

# CONGENITAL ADRENAL HYPERPLASIA

## **Mimi Kim, MD, MSc**

Associate Professor of Clinical Pediatrics  
Keck School of Medicine of USC  
Co-Director, CHLA CAH Comprehensive Care Center  
Los Angeles, California

Friday, July 14, 2023 – 9:00 am – 10:30 am

### ***CAH Overview***

The objectives of this talk are as follows: 1. Discuss the cause of CAH 2. Discuss the impacts on hormone productions 3. Discuss treatments and treatment goals 4. Discuss the different types of CAH

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## **Kyriakie Sarafoglou, MD**

Professor  
Department of Pediatrics – Divisions of Endocrinology and Genetics and Metabolism  
Department of Experimental & Clinical Pharmacology | University of Minnesota  
Director, Center of Congenital Adrenal Hyperplasia and  
Noah's Ark and Weiss Family CAH Research Fund and Endowment  
University of Minnesota Masonic Children's Hospital  
Minneapolis, Minnesota

Friday, July 14, 2023 – 10:45 am – 12:00 pm

### ***Monitoring and Disease Control in Children with CAH***

Description of Segment: Topics covered will include challenges in monitoring disease control in children with CAH; cortisol pharmacokinetic and pharmacodynamic adrenal response studies and the modeling of cortisol, 17-hydroxyprogesterone, androstenedione and testosterone to help guide toward more targeted individualized treatment; challenges in the treatment of young children with CAH; understanding the importance of both circadian and ultradian rhythms of cortisol secretion.

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## **Mitchell E. Geffner, MD**

Professor Emeritus of Pediatrics  
Keck School of Medicine of USC  
Children's Hospital Los Angeles  
Ron Burkle Chair in the Center for Endocrinology, Diabetes and Metabolism  
Los Angeles, California

Friday, July 14, 2023 – 1:30 pm – 2:45 pm

### ***The Future of CAH: An Overview of Clinical Trials and Upcoming Treatments***

The objectives of this talk are to summarize the clinical trial process and review current clinical trials and new treatments as they relate to CAH.

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**Lynn Schema, MS, LGC**

Supervisor of Pediatric Genetic Counseling  
Division of Genetics and Metabolism  
M Health Fairview  
Minneapolis, Minnesota

Friday, July 14, 2023 – 3:00 pm – 4:15 pm

***Genetics and CAH***

The objectives of this talk are as follows: 1. Provide an overview of basic genetic principles 2. Explain autosomal recessive inheritance 3. Review the importance of genetic testing for patients with CAH 4. Address myths and misconceptions about genetic testing 5. Highlight the role of genetic counseling for patients with CAH

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Saturday, July 15, 2023 – 9:00 am – 10:30 am

***Adrenal Insufficiency – Illness Protocols and Emergency Preparedness***

This session will give a deep overview of adrenal insufficiency – causes, symptoms, treatments and how to navigate illness and emergencies.

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**Erin Beamon**

PAN/Tumor Division Consultant for The MAGIC Foundation  
Mother of a child with Panhypopituitarism  
&

**Mimi Kim, MD, MSc**

Associate Professor of Clinical Pediatrics  
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Los Angeles, California

Saturday, July 15, 2023 – 10:45 am – 12:00 pm

***Solu-Cortef Injection Training***

A brief overview of how to be prepared before, during and after an adrenal crisis including step-by-step hands on training on how to administer the Solu-Cortef injection.

## GROWTH HORMONE DEFICIENCY

### **Aristides Maniatis, MD, FAAP**

Board Certified Pediatric Endocrinologist  
Rocky Mountain Pediatric Endocrinology  
Associate Clinical Professor  
University of Colorado Health Sciences Center  
Centennial, Colorado

Friday, July 14, 2023 – 9:00 am – 10:30 am

#### ***The ABC's of GHD***

The objectives of this talk are as follows: 1. Understand potential issues with growth including both the non-hormone and hormone causes of poor growth. 2. Understand the evaluation for Growth Hormone Deficiency. 3. Discuss the process and challenges of growth hormone therapy.

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### **Bradley Miller, MD, PhD**

Division Director, Pediatric Endocrinology  
University of Minnesota Medical School  
Medical Director, Discovery Clinic  
M Health Fairview Masonic Children's Hospital  
Minneapolis, Minnesota

Friday, July 14, 2023 – 10:45 am – 12:00 pm

#### ***Quality of Life with Treatment***

During this segment we will go over the quality of life for children who are treated or denied treatment for Growth Hormone Deficiency. Consequences of non-treatment will be discussed regarding short and long-term outcomes. Finally, discussion will lead to the end of growth hormone therapy for a child and carry through treatment into adulthood.

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### **Aristides Maniatis, MD, FAAP**

Board Certified Pediatric Endocrinologist  
Rocky Mountain Pediatric Endocrinology  
Associate Clinical Professor  
University of Colorado Health Sciences Center  
Centennial, Colorado

Friday, July 14, 2023 – 1:30 pm – 2:45 pm

#### ***Long Acting Growth Hormone***

The objectives of this talk are as follows: 1. Discuss challenges of current daily growth hormone therapy 2. Discuss principles of Long Acting Growth Hormone (LAGH) development 3. Discuss the current landscape of LAGH therapies either approved or in development

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Keck School of Medicine of USC  
Children's Hospital Los Angeles  
Ron Burkle Chair in the Center for Endocrinology, Diabetes and Metabolism  
Los Angeles, California

Friday, July 14, 2023 – 3:00 pm – 4:15 pm

***Puberty Management***

The diagnosis and treatment of delayed puberty, as well as its short and long-term ramifications, will be discussed.

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**Kyle McNerney, MD**

Assistant Professor, Department of Pediatrics  
Division of Endocrinology & Diabetes  
Washington University in St. Louis School of Medicine  
St. Louis, Missouri

Saturday, July 15, 2023 – 9:00 am – 10:30 am

***Evaluating and Treating Different Causes of Short Stature***

In this session we will discuss a typical work-up when considering short stature. We will review examples of patients with short stature with several different diagnoses. We will specifically discuss growth hormone deficiency and growth hormone treatment, and also discuss other modalities and treatment options available for treating short stature.

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**Melissa Davis**

Licensed Clinical Mental Health Counselor  
Owner, Anchor the Soul Counseling, PLLC  
Hubert, North Carolina

Saturday, July 15, 2023 – 10:45 am – 12:00 pm

***When Being Vertically Challenged Takes a Mental Health Toll***

Melissa will discuss what mental health challenges come from short stature. We will discuss these concerns as it relates to elementary, middle and high school as well as transitioning to college. As parents, we will discuss how short stature can impact relationships, friendships and your child's own self-esteem and self-image.

## **MCCUNE ALBRIGHT SYNDROME**

### **Michael T. Collins, MD**

Senior Investigator  
Skeletal Disorders and Mineral Homeostasis Section  
National Institute of Dental and Craniofacial Research  
National Institutes of Health  
Bethesda, Maryland

Friday, July 14, 2023 – 9:00 am – 10:30 am

#### ***What you Need to Know and What to Expect in FD/MAS***

Presenting on the history of McCune-Albright Syndrome and Fibrous Dysplasia and how it is believed to develop. Information on how one becomes diagnosed with MAS/FD, the different testing that needs to be done to confirm diagnosis, as well as testing that should continue to be done throughout development. Will review past and current research that is being done on MAS/FD.

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### **Kirt E. Simmons, D.D.S., Ph.D.**

Director, Craniofacial Orthodontics  
Arkansas Children's Hospital  
University of Arkansas Medical Sciences  
Little Rock, Arkansas

Friday, July 14, 2023 – 10:45 am – 12:00 pm

#### ***Dental Components of Fibrous Dysplasia***

Management and care of fibrous dysplasia within the jaw must be considered when treating patients. This segment will discuss various issues that arise and how treatments can be successfully carried out.

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### **Alison Boyce, MD**

Chief, Metabolic Bone Disorders Unit  
National Institutes of Health  
Bethesda, Maryland

Friday, July 14, 2023 – 1:30 pm – 2:45 pm

#### ***Endocrine Features and Anti-Resorptive Medications in FD/MAS***

This presentation will cover diagnosis and management of endocrine features of FD/MAS. It will also discuss medical management of skeletal disease, including use of bisphosphonates and denosumab.

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**Jason Malone, DO, FAAOS**

Orthopedic Surgeon  
Nemours Children's Hospital  
Orlando, Florida

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**Robert P. Stanton, MD, FACS**

Professor, Orthopedic Surgery  
University of Central Florida  
Orlando, Florida  
Attending Orthopedic Surgeon  
Nemours Children's Hospital  
Orlando, Florida

Friday, July 14, 2023 – 3:00 pm – 4:15 pm

***Orthopedic Treatment of Fibrous Dysplasia: Case Presentations***

Orthopedic management in MAS/FD, the most up to date ways to treat/correct FD, as well as past methods that have been found to not be as successful. Learn about rods/plates and when and when not to surgically intervene.

**Dr. Stanton will be showing some cases of lower limb reconstruction and one case of scoliosis management.**

**Case Presentations by Dr. Stanton:**

Surgical Reconstruction of Limb Deformities from Fibrous Dysplasia  
Scoliosis Management in Fibrous Dysplasia

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**Brittany Anderson**

Fibrous Dysplasia (FD) Patient

Saturday, July 15, 2023 – 9:00 am – 10:30 am

***Fibrous Dysplasia: A Patient Perspective and Advocacy Movements***

This segment will feature a personal journey of resilience and political advocacy from someone using their strength within to change the world for MAS/FD affected people.

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**Family Chat**

Saturday, July 15, 2023 – 10:45 am – 12:00 pm

This is an opportunity for families to network, share stories, discuss treatments and successes that have worked for their family.

# **OPTIC NERVE HYPOPLASIA**

## **Mark Borchert, MD**

Board Certified Pediatric Neuro Ophthalmologist  
Director, Eye Birth Defects Program and Eye Technology Program  
Associate Professor of Clinical Ophthalmology and Neurology  
Keck School of Medicine of USC  
The Vision Center at Children's Hospital  
Los Angeles, California

Friday, July 14, 2023 – 9:00 am – 10:30 am

### ***ONH Overview***

The objectives of this talk are as follows: 1. What is ONH 2. How does ONH happen 3. You have a diagnosis, now what?

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Friday, July 14, 2023 – 10:45 am – 12:00 pm

### ***ONH Current Research***

The objectives of this talk are as follows: 1. What does past research show? 2. What does current research show? 3. What does this mean for our children?

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## **Ellen Lancon Connor, MD**

Professor Pediatric Endocrinology and Diabetes  
Co-Director, Adolescent PCOS Clinic  
American Family Children's Hospital  
University of Wisconsin  
Madison, Wisconsin

Friday, July 14, 2023 – 1:30 pm – 2:45 pm

### ***ONH and Endocrinology***

The objectives of this talk are as follows: 1. How does the pituitary fit in with ONH? 2. How do you manage endocrine issues associated with ONH?

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## **Mark Borchert, MD & Ellen Lancon Connor, MD**

Friday, July 14, 2023 – 3:00 pm – 4:15 pm

### ***ONH Q&A***

The objectives of this talk are as follows: 1. Time to ask all your questions you have associated with ONH and endocrine issues.

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## **PANHYPOPITUITARISM**

### **Mitchell E. Geffner, MD**

Professor Emeritus of Pediatrics  
Keck School of Medicine of USC  
Children's Hospital Los Angeles  
Ron Burkle Chair in the Center for Endocrinology, Diabetes and Metabolism  
Los Angeles, California

Friday, July 14, 2023 – 9:00 am – 10:30 am

#### ***PAN 101***

The session is a broad overview of Hypopituitarism, its causes, presentations, clinical manifestations and treatments.

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### **Mimi Kim, MD, MSc**

Associate Professor of Clinical Pediatrics  
Keck School of Medicine of USC  
Co-Director, CHLA CAH Comprehensive Care Center  
Los Angeles, California

Friday, July 14, 2023 – 10:45 am – 12:00 pm

#### ***What's on the Horizon***

A discussion about the new research, medicine and devices, and other exciting advances in areas of interest to hypopituitarism.

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### **Aristides Maniatis, MD, FAAP**

Board Certified Pediatric Endocrinologist  
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## **RUSSELL SILVER SYNDROME, SMALL FOR GESTATIONAL AGE & TEMPLE SYNDROME**

### **Jennifer B. Salem, MA**

MAGIC RSS/SGA

Research & Education Director

Manhattan Beach, California

Friday, July 14, 2023 – 9:00 am – 10:30 am

#### ***Building Blocks for Success in Understanding RSS/SGA/Temple: The Basics***

This is a MUST SEE foundational presentation for first year attendees and those with young children given by the primary author of the guidebook. You will learn terminology and concepts necessary to understand future presentations. Unless you consider yourself an expert on RSS/SGA/Temple or have a teen, all families should plan to attend this segment PRIOR to attending additional presentations.

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### **Dayna L. Carney, RN, CBC**

Pediatric Nurse & Certified Breastfeeding Counselor

Children's Mercy Hospital

RSS Division Consultant The MAGIC Foundation

Overland Park, Kansas

Friday, July 15, 2022 – 9:00 am – 10:30 am

#### ***Letting Go: Roundtable Discussion for Parents of Teens and Young Adults***

This informal moderated discussion will include several parents of older RSS children available to discuss the challenges during adolescence and young adulthood.

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### **Yvette Q. Getch, PhD, CRC**

Professor and Coordinator, Graduate Counseling Programs

University of South Alabama

Mobile, Alabama

Friday, July 14, 2023 – 9:00 am – 10:30 am

#### ***Finding Balance: The Keys to Self-Care in Adulthood***

Balancing work, relationships and adult life can be challenging for all young adults. When you add in complications arising from a rare disorder, chronic health conditions and insurance challenges, one's mental health can take a toll. This presentation will focus on balancing life, coping strategies, self-care and engaging in practices to improve and maintain mental health.

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### **Madeleine D. Harbison, MD**

Assistant Professor of Pediatrics in Endocrinology

Adrenal and Steroid Disorders Group

Icahn School of Medicine at Mount Sinai

New York, New York

Friday, July 14, 2023 – 10:45 am – 12:00 pm

#### ***Unique Endocrine Issues for the RSS/SGA/Temple Child: Bone Age, Adrenarche & Insulin Resistance***

As always, there are critical topics for all parents to understand fully and a discussion on bone age, adrenarche, and insulin resistance is one of them. This complex and essential presentation explains adrenarche, insulin resistance and the importance of monitoring bone age for advancement to prevent premature cessation of growth.

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**Bethany L. Cook, PsyD, HSP, MT-BC**

Adjunct Professor at NLU-ISPP  
Licensed Clinical Psychologist  
Health Service Psychologist  
Music Therapist Board Certified  
Chicago, Illinois

Friday, July 14, 2023 – 10:45 am – 12:00 pm

***Parenting Road Maps for Navigating ADHD, Anxiety and Bullying***

Anxiety, ADHD and bullying are complex and often intertwined issues that kids face these days. Learn how to spot early signs your child may be struggling, which doctors to seek guidance from, and techniques and skills you can apply in your family.

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**Suparna Jain, MD**

Assistant Clinical Professor of Pediatrics  
Division of Pediatric Endocrinology  
UCLA-Cedars Sinai Medical Center  
Private Practice Pediatric Consultants  
Santa Monica, California

Friday, July 14, 2023 – 10:45 am – 12:00 pm

***Medical Care How to Guide for RSS Adults***

This segment will focus on the changes that come when a young adult/adult independent with their medical care. We will discuss health lifestyle choices, annual testing, doctor appointment recommendations, setting boundaries, weight management, musculoskeletal changes, how to find support and more.

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**Dayna L. Carney, RN, CBC**

Pediatric Nurse & Certified Breastfeeding Counselor  
Children's Mercy Hospital  
RSS Division Consultant The MAGIC Foundation  
Overland Park, Kansas

Friday, July 14, 2023 – 1:30 pm – 2:45 pm

***Understanding the Role of Growth Hormone in RSS/SGA***

While society may sometimes view growth hormone treatment as something they hear about in a news scandal, growth hormone is a hormone necessary for many functions, in addition to linear growth. Learn about what growth hormone is and what it does in the body as well as how growth hormone is related to health for our RSS/SGA/Temple children.

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**Bobbi McGrath, MS, CHC**

Certified Regenerative Health & Detox Practitioner  
Certified Health Coach & Behavioral Change Specialist  
Be For You Health Coaching  
Wheaton, Illinois

Friday, July 14, 2023 – 1:30 pm – 2:45 pm

***Taming the Stress Monster; Practical Resources You'll Actually Use***

Come fill up your self-care toolbox by exploring simple strategies you can weave into your daily self-care routine that can help you manage the stress monster. You'll learn how to improve your health, mood, and overall wellbeing that doesn't cost an arm and a leg or take up mountains of time. This presentation will touch on nutrition, exercise, hormone balancing, nervous system support must-haves, and DIY detoxification strategies that move the needle on mitigating stress one small step at a time.

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**Professor Justin Davies, MD, FRCPCH, MRCP**

Consultant Paediatric Endocrinologist Southampton  
Children's Hospital Faculty of Medicine  
University of Southampton  
Southampton, United Kingdom

&

**Karen Temple, MD**

Professor of Medical Genetics, Faculty of Medicine  
University of Southampton  
Honorary Consultant in Clinical Genetics  
University Hospital Southampton  
NHS Foundation Trust  
Southampton, Hampshire, United Kingdom

Friday, July 14, 2023 – 1:30 pm – 2:45 pm

***Genetics and Diagnostics in Temple Syndrome***

This presentation will include an overview of the current genetic findings that lead to a diagnosis of Temple Syndrome and what those may mean in terms of how each genetic finding may present differently.

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**Lindsay Salem-Bango, MPH**

RSS Adult  
Los Angeles, California

Friday, July 14, 2023 – 3:00 pm – 4:15 pm

***Living with RSS/SGA; A Teen/Adult Perspective***

Join Lindsay Salem-Bango, a young adult with RSS, as she moderates a discussion panel of teens and adults with RSS/SGA. Our panelists will answer pre submitted questions and share how they have handled various topics like bullying, complex medical situations, daily medications and surgical interventions. This is a wonderful opportunity to see into the future if you have a small child or simply have renewed faith in humanity as you are amazed at the resilience of this generation.

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**Professor Justin Davies, MD, FRCPCH, MRCP**  
Consultant Paediatric Endocrinologist Southampton  
Children's Hospital Faculty of Medicine  
University of Southampton  
Southampton, United Kingdom

&

**Karen Temple, MD**  
Professor of Medical Genetics, Faculty of Medicine  
University of Southampton  
Honorary Consultant in Clinical Genetics  
University Hospital Southampton  
NHS Foundation Trust  
Southampton, Hampshire, United Kingdom

Friday, July 14, 2023 – 3:00 pm – 4:15 pm

***Treatment and Management in Temple Syndrome***

This is a must attend presentation for families of children with Temple Syndrome. Learn about treatment options and disorder management from the best!

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**Irène Netchine, MD, PhD**  
Professor of Physiology  
Sorbonne Université Trousseau Hospital  
Paris, France

Saturday, July 15, 2023 – 9:00 am – 10:30 am

***Deep Dive into Study Results***

Take a deep dive with us into the recent study results on topics such as adolescence, sleep disordered breathing, long term studies of adults tracked for height and insulin resistance, cyproheptadine and aromatase inhibitors.

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**Madeleine D. Harbison, MD**  
Assistant Professor of Pediatrics in Endocrinology  
Adrenal and Steroid Disorders Group  
Icahn School of Medicine at Mount Sinai  
New York, New York

Saturday, July 15, 2023 – 9:00 am – 10:30 am

***Common Gut Dysmotility Problems and Solutions***

It is not uncommon for children with RSS/SGA/Temple Syndrome to have a complex relationship with eating that begins with gut dysmotility. This presentation will focus on the physical challenges that interfere and the medical interventions that are available.

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**Deborah JG Mackay, PhD**

Professor of Medical Epigenetics  
University of Southampton  
Southampton, United Kingdom

Saturday, July 15, 2023 – 10:45 am – 12:00 pm

***Born SGA: Molecular Testing and Diagnostic Pathway***

For many parents, finding a "name" to label why their child is not growing is like being given a life preserver. They finally feel as though they are no longer floundering in a sea of uncertainty. The RSS Consensus Statement has provided a clear testing path before a clinical diagnosis of RSS should be given. Learn what diagnostic tests can tell us and the importance of pursuing testing to ensure appropriate treatment as well as avoid contraindicated treatments.

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**Yvette Q. Getch, PhD, CRC**

Professor and Coordinator, Graduate Counseling Programs  
University of South Alabama  
Mobile, Alabama

Saturday, July 15, 2023 – 10:45 am – 12:00 pm

***Education Accommodations; What We Need and How We Get It***

For school-age children or those about to begin school, this presentation will cover the common needs and situations our children may face in school and the accommodations that may help them perform at their best and receive an equal education. We will discuss the 504 Plan and IEP and which of these may be right for your child.

**GENERAL SESSIONS – SATURDAY**

**Linda Rio, MA, MFT**

Licensed Marriage & Family Therapist  
Psychological Care Institute, and Private Practice  
Westlake Village, California

Saturday, July 15, 2023 – 1:00 pm – 2:00 pm

***Emotional and Mental Health for Families with Pituitary Issues***

Pituitary disorders certainly have an impact on the body, but the mind, behaviors, emotions, and relationships can also be deeply implicated and are often overlooked. Medical treatments focus on the body but often do not address what happens after the medical treatments. In this session parents and caregivers of children of various developmental ages and stages, from young children to adults, will learn tools, skills, challenges, and triumphs that pituitary disorders present.

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**Aristides Maniatis, MD, FAAP**

Board Certified Pediatric Endocrinologist  
Rocky Mountain Pediatric Endocrinology  
Associate Clinical Professor  
University of Colorado Health Sciences Center  
Centennial, Colorado

Saturday, July 15, 2023 – 2:15 pm – 3:00 pm

***When Should I Be Concerned about Puberty and What Are My Treatment Options***

Dr. Maniatis will discuss what families should be on the lookout for, concerning early puberty, and bring to their doctor's attention if they happen too early or too late. Dr. Maniatis will also discuss treatment options to better help you decide which option is best for you and your family.

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Saturday, July 15, 2023 – 3:15 pm – 4:15 pm

***Learn More about the Pharmaceutical Patient Support Programs***

Patient support services from growth hormone pharmaceutical companies are an integral part of treatment for families. Representatives from multiple companies will each spend 15 minutes educating you about their assistance programs, which can help you through your journey. You will also have time for Q&A.