What is the OSA?

The Overgrowth Syndromes Alliance (OSA) is a united group of patient advocacy organizations representing overgrowth disorders. Founded by the Malan Syndrome Foundation and the TBRS Community, OSA aims to:

- Align the interests and resources of overgrowth syndrome patient advocacy organizations.
- Prioritize research for treatments and cures, especially those that would benefit the greatest number of overgrowth syndrome patients and address their highest priorities.
- Address/incorporate patient perspectives into clinical and research priorities.

Or visit the Overgrowth Syndromes Alliance on LinkedIn

For more information about OSA, please email us at: info.overgrowth@gmail.com.

Disclaimer

This informational resource was created by Maddy Jones, MS in genetic counseling candidate, for the Overgrowth Syndromes Alliance, 2024, and should not be used for medical decision-making.

Photo references:

Front (from left to right): TBRS, Age 19; TBRS, Age 38

Page 2 (from top to bottom): Malan, Age 2; Malan, Age 9; TBRS, Age 3; TBRS, Age 18; Sotos, Age 18; TBRS, Age 24; TBRS, Age 5; Malan, Age 19

For more information about specific OGID syndromes including support organizations, provider and patient resources, and clinical descriptions, please visit our LinkTree: https://linktr.ee/overgrowthsyndromesalliance

TBRS community
www.tbrsyndrome.org

malan syndrome foundation
www.malansyndrome.org

A Guide to Overgrowth-Intellectual Disability (OGID) Syndromes

OSA Overgrowth Syndromes Alliance

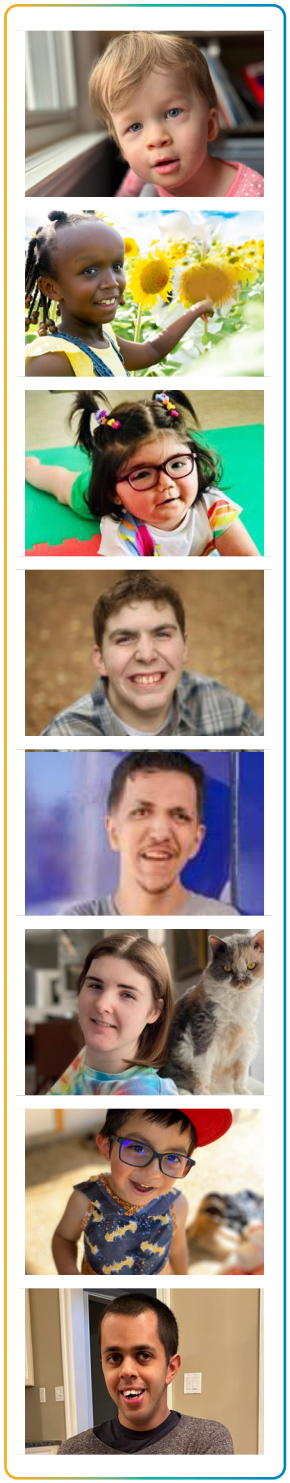
What are OGID syndromes?

Overgrowth-Intellectual Disability (OGID) syndromes are a collection of rare genetic disorders with overlapping clinical features. In addition to the cardinal features of increased growth and some degree of intellectual disability, other clinical findings of OGID syndromes can include:

- Neurodevelopmental and mental health disorders, including autism and anxiety
- Musculoskeletal conditions, including advanced bone age, hypermobility, and scoliosis
- Neurological symptoms, including hypotonia and seizures

Some OGID Syndromes include:

- Beck-Fahrner
- CHD8-related Neurodevelopmental Disorder
- Cohen-Gibson
- Imagawa-Matsumoto
- Kosaki
- Luscan-Lumish
- Malan
- Simpson-Golabi-Behmel
- Smith-Kingsmore
- Sotos
- Tatton Brown Rahman
- Weaver



Diagnosis and treatment of OGID syndromes:

If an OGID syndrome is suspected, a clinical genetics assessment and genetic testing is recommended to confirm a specific diagnosis. Since many OGIDs have overlapping clinical features, it can be difficult to establish a diagnosis from a clinical assessment alone. A molecular diagnosis is important as it allows one to obtain information about the natural history and prognosis of a specific condition and also to develop an optimal management/treatment plan. OGID syndromes are a complex, clinically overlapping group of conditions and inaccurate diagnosis can lead to substandard diagnosis and management of important associated health conditions.

One of the challenges associated with genetic testing is the possibility of a report of a variant of uncertain significance (VUS), i.e., an uncertain diagnosis. Due to a number of factors, VUS results are commonly reported for OGID genes, and present a challenge for clinical management. Patient organizations are equipped to support an individual with a VUS; they can also be referred to the Overgrowth Syndromes Alliance.

Treatments for OGID syndromes focus on managing syndrome-related symptoms, which will vary between the different OGID syndromes. Treatments will also be individualized to each patient, based on their specific presentation. Common specialties for referral include:

- **Neurology:** Evaluation of brain malformations and seizure activity
- **Developmental Pediatrics:** Evaluation/management of developmental and behavioral issues
- **Psychology/Psychiatry:** Assessment/management of behavioral, psychological and psychiatric needs
- **Cardiology:** Evaluation of congenital heart defects, arrhythmias, and other cardiovascular findings
- **Genetics:** Molecular testing, diagnosis, and possible coordination of clinical care
- **Orthopedics:** Assessment of bone, joint, and muscle abnormalities
- **PT/OT/SLP:** Access to early intervention services, academic support, and strengthening function in activities of daily living

This list is not exhaustive and does not include disease-specific recommendations. For professional resources and more information, please visit our LinkTree: