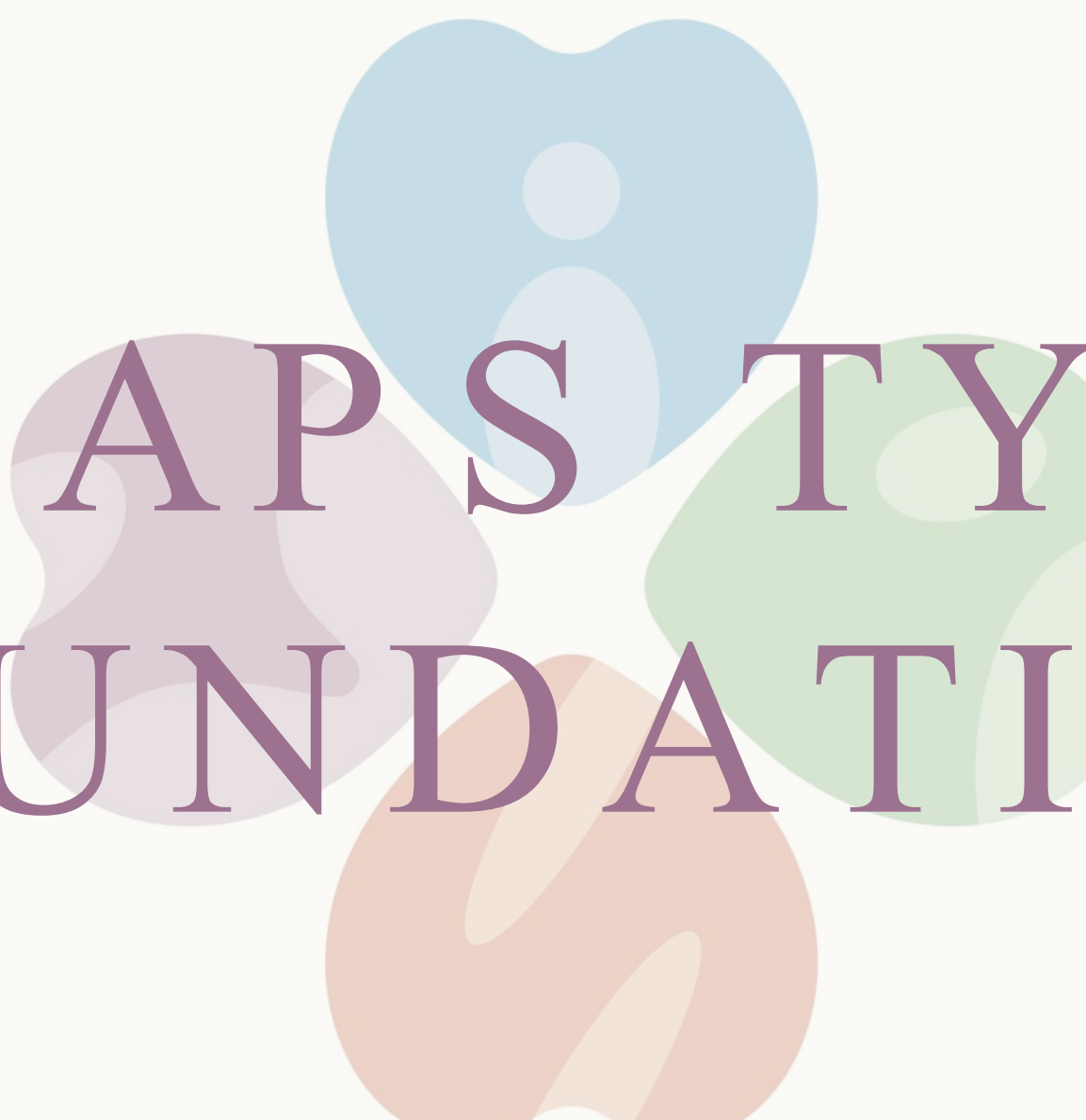




THE APS TYPE 1 FOUNDATION

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

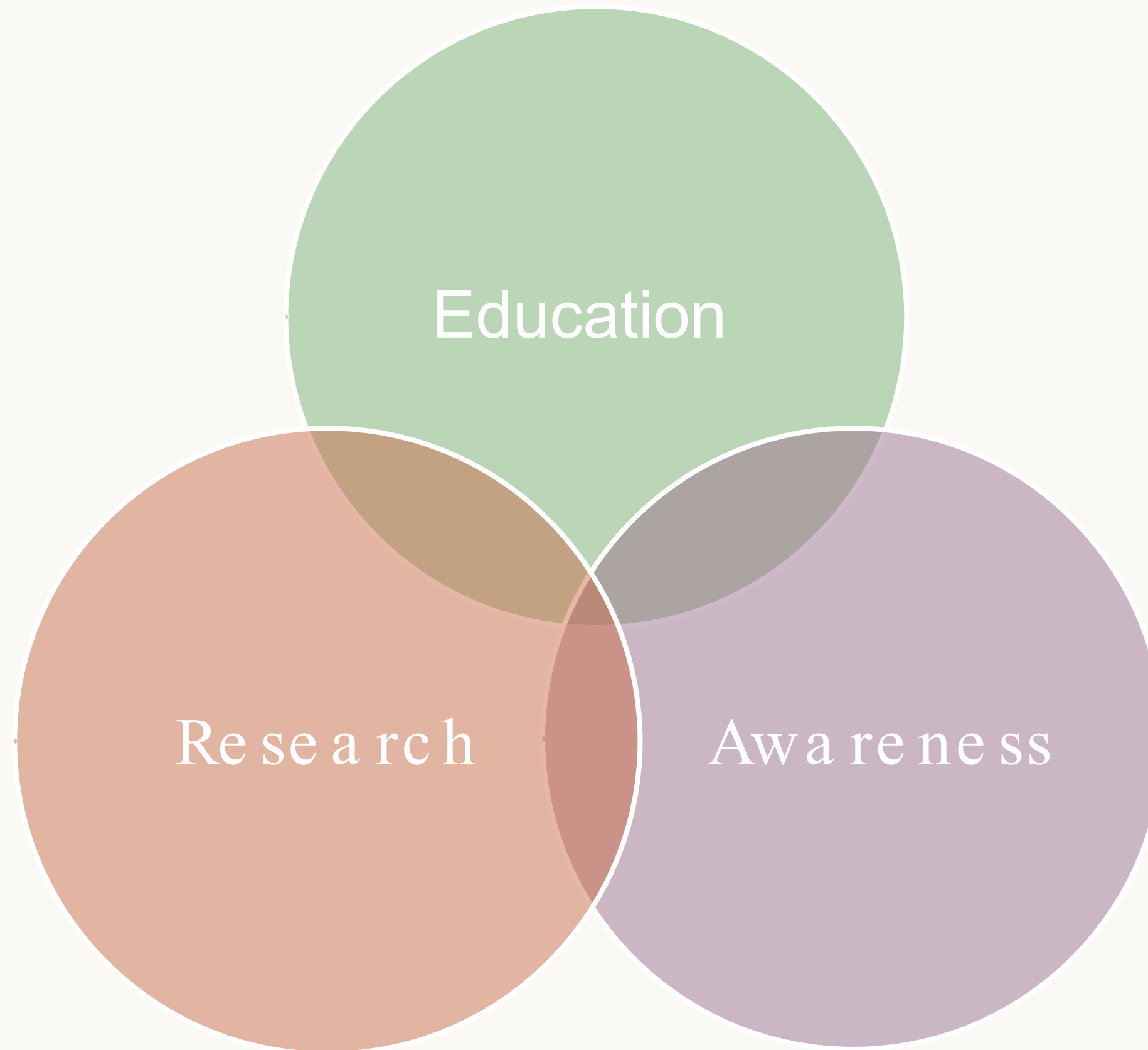
Empower the APS Type 1 Community

HUDDLE Rules of the Road

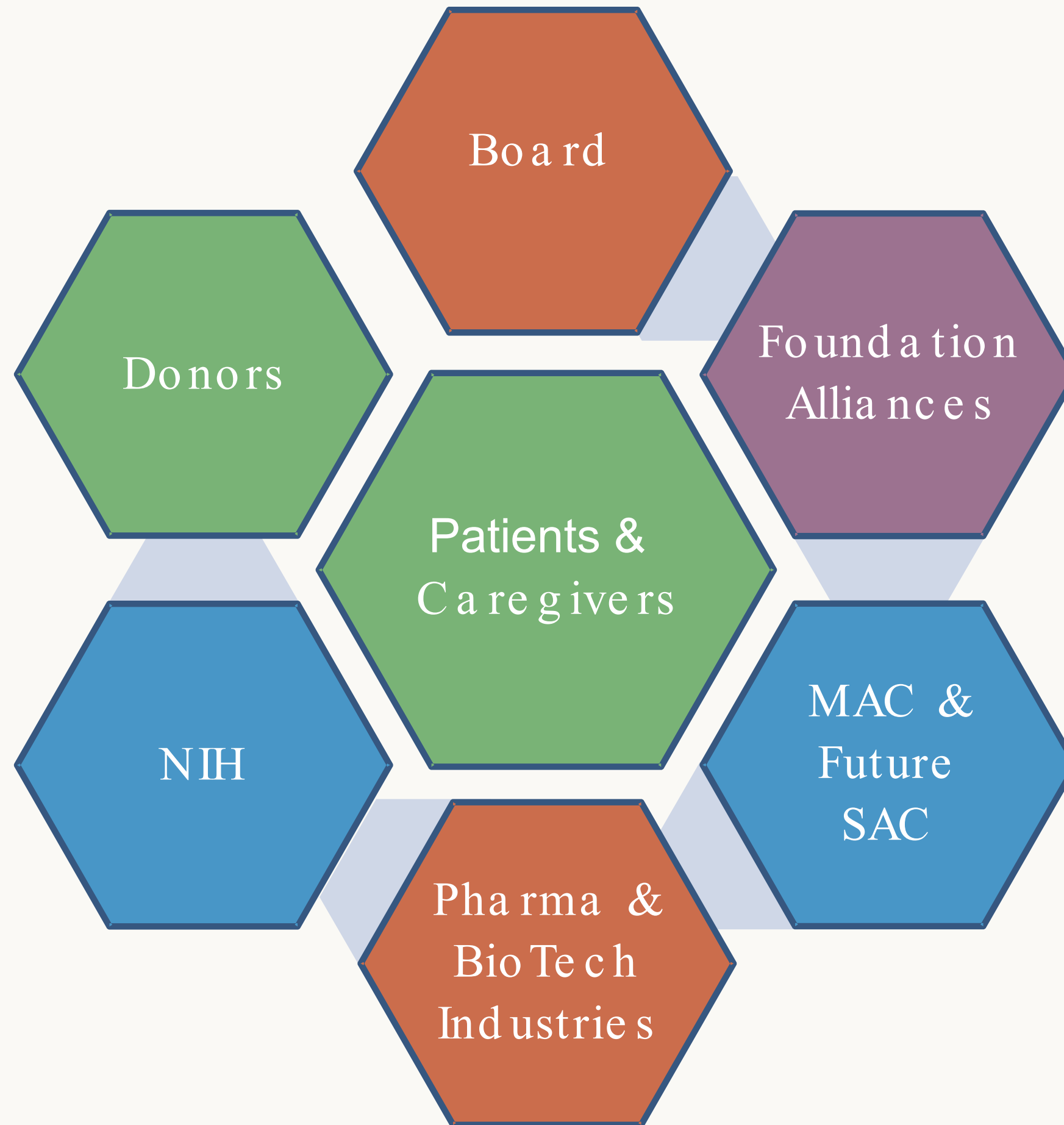
- **Lead with Kindness:** This is a supportive space. Please treat everyone with compassion and respect.
- **Respect First:** Everyone's experience is valid. Respect diverse perspectives and experiences.
- **Honor Confidentiality:** Personal stories shared here stay within this community.
- **Share at 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



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 \$700 K for
Research



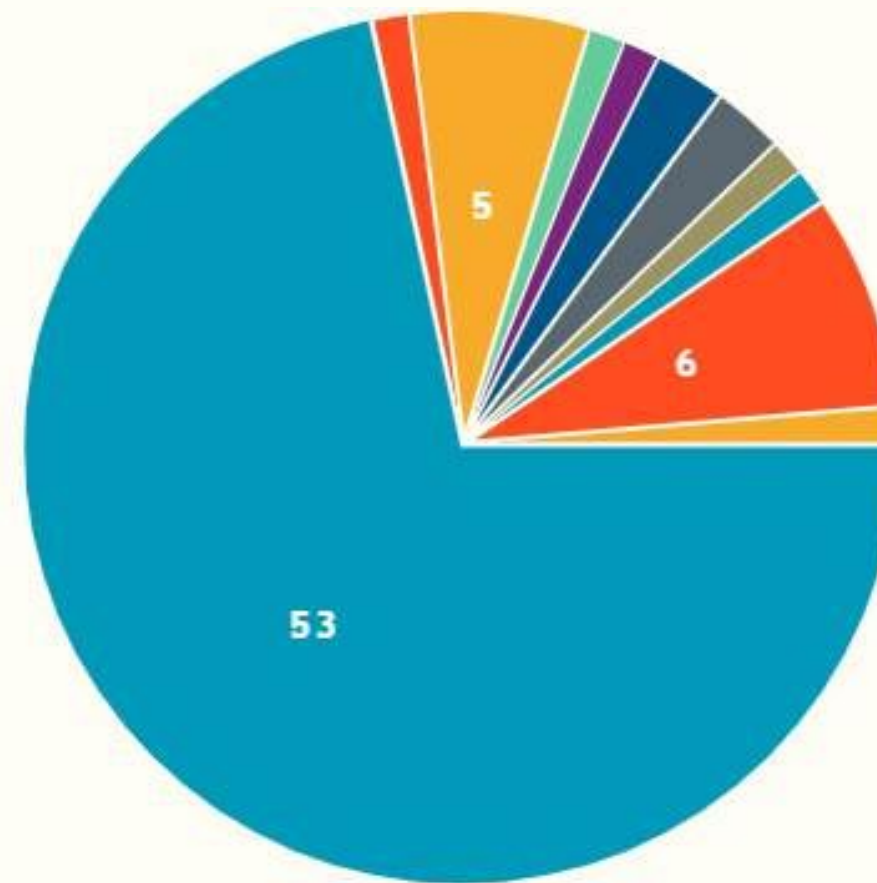
Developed a Web-based
Registry



THE APS TYPE 1 (APECED) REGISTRY

NEW AND IMPROVED PLATFORM LAUNCHING FALL 2026

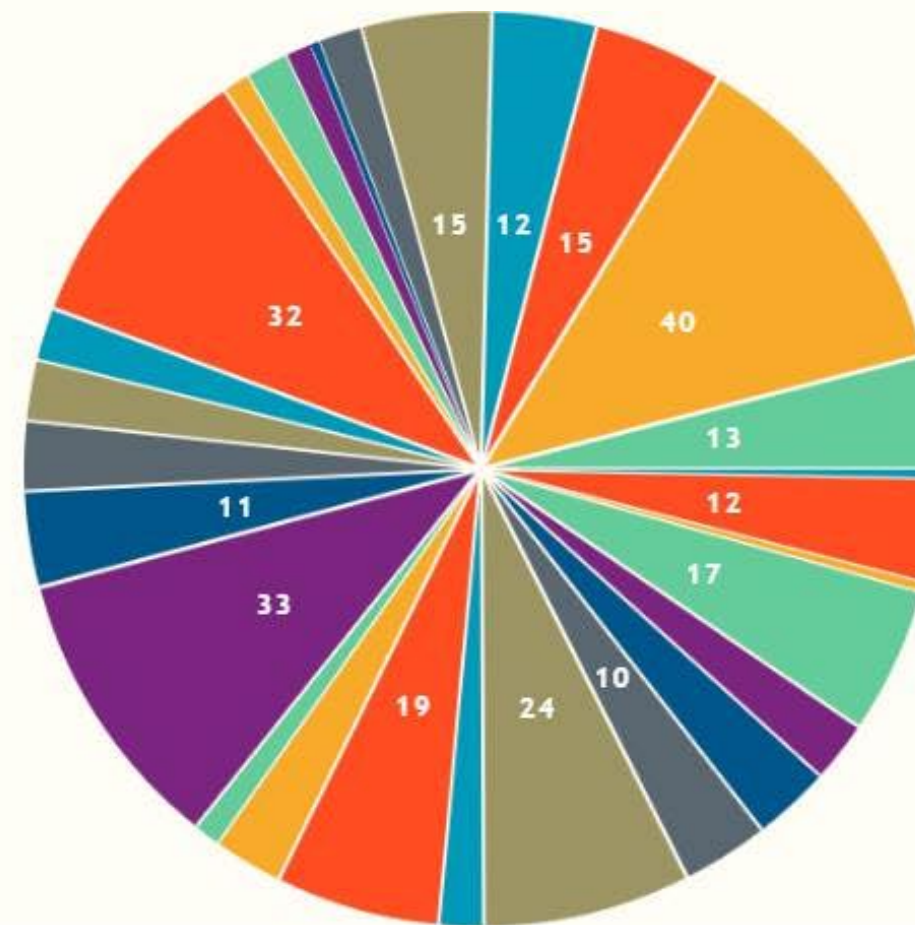
Our registry has approximately 150 patients registered from many countries.



United States Brazil Canada Croatia France Greece Ireland Isle of Man Italy United Kingdom of Great Britain and Northern Ireland (the) Vanuatu

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)



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



APS-1 Arts Festival

APSTYPE1.ORG

Celebrating the life of James Read

Southside Lincoln

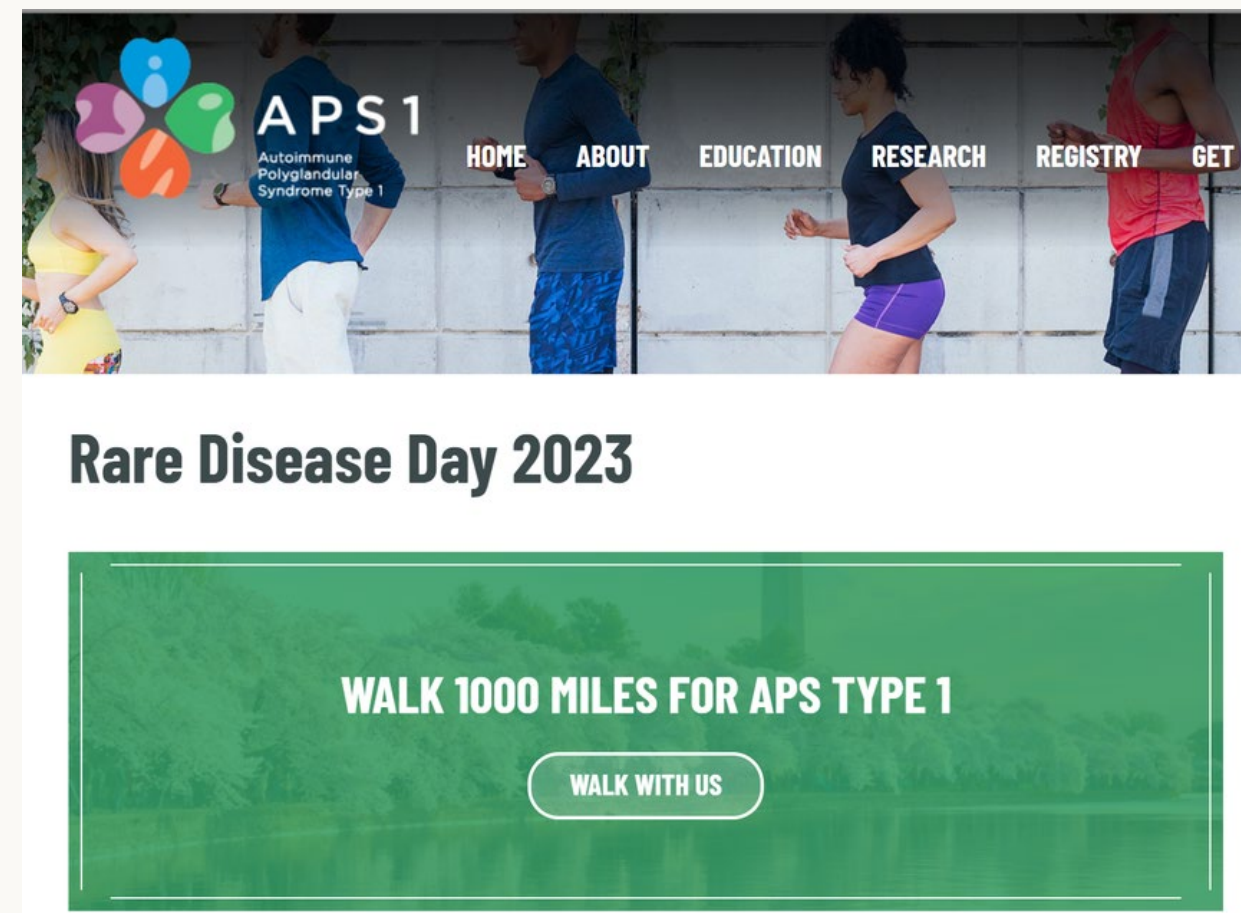
Sunday 9th July 2023, 1pm
St Katherine's Hall LN5 8DW

Fundraising for APS-1 Foundation

- ★ Live music
- ★ Stalls selling art, crafts, books, fashion, baked goods and more
- ★ Film screening about James
- ★ Raffle prizes
- ★ Art auction
- ★ Bar and catering

Reserve free tickets on eventbrite, donations on the door.

The poster features a large orange acoustic guitar on the right side and a photograph of a person performing on stage in a hall.



APS1
Autoimmune Polyglandular Syndrome Type 1

HOME ABOUT EDUCATION RESEARCH REGISTRY GET I

Rare Disease Day 2023

WALK 1000 MILES FOR APS TYPE 1

WALK WITH US

The top section shows a website header with a navigation menu and a group of people walking. The bottom section is a green banner for a walk event.

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

PAST FAMILY FUNDRAISERS



#liveforlovet

SEYFERT FAMILY BASKET RAFFLES



Fact Cards	
Come up with your own creative way of displaying your fact cards at your next Serving Awareness dinner!	
	
APS Type 1 affects about 1 in 2 million people. Now that is rare!	
Serving Awareness	Serving Awareness
The average time between the appearance of symptoms and a correct diagnosis of APS Type 1 is over 10 years!	APS Type 1 is the only known autoimmune disease caused by a single gene pair .
	
Serving Awareness	Awareness
We held the first International Symposium on APS Type 1 in 2015, with world leading researchers and patients all sharing their expertise.	The National Institutes for Health is engaged in the first U.S. natural history study of APS Type 1 with over 50 patients .
	
Serving Awareness	Serving Awareness
The early symptoms of APS Type 1 vary, but can include candida (yeast) infections in the mouth or on the skin, low calcium levels (hypoparathyroidism), fatigue and salt craving (Addison's Disease), fevers and a hive-like rash.	Our APS Type 1 community is full of foodies. We love to eat!
	
Serving Awareness	Serving Awareness



Forever in Blue Jeans for Genes

A concert to benefit The APS Type 1 Foundation & to celebrate Brent Finch's 50th Birthday

Featuring



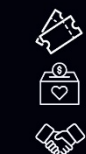
In support of



FEBRUARY 17, 2024 7PM

DRESS CODE: FOREVER IN BLUE JEANS FOR GENES

The Canyon Club
28912 Roadside Drive
Agoura Hills, CA 91301



BUY TICKETS

DONATE

SPONSOR

Scan here



FOUNDATION ALLIANCES



A BOLD SOLUTION TO THE AUTOIMMUNE CRISIS

Over **50 million Americans** live with an autoimmune disease. Cases are rising 3-12% each year. No cures currently exist, but promising research offers near-term solutions.

Join us for a fireside chat, moderated by **Dr. Maureen A. Su**, Department Vice Chair, Microbiology Immunology & Molecular Genetics, JCLA, with **Dr. Victoria Shanmugam**, Director of the Office of Autoimmune Disease Research in the Office of Research on Women's Health at the National Institutes of Health, who will provide an update on the NIH-Wide Strategic Plan for Autoimmune Disease Research.

This is a widely attended event.



2044 RAYBURN BUILDING
WEDNESDAY JULY 22, 2026

BRIEFING: 4-5 PM
RECEPTION: 5-6 PM

RSVP BY
JULY 16TH



DR. SHANMUGAM



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



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?

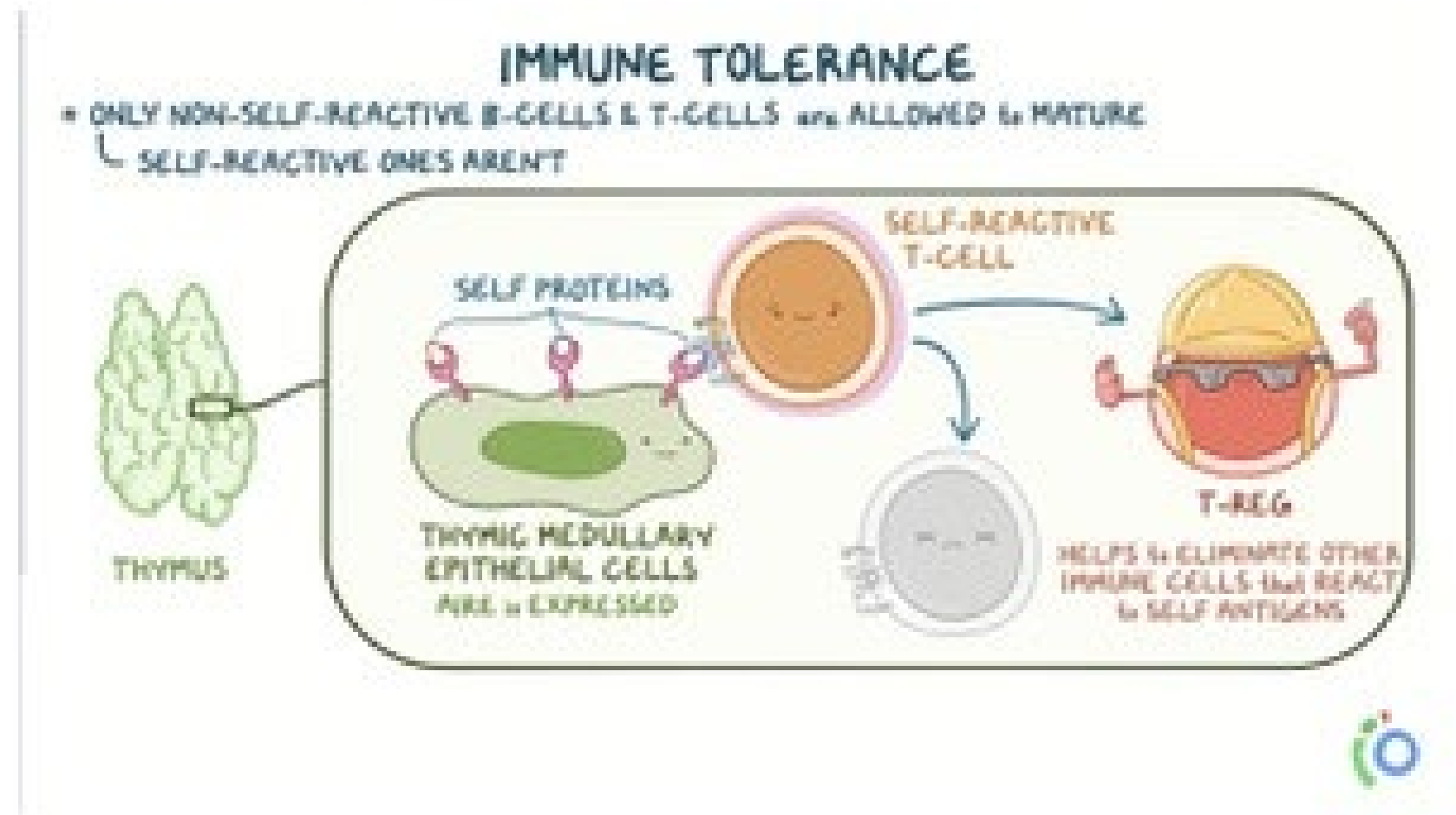
CONNECT YOU TO
OTHER PATIENTS,
FAMILIES & PHYSICIANS

SHARE EDUCATIONAL
MATERIALS, RESEARCH AND
NEW FINDINGS

BE RECEPTIVE TO ALL OF
YOUR IDEAS

What is APS Type 1?

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





THANK YOU!

APS 1