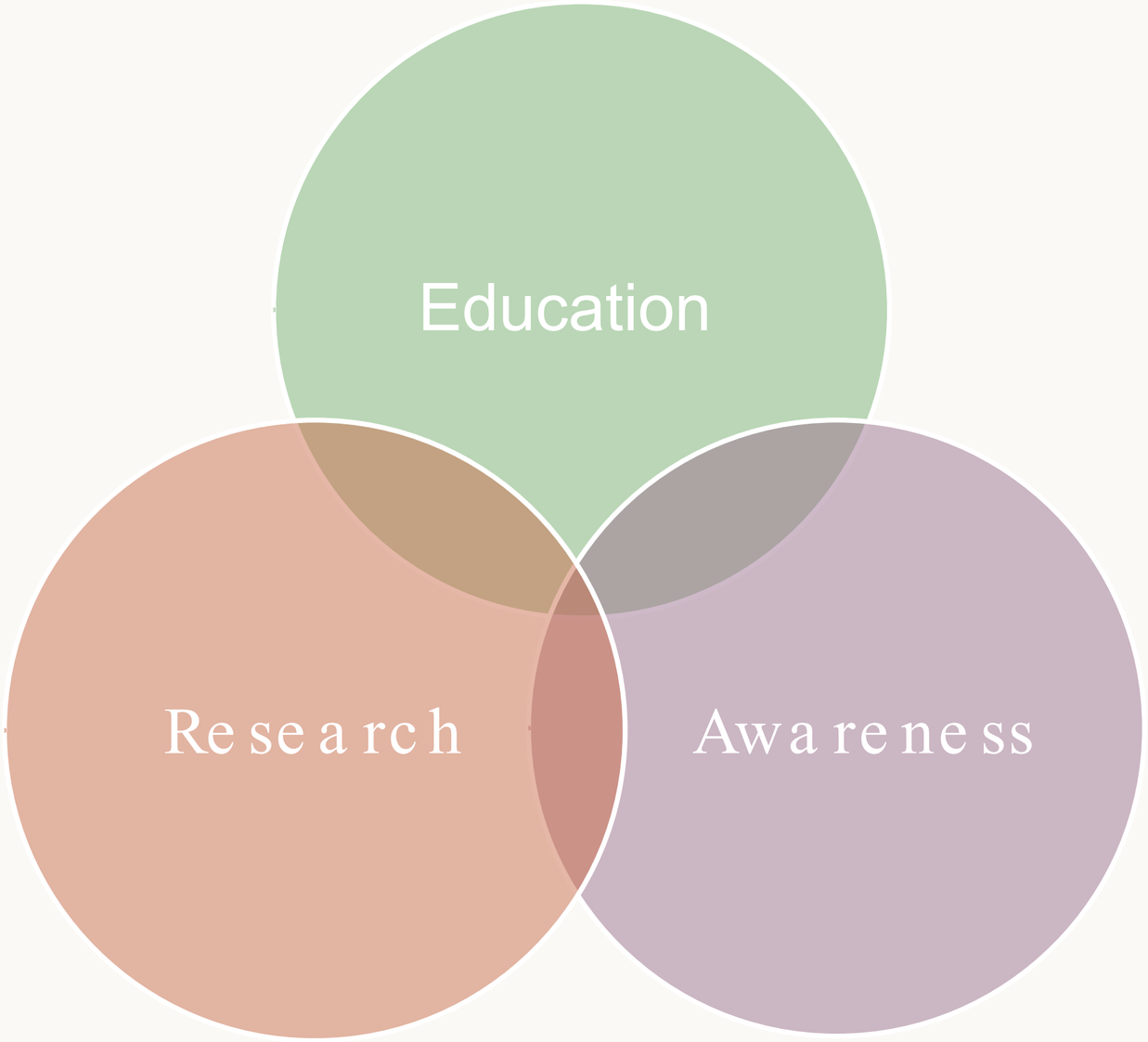
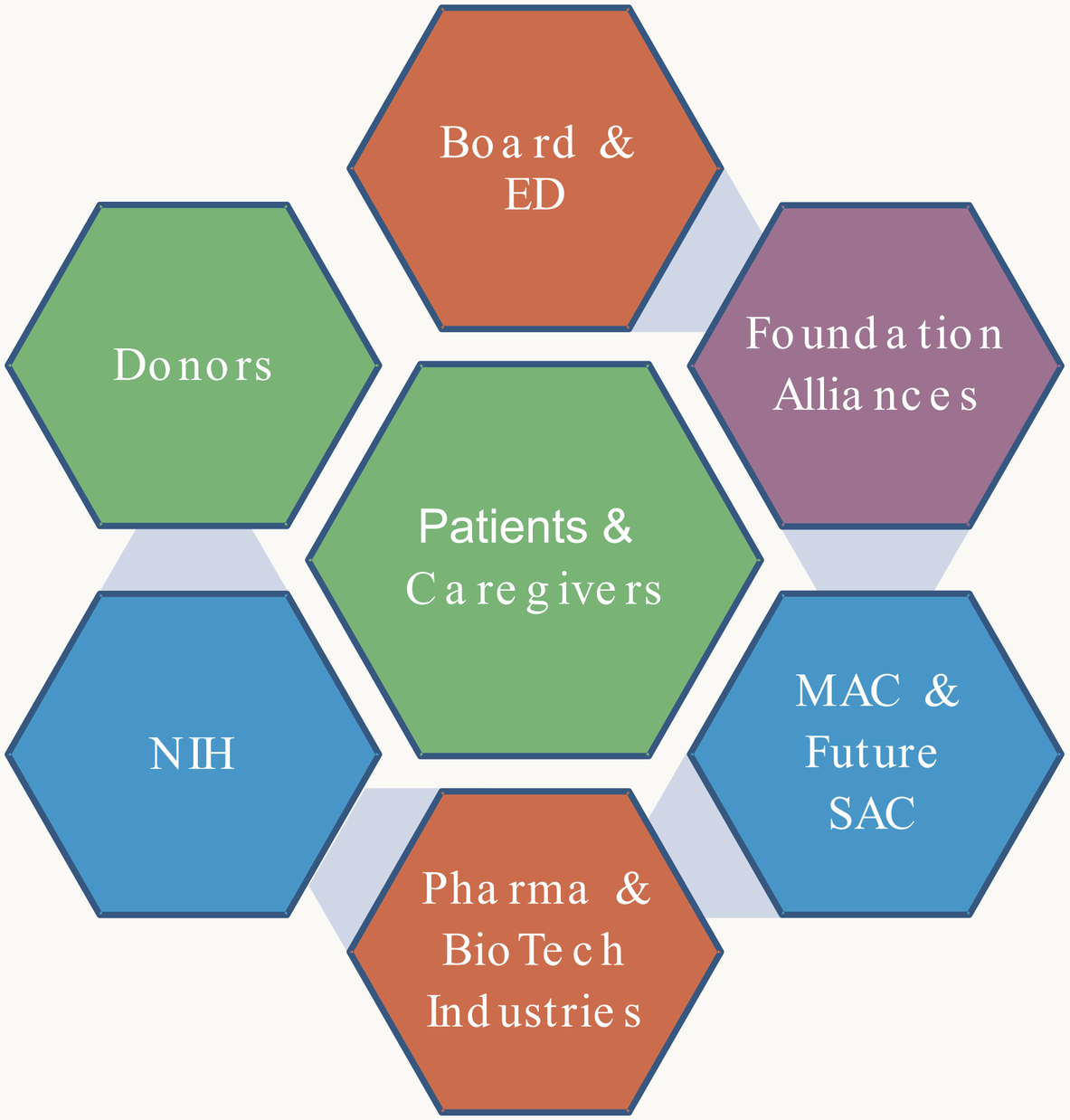
THE APS TYPE 1 FOUNDATION

JUNE 18, 2024 | JENNIFER ORANGE

OUR MISSION



WHO WE ARE


Board & ED
Donors
Foundation Alliances
Patients & Caregivers
NIH
MAC & Future SAC
Pharma & BioTech Industries

APS TYPE 1 FOUNDATION BOARD

NEW EXECUTIVE DIRECTOR

Svetlana Hutfles

MEDICAL ADVISORY COMMITTEE

Dr. Mark Anderson, MD, PhD, is a professor and physician scientist in the UCSF Diabetes Center and a recognized expert in the genetic underpinnings of autoimmune diseases and the control of immune tolerance. He helped establish a genetic mouse model of APS Type 1 to help understand how tolerance is disrupted in APS Type 1 patients.

Dr. Richard J. Auchus, Professor of Pharmacology and Internal Medicine, received his MD and PhD from Washington University. He completed training in internal medicine at the University of Iowa Hospitals and a fellowship in Endocrinology at the Wilford Hall in USAF Hospital and the University of Texas Health Sciences Center in San Antonio. Dr. Auchus' research includes clinical and translational investigation in disorders of the pituitary, adrenal, ovaries and testes.

Dr. Michail S. Lionakis, Chief, Fungal Pathogenesis Section Laboratory of Clinical Immunology and Microbiology, NIAID, NIH, obtained his MD and ScD from the University of Crete, Greece. He did clinical and research training at MD Anderson Cancer Center, Baylor College of Medicine and NIH. His IRB-approved APS Type 1/ APECED clinical research protocol aims to understand the mechanisms of autoimmunity and fungal susceptibility and improve diagnostic and therapeutic strategies for patients.

MEDICAL ADVISORY COMMITTEE

Dr. Dana Orange, MD, MSc, is an Associate Professor at Rockefeller University, received her medical degree from Weill Cornell Medical College, Cornell University, and her MSc from Rockefeller University. She completed her Internal Medicine Residency at New York Presbyterian Hospital and her Rheumatology Fellowship at the Hospital for Special Surgery. Dr. Orange is facilitating research on APS Type 1 as a principal investigator of the APS Type 1 (APECED) Registry, an IRB approved protocol.

Dr. Maureen A. Su, Professor of Microbiology/ Immunology and Medical Genetics and Pediatric Endocrinology at UCLA, received her bachelor's, master's and medical degree from Harvard. She completed her pediatrics residency and fellowship in endocrinology at UCSF. Dr. Su seeks to understand what causes autoimmune diseases in order to develop therapeutics to prevent and treat the underlying immune condition. Her work focuses on APS Type 1, GBS/ CIDP, and T1D.

WHAT WE DO

EDUCATE

International Symposia

Global Scientific Summits

SUPPORT RESEARCH

Web-based Registry

Research Grants

RAISE AWARENESS

Conference Talks

Extensive Website Materials

Social Media Presence

INTERNATIONAL SYMPOSIA

Our patients and families at the 2017 Symposium in Stony Brook, NY

Our patients and families at the 2019 Symposium in National Harbor, Maryland

INTERNATIONAL SYMPOSIA

Our patients, doctors and families at the 2023 Symposium in Washington, DC

GLOBAL SCIENTIFIC SUMMIT 2022

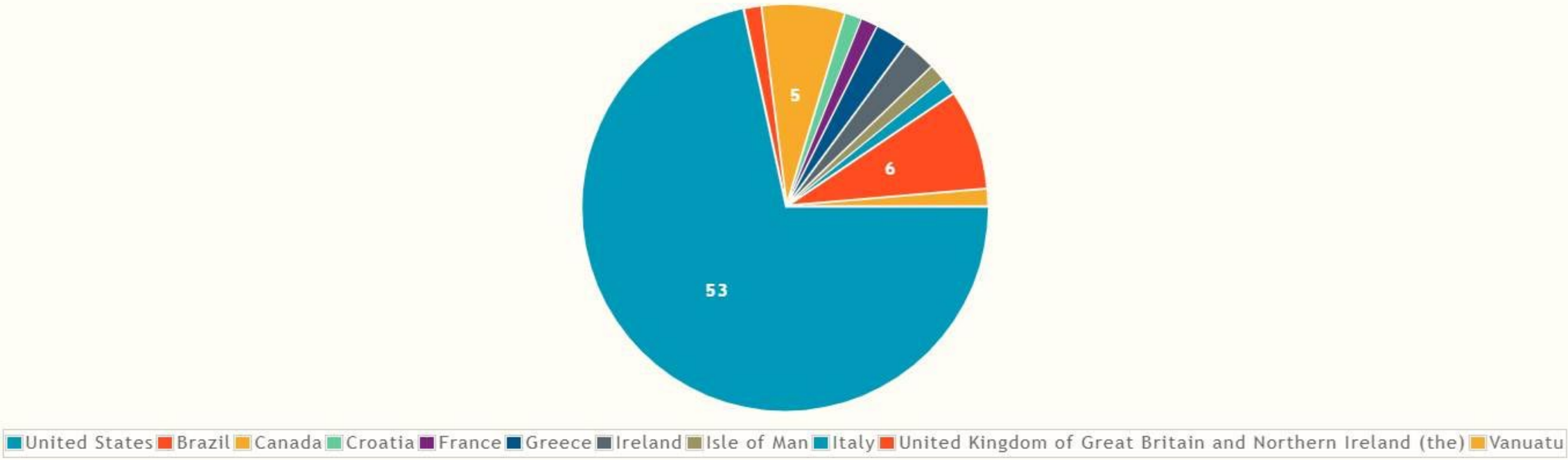
OUR ACCOMPLISHMENTS

Raised over $700K for Research

Developed a Web-based Registry

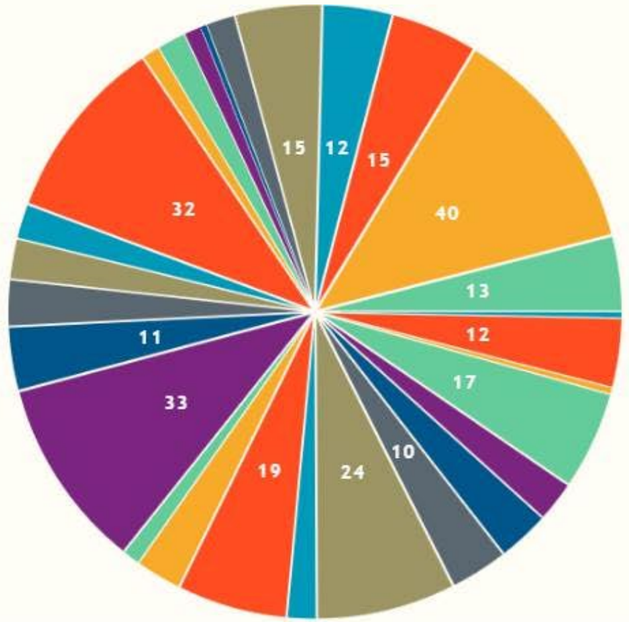
THE APS TYPE 1 (APECED) REGISTRY

Our registry has 133 patients registered from 11 countries.



THE APS TYPE 1 (APECED) REGISTRY

APS Type 1 leads to a large range of different symptoms and conditions.



None	Alopecia (hair loss)	Asplenia (spleen dysfunction and infection susceptibility)	Autoimmune Hepatitis (liver inflammation)
Chronic Bloating	Chronic Constipation	Chronic Diarrhea	Chronic Mucocutaneous Candidiasis
Early onset Hypertension (high blood pressure)	Tooth Enamel Dystrophy	Gastritis (stomach inflammation)	Growth Hormone Deficiency
Hypoparathyroidism	Hypothyroidism	Keratoconjunctivitis (eye inflammation)	Ovarian Failure
Pneumonitis (lung inflammation)	Addison's Disease (Primary Adrenal Insufficiency)	Severe Fat Malabsorption	Sjogrens-Like Syndrome (dry eyes or mouth)
Testicular Failure	Tubulointerstitial Nephritis (kidney problem)	Type 1 Diabetes (juvenile diabetes)	Urticaria (hives)
Vitamin B12 Deficiency	Vitiligo (patches of white skin)	Other	Nail dysplasia

Education Initiative

Genetic Testing Decision Tree

Autoimmune Polyglandular Syndrome Type 1 (APS Type 1)

APS1
The APS Type 1 Foundation Inc.

Autoimmune Polyglandular Syndrome Type 1 (APS Type 1), also known as Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy (APECED), is a rare genetic disorder that is often misdiagnosed. Mutations in the AIRE gene lead to three classic manifestations along with other common symptoms. This resource aims to assist you with diagnosis and referring a patient to a genetics professional. For additional information, visit the APS Type 1 Foundation website or review this research article.

Classic Manifestations

1. **Chronic Mucocutaneous Candidiasis**
Patients may experience recurring and long-lasting fungal infections that affect the skin and mucous membranes.

2. **Autoimmune Hypoparathyroidism**
When the parathyroid glands malfunction, low serum calcium may lead to patients experiencing seizures, fatigue, muscle pain and cramping, and a tingling sensation in lips, fingers, and toes.

3. **Adrenal Insufficiency (Addison's Disease)**
Malfunction of the adrenal glands leads to muscle weakness, loss of appetite, weight loss, low blood pressure, and changes in skin coloring.

Adjunct Triad

1. **Urticarial Eruption (hives)**
Patients often experience a recurrent maculopapular rash that typically resolves on its own and is not usually itchy but can be.

2. **Enamel Hypoplasia**
Patients may have a deficiency in enamel production, making the teeth appear yellow or spotted, with or without cavities.

3. **Intestinal Malabsorption**
When nutrients are not properly absorbed by the small intestine, patients experience increased fecal fat and disruption of the gut microbiome.

Does your patient have...

symptoms for one classical manifestation + symptoms for a manifestation within the adjunct triad?

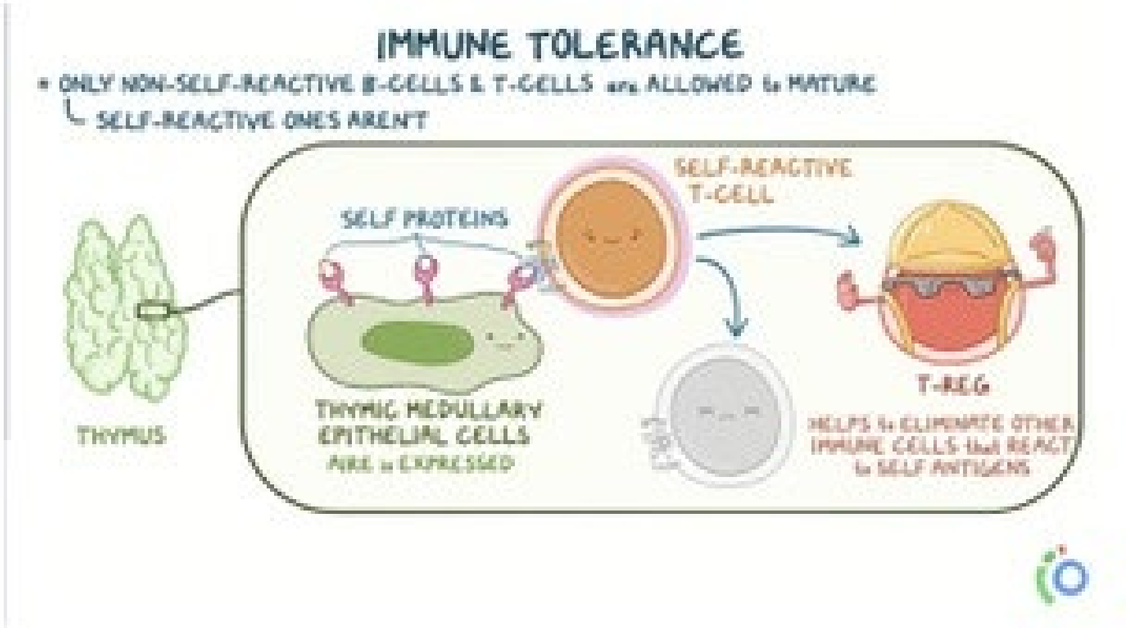
OR

symptoms for two of the classic manifestations?

→ It is recommended that your patient be referred to a genetics professional for genetic testing of the AIRE gene.

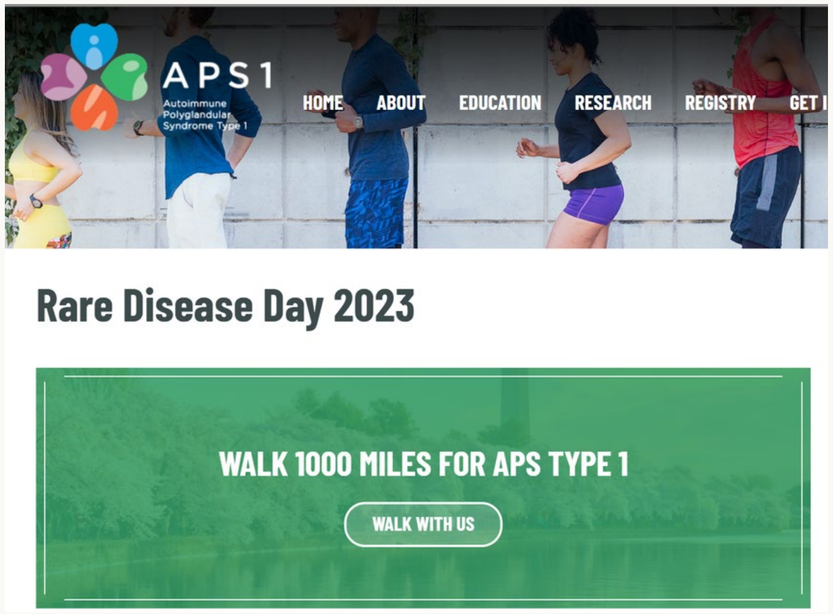
Education Initiative

What is APS Type 1?



Osmosis.org video on “What is APS Type 1?”

Fundraising: Accomplishments & Opportunities



Our community has found creative ways of fundraising and raising awareness for APS Type 1

PAST FAMILY FUNDRAISERS

OUR VISION FOR THE FUTURE

- Find our patients here and around the world
- Shorten time to diagnosis
- Find better treatments
- Find a cure
- Support the LIVED experience of our patients and families
- Build a global research network
- Expand our registry for greater publication
- Fundraise for all of the above

HOW CAN YOU HELP?

CONNECT YOUR PATIENTS WITH US

BRING IN NEW CLINICIANS & SCIENTISTS

FOLLOW THE FOUNDATION

HOW CAN WE HELP?

CONNECT YOU TO OTHER PATIENTS, FAMILIES & PHYSICIANS

SHARE EDUCATIONAL MATERIALS, RESEARCH AND NEW FINDINGS

BE RECEPTIVE TO ALL OF YOUR IDEAS

Thank you!

National Institute of Allergy and Infectious Diseases

And a big thanks to Robin Finch and Julia Band Orange for their help with this presentation!

THANK YOU!

APS1