



PAIN IN FABRY



Thousands of people around the world live with Fabry disease, but no two people experience it in exactly the same way. In addition, some of the symptoms of Fabry may seem “invisible” and difficult to measure through medical tests. This may be frustrating, and can possibly compromise the trust between people with Fabry and their healthcare providers (HCPs).^{1,2}

Pain is one of those symptoms invisible to others, and is one of the most common symptoms of Fabry, affecting almost 75% of those living with the disease.³ It is also usually one of the earliest effects of the disease, often starting in childhood—up to 90% of children with Fabry have pain.⁴

Even though most people with Fabry have some kind of pain, their experiences can be very different. This fact sheet will outline how and where people with Fabry might experience pain and might even help empower people to have productive discussions with their HCPs about pain.

What does Fabry pain feel like?

Burning. Stabbing. Tingling. Agonizing. These are the most common ways that people with Fabry describe their pain.^{4,5}

Some people also say that it is pressing, prickling, squeezing, electrifying, or feels sore.⁶

In addition to many descriptions of pain, there are different ways that people experience pain, and in different parts of the body, as shown on the next page. Researchers have described four main types of Fabry-associated pain.⁶

PAIN ATTACKS



Sudden, intense pain that does not last very long

Experienced by almost 47% of people who have pain with Fabry

PAIN CRISIS



Intense pain that can last for hours or even days

Experienced by more than 17% of people who have pain with Fabry

EVOKED PAIN



Pain caused by something that would normally cause no pain or only slight pain, like a light touch or hot water

Experienced by 66% of people who have pain with Fabry

CHRONIC PAIN



Pain that is always present

Experienced by more than 17% of people who have pain with Fabry

Many people don't have just one type of pain, but different types at different times, or at the same time.⁶ How often people experience pain can vary too. Most people have pain one to four times per month, but some people report pain daily, weekly, monthly, or only a few times a year.⁶ This can change over time: for many people, pain becomes less severe with age, possibly because the longer someone has Fabry, the more damage there is to their nerves, which may make them less able to feel pain.¹ But it does not get better for everyone.

What makes pain happen?

Pain attacks and pain crises can be set off—or “triggered”—by a number of things in everyday life. These include:



acute illness, such as the flu or other viral illness



emotional stress



high or low temperature or a rapid temperature change



exercise



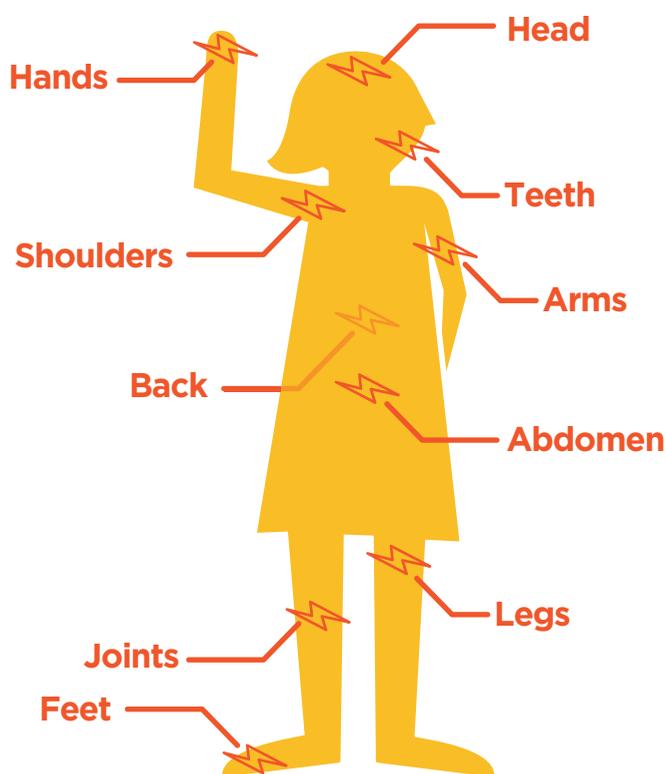
fatigue



certain foods, such as coffee and alcohol^{4,5}

Where do people with Fabry feel pain?

The location of pain is yet another thing that can change from person to person. The most common locations for pain are hands and feet, affecting more than 65% of people who have pain with Fabry.³ Severe pain in the hands, feet, fingers, and toes is called “acroparesthesia” (pronounced ack-roe-pare-us-THEE-zha).⁵ People report pain in many areas besides hands and feet—some in their whole body.⁶



Effects of pain

! Pain hurts physically in a variety of ways, and can also have a bigger effect on daily life. People experiencing significant pain might need to avoid physical activities, or limit daily activities such as working, socializing, or even going to school.^{3,4} Partly because of these limitations, overall quality of life can be lower for people with Fabry who experience pain.⁴ And sometimes pain can be so bad and so constant that it affects mood and state of mind.^{4,5,7}

What can be done about pain?

? If people know what their pain triggers are, of course they can try to stay away from them. For example, people with Fabry might want to avoid extreme temperatures, physical exertion, or caffeine if they notice an onset of pain associated with those factors. A cooling vest may help people keep from being overheated. Some people use prescription medications to manage pain⁸; HCPs can help choose the right medication and pain management plan.

One of the most important things people with Fabry can do is make sure to let their doctors or other HCPs know exactly how, when, and where they are experiencing pain. Researchers have developed a specific questionnaire about pain in Fabry. You can read about this questionnaire in the article “Self-administered version of the Fabry-associated pain questionnaire for adult patients” by Barbara Magg and colleagues, published in the *Orphanet Journal of Rare Diseases* in 2015. You can find information about this article on PubMed: <https://goo.gl/biPD4H>.⁹ This questionnaire could help people experiencing pain explain it and help HCPs understand it, so they can help manage pain along with the other effects of Fabry. Never be afraid to speak up about what you or a loved one is going through!

For those looking to learn as much as possible about Fabry and its effects, the links available at http://amicusrx.com/external_resources.php may be useful.

REFERENCES: 1. Gibas AL, Klatt R, Johnson J, Clarke JTR, Katz J. A survey of the pain experienced by males and females with Fabry disease. *Pain Res Manage.* 2006;11(3):185-192. 2. Gibas AL, Klatt R, Johnson J, Clarke JT, Katz J. Disease rarity, carrier status, and gender: a triple disadvantage for women with Fabry disease. *J Genet Couns.* 2008;17(6):528-537. 3. Hoffmann B, Beck M, Sunder-Plassmann G, et al. Nature and prevalence of pain in Fabry disease and its response to enzyme replacement therapy: a retrospective analysis from the Fabry Outcome Survey. *Clin J Pain.* 2007;23(6):535-542. 4. Hoffmann B. Effects of enzyme replacement therapy on pain and overall quality of life. In: Mehta A, Beck M, Sunder-Plassmann G, eds. *Fabry Disease: Perspectives From 5 Years of FOS.* Oxford, UK: Oxford PharmaGenesis; 2006. 5. Mehta A, Hughes DA. Fabry disease. In: Pagon RA, Adam MP, Ardinger HH, et al, eds. *GeneReviews*® [Internet]. Fabry disease. Seattle, WA: University of Washington; 2002 [Updated October 17, 2013]. 6. Üçeyler N, Ganendiran S, Kramer D, Sommer C. Characterization of pain in Fabry disease. *Clin J Pain.* 2014;30(10):915-920. 7. Wang RY, Lellis A, Mirocha J, Wilcox WR. Heterozygous Fabry women are not just carriers, but have a significant burden of disease and impaired quality of life. *Genet Med.* 2007;9(1):34-45. 8. Schuller Y, Linthorst GE, Hollak CEM, Van Schaik IN, Biegstraaten M. Pain management strategies for neuropathic pain in Fabry disease – a systematic review. *BMC Neurol.* 2016;16:25. 9. Magg B, Riegler C, Wiedmann S, Heuschmann P, Sommer C, Üçeyler N. Self-administered version of the Fabry-associated pain questionnaire for adult patients. *Orphanet J Rare Dis.* 2015;10:113.