

# SYNGAP1

## *SYNGAP1*

### Summary

- *SYNGAP1* (synaptic ras GTPase-activating protein 1) is a gene responsible for making proteins (called SYNGAP), which are essential for cognition and normal synapse function in the brain.
- Changes in the *SYNGAP1* gene can either be *de novo* (new in the person with epilepsy and not inherited from a parent) or be inherited from a parent.
- Epilepsy associated with the *SYNGAP1* gene usually begins during infancy and early childhood, and eyelid myoclonia with absence seizures, myoclonic-atonic seizures, myoclonic seizures and photosensitivity are common.
- Mild to severe intellectual disability, Autism spectrum disorder (ASD), low muscle tone, sleep, behaviour and eating problems are all associated with changes in the *SYNGAP1* gene.
- Diagnosis for *SYNGAP1* epilepsy will require genetic testing.

### Contacts and information

- Website: [www.syngapaaustralia.org/](http://www.syngapaaustralia.org/)
- Facebook: [www.facebook.com/Syngapresearch](https://www.facebook.com/Syngapresearch)
- Email: [syngapaaustralia@gmail.com](mailto:syngapaaustralia@gmail.com)