

RASopathiesNet

Connect ~ Collaborate ~ Cure

A YEAR IN REVIEW

RASOPATHIES NETWORK USA

BOARD OF DIRECTORS:

- ❖ Lisa Schoyer, President
- ❖ Lisa Schill, Vice President
- ❖ Lee K. Johnson, Secretary
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A LOOK BACK ON 2015

Happy New Year! This newsletter contains a monthly highlight of what we accomplished in 2015 and a peek at 2016.

The RASopathies Network USA's volunteer Board of Directors was hard at work during 2015 organizing the 4th International Rasopathies Symposium, which is held biennially. This event brought together 560 family members, researchers and clinicians from 24 states and 11 countries to discuss way to advance research of the Rasopathies.

We continue to work hard educating legislators and pharmaceutical companies about the RASopathies, among other initiatives.

All of this would not be possible without your donations, letters and phone calls to Congress and the Senate, your participation in research studies and initiatives and participation in the Million Dollar Bike Ride to name a few. You are directly helping us fulfill our mission to advance research of the RASopathies. Thank you so much for your continued support!

To find out what you can do to be more involved with or how to support our organization, please email us at info@rasopathiesnet.org.

January

RASopathies Network was mentioned in the Science Daily article, "Researcher's report way to target hard-to-hit site in disease pathway"

<http://www.sciencedaily.com/releases/2014/11/141120123140.htm>

Our text-to-donate campaign hosted by EveryLife Foundation was so successful they asked us to create a toolkit for other organizations to follow. Which can be viewed here: <http://everylifefoundation.org/wp-content/uploads/2012/07/Text-RARE-Campaign-2014.pdf>

February

Our Vice President wrote an article for Rare Disease Report, "Rare Disease Advocacy, more than just selling t-shirts" to educate on some simple things we can do to help advance research. <http://www.raredr.com/contributor/lisa-schill/2015/02/rare-disease-advocacy--more-than-just-selling-t-shirts>



February (continued)

We spent a week in Washington D.C. for Rare Disease Week educating the members of Congress about the RASopathies and specific legislation that will have a great impact on our community, like 21st Century Cures and the OPEN Act. To learn more about legislation that can help advance research of the RASopathies, please visit the Rare Disease Legislative Advocates.

<http://rareadvocates.org/>

March

We attended Patients as Partners, a conference to learn how industry and patients can work together to develop better clinical trials keeping the patients at the center of all the decision making. There are not any active clinical trials for RASopathies yet, but when there are, we want to make sure our family's needs are being met.

David Stevenson, MD, co-chair of the 4th International RASopathies Symposium, spoke about the RASopathies and RASopathies Network at the 2015 American College of Medical Genetics and Genomics' Annual Clinical Genetics Meeting. His talk was entitled - "RASopathies: The Power of Partnering with Advocacy Groups." -

April

RASopathies Network accepted an invitation to speak at the World Orphan Drug Congress. Our talk was titled, - "Tapping on the Brakes: Small Molecule Therapies for the RASopathies." - The focus of our presentation was to delineate the RASopathies and stress the importance of companies investing in therapies for the RASopathies.



Lisa Schill speaking at WODC

Thank you to Bruce Korf, MD, PhD, and our Scientific Advisory Board for publishing the Third International Meeting on Genetic Disorders in the RAS/MAPK pathway: towards a therapeutic approach in the American Journal of Medical Genetics.

- <http://www.ncbi.nlm.nih.gov/pubmed/25900621>

Lisa Schill accepted an invitation to speak on the panel, *Rare Diseases and Orphan Drugs: Publications and Perspectives*, along with Jonathan Goldsmith, MD, from the FDA, Scott Newcomer from Shire Pharmaceuticals and Louise Wyhopen from Novartis Oncology at the 11th Annual Meeting of the International Society for Medical Publication Professionals. In this session, we discussed the importance and challenges associated with rare disease research and publications, using the RASopathies as an example.

May

We enjoyed hosting a Rare Disease Congressional Caucus Briefing at the Capitol called, "The 21st Century Cures Initiative: Priorities for the Rare Disease Community."

June (continued)

What a great turn out for the Million Dollar Bike Ride - <http://www.milliondollarbikeride.org/> to raise money for a RASopathies research grant focused on pain!! We had about 25 riders and a water stop full of volunteers. Bruce Gelb, MD, a member of our scientific advisory board, was one of our riders! Every year the riders tell us our water stop is the best (our water station is more like a RASopathies family reunion)! Thank you to all who helped and participated. A special thank you to the Griest family for being our top fundraiser again this year!! We hope to make it an even bigger event in 2016 so mark May 7, 2016 on your calendar to save the date. Come join us for the ride or the water stop! Please contact us at info@rasopathies.org if you're interested.



Team RASopathies at the Million Dollar Bike Ride



RASopathies Network water stop



Team Cadence for Konnor ringing the cow bell



(L) Lisa Schill and (R) Monica Grund discuss day's events after MDBR

RASopathies Network participated in the - "Lions and Tigers and Genes, Oh My!" - event at the Brandywine Zoo in Wilmington, Delaware to educate participants about genetic syndromes. We handed out lots of RASopathies brochures and educated many families about the RASopathies. If you would like a copy of our brochure, please visit our website <https://rasopathiesnet.org/wp-content/uploads/2014/01/RASNet-brochure-033014.pdf>

June

Our Board was very excited to be one of 1,700 exhibitors at the BIO International Convention, the world's largest biotechnology gathering. Exhibitors and attendees include the leading biotech companies, top 25 pharma companies, top 20 CROs and CMOs, more than 300 academic institutions including the major research labs and government agencies and the

June (continued)

leading consultants and service companies. We offer our sincere gratitude to Bruce Gelb, MD, and Katia Sol-Church, PhD, for helping represent RASopathies Network USA at our booth.



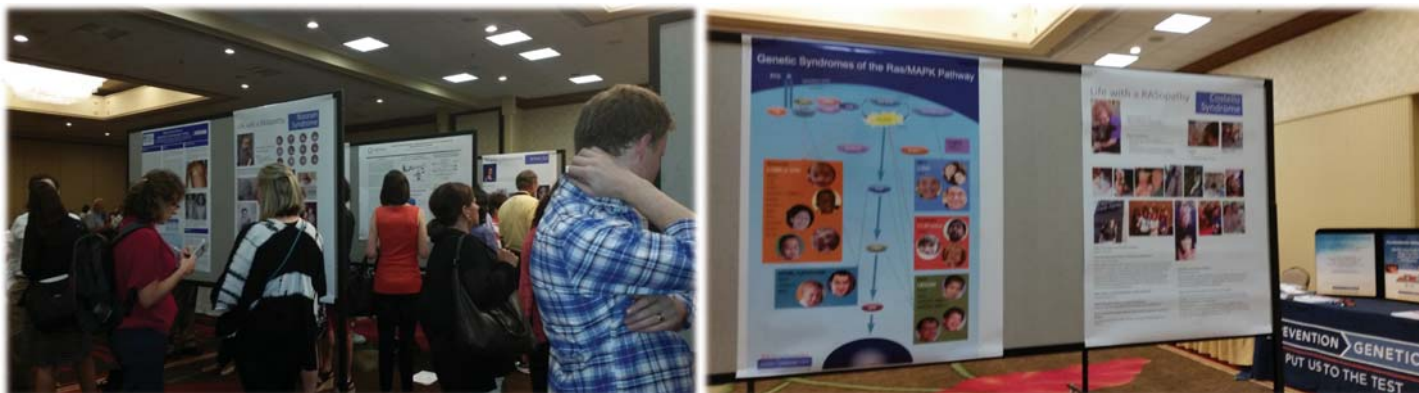
(L) Dr. Gelb and (R) Dr. Sol-Church

Thank you to the Bailey family for hosting the "RIDE or WALK for #RASopathies" benefiting RASopathies Network USA, in Peru, Vermont. We appreciate all of your support!!

Thank you to the staff members of Prevention Genetics for participating in the Chicago Ragnar Relay Race to raise funds for RASopathies Network USA. This team ran approximately 200 miles to raise funds and awareness of the RASopathies!!

July

Our Board organized and held the 4th International RASopathies Symposium, co-chaired by David Stevenson, MD, and Brigitte Widemann, MD. Thank you to the National Institutes of Health, Eunice Kennedy Shriver National Institute Of Child Health & Human Development (NICHD), National Center For Advancing Translational Sciences (NCATS), National Institute Of Neurological Disorders And Stroke (NINDS), and National Cancer Institute (NCI) for supporting our symposium that provided a venue for a scientific conversation between clinicians, researchers, trainees and affected families to share and discuss clinical issues and basic science. These conversations help set forth a framework for future research, translational applications directed towards therapy and best clinical practices for RAS/MAPK pathway syndromes.



Two glimpses at the very successful Poster Session



(L-R) Lisa Schoyer, Dr. Stevenson, Dr. Widemann and Lisa Schill



Rick Guidotti photographing children

At the Symposium, RASopathies Network raffled off **4 FREE GENETIC TESTS** to anyone in the world in need of RASopathies genetic testing. The tests were graciously donated by GeneDx, Invitae, PreventionGenetics and UAB Medical Genomics Laboratory.

This highly scientific event still had many opportunities for RASopathies families, including a dessert reception and poster session which allowed for interaction between families and top doctors and researchers, photo shoot with world famous photographer, Rick Guidotti, and syndrome specific break-out sessions with the doctors who discussed in layman terms what was learned at the Symposium.

We accepted an invitation to attend and speak at the Rare Disease Legislative Advocates NE Conference about the importance of educating legislators on the RASopathies and legislation that will help advance the research of the RASopathies.

September

Thank you to Jack Bailey for riding the Civil War Century Ride as a fundraiser for the RASopathies Network USA. Jack rode this grueling ride that included climbing 8,000 vertical feet. Way to go Jack!!

We helped develop the agenda for the Global Genes Patient Advocacy Summit this year. We also accepted the invitation to talk about working with legislators to advance research of our syndromes. <https://globalgenes.org/2015summit/>

October

Rasopathies Network applied for and received a scholarship to attend the NORD (National Organization of Rare Disorders) Summit. We also accepted an invitation to participate in a PCOR (Patient Centered Outcomes Research) training to learn more about the different types of grants that are available for our organization. It was a very informative meeting and we were very thankful that we were invited to attend.

November

Lisa Schill and her son, Max, received the Federal Level Abbey Award from the EveryLife Foundation. Patient organizations and advocates have many roles and responsibilities in advocating for individuals with rare diseases. These awards recognize organizations and individuals who go above and beyond to become policy leaders and political advocates in their state and nation. Congratulations to them both for helping advance research of the RASopathies by helping change federal policy to increase funding of RASopathies research and help cure the drug development process.

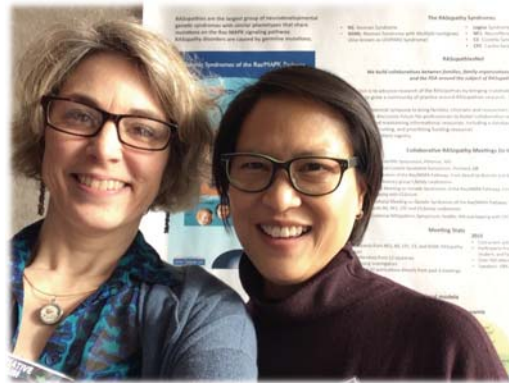
November (continued)

Here is a little update from Maria Kontaridis, PhD, on what she has been doing with the seed money from the 1st Million Dollar Bike Ride Grant awarded to her for *Delineating the cause of gastrointestinal abnormalities in RASopathy disorders using human inducible pluripotent stem cells (iPSCs)*. <https://rasopathiesnet.org/category/rasopathiesnet-usa/>

Thank you Invitae for mentioning the RASopathies Network in your ad in the San Francisco Business Times.

December

Thank you to our President, Lisa Schoyer and parent Beth Stronach for attending and representing RASopathies Network at the 1st RAS Initiative Symposium held by the National Cancer Institute (NCI). This two-day international symposium was the first RAS community-wide meeting sponsored by the National Cancer Institute's RAS Initiative. The overarching goal of the NCI Initiative is to mobilize the cancer research community to develop ways to understand and target cancers driven by mutant RAS in an open model of collaboration among government, academic, and industry researchers. We presented a poster about the RASopathies at the meeting. You can view the poster here. <https://rasopathiesnet.org/wp-content/uploads/2014/01/screen-grab-poster-final.png>



(L) Beth Stronach and (R) Lisa Schoyer

We closed 2015, by announcing the recipient of the \$53,000 Penn Medicine's Million Dollar Bike Ride RASopathies Grant. Thank you again to all that participated and made this grant possible. Don't forget to join us on May 7th, 2016 for the 3rd Annual Million Dollar Bike Ride. 100% of our team's donations go toward the RASopathies research grant and will be matched by Penn Center for Orphan Disease Research-up to \$50,000

And the grant goes to.....

Giuseppe Zampino from the Universita Cattolica Sacro Cuore

Congratulations to Giuseppe Zampino who will be using the grant to fund his research on "Pain in RASopathies: new investigative techniques and treatments."

A PEEK AT 2016

We have already started planning! We are presenting a poster about the RASopathies at the National Institutes of Health for Rare Disease Day and are planning to make the Million Dollar Bike Ride the best yet! If you would like to join us and contribute your time and talents to our organization, we would love your help. Please email us at info@rasopathiesnet.org

If you have any questions or would like to learn more about RASopathies Network, we would love to hear from you.

With the beginning of another year upon us, RASopathies Network USA's Board of Directors wishes you and your loved ones a Happy, Healthy New Year!

Proud members of the following organizations:

