

Transition in Lysosomal Storage Diseases - results of a UK patient and carer survey

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Background

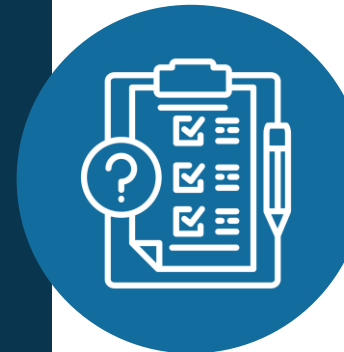
Lysosomal storage diseases (LSDs) are a group of inherited metabolic disorders¹. Early diagnosis, disease-modifying therapies and improved supportive care in childhood has increased survival, necessitating transfer of care to adult providers². Ineffective transition can lead to loss of engagement and non-adherence to treatment with adverse consequences in terms of mortality and morbidity as well as social and educational outcomes³.

The UK LSD Highly Specialised Services set up a multi-centre stakeholder group to review current service provisions and develop LSD specific key principles, standards and resources. This included a national transition pathway, transfer checklist and formal quality assessment process⁴.

To help support and benchmark historical and current patient and carer experiences of transition, the MPS Society co-designed a survey with LSD clinical centres. Associated LSD patient groups then sent out the survey to its members to capture: **Experiences and recommendations for improvement**

Lysosomal storage diseases (LSDs) are inherited metabolic disorders characterized by the accumulation of substrates within cells due to defective lysosomal functioning¹

Methods



The study was open to those:

- People with an LSD condition aged 16 years and above
- Caregivers completing survey on behalf of people with an LSD condition aged 16 years and above
- People who have transitioned or are in the process of transitioning to adult care
- Are a resident of the UK



The study was open from 19/03/2024 until 09/08/2024



Survey was shared online and contained multiple choice and free text questions



Participants gave informed consent to participate

Purpose

- To understand the experiences of patients aged 16 years and above who are or have gone through the transition from paediatric to adult care services
- To evaluate how patient experiences inform the newly implemented transition pathway, principles and standards

Aim & Objectives

- Understand patient experiences of transitioning from paediatric to adult services
- To gather current and previous views on the transition process
- To capture recommendations on what the service should consider going forward

Responses

Demographics

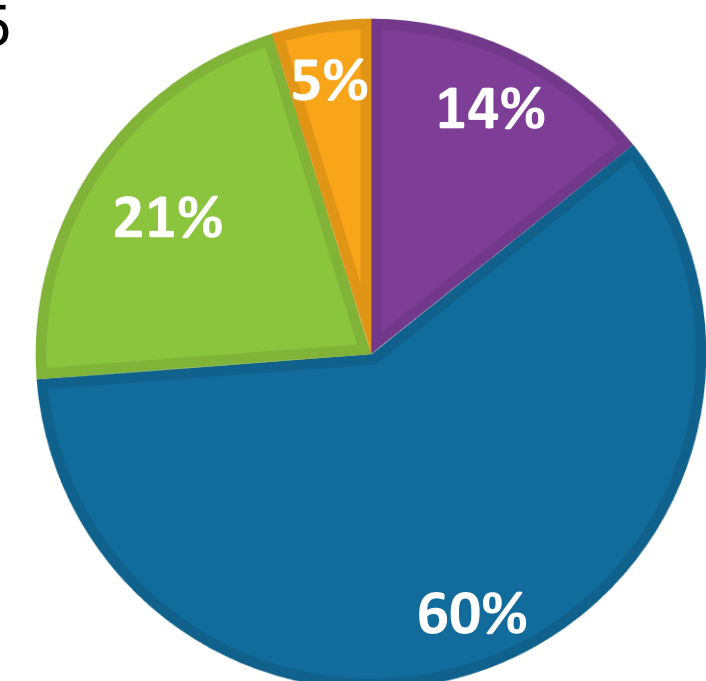
42 responses were received

35 had transitioned and 7 were in the process

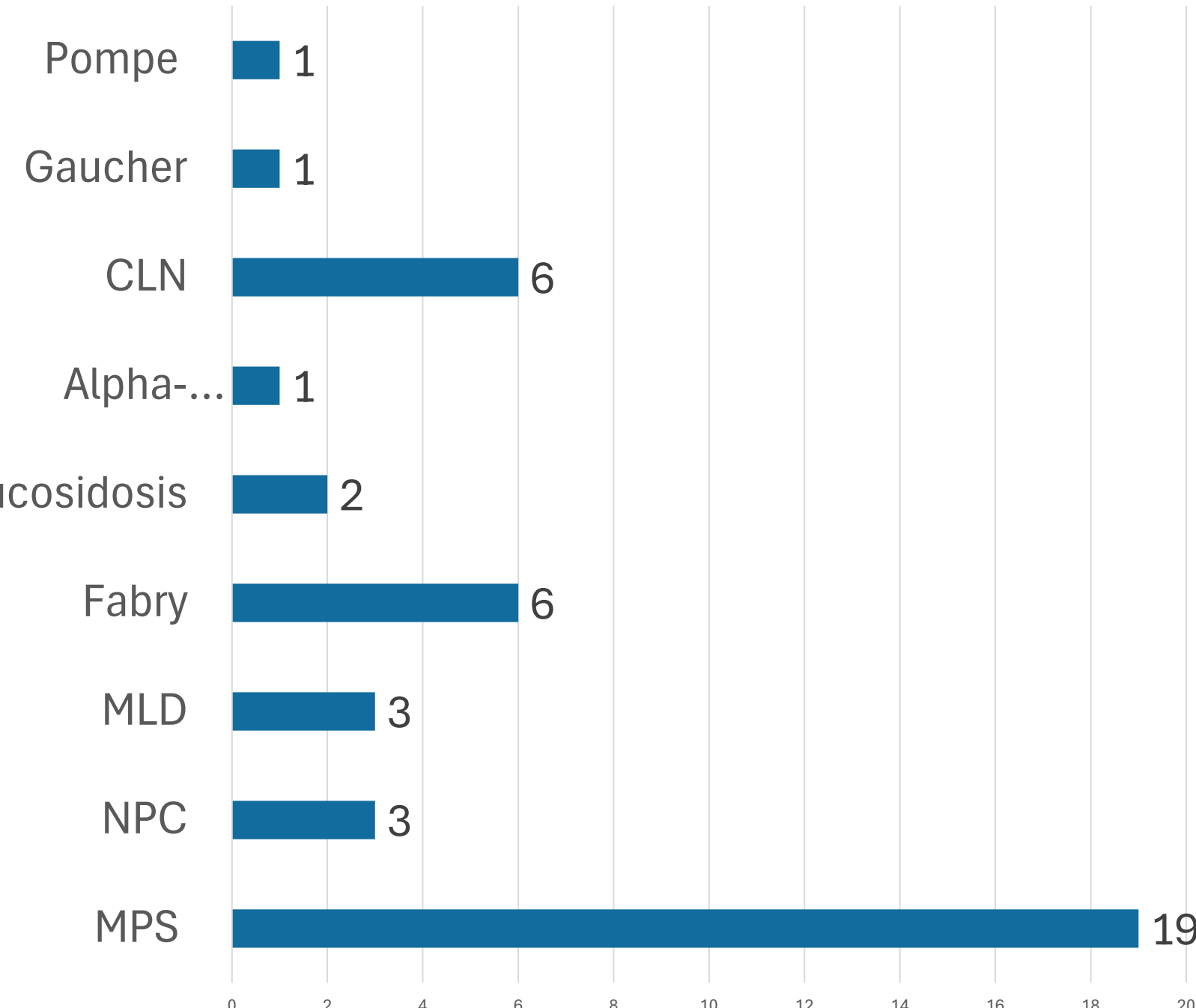
57% males
41% females
2% preferred not to say

Current age of respondents

- 16-17 yrs = 6
- 18-25 yrs = 25
- 26-37 yrs = 9
- 38 & over = 2



Responses by disease type



Who completed the survey

- 19% Patients 16 yrs or older
- 81% Carers of patients 16 yrs or older
- 90% of respondents live in England

Results

Survey responses

	Numbers who have attended or are attending a transition clinic	Numbers who have not attended a transition clinic	Total numbers
Age range of respondents	16-43 years		
Age informed about transition	14-23 years		
Transition completed already	35 (83%)		
Proportion who are or have attended a transition clinic	13 (31%)	0	13 (31%)
Able to meet adult team pretransfer	6 (14%)	12 (29%)	18 (43%)
Provided information about transition	5 (12%)	9 (21%)	14 (33%)
Provided information about adult centres	3 (7%)	2 (5%)	5 (12%)
Transition adequately explained	8 (19%)	9 (21%)	17 (40%)

How could transition process be improved- Feedback from our community

	Need for support and Information during transition	Need for to Paediatric team to be present	Challenges in adult care	Emotional and psychological Impact
Themes	Many respondents highlighted the need for better preparation, information, and emotional support when transitioning from paediatric to adult care	A repeated suggestion was for the paediatric team to be present during first adult appointment	Several statements reflect dissatisfaction with adult healthcare services, such as long waiting times, lack of specialised care, and coordination problems	The emotional impact of transitioning from paediatric to adult care is a recurring theme. The loss of parental involvement and support is frequently mentioned as a challenging aspect.
Examples	"A chance to visit or discuss prior and be given contacts and support info" "Important that the child and parents are introduced via a transition meeting with all professionals involved"	"The paediatric team should be present at the first adult appointment" "It is important that you meet with your existing consultant, specialist nurse, and new care team"	"Adult care services for MPS children do not exist" "Adult teams are not as joined up and they don't speak to each other in the way that they do in child services"	"Patients should not be treated as adults immediately, and parents should not be ignored from the outset." "Would be very helpful and reassuring that you are physically, mentally and emotionally prepared for you transition"
Insights	There is a clear pattern of patients and parents feeling underprepared for the transition and requesting more structured support	Respondents see value in having continuity and presence from the paediatric team during the transition to reduce anxiety and ensure a smoother process	Respondents shared that they felt adult services were less organised, lacked coordination, and don't provide the same level of care or understanding that paediatric services do	The emotional readiness of patients and the gradual introduction of adult care are highlighted as crucial. There's a significant concern about the emotional impact on both patients and their families during the transition.

Summary and recommendations

- Age where transition is introduced needs to improve
- Scheduling of transition appointments would aid preparation
- Joint clinics with both paediatric and adult teams is important
- Having a contact person at the adult centre is important
- Knowing available adult centres and being able to visit prior to transition is helpful
- Need for better information and emotional support
- Clear step by step information that is not too involved, or complex
- Involving parents in transition is important
- Need to ensure young person is physically, mentally and emotionally prepared for transition
- Need to consider whether the young person has capacity to understand the process

Conclusions

Survey results highlighted that those who attended a transition clinic had a more positive transfer, especially when clinics involved individuals from the adult team.

This study helped validate the hypothesis that patients' readiness for the transfer of care needs to be assessed during several transition clinics to ensure they fully engage with the adult services.

In summary, the outcomes from the study underscore the importance for a collaborative approach, with good links with adult teams at an early transition stage.

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References:

- 1 Rajkumar V, Dumpa V. Lysosomal Storage Disease. [Updated 2023 Jul 24]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan-. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK563270/>
- 2 Stepien KM, Kiec-Wilk B, Lampe C, Tangeraas T, Cefalo G, Belmatoug N, Francisco R, Del Toro M, Wagner L, Lauridsen AG, Sestini S, Weinhold N, Hahn A, Montanari C, Rovelli V, Bellettato CM, Paneghetti L, van Lingem C, Scarpa M. Challenges in Transition From Childhood to Adulthood Care in Rare Metabolic Diseases: Results From the First Multi-Center European Survey. *Front Med (Lausanne)*. 2021 Feb 25;8:652358.
- 3 Rigby E et al. Getting health services right for 16-25 year olds. *Arch Dis Child* 2020
- 4 Morris E, Stewart C. Lysosomal Storage Disorders Services: A review of transition from paediatric to adult services in England. *BIMDG*, Nov 2023 [online] accessed 12/03/2024 available at <https://www.bimdg.org.uk/site/guidelines-lsd.asp?t=1>