



Need to talk? Call us **0845 2412173** Monday to Friday  
9am to 5pm



Supporting those affected by  
Inherited Metabolic Disorders

# Adenylosuccinate Lyase Deficiency

Adenylosuccinate Lyase Deficiency is a rare metabolic disorder that causes neurological symptoms. It is caused by a defect in the ADSL gene which codes for an enzyme called Adenylosuccinate Lyase. This enzyme is needed to carry out two key processes to produce purine nucleotides which are the building blocks of DNA, RNA and energy sources within the cells. A deficiency of the enzyme means that both processes are affected, causing a build up of molecules which then go through a different chemical reaction to produce 2 unusual compounds never found in healthy individuals (SAICAR and S-Ado). These compounds are a characteristic feature of this disorder. It is believed that at this point the molecules become toxic and are the likely cause of the neurological manifestations in this disorder which predominantly include varying degrees of brain dysfunction causing developmental delay of mental abilities and movement, autism and seizures. There are three forms classified depending on the severity; the neonatal form (the most severe), type 1 (the severe form) and type 2 (the moderate or mild form). The disorder has been diagnosed in between 90 to 100 individuals worldwide.

## Synonyms

Alternative names for this condition are:

- Adenylosuccinase Deficiency
- ADSL Deficiency
- Succinylpurinemic Autism

Further information about this condition is available from Climb.



## Disclaimer

[Please read our disclaimer and information on data protection.](#)

Updated 05/05/17

Telephone: **0845 241 2173**  
Email: [contact@climb.org.uk](mailto:contact@climb.org.uk)

Freephone: **0800 652 3181**  
Website: [www.climb.org.uk](http://www.climb.org.uk)

Children Living with Inherited Metabolic Diseases is a charity registered in England and Wales (1089588) in Scotland (SC044634) and a Company Limited by Guarantee 4267454



ClimbHQ



ClimbHQ



TeamClimb