

# Gaucher association - who we are



The Gauchers Association is the only registered UK charity providing support to individuals and families affected by Gaucher Disease, acting as the centre point for the Gaucher community working alongside medical professionals, scientists and the pharmaceutical industry to meet our aims;

- To support those individuals and their families affected by Gaucher disease through our patient and family advocacy service
- To provide information to patients and families regarding all aspects of Gaucher disease
- To advocate on behalf of patients and families to ensure access to appropriate treatment and specialised care through the Centres of Excellence in the UK
- To raise awareness and promote education in the medical profession of Gaucher disease to improve diagnosis and the impact of the condition on patients and families lives.

**Gauchers**  
ASSOCIATION



## How can you help?

**Membership** - Become a member of the Gauchers Association. Full application form available to download from our website.

**Organise** - a cake sale, a ball, a sports challenge, tea party.

**Get sponsored** - to run, cycle, swim, skydive, be waxed or shaved. See our website for events we have secured places for.

**Volunteer** - at one of our events, join our board of directors.

**Donate** - at [www.gaucher.org.uk](http://www.gaucher.org.uk) Why not set up a regular donation by setting up a Standing Order Shop - find us on [www.givingabit.com](http://www.givingabit.com)

**Find us** - [www.facebook.com/theGauchersAssociation](http://www.facebook.com/theGauchersAssociation)  
Whatever you want to do to help will be welcomed.  
Contact [ga@gaucher.org.uk](mailto:ga@gaucher.org.uk) or telephone 01453 549231 for a fundraising pack and advice on how to get started.

## Your help provides:

- Support those living with Gaucher disease to reach their full potential
- Visit a family either in their home, hospital and school to help people around them understand their needs promote scientific and medical research into Gaucher disease
- Our aim is to improve therapeutic approaches and to ensure all such recognises the centrality of the Gaucher patient
- Twice yearly newsletter to provide information on Gaucher disease and keep families and medical advisors in touch and up to date

## Have you heard of Gaucher Disease?

*You have now!*

Gaucher disease is so rare that we struggle to get the awareness and fundraising to help those affected.

Help us spread the word and raise awareness so more people can benefit from an early diagnosis and a better quality of life.

Gaucher disease is caused by a genetic mutation resulting in the deficiency, absence or incomplete functioning of an enzyme called glucocerebrosidase.

This leads to a build up of a fatty waste substance in cells, tissues and organs.

There are three types of Gaucher disease

**TYPE 3**  
5%

**TYPE 2** <1%

Affects the central nervous system;  
**typically fatal** during infancy

**TYPE 1** 94%

**Non-neuropathic** - only involves the internal organs and not the central nervous system