CFA
CANADIAN FABRY ASSOCIATION
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NEWSLETTER

News from Idorsia Pharmaceuticals based in Switzerland

For those of you not in the know, Idorsia is the new kid on the block who has joined our fight against Fabry. The people at Idorsia are not so new to Fabry though as they used to work for Actelion Pharmaceuticals who started the work. When Actelion was bought by Johnson & Johnson the researchers took their project to Idorsia to continue their work.

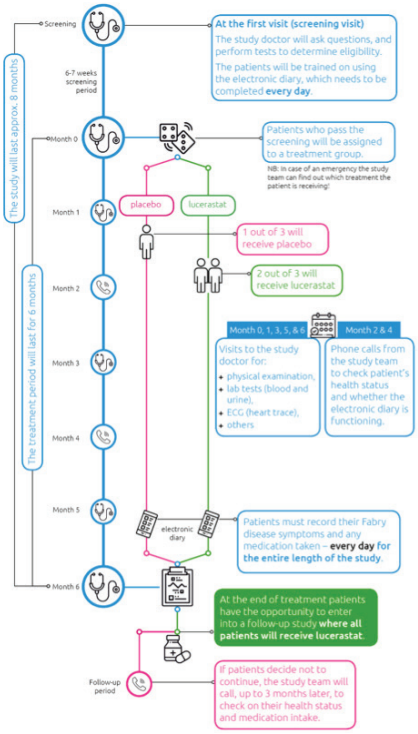
Less than one year later they started "MODIFY" a global clinical research Phase 3 study (that's the last stage in clinical development) for patients with Fabry disease. The purpose of the study is to test whether lucerastat – the new oral investigational medication – can reduce symptoms of Fabry disease, such as pain and stomach problems and to confirm that it is safe to take.

Read on for more information about the study…
MODIFY will enroll over 100 patients who will be split into two groups – two thirds of patients will enter group one and receive lucerastat and the remainder of patients will enter group two and receive placebo. MODIFY is a 'double-blind' study, which means that no one will know who will receive lucerastat or placebo, and who gets what will be decided randomly – like rolling dice.

Lucerastat is taken twice daily in the form of oral capsules.

The MODIFY study is currently open for participation in USA, Canada, UK, The Netherlands, Czech Republic, Germany, and Austria. In addition, sites in Australia, Poland and France are pending approval of their Ethic Committees / Institutional Review Boards.

Want more information?
Patients can find more study details on www.clinicaltrials.gov – using 'lucerastat' or 'NCT03425539' to find the MODIFY study in the list – or speak to their doctor.



So, how was MODIFY designed?
The study is based on an international patient survey (with 367 patients) conducted by Idorsia to better understand the symptoms of patients with Fabry disease. The design also benefitted from input from patient organizations, specialists, and regulatory agencies.

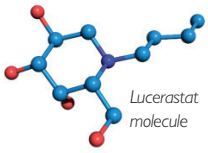
The survey found that 50% of patients with Fabry experience significant "neuropathic pain" which is moderate to severe in intensity, frequent, and located in the hands and feet. This makes it a good marker of the disease that can be measured in a clinical trial. The survey also found that neuropathic pain causes a big impact on a patients' quality of life.

What is neuropathic pain?
a type of pain which feels like burning, shocks or shooting, stabbing, tingling, and/or pins and needles in the hands and feet. …sound familiar?

MODIFY – Who Can Take Part?
Lucerastat is being studied as a monotherapy. As a result, the study is enrolling patients who were never treated with enzyme replacement therapy (ERT), patients who were treated with ERT in the past but stopped it at least 6 months prior to study enrollment, and patients who are currently treated with ERT (for more than 12 months) and agree to stop receiving it for approximately 8 months.

"Since the main aim of the study is to see whether lucerastat reduces neuropathic pain in adult patients with Fabry disease, to enter the study, patients must have genetically confirmed Fabry disease, – irrespective of the genetic mutation! – have neuropathic pain, and be over the age of 18. Importantly patients must agree to complete a daily electronic diary." says Aline Frey, one of the team that is working on the study from Idorsia. "Study physicians will ask questions and perform some health checks to see whether patients are eligible for the study."

How does lucerastat work?

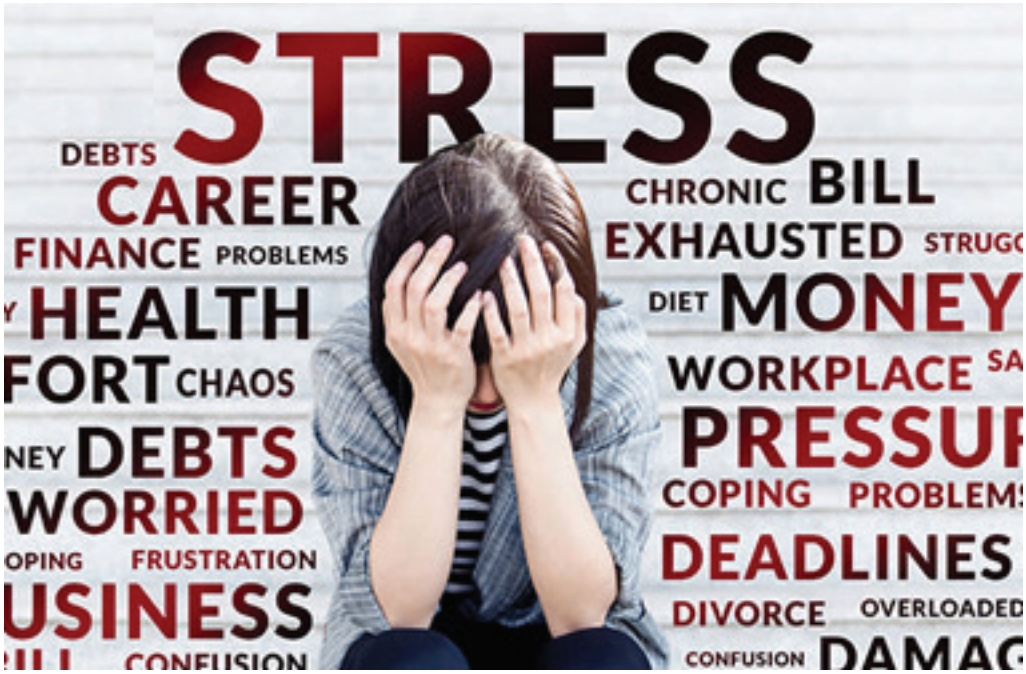

Lucerastat molecule

Fabry disease is a rare inherited disorder in which a particular lipid (a fatty substance) can't be broken down by the body because of a dysfunctional or absent enzyme.

This leads to a build-up of the lipid in the cells of the body organs which results in cell and organ damage. In lab tests, lucerastat blocks an enzyme which is responsible for making the lipid in the first place – this means that the harmful accumulation of the lipid should not occur.

DR. KANWAL'S CORNER

ALLEVIATING THE UBIQUITY OF STRESS



Dr. Seema Kanwal
Board Member

Stress is ubiquitous. Everyone experiences it, almost anything in the environment can cause it (even the internal mental environment), and our bodies have myriad ways of dealing with it. When our bodies are under physical duress or perceived mental distress, our bodies undergo a physiological stress response. Whether the demand is stemming from sitting in an uncomfortable chair in an office all day, worrying about how the next bill is going to be paid, or dwelling about negative events that happened during the day, the response of the body is the same.

So what happens when the body undergoes stress? Hormones, including cortisol and epinephrine, are produced by the adrenal glands. Those hormones are fed into the bloodstream, their purpose being to prepare the body to fight perceived dangers by increasing heart rate and blood pressure and making more energy available for the body to use as fuel.

When physical action is necessary to escape danger, outrunning a large predator for example, that response cascade is exactly what we would have used to escape and save ourselves. Unfortunately, the modern world does not offer much of an outlet for those physiological changes. Thus, with little or no opportunity to vent our stress physically, the body turns the response inward and reduces the effectiveness of our organ systems. That results in a strain on our organisms and causes the symptoms of stress to manifest.

Those symptoms? Fatigue (throughout the day and compounded by a difficulty falling asleep), anxiety, insomnia, perceived body pains or aches, food cravings, weight gain, hair loss, inability to focus, extreme tiredness after exercise, decreased libido, and decreased immune function. That isn't even including the potential increased risk of disease. Chronic stress has been shown to increase the risk of ulcers, heart disease, and even cancer.

There is also a key link between irritable bowel syndrome (IBS) and stress. The underlying chronic stress caused by the IBS can make it worse which can then further compound the already existing digestive issues even more. It's a vicious cycle which can resemble the "chicken and egg" metaphor. One makes the other worse which, in turn, makes the first worse. Stress causes hormones to be released leading to decreased levels of good gut bacteria and inhibits digestion further, causing more stress.

The key to overcoming stress is to recognize its source, understand the physiological response your body is having to it, and pivoting your thoughts and actions to help mitigate it. Because stress can be so dangerous and debilitating, it is important for every one of us to learn how to deal with it effectively as it occurs and, ideally, prevent or reduce its occurrence in the first place. Finding ways to deal with stress, be it through exercise, meditation, or a slew of other relief options is essential. Avoidance doesn't solve anything. Finding stress sources and relieving them, however, does.

For more information, contact Dr. Seema Kanwal at drseema@fabrycanada.com

FABRY PATIENT SURVEY

****An honourarium will be sent to each participant for their time, as well as be entered into win a $150.00 Keg Steakhouse Gift Certificate.

We are seeking individuals with Fabry Disease, and caregivers of individuals with Fabry Disease, to participate in a market research study. The research will aim to gain a better understanding of patients' experiences and opinions about:

- Their journey from initial symptoms to present day
- The impact of Fabry Disease on their lives
- Their awareness and perceptions of current and future treatments

The feedback will be used to help future patients and their caregivers. If you qualify for the study, you will be asked to complete an online survey. You will receive an honorarium check for your time and participation. All information and responses will remain confidential.

INTERESTED IN PARTICIPATING? WANT MORE INFORMATION?

To see if you qualify or to get more information please contact Eva Woo at eva@crcresearch.com

HARVARD RESEARCH STUDY

Do you or someone you know have Fabry disease? Are you treatment naïve or been on treatment for less than 6 months?

Do you have any gastrointestinal symptoms such as abdominal pain, diarrhea or nausea?

If yes, you may be eligible to participate in a research study on people with Fabry disease.

Who? Adults over 18 years old with Fabry disease who have gastrointestinal symptoms

Why? To learn more about the gastrointestinal symptoms in Fabry disease

How? To learn more about this study, contact the study coordinator at Massachusetts General Hospital at mnalmeida@mgh.harvard.edu or at 617-724-0480.

This study is voluntary. Participants will be compensated for their time. The study will use questionnaires to learn more about your symptoms and a device to study the movement of contents through your gut.

Funded by Genzyme Therapeutics to Braden Kuo, MD, Principal Investigator, Massachusetts General Hospital.

Massachusetts General Hospital

Harvard Medical School

BE READY FOR RARE DISEASE DAY

ORDER YOUR TATTOO PACKAGE TODAY!!

Lori Culum
"Be Rare. Be You." Program Manager

Rare Disease Day (February 28, 2019) is fast approaching. Be ready to show your support and raise awareness of the 1 in 12 Canadians being affected by rare disorders.

New English and French tattoo packages have been created for this year's campaign. The information card contains some important facts about rare disorders in addition to how you can show your support on social media. The two tattoos included with the information card incorporate a bilingual message – "Be Rare. Be You." "Sois Rare. Sois Toi.". This year we branded the tattoo with the CFA light and dark blue colours.

An Opportunity For You – visit fabrycanada.com and renew your 2019 membership by February 7, 2019 and you will receive one complimentary tattoo package in the mail. Choose "Member Login" to proceed to renew your membership. If you do not have a Member Login, you can create one and then proceed to pay your membership. Membership fees help to support initiatives such as the Empowerment meetings, website updates, campaigns, etc.

If you wish to receive more than one tattoo package, simply renew your membership and then go to the "Home" page to place an order for the number of packages that you would like to receive – and then we'll include one for free! If you wish to order a French package, remember to click on "Francais" in the top right corner of the Home page. Packages cost $5 for family members and friends and $10 for industry/companies.

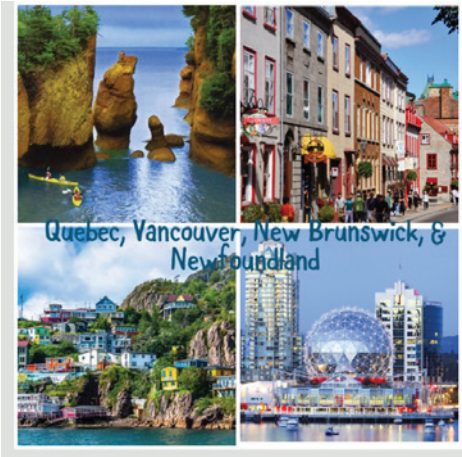
We look forward to seeing you and your tattoo on social media on Rare Disease Day. If you have ideas on how to support this program, questions or wish to place a larger order, you can contact me directly at lori.culum@fabrycanada.com.

Your Support = Successfully Spreading the Message!!

Thank you!

PATIENT EMPOWERMENT SEMINARS 2019

Julia Alton
Executive Director



The CFA continues to spread empowerment throughout the provinces by educating the Canadian Fabry community on the latest therapies, research, and clinical trials. Together as a community we are stronger, and want to give you the opportunity to share stories, ask questions, and meet others living with Fabry.

2019 Locations:
Laval, Quebec - April 27
Vancouver, British Columbia - October 19
New Brunswick - (City & Date TBD)
Newfoundland - (City & Date TBD)

Stay tuned through our social media platforms, member emails, and newsletters for dates and further details.

Donations to the CFA are greatly appreciated

In memory of
Graham Harnish
Mahone Bay, Nova Scotia

THANKS TO OUR SUPPORTERS

We would like to thank all of our supporters that helped make this newsletter possible.

We receive financial support from these Pharmaceutical companies who are currently providing hope for Fabry patients through their research and the products they provide.

Amicus Therapeutics
AVROBIO
genzyme
idorsia
Shire
PROTALIX Biotherapeutics

We would also like to thank all of the physicians, specialists and medical professionals that have helped in so many ways. From providing guidance on medical terms and details to caring for members of our community every day.

And of course we would like to thank all of the patients and family members that have volunteered their time and energy to assist in all the many ways that are necessary in the creation of such a large effort. It is through their efforts that we hope to inform and build a community of Fabry patients for the benefit of patients, their families and caregivers.

MAKE A DONATION

Would you or a family member like to make a donation so that we can continue to educate and advocate for the best treatment as well as communicating with and for Fabry patients in Canada?

The Canadian Fabry Association (CFA) is a registered not-for-profit organization.
If you are interested in making a charitable donation and would like a tax receipt, please make your cheque payable to The Fabry's Charity Association.

100% of donations to the CFA are used to promote education, patient support and access to treatment for Canadian Fabry patients. You can make donation cheques payable to The Fabry's Charity Association and mail the cheque to us.

Send the cheque to:
The Fabry's Charity Association
748 Kelly Street
Thunder Bay, ON
P7E 2A1
or register online by visiting our website:
www.fabrycanada.com

Thanks for your donation to the CFA! It goes to help Canada Fabry patients, their families and caregivers.