Chromosome 15q11.2–13.1 Duplication Syndrome

Chromosome 15q11.2–13.1 Duplication Syndrome (OMIM #608636) "Dup15q Syndrome" is a clinically identifiable syndrome that 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 15q11.2-13.1. It can span past these bands but must contain the 11.2 - 13.1 region to be identified as dup15q syndrome.

Dup15q syndrome is one of the most common copy number variations associated with autism spectrum disorders, intellectual disability, and infantile spasms. Infantile spasms in dup15q often progress to Lennox Gastaut syndrome and other complex seizure patterns that may be difficult to control. Intractable epilepsy in dup15q may result in disabling secondary effects, including falls or developmental regression. This occurs in more than half of individuals with frequent, uncontrolled seizures or non-convulsive status epilepticus.

Physicians: visit www.dup15q.org/physicans for more information.
NIH Gene Review: https://www.ncbi.nlm.nih.gov/books/NBK367946/

Clinical Features

Physical Features

- Minor Unusual Physical Features
- Downslanting palpebral fissures
- Moderate-to-Severe Hypotonia
- Wide-based or Ataxic Gait
- Low-set ears
- High-arched palate
- Dental Issues
- Strabismus
- Growth affected in 20–30% resulting in small stature

These features are typically subtle and may be missed in infancy.

Behavioral

- Autism Spectrum Disorder/Autism Symptomology
- Sensory Processing Disorders
- Attention Deficit Disorders
- Anxiety Disorders

Developmental

- Hypotonia
- Cognitive Disability
- Motor Delays
- Speech Delay
- Learning disabilities

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 small, estimated at 0.5-1% per person per year.

- Sleep Disturbances
- Gastrointestinal Issues

Note:

Patients with dup15q syndrome feature a distinctive electroencephalography (EEG) signature or biomarker in the form of high amplitude spontaneous beta frequency (12–30 Hz) oscillations. * These EEG disturbances are in the absence of seizures.

Patients with dup15q syndrome have shown abnormal sleep physiology with elevated beta power, reduced spindle density, and reduced or absent SWS compared to age-matched neurotypical controls. * These EEG disturbances are in the absence of seizures.

Compared to children with nonsyndromic ASD, children with dup15q-ASD demonstrate a distinctive behavioral profile with relative strength in items related to social interest, including preserved responsive social smile and directed facial expressions towards others.

It is important to note there is a wide range of severity in the developmental disabilities experienced by individuals with dup15q syndrome.

LADDER
Learning Network

For Patients. For Providers. Together, Toward The Cure.

A Partnership Of angelman Dup15q Alliance

LADDER Learning Network

The LADDER Learning Network strives to provide the best possible care to those affected with dup15q syndrome through our Dup15q Clinics while also collecting clinical research data into the LADDER database.

Dup15q Clinics also link providers together to share information about their most challenging cases.

Additionally, the LADDER Learning Network is a collaborative group that offers educational conversations between researchers and medical professionals involved in treating those with dup15q syndrome, along with advocacy groups, and biopharma companies.

4 essential functions of the LADDER Learning Network:

- Connect Patients to Care
- Connect Providers to Providers
- Clinical Trials
- LADDER Database

Get Connected:

Dup15q Alliance

SCAN ME

Professional Advisory Board

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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.

How to connect patients to the Alliance

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