



CANADIAN
FABRY
ASSOCIATION

volume 10 issue 2
spring 2019

NEWSLETTER

in this issue: | Rare Disease Day Campaign | FIN Photo Contest | June Webinar | Mandie's Story | AVROBIO plato™ Platform | The WORLD Symposium | Dr. Kanwal's Corner | Carlton LSD Study | NF Patient Empowerment Seminar

2019 RARE DISEASE DAY CAMPAIGN

Julia Alton
Executive Director

2019 Rare Disease Day Photo Collage - look close to find your picture! The message "Be Rare Be You" reached 1000's and was worn in 16 Countries! Thank you to everyone who participated and look forward to what we can do together next year!

A special thank you to our Campaign Manager Lori Culum for her drive, dedication, and countless hours she put fourth!

If you would like to get involved in next years campaign - please reach out to Lori.culum@fabrycanada.com



PATIENT EMPOWERMENT QUESTION:

Q. How many Canadians have been diagnosed with Fabry Disease?

FABRY AWARENESS MONTH - PHOTO CONTEST

Each year, during April, we aim to increase the awareness of Fabry Disease by increasing understanding of this rare condition and impact of living with Fabry Disease as a patient and family.

Fabry Awareness Month is all about telling the world about this rare disease. The more people that are aware, the more patients we can help get that early diagnosis. We hope that this year you also join us in sharing details about Fabry Disease with your family, friends and doctors!

To celebrate **Fabry Awareness Month** Fabry Board (FIN) is organising a photo contest! Fabry & My Future. Send us a picture during the month of April and let us know how you see your future! Share with us how you make your life valuable every day, despite daily pain or other discomforts. Tell us a bit more about yourself and your picture.

A contest without a prize would not be a contest, so we will choose three pictures and announce the winners at the end of April and make sure the winner receives a nice prize!

Make sure the picture you send in, is a high resolution picture as we plan to print the picture and also present it during our annual Expert Meeting in Barcelona!

Send the picture to: info@fabrynetwork.org before the end of April 2019.

EST 1995

WEBINAR WEDNESDAY

DATE June 12, 2019

SUBJECT New therapies for Fabry Disease

TIME 4pm PDT/ 5pm MDT/ 6pm CDT/
7pm EDT/ 8pm ADT/ 8:30pm NDT

SPEAKER Dr. West

Find instructions to join this
Webinar at
www.fabrycanada.com

MANDIE'S STORY



Let me introduce myself, my name is Mandie. I am a 38-year-old wife, mother of three beautiful daughters, a writer, poet, and artist. I am also a rare disease patient.

Wow, those are a lot of words! Had I introduced myself to you last year, I would have never mentioned being a rare disease patient. Fabry's disease was a little secret I had grown up with, we kept it from the world.

My mother has Fabry's, and she spent her life watching it destroy her family. She lost all her uncles on her dad's side to heart attacks linked to Fabry's. She watched her own father become disabled and confined to a wheelchair. She even spent time visiting him in hospitals from behind glass walls as doctors tried to understand and gain knowledge about this disease. After he died, and after I was diagnosed just after birth, my mom fashioned us a cage from her childhood traumas. That is where we hid all our own

complications and symptoms, never talking to anyone about it, keeping it a secret of sorts.

As I mentioned, I am a mother of three daughters, who have all been witness to the kind of savagery this disease can wield. Our oldest was diagnosed with Fabry's roughly 12 years ago, when she was around eight. Our middle and youngest daughters were tested in the late winter last year, their results came in January of this year, and they both have Fabry's.

Like any parent would be, we were shattered, and we had to wade through some deep emotions like regret and grief. But I had to wrestle with the idea that my cage wasn't an option anymore. I don't want them to ever feel alone or different, or not worthy of true human intimacy based on a diagnosis. And the only way I knew how to teach them that was to open my own cage. So, I got brave with myself and for our girls.

I booked a tattoo appointment to have the "Be Rare, Be You" and the DNA link tattooed on my wrist. And I took to my Instagram to do stories about our disease and to record the daily symptoms we live through. The tattoo was the most impactful way I could come up with for opening my cage and walking in the bravery it takes to be recognized as a rare disease patient, we say warrior in our house. It reminds me daily of my responsibility to push through my rough days, to educate myself and others, to be a role model for our kids and other people, and it starts a conversation as all tattoos do. I've embraced it and wear it as a badge of honour.

Hi, my name is Mandie and I'm a rare disease warrior.

DR. KANWAL'S CORNER INSECT BITES AND SUNBURN

Dr. Seema Kanwal
Board Member

Summer is fast approaching. This is the time we start preparing our gardens for our parties, however, the summer can bring for us insect bites and sunburns. The sun has been long missed however the rays of the sun can age our skin, give us more wrinkles, fine lines and age spots. There is also the increased risk of skin cancer. The good news though there is many natural ways to prevent insect bites, and sun burns.

I. Preventing Insect bites and helping the itch. One of the best methods is to use essential oils to prevent insect bites. You can add them into distilled water and spray them onto your body or you can add them to Grape seed oil or Olive oil and rub into your body. Some of my favorites and that work well are Citronella oil, Tea tree oil, catnip oil, Basil oil, Eucalyptus oil and Thyme oil. Insects tend to be attracted to dark clothing so try and wear light colors. My all time favorite is vitamin B1, thiamine. It is very effective in warding off mosquitoes. Now if you have tried all of these, and still get a bite, try these for the itch:

- A.** Crush an aspirin tablet, wet it, and rub it on the affected area. Aspirin is an anti-inflammatory.
- B.** Rub the inside of a plantain banana on the affected area. It works great to relieve the itch.

2. Preventing sunburns. I get asked a lot by patients about this as there is a lot of confusion as to the labeling of sunscreen. Manufactures are now required to label an SPF number, which indicates how much protection it provides against UVB radiation, the cause of sunburn. However consumers did not know whether the same bottle would protect against UVA, the causer of skin cancer. Here are nutrients that you can add daily to help prevent sunburns.

- A.** Grape seed Extract, which are from Grape seeds, are the best antioxidant in my opinion.
- B.** Resveratrol, which is found in skin of red grapes and red wine.

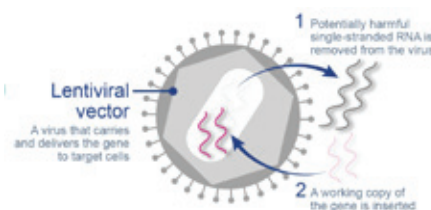
Natural remedies work great for the whole family and of all ages. Remember prevention is key. Keep your immune system strong and working daily.

AVROBIO announces plato™ gene therapy platform

Fernanda Copeland
Avrobio

AVROBIO, a leader in lentiviral-based gene therapies, is a clinical stage company developing disruptive therapies that have the potential to transform patients' lives in a single dose.

Gene therapy involves the transplantation of normal genes into cells in order to correct genetic disorders. Gene therapy uses a vector (see illustration below) to carry and deliver a working copy of a gene to the body's cells. A vector is a deactivated virus that is unable to reproduce and spread (#1 below). There are different types of vectors. **AVROBIO's plato™** is a gene therapy platform which uses a lentiviral vector and is designed to target underlying genetic disease by restoring normal gene function (#2 below) and enzyme production.



For more information about AVROBIO's pipeline and technology please visit <https://www.avrobio.com/>.

WORLD SYMPOSIUM

Julia Alton
Executive Director

The WORLD Symposium is an annual research conference dedicated to lysosomal diseases. WORLD is an acronym for "We're Organizing Research on Lysosomal Diseases." This international research conference attracts more than 1,600 participants from over 50 countries around the world.

WORLD is not only a great opportunity to connect with partners around the world, but also an opportunity to look at how we can work more closely to improve the lives of the communities we serve. Sessions included gene therapy, chaperone therapy, substrate reduction therapy, enzyme replacement therapy, newborn screening, and other high level science that is taking place.

We have some of the best scientific minds working on new treatments for rare diseases and we are pleased to be part of their dedication to finding a way to create a better future for the rare disease community.

REMEMBERING FRIENDS

Would you like to have a note of remembrance included in our newsletter? These notices would be for Fabry Patients and Association Members whom we have lost over the years. Please contact us at: secretary@fabrycanada.com

CARLTON UNIVERSITY

INVITATION TO PARTICIPATE IN RESEARCH STUDY FOR CANADIAN PATIENTS AND FAMILIES WITH LYSOSOMAL STORAGE DISEASES (LSDs)

THIS STUDY IS PART OF THE PHD PROGRAM AT THE CARLETON UNIVERSITY

Invitation to Participate in Research Study for Canadian Patients and Families with Lysosomal Storage Diseases (LSDs)

Study Title:

Access to Orphan Drugs in Canada: Integrating Patient and Policy Perspectives



Dear patients and families with LSDs:

We are conducting interviews as part of a research study to increase our understanding of the lived experiences of patients and families with LSDs with the management of their disease and navigation of health systems in Canada.

Participation:

As a Canadian patient/family carer living with LSD and older than 16 years of age, you are in an ideal position to give us valuable information from your own perspective.

The interview takes around 60 minutes and will be conducted in-person or by phone (using skype, facetime, or any other means that suits you best). Your responses to the questions will be kept confidential. Each interview will be assigned a number code to help ensure that personal identifiers are not revealed during the analysis and write up of findings.

There will be a small greeting gift (\$15 gift card) as a compensation for participating in this study.

Objective of the study:

Your participation will be a valuable component to our research and findings to support establishing a patient-focused orphan drug review approach which may improve the life and health of millions of Canadian patients affected with rare diseases by engaging them in the decision-making processes and ensuring consistency, transparency, and timely access to treatment (which is critical for patients with rare diseases).

If you are willing to participate, please suggest a day and time that suits you and connect with me by email at nahyaawada@cmail.carleton.ca or phone/text (613) 2950622.

Finally, if you would like to raise any concerns or have questions on the interview and overall research processes, please contact me and/or my supervisor.

Interviewer/Researcher Name: Nahya Awada

Affiliation: PhD Candidate, School of Public Policy and Administration, Carleton University

Email: nahyaawada@cmail.carleton.ca **Phone:** +1(613)2950622

Supervisor name: Paul Peters

Affiliation: Associate Professor, Department of Health Sciences, Carleton University

Email: paul.peters@carleton.ca **Phone:** (613) 520-2600. Ext. 2134

Study Title:

Access to Orphan Drugs in Canada: Integrating Patient and Policy Perspectives (Study #110105).

Dear patients and families with Lysosomal Storage Diseases (LSDs):

We are conducting interviews as part of a research study to increase our understanding of the lived experiences of patients and families with LSDs with the management of their disease and navigation of health systems in Canada.

Participation:

As a Canadian patient/family carer living with LSD and older than 16 years of age, you are in an ideal position to give us valuable information from your own perspective.

The interview takes around 60 minutes and will be conducted in-person or by phone (using skype, facetime, or any other means that suits you best). Your responses to the questions will be kept confidential. Each interview will be assigned a number code to help ensure that personal identifiers are not revealed during the analysis and write up of findings.

There will be a small greeting gift (\$15 gift card) as a compensation for participating in this study.

Objective of the study:

Your participation will be a valuable component to our research and findings to support establishing a patient-focused orphan drug review approach which may improve the life and health of millions of Canadian patients affected with rare diseases by engaging them in the decision-making processes and ensuring consistency, transparency, and timely access to treatment (which is critical for patients with rare diseases).

If you are willing to participate, please suggest a day and time that suits you and connect with me by email at nahyaawada@cmail.carleton.ca or phone/text (613) 2950622.

Finally, if you would like to raise any concerns or have questions on the interview and overall research processes, please contact me and/or my supervisor.

Interviewer/Researcher Name: Nahya Awada

Affiliation: PhD Candidate, School of Public Policy and Administration, Carleton University

Email: nahyaawada@cmail.carleton.ca

Phone: +1(613)2950622

Supervisor name: Paul Peters

Affiliation: Associate Professor, Department of Health Sciences, Carleton University

Email: paul.peters@carleton.ca

Phone: (613) 520-2600. Ext. 2134



Carleton
UNIVERSITY

Canada's Capital University



DONATIONS AS MEMORIAM

We have been asked if they can make a donation to the Fabry's Charity Association as a Memoriam for their family member. The answer is Yes. Please contact us at: secretary@fabrycanada.com

RENAL RECIPE

NO BAKE ENERGY BITES

PREPARATION: 10 MINUTES
COOKING: 0 MINUTES
SERVINGS 8

from *Spice It Up!*
Giving Zest to your Renal Diet
www.myspiceitup.ca

INGREDIENTS

- ½ cup natural peanut butter*
- ½ cup liquid honey
- 1 tsp vanilla
- ¼ cup ground flax
- 2 cups rice crisp cereal
- * You can also make this with regular peanut butter.

PREPARATION

- 1 To a mixing bowl, add peanut butter, honey and vanilla. Microwave for 30 seconds.
- 2 To the same mixing bowl, add ground flax and rice crisp cereal.
- 3 Mix well to combine.
- 4 Chill rice crisp mixture for 10 minutes in the freezer.
- 5 Roll into 16 balls.
- 6 Cover and place in a serving container. Refrigerate.



NUTRIENT ANALYSIS

PER SERVING: 2 Balls

RENAL EXCHANGE:

2 Starch	
Calories	213Kcal
Protein	5g
Carbohydrates	29g
Fibre	2.5g
Total Fat	10g
Saturated Fat	1.3g
Cholesterol	0mg
Sodium	41mg
Potassium	157mg
Phosphorus	90mg

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

idorsia

SANOFI GENZYME
Empowering Life

Takeda

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.




you're invited

Patient Empowerment WORKSHOP

PARSONS POND, NFD | May 25, 2019
2-4 Route 430, Parson's Pond, Newfoundland A0X 3Z0

Patients, Families, Caregivers and Health care providers are welcome.
Lunch and Health Snacks are included as part of the day.

JOIN US TO LEARN ABOUT THE EXCITING UPDATES ON FABRY DISEASE
Learn about new treatment options
There will be Canadian Fabry Association members at the meeting to provide support for the patients and families of Fabry in Newfoundland.

Contact:
Sherry Sim
by phone at
1-866-655-8548.




SAVE the DATE
October 19th 2019
VANCOUVER, BC

Patient Empowerment WORKSHOP

VANCOUVER, BC
Pinnacle Hotel Harbourfront, 1133 West Hastings Street

LEARNING OBJECTIVES

- Patients will be able to comprehend what is meant by an LSD.
- Patients leave with a strong understanding of Fabry Foundations – this includes genetics, classic Fabry/variant Fabry, mutations, etc.
- Patients takeaway key learnings of how to interpret their own labs, and diagnostic tests
- Looking at health holistically, exploring mindfulness and the benefits it has both short, and long term.

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.

Open to Newfoundland patients and families

Open to patients and families in BC & Yukon

PATIENT EMPOWERMENT ANSWER:

A): Just over 500 Canadians have been diagnosed with Fabry, however, we believe the numbers are much higher and patients have yet to learn their diagnosis.