



About International Rett Syndrome Foundation

The International Rett Syndrome Foundation (IRSF) is the leading research, advocacy, and family empowerment organization for Rett syndrome. We're on a mission to transform the lives of all those affected by Rett syndrome by accelerating research toward treatment and a cure while empowering families with the information, programs, and services they need to thrive.

Over nearly 40 years, IRSF has invested **more than \$58 million** to fund breakthrough discoveries and life-changing advancements in research while supporting **thousands of individuals and families** around the world at all stages in their Rett journey.

Our vision is to create a world without Rett syndrome.

MISSION STATEMENT

To accelerate full spectrum research to cure Rett syndrome and empower families with information, knowledge, and connectivity.

ABOUT RETT SYNDROME

Rett syndrome is a rare genetic neurological disorder that occurs mostly in females (1 in 10,000 births), with a growing number of males being identified, and leads to severe impairments that affect nearly every aspect of life. Rett syndrome is usually recognized in children between 6 to 18 months as they begin to miss developmental milestones or lose abilities they had gained, including their ability to speak, walk, eat, and even breathe. The hallmark of Rett syndrome is near constant repetitive hand movements while awake, and individuals with Rett may experience seizures, scoliosis, breathing issues, GI issues, and more. Rett syndrome is not a degenerative disorder; individuals can live to middle age or beyond.

IRSF RESEARCH IMPACT

- Funded the research that identified the gene that causes Rett syndrome (MECP2) and proved that it can be reversed in mice.
- Funded the first gene therapy research in Rett syndrome and the first clinical trials on medications to treat Rett.
- Funded early-phase clinical trials of trofinetide, now the first-ever FDA-approved treatment for Rett syndrome known as DAYBUE™ in the United States.
- Created the infrastructure for industry and pharmaceutical partners to invest in Rett research, including the Natural History Study, Rett Syndrome Registry, and Clinical Trial Committee.
- Established a clinical network of 18 U.S. Rett Syndrome Centers of Excellence to ensure everyone with Rett has access to high-quality clinical care.

IRSF FAMILY EMPOWERMENT IMPACT

- Established a network of Family Empowerment Representatives that provide 24/7 support and individualized support for diagnosed families in every state.
- Developed a robust Rett Education program that offers families online, expert-led tutorials on various Rett-related research and care topics, reaching families in 60+ countries.
- Created the IRSF Rett Research Ready™ Program and myRett Trial Finder tool to prepare families for clinical trial involvement.
- Published the first-ever research-based *Rett Syndrome Primary Care Guidelines*, *Rett Syndrome Communication Guidelines*, and *The Rett Syndrome Handbook*, to enhance quality of care and help those living with Rett thrive.
- Created opportunities for families to connect, share, and learn by hosting signature events, including the family RettAway, Raise a Glass for Rett Gala, regional Strollathons, and ASCEND National Summit.

LINKS

- [Website](https://rettsyndrome.org) (rettsyndrome.org)
- [Facebook](https://www.facebook.com/Rettsyndrome/) (@Rettsyndrome)
- [LinkedIn](https://www.linkedin.com/company/rettsyndrome/) (@rettsyndrome.org)
- [Instagram](https://www.instagram.com/rettsyndromeorg/) (@rettsyndromeorg)
- [Twitter](https://twitter.com/Rettsyndrome) (@Rettsyndrome)
- [YouTube](https://www.youtube.com/channel/UC1vHjZgDp0GjYnYdJpYyHw) (@IRSFoundation)



2023 IRSF Talking Points

Thanks to your generous support, the horizon is very bright. Events like this have made a significant difference in advancing Rett syndrome research toward treatments and ultimately a cure. Because of you:

- IRSF committed to fund more than **\$4.4M in research investments** to set up the next wave of Rett breakthroughs, including research that:
 - Investigates genetic treatments as curative approaches
 - Paves the path to new drugs to treat Rett syndrome
 - Ensures clinical trial readiness and success
- The FDA approved Trofinetide as the **first-ever FDA-approved treatment for Rett syndrome**, now known in the U.S. as Daybue™. IRSF invested nearly \$2M to help to fund Phase 2 adult and pediatric clinical trials of this compound and supported the Phase 3 trials through identifying clinical trial sites and promoting participant recruitment.
- The **first adult patient with Rett syndrome was dosed in a gene therapy clinical trial** in Canada, made possible by discoveries by an IRSF-funded researcher. The patient showed clinical improvements at 4-weeks post-treatment, clearing the way for the launch a pediatric study in the U.S. and U.K. soon. And **recruitment began for a gene therapy pediatric clinical trial** at 3 U.S.-based clinical trial sites that are part of IRSF's Center of Excellence clinic network.
- In 2022, 85% of expenses support our core mission, and with income from investments and IRSF reserves, we committed **100% of every dollar donated to accelerating research and empowering families**. We're proud to be a top-rated charity. (FY22)
- There are nearly **40 active Rett syndrome research projects** around the world funded thanks to IRSF donor and supporters.
- More than **20 pharmaceutical companies** are currently working on treatments that we hope could bring us even closer to a world without Rett. Fifteen years ago, there were none.
- IRSF supports a thriving Rett syndrome community, connecting **thousands of families** with the resources they need wherever they are on their Rett journey.
- Experts and families around the world have access to educational resources to help individuals with Rett syndrome thrive. Our RettEd Webinar series has been viewed by families in **more than 200 countries and territories**.
- IRSF just launched **My Rett Ally**, a web-based app to support U.S. parents and caregivers. The free tool helps families simplify, organize, and share their loved one's most important medical and care information all in one secure place.