Mission

SETBP1 Society's mission is to provide support to individuals with SETBP1 disorder and their families, to promote discussion and fund research, and to bring awareness and education to the public. SETBP1 Society is an international organization representing families throughout the world.

SETBP1 SOCIETY

WE ASK FOR YOUR SUPPORT

- Donate at www.setbp1.org
- Shop at Amazon Smile or igive.com to benefit SETBP1 Society

SETBP1 Society is a 501c3 non-profit organization. Donations are tax deductible and should be payable to SETBP1 Society.

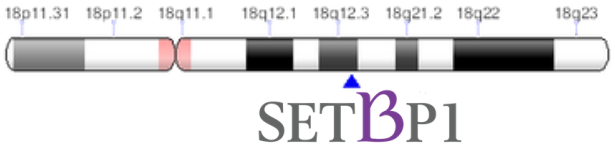
SETBP1 disorder is a newly discovered rare neurodevelopmental disorder that is caused by a loss-of-function mutation in the SETBP1 gene. It is characterized by absent speech/speech delays, intellectual disability, other developmental delays, adhd, autistic traits/autism, & mild dysmorphic features.

Co-existing diagnoses:

- apraxia
- dyspraxia
- autism
- sensory processing disorder

SETBP1 Gene Science

The SETBP1 gene, located at 18q12.3, provides instructions for making a protein called SET binding protein 1 (SETBP1), which is found in cells throughout the body. The SETBP1 protein is known to attach (bind) to another protein called SET. However, the function of the SETBP1 protein and the effect of its binding to the SET nuclear oncogene, which is involved in DNA replication, are unknown.



The Key Genotype-Phenotype Paper

Refining analyses of copy number variation identifies specific genes associated with developmental delay, Nature Genetics 2014

Want to Learn More?

Find out more information about SETBP1 at our website at www.setbp1.org/research

Newly Diagnosed?

You are not alone. You can connect with our community through our private SETBP1 Facebook group facebook.com/groups/setbp1. You can also make a significant impact by registering your child with the Simons VIP database where researchers collect data for their studies. Visit www.setbp1.org/registry

SETBP1 Society needs assistance with:

- Identifying researchers interested in studying the function of the SETBP1 gene and the associated SETBP1 protein
- Identifying researchers interested in creating disorder/disease system models to better understand SETBP1 disorder
- Spreading the word in the neurodevelopmental disorder community
- Finding disorder biomarkers
- Finding affected individuals

Connect with Us!

Join our private SETBP1 Community Group on Facebook. Be sure to "like' the SETBP1 Society Facebook page facebook.com/setbp1 and check it out for information on SETBP1 and important news and updates from the organization. Sign up for our newsletter!

"SETBP1 Society is committed to finding targeted treatments and therapies for individuals affected by SETBP1 disorder."

- Haley Oyler
President of SETBP1 Society

SETBP1 Society Partners

Boston Children's Hospital
Until every child is well

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SETBP1 Society
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