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)

Incidence Rate:
It is estimated that BBSOAS affects 1 out of every 250,000 babies born

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

Clinical Exams/Tests recommended for individuals 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.

CONTACT US:
www.NR2F1.org
hello@nr2f1.org

BBSOAS Parents Support Group (Facebook)
NR2F1 Foundation (Facebook & Instagram)

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

BBSOAS stands for Bosch-Boonstra-Schaaf Optic Atrophy Syndrome which is an ultra-rare neurological condition caused by a variant of the Nr2f1 gene. BBSOAS is characterized by a wide array of clinical features, but the most common are vision impairment, developmental delay, and intellectual disability. There are currently a few hundred known cases of BBSOAS worldwide with diagnoses rapidly increasing every month.

Did you know?

The majority of cases are De Novo, meaning the mutation or deletion was not passed on by either parent. The mutation or deletion occurs spontaneously during DNA replication in the process of cell division in a fetus.

An individual with BBSOAS has a 50% chance of passing the mutation/deletions onto any children they may have. This is due to what's known as autosomal dominant inheritance.

Parents with one child with BBSOAS have a less than 3% chance of having another child with BBSOAS.

Understanding Symptoms:

There are five major impairments or disorders associated with BBSOAS. Learning to recognize the symptoms of each is very helpful not only for a diagnosis, but also for determining the areas where an individual needs therapy or extra help.

1. Autism Spectrum Disorder (ASD) – Over 80% of individuals diagnosed with BBSOAS are also diagnosed with or exhibit features of an autism spectrum disorder.

2. Speech and Language Impairment – One of the most common features of BBSOAS is language or speech impairment, with 91% diagnosed with a speech delay. Types of Speech & Language Disorders: Apraxia of Speech; Expressive Language Disorder; Receptive Language Disorder; Other Common Speech and Language Disorders: Stuttering (stammering), Dysarthria (slurred speech), Lisping, Spasmodic Dysphonia (causes the voice to break or sound strained), Muteness and Selective Mutism.

3. Vision impairment – Vision problems are very common among those diagnosed with Bosch-Boonstra-Schaaf optic atrophy syndrome, as about 90% have some form of visual problem. Common visual impairments among individuals with BBSOAS include: optic atrophy (82%), alacrima (78%), manifest latent nystagmus (52%), optic nerve hypoplasia (49%), and cortical vision impairment (68%).

4. Epilepsy and/or Seizures – About half of everyone diagnosed with BBSOAS has also been diagnosed with epilepsy or a seizure disorder. Some common types of seizures seen in individuals with BBSOAS are infantile spasms, focal seizures, absence seizures, generalized clonic-tonic seizures, atonic seizures, and myoclonic seizures.

5. Hypotonia – Hypotonia, or low muscle tone, is seen in over 90% of those with BBSOAS, and can cause its own set of problems.

Other symptoms can include:

- Developmental/Cognitive Impairment
- Extreme love of music
- High pain tolerance
- Oromotor dysfunction
- Repetitive behavior not associated with ASD
- Attention-deficit hyperactivity disorder
- Hearing impairment
- Spasticity
- Mild and inconsistent dysmorphic facial features
- Thin corpus callosum on brain MRI

A diagnosis is made by genetic testing through full exome sequencing or NR2F1 specific sequencing. Microarray testing will detect full deletions of the NR2F1 gene but will not detect mutations. Genetic testing can be done by taking a small blood sample from the patient or before birth through chorionic villus sampling (CVS).