



## Our Mission

The DESSH Foundation supports individuals and families affected by **DeSanto–Shinawi syndrome (DESSH)** by advancing research responsibly, strengthening the patient community, and advocating for greater understanding and care of this ultra-rare genetic condition.

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## 2025 at a Glance

- **234 individuals with DESSH** connected through the Foundation
- Families represented across **41 countries**
- **962 subscribers** in our family, clinician, and advocate community
- **3,100+ hours (or \$98,000) of volunteer labor**

## Building Community & Support

In 2025, the Foundation focused on connection, education, and trust:

- **3 community calls** for shared learning and updates
- **2 virtual events**, including the **Rare Art Show** for Rare Disease Day and the **Dash 4 DESSH 5K**
- **44 patients seen at multidisciplinary clinic** in collaboration with Washington University, St. Louis Children’s Hospital, and the IDDRRC
- **Family conference** dedicated to education and connection
- Continued partnership with Parent to Parent USA to strengthen peer support

## Research & Progress

DeSanto–Shinawi syndrome is caused by pathogenic variants in the **WAC gene** and currently has **no approved treatment**.



## Research & Strategic Investment

In 2025, the DESSH Foundation made **targeted, high-leverage investments** designed to advance the field of DeSanto–Shinawi syndrome research and position the community for future clinical investigation.

- **\$79,000 invested** in research initiatives supporting foundational science and clinical readiness
- Advanced disease modeling through an **iPSC cell line project** with the **Gabel Lab** and **Kroll Lab**
- Continued support of a **Natural History Study** at **Washington University**, led by **Dr. Shinawi**, to define meaningful clinical outcomes
- Maintained ongoing guidance from the Foundation's **Medical Advisory Board** (7 members across academia, clinical care, and industry)
- Supported a **multidisciplinary DESSH clinic** in collaboration with **Washington University, St. Louis Children's Hospital, and the IDDRC**, strengthening clinical insight and care coordination

Together, these investments move the field from isolated observations toward **trial readiness and translational research**.

## Lean Operations, Maximum Impact

- **\$106,000 raised** in 2025
- **\$79,000** directed to research
- **\$27,000** total operational costs
  - **\$14,400** supporting one part-time staff position
  - **\$12,600** covering all remaining administrative expenses

This structure allows the Foundation to remain **scientifically credible, operationally lean, and mission-focused**.



## Our Broader Network

The DESSH Foundation is an active member of:

- **Rare Epilepsy Network (REN)**
- **NORD**
- **COMBINED Brain**
- **Global Genes**
- **Rare-X**

These partnerships help ensure DESSH is represented within the broader rare disease ecosystem.

Founded in **2019**, the DESSH Foundation has been continuously active since its inception.

## Looking Ahead

In the coming year, the Foundation will continue to:

- Support families with clear, transparent communication
- Advance ethical, patient-centered research pathways
- Strengthen collaboration with clinicians, researchers, and aligned organizations

## With Gratitude

Our progress is made possible by families, donors, researchers, and advocates who believe in thoughtful, responsible advancement. Thank you for standing with the DESSH community.

 [dessh.org](https://dessh.org)