The Gaucher Disease Experience:

An Insight from Gaucher Patients aged 45 and over in the UK

Gauchers ASSOCIATION

June 2019

ACKNOWLEDGEMENTS

The research that informs this report was funded by grants from Sanofi Genzyme and Takeda (formally Shire).

The report is based on questionnaires that were sent, via the specialist centres, to patients with Gaucher disease aged 45 and over, plus additional information that was received from patients and medical professionals at the specialist centres.

A huge thank you to everyone who contributed to this piece of research and shared their experiences to enable us to shape the work of the Gauchers Association

The Gauchers Association

The Gauchers Association, established in 1991, 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 and working alongside medical professionals, scientists and the pharmaceutical industry to meet our aims:

1. To support those individuals and their families affected by Gaucher disease through our patient and family advocacy service;
2. To provide information to patients and families regarding all aspects of Gaucher disease;
3. 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;
4. To promote research into the causes, effects and treatments for the three types of Gaucher disease; and
5. To raise awareness and promote education in the medical profession of Gaucher disease to improve diagnosis and the impact of the condition on patient and families lives.

In addition to providing support from the Gauchers Association office, we publish information leaflets and monthly news bulletins. The Newsletter is circulated to families, friends, donors and professionals worldwide.

CONTENTS

ABOUT GAUCHER DISEASE

Gaucher disease is a rare inherited metabolic disorder, where people lack the enzyme Glucocerebrosidase, making them unable to break down certain fatty substances in their cells. These cells are called Gaucher cells and can accumulate in the liver, spleen, bone marrow and sometimes the nervous system.

There are 3 types of Gaucher disease. Type 1 Gaucher disease, the most common, can result in orthodpaedic problems, such as acute bone pain and bone necrosis, with a possible need for joint replacements and a deterioration in mobility. Other effects include low platelet counts, resulting in fatigue and an increased likelihood of bleeding and bruising. Type 1 Gaucher disease affects approximately 1 in 100,000 births worldwide. It is more prevelant in the Askenazi Jewish population at 1 in 850 births (Ganz. et al. 2017). There are just over 300 known patients in the United Kingdom (taken from the Gauchers Association statistics received from the specialist centres in 2018).

In the less common forms of the disease, Types 2 and 3, the central nervous system is also affected. Type 2 is seen in infants, where the aggressive progression of the condition usually results in death by the age of 2 years. Patients with Type 3 disease may experience similar symptoms to those with Type 1, but with additional neurological symptoms including an eye movement disorder, learning difficulties, seizures, balance problems and difficulties with executive functioning and decision making (e.g. impulsivity).

Effective treatment has been available for patients with Type 1 since the early 1990s. The first treatment was an intravenous enzyme replacement therapy (ERT) to replace the missing enzyme. In the UK there is also oral substrate reduction therapy (SRT) available. Treatment is beneficial for the visercal symptoms in Type 3 Gaucher disease, but at present there is no effective treatment for the neuorogical aspects of this form of the disease.

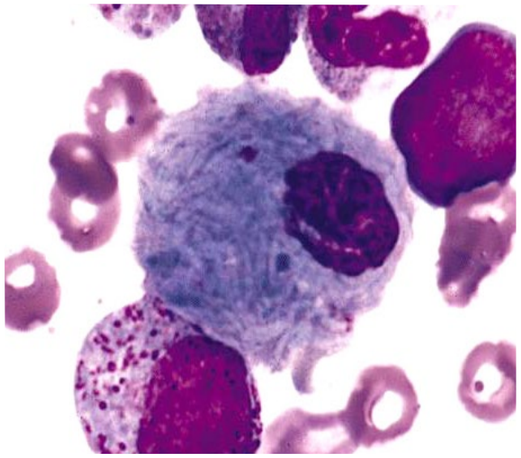
There is currently no effective treatment available for children with Type 2 Gaucher disease apart from palliative care.

Treatment in England is funded by the Department of Health through NHS England and delivered nationally through one of 8 Specialist Centres of Excellence (5 adult, 3 paediatric) that are under 'Highly Specialised Services'. In Wales, funding for treatment is provided via a patients' local health board. Adult and paediatric patients in Wales are treated either in Cardiff or one of the Centres of Excellence in England.

GAUCHER DISEASE & GETTING OLDER

A previous project was carried out in 2012/2013 investigating the non-medical needs of patients and families affected by Gaucher disease, with a focus on newly diagnosed patients and those affected by Neuronopathic (Types 2 and 3) Gaucher disease. From this research, the Gauchers Association's Patient and Family Support Service was developed. The Gauchers Association have a Patient and Family Support Worker who currently works 18.5 hours a week supporting Gaucher patients and families with unmet non-medical needs.

Feedback through the Association membership survey and Patient and Family Support work over the years has indicated that there may be different and changing support needs for the older Gaucher community.



Gaucher cell

There is very little published on the long-term outcomes for Gaucher patients on ERT (Ganz et al. 2017). In 1991 the first UK Gaucher patient recived ERT; for many patients they were diagnosed several years before treatment became available. Prior to ERT the only treatment for Gaucher disease was supportive; splenectomies (removal of the spleen) were common, now known to have a negative impact of bone problems and an increase of Gaucher cells in the liver (van Dussen et al. 2014).

This project is designed to determine what these needs are and how to shape the Patient and Family Support Service moving forwards.

ABOUT THE RESEARCH

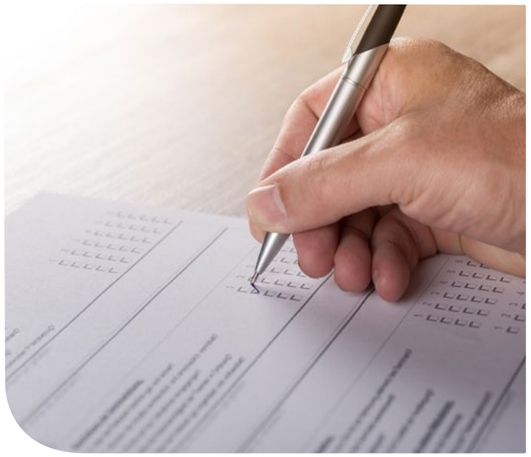
Over the winter of 2018/2019, the Gauchers Association carried out a survey of patients with Gaucher disease aged 45 plus. Why 45 plus? The Association have been aware of the changing needs of the Gaucher population aged 45 onwards and wanted to explore this area further. The aim of the survey was to find out the experience and possible unmet needs of people aged 45 and over living with Gaucher disease in the UK.

The results from this survey will help the Gauchers Association shape their Patient and Family Support Service going forward and feed into possible future research projects.

The questionnaire covered multiple aspects of Gaucher disease that have been highlighted by members of the Gauchers Association in previous feedback. It was sent out, via post, to the 6 UK adult specialist centres (Addenbrooke's Hospital, Queen Elizabeth Hospital, The Royal Free Hospital, Salford Royal Hospital, The National Hospital and University Hospital of Wales) and liaison with the Patient Advocacy Worker in Northern Ireland.

We were delighted to receive 77 returns. 3 had to be discounted as they were not completed. Of the 74 valid responses 96% were from Type 1 patients. The average age being 61, with an age range of 45 to 85. 85% were living in England, 5% in Scotland, 7% in Wales and 3% in Northern Ireland. Thus, all parts of the UK were represented, and this broadly represents the UK population (ONS 2017), suggesting our findings are a good representation for the UK.

There is a huge variation of age when people are diagnosed with the mean age being 28 years old but ranging from 2 to 62 years. The returns were 47% male and 53% female.



SUMMARY OF FINDINGS

The majority of respondents are on Enzyme Replacement Therapy **(69%)** and **27%** are now on oral therapy (SRT)

Concerns were raised regarding traveling to specialist appointments in the future

Only a small number of respondents had used the Patient and Family Support Service

There is a great variation in specific health issues alongside Gaucher disease

18% have poor pain control

43% have fatigue – a huge unmet medical need

There are a significant proportion of respondents who are unable to work due to their health.

The average age for retirement is less than state pension age

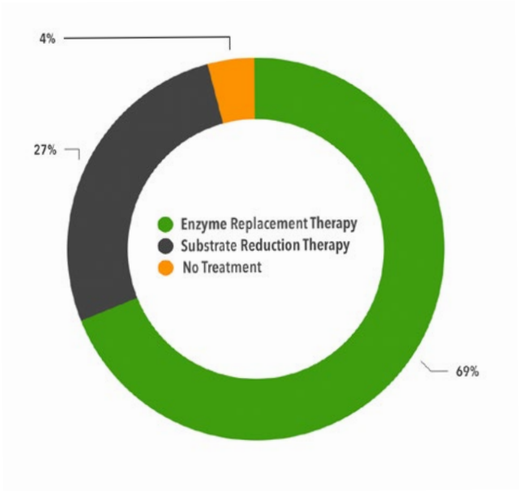
There are still a number of people on historic benefits and will, at some point, have to move onto the newer benefits

41% of respondents affected by Gaucher disease would like a meeting / social event to meet others

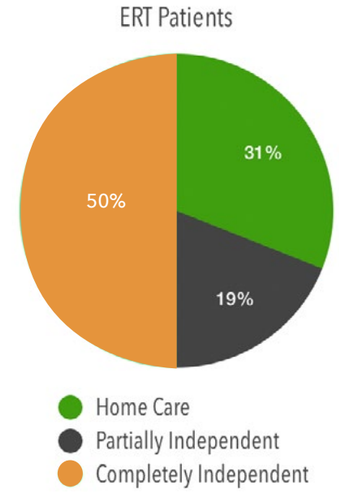
TREATMENT FOR GAUCHER DISEASE

Gaucher patients on ERT receive their infusions at home, traditionally delivered by a homecare nurse. There is a new drive within the NHS to enourage more patients to become more independent with homecare. There is the option for patients to learn to do their infusions themselves, a carer to learn or for them to do part of the treatment, such as removing the canula or getting the treatment ready. There are multiple reasons for this and advantages for the patient – saving the NHS money, increasing the capacity of the home care services so that other treatments can be rolled out and increased independence for the patient so they can do their infusions at a time and place that suits them.

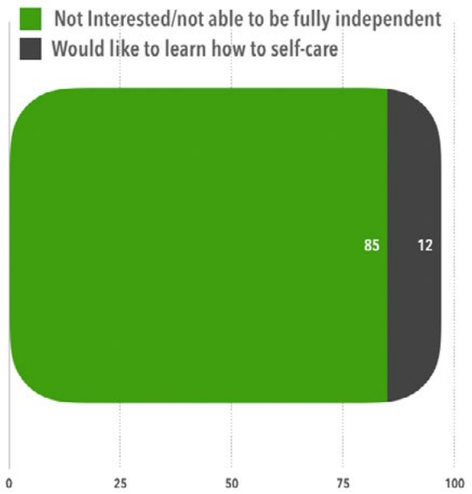
The questionnaire results showed that 69% of respondents are on ERT, 27% on oral tablet SRT and 4% are untreated.



Of those on ERT, 31% rely entirely on homecare for their infusion, 19% are semi- independent (e.g. they make up the infusion and the nurse cannulates) and 50% are completely independent and do the entire infusion themselves or their partner / carer does it.



Of those who aren't fully independent 85% are not interested/unable to be fully independent and 12% are interested in learning to self-infuse.



RECOMMENDATIONS

Many of the 45 plus Gaucher patients are already independent or semi-independent (69%) showing that this is something the Gaucher community have already embraced. For those that have not reached any form of independence with treatment, there are only a small number who may be interested in learning to self-infuse.

The Gauchers Association should:

- **Continue to encourage people to be as independent as they are able but recognise that there will be those, that for a variety of reasons are unable to be more independent**
- **Recognise that needs might change over time and members should feel supported in making those changes becoming both more and less independent**
- **Continue to work with the specialist nurses and home care providers**
- **Continue to share personal stories of people's experiences with their treatment**
- **Ensure the PFSS (Patient and Family Support Service) can support members with any treatment challenges and advocate for them as required.**

SPECIALIST CENTRE APPOINTMENTS

There are 5 adult UK specialist centres and 1 in Wales, meaning that patients often have a long journey to visit their specialist. The average (mean) journey was found to be 3 hrs and 33 minutes (each way). The length of journey ranged from just 20 minutes to 10 hours.

31% were seen once a year and 62% twice a year. This is a huge financial burden for patients. The NHS does have a Healthcare Travel Cost Scheme but you have to meet the strict criteria of being on certain benefits or qualify for the NHS low income scheme (see helpful links at end of document for more information)

Age UK have produced a report 'Painful Journeys' (2017) that highlights many of the challenges faced by older people getting to their hospital appointments. The key issues being, long and uncomfortable journeys on public transport, difficulties with hospital-

provided patient transport and the financial cost to older people and their families.

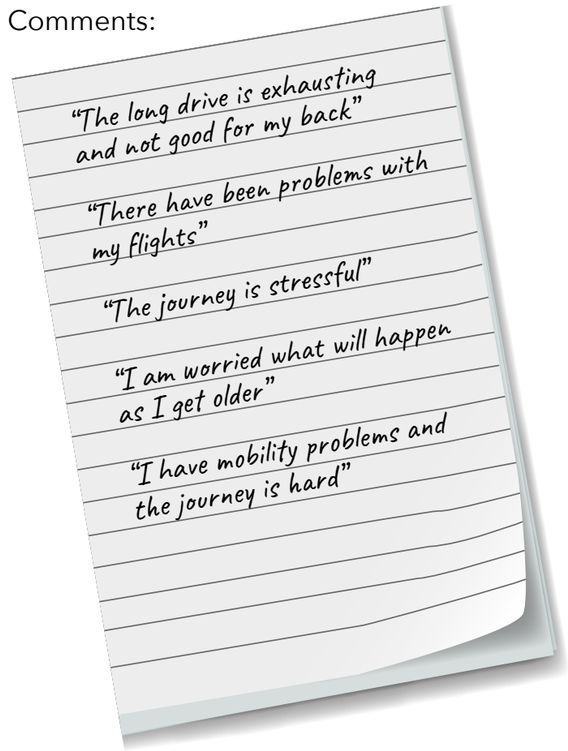
Over recent years there has been a tightening of the eligibility for hospital-provided patient transport, but a patient may be eligible if they have a condition or disability that makes it difficult or impossible to use public transport. The person needs to discuss options directly with their treating centre. Hospital transport can also mean long waits for being collected and for the return journey home.

36% think that they might have a problem getting to appointments in the future. Currently 70% attend their appointments by car, 39% use public transport, 5% come by plane and 1% use hospital transport.

Some specialist centres are aiming to reduce the number of face-to-face visits and have a telephone consultation for alternate appointments.

There is other help available with transport, such as a blue badge, bus passes and railcards. The blue badge scheme is run locally by each council and eligibility varies. Older Person's Bus Passes can be applied for from female state pension age in England and 60 in Wales, Scotland and Northern Ireland. For those younger, there is also a disabled person's bus pass, if certain criteria are met, also managed by local councils. Both disabled person's railcard and senior railcard are managed by National Rail and if you buy a railcard and meet the criteria you get a reduction on your rail fare. For more information on all these schemes please see useful links at the end of this document.

Comments:



RECOMMENDATIONS

The Gauchers Association should:

- **Support members to look at alternative forms of transport as required**
- **Liaise with specialist centres on criteria for hospital transport**
- **Assist people to see if they are entitled to help with transport costs due to qualifying benefits or low income**
- **Support members to access information on blue badge, bus passes and railcards and advise on how to complete forms and appeal decisions if necessary**
- **Encourage telephone consultations where possible**
- **Raise awareness on this issue at NHS meetings in partnership with the LSD Collaborative**

HEALTH & WELLBEING

There are known long-term complications and associated conditions of Type 1 Gaucher disease, including splenectomy, bone complications, pulmonary hypertension, Parkinson's and malignancies (van Dussen et al. 2014). Ayis et al. (2003) highlight that psychological, social and environmental factors are associated with long-term illnesses. People with long-term conditions are high users of health services, accounting for 55% of all GP appointments and 68% of all hospital appointments (George, Martin 2016).

People with long-term health conditions frequently experience mental health problems such as depression or anxiety, having a detrimental effect on the prognosis of their long-term condition and their quality of life (Naylor et al. 2012).

The reported average number of visits to the GP was 4.74 per year ranging from a minimum of 0 to maximum of 30. The average number of consultations for the UK population is 5.5 per year (Hippislex-Cox, Vinogradova, 2019). The average number of hospital appointment, that is not Gaucher specialist is 2.76 per year with the range of 0-25 appointments. This shows huge variation, showing that the range of health issues is diverse.

On specific health issues, 41% have had a splenectomy, 9% heart disease, 12% cancer, 1% multiple myeloma, 32% arthritis, 32% bone fractures, 4% Parkinson's, 43% fatigue. Of those that had splenectomies 37% had previously had bone fractures and 47% arthritis, whereas for those not having a splenectomy this was 27% and 25% respectively. This is just a crude measure but backs up the research that those that have had a splenectomy tend to have more severe bone disease (van Dussen et al. 2014).

Parkinson's disease is a neurological condition with symptoms of slow movement, stiffness and problems with walking. It has been shown that the affected gene in Gaucher disease (GBA) is linked with a higher risk of Parkinson's disease. This small sample shows an incidence of around 1 adult in 25 whereas in the UK it is around 1 in 350 of the general population. Please see links at the end of the reports for more information on Parkinson's disease and the research into this link.

In this questionnaire,

15% *report that their mental health is poor or very poor*

32% *report that their sleep is poor or very poor*

18% *report that pain is poor or very poor*

39% *report that their energy levels are poor or very poor*

24% *report that exercise level is poor or very poor*

RECOMMENDATIONS

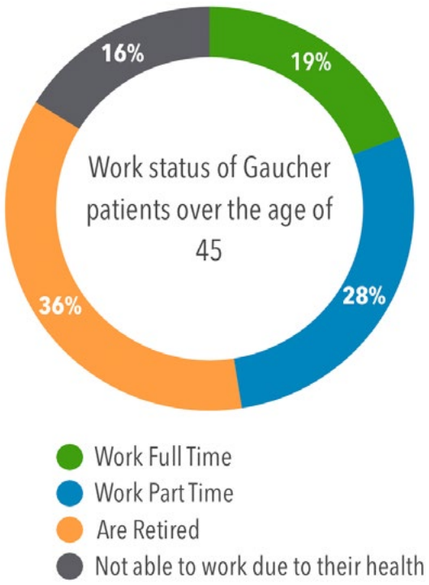
The Gauchers Association should:

- **Encourage research into continuing unmet medical needs especially fatigue, sleep, and pain**
- **Encourage research into other co-morbidities e.g. Parkinson's, malignancies**
- **Link with other patient support groups e.g. Parkinson's UK and direct people to get more specific, specialist information and support**
- **Work alongside specialist team to support patients with mental health challenges.**

EMPLOYMENT & RETIREMENT

The state pension age in the UK will be 66 for men and women by October 2020 and there are further plans to increase it to 67 by 2028. The average age from this survey was 54.6 with a range of 45-70 years. People with Gaucher disease that have already retired, on average, have retired early (54.6). Research has shown that retirement is linked with a reduction in mental and physical fatigue and depressive symptoms in people with chronic disease (Westerlund et al. 2010), therefore having a positive health effect.

It is also worth noting that 28% of this group worked part time and the reason for this should be investigated to further understand the reasons; contributing factors could be health, finances and caring responsibilities.



It is known that there are significant barriers to employment to people with long-term conditions. This leads to lost earnings, lower career prospects, early exit from employment or a prolonged absence from work (Bajorek et al. 2016). Symptoms can be fluctuating, as with Gaucher disease, therefore making it difficult to manage in the workplace. 16% of respondants in this survey were not able to work due to their health.

RECOMMENDATIONS

The Gauchers Association should:

- **Through the PFSS continue to support members with work related issues**
- **Signpost people to other organisations who can provide financial advice e.g. Age UK**
- **Encourage research into Gaucher disease and employment**
- **PFSS to support members to claim for the correct benefits e.g. pension credit, Council Tax reduction**

FINANCE & BENEFITS

Over the last few years the benefits system in the UK has changed significantly. The main disability benefit is Personal Independent Payment (PIP), a point-based system that is based on what you can and can't do rather than what condition a person has. PIP has replaced Disability Living Allowance (DLA) and they are very different.

6 respondants said they are still on DLA – 2 are over 65 and will therefore likely remain on DLA. But 4 are under 65 so will, at some point, be invited to apply for PIP.

PIP is a very different benefit and many Gaucher patients have had challenges when switching over, but the majority have eventually managed to receive what they are entitled to. Some people were previously eligible for DLA but not for PIP and others vice versa.

The Gauchers Association PFSS has to date supported 4 members in court to appeal benefit decisions – all of which have been successful. 19% of respondents are currently claiming PIP. Attendance Allowance is the disability benefit for those over 65 but does not have a mobility element, currently just 4% claim this.

For further information on benefits please see 'Useful Links' at the end of this document.

RECOMMENDATIONS

The Gauchers Association should:

- **Maintain up-to-date information of current disability benefits**
- **Publish in its news bulletins benefits updates and what support is available**
- **The initial application form is vital to success as is the quality of evidence sent with the application. PFSS to support members with this process or signpost to local services**
- **PFSS to liaise with specialist teams to get the best supporting evidence for benefit applications**
- **PFSS to act as representative in court to advocate on the member's behalf as required**

SOCIAL & EMOTIONAL SUPPORT

7% of respondants say their current housing doesn't meet their current needs. Follow-up work needs to be done to understand why their housing doesn't meet their needs and whether they could apply for assessment of needs and adaptations through their local council to access a disability facilities grant. For more information on disability facilities grant please see useful links section at the end of the document.

74% of respondents receive no care and 26% receive care from another person roughly in line with the number of people claiming disability benefits. Therefore, indicating that most people receiving care are claiming the correct benefits.

67% are rarely or never lonely.

33% are always, often or sometimes lonely.

The last meeting the Gauchers Association arranged for all Gaucher patients including Type 1 patients was during the Cerezyme shortage in 2010, and was held at the Royal Free Hospital in London. There were only 20 patients who attended of which 15 were Type 1 patients.

The responses show that 31% would like a patient meeting and 30% would like a social meeting. An overall percentage of 41% would like a meeting.

23% would be interested in an online support group. 38% were not interested in any form of meeting, online or in person. By plotting on a map all those interested in meeting there is a wide geographical spread, with nobody north of Yorkshire. This might be that previous meetings for those in the far north of the UK have always been too far south and therefore by offering a meeting in the North East of England or in Scotland we might attract people.

Comments on patient meeting:

"Depends how far and where the venue is"

"Would need to be London area only"

"Would rather forget about GD"

"It would be good to share experiences, support other Gaucher patients, hear public speaking on issue and get involved in fundraising"

"My disease doesn't define me therefore not interested in meeting others with Gaucher disease"

"I like to distance myself from ill health. I find I am happier when I forget about GD and cancer. Meeting up is not for me."

I have difficulty concentrating and I get confused in group discussions, due to epilepsy, making me panic and stress. I have difficulty interacting with strangers due to my poor cognitive responses to new information

I have attended in the past. It would have been useful with the new tablet. I would like to share views and hear from others

The distance from conferences and meetings is always a problem for me. I am either on crutches or in wheelchair and public transport is inaccessible. Unable to walk steps. Would consider a couple of hours from home.

I would be interested to meet up informally with other local patients, but I found it too far to make visits to the south of England for meetings as I used to attend Gauchers Association meetings years ago, although I enjoyed meeting, such lovely courageous people.

Depends on travelling. Unable to attend if need to travel long distance. Don't drive and depend on others.

I work full time and spare time is limited

RECOMMENDATIONS

The Gauchers Association should:

- **Research holding several separate meetings across the UK, potentially Bristol, Manchester and London, plus a possible North-East meeting or Scottish meeting.**
- **It would still be relevant to include Cerdelga in a programme as members that are already on the tablet can feedback their experience and those that are considering it can talk it through with others.**
- **At least one of the board of trustees should be at every meeting to be able to talk about the work of the Association and to gain feedback from members**
- **Continue to support patients who would like to meet others in their more local area, if that is possible**
- **Continue to liaise with the All-Ireland Advocacy worker to monitor whether a patient meeting would be viable in Ireland / Northern Ireland**

COMMUNICATIONS & MEMBERSHIP

The Gauchers Association have developed their communciation methods over the last decade or so by increasing their email communciations with their members. Whilst the Gauchers Association should continue to encourage their members to receive electronic communications, they must also ensure that their information is accessible to all. There continues to be a number of members without access to email and rely on being kept up to date through hard copy postal communications

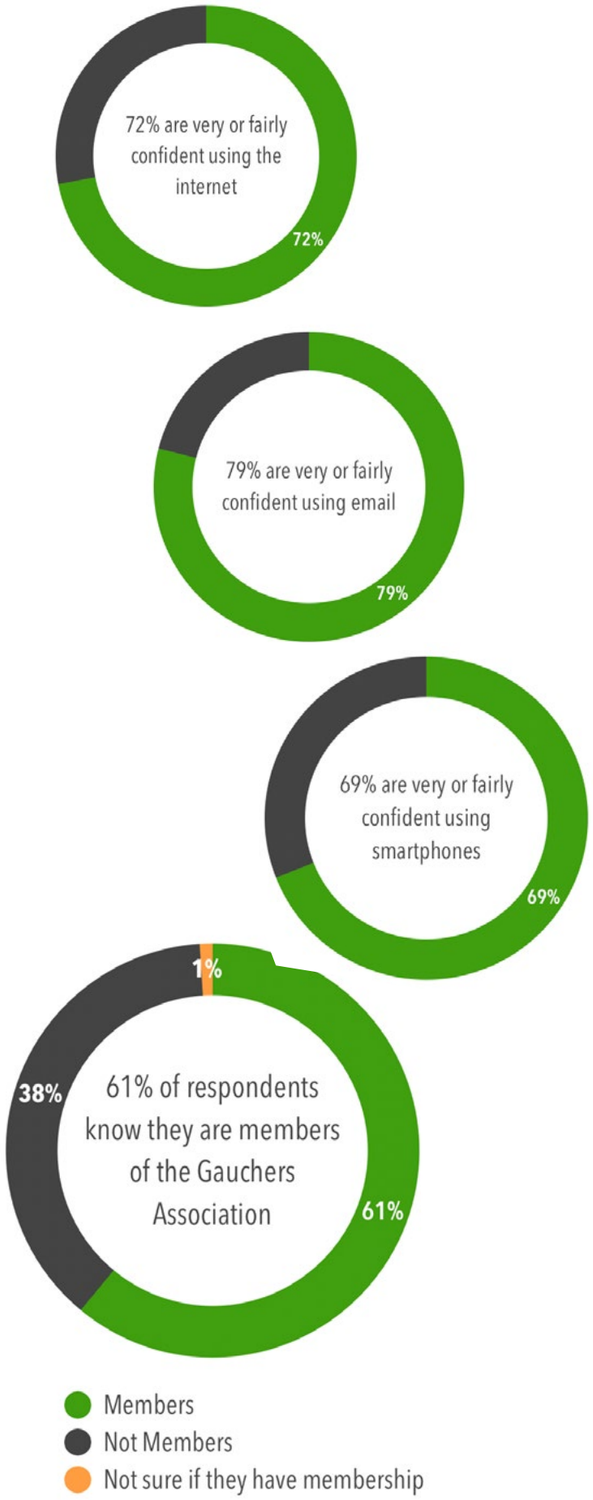
For using the internet, 72% are very / fairly confident. Using email 79% are very / fairly confident and using smartphones, 69% are very / fairly confident. This shows that the Association must continue to provide communications in various forms.

61% say that they are members, 38% say that they are not members, 1% are not sure.

Feedback to the questionnaire shows that work needs to be done around the understanding of membership and this could potentially increase the membership of the Association.

72% receive the newsletter, 14% fundraise, 5% have used PFSS, 14% have had no interaction with Gauchers Association. This shows a discrepancy as you would only automatically get a newsletter if you were a member.

This suggests that a number of people do not realise they are a member.



RECOMMENDATIONS

The Gauchers Association should:

- **Continue to provide its resources in both printed and online forms**
- **Frequently check with its members on how they wish to receive information**
- **Promote the benefits of being a member**
- **Encourage more people affected by Gaucher disease to become members of the Gauchers Association**
- **Continue to promote its Patient and Family Support Service resource**

Wendy

A lot of my memory dates are a bit vague as most of my life I have had several battles with doctors not really understanding all of the unusual things that were happening with my body.

I remember as a young girl I wanted to do so well at athletics, but something in my body was stopping me from reaching the goals I was setting for myself and always felt upset that I could not in my head reach my full potential. I won many challenges but not as fast as I thought I should achieve. I was so frustrated that I could not make my body do more.

I had pains in my shin bones constantly. These were put down as growing pains. If I had a tooth extracted, I always ended up in A & E as it would not stop bleeding or clot. Whenever I got an illness it was always very bad. I nearly died of measles. I picked up infections very easily. My appetite was virtually non-existent

When I reached 13 and started my periods it was horrendous. I bled like a tap for 3 weeks and only had 1 week free. Every month was the same which left me feeling pretty low. Doctors did not want to discuss this.

In 1972 I had my son. Then in 1973 I supposedly contracted infective hepatitis which they said I contracted at the hospital I worked. This actually was Gaucher disease presenting as hepatitis. I was still not diagnosed. 1975 I had my second son. When I was in early stages of pregnancy

with him, I contracted meningitis. The doctors tried to persuade me to terminate my pregnancy due to what problems my son might incur. True to form I refused, and I delivered a perfect son without any of the disabilities that they warned me of.

In 1981 I had a second miscarriage and was shunned at the hospital I was sent to and told to go back home and rest. On reaching my home I passed out and hit my head. Doctor was called who then sent me back to hospital with a stern letter saying they had to admit me. I had several blood transfusions and they could not stop the bleeding so had to perform an emergency hysterectomy. I was only in my early 20s so it was a huge shock.

In 1983 I had my first mini stroke which paralysed me all down my left side. Whilst having an MRI scan they found I had actually had 2 as they found 2 parts of my brain were damaged. I was admitted to Queens Square in London for tests. It was here, after they carried out bone marrow tests, they found out I had a genetic condition called Gaucher disease which was incurable. At least now I knew why I felt so much pain in my bones. Sometimes they got so hot I thought I would self-combust. The pains I had moved around my skeleton sometimes arms, legs, neck but always there.

To try to build my strength up after the strokes I took up running and used a static bike and did exercises every day. Then I started to get horrific pains in my hip. I went to the doctor who was convinced I was making up my condition. He gave me anti-inflammatory drugs and pain relief. I had to put up with this pain for around 2 years, it got so bad I took an overdose as I needed to sleep as I was exhausted looking after 2 young sons.

I ended up saying to the doctor if you X-ray my hip and find nothing wrong, I will never come back again. He arranged this and to his astonishment it showed that I had lost my complete femoral head. Then when they researched my condition they said there was nothing they could do as surgery was not an option.

As I worked full time, I was put on morphine for pain relief which was awful. Most of the time I just felt very sick and the pain was still there. Given two walking sticks so I could get to and from work and a TENs unit (electrodes that you stick on your back) to help with pain management.

A visit to another doctor and a referral to another hospital meant my hip was replaced.

In 1990 I had my spleen removed as it had swelled to 11.4 lbs and my gall bladder was removed. This was due to the condition I had. I was told I had 2 years to live.

To me this was not acceptable so I researched everything I could on my condition and found out that in America they had patients with my condition taking enzymes by infusion. I went to see my consultant and discussed this with him, and he said it would be very unlikely as they came from humans and I could contract HIV or CJD (human equivalent of Mad Cows Disease). I was so incensed I contacted 2 other health authorities who both refused to treat me. I ended up lobbying parliament who authorised my treatment. I had my hip revised and due to the surgeon not understanding my condition and not giving me antibiotics or a clotting agent before surgery he had huge problems. On discharge I was given a box of pills and told just to take them at regular times. What they failed to tell me was when you are prescribed anti-inflammatory drugs you must drink plenty of water. I did not know this, and I ended up in hospital on kidney dialysis and nearly lost both of my kidneys.

I infused myself for 22 years then the veins got temperamental, so I had nurses who visited my home and assisted. I am now monitored by the Royal Free who are a centre of excellence for lysosomal storage disorders. They understand all of the problems I have and feel like family. I can call anytime and there is always someone who I can talk to.

I have been on a very long and sometimes very painful journey but the Royal Free has been my saviour.

Judith

I was born in Penzance, Cornwall in 1955. My general health up to the age of six seemed normal, although I always had a round protruding stomach but with slim arms and legs. From time to time, I did have pains in my legs, but no-one thought anything of it. At age six I fell and broke my elbow, but this seemed to heal normally.

At around age seven or eight, the pains in my legs started to get much worse. My GP at the time attributed this to 'growing pains'. By this time, I was also beginning to have nose bleeds, and began to bruise easily and tire quickly. My GP referred me to our local Paediatrician, who arranged for various tests to be carried out, with Leukaemia being mentioned as a possibility.

One of the most frightening things for me at the time, was the consultants and doctors when examining but not talking to me. They would then want to talk to my mother after I had been told to wait in the waiting room. My active mind was very frightened. When my mother came out of the room, I would always ask what the consultant or doctor had said, and the reply was always 'nothing for you to worry about'. As you can imagine, I was petrified during these appointments.

On my X-rays, the radiologist noticed some unusual marks and thought it might be a rare disease – Gaucher Disease. As a result of the initial tests, I was referred to Great Ormond Street Hospital in 1964 at age 9 for an 'outpatient' appointment. My parents took me to the hospital, and I was admitted for

a period of three weeks. Further tests were conducted and Type 1 Gaucher Disease was eventually confirmed.

One of the most frightening tests I had was a bone marrow sample. This was taken on the general ward with a local anaesthetic, where I was alone without my parents. In those days patient visiting was very restricted. During the procedure, two nurses fainted.

As the size of my spleen had enlarged to the size of a rugby ball rather than a tennis ball, I underwent a splenectomy. This operation has left me with a scar from the middle of my belly to my side. I was very ill at this time and lost a lot of blood during the procedure which required several blood transfusions. I remember the blood was in glass bottles, and the tubing was reusable.

In those days there was no parent accommodation, and no financial assistance for families with one child as there would be now. As my parents could not afford to stay in a hotel, they stayed with my cousin in Surrey. After two weeks, my father returned to Penzance to go back to work. Living in a tied house on a farm, no work meant no house. My mother stayed in London and visited me when allowed and we eventually made the very long trip, for a very sick young girl, back to Penzance.

Eventually I was able to return to school, but the pains in my legs got much worse. I would have spells of agonising pain that required hospitalisation in Truro, our local orthopaedic hospital. The pain was so severe that no pain killer I was given gave me pain relief. I could not bear any weight on my legs or even the weight of a sheet on me. I now realise these pains were bone crises.

The medics looking after me were really at a loss to know how to help me. Gaucher Disease being rare, none of them had experienced this before, there was no internet to research and only limited information in medical research books. The advice given was to take pain relief and bed rest until I was able to walk again. Sometimes they would put my leg in a St Johns Splint to immobilise it.

At age eleven, and still having very bad pain, my Orthopaedic Surgeon referred me to Mount Vernon Hospital in Northwood, Middlesex. My mother and I again travelled by train as we had no car at that time. My father was unable to come with us as they could not afford three train fares.

She again stayed in Surrey with my cousin and caught the bus to see me a few times a week, because of the visiting restrictions. As you can imagine, for a young girl brought up on a farm from Cornwall, it was very frightening. I remember those times at Great Ormond Street and Mt Vernon Hospitals, alone and only wanting my Mummy and Daddy to be with me. Even now, fifty years later, these memories can easily bring me to tears.

The decision was made at Mt Vernon Hospital to give me a series of Radiotherapy treatments in a local Hospital in Redruth. This was to help with my pain relief, and then for me to walk with crutches. I would wear a none weight-bearing metal leg calliper on my left leg, and on my right foot, I had a built-up shoe on a metal frame. This was a hideous contraption for any child. As I lived in a house, and before the days of stair lifts, my father would carry me up and down the stairs every day. I also missed a year of schooling and the local education authority arranged a tutor to visit me for an hour a week to give me schoolwork to do.

After around nine or twelve months, I eventually stopped wearing the calliper, got stronger and was able to return to Secondary School in the second year. Miraculously, I managed to stay in the 'A' stream until I left school at age 17. I still had occasional bone crises and my leg joints would swell, which eventually led to me being hospitalised again for a few weeks.

I started work at age 17 as an accounts clerk and at 19 had met my future husband at work. I was still having regular sick leave due to my ongoing orthopaedic problems in my legs and back. Brian and I married in 1981 and we bought our first home together and were both working hard and were very happy, but my health problems were continuing.

On Friday 8th November 1985 and aged 30, we moved home. When the house we were moving from had been emptied, and Brian was moving everything into our new home, I made the fatal mistake of staying behind to wash the kitchen floor. My next mistake was stepping back onto the floor before it had fully dried. Unfortunately, for the first and only time, I did the splits. I had broken my left femur.

I was taken by ambulance to the casualty department at Truro. I was told you'll be with us for Christmas. It transpired to be five months. Initially I had an operation that night and my leg was put in traction. I was bed ridden for virtually the remaining time in hospital. By the end of January 1986, there was still no bone growth to knit the break. In desperation, and after consultation amongst their colleagues, the surgeons decided to operate again and insert a full-length rod into the femur to aid recovery. To say this was painful in the next few weeks would be an understatement. Again, I was very ill after surgery and needed transfusions.

Eventually, I recovered sufficiently to be able to get out of bed again, but it was decided not to risk another break at this stage, and I was again measured for an iron calliper. Once these were made, I was desperate to get out of bed. Nurses and Doctors were happy to see me standing as they had only seen me lying down. I initially started to walk on a frame, and then progressed to crutches. As I had previous experience of using crutches, I was quite adept. On 20th April 1986, I was finally discharged for Easter and able to walk into my new home for the first time.

I found it very hard to get my mobility back, and I was unable to bend my left knee due to the prolonged period of traction. Because my femur is so fragile, no attempt was made to manipulate and bend my knee under anaesthetic, as the surgeons were concerned the femur would break again. The initial plan was to leave the rod inside my femur to help protect it. However, in 1991 an infection set in, and the rod had to be removed and the infection cleared. I have had many operations for infection in subsequent years and I now have an open wound which I dress daily. Because of this infection, I am now no longer able to have hip and knee replacements, as the body would reject them. As a consequence, my mobility remains very poor. I walk using crutches for short distances, but resort to a wheelchair for longer distances.

In late 1992, I was fortunate to hear of the work of Professor Timothy M Cox, at Addenbrooke's Hospital in Cambridge. I obtained a referral and first saw him in January 1993. My husband and I went with great hope and the blessings of my GP and Rheumatologist and with the caution of my Orthopaedic Surgeon. I took with me my notes and X-Rays.

From the first meeting with this wonderful man, I really felt this was the first person I had met in 37 years who actually knew about Gaucher Disease and understood the associated problems. He had great empathy and was able to draw out feelings that I had bottled up for years. As part of his initial examination he confirmed that I had an enlarged liver, something that I was unaware of, but gave us both hope for the future. He explained that he believed he could help me and would apply for funding from my Area Health Authority for the necessary treatment.

We returned to Cambridge again six weeks later when my Local Health Authority had agreed to fund the treatment. I was admitted for a few days for scans and other tests, and my first two infusions of Ceradase. On a snowy February day, we made the long trek back to Penzance with the rest of the trial medication and delivered them safely to our local hospital. I then began regular infusions three times a week. My local hospital has been wonderful to me over the years and I still go there for my infusions now. Unfortunately, because of the number of infusions over the years, I have had problems with my veins. I am currently receiving ERT infusions fortnightly. We also travel to Addenbrooke's Hospital for the usual check-ups and scans usually every six months.

There are a number of people and organisations I would like to thank including the Gauchers Association of whom I have been a member since 1992. This was when the late and lovely Susan Lewis was such a great help to me. I would also like to thank all the NHS staff who have attended to me over the years including GP's, Surgeons and Medics, and in particular The Gaucher team at Addenbrooks, staff at West Cornwall Hospital in Penzance, the Truro City Hospital and then later the Royal Cornwall Hospital in Truro, Mount Vernon Hospital and finally Great Ormond Street Hospital. Collectively, these are the people and institutions who have managed to get me to now.

On a more personal note, I would like to thank my father and my late mother who had enormous worries to deal with and most of all my husband Brian who I will never be able to thank enough for the love and dedication he has shown me.

The cloud on the horizon is Parkinson's Disease. My mother suffered from this disease as did my father's brother and two sisters. With Parkinson's Disease on both sides of the family, and with its strong links with Gaucher Disease, this remains a worry for me. I have taken part in every research project I am asked to, and urge others to do likewise, as this is the only way cures can be found.

Newly diagnosed Type 1 Gaucher patients are now able to receive treatment relatively quickly. This is as a result of the outstanding research work carried out in the past, and of course those that volunteer to help in research programmes. Soon, I am hopeful that every Gaucher Patient can just take a tablet for their disease.

The purpose of this article is not to ask for sympathy, but to highlight the work that is being done to find a cure for Gaucher Disease. I hope that no-one has to endure the problems and also disabilities that I, and many other sufferers like me, have had to experience.

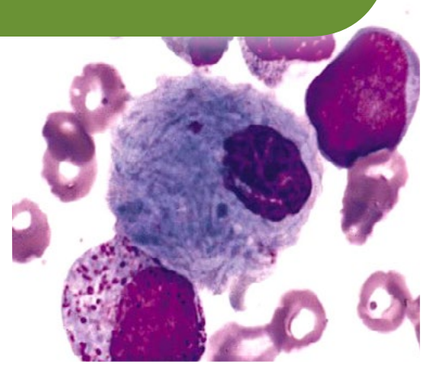
CONCLUSION

Gaucher disease is a complex condition and each person is different.

Those aged 45 and older may have been diagosed long before treatment became available, this therefore influences long term needs, such as bone health, as damage would have been sustained already due to lack of treatment or if a splenectomy was carried out.

Throughout this report there are recommendations and areas for additional work highlighted to meet the needs of patients in the UK with Gaucher disease with the priority areas being:

Encourage members to be as independent with treatment as they are able but recognise that some may be unable to be more independent and that needs change over time.

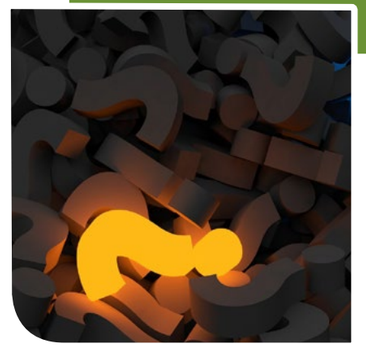


Support members to access appropriate transport, reduced fare travel schemes, to be able to attend specialist clinic appointments

Plan meetings for Gaucher patients in different geographical locations

Encourage research into continuing unmet medical needs, especially fatigue, sleep and pain

Ensure up-to-date information and support in accessing appropriate benefits



PARKINSON'S UK

More links should be made with other patient support groups e.g. Parkinson's UK

Ensure all information is available to members in accessible formats

USEFUL LINKS

If you need any of the following information in printed form please contact the Gauchers Association on 01453 549231.

Help with transport:

Healthcare travel cost scheme https://www.gaucher.org.uk/admin/laravel-filemanager/images/shares/files/Health%20costs%202018.pdf)

The Blue Badge Scheme https://www.gov.uk/blue-badge-scheme-information-council

Disabled person's bus pass https://www.gov.uk/apply-for-disabled-bus-pass

Older person's bus pass https://www.gov.uk/apply-for-elderly-person-bus-pass

Disabled person's railcard https://www.disabledpersons-railcard.co.uk/

Senior railcard https://www.senior-railcard.co.uk/

Parkinson's Disease

https://rapsodistudy.com/en/gaucher-gba-parkinsons

https://www.parkinsons.org.uk/information-and-support/types-parkinsonism

Multiple Myeloma

https://www.myeloma.org.uk/

Benefits

The Gauchers Association Website has information on different benefits

https://www.gaucher.org.uk/patient_family_support_worker

Housing

https://www.gov.uk/disabled-facilities-grants

NOTES

REFERENCES

Age UK (2017), Painful Journey – why getting to hospital appointments is a major issue for older people. Available from https://www.ageuk.org.uk/globalassets/age-uk/documents/reports-and-publications/reports-and-briefings/active-communities/rb_dec17_painful_journeys_indepth_report.pdf *Accessed 17/05/2019*

Ayis et al. (2003) Long-standing and limiting long-standing illness in older people: associations with chronic diseases, psychosocial and environmental factors. Age and Aging. 32 (3) Available from https://pdfs.semanticscholar.org/6bf4/9fc29ec33b83c787bb60e26f94b2211a15c4.pdf *Accessed 17/05/2019*

George, Martin (2016) BMA Briefing Paper – living with long term conditions. Available from

Board of Deputies for British Jews, https://www.bod.org.uk/jewish-facts-info/jews-in-numbers/ (Accessed 10/05/2019) https://www.bma.org.uk/-/media/files/pdfs/collective%20voice/policy%20research/public%20and%20population%20health/living-with-long-term-conditions.pdf
Accessed on 17/05/2019

Ganz et al. (2017) A new framework for evaluating the health impacts of treatment for Gaucher disease type 1. Orphanet Journal of Rare Diseases. [online] 12 (38) Available from: https://ojrd.biomedcentral.com/articles/10.1186/s13023-017-0592-6 *Accessed 28/11/2018*

Hippislex-Cox, Vinogradova (2019) Trends in Consultation Rates in General Practice 1995-2008. NHS. Available from: https://files.digital.nhs.uk/publicationimport/pub01xxx/pub01077/tren-cons-rate-gene-prac-95-09-95-08-rep.pdf *Accessed 16/05/2019*

Naylor et al. (2012) Long-term conditions and mental health – The cost of co-morbidities. The Kings Fund- Centre for Mental Health. Available from https://www.kingsfund.org.uk/sites/default/files/field/field_publication_file/long-term-conditions-mental-health-cost-comorbidities-naylor-feb12.pdf *Accessed on 17/05/2019*

ONS (2017) Population estimates for the UK, England and Wales, Scotland and Northern Ireland: mid-2017. [online] table 1, Available from: https://www.ons.gov.uk/peoplepopulationandcommunity/populationandmigration/populationestimates/bulletins/annualmidyearpopulationestimates/mid2017 *Accessed 09/05/2019*

Van Dussen et al. (2014) Modelling Gaucher disease progression: long-term enzyme replacement therapy reduces the incidence of splenectomy and bone complications. Orphanet Journal of Rare Diseases. [online] 9 (112) Available from: https://ojrd.biomedcentral.com/articles/10.1186/s13023-014-0112-x *Accessed 28/11/2018*

Westerland et al. (2010) Effect of retirement on major chronic conditions and fatigue: French GAZEL occupational cohort Study. BMJ.[online] Available from: https://www.bmj.com/content/341/bmj.c6149 *Accessed 06/06/2019*

SANOFI GENZYME

Takeda

Gauchers ASSOCIATION