# Nervous system issues in 22q11.2 duplication syndrome

The nervous system is **a network that relays messages** from the brain to the rest of the body, and from the rest of the body to the brain. It controls the body's response to what's happening both inside and outside the body. When there are problems in the brain or the nerves, the individual may experience a variety of problems in bodily functions, movements, sense of awareness, learning/memory etc.

The "Brain and Nerves Series" provides explanations on problems that have been found in individuals with 22q11.2 duplication syndrome (22q11.2DupS). The **Health Conditions Explained** page on our website contains all the info sheets listed below.

## Brain and Nerves Issues Found in Individuals with 22q11.2DupS

(1) The links lead to the information sheets on the conditions:

- Seizures and Epilepsy
- Hypotonia
- Other Movement Disorders
  - Dystonia
  - Developmental Coordination Disorder (Dyspraxia)
  - Stereotypies (Motor Stereotypies)
- Structural Differences of the Brain
  - Microcephaly
  - Macrocelphaly
  - Chiari Malformation
  - Craniosynostosis

(2) The following brain/nerve condition is explained in info sheets in another series:

- Restless Legs Syndrome and Periodic Limb Movement Disorder (Sleep Series)

The lists above include the known brain and nerve issues in 22q11.2DupS
but not the ones that have not been reported.
It is recommended that patients **receive neurologic exams** every few years.
It may take time and multiple assessments to diagnose nervous system issues accurately.

## Resources


- 22q11.2 duplications – Unique – Rare Chromosome Disorder Support Group
- Severe Pediatric Dystonia Responding to Deep Brain Stimulation in 22q11.2 Microduplication Syndrome: Rare Clinical Presentation – 2024
- Polysomnographic findings in children with 22q deletion & duplication syndrome: relationship to genetic diagnosis, parent-reported symptoms, and calcium levels – 2024
- Cross-sectional and longitudinal findings in patients with proximal 22q11.2 duplication: A retrospective chart study – 2022
- Atypical nested 22q11.2 duplications between LCR22B and LCR22D are associated with neurodevelopmental phenotypes including autism spectrum disorder with incomplete penetrance – 2019
- 22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening – 2016
- Microduplication 22q11.2: A Benign Polymorphism or a Syndrome With a Very Large Clinical Variability and Reduced Penetrance – 2008



22q International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.


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