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Perspective

Symptoms and Diagnosis of Neurogenetic Disorders

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ABSTRACT

The study of genetics in the development and function of the nervous system is known as neurogenetics. It regards neural characteristics as phenotypes (measureable or not manifestations of an individual's genetic make-up) and is primarily based on the observation that the nervous systems of individuals, even those belonging to the same species, may not be identical. It draws aspects from both neuroscience and genetics studies, as the name implies, focusing on how an organism's genetic code affects its expressed traits.

Keywords: Neurogenetics; Genetic changes; Neurogenetic disorders

DESCRIPTION

Neurogenetic disorders

Neurogenetic disorders are conditions caused by genetic and chromosomal changes. These diseases have an impact on the brain, spinal cord, nerves, and muscles. Neurogenetic disorders can cause health issues at birth or later in life.

Huntington's disease, Kennedy disease, spinocerebellar ataxia, spinal muscular atrophy, hereditary motor neuron disease, early onset muscle disorders, Charcot-Marie-Tooth neuropathies, hereditary spastic paraplegias, Friedreich's ataxia, muscular dystrophies (Duchenne and LGMD), and congenital muscular dystrophies and myopathies are all examples of neurogenetic diseases.

Neurogenetic disease frequently causes severe, often progressive disability. Doctors typically review patients' medical records, study their family history, interview and examine them, and conduct or obtain additional testing, such as genetic testing and neuro-imaging, to diagnose neurogenetic disease.

Gene therapeutic approaches hold out hope for curing some of these diseases (NCBI). The first gene therapy treatment for any neurogenetic disease, for spinal muscular atrophy, was approved in 2019, paving the way for future applications of this approach. Similar treatments are already in the works for Duchenne and other types of muscular dystrophy. This ground-breaking, life-saving treatment exemplifies our core belief that if we can cure one disease, we can cure many.

Neurogenetic disorders come in a variety of forms. While these diseases are all caused by genetic changes, not all of them are inherited.

Neurogenetic disorders are classified as follows:

- Autism Spectrum Disorder (ASD)
- Developmental delay
- Intellectual and cognitive disability
- Epilepsy and seizures (genetic forms)
- Metabolic disorders
- Neurocutaneous disorders (for example,neurofibromatosis)
- Neuromuscular disorders
- Neurodegenerative conditions

Symptoms of neurogenetic disorders

- Children with a neurogenetic disorder may experience the following symptoms:
- Uncontrollable (intractable) epilepsy
- Delayed developmental skills
- Abnormal or uncontrollable movements
- Muscle weakness
- Vision problems (such as retinal abnormalities, cataracts or cortical blindness)
- Hearing issues
- Unusual head growth (such as growing too slowly or too rapidly)

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Received: 01-Aug-2022, Manuscript No. TOA-22-18962; **Editor assigned:** 03-Aug-2022, Pre QC No. TOA-22-18962 (PQ); **Reviewed:** 17-Aug-2022, QC No. TOA-22-18962; **Revised:** 24-Aug-2022, Manuscript No. TOA-22-18962 (R); **Published:** 31-Aug-2022, DOI: 10.35248/2329-8936.22.8.120

Citation: John P (2022) Symptoms and Diagnosis of Neurogenetic Disorders. Transcriptomics. 8:120.

Diagnosis of neurogenetic disorders

To diagnose neurogenetic disorders, doctors at Riley at IU Health perform the following exams and tests:

Clinical history and examination Your child's doctor will examine your child's body and take a detailed history, including an in-depth family history, to look for signs of a neurogenetic disorder, such as certain birthmarks or unusual physical or neurologic features.

A blood test: These could be routine chemistry tests, blood counts, or more specialised metabolic testing to rule out a specific disorder.

Urine analysis: Urine tests can also look for specific chemicals and proteins in the urine that indicate a neurogenetic disorder, which can be used instead of more invasive testing.

Lumbar insertion (spinal tap: If blood and urine tests yield inconclusive results, a lumbar puncture to obtain cerebrospinal fluid may be beneficial.

Biopsy: A skin or muscle biopsy may be required in rare cases to make a specific diagnosis.

Genetic analysis: A child's DNA analysis can reveal changes or abnormalities in genes and chromosomes that result in a neurogenetic condition. Because of advances in molecular genetics, blood tests can now detect specific genetic variations in the blood that are associated with a neurogenetic disorder rather than more invasive lumbar punctures and biopsies.

Magnetic Resonance Imaging (MRI: MRI creates detailed images of internal body tissues such as the brain and nerves by using a magnetic field and radio waves. MRI can detect intracranial signs of certain neurogenetic disorders.

Neurogenetic disorders (such as uncontrollable epilepsy and brain malformations) are first identified through a thorough examination by a specialized doctor who has received additional training to recognize these conditions. Different tests are used to make a specific diagnosis based on these findings. Among these tests are:

- Blood, urine, and cerebrospinal fluid testing.
- Many of the blood tests are genetic tests to see if a child has specific genes that have been linked to neurogenetic disorders.
- Saliva swab.
- Cerebrospinal fluid testing necessitates a lumbar puncture, which is usually done under anaesthesia.
- Brain imaging.
- Muscle, liver, or skin biopsies.

CONCLUSION

Neurogenetic disorders are conditions caused by genetic and chromosomal changes. Neurogenetic disease frequently causes severe, often progressive disability. Gene therapeutic approaches hold out hope for curing some of these diseases. Neurogenetic disorders come in a variety of forms. A child's DNA analysis can reveal changes or abnormalities in genes and chromosomes that result in a neurogenetic condition. Because of advances in molecular genetics, blood tests can now detect specific genetic variations in the blood that are associated with a neurogenetic disorder rather than more invasive lumbar punctures and biopsies. MRI creates detailed images of internal body tissues such as the brain and nerves by using a magnetic field and radio waves.