FABRY MATTERS CONFERENCE 2024

Insights Preview

In March 2024, the MPS Society organized a Fabry conference, bringing together patients, researchers, and clinicians for a weekend of connection and knowledge-sharing within the Fabry community. Insights were gathered during the event from Fabry patients and their caregivers through surveys and interactive post-it note boards, providing valuable perspectives on their experiences and needs.

MPS support research awareness

Rare Disease
Research Partners

The data gathered during the conference can be analysed across various factors, including age, sex, ethnicity, household income, Fabry type, treatment type, level of education, mental health, and much more! For a detailed overview of all the data collected, please refer to **page 6**.

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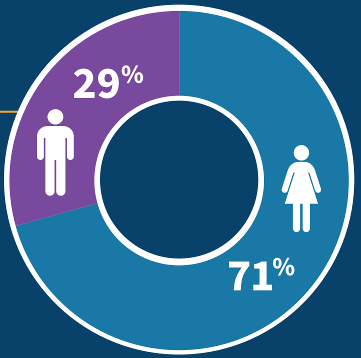
SURVEY 1 – Participant demographics

GENERAL DATA

58 patients completed the patient demographics survey

- **56** were a person with Fabry, including 2 patients awaiting diagnosis
- **2** were caregivers completing the survey on behalf of a person with Fabry (both were parents of children with Fabry not attending the conference)

01 SEX AT BIRTH



02 CURRENT AGE

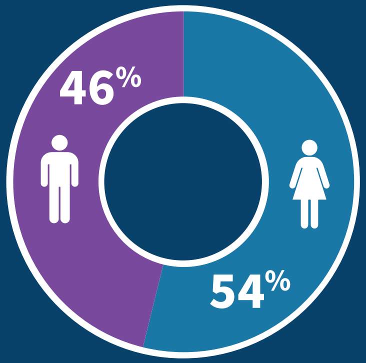
Median age of respondents was 49 years, with patients ranging between 9 and 72 years old.

03 ETHNICITY

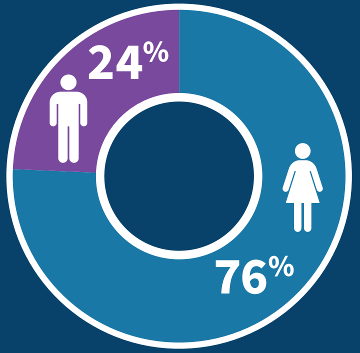
Most participants at the conference were from white backgrounds; however, we also had some representation from Asian and mixed/multiple ethnic backgrounds among the respondents.

04 DIAGNOSIS

Patients diagnosed because they were experiencing symptoms (n = 13)



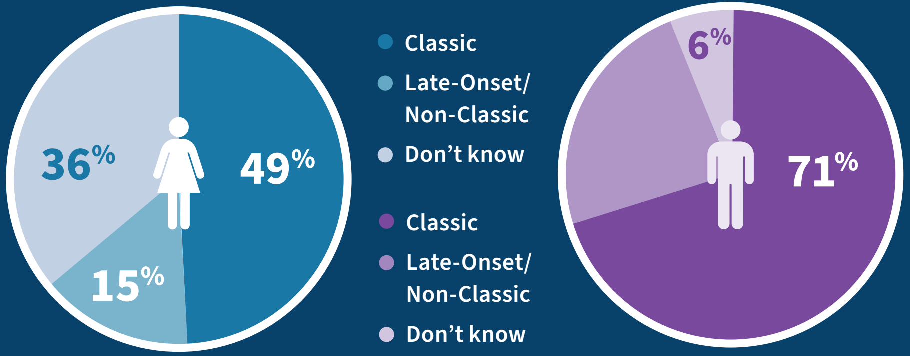
Patients getting diagnosed because of another family member's diagnosis (n = 37)



- Six patients indicated their diagnosis was not due to symptoms they experienced or a family member's diagnosis and selected "other" instead.
- The majority of female Fabry patients are diagnosed following a family member's diagnosis, typically before the age of 30. In contrast, women diagnosed after the age of 40 are more likely to have been identified based on symptoms.

05 TYPE OF FABRY

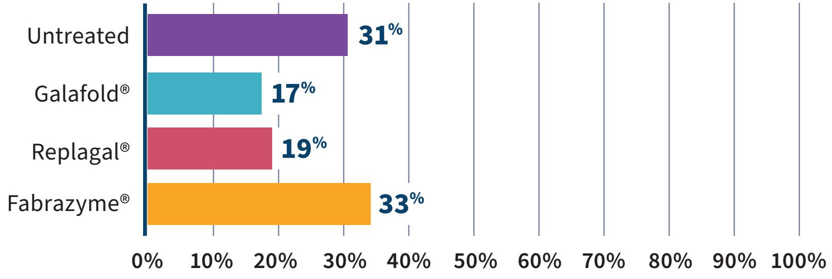
Type of Fabry in females (n=39) **Type of Fabry in males (n=17)**



A significant number of patients are unaware of their specific type of Fabry disease, possibly due to a lack of communication from clinicians to patients or differing classification approaches among professionals. Our survey offered only "Classic" and "Non-classic" categories, but some patients may have been told they have cardiac or renal variants, leading to uncertainty. Further research is needed to clarify Fabry disease subtypes across genders and improve how this information is communicated to patients.

06 TREATMENT

Type of treatment patients are on



SURVEY 2 – Symptom management

GENERAL DATA

52 patients completed the symptom management survey

01 EFFECTIVENESS AT LIMITING PROGRESSION

Among male Fabry patients, **29%** rated their treatment as **very effective** in limiting disease progression, compared to **38%** of female patients. Moreover, **8%** of females reported their treatment as **extremely effective**, while no males selected this option. Additionally, **7%** of males and **4%** of females found their treatment **not effective at all**.

02 EFFECTIVENESS AT MANAGING SYMPTOMS

Among Fabry patients, **29%** of **both** males and females found their treatment to be **very effective** at managing symptoms. Moreover, **4%** of female patients rated their treatment as **extremely effective**, compared to none of the males. Furthermore, **21%** of males reported their treatment as **not effective at all**, significantly higher than the **4%** of females who also reported the same.

03 SYMPTOMS WHICH HAVE THE BIGGEST IMPACT ON PATIENTS

36% Memory problems or brain fog

66% Fatigue

36% Stomach pain and digestive issues

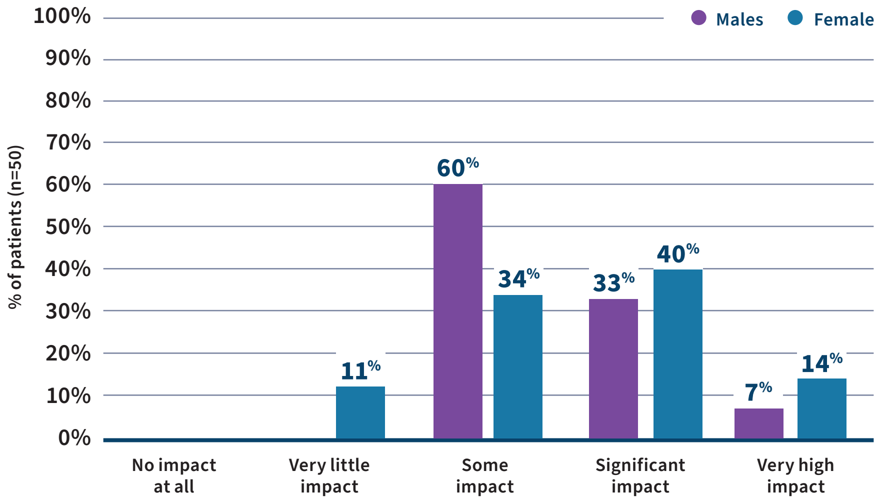
Pain crises were reported as one of the most impactful symptoms by **53%** of male Fabry patients, compared to **3%** of females.

Memory problems emerged as one of the most impactful symptoms for **43%** of female Fabry patients, compared to **20%** of males.

04 LEVEL OF IMPACT SYMPTOMS HAVE ON THEIR DAY-TO-DAY LIFE

42% reported that fatigue had a severe or significant effect on their day-to-day life.

05 MENTAL HEALTH & WELLBEING



06 HOW SUPPORT COULD BE IMPROVED

- Improved communication among different HCPs
- Increased information on lifestyle changes, supportive therapies, Fabry disease, symptoms and treatment
- Inclusion of alternative therapies alongside standard treatments
- Greater emotional support
- More regular follow-up appointments

SURVEY 3 – Attitudes to research and clinical trials

GENERAL DATA

52 patients completed the attitudes to research and clinical trials survey

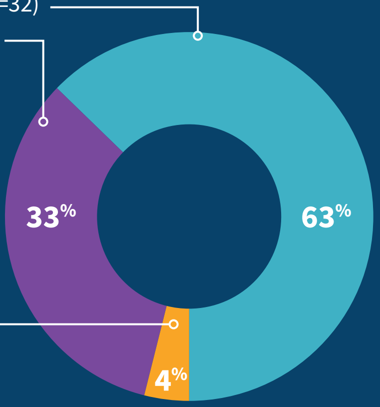
01 WHERE DO YOU FIND OUT ABOUT RESEARCH ACTIVITIES

64% learn about research activities through the MPS Society, while

56% find out through their Fabry specialist centre

02 LIKELY TO PARTICIPATE IN THE FUTURE

- Very likely = **63%** (n=32)
- Likely = **33%** (n=17)
- Neither likely or unlikely = **0%**
- Unlikely = **4%** (n=2)
- Very unlikely = **0%**



03 SUMMARY OF RESULTS

88% indicated receiving a summary of results of the study was important to them

04 KNOW WHEN CLINICAL TRIALS ARE TAKING PLACE & HOW THEY WOULD LIKE TO BE NOTIFIED

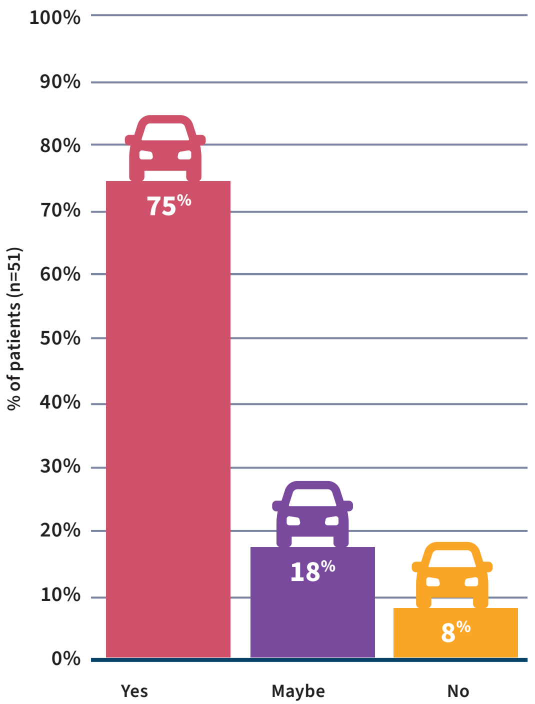
65% were unaware of when clinical trials are taking place.

When asking why patients did not know when clinical trials are taking place, **88%** said they did not know where to find information about clinical trials. This highlights a significant gap in reaching the patient community regarding clinical trial information.

When asked how the patient community would prefer to be notified about clinical trials, **73%** indicated they would like to receive information through their healthcare provider, and **80%** through the MPS Society.

05 LOGISTICS

We asked if the patient community would be more inclined to take part in trials if someone took care of all the logistics:



EMOTIONAL MAPPING
throughout a patient's Fabry journey

We asked patients to share a word or phrase that encapsulated their feelings throughout their journey with Fabry disease. The responses were analysed and categorised into participants' sentiments. Below are a few quotes derived from these categorised responses.

"When I was diagnosed with Fabry I felt..."

PLEASED & COMFORTED
"Relieved that a reason was found for all my symptoms."

ANXIOUS & FEARFUL
"Initially shocked and overwhelmed and I'd never heard of the Fabry condition."

"Supporting a loved one with Fabry, I feel..."

EMOTIONAL DISTRESS
"Frustrated, exhausted, worried for my partner and my future."

ENCOURAGING
"Worried but hopeful as we support and push forward with the treatments and innovations we see coming."

"Living with Fabry now, I feel..."

POSITIVE EMOTIONAL WELL-BEING
"Supported by MPS [Society] and some of the other families I have met. I feel more confident after various discussions at the Fabry conference to open up and ask questions and will be doing so when I next go to my specialist centre. Feel very supported by the therapy I have received from rare minds and very grateful for this."

DISTRESSED
"Scared, confused, worried about children, grandchildren. Out of control. Anxious."

"I feel ... about my future with Fabry"

EMOTIONAL DISTRESS
"Worried as I get older because I don't know how it will affect me."

POSITIVE & EMPOWERED
"More protected and in control with medication and now part of a supportive specialist community."

"Being on treatment for my Fabry, I feel..."

GRATITUDE
"Grateful that I am being given a chance to live as well as I can."

DOUBT
"Better in some areas but less consistent in mental health."

"Not having treatment for my Fabry, I feel..."

DISTRESSED
Dismissed and like my life isn't valued because I'm not ill enough!"

POSITIVE
"Relieved that it's not that bad."

Survey 1

Participant demographics

DEMOGRAPHICS

1. Age (years & months)
2. Sex at birth
3. Ethnicity
4. First language?
5. Where do you live?
6. Total annual household income
7. Highest grade or level of education completed
8. Employment or education status

DIAGNOSIS & TREATMENT

9. Do you have a confirmed diagnosis of Fabry?
10. Age at diagnosis
11. Type of Fabry
12. What led to your diagnosis?
13. What specialist centre are you under the care of?
14. Are you currently on treatment?
15. Age treatment started
16. Have you ever switched treatments?
17. How many times have you switched treatments?
18. Sequence of treatment switches
19. Why did you switch treatments?
20. Have you previously received treatment (if not currently on treatment)?
21. What treatment you most recently on (if not currently on treatment)?
22. Why did you stop treatment (if not currently on treatment)?
23. Why have you never received treatment (if not currently on treatment and not previously received treatment)?

Survey 2

Symptom Management

TREATMENT

1. Are you currently on treatment?
2. How effective is your treatment at limiting progression with Fabry?
3. How effective is your treatment at managing your symptoms overall?

SYMPTOMS

9. In the last 6 months, have you experienced any symptoms?
10. Which of your symptoms have the biggest impact on your life right now (max 3)
11. How much of an impact do the following symptoms have in your day-to-day life?
12. How effective is your treatment at managing symptoms?
13. How do you manage your symptoms outside of standard Fabry treatment?
14. How much do your symptoms impact your ability to work or attend education?
15. How would you rate the impact of Fabry on your overall mental health and emotional well-being?

MANAGING YOUR SYMPTOMS: HEALTH CARE PROFESSIONALS

16. Which specialist(s) do you currently see to monitor and manage your Fabry?
17. Who is your primary healthcare contact for discussing anything Fabry related?
18. How easy is it to get in contact with your primary healthcare provider?
19. How satisfied are you with the support and information you received from healthcare professionals regarding management of your Fabry symptoms?
20. How could the support you receive be improved?

Survey 3

Attitudes to Research and Clinical Trials

ATTITUDES TO RESEARCH

1. How often do you take part in research activities related to Fabry?
2. Where do you usually find out about participating in research activities related to Fabry?
3. What type of research activities do you prefer to participate in?
4. What encourages you to participate in research activities?
5. Please rank your choices in order of those that encourage you the most to the least.
6. What might stop you from participating in research activities?
7. Please rank your choices in order of those that might stop you the most to the least.
8. What is the maximum amount of time you would be willing to spend taking part in a survey?
9. How likely are you to participate in future research activities related to Fabry?
10. If you were offered payment for completing a research study, what would you prefer to receive?
11. If research studies offered no payment or rewards, would you still participate?
12. When participating in research activities, how important is it for you to receive the summary of the results in the study you participated in?

ATTITUDES TO CLINICAL TRIALS

13. Do you know when clinical trials are taking place?
14. How do you find out about these clinical trials?
15. Why do you think you haven't heard anything about clinical trials in Fabry?
16. How would you like to be notified of the current clinical trials?
17. Have you ever participated in a clinical trial for Fabry?
18. If you have participated or are currently in a clinical trial, what would make the experience easier for you?
19. How likely are you to take part in the following clinical trials for Fabry in the future? (ERT clinical trials, Gene therapy, treatment)
20. What would most likely encourage you to take part in a clinical trial?
21. Please rank your choices in order of those that encourage you the most to the least.
22. What would discourage you from taking part in a clinical trial?
23. Please rank your choices in order of those that discourage you the most to the least.
24. Would you be more inclined to participate in clinical trials if someone took care of all the logistics (e.g., travelling and accommodation, financial security, relocation, etc.)?

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If you are interested in understanding more about Fabry Matters and the data gathered, please get in touch with Rare Disease Research Partners:

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