
The Section on

ENDOCRINOLOGY

Newsletter

Fall 2020



Chairperson's Column

Jane L. Lynch, MD, FAAP

Dear Colleagues,

I am writing this column in the midst of an election where we are all inspired to vote and speak up for our beliefs during these difficult times. Elected positions are an honor to hold and I have appreciated the amazing support from our executive committee and the AAP while serving as Chair of Section on Endocrinology (SOEn) over the past four years. As my tenure comes to an end, I would like to take this chance to re-introduce the current AAP SOEn executive committee members. They work tirelessly to improve our subspecialty with a wide range of collaborative projects, policy reviews, publications and many education and advocacy efforts.

Our incoming elected Chair is the wonderful **Kupper Wintergerst, MD**, who is currently the Division Chief and Director of the Wendy Novak Diabetes Center at the University of Louisville and Norton Children's Hospital in Kentucky. He has been instrumental in co-leading many of our subspecialty workforce and advocacy initiatives since elected to the SOEn executive committee in 2012. He is passionate that our pediatric endocrine patients have access to affordable insulin and necessary medication coverage to receive the care that they deserve. He chaired the initial Education in Quality Improvement for Pediatric Practice (EQIPP) online course Growth Surveillance and Linear Growth Failure. He continues to work on the development of several clinical policies and care guidelines. The April 2020 *AAP News Focus on Subspecialties* article, *Type 1 Diabetes May Masquerade as Viral Illness*, (<https://www.aappublications.org/news/2020/04/01/type1diabetesviralillinois040120>) co-authored by his fellow Dr. Ryan Dyess and our past SOEn Chair, Dr. Irene Sills, was incredibly timely as COVID-19 hit our nation.

Kathy Bethin MD, PhD and **Lucy Mastrandrea MD, PhD** are both academic leaders at the University of Buffalo where Dr. Bethin is the Fellowship Program Director and Dr. Mastrandrea is the Division Chief. They have been passionate about the need to support high quality training for our next generation of pediatric endocrinologists both academically and in the research arena. They are our key collaborators with the Pediatric Endocrine Society (PES) Education Subcommittee to develop the ongoing AAP/ PES patient education materials. Dr. Bethin and Mastrandrea are often the first to volunteer to review and critique policies and publications that come to our executive committee for input. Dr Bethin served as a subject matter expert on the revision of the EQIPP course: Growth – Addressing Concerns and Management that launched in 2018 and is included with membership (<https://shop.aap.org/eqipp-growth-addressing-concerns-and-management/>) that is a member benefit and offers CME credit as well as MOC part 2 and part 4 credit. Dr. Mastrandrea has worked with Dr. Brodsky and Dr. Nadia Merchant on annual SOEn proposals for the National Conference and Exhibition education program endocrine topics and speakers and she also heads up the AAP/PES Leona Cutler Award QI application process.

Jill Brodsky, MD, MBA, founded the division of Pediatric Endocrinology at CareMount Medical and is a role model for the private practice model for our subspecialty. She continues to teach and lecture with a stimulation center, insulin pump and sensor program, nutrition services, and research facilities. She has just completed her MBA in Healthcare Management at the Johns Hopkins Carey School of Business. As a full-time private practice clinician, she offers a unique

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Effective November 1, 2020

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Chairperson's Column *Continued from Page 1*

viewpoint on the growing challenges of practicing pediatric endocrinology in the current and emerging healthcare climates and provides guidance and AAP contributions to insurance, billing and coding issues that impact us all. She has worked with PES on a coding webinar and heads up the *Paul Kaplowitz, MD, Endowed Lectureship* award for cost-effective care in Pediatric Endocrinology.

Brittany Bruggeman, MD, is a senior Pediatric Endocrine fellow at the University of Florida who was appointed as the fellowship trainee to our executive committee following several years on the AAP Section on Pediatric Trainees Executive Committee. Dr. Bruggeman has led advocacy initiatives to improve access to medical care for at-risk populations, interned with the AAP Washington Office, and has championed advocacy to reduce the cost of insulin for patients with diabetes. She even published an opinion piece regarding the high cost of insulin in the Gainesville Sun. She is mentoring the growing Pediatric Endocrine trainee's subcommittee along with the Section on Pediatric Trainees Subspecialty Fellowship Trainee Delegate, Dr. Mary McCauley. Our membership has expanded to include over 150 trainees with complementary PREP as a membership benefit for trainees as we welcome and support this next generation of providers to our subspecialty.

Our newest elected members to the SOEn Executive Committee are David Jelley, MD, from Tulsa, Oklahoma and Bess Marshall, MD, from St. Louis. **Dr. Jelley** is the Hille Chair of Diabetes and Senior Deputy Director of the Harold Hamm Diabetes Center at the Oklahoma University (OU) School of

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Community Medicine. He has a special interest in the prevention of diabetic ketoacidosis (DKA) and early diagnosis of diabetes. He recently co-authored a paper on patient/caregiver experience at diagnosis and has conducted a successful regional diabetes awareness campaign. Additionally, he also recently co-authored our 2019 SOEn AAP News Focus on Subspecialties article, "First international consensus statement guides clinicians on diagnosis, management of Silver-Russell syndrome" (<https://www.aappublications.org/news/2019/07/12/focus071219>) with his eldest daughter who is now a Pediatric Endocrine fellow! **Dr. Marshall** is a Professor at the Washington University School of Medicine where she is the Medical Director of the Wolfram Syndrome Research Clinic and Director of the Department of Pediatrics Office of Faculty Development. Dr. Marshall's motivation to join our SOEn executive committee is to advance workforce issues, set pediatric lab normal range standards, and advocate for patients with diabetes, chronic illness, rare diseases, and differences of sex and gender.

My huge thanks go out to **Laura Laskosz, MPH**, as the amazing and experienced AAP staff for SOEn as well as three other pediatric subspecialties. Her knowledge and skills keep us organized and focused on the multitude of opportunities that we juggle. I would also like to acknowledge Dr. Manmohan Kamboj who serves as our Newsletter editor and Dr. Nadia Merchant for her assistance with National Conference program proposals, newsletter articles, policy and payer reviews. As I step down to the role of Past Chair, I realize that this work never ends. Dr. Wintergerst has the leadership skills to address the advocacy, policy, and educational opportunities that come with the privilege of Chairing the SOEn committee. As you read this fall newsletter, please reach out to our SOEn committee with ideas and areas that you would like to be more involved in.

Beyond Genetic Diagnosis, Therapeutic Advancements for Skeletal Disorders

Nadia Merchant, MD, FAAP

Assistant Professor of Pediatrics

Division of Endocrinology and Diabetes

Children's National Hospital

As personalized therapies for rare diseases are becoming a reality, it is extremely important that pediatric endocrinologists are confirming certain genetic disorders and determining if patients may benefit beyond conventional management and treatment. Nosology and Classification of Genetic Skeletal Disorders has expanded over the last few decades, the most recent version in 2019 comprised of 461 different diseases with pathogenic variants affecting 437 different genes. There are over twenty treatments currently being studied, some have been recently approved by the Food and Drug Administration (FDA) for rare genetic skeletal disorders. Even though many of these skeletal disorders are not primarily managed by endocrinologists, this may change as we have more therapeutic treatments. By discussing three specific genetic skeletal disorders, the goal is to emphasize the importance of endocrinologists being up to date and comfortable with new treatments that are rapidly emerging.

Achondroplasia is the most common form of disproportionate skeletal dysplasia. Incidence is about 1 in 25,000 live births with 80% of cases being de novo. Achondroplasia is associated with complications that include foramen magnum stenosis, craniocervical instability, sleep apnea, scoliosis, spinal stenosis, recurrent ear infections, and obesity. Approximately 97% of the cases are caused by a G380R substitution of the fibroblast growth factor receptor 3 (FGFR3) gene, resulting in a gain of function mutation. FGFR3 negatively regulates endochondral ossification, and thus gain of function FGFR3 mutations limit endochondral growth and lead to disproportionate growth. Hypochondroplasia is milder than achondroplasia, and around 70% of hypochondroplasia cases are caused by FGFR3 mutations.

There are no FDA approved treatments for achondroplasia, however this may change soon. BioMarin developed Vosoritide, a C-type natriuretic peptide (CNP) analog that is given as a daily subcutaneous injection. CNP is a known negative regulator of FGFR3. It binds to natriuretic-peptide receptor 2 (NPR2) inducing cyclic guanosine-3',5' monophosphate (cGMP) synthesis, thus inhibiting the MAPK pathway. Vosoritide increases extracellular matrix production which works with chondrocytes to increase endochondral ossification. Last year, BioMarin completed its phase 3 study of Vosoritide. This

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was a randomized, double-blind, placebo-controlled study of 121 children with achondroplasia aged 5 to 14 for 52 weeks. It demonstrated a favorable safety profile with increase in annual growth velocity. In August 2020, BioMarin submitted a New Drug Application (NDA) to the U.S. Food and Drug Administration (FDA) for children with achondroplasia. There is currently also an investigator-initiated clinical trial to determine if Vosoritide improves growth velocity for specific genetic disorders with short stature (hypochondroplasia, CNP deficiency, heterozygous NPR2 variant, rasopathies, SHOX variants). I must disclose that I am a co-instigator for this trial. Another treatment in the pipeline is by Ascendis Pharma that has developed TransCon CNP, a slow release CNP analog that is currently in a phase 2 clinical trial. CNP is bound to and shielded by a TransCon Carrier that allows for sustainable release of CNP over seven days.

Since 2018, there has been a FDA approved treatment for X-linked hypophosphatemia (XLH), also known as vitamin D-resistant rickets, which is the most common heritable form of rickets. It is due to inactivating mutations in the *PHEX* gene leading to FGF23 (fibroblast growth factor 23) over activity. The FGF23 hormone, secreted from osteocytes, inhibits the kidneys' ability to reabsorb phosphate and degrades 1, 25-dihydroxyvitamin D. As a result, patients have low serum phosphate, high urinary phosphate and low or inappropriately normal serum 1,25-dihydroxyvitamin D levels. The abnormal phosphate handling may lead to bowed legs, bone pain, short stature and delayed walking. The FDA approved Burosumab, an anti-FGF23 fully human monoclonal antibody, which targets the underlying pathophysiology of XLH by increasing the levels of the sodium phosphate co-transporter in the proximal kidney tubules and increases 1 alpha hydroxylase. Burosumab has been shown to improve the clinical course of this disease with less side effects than conventional treatment with daily oral phosphate and activated vitamin D.

In 2015, the FDA approved a therapeutic option for Hypophosphatasia (HPP). HPP is caused by loss of function mutations in the *ALPL* gene which encodes the tissue non-specific alkaline phosphatase enzyme (TNSALP). HPP presents with defective bone mineralization and is inherited in both autosomal dominant and recessive patterns. Decreased TNSALP activity results in elevated serum inorganic pyrophosphate (PPi), which is a substrate of TNSALP and inhibits tissue mineralization, thus leading to impaired bone or tooth mineralization. Asfotase Alfa (AA), a recombinant glycoprotein that contains the catalytic active site of TNSALP, enables proper degradation of PPi. Asfotase Alfa has shown improvement in certain forms of HPP, specifically infantile and juvenile-onset HPP. For HPP, it is critical to understand the phenotypes to determine if patient may be a candidate for AA.

These therapies have only emerged over the last decade as more genes were discovered for rare diseases. Since there is limited longitudinal data on new therapies for rare diseases, as subspecialists, pediatric endocrinologist must stay up to date and provide families and patients with information to make the best decision for deciding to pursue novel therapies.

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The T1D Exchange QI Collaborative: A National Network Improving Outcomes for Patients with Type 1 Diabetes

Osagie Ebekeozien¹, Nicole Riales¹, Jake Wexelblatt¹, and Manmohan Kamboj²

Affiliations

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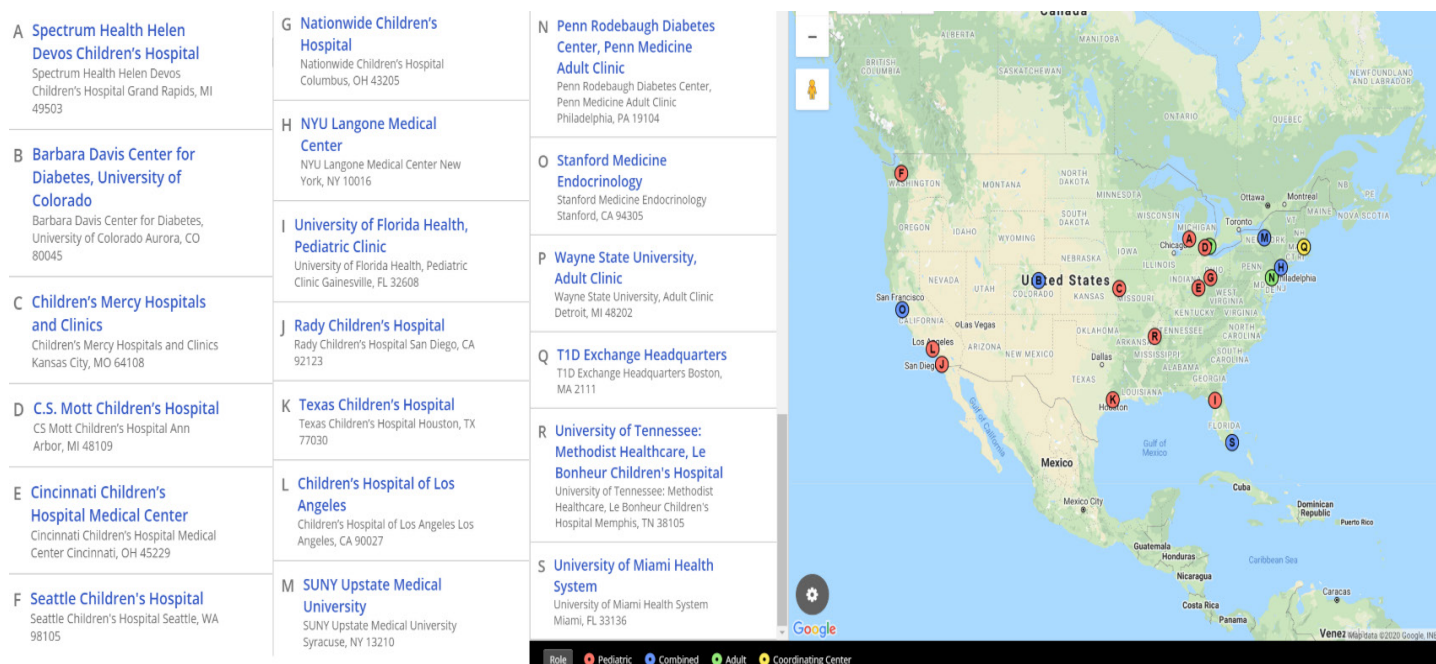
Introduction

In 2016, The T1D Exchange established a learning collaborative which is funded by the Helmsley Charitable Trust. In this Collaborative, the participating clinics share data and best practices to improve care delivery for people with type 1 diabetes (T1D). This effort has led to a foundation for learning from variation in T1D care delivery via QI methodologies and has demonstrated success in improving care processes through iterative testing cycles and transparent sharing of data.¹

Today, the T1D Exchange (T1DX) incorporates more than 20 institutions and hospital systems (Fig 1), collectively known as the T1D Exchange Quality Improvement Collaborative (T1DX-QI). The aim of this network is to improve clinical processes, healthcare outcomes, and care accessibility for all patients with T1D.

Figure 1: List of Centers and Geographical Distribution

Printed with permission from the T1D Exchange. The map was created using BatchGeo software.



Since 2018, T1DX-QI has published and presented on its successful QI methodologies to improve aspects of T1D patient care performed at hospitals within the Collaborative. These interventions frequently occur simultaneously across hospitals, following the same aims, interventions, and measures at each clinic site. Variation and disparities in hospital resources require differences in Plan Do Study Act (PDSA) processes (an iterative framework for developing, testing and implementing changes leading to improvement). Each hospital is unified in the measures and data analyses through a quality infrastructure that the Collaborative provides as well as standard performance and process improvement methods.

Quality Improvement Results: Early in the process of establishing common aims and designing interventions to improve care, T1DX-QI created a change package to support clinics' QI capacity in practice. A change package recommends

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evidenced-based change ideas that support care process improvements. Clinics use the change package recommendations to implement new changes in practice, acting as a pragmatic guide for best practices, change ideas, tools, and strategies that can, agnostically, be adapted and adopted in new settings. The structure helps clinics begin new tests of change faster and the best practices help to ensure success in a variety of settings. The structure helps clinics begin new tests of change faster and the best practices help to ensure success in a variety of settings.²

The clinics set a common goal of improving the use of technology such as insulin pumps and continuous glucose monitors (CGMs). Ten clinics participated in a study published earlier this year in which the different hospitals tested different PDSA cycles, ranging from advocating for Medicaid coverage, to patient education classes, to removing barriers to adoption in an attempt to expand CGM usage in the targeted patient cohort, aged 12 to 26 years.³ This intervention was successful, with a 12% increase in CGM usage across T1DX-QI in the span of 20 months. All participating clinics saw a significant increase in CGM usage of at least 7%, with one clinic even seeing a dramatic 34% improvement over the same 20-month period. Five clinics included in T1DX-QI implemented similar QI interventions to increase insulin pump use in patients ages 12 to 26 years. The transfer of monthly reporting data led to frequent adjustments, resulting in an overall 10% improvement over the span of 20 months, with all clinics seeing at least a 6% increase.⁴

T1DX-QI has additional studies, beyond the increase in use of technology, such as the study of 32,000 patients to determine the impacts of access to care, follow-up touchpoints, and frequency of visits, knowing that care standard of quarterly appointments is a challenge for both patients, families, and care teams. Further analyses into the data of this study and similar studies helped T1DX-QI understand results variation by age, location, and race regarding treatment for T1D. Clinics' attempts to increase visit frequency included limiting the duration of refills, ensuring teenage patients who generally have higher hemoglobin A1c's (HbA1c) can be seen as often as younger patients, and improving clinic and staffing capacity to accommodate four annual visits per patient.⁵

More recent projects at T1DX have focused on reducing the percentage of T1D patients who qualify as high-risk or having fewer patients who have had two HbA1c measurements of at least 9% in the past year. Six of the ten participating clinics reached the target of a 5% reduction, with the remaining four centers still seeking to meet that goal.⁶

Some of the earlier impactful interventions performed by T1DX-QI involved operationalizing depression screening in diabetes care, through the Patient Health Questionnaire-2 and 9- multipurpose instruments for screening, diagnosing, monitoring, and measuring the severity of depression, at seven member clinics. Over 8,000 screenings were conducted on T1D patients at these clinics over the span of two years, increasing screening from 10% to 71% over that period.⁷ Additionally, after two years of focused interventions to support depression screening, T1DX-QI created a Depression Screening Change Package, which can be accessed and downloaded online.⁸

QI Portal: T1DX has created a Type 1 Diabetes QI data Portal (the "*QI Portal*") to collect and report on standardized clinical diabetes data to support diabetes population health management and care. T1DX provides an unlimited number of licenses to the QI Portal, to clinic team members so that they may view practice data, create data visualizations, reports, and compare their data against peer clinics for peer benchmarking and knowledge sharing.

Population Health Data Insights: T1DX-QI's most recent work has been focused on determining the effects of COVID-19 on glycemic management and other symptoms in T1D patients working with 64 centers in the United States.⁹ In studying T1D patients who develop the symptoms of COVID-19 as well as those who were exposed to individuals carrying the virus, T1DX-QI is learning which factors of an individual's identity causes more severe symptoms when living with T1D and experiencing COVID-19. By May 2020, T1DX-QI determined that in addition to living with T1D, other comorbidities such as obesity, occurring in 39.4% of patients, led to more severe symptoms, and many of these patients (45.5%) experienced diabetic ketoacidosis (DKA) while experiencing COVID-19 symptoms. This study involved a heterogenous patient population, comparing length of time with diabetes diagnosis, race, education, and insurance, giving T1DX-QI more information to prepare for future interventions throughout the remainder of the COVID-19 pandemic.

Patient Engagement and Committees: A central tenant of T1DX-QI is co-production with patients and families. All participating clinics are supported to include patients or parents of children with T1D on their QI teams so that the patient voice and perspective is represented. These patient representatives are very active members of the teams and

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participate in reviewing of policies, workflows, PDSAs, surveys, and educational materials. In addition to the Patient/Parent Advisory Committee, T1DX-QI facilitates several committees that support quality leadership and integration; health data analytics; population health; and quality review and accountability. Some of the special committees include Clinical Leadership; Publications; Data Science; and Data Governance committees.

The interventions by T1DX-QI have assisted a broad range of members of the T1D community including people of all ages and from different backgrounds living in the United States. T1DX-QI plans to expand its membership and offerings significantly over the next 12 months. There are opportunities for participation by more clinics.

For more information about joining T1DX-QI, email qi@t1dexchange.org.

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Introducing the NEW AAP Volunteer Network

We are excited to announce a new centralized and easy to access list of national, section, council and chapter [volunteer opportunities](#) for AAP members. There are positions for everyone, whether you are in training, in practice, or retired. You can contribute to the AAP mission even if you only have a limited amount of time to give. This new site:

- Is easily accessible online - just click on “My Collaboration Sites” from aap.org
- Is accessible 24/7 - which means you can access the site in the middle of the night when you’re on call or can’t sleep
- Has micro-engagement opportunities with as little as one hour a month commitment
- Has multi-year leadership opportunities – for members with more time looking to boost their leadership skills and augment their CVs
- Can be sorted by subspecialty, skills needed, membership type and more - customize your view
- Has new opportunities uploaded weekly – be sure to check back often

Now is the time to enhance your career by pursuing a variety of scholarly activities with the AAP. Everything you need to know and [all in one place](#) - volunteering with the AAP just became much, much easier!

Advocacy Resources

- **(NEW!) Interim Guidance on Supporting the Emotional and Behavioral Health Needs of Children, Adolescents and Families During the COVID-19 Pandemic** This new clinical guidance was developed to support the emotional and behavioral health of children, adolescents and families during COVID-19. The interim guidance can be found [here](#).
- **COVID-19 Response–Advocacy Report**– The report summarizes the AAP’s advocacy activities at all levels of government. The October 2020 report can be accessed [here](#) and is posted to the [advocacy subsite](#) of the AAP’s main COVID-19 page.
 - Updates on COVID-19 AAP website- <https://services.aap.org/en/pages/2019-novel-coronavirus-covid-19-infections/>
 - Discussion Board – AAP members can connect with colleagues on the COVID-19 Discussion Board to share resources and discuss critical topics – <https://collaborate.aap.org/COVID-19/Pages/default.aspx>
 - E-mail: members can contact AAP at COVID-19@aap.org
- **Academic and Subspecialty Advocacy Washington Report March 2020** (log-in required) – a biannual publication providing the latest federal policy updates for pediatric medical subspecialists and pediatric surgical specialists.
- **Sign up for Advocacy Emails** – the Academy sends regular advocacy-focused communications to members including timely advocacy action alerts, policy updates and breaking child health news from Washington. To sign up for Advocacy Alerts, email kids1st@aap.org with your name, AAP ID (if known) and preferred e-mail address.
- **Advocacy Training Modules** (log-in required) – information to assist you with becoming an effective advocate
- **FederalAdvocacy.aap.org** – Federal Advocacy Online Resource Center (log-in required) find resources and tools for federal advocacy
- **Connect with Your AAP Chapter** – reach out to your [chapter executive director](#) and let them know about your interest and issues.
- **State Advocacy Resources** – Visit the [State Advocacy page](#) to access briefs on key state issues, advocacy action guides, infographics, advocacy planning and education guides, and the latest news from the states.
- **Become a #Tweetiatician** – stay up to date on child health news, follow and engage with AAP on social media via @AmerAcadPeds, @AAPPres, @AAPNews and @healthychildren.
- **Subscribe to AAP’s official #tweetiatician list** on Twitter at: <https://twitter.com/AmerAcadPeds>

Not a Member? Joining is Easy!

Current members of the Academy in good standing are eligible to join the Section on Endocrinology by contacting the AAP Customer Service at 866/THE-AAP1 (866-843-2271).

New Members

AAP Fellows

Jasmine Gujral, MD, FAAP
Bimota Nambam, MD, FAAP
Martha Revollo, MD, FAAP
Erin Richardson, MD, FAAP

Post Residency Training Members

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Shuai Hao, MD
Noreen Islam, MD, FAAP
Charlene Lai, MD, FAAP
Jennifer May, MD, FAAP
Ghada Naji, MD
Julie Park, MBBS
Lauren Roach, MD, FAAP

Resident Members

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Riordan Ledgerwood, MD

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Olivia Devillers
Cortney Elkin

Marlin Hana
Austin Huang
Meenu Krishnasamy
Nancy Le
Regina Lee
Taylor Lindquist
Ricardo Martinez
Mayra Matar
Alphonette Pinder
Sarah Rachal
Garbiela Ramirez Cuebas
Helly Thakkar
Krizia Trasmonte
Kaiser Valshon
Stephanie Vogel
Jasmine Ware

National Affiliate Member Jamila Smith-Young, CPNP-AC

Call for highlights, notices and special events

This newsletter is intended to highlight important articles, position papers, new guidelines and events for pediatric endocrinologists. If you would like to contribute, please forward your inquiries to Dr Manmohan (Manu) Kamboj.

Pediatric Endocrine Workforce Article

Jane Lynch, MD, FAAP

Hot off the press! Leaders in our field have just published a Pediatric Endocrine Society (PES) workforce taskforce summary¹ As no surprise to any of us, there is a pipeline shortage for Pediatric Endocrinologists as well as an urgent need to both expand our numbers and to recruit more diversity within our subspecialty providers. This article presents a wonderful summary of the contributing factors leading to this situation as well as practical proactive ideas to address the scarcity of providers in our field. These concerns align with similar conclusions made by the AAP that the number of pediatric endocrinologists is greater in academic centers and lower in private practice and other, longer working hours, and longer wait times, therefore, indicating the number of pediatric subspecialists in endocrinology is insufficient (Pletcher BA, et al. *JPeds*. 2010;156:1011–1015, e1). Lastly, similar challenges are impacting our Internal Medicine Endocrine colleagues regarding workforce as summarized in the *Journal of Clinical Endocrinology and Metabolism* (JCEM) article published in June 2020.² These are two must read articles and they will provide sorely needed ammunition for advocacy to improve our subspecialty support!

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Endocrinology Meeting Schedule

2021 Meetings

ENDO 2021 – The Endocrine Society Annual Meeting *Virtual*

March 20-23, 2021

Pediatric Endocrine Society Virtual Meeting

April 30 – May 3, 2020

American Diabetes Association 81st Scientific Sessions

June 25-29, 2021

Washington, DC

AAP National Conference & Exhibition

October 8-12, 2021

Philadelphia, PA

2022 Meetings

American Diabetes Association 82nd Scientific Sessions

June 3-7, 2022

New Orleans, LA

AAP National Conference & Exhibition

October 7-11, 2022

Anaheim, CA