



What is dup15q syndrome?

Chromosome 15q11.2 - q13.1 Duplication Syndrome (OMIM #608636) "Dup15q Syndrome" is a clinically identifiable syndrome which results from the duplication (or multiplication) of a portion of chromosome 15.

Dup15q syndrome is caused by the presence of at least one extra copy of the Prader-Willi/Angelman critical region (PWACR) within chromosome 15q11.2-q13.1. It can span past these bands but must contain the 11.2 - 13.1 region to be identified as dup15q syndrome.

There is a wide range of severity in the developmental disabilities experienced by individuals with dup15q syndrome. Two people with the same dup15q chromosome pattern may be very different in terms of their abilities and skills.

Symptoms and Characteristics

Physical Features

- Hypotonia
- Strabismus
- Minor Unusual Physical Features
- Growth affected in 20-30%
- High Palate
- Dental Issues

Developmental and Behavioral

- Fine Motor Delay
- Gross Motor Delay
- Cognitive Disability
- Behavioral Issues
- Anxiety Disorders
- Sensory Processing Disorder
- Autism Spectrum Disorder/Autism Symptomology
- Speech Delay
- Learning disabilities
- ADHD

Medical

- Seizure Disorders
 - Infantile Spasms, Epilepsy, LGS
 - Developmental Epileptic Encaphaly
 - Over half of the dup15q population will have at least 1 seizure
- Increased Risk for Sudden Death
 - The risk is estimated at 0.5-1% per person per year.
- Sleep Disturbances
- GI Issues

Why use the term dup15q syndrome?

There are many other genetically derived names that are commonly used in the diagnosis for Dup15q Syndrome, including; 15q11.2-q13.1 Duplication Syndrome, Inverted duplication 15 (inv dup15), Partial trisomy 15, Isodicentric chromosome 15 syndrome [Idic(15)], Supernumerary marker chromosome 15 (SMC15), Partial tetrasomy 15q, etc. All of these names describe the genetics of "Dup15q Syndrome".

As with most rare genetic disorders, in the absence of gene therapies, clinical treatment, and support for all the genetic variations of Dup15q Syndrome is based on the presenting symptoms not based upon the genetic mutation. While a genetic diagnosis may not directly change medical care, it often has significant clinical and personal utility in terms of anticipatory guidance, genetic counseling, eligibility for relevant research studies, and access to patient support organizations.



Diagnosing with Genetic Testing

- **Microarray** - DNA tests that are used to measure "copy number" of the chromosomes.
- **Karyotype or Chromosome Study** - chromosomes are counted and the pairs are matched up, then the banding pattern is examined for abnormalities.
- **FISH Test** - uses a fluorescent piece of DNA that marks a specific spot on chromosome 15 to determine whether it is found in the duplication and, if so, how many copies there are.
- **Methylation Assay** - uses a chemical modification of the DNA to distinguish whether the duplication is on a chromosome that came from the mother or the father.



Dup15q Alliance

We are parents, grandparents, professionals, and advocates providing support for patients, families, and caregivers. We are passionate about personal connections, family support, community awareness, and scientific research.

Dup15q Alliance connects families online through our Parent Support Facebook Group and virtual connection groups, face-to-face through regional gatherings, and at our family conferences. In addition to connecting families, Dup15q Alliance has several family support programs for each stage of the dup15q journey.

Dup15q Alliance funds the LADDER Database and works closely with pharmaceutical partners to identify research in varying modalities and endpoints. Our goal is to improve clinical care and discover treatments, such as behavioral and drug therapies, targeted specifically for Dup15q Syndrome.

We have 17 operating clinics providing standardized care for patients with 15q disorders. Our clinics serve as a platform for robust research, including the collection of integral natural history data.



Our Mission

We empower individuals living with dup15q syndrome and other related rare diseases to reach their full potential by advancing breakthrough research and life-changing therapeutic treatments, supporting families affected by dup15q, and promoting advocacy.

We envision a world where families, clinicians, and advocates enable dup15q individuals to thrive.

Research

- Research and Fellowship Grants
- International Scientific Symposia
- Natural History Study
- Targeted Clinical Trials Support

Clinical

- LADDER Learning Network (Clinician and Research)
- Ladder Database
- Dup15q Clinics

Education & Outreach

- International Family Conference
- Ask The Expert
- Programs and Family Support
- Connecting Families and Outreach
- Newly Diagnosed Support and Mentorship
- Targeted information and resources throughout the stages of growth and development
- Family Crisis and Bereavement Support



How to connect patients to the Alliance

Contact Information

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