

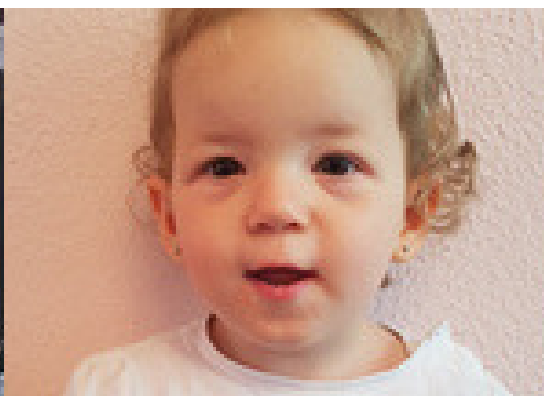
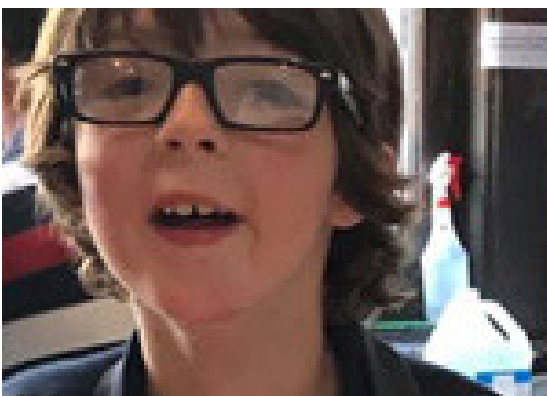
BE THE HOPE! BE THE CHANGE!

ANNUAL REPORT 2018



SETBP1

S O C I E T Y



Dear SETBP1 Friends, Family and Supporters,

Boy has this year flown by. We have seen so much change since this time last year and we are proud to share with you our highlights, joys, and acknowledgements of those who have made a difference in our community. Before we jump into the year end updates, we wanted to take this opportunity to thank our supportive and passionate community who pulls together to help us advance our mission forward and empowers us to reach our goals.

We appreciate your support whether you registered your family and child in the Simons VIP Connect registry, donated to one of our fundraisers, organized a fundraiser, attended our virtual family conference or our virtual family connect, visited a SETBP1 disorder specialist, shared your story on our Facebook support group, volunteered for SETBP1 Society, or simply supported a SETBP1 family. You have made a difference!

We are thankful for the expertise and devotion of our Scientific and Medical Advisory Board members and our distinguished SETBP1 Society Board members. These leaders chart the course for SETBP1 Society and help to ensure we stay on track and successfully make it to our final destination.

This year we raised over \$47,000, initiated a SETBP1 disorder grant funding process, represented our organization at the Global Genes Patient Advocacy Summit conference and Rare Disease Week on Capitol Hill sponsored by EveryLife Foundation, partnered with Simons VIP to host our first virtual family conference, increased our online resource repository, and connected with many new SETBP1 families.

Thank you all for your commitment and motivation! We are looking forward to 2019 and to moving research forward to find the answers we need to help our children!

Sincerely,

Haley

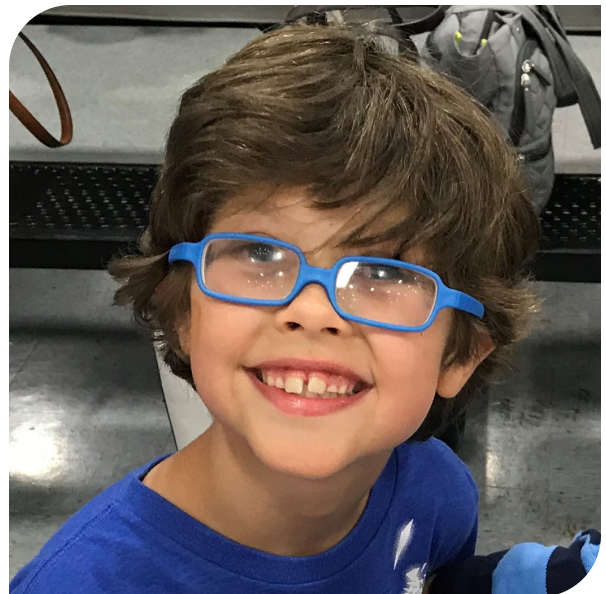
Haley Oyler

President

SETBP1 Society

WHO WE ARE

Our Mission is to provide support to individuals with SETBP1 disorder and their families, to promote discussion and fund research, and to bring awareness and education to the public.



COMMUNITY

- First Virtual SETBP1 disorder Community Virtual Conference hosted in partnership with Simons VIP Connect
- SETBP1 Community grew to 40 families
- Added our newest Scientific and Medical Advisory Board members, Dr. Rocío Acuña Hidalgo & Dr Wendy Chung
- Raised over **\$48,000** primarily through two big fundraisers - Rare Carousel of Possible Dreams and Giving Tuesday

RESEARCH

- SETBP1 registry grew to 21 registered families with 13 medical reports completed.
- Launched our SETBP1 Society Grant Funding Application Process
- Awarded our first Research Grant Award for SETBP1 disorder research
- Supported Dr Bregje van Bon in initiating the SETBP1 disorder phenotype paper
- Supported Dr Angela Morgan with launch of her SETBP1 disorder speech/language phenotype study
- Worked with Dr Carl Ernst to find patients for human iPSC development for SETBP1 disorder and Schinzel-Giedion Syndrome
- Collaborated with Dr Wendy Chung and Dr Cat Lutz from Jackson Labs to launch the SETBP1 disorder mouse model project

AWARENESS

- Joined NORD (National Organization of Rare Disorders) and partnered with NORD with research oversight from Dr Bregje van Bon to develop a SETBP1 disorder guide, planned release in early 2019.
- Joined the TREATMENT center at Columbia University under the leadership of Dr Wendy Chung
- Representation at the Global Genes Rare Patient Advocacy Summit
- Representation at Rare Disease Week at Capitol Hill sponsored by Everylife Foundation

First Virtual SETBP1 Family Conference

Our year kicked off with our first SETBP1 disorder Virtual Family Conference on January 13, 2018. SETBP1 Society partnered with Simons VIP Connect to provide a platform for connecting families virtually to ask questions from professionals familiar with the disorder and to learn potential options for treating certain symptoms of SETBP1 disorder including ADHD and anxiety. Dr Wendy Chung from Columbia University, Dr Siddharth Srivastava from Boston Children's Hospital, and Haley Oyler from SETBP1 Society presented at the conference. The conference was capped off by a meeting of just SETBP1 families to share their stories and to connect with each other.

Rare Carousel of Possible Dreams - Spring Fundraiser

The Rare Carousel of Possible Dreams fundraiser was held online in partnership with Global Genes and the Festival of Children Foundation. They provided the fundraising platform and support that empowered 8 SETBP1 teams to raise a spectacular \$26,589, surpassing our goal of \$25,000. 108 unique donors contributed to our successful fundraiser!



A special thanks to our SETBP1 fundraising teams: Team Olivia, Team Elisabeth, Team Chance, Team Brian, Team Mandy, Team Kathy, Team Cole, and Team Eric. Also a HUGE thanks to our generous top donors: Judeann Warner, Jessica Hoffson, Brian Hoffson, Doug Rogers, Lee & Melanie, Chance Isham, Ann Phillippe, Catherine & Peter, Ray & Andrea Webb, Rick & Sandy Marts, Ross Humphreys, Susan Hoffson, The Hills, and Tim Bollinger.

SETBP1 Facebook Community

Our SETBP1 Facebook community grew from 22 to 40 SETBP1 families in the last 12 months. We are seeing a decrease in the price for the whole exome sequencing (WES) and whole genome sequencing (WGS) genetic tests, which is helping more families to receive an appropriate diagnosis for their child, especially for children who have a monogenetic disorder like SETBP1 disorder. As our community is growing, we are seeing more and more activity within the Facebook group and more opportunities for learning and connecting. Our SETBP1 Facebook Support Community grew to 59 members and our SETBP1 Friends and Family Facebook Group grew to 43 members!

Giving Tuesday - Fall Fundraiser

The Giving Tuesday Facebook fundraiser was another awesome success. We set a goal of \$15,000 and with the support of 14 Facebook Fundraisers and our supportive community, together we raised \$18,468. 162 unique donors contributed to the success of our Giving Tuesday fundraiser!

Special Thanks to SETBP1 Facebook Fundraiser Creators: Nancy Isham, Chance Isham, Linda Boyd, Trina Geye, Nick Munro, Cécile M. Barcelo, Jean-François Barcelo, Nicole Goscilo Wentz, Elisabeth Fiquet, David Markaverich, Erin Floyd Otness, and SETBP1 Society organization fundraisers.

A TREMENDOUS shout out to our top fundraiser creators and top donors: **Top Facebook Creators:** Erin Otness, Eric Oyler, Elisabeth Barcelo & Jean-François Barcelo **Platinum Donors:** Kevin Storli, Erin Otness, Chance Isham, Rita Selvaggi, Elisabeth Fiquet, Cynthia Shepherd, Susan Danziger, Ron Hicks, & Valerie Barcelo **Gold Donors:** Cécile M. Barcelo, Pascale Clement, Denyse Cusson, Nancy Isham, Francois Barcelo, Mikaela Teris, Bridget Mulvenna, Anette Otness, Jane Asher, Carmen Hicks, & Nick Munro **Silver Donors:** Jill Comras, Jennifer Havel, Linda Boyd, Darius Venclauskas, Reed Arnos, Trina Geye, Andrea Webb, Mark Isham, Jason Nakashima, Heidi Prichard, Tom Schalk, Noelle Reed, Dana Dawson, Sally Portera Mankins, Eric Otness, Amy Amick, David Papas, Christi Pramudji, Amber Marie, Christy Hughes, Audrey Beale, Jean-François Barcelo, Doug Rogers

Happy Birthday to You

This year two SETBP1 community members created Facebook Birthday Fundraisers to support SETBP1 Society. Every donation helps and we want to thank them for helping to spread awareness and raise funds to help our organization. Thank you Paige Stephenson and Erika Schimke Reese!

SETBP1 Family Connect

SETBP1 Society hosted our first SETBP1 Family Connect. This opportunity provided families a video platform for communicating with other SETBP1 families online. The attending families shared stories, successes, and challenges our families are facing. We even had a few guest appearances from a few of our children with SETBP1 disorder.

At the beginning of the year, SETBP1 Society established goals to promote the development of one biological model of SETBP1 disorder and the creation of a diagnostic journey report from SETBP1 patient stories. The biological model we planned to help fund was either a mouse model or a human iPSC model of SETBP1 disorder. The human iPSC model is derived from human cells (blood, urine, or skin) from individuals with SETBP1 disorder that are differentiated into neuronal cells with the purpose to learn more about the impact of brain function from the disorder.

We had one partially characterized SETBP1 KO mouse model that was not confirmed to be representative of the disorder and no iPSC work for SETBP1 disorder in development. Funding biological models of any disease is an expensive endeavor and one that our organization can not fund alone. We need partners and larger institution support to help move SETBP1 disorder research forward.

We are proud to say that at this point in time we have human SETBP1 disorder iPSC development underway at two respected institutions - in Dr Carl Ernst's lab at McGill University in Montreal, Canada and in Simon Fisher's lab at the Max Planck Institute in Nijmegen, Netherlands. SETBP1 Society granted Dr Simon Fisher, director of the Max Planck Institute for Psycholinguistics and Professor of Language and Genetics at the Donders Institute for Brain, Cognition and Behaviour in Nijmegen, the Netherlands, \$25,000 for his human iPSC project.

We initiated another project focused on SETBP1 disorder mouse model research with Dr Cat Lutz at Jackson Laboratory. The mouse model project is three-fold. The first phase is verifying whether the current SETBP1 KO mouse model is representative of SETBP1 disorder and produces half of normal amount of the SETBP1 protein. If the mouse model is not representative then the mouse model will be modified to be a true representative of SETBP1 disorder. The modified mouse would be made available to researchers for free on the Jackson Laboratory website. This project is covered by grant money Jackson Laboratory receives by being a Knockout Mouse Project (KOMP) Repository partner.

The second phase is to develop a reversible SETBP1 disorder mouse model. This mouse model will help determine if certain symptoms of SETBP1 disorder i.e. language, intellect, behavior, are reversible and at what stages in life. Jackson Laboratory will produce the reversible SETBP1 disorder mouse model with oversight from Dr Wendy Chung, the director of the TREATMENT center, at Columbia University and make it available for free to researchers on the Jackson Laboratory website. The cost of the reversible mouse model is covered by grant money provided by SETBP1 Society. The third phase of the mouse model project is to perform the needed characterization and experiments of the produced SETBP1 disorder reversible mouse model. Dr Wendy Chung and her team at the TREATMENT center will complete phase three with the financial support of SETBP1 Society.

In addition to biological model development, there are two other research projects in development to help spread awareness of SETBP1 disorder and to help us better understand the impact of the disorder. Dr Angela Morgan, a professor at the University of Melbourne and Murdoch Childrens Research Institute, in collaboration with Dr Bregje van Bon, a clinical geneticist in Nijmegen, Netherlands at Radboud Medical Center, Dr Simon Fisher, director of the Max Planck Institute for Psycholinguistics, and SETBP1 Society developed an extensive SETBP1 disorder speech and language phenotype study. The study is expected to launch in January 2019 with the cooperation and support of the SETBP1 community. The other research project launching soon is a SETBP1 disorder phenotype paper directed by Dr Bregje van Bon with support from Dr Siddharth Srivastava, a pediatric neurologist at Boston Children's with a strong interest in our SETBP1 community, and Dr Wendy Chung as her role as principal investigator for the Simons VIP Connect project. The material for this phenotype paper will come from consented SETBP1 disorder individuals seen by Dr Bregje van Bon, from individuals who completed the medical interview step in the official SETBP1 Society registry, Simons VIP Connect, and from consented individuals seen by Dr Siddharth Srivastava.

Special thanks to the following amazing specialists for their research and scientific contributions to SETBP1 research this year!

Dr Bregje van Bon, Dr Simon E. Fisher, Dr Siddharth (Sid) Srivastava, Dr Wendy Chung, John Ford, Dr Rocío Acuna Hidalgo, Dr Carl Ernst, Lilit Antonyan, & Dr Cat Lutz

Launched Grant Funding Program



With a priority mission to fund research, the SETBP1 Society Board and Medical and Scientific Advisory Board (MSAB) worked together to develop the 2018 SETBP1 Society Grant Funding Application process. The goal of this program is to recruit interest in SETBP1 disorder and to move translational SETBP1 disorder research forward. We care about research that will help bring individuals affected by the life-impacting disorder closer to targeted, meaningful treatment. We ultimately awarded a 1 year

grant for \$25,000 to Dr Simon Fisher and his co-investigator Dr Bregje van Bon to uncover molecular mechanisms & neuronal pathways involved in SETBP1 disorder using human cell-culture models.

The SETBP1 Society Board and MSAB also prioritized SETBP1 disorder mouse model research. SETBP1 Society is awarding Dr Cat Lutz, the director of Jackson Labs, funds to develop a reversible SETBP1 disorder mouse model and to make it freely available to all researchers on their website.



These projects will begin in early 2019.

Special thanks to all our SETBP1 Society grant applicants.

Funding Prioritization Shift Explanation

*We chose to prioritize funding both biological models instead of producing a diagnostic journey guide in 2018 for a few reasons. One reason is that we believe that the biological models will help us find a treatment faster. In order to find a treatment for any disorder, you need models to help test whether the treatment is effective. The other reason is that we believe a diagnostic journey guide will be easier to produce once we have the phenotype studies completed. These studies will serve as tools for the diagnostic journey guide along with input from families and medical specialists focused on SETBP1 disorder.

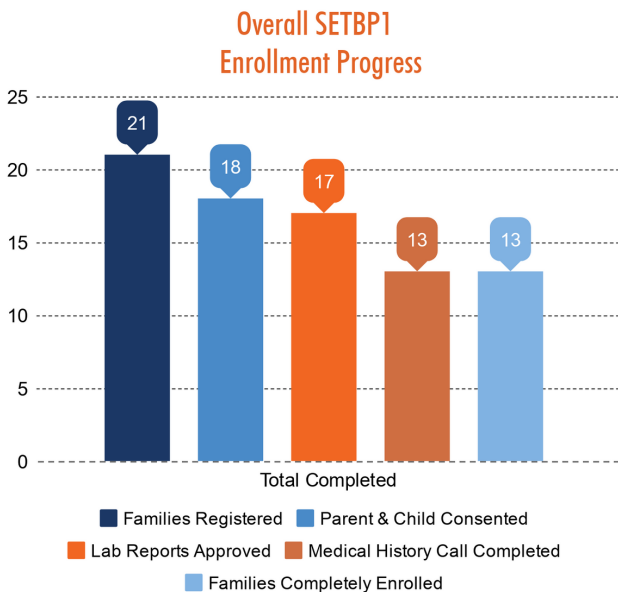
Simons VIP Connect Registry

A valuable way our community can contribute to the advancement of the body of knowledge about SETBP1 disorder is to register their family with Simons VIP Connect, our official SETBP1 Society registry. This registry collects important information about an individual's disorder including medical history, behavior and attention history, developmental history, and other relevant data, de-identifies the registered individual's data, and provides a mechanism for researchers around the world to access and study the data. A phenotype paper spearheaded by Dr Bregje van Bon, a SETBP1 Society Medical and Scientific Advisor and clinical geneticist at Radboud Medical Center in Nijmegen, kicks off soon and will include data from the Simons VIP registry. This data will also be leveraged during our upcoming virtual SETBP1 Family Conference on January 19, 2019 presented by Dr Wendy Chung, the principal investigator for Simons VIP.

Simons VIP Registry #s

Simons VIP Connect: SETBP1 Progress Report





As of December 5, 2018, we currently have **21** SETBP1 families registered for research with Simons VIP, but only **13** families that are fully enrolled and contributing data to researchers.

You can help by completing any steps you are missing in the enrollment process!

Contact the study coordinators with any questions or concerns:

coordinator@simonsvipconnect.org
 1-855-329-5638

Partnerships

SETBP1 Society has added NORD (National Organization for Rare Disorders) to our list of partners. NORD helps rare organizations and patients through education, connection, and empowerment. By joining NORD, our organization's exposure to the rare, research, biotech and pharma communities expands. Other benefits provided include connections to the major influencers in the rare disease community, research and organization guidance, access to relevant educational materials for our community, help spreading awareness of our organization and SETBP1 disorder and priority notification of scholarships and discounts for NORD's annual Rare Disease and Orphan Products Breakthrough Summit. From our partnership, the development of a SETBP1 disorder guide sprouted and is expected for release in early 2019. We are thrilled to be partners with NORD!

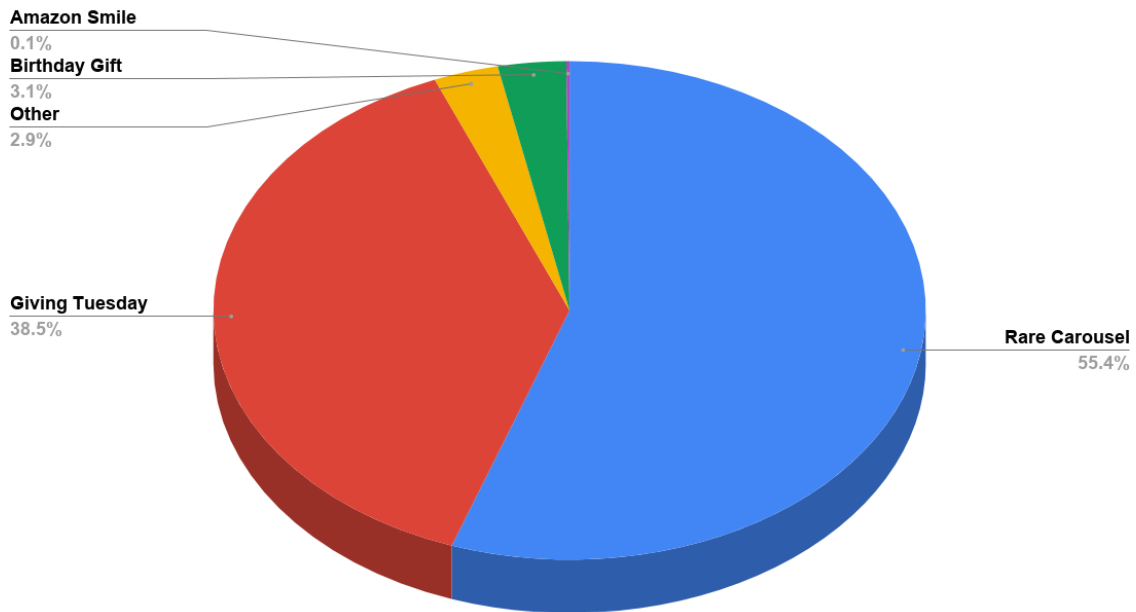
We also formed a partnership with Dr Wendy Chung, the director of the TREATMENT (Targeted Research and Exploration Advancing Trial Models Editing and Next-generation Therapies) program at Columbia University. Columbia's TREATMENT program was developed to transform how rare disorder is understood and how they treat children with rare diseases. The team produces individualized models that allow them to study the identified genetic mutations and biological mechanisms underlying each disease on a personalized basis. This study enables the team to pinpoint windows of opportunity: when in the course of disease development a treatment may be effective, and what it might be.

Representation at Rare Events

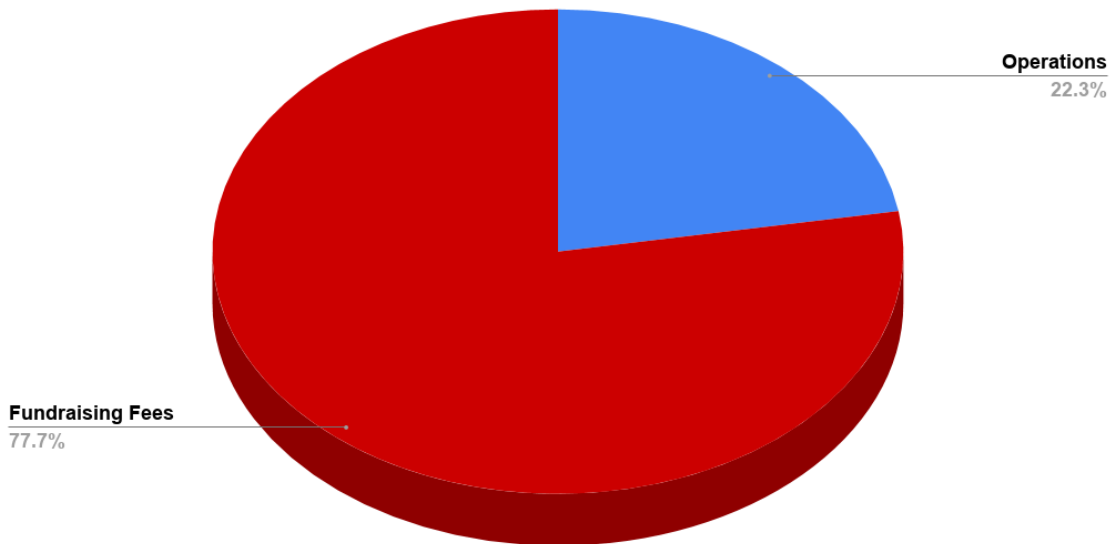
SETBP1 Society is a Foundation Alliance member with Global Genes. Global Genes strives to educate, equip, and empower the global community with tools and resources to influence change in the rare disorder community. In October, the president of SETBP1 Society had the honor of representing SETBP1 Society at the Global Genes Patient Advocacy Summit where she learned about the latest rare disease treatments, connected with researchers and representatives from esteemed universities, met with pharmaceutical companies in the rare disorder space, and learned from other founders of rare patient organizations.

The trip to Washington D.C. for Rare Disease Week on Capital Hill sponsored by Everylife Foundation for Rare Diseases was inspirational, powerful, and helped to move our mission forward. As a small rare disorder organization with limited resources, it is crucial for us to actively spread the word about SETBP1 disorder, to promote legislation that will help ensure there is a treatment pathway available when we reach that point in our journey, and to connect with other patient advocacy organizations to learn and grow.

Revenue - \$48,074



Expenses - \$716.32*



*\$25,000 of revenue is set aside for the 2018 SETBP1 Society Grant Awardee, Dr Simon Fisher.

LOOKING AHEAD

As 2018 comes to a close, we reflect on what we have accomplished and how this momentum flows into 2019. Investing in SETBP1 disorder biological models to help us to better understand the impacted neural pathways and what symptoms of SETBP1 disorder are reversible and at what stages in life will continue to be our focus. We also plan to support researchers currently developing SETBP1 resources for our community. We believe this plan will bring us closer to finding a treatment to help our SETBP1 kids. We aim to provide more resources to support our SETBP1 community and more opportunities for our families to connect and share. Spreading awareness about SETBP1 disorder through targeted campaigns is our third aim for this coming year.

On Saturday, January 19th, we will partner again with Simons VIP for our 2nd Annual Virtual SETBP1 Family Conference! In the first quarter, we plan to launch an online store where SETBP1 disorder merchandise will be available for purchase. We also plan to organize and support 2 big fundraisers aimed at raising funds for SETBP1 disorder research. Our first will be our participation in the Million Dollar Bike Ride sponsored by the University of Pennsylvania Orphan Disease Center (ODC) on Saturday, June 8th and will incorporate a SETBP1 family meetup in Philadelphia, PA.

Our community support enables us to move our mission forward. We would not be here without their generosity. If you would like to make a financial contribution, organize an event, volunteer, donate an item, please reach out to Haley Oyler at haley@setbp1.org.

Thank You!!

Sign up for our newsletter and emails to stay informed of the latest research, upcoming events, and other news that impacts our SETBP1 Community!

<http://www.setbp1.org>

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