STAG1 GENE FOUNDATION

info@stag1gene.org
www.STAG1gene.org

**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.

## 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)**

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

## RESEARCH

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

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.

**To be shared with your medical team**

| SYSTEM | Recommended Initial Evaluations and Treatments |
|---|---|
| **Neurological** | **Eval:** Consider brain MRI and EEG baseline if concern for seizures, evaluate muscle tone<br>**Treatment:** Treat seizures if present, physical therapy, occupational therapy, |
| **Gastrointestinal** | **Eval:** Assess feeding, gastroesophageal reflux, prenatal & postnatal growth delay<br>**Treatment**: Feeding intervention |
| **Psychological & Psychiatric** | **Eval:** Developmental evaluation, autism spectrum disorder, neuropsychological evaluation, sleep disorders<br>**Treatment:** Treat behavioral issues if needed, ABA therapy. |
| **Musculoskeletal** | **Eval**: Growth parameters, lower limb differences, scoliosis, physical therapy evaluation, occupational therapy evaluation.<br>**Treatment:** PT therapy, OT therapy, adaptive equipment if needed. |
| **Speech & Language** | **Eval:** Speech & language evaluation.<br>**Treatment:** Speech therapy, augmentative communication devices |
| **Cardiovascular** | **Eval**: Evaluate for congenital heart malformations<br>**Suggest:** Echocardiogram, pulse oximetry |