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

Climb™

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

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