



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



Supporting those affected by
Inherited Metabolic Disorders

Glutamate Formiminotransferase Deficiency

Glutamate Formiminotransferase Deficiency is a rare inherited metabolic disorder that is caused by defects in the FTCD gene. This gene provides instructions for an enzyme known as formiminotransferase cyclodeaminase which plays a key role in the final breakdown of an amino acid called histidine which is a part of most proteins. The enzyme also helps in the production of the B-Vitamin folate which has many important function in the body.

There are two forms of Glutamate Formiminotransferase Deficiency, a mild and a severe form. Both of which are characterised by delays in reaching mental and physical developmental milestones. Approximately 20 cases have been reported. Approximately 75% of these have the mild form and approximately 25% have the severe form. All of those reported with the severe form have been of Japanese origin.

Synonyms

Alternative names for this condition are:

- Arakawa Syndrome 1
- FLIGLU-uria
- Formiminoglutamic Aciduria
- Formiminotransferase cyclodeaminase Deficiency
- Formiminotransferase Deficiency
- FTCD deficiency



Further information about this condition is available from Climb.

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