Climb™

Supporting those affected by Inherited Metabolic Disorders

# Arginine: Glycine Amidinotransferase Deficiency

Arginine:Glycine Amidinotransferase Deficiency is a very rare disorder that affects creatine metabolism. The body needs creatine to use and store energy properly. The arginine:glycine amidinotransferase enzyme is needed for the first step in the production of creatine from specific amino acids, which are the building blocks of protein. A deficiency of the enzyme results in the body not having enough creatine which affects the organs and tissues in the body, especially those which require high amounts of energy such as the brain. Arginine:Glycine Amidinotransferase Deficiency presents in infancy and is characterised by global developmental delay, autistic behaviour and learning disability. Muscle weakness, fatigue and seizures may also be present.

## Synonyms

Alternative names for this condition are:

- AGAT Deficiency
- Cerebral Creatine Deficiency Syndrome 3
- Creatine Deficiency Syndrome due to AGAT Deficiency
- GATM deficiency
- L-Arginine:Glycine Amidinotransferase Deficiency


Further information about this condition is available from Climb.

## Disclaimer

Please read our disclaimer and information on data protection.

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