

Newsletter



December 2020

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A word from the President:



Dear All

The last newsletter of 2020!

In this edition we share a lot of information which I am sure you will find an interesting read. We would also like to draw your attention to the save the date for our virtual FIN Expert meeting in 2021. We look forward to seeing you all at the meeting.

As we approach the festive season, I would like to pay tribute to everyone who works or volunteers to help others. This year the spirit of the season will likely mean different things to different people. Everyone's 2020 has been incredibly difficult and disruptive. Although we might not be able to spend this time of the year with all the people we love, connecting is now more important than ever. Reach out, call, Zoom, send a card and be thankful for what is and what is yet to come.

On behalf of the FIN Board I would like to thank everyone for all your support and interaction, you are all truly amazing!

Stay safe and healthy!

A handwritten signature in black ink, appearing to read "Lut".

Lut, FIN president



FABRY
International Network

SAVE THE DATE

**2021
VIRTUAL
FIN EXPERT
MEETING**

APRIL 24, 2021

Mental Wellbeing Tips from the FIN Young Adult Community

In light of the COVID-19 pandemic, we want to remind you that taking care of your mental health is as important as looking after your physical health. Good mental health and positive wellbeing can help you better cope with the COVID-19 threat and the uncertainty it's creating.

The FIN Young Adult Community wants to share with you their best tips for mental wellbeing!

- Practice gratitude & forgiveness
- Get some sunshine
- Spend time in nature
- Journaling
- Show kindness to yourself and others
- Being in the moment
- Positive self talk
- Meditation
- Open up about mental health issues and realise you are not alone
- Build a structure and create routines for your daily life
- Listen to music
- Talk to others or go out – spend time with family or friends
- Do fun stuff or give yourself nice things - Buy a plant when you feel down or drink a chai latte, whatever makes you happy
- Get enough sleep and eat nutritious food
- Get professional help when needed
- Exercise regularly and stay active
- Find out what makes you happy - not only what makes others happy
- Spend time alone with yourself - have conversations with yourself
- Know your limits: don't set impossible goals, know your limits in order not to deceive yourself
- Social Media Detox – don't compare yourself to others
- Take care of yourself! Use a facemask or scrub your face once in a while, dress like you are going to the awards... really spend time on yourself!
- Keep your mind busy, don't let intrusive thoughts bother you
- Always know you are valid: when there's someone criticizing because they don't understand the illness, remember it is not your fault and that you are valid in any way, you are not less than any other person in the world.



FIN proudly presents to you new members to the network



A warm welcome to:



Association Tunisienne des Maladies Lysosomales

L'ATML is an association recently founded by patients and relatives of patients suffering from rare and orphan diseases who are engaged in a daily fight against these diseases.



Mexican Federation of Rare Diseases

FEMEXER is the national alliance of rare diseases patients associations. FEMEXER is a non-governmental and totally voluntary alliance of patient organizations, directed exclusively by patients and their families; They represent more than 70 rare disease patient organizations in Mexico, which cover 450 rare diseases (RAID) among the population.

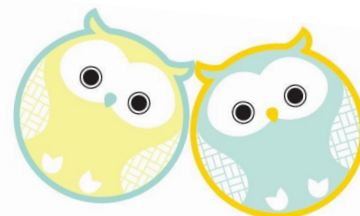


Proyecto Pide un Deseo México

PPuDM is the lysosomal patient association and is an organization dedicated to the support of Gaucher and Fabry patients in Mexico and is part of Palito and Toto Make a Wish AC (PyTPuDac), which is a grantee authorized by the SAT that meets all the requirements of Mexican law to be a non-profit organization. In addition, it belongs to the Mexican Federation of Rare Diseases (FEMEXER) as a founding member.

FOCUS ON FABRY LEADERS AROUND THE WORLD

Interview with Hisao Harada President, Japan Fabry Disease Patients and Family Association (JFA)



Japan Fabry Disease Patients
and Family Association
general incorporated association japan

When did you join your national patient association and what was the reason for joining?

I joined the Japan Fabry Disease Patients and Family Association (JFA) in September 2010.

What was the reason for joining?

There were five reasons for me to join:

- 1. To expand ERT options available for patients including oral therapy*
- 2. To make treatment accessible wherever you live in Japan*
- 3. To make treatment affordable in our medical system*
- 4. To establish a collaborative relationship with bio/pharma industry*
- 5. To build equal relationships with medical service groups and local administrations*

What is the vision and mission of your association?

Our vision is to create communication between members as well as the development of our organization. Our mission is to establish a symbiotic society and a reliable medical system for rare disease patients so we are all able to live with dignity.



What do you consider to be the major achievements or activities you are proud of?

Currently patients in Japan have access to 3 different ERT and 1 oral drug treatment option. Regardless of where patients live, treatment is now fairly affordable. We organise annual educational seminars and symposiums at 7 main cities to cover the entire nation. We built very good relationships with local administrations, medical service groups and the bio/pharma industry.

Can you name some challenges that your association is currently facing?

Because of the global pandemic, we are now working to have home infusion available, for this we are working closely with researchers and congressmen to prepare a proposal for our Ministry of Health, L&W. Seeking radical treatment such as gene therapy and genome medicines. However, we still need to wait until the legislation part is ready.

Can you name some future goals or plans?

- More treatment options which patients and their family can choose between*
- Educate more doctors to become experts in Fabry disease.*
- From the patient centred perspective, we need to promote awareness and provide educational opportunities to learn and seek the best treatment options for themselves. Japanese patients are very passive when it comes to choosing their treatment and only rely on the doctor's advice. But nowadays we see patients also proactively taking control and communicating their preferences more.*
- As several treatment options are available for patients, we need to promote early diagnosis including new-born screening. It's important to have a follow-up system together with the screening.*

Has your association had issues with the national health system or insurance problems?

Due to the national healthcare system's coverage, all patients have access to treatment in general. There is no major problem with medical expenses due to a medical support system based on the patient's household income now. As for private life insurance, there are certain limitations and there are some conditions that need to be cleared still. This is a matter that I expect that will improve.

FOCUS ON FABRY LEADERS AROUND THE WORLD

Interview with Hisao Harada

President, Japan Fabry Disease Patients and Family Association (JFA)

How would you describe the current treatment situation in your country?

Approximately there are 1000 patients receiving treatment. After you get diagnosed, you can start treatment as soon as possible. There is a maximum cap payment system that won't exceed a certain amount. Therefore patients and family can visit multiple medical specialists if they need additional care or other medication for symptomatic treatment.



What are the major issues with Fabry disease in your country?

Many Fabry patients wish to have a pain-free life although they take pain relievers from overseas. The pain management still seems to be the biggest issue for Fabry patients I know. There is a large need for specialized medical care facilities for rare diseases. We need a prompt implementation for telemedicine including online consultation in case of future natural disasters and/or pandemics.

How would you further raise global awareness for Fabry disease?

I think it's much better to focus on children rather than adults. There is a report that says that one in 7000 patients could be diagnosed through new-born screening. The age that patients can start ERT can be as low as 4 or 5 years old. Therefore, it's ideal to work with physicians who check both kidney failures and heart failures and apply early screening interventions.

What are your suggestions for future projects for FIN?

It's more important to work together with Fabry specialists especially on gene therapy and genome medicine. I also like to see the potential which we can develop more. I think it might be nice to invite a genome specialist at your next expert meeting. Since Fabry disease is rare and doesn't meet the quota, it's important to unite the patients on a global scale.

What would you like to explain to other people / doctors / nurses /decisionmakers about Fabry disease?

Although Fabry disease is a rare disease, it's very important to think of it as something that could also happen to you or to someone close to you.

What kind of information are we still missing about Fabry disease?

Many patients worry about their future. Especially young adults may experience it more often. It tends to have their income lowered as their symptoms progress. Then it will be hard to even consider married life with children. It requires more support for them.

Is there anything else you would like to add?

I think it's important to make our voice heard. I think FIN holds a key role for the global movement in the Fabry community in the world. Thank you for your dedication! With Ikuko Kaku as the JFA representative for FIN we look forward to continue to work together.

Thank you for your time!



**Hisao Harada hands the pen to David Peña Castillo
from FEMEXER Mexico**

Supporting Cellular Resiliency

By Dr. Seema Kanwal, ND

News regarding COVID-19 is breaking daily. With all the media attention on this emergent pandemic, there is an extraordinary concern in supporting our health. We can all take basic steps to be responsible to ourselves as well as people around us in preparing and supporting our cells to try and ensure a good outcome.

COVID-19 is thought to have an incubation period of 1-14 days with signs and symptoms that last 1-4 weeks and include:

- ⇒ Nasal congestion
- ⇒ Febrile Respiratory Illness (greater than 100F (37.8C))
- ⇒ Fatigue, Headache, Cough
- ⇒ Sore throat
- ⇒ Gastrointestinal manifestations such as nausea, vomiting, and diarrhea have also been commonly found which are uncharacteristic of the seasonal flu
- ⇒ Infants and toddlers may show with only fever and lethargy

Although most sources are estimating that an individual remains contagious for seven days from initial onset of symptoms (especially with coughing and sneezing) individuals have been found to be contagious for up to 16 days after the first symptoms appeared.

SUPPORTING OUR IMMUNE SYSTEM, THUS OUR CELLULAR RESILIENCY

A healthy immune system will not only offer increased protection against the regular seasonal flu and other variations of the common cold, but also shown to minimize and improve the immune response when you are now in bed under the weather.

There is good evidence, albeit not large clinical trials, to show that persons with higher serum 25(OH)₂ D₃ levels are protected from upper respiratory tract infections. There is also good mechanistic evidence that high doses could be used to treat the flu. 25 Hydroxy Vitamin D tests are an excellent resource for obtaining a person's vitamin D levels. Other evidence also exists for the efficacious use of vitamin C, zinc, selenium, vitamin A, and Elderberry extracts.

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Supporting Cellular Resiliency

By Dr. Seema Kanwal, ND

Here are now a few things you can do to keep your immune system healthy and strong.



First, let's review the basics. Hand washing, is by far one of the key strategies for preventing viruses from spreading. Viruses are easily spread from person to person. Think of how many times a day you touch a doorknob, keypad or pick up the telephone. Wash your hands in soapy warm water for at least 30 seconds (or the time it takes to sing happy birthday) and try to avoid touching the faucet after doing so. Paper towels, although less environmentally friendly are more hygienic than cloth towels, as are hot air dryers.

Nutrition, nutrition, nutrition. Proper nutrition is the key to a healthy immune system. Would you put diesel in a gas engine? Eating fried, sugary or highly processed foods can hinder your immune system's ability to ward off infection. Brightly coloured and dark green leafy vegetables are loaded with immune boosting phytonutrients. Phytonutrients are nutrients that come from plants. Bioflavonoids, found in fruits and vegetables work with Vitamin C to help strengthen the immune system.



Sugar, consumed in even moderate amounts impairs your immune system's germ-fighting ability. Our white blood cells, which engulf bacteria and viruses, cannot function optimally when we eat high amounts of sugar, especially white refined sugar. Drink lots of water and herbal teas when you're under the weather. Stay clear of soda pop, as they are very high in sugar. Even Diet pop is a no no! Avoid sports drinks because they contain excess amounts of sugar and salt. Warm herbal teas such as peppermint, can be made cooler as a flavourful iced tea, will also boost the immune system.

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Supporting Cellular Resiliency

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Hydration hydration! This is much easier to do when the weather is warmer, versus when it is cold outside. I tend to drink less water in the winter. If water is difficult for you as well, drink lots of herbal, non-caffeinated teas as this will count towards water intake. Fall and Winter is a very drying time of year. If you are prone to nose bleeds, or sinus infections, you want to ensure you maintain hydration to moisten the mucus membranes, to prevent dryness in the first place. Do you know how much water you need to drink? Take your weight in pounds, divide by 2, this equals to the number of ounces required by your body. The 8 glasses of water a day I cannot disagree with, as this is a random number that I have no idea who came up with.

When treating any illness, herbs are best used in a combination that is individually tailored to you and your symptoms. How about taking a trip to your kitchen? Garlic is one of nature's most potent herb. Not only does it add wonderful flavour to foods, it is a powerful anti-microbial agent. It has been shown to reduce bacterial AND viral growth, thus making it an effective agent in fighting many infections. Add it to soups or stir-fry's at the end of cooking which will maintain the anti-microbial properties of garlic. Adding it to the beginning of cooking will make it lose efficacy of its antimicrobial properties.





9 in 10 people living with a rare disease experiencing interruption in care because of COVID-19 Survey shows detrimental impact of coronavirus on rare disease community.

[Out now: the COVID-19 European results in seven languages!](#)

How did COVID-19 affect people with rare diseases? Find out their [infographic](#) with the key findings from their biggest ever survey in Europe, now ready to share in French, Spanish, German, Italian, Portuguese and Russian!

Share it on social media to show the impact of COVID-19 on people with rare diseases as the pandemic continues to unfold.



**RARE
DISEASES
INTERNATIONAL**

Rare Diseases in UHC2030 Synthesis

Rare diseases have been mentioned in UHC2030's "State of Commitment to UHC" synthesis. This is the first edition of an annual report monitoring action towards Universal Health Coverage in all UN Member states.

*The synthesis report notes that the COVID-19 pandemic has magnified inequities for vulnerable groups including people living with a rare disease. (page 28)

*It highlights the need, not only for better coverage of health services, but also for improved specialized health services, making particular mention of the rare disease community. (page 34)

RDI is a member of UHC2030, a multi-stakeholder platform convened by the WHO and the World Bank. We would like to thank RDI member organisations who answered our call this September to participate in the UHC2030 survey on national UHC contexts. The recognition of the rare disease community in this seminal report is a great achievement.

[Click here to read the full report](#)



Dear

I am very pleased to introduce you to the 2019 update on the Fabry Outcome Survey (FOS). FOS is a disease registry sponsored by Takeda that collects information on patients with Fabry disease across the world. Information on symptoms, disease progression and treatment has been collected since the first patients joined FOS in 2001, and there are now over 4000 patients enrolled. The aim of FOS is to improve our understanding of Fabry disease, which will help healthcare professionals to provide the best care possible for their patients.

We hope that this report will help patients and caregivers who are involved or interested in FOS to understand better how the registry works and what has been achieved so far. It provides an update on the patients enrolled in FOS up to January 2020 and a summary of published findings from the registry. The report has a new layout with colourful infographics, is written in easy-to-understand language and provides answers to commonly asked questions about FOS.

On behalf of the FOS Steering Committee, I would like to thank patients and caregivers involved in the registry for their extremely valuable contributions. The more information we can collect on Fabry disease, the more we can learn and the more we can improve the care of patients in the future.

Mary Pavlou
Patient organization representative on the FOS Steering Committee
FIN secretary

[Click here to read the report](#)



Sleep Hygiene for Fabry Disease

People with Fabry disease frequently experience sleep problems. Because a good night's sleep is essential to physical and mental health, as well as your quality of life, it may be beneficial for you to practice sleep hygiene.

[Click here to read the article](#)



Enter the EURORDIS PHOTO AWARD 2021

Do you have a photograph that captures life with a rare disease during the COVID-19 pandemic? Perhaps you have the perfect picture already, or perhaps you haven't taken it yet. Either way, don't forget to submit it to the [EURORDIS Photo Award 2021](#)! This annual competition is your chance to show the world what it means to live with a rare disease.

AVROBIO held a webcast on November 18th where the company announced:

- 3 month Gaucher type 1 data for patient #1. AVR-RD-02 for Gaucher disease type 1: Positive early reductions in plasma lyso-Gb1 and chitotriosidase activity at three months as compared to baseline, when Patient 1 was on ERT; additional positive trends observed across multiple other measures
- AVR-RD-06 for Gaucher disease type 3: New program leveraging the same vector as AVR-RD-02 for Gaucher disease type 1
- AVR-RD-01 for Fabry disease: Potential accelerated approval strategy planning underway as clinical data across Phase 1 and Phase 2 trials continue to show positive and durable clinical activity and safety data. 42 month data for the phase 1 Fabry program
- AVR-RD-04 for cystinosis: Functional and clinical improvements for the first patient at 1 year; third patient in the trial dosed
- New preclinical data for AVR-RD-03 for Pompe disease: Preclinical data show normalization of substrate levels in multiple hard-to-reach organs
- Pre-clinical update for AVR-RD-05 for Hunter syndrome: Normalization of multiple biomarkers in mouse model of the disease

The company also announced End-to-end plato[®] platform ready to enable global commercialization and data on its industry-leading plato[®] platform highlighting advances in chemistry, manufacturing and controls (CMC) to prepare for planned upcoming trials and potential global commercialization

[Click here to read the full press release](#)



This holiday season is unlike any other, to cap off a year unlike any other. Throughout this season, and as we move into a new (and hopefully better) year, we wish you moments of peace amid the difficulties, connections with family and friends even if they can't be in person, the warmth of memories from holidays past, and wonderful glimpses of the joy that still lives under the surface.

We are here for you through it all. We wish you endurance, strength, health, and as much happiness as these times can allow!

Best wishes for the holidays and the coming year from the entire FIN Board.



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International Network

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