Gauchers ASSOCIATION

newsletter

February 2018

8th neuronopathic Gaucher disease meeting

Type 3 Young adults and siblings

Parents enjoying a game of Giant Monopoly

On 18th November 2017, 89 delegates attended the 8th neuronopathic Gaucher disease meeting at Centre Parcs, Woburn, Bedfordshire. We welcomed some overseas families from Denmark, Canada and USA.

Presentation topics included: symptoms & management of nGD, genetic modifiers, potential new therapies and several powerful and emotional personal stories.

The children all enjoyed a full packed activities programme; including bowling, cup cake decorating & habitat box building.

On Saturday evening families enjoyed a gala dinner with the opportunity to catch up with old friends and meet new.

On Sunday morning parents enjoyed a giant game of Monopoly as part of a team building exercise.

Quote from type 3 parent: '*Just like to say a BIG thank you to you and your team for organising a wonderful weekend. It was really nice to catch up with old friends and also meeting new parents. You all deserve a medal!!*'

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Scottish Medicines Agency (SMC) says 'YES' to paying for Cerdelga

The SMC announced that Cerdelga is recommended within its marketing authorisation for treating type 1 Gaucher disease, that is, for long-term treatment in adults who are cytochrome CYP2D6 poor, intermediate or extensive metabolisers. This recommendation means that the SMC must make Cerdelga available to patients in Scotland, where the doctor responsible for their care thinks Cerdelga is the right treatment.

Chief Executive, Tanya Collin-Histed said '*The Association is delighted with the outcome, which will give patients a choice of treatment in consultation with their treating doctor*'

Type 1 Gaucher patient, Anne says '*I was excited to learn the Scottish Medicines Consortium has agreed that Cerdelga can also be prescribed for patients in Scotland. Life will become so much easier for people who have been infusing for a number of years if they are able to move onto a new drug. They will be able to swap a drip stand for an easy to take pill*'.

Inside this issue:

SMC says 'YES' continued

The Association represented Gaucher patients at two SMC meetings in October and November where they presented evidence about the impact of living with Gaucher disease and the challenges that regular infusions of Enzyme Replacement Therapy have on some patients.

This decision comes seven months after Cerdelga was approved by NICE for patients living in England.

For type I Gaucher patients living in Wales they will be following the same decision as National Institute for Health and Care Excellence (NICE) who approved Cerdelga for patients living in England.

Helen Whitehead

Patient & Family Support Worker update

I have continued to be contacted since my last update by many people experiencing difficulties when moving onto the newer benefits such as PIP (Personal Independent Payment) and ESA (Employment Support Allowance). Both of these benefits require people to have a face-to-face assessment where you gain points for being able to do certain tasks and have to score a set number of points to be eligible.

If it comes back that you are not eligible all is not lost. The first step is to ask for a Mandatory Reconsideration; this is just asking them to relook at your case. For this you can provide extra evidence such as specialist letters, a diary and statements from carers etc. But it is very important to get this back to them in time within 1 calendar month. If this is still a no you can take this to appeal, again within 1 calendar month.

Statistics show that around 60% of appeals are successful (for PIP and ESA). Also it is better to attend the hearing in person, rather than opt to have it done on paper. You can take someone with you into the appeal, it could be someone who knows you well or it can be a representative, for example me from the Gauchers Association or someone from Citizens Advice Bureau.

How can I help?

I can support you through any stage of the process, from completing the initial form, through the mandatory reconsideration process and appeal.

It can be helpful to be able to meet face-to-face to go over all the details and make sure that you have the evidence is required and get you fully prepared. If this affects you please get in touch.

If you have any unmet support needs, I work Monday to Thursday. To get in touch you can call me at the Gauchers Association on 01453 549231 or call or text me on my mobile on 07795 192311, email me helen@gaucher.org.uk or find me on Facebook by searching for Helen Whitehead and the Gaucher logo. If you would rather meet in person we can arrange this too either at your home or a coffee shop.

Susan Lewis memorial Award

The Association has supported Dr Gali Maor and Dr Or Cabasso from Israel to participate in the European Study Group on Lysosomal Diseases (ESGLD) during September in Lyon, France and Dr Debora Rapaport to participate in the annual American Society of Cell Biology (ASCB) meeting in Philadelphia, USA in December

Top; Susan Lewis
Botton; Gali Maor, Or

Dr Gali Maor & Dr Or Cabasso write -

The meeting focused on different lysosomal diseases such as Batten disease, Mucopolysaccharidoses types 2 and 3A, Fabry disease and Neiman Pick type C. A vast number of the presentations concentrated on therapeutic modalities such as gene therapy and chaperones.

Concerning Gaucher disease, a talk and two posters were presented; the talk was by Gali Maor, from the group of Professor Mia Horowitz. She used a *Drosophila melanogaster* as a model system to study the contribution of mutant glucocerebrosidase (GCase) to the development of Parkinson disease (PD). She showed data indicating that the presence of mutant GCase stabilizes α-synuclein, causes its accumulation and aggregation.

A poster presented by Or Cabasso from the group of Mia Horowitz used *Drosophila melanogaster* as an animal model to study both neuronopathic and non-neuronopathic Gaucher disease. The flies presented substrate accumulation, inflammation and neuroinflammation, activation of the Unfolded Protein Response (UPR) and premature death, recapitulating the human disease. In a poster presented by Beccari (Paciotti S, Eusebi P, Dardis, A, Zampieri S, Chiasserini D, Tasegian A, Bembi B, Ceccarini MR, Calabresi P, PArentti L, Beccari T, CSF Lysosomal Enzyme Activity and GBA1 Genotyping in Parkinson's Disease) the authors found decreased GCase activity in the cerebral spinal fluid (CSF) not only in GD carriers who developed PD but also in the CSF of PD patients, independent of the presence of a *GBA1* mutation. They also showed that GCase and Cathepsin D (another lysosomal enzyme) activity are lower in more advanced stages, suggesting their possible role as PD prognostic markers.'

Dr Debora Rapaport writes -

The meeting took place at the Philadelphia Convention Centre, Philadelphia, USA, between 2-6 December, 2017.

It included plenary sessions, mini symposia and workshops, in which up to date technologies and scientific achievements were presented. These included very advanced imaging methods (Cryo-electron microscopy, cryo-EM, EM tomography, super resolution confocal microscopy) to follow and study the dynamics of different interactions between intracellular organelles, for example; interactions between the plasma membrane and the endoplasmic reticulum (ER) or between mitochondria and ER membrane.

The CRISPR-Cas9 based editing was discussed in a very informative workshop by world leaders in the field. The CRISPR-Cas9 is a new technology that allows editing of genomes, such that mutations can be introduced or mutant genomes can, hopefully, be corrected. The major problem with the latter is editing of "OFF target" sequences, which cause, beside correction of a mutation, creation of undesired mutations.

This problem is under investigation and will have to be solved before this technique can be applied to genome correction, as a new tool for Gene Therapy.

James Cox

Go with Gaucher

'Go with Gaucher – taking forward the next generation' is a European Gaucher Alliance (EGA) project. The EGA organised the event to bring young people with Gaucher disease from all over the world together to exchange information and ideas and also identifying future leaders for the Gaucher community. Type I Gaucher patient James Cox attended this event and writes—

Well firstly, I had a fantastic time. It was really good to be able to get to know other people who suffer with type 1 Gaucher disease and be able to share our stories with one another. It was reassuring to know other people get the same problems I do and it gave me a perspective. This being that my condition could be a lot worse, I was very lucky in comparison to others.

I learnt so much from them and the guest speakers about my condition. The event prepared me for when I informed my current employer about my illness, as I was able to practice during a workshop. Everyone who attended the symposium were a joy to be around and I couldn't have asked to spend these few days with anybody more amazing. I hope to attend more of these meetings in the future.

I would like to thank the Association, Chris and the European Gaucher Alliance (EGA) for giving me the opportunity to attend in Frankfurt.

Rare Disease Day 28th February 2018

28th February 2018 marks the eleventh international Rare Disease Day coordinated by EURORDIS.

On and around this day hundreds of patient organisations from countries and regions all over the world will hold awareness-raising activities. The 2018 theme is a continuation on Research from 2017.

There are lots of ways to get involved; to find out more visit www.rarediseaseday.org

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