## CONTENTS

<table>
<thead>
<tr>
<th>Topic</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>SAVE THE DATE FOR OUR 3RD INTERNATIONAL SYMPOSIUM JUNE 20-23, 2019</td>
<td>2</td>
</tr>
<tr>
<td>INTRODUCING THE APS TYPE 1 (APECED) REGISTRY</td>
<td>3</td>
</tr>
<tr>
<td>PRESTIGIOUS ROCKEFELLER UNIVERSITY AWARDS APS TYPE 1 RESEARCH GRANT</td>
<td>4</td>
</tr>
<tr>
<td>EVER WONDER HOW YOU COULD HELP?</td>
<td>5</td>
</tr>
<tr>
<td>INTRODUCING OUR MEDICAL ADVISORY COMMITTEE</td>
<td>6</td>
</tr>
<tr>
<td>PABLO RAMIREZ ATTENDS NORD’S ANNUAL RARE DISEASES AND ORPHAN PRODUCTS SUMMIT</td>
<td>7</td>
</tr>
<tr>
<td>ENSURING LASTING SMILES ACT</td>
<td>7</td>
</tr>
<tr>
<td>2108 FINANCIAL REPORT</td>
<td>8</td>
</tr>
<tr>
<td>THANK YOU FROM FOUNDATION PRESIDENT, TODD TALARICO</td>
<td>9</td>
</tr>
<tr>
<td>BOARD CONTACTS</td>
<td>10</td>
</tr>
</tbody>
</table>
SAVE THE DATE FOR OUR
3RD INTERNATIONAL SYMPOSIUM
JUNE 20-23, 2019

In collaboration with the Immune Deficiency Foundation’s IDF National Conference, we will host our 3rd International Symposium on APS Type 1. We hope you can join us June 20-23, 2019, at the Gaylord National Resort and Convention Center in National Harbor, Maryland, located outside Washington, D.C. Here’s a link to the amazing hotel, which your family is sure to enjoy: https://www.nationalharbor.com/gaylord-national/.

From physicians to nurses to specialized life management experts, patients and families will be presented with an unsurpassed opportunity to network on the cutting edge of research and treatment and learn more about issues that affect all of the primary immune deficiency (PI) community.

The Conference will have a day dedicated to APS Type 1, which will kick off with an introduction to APS Type 1 presentation by Dr. Gregory Constantine from the NIH and a talk by Dr. Mihalis Lionakis titled “APS-1/APECED in 2019: new knowledge on managing the individual patient.” Additional presenters include Drs. Theo Heller and Karen Winer and Ms. Elise Ferre of the NIH, as well as Drs. Catherine Gordon and Yee-Ming Chan of Boston Children’s Hospital and Dr. Richard Auchus from the University of Michigan.

The Foundation is pleased to offer travel scholarships to help as many as possible attend. Registration and more information will be available in early February.
INTRODUCING THE APS TYPE 1 (APECED) REGISTRY

A NEW DYNAMIC TOOL TO FACILITATE THE DEVELOPMENT OF NEW DIAGNOSTIC AND TREATMENT OPTIONS

The APS Type 1 (APECED) Registry is a powerful opportunity for individuals with APS Type 1 and their family members to contribute directly to research that will enhance our understanding of APS Type 1, thus facilitating the development of new diagnostic and treatment options.

Participation is especially vital given the rarity of APS Type 1. Every patient experience is a unique and valuable part of the natural history of APS Type 1. We encourage you to participate to help improve management and discover a cure for APS Type 1.

We are in the final stages of preparing the registry. Please watch for another announcement in February.
PRESTIGIOUS ROCKEFELLER UNIVERSITY AWARDS
APS TYPE 1 RESEARCH GRANT

The Rockefeller University Center for Clinical Translational Science has awarded Dr. Dana Orange and her collaborators, Drs. Rhonda Kost and Roger Vaughn, a grant for her new study entitled “Leveraging Phenotype and Genotype Frequency Data to Accelerate Diagnosis of Autoimmune Polyglandular Syndrome Type 1.” (Supported by NCATS/NIH grant #UL1TR001866.)

This new study will help us estimate the incidence of APS Type 1 in North America by computing: (i) the frequency of patients meeting standard or expanded diagnostic criteria for the APS Type 1 phenotype data from the New York City Clinical Data Research Network (NYC-CDRN) database of up to 6 million patients, and (ii) the frequency of pathogenic AIRE variants using publicly available genomic data. The study hopes to identify the optimal clinical criteria for earlier diagnosis.
EVER WONDER HOW YOU COULD HELP?

We welcome your help! There are some easy ways to help financially. And, if you’re interested in sharing your time helping the Foundation grow its capacity, please contact any member of the board of directors. Find their email addresses on the last page.

• Our Website – You can make a simple tax-deductible donation on our website. Remember the organization is 100% volunteer, so contributions directly support Foundation activities! This link below will take you right to the donation section of our website. [http://apstype1.org/donate/](http://apstype1.org/donate/)

• Amazon Smile – It’s simple: just log on to [https://smile.amazon.com/](https://smile.amazon.com/) and put our Foundation’s name in your account. The Foundation will receive a small portion of the proceeds from everything YOU spend! So far, Amazon has donated nearly $100 million to registered charities. With your help, we can raise our profile among these charities.

• Serving Awareness Fundraiser – If you are planning a dinner event, make it a Serving Awareness Fundraiser! All the materials you need are located at [http://apstype1.org/serving-awareness](http://apstype1.org/serving-awareness) with an easy point, click and print. Your friends and family will be happy to support the Foundation after the incredible dinner you provide. If you don’t like that, make it a lunch date! We hope these meals can fund our symposium travel grants. Help your fellow community members get to the symposium.

• Company Matching – Many employers match their employees’ charitable donations. The Foundation is a non-profit 501(c)(3) organization. Last year we received a good amount of company matching. Let’s keep that going. If you or other donors within your APS Type 1 circle are looking for a great way to increase financial support, ask your employers about company matching.
INTRODUCING OUR MEDICAL ADVISORY COMMITTEE

We are pleased to introduce Dr. Richard Auchus as the newest member of our Medical Advisory Committee (MAC).

Dr. Auchus joins Dr. Michael Lionakis (NIH), Dr. Mark Anderson (UC San Francisco), and Dr. Dana Orange (Rockefeller University) on the MAC. The MAC acts as an advisory committee to the Foundation’s board of directors and helps with the development of initiatives and strategies to promote awareness, education and research into APS Type 1.

Dr. Auchus is Professor of Pharmacology and Internal Medicine in the Division of Metabolism, Endocrinology, and Diabetes at the University of Michigan and Director of the Diabetes, Endocrinology, & Metabolism Fellowship Program at Michigan.

Dr. Auchus and his group are active in research projects ranging from basic chemical principles of steroid biosynthetic enzymes and steroid mass spectrometry to clinical and translational investigation in disorders of the pituitary, adrenal, ovaries, and testes that cause hypertension, infertility, and obesity. His clinical interests also focus on pituitary, adrenal, and reproductive diseases that involve disorders of steroid production, and he is particularly interested in the care of adults with genetic disorders of steroid biosynthesis and action. He has authored over 230 journal articles and 30 book chapters, and he has presented at a diverse range of national and international conferences.

In addition to enjoying the MAC’s support, the Foundation hosts semi-annual conference calls with a broader group of U.S. and international researchers and clinicians to brainstorm and collaborate with each other.
PABLO RAMIREZ ATTENDS NORD’S ANNUAL RARE DISEASES AND ORPHAN PRODUCTS SUMMIT

On behalf of the Foundation, community member, Pablo Ramirez, attended the NORD Summit in Washington D.C. this past October. During the two day meeting, he was able to meet with NORD State Ambassadors who gave tips on how to use the NORD volunteer network across the country. He also connected with representatives of other rare disease organizations, including the Immune Deficiency Foundation, with whom we are partnering for our 2019 APS Type 1 Symposium.

Finally, Pablo met many of the pharmaceutical company representatives who attended. All of these relationships will help us build networks and work towards better management of APS Type 1.

Thank you, Pablo!

ENSURING LASTING SMILES ACT

In 2018, with the support of the National Foundation for Ectodermal Dysplasias (NFED), a bill was put before Congress, called the Ensuring Lasting Smiles Act, to require all group and individual health insurance plans to cover medically necessary dental services as a result of congenital abnormalities. Please register your support for this legislation by accessing this link:

https://www.nfed.org/get-involved/advocate/ways-to-advocate/-/

NFED’s website (http://www.nfed.org) is also tremendous resources for those of us affected by ectodermal dysplasias. You may also consider participating in NFED’s Ectodermal Dysplasias International Registry (https://nfed.patientcrossroads.org/) to help connect researchers with individuals affected by ectodermal dysplasias.
2018 was a light year for donation income as there was only one fundraiser held in 2018; a serving awareness event held in Toronto by the Band Orange family. We had a significant rise in company match donations this year - way to go members! We continue to receive checks from Amazon Smile as well. Our overall general donation income in 2018 reached $58,000 (2017 was over $100,000).

This was also a light year for expenses, as we did not host a symposium or fund research in 2018. Our overall expenses for 2018 were only $9,000.

Looking ahead to 2019, our expense budget will be much higher, and we hope that our donation income will be too. In 2019, we will incur expenses related to:

- Our 3rd International Symposium, including travel assistance for our members in need.
- The soon to be launched APS Type 1 (APECED) Registry, including quarterly maintenance fees.
- A brand-new Foundation website to better meet the needs of our community, including patients, families, caregivers, researchers, clinicians and scientists.
- General expenses, including accounting and bookkeeping fees, insurance premiums and non-profit membership fees.

In addition, we hope to take the great leap of hiring a part-time employee in 2019 to help move our key initiatives forward more quickly, including the Registry, fundraising, community outreach and website maintenance. Finally, we plan to fund our 7th research grant through NORD in 2019.

With assets of $220,000 at the end of 2018, we are in great shape for such a young foundation, but we have a long way to go! Keep up the good work and fundraise, fundraise, fundraise. The Talarico family and the Finch family are planning their 2019 fundraisers now. More details to come. With your support, we can continue to advance our mission of raising awareness and promoting research.

All the Best in Health and Happiness,
Sherri Seyfert
Treasurer
THANK YOU FROM FOUNDATION
PRESIDENT TODD TALARICO

As I look at the past year, our fourth year as a Foundation, I’m taken aback by the large steps we have taken as a Foundation, and at the same time I am reminded of just how far we need to go.

First, I want to recognize all of you for your amazing level of networking and communicating with one another. You always make new families feel welcome and jump in to answer the tough questions. You’re quick with a note on a good packing trick for the NIH, addressing a challenging clinical manifestation, and cheering each other along through the good and the bad days of APS Type 1. Let’s keep that sense of family going. It’s critical to who we are today and who we will be in the future.

Our Foundation wouldn’t exist without our generous donors. Thank you to everyone who has donated to the Foundation and to the APS Type 1 cause. In 2017, in partnership with the National Organization for Rare Disorders (NORD), the Foundation awarded its 6th research grant to Dr. Maureen Su from the University of North Carolina at Chapel Hill for her study entitled, “Thymus Transplantation for APS1.” Dr. Su recently moved her lab (and our grant) to the University of California, Los Angeles (UCLA). We hope to award our 7th grant in 2019.

I also want to thank this amazing board. I know you may not see it, but Jennifer Orange, Robin Finch, Heather Talarico, Sherri Seyfert, and Dave Seyfert devote countless hours to moving this Foundation forward. They do this voluntarily while raising families, working their careers, and dealing personally with their own APS Type 1 challenges. Each month when we have our board meeting, I can feel the sense of pride they take in the work they do and the dedication they have to do it RIGHT for each and every one of us. They are a great team, and I’m humbled by their dedication in support of all of us challenged with this disorder.

Finally, I want to thank our Medical Advisory Committee: Dr. Anderson, Dr. Lionakis, Dr. Orange, and our newest addition to the team, Dr. Auchus! Without their guidance, direction, and clinical support, we wouldn’t have reached many of the milestones we have in just a few short years. Thank you!

I hope that you have enjoyed our first newsletter. We’re working to make the updates more frequent and comprehensive as we share not only the accomplishments of 2018, but also the excitement of what’s planned for 2019. I look forward to seeing all of you in Maryland in June at our 3rd International Symposium on APS Type 1, which we will host in partnership with the Immune Deficiency Foundation (IDF). I’m confident this conference will be a wonderful experience that will provide new memories and friendships to last a lifetime.

Cheers! Todd
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