

LSD Community survey

If you or a family member is affected by a Lysosomal Storage Disorder (LSD), we invite you to share your experiences of the care and services you receive.

The survey is open to people affected by LSDs, their family members and carers. It is being conducted by the LSD Collaborative, a network of UK based patient organisations who have joined together to raise awareness of LSDs, advance standards of care and improve the wellbeing of those affected.

It will take about 20 minutes to complete and gives you the opportunity to share your views, anonymously and in confidence, about the services you receive now, including clinical care, home care, social care and patient advocacy. Your participation will help influence future LSD care and services.

For questions about the survey, please contact:

The UK LSD Collaborative

Email: info@lsdcollaborative.org.uk

Website: www.lsdcollaborative.org.uk

About you

* 1. Consent

Your responses to questions will be confidential and we will not collect identifying information such as your name, email address or IP address. All responses will be stored in a password-protected electronic format.

The results of this survey will be used to inform and influence future care for those affected by Lysosomal Storage Disorders (LSDs). To support this objective, anonymous data may be shared with NHS England, Specialist Centres, Home Care providers and rare disease patient advocacy groups..

Please indicate your agreement to participate in this survey by checking the statements below:

- I understand the purpose and nature of this survey and I am participating voluntarily.
- I understand that I can withdraw from the survey at any time, without any penalty or consequences.
- I grant permission for the anonymous data generated from this survey to be shared and used in publications
- I agree to participate in this survey.

* 2. Please provide the first three to four letters of your postcode, i.e. NE37, OX4.

3. In which country within the British Isles do you live?

- England
- Wales
- Scotland
- Northern Ireland
- Isle of Man
- Republic of Ireland
- Other (please specify)

About You

* 4. Which region within England do you live

- East Midlands
- East of England
- London
- North East
- North West
- South East
- South West
- West Midlands
- Yorkshire and the Humber

* 5. In what capacity you are participating in this study.

- Patient
- Parent
- Partner
- Carer
- Sibling
- Extended Family Member
- Patient Advocate
- Other (please specify)

PLEASE ANSWER THE FOLLOWING QUESTIONS AS THEY AFFECT YOU OR THE PERSON YOU CARE FOR (e.g. CHILD OR FAMILY MEMBER)

* 6. Which Lysosomal Storage Disorder affects you?

Batten disease

* 7. Which form of Batten Disease affects you?

8. Have you taken part, or are you participating in a clinical trial?

- No, I have not participated in a clinical trial
- CLN2 Intrathecal Enzyme Replacement Therapy (ERT)
- CLN2 Intrathecal Enzyme Replacement Therapy (ERT) (Compassionate Use)
- CLN2 Gene Therapy
- CLN6 Gene Therapy
- Other (please specify)

9. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- DEM-Child Registry
- I'm not sure
- Other (please specify)

Fabry disease

10. Have you taken part, or are you participating in a clinical trial?

- No, I have not participated in a clinical trial
- Fabry Intravenous Enzyme Replacement Therapy (ERT)
- Fabry Oral Therapy
- Fabry Gene Therapy
- Other (please specify)

11. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- Fabry (Amicus)
- Fabry (Sanofi Genzyme)
- Fabry (Shire)
- I'm not sure
- Other (please specify)

Gaucher Disease

* 12. Which Gaucher Disease Type affects you?

- type 1
- type 2
- type 3
- Other (please specify)

13. Have you taken part, or are you participating in a clinical trial?

- No, I have not participated in a clinical trial?
- Gaucher Disease Type 1 Substrate Reduction Therapy (SRT) Paediatric
- Gaucher disease Type 2 Gene Therapy (PROVIDE)
- Gaucher Disease Type 3 Substrate Reduction Therapy (SRT) Adult
- Other (please specify)

14. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- Gaucher: GAUCHERITE
- Gaucher: GARDIAN - neuronopathic Gaucher disease registry
- Gaucher: (ICGG - Sanofi)
- Gaucher: (GOS - Takeda)
- I'm not sure
- Other (please specify)

LAL-D - Lysosomal Acid Lipase Deficiency

15. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- LAL-D (Alexion)
- I'm not sure
- Other (please specify)

Mannosidosis

* 16. Which Mannosidosis affects you?

- Alpha
- Beta
- Other (please specify)

17. Have you taken part, or are you participating in a clinical trial?

- No, I have not participated in a clinical trial
- Mannosidosis Intravenous Enzyme Replacement Therapy (ERT)
- Other (please specify)

18. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- Mannosidosis Alpha: Sparkle (Chiesi)
- I'm not sure
- Other (please specify)

MLD - Metachromatic Leukodystrophy

19. Have you taken part, or are you participating in a clinical trial?

- No, I have not participated in a clinical trial
- Metachromatic Leukodystrophy (MLD) Intrathecal Gene Therapy (OTL-200)
- Other (please specify)

20. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- Metachromatic Leukodystrophy (MLD)
- I'm not sure
- Other (please specify)

Mucopolipidosis

* 21. Which Mucopolipidosis affects you?

- type I (ML 1) Neuramidase Deficiency/Sialidosis
- type II (ML 2) I-Cell N-acetylglucosamine-1-phosphotransferase
- type III (ML 3) Pseudo-Hurler polydystrophy N-acetylglucosamine-1-phosphotransferase
- type IV (ML 4)
- Other (please specify)

MPS (Mucopolysaccharidosis)

* 22. Which MPS (Mucopolysaccharidosis) affects you?

23. Have you taken part, or are you participating in a clinical trial?

- No, I have not participated in a clinical trial
- MPS I Intravenous Enzyme Replacement Therapy (ERT)
- MPS I Intravenous Enzyme Replacement Therapy (post Haemopoetic Stem Cell Transplant (HSCT))
- MPS II Intravenous Enzyme Replacement Therapy (ERT)
- MPS II Intrathecal Enzyme Replacement Therapy (ERT)
- MPS IIIA Intrathecal Safety Study
- MPS IIIA Gene Therapy
- MPS IIIB Intrathecal / Intracerebroventricular
- MPS IIIB Gene Therapy
- MPS IVA Intravenous Enzyme Replacement Therapy (ERT)
- MPS VI Intravenous Enzyme Replacement Therapy (ERT)
- MPS VI Gene Therapy
- Other (please specify)

24. Have you received or are you receiving Haemopoetic Stem Cell Therapy / Bone Marrow Transplant?

- No, never
- Yes, Once
- Yes, Twice
- Other (please specify)

25. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- MPS I (Sanofi Genzyme)
- MPS II (Shire)
- MPS IVA (Biomarin)
- MPS VI (BioMarin)
- MPS Society Database
- I'm not sure
- Other (please specify)

Niemann-Pick

* 26. Which Niemann-Pick Disease affects you?

- ASMD type A
- ASMD type B
- type C
- Other (please specify)

27. Have you taken part, or are you participating in a clinical trial?

- No, I have not participated in a clinical trial
- ASMD Niemann-Pick type B - Enzyme Replacement Therapy (ERT) Adult
- ASMD Niemann-Pick type B - Enzyme Replacement Therapy (ERT) Paediatric
- Niemann-Pick type C Oral Therapy (IB1000 - IntraBio)
- Niemann-Pick type C Intrathecal Cyclodextrin (Adrabetadex - Mallinckrodt)
- Niemann-Pick type C Intravenous Cyclodextrin (Trappsol - Cyclo Therapeutics)
- Niemann-Pick type C Oral Heat Shock Protein (Arimoclomol - KenPharm [formerly Orphazyme])
- Other (please specify)

28. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- Niemann-Pick Disease - International Niemann-Pick Disease Registry (INPDR)
- I'm not sure
- Other (please specify)

Pompe Disease

* 29. Which form of Pompe disease affects you?

- IOPD: Infantile onset Pompe Disease
- LOPD: Late onset Pompe Disease
- Other (please specify)

30. Have you taken part, or are you participating in a clinical trial?

- No, I have not participated in a clinical trial
- Pompe Intravenous Enzyme Replacement Therapy (ERT)
- Pompe Intravenous Chaperone with Enzyme Replacement Therapy (ERT)
- Pompe Gene Therapy
- Other (please specify)

31. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- Pompe (Sanofi Genzyme)
- Pompe (IPA/Erasmus Survey)
- I'm not sure
- Other (please specify)

Tay Sachs and Sandhoff Diseases

32. Are you enrolled in a registry? Please select all that apply.

- No, I am not enrolled in a registry
- CATS (Cure and Action for Tay Sachs) Registry
- I'm not sure
- Other (please specify)

Treatments

* 33. Do you have access to a treatment or therapy specific to your condition? Please tick all that apply.

- No access to therapy
- Intravenous therapy i.e. Enzyme Replacement Therapy (ERT)
- Oral therapy i.e. Chaperone Therapy / Heat Shock protein / Substrate Reduction Therapy (SRT)
- Intravenous therapy plus oral therapy i.e. ERT and Chaperone Therapy
- Intrathecal therapy
- Intrathecal therapy plus Intravenous therapy (i.e. ERT)
- Haemopoetic Stem Cell Transplant / Bone Marrow Transplant (BMT)
- Intracerebroventricular (ICV) infusions
- Gene therapy
- Therapy received through a compassionate use or expanded access programme
- Not sure

Treatments

34. If you do not have access to therapy, please tell us why:

- There is no therapy available
- A therapy is available, but not in my country/region
- Personal choice not to access therapy
- Do not meet the clinical criteria for therapy
- Other (please specify)

35. Where do you usually receive treatment. Please tick all that apply.

- At a National Specialist Centre
- At work
- At School/University
- At Home
- At a local hospital/health centre
- At another family member's home
- Other (please specify)

Advocacy

* 36. Do you regularly interact with a patient advocacy group or charity linked to your condition?

- Yes
- No

Advocacy

37. Why don't you interact with a Patient Advocacy Group or Charity?

- I did not know there was a patient advocacy group or charity for my condition
- I don't think a patient advocacy group or charity could help me
- I don't want to interact with a patient advocacy group or charity for my condition
- Other (please specify)

Advocacy

38. If you would like to know more about the Patient Advocacy Groups that supports your condition and the ways in which they may be able to assist, please provide your preferred contact details below.

To share your contact details with the relevant group, we will need your explicit consent.

- I give explicit consent for my contact details to be securely shared with the relevant group. I understand that my information will only be used for this purpose and will not be used or shared in any other way.

My contact details are:

Advocacy

39. Please tell us which patient advocacy groups or charities you interact with.

Tick all that apply.

- AGSD UK - Pompe and Danon Disease
- Alex, The Leukodystrophy Charity
- Batten Disease Family Association - Batten Disease
- CATS Foundation - Tay-Sachs and Sandhoff Diseases
- Gauchers Association - Gaucher Disease
- Genetic Alliance UK
- Krabbe UK - Krabbe Disease
- Metabolic Support UK
- MLD Support Association UK (Metachromatic Leukodystrophy)
- MPS Society - MPS, ML, MLD, Fabry and related Lysosomal Storage Disorders
- Muscular Dystrophy UK - Pompe Disease
- Niemann-Pick UK - Niemann-Pick Diseases
- Pompe Support Network - Pompe Disease
- Rare Disease UK
- Other (please specify)

40. Please rate the services you receive from the patient advocacy groups / charities

	Not applicable	Not at all satisfied	Somewhat satisfied	Satisfied	Highly satisfied
Information provision	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Telephone or online support	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Practical support	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Emotional / mental health support	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Support or Advocacy team	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Nursing support	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Events and conferences	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Fundraising support	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Support during the COVID-19 crisis	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Comments:

Specialist Centres

* 41. Which Specialist Centre do you attend?

Specialist Centres

42. Why don't you attend a Specialist Centre?

- There is no Specialist Centre near me
- It is too difficult to travel to a Specialist Centre
- I haven't been referred to a Specialist Centre
- Prefer to receive care elsewhere

If you receive care elsewhere, please tell us where you usually access healthcare services?

48. Do you communicate with your Specialist Centre between appointments?

- Regularly
- Sometimes
- Rarely
- Never

49. Please tell us how you prefer to communicate with your Specialist Centre:

- Dedicated team email address
- Dedicated telephone help line
- Direct contact with nurse specialist
- Direct contact with consultant
- Other, please specify:

50. Please rate your experience at your Specialist Centre:

	Strongly Disagree	Disagree	Not Applicable	Agree	Strongly Agree
It is easy to park at the Specialist Centre	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
The Specialist Centre is clean	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
The facilities (restaurants, cafes, shops etc) at the Specialist Centre are good	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Staff are friendly and helpful	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Staff are competent and knowledgeable about my condition	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I can access all of the healthcare specialities and services needed in one place	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I have a good relationship with the Specialist Centre healthcare team	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I am treated with dignity and respect	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I am encouraged to ask questions and give feedback	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Please add comments here:

51. Overall, how would you rate your experience at your Specialist Centre?

- Not at all satisfied
- Somewhat satisfied
- Satisfied
- Very satisfied

52. Is there any aspect of your care that is not being met by your Specialist centre?

53. What could be done to improve your experience of attending your Specialist Centre?

54. Many LSD patients receive care across different clinical specialities. Please tell us which specialist areas you have experience of:

- | | |
|---------------------------------------------------------------|----------------------------------------------------------------------|
| <input type="checkbox"/> Audiology (hearing) | <input type="checkbox"/> Orthopaedics - bone health/Orthotics |
| <input type="checkbox"/> Cardiology (Heart) | <input type="checkbox"/> Ophthalmology - Eyes |
| <input type="checkbox"/> Dentistry/maxillofacial/Orthodontics | <input type="checkbox"/> Physiotherapy |
| <input type="checkbox"/> Dermatology (Skin) | <input type="checkbox"/> Psychology - mental health |
| <input type="checkbox"/> Dietitian or Nutritionist | <input type="checkbox"/> Radiology (MRI, CT Scan, X-Ray, Ultrasound) |
| <input type="checkbox"/> Metabolic medicine | <input type="checkbox"/> Respiratory, Lung function |
| <input type="checkbox"/> Neurology | <input type="checkbox"/> Speech and Language Therapy (SALT) |
| <input type="checkbox"/> Occupational Therapy (OT) | <input type="checkbox"/> Urology |
| <input type="checkbox"/> Other (please specify) | |

Home Care Services

55. Do you access Home Care Services for specialist treatment and/or medical deliveries?

- Yes
 No

Home Care Services

56. Please tell us the name of your Home Care service provider:

- Lloyds Pharmacy Clinical Homecare (LPCH)
 Healthcare at Home
 Pharmaxo
 HealthNet Homecare
 Other (please specify)

57. How satisfied are you with the service provided?

- Not at all satisfied
- Somewhat satisfied
- Satisfied
- Very satisfied

58. How is home-care working for you? (check all that apply)

	Working well	Not working	Not applicable
Telephone booking	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Nursing care	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Deliveries	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

59. If you have complained about your Home Care service, how was it made (check all that apply):

- Directly to home care company
- Through NHS specialist centre
- Through patient organisation

60. Did you receive a response to your complaint?

- Yes
- No

Comments:

61. If you have a medical fridge supplied by the home-care company, is it checked/serviced regularly (at least once a year)?

- Yes
- No
- Not applicable

62. What would you like to see included in your home-care service that is not already provided?

63. How do you think the home-care service could be improved?



The Impact of Coronavirus (COVID-19)

The Coronavirus pandemic continues to affect medical and social care services. In the following questions, we are seeking to specifically understand the impact on patients and families affected by Lysosomal Storage Disorders and how it may have affected the care, treatment, and services you receive.

64. As society fully reopens after the pandemic, are you:

- Continuing to Shield
- Starting to go out
- Unsure, the advice is confusing
- Question is not applicable to me

Comments

65. Are you now able to access the following?

Please select all that apply.

- | | |
|-------------------------------------------------------------------------|------------------------------------------------------------------------------|
| <input type="checkbox"/> Regular medical care and appointments | <input type="checkbox"/> Personal care / support at home (external provider) |
| <input type="checkbox"/> Urgent medical care | <input type="checkbox"/> Social care |
| <input type="checkbox"/> Medical tests or scans | <input type="checkbox"/> Respite care |
| <input type="checkbox"/> Prescribed treatment or therapy | <input type="checkbox"/> Hospice care |
| <input type="checkbox"/> Treatment received as part of a Clinical Trial | <input type="checkbox"/> Medical supplies |
| <input type="checkbox"/> Monitoring visit as part of a Clinical Trial | <input type="checkbox"/> Groceries |
| <input type="checkbox"/> Physiotherapy | <input type="checkbox"/> Work |
| <input type="checkbox"/> Speech and Language Therapy (SALT) | <input type="checkbox"/> School / Higher Education |
| <input type="checkbox"/> Psychological / mental health support | |
| <input type="checkbox"/> Other (please specify) | |

66. If your routine appointments were changed to video and/or telephone call, please tell us how satisfied you were with this option:

	Not at all satisfied	Somewhat satisfied	Highly satisfied
Video appointment	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Telephone appointment	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Would you like these services to continue?

Mental wellbeing

67. Since the pandemic, what has caused you to feel worried, anxious or scared?

Please tick all that apply.

- Affected family member(s) catching Covid-19
- Other family members catching COVID-19
- Whether to continue to shield or to start going out
- Allowing homecare nurses back into the home
- Accessing hospital for appointments
- Financial worries
- Employment issues
- School / college / university attendance
- How things are going to return to normal

Other (please specify)

68. Do you or a member of your household suffer from poor mental health?

- Yes
- No
- Not sure

69. If yes, do they receive counselling or emotional support?

- Yes
- No

70. What is the nature of the emotional support?

- GP
- Clinical psychologist
- LSD Specialist Centre
- Patient Organisation
- Counselling
- Other (please specify)

71. Is the emotional support satisfactory?

- Yes
- No

72. Does your mental wellbeing benefit from:

- Music - playing or listening
- Exercise
- Gardening
- Art
- Crafts and hobbies
- Dancing
- Social networks
- Reading
- Volunteering
- What would you recommend?

73. Can you describe some of the key emotions you have experienced (i.e. anxiety, worry, loneliness, anger)?

74. Have you had genetic counselling?

- Yes after diagnosis
- Yes for advice on reproductive choices
- No
- Other reasons? (please specify)

Social Care

75. Do you receive support from social services?

- Yes
- No

76. Are you satisfied with the social care support you receive?

Yes

No

77. Can you describe any issues you have with social care services?

78. What care services do you receive

Social care and support

Household gadgets and equipment

Personal alarms and security systems

Home adaptations

Walking and mobility aids

Home help

Transport assistance

None of the above

Respite Care

79. Do you use respite care services?

Yes

No

80. Are you satisfied with the Respite care support you receive?

Yes

No

81. Can you describe any issues you have with Respite care services?

Hospice Care

82. Do you use Hospice care services?

Yes

No

83. Are you satisfied with the Hospice care support you receive?

Yes

No

84. Can you describe any issues you have with Hospice care services?

Survey comments

85. Finally, please feel free to add any further comment relating to the subject areas covered by this survey: