



# 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\*\*





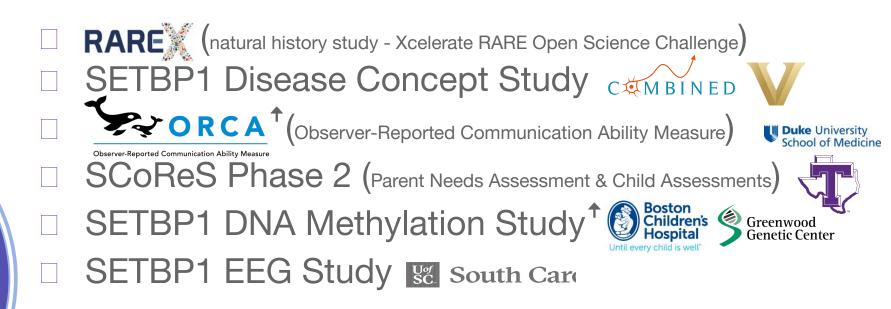
 Simons Searchlight Grants for SETBP1
 Research at Conference: ~\$40,000 SIMONS SEARCHL\$GHT
 Types: EEGs, DNA Methylation Studies, Neuropsych testing, Speech and Language testing, Additional surveys Total Funds Million Dollar Bike Ride \$331,819

SETBP1 Society Directly or Co-Funded:
 Grants ~\$58,400 (ss - \$49,200\*)

Total SETBP1 Society Funds to Date:
 \$269,780\*\*



#### **New Studies**



## **Ongoing Studies**

2022

Patient

Research

- Speech Tracker (3 year natural history speech study)
- SIMONS SEARCHLØGHT (natural history study)
- Patient Data Collection Boston Children's Radboudumc



#### **New Studies**

2022

Model

Research

Mice (Cell-type function of SETBP1) UTSouthwestern Medical Center SETBP1 Protein-Binding Drug Discovery THE UNIVERSITY OF ALABAMA IN HUNTSVILLE Oueens SETBP1 LOF Zebrafish (Creation & Characterization) McMaster University 🔛 Classification of SETBP1 missense variants TELETHON **Ongoing Studies** мах NCK MAX PLANCK INSTITUT 2D & 3D SETBP1 Cellular Models St McGill CARDIF UNIVERSITY Mice (Reversible Mouse Model Study, Behavior Characterization, Brain Circuitry / Jan and Dan Duncan Neuronal Impact) Baylor College of Medicine Neurological Texas Children's Hospital\* Research Institute SETBP1 Protein Structure **DEGLI STUDI** DI PADOVA Classification of SETBP1 missense variants MAX NCK

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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)





#### **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)





# BE THE HOPE BE THE CHANGE

