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

Finding One Another Since 2017

SUPPORT • ADVOCATE • EDUCATE

STAG1

GENE FOUNDATION

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

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 including Speech
Variable Degrees of Intellectual Disability
Facial Features
- **High Nasal Bridge**
- **Deep-set Eyes**
- **Wide Mouth**

Feeding difficulties and/or GERD
Seizures
Hypotonia
Autistic features
Joint laxity
Brain MRI or ultrasound findings
Microcephaly
Growth (Small Size)

SYSTEM

Recommended Initial Evaluations and Treatments

To be shared with your medical team

Neurological

Eval: Consider brain MRI and EEG baseline if concern for seizures, evaluate muscle tone
Treatment: Treat seizures if present, physical therapy, occupational therapy,

Gastrointestinal

Eval: Assess feeding, gastroesophageal reflux, prenatal & postnatal growth delay
Treatment: Feeding intervention

Psychological & Psychiatric

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

Speech & Language

Eval: Speech & language evaluation.
Treatment: Speech therapy, augmentative communication devices

Musculoskeletal

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

Cardiovascular

Eval: Evaluate for congenital heart malformations
Suggest: Echocardiogram, pulse oximetry

RESEARCH

Mutations in the STAG1 gene cause a multisystem development disorder. The clinical phenotype associated with STAG1 shares some but not all features with a broader class of related conditions called "cohesionopathies," which include Cornelia De Lange syndrome.

The STAG1-related condition and the gene variants that cause it remain poorly characterized. More patients and phenotype correlation studies are needed to characterize this rare cohesionopathy.

Scan the QR code to access the CORDS Registry & the Cohesinopathy Biobank Research Study