Dup15q Alliance

What is dup15q syndrome?

Chromosome 15q11.2 – q13.1 Duplication Syndrome (OMIM #608636) "Dup15q Syndrome" 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
- High Palate
- Dental Issues
- Minor Unusual Physical Features
- Growth affected in 20–30%

Developmental

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

Medical

- Seizure Disorders
  Over half of the dup15q population will have at least 1 seizure
- Increased Risk for Sudden Death
  The risk is small, estimated at 0.5-1% per person per year.

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.

together we believe

Dup15q Alliance

together we achieve

Dup15q Alliance

Dup15q Alliance provides family support and promotes awareness, research and targeted treatments for dup15q syndrome.

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

Dup15q Alliance connects families online through our Parent Support Facebook Group and face-to-face through regional gatherings and conferences. In addition to connecting families, Dup15q Alliance funds and encourages a variety of research studies. Our goal is to improve clinical care and to discover treatments, such as behavioral and drug therapies, targeted specifically for Dup15q Syndrome.

We have 20 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

Through our work to raise awareness and promote research into chromosome 15q duplications, we seek to find targeted treatments so that affected individuals can live full and productive lives. Together with our families, Dup15q Alliance is working towards a better tomorrow for children with chromosome 15q11.2-13.1 duplication (dup15q) syndrome.

Research

- Research and Fellowship Grants
- Scientific meetings
- Clinic trials

Clinical

- Clinical Research Network
- Database

Education & Outreach

- International Family Conference
- Newsletter
- Website

How to connect patients to the Alliance

Contact Information
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