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
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

- RAREX (natural history study - Xcelerate RARE Open Science Challenge)
- SETBP1 Disease Concept Study
- ORCA (Observer-Reported Communication Ability Measure)
- SCoReS Phase 2 (Parent Needs Assessment & Child Assessments)
- SETBP1 DNA Methylation Study
- SETBP1 EEG Study

Ongoing Studies

- Speech Tracker (3 year natural history speech study)
- Patient Data Collection
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)
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