

February 9-11, 2023

Le Westin Montreal Hotel

17th Annual

CPEG Scientific Meeting

Hosted in Montréal by:

Division of Pediatric Endocrinology at the Université de Montréal and CHU Sainte-Justine

Full Program



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We wish to extend our warmest welcome to you all to the 17th annual Scientific Meeting of the Canadian Pediatric Endocrinology Group (CPEG). It is such a pleasure to be hosting the first in-person CPEG meeting since 2020! After three years of pandemic challenges, it will be so nice to be able to share and catch up «live»!

This year's scientific program is exciting and rich, inspired by your suggestions for symposium themes and topics, with excellent speakers to share their insights and expertise. Don't forget to visit our high-quality posters (the highest number ever submitted to CPEG!), as well as the booths of the industry sponsors who support our conference. We also hope to see you at our Friday evening activity, held at the PHI Center, an innovative and unique modern art center in Montreal, where multimedia and virtual reality intersect with art.

Enjoy the 2023 CPEG Meeting!

Mélanie, Despoina, Lyne, Caroline and Geneviève (the local organizing committee)

Dear Attendees,

On the behalf of the Scientific Committee and the Canadian Pediatric Endocrine Group (CPEG) Executive, I would like to welcome you all to the 17th Annual CPEG Scientific Meeting.

Our annual CPEG meetings provide a wonderful opportunity for the Canadian pediatric endocrine community to come together to learn, network, share ideas, visit old friends, and make new ones. The last two years of virtual meetings provided a great but different experience. The learning and science were still excellent but, despite some great programming (like sea shanties, and virtual chocolate and wine tasting), we were limited in what we could provide for social networking and interaction. I am truly excited to again see you all in person this year.

With our partners at the University of Toronto Continuing Professional Development, the Scientific Committee has developed a fantastic meeting and program. The meeting highlights work from our local hosts from Université de Montréal and CHU Sainte-Justine, and also includes presentations by other national and international experts. We will also enjoy the now infamous CPEG debate. As always, learners and others will present their work in scheduled oral and poster abstract sessions. This year, we have the most abstracts in our meeting's history. Back in person this year, we will restart the fun and informative one-minute poster highlights. We hope that our efforts have produced a program that meets the needs of all attendees including nurses, scientists, endocrinologists, other care providers, and trainees.

I would like to thank the members of the Scientific Committee for their hard work in planning this meeting. A special acknowledgement to Mélanie Henderson, the Local Chair, and her local committee who were instrumental in arranging the scientific program, and of course planning and hosting the Friday night social event.

Finally, thank you to our sponsors who continued to support us the last two virtual years and again this year. Their support is vital in making this meeting possible and I encourage you to explore their exhibits. CPEG would also like to thank those sponsors who also support our CPEG Fellowship Awards allowing us to train our future endocrinologists. This year's awardees will be announced on Saturday.

I hope you all have a stimulating, fun, and collegial meeting.

Sincerely,

Seth Marks MD, MSc, FRCPC

Det MAK

Chair, 2023 CPEG Scientific Committee

Scientific Committee

Seth Marks (Chair)

Melanie Henderson (Local Chair)

Rebecca Perry (Incoming Chair)

Diane Wherrett

Manpreet Doulla

Sharon Costantini

Mark Inman

Raelynn Friesen

Lyne Chiniara

Despoina Manousaki

Annie Gervais

Caroline Boucher

Wendy Schwarz

Paola Luca

Kate Potter

Geneviève Nadeau

Munier Nour (Ex Officio Member)

Faculty Disclosure

It is the policy of the University of Toronto, Temerty Faculty of Medicine, Continuing Professional Development to ensure balance, independence, objectivity, and scientific rigor in all its individually accredited or jointly accredited educational programs. All speakers, moderators, facilitators, authors and scientific planning committee members participating in University of Toronto accredited programs, are required to disclose to the program audience any real or apparent conflict(s) of interest that may have a direct bearing on the subject matter of the continuing education program. This pertains but is not limited to relationships within the last FIVE (5) years with for-profit organizations, not-for-profit and public sector sponsors and donors, biomedical device manufacturers, or other corporations whose products or services are related to the subject matter of the presentation. The intent of this policy is not to prevent a speaker with a potential conflict of interest from making a presentation. It is

merely intended that any potential conflict of interest should be identified openly so that the listeners may form their own judgements about the presentation with the full disclosure of facts. It remains for the audience to determine whether the speaker's outside interests may reflect a possible bias in either the exposition or the conclusions presented.

Accreditation

Royal College of Physicians and Surgeons of Canada - Section 1

This event is an Accredited Group Learning Activity (Section 1) as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada, approved by Continuing Professional Development, Temerty Faculty of Medicine, University of Toronto. You may claim a maximum of 10.5 hours (credits are automatically calculated).

Session Polling and Q&A

Visit slido.com and enter the code **CPEG** (not case-sensitive) or scan the QR code on the right to participate in polling questions and to submit your question during each session. The moderator will review the questions and ask the speaker during Q&A.

Wi-Fi Internet Access

Network: Westin Conference

Password: westin2023



Full Conference Program

You can download the full conference program from the link in your conference reminder email or from the conference home page at www.cpd.utoronto.ca/cpeg-gcep/

Program Overview

The 17th Annual Scientific Meeting of Canadian Pediatric Endocrine Group (CPEG) includes a program of current and high-level content in pediatric endocrinology. The meeting also provides an opportunity for the Canadian pediatric endocrine community to come together, network and share ideas.

For the first time in three years, after two years of successful but perhaps lonely virtual format meetings, this year's meeting will be in-person! The organizing committee has worked hard to build on the successes of both the past in-person and virtual meetings. We hope to provide attendees with an exceptional meeting experience.

The program includes theme-based symposia, an annual debate, oral abstracts, and poster presentations. Presenters include national and international experts. The meeting also provides a forum for trainees to present their work.

We have an exciting program planned for this year that should meet your educational needs as it has in past years. We look forward to visiting and learning with you.

Program Learning Objectives

At the conclusion of this conference, the participants will be able to:

- 1. Utilize clinical and basic science knowledge to further develop a diagnostic and treatment approach to neonates and children with calcium disorders
- 2. Recognize the complexities of care of transgender, non-binary, and gender diverse youth
- 3. Appreciate the broad risk of complications in youth living with diabetes
- 4. Identify a variety of treatment approaches for youth living with type 2 diabetes
- 5. Formulate a comprehensive diagnostic and management approach for children with Cushing's syndrome
- 6. Compare different treatment options in youth with autoimmune hyperthyroidism (Graves' disease)

Session Learning Objectives

Symposium I: Calcium Disorders

Neonatal Hypercalcemia - Celia Rodd

Objectives:

- 1. Understand the complexities of calcium homeostasis and the role of calcitropic hormones in the first weeks of life
- 2. Diagnose neonates with hypercalcemia
- 3. Develop a comprehensive investigative approach to these neonates
- 4. Understand advances in the diagnosis of underlying etiologies, especially in the Canadian context
- 5. Understand newer targeted therapies

Phosphate's Role in Extracellular Matrix Mineralization in Bone Health and Disease: The Stenciling Principle - Marc McKee

Objectives:

- 1. After this presentation, participants will be able to explain how mineralization occurs in normal bone at both the micro- and nano-scale, and how mineral packing patterns contribute to bone strength
- 2. After this presentation, participants will be able to describe the mineralization defects that occur in bones and teeth in the osteomalacia/odontomalacia diseases
- 3. After this presentation, participants will be able to better inform their osteomalacic patients (particularly those having X-linked hypophosphatemia) about the reasons underlying their deforming and/or fracturing soft skeletons and teeth.

Renal Complications in Children with Calcium Disorders - Véronique Phan

Objectives:

- 1. Recognized which calcium disorders can be associated with renal complications
- 2. Initiate a workup to follow potential renal complications associated with those Calcium Disorders
- 3. Identify those who would need a consultation to a nephrologist

Symposium II: Transgender Health

Challenges for the Pediatric Endocrinologist in the Care of Transgender and Gender Diverse Youth - Stephen Rosenthal Objectives:

- 1. Review evidence for biological underpinnings of gender identity
- 2. Review Mental Health outcomes in TYC-U.S. multi-center study
- 3. Review Fertility Preservation in Transgender Youth
- 4. Review outcomes of current treatment models for transgender youth
- 5. Bone Health
- 6. Physiologic and metabolic parameters

An Up to Date Approach to the Care of the Non-Binary Adolescent - Dan Metzger, Karine Khatchadourian Objectives:

- 1. Review the Canada Census 2021 demographic information about trans and nonbinary Canadians
- 2. Understand clinical challenges in endocrine management of nonbinary adolescents
- 3. Review current treatment options for the nonbinary adolescent based on young person's pubertal status and goals

Symposium III: Diabetes

Cardiovascular Complications in Youth with Type 1 Diabetes - *Mélanie Henderson* Objectives:

- 1. Understand the epidemiology of cardiovascular disease among individuals living with T1D
- 2. Understand sex differences in cardiovascular disease risk among individuals living with T1D
- 3. Identify potential determinants of early cardiovascular disease risk among youth living with T1D

Complications in Youth With Type 2 Diabetes: The Widening Circle - *Elizabeth Sellers* Objectives:

- 1. Describe the complications of childhood onset type 2 diabetes
- 2. Recognize the broad domains of complications in childhood onset type 2 diabetes

Treatment Approaches in Youth with Type 2 Diabetes - Phil Zeitler

Objectives:

- 1. Identify unique features of the pathophysiology of type 2 diabetes in youth
- 2. Develop an individualized approach to treatment of type 2 diabetes in youth based on variability in pathophysiology

Symposium IV: Cushing's Syndrome

Cushing's Syndrome: Case 1,2,3 & Approach to Cushing's Syndrome and Review of Cases - Despoina Manousaki, Lyne Chiniara, Cheri Deal, André Lacroix

Objectives:

- 1. Improve their capacity to diagnose Cushing's syndrome
- 2. Be familiar with the strategies to identify the etiology of Cushing's syndrome
- 3. Select the most adequate therapeutic approaches for the various etiologies of Cushing's syndrome
- 4. Be familiar with required long-term follow-up of their patients treated for Cushing's syndrome

Debate

Be It Resolved That Long Term Antithyroid Medication Should Be Used Instead of Radioactive Iodine in the Treatment of Youth With Autoimmune Hyperthyroidism (Graves' Disease) - Pro: *Elizabeth Rosolowsky* Con: *Preetha Krishnamoorthy* Objectives:

- 1. Outline the benefits and risks of antithyroidal medications in the treatment of pediatric Graves' disease
- 2. Outline the benefits and risks of radioactive iodine in the treatment of pediatric Graves' disease
- 3. Tailor long-term treatment options to the individual pediatric patient with Graves' disease
- 4. To describe the pros and cons of use of medication for treatment of pediatric obesity

CPEN Symposium

Endocrine Management of the Transgender Youth - *Karine Khatchadourian* Objectives:

- 1. Understand clinical challenges in endocrine management of nonbinary adolescents
- 2. Review current treatment options for the nonbinary adolescent based on young person's pubertal status and goals

Journey from Youth to Adult - Liam Rodrigues

Objectives:

- 1. To review and refine communication skills: How to interact appropriately with transgender people; and How to approach different subjects that can be delicate
- 2. To understand the different transition stages and processes that the individual experiences
- 3. To gain a better understanding of the impact of transition on the individual and family: Inform and understand the situation/reality of the patient
- 4. Access resources available to support their patients and families requiring thyroid ablation treatment

Gender Affirming Surgery at GrS Montreal: Process and Surgical Pathway - Evelyne Sheitoyan, Carla Geambasu Objectives:

- 1. Understand the process to access gender-affirming surgery at GrS Montreal
- 2. Understand the possible results with the procedures offered
- 3. Learn how to support a patient after a gender-affirming surgery at GrS MontrealAccess resources available to support their patients and families requiring thyroid ablation treatment

Fellows' Symposium

How to Successfully Tell Your Story: Tips for Oral Research Communications From a Clinician Scientist's Perspective - Despoina Manousaki

Objectives:

- 1. General rules of research communication
- 2. Tips for specific types of talks
- 3. The Do's and Don'ts in scientific presentations

A Clinical Approach to Differences of Sex Development : Navigating the Ambiguity - Lyne Chiniara Objectives:

- 1. Review the genetic and biochemical aspects of normal sexual determination, differentiation and development
- 2. Perform a standardized physical examination of a newborn with ambiguous genitalia and formulate findings using the correct terminology
- 3. Develop a logical approach to investigations
- 4. Provide full disclosure and optimal guidance for parents

Invited CPEG Speakers

Lyne Chiniara MD MA FRCPC

Assistant Professor Université de Montréal Pediatric Endocrinologist CHU Sainte-Justine Montreal, QC

Cheri Deal MD PhD FRCPC

Emeritus Professor, Université de Montréal Pediatric Endocrinologist and Associate Member Research Center, CHU Ste-Justine Montreal, QC

Mélanie Henderson MD PhD FRCPC

Pediatric Endocrinologist Associate Clinical Professor Division of Endocrinology and Diabetes University of Montreal Centre Hospitalier Universitaire Sainte-Justine Montreal, QC

Karine Khatchadourian MD MSc FRCPC

Pediatric Endocrinologist / Endocrinologue Pédiatrique Children's Hospital of Eastern Ontario (CHEO) Ottawa, ON

Preetha Krishnamoorthy MDCM FRCPC

Associate Professor McGill University Pediatric Endocrinologist The Montreal Children's Hospital Montreal, QC

André Lacroix MD FCAHS

Professor of Medicine Department of Medicine, Endocrine Division Centre hospitalier de l'Université de Montréal (CHUM) Montreal, QC

Despoina Manousaki MD PhD FRCPC

Department of Pediatrics Department of Biochemistry and Molecular Medicine University of Montreal Research Center of the Sainte-Justine University Hospital Montreal, QC

Marc McKee PhD

Professor
Faculty of Dental Medicine and Oral Health Sciences
Faculty of Medicine and Health Sciences
McGill University
Montreal, QC

Dan Metzger MD FAAP FRCPC

British Columbia's Children's Hospital Endocrinology & Diabetes Unit Vancouver, BC

Véronique Phan MDCM MSc FRCPC

Professor of Pediatrics Faculté de Médecine Université de Montréal Pediatric Nephrologist CHU Ste-Justin Montreal, QC

Celia Rodd MD MSc FRCPC

Professor

Section of Pediatric Endocrinology & Metabolism
Department of Pediatrics and Child Health
Max Rady College of Medicine
University of Manitoba
Clinician Scientist
Children's Hospital Research Institute of Manitoba
Winnipeg, MB

Stephen Rosenthal MD

Professor of Pediatrics
Division of Pediatric Endocrinology
Medical Director
Child and Adolescent Gender Center
University of California
Mission Hall: Global Health and Clinical Sciences
San Francisco, CA

Elizabeth Rosolowsky MD MPH FAAP FRCPC

Associate Professor of Pediatrics Faculty of Medicine & Dentistry University of Alberta Pediatric Endocrinologist Stollery Children's Hospital Edmonton, AB

Elizabeth Sellers MD MSc FRCPC

Professor, Pediatric Endocrinology and Metabolism Department of Pediatrics and Child Health University of Manitoba Winnipeg, MB

Phil Zeitler MD PhD

Professor of Pediatrics and Clinical Sciences Head, Section of Pediatric Endocrinology University of Colorado School of Medicine Chair, Department of Endocrinology Children's Hospital Colorado Aurora, CO

Invited CPEN Speakers

Carla Geambasu RN Registered Nurse Clinician Postoperative Clinic GrS Montreal

Montreal, QC

Karine Khatchadourian MD Msc FRCPC Pediatric Endocrinologist / Endocrinologue Pédiatrique Children's Hospital of Eastern Ontario (CHEO) Ottawa, ON **Liam Rodrigues**Patient Advocate
Gatineau, OC

Evelyne Sheitoyan RN Registered Nurse Clinician Postoperative Clinic GrS Montreal Montreal, QC

Invited Fellows' Symposium Speakers

Lyne Chiniara MD MA FRCPC Assistant Professor Université de Montréal Pediatric Endocrinologist CHU Sainte-Justine Montreal, QC **Despoina Manousaki** MD PhD FRCPC
Department of Pediatrics
Department of Biochemistry and Molecular Medicine
University of Montreal
Research Center of the Sainte-Justine University Hospital
Montreal, QC

Invited CPEG Speaker Biographies

Lyne Chiniara

Dre Chiniara is a pediatric endocrinologist at the CHU Sainte-Justine, and an assistant professor at Université de Montréal. She obtained her medical degree from Université de Montréal in 2008 and completed her training in pediatric endocrinology in 2013. She is the co-founder and co-director of the CHU Sainte-Justine gender diversity clinic.

Cheri Deal

Cheri Deal, PhD,MD,FRCPC and Professor of Pediatrics completed her mandate as the Chief of Endocrinology and Diabetes at the Sainte-Justine Mother-Child University of Montreal Hospital in 2020, where she worked since 1992 as a Pediatric Endocrinologist and Clinical Investigator at the Ste-Justine Research Institute. She was recently made an Emeritus Professor at the University of Montreal, where she continues to teach, and she maintains her affiliation with the research center. She is also a Pediatric Endocrinology Consultant for the ELNA Tiny Tots multispecialty pediatric clinic in Montreal, Canada.

Dr. Deal has completed mandates as the President of the Canadian Pediatric Endocrine Group and of the Canadian Society of Endocrinology and Metabolism (CSEM), as Council Member for the Growth Hormone Research Society and the International Society of Endocrinology (ISE), as the Training Director for the Royal College program in Pediatric Endocrinology and Diabetes for the University of Montreal, and as the site leader for the pan-Canadian Child Health Clinical Scientist Training Program (CCHC-SP). She is currently co-chair of the Education Committee of ISE, and is on the curriculum committee of CCHCSP (now renamed ENRICH-Empowering Next-generation Researchers in perinatal and Child Health) and the European Society of Pediatric Endocrinology (ESPE) summer school organizing committee.

Mélanie Henderson

Mélanie Henderson is a Pediatric Endocrinologist and researcher at CHU Sainte-Justine and Clinical Associate Professor in the Department of Pediatrics at the Université de Montréal. Her research focuses on pediatric cardiometabolic health and its epidemiology. She is particularly interested in the influence of lifestyle habits (physical activity, sedentary behavior, diet and sleep) on metabolic

health in various population, including healthy children and those living with Type 1 and Type 2 Diabetes, in order to identify the best preventive strategies. She is co-director of the "Centre CIRCUIT", that proposes novel strategies for the treatment or prevention of cardiovascular disease risk in children. She is also director of the "Cardiovascular and Metabolic Health" research axis at the CHU Sainte-Justine. Dr Henderson has received several awards, including the «Young Investigator Award» from the CSEM (Canadian Society for Endocrinology and Metabolism).

Karine Khatchadourian

Dr Karine Khatchadourian is a pediatric endocrinologist at the Children's Hospital of Eastern Ontario and an Assistant Professor of Pediatrics at the University of Ottawa. She's the co-director of CHEO's Diversity Clinic. As a fellow, she trained with Dr Metzger at BC Children's Hospital and completed an elective at the VU University Medical Center in Amsterdam. She published the first Canadian study characterizing the endocrine management of transgender adolescents. She has been providing hormone therapy and support to transgender adolescents since 2014 at CHEO's Diversity Clinic. She has supervised three pediatric endocrine fellows in research projects examining effects of GnRH agonist therapy in transgender adolescents. She was also a site co-Investigator for the TransYouth Can! study.

Preetha Krishnamoorthy

Preetha Krishnamoorthy completed her pediatrics residency and fellowship in Pediatric Endocrinology at the Montreal Children's Hospital, and has been an attending staff there since 2003. She is the Director of Pediatric Undergraduate Education at McGill. She won the Paige and Bernard Kaplan Award for excellence in teaching in 2004, the Osler Award by McGill Medicine's graduating class of 2012 of 2022, and was named to the Faculty Honour List for Educational Excellence. She is one of the recipients of the 2016 CAME Certificate of Merit Awards. In addition to her medical-related teaching, she loves to teach Zumba!.

André Lacroix

Prof Lacroix's major areas of research: molecular and genetic mechanisms of adrenal tumors and hyperplasias leading to Cushing's syndrome, primary aldosteronism and adrenal tumorigenesis. Role of aberrant adrenal hormone receptors in adrenal overfunction. New drugs in the therapy of Cushing's disease and primary aldosteronism, of adrenocortical cancer and pheochromocytomas.

Prof Lacroix's major areas of research: molecular and genetic mechanisms of adrenal tumors and hyperplasias leading to Cushing's syndrome, primary aldosteronism and adrenal tumorigenesis. Role of aberrant adrenal hormone receptors in adrenal overfunction. New drugs in the therapy of Cushing's disease and primary aldosteronism, of adrenocortical cancer and pheochromocytomas.

Despoina Manousaki

Dr. Despoina Manousaki received her MD from the University of Athens, Greece, her post-graduate training in pediatrics in Switzerland (University Hospitals of Geneva) and in pediatric endocrinology in Canada (Sainte-Justine Hospital in Montreal). Subsequently, she obtained a PhD in genetic epidemiology from McGill University. Dr. Manousaki's research focuses on the genetics of complex disease in childhood. Her team uses genomics, bioinformatics, and genetic epidemiology methods to better understand the genetic architecture of complex disease, and to apply these findings to predict disease risk and develop new disease biomarkers. Her research interests lie in the field of genetics of type 1 and type 2 diabetes in children, vitamin D levels, youth-onset osteoporosis, pediatric obesity, growth and puberty. Dr. Manousaki is an FRQS Junior 1 Clinician researcher and holds an ENRICH career development award and a JDRF strategic research award.

Marc McKee

Marc McKee is a full professor at McGill University in Montreal where he holds the Canada Research Chair in Biomineralization. He received his Ph.D. degree from McGill University in cell biology, followed by postdoctoral training at Harvard / The Children's Hospital Boston, and then he held appointments at the Forsyth Institute in Boston and University of Montreal. His research focuses on biomineralization in bones, teeth, otoconia and eggshells, and in pathologic calcification. He has received two Distinguished Scientist Awards from the International Association for Dental Research (1996 Young Investigator Award, 2003 Biological Mineralization Award), the 2018 Adele Boskey Esteemed Scientist Award from the American Society for Bone and Mineral Research, and the 2018 C.P. Leblond Award From the Quebec Network for Bone and Oral Health Research.

Dan Metzger

Dr. Metzger is a Clinical Professor of Pediatrics at the University of British Columbia, and a Pediatric Endocrinologist working on the Endocrinology & Diabetes Unit of BC Children's Hospital.

Dr. Metzger and his colleagues—in collaboration with hospital- and community-based mental health professionals—began seeing transgender kids in 1998, and they have now seen nearly 800 trans and gender-diverse children and youth in the BCCH Gender Clinic, one of the busiest in Canada.

Véronique Phan

Dre Véronique Phan is a full professor of Pediatrics at l'Université de Montréal, practicing as a nephrologist at the CHU Ste-Justine. Her main interests are kidney transplantation and medical education. She is the educational assistant of the UGME program of the Université de Montréal. She is involved in many national committees.

Celia Rodd

Dr. Rodd's interests focus on calcium homeostasis, vitamin D metabolism and adequacy, bone physiology, growth charts, and the epidemiology of endocrine disorders. She has published over 120 peer-reviewed manuscripts in these areas and has been funded by multiple agencies, including CIHR. Even in retirement, she continues to love lifelong learning.

Stephen Rosenthal

Stephen M. Rosenthal, MD, Professor of Pediatrics at University of California San Francisco (UCSF), has served as Program Director for Pediatric Endocrinology and Director of the Pediatric Endocrine Clinics, and currently serves as co-founder and Medical Director of the multidisciplinary UCSF Child and Adolescent Gender Center (CAGC). He is Principal Investigator (PI) (multiple PI format) for NIH/NICHD "The Impact of Early Medical Treatment in Transgender Youth" and co-Investigator on two additional NIH-funded studies focused on optimizing care of transgender/gender diverse youth. He has also co-directed a collaboration between the UCSF CAGC and the San Francisco Department of Public Health to develop community outreach services for gender diverse youth. Dr. Rosenthal has recently completed his terms as President of the Pediatric Endocrine Society and as Vice President and member of the Board of Directors of the Endocrine Society. He is currently a member of the Board of Directors of the World Professional Association for Transgender Health.

Dr. Rosenthal's principal non-work passion is figure skating. He has participated in various adult national and international competitions, most recently in Paris, August, 2018.

Elizabeth Rosolowsky

Liz Rosolowsky is an academic pediatric endocrinologist with a substantial clinical diabetes and endocrinology practice. She has served as the former Residency Program Director in Pediatric Endocrinology & Metabolism and is the current Endocrinology Course Coordinator for the undergraduate medical education program at the University of Alberta. She also chairs the Fellows' Retreat Committee for the Pediatric Endocrine Society and the Continuing Professional Development Committee for the Canadian Society of Endocrinology and Metabolism.

Elizabeth Sellers

Elizabeth Sellers is a Pediatric Endocrinologist at the Winnipeg Children's Hospital . As a clinician and clinician scientist, her primary focus has been the epidemiology, pathophysiology, complications, treatment, and support of youth with type 2 diabetes with a particular interest in Indigenous populations.

Phil Zeitler

Research in Dr. Zeitler's group focuses on understanding obesity, insulin resistance, renal function, and diabetes in adolescents, and the physiology of insulin resistance and cardiorenal risk in special populations. Dr. Zeitler has been Chair of TODAY, an NIH-funded, multi-center longitudinal study of adolescents with type 2 diabetes since 2003.

Invited CPEN Speaker Biographies

Carla Geambasu

Carla Geambasu, is a registered nurse clinician who graduated from the University of Montreal in 2005. She began her career at the mother and newborn department of the Royal Victoria Hospital, a specialized unit for high-risk pregnancy. After working there for 8 years, she went on to work as a nursing teacher at the Sir Wilfrid Laurier School Board for another 5 years before joining the GRS team.

She is a preoperative nurse working at the GRS clinic since 2018. She evaluates all the different components of the client's files to facilitate their journey toward surgery. This evaluation includes the WPATH standards of care criteria, physical and mental health readiness, and teaching through phone or virtual consultations.

Karine Khatchadourian

Dr Karine Khatchadourian is a pediatric endocrinologist at the Children's Hospital of Eastern Ontario and an Assistant Professor of Pediatrics at the University of Ottawa. She's the co-director of CHEO's Diversity Clinic. As a fellow, she trained with Dr Metzger at BC Children's Hospital and completed an elective at the VU University Medical Center in Amsterdam. She published the first Canadian study characterizing the endocrine management of transgender adolescents. She has been providing hormone therapy and support to transgender adolescents since 2014 at CHEO's Diversity Clinic. She has supervised three pediatric endocrine fellows in research projects examining effects of GnRH agonist therapy in transgender adolescents. She was also a site co-Investigator for the TransYouth Can! study.

Liam Rodrigues

My name is Liam Rodrigues, I am 25 years old and I am originally from Gatineau, Qc. I have been working as a tattoo artist for a few years. In 2014 I made the most important decision of my life, that of starting my transition from woman to man.

Evelyne Sheitoyan

Evelyne Sheitoyan, is a registered nurse clinician who graduated from the University of Montreal. She has some experience in the OR in orthopedics before starting at GrS Montreal as an assistant head nurse at Asclépiade in 2018. She has been working at the GrS Montreal postoperative Clinic since September 2019. She is providing a close follow-up to patients who require it (all surgeries combined), and she does virtual consultations, as well as examinations and treatments to those who come for a consultation at the postoperative clinic.

Invited Fellows' Day Speaker Biographies

Lyne Chiniara

Dre Chiniara is a pediatric endocrinologist at the CHU Sainte-Justine, and an assistant professor at Université de Montréal. She obtained her medical degree from Université de Montréal in 2008 and completed her training in pediatric endocrinology in 2013. She is the co-founder and co-director of the CHU Sainte-Justine gender diversity clinic.

Despoina Manousaki

Dr. Despoina Manousaki received her MD from the University of Athens, Greece, her post-graduate training in pediatrics in Switzerland (University Hospitals of Geneva) and in pediatric endocrinology in Canada (Sainte-Justine Hospital in Montreal). Subsequently, she obtained a PhD in genetic epidemiology from McGill University. Dr. Manousaki's research focuses on the genetics of complex disease in childhood. Her team uses genomics, bioinformatics, and genetic epidemiology methods to better understand the genetic architecture of complex disease, and to apply these findings to predict disease risk and develop new disease biomarkers. Her research interests lie in the field of genetics of type 1 and type 2 diabetes in children, vitamin D levels, youth-onset osteoporosis, pediatric obesity, growth and puberty. Dr. Manousaki is an FRQS Junior 1 Clinician researcher and holds an ENRICH career development award and a JDRF strategic research award.

M. Jetha

Awarded Fellowship Listing

1992-1993	M. Lawson	2007-2008	B. Wicklow	2017-2018	C. Nugent
			T. Pinto, B. Babic		S. Fuchs
1993-1994	S. Lawrence		J. Deladoey		
	M. Lawson			2018-2019	J. Sorbara
	A. Simone	2008-2009	A.M. Sbrocchi		
			P Olivier	2019-2020	A. Chesover
1994-1995	S. Lawrence		T. Pinto		B. Navabi
	S.Taback				
	A. Simone	2009-2010	R. Shulman	2020-2021	A. Marr
			P Olivier		M. Lautatzis
1995-1996	C. Vaz		T. Edouard		J. Ladd
	S.Taback		S. Runge-Wildi		H. Geddie (declined)
	B. Cummings		C. Saaman		
				2021-2022	F. Babalola
1996-1997	J. Hamilton	2010-2011	E. Bassilious		M. Jiang
	E. Sellers		J. Wasserman		
	B. Cummings		Y. Yeshayahu	2022-2023	K. Oei
			S. Tsai		T. Dyer
1997-1998	J. Hamilton				K. Pabedinskas
	E. Sellers	2011-2012	M. Millete		M. Feldman
	B. Cummings		J. Wasserman		
			C. Zuijdwijk		
1998-1999	J. Curtis		M. Cohen		
	J. Hamilton				
		2012-2013	J. Harrington		
1999-2000	J. Curtis		T. Oron		
	J. Hamilton		P. Luca		
			M. Nour		
2000-2001	C. Panagiotopoulos		D. Manousaki		
	C. Huang				
		2013-2014	K. Winston		
2001-2002	C. Panagiotopoulos		C. Leblicq		
	S. Stock		A. Ens		
			B. Hursh		
2002-2003	P Krishnamoorthy		I. Rousseau-Nepton		
	P Zimakas				
	R. McEachern	2014-2015	I. Levy		
			D. Manousaki		
2003-2004	P Krishnamoorthy				
	H. Bui	2015-2016	L. Chiniara		
			S. Basak		
2004-2005	M. Nakhla		K. Verbeeten		
	J. Simoneau-Roy				
		2016-2017	C. Nugent		
2005-2006	M. Nakhla		K. Pundyk		
	I. Chapados		N. Coles		

Dr. John Bailey Resident Research Award

The John Bailey Award is named after Dr. John D. Bailey. Dr. Bailey graduated from the University of Toronto Medical School in 1949, did his training in pediatrics at the Hospital for Sick Children, Toronto and the University of Manitoba in Winnipeg. By the mid-1950s he was firmly established as a major contributor to pediatric care and education at the Hospital for Sick Children and University of Toronto. His major area of interest was in understanding normal and abnormal growth patterns in childhood and adolescence. He was the first chief of the Endocrine Division at the Hospital for Sick Children and an important contributor to the development of the Canadian national growth hormone treatment program. He was the consummate academic: clinically fastidious, intellectually challenging and constantly learning and sharing.

Below is a list of the recipients of the Dr. John Bailey Resident Research Award:

2007	Meranda Nakhla	2013	Karine Khatchadourian	2019	Julia Sorbara
2008	Meranda Nakhla	2014	Akash Sinha	2020	Christine Tenedero
2009	David Saleh	2015	Rayzel Shulman	2021	Richelle Waldner
2010	Brandy Wicklow	2016	Sanjukta Basak	2022	Funmbi Babalola
2011	Jonathan Wasserman	2017	Stephen Zborovski		
2012	Jennifer Harrington	2018	Marie Eve-Robinson		

CPEG Distinguished Service Award

The CPEG Distinguished Service Award will be awarded periodically (not annually) to a member who has shown exemplary service to the organization or to the discipline of pediatric endocrinology in Canada. The award will be focused on work that furthers the aims of CPEG and can be in one or more of the following areas: administration, teaching, research, clinical service. Nominations will be solicited by the CPEG Executive Committee every 1- 3 years. CPEG members can put forward a name for nomination at any time. The nomination should include a letter signed by two CPEG members in good standing describing the contributions of the nominee. The award will be presented at the annual CPEG business meeting. The awardee will receive a certificate and a \$1,000 donation to a charity of their choice.

Below is a list of past recipients of the CPEG Distinguished Service Award:

2017 Daniel Metzger 2019 Denis Daneman 2022 Cheril Clarson

Fellows' Symposium Thursday, February 9, 2023 (Room: St-Antoine B)

1300	Fellows Welcome & Lunch	
	Fellows' Symposium Chairs: Geneviève Nadeau (Montreal), Tracey Dyer (Montreal)	
1320	How to Successfully Tell Your Story: Tips for Oral Research Communications From a Clinician Scientist's Perspective	Despoina Manousaki
1420	Break	
1450	A Clinical Approach to Differences of Sex Development : Navigating the Ambiguity	Lyne Chiniara
1550	Fellows Closing Remarks	Geneviève Nadeau, Tracey Dyer
1600	Symposium Adjourns	

Welcome Reception Thursday, February 9, 2023 (Room: Ville-Marie B)

1600	On-Site Registration Opens
1700	Welcome Reception & Exhibits
1900	Adjourn for the Day

CPEG Program Friday, February 10, 2023 (Room: Fortifications)

Each presentation will include a 25% (minimum) of interactivity comprised of audience response questions (polling) and audience submitted Q&A via Slido.com.

0730	On-Site Registration & Breakfast		
0830	Opening Remarks & Friday Poster Highlights (Odd numbered Posters)		
	Symposium I: Calcium Disorders Chairs: Geneviève Nadeau (Montreal), Nathalie Alos (Montreal)		
0900	Neonatal Hypercalcemia	Celia Rodd	
0930	Phosphate's Role in Extracellular Matrix Mineralization in Bone Health and Disease: The Stenciling Principle	Marc McKee	
1000	Renal Complications in Children with Calcium Disorders	Véronique Phan	
1030	Break & Exhibits		

	Symposium II: Transgender Health Chairs: Lyne Chiniara (Montreal), Mark Palmert (Toronto)	
1100	Challenges for the Pediatric Endocrinologist in the Care of Transgender and Gender Diverse Youth	Stephen Rosenthal
1145	An Up to Date Approach to the Care of the Non-Binary Adolescent	Dan Metzger, Karine Khatchadourian
1230	Poster Viewing I (Odd numbered Posters)	
P1	Pediatric Diabetes: An Exploration of Shared Care in Nova Scotia	Maggie Flemming (IWK Health Center)
P3	Atypical Presentation of New Onset Diabetes With Hyperosmolar Hyperglycemic State in a Toddler	Jennifer Ladd (Nationwide Children's Hospital)
P5	Double Diagnosis: Acetaminophen Overdose and Diabetic Ketoacidosis?	Supraja Rengan (BC Children's Hospital)
P9	The Impact of Family Structure of Adolescents With T1D in Diabetes Management Aspects Before Transition to Adult Care.	Marie-Laurence Brunet (McGill University)
P11	Double the Trouble: Dual Presentation of Diabetic Ketoacidosis and Thyroid Storm in an Adolescent Patient	Nour Almutairi (Montreal Children's Hospital)
P13	Pediatric Endocrinology Education Amongst Trainees: A Scoping Review	Muhammed Abdulshakour (McMaster Children's Hospital)
P15	A Case of Familial Glucocorticoid Deficiency Type I With a Newly Reported Mutation in the MC2R Receptor	Alaa Alharbi (BC Children's Hospital)
P17	Pituitary Adenoma or Not? A Case of a Pediatric Pituitary Abscess – A Rare Adenoma Mimicker	Afnan Alawi (McMaster Children's Hospital)
P19	Hyperfunctioning Thyroid Nodules in Pediatric Patients: Rare and Unpredictable!	Pascaline Cattrysse (CHU Sainte Justine)
P21	Improving Healthcare Provider Education on High-Quality, Inclusive, Affirming, Trauma Informed and Culturally Sensitive Care for 2SLGBTQ+ Patients	Emma Metivier (Children's Hospital, London Health Science Centre)
P23	An Interesting Case of Persistent Low IGF-1 in Two Siblings	Kristina Pabedinskas (BC Children's Hospital)
P25	ACTH-Dependent Cushing's in an 8-Year-Old: A Diagnostic Challenge	Marian Thorpe (Stollery Children's Hospital, University of Alberta)
P27	Rebound Hyperkalemia After Focal Resection for Congenital Hyperinsulinism	Samantha Gerber (Alberta Children's Hospital)
P29	Metastatic Pheochromocytoma/Paraganglioma in Von-Hippel-Lindau Disease: An Uncommon Cause of Paediatric Hypertension	Joshua Stanley (The Hospital for Sick Children)

P31	Transition From Pediatric to Adult Obesity Care – A Retrospective Review	Mary Jiang (Children's Hospital of Eastern Ontario)	
P33	Associations Between Clusters of Parental Characteristics and Offspring Adiposity in Late Adolescence	Marina Ybarra (London Health Science Centre – Children's Hospital, Western University)	
P35	Outcomes of Growth Hormone Therapy for Children With Short Stature	Matthew Feldman (Alberta Children's Hospital)	
1300	Lunch & Exhibits		
1400	Oral Abstracts I Chairs: Rebecca Perry (Calgary), Trisha Patel (Vancouver)		
OR1	Nature Versus Nurture of the Puberty: A Combined Clinical and Polygenic Risk Score to Predict Pubertal Timing in Girls	Sara Moline (CHU Sainte- Justine)	
OR2	Mutations in the WFS1 Gene as a Cause of Non-syndromic Diabetes	Alix Van Poperinghe (Montreal Children's Hospital)	
OR3	Usability and Feasibility Testing of Eating for Wellness (E4W): An Image-Based Dietary Assessment App for Adolescents With Obesity	Krista Oei (The Hospital for Sick Children)	
OR4	Factors Associated With the Development of Dyslipidemia Among Pediatric Patients With Diabetes: A Single Center-Based Study	Fahd Alshammri (McMaster Children's Hospital)	
OR5	Pediatric Diabetes Virtual Visit Quality and Outcome Not Associated With Social Determinants of Health	Laurence Bastien (Children's Hospital of Eastern Ontario)	
OR6	Associations of Diabetes-Related and Health-Related Quality of Life With Glycemic Levels in Adolescents With Type 1 Diabetes Preparing to Transition to Adult Care	Simon Lafontaine (McGill University Health Centre)	
1530	Break & Exhibits		
	Symposium III: Diabetes Chairs: Manpreet Doulla (Edmonton), Daphne Yau (Saskatoon)		
1600	Cardiovascular Complications in Youth with Type 1 Diabetes	Mélanie Henderson	
1625	Complications in Youth With Type 2 Diabetes: The Widening Circle	Elizabeth Sellers	
1650	Treatment Approaches in Youth with Type 2 Diabetes	Phil Zeitler	
1730	Adjourn for the Day		
1800	ININ HE TOY 2 OTINK 2ND C2CH2L DINNOY 2T THE PHI	15 Rue Saint-Paul O, Montréal nute walk from Le Westin hotel Dinner will start at 7:00 PM	

CPEG Program Saturday, February 11, 2023 (Room: Fortifications)

Each presentation will include a 25% (minimum) of interactivity comprised of audience response questions (polling) and audience submitted Q&A via Slido.com.

0730	On-Site Registration & Breakfast			
0830	CPEG Business Meeting [members only]			
1030	Break & Exhibits			
	Symposium IV: Cushing's Syndrome Chairs: Paola Luca (Calgary), Katie Pundyk (Winnipeg)			
1100	Cushing's Syndrome: Case 1	Despoina Manousaki		
1110	Cushing's Syndrome: Case 2	Lyne Chiniara		
1120	Cushing's Syndrome: Case 3	Cheri Deal		
1130	Approach to Cushing's Syndrome and Review of Cases	André Lacroix		
1215	Panel Q&A			
1230	Saturday Poster Highlights (Even numbered Posters)			
1245	Lunch & Exhibits			
1315	Poster Viewing II (Even numbered Posters)			
P2	Adrenal Insufficiency in the Pediatric Emergency Department	Marian Thorpe (University of Alberta)		
P4	Delayed Puberty and Hypogonadotropic Hypogonadism Caused by 17-Beta-Hydroxysteroid Dehydrogenase Type 3 Deficiency	Gonzalo Alfonso Dominguez Menendez (BC Children's Hospital)		
P6	Evaluation of the First Two Years of Treatment of Children With Congenital Hypothyroidism Identified Through the Alberta Newborn Screening Program	Elizabeth Rosolowsky (University of Alberta)		
P8	Hypocalcemia After Total Thyroidectomy: It May Not Be What You Think	Rachel Parker (The Hospital for Sick Children)		
P10	Hypoglycemia During Treatment of Acute Lymphoblastic Leukemia – A Canadian Pediatric Surveillance Program Study Protocol	Mary Jiang (Children's Hospital of Eastern Ontario)		
P12	A Pediatric Presentation of a Composite Pheochromocytoma	Samantha Nordlund (McMaster Children's Hospital)		
P14	A Rare Case of Hypercalcemia of Malignancy of Unknown Etiology	Samantha Gerber (Alberta Children's Hospital)		

P16	Invasive Macroprolactinomas in Two Adolescents: Hypogonadotropic Hypogonadism Varies in Severity and Persistence	Geneviève Nadeau (CHU Sainte-Justine)
P18	A Case of NROB1-Related Adrenal Hypoplasia Congenita: A Challenging Diagnosis Mimicking Isolated Hypoaldosteronism	Duha Hejla (BC Children's Hospital)
P20	Primary Ovarian Insufficiency in Coffin-Siris Syndrome Type 8 With a Novel SMARCC2 Variant: A Case Report	Fahd Alshammri (BC Children's Hospital)
P22	A Unique Case of Hypophosphatemic Rickets	Jacqueline Mendes (The Hospital for Sick Children)
P24	It Takes Two to Tango: A Case of Hyperglycemia in Multiple Endocrine Neoplasia Type 1	Sruthi Thomas (The Hospital for Sick Children)
P26	Diabetes Duration and Glycemic Control in Adolescents With Type 1 Diabetes: A Cross-sectional Study	Olivier Renaud-Charest (Research Institute of the McGill University Health Centre)
P28	Diabetic Ketoacidosis Associated With Severe Hypertriglyceridemia and Acute Pancreatitis in Type 1 Diabetes: 4 Pediatric Cases	Sara Moline (CHU Sainte- Justine)
P30	Gaps in Type 1 Diabetes Care After Transfer From Pediatric to Adult Care and Longitudinal Associations With Psychosocial Factors	Simon Lafontaine (McGill University Health Centre)
P32	Effect of COVID-19 on Glycemic Control in Children Living With Type 1 Diabetes Mellitus	Emma Metivier (Children's Hospital, London Health Science Centre)
P34	Bone Health in Adolescents With Type 2 Diabetes	Poonam Jariwala (Jim Pattison Children's Hospital)
P36	Evaluation of the Impact of Attending Camp Banting (for Children and Youth With Diabetes) on Diabetes Distress, Quality of Life and Mental Health	Carly Baxter (Children's Hospital of Eastern Ontario (CHEO))
1415	Oral Abstracts II Chairs: Julia Sorbara (Toronto), Alexandra Ahmet (Ottawa)	
OR7	Association of Diabetes Knowledge and Self-efficacy With Stigma in Adolescents With T1D Before Transitioning to Adult Care	Nour Almutairi (Montreal Children's Hospital)
OR8	Outcomes of a Novel Subcutaneous Insulin Protocol for Management of Diabetic Ketoacidosis	Tracey Dyer (Montreal Children's Hospital)
OR9	Gonadal and Pituitary Outcomes of Young Children With Intracranial Tumors Treated With Radiation Sparing Therapy	Sarah Riedlinger (BC Children's Hospital)
OR10	Medication Induced Diabetes During Induction Therapy in Pediatric Acute Lymphoblastic Leukemia: Impact of Risk Grouping	Katie Ross (IWK Health)

OR11	Dried Blood Spot (DBS) Test for HbA1c Measurement in Pediatric Diabetes Care in Saskatchewan	Mallory McNiven (Jim Pattison Children's Hospital)
OR12	Knowledge of Time in Range and Time Below Range Among Pediatric Patients Living With Type 1 Diabetes (PLWD) and Their Caregivers	Ashlee Yang (Stollery Children's Hospital)
1545	Break & Exhibits	
	Debate	
	Chair: Jill Hamilton (Toronto)	
1615	Chair: Jill Hamilton (Toronto) Debate: Be It Resolved That Long Term Antithyroid Medication Should Be Used Instead of Radioactive Iodine in the Treatment of Youth With Autoimmune Hyperthyroidism (Graves' Disease)	Pro: Elizabeth Rosolowsky Con: Preetha Krishnamoorthy
1615 1715	Debate: Be It Resolved That Long Term Antithyroid Medication Should Be Used Instead of Radioactive Iodine in the Treatment of	Con: Preetha Krishnamoorthy

CPEN Program Friday February 10, 2023 (Room: St-Antoine B)

	CPEN Symposium Chair: Wendy Schwarz (Calgary), Caroline Boucher (Montreal)	
1400	Endocrine Management of the Transgender Youth	Karine Khatchadourian
1445	Journey from Youth to Adult	Liam Rodrigues
1530	Break & Exhibits	
1600	Gender Affirming Surgery at GrS Montreal: Process and Surgical Pathway	Evelyne Sheitoyan, Carla Geambasu
1730	Rejoin CPEG Group	

CPEN Program Saturday, February 11, 2023 (Room: St-Antoine B)

1415	CPEN Business Meeting [members only]
1545	Rejoin CPEG Group

Oral Abstracts

OR₁

Nature versus nurture of the puberty: a combined clinical and polygenic risk score to predict pubertal timing in girls

Mojgan Yazdanpanah (1), Nahid Yazdanpanah (1), Sara Moline (2), Ken Ong (3,4), John Perry (3) and Despoina Manousaki (1, 2, 5)

Research Center of Sainte-Justine University Hospital, University of Montreal, Montreal, Quebec, Canada

Department of Pediatrics, University of Montreal, Montreal, Quebec, Canada

MRC Epidemiology Unit, University of Cambridge School of Clinical Medicine, Institute of Metabolic Science, Cambridge Biomedical Campus, Cambridge, UK

Department of Paediatrics, University of Cambridge, Cambridge, UK

Departments of Pediatrics, Biochemistry and Molecular Medicine, University of Montreal, Montreal, Quebec, Canada

Context: Precocious or late puberty are common causes of referral to pediatric endocrinology. Most youth presenting such pubertal variations do not present underlying endocrine, metabolic, neurologic or neurosurgical conditions and the cause of their extreme pubertal timing is defined as "idiopathic". However, these children undergo extensive work-up, which causes significant distress and societal cost. Identifying which children with precocious or late puberty lay in the extremes of a normal distribution for pubertal timing could help avoid unnecessary investigations. Pubertal traits are highly heritable. Genome-wide association studies (GWAS) have identified hundreds of variants associated with age at menarche (AAM), while epidemiological risk factors have also been linked to pubertal timing.

Objective: To examine if a polygenic risk score (PRS) for AAM combined with clinical risk factors associate with AAM or age at peak height velocity (APHV) and predict early or late menarche in girls.

Exposures: We tested regression models using as exposure an AAM PRS by Privé et al. derived from a GWAS in UK Biobank, adjusting for clinical variables (BMI, maternal AAM, gestational age, maternal education, parental BMIs) in 3,140 girls from the Avon Longitudinal Study of Parents and Children (ALSPAC). We calculated areas under the receiver-operating curve (AUROC) for early and late AAM for the clinical and combined predictors.

Outcomes: Early menarche was defined as AAM \leq 10.4y and late menarche as AAM \geq 15y (2SD around the population-specific mean). APHV was evaluated by serial height measurements between ages 8y and 20y.

Results: In the univariate models, the standardized PRS strongly associated with AAM (beta 0.35y; Pvalue 1.27x10-68; R² 0.09) and APHV (beta 0.26y; Pvalue 1.72x10-58; R² 0.10). In the adjusted models, the association of the PRS persisted for both outcomes (betaAAM 0.30y; Pvalue 8.58-5; R² 0.24; betaAPHV 0.19y; Pvalue 2.44x10-5; R² 0.24). The AUROC of the combined predictor was 0.73 (Pvalue 1.2x10-5) for early menarche and 0.86 (Pvalue2.83x10-8) for late menarche, compared to 0.71 and 0.79 respectively of the clinical predictor alone.

Conclusion: A combined polygenic and clinical risk score could identify girls at risk of extreme pubertal timing and in whom further testing is unnecessary.

OR₂

Mutations in the WFS1 gene as a cause of non-syndromic diabetes

Alix Vanpoperinghe(1,2), Natalija Popovic (1), Angeliki Makri (1), Constantin Polychronakos (1,2)

Endocrine Genetics Laboratory, Child Health and Human Development, Research Program of the RI-MUHC Department of Human Genetics, McGill University

Background: Type 1 diabetes (T1D) is due to the autoimmune destruction of insulin-producing pancreatic beta cells. However, there is a group of non- autoimmune monogenic forms of the disease, like maturity-onset diabetes of the young (MODY), which can lead to hyperglycemia, often misdiagnosed as T1D. Correct diagnosis is essential as some forms can be treated with oral medication instead of insulin. The current inheritance model is exclusively autosomal dominant and includes 14 validated MODY genes (OMIM #606391). In addition, rare cases of monogenic diabetes can be syndromic and/or neonatal. We hypothesize that non- syndromic, non-neonatal forms of diabetes exist that have not been discovered because of lack of compelling family history. Such lack can be due either to recessive inheritance (diabetes in only ¼ of siblings, not in parents or extended family) or very low penetrance.

Approach: As part of our cross-Canada ADDAM project (Accurate Diagnosis in Diabetes for Appropriate Management, clinical trial NCT03988764), we sequenced the exomes of the first 192 autoantibody-negative patients with a clinical diagnosis of T1D. Results: In addition to 8 subjects who carried pathogenic or potentially pathogenic mutations in the known MODY genes, we found 5 biallelic (homozygous or compound heterozygous) WFS1 variants in patients with no evidence of Wolfram syndrome, confirming our previously published finding of a non-syndromic, recessive WFS1 MODY phenotype, as frequent as MODY3 (the most common MODY). More interestingly, we also found seven high-scoring heterozygous WFS1 variants which, by mutation-burden analysis, are highly unlikely to occur by chance alone (p-value = 0.014), two with affected siblings but none with affected parents.

Conclusion: We confirm our previous finding of a recessive MODY phenotype due to non-syndromic WFS1 mutations, More interestingly, we generate evidence of WFS1 mutations causing dominant diabetes, with penetrance too low to generate dominant family history. Both observations constitute proof of principle for our hypothesis, and we are currently scrutinizing the exomes of negative cases for additional novel genes. We are also testing the hypothesis that the low penetrance of the dominant form is due to digenic inheritance and we are evaluating possible candidates for the second gene that needs to be mutated.

OR₃

Usability and Feasibility Testing of Eating for Wellness (E4W): An Image- Based Dietary Assessment App for Adolescents with Obesity

Krista Oei (1,2), Elizabeth Choi (3), Alisa Bar-Dayan (4), Jennifer Stinson (5,6), Mark Palmert (1,2,5), Jeffrey Alfonsi (3,7), Jill Hamilton (1,2,5)

(1) Division of Endocrinology, The Hospital for Sick Children, Toronto, ON. (2) Department of Pediatrics, University of Toronto, Toronto, ON. (3) Research Department, RxFood Corporation, Toronto, ON. (4) Clinical Dietetics, The Hospital for Sick Children, Toronto, ON. (5) Research Institute, The Hospital for Sick Children, Toronto, ON. (6) Lawrence S. Bloomberg Faculty of Nursing, University of Toronto, Toronto, ON. (7) Department of Medicine, University of Toronto, Toronto, ON.

Introduction: Adolescence is a period of increased susceptibility for developing obesity-related health issues due to poor eating patterns and increased sedentary behaviours. Recommendations for pediatric obesity management include dietary assessments. However, adolescents often avoid food logging through traditional methods. E4W is a mobile app that determines the nutritional content of meals from photos and incorporates nutritional goal-setting. Nutritional data is transmitted to the dietitian.

Objectives: To determine usability, feasibility and preliminary effectiveness of E4W in assessing and improving dietary intake in adolescents with obesity.

Phase 2: To assess feasibility and preliminary effectiveness of E4W, a pilot randomized controlled trial of 32 patients (16 E4W, 16 controls) is being completed over one month. Both groups met with their dietitian at baseline, halfway and one month following their baseline visit to discuss goals and eating patterns. Primary outcome is the feasibility of implementation. Secondary outcomes will examine overall change in dietary intake and achievement of nutritional goals.

Results: Interview and acceptability results from usability tests were positive. Patients found the app easy to use and expressed a desire to continue using it. The most-liked feature was the ability to identify food items, portion sizes and nutritional content. Usability tests revealed some issues which were resolved. No critical issues were identified in the final cycle.

Conclusion: Results from usability testing of E4W are promising with high acceptance, supporting its use in adolescent weight management programs. Further testing of feasibility and preliminary effectiveness in improving dietary intake in adolescents with obesity is currently underway with results expected before the conference.

OR4

Factors associated with the development of dyslipidemia among pediatric patients with diabetes: A single center-based study

Fahd Alshammri(1), Hannah Geddie(1), Noor Sawalha(1), Gloria Kim(1), Karen McAssey(1), Katherine Morrison(1)

Department of Pediatrics, Division of Endocrinology, McMaster University, Hamilton, ON.

Background: The prevalence of pediatric diabetes is increasing. Dyslipidemia is an important modifiable cardiovascular disease (CVD) risk factor often presents in children with diabetes. Diabetes Canada (DC) clinical practice guidelines recommend screening for dyslipidemia in youth with type 1 diabetes (T1DM) beginning at 12 years of age or at diagnosis in youth with type 2 diabetes (T2DM).

Objectives: To determine the prevalence of dyslipidemia in youth with diabetes and identify risk factors related to dyslipidemia. Study Design and Methods: This retrospective chart review included patients at McMaster Children's Hospital with T1DM or T2DM, who were 12 years of age or older as of January 1st, 2019, and had been diagnosed with diabetes for at least 6 months. Extracted data included age, sex, family history of diabetes or dyslipidemia, date of diagnosis, body mass index (BMI), type of glycemic monitoring system used, lipid profile, A1C, and TSH values. Statistical methods included descriptive statistics and logistic regression modeling.

Results: Of the 275 patients included, 45% (n=125) met criteria for dyslipidemia. The most common type of dyslipidemia was hypertriglyceridemia (35%; n=95), followed by elevated non-high density lipoprotein cholesterol (non-HDL-C) (20%; n=54), low HDL-C (12%; n=32), and elevated low-density lipoprotein cholesterol (LDL-C) (10%; n=27). The prevalence of dyslipidemia was highest amongst those with T2DM, older age, short duration of diabetes, higher A1C, obesity, and use of finger stick for monitoring (p<0.05).

Conclusion: Dyslipidemia, mainly hypertriglyceridemia, is highly prevalent in our population. Dyslipidemia was associated with obesity in this patient population, though 44% of patients without obesity also had dyslipidemia. While dyslipidemia in this population is expected, our findings suggest the need for improved recognition of this condition given the lifetime risk of CVD in these patients.

OR₅

Pediatric diabetes virtual visit quality and outcome not associated with social determinants of health

Laurence Bastien (1), Ellen Goldboom (1,2,3), Ewa Sucha (3), Richard Webster (3), IvanTerekov (3), Caroline Zuijdwijk (1,2,3)

Department of Pediatrics, Faculty of Medicine, University of Ottawa, Ottawa, ON Division of Endocrinology and Metabolism, Children's Hospital of Eastern Ontario, Ottawa, ON Children's Hospital of Eastern Ontario Research Institute, Ottawa, ON

Objectives: Social determinants of health (SDH) impact diabetes short- and long- term outcomes. In response to COVID-19, virtual care has become a solution to ensure ongoing healthcare access. However, socially disadvantaged groups have less access and skills related to technology, leading to potential increases in health disparities. We aimed to determine the association between SDH and physician- perceived virtual visit quality and outcome in pediatric diabetes patients.

Methods: Retrospective chart review of patients who attended a scheduled virtual diabetes follow-up visit at a pediatric tertiary care centre from December 1, 2020 to March 31, 2021. During this time, a quality improvement study required physicians to rate the quality (same/better or worse than in-person) and outcome (successfully replaced in-person visit or not) of each virtual visit. These data, along with patient characteristics, were extracted from the electronic health record. Institut national de santé public du Québec Material and Social Deprivation Index and the Ontario Marginalization Index (Ethnic Concentration) were used to determine SDH based on postal code. Statistical analysis tested for an association between deprivation index quintiles and virtual visit quality and outcome.

Results: Data were obtained for 447 patients with mean age 12.7±3.8 years; 47.7% female; 93.1% type 1 diabetes; and mean glucose management indicator 7.96±1.41%. During the study period, 17.9% visits were evaluated as worse than in person, 13.1% visits were unsuccessful in replacing an in-person visit, and 20.7% were rated worse or unsuccessful. The odds of having a worse or unsuccessful visit were not different in those with the highest vs. lowest degree of material deprivation (odds ratio [OR] 1.59, 95% confidence interval [CI] 0.53, 4.76), social deprivation (OR 1.36, 95%CI 0.48, 3.83), or ethnic concentration (OR 0.30, 95%CI 0.30, 1.77).

Conclusion: In our pediatric diabetes population, virtual visit quality and outcome were not associated with material or social deprivation. These results show promising success in equitable access and delivery of high quality virtual diabetes care for patients regardless of socioeconomic status, which is important in the context of the current pandemic and sustainable widespread integration of virtual care. Further studies are required to assess this association in other populations.

OR₆

Associations of Diabetes-Related and Health-Related Quality of Life with Glycemic Levels in Adolescents with Type 1 Diabetes Preparing to Transition to Adult Care

Simon Lafontaine (1), Elise Mok, PhD (2), Jennifer Frei, BA (2), Mélanie Henderson, MD, PhD (3), Elham Rahme, PhD (2), Kaberi Dasgupta, MD, MSc (2), Meranda Nakhla, MD, MSc (1,2)

Department of Pediatrics, Division of Endocrinology, McGill University Health Centre, Montreal, QC Research Institute of the McGill University Health Centre, 5252 de Maisonneuve Ouest, Montreal, QC Department of Pediatrics, Université de Montréal, Montreal, QC. Centre de Recherche CHU Sainte-Justine, Montreal, QC. School of Public Health, Department of Social and Preventive Medicine, Université de Montréal, Montreal, QC.

Aims: As adolescents with type 1 diabetes (T1D) progress to adulthood they assume responsibility for diabetes self-management while dealing with competing life demands, decreasing parental support, and the transfer to adult care. Lower perceived quality of life (QOL) may hamper diabetes management which is associated with suboptimal glycemic levels. Our objective was to determine associations of diabetes-related and health-related QOL with glycemic control (hemoglobin A1c (A1C)) in adolescents with T1D

before their transfer to adult care. Methods: We conducted a cross-sectional analysis of baseline data from the Group Education Trial to Improve Transition (GET-IT) in adolescents with T1D (ages 16-17 years). Participants completed validated questionnaires measuring diabetes-related (PedsQL 3.2 Diabetes Module) and health-related QOL (PedsQL 4.0 Generic Core Scales). Associations of QOL Total and Subscale Scores with A1C were assessed using linear regression models adjusted for sex, diabetes duration, socioeconomic status, insulin pump use and mental health comorbidity.

Results: 153 adolescents with T1D were included (mean (standard deviation) age: 16.5 (0.3) years). Diabetes-related QOL Total Scores (adjusted β -0.036; 95% Confidence Interval (CI) -0.05, -0.02) as well as the subscale scores for diabetes symptoms (adjusted β -0.021; 95%CI -0.04, -0.005) and diabetes management (adjusted β -0.035; 95%CI -0.05, -0.02) were inversely associated with A1C. Health- related QOL Total Scores were not associated with A1C, but Psychosocial Health Subscale Scores were (adjusted β -0.015; 95%CI -0.03, -0.0002).

Conclusions: Our results suggest that strategies focusing on diabetes related QOL are important and may help prepare adolescents for the increasing responsibility of diabetes self-management.

OR7

Association of Diabetes Knowledge and Self-Efficacy with Stigma in Adolescents with T1D Before Transitioning to Adult Care

Meranda Nakhla, Elise Mok, Nour Almutairi

Pediatrics, Endocrinology, McGill University Health Centre, Montreal, Québec, Canada.

Centre for Outcomes Research & Evaluation, Research Institute of the McGill University Health Centre, Montreal, Québec, Canada.

BACKGROUND/RATIONALE: Type 1 Diabetes (T1D) is one of the most common chronic diseases in childhood and is associated with significant morbidity and mortality. T1D is a chronic illness that requires a multidisciplinary team and intensive self-management to achieve optimal glycemic control., Approximately 2/3 of adolescents with T1D have diabetes-related stigma which is associated with suboptimal glycemic control and poorer quality of life. The aim of our current study was to explore perception of diabetes-related stigma among adolescents with T1D before transfer to adult care. Specifically, we assessed the associations of diabetes knowledge and self-efficacy with stigma. Results may identify potential mitigating strategies to reduce diabetes-related stigma and enhance coping skills amongst young adults with diabetes.

OBJECTIVES: 1) To describe stigma in adolescents with T1D before transition to adult care. 2) To determine if diabetes knowledge and self-efficacy are associated with stigma in the same population.

METHODS: We conducted a cross-sectional study of T1D adolescents (ages 16-17 years) receiving care at a pediatric diabetes clinic at two academic hospitals in Montreal, Canada. We conducted a secondary analysis of baseline data from a subsample of adolescents enrolled in The Group Education Trial to Improve Transition in Adolescents with Type 1 Diabetes (GET-IT-T1D). We used laboratory results and participant characteristics collected from medical records and validated self-reported questionnaires assessing stigma (defined as an affirmative response to at least one of 3 key items on the Barriers to Diabetes Adherence [BDA] stigma subscale in Adolescence), diabetes knowledge (L'Aide aux Jeunes Diabétiques Diabetes Knowledge and Skills [AJD DKS], score 0-50), and self-efficacy (Self- Efficacy for Diabetes Self-Management Measure [SEDM], score 1-10). We used logistic regression models to analyze the associations of diabetes knowledge and self-efficacy with stigma adjusted for HbA1c, diabetes duration, sex, mental health comorbidities and socioeconomic status.

RESULTS: Of 198 adolescents with T1D, 125 (63%) self-reported having diabetes-related stigma. Self-efficacy in diabetes management was associated with lower stigma, Odds Ratio (OR): 0.60, (95% confidence interval (95% CI) 0.47-0.76). Diabetes knowledge was not associated with stigma, OR: 0.98, (95% Cl 0.93-1.03).

OR8

Outcomes of a Novel Subcutaneous Insulin Protocol for Management of Diabetic Ketoacidosis

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Background: Intravenous (IV) insulin infusion has been the treatment of choice for diabetic ketoacidosis (DKA) but is both cost and resource-intensive. Subcutaneous (SC) insulin administration is a potentially feasible, safe, and effective alternative in the management of uncomplicated mild and moderate DKA. It can decrease resource utilization such as intensive care unit (ICU) beds. Pediatric studies on outcomes of SC insulin in the treatment of DKA are scarce.

Objectives: To evaluate a novel DKA protocol introducing SC insulin for the management of pediatric mild and moderate DKA.

Methodology: Retrospective chart review of patients 0-18 years old presenting to the Montreal Children's Hospital (MCH) Emergency Department (ED) in DKA during the first year of implementation of a novel protocol that uses SC insulin aspart or lispro 0.15 U/kg SC every 2 hours. Primary outcome was time to DKA resolution (pH \geq 7.3) from detection of DKA (time of initial gas result). Secondary outcomes were time to insulin administration from DKA detection, hospital admission, and safety (hypoglycemia, hypokalemia, hypophosphatemia, DKA recurrence, acute kidney injury (AKI), cerebral edema, and mortality). Demographic, laboratory and clinical data were extracted from medical records.

Results: Between September 1, 2021 and August 31, 2022, 30 patients presented to the MCH ED in mild (n=9) or moderate (n=21) DKA. 26 of these patients were treated with the SC insulin protocol; 2 were not due to young age, and 2 due to borderline mild DKA. Median time to insulin administration was 99 minutes (IQR 70, 123). Median time to DKA resolution was 8h58min (IQR 6h03, 11h07). Twenty-five of the 26 patients avoided an ICU admission, as they were treated in the ED (n=12) or wards (n=13). 15.4% of patients had hypokalemia and 7.7% had hypophosphatemia. No severe safety events occurred including no DKA recurrence, AKI, cerebral edema, severe hypoglycemia, or mortality.

Conclusions: This innovative SC protocol for the treatment of mild and moderate DKA in children was effective and safe, with minimal complications and reduction in ICU admissions. Further research comparing our outcomes to a historic control using IV insulin is ongoing.

OR9

Gonadal and pituitary outcomes of young children with intracranial tumors treated with radiation sparing therapy

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BACKGROUND: Cranial radiation therapy (RT) is known to have detrimental effects to the developing brain. Therefore, a RT-sparing approach with higher doses of alkylating agents (AA) has been used in young children with intracranial tumors at BC Children's Hospital (BCCH). While primary gonadal failure (PGF) is a known complication of alkylating agents, the risk of PGF in the population treated with an RT-sparing approach remains unclear.

OBJECTIVE: The primary objective is to describe the incidence and associated risk factors of PGF in children with an intracranial tumor treated with an initial RT- sparing approach. The secondary objective is to determine the incidence of additional endocrinopathies.

METHODS: This is a retrospective cohort study of children 0-8 years of age, who presented to BCCH between 1998 – 2018 and were treated for an intracranial tumor using a radiation-sparing approach. Data collection included demographics, cancer diagnosis and treatment, endocrine diagnoses and treatment, as well as endocrine laboratory values. Kaplan Meier survival curves, regression analysis, and T-tests were used in data analysis.

RESULTS: Eighteen patients met inclusion criteria; 8/18 (44 %) developed PGF, and one patient later recovered gonadal function. Of the 18 patients, 14 remained RT- naïve throughout the entire duration of follow-up; 5/14 (36 %) RT-naïve patients developed PGF. The probability of developing PGF at 5 years, and 10 years after diagnosis is 11% and 31%, respectively (Figure 1). Compared to the non-PGF group, the PGF group has a higher CED dose (45.2 vs. 56.1 g/m2, p = 0.02) but also has a longer duration of follow up (p = 0.01). Of the 8 patients with PGF, 6 developed additional endocrinopathies. The 10 patients without PGF did not develop other endocrinopathies.

CONCLUSIONS: High CED is a risk factor for the development of PGF in patients treated for brain tumors with a RT-sparing approach. Within this cohort, additional endocrinopathies are uncommon in patients who do not receive cranial RT and do not develop PGF. Correlation between CED dose and time to onset of PGF requires further study.

OR10

Medication induced diabetes during induction therapy in pediatric acute lymphoblastic leukemia: Impact of risk grouping

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Methods: We performed a retrospective single-center study of 262 patients (142 males, 120 females) diagnosed with ALL at the IWK Health Centre from 2000 to 2019. Demographic and treatment data was extracted from the Pediatric Oncology Research Database and EMRs.

Results: Twenty-two patients developed MID (8.4%) and more than two-thirds (68.2%) required treatment with insulin. Patients with MID were significantly older (10.3 vs 6.2 years, p < 0.001), had higher BMI z-scores (1.2 vs 0.3, p=0.003), and had higher rates of Trisomy 21 (9.5% vs 1.3%, p=0.012). Patients with MID had significantly higher rates of CNS disease (36.4% vs 14.2%, p=0.007) but did not have increased rates of infection, relapsed disease, or death. HR patients (n=122) had significantly more complications than SR patients (n=140) including MID (13.1% vs 4.3%, p=0.01), CNS disease (23.8% vs 9.3%, p=0.001), infection (68.3% vs 45.7%, p<0.001), relapsed disease (10.7% vs 4.3%, p=0.047), and death (11.5% vs 1.4%,<0.001). HR patients treated with 28-days of prednisone developed significantly more MID than those treated with 14-28 days of dexamethasone (21.5% vs 3.5%, p=0.003) and were significantly older (12.7 vs 4.2 years, p<0.001).

Conclusion: Older age, higher BMI, CNS disease, Trisomy 21, and steroid type were risk factors associated with MID in our cohort. HR patients developed significantly more complications including MID. As hyperglycemia can be easily identified through routine screening, protocols that consider individual patient risk factors and treatment regimens should be developed and implemented, particularly for HR leukemia patients receiving prednisone.

OR11

Dried Blood Spot (DBS) Test for HbA1c Measurement in Pediatric Diabetes Care in Saskatchewan

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Background: Glycated hemoglobin (HbA1c) is a basis for diagnosis, monitoring, and screening of pediatric diabetes. However, the frequency of HbA1c testing, remote laboratory access, needle phobia, and, most recently, COVID19 laboratory restrictions, all impede timely, consistent access to HbA1c testing. The dried blood spot (DBS) card is a novel method for measuring HbA1c, allowing patients to collect small volumes of blood through a self-initiated, at-home capillary sample. DBS cards for HbA1c measurement have been validated in the adult population, but there is an absence of pediatric data.

Aim: This study's aim is to validate the use of DBS cards in measurement of HbA1c in comparison to the standard venous approach and to identify barriers to implementing this method provincially.

Methods: Venous and DBS card samples were collected simultaneously from 62 patients. Venous samples were collected as per protocol. DBS samples were collected by patients upon presentation to their laboratory, time stamped, and mailed between the local and provincial laboratory for single-site DBS card analysis and reporting. Correlation analyses were conducted to assess inter-assay agreement by Pearson correlation coefficient and Bland-Altman plot.

Results: 55 of 62 paired samples were analyzed (exclusions for elevated hemoglobin F, insufficient sample, and unavailable sample). Mean venous HbA1c was 7.49%; DBS was 7.26%; inter-assay difference of 0.23%. Data shows a strong positive correlation between HbA1c collection methods (r=0.87, p<0.001). A standardized formula incorporating the ranges of both data sets was used to rescale the DBS data using a correction factor. The rescaled data showed an even stronger correlation between the two methods (r=0.8935, p<0.001). Analysis of time to DBS processing and effect of HbA1c variation on inter-assay agreement is forthcoming.

Discussion: Our study demonstrates a strong inter-assay agreement between DBS and venous HbA1c measurements. Preliminary data suggests this is further strengthened with lower HbA1c values. Given the DBS feasibility, cost effectiveness, and performance characteristics at a lower HbA1c, this study provides support for DBS testing in pediatric diabetes screening under the right conditions. Our next phase is to assess the DBS HbA1c test as a screening tool for type 2 diabetes in remote and underserved populations.

OR12

Knowledge of Time In Range and Time Below Range among pediatric patients living with type 1 diabetes (PLWD) and their caregivers

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Objective: Continuous Glucose Monitoring (CGM) metrics, including Time In Range (TIR) and Time Below Range (TBR), provide insight into glycemic variability and correlate with long-term outcomes. We aimed to explore knowledge of TIR and TBR among pediatric patients living with type 1 diabetes (PLWD) and their caregivers.

Methods: Participants completed a cross-sectional questionnaire. Questionnaire items were informed by the literature, and reviewed by content experts, patient advisors, and a survey methodologist. The questionnaire was piloted for accessibility, comprehensibility, and validity.

Results: 216 (77% caregivers; 23% patients) participants took part in the study from January -June 2022. The top 3 main reasons for CGM use identified by respondents were to improve glycemic control, reduce fingerstick testing, and reduce hypoglycemia. Among those who reported knowing TIR and TBR, a third (34%) and more than three-quarters (89%) correctly defined TIR and TBR, respectively. While two-thirds (68%) were able to identify the recommended TIR for most pediatric PLWD, less than a quarter (19%) were able to identify the recommended TBR. Among those who reported knowing the differences between CGM metrics and hemoglobin A1c (HbA1c), three-quarters (75%) knew that a key advantage of CGM metrics over HbA1c is the insight on glucose variability. However, only a quarter (25%) understood that both HbA1c and TIR correlate with long-term outcomes. Better knowledge was observed among those who review CGM reports frequently and those who use CGM mainly to improve glycemic control.

Conclusion: Our study identified large knowledge gaps among pediatric PLWD and their caregivers in the definitions, recommended targets, and long-term impacts of TIR and TBR. Such knowledge gaps can be narrowed by gearing education to improve understanding and creating patient-centered educational tools.

Poster Abstracts

P1

Pediatric Diabetes: An Exploration of Shared Care in Nova Scotia

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Introduction: Care for pediatric type 1 diabetes (T1D) in Nova Scotia (NS) occurs at one tertiary care pediatric centre in Halifax (central zone) and several regional diabetes centres (DC) serving all ages. Ideally, children receive diabetes care close to home, however, the increasing complexity of diabetes care means shared care between tertiary and regional centres is often sought. We aimed to assess patterns in the provision of pediatric T1D care throughout NS and to understand the perspectives of DC staff.

Methods: The Diabetes Care Program of NS registry contains records for all DC visits in NS. Patients living outside of the central zone at diagnosis, aged <15 years with T1D diagnosed between 2007-2016 and 2+ years of follow-up were classified as receiving shared, tertiary-only, or regional-only care. Shared care was defined as having regular or intermittent visits at both the tertiary and a regional DC. Semi- structured qualitative interviews were conducted with 20 staff and physicians working in regional and tertiary DCs to understand facilitators and challenges in shared care. Interviews were transcribed, coded, and analyzed for themes.

Results: Of 267 patients living outside central zone, 20% received tertiary care only, 37% regional only, and 31% received on-going shared care with rates varying by region. Twelve percent saw the tertiary centre only at initial diagnosis. Children <5 years old were more likely to have some shared care compared to older children. Two overarching themes from interviews were: communication and resources. Communication concerns revolved around variable communication between regional and tertiary DCs as well as intra-centre communication between physicians and diabetes educators. Resource needs included mental health/social work support, after hours phone support, and dedicated time for continuing professional development in pediatrics.

Conclusion: Four in 10 patients living in predominantly rural NS received some degree of shared care. Our findings highlight the need for formalized shared care processes. This could be supported with standardized forms on a province-wide electronic health record to enhance consistency of information collection and sharing across providers. Provincial support for regional DCs is needed to facilitate both continuing professional development and province-wide psychosocial supports with equitable access.

P2

Adrenal Insufficiency in the Pediatric Emergency Department

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Background: Children with adrenal insufficiency (AI) are at risk of life-threatening adrenal crisis without adequate steroid treatment during illness. Our center has introduced a clinical decision support (CDS) tool in the electronic health record (EHR) which alerts providers to the diagnosis and need for emergency treatment. The aim of this study is to determine how promptly children with AI received parenteral steroids when they visited the emergency department (ED) prior to the CDS, and whether there is any improvement after its introduction.

Results: There were 217 retrospective and 31 prospective encounters. Home hydrocortisone injection was used prior to 3% of all encounters. Prior to the CDS, parenteral steroids were given during 59 (27%) of encounters, and median time to steroid was 1.8 hours (SD 1.1-3.4). Comparison of outcomes before versus after the CDS did not show statistical significance, with proportion who received steroid 27% before and 32% after (p=0.529), and median time to steroid 1.8 hours before and 2.2 hours after (p=0.916).

Discussion: Although the prospective sample size was not sufficient to assess impact of the CDS currently, the study provides insight into the emergency management of pediatric AI at our tertiary center prior to CDS implementation. Among those who received parenteral steroids in the ED, the time from triage to first injection was considerably longer than published recommendations, highlighting the need for ongoing quality improvement. The study also demonstrates that despite a third of patients receiving parenteral steroids in the ED, only 3% of all patients had used their emergency hydrocortisone injection prior to arrival. These findings provide impetus for further study not only on ED management of pediatric AI, but also on barriers to appropriate home management prior to presentation.

P3

Atypical Presentation of New Onset Diabetes with Hyperosmolar Hyperglycemic State in a Toddler

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Introduction: Hyperosmolar hyperglycemic state (HHS) is an uncommon presentation of new onset diabetes in children and is most often seen in adolescents with type 2 diabetes. Here, we present a case of severe HHS with ketosis in a toddler.

Case: A 4-year-7-month-old nonverbal male with Trisomy 21, duodenal atresia status post repair, bilateral cystic kidney disease, and autism spectrum disorder presented to the emergency department with lethargy and respiratory distress. Examination was notable for tachycardia, somnolence, dry oral mucosa, and diffusely tender abdomen. Initial laboratory evaluation revealed markedly elevated serum glucose of 117mmol/L (2114mg/dL) with hypernatremia (corrected sodium 166mmol/L). Blood gas was consistent with a predominant respiratory acidosis with high anion gap metabolic acidosis (bicarbonate 19mmol/L; serum - hydroxybutyrate 4.4mmol/L). Rapid hemoglobin A1c was 10.3%. Additional laboratory results were consistent with pancreatitis and acute kidney injury. Upon more detailed history, parents noted polyuria over the preceding week and had offered ice water and diluted juice. Due to developmental delays, the patient was unable to express increased thirst.

Initial management included aggressive intravenous (IV) fluid rehydration with initial boluses of 40ml/kg followed by twice maintenance rate. To prevent rapid decreases in glucose, IV insulin infusion was started at 0.025units/kg/hr and continued for 36 hours. His kidney function and symptoms of pancreatitis slowly improved. He did not have evidence of cerebral edema, rhabdomyolysis, or deep vein thrombosis. He was extremely insulin resistant, requiring 1.7 units/kg/day of basal-bolus subcutaneous insulin on discharge. Autoantibodies including islet antigen 2, insulin, glutamic acid decarboxylase, and zinc transporter 8 were all negative. A panel for genetic causes of diabetes (MODY panel) is pending.

Discussion: This is one of very few reported cases of HHS in a toddler and highlights differences in presentation and management from more typical diabetic ketoacidosis. Developmental delay likely contributed to the severity of the initial presentation with inability to adequately increase fluid intake at home. Based on age and underlying Trisomy 21, type 1 diabetes was initially thought to be the most likely diagnosis. However, given four negative autoantibodies and underlying renal disease, genetic forms of diabetes, such as MODY5, must be considered.

Delayed Puberty and Hypogonadotropic Hypogonadism Caused by 17-beta- hydroxysteroid Dehydrogenase Type 3 Deficiency

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Introduction: The 17β-hydroxysteroid dehydrogenase type 3 enzyme (17-β-HSD3), primarily found in testis, converts androstene-dione to testosterone. 17-β-HSD3 deficiency has autosomal recessive inheritance pattern, and it is caused by defects in the HSD17B3 gene, resulting in undervirilized male newborns. Phenotype varies from typical external female genitalia to hypospadias and crypt-orchidism. In puberty, gonadotropins stimulate the secretion of androstenedione, which has low androgenic potency. Androstenedione is peripherally converted to testosterone by other hydroxysteroid dehydrogenase isoenzymes. We described a case of 17-β-HSD3 deficiency associated with delayed puberty, hypogonadotropic hypogonadism, hypospadias, and cryptorchidism.

Case: A 14-year-old male with delayed puberty and history of perineoscrotal hypospadias and unilateral cryptorchidism. The newborn period investigations showed a normal male karyotype (46,XY). The gonadotropins demonstrated low FSH, LH and testosterone; androstenedione was not measured at that time. In the genetic evaluation, no variants were present for 5-a reductase deficiency or androgen insensitivity syndrome. Chromosome microarray analysis showed a variant of unknown significance (956.6 Kb copy gain on chromosome 7 at 7p22.3-p22.2).

On physical exam, he presented with scarce pubic hair and prepubertal genitalia. Supplementation with intramuscular testosterone was recommended. He underwent appropriate development of secondary sexual characteristics, but with no testicular growth. Whole exome sequencing presented compound heterozygous changes in 17BHSD3: a pathogenic variant, c.277+A4A>Y (p.?), and a variant of uncertain significance not previously reported, c.824C>T (p.Ala275Val). Additionally, the analysis was consistent with heterozygosity for a CTLA4 variant of uncertain significance, c.160G>A (p.Ala54Thr). Between the blood draw for the exome and the return of the results, the proband developed immune thrombocytopenia and recurrent sinopulmonary infections, consistent with CTLA4 haploinsufficiency.

Discussion: $17-\beta$ -HSD3 deficiency is a rare cause of differences in sexual development in males. Severe cases present female-appearing external genitalia at birth and virilization at puberty. More infrequent mild forms exhibit undervirilized male-appearing genitalia. The conversion of androstenedione to testosterone by other $17-\beta$ -HSD isoforms or residual enzymatic activity of $17-\beta$ -HSD3 in the testis may explain pubertal progression. Our case did not have pubertal development, and gonadotropins were inhibited. High concentrations of androstenedione relative to testosterone may suggest the diagnosis $17-\beta$ -HSD3 deficiency, and it is confirmed with genetic testing.

P5

Double Diagnosis: Acetaminophen Overdose and Diabetic Ketoacidosis?

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Case Presentation: A previously healthy 16-year-old male presented to a community emergency department with nausea and emesis with a history of intentional acetaminophen overdose. He was noted to be tachycardic and hypertensive on presentation. Initial investigations revealed an elevated acetaminophen level of 724 umol/L (normal <65). He was started on N-acetylcysteine (NAC) protocol for treatment of the acetaminophen overdose. As part of his initial work up, his blood glucose was noted to be persistently elevated at 32.2 mmol/L and 35.9 mmol/L. A venous blood gas revealed metabolic acidosis (pH 7.18, bicarbonate 12.4 mmol/L, base excess -12 mmol/L, and anion gap 44 mmol/L) with hyperglycemia and ketonuria but no glucosuria. A diagnosis of diabetic ketoacidosis with presumed new onset type 1 diabetes was made. Endocrinology was consulted for guidance on fluid management given the unusual presentation. DKA protocol was initiated as per TREK guidelines. The hyperglycemia quickly resolved to 8.5 mmol/L after the initial IV fluid boluses and insulin infusion was not initiated. Subsequent blood glucose monitoring revealed blood glucoses in the normal range (4.9-9.4 mmol/L) with resolution of anion gap. Of note, liver function and lipase were normal during admission. No other co-ingestions were identified. Hemoglobin A1C was drawn at presentation and was reported at 5.1%. Our patient was admitted to the pediatric ward for the duration of treatment and had an uneventful recovery.

Discussion: We describe a pediatric case of hyperglycemia with metabolic acidosis and ketonuria, in the context of an acetaminophen overdose and absence of hepatic dysfunction. It is well known that high levels of acetaminophen can interfere with certain laboratory assays for measuring blood glucose. The adult literature also describes hyperglycemia in the setting of acetaminophen overdoses however all in the setting of liver failure. We also highlight in this case that treatment with IV fluids alone is sufficient management in such cases.

P6

Evaluation of the first two years of treatment of children with Congenital Hypothyroidism identified through the Alberta Newborn Screening Program

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Background: Congenital Hypothyroidism (CH) is the most common yet preventable cause of intellectual disability. Universal newborn screening allows for early identification of affected infants so that replacement of thyroid hormone can begin promptly. The effectiveness of the screening program relies on timely screening, confirmation of diagnosis, and initiation and ongoing monitoring of treatment, particularly during the first 2 years of age.

Objectives: The primary objective of this study is to ascertain the extent to which infants with CH have received timely and appropriate management within the first 2 years of life, following diagnosis through newborn screening (NBS) in Alberta. Methods— Deidentified laboratory data were extracted from Alberta Health administrative databases between April 1, 2014 and March 30, 2019. All confirmed cases of CH identified during this time frame were included in the initial analyses. Time to lab collection was anchored from date of birth. Timeliness was assessed as the frequency of monitoring of thyroid function indices; appropriateness as the frequency of children maintaining biochemical euthyroidism.

Results: Alberta NBS confirmed 185 cases of CH in this time frame. (Table 1). Overall, the majority of infants had confirmation of diagnosis within 10 days (median 7 days; range 1-69) of age, and 84% within 16 days. Most babies had 2 TSH measurements done in the time interval from 0 to 1 month, 4 measurements from 1 to 6 months, and 2 measurements from 6 to 12 months of age. Approximately half of the cohort were still hypothyroid at 1 month of age. Subsequent to becoming euthyroid, at least some period of hypo- (57%) or hyperthyroidism (40%) was commonly experienced.

P8

Hypocalcemia after total thyroidectomy: It may not be what you think...

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Hypocalcemia is a common complication following total thyroidectomy. Most frequently it reflects compromise of the parathyroid glands or their vascular supply. We present a unique case of post-thyroidectomy hypocalcemia due to an additional etiology, highlighting synergistic pathology and discuss potential pathophysiologic mechanisms underlying the challenging management. A normocalcemic 15-year-old male with papillary thyroid carcinoma and bulky metastatic cervical adenopathy underwent a total thyroidectomy with bilateral central and left lateral neck dissection (levels II-IV). Intra-operatively, two parathyroid glands were visualized and preserved while a third was re-implanted after concern for devascularization. There were no concerns for intra-operative complications.

Post-operatively, early and profound hypocalcemia was noted and parenteral and enteral calcium and calcitriol were initiated. On post-operative day (POD) #1, milky effusion was noted from the surgical drain, consistent with a chyle leak, and treatment with octreotide and fat-restriction were initiated. The patient continued to demonstrate extremely high requirements for calcium replacement and calcitriol through POD#9 with 4 failed attempts at discontinuing calcium gluconate infusion despite escalation of enteral supplementation.

The chyle leak resolved on POD #7 and subsequently, he was weaned to enteral calcium and discharged on POD#11. He subsequently weaned from oral calcium and calcitriol by 7 months post-operatively and remains well 15 months later. Thoracic duct (TD) injury leading to chyle leak is a rare but recognized complication of neck surgery and has been associated with prolonged and severe hypocalcemia following thyroidectomy in adults. Dense metastatic adenopathy in the left lateral neck constitutes a specific risk for TD injury. The mechanisms of hypocalcemia secondary to chyle leak are likely multifactorial, including lymphatic loss of calcium and vitamin D, and the therapeutic use of octreotide as management for chyle leak, which decreases the absorption of enteral calcium through reducing both gastrointestinal motility and acidification required for the absorption of calcium carbonate. These factors, coupled with post-surgical hypoparathyroidism, can prolong post-thyroidectomy hypocalcemia and complicate its management. In the setting of TD injury, high dose oral supplementation and prolonged IV requirements should be anticipated and rapid escalation of oral replacement should be considered.

P9

The impact of family structure of adolescents with T1D in diabetes management aspects before transition to adult care

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Objectives: Adolescence is associated with a suboptimal glycemic control and decreased adherence to care In young adults with T1D. Marital status of parents (separated/deceased parents) has been linked to suboptimal glycemic control in children. Our aim is to examine whether marital status of parents is associated with diabetes management aspects in adolescents with T1D transferring to adult care.

Methods: We conducted a cross-sectional study of adolescents (aged 16-17) with T1D followed at the MCH or the CHU Ste-Justine. Participants were asked to complete validated questionnaires on self-efficacy (SEDM, score 1-10), transition readiness (TRAC, cutoff ≥ 8), diabetes distress (T1-DDS, cutoff ≥ 3) and stigma perception (cutoff 1). The exposure was parent's marital status. We examined associations of family structure with self-efficacy, A1c, diabetes knowledge and distress, transition readiness and stigma perception using multivariate linear and logistic regression models adjusted for sex, diabetes duration, socioeconomic status, technology use and A1c.

Results: Of 202 adolescents with T1D (47% male and 53% female), 35,6% had separated, divorced or deceased parents while 64,4% did not. Non-traditional family structure was associated with lower self-efficacy (B=-0,752; 95% CI-1,22 to -0,27) and higher A1c (B= 0,540; 95% CI 0,064 to 1,017)

Conclusion: Improving health-care transition for teenagers with T1D should focus on those with non-traditional family structure to help them achieve a better transition to adult care.

P10

Hypoglycemia during treatment of acute lymphoblastic leukemia – A Canadian Pediatric Surveillance Program Study Protocol

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associated with severe hypoglycemia, especially in those under the age of 6 years.

The mechanism and temporal relationship between hypoglycemic events and paediatric ALL treatment has not been well described. In collaboration with the Public Health Agency of Canada, a national public health surveillance project was developed through the Canadian Pediatric Surveillance Program (CPSP) to understand the frequency of this rare ADR by systematically documenting biochemically proven hypoglycemia associated with ALL therapy.

Methods: Through the established methodology of the CPSP, paediatricians and paediatric subspecialists who are part of the CPSP network will be asked on a monthly basis to report on any patient less than age of 18 years (up to the 18th birthday) with a first known episode of biochemically proven hypoglycemia via laboratory serum (if not available, then point-of-care) glucose sample with blood glucose level below 3.0 mmol/L during chemotherapy for ALL. Data captured will include patient clinical characteristics, specific demographic features, treatment information, timing of onset, and duration in relation to exposure and management strategies being utilized.

Discussion: This is a 2-year surveillance study, data collection initiated on October 1, 2022. Our study will report the frequency, clinical features and management strategies associated with first known hypoglycemic events associated with ALL therapy and will help to inform the need for a screening protocol for this ADR. The development of a screening protocol would help define the scope of this issue and could help to eventually inform guideline development. CPEG members are encouraged to report patients through CPSP.

P11

Double The Trouble: Dual presentation of diabetic ketoacidosis and thyroid storm in an adolescent patient

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Background: Diabetic ketoacidosis (DKA) and thyroid storm (TS) are both endocrine emergencies with potentially serious consequences. While the two endocrine conditions are known to be associated, simultaneous presentation in the pediatric population is extremely rare and is limited to few case reports.

Case Presentation: A 14-year-old young Inuit male known to have type 1 diabetes (T1D) that was diagnosed 2 months prior, presented to his village health post with persistent vomiting and hyperglycemia. Patient is known for a difficult psychosocial situation that has resulted in difficulty with diabetes management and adherence. He was subsequently med-evacuated to the closest hospital where he was found to be in severe DKA (pH: 7.10, HCO3: 4 mmol/L, blood glucose: 19.9 mmol/L, urine ketones>7.8 mmol/L). Treatment with IV fluids and IV insulin as per local protocol was started. Transfer to our emergency department was initiated. Upon arrival, DKA management was continued with IV fluids and SC insulin following our institution's protocol. Despite near resolution of the DKA over the course of 22 hours, patient remained symptomatic with an unusual constellation of findings including fever 38.1°C, altered level of consciousness (GCS 14/15), prominent abdominal pain, and cardiovascular dysfunction (tachycardia up to 160 bpm, with BP: 146/111).

Conclusion: Simultaneous presentation of DKA and TS in pediatric patients with a prior history of T1D is rare but may arise. Prompt diagnosis and management are needed given the high risk of mortality and morbidity. Both conditions have similar predispositions and either condition may trigger the other. Increased awareness of the possibility for copresentation is important.

P12

A Pediatric Presentation of a Composite Pheochromocytoma

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Composite pheochromocytomas are rare tumours arising from the adrenal medulla, consisting of both neuroendocrine and neural components. Neural tumours (i.e. neuroblastoma) and neuroendocrine tumours (i.e. pheochromocytoma) have a common embryological origin from the neural crest cells, although are distinct entities. A 4-year-old girl presented to the emergency room with a 5-month history of recurrent fevers with no infectious source and worsening abdominal pain. Exam did not reveal any diagnostic clues. Initial abdominal ultrasound showed a left renal mass. Abdomen and pelvic computed tomography revealed an 8.5 x 8.9 x 5.5 cm partially calcified mass anterior to the left kidney and extending to the suprarenal zone, with extensive lymph node metastases. There was evidence of multifocal bone metastases on the MRI abdomen and pelvis. MIBG had evidence of intra- abdominal and retroperitoneal neuroblastoma with bony metastases. Initial pathology of the left adrenal reported findings consistent with a peripheral neuroblastic tumour with variable morphology. Urine testing for VMA and HMA were positive in keeping with a diagnosis of neuroblastoma. A diagnosis of a high-risk neuroblastoma was made. Treatment including chemotherapy was started for neuroblastoma. On pathology review by the Canadian Oncology Group, the adrenal biopsy was identified to be composed of peripheral neuroblastic tumour (ganglioneuroblastoma) and pheochromocytoma. 24-hour urine metanephrines were also elevated, in keeping with a diagnosis of a composite pheochromocytoma.

The patient has been asymptomatic from any symptoms consistent with elevated catecholamines. Tumour debulking was arranged and alpha blockade was achieved with doxazocin. Post-operatively, plasma metanephrines have remained elevated. Imaging with I125 MIBG reported improvement in pre-existing lesions with evidence of a new lesion. It is not known whether the progressive disease or metastases seen are of neuroblastoma or pheochromocytoma origin. The patient is currently undergoing treatment for the high-risk neuroblastoma with a combination of chemotherapy, radiation, stem cell transplantation and immunotherapy. The question remains how does an asymptomatic composite pheochromocytoma fit into the larger picture of treatment decision making with a diagnosis of a high-risk neuroblastoma and is there an overall benefit to treatment of the pheochromocytoma.

Pediatric Endocrinology Education Amongst Trainees: A Scoping Review

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Background: Pediatric endocrinology education is a fundamental part of all pediatric residency and pediatric endocrinology fellowship programs. Hence, understanding the current methods used for training trainees on skills and abilities required in pediatric endocrinology will help improve the quality of training, and consequently help improve patient care.

Objectives: To explore training and assessment strategies used in pediatric endocrinology training across undergraduate and post-graduate medical education.

Methods: First, main topics of pediatric endocrinology were reviewed and identified. Second, using the topics as a reference, a search strategy was developed with the help of a librarian, and bibliographic databases (e.g. MEDLINE, EMBASE, PsycINFO, Compendex) were searched from January 2005 to September 2022. Before the title/abstract and full-text screening, pilot screenings were conducted until an agreement of 75% was achieved amongst the reviewers. Full-text articles were included if they were empirical studies, editorials, letters and commentaries related to pediatric endocrinology education, and focused on medical students, interns, residents and/or fellows.

Main Results: After the removal of duplicates, our scoping review yielded a total 5017 sources of evidence, of which 4935 were excluded during the title/abstract screening. After full- text review, we included 39 articles, and we are currently in the process of extracting data from all of the studies. We found that all studies were empirical, and majority focused on knowledge of type 1 diabetes and DKA (N= 15), followed by genitalia examination and pubertal assessment (N=8). Most frequently used training method was through didactics. Additionally, most frequently used assessment measures included knowledge tests (N=26). Also, limited number of studies targeted diabetes technology related skills and abilities that trainees should be competent at, even though technology such as insulin pumps for type 1 diabetes is widely used. Finally, no studies targeted common pediatric endocrine topics such as thyroid, bone health and adrenal insufficiency.

Conclusion: We identified that most studies targeting pediatric endocrinology education focused on knowledge about type 1 diabetes and DKA, without any training related to the common technologies used in these fields. Additionally, majority of the studies focused on knowledge as outcome, and assessed knowledge gain through written tests. Hence, future research should (a) explore educational innovation in other pediatric endocrinology topics, (b) target skills and abilities that go beyond knowledge that are important for patient care, such as technical skills, teamwork, and communication, and (c) link performance outside of the clinical setting to performance in the clinical setting.

P14

A Rare Case of Hypercalcemia of Malignancy of Unknown Etiology

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Division of Endocrinology, Alberta Children's Hospital, Calgary AB, Canada Department of Pediatrics, University of Calgary, Calgary AB, Canada Division of Oncology, University of Calgary, Calgary AB, Canada Department of Pathology and Laboratory Medicine, University of Calgary, Calgary AB, Canada Case: A 3-month-old girl presented with a 3-day history of emesis, decreased feeding, and decreased urine output. She was found to have a left renal mass and hypercalcemia (total calcium 4.72 nmol/L). Repeat labs confirmed hypercalcemia (total calcium 4.22 nmol/L, ionized calcium 2.47 mmol/L) with suppressed PTH, appropriately high urine calcium/creatinine ratio (4.3), and normal phosphate, magnesium, albumin, and creatinine. Her 25-OH vitamin D was 47.7 nmol/L and her 1,25-OH vitamin D was 52 pmol/L. Her PTHrP level was undetectable. Staging imaging showed metastases to the liver and lung, but not bone. After insufficient response to 2 days of IV fluids at 1.5 times maintenance and furosemide, she received 2 doses of Pamidronate. These therapies normalized her calcium levels prior to her surgery to remove the tumour. On pathology, the tumor was identified as a malignant rhabdoid tumour. She required some IV calcium in the first 3 days post-operatively. Follow-up PTH levels rose appropriately to 72 (ng/L), 74 during hypocalcemia but fell to 22 (normal) with normalization of her calcium levels.

Despite having tumour metastasis, her PTH and calcium levels have remained normal. Further investigations for other tumour markers that may have caused the hypercalcemia are ongoing.

Discussion: This patient's hypercalcemia of malignancy remains under investigation. The most common humoral factors were ruled out with laboratory testing, and osteolytic effects were ruled out by imaging. In addition, hypercalcemia unrelated to malignancy is unlikely given the resolution of hypercalcemia with tumour excision.

Conclusion: Hypercalcemia of malignancy occasionally cannot be explained by conventional markers, warranting further investigations.

P15

A case of Familial Glucocorticoid Deficiency Type I with a Newly Reported Mutation in the MC2R Receptor

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Background: Primary adrenal insufficiency (PAI) in children is a potentially fatal disorder if left untreated. Most commonly, this is due to congenital adrenal hyperplasia or Addison disease, but there are other less common causes that require a high index of suspicion to establish an early diagnosis and management. One of the rare causes of PAI are ACTH insensitivity syndromes such as familial glucocorticoid deficiency or triple A syndrome. Familial glucocorticoid deficiency is a nautosomal recessive condition involving ACTH signalling due to mutations in the genes coding for the melanocortin 2 receptor (MC2R) or its accessory protein 1 (MRAP). Inactivating mutations of the MC2R account for approximately 25% of these cases.

Case: We are presenting a case of a 2-year-old boy with macrocephaly, tall stature (>99th percentile), subtle distinctive facial features, developmental delay and diffuse bronze skin who was referred for overgrowth and a suppressed cortisol value (<22 nmol/L). Prior to this, he did not have any episodes of adrenal crises, except one isolated episode of hypoglycemia with RSV bronchiolitis. A high dose ACTH stimulation test revealed an undetectable stimulated cortisol value (<11 nmol/L) and he was started on hydrocortisone at 10mg/m2/day pending other investigations. It was later revealed he had an elevated ACTH (>440 pmol/l), with an intact mineralocorticoid axis (Aldosterone 283 pmol/L, Renin 1.09 ng/L/s) and normal androgen levels. Genetics obtained whole exome sequencing that showed two variants in the MC2R gene (in trans):c.437G>A, p.(Arg146His) and c.593T>C, p.(Leu198Pro), consistent with familial glucocorticoid deficiency type I. Interestingly, the latter one has not been previously reported. In subsequent clinic visits, his height velocity slowed down and the bronze skin colour has diminished. Family has been unable to obtain repeat ACTH testing.

Discussion: Familial glucocorticoid deficiency is a rare cause of PAI. Its clinical presentation is quite different including tall stature and lack of mineralocorticoid deficiency, which can delay diagnosis and management. Therefore, it is important to consider it as a differential diagnosis at an early stage when assessing a patient with PAI.

P16

Invasive Macroprolactinomas in Two Adolescents: Hypogonadotropic Hypogonadism Varies in Severity and Persistence

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Prolactin-secreting adenomas are the second most frequent functional pituitary adenomas in children and adolescents (37% VS 43% ACTH-secreting adenomas). [1] However, pediatric macroprolactinomas remain a rare entity, and there are only a few reported cases of giant macroprolactinomas in children, defined by a diameter greater than 4mm, often characterized by massive extrasellar extension and associated to severe hyperprolactinemia (> 1000mcg/L). [2] Adult studies have demonstrated that in macroprolactinomas specifically, a tumor volume shrinkage of 30% or more in the first 3–4 months after initiation of a dopamine agonist are predictive of the long-term response to a pharmacological approach. [3]. We report two cases of prolactin-secreting adenomas: one macroprolactinoma and one giant macroprolactinoma in a female and a male adolescent respectively.

The first patient is a 16 year old female who presented with a complaint of primary amenorrhea. A biochemical evaluation unveiled a prolactin level superior to 2000 mcg/L and a brain MRI demonstrated a pituitary macroadenoma of $20 \times 17 \times 22 \text{ mm}$, with slight invasion of the cavernous sinuses. She was started on cabergoline, and the control MRI showed a small reduction in size of the tumor (18 x 18 x 26mm) 2 months after treatment was initiated, time at which the prolactin level normalized. She remained amenorrheic 2 months post treatment initiation, but presented with progressively increasing LH and estradiol levels at that time.

The second case is a 17 year old male patient who presented with a history of worsening headaches and diplopia. A brain MRI showed an invasive giant macroadenoma ($64 \times 55 \times 52$ mm) with lateral invasion of cavernous sinuses, with an initial prolactin level > 2000mcg/L and asymptomatic biochemical hypogonadotropic hypogonadism. There was an excellent response to treatment, with a 85% reduction in the volume of the lesion ($23 \times 42 \times 28$ mm) after a 3 month course of cabergoline, but he remained with low testosterone and gonadotropin levels in the long term.

This case report will add to the literature on macroprolactinomas in the pediatric population and will hopefully help us further predict their clinical course, specifically regarding factors contributing to persistent hypogonadotropic hypogonadism.

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Pituitary Adenoma or Not? A Case of a Pediatric Pituitary Abscess - A Rare Adenoma Mimicker

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Background: A pituitary abscess is a rare pediatric pituitary lesion seldomly described in pediatric case reports. Here we report a case of primary pituitary abscess initially diagnosed as macroadenoma.

The Case: Our case is a 12-year-old female who presented with periodic episodes of headache, fever and vomiting associated with visual changes. The episodes were self-limited over several months. Following the fifth episode, she was hospitalized. Neuroimaging revealed a sellar mass measuring $1.2 \times 1.9 \times 1.4$ cm in size with a differential diagnosis of a macroadenoma, atypical Rathke's cleft cyst or craniopharyngioma. During admission, she was diagnosed with panhypopituitarism (PHP) with central diabetes insipidus and hyperprolactinemia secondary to the stalk effect. Multidisciplinary assessment with neurosurgery, rheumatology and ID resulted in a plan for biopsy due to the patient's abnormal pituitary function and concern for impingement of the optic chiasm.

Management: Surgical resection was performed via an endoscopic trans-sphenoidal approach, revealing purulent material and necrotic tissue. Tissue culture was positive for c ultibacterium acne infection. However, no pus cells were seen on the gram stain. The patient received a 3 -day course of ceftriaxone 6 weeks before surgery; therefore, partial treatment of the infection was suspected versus sample contamination. The patient was managed with 9 days of ceftriaxone and metronidazole and discharged with a plan to complete a total of 5-week course of both antimicrobials.

Discussion Points: Based on the literature review, pituitary abscesses are very rare in pediatrics compared to adults, accounting for only 0.11 % of pituitary lesions. Distinguishing pituitary abscesses from other lesions with MRI remains challenging, with accurate diagnosis ranging between 24 to 50 percent. Most cases are diagnosed intraoperatively. There is a 37 % chance of endocrine recovery (partial or complete). Thus periodic monitoring of pituitary hormones post-operatively is warranted.

Conclusion: Pituitary abscess should be suspected in patients with sellar lesions with atypical inflammatory symptomatology to ensure timely medical and surgical management

P18

A Case of NR0B1-related Adrenal Hypoplasia Congenita: a challenging diagnosis mimicking isolated hypoaldosteronism

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Introduction: X-linked adrenal hypoplasia congenita (AHC) is a potentially life- threatening condition characterized by primary adrenal insufficiency associated with hypogonadotropic hypogonadism (HHG). This genetic disorder is caused by mutations or deletions in NR0B1 gene located on the short arm of chromosome X. NR0B1 gene encodes a nuclear receptor protein (DAX-1) which plays a key role in regulating the development of the adrenal cortex, gonads, hypothalamus, and pituitary gland. Salt-wasting crisis is often the first clinical manifestation of AHC occurring during the neonatal period. However, timing and clinical presentations may be variable and characterized by nonspecific symptoms, which can occur during childhood or adolescence. HHG typically develops during adolescence. Abnormal spermatogenesis has also been observed. We present an unusual case of AHC, which presented initially as isolated hypoaldosteronism.

Conclusion: This case demonstrates that AHC should be considered in male infants presenting with signs of isolated hypoaldosteronism as salt loss may be the presenting feature, with cortisol insufficiency developing with time. Therefore, a basal cortisol level within normal limits should be interpreted with caution and possibly reassessed with provocative testing, particularly when there are signs and symptoms of adrenal insufficiency in an unwell child.

P19

Hyperfunctioning thyroid nodules in pediatric patients: rare and unpredictable!

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Although thyroid nodules are rare in childhood, malignancy rates are higher in children than in adults. Up to 26% of all pediatric thyroid nodules are malignant compared to 7-15% in adults(1,2,3). Pediatric thyroid carcinomas are almost always well differentiated(4) and almost 90% of those are papillary thyroid carcinomas while less than 10% are follicular thyroid cancers(5). Hyperfunctioning thyroid nodules (HFTN) account for 5% of all pediatric thyroid nodules(4). Histology of HFTN in children is most often benign but papillary carcinoma has been found in approximately 5% of them(4).

We describe two adolescent females presenting with clinically mild hyperthyroidism (suppressed TSH, elevated total T3 with moderately elevated FT4 levels). The second patient had thyroid antibodies including TSI (Thyroid Stimulating Immunoglobulin). Both patients had a palpable HFTN in the left lobe characterized as TIRADS 4 on ultrasonography. Thyroid scintigraphy scan performed in the workup of the second patient who had thyroid antibodies confirmed the presence of the HFTN in the left lobe. A Fine Needle Aspiration Biopsy of the first patient's HFTN was interpreted as a follicular lesion of undetermined significance. Both patients underwent left hemithyroidectomy. Pathology of the resected lobe showed an encapsulated follicular neoplasm with 0,1cm of papillary microcarcinoma and a follicular Hürthle (oncocytic) cell adenoma in respectively the first and second patient. The post- operative period was complicated by hypothyroidism necessitating temporary suppletion with levothyroxine only for the first patient. Since both patients present infracentrimetric lesions in the contralateral lobe, careful follow-up is recommended.

These cases highlight the following points:

The most sensitive marker for hyperthyroidism is an elevated FT3 level rather than an elevated FT4 level(6), especially for HFTN The challenge in risk classification of HFTN in children

Management of HFTN in children should be different compared to adults: According to current adult guidelines, HFTN should not undergo malignancy stratification since malignancy in these nodules is extremely rare(7), whereas in pediatrics, surgery is recommended in all pediatric HFTN by The American Thyroid Association guidelines(5).

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P20

Primary ovarian insufficiency in Coffin-Siris syndrome type 8 with a novel SMARCC2 variant: a case report

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Background: Coffin-Siris Syndrome type 8 (CSS8) is an autosomal dominant condition caused by inactivation of one copy of the SMARCC2 gene. SMARCC2 is one of the invariable core subunits of the ATP-dependent chromatin remodeling BAF (BRGA1-associated factor) complex and plays a critical role in embryogenesis and corticogenesis. Intellectual disability, developmental delay, growth retardation, and dysmorphic features are the main presenting signs for the 15 reported CSS8 individuals. Primary ovarian insufficiency has not been previously reported among any of the total 208 CSS cases.

Case Description: We present a 15-year-old girl referred for endocrinologic evaluation of delayed puberty, primary amenorrhea, and short stature (height 3rd percentile, -1.85 SD; mid-parental height at 14.8th percentile). Significant medical history included global developmental delay, ADHD, anxiety, speech delay, and dysmorphic features. Ophthalmological assessment for abnormal eye movements revealed bilateral mild optic nerve hypoplasia (ONH). Workup for primary amenorrhea revealed repeatedly elevated FSH, LH and undetectable levels of estradiol and anti-Mullerian hormone (AMH). Adrenal autoantibodies were negative. Genetic testing was negative for Turner and Fragile X syndrome. Chromosomal microarray was normal female. Evaluation for short stature and possible hypopituitarism given clinical finding of mild ONH included normal IGF-1, sodium, free T4, and low-dose ACTH stimulation test; bone age was consistent with chronological age. MRI head is pending. Whole exome sequencing (WES) showed a likely pathogenic de novo heterozygous nonsense variant in the SMARCC2 gene, c.635dup, p.(Tyr212*), in keeping with Coffin-Siris type 8. Parents are non-consanguineous from India.

Conclusion: This patient highlights the broad phenotype of individuals with CSS8 and suggests that SMARCC2 should be added to primary amenorrhea gene panels. Comprehensive investigations for primary ovarian insufficiency are warranted given the negative impact syndromic causes like CSS may have on an individual.

Improving Healthcare Provider Education on High-Quality, Inclusive, Affirming, Trauma Informed and Culturally Sensitive Care for 2SLGBTQ+ Patients

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Background: Individuals who are two spirit, lesbian, gay, bisexual, transgender, queer/questioning (2SLGBTQ+) experience more discrimination, victimization and violence compared to their heterosexual, cisgendered peers. Negative healthcare experiences faced by 2SLGBTQ+ patients contribute to disproportionately poor health outcomes and barriers accessing care. Medical educators in Canada have acknowledged the need to prepare trainees to provide informed, compassionate, affirming care to the 2SLGBTQ+ patient population but curriculum has not been well established and curriculum that has been developed remains sparse. This knowledge gap puts patients at risk and leaves them with an unfair burden to educate their care teams.

Methods: A Gender Sex Working Group was formed at London Health Science Centre (LHSC), comprised of physicians, residents, nurses, allied health and members of the 2SLGBTQ+ community. This group is improving care provided to the 2SLGBTQ+ community through better documentation practices within the patient's hospital records. The aim of this advocacy project is to improve healthcare provider education on providing inclusive, affirming, trauma informed and culturally sensitive care to 2SLGBTQ+ patients. This education roll out is in three phases.

First, infographics were circulated through email to LHSC staff explaining key terms such as sex, gender and pronouns. Second, the Working Group is establishing a partnership with leaders in 2SLGBTQ+ healthcare education to provide all staff with an online course to build foundational knowledge when providing care to 2SLGBTQ+ patients. Third, resident curriculum is being developed to involve patient panels to discuss ongoing health care disparities, attitudes and discrimination along with simulated cases to practice navigating care scenarios. Curricular changes will be assessed with pre and post resident OSCEs.

Results: This advocacy project is ongoing. Changes to the hospital's EMR have led to more inclusive patient identification. Roll out of the infographics has been very well received by frontline workers, who have been excited to see LHSC provide resources on 2SLGBTQ+ specific health care needs and have found the resources helpful.

Conclusion: These initiatives will not fix the oppressive structures that ultimately produced this health gap but are an essential strategy to improve patient experiences and reduce the suboptimal health outcomes amongst 2SLGBTQ+ individuals.

P22

A Unique Case of Hypophosphatemic Rickets

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Bjornstad syndrome is a rare autosomal recessive condition characterized by s ensorineural hearing loss and pili torti (twisted hairs). It is caused by mutations in the BCS1L gene, which encodes a member of the AAA family of ATPases that is necessary for the assembly of complex III in the mitochondria.

We report on a 5-year-old boy with Bjornstad syndrome that was referred to the endocrine clinic for poor growth and complaints of leg pain and difficulty climbing stairs. His height was 88.9 cm (<1 %, Z= -4.60) and weight 1.4 kg (<1 %, Z= -3.78). The physical examination confirmed normal body proportions, widened wrists, genu valgum, no scoliosis and bony tenderness. Generalized alopecia identified as pili torti was evident and as well as bilateral cryptorchidism. Initial investigations revealed normocalcemia (2.45 mmol/L), hypophosphatemia (0.81 mmol/L) and an elevated alkaline phosphatase (911 U/L) with a normal PTH (44ng/L) Vitamin D was replete (75 nmol/L) and the 1.25 dihydroxy vitamin D was elevated (272 pmol/L). He had normal TFTS, celiac screen and his IGF-1 was low normal at 20 ug/L. Metaphyseal irregularity and physeal widening in keeping with rickets were seen on radio-imaging. His bone age was delayed at 2 years. A growth hormone stimulation test was suboptimal (4.99 IU/L), however MRI delineated a normal pituitary gland. He was commenced on growth hormone therapy and the etiology of hypophosphatemic rickets ensued as it was unlikely to be FGF23- mediated.

Additional investigations identified a metabolic acidosis, glucosuria, high urine phosphate loss, and high renal tubular protein loss, consistent with a renal tubulopathy. This is a rarely reported complication of Bjornstad syndrome.

He was referred to nephrology for ongoing management and we continue to monitor his progress.

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An Interesting Case of Persistent Low IGF-1 in Two Siblings

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Background: The differential diagnosis for short stature includes abnormalities in the growth hormone (GH) axis, such as rare GH insensitivity syndromes. We present a case of two brothers with low IGF-1 suspicious for GH insensitivity.

Case: Two brothers were referred for assessment of short stature. The older brother was initially seen at age 6 years. On exam, he was non-dysmorphic and proportionate with height 106.0 cm (-2.6 SDs) and weight 15.7 kg (-2.6 SDs). Height velocity was 5.8 cm/year over the preceding 3 years. The younger brother was seen at age 5 years and was non-dysmorphic and proportionate with height 105.4 cm (- 1.9 SDs) and weight 16.0 kg (-1.7 SDs). Height velocity was 5.6 cm/year over 2.5 years. Midparental height was 170.1 cm (-0.9 SDs). Initial investigations were unremarkable except for very low IGF-1, 15 (47–184) mcg/L and 14 (27–134) mcg/L in the older and younger brother, respectively. Bone age was within 2 SDs for both siblings. Peak GH on stimulation testing was 10.64 mcg/L in the older brother and 7.85 mcg/L in the younger brother.

Despite ensuring adequate nutrition, IGF-1 levels remained low in both siblings (11 and 14 mcg/L). Assay results were verified at another centre and IGFBP-3 levels were obtained – 0.4 (1.3–6.5) mg/L and 0.3 (1.3–6.5) mg/L in the older and younger sibling, respectively. Given profoundly low IGF-1 and IGFBP-3 with normal GH stimulation tests, GH insensitivity was suspected. IGF-1 generation test (IGFGT) was completed in the older brother. After 7 days of recombinant GH at 0.18 mg/kg/week, IGF-1 was 34 (67–254) mcg/L, likely consistent with GH insensitivity. Genetic testing is being pursued.

Discussion: In this case, the suboptimal rise in IGF-1 with recombinant GH is suspicious for GH insensitivity, though not diagnostic given the heterogeneity in IGFGT protocols and cut-offs. This illustrates one of the challenges in diagnosing GH insensitivity. For accurate diagnosis, genetic testing may be required. In these siblings, we suspect an acid-labile subunit deficiency based on the characteristic pattern of low IGF-1 and IGFBP-3 with normal GH, adequate growth velocity, and less severe short stature.

It takes Two to Tango: A Case of Hyperglycemia in Multiple Endocrine Neoplasia Type 1

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Hyperglycemia in multiple neuroendocrine neoplasia type 1 (MEN1) can be related to neuroendocrine tumors or pituitary hormone excess secondary to pituitary adenomas. We present another etiology of hyperglycemia in the setting of MEN1 and suggest that both conditions interact.

A 12-year-boy with known MEN1 was found on annual neuroendocrine tumor screening to have impaired fasting blood glucose and a hemoglobin A1C of 6.4%. Over the next three months, he developed polyuria, polydipsia, and weight loss. Continuous glucose monitoring revealed normal post-prandial glucose levels. There was a family history of type 1 diabetes (paternal grandfather) and no risk factors for type 2 diabetes other than elevated glucose levels, there was no clinical or biochemical evidence of glucagonoma, growth hormone excess, hypercortisolism, or neuroendocrine tumors. He was diagnosed with type 1 diabetes in evolution and started on once daily basal insulin. However, anti-GAD, anti-insulin and islet cell antibodies were negative. and insulin requirements remained low. Genetic testing for monogenic diabetes revealed a heterozygous pathogenic missense variant in the glucokinase gene (GCK), confirming a diagnosis of Maturity-onset diabetes of the young type 2 (MODY 2). Attempts to discontinue basal insulin, which he remains on, were unsuccessful with further elevation of fasting blood glucose with mild symptoms of hyperglycemia. Post-prandial blood glucose readings remained normal. MODY2 is characterized by non-progressive, mild fasting hyperglycemia and minimal post-prandial blood glucose excursions. Insulin therapy is rarely necessary and, when required, is more likely in the setting of obesity or older age. MEN1 is a neuroendocrine tumor syndrome resulting from mutations in MEN1, a tumor suppressor gene that encodes menin. MEN1 has been associated with insulin resistance with the suggestion that menin has downstream modulatory effects on insulin action. It is possible that pathogenic mutations to both MEN1 and GCK have additive effects on glucose dysregulation, underlying the persistent basal insulin requirements observed in this patient with both MEN1 and MODY2. This case depicts the interaction of two rare conditions, emphasizing the impact of MEN1 on glucose metabolism and highlighting that persistent insulin needs in MODY2 may be a result of additional underlying pathology.

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ACTH-dependent Cushing's in an 8-year-old: a diagnostic challenge

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Background: Cushing's syndrome (CS) is rare, with 2-5 cases per million annually, and only 10% of these occurring in children. Cushing's disease (CD) is the most common organic etiology of CS in children (75-90%), while ectopic ACTH syndrome (EAS) accounts for less than 1%.

Case Presentation: An 8-year-old male presented with extensive vertebral compression fractures, rapid weight gain, and headaches. He was found to have hypertensive urgency (blood pressure 170/110 mmHg, > 99.9 percentile) and Cushingoid features. Investigations revealed hypokalemia (2.0 mmol/L), elevated serum cortisol (1654 nmol/L, ref 85-620) and elevated 24-hour urine cortisol (27244 nmol/day, ref < 25). An elevated ACTH (35pmol/L, ref 1.6-13.9) confirmed ACTH- dependent Cushing's. Although non-invasive dynamic testing (including high dose dexamethasone suppression and desmopressin stimulation tests) suggested an ectopic source, inferior petrosal sinus sampling (IPSS) was suggestive of a pituitary source. Unfortunately, anatomical (3T MRI Sella) and functional imaging (18 F-DOPA PET/CT and Gallium-68 Dotatate PET/CT scans) have not identified a causative lesion to date. As such, he has been started on ketoconazole to control the hypercortisolemia and to reduce morbidity. This has been mainly effective, with 24- hour urine cortisol reduced to 70nmol/day. He will undergo serial imaging and continue medical therapy as long as this is tolerated. Exploratory pituitary surgery may be considered if medical therapy proves ineffective or overly burdensome over time.

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Diabetes Duration and Glycemic Control in Adolescents with Type 1 Diabetes: A Cross-Sectional Study

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Objectives: Evidence is lacking on whether diabetes duration influences comfort with diabetes management in adolescents with type 1 diabetes (T1D) prior to the transfer from pediatric to adult care. We examined associations of diabetes duration with diabetes management dimensions (self-efficacy, transition readiness, diabetes distress) as well as glycemic control in late adolescence.

Methods: Using a cross-sectional design, we conducted a secondary analysis of baseline data of adolescents (ages 16-17 years) with T1D followed at pediatric diabetes academic hospitals in Montreal and enrolled in the Group Education Trial to Improve Transition (GET-IT-T1D). Participants completed validated questionnaires on self-efficacy (Self-Efficacy for Diabetes Self-Management Measure [SEDM], score 1 to 10), diabetes distress and transition readiness, as well as an A1c capillary blood test. The primary outcome was self-efficacy. We examined associations of diabetes duration with diabetes management dimensions and A1c using regression models adjusted for sex, socioeconomic status, insulin pump use, glucose sensor use, and psychiatric comorbidity (eating disorders, depression, anxiety, attention deficit disorder, other).

Results: Of 203 adolescents with T1D, mean diabetes duration (SD) was 7.57 (4.44) years. SEDM score was 6.83 (1.62). Diabetes duration was not associated with self- efficacy, diabetes distress or transition readiness. Adolescents with a longer diabetes duration had higher A1c (adjusted β , 0.107; 95% CI, 0.053 to 0.161).

Conclusions: Whereas diabetes duration is not associated with diabetes management dimensions, adolescents with longer diabetes duration are at risk for higher A1c and may need additional support to improve glycemic control before transition to adult care.

Rebound Hyperkalemia After Focal Resection for Congenital Hyperinsulinism

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Background: Congenital hyperinsulinism (CHI) is characterized by dysregulated insulin secretion and can cause significant brain injury. Inactivating mutations in ABCC8 and KCNJ11, the genes encoding the beta-cell adenosine triphosphate (ATP)- sensitive potassium (K–ATP) channel, located on chromosome 11p15.1, cause the most common forms of CHI, which can be diffuse or focal. Focal CHI occurs by a "two-hit" process - there is a paternally inherited loss-of-function mutation in ABCC8 or KCNJ11 and a somatic loss of the maternal 11p15 region limited to a specific area within the pancreas.

Case: A 3-month-old boy was diagnosed with hyperinsulinism in India, which was confirmed at our centre with a critical sample showing hypoketotic hypoglycemia (glucose 2.6 mmol/L) with a detectable insulin level (16.2 pmol/L). An F-DOPA PET scan demonstrated a focal lesion in the pancreas. Genetic testing showed he was heterozygous for a paternally inherited loss of function variant in ABCC8, consistent with focal CHI due to an additional somatic loss of the maternal allele. The patient had surgery to remove the pancreatic lesion and pathology demonstrated a 0.5cm focus of endocrine cell adenomatous hyperplasia that was insulin immunopositive but p57 negative, consistent with focal hyperinsulinism. Starting intra-operatively, he developed hyperglycemia and hyperkalemia (K peak of 8.9 mmol/L immediately post-operative with peaked t-waves on electrocardiogram (ECG)), requiring treatment with calcium chloride, insulin and dextrose, salbutamol, and sodium bicarbonate. Within the first 24 hours post-operative, potassium and blood sugars normalized without further intervention.

Discussion: Rebound hyperkalemia following abrupt withdrawal of excessive endogenous insulin has been reported only once previously in a 19-month-old following partial (80%) pancreatic resection for CHI. The post-operative potassium peaked at 12.3 mmol/L and produced ECG changes. The patient also had hyperglycemia up to 21.7 mmol/L post-operatively. Both the hyperkalemia and hyperglycemia resolved by the next morning, similar to our case. The mechanism of hyperkalemia is thought to be excessive total intracellular potassium that, following a reduction in serum insulin, effluxed back into the extracellular compartment.

Conclusion: We report a case of critical hyperkalemia following surgery for focal hyperinsulinism. This is a rare complication that should be monitored for post-operatively.

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Diabetic ketoacidosis associated with severe hypertriglyceridemia and acute pancreatitis in type 1 diabetes: 4 pediatric cases

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Severe hypertriglyceridemia due to diabetic ketoacidosis is rare, particularly in pediatrics. Most patients have a secondary cause or type 2 diabetes. We report 3 cases of young patients with new onset type 1 diabetes (DT1) with no secondary cause for their diabetes, presenting with severe hypertriglyceridemia.

The first patient, a 17 year old girl with obesity, presented with DKA, acute abdominal pain and vomiting with elevated lipase= 1725U/L (N: 4-39U/L) and TG = 114.96mmol/L (N: 0.4-1.3mmol/L). Abdominal CT-scan confirmed acute pancreatitis. She received plasmapheresis, with significant improvement of her clinical picture. DT1 was confirmed by antibody testing and no underlying lipid disorder was found at the genetic panel. The second patient was a 13 year old boy with persistent DKA and abdominal pain despite normalization of blood glucose. Lipemic appearing serum prompted TG measurement, which was 75.65mmol/L. Subsequent genetic analysis found a variant in the APOA5 gene that may be associated with increased susceptibility for elevated triglyceride levels. The 3rd patient was a 13 year old girl who presented with classic symptoms of DKA and very elevated TG = 159.8 mmol/L. Given persistence of her acidosis and significant abdominal pain without radiological evidence of pancreatitis, plasmapheresis was performed with rapid normalization of symptoms. Molecular screening did not demonstrate any mutation associated with hypertriglyceridemia. On follow-up, clinical and biological parameters normalized in all patients, with no further hypertriglyceridemia upon resolution of the acute event.

The underlying pathophysiology of the triad DKA-HyperTG-AP is linked to prolonged lack of insulin, leading to increased lipolysis and inhibition of lipoprotein lipase activity, with subsequent increases free fatty acids and triglycerides. This in turn leads to pancreatic cell free radical injury, edema, and subsequent ischemia, as well as pancreatic ischemia directly from hyperchylomicronemia induced capillary hyperviscosity. Some underlying mechanisms still remain unclear, in particular individual susceptibility factors. Management strategies include high-dose insulin infusion and plasmapheresis; to date there are no consensus guidelines for the management of this rare but serious complication of DKA.

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Metastatic Pheochromocytoma/Paraganglioma in Von-Hippel-Lindau disease: An Uncommon Cause of Paediatric Hypertension

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Catecholamine-secreting chromaffin tumours are common among individuals with germline predisposition. Von Hippel-Lindau (VHL) disease is the most common genetic cause of pediatric pheochromocytoma/paraganglioma (PPGLs). While tumours may be multifocal (either synchronous or metachronous), metastatic PGGL are rare outside of carriers of germline SDHB variants, with an estimated rate of 5–8% in VHL disease. We present the case of a 6-year-old previously healthy male with multiple incidental secreting tumours, including a suspected metastatic lesion. We describe his diagnostic evaluation and treatment course.

A 6-year-old boy was referred for evaluation of bilateral undescended testicles. Abdominal-pelvic ultrasound demonstrated an incidental retroperitoneal mass in the right supra-renal fossa. More detailed evaluation demonstrated three retroperitoneal masses. During his evaluation, he had asymptomatic hypertension with peak systolic blood pressure of 149 mmHg. He had no other symptoms of catecholamine excess or family history of PGGLs or endocrine neoplasias. Bloodwork was notable for elevated serum normetanephrine (>7.5 nmol/L; reference range <0.9 nmol/L) and urine VMA (12.8 mmol/mol Cr; reference range ≤5.0 mmol/mol Cr) with normal remaining serum metanephrines. He was diagnosed with PGGL. MRI abdomen confirmed three discrete heterogeneously enhancing lesions, ranging from 1.1-2.6 cm in the right adrenal gland, midline retroperitoneum and adjacent to the right liver. 68Ga--DOTATATE scan showed similar somatostatin-avid lesions and an additional mid-retroperitoneal focal uptake.

Pre-operative alpha-blockade was initiated with doxazosin, as was salt-loading and hyperhydration. He underwent an uncomplicated complete right adrenalectomy and excision of multiple paragangliomas. Multiple intra-adrenal pheochromocytomas, one extra-adrenal paraganglioma with negative excision margins and one subdiaphragmatic lesion with positive resection margins were identified. Given the rarity of metastatic disease in VHL, particularly at this young age, careful histological examination of the subdiaphragmatic lesion was performed. This was felt to be consistent with metastatic, as opposed to synchronous, tumour due to its atypical site, extensive infiltration of connective tissue and vascular space, and a solitary lymph node containing tumour cells. Germline analysis demonstrated a de novo c482G>A variant in the VHL gene and a variant of uncertain significance in the FH gene. Post-operatively, he remains normotensive with normal metanephrines and no residual disease on repeat imaging.

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Gaps in Type 1 Diabetes Care After Transfer from Pediatric to Adult Care and Longitudinal Associations with Psychosocial Factors

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Aims: Adolescents with type 1 diabetes (T1D) progressing to adulthood assume responsibility for diabetes self-care management while dealing with competing life demands. Psychosocial factors that may improve diabetes-related outcomes during the transition to adult care remain understudied. In emerging adults with T1D, our objectives were to 1) estimate prevalence of gaps in care after transfer to adult care and 2) evaluate longitudinal associations of psychosocial factors with gaps in care.

Methods: Prospective cohort study of adolescents with T1D followed 18 months from their last pediatric visit, from 2017 to 2021. Before transfer, adolescents (ages 17 years) completed validated psychosocial questionnaires on self-efficacy, transition readiness and diabetes distress. Diabetes care visits were assessed with administrative health records. Primary outcome: gap in care, defined as ≥6 months between the last pediatric and first adult care visit. Secondary outcome: ≥1 gaps in care ≥6 months between physician visits over 18 months after transfer to adult care. Associations of psychosocial measures with gaps in care were evaluated using logistic regression adjusted for sex, diabetes duration, socioeconomic status, mental health comorbidities, use of insulin pump or continuous glucose monitoring, and Hemoglobin A1c (HbA1c) before transfer.

Results: 74 adolescents with T1D were included. 11 participants (15%) had a delay ≥6 months in establishing adult diabetes care. During the 18 months after transfer, 34 participants (46%) had ≥1 gaps in care ≥6 months. No evidence of associations was found between psychosocial measures and gaps in care.

Conclusions: Whereas only 15% had delays in establishing adult diabetes care, almost half developed gaps in adult care after transfer. No evidence of longitudinal associations was found between psychosocial factors in adolescence and gaps in care after transfer. Strategies should support young adults with T1D in maintaining regular access to specialized healthcare services.

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Transition from pediatric to adult obesity care - a retrospective review

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Background: Transitioning youth with complex health needs remains challenging. Obesity is a complex medical disorder that can begin in childhood with over 90% continuing to have obesity into young adulthood. Despite increasing dialogue for transition care in obesity management, there is minimal empirical research and only few clinical guidelines contained brief recommendations. Given such, risk factors and areas of care deficiencies during transition remain unclear. This study aims to review the characteristics of patients who transitioned from pediatric to adult obesity care.

Results: Of 280 patients (68% female), 23% were referred from a pediatric obesity program or provider and remaining from a family physician or adult provider. 69.3% attended their consultation, 20% did not attend, and 10.7% were booked for consultation after September 2022. There was no difference in consult attendance based on referral source. Females were more likely to attend their consultation (78% vs 60%), but ongoing follow-up was approximately 50% for both genders. Of those who attended their consultation, in-person or video visits were more likely to follow-up (57% and 62%, respectively), compared to 20% follow-up for telephone visits. 56.5% attended the consultation with a parent. Those who attended the consultation alone were more likely to have at least one follow-up compared to those attended with a parent (63% and 50%, respectively).

Discussion: Whether referred by a pediatric obesity program or family physician, about 2/3 of patients presented for their initial consultation (females more than males) and half of these patients were likely to have at least one follow-up when the consultation had been in person or by video, as opposed to telephone. Attendance at consultation without a parent also increased follow-up attendance. Results of the study will help inform how best to structure and evaluate a future bariatric transition program.

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Effect of COVID-19 on Glycemic Control in Children Living with Type 1 Diabetes Mellitus

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Background: The world began to see effects of the SARS-CoV-2 virus (COVID-19) in December 2019. A state of emergency was declared in Ontario, March 2020 resulting in sweeping changes to health care provision. Reports from Europe, the United States and Canada found an increase in presentations of diabetic ketoacidosis (DKA), and an increase in severity of presentations. There are emerging data by region on glycemic control in patients with established Type 1 Diabetes Mellitus (T1DM) during the pandemic.

Objectives: The primary aim was to assess glycemic control during the COVID-19 pandemic in a cohort of patients living with T1DM followed at London Health Science Centre (LHSC).

Methods: A retrospective chart review was completed evaluating glycemic control in patients with established T1DM and assessing severity of presentation of new patients with T1DM from March 2020 to February 2021. Data were compared to the year prior. Postal code data were collected for secondary location data analysis.

Results: There were 65 new presentations of T1DM during the pandemic year compared to 41 presentations the year prior. No significant difference in the number of new presentations of DKA or severity of presentation was found in patients with new onset T1DM. In patients with established T1DM (n=132), there was an average increase in HbA1C of 0.35% (p<0.001) during the pandemic year. There was also a significant increase in BMI percentile by 2.3% (p=0.024) and weight percentile by 2.17% (p=0.008). A secondary location data analysis is underway to assess HbA1c and growth parameters by postal code region.

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Associations between Clusters of Parental Characteristics and Offspring Adiposity in Late Adolescence

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Introduction: Several parental lifestyle behaviours, such as physical activity (PA), may determine offspring obesity; however, no prospective studies have examined the joint impact of combined parental characteristics including lifestyle habits on offspring adiposity. We examined whether clusters of selected parental characteristics were associated with subsequent offspring adiposity in late adolescence.

Methods: Data were from the QUALITY Cohort, a longitudinal study of children with at least one biological parent with obesity (n=630). Parental characteristics were collected at baseline when participants were 8-10y and offspring adiposity outcome measures (BMI z-score, android/gynoid fat ratio, % body fat, fat mass index) were obtained at 15-17y. Parental smoking, PA and education were self-reported. Parental and child weight and height were measured by trained nurses and BMI was computed. Additional child adiposity measures were obtained using dual energy x- ray absorptiometry. Cluster analysis was used to identify distinct maternal and paternal clusters based on parental BMI, PA, education and smoking habits at baseline in 209 families with complete data across both evaluation cycles. Multivariable regression models, adjusted for offspring age, sex and Tanner stage, were used to assess associations between maternal and paternal clusters and offspring adiposity outcomes.

Results: Three distinct clusters were identified among mothers and four among fathers. Mothers in cluster 1 (n=18) had obesity, lower educational attainment, smoked, and were more physically active; in cluster 2 (n=109) had overweight, higher educational attainment and were non-smokers; and in cluster 3 (n=82) had overweight, lower educational attainment, were non-smokers, and were less physically active. Offspring of mothers in cluster 1 had higher adiposity on all outcomes examined when compared to offspring from mothers in cluster 2 (e.g. for BMI z-score β =0.94, [0.35,1.53], p-value=0.01). Offspring adiposity levels were comparable across all four paternal clusters.

Conclusions: The offspring of mothers who presented a specific combination of risk factors (obesity, lower educational attainment and smoking habits) had higher adiposity measures. A similar risk profile for paternal characteristics and offspring adiposity was not observed. The context within which mothers are more likely to exhibit a deleterious profile of modifiable risk factors could be targeted for paediatric obesity prevention strategies.

Bone health in adolescents with Type 2 diabetes

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Introduction: The prevalence of type 2 diabetes (T2D) has increased alongside the global obesity epidemic. While once considered a disease of adulthood, T2D is being diagnosed at an increasing rate in children and adolescents. Adults with T2D have been found to have an increased fracture risk without reductions in bone density. As childhood and adolescent T2D is a relatively new entity, the effect on bone density and microarchitecture are currently unknown.

Objective: The primary objective of this study was to examine the bone microarchitectural properties of adolescents with T2D compared to a healthy reference population.

Methods: To achieve this objective a prospective, cross-sectional study was conducted. Participants were recruited from the Pediatric Diabetes clinic in Saskatoon, Canada, and compared with a previously published healthy pediatric reference sample. Anthropometric measurements, medical record review, questionnaires, and HR-pQCT scans of the non-dominant radius and tibia were obtained for each T2D participant. Demographics were compared between the groups using independent sample t-tests. Z-scores were calculated for the HR-pQCT bone parameters for the T2D participants using previously published sex specific healthy reference values. Reference curves were also created to compare the T2D participants to the healthy reference population. Results: 5 participants with T2D (4 female) were recruited with a mean age of 14.5 \pm 2.3 years (range 10.4-16.3), duration of diabetes 19 \pm 15.7 months (range 3-36), BMI of 32.7 \pm 7.3 kg/m² (range 22.4-40.1), and A1C of 8.6 \pm 3.9 % (range 5.4-13.1). Female adolescents with T2D had trabecular thickness 2SD below the reference sample population at both the distal radius and tibia.

Conclusion: To our knowledge, this is the first study to use HR-pQCT to examine bone density and microarchitectural properties, in a cohort of adolescents with T2D. Our preliminary data found that females with T2D had differences in bone microarchitecture, specifically in the trabecular compartment. These findings suggest differences in bone development may be present in female adolescents with T2D. Future studies with a larger cohort are required to confirm and expand upon our findings, and aid in understanding the clinical implications of the differences identified.

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Outcomes of Growth Hormone Therapy for Children with Short Stature

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Background: Growth hormone therapy (GHT) has been used in the practice of Endocrinology for over 70 years. In Canada, GHT is indicated in 8 distinct conditions in addition to growth hormone deficiency (GHD). While efficacy is well demonstrated in children with GHD, outcomes in children on GHT for other indications are more variable. The goal of this study is to examine the outcome of GHT in children with short stature and identify predictors of response.

Results: A mean follow-up of 4.77 years was collected per patient (range 1 to 7 years). Over the first year of GHT, there was a weak negative correlation between the change in Z-score and the baseline IGF-1 value (n= 112, Spearman's rho = -0.314, p-value < 0.001) but no significant correlation between the change in Z-score and the GH-peak value (n= 101, p-value = 0.348). In the multi-year follow-up, baseline mid-parental height and IGF-1 levels were significant positive predictors and pubertal status a negative predictor of the change in height Z-score for patients receiving GHT. A patient's diagnosis had a statistically significant interaction with time, indicating diagnoses respond to GHT differently over the course of treatment. Sex and age at GHT initiation were determined to be insignificant predictors as they are accounted for by the Z-score calculation and are co-linear with change over time.

Conclusions: Outcomes in children with short stature undergoing GHT are significantly affected by the underlying diagnosis and associated with several correlated biochemical and genetic variables indicating that children with different diagnoses respond to GHT differently over the course of treatment.

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Evaluation of the impact of attending Camp Banting (for children and youth with diabetes) on diabetes distress, quality of life and mental health

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Background: Depression and anxiety have been observed with a higher incidence in individuals with type 1 diabetes (T1DM). Diabetes distress (DD) and a sense of isolation are commonly reported.

Objectives: Primary: To assess change in DD for youth one month after attending diabetes camp. Secondary: To assess the impact over time of camp attendance on DD, health-related quality of life (HRQOL), symptoms of anxiety, depression, stress and family relationships.

Methods: A single center prospective study of youth with T1DM at Camp Banting. Individuals 8-16 years old and a caregiver completed validated online questionnaires assessing diabetes distress (PAID), HRQOL (T1DAL), mood and anxiety (PROMIS), immediately before and one month following camp attendance.

Results: Seventy campers and their caregivers completed baseline surveys with 2 8 campers and 48 caregivers completing surveys at one month, representing at least one respondent from 78% of camper-caregiver dyads. Camper mean (standard deviation [SD]) age was 12.4 (2.3) years with mean (SD) diabetes duration of 5.1 (3.9) years. Fifty-one (73%) campers were on pump therapy and 66 (94%) used continuous glucose monitoring. Mean (SD) baseline and one month PAID scores decreased from 25.9 (9.0) to 24.5 (9.2) for children and from 37.2 (13.7) to 34.0 (10.9) for adolescents, indicating a slight reduction in DD (though not statistically significant). A similar trend was seen in caregiver PAID scores. Baseline DD was higher in adolescents who did not complete follow-up versus respondents and similar for the other subgroups. Mean (SD) T1DAL scores at baseline and one month improved from 72.8 (11.0) to 76.0 (11.2) for children and from 64.7 (12.7) to 67.3 (12.8) for adolescents (also not statistically significant). The scores on the PROMIS measures for anxiety, depression, stress and family relationships were comparable to the general population for all groups.

Conclusion: Our study on a small convenience sample of campers and their caregivers showed a trend of decreasing DD and increasing diabetes HRQOL one month following diabetes camp. Our preliminary results suggest that a definitive study with a larger sample size should be conducted to further delineate the positive impact of diabetes camp for youth with T1DM.

Optional Non-Accredited Industry Symposia

Friday February 10, 2023 (Room: St-Antoine B)

Breakfast Session: 0745-0815

Navigating Tech Use in Tots: Emerging Evidence and Practical Tips

Toddlers and young children by their nature are fully dependent on parents for daily (sometimes hourly) diabetes management. Do tots and their families have a unique perspective on diabetes technology? You bet! This session will share evidence, recommendations, practical strategies and tips learned from the front lines of supporting families to navigate daily diabetes decisions.

Dr. Dan DeSalvo (Texas Children's Hospital)

Friday February 10, 2023 (Room: St-Antoine B)

Lunch Session: 1315-1345

Emerging Therapies of Patients with Prader-Willi Syndrome (PWS)

At the end of this session, participants will be able to:

- Outline treatment updates and approaches for patients with PWS
- Describe updates to the management of comorbidities in patients with PWS

Dr. Sanjukta Basak (BC Children's Hospital)

Saturday February 11, 2023 (Room: St-Antoine B)

Lunch Session: 1300-1330

Long-acting GH therapy: What are the current safety and efficacy data?

At the end of this session, participants will be able to:

- Discuss challenges of current daily GH therapy
- Discuss principles of long-acting GH therapy development
- Discuss current landscape of long-acting GH therapies either approved or in development

Dr. Aristides Maniatis (Rocky Mountain Pediatric Endocrinology)

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