Join Our Community

Our website contains resources for families and professionals who want to learn more about STAG1

- **Connect with our private Facebook support group**
- **Read family stories**
- **Learn about research and STAG1 published medical articles**
- **Enroll in our CORDs patient registry**
- **View our STAG1 merchandise store**
- **Stay up to date on news by Facebook and Instagram social media**
- **Volunteer / host a fundraiser**
- **Make a donation**

A Patient Led 501(c)3 Nonprofit Organization

We envision a world in which those with the STAG1 gene mutation are known, valued, and equipped to meet their greatest potential.

www.stag1gene.org

SUPPORT • ADVOCATE • EDUCATE

STAG1

GENE FOUNDATION

Finding One Another Since 2017

www.stag1gene.org

What is STAG1?

A rare genetic neurodevelopmental syndrome involving multiple congenital anomalies in a spectrum of mild to severe symptoms such as:

- **Global Developmental Delay**
- **Variable Degrees of Intellectual Disability**
- **Facial Dysmorphism (High Nasal Bridge, Deep-set Eyes, Wide Mouth)**
- **Feeding difficulties and/or Gastro-esophageal reflux**
- **Seizures**
- **Hypotonia**
- **Autistic features**
- **Joint laxity**
- **Brain imaging may show non-specific features such as Cerebral atrophy**

SYSTEM Recommended Initial Evaluations and Treatments

To be shared with your medical team

Neurological

Initial: Consider brain MRI and EEG baseline if seizures are present, evaluate muscle tone
Treatment: Treat seizures if present, physical therapy, occupational therapy,

Gastrointestinal

Initial: Assess feeding, gastroesophageal reflux, prenatal & postnatal growth delay
Treatment: Feeding education

Psychological & Psychiatric

Initial: Developmental evaluation, autism spectrum disorder, neuropsychological evaluation, sleep disorders
Treatment: Treat behavioral issues if needed, ABA therapy.

Speech & Language

Initial: Speech & language evaluation.
Treatment: Intensive speech and language therapy aimed at speech apraxia. PROMPT therapy.
Augmentative and alternative communication devices.

Musculoskeletal

Initial: Growth parameters, lower limb differences, scoliosis, physical therapy evaluation, occupational therapy evaluation.
Treatment: PT therapy, OT therapy, adaptive equipment if needed.

Cardiovascular

Initial: Evaluate for congenital heart malformations
Treatment: Echocardiogram, pulse oximetry

Research

Mutations in the STAG1 gene protein have been demonstrated to cause multisystem developmental disorders known as “cohesinopathies”. The clinical phenotype associated with STAG1 seems to be milder compared to other cohesinopathies, such as, Cornelia de Lange syndrome. STAG1 pathogenic variants are to date poorly characterized. More patients and phenotype correlation studies are needed to collaborate this data and better characterize the clinical spectrum of this rare cohesionopathy.

Scan the QR code or visit our webpage to access the CORDS Registry & the Cohesinopathy Biobank Research Study