Living with Bainbridge-Ropers Syndrome

ASXL3-related disorder

A diagnosis of ASXL3-related disorder (Bainbridge-Ropers Syndrome, BRS) for your loved one likely brings a range of feelings about the future. This rare genetic condition impacts many systems of the body and can cause significant, lifelong challenges. Although there is still much to be learned about BRS, the ASXL community of families, doctors, and researchers have learned and continue to learn many lessons about how to best manage symptoms and complications of BRS.

How to use this guide: Although not all of the symptoms discussed in this guide will present for each person with BRS, the various systems highlighted throughout are closely interconnected, so treatment of one symptom may work to improve others.

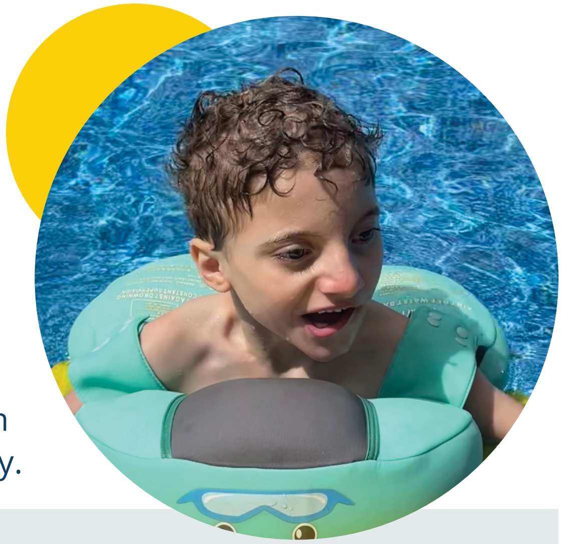
This guide provides an overview of the most common symptoms and their management. Medical appointments can be overwhelming, so refer to the education provided in this guide at your own pace and as often as necessary. The ASXL Rare Research Endowment (ARRE) Foundation is here to support your family so that you can feel more confident about today, tomorrow, and all of the tomorrows to come.

ARRE
ASXL RARE RESEARCH ENDOWMENT
arrefoundation.org/bainbridge-ropers

Communication & learning

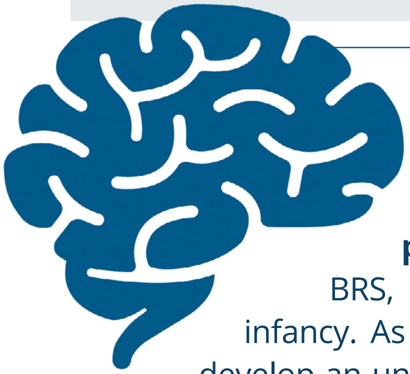
All known individuals with BRS have **delayed speech and language development or absent speech.** Research has shown that receptive language skills (the ability to understand language) may be stronger or easier to obtain than expressive language skills (the ability to communicate wants or needs).

Many affected individuals may be able to attend mainstream schools with special education provisions, although the majority have moderate-to-severe intellectual disability.



What you or your doctor can do

- Evaluate motor, adaptive, cognitive, and speech skills
- Early intervention/special education/individualized education plan (IEP) evaluation
- **Physical, occupational, and speech therapy** (which may be included in an IEP)
- Encourage basic vocalizations and try to associate gestures with vocalizations
- Augmentative and alternative communication devices (AAC) evaluation
- Specialized school instruction that challenges using a 504 plan (in the United States)
- Assess developmental progress and educational needs at each medical visit



Brain-body connection

Low muscle tone (hypotonia) is common in BRS, especially during early infancy. As children age, they may develop an unusual posture with bent elbows and wrists and flexed fingers. Skeletal abnormalities are common and may include scoliosis and an unusually large range of movement (joint hypermobility). Some individuals have extremely long arms for their height (marfanoid habitus), abnormalities of their fingers and toes, and delayed aging of their bones.

Although one-third of individuals with BRS have some sort of **seizure**, they typically respond well to standard medications. Some individuals do have abnormal MRI findings of the brain.

What you or your doctor can do

- Neurologic evaluation with brain MRI and EEG if seizures are present
- Evaluation of motor skills and for skeletal abnormalities/mobility
- **Physical and occupational therapy** to improve both gross and fine motor skills
- Use of adaptive strollers, wheelchairs, bath chairs, walkers, and orthotics
- Orthopedic procedures, medications used in Parkinson's disease or multiple sclerosis, or common muscle relaxants or paralyzers may aid muscle tone abnormalities

Behavioral concerns & sleep

There is a wide range of behavioral and social difficulties for people with BRS. Research shows that approximately 50% of affected individuals meet the formal clinical diagnostic criteria for **autism spectrum disorder**.

Sleep disturbances including **interrupted breathing during sleep (sleep apnea)** and irregular breathing patterns are common in BRS and may have an effect on behavior. These issues can be managed with standard treatments if present.

For those individuals displaying traits shared with autism spectrum disorder, applied behavior analysis-based therapy, which focuses on behavioral, social, and adaptive skills, may be helpful.

Some associated behaviors may include hand flapping, agitation, hyperventilation episodes, and teeth grinding (bruxism). Self-harm behaviors such as head banging, face scratching, and self-biting have been noted and may begin as early as 2 years of age or later in life. **Serious aggressive or destructive behavior can be assessed and managed by a pediatric psychiatrist.**

What you or your doctor can do

- Neuropsychiatric evaluation
- For those older than 1 year, monitor for sleep disturbances. Any sleep apnea should be treated with standard management.
- For those older than 1 year, screen for attention deficit disorder and traits suggestive for autism spectrum disorder. **Children may benefit from applied behavior analysis-based therapy** if any autistic-like traits are discovered.

Family & community

Families caring for an individual with BRS may **benefit from the involvement of social workers/case managers** to aid in access to local resources, respite, and support. Most individuals with BRS will require support throughout their lives.

Individuals with BRS should be referred to early-intervention therapies. ***They need as much therapy—physical, occupational, speech, etc.—as possible to challenge them and prevent developmental regression. Slow or minimal developmental gains are okay; the goal is to avoid regression.***

Growth & feeding

Most babies with BRS have normal birth weights, but their postnatal growth often declines due to feeding issues during infancy, such as **high arched palate, poor suck and swallow, recurrent vomiting, and gastroesophageal reflux disease (GERD)**. Because the range of severity of feeding-related issues vary, some issues may be improved with the use of slow-flow nipples but others may require the surgical placement of a tube directly into the stomach (G tube) or the small intestine (GJ tube) to allow liquid nutrition and medications to be given. Even for those individuals who show improvement in feeding issues with age, there may be ongoing food aversion, texture sensitivity, and behavioral issues that affect eating. Both weight and height tend to normalize for age after feeding issues are managed appropriately.

What you or your doctor can do

- Gastroenterology/nutrition/feeding team evaluation to determine the risk of the rerouting of vomit into the trachea and then the lungs (aspiration), nutritional status, and presence of GERD
- Swallow study
- **Feeding therapy** to improve coordination and food-texture sensitivity
- For those with mild or moderate gastroesophageal reflux, anti-reflux medications should be considered. For more severe reflux, consult with a gastroenterology specialist
- For those with difficulty swallowing (dysphagia) or a risk of aspiration, placement of a gastric tube should be considered

*Many families have found that a **blended, whole-food diet therapy** may lessen vomiting episodes and supports growth.*

Teeth

Dental and palate abnormalities also occur in up to 50% of affected individuals. These range from overcrowding to irregular spacing due to missing permanent teeth (hypodontia). Large teeth and imperfect alignment also have been noted.

What you or your doctor can do

- Regular, age-appropriate dental exams
- Standard orthodontic treatments to manage crowding and alignment
- Regular eye exams

Eyes

Misalignment of eyes resulting in inward or outward positioning (strabismus) affects up to 50% of individuals with BRS and can be intermittent or persistent. **Upper eyelid drooping (ptosis) and visual difficulties** also may occur and require corrective lenses.

*Some individuals with BRS have **difficulty with temperature regulation or are insensitive to cold and heat**. Others have an extremely high tolerance for pain.*

Resources

The information in this guide is based on Balasubramanian M, Schirwani S. ASXL3-Related Disorder. 2020 Nov 5. In: Adam MP, Feldman J, Mirzaa GM, et al., eds. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024.

This resource was developed with financial support from the American Legion Child Welfare Foundation.

This information is not medical advice and should not replace a consultation with a physician.

Connect & learn more

Find caregiver information and resources on the ARRE Foundation website:
- Educational resources & tools
- Directory of doctors & therapists
- Caregiver support
- Resource Library

Education for healthcare professionals also is available.

Visit arrefoundation.org/resource-library

ARRE
ASXL RARE RESEARCH ENDOWMENT
arrefoundation.org/bainbridge-ropers

@arrefoundation @arrefoundation @arrefoundation