

# Information Sheet

## SETBP1 HAPLOINSUFFICIENCY DISORDER (SETBP1-HD)



Other Names: SETBP1 disorder | SETBP1-related disorder | SETBP1-related developmental delay | MRD29

### WHAT IS SETBP1-HD

SETBP1 haploinsufficiency disorder (SETBP1-HD) is an extremely rare genetic neurodevelopmental disorder in which there is a variation or a small loss (microdeletion) of genetic material within the *SETBP1* gene. The variation or microdeletion causes the body to produce insufficient levels of the SETBP1 protein. In most instances, variations in the *SETBP1* gene occur spontaneously and there is no family history of the disorder (de novo variations). The *SETBP1* gene is located on Chromosome 18 within the 12.3 region.

### FEATURES

- Mild-significant neurodevelopmental disorders in all affected individuals, which may include: global developmental delay including mild-severe speech delay (apraxia of speech\*, absence of speech), gross and fine motor delays, developmental coordination disorder (dyspraxia), mild-severe intellectual disability, hypotonia (low muscle tone)
- Behavioral issues may include: autism or autistic features, ADHD, aggression, seizures (less common)
- Other traits: sleep issues, vision issues, and gastro issues
- Some individuals may have mild facial differences including: long, pointed chin, mild ptosis (drooping of upper eyelid), thin upper lip

\*formally diagnosed by a specialist such as a speech and language pathologist

### DIAGNOSIS

A diagnosis of SETBP1-HD is based upon identification of characteristic symptoms, a detailed patient and family history, a thorough clinical evaluation and a variety of specialized tests, including genetic testing. Children with mild or moderate intellectual disability or developmental delay, and speech development problems, but no other anomalies may be suspect of having SETBP1-HD. In most instances, SETBP1-HD mutations are discovered through a genetic blood test called Whole Exome Sequencing (WES).

### THERAPIES

Following an initial diagnosis, a developmental assessment may be performed and appropriate occupational, physical, and speech therapies be instituted. Speech therapy is required and can include one-on-one sessions with a speech therapist, combined sessions where children learn language and social skills as a group, and the use of augmentative and alternative communication (AAC) devices. A full behavioral assessment may be necessary and can help to identify triggers of certain behaviors.

### TREATMENT

The treatment of SETBP1-HD is directed toward the specific symptoms that are apparent in each individual. Treatment may require the coordinated efforts of a team of specialists.

- Pediatricians
  - Geneticists
  - Physicians who specialize in diagnosing and treating disorders of the brain and central nervous system in children (pediatric neurologists) and in adults (neurologists)
  - Developmental pediatricians
  - Speech therapists
  - Physical therapists
  - Occupational therapists
  - Ophthalmologists
- . Other healthcare professionals may need to systematically and comprehensively plan treatment. Genetic counseling is of benefit for affected individuals and their families. Psychosocial support for the entire family is essential as well. Currently, there are no standardized treatment protocols or guidelines for affected individuals.

### Resources

Additional medical, scientific & registry information: [www.setbp1.org](http://www.setbp1.org).

Information about SETBP1 Society: [www.setbp1.org](http://www.setbp1.org)

Private Facebook group for families to connect - <https://facebook.com/groups/SETBP1>

SETBP1-HD Resource Guide: <https://www.setbp1.org/setbp1-hd-resource-pocket/>



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