

Neurogenetics

Genetics is the study of genes, genetic variation, and heredity in living organisms.

It is generally considered a field of biology, but intersects frequently with many other life sciences and is strongly linked with the study of information systems.

The discoverer of genetics is Gregor Mendel, a late 19th-century scientist and Augustinian friar. Mendel studied “trait inheritance”, patterns in the way traits are handed down from parents to offspring. He observed that organisms (pea plants) inherit traits by way of discrete “units of inheritance”. This term, still used today, is a somewhat ambiguous definition of what is referred to as a gene.

Neurogenetics is a branch of [genetics](#) that analyzes the impact of [genes](#) on the structure and function of the brain and peripheral nervous system.

7,000 [rare diseases](#) have been identified, most of them are of genetic origin. The diagnosis of a neurogenetic disease is difficult, and management and training programs are not well defined through Europe. To capture and assess care needs, the Neurogenetics Panel of the European Academy of Neurology (EAN) has performed an explorative survey.

The survey covering multiple topics of neurogenetics was sent to all neurologists and neuropsychiatrists affiliated to the EAN practicing in Europe.

Mancuso et al. collected answers from 239 members based in 40 European member states. Even though most of the responders are aware on neurogenetic diseases, when we come to amenability of carrying out a complete genetic diagnosis, almost one third of the responders declare that are not happy with the current way of ordering genetic analyses in their Countries. Furthermore, while single gene analysis is diffusely present in Europe, whole exome and genome sequencing are not easily accessible, with considerable variabilities between Countries. Almost 10% of the responders didn't know if pre-symptomatic and prenatal diagnosis is available in their Countries, and 47,3% were not aware of which newborn screening programs are available. Finally, 96,3% of responders declare that there is a need for education and training in neurogenetics.

They believe that this survey may be of importance for all European stakeholders in neurogenetics in identifying key priorities, targeting areas to encourage education/travel fellowships and educational seminars in the future as this area will only accelerate and diagnostic requirements expand ¹⁾.

¹⁾

Mancuso M, Houlden H, Molnar MJ, Filla A, Breza M, Graessner H, Bassetti C, Boesch S. How to approach a neurogenetics diagnosis in different European countries - The EAN Neurogenetics Panel survey. Eur J Neurol. 2022 Mar 10. doi: 10.1111/ene.15320. Epub ahead of print. PMID: 35271759.

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