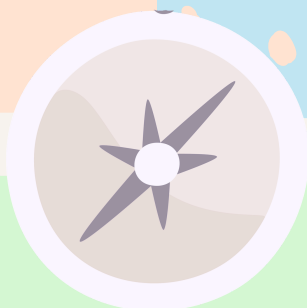


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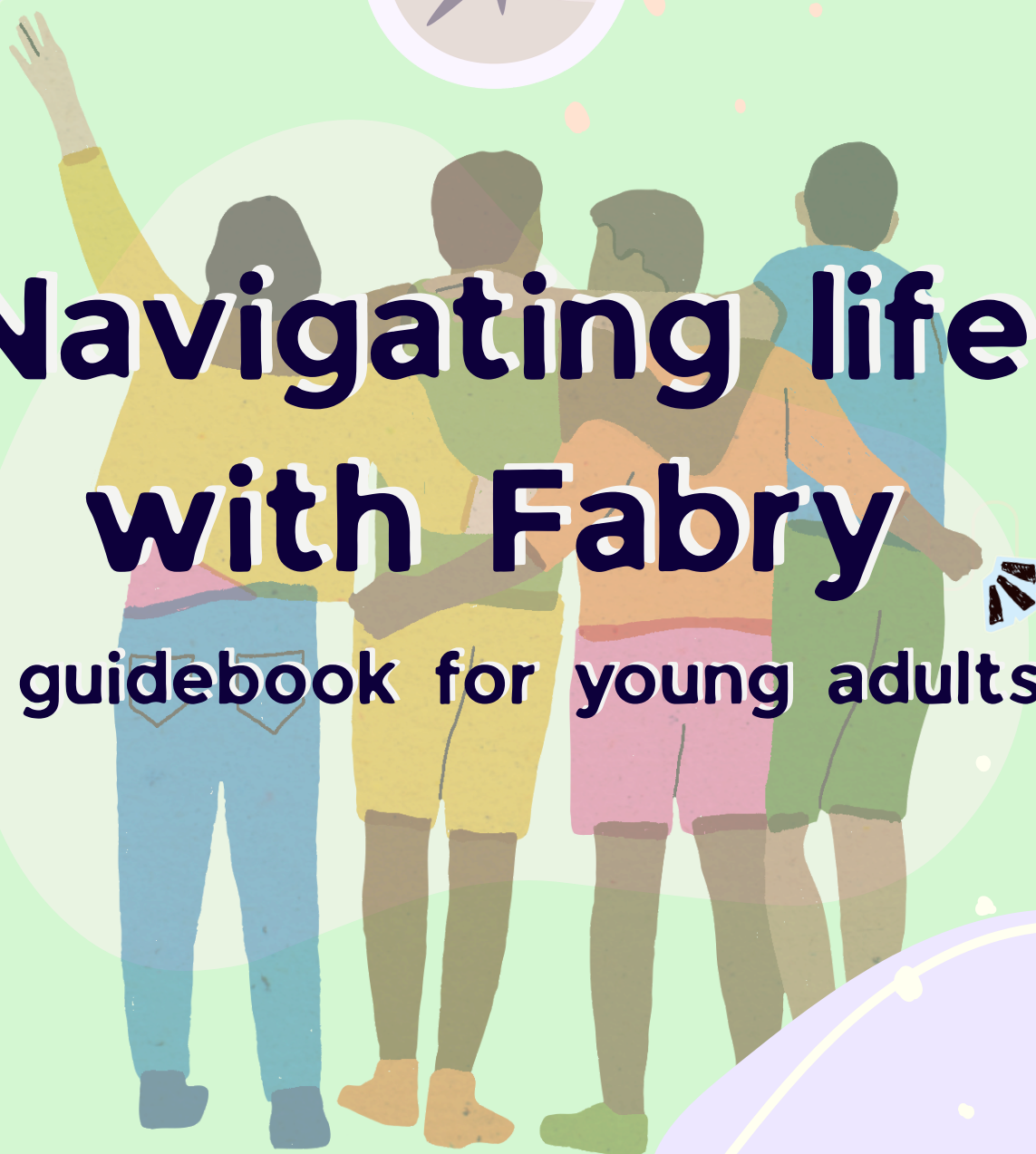
Fabry International Network

FIN young adults



Navigating life with Fabry

a guidebook for young adults



Foreword

by Anna Meriluoto,

Mentor of the FIN Young Adults Program



If you're reading this, chances are you're a young adult living with Fabry or you're close to someone who is. Either way, I want you to know one thing first: you're not alone, even if it sometimes feels that way.

The FIN Young Adults program was created because we saw a gap. Not in information, there's plenty of that, but in support that fits this phase of life. Being a young adult with Fabry comes with its own questions, challenges, and contradictions. You're figuring out who you are, where you're going, what you want from life... all while carrying something that most of your peers don't have to think about at all.

One thing we want to say clearly is this: you are more than your Fabry. It may be a significant part of your life, and there's no denying its impact, but it does not define your whole identity. We see you as a full person: with dreams, doubts, talents, humour, relationships, and a future that is uniquely yours.

When I was younger, I would have loved to have something like this guide. Not a textbook. Not a checklist. But a place where I could recognize myself. Where I could find answers and reassurance. Where someone would say: yes, this is hard and yes, you're allowed to take your time.

We believe deeply in young adults. In your strength, your resilience, your creativity, and your ability to shape your own path even when that path looks different from what you imagined. We also know that your needs will change over time. What you need when you're newly diagnosed is not the same as

Foreword

by Anna Meriluoto,

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what you need when you're navigating studies, work, relationships, family planning, or simply learning how to live well in your own body.

That's why this guide isn't meant to be read from start to finish in one go. Think of it as something you can come back to at different moments in your life, taking what's useful now and leaving the rest for later. There's no right pace, no right order, and no "right way" to be a young adult with Fabry.

This guide was created with young adults, not just for them. It brings together lived experience, practical tools, and honest conversation, the kind many of us wish we'd had earlier. Our hope is that it helps you feel more informed, more confident, and more supported as you make decisions that are right for you.

While every effort has been made to ensure the accuracy of the information shared, this guide reflects knowledge at the time of writing and is not a substitute for personalised medical advice. We encourage you to consult your healthcare team and refer you to the [full disclaimer](#) for further information.

If there's one gentle invitation we'd like to offer, it's this: don't do this alone. Whether that means connecting with other young adults, joining a FIN activity, reaching out for support, or simply knowing that this community exists, you are welcome here, exactly as you are.

Take what you need. Skip what doesn't resonate. And remember: your story is still unfolding, and it matters.

Anna ✨

A note of thanks to contributors and reviewers



This guide would not exist without the many people who shared their time, knowledge, experience, and care to help shape it.

Our heartfelt thanks go to the young adults who contributed their voices, stories, questions, and reflections. Your openness, honesty, and courage are at the core of this guide, and they remind us why this work matters.

We are deeply grateful to the healthcare professionals, experts, advocates, and community members who reviewed the content, shared their insights, and helped ensure that this guide is accurate, relevant, and grounded in real life. Your thoughtful feedback and willingness to engage made this resource stronger, clearer, and more meaningful.

We would also like to acknowledge the dedicated work behind bringing this guide to life. We are deeply grateful to Anna Meriluoto, FIN Vice President, and Tatevik Yezekyan, FIN Director, for their meaningful contributions, thoughtful input, relentless dedication, and ongoing collaboration throughout the development of this guide. Their perspectives, care, and engagement have helped shape its direction and depth.

We would also like to thank Nicolas Convertini and Narine Fischer from the FIN Young Adult Community who generously dedicated their time, reflections, and lived experience to extensively review and contribute to the guide. Their honest feedback, insights, and perspectives as young adults living with Fabry disease helped strengthen and enrich this resource in invaluable ways.



A note of thanks to contributors and reviewers



In particular, our sincere thanks to Charlotte Wauters, FIN Coordinator, for shaping the vision and leading the development of the guide, guiding the content, and weaving together the many voices and contributions into the guide you see today.

We also want to thank those who inspired this work through conversations, shared experiences, mentorship, and sometimes simply by showing up and being real. Many of you may not see yourselves named on these pages, but your influence is felt throughout.

This guide is the result of collaboration, trust, and a shared belief that young adults living with Fabry deserve resources that truly reflect their lives, in all their complexity, strength, and individuality.

Thank you for believing in this project, and in the young adult community it was created for.

thank you!



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Start here ...



*"Everyone's journey is different
and that's okay"*

No two experiences with Fabry disease are exactly the same.

You might have been diagnosed recently or years ago. Maybe it came as a surprise during routine tests, or maybe you've known for a while through family history. Some people receive clear explanations from doctors, others are left with more questions than answers. You might be a young adult navigating this on your own, or supporting a loved one through their diagnosis.

Whatever your story looks like, one thing is true for everyone: we all need answers, support, and tools to move forward.

That's exactly what this guidebook is here for.

It was created to help you understand, manage, and live well with Fabry disease, on your terms.



Start here ...

Inside, you'll find:



- **Practical templates to help you prepare for medical visits or request support at work/school;**
- **Tips for sharing your diagnosis with friends, family, or colleagues;**
- **Travel advice and planning tools;**
- **Career suggestions and study resources;**
- **Reference links, support networks, and trusted sources of information.**

Whether you're just getting started or looking to take the next step in your journey, this guide is here to walk alongside you.

Take what you need. Come back when you're ready.

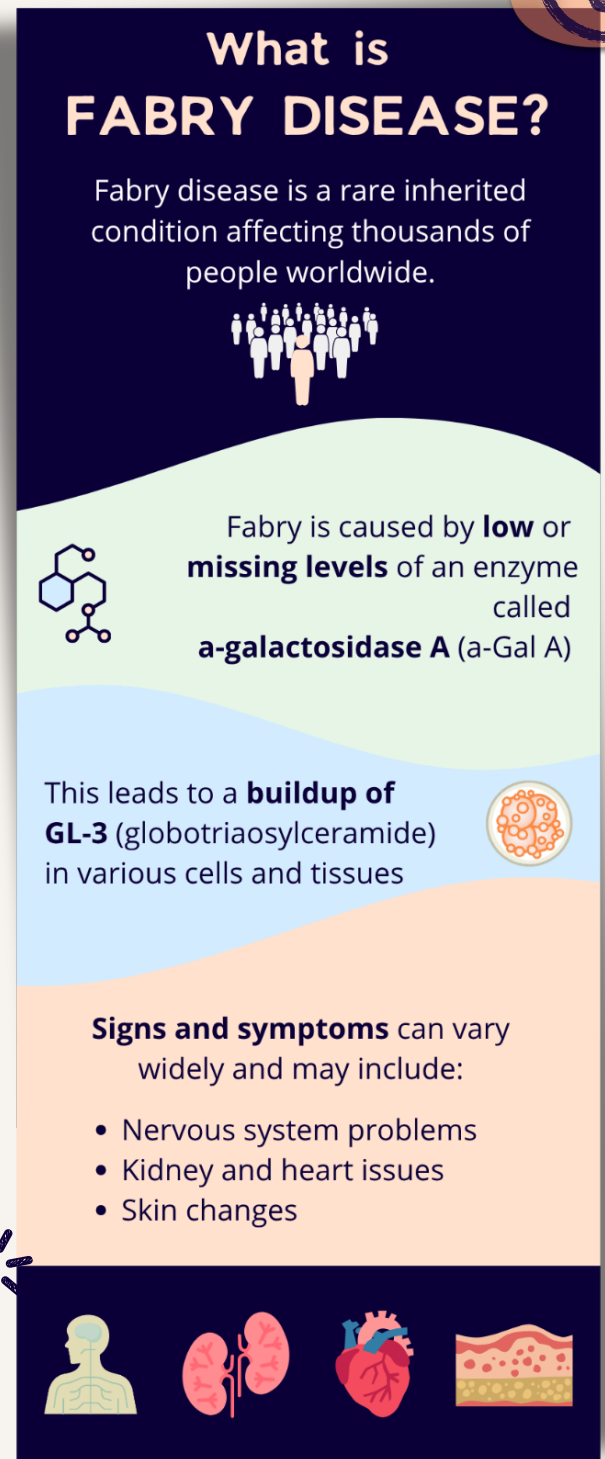
 *You're not alone.* 

1 Introduction to Fabry

1.1 What is Fabry?

Fabry disease is a rare, inherited condition affecting thousands of people worldwide. It's caused by a change in a gene that leads to low or missing levels of an enzyme called α -galactosidase A (α -Gal A). Without enough of this enzyme, the body isn't able to properly break down a type of fat called GL-3 (globotriaosylceramide).

Over time, this fat can build up in various cells and tissues, leading to damage throughout the body. Because Fabry can affect many systems, such as the kidneys, heart, nervous system, and skin, the symptoms can be very different from one person to another.

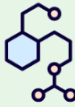


What is FABRY DISEASE?

Fabry disease is a rare inherited condition affecting thousands of people worldwide.



Fabry is caused by **low** or **missing levels** of an enzyme called **α -galactosidase A** (α -Gal A)



This leads to a **buildup of GL-3** (globotriaosylceramide) in various cells and tissues



Signs and symptoms can vary widely and may include:

- Nervous system problems
- Kidney and heart issues
- Skin changes

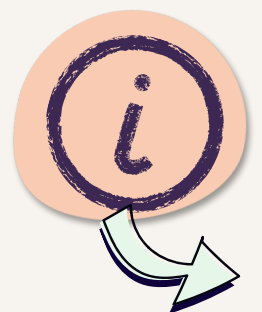


1 Introduction to Fabry

Some people begin to experience symptoms early in life, while others may not notice anything until adulthood. The way Fabry progresses can also depend on factors such as sex assigned at birth and the type (or phenotype) of Fabry someone has. There are forms often described as classical (typically more severe and earlier onset) and non-classical (usually milder or with later onset), though experiences vary widely.

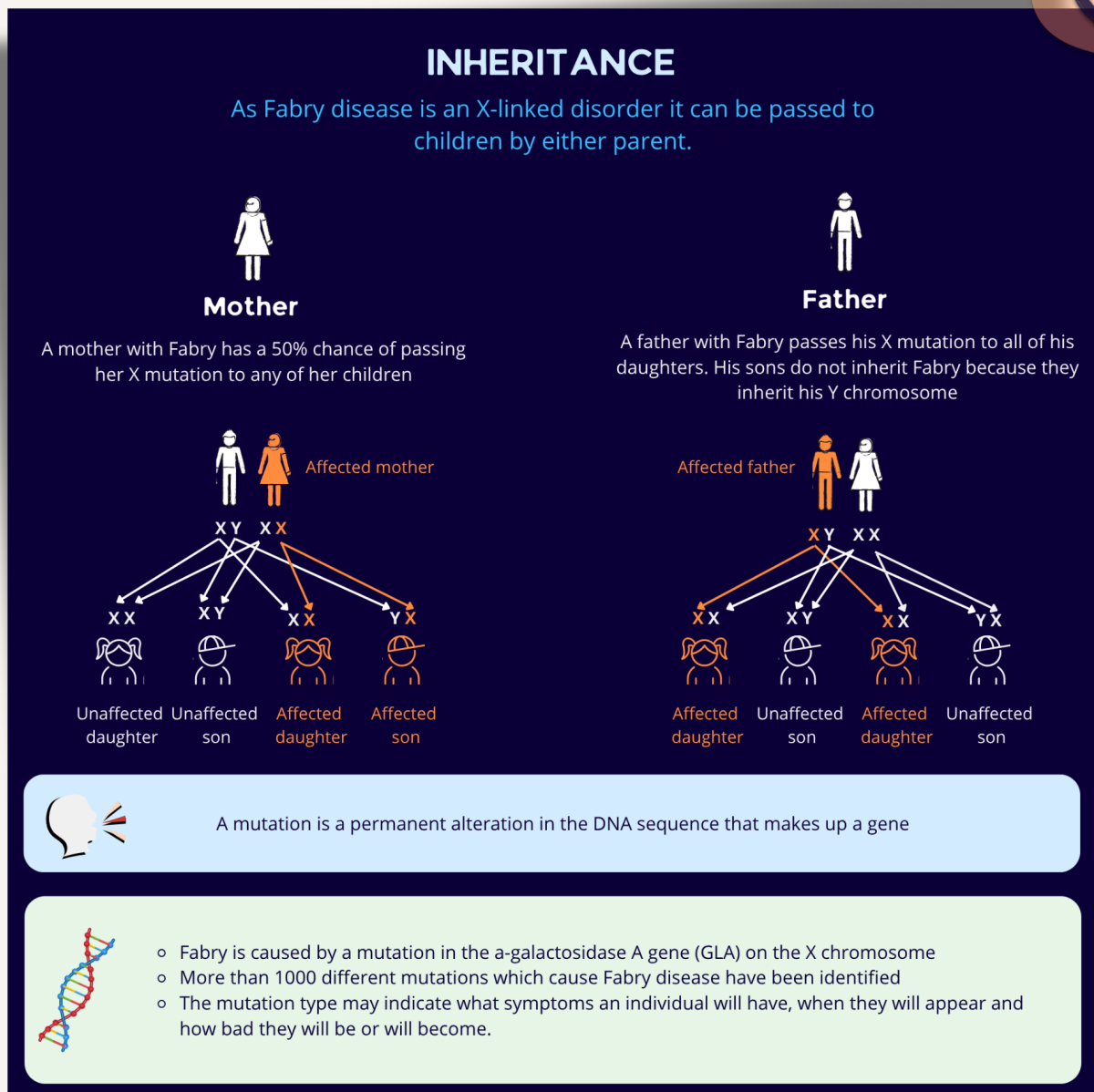


In your journey with Fabry, it's important to know that there are ways to manage symptoms and live well.



1 Introduction to Fabry

As Fabry is an X-linked condition, the way it is passed on depends on whether the mother or father carries the gene. The diagram below shows the possible inheritance patterns.



1 Introduction to Fabry

1.2 Testing and diagnosis

Getting diagnosed with Fabry disease early can really help. It allows you to start managing symptoms sooner, reduce the risk of future complications, and even find out if other family members might also be affected.

Doctors usually start by looking at your symptoms and medical history. If they suspect Fabry disease, they may refer you to a specialist centre for more testing and support. A diagnosis can be confirmed through lab tests.

Enzyme & genetic testing

One common test is called an enzyme assay, which checks how much of the alpha-GAL (α -Gal A) enzyme is present in your blood.

- In males, this test is often enough to confirm Fabry disease. A very low level of the enzyme usually means the person has the condition.
- In females, the test might not always give a clear answer. In those cases, especially if there's a family history of Fabry, a genetic test is often recommended. This test looks at your DNA to see if there are changes (mutations) in the gene that causes Fabry disease.

1 Introduction to Fabry

Prenatal testing

There are also tests available during pregnancy (called prenatal testing) for those who are already known to have Fabry in the family. These tests look at enzyme activity or DNA from fluid around the baby. If this is something you're considering, a specialist can guide and support you through the process.



See also chapter 7: Family planning.

Family history

Sometimes, looking at your family's medical history, especially if a male relative had kidney problems, heart disease, or a stroke at a young age, can help doctors figure out if Fabry might be running in your family. Filling out a simple family medical tree can be a helpful step.

Misdiagnosis

Because Fabry disease is so rare and its symptoms can look like many other conditions, it is often misdiagnosed at first. Some people are told they have:

→ Meniere's disease	→ Arthritis
→ Crohn's disease	→ Raynaud syndrome
→ Multiple sclerosis	→ Irritable bowel syndrome
→ Rheumatic fever	→ Erythromelalgia

1 Introduction to Fabry

A note about Fabry and women

For a long time, women living with Fabry were described as "carriers" a word that quietly suggested they passed the disease on without truly being affected by it. We now know this isn't true. Women with Fabry can experience the full range of symptoms, including pain, fatigue, gastrointestinal issues, kidney and heart involvement, and Fabry fog.

The lived reality, though, is that women are often diagnosed later than men. Many only learn they have Fabry after a male relative's diagnosis brings them into testing. Others find out during pregnancy, through prenatal genetic counselling, or when symptoms in their own children raise questions.

Many women also describe being told, for years, that their symptoms were anxiety, stress, hormones, IBS, or "growing pains" when in fact their pain and fatigue were already early signs of Fabry. That experience can be exhausting and isolating, and it's easy to start doubting yourself.

If this sounds familiar: your symptoms are real, even if they have been dismissed. Women living with Fabry deserve the same level of monitoring, treatment, and care as anyone else. Pushing for proper testing and for a healthcare team who takes you seriously is not being difficult. It is being a good advocate for yourself.



1 Introduction to Fabry

1.3 Treatment options for Fabry disease

1.3.1 Enzyme Replacement Therapy (ERT)

ERT provides the body with the missing alpha-galactosidase A (α -Gal A) enzyme through regular intravenous (IV) infusions, usually every two weeks. It helps clear the build-up of harmful fats (GL-3) in cells. ERT is a lifelong treatment and is suitable for most people with Fabry.

Currently, three ERT options are approved:

- Agalsidase alfa (Replagal)
- Agalsidase beta (Fabrazyme)
- Pegunigalsidase alfa (Elfabrio) – the newest formulation, approved in both the EU and U.S. in May 2023.

ERT is often started after diagnosis to manage symptoms and protect organs.

// **How often:** Every 2 weeks

Time needed: Each infusion takes about 1.5 to 4 hours, plus travel/prep time.

Where: Usually at a hospital or infusion centre, though home infusion may be available in some countries.

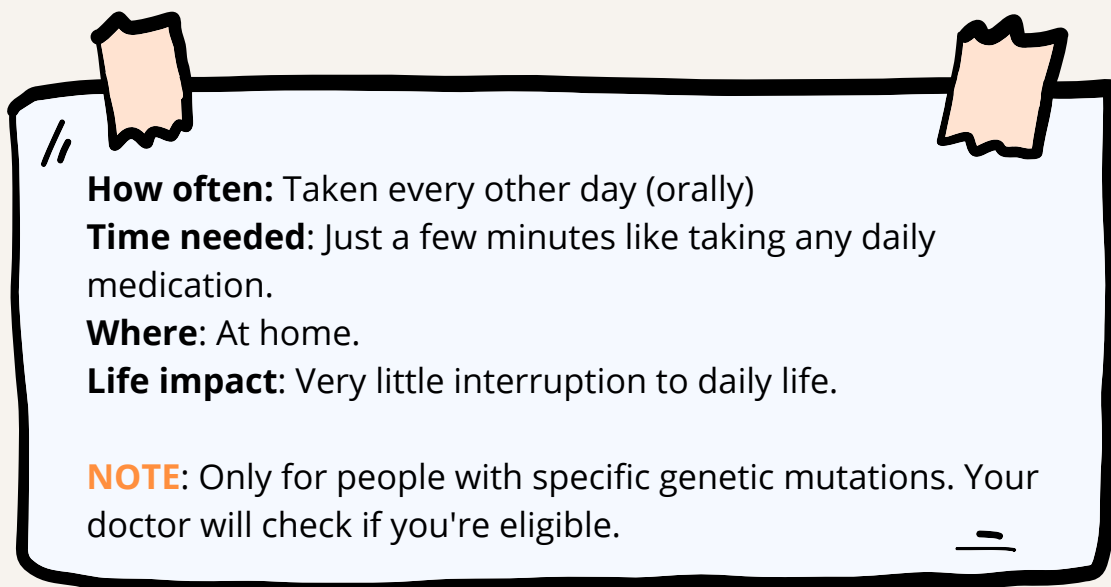
Life impact: You may need to take time off work or school for infusions, especially if travel is needed. Some people plan infusions in the evening or on their day off.

1 Introduction to Fabry

1.3.2 Oral Chaperone Therapy

Chaperone therapy is a daily capsule that helps the body's own α -Gal A enzyme fold correctly so it can work better.

- It's only effective for people with specific genetic mutations.
- Currently, the only approved chaperone therapy is migalastat (Galafold).
- It's taken orally at home, making it more convenient than infusions.
- Your doctor will do a genetic test to see if you're eligible.



//

How often: Taken every other day (orally)

Time needed: Just a few minutes like taking any daily medication.

Where: At home.

Life impact: Very little interruption to daily life.

NOTE: Only for people with specific genetic mutations. Your doctor will check if you're eligible.

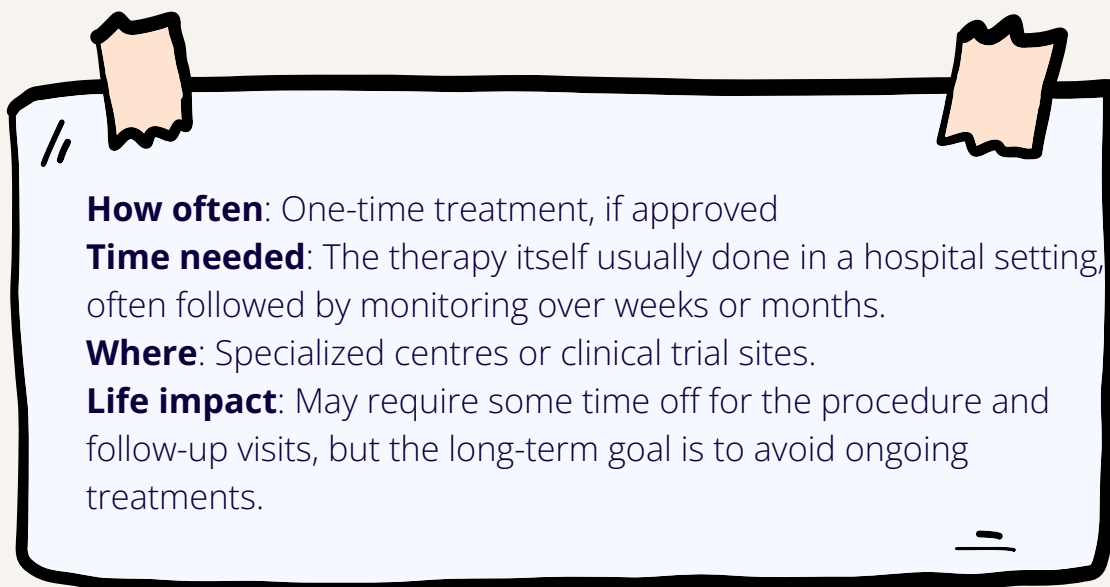
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1 Introduction to Fabry

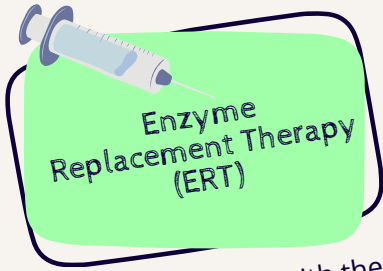
1.3.3 Gene Therapy (in development)

Gene therapy is being studied as a potential one-time treatment that addresses the root cause of Fabry disease.

- It delivers a healthy copy of the GLA gene to the body's cells.
- This new gene helps cells start producing the α -Gal A enzyme on their own.
- The goal is to offer a long-term or even permanent solution.
- Clinical trials are ongoing, and it's not yet widely available.



1 Introduction to Fabry



ERT provides the body with the missing α -Gal A enzyme through regular intravenous (IV) infusions.

- ERT helps clear the build-up of harmful fats (GL-3) in cells.
- ERT is often started after diagnosis to manage symptoms and protect organs.

How often:

Every 2 weeks, each infusion takes about 1.5 to 4 hours plus travel/prep time.

Where:

Usually at hospital or infusion centre, home treatment is available in some countries.



Gene therapy is being studied as a potential one-time treatment that addresses the root cause of Fabry disease.

How often:

One-time treatment if approved, followed by monitoring over weeks or months.

Where:

Specialized centres or clinical trial sites.



Chaperone therapy is a daily capsule that helps the body's own α -Gal A enzyme fold correctly so it can work better.

How often:

Take every other day orally, like taking any oral medication.

Where:

At home.

TREATMENT OPTIONS for Fabry disease

2 Finding out you have Fabry: what now?



"Your diagnosis doesn't change who you are"

Being diagnosed with Fabry disease can be overwhelming. For many, it comes after years of unexplained symptoms and uncertainty. And for some, the diagnosis may come unexpected or may be discovered by chance. But here's the thing, your diagnosis doesn't change who you are. You've always had Fabry; now you just have a name for it. And that name gives you power: the power to learn, manage, adapt, and take control of your health.

Some young adults have known about Fabry since childhood, but are only now beginning to understand what it actually means to live with it. If you're newly diagnosed or just starting to explore what Fabry means for you, you're not alone. Here's where to begin:

2.1 First steps after diagnosis

Being diagnosed with Fabry can come with a lot of emotions and there's no "right" way to respond. Some people want to learn everything right away, while others need time to process. Wherever you are in your journey, here are some options you can explore at your own pace, when and if you feel ready.



2 Finding out you have Fabry: what now?

Find your community

You don't have to go through this alone. Some people find it helpful to connect with others living with Fabry or other rare conditions. If or when you feel ready, you might consider:

- Exploring Fabry-specific patient groups, lysosomal storage disorder associations, or rare disease communities.
- Checking out organizations like the European Patient Forum or national Fabry associations.
- Joining online communities, these can be supportive spaces, but always cross-check medical information with trusted sources.

Learn more about Fabry

When you feel ready, you might want to understand more about your condition and what to expect:

- Patient support departments from pharmaceutical companies may provide helpful resources.
- Look for information from trusted medical sources.
- If you're preparing for a medical appointment, writing down your questions or bringing an information leaflet can help.



See also checklist page 69, 70 and 71



2 Finding out you have Fabry: what now?

Care for your mental health

- A diagnosis can bring a mix of emotions; shock, sadness, relief, confusion. You might go through stages of grief or uncertainty, and that's completely normal.
- Talking to a psychologist, counselor, or support group can be helpful. (See our list of support groups.)
- If things feel overwhelming, reaching out to someone who understands whether it's a friend, family member, or another Fabry patient can make a big difference.

Explore practical support

When you're ready, there may be support systems to help with the practical side of things:

- Some countries offer social counseling or patient navigation services.
- Financial aid, insurance options, or medical subsidies might be available, your doctor or patient advocacy group can help guide you.



2 Finding out you have Fabry: what now?

Adjusting to life with Fabry

A chronic illness can feel like an unwanted guest that never leaves. But remember:

You are not Fabry.

It's part of your life, but it doesn't define you.

Your experience is unique.

Even if Fabry runs in your family, your symptoms, challenges, and journey will be your own.

Fabry brings possibilities, not just limitations

Many patients find new perspectives, resilience, and unexpected strength along the way.

2 Finding out you have Fabry: what now?



"If today's goal is just getting through the day, that's enough."

2.2 How to cope with the diagnosis



Journaling can really help whether it's writing about your feelings, tracking symptoms, or simply noting your good and bad days. Some people prefer pen and paper, while others use apps to make it easier. Here are a few options you might explore:

- **For emotional & mental health journaling:** Daylio, Reflectly (Android & iOS – free)
- **For health & symptom tracking + journaling:** Bearable, CareClinic (Android & iOS – free)
- **For privacy-conscious users:** Penzu (encrypted journal), Day One (end-to-end encryption – free)

Choose what feels right for you, there's no one-size-fits-all approach.



Learn how your body reacts. Keep notes on what triggers symptoms certain foods, stress, temperature changes ...



Bad days happen. And that's okay. Try not to feel guilty for resting or relying on comfort food (yes, even if it's frozen pizza).



2 Finding out you have Fabry: what now?

- **Build your support system.** A support system can take many forms. It might be your family, close friends, an online community or a combination of all three. If family support isn't strong, know that meaningful support can still be found elsewhere, from people who truly understand what you're going through.
- **Denial is normal.** You don't have to accept everything at once, give yourself time.
- **Set your own goals.** If today's goal is just getting through the day, that's enough. Tomorrow might feel different.

2.3 Navigating family dynamics

Fabry is often a family disease, which can be both a source of support and frustration.

- **Family can be your biggest support but also a challenge.** Some family members may be understanding, while others may struggle to accept it.
- **It's okay to have negative feelings.** Anger, sadness, even jealousy among siblings or parents, these emotions are normal.
- **You don't have to be a 'textbook perfect Fabry patient.'** There's no right or wrong way to handle your diagnosis. Find what works for you.

2 Finding out you have Fabry: what now?

2.4 Living with a chronic condition

- **You don't need to be a hero every day.** Some days will be harder than others, listen to your body.
- **Self-pity happens and that's okay.** Be gentle with yourself; it's completely normal to feel this way at times. Just try not to stay there too long and look for small ways to move forward when you're ready.
- **Fear is real.** Fabry can feel like a "ticking time bomb," but knowledge and treatment options are evolving.
- You might encounter people who struggle to understand what you're going through, or even invalidate your experience because you don't "look sick." Having to constantly explain your condition can be exhausting. Know that **your experience is real**, even if others don't always see it.
- **It's okay to feel jealous.** Especially when people recognize diseases like cancer but not Fabry. It's frustrating, but awareness is growing.

2 Finding out you have Fabry: what now?

2.5 Moving forward

Your diagnosis is just the beginning of a new chapter. There will be challenges, but also victories. Every Fabry patient's journey is different, but one thing remains true: you're not alone. Seek support, gather knowledge, and most importantly live life on your terms.

Some next steps you can take are:

- Connect with a **Fabry support group**.
- Schedule a **follow-up** with a **specialist**.
- Start **tracking** your **symptoms**.

Remember: Fabry is just one part of your story, not the whole book. It doesn't define who you are.



You've got this



3 Living with Fabry & understanding daily life with Fabry



"Fabry doesn't define who you are, it's just one part of your journey."

Living with Fabry disease means navigating daily challenges that may affect your energy levels, work, school, social life, and overall well-being. But Fabry doesn't define who you are it's just one part of your journey. Learning how to adapt, advocate for yourself, and creating a fulfilling life is possible, and you're not alone in this.

3.1 Managing symptoms in daily life

- **Fatigue:** Many people with Fabry experience extreme tiredness, even after rest. Managing energy levels through pacing, proper nutrition, and good sleep hygiene can help.
- **Heat & cold sensitivity:** Temperature extremes can trigger pain and discomfort. Wearing appropriate clothing, using cooling towels, or avoiding extreme temperatures can help.
- **Gastrointestinal issues:** Nausea, diarrhea, or bloating are common. A food diary can help track triggers, and working with a dietitian may provide relief.
- **Skin & sweat regulation:** Some people with Fabry sweat less or not at all. Staying hydrated and avoiding overheating is key.

3 Living with Fabry & understanding daily life with Fabry

3.2 Talking about Fabry: how to share your diagnosis with people in your life

At some point, you'll probably need to talk to someone about your diagnosis whether it's a friend, partner, family member, teacher, boss, or colleague. These conversations can feel awkward or even scary at first. And it's good to keep in mind: people are different. Some will react with kindness and curiosity. Others might be confused, unsure, or even say the wrong thing. That's totally normal and not a reflection of you.

What you say and how you say it can really help set the tone. Being clear, calm, and real (without diving into too much medical detail) can make a big difference. And if you feel nervous, that's okay too. This is personal stuff. You're allowed to go at your own pace.

3.2.1 Telling a friend

Should you tell your friend? That's up to you. If it's someone close, someone you trust, opening up can actually bring you closer. It helps them understand why you might be tired, need flexibility, or just have days where you're not your usual self. You don't have to share every detail. Just what feels right to you.



3 Living with Fabry & understanding daily life with Fabry

5 simple **tips** when talking to a friend:



1. Pick the **right moment**. Somewhere quiet where you won't be interrupted is best.
2. Keep it **simple**; use your own words, skip the medical terms. Say what it is and how it affects you.
3. Expect **all kinds of reactions**; some people need time. That's not a reflection on you.
4. Be **clear about what you might need**; let them know if you might cancel plans sometimes or need rest.
5. **Reassure** them; you've got a plan, you're managing your health, and you just want them to know what's up.

3 Living with Fabry & understanding daily life with Fabry

Here's something you could say:

Hey [Name], I've been meaning to tell you something. I have a condition called Fabry disease. It's genetic and affects different parts of my body like my nerves, kidneys, and sometimes my energy. Some days I feel totally fine, and other days I might be dealing with pain or fatigue. I'm getting treatment and managing it, but I wanted you to know because you're important to me, and I think it helps when people understand. If you ever have questions, I'm happy to chat about it.



3 Living with Fabry & understanding daily life with Fabry

3.2.2 Sharing with a partner

Opening up to a partner about Fabry disease can feel vulnerable but it's also a chance to build deeper trust. Whether you've been together a long time or the relationship is just starting, honesty about your condition can help avoid misunderstandings and bring you closer.

That doesn't mean you need to spill everything all at once. Start with what feels right. Think of it as an ongoing conversation, not a one-time confession.



Why it's worth talking about

- It gives your partner context: if you're feeling low-energy, in pain, or need to cancel plans, they'll understand it's not personal.
- It allows them to support you: most people want to show up for their partner, but they need to know how.
- It reduces pressure on you to pretend you're fine when you're not.



When and how to have the conversation

- Choose a moment where you both have time and space, somewhere quiet and low-stress, like a walk or relaxed dinner at home.
- Be honest, but keep it real, share how Fabry shows up in your life, not just what it says in a brochure.
- Let them ask questions, even if they don't know what to say right away.
- Reassure them where you can, you're managing your health and you're not expecting them to "fix" anything.

3 Living with Fabry & understanding daily life with Fabry



Things you might want to share

- What Fabry is and how it affects you.
- That you're under medical care.
- Any symptoms that might impact daily life or future plans (like fatigue, pain, needing flexibility).
- That you're sharing this because you care and want to be open, not because you expect a certain reaction.

Example conversation starter:

There's something personal I've been meaning to talk to you about. I have a rare genetic condition called Fabry disease. It affects things like my energy, pain levels, and sometimes my organs, but I'm managing it with medical support and I'm doing okay.

I wanted to tell you because you're important to me, and I think it's better to be open about these things. It might mean I need to rest more sometimes, or be flexible with plans. But I'm still me, and I don't want this to feel like a big scary thing.

If you ever want to know more or have questions, I'm happy to talk about it or take it one step at a time.

3 Living with Fabry & understanding daily life with Fabry



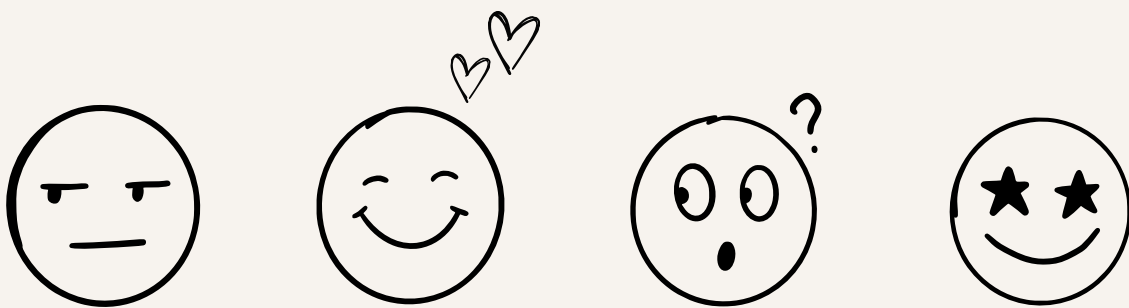
How a supportive partner might react

- They ask thoughtful questions.
- They say thank you for trusting them.
- They want to understand more so they can support you better.

And sometimes... they just listen quietly. That's okay too. Let it breathe. What matters most is that you were honest, and you opened the door for deeper connection.

3.2.3 How people might react, and what that means

When you share your diagnosis, people might not always react the way you expect. Some will get it right away. Others might struggle or overdo it. Here are a few common types of responses.

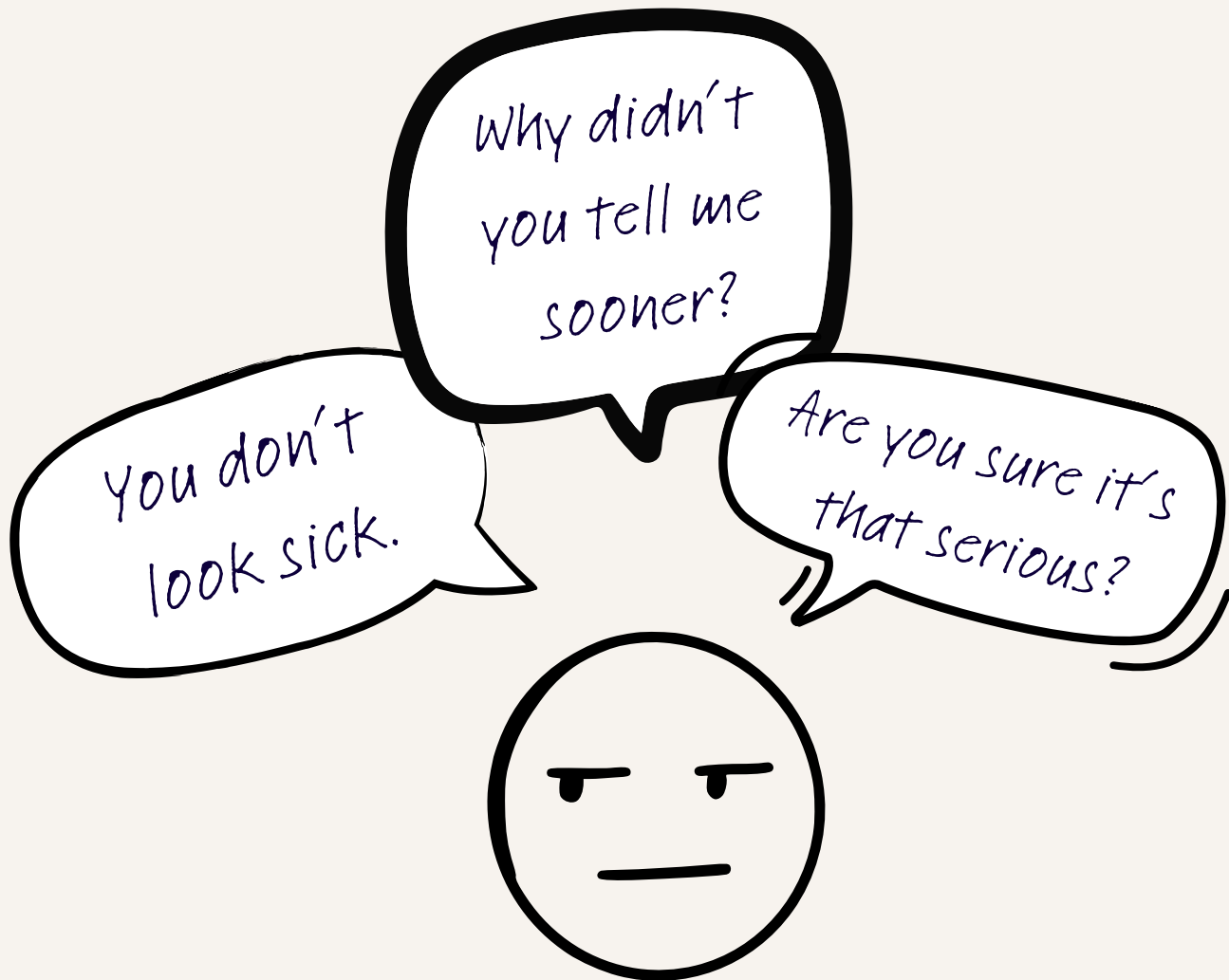


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The complicated one

Not everyone knows how to respond to something like this. Some might deny it, ask weird questions, or minimize what you're going through. It's tough but also shows you who's ready to show up for you and who might not be.

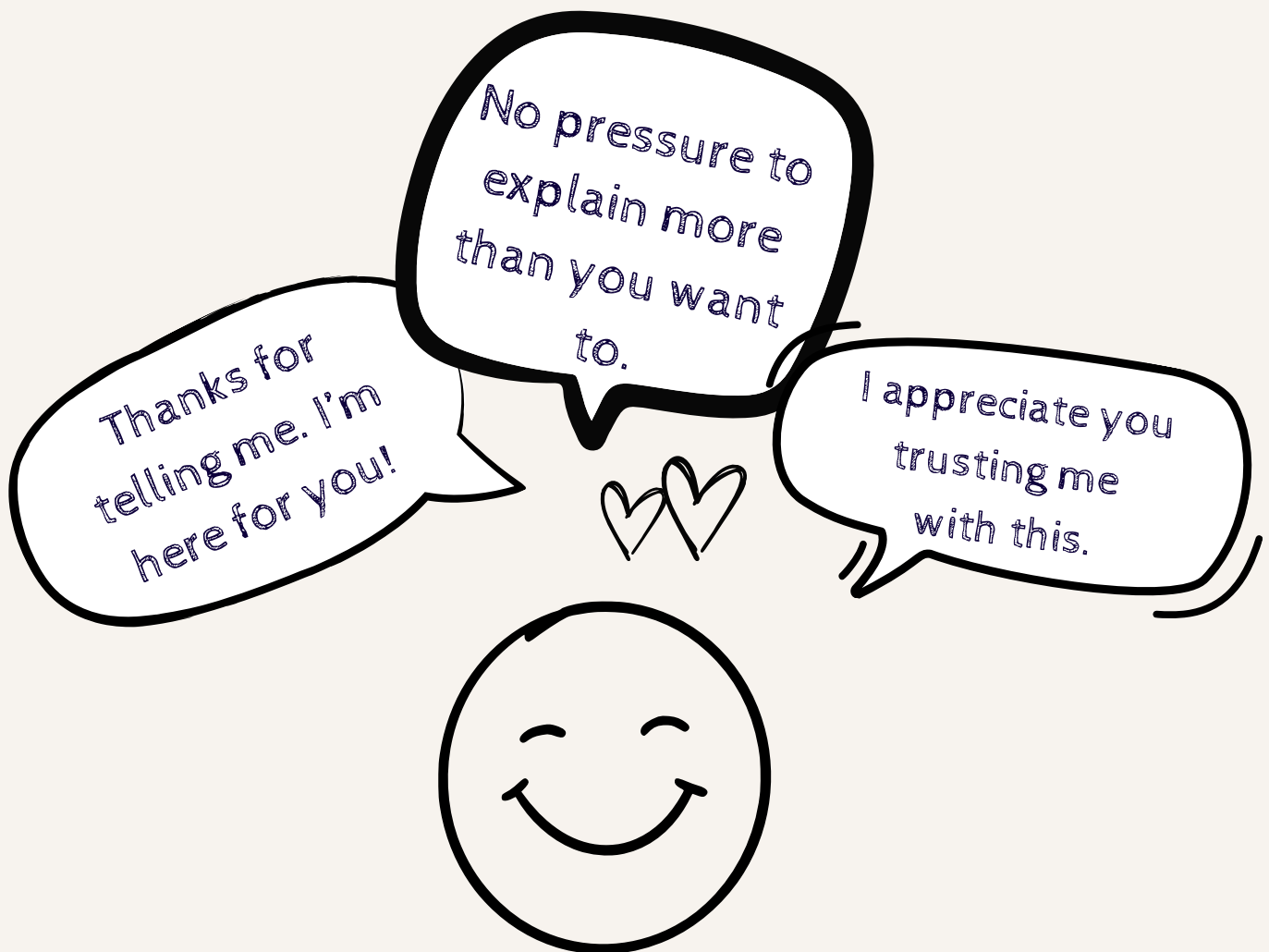


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The supportive one

These are the gems. They respect your boundaries, ask how they can help, and don't make it all about them.



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The unsure one

Some people just need time to understand what Fabry is. They might not say much at first but could turn out to be great supporters once they learn more.



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The over-helper

Their heart is usually in the right place, but they go into hypercare mode or offering too much help, excluding you from things to "protect" you, or treating you differently.



It's okay to gently set boundaries with people in this group.

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3.3 Dating & social life

Talking about Fabry in relationships

- **When to share:** It's up to you! Some prefer to share early, while others wait until they trust the person.
- **How to explain it:** Keep it simple and avoid overwhelming details at first. You can always share more later, as the relationship deepens. Remember how you felt when you were diagnosed. While it's not exactly the same, the people you share this with may also need time to adjust how they perceive you, just as you did. Be gentle with them, and most importantly, with yourself. These conversations can be emotionally tough for everyone involved.
- **Managing reactions:** not everyone will understand immediately... and some may never fully understand. If that happens, please remember: it's not because of anything you did wrong. You shared something personal and important, and if someone can't accept or process that, it's a reflection of their limitations, not yours. One of the unexpected truths of living with Fabry is that it often reveals who's truly there for you. Be patient with others when you can, and give space for their questions but be kind to yourself, too.
- **Discussing family planning:** If a relationship gets serious, it may be worth discussing the genetic aspect.



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Socializing & friendships

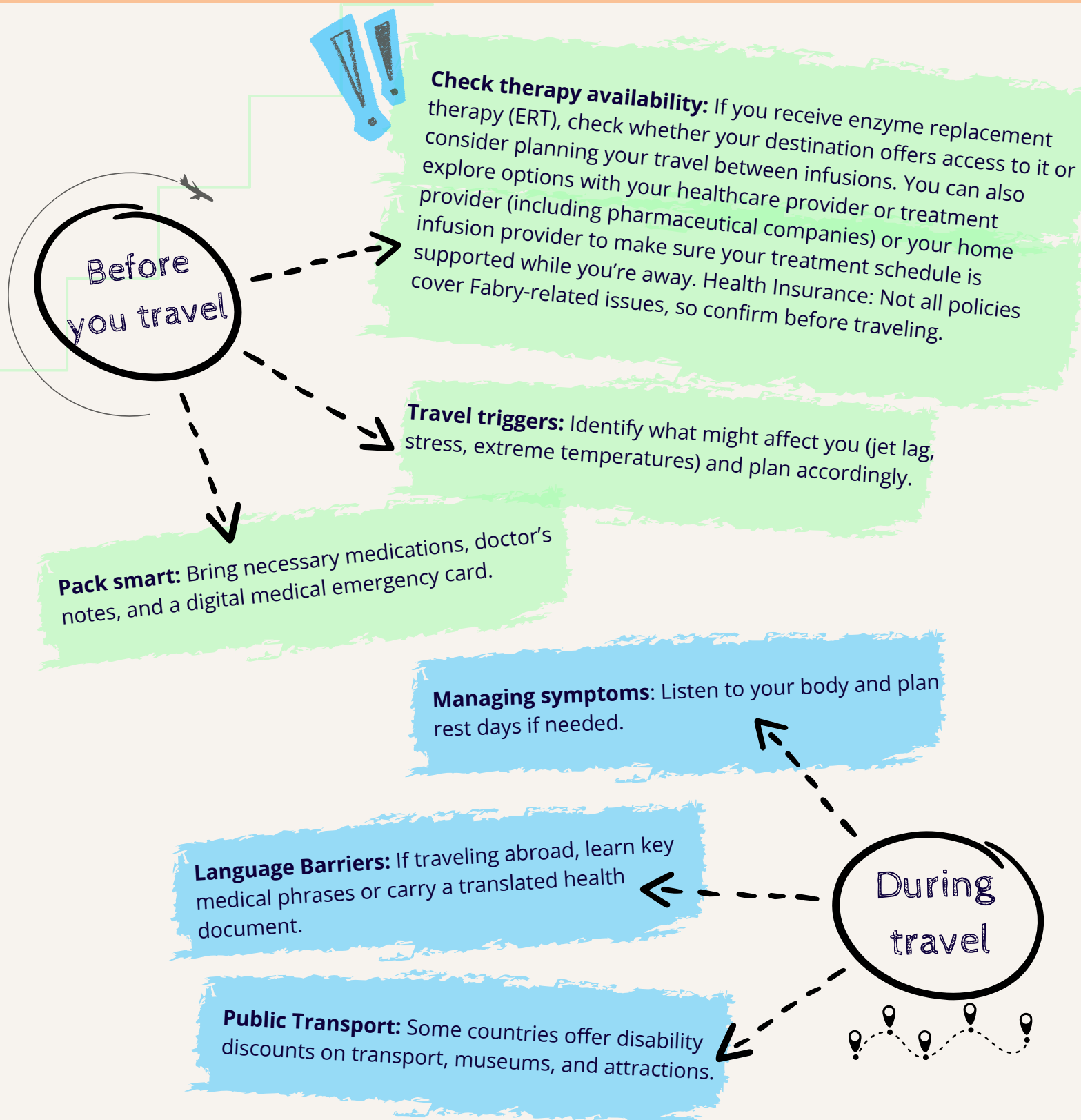
- **Setting boundaries:** Let people know if certain activities (hiking, sauna, extreme sports) might be difficult for you.
- **Traveling with Fabry:** Make sure at least one friend or coworker knows about your condition in case of emergencies.
- **Handling misunderstandings:** People might say things like "*but you don't look sick!*" Having a prepared response can help, e.g., "*Fabry is an invisible illness, some days are better than others.*"

Traveling with Fabry

Fabry shouldn't stop you from exploring the world! However, some planning is necessary.



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3.4 Navigating school & work with Fabry

Balancing education and career while managing symptoms can feel overwhelming, but accommodations can make things easier.

School & University

- **Know your rights:** Many countries have disability protections that allow for accommodations in education.
- **Request adjustments:** You may be eligible for accommodations like extra time for assignments, flexible attendance policies, or remote learning options. These can make a big difference in managing both your education and your health. Start by checking if your school or university has an office or department for student support, this may be called “Services for Inclusion,” “Disability Services,” or “Student Accessibility Services.” They can help coordinate accommodations and provide official documentation for your professors or faculty administration.
- **Communicate with professors:** If needed, inform your professors or school administrators about your condition. A calm, clear conversation (or using a template) can help.
- **Bullying & stigma:** If you experience discrimination, reach out to a trusted teacher, counselor, or student services.
- **Taking your time:** There’s no shame in taking a gap year or extending your studies. Everyone’s journey is different.

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Work & Career

- **Disclosing Fabry:** You don't have to tell your employer unless you want to, but if you need accommodations, a clear and professional explanation can help.
- **Workplace adjustments:** Flexible schedules, remote work, or extra breaks can help manage symptoms.
- **Legal Protections:** Some countries offer disability status, which can provide benefits, protections, or employer incentives.
- **Choosing the right career:** You know yourself better than anyone else. It's important to recognize your own limits and what you feel capable of doing. While flexibility and less physically demanding roles can be beneficial for many people living with Fabry, the choice is entirely yours. Your path doesn't have to look like anyone else's, what matters most is that it works for you.
- **Workers' Unions & Rights:** Unions can provide legal guidance and workplace support.



3 Living with Fabry & understanding daily life with Fabry

The hidden bonus: scholarships & opportunities for students with disabilities or rare conditions



So ... how does it work?

These scholarships and grants usually come from:

- Governments
- Universities
- Non-profit organizations
- Private companies or foundations



Every program is a little different, but common requirements might include:

- Proof of a disability or rare disease (like a doctor's letter)
- Enrollment in a recognized school or university
- Decent grades (some have minimum GPA requirements)
- Financial need (for certain programs)
- Your field of study (especially if it's health-related)

Most applications ask for a few things like:

- An application form
- Proof of diagnosis
- Academic transcripts
- Maybe a personal statement or a letter of recommendation

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Where can you find these opportunities?

Here are a few great places to start looking:

- **Your university's financial aid office**

They'll often know about internal grants or partnerships with organizations that support students with disabilities.

- **Government websites**

National education ministries or student support platforms usually list options. In Italy, for example, check with the Ministry of Education, Universities and Research.

- **Rare disease & disability organizations**

Look at groups focused on rare diseases or chronic illness. Many collaborate with schools or offer their own scholarship programs.

- **Online scholarship search tools**

Use platforms like Scholarship.com or Fastweb, and filter by "disability" or "medical condition."

- **Local support groups**

Sometimes smaller or regional orgs have their own scholarships or know about ones that aren't widely advertised.

- **Education fairs & workshops**

Disability-focused education events often include booths or talks about scholarships and how to apply for them.

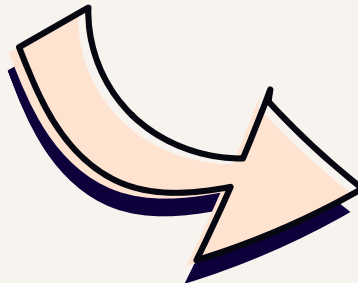
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TIP

Always double-check the eligibility criteria and deadlines.
And if you're not sure, don't hesitate to email or call the organization, they're usually happy to help.



Also check the success story from Wojciech Nadolski on page 57, 58 and 59



3 Living with Fabry & understanding daily life with Fabry

3.5 Communicating at school & work

3.5.1 Short explanation template

It's not always easy to talk about your health, but having a short, clear way to explain Fabry can make things easier especially with professors, colleagues, or supervisors. Here are some examples and templates you can adapt to your style and situation.

"I have a rare genetic condition called Fabry disease, which can sometimes affect my energy levels and cause pain episodes. I manage it well, but I may occasionally need flexibility with deadlines or work hours. I appreciate your understanding."

template

You don't owe anyone every detail but sharing just enough can open the door to support and accommodations when you need them.

If Fabry sometimes affects your work, like needing breaks, having low-energy days, or missing time for medical appointments, it can be helpful to let a colleague know. Especially if they're someone you work closely with and trust.

You don't need to go into detail. Just enough so they understand where you're coming from if something's off.

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Tips for sharing at work:



- Keep it professional and focused on how it might impact work (not a full health history).
- Choose a private moment, never in a group setting.
- Let them know you're still committed and on top of things.
- Only share what you're comfortable with.



3 Living with Fabry & understanding daily life with Fabry

3.5.2 E-mail templates

Informing school or university

Being a student with Fabry can be challenging especially with symptoms like pain, fatigue, or brain fog. It can affect how you focus, get to class, meet deadlines, or join group projects.

You're allowed to ask for support and flexibility. Most schools and universities have disability services or support departments you can contact. You don't have to face this alone.



// Things you might ask for:

- extended deadlines;
- excused absences;
- recorded lectures;
- exam rescheduling;
- flexible class participation.

—



// Who to contact:

- disability office or student support services;
- your professors or academic advisor;
- the dean's office, if needed.

—



3 Living with Fabry & understanding daily life with Fabry

E-mail example you could use:



Subject: Request for academic accommodations



template

Dear [Dean's Name],

I hope you're doing well. I'm writing to let you know that I live with a medical condition called Fabry disease. It sometimes causes fatigue, pain, and other symptoms that can affect my ability to attend classes, meet deadlines, or perform at my best academically.

I remain committed to my studies but may occasionally need flexibility due to medical appointments or symptom flare-ups.

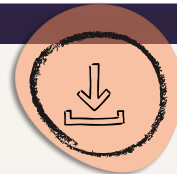
I would really appreciate the opportunity to discuss potential accommodations like extended deadlines or flexible attendance if needed. I'm also happy to provide documentation if required.

Thank you for your time and support.

Best regards,

[Your Name]

[Student ID]



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3 Living with Fabry & understanding daily life with Fabry

Informing a colleague

Example message you can adapt:



Subject: Quick note about my health



template

Hi [Name],

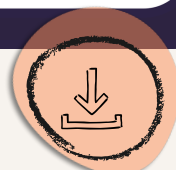
I wanted to share something personal with you so you understand where I'm coming from if I ever seem a bit off.

I have a condition called Fabry disease. It can cause fatigue or sometimes make it harder to keep up with everything at full speed. I'm managing it with treatment, but now and then, I might need a bit of flexibility or understanding.

Just wanted you to know, thanks for your support, and feel free to ask if you're ever curious.

Best,

[Your Name]



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Informing a manager

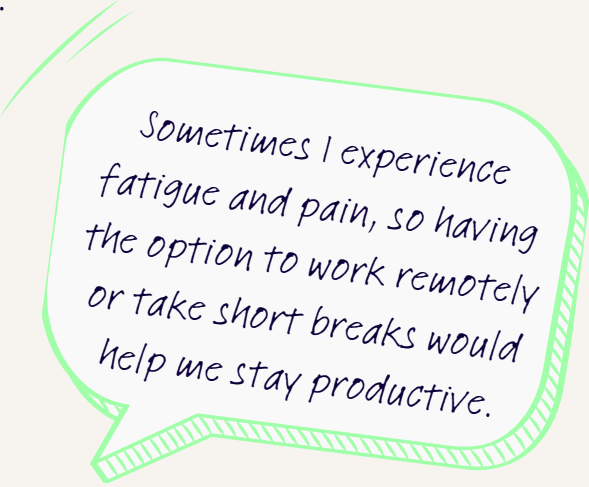
Navigating Fabry disease at work can be tricky, especially when it affects your energy, concentration, or flexibility. Whether or not you have a recognized disability, you still have the right to ask for support or accommodations that help you do your job well.

Here are two main scenarios, depending on your situation:

Option 1: You don't have official disability status, but need accommodations

If you haven't been formally recognized as having a disability, you don't have to share your diagnosis. You can just focus on how your symptoms affect your work and what might help you do your job better.

A practical way to ask:



Sometimes I experience fatigue and pain, so having the option to work remotely or take short breaks would help me stay productive.

You're not hiding anything, you're just leading with what you need, rather than a full explanation of your health.



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E-mail example (no formal disability status)



template

Subject: Request for workplace flexibility



Dear [Manager's Name],

I hope you're doing well. I wanted to talk about something that could help me maintain my productivity and well-being at work. I occasionally experience symptoms such as fatigue and pain, which can impact my energy levels during the day.

To help manage this while staying on top of my work, I'd like to explore the possibility of small adjustments such as the option to work from home occasionally or take short breaks when needed.

I'd be happy to discuss this in more detail and find a solution that works for both me and the team. Let me know when would be a good time to talk.

Thanks for your time and understanding.

Best regards,

[Your Name]



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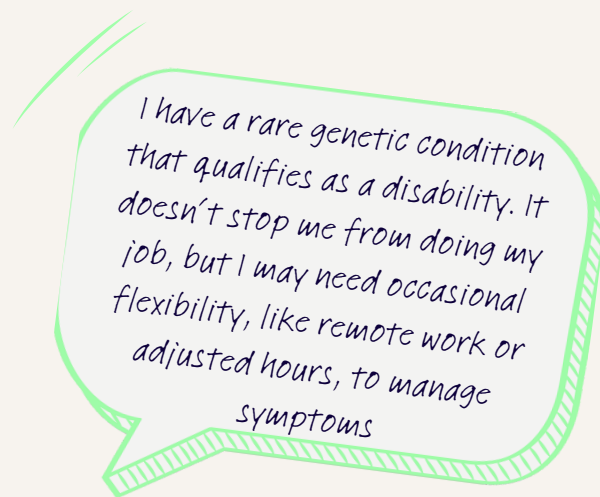
3 Living with Fabry & understanding daily life with Fabry

Option 2: You do have a recognized disability

In some countries, Fabry qualifies for disability status, this means you may have legal rights to ask for accommodations, like flexible hours or remote work.

If you're in a setting that supports disability inclusion, being open about your diagnosis might give you access to formal resources or extra support.

What you might say:



It's still up to you how much to share but knowing your rights and what's available can make a big difference.



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E-mail example (with recognized disability status)



template

Subject: Request for reasonable accommodations



Dear [Manager's Name],

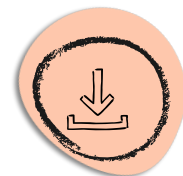
I hope you're well. I wanted to reach out to discuss a small adjustment request related to my health. I have a rare genetic condition that qualifies as a disability. While it doesn't affect my ability to perform in my role, I do sometimes experience symptoms such as fatigue and chronic pain.

To help me stay productive and maintain a good workflow, I'd like to request accommodations such as [e.g., flexible hours, occasional remote work, short breaks].

I'd really appreciate the chance to talk this over with you and find a way that works for everyone. Let me know when would be a good time to meet.

*Thank you for your understanding and support.
Best regards,*

[Your Name]



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Keep in mind that it depends on the company culture, legal protections in the country, and how much the individual wants to disclose. Many people choose to focus on their needs rather than the diagnosis itself unless formal accommodations are required.



3 Living with Fabry & understanding daily life with Fabry



Living with Fabry means adapting, but it also means finding strength in resilience. You're not alone, there are communities, resources, and strategies to help you live fully and confidently.



Wojciech's story: Turning Fabry into an opportunity



SUCCESS!!



Talk to Wojciech about his student years and you'll notice a kind of quiet pride, not because everything was easy, but because he built a path that once felt completely out of reach.

He grew up in Poland, and now holds two degrees: a bachelor's in Branding, Marketing & Public Relations, and a master's in International Entrepreneurship from Coventry University in the UK.

Most of the master's was online, but he still remembers stepping onto campus for graduation, finally meeting in person the classmates he had only seen on screen.

"It felt like closing a chapter," he says. "A big one."

What might surprise you is how much Fabry disease shaped that path, not as the obstacle most people might assume, but as something that quietly opened doors Wojciech hadn't known existed.

When Wojciech got his disability card at 15, a woman on the board told him something he didn't fully understand then: "Plan to go to university. This card will make it easier."

Years later, when he began preparing for university applications, he remembered her words and discovered she had been right.

Wojciech's story: Turning Fabry into an opportunity



Because of his disability status, doors opened that he hadn't even known existed. Universities offered extra admission points, scholarships, reduced tuition fees, and even the option for a single dorm room.

On top of that, Poland has government programs that help students with disabilities pay for private universities programs that can make the impossible feel suddenly, surprisingly possible.

But accessing these opportunities wasn't always simple. When Wojciech tried to navigate government websites, he found a lot of outdated or confusing information.

The turning point came when he reached his university's Support Centre, a place he hadn't known existed. "They explained everything," he says. "Like, step by step. Suddenly it all made sense."

The application process itself was mixed. The university scholarship was quick and easy, just a short form and his disability certificate. The government application, though... that was another story.

"It was like a mini-puzzle," he laughs. "So many documents. I messed it up the first time."

But instead of rejecting him, the government office sent the application back with line-by-line instructions.

Wojciech's story: Turning Fabry into an opportunity



And with each semester, it became easier. Eventually, it covered 80% of his tuition and even provided extra funding for books and living costs.

This support didn't just help financially; it changed what Wojciech believed was possible for him.

It allowed him to choose a private university he genuinely wanted. It made studying abroad, something he once viewed as just a dream, a real, tangible experience. It built his confidence, step by step.

When asked what he would tell his younger self, he doesn't hesitate:

“Don't be scared. There's more support out there than you think.”

He would tell him that Fabry, while challenging, can also unlock opportunities. That complicated paperwork shouldn't stop him. And that there are people, entire teams even, dedicated to helping him succeed.

Today, looking back, Wojciech knows that the support he received didn't just fund his education. It shaped the life he's building now.

“It gave me a chance,” he says. “A real one.”



Wojciech is a proud member of the **FIN Young adults Community** and a **board member** of the **Polish Fabry Organisation**.



3 Living with Fabry & understanding daily life with Fabry

Quick Q & A

Funding & support as a student living with a rare condition



What kinds of support are available?

- University scholarships for students with disabilities;
- Government funding for tuition (private universities included);
- Grants for living expenses, books, and sometimes travel;
- Academic accommodations (extra exam time, flexible deadlines, etc.);
- In some countries: extra admission points when applying to university.

Where do I start looking?

- The disability support office or student support centre at the university;
- University admissions office (ask before you even apply!);
- Government websites (even if messy, the information is usually there)
- Foundations or charities supporting rare-disease students, especially for study abroad.

What documents are usually needed?

- Disability certificate;
- Confirmation of current studies;
- Proof you completed the last semester;
- Tuition invoices;
- Sometimes: confirmation from your employer (if you work).

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Quick Q & A

Funding & support as a student living with a rare condition



Is the application difficult?

It can be, especially government forms, but staff will help you, and mistakes don't mean rejection. Many students learn the process step by step.

Does financial support make a big difference?

Absolutely. It can:

- Reduce or cover tuition;
- Make living away from home possible;
- Help fund study abroad;
- Make specialized programs affordable;
- Reduce stress so you can focus on studying.



Top tips from Wojciech

- **Be proactive**, ask universities early about support;
- **Don't rely only on government aid**, university scholarships might be even better;
- **Look for accommodations, not just money**;
- **Don't get discouraged by paperwork**, the first application is the hardest;
- **DREAM BIG**, support can open doors you don't expect.

3 Living with Fabry & understanding daily life with Fabry

3.6 Choosing a career with Fabry: a guide, not a limit

Living with Fabry disease can shape parts of your life but it doesn't define who you are or what you're capable of.



You still get to dream big, follow your passions, and choose a career that feels right for you.

The suggestions below aren't about limiting your options. They're here to help you think through what might work best for your body, your goals, and your lifestyle. You can absolutely go after any path that excites you. These ideas are just meant to help you navigate things with a bit more comfort and ease when symptoms show up.

3.6.1 Things to consider when planning your career

As you think about what kind of work fits best for you, here are a few helpful factors to keep in mind:

- **Remote or hybrid options** – Jobs that let you work from home (even some of the time) can be a game-changer.
- **Flexible hours** – Being able to schedule breaks or shift your hours can help manage energy levels.
- **Low physical demands** – Avoiding super physical roles can reduce stress on your body.



3 Living with Fabry & understanding daily life with Fabry

- **Comfortable environments** – Office settings with heating/AC can be better if you're sensitive to temperature.
- **Good benefits** – Access to quality health insurance and support programs is a major plus.

Some roles might be more physically demanding

Jobs that involve a lot of physical activity, exposure to extreme temperatures, or strict schedules might be harder to manage with Fabry symptoms. That doesn't mean they're off-limits, but they may need some creative planning, adaptations, or backup support. If you love these kinds of jobs, go for it, just look into how you can make them work for you.

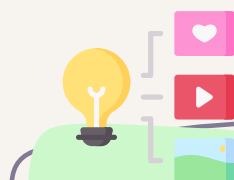


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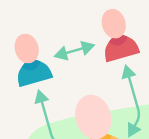
Tech & IT

From coding to data analysis, tech roles are often remote-friendly and deadline-based. As long as you've got internet, you're good to go.



Writing & Content Creation

Writing, editing, social media management, creative content roles let you express yourself, often on your own schedule and from wherever you feel best.



Customer Support & Client Services

Support roles are often done from home or in hybrid setups, especially when you're answering questions, solving problems, or helping people online or over the phone.



Business & Project Management

Organizing workflows, managing teams, or analyzing business processes? These jobs usually offer flexibility and can be done partly or fully from home

Careers WITH GREAT FLEXIBILITY (and remote options)



Finance & Accounting

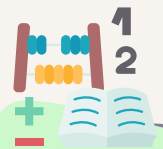
Many finance-related jobs are office- or remote-based, involve focused tasks, and allow for quiet, structured work.

3 Living with Fabry & understanding daily life with Fabry



Creative & Design Fields

Graphic design, video editing, UX/UI work, these fields tend to blend independent work with collaboration, giving you a nice balance of structure and freedom.



Education & Training

Teaching online, tutoring, or developing e-learning materials are all ways to work in education while keeping flexibility in your day.



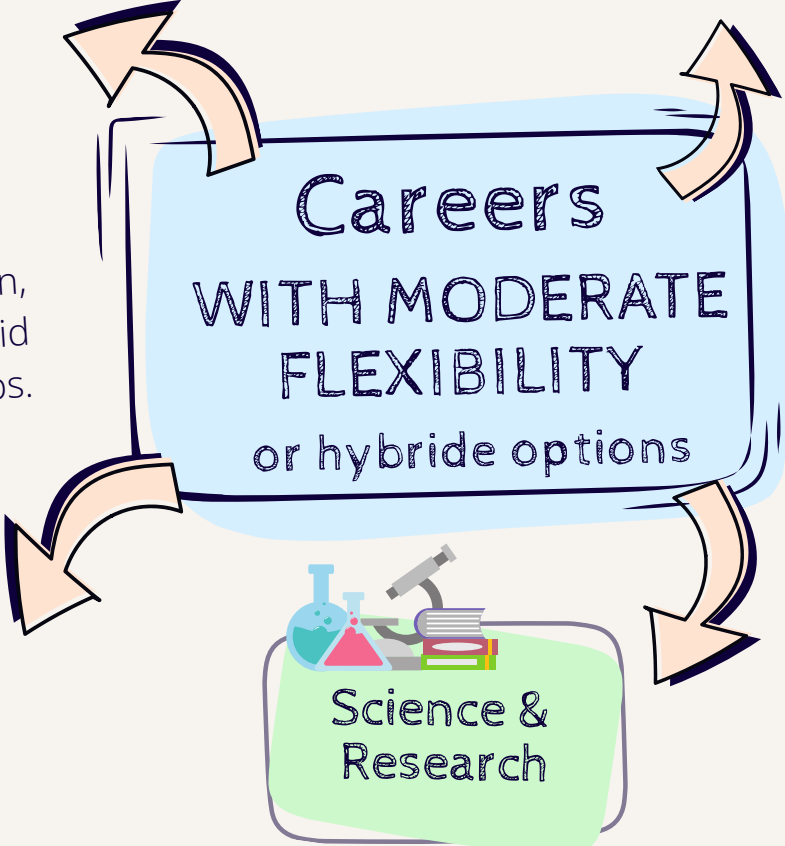
Engineering & Technical Fields

While some engineering jobs are hands-on, others, like design or consulting, offer hybrid options and flexible collaboration setups.



Legal & Consulting

Many legal and consulting jobs can be tailored with remote days and flexible hours depending on the client or casework.



Careers WITH MODERATE FLEXIBILITY or hybride options



Science & Research

Not all lab jobs are rigid! Research roles in data analysis, grant writing, or academic settings often allow for flexible work structures.

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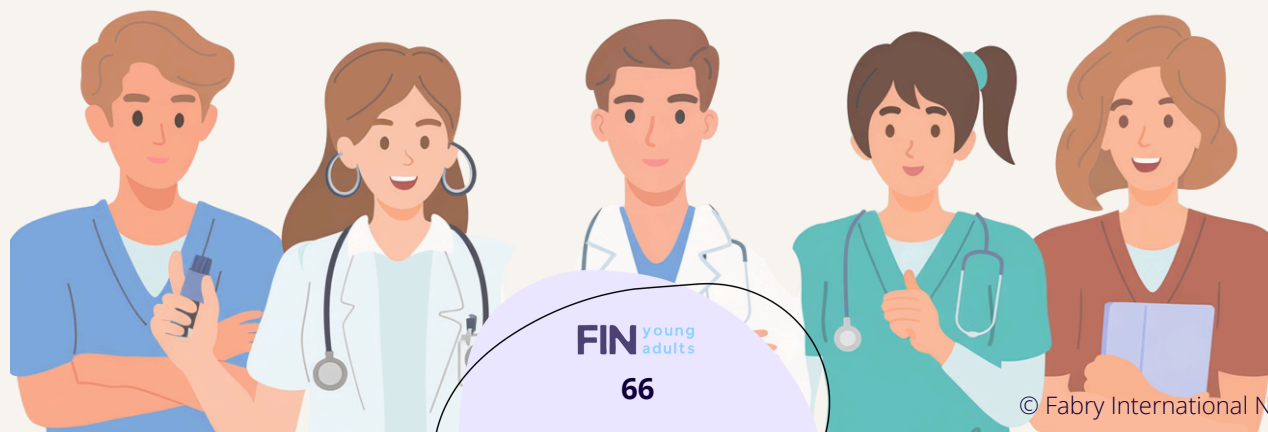
For many people with Fabry, choosing a healthcare career feels deeply personal. It's about using your lived experience to understand your own health better and support others. Whether you become a nurse, doctor, psychologist, researcher, or therapist your insight is powerful.

What makes you unique

You bring something incredibly valuable to the table: empathy. You get what it's like to navigate a diagnosis. That makes your voice important in shaping better care, raising awareness, and pushing for more patient-centered approaches.

The rewards

Helping others while growing your own knowledge can be incredibly fulfilling. Many people with rare diseases find strength and meaning in supporting others on similar paths. You're not just working a job, you're building something with heart.



3 Living with Fabry & understanding daily life with Fabry



If you feel drawn to helping people emotionally, careers like psychology, counseling, or mental health coaching can be a great fit. Your experience gives you a deeper understanding of resilience, grief, fear, and hope and that kind of insight is rare and deeply needed.

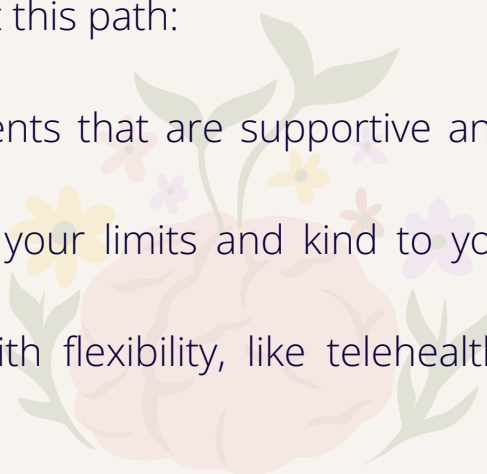
Real Talk: Challenges in healthcare careers



Working in healthcare can be demanding, physically and emotionally. Long shifts, high stress, and unpredictable days can be tough, especially when managing your own health.

If you're thinking about this path:

- Choose environments that are supportive and allow space for self-care.
- Be realistic about your limits and kind to yourself when you need rest.
- Look into roles with flexibility, like telehealth, admin, research, or coaching.



3 Living with Fabry & understanding daily life with Fabry



A message for the changemakers

To anyone with Fabry who chooses to step into healthcare, support, education, or any other purpose-driven career: you are bold, resilient, and inspiring.

You're not just managing your own journey you're using it to make things better for others. Whether you're advocating for patient rights, developing better systems, or simply showing up every day with empathy, you're helping shape a more understanding world.



**You don't have to do it all.
But what you do matters.**



4 Fabry & you: your doctor visit

4.1 Your doctor visit checklist

A friendly guide to help you get the most out of your medical appointments.

Whether you're newly diagnosed or living with Fabry for a while, it's not always easy to know what to ask at the doctor's office. This checklist is here to help you feel more prepared, confident, and in control of your care.

This is an indicative list of the questions that you may get inspired by, you can tailor it for yourself, since doctors might have a 180-degree different approach in different countries.

Use it before each visit to make sure all your questions and concerns are covered, and don't be afraid to add your own!

Tip:

Click on the checklists on the following pages to download them. You'll receive a text file that you can edit, add to, and so on.



4 Fabry & you: your doctor visit

CHECKLIST doctor's visit



A. Understanding what's going on with your body

- ☐ Can you explain where I currently stand with Fabry?
- ☐ Have there been any changes in my test results?
- ☐ Are there new symptoms or complications I should know about?
- ☐ How might my condition change over time, and what should I watch for?
- ☐ Are there things I'm doing (or could do) that might impact how fast it progresses?



B. Treatment & therapy check-in

- ☐ Is my current treatment still the best option for me?
- ☐ How well is it working, and are there other options I should consider?
- ☐ What side effects should I expect, and how can I manage them?
- ☐ Are there any new treatments or clinical trials I should know about?
- ☐ How often should I be getting checkups or specific tests?
- ☐ Are there any meds or supplements I should stay away from?



click to download

4 Fabry & you: your doctor visit

CHECKLIST doctor's visit



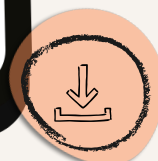
C. Looking at my whole health

- ☐ How is Fabry affecting my general health right now?
- ☐ Are there specific organs or systems I should keep an eye on (like heart, kidneys, or nervous system)?
- ☐ Are there changes I can make to my lifestyle, like diet, exercise, or stress, to help me feel better?
- ☐ What can I do about fatigue or pain when they get in the way?
- ☐ Should I be seeing any specialists regularly (like a cardiologist or nephrologist)?



D. Life beyond the lab results

- ☐ How can I balance work, studies, or daily life with Fabry?
- ☐ What support or flexibility can I ask for at school or work?
- ☐ What's the best way to talk to others like friends, family, or coworkers about my condition?
- ☐ Are there patient groups, communities, or online spaces I can join?
- ☐ Can Fabry affect my mental health, and if so, where can I go for support?
- ☐ Are there any social or financial resources I should look into?



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4 Fabry & you: your doctor visit

CHECKLIST doctor's visit



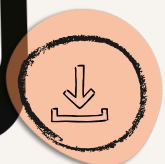
E. Looking ahead & staying prepared

- ☐ What should I know about family planning or genetic counseling?
- ☐ How can I stay ahead of potential complications long-term?
- ☐ What do I need to know in case of a Fabry-related emergency?
- ☐ Are there habits or routines I should follow to help prevent flare-ups?
- ☐ Are there any exciting developments or research updates I should know about?
- ☐
- ☐
- ☐
- ☐



Bonus tip:

Write down your own questions before each visit, no concern is too small. Your health journey is personal, and your voice matters in every appointment.



click to download

4 Fabry & you: your doctor visit

4.2 How to advocate for yourself: at the doctor's office and beyond

When you're living with a rare condition like Fabry disease, one of the most powerful skills you can build is self-advocacy, learning to speak up for your needs, ask the right questions, and make sure your voice is heard.

That might sound intimidating at first but the good news is, you don't have to be loud or know all the medical terms. Self-advocacy is simply about being confident in what you know about your own body, experiences, and needs.

4.2.1 Advocating for yourself during medical visits

Doctors are experts in medicine but **you are the expert in your own life**. Here's how to make the most of your appointments:



Prepare before your visit

- Write down symptoms, questions, or changes since your last appointment.
- Use a checklist (like the one in this guide) to make sure you don't forget important topics.
- Bring someone with you if it helps to have support or a second set of ears.

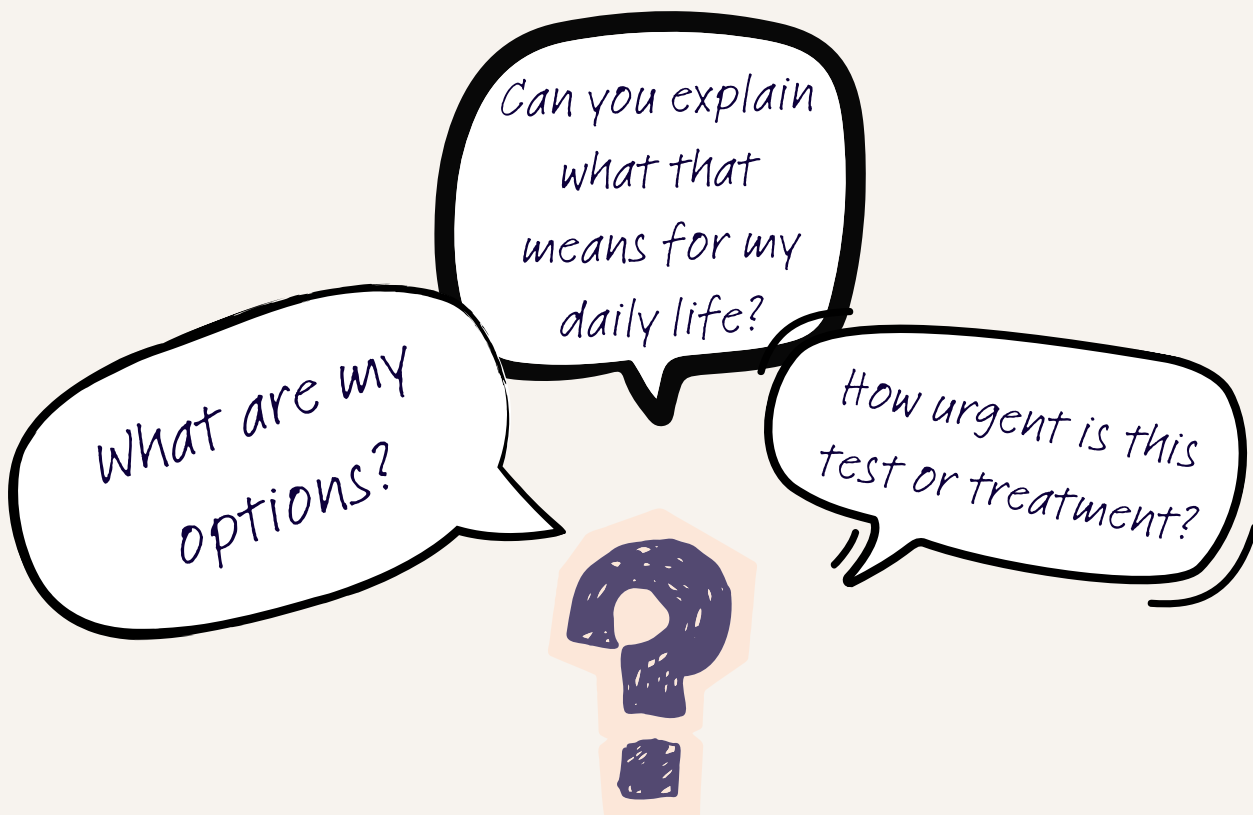
4 Fabry & you: your doctor visit



During the appointment

Try not to hesitate to ask your doctor to slow down or explain something again.

Use phrases like:



If something doesn't feel right, say so. It's okay to ask for a second opinion or more time to decide.

4 Fabry & you: your doctor visit

➤ After the appointment

- Summarize what was discussed in a notebook or app.
- Follow up if something wasn't clear.
- Track how you're feeling or reacting to treatment, this helps in future visits.

➤ Talking to others about your condition

Self-advocacy also includes communicating your diagnosis to others in ways that feel honest and comfortable to you. This might include friends, teachers, employers, or romantic partners.

You don't have to share everything but being open (when you feel ready) can make your relationships stronger and help people better support you.

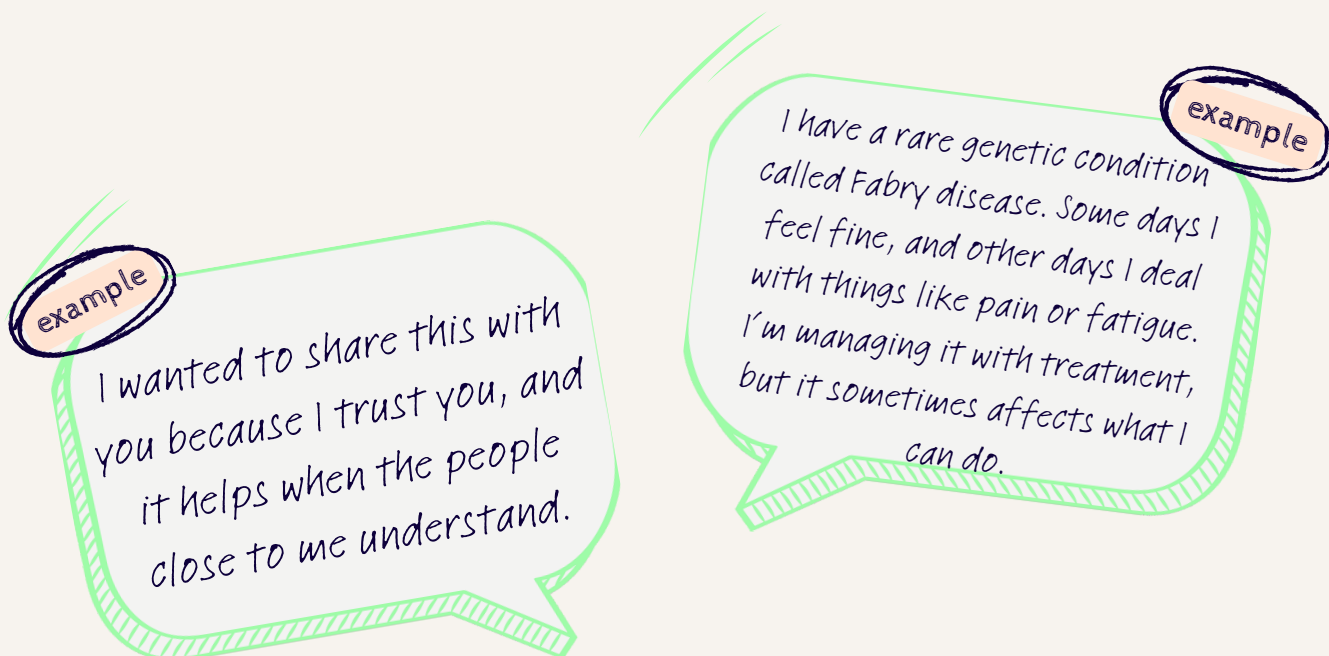
➤ Tips for talking about Fabry

- Choose a time and place that feels right for you, somewhere calm, private, and where you feel at ease.
- Share what feels important to you, at your own pace. You don't have to explain everything at once.
- Keep it simple if that helps and focus on what matters most for your daily life and needs.
- Remember that this is your story, you can decide what to share, how much, and with whom.
- It's okay to set boundaries. You don't have to answer every question or go into details you're not comfortable with.
- Be prepared for different reactions. Some people may need time to process, and that's okay, but your experience remains valid.



4 Fabry & you: your doctor visit

Example phrases:



It can help you to remember that:

- Your experiences and needs are valid, even if others don't fully understand right away.
- You have the right to ask questions, take your time, and be involved in decisions about your care.
- Advocating for yourself is a skill and like any skill, it gets easier with practice.

Being an advocate for yourself doesn't mean having all the answers. It just means showing up for yourself, and giving others the chance to show up for you, too.

4 Fabry & you: your doctor visit

4.3 Fabry patient digital card, needed for emergency, travel, safety

MEDICAL EMERGENCY CARD ANDERSON-FABRY DISEASE

FIN
Fabry International Network

- Full name:
- Date of birth:
- Emergency contact:
 - Name:
 - Relationship:
 - Phone number:
- Doctor/Specialist contact:
 - Name:
 - Phone number:
- **Diagnosis:** Anderson-Fabry disease (Genetic condition)
- **Current treatment** (e.g. ERT/Chaperone)

MEDICAL EMERGENCY CARD ANDERSON-FABRY DISEASE

FIN
Fabry International Network

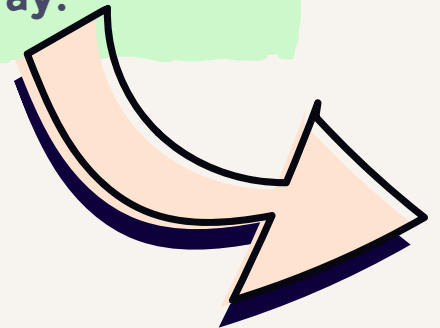
- **Key health risks** (e.g. cardiovascular, renal, neurological issues):
- **Allergies & medication sensitivities:**
- **Current medications:**
- **Important notes for medical staff** (e.g. drug precautions, treatment considerations):
- **Additional information** (e.g. blood type, Fabry Disease Association Contact, ...):

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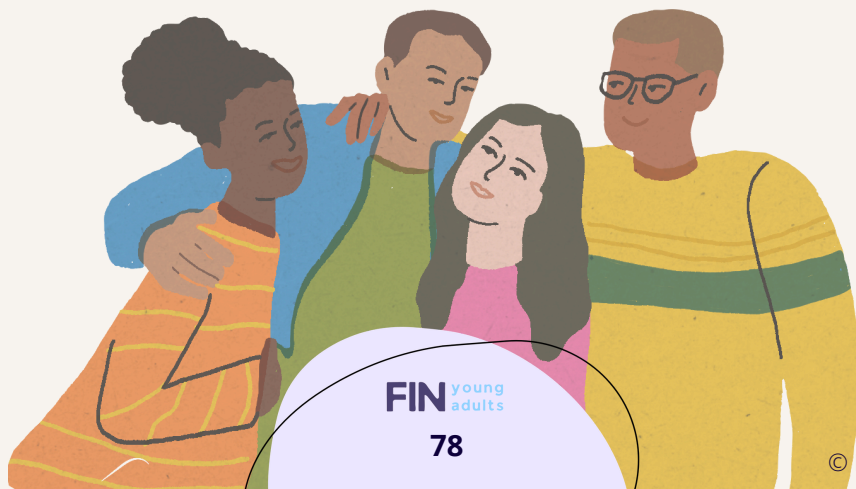
The following chapters aim to provide support, guidance, and actionable steps to help young adults with Fabry navigate mental health, pain management, and family planning.

You're not alone in this journey, there are resources, professionals, and communities ready to support you every step of the way.



Disclaimer:

The questions and responses in this chapter were developed together with members of the FIN Young Adults community and reflect topics that young adults commonly raise when navigating life with Fabry. The expert responses provide general guidance and insight based on professional experience and community discussion. As every individual's experience with Fabry is unique, readers are encouraged to discuss specific medical questions or decisions with their healthcare team.



Conversations with experts: Answering your questions



At Fabry International Network (FIN), our work is shaped by the voices and experiences of our community. We work closely with people living with Fabry around the world, and the Young Adults community continues to play a particularly important role in guiding our projects and resources.

Through conversations, meetings, and shared experiences, young adults have raised thoughtful questions about living with Fabry, from medical and lifestyle topics to emotional wellbeing and planning for the future. Together with members of the FIN Young Adults community, we gathered these questions and invited trusted medical and allied health experts, who work closely with individuals living with Fabry, to share their insights.

The following section presents these community-driven questions alongside expert reflections. We hope these perspectives help provide reassurance, knowledge, and a starting point for further conversations with your own healthcare team. These chapters go a bit deeper into the medical side. If it feels like a lot, that's normal, you don't have to read it in one sitting.

Before moving into the questions and answers, we would like to introduce the experts who contributed to this chapter. All of them have worked closely with the Young Adults group and have become trusted allies and steady supporters of our community.



Conversations with experts: Answering your questions



Dr. Nadia Ali (USA)



Dr Nadia Ali is a U.S.-based psychologist specialising in Fabry disease and related metabolic conditions. She is involved in both clinical care and research, with a focus on neurological symptoms and long-term disease management. Dr Ali is committed to advancing education and collaboration to improve outcomes for people living with Fabry.

Dr. Seema Kanwal (Canada)



Dr Seema Kanwal is a Canadian physician working in the field of rare and metabolic diseases, including Fabry disease. She is dedicated to providing patient-centred care and works closely with individuals and families to navigate diagnosis, treatment, and long-term monitoring. Her approach combines clinical expertise with compassionate communication.

Dawn Laney (USA)



Dawn Laney is a genetic counselor and long-standing leader in the Fabry community in the United States. She has worked closely with individuals and families affected by Fabry disease for many years, supporting them through diagnosis, education, and long-term care decisions. Dawn is passionate about patient advocacy, research, and ensuring that lived experience remains at the heart of clinical conversations.



Conversations with experts: Answering your questions



Dr. Uma Ramaswami (UK)



Dr Uma Ramaswami is a leading Fabry specialist based in the United Kingdom, with decades of experience caring for children and adults living with Fabry disease. She has been closely involved in international research and registries and has contributed significantly to our understanding of Fabry pain, early diagnosis, and long-term treatment outcomes. Dr Ramaswami is known not only for her clinical expertise, but also for her deep commitment to improving quality of life for patients and families.



5 Fabry & mental health



Dr. Nadia Ali

5.1 Questions we asked Dr. Nadia Ali

- What is the link between Fabry Fog and emotional regulation?
- Would treatment for ADHD (stimulants) work for Fabry Fog?
- Should I get psychological help?
- How do I get through the grief?
- How do I deal with the grief of my family?
- How to deal with the medical overwhelm?
- How do I accept the progress of the disease?
- How to deal with the invisibility of the disease?
- How to talk about the disease with my peers and close ones?
- How can I deal with the loneliness and powerlessness?
- What are the best coping skills?
- Acceptance of the disease is very difficult for newly diagnosed patients. How do I cope?
- How do outcomes (depression/anxiety) differ between Fabry & Non Fabry patients?
- Tips for acceptance and managing anxiety that comes with a diagnosis and limitations
- What is the impact of Fabry Fog on daily life?
- How/where to get help if feeling depressed or anxious?



5 Fabry & mental health



Dr. Nadia Ali

- Am I eligible for subsidy?
 - How and where can I find this information in my country?
- How might chronic illness impact my mental health in the future? Or those around me? (family/carers/friends)?
- What could be the sources of depression: mortality /pain/limitations (social and physical)?
- Are there any routines or habits that can make day-to-day life with Fabry easier?
- What role do practices like mindfulness, yoga, or meditation play in managing Fabry?
- How can I build sustainable habits when I'm overwhelmed or inconsistent?
- Is there any link between nutrition and brain fog or concentration problems?

Living with Fabry disease isn't just about managing physical symptoms, it also comes with emotional and mental health challenges. Many young adults with Fabry experience stress, anxiety, or depression, often linked to uncertainty about the future, chronic pain, or feeling isolated. You are not alone in this, and there are ways to navigate these challenges.

5 Fabry & mental health



5.2 Understanding the mental health impact of Fabry

Fabry fog & emotional regulation

Many people with Fabry experience what's often called "Fabry fog", a state of mental cloudiness that can include confusion, forgetfulness, trouble concentrating, or feeling mentally slow. It's a real and common symptom that can make everyday tasks feel harder. Fabry fog can also affect your mood and make it more challenging to regulate stress, frustration, or emotions in general.

Grieving the diagnosis

Whether diagnosed recently or as a child, grief is natural. You might mourn the idea of a life without chronic illness, and that's okay.

Medical overwhelm

Appointments, treatments, and lifestyle changes can feel like too much at times. Learning how to manage these challenges can help reduce anxiety.

Loneliness & isolation

Because Fabry is rare, it may be hard to find people who understand what you're going through. Connecting with others can help.

5 Fabry & mental health



Dr. Nadia Ali

5.3 How to cope with mental health challenges

Seek professional support

Therapy can help, especially with professionals experienced in chronic illness.

Talk about It

Opening up to friends, family, or peer support groups can make a huge difference.

Develop coping skills

Mindfulness, journaling, or engaging in hobbies can help manage stress.

Manage anxiety & depression

If you experience symptoms of anxiety or depression, don't hesitate to seek medical help.

Set boundaries

It's okay to say no to things that drain your energy.

5 Fabry & mental health



5.4 Resources for mental health

Online therapy platforms

Some people explore platforms like BetterHelp or Talkspace, which offer support for people living with chronic illness. Experiences vary, so it's important to choose what feels right for you.

Therapist

Things to consider when looking for a therapist:

- Do they have experience with chronic or rare illnesses?
- Are you comfortable with video sessions or do you prefer in-person support?
- Do you feel safe and understood during your sessions? A good fit matters, don't hesitate to try a few options if the first one doesn't feel right.
- If cost is a concern, check for free or subsidized mental health services in your country or through patient organizations.

Fabry-specific mental health groups

Check Fabry support organizations.

Crisis support

National mental health helplines.

6 Fabry & pain



Dr. Uma Ramaswami

6.1 Questions we asked Dr. Uma Ramaswami

- What is the difference between diabetic neuropathy and Fabry neuropathy?
- Why is there so much variation in neurological pain symptoms between patients?
- How do I identify Fabry pain?
- What is a Fabry pain crisis?
- How should I talk to my doctor about pain?
- Should I take medication for pain and when?
- How do different pain medications work?
- What are the pros and cons of pain medication?
- Does enzyme replacement therapy (ERT) relieve pain?
- Are there natural ways to help manage pain?
- What about cannabis or CBD?
- Do supplements support pain control?
- Can I adjust my medication dosage myself?
- Are some pain treatments more effective than others?
- How can I identify my personal pain triggers?

6 Fabry & pain



Dr. Uma Ramaswami

6.2 Understanding Fabry pain

Fabry pain is primarily **a small fibre neuropathy**.

In Fabry disease, pain occurs because the enzyme **alpha-galactosidase A** is deficient. This leads to the accumulation of sphingolipids, especially **globotriaosylceramide (Gb3)** and **lyso-Gb3**, in nerve endings. This build-up disrupts normal nerve function and causes neuropathic pain.

In classic Fabry disease, this process often begins in **early childhood**.

Small fibres vs. large fibres

Fabry disease mainly affects:

- **Unmyelinated C fibres** → dull, burning pain, heat intolerance
- **Myelinated A-delta fibres** → sharp, stabbing pain, cold sensitivity

This explains why Fabry pain is often described as:

- Burning
- Sharp or stabbing
- Electric shock-like
- "Walking on needles"
- Extreme discomfort with temperature changes

Large nerve fibres are usually spared in Fabry, which means nerve conduction tests may appear normal especially early in the disease.

6 Fabry & pain



Dr. Uma Ramaswami

How is this different from diabetic neuropathy?

Diabetic neuropathy:

- Usually develops after many years of poorly controlled diabetes
- Typically appears in mid to late adulthood
- Often affects large fibres
- Progresses upward (from feet toward legs)
- Is associated with numbness and risk of ulcers

Fabry neuropathy:

- Often begins in childhood
- Is caused by enzyme deficiency and lipid accumulation
- Is mainly small fibre neuropathy
- Typically remains in hands and feet (acroparaesthesia)
- Is not fully preventable, though treatment may reduce severity

Unlike diabetic neuropathy, Fabry pain does not usually “travel up” the limbs in a classic stocking pattern.

What is a Fabry pain crisis?

A Fabry pain crisis is a severe, acute episode of neuropathic pain.

It can:

- Start suddenly
- Last hours or days
- Be triggered by infection, fever, hot or cold weather, exercise, stress, dehydration

6 Fabry & pain



Dr. Uma Ramaswami

Pain crises are more common in children with classic Fabry disease, particularly boys.

The pain can be intense enough to require hospitalization. Simple analgesics often do not help. Stronger medications or intravenous treatment may be necessary.

Dr Ramaswami emphasizes the importance of:

- Preventing overheating
- Maintaining hydration
- Educating schools about Fabry
- Recognizing that abdominal pain crises may mimic conditions like appendicitis

Why do symptoms vary so much?

Fabry disease is heterogeneous. Pain severity and pattern depend on:

- Genotype
- Degree of enzyme deficiency
- Age of onset
- Co-morbidities (e.g., Raynaud's, diabetes)
- Individual nervous system sensitivity

Even within the same family, symptoms can differ significantly.

6 Fabry & pain



Dr. Uma Ramaswami

Associated symptoms

Because small fibres and autonomic fibres are involved, patients may experience:

- Abnormal or reduced sweating
- Heat/cold intolerance
- Impaired heart rate variability
- Dizziness or postural hypotension
- Gastrointestinal symptoms
- Cornea verticillata (eye finding)
- Angiokeratoma rash
- Fatigue

Heart, kidney, and stroke complications usually occur in adulthood, but early organ involvement should always be evaluated carefully.

6.3 How pain medications work

Different medications target different mechanisms:

Sodium channel dysfunction

Gb3 accumulation alters ion channels in nerve membranes → shooting pain

→ **Carbamazepine** may help

6 Fabry & pain



Dr. Uma Ramaswami

Peripheral sensitisation (C-fibre damage)

Burning pain, heat sensitivity

→ **Opioids** (carefully monitored) may help

A-delta fibre disinhibition

Burning pain triggered by cold

→ **Gabapentin** or GABA analogues

Sympathetic-mediated pain

Cold allodynia

→ **Clonidine** (in selected cases)

Tricyclic antidepressants

Can reduce sharp/shooting pain and support mood

Standard analgesics like paracetamol are generally ineffective for Fabry neuropathic pain. NSAIDs are used cautiously due to kidney risk.

6 Fabry & pain



Dr. Uma Ramaswami

6.4 Should I take medication?

Medication decisions depend on:

- Type of pain
- Severity
- Frequency
- Impact on daily life

Doctors usually:

1. Start low
2. Increase gradually
3. Switch or combine if needed

Psychological support can significantly improve coping and quality of life. Patients should never adjust doses independently.

Does ERT help?

Enzyme replacement therapy may:

- Reduce frequency of pain crises
- Lower severity
- Allow reduction of pain medication

However, it does not usually eliminate pain completely.

6 Fabry & pain



Dr. Uma Ramaswami

6.5 Natural and lifestyle approaches

Identifying triggers is crucial.

Common triggers:

- Heat (baths, sauna, humidity)
- Cold exposure
- Exercise
- Fever/infection
- Stress
- Fatigue

Supportive strategies include:

- Hydration
- Temperature management
- Pacing activities
- Avoiding overheating
- Stress reduction
- Dietetic review for abdominal symptoms (e.g., supervised low FODMAP if appropriate)

These approaches help reduce frequency but may not replace medication.

6 Fabry & pain



6.6 Cannabis, CBD & supplements *Dr. Uma Ramaswami*

Cannabinoids have shown benefit in neuropathic pain in general populations. However:

- Evidence in Fabry is limited
- Dosing and quality vary widely
- Side effects and interactions must be considered
- Regulations vary per country and region

If explored, it should be done under specialist supervision.

Supplements

- Vitamin D (if deficient) → bone pain
- Vitamin B12 (if deficient) → tingling symptoms
- Magnesium → muscle cramps

There is limited evidence that supplements alone improve small fibre neuropathy in Fabry.

7 Fabry & family planning



Dawn Laney

7.1 Questions we asked Dawn Laney

- What is family planning and why does it matter when a parent has Fabry disease? When should you start thinking about it?
- What are the risks of being pregnant or giving birth to a baby when you have Fabry disease? Are there risks to the baby?
- Should I or my partner do carrier screening for genetic conditions?
- What are alternative conceiving methods when you have Fabry disease? What are the pro's and con's to using them or not?
- In which countries are alternative conceiving methods available?
- How do I talk to my (potential) partner and his/her/their family about Fabry? And when?
- Does being on enzyme replacement therapy (ERT) affect pregnancy?
- Does being on chaperone therapy affect pregnancy?
- If you know that a baby born as a boy or girl will have Fabry, can a parent select gender in order to not passing on the condition?
- What are the options are available for individuals with Fabry disease and what support is available?
 - Adoption – is this option limited because of having Fabry?
 - In Vitro Fertilization (IVF) and preimplantation genetic diagnosis (PGD)
 - Natural – how to deal with stigma associated with Fabry?

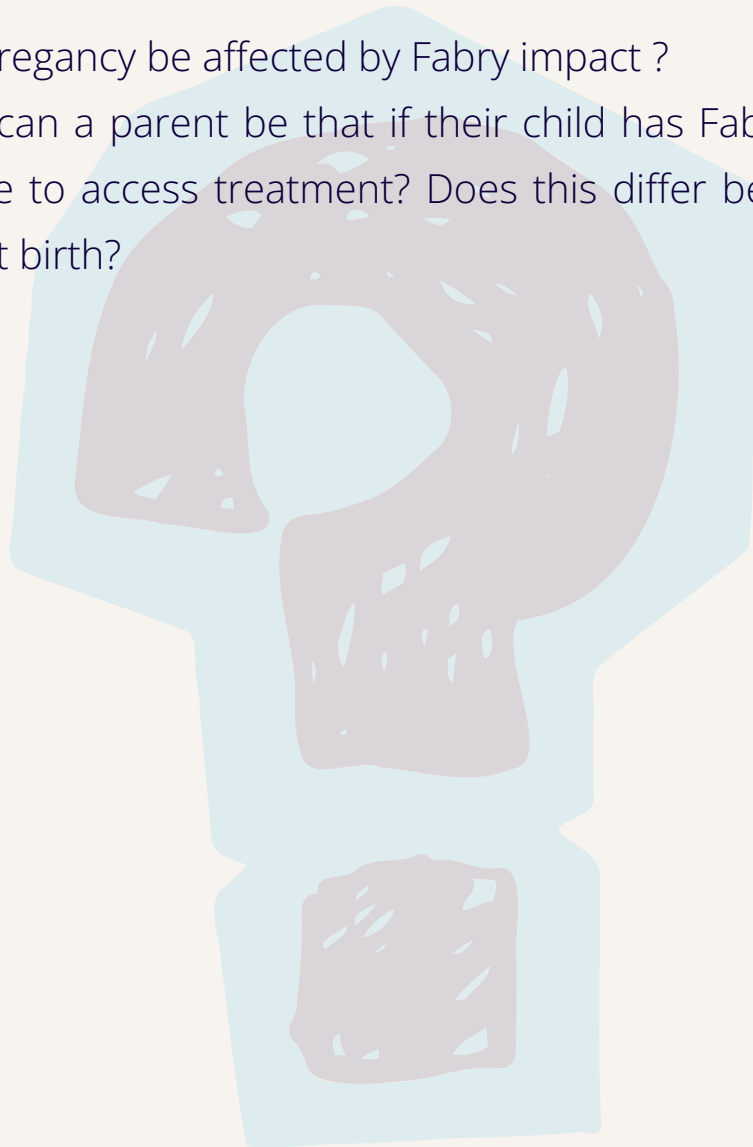


7 Fabry & family planning



Dawn Laney

- Should care provided to a pregnant person with Fabry differ to a non-Fabry person?
- Should there be extra care for baby AND pregnant person with Fabry?
- How can pregnancy be affected by Fabry impact ?
- How sure can a parent be that if their child has Fabry disease they will be able to access treatment? Does this differ between genders assigned at birth?



7 Fabry & family planning



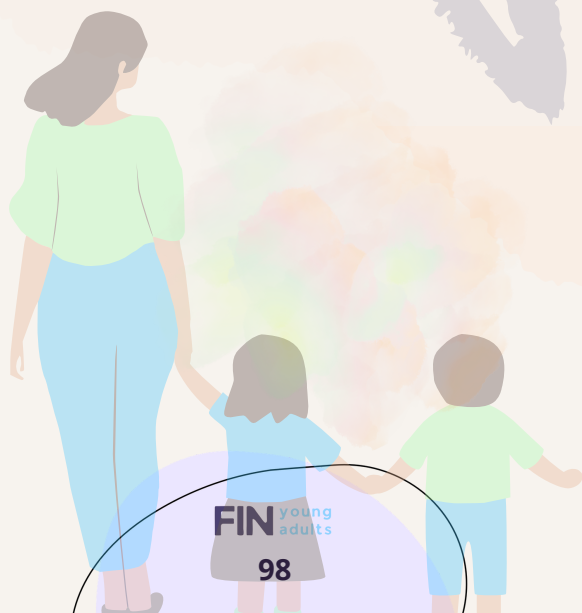
Dawn Laney

Thinking about the future, relationships, and having children while living with Fabry disease can feel complex. There are many options, and understanding them can help you make decisions that feel right for your situation.

It's important to remember that the decision to have children and how you choose to have them, is deeply personal. It may involve you, your partner, and your wider support system. There is no single "right" path.

Understanding how Fabry disease may affect you, how it can be inherited, and what treatment or support options exist can help you feel more informed and confident in these decisions. If needed, you can always seek further information and guidance from healthcare professionals, Fabry specialists, and patient organisations.

In this chapter, we highlight some key aspects to consider as someone living with Fabry disease.



7 Fabry & family planning



7.2 Key considerations in family planning

Genetic risks

If you have Fabry disease, there is a possibility that your children may inherit it. Fabry disease is an X-linked condition, which means inheritance works differently depending on your genetic makeup.

- Individuals with one X chromosome (typically biological males) will pass the altered gene to all their daughters and none of their sons.
- Individuals with two X chromosomes (typically biological females) have a 50% chance of passing the altered gene to each child, regardless of the child's sex.

These patterns can feel complex at first. Many people find it helpful to use visual tools or diagrams to better understand how Fabry disease is passed through families. Your healthcare provider or patient organisations can guide you to resources that make this easier to understand and explain.

Fabry-specific treatment and pregnancy

The decision to start, continue, or pause Fabry-specific treatments such as enzyme replacement therapy or chaperone therapy during pregnancy is a personal one. It is usually made together with your healthcare team and, if relevant, your partner.

7 Fabry & family planning



Dawn Laney

There is growing information and research about pregnancy outcomes for individuals using these treatments, which can help guide these conversations and decisions.

Medications to review during pregnancy

Some medications may not be recommended during pregnancy due to potential risks to the developing baby. It's important to review your medications with your doctor, ideally before becoming pregnant, to understand what may need to be adjusted and what alternatives are available.

If you are pregnant, do not stop any medication abruptly without medical guidance, as this can also have an impact on your health and the pregnancy.

Exploring different ways to build a family

Depending on where you live, there may be different options available if you wish to reduce the chance of passing on Fabry disease. These can include approaches such as IVF (in vitro fertilisation) with preimplantation genetic diagnosis (PGD), or the use of egg or sperm donation.

For some people, these options feel right; for others, they may not. Access, availability, and personal preferences can all play a role.

7 Fabry & family planning



Dawn Laney

Adoption

Some individuals or couples choose to grow their families through adoption. This can be a meaningful path to parenthood and may be something you wish to explore as part of your decision-making.

Prenatal testing

In many countries, there are tests available during pregnancy that can help determine whether a baby is affected by Fabry disease. These may include procedures such as chorionic villus sampling (CVS) or amniocentesis.

These tests are carried out at specific stages of pregnancy and can involve certain risks, so it's important to discuss them carefully with your healthcare provider.

At present, there are no treatments that can change the course of Fabry disease before birth, so deciding whether to pursue testing is a personal choice.

Talking about Fabry in relationships

Sharing information about Fabry disease in a relationship can feel vulnerable, and there is no perfect moment to do so. You may choose to talk about it early, or wait until you feel ready, both are valid.

- **When to talk about it**

There is no right timing, but openness can help build understanding and trust over time.

7 Fabry & family planning



Dawn Laney

- **How to explain it**

You don't need to explain everything at once. Keeping things simple and sharing what feels most important to you can be enough.

If helpful, there are resources and healthcare professionals who can support these conversations, and some people choose to bring their partner to appointments to ask questions together.

- **Addressing concerns**

It's natural for both you and your partner to have questions, concerns, or different reactions. Giving space for these conversations, and approaching them with honesty and care, can help you navigate them together.

The emotional side of family planning

Thinking about family planning while living with Fabry disease can bring up many different emotions. You might feel uncertain, hopeful, overwhelmed, or even conflicted at times. These feelings are valid, and you are not alone in experiencing them.

There is no need to have everything figured out right away. Taking time to reflect, ask questions, and explore what feels right for you is an important part of the process. Some people find it helpful to talk things through with a partner, a trusted person, or a professional such as a counsellor or psychologist.

7 Fabry & family planning



Dawn Laney

Finding support and information

If you would like to explore your options further or speak to someone, the following resources may be helpful:

- **Genetic counselling services:** to better understand inheritance, risks, and available options
- **Fabry specialist centres and healthcare providers:** for personalised medical advice and support
- **Patient organisations:** such as the Fabry International Network (FIN) and national Fabry groups: offering information, peer support, and connection
- **Rare disease organisations:** such as EURORDIS, providing broader resources and support for individuals and families
- **Fertility clinics with experience in genetic conditions:** for guidance on options such as IVF and PGD

A gentle reminder

You don't have to go through this alone. Reaching out for information or support, when you feel ready, can help you make decisions that feel informed, supported, and right for you.

8 Nutrition & lifestyle



Dr. Seema Kanwal

8.1 Questions we asked Dr. Seema Kanwal

- Are there specific foods or diets that help manage Fabry symptoms, like fatigue, GI issues, or pain?
- Are there foods that can make Fabry symptoms worse or should be avoided?
- What role does hydration play in managing Fabry symptoms?
- How can I eat well if I don't have much energy to cook or shop regularly?
- Can diet impact inflammation or pain levels in Fabry?
- Are there supplements that might help (e.g., for energy, digestion, immune health)? Should I talk to a doctor before using them?
- What are some easy, go-to meals or snacks that support overall health with Fabry?
- How important is physical activity when you have Fabry? What types of movement are safe and beneficial?
- What can I do to improve my energy levels on a daily basis?
- How do I balance rest and activity when I feel too tired but want to stay active?
- Are there strategies to improve sleep, especially when I have pain or anxiety?



8 Nutrition & lifestyle



Dr. Seema Kanwal

- How do stress and emotional health affect physical symptoms, and what can help?
- Are there any routines or habits that can make day-to-day life with Fabry easier?
- What role do practices like mindfulness, yoga, or meditation play in managing Fabry?
- How can I build sustainable habits when I'm overwhelmed or inconsistent?
- Is there any link between nutrition and brain fog or concentration problems?
- What advice would you give to someone trying to live a "healthy" life with a chronic illness like Fabry, without feeling guilty or pressured?

8 Nutrition & lifestyle



Dr. Seema Kanwal

8.2 Nourishing your body while living with Fabry

Living with Fabry disease can bring many physical and emotional challenges, and nutrition and daily lifestyle choices can play an important role in how you feel, both day to day and long-term. While there is no specific "Fabry diet," certain foods and habits may help support your energy levels, ease digestive symptoms, and reduce discomfort.

8.2.1 Food as a foundation, not a fix

This chapter offers gentle guidance and ideas, not rules. We know that life is busy, and not everyone has the time, energy, or resources to prepare elaborate meals or follow strict wellness routines.

Whether you're studying, working, managing symptoms, or simply trying to keep up with daily life, we hope the tips here feel supportive and realistic.

Start by tuning into your body. Keep track of what helps and what doesn't. Everyone with Fabry is different, but here are some general tips shared by others in the community:

8 Nutrition & lifestyle



Dr. Seema Kanwal

- **Stay hydrated:** Drinking enough water can help with fatigue and flushing out toxins.
- **Eat regularly:** Skipping meals can sometimes worsen fatigue or cause dizziness.
- **Focus on whole foods:** When possible, try to include fruits, vegetables, whole grains, and lean proteins.



8 Nutrition & lifestyle



Dr. Seema Kanwal

Foods that may help with common symptoms:

Symptom		Helpful foods
Nausea	→	Ginger tea, plain crackers, bananas, boiled potatoes, applesauce
Fatigue	→	Whole grains, nuts, eggs, iron-rich foods (e.g. lentils, spinach), complex carbs
Digestive discomfort	→	Oats, steamed vegetables, probiotic foods (e.g. yogurt), chamomile tea
Constipation	→	Fiber-rich foods (berries, pears, legumes), water, flaxseed
Bloating / Gas	→	Cooked (vs. raw) vegetables, peppermint tea, small frequent meals

Keep in mind that everyone reacts differently to food. Some people with Fabry find certain high-fiber foods or dairy irritating for instance. It's okay.

8 Nutrition & lifestyle



Dr. Seema Kanwal

8.2.2 Foods to watch out for

Some people with Fabry find that dairy, very high-fiber raw foods, or overly processed meals can increase discomfort. Keep a gentle food diary if you're unsure what's affecting you, it's about learning, not judging.

8.2.3 Hydration is not just about water

Hydration is essential for everyone, but it's especially important for people living with Fabry disease. Due to gastrointestinal symptoms like diarrhea, vomiting, or decreased sweating, many individuals with Fabry are at a higher risk of dehydration, sometimes without even realizing it. In addition, issues with autonomic function can lead to low blood pressure or low blood volume, which can make fatigue, dizziness, and brain fog worse.

Staying hydrated doesn't just mean drinking more water, it means making sure your electrolytes (like sodium, potassium, and magnesium) are balanced, too.

8 Nutrition & lifestyle



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Here are some ways to stay on top of hydration, even on low-energy days:

Daily hydration tips

- **Carry a reusable water bottle** with you and sip throughout the day, even small sips help.
- **Start your day with a glass of water** before caffeine or breakfast to rehydrate after sleep.
- Add **electrolytes** to your water if you're experiencing symptoms like diarrhea, excessive sweating, or fatigue. Look for clean options without added sugar or artificial dyes.
- Options include: **coconut water**, electrolyte powders or tablets (e.g., LMNT, Nuun, or Hydralyte), or homemade electrolyte drinks with sea salt, lemon, and a bit of honey.
- **Warm herbal teas** such as chamomile, peppermint, or ginger can be soothing and hydrating, especially if you're feeling nauseated or anxious.
- If plain water is hard to drink, **infuse it with fruit, cucumber, or mint** for flavor without added sugar.
- **Monitor your urine color:** pale yellow usually means you're well-hydrated, while darker shades may signal dehydration.

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Helpful tip for hot days after exercise

If you're outdoors or active, pre-hydrate with water and a small pinch of salt or an electrolyte tablet 20–30 minutes before activity. Then continue to sip fluids during and after.

What to watch for

Dehydration symptoms can include:

- Headaches
- Dizziness or lightheadedness
- Dry mouth or lips
- Fatigue or sluggish thinking
- Muscle cramps
- Constipation

Hydration needs can vary day to day. **Listen to your body**, and if you're losing fluids through sweating or GI symptoms, make it a priority to replenish both **water and minerals**.

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8.2.4 When you're too tired to cook

Fatigue is one of the most common and challenging symptoms of Fabry disease. On low-energy days, even the thought of preparing a meal can feel overwhelming. This is where simple, flexible, and forgiving strategies can help keep your nutrition on track, without draining your energy or willpower.

You don't need to cook every day to eat well. A little planning when you feel up to it can go a long way.



Smart shortcuts for low-energy days

- **Batch cook and freeze:** Make a big pot of soup, chili, or stir-fry when you're feeling well. Freeze single-serving portions in containers so you have something ready to go later.
- **Snack plates can be meals:** A boiled egg, a few crackers, cut-up veggies, and some hummus or avocado is a real meal, no stove required.
- **Protein smoothies:** Keep ingredients like frozen berries, greens, protein powder, nut butter, and oat milk on hand. Blend and sip, even lying on the couch.

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- **No shame in shortcuts:** Pre-chopped veggies, frozen brown rice, canned lentils, or rotisserie chicken are tools, not cheats.
- **Overnight oats:** Mix oats, plant milk, chia seeds, and berries in a jar, let it sit overnight for a grab-and-go breakfast.
- **Toast is a canvas:** Try nut butter + banana, hummus + cucumber, or avocado + hemp seeds for easy nutrition.

Low-energy meal ideas

Here are some ideas that **take 5 minutes or less:**

- Greek yogurt with granola and berries
- Rice cakes with nut butter and sliced apple
- Hard-boiled eggs and a handful of cherry tomatoes
- Pre-washed greens + canned beans + olive oil + lemon juice = instant salad
- Crackers with cheese and sliced cucumber



Meal support options

- **Meal delivery services** (e.g. Fresh Prep, HelloFresh, or local delivery) can take the stress out of planning and shopping.
- Ask a friend or family member to help with a weekly prep session or grocery pickup. Many people want to help, you just have to let them in.

8 Nutrition & lifestyle



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- Keep a small “Fabry fatigue food stash” of shelf-stable or frozen essentials you can turn to when needed.

Reminder: Eating something is always better than skipping a meal entirely. Gentle nutrition can look like soup from a can or toast with peanut butter.

This isn't about perfection - it's about sustaining yourself with kindness.



8 Nutrition & lifestyle



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8.2.5 Inflammation, pain & diet

Pain in Fabry disease can stem from several sources: nerve sensitivity, small vessel changes, or chronic inflammation in the body. While medication plays a role, your daily food choices can influence how much inflammation is present and how your body responds to pain.

The goal isn't perfection. It's **supporting your body** with foods that calm inflammation while reducing those that may fuel it. Think of it as adding in more of the "good stuff" rather than taking everything away.

Anti-inflammatory foods to embrace

Incorporating these foods consistently can help support your body's natural healing systems:

- **Colorful fruits and vegetables** – Especially berries, leafy greens, and cruciferous veggies like broccoli and cauliflower. These are rich in antioxidants that help repair cellular damage.
- **Healthy fats** – Found in olive oil, avocado, nuts, seeds, and fatty fish like salmon, mackerel, and sardines (omega-3s help reduce inflammation in the nervous and cardiovascular systems).
- **Herbs and spices** – Turmeric, ginger, cinnamon, and garlic all have natural anti-inflammatory properties. Use them often in cooking or teas.

8 Nutrition & lifestyle



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- **Whole grains** – Brown rice, oats, quinoa, and buckwheat provide fiber that helps regulate blood sugar and support gut health.
- **Fermented foods** – Such as kimchi, sauerkraut, kefir, and yogurt (if tolerated). A healthy gut can help regulate the immune system and inflammation.

Foods to reduce (not eliminate)

Some foods are known to increase inflammation when consumed in excess. These don't need to be avoided completely, but being mindful of them can help:

- **Refined sugar** – Especially in sodas, candies, baked goods, and sweetened cereals
- **Highly processed foods** – Packaged snacks, microwave meals, fast food
- **Refined oils** – Such as soybean, corn, or canola oil, which are often found in processed foods
- **Excess red or processed meats** – Limit deli meats, bacon, sausages if they worsen your symptoms

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Instead of focusing on restriction, **try crowding in nutrient-rich options**; you may find the more you add the good stuff, the less you crave the rest.

Inflammation affects more than just the body

Chronic inflammation isn't just physical. It can also impact mood, energy, and cognition, contributing to brain fog or emotional ups and downs. Supporting your body with nourishing foods may help create more stability across the board.

Small, sustainable shift that helps

- Start your day with a **smoothie** packed with greens, berries, and flaxseed
- Swap out one sugary drink for a **herbal tea** or **infused water**
- Add a handful of **walnuts or chia seeds** to your meals for omega-3s
- Cook with **olive oil or avocado oil** instead of vegetable oil
- Make a **colorful plate** at each meal: aim for 3+ different colors

8 Nutrition & lifestyle



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You don't need a perfect diet to reduce inflammation.

What matters most is **consistency and compassion**, choosing foods that support you, not stress you out.

8.2.6 Supplements: support with caution

Supplements can sometimes offer gentle support for common symptoms associated with Fabry disease, especially when nutrient levels are low or digestion is compromised. Some young adults may find benefit from:

- **Magnesium** – to ease muscle cramps, support bowel movements, and promote restful sleep
- **B-complex & iron** – to improve low energy, especially if bloodwork shows a deficiency
- **Probiotics** – to support a healthy gut environment and ease digestive symptoms like bloating or irregularity
- **Vitamin D & Omega-3s** – to reduce inflammation, support immune function, and stabilize mood

8 Nutrition & lifestyle



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That said, **more isn't always better**. Taking supplements without clear need or professional guidance can lead to imbalances or unintended side effects.



Important: Always speak with your Fabry care team before starting anything new. Some supplements may interfere with medications, affect kidney function, or simply be unnecessary based on your current labs or health status.

Remember: your body is already doing a lot. Supplements are meant to support, not replace, a well-rounded care plan.

8.2.7 Nutrition & brain fog

Many with Fabry report trouble concentrating or feeling mentally sluggish. Stable blood sugar, hydration, and nutrient-rich meals (especially with healthy fats like avocado and fish) can help support cognitive clarity.

8 Nutrition & lifestyle



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8.3 Movement, rest & emotional health

Exercise helps with circulation, mood, and energy, but it needs to match your current capacity.

- On good days: walking, yoga, light strength training
- On tough days: gentle stretching, breathwork, or just focusing on rest
- Sleep hygiene: avoid screens before bed (at least 1 hour), keep a consistent routine, magnesium or herbal teas can support better sleep
- Stress and pain often go hand-in-hand. Consider:
 - Mindfulness apps (like Calm or Insight Timer)
 - Gentle yoga or tai chi
 - Journaling, creative outlets, or counseling support when needed

8 Nutrition & lifestyle



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8.4 Habits when you feel overwhelmed

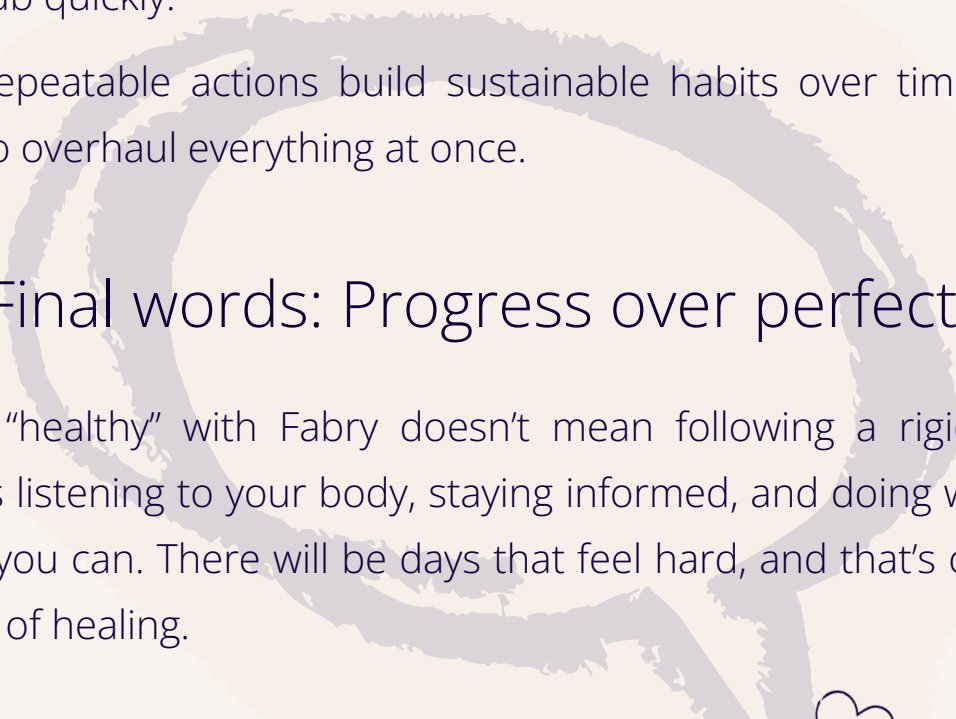
Start with just one small action:

Example: "I'll drink one extra glass of water today," or "I'll prep snacks I can grab quickly."

Tiny, repeatable actions build sustainable habits over time, you don't have to overhaul everything at once.

8.5 Final words: Progress over perfection

Living "healthy" with Fabry doesn't mean following a rigid routine. It means listening to your body, staying informed, and doing what you can when you can. There will be days that feel hard, and that's okay. Resting is part of healing.



*Be kind to yourself.
You're not failing - you're adapting.
And that is powerful.*



9 Patient advocacy: your voice matters

Living with Fabry disease or any rare condition can sometimes feel isolating or overwhelming. But here's something powerful to remember: your story, your experience, and your voice matter. And when you choose to speak up not just for yourself, but for others you're stepping into something important: patient advocacy.

You don't have to be an expert or have a big platform to be an advocate. It starts small, and it starts with you.

9.1 What is patient advocacy?

Patient advocacy means using your experience to make things better for yourself, and for others living with the same or similar challenges. It's about:

- Speaking up for your needs in healthcare, school, or work
- Telling your story can be a powerful way to raise awareness and reduce stigma around Fabry disease. If you feel ready to share, remember that you don't have to do it alone. This guide includes tools and examples throughout like communication tips, mental health resources, and advocacy ideas that can support you in deciding how, when, and what to share.
- Helping others feel seen, heard, and less alone
- Working toward better access to treatments, support, and rights

Advocates come in all forms, students, professionals, parents, patients. You don't need a title to be one. You just need the willingness to be involved.

9 Patient advocacy: your voice matters

9.2 Why is advocacy important?

It drives change. Policies, treatments, and services improve when people living with rare conditions speak out.

- **It creates community:** Sharing your story can help others feel less alone.
- **It gives you power:** Understanding your rights and options means you can make informed choices.
- **It helps healthcare professionals listen:** Your insight helps doctors and researchers understand what really matters to patients.

Whether you're writing a blog post, giving feedback to your doctor, speaking at an event, or helping someone one-on-one, you're making an impact.



9 Patient advocacy: your voice matters

9.3 How to start your advocacy journey?

Start with your story

Everyone's experience with Fabry is unique. Take time to reflect:

- What's something you wish people understood about your condition?
- What challenges have you faced, and how have you overcome them?
- What advice would you give to someone newly diagnosed?
- Sharing your experience even just with friends or online peers, is advocacy in action.

Learn the basics

Understanding how Fabry works and how the healthcare system supports rare diseases can help you feel more confident in conversations with doctors, teachers, or employers.

You can explore:

- Patient-friendly info sites like Orphanet, NORD, or EURORDIS
- Fabry-specific platforms (like the Fabry International Network)
- Webinars or workshops for young patient advocates

9 Patient advocacy: your voice matters

Connect with others

You're not alone. Joining a community can give you strength, new ideas, and opportunities to get involved.

You can:

- Join a patient organization (like FIN's Young Adults Community)
- Attend meetups, conferences, or summer schools
- Follow rare disease advocates and groups on Facebook, Instagram, TikTok, or LinkedIn
- Join peer networks or WhatsApp/Discord groups for Fabry or rare diseases

Take small actions

Advocacy can be simple. You could:

- Write a social media post or personal blog
- Share a resource with a newly diagnosed person
- Ask your school or workplace for better support
- Speak at an event or volunteer with a Fabry organization
- Attend an advocacy training or summer school to grow your skills

Final thoughts

You don't need to have it all figured out to start. Advocacy is about being real, being curious, and showing up in your own way. Whether you want to help others, raise awareness, or push for better care, your experience has value.



Success story from Tatevik Yezekyan



Tatevik Yezekyan's success story



SUCCESS!!



From diagnosis to advocacy: Finding strength through connection

Tatevik is curious; about people, places, and ideas. She likes meaningful conversations, simple things done well, and the kind of quiet beauty you only notice if you slow down: a good cup of tea, a poem that lingers, a photograph that catches a moment exactly as it was. She has a PhD in engineering, works as a project manager, and has lived in more than eight cities across four countries. Four languages have shaped the way she thinks. She calls herself a fighter and an aesthete and the two sit comfortably side by side.

She is authentic and deeply caring, highly sensitive in the best sense of the word, thoughtful, sharp, and considered in everything she does. And like everyone else, she is simply working hard, every day, to build a beautiful life.

Fabry disease is part of her story, but it is not where her story begins.

Tatevik was 27 when she was diagnosed, after years of childhood symptoms, doctor visits across multiple countries, and several misdiagnoses. The answer finally came from a curious and determined emergency room doctor in northern Italy who connected the dots no one else had. Looking back, she believes she could have been diagnosed much earlier and that experience shaped her commitment to raising awareness, both among healthcare professionals and in the wider world.



Tatevik Yezekyan's success story



When everything changes overnight

Hearing the diagnosis felt like the world had turned upside down. Suddenly, life seemed to revolve around one thing: understanding Fabry, learning to live with it, and trying to imagine what it meant for the future.

She was studying abroad at the time, far from family and close friends, in a country whose language she didn't yet speak.

"Understanding medical reports in your own language is already difficult. Imagine trying to understand your life-changing diagnosis in a foreign language."

She turned to the internet, hoping for clarity. What she found instead were overwhelming statistics, risks, and frightening scenarios.

"There are rarely easy diagnosis stories. Mine was a very challenging one."

Choosing silence and why that's okay

For a long time, Tatevik didn't talk openly about her diagnosis. Living with a rare disease felt overwhelming, and the unfiltered information online pushed her further into herself. She was afraid of the disease, its progression, and the uncertainty it brought.



Tatevik Yezekyan's success story



The timing made it harder. The diagnosis arrived at a crucial moment in her studies and career, and life decisions started to feel as much about medical necessity as ambition. She worried about being seen differently. She worried that Fabry might come to define her. Most of all, she needed time to accept it herself.

"To share your story, you first have to accept it for yourself. And that takes time and courage."

It was difficult to talk about something she didn't yet fully understand. She wasn't ready for questions she couldn't answer, or to confront every possible outcome of the disease. For several years, she stayed silent.

There were deeper fears, too. She later discovered that she had lost her father to the same disease. Thoughts about inheritance, future family planning, guilt, responsibility, and possible stigma weighed heavily on her.

"It felt like carrying a huge bag full of fears."

The moment that changed everything

After years of quietly carrying her diagnosis, life brought another heartbreak. Tatevik lost members of her family during the COVID pandemic. That loss made her pause and ask herself what truly mattered and which parts of her life she still had the power to shape.



Tatevik Yezekyan's success story



In August 2020, she saw a flyer for a new initiative for young adults living with Fabry disease, organised by the Fabry International Network (FIN).

"The flyer felt like the light at the end of the tunnel. And it really became one."

For the first time, she found a space to connect, share, and listen without needing to hide behind a brave face. Community. Belonging. People who understood.

"That was my turning point. That's when everything started changing."

Finding her voice, one step at a time

She still remembers her first online meeting with the FIN Young Adults community. Saying out loud that she was living with Fabry disease felt emotional and incredibly relieving.

Another unforgettable moment came when she shared her story at the FIN Expert Meeting in 2021. She was nervous, but proud. For the first time, she realised she could speak openly about her journey without being overtaken by emotion.

There was also a quiet moment, during a patient group activity, when she spoke with someone around her father's age. For a brief second, she let herself imagine it could have been him.



Tatevik Yezekyan's success story



"That moment broke my heart. But it also gave me strength and a sense of purpose that stays with me."

From community member to mentor and advocate

As Tatevik kept showing up to meetings, to webinars, to conversations and something quietly began to shift. Fear started to turn into understanding. Learning about Fabry in a supportive environment, without pressure or overwhelm, let her see her own experience in a new light. Other people's stories opened up her own.

She began to feel unblocked. And with that came a wish to help others who might be feeling the same fear or uncertainty she once carried.

Today, Tatevik is a **Board Member** of **the Fabry International Network (FIN)** and a mentor within the **FIN Young Adults** community, supporting others as they navigate their own journeys with Fabry disease. She describes her board role as both a responsibility and a reflection of resilience.

"Being on the board is proof of the power of not giving up and finding purpose."

Through her advocacy, she works to improve quality of life for people living with Fabry, highlight unmet needs, push for equal rights, and break down stigma and invisible barriers.



Tatevik Yezekyan's success story



Advocacy that starts with empathy

What motivates her most is seeing small positive changes in people's lives; someone opening up, gaining confidence, or feeling understood for the first time.

She is especially passionate about:

- Equal rights for people living with Fabry disease
- Better recognition and support for female patients
- Stronger, more empathetic communication between doctors and patients
- Addressing social stigma and invisible barriers
- Improving patient-centred care

The power of sharing

Sharing her story helped Tatevik discover strength she didn't know she had. It taught her how much courage it takes to speak openly about something deeply personal and how much of yourself you can rediscover in the process. Her empathy. Her resilience. Her passion for helping others.

Her message to you, if you're newly diagnosed

Her advice is gentle, honest, and reassuring: "Give yourself time. Be patient with your emotions. I stayed silent for three years because that was what I needed. Everyone has their own timing."



Tatevik Yezekyan's success story



She encourages young adults to explore the Fabry community whenever they feel ready. Support doesn't always mean speaking right away, sometimes simply listening, learning, or connecting quietly is already enough.

And if there is one message, she hopes every young person living with Fabry carries with them, it is this:

"Before advocating for others, learn to advocate for yourself."

For Tatevik, learning to live with Fabry has slowly transformed fear into strength, and isolation into connection without ever letting the disease define who she is.



Tatevik is now a **Board Member of the Fabry International Network (FIN)** and also acts as a **mentor within the FIN Young Adults community**



9 Patient advocacy: your voice matters

Quick Q & A From diagnosis to advocacy



How did she react to her diagnosis?

She felt shocked, scared, and overwhelmed, especially while living abroad without close support and facing language barriers.

Why didn't she talk openly about Fabry at first?

- She needed time to understand and accept the diagnosis
- She feared judgement and being defined by the disease
- She felt unprepared to answer questions
- Personal family history made the diagnosis emotionally complex

What helped her change her perspective?

Joining the FIN Young Adults community gave her connection, education, and a safe space to share and grow.

How is she involved today?

- FIN Board Member
- Mentor within the FIN Young Adults community
- Active advocate for patient rights, awareness, and support

What has sharing her story taught her?

That vulnerability takes courage and can help others feel less alone.



9 Patient advocacy: your voice matters

Quick Q & A From diagnosis to advocacy

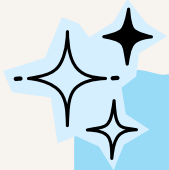


Key advice from Tatevik

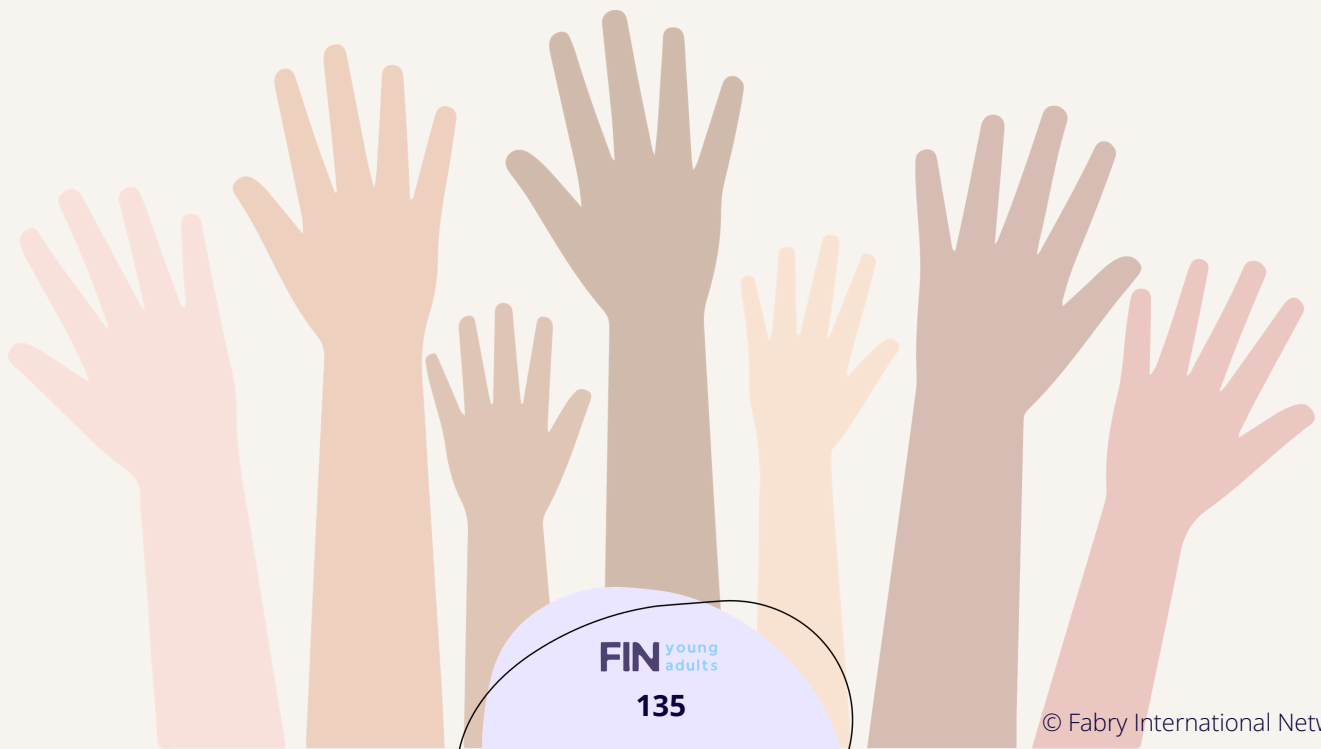


- Give yourself **time**
- Connect with others **when you feel ready**
- You are **not alone** in this journey
- Learn to **advocate for yourself first**
- Your **experience** can become your strength

9 Patient advocacy: your voice matters



*Start small. Stay curious. And remember
you're not just living with Fabry.
You're helping shape what life with Fabry
can look like for others too.*



10 Fabry disease: global initiatives, trainings & summer schools

Living with Fabry disease means navigating a complex healthcare journey, but it also opens the door to a global network of support, education, and advocacy. Across the world, there are specialized programs and initiatives designed to help young people like you become informed, connected, and empowered.

Below is a summary of key resources that offer training, leadership development, and support for young adults with Fabry disease or other rare conditions.

10.1 Community initiatives & networks

Fabry International Network (FIN) - Young Adults Community

A global platform for young people living with Fabry disease. Offers peer support, educational tools, and leadership opportunities.



[Visit website](#)



10 Fabry disease: global initiatives, trainings & summer schools

10.2 Patient advocacy & training programs

These trainings help patients, caregivers, and advocates develop knowledge and skills to actively engage in healthcare systems, research, and policy-making.

EURORDIS Open Academy Schools

Offers intensive training in rare disease advocacy, research, and policy. Participants gain skills in patient engagement and communication with stakeholders. Focused on advocacy, ethics, and clinical research. Ideal for young patient advocates ready to take their engagement to the next level.

 [Eurordis Open Academy](#)

Rare Disease Day Young Ambassador Group

Rare Diseases Europe has established the Rare Disease Day Young Ambassador Group to harness the power of youth advocates and professionals across the world in shaping the future of the rare disease movement.

 [Go to RDI Young Ambassadors](#)

RDI Youth Leadership Programme

The RDI Youth Leadership Programme is designed to engage and empower young people within the rare disease community.

 [Learn about RDI Youth Leadership Programme](#)



10 Fabry disease: global initiatives, trainings & summer schools

Rare Gene's Youth Research Program

RareGen's Youth Research Program bridges the gap between young individuals living with rare diseases and the cutting-edge scientific research needed to improve their lives.



[Check out Rare Gene](#)

Global Genes – Advocacy Training & RARE Advocacy Exchange

Provides leadership training, webinars, and annual summits to support rare disease advocates.



[Global Genes Training](#)

EveryLife Foundation – Education Series

Educational tools for patients and caregivers focusing on policy, treatments, and navigating rare disease care.



[Visit EveryLife Foundation](#)

Rare EDU (by NORD)

Free online courses for patients and professionals to deepen knowledge in rare disease research and advocacy.



[Explore Rare EDU](#)

IRDiRC – Rare Disease Research Summer School

Focuses on research, innovation, and collaboration between patients and scientists.



[Visit IRDiRC](#)



10 Fabry disease: global initiatives, trainings & summer schools

Orphanet Summer School

Covers rare disease research, diagnosis, and policy. A great opportunity for young advocates interested in scientific perspectives.



[More on Orphanet](#)

European Rare Disease Research Alliance (ERDERA)

ERDERA brings together researchers, healthcare professionals, and patient communities across Europe to advance research and improve care for people living with rare diseases. It aims to strengthen collaboration and accelerate the development of new treatments and solutions.



[Check out ERDERA](#)

European Patients' Forum (EPF)

The European Patients' Forum is an umbrella organisation that represents patient organisations across Europe. It works to ensure that patients' voices are heard in healthcare policies and supports patient involvement, empowerment, and access to quality care.



[Visit EPF](#)



10 Fabry disease: global initiatives, trainings & summer schools

European Patients' Academy on Therapeutic Innovation (EUPATI)

EUPATI provides education and training for patients and the public on medicines development and research. It aims to empower patients to engage confidently in discussions about treatments, clinical trials, and healthcare decisions.



[Check EUPATI out](#)

10.3 Mental health & emotional support

RareMinds

Offers counseling and mental health resources tailored to the emotional needs of rare disease patients.



[Visit RareMinds](#)

National Youth Advocacy Service (NYAS)

Supports youth with chronic conditions in making informed decisions about education, health, and social care.



[Explore NYAS](#)



11 Finding your community

11.1 Patient support organizations & networks

Living with Fabry as a young adult can sometimes bring questions that feel difficult to share with friends, family, or even healthcare professionals. You may feel caught between wanting independence and still needing support and this is where patient organisations can sometimes make a meaningful difference for you.

Patient organisations are communities created by and for people living with Fabry and their families. They often provide reliable information about Fabry, opportunities to connect with others who understand the condition, advocacy initiatives that help ensure patient voices are heard, and sometimes practical support such as educational events, peer mentoring, or guidance on navigating healthcare and life transitions.

Joining a patient organisation does not mean you need to be highly involved or take on responsibilities. For many people, it simply becomes a trusted place to find information, listen to shared experiences, or know that support is available if and when they need it. Some people feel an immediate sense of connection, while others may feel unsure or even overwhelmed at first. Both reactions are completely normal.

Meeting others who live with Fabry can sometimes lead to friendships, peer support, and reassurance that you are not navigating this journey alone. It can also give you the chance to learn from people who may be at a different stage of life, while sharing your own experiences whenever you feel ready.



11 Finding your community

There are many Fabry-specific patient organisations worldwide. In some countries, support may also be available through rare disease organisations or Lysosomal Storage Disease (LSD) networks. These groups often collaborate and can help connect you with additional support, resources, or community opportunities.

If you would like to explore patient organisations and support networks, you can find an overview of Fabry patient organisations around the world on the Fabry International Network website: www.fabrynetwork.org.



There is no “right” way to be part of a community. You can engage as much or as little as feels comfortable, and your needs and preferences may change over time.

You might consider reaching out if ...

- You want to meet others who understand life with Fabry
- You have questions you find difficult to ask elsewhere
- You are going through a life transition (starting work, relationships, moving out, family planning, etc.)
- You would like to learn from shared experiences
- You are looking for reliable information or resources
- You are interested in advocacy or raising awareness
- You simply want to know that support is available, even if you don't use it right away

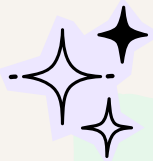


11 Finding your community

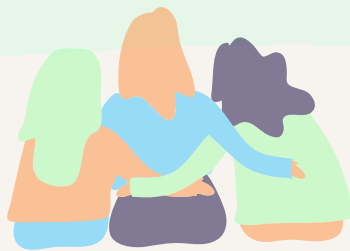
It's okay if you feel unsure

Joining a patient community can sometimes feel confronting. You may worry about being reminded of Fabry, hearing other people's experiences, or not knowing where you fit in. These feelings are very common.

You can take your time. Many organisations offer different ways to engage from reading information online, joining webinars, attending social activities, or connecting one-to-one with peers. You can choose what feels safe and comfortable for you.



Being part of a community is not about how active you are. It is simply about knowing that there is a place where people understand and support each other.



11 Finding your community

11.2 Connecting with others: The FIN Young Adults Community

Young adulthood can be a time of big transitions building independence, shaping relationships, exploring career paths, and thinking about the future. Living with Fabry during this stage can sometimes bring unique questions and experiences that are difficult to explain to others who have not lived them.

Why the panda?

Because it's rare, a little quirky, and quietly strong and somehow deeply lovable. Over time, it became our symbol: playful, resilient, and uniquely ourselves just like our community.



The FIN Young Adults community was created as a supportive space specifically for people aged approximately 18 to 32 who are living with Fabry. The community connects young adults from around the world, offering opportunities to share experiences, learn together, and support one another in a safe and understanding environment.

Within the community, FIN organises a variety of activities, including online meet-ups, educational programmes, and an annual in-person Young Adults Weekend where participants can connect, learn, and spend time together in a relaxed and supportive setting. We also develop resources created together with young adults, focusing on topics that reflect real-life needs and experiences.



11 Finding your community

For many participants, the community becomes a place where they meet others navigating similar life stages for the first time. One young adult described it as:



"It feels like the family I never knew I had."



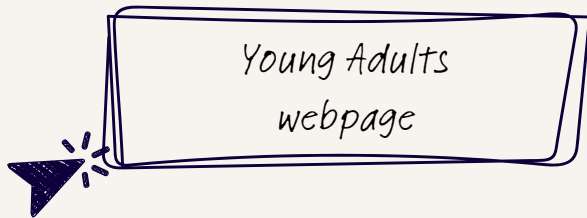
The community brings together individuals from different countries and backgrounds, creating a worldwide network of peer support, shared learning, and friendship. Some members join simply to stay informed or to know that support is available. Others choose to become more actively involved in projects, events, or community initiatives. Both approaches are equally valued.

The FIN Young Adults community is built on trust, openness and respect. There is no expectation about how involved you should be, participation can grow naturally over time, depending on what feels comfortable and meaningful for you.



11 Finding your community

If you feel curious and would like to learn more or join the community, you can find additional information and registration details here:





You might consider joining if ...

- You would like to **meet other young adults** living with Fabry
- You are curious about **shared experiences** during this stage of life
- You want to learn more through **educational** or **social activities**
- You are looking for **peer support** or **connection**
- You want to **contribute** to **projects** or advocacy initiatives
- You simply want to know that a **supportive community** exists





11 Finding your community

It's okay if you're not ready yet

Joining a community connected to Fabry can sometimes feel like a big step. You may feel unsure, curious, or hesitant all of these feelings are completely valid.

You can take your time. Some people begin by reading resources or attending an online session quietly, while others join activities when they feel ready. There is no pressure and no expectation. The community is there whenever and however it feels right for you.



"For the first time, I didn't have to explain Fabry before talking about my life. Everyone just understood."



11 Finding your community

11.3 Additional resources for daily life with Fabry

11.3.1 Various

- Medical & Scientific Information
- Mental Health & Coping
- Rare disease peer support groups (Facebook, Discord, Reddit)
- Online therapy platforms (e.g., BetterHelp, Talkspace)
- Financial & Practical Help
- Disability benefits and rare disease subsidies (varies by country)
- Support from pharma companies (assistance programs)
- Guidance from patient advocacy groups
- Lifestyle, Diet & Tools
- Registered dietitians familiar with Fabry disease
- Symptom tracking apps and self-care planners
- Legal & Work-Life Support
- Disability rights organizations
- University accessibility offices
- Unions and employee support networks
- Travel & Insurance Guidance
- Rare disease-friendly insurance options
- Medical travel tips and checklists

11 Finding your community

11.3.2 Podcasts

- “Rare Together” by EURORDIS – Conversations with patients, caregivers, and advocates from across Europe.
- “The FabryCast” by Fabry Canada - The FabryCast brings the human element of living with a rare disease. It’s a podcast for important conversations that teaches small bits of information, practical insights, and things you can do today to positively impact your life with Fabry.
- “Rare Disease Discussions” by NORD – Insightful episodes from patients, scientists, and policy leaders.
- “Patient Worthy Podcast” – Authentic patient stories on living with rare or chronic illnesses.
- “Major Pain Podcast” – Stories from people navigating difficult medical mysteries and rare conditions.
- “Uninvisible Pod” – Focused on invisible and rare illnesses, with advocacy and lifestyle tips.

The Mighty – themighty.com: Huge online community sharing stories about chronic illness, mental health, and rare disease.

SickGirlSpeaks (Tiffany Christensen) – sickgirlspeaks.com: A rare patient and speaker on navigating young adulthood with illness.

11 Finding your community

11.3.3 Videos

- **Fabry disease Patient Story – Maya Kineen**

 [Watch on YouTube](#)

- **Fabry disease: Melissa's Story**

 [Watch on YouTube](#)

- **What is Fabry disease? (Doctor explains)**

 [Watch on YouTube](#)

- **Living with Fabry – Real People, Real Stories**

 [Watch on YouTube](#)

11.4 Find what works for you

These programs and resources were created with you in mind to support your voice, your journey, and your impact. Whether you want to learn more about your condition, get involved in advocacy, explore research, or simply connect with others who get it, there's something here for you.

Take your time exploring what feels right. You don't have to do everything but you do have the power to make informed, confident choices about your path forward.

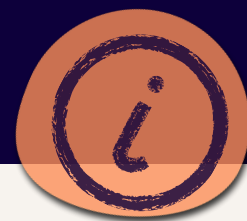
12 Final note: you are more than your diagnosis



Fabry disease is just one part of your story.
With the right information, support, and care, you can live a
full, meaningful life on your own terms.
Whether you're navigating studies, relationships, work, or
health decisions, remember that you have options, and you
have a voice.



12 Glossary



About Fabry: the basics

- **α-Gal A (alpha-galactosidase A)**

An enzyme that breaks down certain fats in the body. People with Fabry disease don't have enough of this enzyme.

- **Enzyme**

A type of protein that helps the body carry out important chemical processes, like breaking down food or other substances. In Fabry disease, the body doesn't make enough of α-Gal A, which leads to a buildup of certain fats.

- **GL-3 (globotriaosylceramide)**

A type of fat that builds up in cells when you have Fabry disease. This buildup causes damage to organs over time.

- **Sphingolipids / Lyso-Gb3**

Fats that build up in cells when α-Gal A is missing; Lyso-Gb3 is a related substance often measured in blood tests.

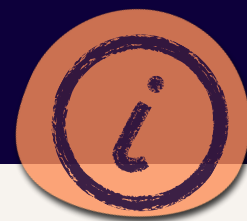
- **GLA Gene**

The gene that gives instructions to make the α-Gal A enzyme.

- **Mutation (Genetic Mutation)**

A small change in a gene that can cause disease. In Fabry, a mutation in the GLA gene affects enzyme production.
n childhood. "Non-classical" is a milder form that may appear later in life.

12 Glossary



- **Lysosomal Storage Disorder (LSD)**

A group of rare diseases where substances build up in cells because the body lacks certain enzymes. Fabry is one of them.

- **Genotype**

The specific genetic makeup someone has, the underlying gene changes, paired with phenotype (the symptoms that show).

- **Phenotype**

The observable traits or symptoms of a disease. In Fabry, this can range from mild to severe.

- **Classical Fabry / Non-classical Fabry**

“Classical” is the more severe form of Fabry, usually starting in childhood. “Non-classical” is a milder form that may appear later in life.

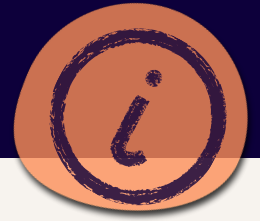
- **X-linked inheritance**

A pattern where a gene on the X chromosome is passed from parent to child; explains why Fabry shows up differently depending on biological sex.

- **Rare Disease**

A condition that affects a small number of people. Often needs specialised care and support.

12 Glossary



Diagnosis and testing

- **Enzyme Assay**

A lab test that measures how much enzyme activity you have. Used to help diagnose Fabry.

- **Genetic Testing**

A test that looks at your DNA to check for changes that might cause Fabry disease.

- **Carrier / Carrier testing**

Testing to see whether someone carries a gene change that they could pass to a child.

- **Genetic counsellor**

A specialist who helps people understand genetic conditions and what they mean for them and their family.

Symptoms: pain and neurology

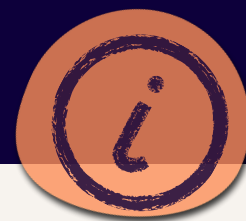
- **Neuropathic pain / Neuropathy**

Pain caused by nerves not working properly, rather than by injury or inflammation.

- **Small fibre neuropathy**

Damage to the smallest nerves; the main type of nerve pain in Fabry.

12 Glossary



- **Acroparaesthesia**

The burning, tingling or stinging pain in hands and feet that's often the earliest Fabry symptom.

- **Pain crisis (Fabry pain crisis)**

A sudden, severe episode of nerve pain, often triggered by heat, fever, exercise or stress.

- **Allodynia**

Pain from something that shouldn't normally hurt (like a light touch or cold air).

- **Postural hypotension**

Feeling dizzy or faint when standing up because of a drop in blood pressure.

- **Autonomic function / autonomic nervous system**

The part of the nervous system that controls things you don't think about (heart rate, sweating, digestion).

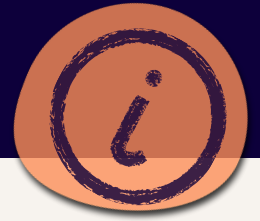
- **Fabry Fog (also called brain fog)**

A term some people use to describe forgetfulness, trouble concentrating, or mental tiredness caused by Fabry.

- **Kidney function / Proteinuria**

Kidney function means how well your kidneys work. Proteinuria means protein in the urine, which can be a sign of kidney damage.

12 Glossary



Physical signs

- **Angiokeratoma**

Small dark red or purple skin spots, common in classical Fabry, usually around the lower body.

- **Cornea verticillata**

A harmless whorl-shaped pattern in the cornea (the front of the eye) that an eye doctor can see; often a sign of Fabry.

Treatment and medications

- **Enzyme Replacement Therapy (ERT)**

A treatment where missing enzymes (like α -Gal A) are replaced through regular infusions.

- **Agalsidase alfa / beta / Pegunigalsidase alfa (Replagal / Fabrazyme / Elfabrio)**

The three currently approved enzyme replacement therapies.

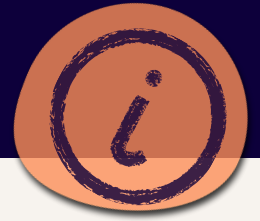
- **Chaperone Therapy**

A pill that helps your body's own enzyme work better. Only works for certain genetic mutations.

- **Migalastat**

The brand and generic name of the currently approved chaperone therapy for Fabry.

12 Glossary



- **Gene Therapy**

A new type of treatment being studied. It aims to fix the root cause of Fabry by adding a healthy gene to the body's cells.

- **Infusion**

A way to deliver medicine through a needle into your vein. Often done at a hospital or infusion centre.

- **Disease-modifying therapy**

Treatment aimed at the underlying cause of a disease, not just symptoms.

- **NSAIDs (Non-Steroidal Anti-Inflammatory Drugs)**

Common painkillers like ibuprofen; need extra caution with Fabry because of kidney impact.

- **Tricyclic antidepressants**

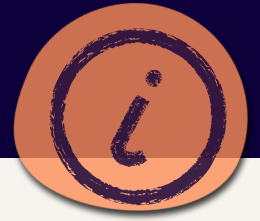
A class of older antidepressants sometimes used in low doses for nerve pain.

Family planning

- **In Vitro Fertilization (IVF)**

A method of conceiving outside the body.

12 Glossary



- **Preimplantation Genetic Diagnosis (PGD)**

A test done on embryos created through IVF to check for specific genetic conditions before pregnancy.

- **Chorionic Villus Sampling (CVS)**

A prenatal test that takes a small sample from the placenta to check for genetic conditions.

- **Amniocentesis**

A prenatal test that takes a small sample of fluid from around the baby.

Healthcare, advocacy and rights

- **Multidisciplinary Team**

A healthcare team made up of doctors, nurses, therapists, and other specialists who work together to support you.

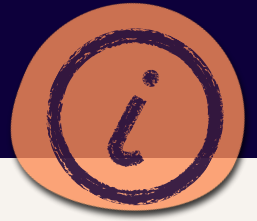
- **Informed Consent**

Agreeing to medical treatment or participation in a study after understanding all the facts and risks.

- **Clinical Trial**

A research study where people try new treatments to see if they work and are safe.

12 Glossary



- **Support Group**

A group of people who meet (online or in person) to share experiences and help each other cope with challenges.

- **Self-advocacy**

Speaking up for your own needs, asking the right questions, and making sure your voice is heard.

- **Reasonable accommodations**

Adjustments at school or work (flexible hours, remote options, extra time) that allow someone with a health condition to perform fully.

- **Disability status / disability recognition**

Formal legal recognition that someone has a condition that qualifies for protections and supports; the criteria vary widely by country.



DISCLAIMER



This guide has been developed by Fabry International Network (FIN) in close collaboration with members of the Fabry community, healthcare professionals, and expert contributors. It reflects shared knowledge, lived experiences, and expert perspectives available at the time of publication.

Our goal is to provide supportive, accessible, and community-informed information. However, Fabry disease affects individuals differently, and medical knowledge and recommendations continue to evolve. The content in this guide is intended to support awareness, understanding, and conversation, but it should not be considered medical, legal, or psychological advice.

Decisions regarding treatment, health management, and lifestyle choices should always be made in consultation with qualified healthcare professionals who are familiar with an individual's medical history and personal circumstances.

References to organisations, services, or resources are included to support awareness and connection within the Fabry and rare disease communities. Inclusion does not represent endorsement, and readers are encouraged to explore resources that best meet their personal needs.

FIN is committed to supporting and empowering the Fabry community through collaboration, shared learning, and trusted information. Readers are encouraged to use this guide as a starting point for further conversations, support, and connection.

About Fabry International Network (FIN)

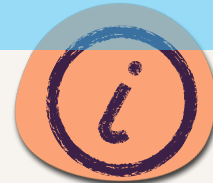


Fabry International Network (FIN) is a global patient-led umbrella organisation that connects Fabry patient organisations, individuals, families, healthcare professionals, and partners around the world.

FIN works to strengthen the global Fabry community by creating opportunities for connection, shared learning, and collaboration. We support patient organisations across different countries while developing educational resources, community programmes, and international initiatives that help ensure the voices and experiences of people living with Fabry are heard and represented.

A central part of our work is bringing the community together creating safe and supportive spaces where individuals and families can share experiences, learn from one another, and access reliable information. We work closely with healthcare professionals, researchers, and rare disease partners to help improve awareness, understanding, and support for people living with Fabry worldwide.

About Fabry International Network (FIN)



Our Commitment

FIN is committed to building a trusted and inclusive global Fabry community by:

- Supporting connection between individuals and patient organisations
- Providing educational and community-driven resources
- Creating safe spaces for shared experiences and peer support
- Promoting patient voices in advocacy, research, and healthcare discussions
- Working collaboratively with the global Fabry and rare disease community

Staying Connected & Contact

If you would like to learn more about FIN, explore resources, or connect with patient organisations worldwide, you can visit:

- Website: www.fabrynetwork.org
- Email: coordinator@fabrynetwork.org
- Social Media:



If you are looking for support in your region, FIN is happy to help connect you with local patient organisations and community resources.

A final thought[!]

Wherever you are in your journey with Fabry,
you are part of a global community.
Support, knowledge and connection are there
whenever you feel ready to reach out.

