognitive deterioration, behavioural problems and affects the ability to undertake daily activities. Between 1 and 5% of cases are early onset Alzheimer's disease (<60-65 years old). The majority of these patients are sporadic cases and approximately 2% are inherited in an autosomal dominant manner, where risk variants in genes *PSEN1, PSEN2* and *APP* are described as the most frequent causes. Signs and symptoms of this (hereditary) type follow an aggressive course and generally appear between the ages of 30 and 40.





On the other hand, approximately 95% of cases are delayed onset Alzheimer's (>60-65 years old). And although variants have been identified in ~20 genes associated with delayed onset Alzheimer's, the apolipoprotein E *(APOE)* genotype has been deemed as a significant predictive factor. This is because a specific genetic profile presents an increased risk of developing the illness. It is important to highlight that a positive result for risk variants in associated genes does not necessarily mean that you will develop the disease, as there are other factors that can operate alongside this such as environmental and lifestyle factors.

This genetic analysis allows us to detect the genetic predisposition of developing the disease, to take measures to delay the symptoms and improve quality of life, as well as detect family history and alert other family members.

Amyotrophic lateral sclerosis

Amyotrophic lateral sclerosis or ALS is a rare neurodegenerative disease that affects neurons in the brain, the brainstem and the spinal cord, which control the voluntary muscle movement. In 10% of cases, the cause is genetic.

Some of the genes studied that are associated with neurodegenerative conditions:

In total, more than 450 genes are analyzed.

Main genes of Parkinson's Disease, Alzheimer's and Amyotrophic Lateral Disease::

APOE, ALS2, APP, ATP, ATP13A2, ATP7B, CHCHD10, DCTN1, DNAJC6, FBXO7, FUS, GCH1, GRN, KIF5A, LRRK2, MAPT, OPTN, PARK7, PFN1, PINK1, PRKN, PRKRA, PRNP, PSEN1, PSEN2, SETX, SLC6A3, SNCA, SOD1, SPG11, SPR, TARDBP, TBK1, TFG, TH, UBQLN2, VAPB, VCP, VPS35

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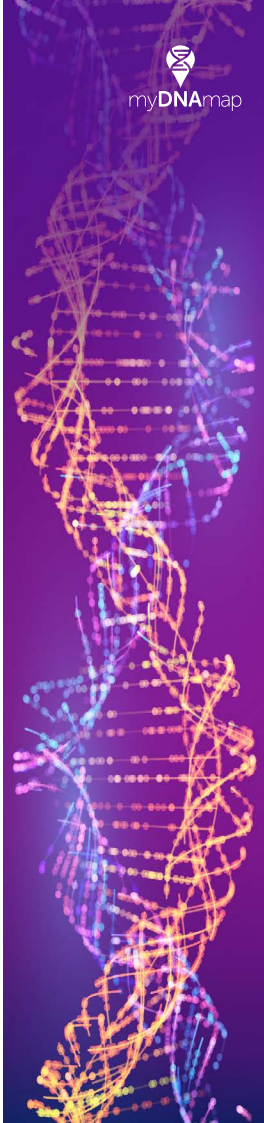
Technology used
Whole genome sequencing (WGS) using Illumina HiSeq X10 or NovaSeq 6000 systems. Illumina PE150, QQ30 ≥80%, aligned with the human reference genome GRCh37/hg19. The classification and analysis of variants is carried out based on the recommendations of the American College of Medical Genetics and Genomics (ACMG). The variants reported are mentioned based on the recommendations of the Human Genome Variation Society (HGVS).

Our report: is the scientific-technical communication of the results. It is written in a precise yet simple manner, intended for both our users and health professionals. It is provided within the framework of a consultation with one of our genetic counsellors. This can take place via videoconference. During the consultation the specialised professional will explain the details, answer any user queries and clear up any doubts that may occur.

Note: the detection of genetic variants only provides a predisposition or potential, but never provides a certainty of developing diseases. Under no circumstances is the genetic information provided valid for diagnostic use. It does not imply the potential to determine the age you will be when it starts nor the type or severity of the disease(s); nor does it allow us to rule out the existence of clinical or genetic heterogeneity.

Bibliography:

- Orphanet, OMIM, Pubmed
- Alzheimer Disease Overview, GeneReviews. Last Update: December 20, 2018.
- Parkinson Disease Overview, GeneReviews. Last Update: February 27, 2014.
- Amyotrophic Lateral Sclerosis Overview, GeneReviews. Last Revision: February 12, 2015.



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