



Fabry Focus e-Newsletter

June 30, 2015

Greetings!

Welcome to Summer in our part of the world,

North Carolina, where I live, has been a mix of many days over 90 degrees Fahrenheit and lots of thunderstorms. We are always eager to have the warm weather but it gets very hot very quickly in the summer.

Our weather situation provides a good analogy for our upcoming annual conference and camp in September. We are very eager to see the annual application process begin but our efforts (yours and mine) to make it happen smoothly are heating up very quickly.

Please begin your online application to the Charles Kleinschmidt Fabry Family Camp in September as soon as possible beginning tomorrow. I just spoke with the Victory Junction staff and they will have our online camp application process open July 1st as planned.

Weekend camp volunteers, please complete your online application as soon as possible starting tomorrow also.

In addition to conference and camp planning we have lots of other information to share with you.

This issue includes:

- Camp & Conference Application Process Starts Tomorrow!
- Other important meetings - FSIG 2016 conference Save the Date
- NFDF Programs Handout
- Clinical Center Initiatives
- New Market Research Opportunities
- Clinical Trial News
- NFDF Information Update Survey
- 2014 Take Our Surveys Contest Update
- February 2014 Survey Results
- April 2015 Matching Gift Campaign Update
- The Shining Star Campaign Donor Acknowledgements
- Clarifying Our Future Funding Strategy

Enjoy the Summer, or whatever season you are in in your country.

Thank you for your continued participation and support!

Proudly serving the Fabry disease community,

Jerry Walter

Camp & Conference Application Process Starts Tomorrow!



The 6th annual Charles Kleinschmidt Fabry Family Weekend Camp and the 5th annual Fabry Family Conference
... information for families and volunteers

The Victory Junction Camp and volunteer application processes begin tomorrow, July 1st. As most of you already know, the application start date changed after our last newsletter announcement.

If you have a child with Fabry age 6 to 16, please submit an application for your entire immediate family to attend this expense-free event.

Earlier this month we sent an email with guidance for submitting camp and conference applications and for submitting volunteer applications to everyone on our list who previously expressed an interest in attending the camp and/or conference. If we missed you, please email Jerry at jerry.walter@fabrydisease.org right away so we can send you the guidance. Please do not respond via face book.

Our goal is to get applications approved as soon as possible to enable us to purchase airplane tickets at the best possible prices. Please submit your applications promptly and follow up as quickly as possible with remaining requirements.

Educational Conference Dates: 24-25 September (Thursday afternoon to Friday afternoon) in Greensboro, North Carolina. All individuals with Fabry and families are welcome to attend. Travel and lodging assistance is available for those with financial constraints. If you have not already emailed with your intent to attend, please let us know as soon as possible.

Camp Dates: 25 -27 September (Friday at 5:30 PM until about noon on Sunday) at Victory Junction Camp in Randleman, North Carolina (45 minutes from Greensboro).

Camp volunteers: Volunteer applications will be accepted on July 1st. The volunteer

time commitment is from 2:00 PM (at camp) on Friday until about noon on Sunday (depart camp). The Victory Junction volunteer application process has recently changed. Please do not use the old forms. If you have not already, please email Jerry with your intent to volunteer so we may send pre-application and application information.

Conference volunteers:

If you are unable to volunteer for camp and you do not plan to attend the conference as an individual with Fabry or family member but would like to help, we can always use a few conference volunteers. Please let us know.

In all categories above, an important first step is to email Jerry Walter as soon as possible to let us know your intentions to attend the conference, to attend the camp, or to volunteer. We'll send you important pre-application information.

We look forward to seeing you all there!

Other important meetings

2016 FSIG Fabry Expert Conference

Save the Date!

As many of you know, the Fabry Support and Information Group (FSIG) recently held their 2015 Fabry Expert Conference.

Please put the 2016 FSIG conference on your calendar so you won't miss this important event. We hope everyone gets a chance to attend at least one of the September (NFDF) or March (FSIG) annual Fabry community conferences.

Save The Date!!

2016 FSIG Expert Fabry Conference

March 4th, 5th, & 6th

*Wyndham San Diego Bayside
1355 North Harbor Drive
San Diego, CA 92101*



*Speakers
Excellent Information
You Don't Want To Miss It!*

Excellent

Registration is \$75.00 for the first person and \$25.00 for each additional person over age 10

For more information, contact FSIG at info@fabry.org or (660) 463-1355.

NFDF Programs - don't miss anything!

Our program handout is now located on our website.



Last month was our 10 year anniversary as a 501(c)(3) charitable non-profit organization. Still, we are often told by community members they have never heard of many of our programs and services.

Everyone should have the opportunity to use all resources available to you, NFDF programs and services and other organization's resources.

To ensure you all know what we are up to, a PDF file our program handout is now located in the "Featured Resources" section near the bottom of our website home

page.

Please share the program handout with your patients, family and friends!

Clinical Center Initiatives



An important initiative from Emory .. we need your help!

Many of you know how much Dawn Laney does for the Fabry community. She is awesome! Let's show our gratitude in a small way by participating in this initiative that may really benefit our community. Thank you! Jerry

Message below from Dawn:

Are you living with Fabry disease or are the family member of someone living with Fabry disease? If so, we are interested in asking you a few questions through a brief SurveyMonkey survey.

Emory Genetics, Davalen, Inc., and the National Organization for Rare Disorders (NORD) are developing a webpage/application called GeneThink that uses the power of IBM's Watson supercomputer to do a better job of answering questions about genetic conditions than Google. In order to make this website useful, we're asking you to tell us about your questions related to Fabry disease. All individuals who complete the survey will be entered into a drawing to win a \$25 Amazon gift card.

If you are interested in doing this survey, please click on the SurveyMonkey link below and complete the questions:

[Emory's GeneThink Survey](#)

New market research opportunities

Participants for the two initiatives described below will be accepted on a first-come, first-served basis. The survey duration and number of participants are limited.

Initiative #1: Fabry Patient Journey Research

We recently distributed information about a market research initiative on our face book page. We are seeking 30 participants for a survey consisting of completing a questionnaire that should take about 15 minutes followed by a 45 minute phone interview. Participants will receive a \$50.00 gift card for your participation.

Criteria:

- Lives in the U.S.
- 18-75 years old
- Diagnosed with Fabry disease in the last 5 years **OR** started Enzyme Replacement Therapy in the last 3 years.

The study consists of completing a questionnaire that should take about 15 minutes followed by a 45 minute phone interview.

To thank you for your participation, you will receive a check in the amount of \$50.

Initiative #2: Treatments in Development Research

Criteria:

- Lives in the U.S.
- 18-75 years old
- Diagnosed with Fabry disease
- Must experience at least very mild symptoms of Fabry disease (excludes patients with no symptoms)
- Must be at least a little interested in learning more about new treatments for Fabry disease
- Patient and immediate family must not be employed by FDA or any pharmaceutical company

Participants currently on a treatment or not on treatment are needed,

After a few screening questions, the study consists of a 60-minute telephone interview to be conducted between June 29 and July 10th.

To thank you for your participation, you will receive a check in the amount of \$85.

Contact us: If you are interested in either study, please email Jerry at jerry.walter@fabrydisease.org and let me know if you are interested in initiative #1 or #2 or both. Please do not respond via face book.

Clinical Trial News



Exciting Progress with Research ...

Amicus Therapeutics announced that the European Medicines Agency (EMA) has validated the Marketing

Authorization Application (MAA) for the oral small molecule pharmacological chaperone Galafold (migalastat HCl) to now enter the formal regulatory review process in the European Union.

Amicus states they remain on track to hold a pre-NDA (new drug application) meeting with the United States Food and Drug Administration (FDA) and to submit an NDA for Galafold in the second half of 2015.

This brings the Fabry community closer to a 3rd approved Fabry disease therapy outside the U.S. and a 2nd approved therapy in the U.S.

See more at [Amicus investigational drug approval update](#)

Individuals with Fabry and parents ... please help with this important information update

Six \$200 prizes waiting to be awarded ... for a great cause!

We are continuing to extend this survey for a while to ensure everyone has a chance to take it. And, especially because the results will really help others!



All families with Fabry disease should have a chance to benefit from our many programs and services. To accomplish this, we need everyone's help!

We published a new survey last month to update several important resources so we can provide the updated resources and tools to the Fabry community and ...

... to make this important effort more interesting and more successful we published the survey in a contest format following the success of the 2014 survey program.

Individuals with Fabry disease 18 and older or parents with kids under 18 with Fabry who do not have a parent in their home who also has Fabry disease may participate.

To thank you for your participation, five prizes of \$200 will be awarded in a random drawing of survey participants. Also, a bonus drawing for \$200 will be held for those answering the family mutation question.

This survey is open to the global community. Please forward this newsletter and share the survey link widely.

The National Fabry Disease Foundation (NFDF) is gathering information to:

- update our Find a Doctor database (a family resource)
- update our website population map
- update the NFDF contact database to send important news and information
- update contact information for those who subscribe to our newsletter
- provide our current patient/family member handout explaining our programs
- send your wallet size Alert Card USB Drives to everyone with Fabry

This is a very short and easy survey to take.

WE DO NOT SHARE YOUR PERSONAL INFORMATION WITH ANYONE!

Survey link below:

<https://www.surveymonkey.com/r/March2015InformationUpdateSurvey>

Thank you for your support! Please share the survey widely.

2014 Take Our Surveys Contest

Thank you again to everyone who participated in the 2014 monthly survey program!

We continue to analyze the results of the 2014 monthly surveys. See the February 2014 results below. We will publish the results for each month in our upcoming newsletters.



We look forward to resuming the survey program later in the year!

February 2014 Survey Results



February 2014 survey summary

After the February survey was initially distributed it remained opened the rest of the year. A total of 243 people took the survey. Here are the results which serve to provide insights to the Fabry community.

Total number of adult participants with Fabry disease = 243

Females participants = 76% of the total participants

Male participants = 24% of the total participants

U.S. participants = 80%

Non-U.S. participants = 20%

Adults with Fabry disease from the United States, Canada, England, Scotland, Ireland, Australia, New Zealand, Germany, The Netherlands, Norway and Poland participated.

Question 1. What type of physician originally suspected/discovered Fabry disease in your family?

Eye Doctor = 19%

Nephrologist (kidney doctor) = 18%
Geneticist = 10%
Dermatologist = 7%
Self-Diagnosed = 7%
Primary Care Physician = 6%
Neurology = 4%
Cardiology = 4%
Internal Medicine = 2%
Newborn screening = 1%
Autopsy = 1%
Other physicians = 6%
Didn't know = 14%

Question 2. What types of physician prescribes your approved or investigational Fabry treatment medication?

Participants on an approved or investigational treatment = 184 (76%)

Participants not on an approved or investigational treatment = 59 (24%)

Breakdown of those on treatment - medication prescribed by:

Geneticist = 49%
Nephrologist (kidney doctor) = 20%
Primary Care Physician = 13%
Internal medicine = 5%
Neurology = 4%
Endocrinologist = 3%
Various others answers less than 1% each = 6%

Question 3. How often do you visit a major medical center (MMC) with Fabry disease expertise?

- I routinely visit a MMC for all my Fabry disease needs = 25%
- I visit a MMC with Fabry disease expertise one to four times a year = 47%
- I have visited a MMC with Fabry disease expertise but I'm no longer in touch with them = 15%
- I have never visited a MMC with Fabry disease expertise = 5%
- Other answers like I visit a MMC every two years or less frequently, I have only visited a MMC with expertise once, etc. = 8%

Question 4. Does your **local** physician managing your Fabry disease symptoms have knowledge of Fabry disease?

- My primary local physician managing my Fabry disease has a good knowledge of Fabry = 21%
- My primary local physician seems to have little/no knowledge of Fabry disease but seems eager to learn = 13%
- My primary local physician only has some knowledge of Fabry disease. = 22%
- My primary local physician seems to have little/no knowledge of Fabry disease and doesn't seem very interested in learning = 7%
- I only go to a major medical center with Fabry disease expertise, not any other local clinics for primary Fabry management = 23%

- I don't have a local physician managing my Fabry disease symptoms =10%

- Other answers in addition to the above answer choices = 6%

Question 5. How far do you travel to a major medical center (MMC) with Fabry expertise, a local clinic for routine management, or a different clinic for treatment or management other than the MTC or local clinic you reported?

5a. Of the 206 people who reported how far they travel to a MTC with Fabry disease expertise, they report:

- Travel 10 miles (16 km) or less to their Fabry MTC = 19%
- Travel between 11 and 30 miles (>16-48 km) to their Fabry MTC = 28%
- Travel between 31 and 60 miles (>48-97 km) to their Fabry MTC = 18%
- Travel between 61 and 120 miles (>97-123 km) to their Fabry MTC = 17%
- Travel between 121 and 200 miles (123-322 km) to their Fabry MTC = 8%
- Travel between 201 and 420 miles (>322-676 km) to their Fabry MTC = 7%
- Travel between 421 and 900 miles (>676- 1448 km) to their Fabry MTC = 2%

** Of course, some folks fly to their major treatment center and a couple had ferry trips as part of their journey.

5b. Of the 154 people who reported they visit a local clinic in their area for routine Fabry disease management, they report:

- Travel 10 miles (16 km) or less to their local clinic for Fabry symptoms = 54%
- Travel between 11 and 30 miles (>16-48 km) to their local clinic for Fabry symptoms = 31%
- Travel between 31 and 60 miles (>48-97 km) to their local clinic for Fabry symptoms = 8%
- Travel between 61 and 120 miles (>97-123 km) to their local clinic for Fabry symptoms = 4%
- Travel between 121 and 200 miles (>123- 322 km) to their local clinic for Fabry symptoms = 1%
- Travel over 200 miles (322 km) to their local clinic for Fabry symptoms = less than 1%

5c. Of the 96 people who reported they receive treatment at a different location than their major or local treatment center, they report:

- Receive treatment at a center within 25 miles from home = 40%
- Receive treatment at a center within 30 to 60 miles from home = 12%
- Receive treatment at a center within 75 to 100 miles from home = 3%
- Receive treatment at home = 45%

Question 6. Who is on your health care team?

- A cardiologist (heart specialist) = 55%
- A primary care physician/family physician = 52%
- A nephrologist (kidney specialist) = 48%
- A geneticist (genetic disease specialist) = 42%
- An ophthalmologist or optometrist (eye specialist) = 41%
- A neurologist (nervous system specialist) = 23%
- A dermatologist (skin specialist) = 14%
- An internal medicine physician (general medicine specialist) = 11%
- A psychological health specialist (anxiety, depression or other psychological or

psychosocial needs) = 9%

- A gastroenterologist (digestive system specialist) = 9%

- A pulmonologist (lung specialist) = 9%

- A rheumatologist (rheumatic diseases specialist such as arthritis) = 4%

- A pain specialist = 4%

While we unintentionally omitted audiologist/ENT, 3% of participants manually entered "An Audiologist/ENT". We suspect this number is actually higher and will ask again in a future survey.

We asked which other physicians people visited:

- A hematologist = 2%

- OBGYN, nurse, chiropractor, naturopath, endocrinologist, electrophysiologist, and a neurosensory specialist were also mentioned by at least one person.

Question 7. Can you get referrals to other physicians when you need them?

- I don't see any Fabry treating physicians to request referrals from = 9%

- My physician is very reluctant to make referral to other clinics = 3%

- Sometimes my physician is reluctant to make a referral to some of the specialty clinics I need = 8%

- I always receive the referrals to other clinics I need = 80%

Question 8. What are your 3 primary resources to learn about Fabry disease?

The percentages below indicate the number of participants who listed each resource indicated below as one of their top 3 resources.

- My primary Fabry disease physician = 60%

- The non-profit Fabry disease patient advocacy/support organizations = 46%

- My genetic counselor or clinic nurse = 40%

- Face book support groups and pages for Fabry disease = 30%

- Other internet resources = 21%

- A family member or friend who knows a lot about Fabry disease = 19%

- My pharmaceutical industry case manager = 19%

- My infusion nurse = 15%

- I've reported the resources I use but I don't have 3 good resources = 14%

- My regional or local pharmaceutical industry patient liaison = 11%

- Another physician on my healthcare team = 10%

- Other non-profit patient support organizations = 8%

- Pharmaceutical industry internet sites = 6%

- My case manager from my hospital or clinic = 5%

- Other face book support groups and pages = 5%

- The PubMed journal article library = 4%

Question 9. Do you know your Fabry disease mutation?

- 44% of participants reported their family mutation in the January or February survey.

- 47% of participants reported they would attempt get their mutation from their physician and provide it at a later date.

- 9% of participants responded they did not know it or did not respond to the question.

We asked the question about family mutations in all twelve 2014 monthly surveys. We will report the final results in the December survey analysis later.

Other Questions.

We also asked questions about participant's physicians and healthcare team members contact information to help rebuild our "Find A Doctor/Clinic" searchable database - a patient resource. We will update the database/resource this year.

End of February 2014 survey results.

April Matching Gift Campaign

Thank you for helping us to exceed our April matching gift campaign goal and thank you to our generous sponsors!



During April the Kleinschmidt Family Foundation generously offered to match all individual donations up to a total of \$10,000 to support the Charles Kleinschmidt Fabry Family Weekend Camp in September.

A second family who prefers to remain anonymous generously offered to match a total of \$3,000 to support NFDF education and community support programs.

We exceeded our goal of \$13,000 by raising \$13,645 for a total of \$26,645 with the matching funds.

Together, we are making a difference for people with Fabry disease and their families!

Please see the donor acknowledgements below.

An Essential Ingredient to our Success is You!



Shining Star Campaign

Be the Brightest star!

Thank you to all of the shining stars listed below who have contributed since our last regular newsletter in April 2015.

We are extremely grateful for your generous charitable contributions!

The brightest stars for this period are:

Champion Level (\$1,000 or more) -

HSLopez Family Foundation
Ron Dukenski for our Victory Junction Camp

Recurring monthly donations:

Jamie Hanrahan in honor of the Bohn family
Warren Tausch
Alex Lane

Our many other generous Shining Stars for this period are:

Leader Level Donor (\$500 to \$999):

Stacy Ho

Advocate Level Donor (\$100 to \$499):

Tamara Klein
Steve McLaughlin
Rob and April Hopkin
Genzyme East Team
Lisa Bacon
Theresa Cosentino
Deborah Schultz
Christine Francey
Dawn Laney in memory of Wayne Kennedy

Supporter Level Donors (up to \$99)

Tracy Myatt in memory of Craigg Cordell
Maribeth Greene
Uzma Atif
Lois Riley dedicated to Cheryl Barcomb
Becky Winstell
Michelle Fowler in memory of Steven J. Nelson and Robert Nelson
Lisa Berry
Danielle Fischl
Erin Leise
Barbara Earley
Kelly Otter
Whitney Shanks
Kimberly Michael in honor of Cheryl Barcomb
Lori Wise in honor of the Larese family for Fabry Awareness Month
Sandra Atwood
Katie McWilliams
Kacey Nielson for Katie Jensen Reese, a beautiful person and an amazing mother
Cheryl Barcomb in honor of Michelle and Zach
Katie Reese

Paul Rakoski
Irene Allen
Maribeth Greene
Suzanne Ferguson

Thank you all for your tremendous support! Every size contribution adds up to make a huge difference. You have our deepest gratitude for being the brightest star you can be.

On behalf of everyone who benefits from our many programs, thank you so much for your generosity!

We would also like to thank our Sirius sustainers!

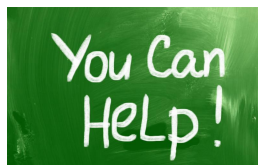
Sirius is the brightest star (in fact, star system) in the Earth's night sky. Our Sirius category donors make major contributions to provide our annual funding base on which to build. Our Sirius sustainers provide the starting point to make our programs and services possible.

Thank you!

to:

The Kleinschmidt Family Foundation
Genzyme Corporation
Shire Human Genetic Therapies
Amicus Therapeutics

Our future funding strategy



Clarifying our funding program ...

As many of you know, non-profit charitable organizations rely solely on contributions from our supporters to fund our many important programs and services. And, per U.S. Internal Revenue Service (IRS) requirements at least 10% (increasing to 33% over time) of our total funds must come from the public rather than from industry or family foundations.

Providing programs and services to support the Fabry community is our most important job, always. In parallel with that effort we must also raise funds along the way to support our programs.

Our future fundraising strategy is to hold two major fundraising efforts a year in April and October. We hope to continue a matching gift campaign in April like we did this year. It was a great success! This is also a critical time to ensure we have enough funding to support our conference and camp.

To give us the best opportunity of implementing a sustainable annual program, we urge everyone to make a tax deductible charitable contribution once or twice a year. Of course, we graciously accept contributions any time of the year, but we'll send out invitations to donate in April and October.

Another great way to help us maintain a sustainable program is to start a recurring monthly donation from our website fundraising button in the upper right corner of the home page at www.fabrydisease.org. You can automatically donate \$5, \$10, \$15, \$20 dollars a month. You don't feel the pain of a lump sum donation and we can rely on your generous support to provide stable programs and services to our community. Please consider it!

Also, please keep us in mind for memorial contributions.

An important note for our global community members: We share information and provide some services to our Fabry friends in 49 countries. We want you to use as many of our programs and services and we are able to share globally.

However, as we implement fundraising initiatives and announce them in our newsletter, on our face book page and by other means, **please use our fundraising initiatives as a reminder to make a charitable contribution to the Fabry support organization in your country rather than to the National Fabry Disease Foundation.** They need your help!

Until Next Time

Thank you all for your continued participation and support! Please contact me anytime at the phone number or email address below.

We look forward to publishing the remaining monthly survey results in the upcoming months.

Please share this newsletter among your friends and family members, and others who may be interested.

If you have not "liked" our face book page at www.facebook.com/FabryDisease please like our page when you get a chance and share it with your family and friends.

Proudly serving the Fabry community!

Sincerely, Jerry

Jerry Walter
Founder and President
National Fabry Disease Foundation

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