THE APS TYPE 1 FOUNDATION

APS1

H Help fellow APS Type 1 Families and Patients

U Understand Shared Challenges

D Develop Connections

D Dispense Knowledge

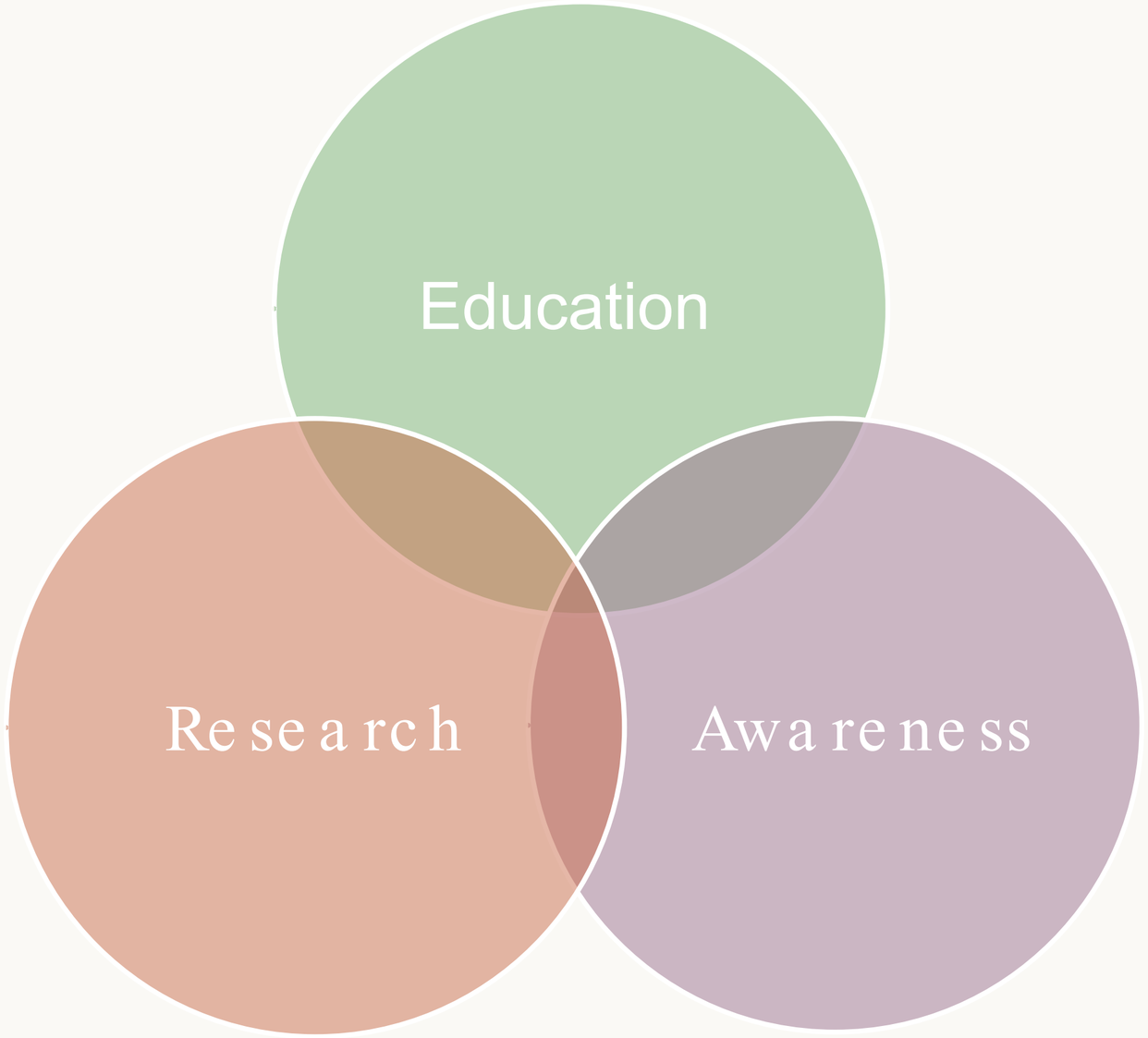
L Learn from Each Other

E Empower the APS Type 1 Community

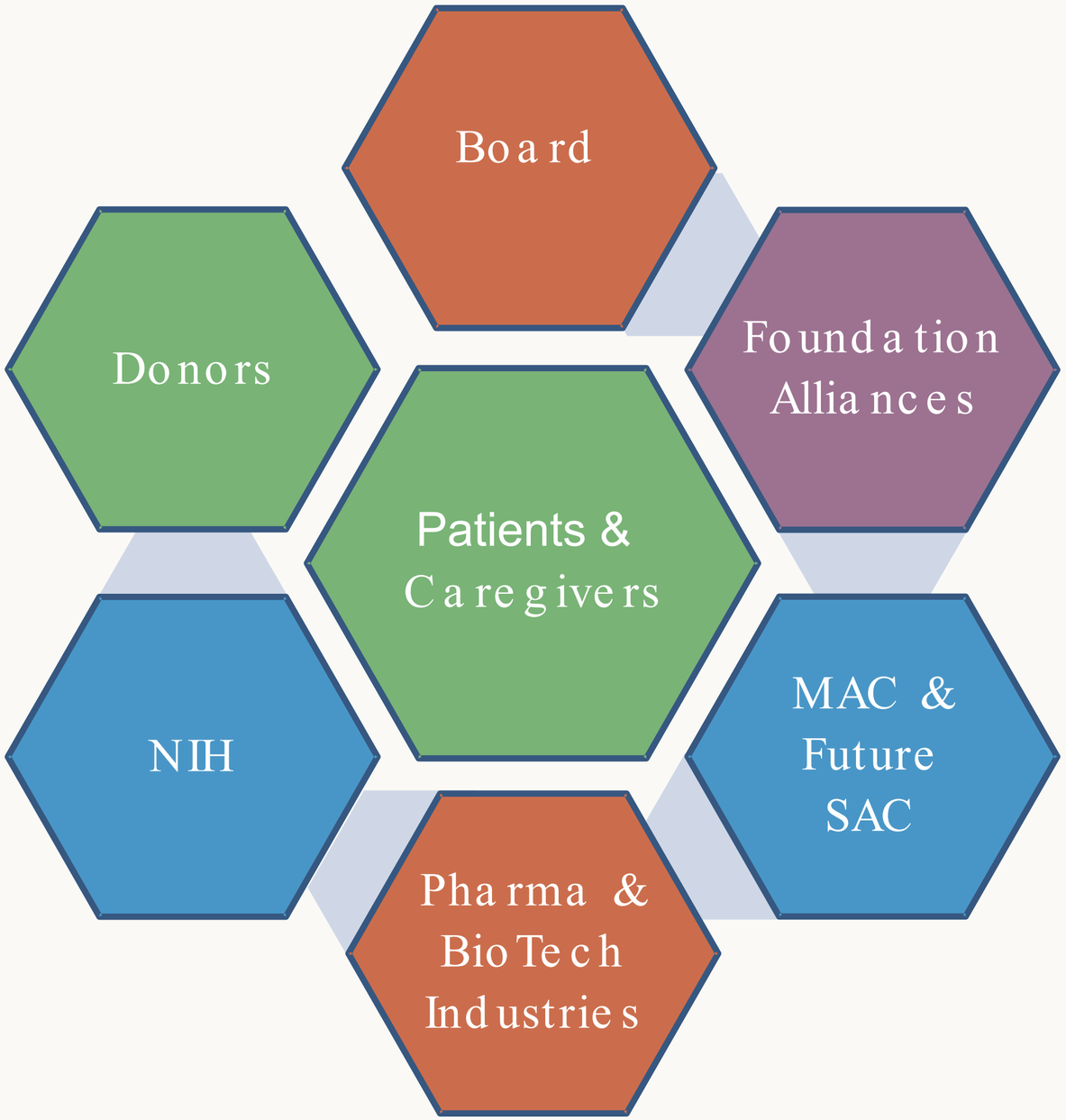
HUDDLE Rules of the Road

- Lead with Kindness: This is a supportive space. Please treat everyone with compassion and respect.
- Respect First: Everyone’s experience is valid. Respect diverse perspectives and experiences.
- Honor Confidentiality: Personal stories shared here stay within this community.
- Share at Your Comfort Level: You are welcome to speak or simply listen. Both are valid ways of participating.
- Experiences, Not Medical Advice: We share personal experiences only. The Foundation does not provide medical advice. Please consult your own healthcare professionals for guidance.
- One Voice at a Time: Allow each person the space to be heard without interruption. Mute when not speaking.
- Stay Present & On Topic: Help us create a meaningful and focused conversation for all.

OUR MISSION



WHO WE ARE


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

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 2025 Symposium in San Francisco, CA

Our patients and families at the 2023 Symposium in Washington, D.C.

Next Symposium will be in Summer 2027

OUR ACCOMPLISHMENTS

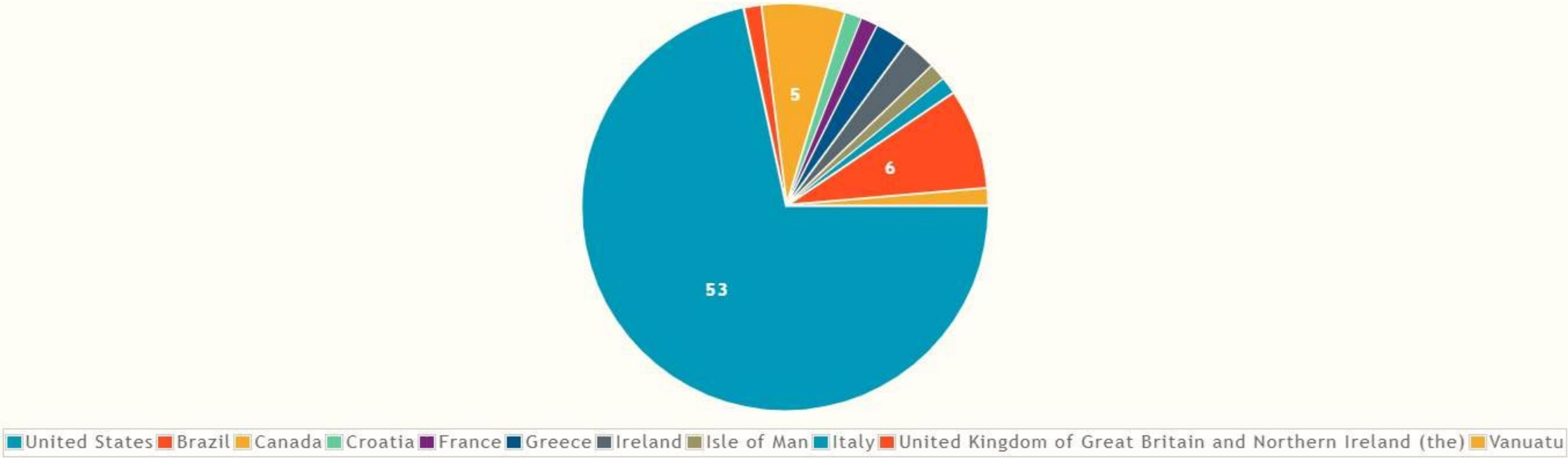
Raised over $700K for Research

Developed a Web-based Registry

THE APS TYPE 1 (APECED) REGISTRY

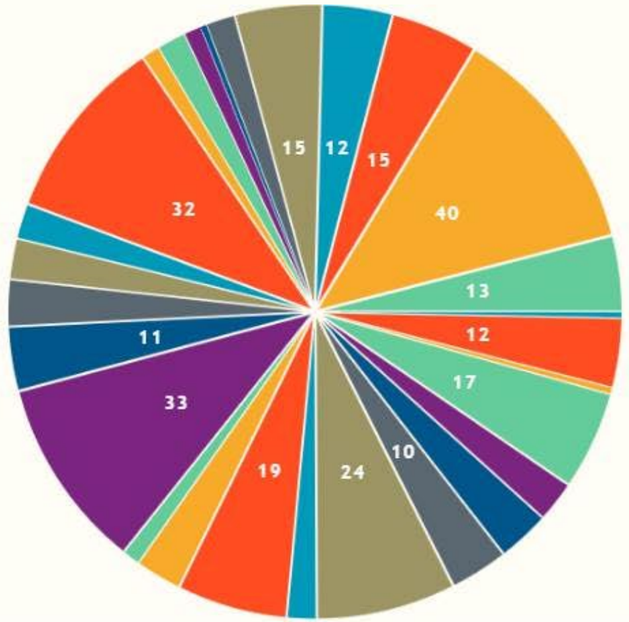
NEW AND IMPROVED PLATFORM LAUNCHING FALL 2026

Our registry has approximately 150 patients registered
from many 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...

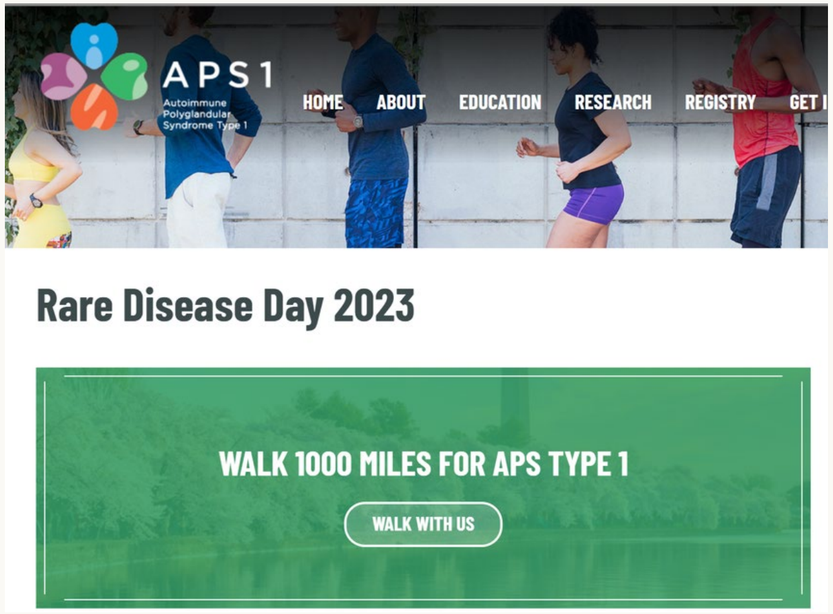
symptoms for one classical manifestation + symptoms for a manifestation within the adjunct triad?

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.

Fundraising: Accomplishments & Opportunities

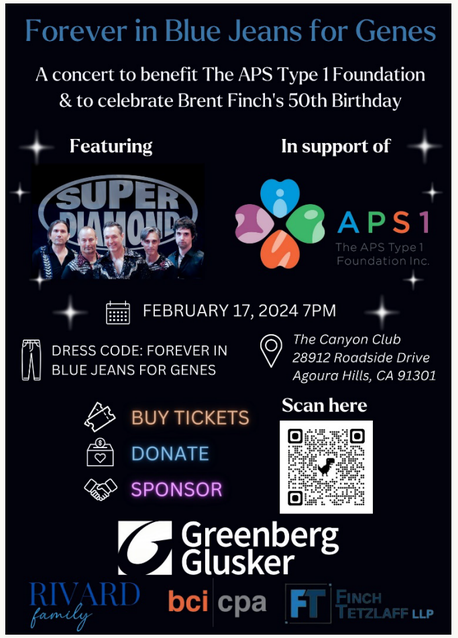


Our community has found creative ways of fundraising and raising awareness for APS Type 1

PAST FAMILY FUNDRAISERS

#liveforlovett

SEYFERT FAMILY BASKET RAFFLES



FOUNDATION ALLIANCES

CORD Canadian Organization for Rare Disorders

indousrare Indo US Organization for Rare Diseases

EVERYLIFE FOUNDATION FOR RARE DISEASES

NORD® National Organization for Rare Disorders

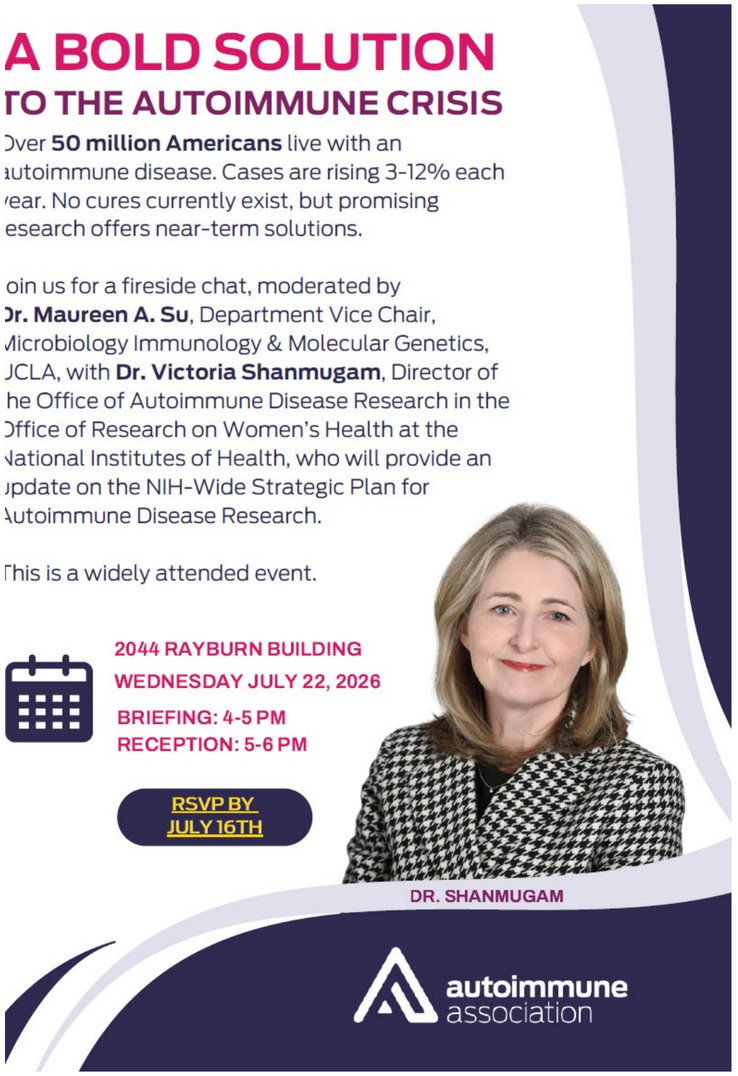
autoimmune association

IDF Immune Deficiency Foundation

Autoimmune Registry

Global Genes® Allies in Rare Disease

Raregivers™ .global



Policy Solutions for Rare Diseases: Improving Access to Care, Treatment, and Community Supports

Wednesday, July 22, 2026 from 12:00 – 1:00 pm ET

Russell Senate Office Building, Room 188 or virtually.

Rare Disease Legislative Advocates, in cooperation with the Rare Disease Congressional Caucus, invite you to a Rare Disease Congressional Caucus Briefing:

Policy Solutions for Rare Diseases:
Improving Access to Care, Treatment, and Community Supports

Wednesday, July 22, 2026 from 12:00 - 1:00 pm ET
Russell Senate Office Building, Room 188 or virtually.
Lunch will be available for in person attendees.

Speakers

- **Carolyn Applegate**, **MGC**, Johns Hopkins Medicine
- **Sarah Chamberlain**, Flok
- **Maynard Friesz**, Cure SMA
- **Jason Harris**, National Psoriasis Foundation
- **Shannon Wood,** Muscular Dystrophy Association

RDLA
Rare Disease Legislative Advocates
POWERED BY THE EVERYLIFE FOUNDATION

JOIN US IN JULY IN D.C.

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?

JOIN FUTURE HUDDLES

ORGANIZE A FUNDRAISER

FOLLOW THE FOUNDATION

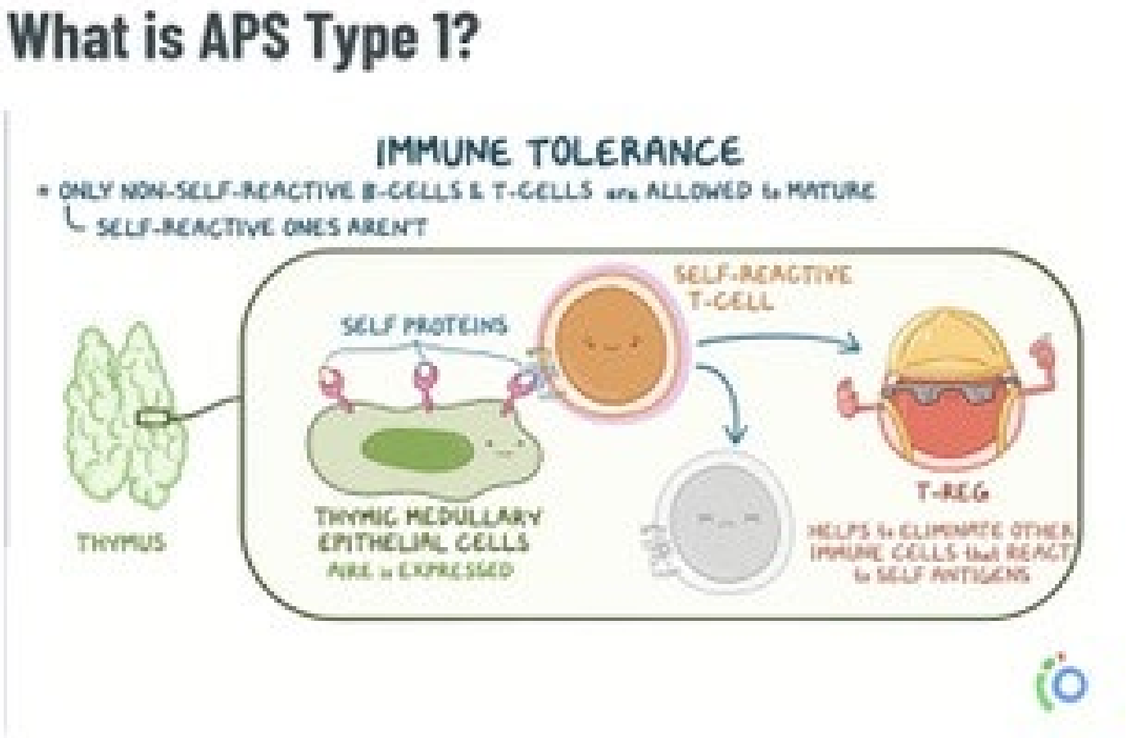
HOW CAN WE HELP?

CONNECT YOU TO OTHER PATIENTS, FAMILIES & PHYSICIANS

SHARE EDUCATIONAL MATERIALS, RESEARCH AND NEW FINDINGS

BE RECEPTIVE TO ALL OF YOUR IDEAS

Education Initiative



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

THANK YOU!

APS1