2021 SETBP1 Society
End of Year Report

Total SETBP1 Funds Raised - $426,135*

SETBP1 Society Funds Raised - $205,345

2021 SETBP1 Society $84,633

*MDBR - 2021 $91,466, 2020 $80,746, & 2019 $67,943

Highlights

COMMUNITY

- Hosted our 4th Annual Virtual SETBP1 Conference in partnership with Simons Searchilght
- Developed a SETBP1-HD Resource Guide in English and French for parents and support team
- SETBP1 Registry data provided in 3 quarterly reports by Simons Searchlight

RESEARCH

Publications

- SETBP1-HD phenotype paper published by Bregje van Bon & team
- SETBP1-HD speech/language phenotype paper published by Angela Morgan & team

Disorder Models

- Simons Searchlight released 3 patient-derived SETBP1 iPSC (induced pluripotent stem cell) lines for researcher access (2 additional iPSC lines under development)
- SETBP1-HD patient blood sample banked with IBX repository for iPSC line creation
- Reversible SETBP1 mouse model development underway from MDBR funding
- Characterization of SETBP1 indel and KO Conditional mouse model study to validate they are valid SETBP1-HD models
- SETBP1 indel mouse prefrontal cortex brain study to help understand why individuals with SETBP1-HD have challenges with executive functioning underway from MDBR funding

Collaboration and/or Studies

- Hosted 2 SETBP1 Collaboration Calls to unite our SETBP1 research and medical community
- Hosted 2nd SETBP1 Research & Medical Conference in partnership with Simons Searchlight
- Participated with 30+ rare disease groups in the Million Dollar Bike Ride for the 3rd year and raised $91,466 to fund 2 new SETBP1-HD research projects
- Launched 2 new SETBP1 community studies - SCoReS - collaborative SETBP1 parent impact study & SpeechTracker - study to track SETBP1-HD speech and language development over 3 years

AWARENESS

- SETBP1-HD NIH GeneReview published, which provides SETBP1-HD guidance to medical professionals
- Launched SETBP1 Alliance website at setbp1alliance.org to strengthen and expand SETBP1 collaboration
- ApraxiaKids Childhood Apraxia of Speech genetics video released for CEC credit hosted by SETBP1 Society president & Angela Morgan of MCRI
- Presented at GlobalGenes conference
- Launched the Meet the SETBP1 specialist video series
- Launched new SETBP1 Society Store

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2021 Primary Focuses

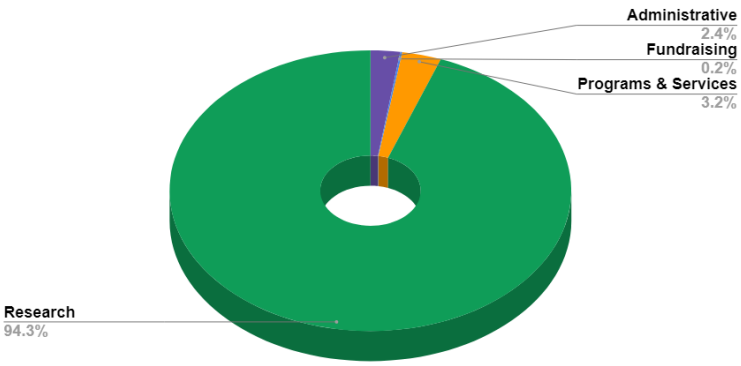
- Educate, Unite Community & Drive Engagement with Family & Research Conference, Social Media Presence, and Education Events
- Commit to at least 2 Fundraisers (Million Dollar Bike Ride & Giving Tuesday)
- Build the SETBP1 research network through organization of SETBP1Alliance Collaborative Research Network, SETBP1 Collaboration Calls, and Funding & Promoting Research
- Continue to promote and fund research focused on expanding research tools for studying SETBP1-HD, identifying affected pathways or targets for treatment, and validation of identified molecular targets (biomarkers & surrogate endpoints)

141 Families — SETBP1

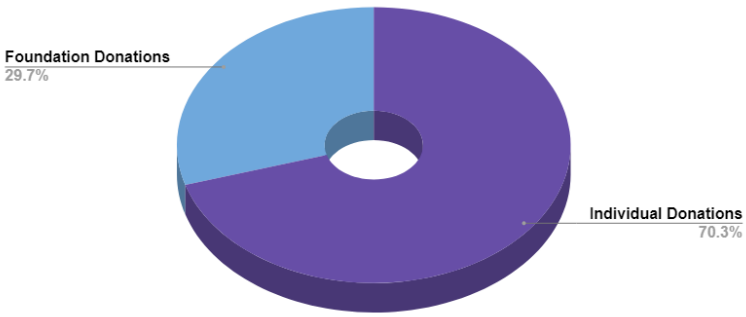
60/25 Families — SIMONS SEARCHLIGHT

65 Families — SETBP1 REGISTRY

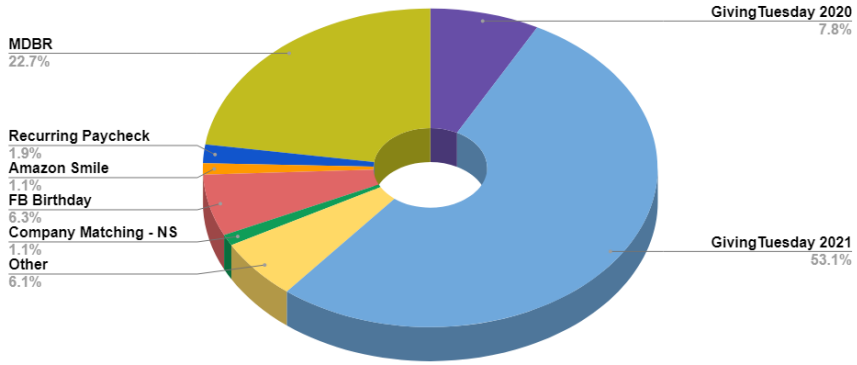
Expenses - $22,559



Income Received - $84,633



2021 Donations Committed by Program



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2022 Looking Ahead

1st Qtr
- Kick-off new disease model and pathway discovery SETBP1 research
- Begin partnership with Vanderbilt for SETBP1-HD Disease Concept Study
- Expand the Meet a SETBP1 Researcher program to include community engagement and Q&A
- Rare Disease Day - Opportunity to Spread Awareness
- ORCA (Observer-Reported Communication Ability) outcome measure study partnership expansion
- Quarterly SETBP1Alliance Collaboration Call

2nd Qtr
- Launch Rare-X Data Collection - March
- Launch SETBP1 Educational Series in collaboration with IDefine and KDVS
- Quarterly SETBP1Alliance Collaboration Call

3rd Qtr
- MDBR hosted by UPenn ODC in Philly
- ORCA study family participation
- SETBP1 & Simons Searchlight Family & Research Conference
- Global Genes Summit and/or NORD Conference

4th Qtr
- Launch 2nd phase of SCoReS Collaboration project with Tarleton State University
- Establish multidisciplinary SETBP1 clinic
- Review MDBR grants & announce grant recipients
- Quarterly SETBP1Alliance Collaboration Call
- GivingTuesday Fundraiser

2022 Primary Focuses

- Educate, Unite Community & Drive Engagement with the in-person Family & Research Conference, Social Media Presence, and Education Events
- Commit to 2 Fundraisers (Million Dollar Bike Ride & GivingTuesday)
- Expand & Unite the SETBP1 research network out further under the SETBP1Alliance Collaborative Research Network
- Establish the first multidisciplinary SETBP1-related disorders clinic
- Continue to promote and fund research focused on expanding research tools for studying SETBP1-HD, identifying affected pathways or targets for treatment, and validation of identified molecular targets (biomarkers & surrogate endpoints)

We are fighting everyday to give our kids their best life. SETBP1 Society is there beside you providing resources, community, and encouragement. We are supporting & funding research to identify more ways to help our kids. Thank you community for your love & support!

Be the Hope! Be the Change!
Warm regards, Haley Oyler - President & Founder