



Clinic and Family Conference

September 30, 2023





The DESSH Foundation

Introduction to the foundation

- Dedicated to the care and advocacy of DeSanto-Shinawi Syndrome patients
- Founded in 2019 by the Piccirillo family
- 501c3 non-profit status in 2020
- Raised ~\$100,000 since inception



History

2019

- The Dash 4 DESSH founding event takes place in April.
- The first family meeting in Houston, TX in October.

2020

- Achieve 501c3 non-profit status
- Form our first Board of Directors and Medical Advisory Board.

2021-2022

- Webinar for families on the importance of research participation.
- Board exhibits at ACMG
- First DESSH Clinic and conference!

Present

- Formalize our research roadmap
- Launch patient registry
- Engage the DESSH community and supporters through active social media presence



Driven by the desire to make a positive impact on the lives of those affected by DeSanto-Shinawi Syndrome.

We are committed to improving the well-being of our community, advocating for research and treatments, and raising awareness about DeSanto-Shinawi Syndrome.



Values



We believe in the power of the patient

The DESSH Foundation bridges the gap between science and the human experience, improving the lives of our community.

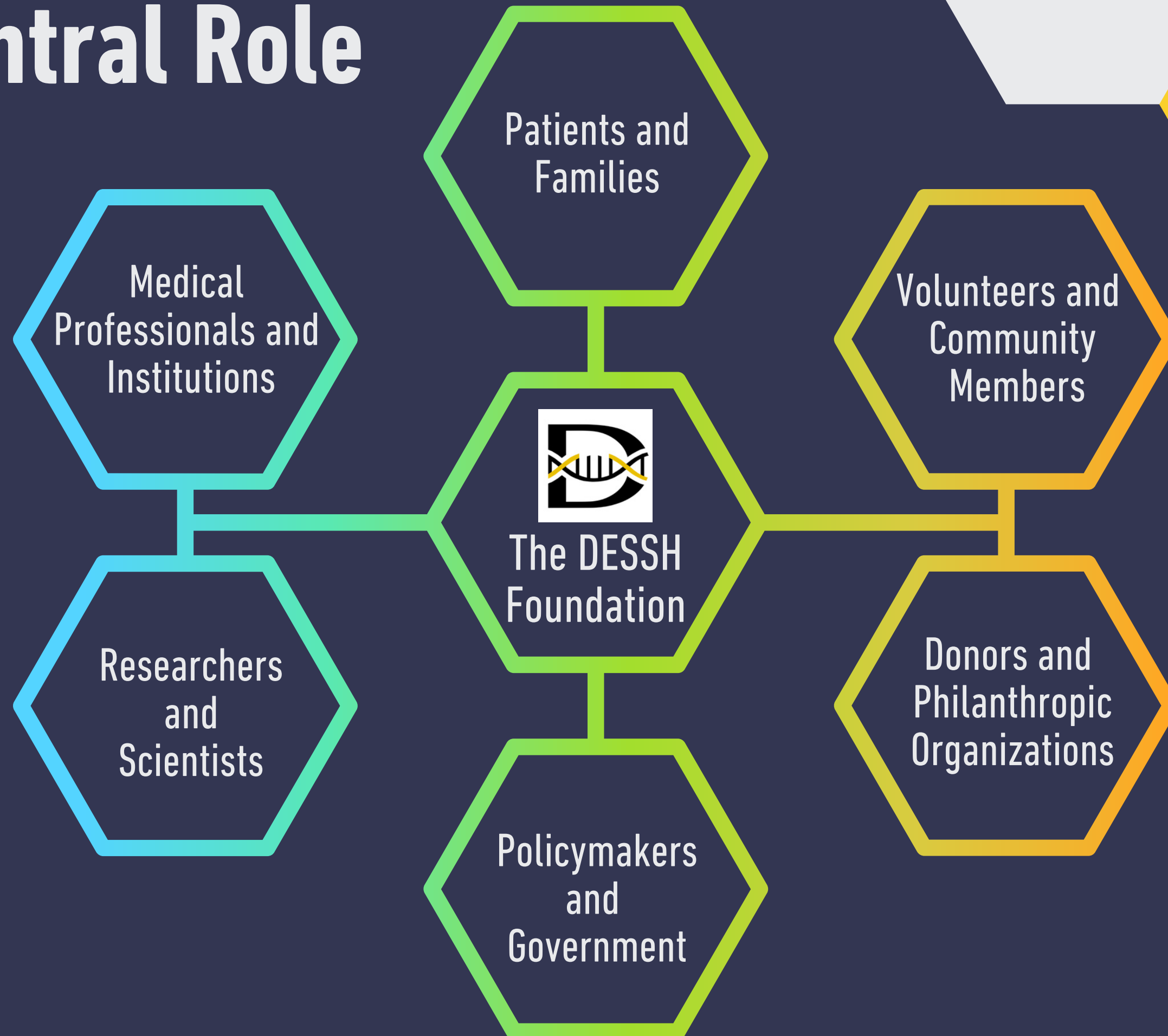
We believe in the power of data

Our collection of data serves as the compass guiding DeSanto-Shinawi Syndrome research, illuminating the path toward treatments and cures.

We believe in collaboration

The DESSH Foundation serves as a unifying force, connecting stakeholders who are committed to addressing the needs of those impacted by DESSH.

PAG's Central Role



Goals and Strategy

Patient and Family Support

- Build a network through social platforms
- Provide resources and opportunities, including meetups
- Partner with the broader rare disease community

Raise Awareness

- Exhibit / attend rare disease conferences
- Participate in global initiatives like Rare Disease Day
- Leverage news, social media, and website

Advance Research

- Establish research roadmap and follow with fidelity
- Build natural history study (DESSH Clinic)
- Launch patient registry

Fundraise

- Nurture donor relationships
- Seek grant opportunities
- Host events like Dash 4 DESSH
- Pursue passive avenues like Spreadshirt and minted.com

Organizational Excellence

- Acquire/retain quality Board of Directors and committee volunteers
- Listen to our patient community
- Follow the guidance of our medical advisors

RARE RESEARCH ROADMAP

Let's Collaborate!



We Are Here

Basic Research

Functions of WAC Gene

animal models (2000 - ongoing)

- drosophila models
- mouse models

Impact of Mutations / Loss of WAC Protein

case studies (2015 - ongoing)

animal models (2020 - ongoing)

- zebrafish models
- mouse models

natural history studies (2022 - ongoing)

baseline clinical data (2022 - ongoing)

biobank / tissue (2022 - ongoing)

patient registry (2023 start)



Toolbox

animal models

- zebrafish models
- mouse models

biobank/tissue

baseline clinical data

natural history studies

patient registry

Join Our Patient Registry!



Key Dates and Upcoming Events

October 21st-22nd

Dash 4 DESSH

November 28th

Giving Tuesday

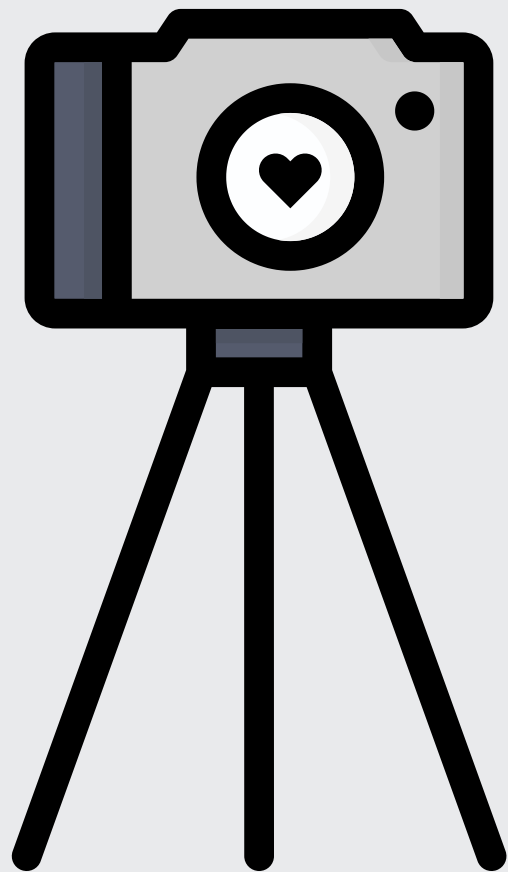
February 29th, 2024

Rare Disease Day

**Patient
Registry
Launches
Late Fall**



Join us for a
group photo on
the 3rd floor!



Thank You!

