

Clinical Exams/Tests recommended for children with BBSOAS

- A developmental assessment to identify areas of impairment and allow for early intervention.
- A comprehensive psychological evaluation for autism
 - *ADI-R and ADOS testing performed by a certified clinical psychologist.
- EEG if seizures are suspected.
- Brain MRI, recommended at age three years or older.
- Full, dilated eye examination by an ophthalmologist every two years.
- Full hearing evaluation every two years.



Treatment:

Currently there is no treatment for BBSOAS.

However, there are treatments for some of the symptoms:

- Anti epileptic drugs (AEDs) for seizures/epilepsy
 - ACTH or vigabatrin (Sabril TM) for infantile Spasms
- ADHD medications (guanfacine and clonidine preferred for patients with epilepsy)

Prevalence:

There are roughly 200 known cases worldwide as of 2022.

Therapies to consider:

- Visual services and/or vision therapy, focused on CVI
- Physical therapy
- Occupational therapy
- Speech therapy, consideration of sign language and alternative communication devices
- ABA therapy if a diagnosis of autism spectrum disorder is made (can also be considered for ADHD)
- Music therapy
- Hippotherapy

CONTACT US:

BBSOAS Parents Only Support Group (Facebook)
NR2F1 Foundation (Facebook)
NR2F1 Foundation webpage - www.NR2F1.org

Does someone you love have BBSOAS?
 December 7th is our worldwide awareness day.
 Show support by wearing teal, coral, or stripes.



UNDERSTANDING BBSOAS

Bosch-Boonstra-Schaaf
Optic Atrophy Syndrome