
SETBP1 SOCIETY

# 2022 Highlights
# President's EOY Report

# 2022 Community

- **178** families connected to SETBP1 Society from **34** countries
- **190** documented cases of SETBP1-HD or SETBP1-related disorder*
- **68** signed up for Simons Searchlight with **54** genetic reports provided & **29** SETBP1-HD variants (~54-59%)
- **9** SETBP1 families attended the SETBP1 Family & Research Conference
- **7** signed up for RARE-X

# 2022 Fundraising

- Million Dollar Bike Ride **$91,664**

  Goal: Fund pathways, novel therapies, natural history studies, and.or biomarkers* (1 or 2 grants)

  - **12** Fundraisers
  - **14** In-Person Riders**

- GivingTuesday >**$28,500**

  Goal: Fund first SETBP1 Multidisciplinary Clinic

  - **11** Fundraisers
  - **148** Unique Donors**

- Simons Searchlight Grants for SETBP1 Research at Conference: ~**$40,000**

  Types: EEGs, DNA Methylation Studies, Neuropsych testing, Speech and Language testing, Additional surveys

# Total Funds

- Million Dollar Bike Ride **$331,819**
- SETBP1 Society Directly or Co-Funded:
  Grants ~**$58,400 (SS - $49,200*)**
- Total SETBP1 Society Funds to Date:
  **$269,780****

2022 Patient Research

## New Studies

- RARE X (natural history study - Xcelerate RARE Open Science Challenge)
- SETBP1 Disease Concept Study COMBINED
- ORCA Observer-Reported Communication Ability Measure (Observer-Reported Communication Ability Measure) Duke University School of Medicine
- SCoReS Phase 2 (Parent Needs Assessment & Child Assessments)
- SETBP1 DNA Methylation Study Boston Children's Hospital Until every child is well Greenwood Genetic Center
- SETBP1 EEG Study USC South Carolina

## Ongoing Studies

- Speech Tracker (3 year natural history speech study) murdoch children's research institute
- SIMONS SEARCHLIGHT (natural history study)
- Patient Data Collection Boston Children's Hospital Until every child is well Radboudumc

2022
Model
Research

## New Studies

- Mice (Cell-type function of SETBP1)
- SETBP1 Protein-Binding Drug Discovery
- SETBP1 LOF Zebrafish (Creation & Characterization)
- Classification of SETBP1 missense variants

## Ongoing Studies

- 2D & 3D SETBP1 Cellular Models
- Mice (Reversible Mouse Model Study, Behavior Characterization, Brain Circuitry / Neuronal Impact)
- SETBP1 Protein Structure
- Classification of SETBP1 missense variants

# 2022 Awareness / Education

- SETBP1 Family & Research Conference hosted by Simons Searchlight
- Hosted/Co-hosted **7** Educational Sessions
  - **2** It Takes a Village Educational Series
  - **2** SETBP1 Family Chats
  - **2** RARE-X Informational Interactive Sessions
  - **1** Speech/Language/Speech Tracker Session
- Released **2** Press Releases
- Represented SETBP1 Society at Global Genes Summit & RARE-X Summit
- SETBP1 Research presented at Society for Neuroscience (poster) & Southwestern Psychological Association (presentation)

# GOALS for 2023

## Identify & Support Parent Needs

- Build out Board Underrepresentation
- SCoReS collaboration
- 2023 Community Assessment Survey
- Provide Resources based off Results

## Host/Support In-Person Meetups

- In-person meetups promote community and connection
- Proposed locations: Dallas, TX (February), Philadelphia, PA (June), ? (Fall)
- Plan for 2024 In-person Conference

## Refine, Disseminate & Execute Research Strategy

- Establish SETBP1 Multidisciplinary Clinic
- Refine Strategy for Repurposing Approved Drugs
- Collaborate with existing SETBP1 Team
- Fundraise to Support Efforts (MDBR & GivingTuesday)

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# BE THE HOPE
# BE THE CHANGE

SETBP1 SOCIETY