

GROWTH HORMONE DEFICIENCY

Aristides Maniatis, MD

Rocky Mountain Pediatric Endocrinology
University of Colorado Health Sciences Center
Assistant Clinical Professor
Centennial, Colorado

"The ABC's of GHD"

The objectives of this presentation are to understand potential issues with growth as well as the non-endocrine and endocrine causes of poor growth. We will also cover the evaluation for GH deficiency and explore the process and challenges of GH therapy. Finally, we will discuss potential new GH treatment options.

Larry Kurt Midyett, MD

Medical Director, Midwest Pediatric Specialists
Chief Medical Officer, Pediatrics
Overland Park Regional Medical Center
Overland Park, Kansas

"GHT During the Pubertal Years"

This presentation will provide an overview of GH therapy during mid to late adolescence. Topics will include an overview of GH dosing recommendations during puberty, the effects of puberty on growth patterns, and the importance of closely monitoring the pace of puberty while on GH. We will also discuss and review GH dosing at the end of adolescence and how to determine if GH would be needed after the end of the adolescent growth phase.

Bradley S. Miller, MD, PhD

Professor
Division Director, Pediatric Endocrinology
Medical Director, Discovery Clinic
University of Minnesota Masonic Children's Hospital
Minneapolis, Minnesota

"How do we decide between long-acting and daily GH?"

Dr. Miller will discuss the different options for treatment of Growth Hormone Deficiency. Focusing on the new Long-Acting Growth Hormone products and how they will be a good treatment option for some children but not for others, and why. He will also tell us about the oral secretagogue product that is in trials and which kids could benefit from this product.

Kara Schmid, APRN, CNP

University of Minnesota Masonic Children's Hospital
Minneapolis, Minnesota

"What to Expect When Coming to the Endocrine Clinic"

In this presentation we will discuss what to expect when coming to an endocrine clinic appointment. You will learn the typical steps performed during each office visit with explanation of why we do the things we do in clinic. We will review the importance of accurate height measurements, what a typical exam from the doctor/provider will look like, and typical testing that may be performed.

Peter E Clayton, MD, FRCPCH

Deputy Vice-President & Deputy Dean Faculty of Biology
Medicine & Health, University of Manchester

“Simultaneous Presence of Other Medical Conditions Associated with GHD”

Dr. Clayton will speak about other medical conditions that are associated with children who live with Growth Hormone Deficiency. These will include both relatively common and less common conditions. He will discuss why it is that these seem to affect our children.

Andrew Dauber, MD, MMSc

Chief of Endocrinology
Children’s National Hospital
Washington, DC

“Introduction to Genetic Principles of Short Stature”

In this talk, we will begin with an overview of the different types of genetic variation present in the human genome. We will discuss how these variants can contribute to determining an individual’s stature. We will then review some of the major genetic causes of different growth disorders including growth hormone deficiency, Russell-Silver Syndrome, Noonan Syndrome, and “Idiopathic” Short Stature. We will discuss how new insights into the genetics of short stature are opening the door for new types of treatments.

HYPOPHOSPHATASIA

Howard M. Saal, M.D., FACMG

Professor
Director, Clinical Genetics
Division of Human Genetics
Cincinnati Children’s Hospital
Cincinnati, Ohio

“Genetics in HPP”

The history, diagnosis, management, and treatment options in HPP, including the new therapy Strensiq.

Robert J. Hopkin, M.D.

Associate Professor, Clinical Pediatrics
Director, Medical Genetics Training Program
Division of Human Genetics
Cincinnati Children’s Hospital

Cincinnati, Ohio

“New Developments in HPP”

A presentation of information on recently published HPP research.

Susan Millner, M.Ed., MSW, LCSW

Susan Millner Therapeutic Services

Skokie, Illinois

Vice President of the Chicago Chapter of the Association of Contextual Behavioral Sciences

“Mental Health While Raising a Child with a Rare Disease”

Most parents imagine a parenting journey that is very different from the reality of parenting a child with complex medical and/or developmental needs. As a result, there are many unique emotional challenges that arise. We will talk about the wide range of feelings and experiences and ways to endure and thrive through this unique parenting marathon.

Mark L. Cannon D.D.S., M.S.

Northwestern University

Feinberg School of Medicine

Division of Dentistry

Ann and Robert Lurie Children’s Hospital

Chicago, Illinois

“Dental Aspects of HPP”

Discussion of diagnosis, oral rehabilitation, and medical advancement in dental concerns for those with Hypophosphatasia.

Sarah Mumper

MAS/FD Patient

Disability Advocate

Instagram: @smumps

Website: SarahMumper.com *“Sarah Elizabeth: One in Million”*

One of our own MAS/FD MAGIC family members, Sarah Mumper, will be sharing her story with us! Sarah will be sharing her journey of being diagnosed with MAS/FD, transitioning to adulthood and having to be more in charge of her own care and advocating for herself. She will also be telling us about her experiences at NIH being a part of the clinical challenges. Sarah will also tell us about other challenges she has faced, like getting a new wheelchair!

Scott M. Paul, MD

Physiatrist/ Associate Research Physician

Rehabilitation Medicine Department

National Institutes of Health

Bethesda, Maryland

“Getting the Most Out of Rehab to Optimize Physical Function for Persons with Bone Disorders”

Dr. Paul, from the NIH, our last speaker of the convention will be discussing rehab and physical function for those with bone disorders. He will be teaching us how to be more adaptive to our physical needs and ways to optimize physical function. He will also touch upon rehab and how to get the most out of it.

MCCUNE ALBRIGHT SYNDROME

Michael T. Collins, MD

Senior Investigator Chief, Skeletal Disorders and Mineral Homeostasis Section
National Institute of Dental and Craniofacial Research
Director, Career Development and Research Training
NIH Interinstitute Endocrine Training Program
National Institutes of Health
Bethesda, Maryland

“Reaching an MAS/FD Diagnosis and What to Further Expect”

Dr. Collins from the National Institute of Health will begin our convention by discussing McCune-Albright Syndrome and Fibrous Dysplasia. He will explain how one is diagnosed with MAS/FD and the tests that should and may be done to confirm diagnosis. He will tell us what we need to know, and what to expect with time. He will update us on current research, informing us on how this research is progressing and what information we are learning from it!

Alison Boyce, MD

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

“Endocrine Care in FD/MAS” & “Pain in FD/MAS”

Dr. Boyce, also from the NIH, will follow Dr. Collins by presenting endocrine care and pain in FD/ MAS. During her presentation she will explain how the endocrine system is involved in FD/ MAS, what test should be done and continued throughout treatment. The importance of having an endocrinologist that is familiar with FD/MAS. She will also touch upon pain in FD/MS and ways to help/treat pain that patient’s may experience. She will talk about medications that we have found to help with pain in MAS/FD as well as ones that did not work as well.

Jason Malone, DO

Department of Pediatric Orthopedics
Nemours Children's Hospital, Orlando, Florida
Assistant Professor University of Central Florida

"Orthopedic Management of McCune-Albright/Fibrous Dysplasia"

Dr. Malone from Nemours Children's Hospital, will be presenting to us after lunch on orthopedic management in MAS/FD. He will be teaching us about past and current surgical management options; what has worked and what hasn't. He will help us gain a better understanding of the limitations of surgical techniques with fibrous dysplasia.

P.G. Shelton, MD DFAACAP, DFAPA

Child and Adolescent Psychiatrist, Senior Medical Officer
Child, Adolescent, and Family Service
MidCentral District Health Board, Palmerston North, New Zealand

A Year Lost, Examining the Effects of the COVID19 Pandemic on Youth

Navigating the stresses of a complex medical condition in light of a global pandemic. How to help ourselves and our children.

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MAS/FD Patient
Disability Advocate
Instagram: @smumps
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RUSSELL SILVER SYNDROME & SMALL FOR GESTATIONAL AGE

Jennifer B. Salem, MA

MAGIC RSS/SGA Director of Research & Education
Manhattan Beach, California

“Building Blocks for Success in Understanding RSS/SGA: 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, Jennifer Salem. You will learn terminology and concepts necessary to understand future presentations. Unless you consider yourself an expert on RSS/SGA or have an RSS/SGA teen, all families should plan to attend this presentation PRIOR to attending additional presentations.

Suparna Jain, MD

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

“All Grown Up with RSS/SGA; Now What?”

We have a large group of RSS/SGA kids who are now in their teens and 20's, and are joined by a number of adults who are just now finding others like them. Living with RSS/SGA means that trips to the endocrinologist do not cease just because growth hormone treatment is finished. There are many things our teens/adults can monitor in anticipation of what might occur as they age. While this segment is aimed at teens and adults, parents of younger children can certainly tune in to see what they can expect later on!

Madeleine D. Harbison, MD

Assistant Professor of Pediatrics
Adrenal and Steroid Disorders
Pediatric Endocrinology
Mt. Sinai School of Medicine of NY University
New York, New York

“Hypoglycemia, Ketonuria & Utilizing the Emergency Protocol”

Dr. Harbison will focus on recognizing and treating low blood sugar as well as testing for ketones to prevent lows that can occur with RSS/SGA children. In addition, you will learn how to utilize the protocol designed to quickly educate in emergency situations. This is a must see for all parents of RSS/SGA infants and toddlers, particularly those who are thin with low muscle mass and larger head for body. Please note that low blood sugar can continue through teens for some, so this is appropriate for all.

Eloïse Giabicani, MD, PhD

Physician Scientist
Sorbonne Université,
Hôpital Armand Trousseau
Paris, France

“RSS/SGA New Study Results”

Dr. Giabicani will present on a few new research topics including sleep disordered breathing, stem cell research as well as imprinting disorders. We hope to be able to add some additional information as well that may cover the new results regarding orthopedics and ENT.

Madeleine D. Harbison, MD

Assistant Professor of Pediatrics
Adrenal and Steroid Disorders
Pediatric Endocrinology
Mt. Sinai School of Medicine of NY University
New York, New York

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

As always, these are critical and mandatory topics for all parents to understand fully - join Dr. Harbison for a discussion on bone age and adrenarche and why you always have to monitor bone age and also bone age advancement to prevent premature cessation of growth.

S. Robert Rozbruch, MD

Pediatric Orthopaedic Surgeon | Hospital for Special Surgery;
Chief | Limb Lengthening and Complex Reconstruction Service (LLCRS);
Professor of Clinical Orthopaedic Surgery;
Weill Cornell Medicine, Cornell University
New York, New York

“Strategies for Treating Leg Length Discrepancy in RSS”

Join Dr. Rozbruch to understand the newest methods used for treating leg length discrepancies. This is a must see presentation for any family with a child with an LLD. Learn what the options are and what to expect on this part of your journey - as well as what you may be able to do now to best help your child prevent long term back, spine, hip and joint problems.

Irène Netchine, MD, PhD

Professor of Physiology
Sorbonne Université
Trousseau Hospital
Paris, France

“The Diagnostic Map for SGA to RSS with Treatment Recommendations”

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. We have seen an increase in the number of SGA children who are being given a clinical diagnosis before exhausting the recommended testing. Children who continue on with testing may be able to find a relevant diagnosis for a different growth disorder. Join Dr. Netchine to understand what diagnostic tests can tell us and the importance of pursuing testing to ensure proper treatment as well as prevent using treatments that may be contraindicated or cause harm.

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

Licensed Clinical Psychologist

Health Service Psychologist

Board Certified Music Therapist

Author of For What It's Worth: A Perspective on How to Thrive and Survive Parenting

Mom of 2 humans (6 and 7) and 2 dogs

Chicago, Illinois

"Understanding The Impact of ADHD & Anxiety on the Developing Brain & Your Child's Self-Esteem, Plus Tips on What You Can Do to Help"

Dr. Cook will present on the often overlooked diagnoses that affect many more of our kids than previously thought; anxiety and/or ADHD. Studies have shown that medically complex kids have more anxiety than other kids. In addition, many of our kids are experiencing learning difficulties including, but not limited to, ADHD. Dr. Cook will provide insight on what symptoms parents should watch for as well as treatment options.

Lindsay Salem, MPH Candidate

RSS Adult

Returned Peace Corps Volunteer

Baltimore, Maryland

"Living with RSS/SGA/Temple Syndrome: A Teen/Adult Perspective"

Join Lindsay Salem, a young adult with RSS, as she moderates a discussion panel of teens and adults with RSS/SGA/Temple Syndrome. 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.

Veronica Mericq, MD

Professor of Pediatrics

Institute of Maternal and Child Research, Faculty of Medicine University of Chile

Department of Pediatrics, Clinica las Condes

Santiago, Chile

"Consequences of Being Born SGA & Mitigating Risks with GHT"

The evidence has long been published that being born SGA has potential long term consequences on health; particularly in regards to metabolic syndrome. Growing research over the past decade has shown that growth hormone therapy can mitigate several of these risk factors related to being born SGA. This segment with Dr. Mericq will discuss those risks and how growth hormone can impact long term health.

Dayna Carney, RN

Pediatric Nurse

Children's Mercy Hospital

MAGIC RSS Division Consultant

"All About Growth Hormone for RSS/SGA/Temple Syndrome"

While society may sometimes view growth hormone treatment as something they hear about in a news scandal, GH is something everyone produces in their body and it is a hormone necessary for many functions, in addition to linear

growth. Join RSS Division Consultant and registered nurse Dayna Carney for a segment on how growth hormone is related to health for our RSS/SGA/Temple children.

Jennifer M. Pusins, CScD, CCC-SLP, BCS-S, IBCLC

Associate Professor/Clinical Supervisor
Board Certified Specialist in Pediatric Swallowing & Swallowing Disorders
International Board Certified Lactation Consultant
Department of Speech-Language Pathology
Nova Southeastern University
Ft. Lauderdale, Florida

“The Mechanics of Eating; Feeding and Swallowing Difficulties in RSS/SGA/Temple Syndrome”

While feeding difficulties are most common in the first several years, some will experience these longer. Dr. Pusins will help us understand the physical mechanics of eating and swallowing as well as provide information to address common feeding issues in this group of children.

TEMPLE SYNDROME

Prof Karen Temple, MD

Professor of Medical Genetics, Faculty of Medicine
University of Southampton
Honorary Consultant in Clinical Genetics
University Hospital Southampton
Southampton, Hampshire, UK

“Temple Syndrome; Genetics & Diagnostics”

Dr. Temple will provide 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.

Justin Davies, MD, FRCPCH, MRCP,

Consultant Paediatric Endocrinologist
Honorary Associate Professor, University of Southampton
Southampton Children’s Hospital,
University Hospital Southampton, Southampton, UK.

Prof Karen Temple, MD

Professor of Medical Genetics, Faculty of Medicine
University of Southampton
Honorary Consultant in Clinical Genetics
University Hospital Southampton
Southampton, Hampshire, UK

“Treatment & Management of Temple Syndrome”

Dr. Davies will be joined by Dr. Temple to present the most current treatment and management strategies for Temple Syndrome. This is a must see presentation for all families with children with Temple Syndrome.

Deborah Mackay PhD

Professor of Medical Epigenetics
University of Southampton, UK

“Genome Sequencing and Causes of RSS/SGA: Hunting for the Pearl of Great Price”

Dr. Mackay returns to fascinate families with her tales from the genetics lab! Whole genome sequencing has revealed some surprising and interesting results for families of children with growth disorders and Dr. Mackay is here to share the latest information.

Danielle Binns CNP, BA

Certified Nutritionist & Picky Eating Expert
Mother of RSS daughter, Sienna (age 7)
Toronto, Ontario

“Practical Nutrition for RSS/SGA & Temple Syndrome; Underweight vs. Overweight”

Presented by a certified nutritionist who also happens to be mom to a daughter with RSS, this segment will be geared towards families struggling to ensure appropriate caloric intake and good nutrition. Danielle knows first-hand the difficulties of feeding an RSS/SGA child and has great practical tips you can implement now to either increase or decrease intake. This presentation will be helpful for our families of children with Temple Syndrome who may initially present with lack of interest in eating, and then later over eat.

OPTIC NERVE HYPOPLASIA/SEPTO OPTIC DYSPLASIA

Mark Borchert, M.D.

Director of the Eye Birth Defects Program and Eye Technology Program
Associate Professor of Clinical Ophthalmology and Neurology
Children's Hospital Los Angeles
Borchert Laboratory
Los Angeles, California

“Correcting Misconceptions about ONH”

What is ONH, what are the true and fictitious clinical associations. How to navigate an ONH diagnosis.

Mark Borchert, M.D.

Director of the Eye Birth Defects Program and Eye Technology Program
Associate Professor of Clinical Ophthalmology and Neurology
Children's Hospital Los Angeles
Borchert Laboratory
Los Angeles, California

“State of the Art Research on ONH”

What does Dr. Borchert's newest research say about clinical risks and causes of ONH.

Ellen Lancon Connor, M.D.

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

"Endocrinology 101 and ONH"

In this talk, we will begin with an overview of the endocrine system, how it works and how to address the endocrine issues associated with ONH.

Mark Borchert, M.D. & Ellen Lancon Connor, M.D.

"ONH Q&A"

This session will be the time for you to ask all the questions you have about ONH. (endocrine, vision, medical management, school issues)

Mimi Kim, MD MSc

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

"Adrenal Insufficiency"

This session will give a deep overview of Adrenal Insufficiency - causes, symptoms, treatments, and how to navigate illness and emergencies.

Erin Beamon

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

"Solu-Cortef Training"

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

PANHYPOPITUITARISM

Mitchell E. Geffner, MD

Professor 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

"Hypopituitarism: Out of the PAN and into the Fire"

This session is a broad overview of Hypopituitarism, its causes, symptoms, and treatments.

Mitchell E. Geffner, MD

Professor 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

“New Treatments for Hypopituitarism”

This session will highlight new and upcoming treatments for many of the hormone deficiencies that patients with hypopituitarism suffer from. Medications to be highlighted: growth hormone, thyroid hormone, hydrocortisone, and testosterone.

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

“Transitioning to Adulthood”

A brief look at the ins and outs of transitioning from pediatric care to adult endocrine care.

P.G. Shelton, MD DFAACAP, DFAPA

Child and Adolescent Psychiatrist, Senior Medical Officer
Child, Adolescent, and Family Service
MidCentral District Health Board, Palmerston North, New Zealand

“A Year Lost, Examining the Effects of the COVID19 Pandemic on Youth”

Navigating the stresses of a complex medical condition in light of a global pandemic. How to help ourselves and our children.

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GENERAL SESSIONS – SUNDAY AFTERNOON

Pamela Alma Weymouth, MSW, MFA

Health Advocate

Writer

www.mightykidscan.com

“Fearless Needle Pokes: How to Make Humans of All Ages Feel Safe During Injections, Blood Draws and Most Pokes”

Needle Phobia is a real and often overlooked condition that can lead to a great deal of stress for children and adults. Pamela Alma Weymouth is a mother and social worker who experienced firsthand the toll this took on her own son, who was born with Congenital Hypothyroidism. After years of being told there was no solution, she learned there were pain blocking tools and 7 key steps that would transform her son’s experience. Through AngelAidCares.com and MightyKidsCan.com she is bringing this groundbreaking program to patients in the rare disease community.

Megan Donnell, MBA, SHRM-CP, PHR

SGA Division Consultant, MAGIC Foundation

Massachusetts License - Benefits Broker

Norwell, Massachusetts

“Health Insurance 101”

Health insurance is complex and often frustrating. Health Insurance 101 will give you the basics - but we will also cover fully insured vs. self funding, PBMs (Pharmacy Benefit Managers) and prescription drugs, how to evaluate and pick your plan and appeals.

Ryan Lehfeldt

Actor

Russell-Silver Syndrome adult

Fullerton, California

“Built for This...My Story”

As a young man with Russell-Silver Syndrome who always seemed to straddle the grey area between "typical" and "disability", he has been bullied, discounted and doubted plenty of times, but Ryan has a positive attitude and relentless pursuit of excellence doing what he loves. Listen in for an encouraging word and reminder that we are only limited by our dreams.

Yvette Q. Getch, Ph.D., CRC

Professor and Coordinator, Graduate Counseling Programs

The University of South Alabama

Mobile, Alabama

“Success at School: Section 504 or an IEP...Which is Best?”

When children struggle at school whether it's academically, physically, or emotionally, parents and teachers become concerned. We all want our children to learn, grow, and be as healthy and happy as possible. Investigating what is behind children's struggles and then determining how to help them can be confusing and frustrating especially when the lingo used is unfamiliar. Dr. Yvette Getch will help you learn about IEPs and 504s and how our children can receive the accommodations and supports needed to be successful at school. Dr. Getch is a parent of an adult with RSS, an adult with GHD, and 4 of her 5 children needed either an IEP or a 504 Plan to be successful in school. She understands the questions parents have and the frustration they often feel when trying to navigate the educational system. This presentation will provide you with the knowledge and tools you need to work with school personnel to provide the supports your child needs to be successful.