# SYNGAP1 gene

synaptic Ras GTPase activating protein 1

## Normal Function

The *SYNGAP1* gene provides instructions for making a protein, called SynGAP, that plays an important role in nerve cells in the brain. SynGAP is found at the junctions between nerve cells (synapses) where cell-to-cell communication takes place. Connected nerve cells act as the "wiring" in the circuitry of the brain. Synapses are able to change and adapt over time, rewiring brain circuits, which is critical for learning and memory. SynGAP helps regulate synapse adaptations and promotes proper brain wiring. The protein's function is particularly important during a critical period of early brain development that affects future cognitive ability.

## Health Conditions Related to Genetic Changes

SYNGAP1-related intellectual disability

At least 40 mutations in the *SYNGAP1* gene have been found to cause *SYNGAP1*-related intellectual disability. In addition to mild-to-moderate intellectual disability, this condition commonly features other neurological problems, including recurrent seizures ( epilepsy) and autism spectrum disorder, which affects communication and social interaction.

Gene mutations involved in *SYNGAP1*-related intellectual disability prevent the production of functional SynGAP protein from one copy of the gene, reducing the protein's activity in cells. Studies show that a reduction of SynGAP activity can have multiple effects in nerve cells, including pushing synapses to develop (mature) too early. The changes triggered by a reduction of SynGAP activity disrupt the synaptic adaptations in the brain that underlie learning and memory, leading to cognitive impairment and other neurological problems characteristic of *SYNGAP1*-related intellectual disability.

Autism spectrum disorder

At least five *SYNGAP1* gene mutations have been identified in people with autism spectrum disorder (ASD), a condition that appears early in childhood development, varies in severity, and is characterized by impaired social skills, communication problems, and repetitive behaviors. These mutations result in a SynGAP protein with

impaired function or prevent the production of the protein. Changes in synaptic adaptation in individuals with these mutations may underlie the behavioral abnormalities characteristic of ASD. It is not known why some people with *SYNGAP1* gene mutations develop ASD while others have the additional features of *SYNCAP1*-related intellectual disability (described above).

## Other Names for This Gene

- KIAA1938
- MRD5
- neuronal RasGAP
- Ras GTPase-activating protein SynGAP
- ras/Rap GTPase-activating protein SynGAP
- RASA5
- synaptic Ras GTPase activating protein 1 homolog
- synaptic Ras GTPase activating protein, 135kDa
- synaptic Ras GTPase-activating protein 1
- SYNGAP

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SYNGAP1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8831[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SYNGAP1%5BTIAB%5D%29+OR+%28synaptic+Ras+GTPase+activating+protein+1%5BTIAB%5D%29%29+OR+%28%28Ras+GTPase-activating+protein+SynGAP%5BTIAB%5D%29+OR+%28SYNGAP%5BTIAB%5D%29+OR+%28neuronal+RasGAP%5BTIAB%5D%29+OR+%28ras/Rap+GTPase-activating+protein+SynGAP%5BTIAB%5D%29+OR+%28synaptic+Ras+GTPase+activating+protein+1+homolog%5BTIAB%5D%29+OR+%28synaptic+Ras+GTPase+activating+protein,+135kDa%5BTIAB%5D%29+OR+%28synaptic+Ras+GTPase-activating+protein+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SYNAPTIC RAS-GTPase-ACTIVATING PROTEIN 1; SYNGAP1 (https://omim.org/entry/603384)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/8831)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SYNGAP1[gene])

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## Genomic Location

The *SYNGAP1* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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