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

Climb™

Supporting those affected by Inherited Metabolic Disorders

Aspartylglucosaminuria

Aspartylglucosaminuria (AGU) is a rare disorder which is most prevalent in Finland. It is caused by a defect in the AGA gene which provides instructions for the enzyme aspartylglucosaminidase. This enzyme is active in lysosomes, which have a role akin to recycling centres within the body's cells. The enzyme is involved in breaking down certain proteins called glycoproteins. An enzyme deficiency causes glycoproteins to accumulate within the lysosomes, meaning the cell is unable to work properly, sometimes resulting in the cell being destroyed. This accumulation is most apparent in the brains nerve cells and causes many of the symptoms associated with the progressive decline in mental function which characterises this disorder. Osteoporosis, movement problems, seizures and characteristic facial features may also seen in patients with AGU.

Synonyms

Alternative names for this condition are:

- AGA deficiency
- AGU
- Aspartylglucosamidase Deficiency
- Aspartylglucosaminidase Deficiency
- Aspartylglycosaminuria
- Glycosylasparaginase deficiency

Further information about this condition is available from Climb.

Disclaimer

Updated 02/06/17

Telephone: **0845 241 2173** Freephone: **0800 652 3181**
Email: contact@climb.org.uk Website: www.climb.org.uk