

LSD Collaborative Community Survey

2020 Report on responses from the Pompe community

Allan Muir, 12 February 2021, Updated 24 March 2021

Introduction

In the Summer of 2020, the UK LDS Collaborative distributed a survey to individuals and families who live with any one of the Lysosomal Storage Disorders (LSDs). The Survey is intended to be undertaken every one or two years, the last one was in 2017, and gathers information about how people experience their care and treatment by the NHS Highly Specialised Services for LSDs throughout the UK.

For the 2020 survey we received an incredible 112 Pompe responses which provides a huge expanse of data to help the NHS and patient support groups to understand how well things are going, and what might be improved.

Just as we were planning the Survey launch, we suffered from the first wave of COVID-19 epidemic and all the attendant measures put in place to protect the health of the population and the NHS. The Survey was therefore amended to include a number of questions about the impact of the epidemic, responses to which are reflected within this report.

A tremendous amount of high-quality data was collected from the survey, and this report will summarise the data collected from the Pompe community only; data that amounted to 141 pages, so please excuse this summary for being a mere 15 pages!

Response and demographics of the Pompe respondents

A total of 112 people reported that they are either an individual, parent or carer living with Pompe disease, their distribution across the UK and Ireland were:

Region	Responses	Devolved Nations	Responses
North East	1	Scotland	4
North West	10	Wales	8
Yorkshire and the Humber	4	Northern Ireland	1
East of England	1		
East Midlands	9	Republic of Ireland	1
West Midlands	17		
London	11		
South East	32		
South West	13		

Respondents were primarily individuals (88), with parents (20) completing the form on behalf of the child and, others were completed by partners (4) and carers (2).

Access to Therapy

Of those responding, 106 (93%) had access to ERT and 3 reported that they received ERT with a chaperone under a clinical trial. One person received ERT through a company access programme.

One person had chosen not to receive therapy, and one person did not meet the clinical criteria required.

Most people (89) receive their ERT infusions at home, but a number were not; largely because 17 of them were part of a clinical trial.

ANSWER CHOICES	RESPONSES	
At a National Specialist Centre	12.96%	14
At School/University	1.85%	2
At a local hospital/health centre	7.41%	8
At work	1.85%	2
At Home	82.41%	89

Of those participating in a clinical trial, 13 were receiving ERT, and 4 reported that they are receiving ERT with a chaperone.

Patient Registries

Registries included those operated by companies (Sanofi Genzyme) and the patient-reported registry run by the International Pompe Association and the Erasmus Medical Centre in Rotterdam.

34 people surveyed are in the Sanofi Pompe registry and 29 complete the IPA/Erasmus survey. Another 19 people were not sure if they were included.

Patient Advocacy Groups

Of those reporting, 83 said that they interact with Patient advocacy groups, and 27 did not feel the need to engage regularly with any of the available support groups.

All areas of support were valued, with the order of priority by those responding was:

1. Information provision
2. Telephone or online support
3. Support or Advocacy team
4. Support during the COVID-19 crisis
5. Practical support
6. Fundraising support
7. Nursing support
8. Events and conferences
9. Emotional / mental health support

NHS Specialist Centres

A small number of people reported that they do not attend one of the recognised specialist centres, giving several reasons, including the need to attend a clinical trial at the Centre for Life, Newcastle. The majority, however, attend the centres listed below

Paediatric Specialist Centre	Number
BIRMINGHAM - Birmingham Children's Hospital	4
LONDON - Great Ormond Street Hospital for Children	9
MANCHESTER - Royal Manchester Children's Hospital	3
Adult Specialist Centre	Number
BIRMINGHAM - University Hospitals Birmingham	17
CAMBRIDGE - Addenbrooke's, Cambridge University Hospitals	8
CARDIFF - University Hospital of Wales, Cardiff, Wales	9
GLASGOW - The Royal Hospital for Children (previously Yorkhill)	1
LONDON - The National Hospital - UCLH	24
LONDON - Royal Free Hampstead	11
MANCHESTER - Salford Royal	13

Not everyone found the location of their specialist centre to be convenient, 28% reported that it was not, and many respondents commented on the stress, difficulties and cost of distance, mobility, public transport, parking, and having to rely on other family members.

Communication

Communicating with the specialist team between appointments did not seem to pose any difficulties; only 6 people reported that they never do so.

Common forms of communication were by phone or email to the nurse or consultant, and some noted the recent use of video calls during the Covid-19 pandemic.

Patient Experience at the Specialist Centre

Satisfaction with the specialist centres seemed to be very high, except for the parking arrangements:

“Buildings and location are inappropriate for disabled people to visit. Disabled people need to be able to park without being run over and have all the things they are required to do in one location which is flat and accessible.”

A number of other comments about the experience at centres, the survey itself and clinical trials were reported, all of which we will try to address in our future meetings with the medical teams.

	Strongly Disagree	Disagree	N/A	Agree	Strongly Agree
It is easy to park at the Specialist Centre	27	27	15	23	6
The Specialist Centre is clean	2	0	5	41	49
The facilities (restaurants, cafes, shops etc.) at the Specialist Centre are good	1	7	16	45	29
Staff are friendly and helpful	1	1	2	25	66
Staff competent and knowledgeable about the condition	1	0	3	26	66
Can access all of the specialties and services needed in one place	2	7	9	39	40
Good relationship with the Specialist Centre healthcare team	1	4	3	29	57
Treated with dignity and respect	1	2	2	26	64
Encouraged to ask questions and give feedback	1	3	5	30	56

Overall, most people were satisfied or very satisfied with their specialist centre:

Overall Experience	
Not at all satisfied	1%
Somewhat satisfied	6%
Satisfied	21%
Very satisfied	71%

There were areas where people reported that they would like more support from their teams, notable examples are:

- Mental health
- Dietary advice
- Physiotherapy
- Respiratory support

When asked to suggest improvements to the service, in addition to the additional support, a number of examples were offered:

- More time between tests
- Improve toilet facilities
- Appointments with different specialties on same day
- Improved parking facilities

Clinical Specialties

Most people have experience of a wide range of specialists:

Clinical Specialties	Respondents
Speech and Language Therapy (SALT)	28
Occupational Therapy (OT)	36
Physiotherapy	78
Dietitian or Nutritionist	44
Neurologist	29
Psychologist	20

Specialties mentioned in addition to those surveyed include:

- Cardiology
- Respiratory (Long-term ventilation, ENT)
- Metabolic
- Neuromuscular physiotherapist

Homecare

It was interesting to note that 35% of respondents do not use Homecare services for ERT infusions. Of those who do use Homecare services, the distribution across homecare companies was reported as:

Company	Respondents
Lloyds Pharmacy Clinical Homecare	59%
Healthcare at Home	27%
Pharmaxo	3%
HealthNet Homecare	0.00%

Of those using Homecare services, 42% used the provider's telephone ordering service. In general people found the ordering staff to be helpful. Most were satisfied with the service although the level of satisfaction was quite variable. There were a small number of complaints about deliveries and scheduling.

Satisfaction with the Homecare delivery staff were also generally high, with deliveries being punctual, accurate and complete. But there were a number that did not meet the highest standards expected. Complaints were noted about:

- Missing items
- Time and date
- Scheduling
- Wrong medication
- Sharps collection

Homecare Nursing Support

Most of those receiving homecare rely on the homecare nursing support service (84%), although only 56% are fully dependant on the nurse for drug preparation and cannulation. 16% of respondents are fully independent, so need no support, and 29% are semi-dependant, and so do not require the nurse to stay throughout the infusion.

Of those independent respondents, 90% reported that it was their own decision, only one person did not undergo training and felt unsupported throughout the process of becoming independent. Reasons for becoming independent from the nursing service varied:

- To reduce home visits from nurses during the pandemic.
- Offered more flexibility and self-sufficiency.
- To better care for my son.
- Convenience and flexibility.
- Inconvenience of travel into London for every infusion.
- Freedom to choose treatment days and times.
- Flexibility to infuse over a weekend to avoid disruption of school schedule.
- Disagreement with the homecare nurse.

There were many reasons stopping people from choosing to become more independent from the homecare nursing support:

What stops you becoming independent?	Percentage affected
Worried about self-cannulation	44%
Nurses often struggle to cannulate immediately	31%
Not enough dexterity / strength	23%
Not confident about reconstituting the drug	21%
Needle Phobia	19%
Previous bad experience	13%
Enjoy the company of the nurse	6%
Therapy administered at school	4%

Other reasons given separately include:

- Nurse must stay to cannulate and do observations
- Blood phobia, would feel nervous in case something went wrong during the infusion.
- As a parent with other children, I have many other commitments.
- Cannot cannulate
- Requires the nurse to unpack the vials only so that if damaged they will be replaced. If I damage them, they may not be replaced.
- Other children, so it's not practical. Once had a mild reaction to ERT.
- Patient is not well enough, and I could not do it for him.
- I am a carer for a friend and for my brother who has Pompe
- Daughter is very tiny, so I could not cannulate.
- Physically unable.
- Find the idea very stressful. Nurses train for years for good reason! We value them!
- Child is severely disabled we have enough to cope with and do on a daily basis.
- Problems cannulating without tissing

Of those who do not have a level of independence, six said that they would be willing to be trained to prepare the drug and self-cannulate, and four said that they would learn to self-cannulate if the drug was delivered in a reconstituted form.

Homecare experiences

People seemed to be very happy with the nursing services provided, finding their nurses to be polite, friendly and helpful. A small number would have liked their nurse to be more knowledgeable and work more flexibly to meet their needs. Only 4% were dissatisfied with the service they received, and most others were very satisfied.

Complaints to the homecare companies were mostly about the home delivery service, but a number were also regarding the telephone ordering system (8) and the nursing service (7). Only four complaints were not handled satisfactorily.

Other comments about complaints to the companies included:

- I would like to directly arrange infusion dates and times with the infusion nurse rather than going through a third party.
- The scheduling department is hard to work with. I would like the nurses to handle their own scheduling.
- For several months, the provider has sent a different nurse and continuity has been broken with disastrous results for the patient. It has meant extra visits from another nurse and more visits. A regular nurse becomes familiar with the patient and leads to more successful infusions.
- Complaints about delivery, nurses and running a refrigerator in a crowded room.
- Too many different nurses - unfamiliar with my tricky veins

Coronavirus Experiences

Thankfully only three people reported that there had been cases of Covid-19 in their family, although another nine did not have a confirmed diagnosis. Together they experienced the recognised symptoms:

Fever (temperature higher than it is usually)	80%
Unusual Fatigue	70%
Headache	50%
New, continuous cough	40%
Loss of smell or taste	30%
Unusual Breathing difficulties	30%
Painful sore throat	20%

How the pandemic affected Pompe individuals and families

Like the general population, the pandemic has affected people in different ways, the list below highlights the particular impact on social isolation and mental wellbeing:

Effect of Covid-19 pandemic	RESPONSES
Social Isolation (not seeing family or friends)	65
Practical issues, i.e. access to food supplies	48
Mental and emotional issues	42

Reduced access to treatment and therapy	33
Reduced level of clinical care	20
Financial hardship	19
Employment - furloughed	16
School / Higher education paused	16
Reduced access to hospice services	11
Reduced level of personal care	8
Reduced access to respite services	6
Reduced access to social care	6
Employment - Made redundant	1

Other individuals reported a mixed impact if the first wave:

- It was not safe for me to go to work.
- I didn't feel too affected by the virus as staying indoors was not a problem for any of us.
- It is easier as I have family living with me to look after me
- Childcare issues because they had to stay with us at all times
- It was a nice holiday in the main. Had plenty to do at home and am used to being self- sufficient.
- Managing shielding combined with working from home and also doing home schooling is a challenge.
- Closure of all other sports and social facilities, restrictions on movements
- I was missed off the vulnerable list for five weeks and had no access to usual food deliveries, so struggled to get food.
- Prescription items missing, so went without meds for a few weeks.

Information about shielding

It seems that people were informed of their shielding status in a number of different ways, but most received a letter from the central NHS and their specialist centre:

How were you informed about shielding?	RESPONSES
Letter from NHS England	80
Letter from Specialist Centre	62
Letter from GP	34
Telephone call	27
Text message	23
Not at all	2

Most felt that the message was clear, helpful, timely and appropriate, although a significant number (22%) were confused by some of the messaging. Many comments were received suggesting how the process could be improved:

- More information regarding how it affects certain conditions
- Guidance should have been given in the start of the pandemic.

- There were several letters not just one. The first was clear but subsequent ones were more confusing
- Filter those for whom shielding was right for
- Priority deliveries was confusing and some advice if followed would have totally isolated me from my family that I live with.
- I didn't need 6 letters as I watch the news and can think for myself.
- Letter from local hospital was too blunt and upsetting.
- It would have felt better having someone to speak to about it and explain it to me,
- Stopped NHS texts because there were so many and so long as to be unusable.
- They could have texted or emailed immediately
- Too many letters, too many texts, too much information.
- I received more than one letter and they contradicted each other
- I was told that if my son got sick, he would not be given a ventilator. I thought that was an extremely inappropriate thing to say.
- I have had quite a lot of letters including the first one twice telling me to stay in for 3 months in March and the end of April
- Postal letter would've been better
- Target at-risk groups at risk as identified by specialists rather than broad brush by GPs
- Improve access to food deliveries.
- The government should have reacted quicker, should have enforced lockdown, be more open about underlying conditions related to Covid deaths

After taking advice, most respondents followed the advice given (95%), but a few felt that they could not:

- checked with consultant: shielding wasn't necessary, but still mostly followed guidance
- I need someone to help me move, so it seemed stupid to isolate and stay in my room when I can't be independent.
- I did what i felt was right for me personally. I made my own mind up and made my own decisions.
- I don't consider that I need to shield.

There were people who did not receive direct advice, of those most chose to shield because of the vulnerability they felt.

Once the shielding advice was changed, about 50% of respondents started to venture outside again, but during the first wave of the crisis, it is clear that access to many facilities ceased:

Which services could you access during the crisis?	RESPONSES
Groceries	65%
Prescribed treatment or therapy	54%
Medical supplies	44%
Regular medical care and appointments	43%

Urgent medical care	24%
Work	15%
Treatment received as part of a Clinical Trial	12%
Psychological / mental health support	11%
Personal care / support at home (external provider)	10%
Medical tests or scans	9%
Speech and Language Therapy (SALT)	7%
Physiotherapy	6%
Monitoring visit as part of a Clinical Trial	4%
Social care	1%
Respite care	1%
School / Higher Education	1%
Hospice care	0%

Routine medical appointments suffered with 7% choosing not to attend, 24% were cancelled by the healthcare provider, and the remainder (44%) were changed to either a video or telephone call. A small number of people were not happy with their video/telephone appointments, but the majority seemed to be reasonably satisfied with them as alternatives. Indeed, many people suggested that they would like to continue with these remote appointments, although the benefits of face-to-face appointments were highlighted.

Treatment during the Pandemic (first wave)

OF those whose treatment was suspended during the pandemic, 3% were not shielding but chose to take a treatment holiday. 34% said that the treatment centre suspended treatment and 15% said that they could not attend a treatment centre while they were shielding.

Below is a selection of comments about the impact of treatment disruption:

• WHAT IMPACT DID THE TREATMENT DISRUPTION HAVE?

- Went without for 12 weeks as was not safe to have the nurse come into my house.
- I missed 2 before hospital arranged home visits
- I decided to carry on with treatment throughout. I was contacted by the centre who informed me that it was my choice. I asked that only one nurse attend; I did not want a training nurse attend.
- After three months I felt I was suffering due to lack of infusions, so they were reinstated.
- After missing some treatments, I asked for them to be reinstated as I was physically deteriorating
- Weaker and more pain for a few weeks then restarted treatment
- No apparent affect has been noticed to date. However, the treatment has been resumed by the patient during the middle of July.
- Deterioration occurred after two months and infusions restarted immediately.
- I experienced increased pain, but this resolved since restarting.
- I became weaker, more tired & muscle pain increased
- Loss of treatment, muscle twinges especially in stomach

- Declined and had several falls, pain increased and wasn't sleeping as in pain. Felt more fatigued and struggled to move safely. Use a wheelchair but can walk a few steps in the house holding on to furniture. Was told to miss 6 treatments, after 3 I asked to restart with extra precautions and PPE, so I missed 8 weeks treatment of Myozyme
- Failed to start ERT. Now diagnosed 6 months and still not started. Very depressing and worrying.
- Condition deteriorated especially the breathing quite noticeably.
- Became tired achy & weaker. Treatment restarted.

Disruption to clinical trials

Eleven people responded to comment on their experience as part of a clinical trial. Five reported that the study continued as normal, for two people the trial was suspended, and one suffered some disruption. For three people, the treatment was moved from a hospital setting to homecare.

Travel to access care at a Specialist Centre

Most people had concerns about accessing care at their specialist centre, particularly about parking, toilets, PPE and infection control, and social distancing in waiting rooms. Others were less worried and will be happy to return in future.

Once the pandemic is over, and clinical care returns to normal, 18% of respondents said that they would consider transferring to a specialist centre closer to home. 32% of respondents were interested in a shared care arrangement with a local clinic or hospital, and 50% would be interesting un undertaking some medical monitoring procedures at home. 75% showed a preference for continuing with video or telephone appointments at home.

Mental Wellbeing

Understandably, we all have concerns about the pandemic, these are how people responded to a list of suggestions:

What causes you to feel worried, anxious or scared?	
Other family members catching COVID-19	54%
How things are going to return to normal	45%
Whether to continue to shield or to start going out	44%
Affected family member(s) catching Covid-19	39%
Not being able to secure home deliveries of food / medication	39%
Stopping treatment	35%
Accessing hospital for appointments	32%
Lifting of social distancing measures	27%
Allowing homecare nurses back into the home	21%
Financial worries	20%
Whether to shield or not	16%
School / college / university attendance	14%
Employment issues	13%
Exams being cancelled	4%

Information Sources

The information people found most useful was rated by respondents as:

1. NHS Specialist Centre
2. Patient Advocacy Group / Charity
3. Television
4. Government website (gov.uk)
5. Local Health Care team. i.e. GP, community nurse
6. Public Health England
7. NHS website
8. Social media
9. Press
10. Radio
11. Social care teams

Overall, respondents said that they were concerned about the Coronavirus pandemic at a level of 7 out of 10, on average. But many made specific observations about their concerns as sampled below:

- I was worried my daughter would catch it and end up in hospital
- Life has changed radically. My interaction with other people is totally reduced. Very difficult to meet new people. May never shop again in the ways I used to.
- High risk for my family and with no respite it is very stressful.
- When, if or how we will get back to normal. Family / friends becoming ill. Returning to work. Children returning to school and travelling on the bus
- I wear a mask and have done since going out again, I check places first before going in - if it's busy I leave it. I do not like crowded places. I'm not at the stage to feel comfortable going into a hospital.
- Whether my underlying condition would cause a severe reaction and serious illness, possibly death, should I catch COVID-19
- Worried about my venerable partner and family members
- Return to work safely
- Long term, progress versus complacency
- How long will the threat last?
- Treatment stopping due to a second spike
- My manners have changed, and I worried about going back to 'normal', can't imagine going on public transport and not constantly use antibac.
- Working in a school, how to manage children returning
- Death for myself or others shielding in my family
- That Covid might be with us for some considerable time. That due to my medical condition I might not survive if I was to become infected and that I would have to face it without the support of my family around.
- This affects everyone and how we all live all the time until there is a vaccine
- Other people do not follow advice, and this is frightening
- Mixed feelings vary each day

- Starting treatment again. Really worried about allowing nurses back into my home where we have had no visitors for nearly 6 months. Concern on the quality of nurses provided.
- I wonder when it will be safe to go out and whether we'll have to wait for a vaccine
- That virus will take hold again. Hard to see an end to it all.
- Others are being irresponsible and putting us at risk
- Virus has the ability to kill me very quickly or if I did survive, reduce my independence completely, putting me in a wheelchair and requiring help to breathe.
- Biggest thing that has happened in our lifetime and has affected so many people in different ways. Hard to see what the future will look like.
- Will anything be the same again?
- Confusing lack of accurate information as we have been told to continue shielding but no end date in sight. I am worried about my child's mental health as for 4 months he has had access to the garden and two rooms in the property.
- Breathing problems when lying down would make successful treatment unlikely.
- If we cannot find a vaccine. Also travel concerns going abroad.
- How safe can we be now, when can I hug family/friends, will I ever feel safe in public?
- As my breathing capacity is low and I've had other non-Pompe health issues I don't think I'd survive Covid
- Concerned about how and when and if life can return to some kind of normal. Allowing myself to go back into friends' houses for a cuppa etc.
- Virus does not concern me, the crazy world aftermath does
- Pandemic will be with us globally for a long time and will affect absolutely everything. Will be no return to the pre-covid situation socially or economically anywhere on the globe.
- My main concern is how we get back to a new "normal"
- Patient commenced on autoimmune suppressants. We as parents were worried
- Only had an FVC of 53% last year. Don't think I would survive ventilators etc.
- Concerns for my health and of those I know.
- Until there is a vaccine, I'm concerned I may be severely impacted.
- There being another spike means all this transition back to the new normal, all the stress and we have to start all over again
- Catching the disease and the complications this would cause to myself and my family.
- I'm most concerned about my elderly parents and the economy going forward
- Uncertainty of severity of illness if I or my wife get it. as could be very bad or insignificant
- I struggle to breath with a mask on & concerned about going out without a mask.
- When it will be truly safe to go out and about for someone with a respiratory condition
- With a health condition I am worried about the effect on me, and my wife and children
- Concerned about older and disabled people who do not have as much support as I have. I am concerned that the society around us will be less concerned about disabled people as things get more difficult. Wealth inequality has been increasing and the pandemic has only increased it, leading to a more stratified society which is not good or sustainable.
- Whether I can stop shielding safely

Any other comments about the survey

The final question asked for comments about the survey itself and the subjects it covered. These were the responses:

- Thank you for asking me to participate in this survey. I prefer to write than to talk so it has been good to express how I feel in this way.
- I will venture out more now but conscious that I need to ensure that I don't put myself in places/ situations where social distancing may be an issue and continue to follow all guidance around hygiene etc.
- Generally, I have coped well, family bring me food and I do not go out much anyway.
- Some parts of the survey are not clear. I have provided my answers on my interpretation of the questions.
- Excellent data to be capturing at the present time.
- It covered almost everything.
- This survey needs to include the n/a option on every question and allow you to choose more than one response on others.
- Patient group should have contacted all vulnerable members; they would have realised that many were alone and not on the vulnerable list. Many struggled through on limited food and missing essential meds. Shared info with others on Messenger, so friends kept me more updated than the charity. Felt vulnerable and left behind.
- No contact from my Hospital beyond first shielding email. A personal letter when shielding was being reduced would have been nice, as mentioned in the webinar.
- I know it seems an odd thing to say, but I've actually quite enjoyed the lock-down. The great weather has helped. I've enjoyed going for long daily bike rides with my son.
- Who is pushing for home treatment to be carried out by the patient, ie self-cannulation? My health care provider raises it at every infusion
- The use of the term "vulnerable" for those with medical conditions and disabilities is not very positive for the populations to which it has been applied.

Conclusion

The questionnaire developed by the UK LSD collaborative was a major survey with 78 questions that revealed a tremendous amount of valuable information to help both patient organisations and NHS providers understand the patient/family experience of NHS specialised services, homecare and the effect of the Covid-19 pandemic. The data will be used to shape future services and, in particular, we hope to see improvements in the provision of services that help the mental wellbeing of individuals and families, as well as their physical wellbeing.

I would personally like to thank everyone who took time to complete the survey, it was a significant investment of their time, and also all those who helped to distribute the survey so that we were able to collect such a rich cross-section of the Pompe community.

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18 February 2021