

ANNUAL IMPACT



REPORT



LOVE FEARLESSLY, SHINE FEARLESSLY!

Our mission is to improve the quality of life for children and families impacted by SHINE Syndrome through supporting collaborative research among international medical professionals, nurturing a family community, and raising worldwide awareness.

	Projects	Details	Outcome
Advocacy	ICD-10 Code, Medical Considerations	We applied and presented to the CDC in September	Results still pending; Medical Considerations document to be published
Research	AAV9, mice model, CB, epilepsy	Fully funded an AAV9 project with HFHF	One successful DLG4 mouse colony, AAV9 research in progress
Community	Rare Bears, Meet-up, Virtual Meetups	Excellent family participation in various activities	Largest DLG4 family meetup to date at the Cincinnati Zoo.



When you have a rare disease,
finding people who truly get it
is everything!

THE YEAR IN REVIEW



The SHINE Syndrome Foundation (SSF) supported several research initiatives this year, including an AAV9 project at Hebrew University in collaboration with the HOPE for Harvey Foundation. We also supported a full-year salary for a postdoc student in Denmark under the leadership of Dr. Zeynep Tümer. Amanda Levy has been critical in running various studies for DLG4 as we learn more about this disorder.

The SSF also supported the collaboration of medical professionals to publish a chapter in Gene Reviews, author a Medical Considerations document for families, and applied and presented for a unique ICD-10 code for DLG4-related synaptopathy. Additionally, we've supported partnerships for families, including Rare Bears, SAMi cameras, joined REN, DEE-P Connections, NORD, and more!

Spring Highlight

Seven DLG4 families met in-person at the Cincinnati Zoo for a fun afternoon and dinner. The next day, Laura Palmer pushed her son with SHINE Syndrome, Nolan, in the Flying Pig Marathon to raise awareness and funds for SHINE.

Summer Highlight

The SHINE Syndrome Foundation hosted our annual awareness day on July 17 alongside our annual sunSHINE Campaign. Several families participated in the COMBINEDBrain biomarker project and donated samples.

Fall Highlight

Dr. Tümer presented to the CDC on behalf of the DLG4 community to advocate for a unique ICD-10 code for DLG4. Several board members attended the Global Genes and NORD conferences.

Winter Highlight

Our first mouse colony was deemed a success and researchers started testing and running experiments on the mice. The first paper describing the epilepsy phenotype in DLG4 was published in December.



FINANCES

Current Assets

Cash as of 12/31 \$38,979

Liabilities

None as of 12/31 \$0

Revenues

Fundraising \$78,806
Grants \$5,000

Expenses & Deductions

Research Projects \$146,413
Operating Expenses \$2,476

FUNDING SUMMARY

\$78,806

Donations

\$5,000

Grants

WE'D LIKE TO THANK ALL OUR DONORS FOR THEIR GENEROUS SUPPORT

ACKNOWLEDGEMENTS

Thank you so much to the entire board of the SHINE Syndrome Foundation, to the DLG4 families who hosted fundraisers within their communities, and to our Medical Advisory Board for their expertise and unwavering support as we work towards knowledge and a treatment for the DLG4 community.

