



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



Supporting those affected by
Inherited Metabolic Disorders

AICAR Transformylase/IMP Cyclohydrolase Deficiency

AICAR Transformylase/IMP Cyclohydrolase Deficiency is caused by a defect in the ATIC gene and leads to severe neurological problems including learning difficulties, developmental delay and behavioural problems, epilepsy, and blindness that is apparent from birth. Individuals may have dysmorphic symptoms including dimples on the skin especially around the knees, elbows and shoulders.

Synonyms

Alternative names for this condition are:

- ATIC Deficiency
- AICA-Ribosuria Due to ATIC Deficiency
- AICA-Ribosiduria
- AICA-Ribosuria

Further information about this condition is available from Climb.

Disclaimer

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



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Telephone: **0845 241 2173**
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