**Medical Seizure Disorders**

- Increased Risk for Sudden Death
- Sleep Disturbances
- Gastrointestinal Issues

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.

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**Clinical Features**

**Physical Features**
- Minor Unusual Physical Features
- Downslanting palpebral fissures
- Moderate-to-Severe Hypotonia
- Wide-based or Ataxic Gait
- Growth affected in 20–30% resulting in small stature

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

**Medical**
- Seizure Disorders
  - Infantile Spasms, Epilepsy, LGS
  - Developmental Epileptic Encephalopathy
  - 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:**

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**See Physicians Advisory:** https://dup15q.org/physician-advisory/
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:

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

Contact Information
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