THECUTESYNDROME

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Tracey Buyce
Photography

ANNUALREPORT

August 2014

A word from Esmé's Maman

On The Cute Syndrome Foundation's first year

When we founded The Cute Syndrome last year our goals were rather modest. We wanted to contribute to the work our partners in Italy were doing funding PCDH19 research, and we wanted to help raise awareness of PCDH19 by telling Esmé's story. However, thanks to a tremendous outpouring of support we exceeded our initial fundraising goals, raising over $80,000 to date. As a result we expanded our commitment to our partners in Italy to co-fund a two-year $135,000 research grant, which has been awarded to the CNR Institute of Neuroscience Milan, Italy for Dr. Maria Passafaro and Dr. Elena Battaglioli's research, discussed below. We were also able to fund a $25,000 research grant to Dr. Poduri's team at Boston Children's Hospital in order to help support their work with zebrafish, also discussed below. Additionally, we are working with our partners in Italy and a new PCDH19 Epilepsy foundation in Spain in order to continue to fund promising research together.

Our work with The Cute Syndrome is proving to be more important than ever. While PCDH19 FLE is a relatively new genetic diagnosis, it has been projected to affect between 15,000-30,000 patients in the US. Many of these patients remain undiagnosed due to a lack of information about this disorder. In addition to our fundraising, we have helped to accomplish significant gains in researcher/foundation collaboration, PCDH19 FLE awareness, and raising The Cute Syndrome's profile. We want to use this annual report to let our supporters know more about all that we have been able to do in the last year with their help!

Thank you for your continued support and we look forward to big things in the coming year!

-Hillary Savoie, PhD
Founder of The Cute Syndrome (and Esmé's Maman)
www.thecutesyndrome.com

The Cute Syndrome Foundation is is tax-exempt under Section 501(c)(3) of the Internal Revenue Code and donations are tax deductible to the extent permitted by law.

The first internationally-funded competitive PCDH19 research grant

We are pleased to announce that The Cute Syndrome Foundation, together with our partners in Italy, Insieme per la Ricerca PCDH19 (Together for PCDH19 Research) are funding exciting research of Dr. Maria Passafaro and Dr. Elena Battaglioli at the CNR Institute of Neuroscience Milan, Italy. The $135,000 grant is for two-years to support their proposal to better understand how the PCDH19 mutation disrupts molecular function in the body--resulting in seizures and developmental delays.

Their work shows great promise because understanding what is "wrong" at a cellular level can help in the development of a targeted treatment for PCDH19. This proposal also includes some very innovative and exciting work using exon skipping, which allows cells to "skip" over the mutated genes. Exon skipping is producing promising results in treating muscular dystrophy and spinal muscular atrophy, but, as far as we know, this would be the first use of this method in treating epilepsy.

Our partnership with Insieme per la ricerca PCDH19 has been very fruitful. This year we will expand our funding partnership to include the PCDH19 foundation from Spain with the goal of continuing to fund research together.

Members of Insieme per la Ricerca, international researchers, and Hillary at dinner before the conference in Rome (Oct 2013)

Paola, HIllary, and Esmé Meeting in (March

Our organizations joined forces as partners last year with this grant in mind. We believe it is essential to the cause of better understanding, treating, and, ultimately, curing PCDH19 FLE that organizations and researchers around the world work together closely and collaboratively.

Zebrafish and Boston Children's Hospital

The Cute Syndrome Foundation has also committed $25,000 to help Dr. Annapurna Poduri at Children's Hospital Boston study the effects of existing FDA-approved compounds on zebrafish with PCDH19 mutations.

Dr. Poduri's research is still in the early stages, but her team has made very good progress their studies of seizure activity in zebrafish.

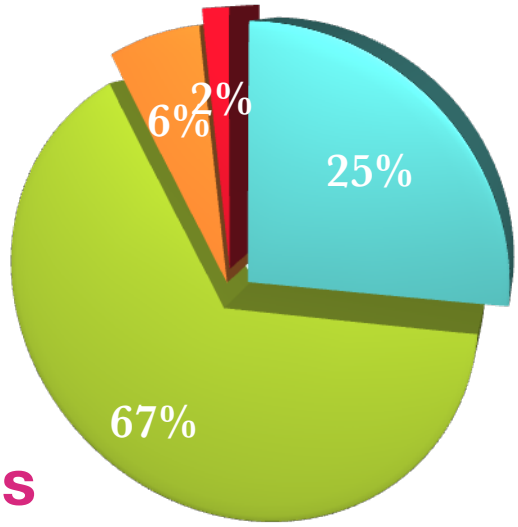
They are also working with the newest genome editing techniques (CRISPR/Cas) in order to create a zebrafish with a PCDH19 mutation that exhibits symptoms that are most closely related to those that girls with PCDH19 FLE experience. Once they have the best model then they can trial existing pharmaceutical agents to rectify the neuro-developmental and seizure abnormalities. This is a method that was successful in finding a new treatment for a similar disorder, Dravet, with an antihistamine, clemizole.

We have been able to fund this research with the help of a $10,000 matching grant from an anonymous donor. So far, we have raised approximately $17,000 with the help of this donor!

Our visit to Boston Children's Hospital Zebrafish Lab (April 2014)

Fund Allocation

- BCH Zebrafish
- Competative Grant
- Special Projects
- Administration costs



Upcoming Clinical Trials

Insieme per la ricerca PCDH19 has history of funding very promising research. They funded Prof. Gecz at the University of Adelaide in Australia. Gecz's research into the effects of neurosteriod hormones on the expression of PCDH19 epilepsy has lead to an upcoming drug trial by Marinus Pharma of a new drug, ganaxalone. In their announcement of the trial Marinus predicts that the US population effected by PCDH19 Epilepsy will be 15,000-30,000 patients.

Esmé and Hillary were able to join Paola Squillante from Insieme per la ricerca PCDH19 for a meeting with Marinus Pharma last year.

Awareness Raising Activities

Raising the profile of PCDH19 Epilepsy is essential to our mission. Over the last year we have engaged in several campaigns to help raise awareness so that more girls will be able to be diagnosed and receive the support they need once they are diagnosed.

Follow our activities on facebook.com/thecutesyndrome.com

Photos and TV

In the last year Esmé has also had three photoshoots! TCS organized a photoshoot for Esmé and two other girls with PCDH19 with Rick Guidotti of Postive Exposure. Tracey Buyce from Tracey Buyce Photography also donated a day of her time to take some beautiful photos of Esmé for TCS and the Spring 2014 edition of Mamatoga Magazine. Esmé also appeared on WNYT with Benita Zahn in February. The Cute Syndrome was a finalist in the Times Union newspaper promotion Capital Region Gives.

Novartis Annual Report

Thanks to Dr. Ricardo Dolmetsch, the Global Head of Neuroscience at Novartis, and world-renowned photographer Brent Stirton, Esmé will be featured in the Novartis annual report as an example of a person who might benefit from Novartis's focus on finding treatments to rare disorders.

PCDH19 Awareness Day

With the help of our partners, PCDH19 families, and The Cute Syndrome fans on Facebook, we successfully helped launch the first annual PCDH19 Awareness Day on November 9th, 2013. TCS Facebook page was covered with photos taken by friends and family all around the world holding purple balloons.

Capital Care Pediatrics in Clifton Park, NY celebrating PCDH19 Awareness Day with a visit from Esmé

Esmé during speech therapy, Photo credit Tracey Buyce Photography

We would like to extend our thanks!

2013 Catch the CUTE Auction Donations

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YTK Photography
Artcentric Gallery
Doug Klein Pottery
IvyRose
Neil Levine
Brian and Heather Straughter
Drue Sanders
Alex and Ani
Kristen Barrows
Mazzone Hospitality
Cantina Restaurant
Leah's Cakery
Old Saratoga Wine and Spirits
Bob and Mary Pratt of Elihu Farm
Schuyler Creek Chiropractic
Stiletto Salon
Truly Rhe
Anchor No 5
Pennywise Shoppe
Billie Giberson Moloney

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Matina Skipski
Justin Donohoe
Angela and Marc Savoie
Truly Rhe
Pennywise Shoppe
The Meinweiser Family
The Mixter Family in Memory of Edith Mixter
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Hillary Savoie and Philip Gitlen at the 2013 Catch the CUTE fundraiser. This event raised almost $20,000 last year.

www.thecutesyndrome.com