CPEG GCEP

Canadian Pediatric Endocrine Group
Groupe canadien d'endocrinologie pédiatrique

February 6 - 8, 2025

RBC Place London

19th Annual

CPEG Scientific Meeting

Hosted in London by:

Division of Pediatric Endocrinology & Diabetes
at Western University and Children's Hospital,
London Health Sciences Centre

Digital Program

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Welcome to London!

On behalf of the Division of Pediatric Diabetes and Endocrinology at Western University, we are pleased to welcome you to London, Ontario!!

The first independent Canadian Pediatric Endocrine Group (CPEG) meeting occurred in London, Ontario in 2007. We are privileged to host our colleagues from across Canada for our 19th annual Scientific Meeting. We are excited to host the CPEG dinner at the Old Court House, a beautiful and historic building. If you have time, we hope you enjoy an outdoor skate at Victoria park, a few blocks away or take a tour of Western University. We are pleased to provide private tours of Banting House, National Historic Site; explore the legacy of Sir Federick Banting, birthplace of insulin discovery and tribute to medical innovation.

The scientific program for CPEG 2025 is inspired by what is new and upcoming in the field of Pediatric Endocrinology. Based on feedback from our recent conferences, our invited symposia speakers and debaters will share their expertise in novel understandings of clinical diseases, diagnostics, and therapeutics. Furthermore, we are excited to hear from our array of oral and poster presenters who will be showcasing their research.

As we cannot hold such a conference without the support of our sponsors, we encourage you to visit our industry booths in the conference foyer.

As your hosts, we would be pleased to share any local information with you – whether it be suggestions of where to eat, grab a coffee, or engage in outdoor activities.

We hope you have an engaging and memorable experience at CPEG 2025.

Funmbi Babalola & Patricia Gallego (CPEG 2025 local organizing committee)

Dear Attendees,

On behalf of the Scientific Committee and the Canadian Pediatric Endocrine Group (CPEG) Executive, I would like to welcome you all to the 19th Annual CPEG Scientific Meeting.

With our partners at the University of Toronto Continuing Professional Development, I hope that you will find that, the Scientific Committee has developed a remarkable meeting and program. In CPEG tradition we will continue to enjoy the habitually dynamic CPEG debate. As always, learners and others will have the opportunity to present their research in the oral and poster abstract sessions. The catchy one-minute poster highlights will also return this year.

I would like to thank the Scientific Committee for their hard work in planning this meeting with a special acknowledgement to Funmbi Babalola, Patricia Gallego and Kristen Langdon our local hosts.

Finally, a big thank you to our sponsors whose continued support makes 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 future endocrinologists.

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

Rebecca Perry

Chair, 2025 CPEG Scientific Committee

Scientific Committee

Rebecca Perry (Chair)
Funmbi Babalola (Local Chair)
Rose Girgis
Seth Marks
Manpreet Doulla
Paola Luca
Kate Potter
Patricia Gallego
Kristen Langdon
Sharon Costantini
Jana Haylor
Marwa El Masri
Sruthi Thomas
Mark Inman
Raelynn Friesen
Mallory McNiven
Chelsea Grimbly
Andrea Ens (Ex Officio Member)

Accreditation

Royal College of Physicians and Surgeons of Canada - Section 1

This event has been approved by the Canadian Paediatric Society for a maximum of 10 credit hours as 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. The specific opinions and content of this event are not necessarily those of the CPS, and are the responsibility of the organizer(s) alone.

Wi-Fi Internet Access

Network: **CPEG2025**

Password: **CPEG2025**

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/

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.

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.

Program Overview

The 19th 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.

We are pleased to gather again for our 2025 CPEG Meeting in London, Ontario. The scientific 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. Please note this year's meeting will commence midday on Thursday and conclude midday on Saturday, enabling attendees to return home on Saturday afternoon.

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. Apply knowledge of newer therapeutic options to optimize the management of autoimmune polyglandular syndrome 1
2. Apply knowledge of newer diagnostic options including genetic investigations to optimize the management of hypoparathyroidism
3. Recognize the importance of longitudinal thyroid screening in patients who have received iodinated contrast agents
4. Apply knowledge of newer diagnostic options including genetic investigations to optimize the management of pediatric thyroid cancer
5. Formulate a comprehensive approach to assessment and management of atypical congenital adrenal hyperplasia
6. Identify children and adolescents with monogenic dyslipidemia and maturity onset diabetes of the young (monogenic diabetes) to customize therapy

Session Learning Objectives

Symposium I - Calcium/PG Disorders

Hypoparathyroidism: Clinical Management Including Role of Genetics - Michael Levine

Objectives:

1. Distinguish between various causes of hypocalcemia
2. Recognize and diagnose the basis for hypoparathyroidism
3. Explain treatment options and goals of therapy for hypoparathyroidism

Symposium II - Thyroid

Thyroid Dysfunction After Iodinated Contrast Media - Jonathan Wasserman

Objectives:

1. Recognize the risk of thyroid dysfunction following use of Iodinated Contrast Media
2. Summarize newly described screening strategies to mitigate harm from unrecognized thyroid dysfunction

Thyroid Cancer in Pediatrics - Andy Bauer

Objectives:

1. Recognize and diagnose Thyroid Cancer in Pediatrics
2. List treatment options and goals of therapy for Thyroid Cancer in Pediatrics

Symposium III - Atypical Congenital Adrenal Hyperplasia (CAH)

3 Challenging CAH Cases - Funmbi Babalola, Patricia Gallego

Objectives:

1. Describe the pitfalls of prenatal screening in CAH

Genetic Review of Atypical CAH - Maha Saleh

Objectives:

1. Describe the genetics of CAH
2. Summarize molecular test modalities and limitations in diagnosis of CAH

Ethical Considerations for CAH - Jeanne Webber

Objectives:

1. Identify legislative and ethical considerations of pediatric capacity for decision-making within an intracultural context
2. Examine ethical considerations related to disclosure/withholding of information in treatment of Congenital Adrenal Hyperplasia

Symposium IV – Diabetes/Lipids

MODY - Amanda Berberich

Objectives:

1. Recognize the clinical presentations and consequences of the most common subtypes of monogenic diabetes
2. Recognize when to consider and how to confirm a diagnosis of monogenic diabetes

Monogenic Cases of Dyslipidemia Including Familial Hypercholesterolemia - Robert Hegele

Objectives:

1. Explain new concepts in the diagnosis and management of familial hypercholesterolemia
2. Formulate a clinical approach to other monogenic dyslipidemias

Debate

Use of GnRH Analogues in Later Early Puberty (6–8 Years) - Ereny Bassilious, Mark Palmert

Objectives:

1. List the pros of using GnRH Analogues to treat later early puberty (6-8 years)
2. List the cons of using GnRH Analogues to treat later early puberty (6-8 years)
3. Weigh the pros and cons of treatment and apply these to clinical practice

Invited CPEG Speakers

Funmbi Babalola MD MSc FRCPC
Pediatric Endocrinologist
Assistant Professor
Univeristy of Western Ontario
Children's Hospital
London Health Sciences Centre

Ereny Bassilious MD FRCPC MHPE
Associate Professor
Division of Endocrinology
Associate Chair Education,
Department of Pediatrics,
McMaster University Faculty of Health Science

Andy Bauer MD
Professor, Department of Pediatrics
Perelman School of Medicine
University of Pennsylvania
Director, The Thyroid Center
The Children's Hospital of Philadelphia

Amanda Berberich MD PhD FRCPC
Cert Endo
Assistant Professor
Schulich School of Medicine and Dentistry, Western University
Endocrinologist
Division of Endocrinology & Metabolism
St. Joseph's Hospital

Robert Hegele MD FRCPC Cert Endo FACP FCAHS FAHA
Jacob J. Wolfe Distinguished Medical Research Chair
Martha Blackburn Chair in Cardiovascular Research
Distinguished University Professor of Medicine and Biochemistry
Scientist, Robarts Research Institute
University of Western Ontario

Michael Levine MD ML MACE
Professor Emeritus, Pediatrics and Medicine
Chief Emeritus, Division of Endocrinology and Diabetes
Center for Bone Health

Mark Palmert MD PhD
Division of Endocrinology
The Hospital for Sick Children
Associate Professor of Pediatrics and Physiology, The University of Toronto

Maha Saleh MD FRCPC FCCMG
Co-Section Head, Southwest Ontario Regional Genetics Program
Medical Geneticist and Associate Professor
Genetics and Pediatrics

Jonathan Wasserman MD FRCPC
Staff Physician, Endocrinology
Project Investigator, Genetics and Genome Biology
Hospital for Sick Children
Associate Professor, Paediatrics

Jeanne Webber MHSc MSL MSW RSW
Clinical and Organizational Ethicist
London Health Sciences Centre

Invited CPEN Speakers

Andrea Ens MD MEd FRCPC
CPEG Treasurer
Pediatric Endocrinologist
Children's Hospital London Health Sciences Center
Assistant Professor, Department of Paediatrics
Schulich School of Medicine
Western University

Andrea Gore PhD
Professor and Vacek Chair in Pharmacology
The University of Texas at Austin

Carol King MD FRCSC
Obstetrics & Gynecology, Pediatric & Adolescent Gynecology
London Health Sciences Centre
Assistant Professor, Schulich School of Medicine & Dentistry
Western University

Invited Fellows' Symposium Speakers

Scott Somerville MD FRCPC
Pediatric Endocrinologist
Children's Hospital of Eastern Ontario

Caroline Zuijdwijk MD FRCPC
Assistant Professor, Department of Pediatrics
University of Ottawa
Pediatric Endocrinologist
Children's Hospital of Eastern Ontario (CHEO)
Program Director, Pediatric Endocrinology & Metabolism, University of Ottawa

Invited CPEG Speaker Biographies

Funmbi Babalola

Dr. Funmbi Babalola is a Pediatric Endocrinologist at Children's Hospital, London Health Sciences Centre and an Assistant Professor in the Department of Pediatrics and Epidemiology at The University of Western Ontario. She is an Associate Scientist at Lawson Health Research Institute.

Dr. Babalola joined Children's Hospital, August 2022. She completed her Doctor of Medicine in 2016 at the Northern Ontario School of Medicine. She completed her 3-year general pediatric training at The University of Western Ontario in 2019, and, 3-year pediatric endocrinology fellowship at the Hospital for Sick Children, University of Toronto in 2022. She completed a Master of Science through the Institute of Medical Science and Clinical Investigator Program at the University of Toronto in 2022. Dr Babalola's research areas of interest are diabetes and disorders of calcium and bone.

Ereny Bassilious

Dr. Ereny Bassilious completed medical school at McMaster University, pediatric residency and fellowship in pediatric endocrinology at the Hospital for Sick Children, and Master of Education at the University of Illinois in Chicago. She was program director for the Pediatric Endocrinology Subspecialty Residency program and is currently the Associate Chair of Education in the Department of Pediatrics at McMaster university. Dr. Bassilious has a clinical interest in disorders of sexual differentiation, endo/gyne, and disorders of growth.

Andy Bauer

Dr. Bauer's research interests span the spectrum of thyroid disorders, with a focus on thyroid carcinoma. He serves as executive director of the Child and Adolescent Thyroid Consortium (www.ThyroidCATC.org), co-chair of the ATA pediatric thyroid cancer guidelines, and consultant for ThyCa, the DICER1 registry and the PHTS foundation.

Amanda Berberich

Amanda attended medical school at the State University of New York in Syracuse, NY and completed her Internal medicine and Endocrinology training at Western University in London, Ontario. She has a clinical Endocrinology practice at St. Joseph's Healthcare, is an assistant professor at Western in the Department of Medicine and completed Biochemistry PhD Program focusing on the clinical applications of genetic testing within Endocrinology. Her research interests are in Endocrine genetics and the clinical applications of genetic testing in adult chronic disease. Her published works include those related to inherited lipid and diabetes disorders and have appeared in several journals including Nature Reviews in Endocrinology, Lancet Diabetes and Endocrinology, Current Opinion in Lipidology and the Canadian Journal of Diabetes.

Robert Hegele

Rob Hegele cares for >2800 patients in his lipid clinic at University Hospital in London, Ontario. His laboratory discovered the causal genes for >20 human diseases and also developed the world's first targeted next-generation sequencing panel for dyslipidemias. He is also known for characterizing the polygenic basis of human lipid disorders. He was among the first in the world to use five medications that are now routinely prescribed to treat dyslipidemia or diabetes. He has published 960 papers with >88,000 citations on Google Scholar and is in the top 1% of highly cited scientists in the world. The website Expertscape.com in 2023 ranked him #1 globally in the area of "hypertriglyceridemia" and #2 for "disorders of lipid metabolism". He received the 2019 American Heart

Association Lyman Duff Award, the 2020 FH Foundation Pioneer Award and 2024 US National Lipid Association Virgil Brown Distinguished Achievement Award. He has co-authored many clinical practice guidelines for cholesterol, blood pressure and diabetes. He has trained numerous physicians, medical students and graduate students.

Michael Levine

Dr. Michael A. Levine is Chief Emeritus of Endocrinology and Diabetes at The Children's Hospital of Philadelphia. Dr. Levine holds the Lester Baker Endowed Chair and is Professor Emeritus of Pediatrics and Medicine at the University of Pennsylvania Perelman School of Medicine. Dr. Levine's research has focused on the genetic basis of endocrine signaling defects. His primary clinical interests are endocrine diseases that affect bone and mineral metabolism, particularly osteoporosis, primary hyperparathyroidism, and hypoparathyroidism.

Mark Palmert

Dr. Palmert is a staff endocrinologist at the Hospital for Sick Children (SickKids). He has a long-standing interest in the regulation and disorders of pubertal timing. He has conducted clinical studies of precocious and delayed puberty and in parallel has directed a laboratory-based program designed to identify and understand genetic factors that regulate the onset of puberty.

Maha Saleh

Dr. Maha Saleh is a Physician Geneticist at London Health Sciences Centre. She completed her Medical Genetics Residency at the University of Toronto, SickKids Hospital. She later joined London as staff in 2017 and became an Associate Professor at Schulich School of Medicine, Department of Pediatrics. Dr. Saleh's practice is board and includes prenatal Genetics, both Adult and Pediatric General Genetics as well as Cancer Genetics.

Dr. Saleh is currently Co-Section head of the Southwest Ontario Regional Genetics Program. She is also the Program Director of the Canadian College for Medical Genetics (CCMG) fellowship in London.

Jonathan Wasserman

Dr. Jonathan Wasserman joined the endocrine staff at SickKids in July 2012 after completing a clinical and research fellowship in the Division of Endocrinology. He undertook his medical training at Harvard Medical School and the Massachusetts Institute of Technology and subsequently pursued an internship and residency in paediatrics at Children's Hospital Boston where he also served as a General Paediatric Hospitalist. He previously earned a PhD and completed postdoctoral training in genetics at the University of Cambridge, England.

Jeanne Webber

Jeanne is a Clinical and Organizational Ethicist at London Health Sciences Centre where she provides ethics consultation across three hospitals and a diverse range of programs. A social worker by background, Jeanne supports value-based decision-making grounded in 15 years of clinical experience in hospital and community-based heath care.

Invited CPEN Speaker Biography

Andrea Ens

Andrea Ens has been a pediatric endocrinologist at LHSC Children's Hospital since 2018. She completed her pediatric residency at the IWK Health Centre in Halifax and her pediatric endocrine fellowship at The Hospital for Sick Children in Toronto. During her fellowship, she did a Master's in Education at the University of Toronto. Andrea has co-developed and co-lead the Pediatric Gynecology-Endocrine clinic at LHSC Children's Hospital, gaining invaluable experience working with teens with Polycystic Ovarian Syndrome.

Andrea Gore

Dr. Gore researches mechanisms of how environmental endocrine-disrupting chemicals (EDCs) perturb the developing brain and cause transgenerational epigenetic effects. She has published 4 books and over 200 scientific papers. She was Editor-in-Chief of Endocrinology and was lead author of the Endocrine Society’s two Scientific Statements on EDC.

Carol King

Dr. Carol King is an Obstetrician/Gynecologist in London, Ontario with fellowship training and sub specialization in Pediatric & Adolescent Gynecology. She is the co-lead of the Joint Pediatric Gynecology and Endocrinology Clinic at London Health Sciences Centre.

Invited Fellows’ Symposium Speaker Biographies

Scott Somerville

TBA

Caroline Zuijdwijk

Caroline Zuijdwijk is a pediatric endocrinologist at CHEO and has been the co-lead for the Joint Pediatric Endocrine & Gynecology clinic since 2017. She received her medical degree from McMaster, completed her pediatric residency at Memorial and her Pediatric Endocrinology residency at CHEO, followed by a research fellowship at SickKids.

Awarded Fellowship Listing

1992-1993	M. Lawson
1993-1994	S. Lawrence M. Lawson A. Simone
1994-1995	S. Lawrence S.Taback A. Simone
1995-1996	C. Vaz S.Taback B. Cummings
1996-1997	J. Hamilton E. Sellers B. Cummings
1997-1998	J. Hamilton E. Sellers B. Cummings
1998-1999	J. Curtis J. Hamilton
1999-2000	J. Curtis J. Hamilton
2000-2001	C. Panagiotopoulos C. Huang
2001-2002	C. Panagiotopoulos S. Stock
2002-2003	P Krishnamoorthy P Zimakas R. McEachern
2003-2004	P Krishnamoorthy H. Bui
2004-2005	M. Nakhla J. Simoneau-Roy
2005-2006	M. Nakhla I. Chapados M. Jetha
2007-2008	B. Wicklow T. Pinto, B. Babic J. Deladoey
2008-2009	A.M. Sbrocchi P Olivier T. Pinto
2009-2010	R. Shulman P Olivier T. Edouard S. Runge-Wildi C. Saaman
2010-2011	E. Bassilious J. Wasserman Y. Yeshayahu S. Tsai
2011-2012	M. Millete J. Wasserman C. Zuijdwijk M. Cohen
2012-2013	J. Harrington T. Oron P. Luca M. Nour D. Manousaki
2013-2014	K. Winston C. Leblicq A. Ens B. Hursh I. Rousseau-Nepton
2014-2015	I. Levy D. Manousaki
2015-2016	L. Chiniara S. Basak K. Verbeeten
2016-2017	C. Nugent K. Pundyk N. Coles
2017-2018	C. Nugent S. Fuchs
2018-2019	J. Sorbara
2019-2020	A. Chesover B. Navabi
2020-2021	A. Marr M. Lautatzis J. Ladd H. Geddie (declined)
2021-2022	F. Babalola M. Jiang
2022-2023	K. Oei T. Dyer K. Pabedinskas M. Feldman
2023-2024	J. Stanley S. Rengan
2024-2025	G. Nadeau S. Lenet

Within the last 5 years, the CPEG Fellowship Program was and/or is supported by:
Eli Lilly, EMD Serono, Ipsen, Novo Nordisk, Pfizer, Sandoz, and Ultragenyx.

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	2023	Tracy Dyer
2012	Jennifer Harrington	2018	Marie Eve-Robinson	2024	Joshua Stanley

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	2022	Cheril Clarson	2023	Sarah Lawrence
2019	Denis Daneman	2023	Heather Dean		

Day 1 Agenda (All times are listed in local time)

Fellows' Symposium Thursday, February 6, 2025 (Room: Salon B1)

0830	Fellows Welcome & Breakfast	
	Fellows' Symposium **Chairs:** *Marwa El Masri (Ottawa) & Sruthi Thomas (Vancouver)*	
0850	Polycystic Ovary Syndrome	*Caroline Zuijdwijk*
0950	Coffee Break	
1020	Career/Transition to Practice	*Scott Somerville*
1120	Fellows Closing Remarks	*Marwa El Masri, Sruthi Thomas*

CPEG Program Thursday February 6, 2025 (Room: Salon D)

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

1100	On-Site Registration & Lunch	
1230	Opening Remarks & Thursday Poster Highlights (Odd Numbered Posters)	*Funmbi Babalola, Rebecca Perry*
	Symposium I: Calcium/PG Disorders **Chairs:** *Carol Lam (Toronto) & Anne Marie Sbrocchi (Montreal)*	
1300	Hypoparathyroidism: Clinical Management Including Role of Genetics	*Michael Levine*
1415	Break & Exhibits	
1500	**Poster Viewing I (Odd Numbered Posters)**	
P1	Siblings of Laron Syndrome	*Muath Abu Abah*
P3	Beyond Salt-Wasting CAH: The Broader Differential for a Positive Newborn Screen	*Aliya Allahwala*
P5	Serum Sickness Like Syndrome Due to Methimazole in Graves' Disease	*Alanoud Aman*
P7	Case Report of an Adolescent With Normal Breast Development, Suppressed Gonadotropins, Abnormal Ovaries, and Infantile Uterus	*Tali Baird*
P9	Pediatric Cushing's Syndrome Secondary to Ectopic ACTH Secretion by a Pancreatic Neuroendocrine Tumor: A Case Report	*Gabrielle Doré-Brabant*

Day 1 Agenda (All times are listed in local time)

CPEG Program Thursday February 6, 2025 (Room: Salon D)

P11	TMEM38B Gene Mutation Associated With Osteogenesis Imperfecta	*Abdulrahman Habib*
P13	Examining the Moderators of Diabetes Distress Among Adolescents With Type 1 Diabetes on the Effect of a Mindful-Self Compassion Intervention	*Alanna Jane*
P17	Improving Tools and Clinical Processes to Identify and Address Food Insecurity	*Mara McNeil*
P19	Early Detection and Treatment of Vertebral Fractures, a Sign of Accelerated Senescence in Hutchinson-Gilford Progeria Syndrome, Is Associated With Stabilization of Vertebral Fractures	*Ulrich Montcho*
P21	Weight Stigma in the Pediatric Diabetes Population: Evaluating Patient Level Factors and Diabetes Related Outcomes	*Supraja Rengan*
P23	Diabetes Education During Pediatric Residency Training: Are We Doing Enough?	*Gabrielle Scantlebury*
P25	A Case of Type 1 Diabetes Mellitus Presenting With Hypothyroidism	*Marie St Jacques*
P27	Prevalence and Risk Factors for Bladder and Bowel Dysfunction in Children With Type 1 Diabetes	*Sruthi Thomas*
P29	Echoes of the Past: A Case of Multiple Endocrinopathies Due to Iron Overload	*Abigail Wittenberg*
P31	Unrecognized Social Needs in a Pediatric Diabetes Clinic: Insights From Navigating Social Resources for Children's Health (NSRCH) Study	*Reem Al-Obiade*
	Symposium II: Thyroid **Chairs:** *Jill Hamilton (Toronto) & Danièle Pacaud (Calgary)*	
1530	Thyroid Dysfunction After Iodinated Contrast Media	*Jonathan Wasserman*
1600	Thyroid Cancer in Pediatrics	*Andy Bauer*
1630	Thyroid Tumour Board Discussion	*Jonathan Wasserman, Andy Bauer*
1700	Welcome Reception & Exhibits	
1800	Thyroid Ultrasound in Clinical Care Workshop [pre-registration required - held in Salon B1]	*Jonathan Wasserman, Andy Bauer*
1900	Adjourn for the Day	

Day 2 Agenda (All times are listed in local time)

CPEG Program Friday, February 7, 2025 (Room: Salon D)

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

0730	Registration & Breakfast	
0830	Opening Remarks & Friday Poster Highlights (Even Numbered Posters)	*Rebecca Perry, Funmbi Babalola*
0900	**Oral Abstracts I** **Chairs:** *Beth Cummings (Halifax) & Dina Panagiotopoulos (Vancouver)*	
OR1	A Rare Case of Acromegaly Caused by Ectopic Growth Hormone Releasing Hormone Secretion	*Ali Alghamdi*
OR2	The Anti-Tumoural and Metabolic Bone Effects of High-Dose Denosumab in a Child With an Aggressive, Inoperable Aneurysmal Bone Cyst	*Esraa AlQasim*
OR3	Adrenal Insufficiency and the Maternal Serum Estriol Connection	*Saheba Bajwa*
OR4	Pituitary Hormone Dysfunction and Screening in Children With Congenital Intracranial Structural Midline Anomalies	*Laurence Bastien*
OR5	Remission in Youth Onset Type 2 Diabetes, a Case Series	*Gonzalo Dominguez-Menendez*
OR6	Teaching Adolescents With Type 1 Diabetes Self-Compassion (TADS) to Reduce Diabetes Distress: A Randomized Controlled Trial	*Marwa El Masri*
1030	Break & Exhibits	
	Symposium III: Atypical Congenital Adrenal Hyperplasia (CAH) **Chairs:** *Rose Girgis (Edmonton) & Laura Stewart (Vancouver)*	
1100	3 Challenging CAH Cases	*Funmbi Babalola, Patricia Gallego*
1120	Genetic Review of Atypical CAH	*Maha Saleh*
1155	Ethical Considerations for CAH	*Jeanne Webber*

Day 2 Agenda (All times are listed in local time)

CPEG Program Friday, February 7, 2025 (Room: Salon D)

1230	Poster Viewing II (Even Numbered Posters)	
P2	Severe Hypertriglyceridemia During Chemotherapy for Childhood Acute Lymphoblastic Leukemia	*Nasra Al Jaafari*
P4	Low Bone Turnover and Intravenous Zoledronic Acid Are Insufficient to Prevent Denosumab-Induced-Rebound Hypercalcemia in Duchenne Muscular Dystrophy: A Case Report	*Fahd Alshammri*
P6	The Diagnostic Odyssey of Cyclical Cushing Syndrome: A Case Report	*Mary Zhao*
P8	Treatment of Persistent Hypertension in a Patient With 17-Alpha-Hydroxylase Deficiency	*Laurence Bastien*
P10	Hidden Cause of Fragile Bones: Scurvy-Induced Vertebral Compression Fractures in an Apparent Healthy 11-Year-Old—a Case Report	*Regina Duperval*
P12	An Unlikely Pair-A Presentation of Hypercalcaemia and Hyponatraemia	*Annabelle Hobbs*
P14	Hyperosmolar Hyperglycemic State and Significant Microvascular Complications: A Rare Presentation of Adolescent Type 2 Diabetes	*Alyssa Kahane*
P16	Two Siblings With Laron Syndrome and Their Response to Mecasermin Therapy	*Elise Martin*
P18	The AB(C)s of Thyroid Hormone Resistance	*Mallory McNiven*
P20	6-Month Leuprolide Acetate for Monitoring Central Precocious Puberty Treatment Efficacy	*Rebecca Perry*
P22	Primary Hyperparathyroidism Due to a Single Mutation in CDC73 in a 14-Year-Old Girl: A Case-Report	*Solène Rérat*
P24	A Case of Truly “Idiopathic” Infantile Hypercalcemia – When Calcium Is High but Everything Else Is Low	*Sulafa Sindi*
P26	Not All Salt Wasting Is Congenital Adrenal HYPERplasia: Case Report of Delayed Presentation With NR0B1 Mutation	*Shwetha Suresh*
P28	A Description of ACTH Stimulation Testing Results in Term Neonates	*Sruthi Thomas*
P30	The Genetics of Mild Isolated Neonatal Hyperthyrotropinemia – an Additional Tool to Help Predict Transient vs. Permanent Congenital Hypothyroidism?	*Nicole Yokubynas*
P32	The Causal Role of Endocrine Disrupting Chemicals in Pubertal Timing: A Mendelian Randomization Study	*Melody Zuo*

Day 2 Agenda (All times are listed in local time)

CPEG Program Friday, February 7, 2025 (Room: Salon D)

1300	Lunch & Exhibits	
1400	**Oral Abstracts II** **Chairs:** *Katie Pundyk (Winnipeg) & Mark Inman (Saskatoon)*	
OR7	Assessment of Glycemic Control in Children With Type 1 Diabetes (T1D) Using Full Time Exercise Mode With Automated Insulin Delivery (AID) Systems at Diabetes Camp	*Marwa El Masri*
OR8	The Skeletal Phenotype in Becker Muscular Dystrophy: The Under-Studied Cousin of Duchenne	*Rana Halloun*
OR9	Does Hybrid Closed-Loop System Use Mediate the Relationship Between Social Determinants of Health and Glycemic Outcomes in Pediatric Type 1 Diabetes?	*Nacera Hanzal*
OR10	Radioactive Iodine Therapy in N1b Papillary Thyroid Cancer: Is It Always Necessary?	*Annabelle Hobbs*
OR11	Hypoglycemia During Treatment of Acute Lymphoblastic Leukemia – a Canadian Pediatric Surveillance Program Study	*Mary Jiang*
OR12	Evaluating the Implementation and Safety of a Subcutaneous Insulin Protocol Versus Standard Insulin Infusion for the Management of Mild to Moderate Diabetic Ketoacidosis: A Single-Center Experience	*Emma Metivier*
1530	Break & Exhibits	
	Symposium IV: Diabetes/Lipids **Chairs:** *Sanjukta Basak (Vancouver) & Katherine Morrison (Hamilton)*	
1600	MODY	*Amanda Berberich*
1645	Monogenic Cases of Dyslipidemia Including Familial Hypercholesterolemia	*Robert Hegele*
1730	Adjourn for the Day	
1800	**Conference Social Event [pre-registration required]** Join us for a drink and casual dinner at the Old Court House. Dinner will start at 7:30 PM.	**Old Court House, 399 Ridout St N, London, ON** Located a 14-minute walk from the DoubleTree hotel

Day 2 & 3 Agenda (All times are listed in local time)

CPEN Program Friday, February 7, 2025 (Room: Salon B1)

0900	**CPEN Symposium 1** **Chair:** *Jana Haylor (London)*	
	PCOS: Gynecological and Endocrine Perspective	*Carol King, Andrea Ens*
1030	Rejoin CPEG Group	
1400	**CPEN Symposium 2** **Chair:** *Kristen Langdon (London)*	
	Endocrine Disruptors	*Andrea Gore*
1530	Rejoin CPEG Group	
1600	CPEN Business Meeting [members only]	
1730	Adjourn for the Day	

CPEG Program Saturday, February 8, 2025 (Room: Salon D)

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

0730	AGM Check-In & Breakfast	
0830	CPEG Business Meeting [members only]	
1030	**Poster Viewing III (All Posters)**	
1045	Break & Exhibits	
1100	**Debate** **Chairs:** *Funmbi Babalola (London) & Andrea Ens (London)*	
	Use of GnRH Analogues in Later Early Puberty (6–8 Years)	**Pro:** *Ereny Bassilious* **Con:** *Mark Palmert*
1200	John Bailey Award, CPEG Fellowship Awards, Dan Metzger Distinguished Service Award & Closing Remarks	
1230	Meeting Adjourns	

Oral Abstracts

OR1

A Rare Case of Acromegaly Caused by Ectopic Growth Hormone Releasing Hormone Secretion

Ali Alghamdi(1) and Krista Oei(1)

(1) Department of Paediatrics, Division of Endocrinology, The Hospital for Sick Children, Toronto, ON

A 15-year-old female presented with a five-year history of worsening migraines, 6- months of peripheral vision loss, and 10-months of secondary amenorrhea. She had a significant growth spurt from 10-13 years (crossing from 50th to >97th percentile and well-surpassing her parents), followed by enlargement of her hands, feet, and facial features post-menarche. Physical exam revealed mild coarse facial features, hands and feet appearing larger-for-age, and height 174cm (96%, Z= 1.78) (mid- parental height: 159.5cm). Eye exam revealed left optic disc edema and multifocal choroidal lesions bilaterally.

IGF-1 was elevated (782 (121-606ug/L)) and growth hormone(GH) paradoxically increased after 75g dextrose load (GH Baseline: 77.70ug/L, 3h: 94ug/L; peak glucose: 11.0mmol/L). GH-releasing-hormone(GHRH) was inappropriately elevated 107 (5-18pg/mL). Prolactin was elevated (70.4 (4.2-23ug/L)). Other pituitary labs were unremarkable. Initial MRIs/CTs identified a sellar/suprasellar mass, bilateral choroidal lesions, large left upper lobe mass, extensive cervical and mediastinal lymphadenopathy and vertebral involvement. PET scan showed widespread disease (many Ga-68 DOTATATE-avid lesions) involving aforementioned lesions identified on MRI/CT, many lymph nodes, pancreas, left adrenal gland, dura, and extensive involvement of axial/appendicular skeleton. Biopsy of bronchial lesion suggested a low grade, well-differentiated neuroendocrine tumor (NET), leading to a diagnosis of a presumed lung primary NET secreting GHRH with widespread metastases.

Sellar/suprasellar mass was thought to be pituitary hypertrophy secondary to ectopic GHRH stimulation. Visual field deficits were secondary to retinal detachment due to choroidal lesions.

Genetic testing was negative for FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL, TSC1, TSC2.

Lanreotide was started (90mg increased to 120mg monthly) and well-tolerated. There was minimal improvement in vision following external ocular radiotherapy. 5- month follow-up showed decrease in sellar/suprasellar mass, small improvement in primary lung lesion, qualitative improvement in bony lesions and return of menses with gradual reduction of IGF-1, GH and GHRH. She remains largely asymptomatic aside from prior vision loss.

Adolescent-onset of ectopic GHRH-secreting NETs are extremely rare. To our knowledge, this is the first reported pediatric case of genetic-negative metastatic GHRH secreting NET with a presumed lung primary and highlights unique clinical stability with somatostatin analog alone without surgery or chemotherapy to date.

OR2

The Anti-Tumoural and Metabolic Bone Effects of High-Dose Denosumab in a Child with an Aggressive, Inoperable Aneurysmal Bone Cyst

Esraa AlQasim, Fahd Alshammri, Leanne M. Ward

Division of Endocrinology & Metabolism, Children's Hospital of Eastern Ontario, Ottawa, ON, Canada.

Background: Aneurysmal bone cysts (ABC) are benign osteolytic tumours characterized by locally aggressive, expansile growth. RANKL expression is increased in ABC, which contributes to tumoural growth and causes excessive bone resorption. Denosumab, a monoclonal antibody targeting RANKL, effectively halts osteoclast activity, promoting peri- and within-tumour ABC ossification. Denosumab demonstrates efficacy in reducing tumour size, facilitating bone reconstitution, and improving clinical outcomes in pediatric ABC. However, denosumab also causes rebound hypercalcemia both during treatment and following discontinuation.

Intravenous zoledronic acid (IV ZA) during, or immediately after, denosumab tapering is a recommended strategy to prevent rebound hypercalcemia. We describe a young girl with an inoperable ABC who experienced tumoural regression on denosumab but who developed post-denosumab rebound hypercalcemia despite prophylactic IV ZA administration.

Case description: Denosumab was started on a 4-year-old girl with a surgically unresectable ABC at L5 to achieve tumoural regression and prevent neurological sequelae. She received five weekly doses of 70 mg/m^2, followed by monthly administration for 14 months (with gradual tapering over the last 6 months). This led to complete resolution of her regional bone pain and significant reductions in ABC dimensions. Her lumbar spine areal BMD Z-score (LSaBMDZ) increased significantly during treatment, as expected (-1.2 at baseline to +3.1 at completion of treatment). One month following her last planned denosumab dose, she received IV ZA (0.0125 mg/kg) in an effort to prevent the rebound phenomenon. Two and half months following the single prophylactic IV ZA dose, she developed asymptomatic hypercalcemia (ionized calcium 1.76 mmol/L, normal: 1.16-1.36) requiring three additional IV bisphosphonate doses to achieve eucalcemia. Nine months following the rebound hypercalcemia, her LSaBMDZ dropped to +2.7 (height Z-score +1.73) and she remained eucalcemic with stable, reduced ABC dimensions.

Conclusions: We highlight the positive treatment effect of denosumab on an inoperable L5 ABC, plus rebound hypercalcemia post-denosumab discontinuation despite prophylactic IV ZA therapy. Although the precise etiology of denosumab- induced rebound hypercalcemia remains unclear, we postulate that supraphysiological increases in LSaBMDZ may predict the rebound risk, an hypothesis which requires further testing. Careful monitoring and treatment strategies are needed to successfully manage this serious complication of denosumab therapy.

OR3

Adrenal Insufficiency and the Maternal Serum Estriol Connection

Saheba Bajwa (1), Laura Stewart (1,2), Carolina Silva (1,2).

(1) Division of Endocrinology, Department of Pediatrics, BC Children's Hospital, Vancouver BC. (2) Faculty of Medicine, University of British Columbia, Vancouver BC.

Adrenal insufficiency is a serious and potentially fatal condition, particularly in the newborn period when infants can be completely asymptomatic prior to the occurrence of an adrenal crisis. Maternal serum screening aims to identify pregnancies with higher risk of aneuploidies or neural tube defects. This includes measurement of estriol, which derives from the placental aromatization of fetal adrenal androgens. Isolated low maternal serum estriol levels should raise suspicion for deficient fetal steroidogenesis, however, to-date, recommendations for follow-up are variable. We have reviewed three cases of newborn infants with adrenal insufficiency born to mothers with low serum estriol, detected prenatally.

The patients in all three cases were term, male infants, ranging from 6 to 9 days of age at presentation. All three patients had normal male genitalia. The first case presented with dehydration and failure to thrive. The other two cases presented completely asymptomatically, with hyponatremia and hyperkalemia on bloodwork done at one week of life to monitor for electrolyte disturbances due to the finding of low maternal estriol prenatally. All three patients underwent stimulated cortisol testing and had suboptimal responses. Treatment with maintenance glucocorticoid therapy, as well as fludrocortisone was initiated. In all cases, the patients responded well to treatment, as evidenced by clinical improvement and resolution of electrolyte disturbances. In the first two cases, the etiology of adrenal insufficiency was found to be secondary to congenital adrenal hypoplasia, due to pathologic variants in the DAX1 gene. The etiology of the adrenal insufficiency in the third and most recent case, is still under investigation, with genetic testing pending.

This case series demonstrates an association of low maternal serum estriol levels and adrenal insufficiency in the newborn. In each of these cases, close follow up, prompt investigation and management were paramount to the positive outcomes. This case series highlights the need for clear clinical protocols for monitoring newborns with a history of low serum maternal estriol. To inform such recommendations, larger scale studies are needed to assess the frequency of this association.

OR4

Pituitary Hormone Dysfunction and Screening in Children with Congenital Intracranial Structural Midline Anomalies

Laurence Bastien (1), Caroline Zuijdwijk (1,2), Nicholas Mitsakakis (3), Alexandra Ahmet (1,2)

(1) Division of Endocrinology and Metabolism, Children's Hospital of Eastern Ontario, Ottawa, ON

(2) Department of Pediatrics, Faculty of Medicine, University of Ottawa, Ottawa, ON

(3) Children's Hospital of Eastern Ontario Research Institute, Ottawa, ON

Background: Pituitary hormone dysfunction (PHD) is a common finding in children with congenital intracranial midline anomalies; however, the degree of risk associated with specific anomalies (e.g., corpus callosum agenesis, absent septum pellucidum) is not well understood, especially when there is a structurally normal pituitary gland. As a result, screening guidelines are lacking. Early detection of PHD reduces morbidity and mortality, however frequent and unnecessary medical visits and screening may lead to higher family and healthcare burden.

Objective: We aim to determine the incidence, type, and age at diagnosis of PHD, in children and adolescents with congenital intracranial midline anomalies, including isolated or multiple congenital midline anomalies, congenital pituitary anomalies, and congenital hypothalamic anomalies, followed at the Children's Hospital of Eastern Ontario (CHEO). We also aim to describe the frequency and type of screening done in this population.

Methods: Retrospective chart review of children (0-18 years) followed at CHEO between July 1st 2014 and July 1st 2024 with a congenital intracranial midline anomaly was conducted. Children with transient, acquired, or non-specific anomalies were excluded. Data collection included: (1) patient characteristics; (2) midline anomaly diagnosis; (3) endocrinology involvement; (4) pituitary screening – type, age, and frequency; (5) PHD diagnosis – type(s), associated symptoms, and age(s) of diagnosis; and (6) treatment of PHD.

Results: During the study period, 421 children were followed at CHEO with a congenital intracranial midline anomaly. Of these, 282 (67%) were followed in endocrinology for pituitary screening and 98 (34%) of those followed for screening were diagnosed with PHD(s). In children who had >1 visit with endocrinology (91%), average follow-up duration was 5.9 years.

Conclusion: Our study demonstrates that children and adolescents with congenital intracranial midline anomalies are at risk of PHD. Further analysis is planned to evaluate the association between patient characteristics, type of midline anomaly, and type and timing of PHD diagnosis; these results will be available to be presented at CPEG 2025. We anticipate that through a better understanding of risk factors for and timing of PHD with various midline anomalies, our study will help to inform the development of a structured approach to screening at-risk children.

OR5

Remission in Youth Onset Type 2 diabetes, a case series

Gonzalo Dominguez-Menendez (1), Rachel Dun (1), Justin Ma (1), Sanjukta Basak (1).

(1) Department of Pediatrics, BC Children's Hospital, University of British Columbia, BC, Canada.

Background: Youth onset type 2 diabetes (T2D) is described as having an aggressive clinical course with higher rates of progression of beta-cell dysfunction, earlier onset and a higher number of complications despite intensive intervention. Diabetes remission has been proposed in adult-onset T2D, defined as a normal glycemia level (HbA1c <6.5%) without anti-hyperglycemic medications for at least 3 months as per Diabetes Canada guidelines. The most significant contributors to remission were sustained weight loss and shorter disease duration. Our aim is to describe a case series of youth-onset T2D remission followed in the BC Children's Hospital dedicated Insulin resistance/Type 2 Diabetes program.

Case series: We describe 6 youth (2 female) with median age of diagnosis 13.0 years (range 12.2-15.1 years), HbA1C at diagnosis 8.1% (6.6 – 13.7%), baseline BMI Z-score 2.6 (+1.90 – +5.81) with initial diagnosis in 2021-2022. All youth were treated as per the ISPAD 2022 guidelines with intensive lifestyle education, and 2 cases requiring metformin therapy and basal and bolus insulin therapy for 3-6 months. Median time of follow-up was 1.4 years (0.3 to 2.4), with an average of 1.7 visits per year. All individuals increased physical activity, decreased calorie-rich food and increased fibre intake. HbA1c reduced to -4.4% (-0.5- -7.6%) within 3 months to prediabetes level (5.8-6.1%), and -2.4% (range -7.3 to -0.8%) within 12 months follow-up. The median BMI Z-score decreased in 12 months was -0.9 SD (-0.5 to -3.8). One case relapsed after 18 months in remission.

Discussion: T2D has been traditionally viewed as a life-long condition, emerging cases demonstrates that remission of the disease is possible in youth, as seen in the adult population, incorporating early interventions, including physical activity and dietary modifications.

OR6

Teaching Adolescents with type 1 Diabetes Self-compassion (TADS) to reduce diabetes distress: A randomized controlled trial

Marwa El Masri (1), Saunya Dover (2), Adam Khalif (2), Karen Bluth (3), Corien Peeters (4), Andrew Leonard (5), Alexandra Ahmet (1,2,6), Ellen B Goldbloom (1,2,6), Karine Khatchadourian (1,2,6), Caroline Zuijdwijk (1,2,6), Sarah Lawrence (1,2,6), Gary S Goldfield (1,6), Sarah Hamilton (1), Omar Imran (1), Yongdong Ouyang (7), Arlene Jiang (7), Anna Heath (7), Brian M Feldman (8,9), Kuan Liu (8), Jai Shah (10,11), Marie-Eve Robinson (1,2,6)

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Objective: Diabetes distress is defined as the emotional burden and stress associated with managing diabetes. Youth with type 1 diabetes (T1D) have increased levels of diabetes distress, underscoring the need for targeted interventions to improve their psychological wellbeing. Mindful Self-Compassion (MSC) is a psychological intervention that combines mindfulness practices and self- compassion techniques. This study evaluates the effectiveness of the virtual MSC for Teens (MSC-T) intervention in reducing diabetes distress among youth with T1D.

Methods: We conducted a single-center parallel-group randomized controlled trial among adolescents aged 12-17 years with T1D in Ontario, Canada. Participants were assigned to either the virtual 8-week MSC-T program or standard of care. Our primary outcome was diabetes distress as measured through the Problem Areas in Diabetes-Teen (PAID-T) at 3-months post-baseline. The PAID-T calculates an overall distress score of 26-156. Changes of more than 7 points are considered clinically meaningful. This was analyzed using a linear regression model adjusted for age and baseline diabetes distress.

Results: 141 youth completed the study (70 in the MSC-T group and 71 in the control group). Mean age was 15.2 ± 1.7 years for MSC-T and 15.1 ± 1.4 years for controls. Baseline mean PAID-T scores were 74 ± 27 in MSC-T and 72 ± 25 in controls. The intention to treat analysis indicated a significant effect of MSC-T in reducing PAID-T raw scores at 3 months (β [regression coefficient] -12.33; 95% confidence interval [CI] -18.54 to -6.11; $P < 0.001$). The multiple imputation analysis to account for missing data showed maintenance of a significant effect for the intervention (β -12.03; 95% CI -18.33 to -5.74; $P < 0.001$).

Conclusion: MSC-T reduced diabetes distress in youth with T1D at 3 months post- baseline in a clinically and statistically significant manner compared to standard of care. This finding underscores the potential of MSC-T as an intervention that addresses the emotional burden associated with T1D. The remainder of our analysis will evaluate the impact of MSC-T on anxiety, depressive symptoms, disordered eating, suicidal ideation, and metabolic outcomes.

OR7

Assessment of Glycemic Control in Children with Type 1 Diabetes (T1D) Using Full Time Exercise Mode with Automated Insulin Delivery (AID) Systems at Diabetes Camp

Marwa El Masri (1), Mary Jiang (1), Scott Somerville (1), Sarah Lawrence (1)

(1) Division of Endocrinology & Metabolism, Children's Hospital of Eastern Ontario, Ottawa, ON, Canada

Background: Diabetes camp provides a chance for youth with T1D to enjoy traditional camp activities while receiving support with diabetes management. The changes in activity levels typically require a reduction in insulin dosing; through both basal and insulin to carbohydrate-to-insulin ratio (ICR) adjustments. Empiric adjustments need to be adapted for the use of AID pumps. In 2024, Camp Banting adopted a new protocol of maintaining home basal settings for AID pumps and only changing ICRs while putting pumps in exercise mode (EM) for the duration of camp. This study aims to evaluate changes in glycemic control and analyze basal and bolus insulin delivery to inform future camp recommendations.

Methods: Retrospective review of 2023 and 2024 Camp Banting campers in Ottawa, Ontario on AID pumps. Data from continuous glucose monitoring (CGM) and pump uploads was collected from the week before and during camp. Glycemic control was assessed by comparing mean time in range (TIR) and time below range (TBR) between these periods. TBR was compared for 2023 and 2024 in AID users.

Results: Of 48 participants (21 males, 27 females), all had CGM data, and 37 (71%) had pump upload dataavailable. Mean age was 12.2 ± 2.8 years. For campers with GMI ≥ 7.5, TIR improved significantly from 49.9 ± 10.8% precamp to 66.5 ± 13.5% during camp ($P<0.001$), without a significant increase in TBR (1.5 ± 2.2% to 2.6 ± 3.5%, $P=0.09$). Daily basal insulin delivery decreased 16.6% and bolus increased 19.9% during the camp with a significant increase in carbohydrate (CHO) entries ($p=0.045$). Campers with GMI < 7.5% showed no significant TIR change (72.3 ± 8.6 to 75.5 ± 8.4, $P=0.2$) with basal insulin delivery decreased by 10.6% and bolus increased by 8.2%. There was no difference in the TBR between 2023 (without full time EM) and 2024 (with full time EM).

Conclusion: Using home basal rates, with full time exercise mode in AID pump users, improved TIR without a significant rise in hypoglycemia. The increase in total daily bolus, even with weaker ICR, likely reflects more consistent CHO entries, especially among those with a higher baseline GMI.

OR 8

The Skeletal Phenotype in Becker Muscular Dystrophy: The Under-Studied Cousin of Duchenne

Rana Halloun (1,2), Stefan Jackowski (1), Maya Scharke (1), Jinhui Ma (1, 3), Ken Gaither (4), Thomas Fuerst (4), Hugh McMillan (1,2), James MacDougall (5), Joanne Donovan (5), Leanne Ward (1,2) for the CANYON Investigators

(1) The Ottawa Pediatric Bone Health Research Group, Ottawa, Ontario, Canada; (2) University of Ottawa, Ottawa, Ontario, Canada; (3) McMaster University, Hamilton, Ontario, Canada; (4) Clario, Philadelphia, Pennsylvania, USA; (5) Edgewise Therapeutics, Boulder, Colorado, USA

Background: Becker muscular dystrophy (Becker) is an X-linked, progressive myopathy caused by pathogenic, usually in-frame, variants in the *DMD* (dystrophin) gene. Contraction-induced injury leads to replacement of muscle by fat and fibrosis, with progressive loss of ambulation. To date, information has been scarce on the skeletal phenotype in Becker, although low-trauma fractures are a feature.

Aim: To describe the multi-site bone mineral density (BMD) phenotype by DXA in a large cohort of adolescents and adults with Becker, and to determine factors associated with BMD reductions.

Methods: In this cross-sectional study of 66 individuals with Becker, ambulatory at baseline (pre-treatment), were enrolled in a multicenter phase 2 clinical trial (NCT05291091). Participants underwent areal BMD (aBMD) at the lumbar spine (LS1-4), proximal (total) hip and total body (TB); TB lean mass was also captured. aBMD results were converted to age- and sex-matched Z-scores; lean mass was expressed as the lean mass index (LMI, weight in kg/height in m2). LMI and muscle function tests including the North Star Ambulatory Assessment (NSAA) and Timed Up and Go (TUG, seconds) were studied for their independent relationships with aBMD Z-scores.

Results: Mean age was 24.6 ± 11 years (range 12 to 50). The aBMD Z-score was lowest at the total hip (-1.7 ± 1.1), followed by the TB (-1.5 ± SD 1.2) and highest at the LS (-0.3 ± SD 1.3). A multiple linear regression model adjusted for height and weight Z-scores showed that low LMI was associated with low total hip aBMD Z-score (β=0.2, 95% CI 0.04 to 0.33). Adjusting for NSAA or TUG did not improve the model. In a multiple linear regression model, adjusted for height and weight Z scores, higher LMI was highly correlated with higher NSAA score (β=3.66, 95% CI 2.66 to 4.66) and shorter TUG (β= -1.05, 95% CI -1.57 to -0.52).

Conclusions: In this largest Becker cohort, we show reductions in multi-site aBMD Z-scores that were present despite retained ambulation, and that LMI was independently associated with low total hip aBMD Z-scores (the most affected skeletal site). In addition, LMI was highly correlated with muscle function, suggesting LMI may be a surrogate for muscle strength in the dystrophinopathy setting. These findings underscore the importance of routine bone health surveillance in Becker despite preserved ambulation.

OR 9

Does Hybrid Closed-Loop System Use Mediate the Relationship Between Social Determinants of Health and Glycemic Outcomes in Pediatric Type 1 Diabetes?

Nacera Hanzal (1), Alexandra Ahmet (1-3), Ellen Goldbloom (1-3), Nicholas Mitsakakis (3), Ewa Sucha (3), Caroline Zuijdwijk (1-3)

(1) Department of Pediatrics, Faculty of Medicine, University of Ottawa, Ottawa, ON; (2) Department of Endocrinology and Metabolism, Children's Hospital of Eastern Ontario, Ottawa, ON; (3) Children's Hospital of Eastern Ontario Research Institute, Ottawa, ON

Background: Sociodemographic disparities continue to impact people living with type 1 diabetes (T1D), despite substantial diabetes technological advancements like hybrid closed-loop systems (HCL), which improve glycemic outcomes and quality of life. Funding for diabetes technology differs widely between health plans and provinces, which may result in inequitable access to HCL systems.

Objective: To evaluate the mediating role of HCL systems on the relationship between social determinants of health (SDH) and glycemic outcomes in children with T1D.

Design and Method: A retrospective chart review was conducted for children with T1D who attended a physician follow-up visit at the Children's Hospital of Eastern Ontario from May 1, 2023 to May 1, 2024. Glycemic outcomes were measured using hemoglobin A1c (HbA1c) and percent time in range (%TIR). The Canadian Index of Multiple Deprivation (CIMD) measured SDH by postal code. Regression models were used to describe mediation effects.

Results: Data were obtained for 581 patients with mean age 13.4±3.8 years (46.6% female), mean T1D duration 6.1±3.8 years and mean HbA1c 8.0±1.5 mmol/L at baseline. 91% used CGM (76% Dexcom) and 67% were on insulin pump, with 69% of those (46% of all patients) using HCL systems. We found an association between HbA1c and the CIMD dimensions of situational vulnerability (b 0.27, 95%CI 0.15, 0.38, $p<0.001$) and residential instability (b 0.39, 95%CI 0.28, 0.58, $p<0.001$), but no mediating effect of HCL system use on this relationship. There was no association found between HbA1c and CIMD dimensions of economic dependency or ethnocultural composition. Results were consistent when this analysis was repeated using %TIR for the glycemic outcome.

Conclusion: Although residential instability and situational vulnerability were associated with glycemic outcomes, these relationships were not mediated by HCL system use in our clinic. We found no association between glycemic outcomes and economic dependency or ethnocultural composition. In the last few years, our centre has prioritized equitable access to HCL for patients of all socioeconomic backgrounds regardless of baseline glycemic control; this may in part explain our findings. Further analysis will be performed to better understand these relationships prior to CPEG 2025.

OR10

Radioactive iodine therapy in N1b papillary thyroid cancer: is it always necessary?

Annabelle Hobbs (1), Jonathan Wasserman (1), Child and Adolescent Thyroid Consortium

(1) Division of Endocrinology, The Hospital for Sick Children and Department of Paediatrics, University of Toronto

Introduction: Papillary thyroid cancer (PTC) constitutes up 90% of thyroid cancer in children and is associated with higher rates of locoregional and distant metastasis than in adults. Despite this, long term prognosis is excellent and 30-year disease-specific survival exceeds 95%.

Given these favourable outcomes, debate persists regarding the optimal approach to treatment and the balance between disease control and adverse effects of therapies such as radioactive iodine (RAI). We reviewed outcomes of children with N1b disease (metastasis to unilateral, bilateral or contralateral cervical levels I,II, III, IV or V or retropharyngeal or superior mediastinal lymph nodes) who underwent RAI vs those who did not.

Methods: Data were collected from the Child and Adolescent Thyroid Consortium, an international, multi-site registry of children treated for differentiated thyroid cancer. A retrospective review was conducted on 746 pediatric patients diagnosed with PTC and treated between 2010 and 2020. Those with a minimum 6 months follow-up were included for analysis.

Results: 288 children with N1b disease were identified, and 103 excluded due to known distant metastases. Of the remainder, 168/185 (91%) underwent RAI, with a median of 1 treatment (1-4) and a median cumulative activity of 102.5mCi (21-405.5), while 17 were not treated.

The median age of patients was 16.4 (6.4-18.7) years for the non-RAI group, versus 14.6 years (8.4-18.7) in the RAI group. Most patients had classic subtype PTC (57%).

Among those untreated with RAI, 15/17 (88%) demonstrated excellent response to therapy at final follow-up, without additional intervention (median follow up 4.1 years, range 0.57-9.6), versus 66/164 (40%) in the RAI-treated group. Of the two who did not attain excellent response, one had an elevated TSH-stimulated thyroglobulin level 4.5 years post surgery (without structural correlate) and the second had persistently elevated Tg antibodies after 4 years.

Discussion: Radioactive iodine remains standard therapy for most children with PTC and N1b disease. Nonetheless, our review demonstrates that a select subgroup of children may achieve an excellent response to therapy without RAI, despite N1b disease.

Further study is required to identify which children warrant more aggressive therapy, and to define longer-term outcomes for this population.

OR11

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

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Background: An important but under-recognized adverse drug reaction (ADR) associated with acute lymphoblastic leukemia (ALL) treatment is hypoglycemia, which can lead to significant morbidity. There are reports of hypoglycemia associated with asparaginase and 6-mercaptopurine (6MP), which are commonly used in ALL protocols. The association, mechanism, and temporal relationship between these agents and hypoglycemic events are not well described. A national Canadian Pediatric Surveillance Program (CPSP) study was completed to further understand this rare ADR.

Methods: Through CPSP methodology, a network of pediatricians were asked monthly to report patients less than 18-years-old with a first known episode of biochemically proven hypoglycemia (<3mmol/L) during ALL therapy from October 2022–September 2024. Descriptive analysis was completed for demographics, timing of onset and duration of hypoglycemia, and management.

Results: Fourteen patients met criteria prior to abstract submission. Median age was 5.51(IQR 2.74, 7.53) years. Median BMI percentile was 44%(29, 95). Six patients were female (43%). Median initial blood glucose value was 2.60(2.30, 2.90) mmol/L. Seven (50%) patients had symptoms including altered level of consciousness, seizure, tremor/jitteriness, diaphoresis, hunger, fatigue, dizziness, and nausea. Seven patients had hypoglycemia within 40 days of PEG-asparaginase exposure; onset ranged from five to 38 days (median 16) after most recent exposure. Of those with a critical sample (3/6), 100% were hypoketotic. Median venous glucose level was 3.1(2.95, 3.1) mmol/L and median insulin level was 7.0(5.5, 23) pmol/L. Six patients had hypoglycemia within seven days of 6MP completion. Onset of hypoglycemia ranged from 2-697 days (median 26) from initiation of the most recent 6MP course. One patient had a critical sample and was ketotic. One patient had hypoglycemia following exposure to both PEG-asparaginase and 6MP. Thirteen (93%) patients needed treatment for hypoglycemia. Median duration of treatment was two days (range 1-8 days) with six of thirteen (46%) patients requiring ongoing treatment at the time of reporting.

Discussion: Although the frequency of hypoglycemia was rare, a significant number of patients were symptomatic and almost all required treatment. This study highlights the need for increased awareness of this potential treatment complication and provides a better understanding of mechanism to inform treatment.

OR12

Evaluating the Implementation and Safety of a Subcutaneous Insulin Protocol Versus Standard Insulin Infusion for the Management of Mild to Moderate Diabetic Ketoacidosis: A Single-Center Experience

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Background: Intravenous (IV) insulin therapy is the standard-of-care for managing pediatric diabetic ketoacidosis (DKA). Subcutaneous (SC) insulin is an alternative for the management of mild-to-moderate DKA when IV insulin infusions are not feasible. Studies have shown SC protocols used in this context are safe, as effective as IV insulin, and resource-efficient. A quality improvement project at the Hospital for Sick Children (HSC) sought to develop and pilot a SC Insulin Protocol for the management of uncomplicated mild-to-moderate DKA.

Aim: Describe the patient and clinical characteristics treated with SC and IV insulin, rate of uptake and safety. We aimed to safely promote better resource utilization and decrease time-to-correction and time-to-transition with implementation of a SC protocol.

Methods: The protocol was developed based on ISPAD guidelines using SC insulin lispro or aspart, 0.15 U/kg, every 2 hours. It was implemented in December 2022 for children 5-18 years. Protocol use was subject to physician discretion. Division-wide education and weekly discussion of adverse events was performed. We conducted a retrospective chart review of all children 5-18 years old who presented to HSC in mild-to-moderate DKA between January-December 2023.

Results: 26 patients (pre-existing type 1 diabetes(T1DM): n=7, 26.9%), presented in mild-to-moderate DKA in 2023. SC Protocol: 46.1% (n=12), IV insulin: 53.8% (n=14). Both groups had similar baseline characteristics (age, sex, pre-existing T1DM, initial pH), despite choice of protocol being subject to physician discretion. Patients treated with the SC protocol corrected and transitioned significantly faster. Median time-to- correction, 4.9hrs (IQR 1.3hrs) versus 6.7hrs (IQR 2.2hrs) in the IV group (p=*0.027)*. Median time-to-transition in the SC group 8.5hrs (IQR 2.3hrs) versus 14.4hrs (IQR 5.5hrs) in the IV insulin group (p<0.001). There were no severe adverse events.

Conclusions: Implementation of a SC protocol for managing uncomplicated mild-to- moderate DKA in a tertiary pediatric hospital was successful, safe and resulted in faster DKA correction and transition. Testing further protocol iterations in community hospitals who manage a large proportion of patients presenting in mild-moderate DKA is warranted. The SC protocol could allow for better cost-effective resource utilization and reduce need for patient transfers to tertiary centers, particularly in resource-limited settings.

Poster Abstracts

P1

Siblings of Laron Syndrome

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This abstract details the case of a 12-year-old male with severe short stature, ultimately diagnosed with Laron Syndrome, a rare genetic disorder characterized by growth hormone (GH) insensitivity. Initially managed in family medicine for subclinical hypothyroidism, the patient presented to the endocrine clinic with marked short stature (height: 116.3 cm, >-5 SD below the mean), delayed bone age, a history of transient neonatal hypoglycemia, recurrent fractures, poor appetite, and additional findings, including facial dysmorphism and an undescended testis.

Despite an initial GH therapy trial prompted by low IGF-1 and IGF Binding Protein-3 levels, the patient showed inadequate growth response, raising suspicion for GH insensitivity.

Further investigations, including genetic testing, confirmed the diagnosis of Laron Syndrome, an autosomal recessive disorder caused by mutations in the GH receptor gene, which disrupts GH signaling and results in primary IGF-1 deficiency. As part of the management, recombinant IGF-1 therapy (Increlex) was initiated and gradually increased to therapeutic levels to promote growth while carefully monitoring for hypoglycemia, a known risk with IGF-1 treatment.

This case underscores the need for heightened clinical awareness of Laron Syndrome, especially in children exhibiting features resembling GH deficiency but who demonstrate resistance to GH therapy. Early recognition and appropriate diagnostic measures are essential to distinguish Laron Syndrome from more common causes of short stature and ensure timely intervention. The presentation also highlights the clinical nuances in managing GH insensitivity, emphasizing the importance of individualized treatment and close monitoring in optimizing patient outcomes.

P2

Severe Hypertriglyceridemia During Chemotherapy for Childhood Acute Lymphoblastic Leukemia

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Abstract: Severe hypertriglyceridemia (HTG) is a rare but serious complication that can arise in pediatric patients undergoing chemotherapy for acute lymphoblastic leukemia (ALL). This case describes a 15-year-old male with high-risk T-cell ALL who developed severe HTG during induction chemotherapy, particularly after receiving PEG-asparaginase and dexamethasone. Although he did not experience typical HTG symptoms like abdominal pain or visual disturbances, he presented at a routine follow-up with lethargy, poor appetite, and significant weight loss, leading to his hospitalization.

Case Presentation: Eighteen days into chemotherapy, the patient showed signs of fatigue, decreased energy, weight loss, and poor appetite. Laboratory tests confirmed hypertriglyceridemia, abnormal liver enzymes, and coagulopathy. Notably, he lacked symptoms like nausea, vomiting, or headaches, which are commonly associated with high triglyceride levels. The absence of a family history of dyslipidemia or other metabolic conditions, along with normal glucose levels, suggested that his HTG was likely induced by chemotherapy.

Clinical Course and Management: Following his admission, management efforts centered on mitigating the severe HTG and associated risks. Given the high triglyceride levels and potential pancreatitis risk, a multidisciplinary approach was implemented, which included careful monitoring and nutritional support to address his weight loss. Lipid-lowering therapies were considered cautiously due to his fragile health status.

Discussion: This case illustrates the impact of PEG-asparaginase and dexamethasone on lipid metabolism, with asparaginase increasing VLDL production and triglyceride levels.

Currently, HTG in pediatric oncology is often managed reactively, with no standardized guidelines to prevent or manage severe cases proactively.

Conclusion: This case highlights the urgent need for evidence-based guidelines to manage HTG in pediatric oncology patients. Treatment practices remain inconsistent, often relying on anecdotal experiences, underscoring the importance of formal management protocols. Establishing comprehensive guidelines would facilitate early detection and consistent management, improving patient outcomes and reducing the risk of severe complications. Further research into dietary, pharmacologic, and supportive interventions is essential for developing standardized care approaches for HTG in pediatric chemotherapy settings.

P3

Beyond Salt-wasting CAH: The Broader Differential for a Positive Newborn Screen

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Background: Cytochrome P450 oxidoreductase (POR) plays a key role in steroidogenesis. POR mutations can cause a broad range of phenotypic effects, including a rare form of congenital adrenal hyperplasia that is not included in the newborn screen (NBS), genital anomalies, and in utero androgen excess leading to maternal virilization. Some may also have skeletal abnormalities. We report two siblings with POR deficiency, born to non-consanguineous Japanese parents.

Case 1 (older sibling, now 6 years old): 3-month-old male with birthweight

<10th percentile and normoglycemia referred for microphallus. Examination revealed midface hypoplasia, flattened nasal bridge, small ears, radio-humeral synostosis, bifid scrotum, a high penoscrotal junction, and later diagnosed with moderate-severe bilateral hearing loss. Pituitary screening revealed low cortisol, prompting an ACTH stimulation test.

Investigations:

NBS: Negative; **ACTH stimulation:** Peak cortisol = 191 nmol/L, stimulated 17-OHP = 43.8 nmol/L (0.1-3.4); **Testosterone:** 7.4 nmol/L (0.3-10.4); **Androstenedione:** 0.2 nmol/L (0.0-2.1); **DHEA-sulphate:** 1.7 umol/L (0.7-15.6); **Genetic testing:** Antley-Bixler syndrome due to autosomal recessive POR mutation inherited from both parents

Case 2 (younger sibling): 11-day-old male referred following positive NBS for CAH. Mom experienced voice deepening and acne during pregnancy, but antenatal ultrasounds were unremarkable. He had facial dysmorphisms, a microphallus and normoglycemia. Genetic testing for POR deficiency was sent due to the clinical presentation and sibling's history.

Investigations:

NBS: 17-OHP = 26.8 (cutoff 10.0), (17OHP + 4A)/Cortisol = 1.17 (cutoff 0.33); **ACTH stimulation:** Peak cortisol = 236 nmol/L, stimulated 17-OHP = 32.9 nmol/L (0.0-4.8); **Testosterone:** 3.2 nmol/L (0.3-6.4); **Androstenedione:** 1.3 nmol/L (0.0-2.5); **Genetic testing:** pending

Both siblings were prescribed stress-dose hydrocortisone but not started on maintenance hydrocortisone. Neither had concern for salt-wasting given normal sodium, potassium, renin, and blood pressures.

Discussion: NBS is designed to detect life-threatening salt-wasting CAH but can occasionally yield positive results in other cases, often due to mild 17-OHP elevations. In case 2, the positive NBS prompted further investigations, and the diagnosis was facilitated by the sibling's history of Antley-Bixler syndrome and POR deficiency. This underscores that while NBS is not intended to detect such conditions, it may occasionally flag cases where further investigation reveals other underlying conditions.

P4

Low Bone Turnover and Intravenous Zoledronic Acid are Insufficient to Prevent Denosumab-Induced-Rebound Hypercalcemia in Duchenne Muscular Dystrophy: A Case Report

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Background: Denosumab, a monoclonal antibody targeting RANK-L, is a potent anti-resorptive inhibiting osteoclast formation. It is an attractive osteoporosis therapy in children due to its sub-cutaneous route of administration and lack of first exposure adverse effects. However, denosumab is also associated with "rebound hypercalcemia", a phenomenon which can occur following cessation, but also while actively receiving denosumab. Children appear particularly prone to the rebound phenomenon, hypothesized to result from the higher (than adult) bone turnover inherent to the juvenile skeleton. However, we recently observed hypercalcemia in a boy with Duchenne muscular dystrophy (DMD) on denosumab for seven years, highlighting that low bone turnover, combined with prophylactic intravenous zoledronic acid (IV-ZA), are insufficient to prevent this serious denosumab-related complication.

Case Description: A 17-year-old male with DMD and glucocorticoid (GC)-induced osteoporosis began denosumab at age 9. Initially enrolled in a two-year clinical trial, he received denosumab every 4-6 months over seven years, followed by IV-ZA (the latter, prescribed in response to declining lumbar spine bone mineral density Z- scores). As a post-denosumab hypercalcemia prevention measure, oral risedronate was started immediately after the last denosumab dose, followed by IV-ZA five months later. However, 11 months post-denosumab and 6 months after a single IV- ZA dose (0.0125 mg/kg), he developed asymptomatic hypercalcemia (serum ionized calcium 1.65 mmol/L; normal: 1.16-1.36), an increase in serum c-telopeptide of type I collagen (CTX) Z-score (from -3.1 at baseline to -0.5 at the time of hypercalcemia), with rising urinary calcium excretion and a suppressed PTH. The hypercalcemia caused acute kidney injury (with a 140% increase in serum creatinine from baseline), necessitating aggressive hydration and 2 doses of IV pamidronate to achieve eucalcemia. At the time of the hypercalcemia, he also sustained a right forearm fracture. Four months after the last dose of pamidronate given to restore eucalcemia, he remains on IV bisphosphonate therapy to treat his high-risk osteoporosis.

Conclusions: This case highlights that increases in serum CTX, hypercalcemia and suppressed PTH portend the rebound phenomenon, and that the low bone turnover of GC-treated DMD plus a single dose of IV-ZA are insufficient to prevent this aggressive hypercalcemic complication of denosumab.

P5

Serum Sickness Like Syndrome Due to Methimazole in Graves' disease

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Graves' disease (GD) is a common cause of hyperthyroidism in children.

This case report describes a 3-year-old female with trisomy 21 diagnosed with Graves' disease (GD), initially managed with methimazole (0.4 mg/kg/day) and propranolol (1 mg/kg, three times daily). Two weeks after starting treatment, she presented to the emergency department with a pruritic rash, bilateral knee swelling, subjective fever, and fatigue, necessitating hospital admission for further evaluation. Initial management included cetirizine for symptomatic relief, and a differential diagnosis was considered, including serum sickness-like reaction from methimazole, acute rheumatic fever, and Kawasaki disease. Methimazole was discontinued. As her rash and fever subsided but free T4 levels increased, methimazole was cautiously reintroduced at a reduced dose (2.5 mg daily). However, she soon developed vomiting, diarrhea, and fever, prompting the discontinuation of methimazole once more.

Extensive workup ruled out other inflammatory or infectious causes: complement levels (C3/4) and ANCA/PANCA were within normal limits. That led to the diagnosis of a serum sickness-like reaction from methimazole.

Due to worsening hyperthyroid symptoms, the medical team decided to proceed with a total thyroidectomy. The patient was pre-treated with Lugol's iodine solution, propylthiouracil (PTU), and hydrocortisone to stabilize thyroid hormone levels and prevent a thyroid storm during surgery. Thyroid hormone levels were successfully controlled, and a total thyroidectomy was performed with preservation of the parathyroid glands.

Postoperatively, the patient was managed with thyroid hormone replacement using levothyroxine, calcium supplementation, and alfacalcidol.

This case underscores a rare but potentially severe adverse reaction to methimazole, highlighting the importance of early recognition and prompt withdrawal of the offending drug. When drug intolerance complicates hyperthyroid management, alternative therapeutic strategies, including surgery, may be necessary to prevent severe complications.

P6

The diagnostic odyssey of cyclical Cushing syndrome: A case report

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Introduction: Cyclical Cushing can be very challenging to diagnose due to the cyclical nature of the disease and flaws with current screening methods. This case presents a diagnostic odyssey that took five years to diagnose.

Case: We present a case of a 9 year old male with 5 year history of cyclical episodes q4 - 6months of hyperphagia, insatiable appetite, rapid weight gain of 20 – 50 pounds, facial flushing, abdominal distension and mood changes. Physical exam revealed elevated weight >97th percentile and normal growth velocity for the past five years. Investigations were the following: 1mg dexamethasone suppression test, cortisol of 786 --> 772nmol/L. 24 hour urinary cortisol x 2, <274nmol/day, midnight salivary cortisol 103, 55.1, 61.9 (normal range is <11.3nmol/L). 8mg dexamethasone test, 87 --> 83nmol/L. ACTH 1.7 --> <0.7pmol/L. Liddle's test showed progressive increase in urinary cortisol with day 6, 24 hour urinary cortisol, 1109nmol/L; 3.3x greater than baseline cortisol. Ultrasound testes showed small discrete irregular foci of hyperechoic material. Genetic testing showed a pathogenic change in PRKAR1A gene consistent with diagnosis of Carney complex. Parents preferred a unilateral adrenalectomy understanding its likely not curative. Patient had a left adrenalectomy as CT adrenals showed a more bulky left adrenal gland, January 2024, and continues to remain symptom free.

Discussion: Carney complex is a rare multiple endocrine neoplasia syndrome characterized by multi-organ tumors, two of which were present in this case, primary pigmented nodular adrenocortical disease (PPAND) and testicular tumor. The diagnostic challenge of this case stemmed from normal 24 hour urinary cortisol, and the lack of halted height. Additionally, the CT adrenal glands were read as normal.

Liddle's test and genetic testing ended up confirming the suspected case of cyclical Cushing.

Conclusion: This case highlights the challenge in diagnosis of cyclical Cushing. It emphasizes the utility of Liddle's test and genetic testing in challenging cases like this. Although bilateral adrenalectomy is the standard of care for cyclical Cushing, this case adds to other case reports that have had success with unilateral adrenalectomy; albeit the likelihood of recurrence is high.

P7

Case Report of an Adolescent with Normal Breast Development, Suppressed Gonadotropins, Abnormal Ovaries, and Infantile Uterus

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Case: We present a case of a 14-year-old female referred for delayed puberty. On initial examination, she was Tanner two for breast development and Tanner two for pubic hair. At age 15, she had progressed to breast Tanner 5. Ultrasound pelvis was ordered and neither uterus nor right ovary were identified. MRI abdomen and pelvis revealed an infantile uterus and absent or abnormal, small ovaries on MRI. The lower one-third vagina appeared normal, however, the upper two-thirds of the vagina was not well developed. Kidneys were normal. The patient has no known vertebral, cardiac, or hearing anomalies.

Gonadotropins were suppressed, despite low estradiol levels, though an appropriate response to the gonadotropin stimulation test was demonstrated. Anti-mullerian hormone, androgens, 17-OHP, thyroid function and prolactin levels were all normal. Celiac screen was negative. Given the incongruence between breast development and suppressed gonadotrpins and low estradiol levels, MRI pituitary and chest were ordered. A normal pituitary gland and stalk were visualized and glandular breast tissue was confirmed. Chromosomal microarray demonstrated two X chromosomes, without any clinically relevant copy number variants. Bone age was delayed.

Transdermal estrogen supplementation was initiated, and the dose was increased to 50 mcg twice weekly over 11 months. A repeat MRI pelvis now identifies a normal sized uterus with normal post pubertal appearance, normal sized ovaries, with evidence of follicles, and normal appearing cervix and vagina. Unstimulated gonadotropins were remeasured and were normal.

Discussion: The source of estrogen to support breast development is unclear, given limited ovarian tissue on initial imaging, paired with low estradiol levels. Suppressed gonadotropins in the presence of small, and presumably dysfunctional ovaries is unexpected. Further, the mechanism through which this patient developed normal female reproductive anatomy following estrogen supplementation, is unclear. We suspect an underlying genetic etiology associated with gonadal dysgenesis and ovarian insufficiency which responded to exogenous estrogen supplementation. We have referred our patient to the genetics team, and await investigation results in the near future.

P8

Treatment of Persistent Hypertension in a Patient with 17-Alpha- Hydroxylase Deficiency

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Background: 17-alpha-hydroxylase deficiency (17OHD) is very rare form of congenital adrenal hyperplasia (CAH) caused by autosomal recessive mutations in the cytochrome P450 family (CYP17A1). 17OHD causes decreased cortisol and overproduction of deoxycorticosterone and corticosterone. Despite low cortisol, adrenal insufficiency does not typically manifest because of elevated corticosterone, a weaker glucocorticoid. The typical presentation is a phenotypic female with delayed puberty, low-renin hypertension, and hypokalemia. Given the rarity of this condition, limited literature about management is available. We describe a female adolescent with 17OHD who required combination of glucocorticoids and mineralocorticoid antagonists to control hypertension.

Case Presentation: An otherwise healthy 14-year-old Chinese female was referred for delayed puberty. Investigations showed primary ovarian insufficiency (LH 41.6IU/L, FSH 36IU/L, estradiol <19pmol/L). Karyotype was 46XX; anti-adrenal antibodies were negative. Estrogen replacement was initiated. Genetic testing identified 2 heterozygous CYP17A1 variants: c.985_987delinsAA,p.Tyr329Lysfs*90 (paternally inherited) and c.316T>C,p.Ser106Pro (maternally inherited). An 8AM panel confirmed low renin (<1.0ng/L), low-normal potassium (3.7mmol/L), high deoxycorticosterone (4.73nmol/L), low serum cortisol (21nmol/L), and high ACTH (57.3pmol/L). She had mild hypertension on ambulatory readings (121-131/83-90), though 24h blood pressure monitoring showed significant nocturnal hypertension (129/85). Several therapeutical approaches were attempted, while aiming to avoid adrenal suppression. Physiologic hydrocortisone replacement given once daily (8.8 mg/m2/day) and spironolactone 100 mg/day separately and combined did not improve hypertension. Eventually, a combination of physiological hydrocortisone once daily and slow titration to higher dose spironolactone (100 mg bid) normalized hypertension (115/75) with no side effect.

Conclusions: In conclusion, 17OHD is a rare form of CAH, with about 100 published cases and limited established clear management guidance. Our case highlights the value of genetic testing in primary ovarian insufficiency when no auto-immune etiology is found. Reliance solely on office blood pressures may miss significant hypertension. Therefore, 24h blood pressure monitoring should be considered. While physiological dosing of hydrocortisone alone may be insufficient to control hypertension, addition and titration of spironolactone may help achieve normotension. Partial (once daily) glucocorticoid replacement therapy mitigates long-term consequences of glucocorticoid therapy and likely reduces but fails to normalize cortisol precursors. This approach avoids both symptomatic adrenal suppression and adrenal crisis in 17OHD.

P09

Pediatric Cushing's Syndrome secondary to Ectopic ACTH Secretion by a Pancreatic Neuroendocrine Tumor: a case report

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Neuroendocrine tumors and ectopic Cushing's syndrome are rare entities, especially in pediatrics, and management remains empirical.

We report a 10-year-old girl who presented with a 2-week history of facial plethora and acne, weight gain and abdominal distension. On examination, liver was perceptible 4 cm below the costal margin. Imaging showed multiple hepatic masses, retroperitoneal lymphadenopathies and multiple metastatic pulmonary nodules. A liver biopsy revealed a well-differentiated neuroendocrine tumor with CDX2 positivity (suggestive of jejunoileal versus pancreatic neuroendocrine origin).

Biological investigations revealed an abnormal cortisol cycleand a non-suppressed cortisol and ATCH levels following an 8 mg dexamethasone suppression test. A dotatate PET scan was performed, followed by an abdominal MRI that showed the presence of a single uptaking lesion in the body of the pancreas with normal adrenal glands and pituitary, confirming the diagnosis of a neuroendocrine tumor of the pancreas with ACTH secretion explaining the Cushing's syndrome.

She developed diabetes requiring insulin, hypertension and hypokalemia requiring high dose of potassium supplements, and diuretics and osteopenia without fracture. Prophylactically, enoxaparine and trimethoprime/sulfamethoxazole were initiated for the risk of thrombosis and infection, which she never experienced.

Chemotherapy with octreotide, temozolamide and capecitabine was used. While chemotherapy had begun, adjunctive medical therapy was deemed necessary to control the Cushing's. Ketoconazole alone was used initially and was ineffective. Treatment was changed to metyrapone and mitotane, which was effective to lower but not to normalize cortisol values. Bilateral adrenal embolization was chosen instead of adrenalectomy, firstly, because of the surgical risks with uncontrolled Cushing's and secondly because the right adrenal gland was not accessible due to abdominal masses. Normalization of cortisol values required three embolizations.

Three years after her diagnosis, she still receives chemotherapy. Lesions are stable on imaging. She is now only on metyrapone with normal cortisol levels and stress dose hydrocortisone only. All comorbidities have resolved.

In conclusion, unresectable NETs may require a multipronged approach with chemotherapy, steroidogenesis inhibitors and adrenal resection or embolization used concomitantly or in a stepwise manner with interdisciplinary involvement of endocrinology, imaging, surgery, oncology, pharmacy. Given the rarity of NETs in children, an individualized approach is mandated.

P10

Hidden Cause of Fragile Bones: Scurvy-Induced Vertebral Compression Fractures in an Apparent Healthy 11-Year-Old—A Case Report

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INTRODUCTION: Scurvy, one of the oldest known diseases, results from severe Vitamin C deficiency. It is typically associated with specific populations, including individuals on the autism spectrum or those with limited diets due to food avoidance or severe economic hardship (1,2) making it uncommon in otherwise healthy children (3). Signs and symptoms are often subtle (4).

CASE PRESENTATION: An otherwise healthy 11-year-old refugee boy presented to the Emergency Department at Montreal Children's Hospital with severe lower back pain and progressive difficulty moving.

Imaging revealed vertebral compression fractures and mild diffuse osteopenia, and he was diagnosed with osteoporosis. While 25OHVitamin D levels were severely low (<15 nmol/L; N 50-150), calcium, phosphate, magnesium, ALP, and TSH were normal. A rickets survey revealed findings of decreased bone mineralization with indistinctness and irregularity at the distal femoral and proximal tibial metaphysis, suggestive of a systemic or infiltrative etiology. MRI of the spine suggested an infiltrative process with pelvic lesions, initially raising suspicion of lymphoma.

The patient received intravenous Pamidronate for his vertebral compression fractures and bone pain after correcting his vitamin D deficiency.

On reassessment, the patient was noted to have gingival hyperplasia, and follicular hyperkeratosis. A more detailed dietary history revealed that he had a restrictive diet consisting of rice and beans and a lack of fruits and vegetables. Taken together, he was diagnosed with scurvy and treated with 300 mg daily of vitamin C, leading to notable improvement in bone pain and skin symptoms. Post-discharge labs confirmed severe Vitamin C deficiency (<4 ng/mL), validating the diagnosis of scurvy. Concerns were raised for a possible autism spectrum disorder diagnosis, given his dietary patterns and behaviors.

CONCLUSION: This case highlights scurvy as a rare but significant cause of vertebral compression fractures, underscoring the importance of including it in the differential diagnosis for such presentations. Scurvy can mimic various conditions, leading to extensive investigations and unnecessary treatments if not promptly recognized. A detailed dietary history and thorough physical examination can be crucial in identifying this treatable condition early, to avoid the pitfalls of misdiagnosis and ensure timely intervention for this overlooked cause of bone fragility.

P11

TMEM38B Gene Mutation Associated With Osteogenesis Imperfecta

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Osteogenesis imperfecta is a genetic disorder characterized by decreased bone density, bone deformities, and fractures. It results from mutations in different genes, including all steps of collagen 1 synthesis and modifications. In addition, the gene is involved in the homeostasis of intracellular calcium. TMEM38B is a gene involved in the formation of a cation channel responsible for calcium entry intracellularly.

Mutations in this gene are associated with osteogenesis imperfecta. However, this mutation has not been frequently discussed in the literature. In our study, we report a case of TMEM38B-associated autosomal recessive osteogenesis imperfecta in a child of a consanguineous family presented with a history of multiple prenatal and postnatal fractures.

P12

An Unlikely Pair-a Presentation of Hypercalcaemia and Hyponatraemia

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A previously well, developmentally normal 7-month-old presented with a one-month history of lethargy, reduced oral intake, and weight loss. She was neurologically appropriate, non-dysmorphic, normotensive with mild tachycardia. Family history was positive for nephrolithiasis in mother, maternal grandmother and maternal great grandmother. Initial electrolytes demonstrated a sodium of 116 mmol/L (132–142), potassium 7.3 mmol/L (3.5–6) and chloride 84 mmol/L (96–108).

Total calcium was 3.64 mmol/L (2.08–2.64), phosphate was low 1.51 mmol/L (1.63 – 2.83), urea and creatinine were elevated (33 umol/L, (8-29)), ALP was low (76 U/L (125–440)) and PTH was appropriately suppressed (<5 ng/L). Urine calcium:creatinine ratio was elevated (4.36 mmol/mmol) and urine sodium was <20 mmol/L. A renal US was performed which demonstrated bilateral nephrocalcinosis, indicating longstanding hypercalcaemia. Urine culture was negative. Both renin (>2,500 uIU/mL (9.2–86.9)) and aldosterone levels (52,000 pmol/L (140 – 2500)) were high in keeping with pseudohypoaldosteronism, with likely idiopathic infantile hypercalcaemia (IIH).

She was treated with a 0.9% NaCl bolus and maintenance fluids (0.9% NaCl with 5% dextrose). As hyperhydration was contraindicated given the severe hyponatraemia, calcitonin at a dose of 4u/kg every 12 hours was commenced. With maintenance fluids, there was slow improvement of her sodium and renal function. Calcium also improved, although was still elevated (2.86 mmol/L). The decision was made to proceed with zoledronate (0.02 mg/kg). She was discharged once electrolytes normalized on formula feeds with age-appropriate dietary reference intake of calcium. Nephrocalcinosis panel post discharge demonstrated two heterozygous variants of uncertain significance in the SLC34A1 gene (c.1325 C>T p.[P442L]) and SLC26A1 genet (c.859 C>T p.[R287C]).

Our case describes a novel variant in SLC34A1 in an infant presenting with IIH and pseudohypoaldosteronism. The SLC34A1 gene codes for the type 2a sodium- phosphate transporter NaPi-IIa. Defects within this gene are associated with IIH, nephrolithiasis and hypophosphataemia. Pseudohypoaldosteronism is not well described in association with IIH, although a case report by Kurnaz et al. describes a similar presentation in an infant with homozygous mutations in SLC34A1 and associated hyponatraemia and hyperkalaemia. A clear mechanism for this association is not yet established.

P13

Examining the Moderators of Diabetes Distress among Adolescents with Type 1 Diabetes on the Effect of a Mindful-Self Compassion Intervention

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Introduction: Diabetes distress, defined as emotional distress from living with and managing type 1 diabetes (T1D), is common amongst adolescents with T1D. We completed the "Teaching Adolescents with Type 1 Diabetes Self-compassion" (TADS) study at the Children's Hospital of Eastern Ontario, a parallel group randomized controlled trial involving 141 adolescents aged 12-17 years with T1D. The study evaluated the effectiveness of an 8-week virtual mindful self-compassion for teens (MSC-T) intervention on reducing diabetes distress.

In this secondary analysis, we aimed to identify clinically relevant baseline moderators contributing to inter-individual variability in reduction of diabetes distress, with the ultimate goal of helping clinicians to determine which adolescents may benefit most from the MSC-T intervention.

Methods: Using descriptive statistics, baseline demographic data was summarized. Potential moderators of diabetes distress (TADS primary outcome), as measured by the Problem Areas in Diabetes Teen Version (PAID-T, range 26-156) score 3 months post-baseline, were identified by clinical relevance and literature review. These included depression and anxiety, as measured by the Patient Health Questionnaire (PHQ-9, range 0-27) and Generalized Anxiety Disorder 7-item scale (GAD-7, range 0- 21), respectively, and glycemic metabolic outcomes, as measured by HbA1C and time in range. These potential moderators were assessed at baseline and at 3 months post-MSC-T intervention. Separate linear regression models were constructed for each moderator to determine if selected variables moderated the effects of the MSC-T intervention on diabetes distress.

Results: Mean scores and standard deviations are reported for baseline diabetes distress as measured by PAID-T in the intervention group (74 ± 27, N=70) and control group (72 ± 25, N=71). Baseline measures of moderators are reported for the intervention and control groups, respectively: PHQ-9: 7.7 ± 6.3 and 7.0 ± 5.7, GAD-7: 7.2 ± 5.8 and 5.9 ± 4.7, percentage of time in range among CGM users: $57\% \pm 22$ and $51\% \pm 19$. Results of linear regression models will be available at the time of the 2025 CPEG meeting.

Conclusion: Findings will identify baseline features of adolescents with T1D who are most likely to benefit from the MSC-T intervention, thereby assisting in targeting those adolescents in the clinical setting.

P14

Hyperosmolar hyperglycemic state and significant microvascular complications: a rare presentation of adolescent type 2 diabetes

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Background: Youth-onset Type 2 Diabetes (YO-T2D) is on the rise and disproportionately affects those from low socioeconomic status and specific ethnic and racial minority groups. The phenotype, comorbidities, and complications of YO- T2D are more severe than later onset T2D. One such rare, but serious complication is Hyperosmolar Hyperglycemic State (HHS), which is characterized by significant hyperglycemia, hyperosmolarity, lack of ketosis, and profound dehydration.

Case: The patient is a 14-year-old who presented after a fall in the context of a 1 month history of polyuria and polydipsia. She was found to be acutely unwell with significant peripheral edema. Initial investigations indicated HHS with a serum glucose of 44 mmol/L, serum osmolality 330 mmol/kg, Na 129 mmol/L, and no significant metabolic acidosis (venous blood gas pH 7.31, bicarbonate 21.8 mmol/L) and negative ketones. Additionally, investigations indicated acute kidney injury (creatinine 156 umol/L), and nephrotic range proteinuria with 3+ proteinuria on dipstick, and hypoalbuminemia (23 g/L). Sixteen months prior, hemoglobin A1C was greater than 16% and random glucose was 23.1 mmol/L, with no treatment initiated at that time. She was initially managed in the pediatric intensive care unit. Due to her low albumin state, fluid resuscitation was commenced at a lower rate than typical for HHS. Intravenous rehydration followed by insulin infusion combined with albumin infusion and furosemide corrected HHS. Following the resolution of HHS, further evaluation revealed mild retinopathy and stage 3 diabetic nephropathy. The patient started multiple daily insulin injections prior to beginning metformin and empagliflozin, which was chosen as an adjunct agent due to the renal protective effects of SGLT-2 inhibitors. The presence of developmental delays, short stature, and hypogonadism prompted genetic testing, which confirmed a diagnosis of Prader Willi Syndrome (PWS).

Discussion: HHS is a rare and potentially fatal acute complication of diabetes. There is an increased incidence of T2D in PWS which is often diagnosed incidentally with screening. The prolonged prodrome of hyperglycemia contributed to the severity of this patient's condition at presentation. This case underscores the severity of YO-T2D, the risks of delayed healthcare access, and the important considerations for optimal long-term diabetes management.

P16

Two siblings with Laron syndrome and their response to mecasermin therapy.

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Severe primary insulin-like growth factor-1 deficiency (SPIGFD) is primarily secondary to growth hormone receptor(GHR) mutations also known as Laron syndrome (LS). There are about 350 reported cases worldwide. Recombinant IGF-1 (rIGF-1) has been approved in Canada since 2022, with reported growth velocity of up to 8cm/yr in the first year of treatment. We present two sisters' responses to rIGF-1. They were born to consanguineous parents of East Indian heritage with a mid-parental height of 156.06 cm +/- 8 cm.

For patient 1, investigations were initiated at 12 months of age due to postnatal growth failure (Ht -5.52 SD, Wt -4 SD). Genetic workup revealed a homozygous variant in GHR c.508G>C, p.Asp170 and two variants of unknown significance (a paternally inherited 15q11.2 duplication and a heterozygous variant in the IGF1 receptor gene: c.1381C>T, p.Arg461Cys). She was growth hormone sufficient (GH 31.3 ug/L), but had low IGF-1 (<10 ug/L) and IGFBP3 (<0.5 mg/L) levels. Treatment with rIGF-1 was started at 2 years of age (GV 4.5cm/yr, Ht -4.10 SD). Adherence was challenging, with height gain of 3 cm/yr (SD -5.23), therefore treatment was stopped. While off rIGF-1, her growth velocity slowed significantly (0 cm in 8 months), and she restarted therapy. Height velocity was maintained for the next two years (4.5cm/yr, 5.9cm/yr).

Patient 2's birth weight and height were on the 3rd percentile. At 31 months her growth velocity was 2.8cm/yr (Ht -5.02 SD). Testing revealed the same homozygous GHR mutation as her sister. rIGF-1 was started at 3 years 6 months of age (Ht - 4.79 SD). After one and two years of treatment, her height velocity was 6.9cm/yr and 9.1cm/yr (Ht -3.85 SD).

Studies on rIGF-1 treatment showed that the highest growth velocity is expected within the first year, though Patient 2's height velocity continued improving beyond two years. Sibling responses varied, potentially due to adherence and an additional heterozygous IGF1R variant of unknown significance in the eldest. Over 70 mutations linked to Laron syndrome have been identified. Further research is needed to determine if specific genetic variants affect treatment outcomes and guide clinical practice.

P17

Improving Tools and Clinical Processes to Identify and Address Food Insecurity

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Background: Food insecurity is inadequate access to food due to financial constraints. The prevalence of food insecurity is higher in families of children with diabetes, compared to the general population in Nova Scotia. Household food insecurity has been associated with an increased risk of acute diabetes complications. We aimed to identify barriers and facilitators to discussing food security (FS) status and to improve clinic processes that support identification of, and coping with, food insecurity among families of children with diabetes.

Methods: The Plan-Do-Study-Act model is guiding this quality improvement project. We started with a focus group (FG) with the pediatric diabetes team to understand the current screening process and guide development of FG questions for families. Next, the pediatric diabetes clinic population were invited to participate in FGs, and interested families provided consent to be contacted. Results from the FGs informed development of an anonymous survey, that was distributed to the clinic population via email. This feedback informed improvements made to the screening process and the development of a resource guide for families. These interventions will be examined iteratively throughout the process using the Plan-Do-Study-Act model.

Results: The diabetes team lacked understanding of and processes to assess FS. FG participants also did not understand the term *food insecurity* or how the clinic could offer support but identified the dietician and social worker as most appropriate to lead FS discussions. Surveys revealed that most families (71%) agree that identifying FS status is important but very few (5.1%) have discussed concerns about affording groceries in clinic. Acceptable improvements to screening include adding FS screen to the existing pre-appointment questionnaire (63%) and appointing the dietitian (61%) or social worker (49%) to initiate FS discussions.

Families agreed helpful resources would include information on financial programs, recipes, meal plans, cooking workshops, and access to local programs and supports. Conclusion: Improvements that are acceptable to families can be made to the diabetes clinic screening process for FS. These results are being used to adjust and then re-assess clinic processes and develop a resource guide for clinic use.

P18

The AB(C)s of Thyroid Hormone Resistance

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Background: Thyroid hormone resistance results from mutations in the thyroid hormone receptor genes, THRA and THRB. The presentation is gene-dependent and limited data exists on management. We describe our management of thyroid hormone resistance alpha and beta.

Case Presentation: Patient A presented at 34 months with short stature, severe constipation and global developmental delay. Her height was - 3.3 SD, anterior fontanelle was widely open and she had relative macrocephaly. Labs showed free T4 9.6 pmol/L (10 - 25 pmol/L) and TSH 1.95 pmol/L (0.2-6.5 mIU/L). Pituitary MRI was normal. Genetic testing showed a heterozygous mutation in THRA, c921T>G, p(Leu274Arg), a variant of unknown significance. Levothyroxine was started to overcome resistance, with significant clinical improvement. Free T4 normalized, with a suppressed TSH and an elevated free T3.

Patient B presented at 22 months with short stature (-2.4 SD), tachycardia, and hyperactivity. Family history revealed thyroid hormone resistance. Free T4 and free T3 were elevated at 44.6 pmol/L and 15.5 pmol/L, respectively. TSH was 2.02 mU/L . Genetic testing showed a likely pathogenic heterozygous mutation for THRB, c980C>A p(Thr327Asn). Atenolol was started for tachycardia. At the age of five, he continued to struggle with hyperactivity. The literature suggests treatment with high-dose T3 to suppress TSH. He was started on liothyronine 50 mcg every other day. The family reported improved behaviour on medication days. Subsequently, he was tried on daily dosing. This improved his symptoms, but suppressed his free T4. A trial of splitting the dose to BID dosing resulted in a high free T3, and the patient was clinically hyperthyroid. The patient is back on liothyronine 50 mcg daily, which is keeping him clinically euthyroid, but he remains biochemically abnormal.

Conclusion: In our experience, thyroid hormone resistance due to mutations in THRA, it is possible to achieve clinical improvement with levothyroxine. However, thyroid hormone resistance due to mutations in THRB, management is more challenging.

P19

Early detection and treatment of vertebral fractures, a sign of accelerated senescence in Hutchinson-Gilford Progeria Syndrome, is associated with stabilization of vertebral fractures

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Background: Hutchinson-Gilford Progeria Syndrome (HGPS) is a rare, autosomal dominant disorder of accelerated senescence caused by variants in the Lamin A (*LMNA*) gene. HGPS is characterized by signs of premature aging including low bone mineral density (BMD) and atherosclerosis, with patients typically succumbing to cardiovascular events in the second or third decade of life. Lonafarnib, a protein farnesyltransferase inhibitor, is standard of care for HGPS (in addition to pravastatin), with evidence for improved BMD when given with intravenous zoledronic acid (IV ZA). Vertebral fractures, hallmarks of osteoporosis in natural aging, have not previously been reported in HGPS. We describe early detection of vertebral fractures in a young girl with HGPS, along with the structural and BMD at the spine response to IV ZA.

Case Description: A 3.5-year-old girl with HGPS was referred to the Pediatric OsteologyClinic for routine bone health surveillance. She was diagnosed with HGPS at 4 months of age after a lipodystrophy gene panel revealed a pathogenic *de novo* variant in *LMNA*, and subsequently treated with lonafarnib. Her physical examination revealed short stature (height Z-score -3.3), tight skin, prominent veins, minimal muscle and sub-cutaneous fat, near-complete alopecia, dental crowding, and abnormal skull shape. A lateral spine X-ray showed multiple asymptomatic vertebral fractures from T2 to L1 (total Spinal Deformity Index = 6) with low bone turnover (serum c-telopeptide of type I collagen Z-score -2.6, as expected with premature aging). Given the progressive nature of her condition, IV ZA was initiated at standard doses. Over the next four years, she demonstrated stabilization of vertebral fractures, with a marginal increase in the lumbar spine areal BMD Z score (from -3.5 to -3.3).

Conclusions: This case highlights that individuals with HGPS can present with asymptomatic vertebral fractures, underscoring the need for routine surveillance at a young age. Despite the progressive nature of HGPS, there is potential for stabilization of vertebral collapse and marginal improvements in LS areal BMD Z- scores on IV ZA. Whether timely initiation of IV ZA in HGPS prolongs longevity (as it does in natural aging) requires further study.

P20

6-month Leuprolide Acetate for Monitoring Central Precocious Puberty Treatment Efficacy

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Introduction: Gonadotropin-releasing hormone (GnRH) stimulation testing is used in the diagnosis and monitoring of treatment efficacy in children with central precocious puberty (CPP). The recent discontinuation of daily subcutaneous leuprolide acetate (LA) in Canada prevents the use of stimulation testing in clinical practice. Moreover, these tests are costly and necessitate additional injections, which may negatively impact children's experience. Therapeutics that can also function as stimulation tests can resolve issues related to stimulation testing accessibility and improve care. Some long-acting GnRH agonists (GnRHa) have been used as such. We present secondary analyses of pharmacokinetic data from the pivotal trial of the first small-volume, long-acting, subcutaneously administered GnRH agonist for CPP, approved in 2023, that support this possibility.

Methods: 62 treatment-naïve children (60 girls) with CPP received 2 doses of 45 mg subcutaneous LA at 24-week intervals. Blood samples were used to characterize the initial luteinizing hormone (LH) burst phase and baseline and trough concentrations of LA following study drug administration.

Results: Mean serum LH concentrations reached a peak of 43.4 IU/L (SD: 43.7 IU/L) 4 hours after the first study drug injection. Mean serum LA concentration increased rapidly after the first study drug injection, peaking at 215.7 ng/mL (SD: 163.2 ng/mL) 4 hours post-injection. Area under the leuprolide concentration-time curve was 39.1 ng*day/mL 0-6 hours after initial dose.

Conclusions: A therapeutic dose of 45 mg 6-month subcutaneous LA demonstrated a pharmacokinetic profile consistent with long-acting injectable LA therapies. The burst kinetics, due to the unique polymeric gel extended-release drug delivery system, reflect those of other GnRHa preparations that have been shown to be an effective substitute for native GnRH or GnRHa in stimulation tests. Peak stimulated LH concentrations (43.4 IU/L) were higher than those seen following a traditional GnRH stimulation test (21.5-25 IU/L), which is expected given the greater potency of LA compared to native GnRH. These data provide preliminary evidence that this new therapy can be used as a stimulation test for subsequent injections to monitor therapeutic response, alleviating challenges created by the discontinuation of daily subcutaneous LA and improving the child's therapeutic experience.

P21

Weight Stigma in the Pediatric Diabetes Population: Evaluating Patient Level Factors and Diabetes Related Outcomes

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Background: Weight stigma is defined as discrimination based on weight and body size and is well-documented among adults and children. Individuals who experience weight stigma are at risk of adverse physical and mental health outcomes. To our knowledge, there are no studies evaluating weight stigma in the pediatric diabetes population despite weight often being reviewed and discussed in diabetes clinic follow-ups.

Objective: Our study aimed to evaluate the prevalence of weight stigma in our pediatric diabetes population and to identify any patient level factors associated with higher prevalence of weight stigma. We also aimed to determine if weight stigma is associated with poorer diabetes related health outcomes and increased diabetes distress.

Methods: A cross-sectional study was conducted among pediatric diabetes patients at BC Children's Hospital. Participants completed questionnaires that included experienced weight stigma (EWS), weight bias internalization scale (WBIS), weight self-stigma questionnaire (WSSQ), modified brief illness perception questionnaire (BIPQ) and diabetes distress scale (DDS). A chart review was conducted to gather patient level data and outcomes. Descriptive statistics and logistic regression analysis were used to analyze the data.

Results: There were a total of 103 participants (95 with type 1 diabetes and 8 with type 2 diabetes). The response rate was 33.2%. The prevalence of experienced weight stigma was found to be 19.4%, of which 20% identified family members to be the leading source of stigma. Of the participants endorsing weight stigma, 28.5% scored high for weight self stigma, and 57.1% scored high for internalized weight stigma.

Female sex was found to be associated with weight stigma (p-value <0.05). Participants experiencing weight stigma had higher diabetes distress (p-value <0.001), and presented a non-significant trend towards increased residential instability (OR 1.46, 95%CI 0.98-2.17), and higher A1c (OR 1.34, 95%CI 0.98-1.84).

Discussion: Weight stigma is prevalent among pediatric patients with diabetes and is associated with higher diabetes distress. While limited in sample size, and strongly biased towards type 1 diabetes, these findings highlight the need for further studies to better understand the role of weight stigma in management of pediatric patients with diabetes to optimize their health outcomes.

P22

Primary hyperparathyroidism due to a single mutation in CDC73 in a 14- year-old girl: a case-report

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Primary hyperparathyroidism (PHPT) is a rare condition in children, with an incidence estimated at 2–5 per 100,000. Hyperparathyroidism and hypercalcemia can be subtle, making early detection challenging to prevent complications such as renal lithiasis, nephrocalcinosis and renal failure, as well as bone demineralization.

We report the case of a 14-year-old girl, known for focal epilepsy, who presented with persistent hematuria and abdominal pain. Abdominal ultrasound revealed numerous non-obstructive kidney stones. Investigations showed levels of ionized calcium of 1.74 mmol/L and PTH of 59.5 pmol/L. Kidney function was normal. She did not report polyuria-polydipsia, vomiting, or asthenia. Neck ultrasound showed a lesion measuring 30x16 mm in the left III chain above the thyroid while technetium99m scintigraphy confirmed the presence of a parathyroid adenoma in the upper part of the left thyroid lobe. Initial management included hyperhydration and restriction of dairy products intake, but the initiation of cinacalcet was secondarily necessary. Vitamin D was prescribed due to severe vitamin D deficiency.

Excision of the adenoma resulted in transient hypoparathyroidism, requiring calcitriol and calcium supplements for less than a week. The pathology report indicated complete excision of a parathyroid adenoma weighing 5 g. Eosinophilic trabeculae with enlarged nuclei (sometimes multinucleated) were observed, suggestive of a germline pathogenic variant or deletion of CDC73.

Genetic analysis revealed an autosomal dominant CDC73 mutation, with increased risk for primary hyperparathyroidism, jaw tumors (known as PHPT-jaw tumor syndrome) as well as kidney and uterine tumors (mostly benign). In ≈15% of cases, hyperparathyroidism is caused by parathyroid carcinoma. Penetrance is variable and primary hyperparathyroidism may be isolated. Although pediatric cases in have been described, studies indicate a median age at diagnosis between 23 and 38 years. There is no known case of primary hyperparathydoism in this family, but genetic counselling is underway. Interdisciplinary follow-up involving endocrinology, genetics, surgery and imaging is mandatory for optimal monitoring of tumor risk.

In conclusion, a genetic evaluation is recommended in any case of pediatric primary hyperparathyroidism to identify potential genetic causes and manage associated risks effectively.

P23

Diabetes Education during Pediatric Residency Training: Are we doing enough?

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Objective: To evaluate perceived competency and general diabetes knowledge of pediatric residents from the University of British Columbia (UBC)

Introduction: The incidence of type 1 diabetes mellitus (T1D) is predicted to rise 3 to 4-fold in the coming decades. In British Columbia, approximately one third of children with T1D receive care at BC Children's Hospital (BCCH), while the rest are followed in their communities via local pediatric endocrinologists, pediatricians, and outreach clinics. To ensure equitable and high-quality care, community pediatricians must have the competency to care for these patients.

Methodology: A survey was created and distributed via REDCap to pediatric residents at BCCH who had completed their core rotation in pediatric endocrinology. This survey included questions on the characteristics of their training, perceived competence and knowledge of general diabetes. Their knowledge was also assessed using brief clinical scenarios. Feedback on their educational needs was also ascertained.

Results: Twenty-three out of eighty pediatric residents responded to the survey (response rate 28%). Of these, 26% were senior residents (third or fourth year). 35% had personal experience with diabetes and 43% had additional exposure (diabetes camp, electives). 52% were very confident in managing hypoglycemia. 65% were somewhat confident regarding general diabetes management. Only 13% were very confident in making insulin dose adjustments. Further, on assessment of knowledge, 87% of participants were not able to recommend an appropriate insulin dose in a case scenario of hyperglycemia with ketones. Residents expressed a clear interest in receiving more hands-on clinical experience in diabetes management.

Conclusion: Adequate care and follow up is needed to ensure good outcomes in pediatric patients living with diabetes. Pediatricians and pediatric trainees will care for families living with diabetes throughout their professional career. While such competences should be acquired during medical school and residency, there is a mismatch between expectations and reality. Pediatric residents trained at our tertiary care pediatric center were not fully confident in their ability to manage patients with diabetes and requested more practical exposure. This study is ongoing and survey results of graduates and educators are to follow. Results will inform further educational initiatives.

P24

A case of truly “idiopathic” infantile hypercalcemia – when calcium is high but everything else is low

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Hypercalcemia in infants is a rare but potentially serious condition with a broad differential diagnosis that includes both parathyroid hormone (PTH)-mediated and non-PTH-mediated etiologies.

Case: A 10-month-old male with a known history of X-linked ichthyosis and developmental delay, on no pre-existing medications, presented with a several month history of poor weight gain, feeding difficulties, and irritability. He was admitted initially for work-up of his failure to thrive, and was found to have severe hypercalcemia, with initial total calcium 5.62 mmol/L and ionized calcium 2.45 mmol/L. Phosphate was low 1.37 mmol/L and creatinine elevated 50 umol/L. He was noted to be hypertensive with blood pressure of 161/108, thought to be due to vasoconstrictive effects of hypercalcemia.

Management of his hypercalcemia included IV hyperhydration and loop diuretics, followed by a single dose of calcitonin for rebound hypercalcemia. Calcium was then maintained within normal on low-calcium formula. Oral phosphate supplementation was also started for persistently low phosphate.

Work-up to determine causes of his hypercalcemia revealed undetectable PTH < 0.6 pmol/L, with elevated urine calcium:Cr ratio, and normal 25-hydroxyvitamin D 46.4 nmol/L. Thyroid function tests and cortisol were normal. Renal ultrasound showed diffuse medullary nephrocalcinosis, but otherwise structurally normal kidneys. Given his clinical picture, a diagnosis of idiopathic infantile hypercalcemia was suspected. However, he had an undetectable 1,25-dihydroxyvitamin D < 12 pmol/L. PTH-related peptide was also normal and he had no other signs of malignancy. Skeletal survey was completely unremarkable. ECHO was normal and microarray findings were not consistent with Williams Syndrome. Whole exome sequencing was sent.

This case highlights the diagnostic challenge in pediatric hypercalcemia with suppressed PTH and normal/low vitamin D levels. Genetic testing will hopefully reveal the etiology of his hypercalcemia and guide long-term management.

P25

A case of type 1 diabetes mellitus presenting with hypothyroidism

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Background: Clinical manifestations of hypothyroidism may vary widely from severe, life- threatening conditions to completely asymptomatic presentations. While the risk of hypothyroidism is elevated in children with type 1 diabetes (T1D), the simultaneous onset of both conditions remains relatively uncommon.

Case presentation: A 9-year-old girl was referred to endocrinology due to height deceleration with a growth velocity of 1.5 cm/year associated with obvious weight gain (+10 kg) over the past year. She was otherwise asymptomatic, with no reported changes in appetite, diet, physical activity, urination or bowel movements. Her past medical history was notable for alopecia areata. Family history did not reveal any autoimmune disease. On physical examination height was: 124.5 cm (5th - 15th %) and weight was:32.9 kg (75th %). Cheeks were erythematous and puffy but she had no dysmorphic features. Thyroid was not enlarged and visual fields were normal.

Initial investigations were notable for elevated random blood glucose level:27.6 mmol/L (3.9 – 11mmol/L), normal blood gas, negative ketones, HbA1C:9.5% (5.0- 6.0%); and TSH: 473.43 (0.34–5.60 mU/L) with FT4: < 2.50 (8.0-18.0 pmol/L). TPO and GAD autoantibodies were positive. Treatment was initiated with a low dose of Levothyroxine (12.5 mcg) which was progressively increased alongside insulin therapy (total daily dose 0.4 Ui/kg/day). During treatment, TSH decreased gradually and normalized after 8 weeks, HbA1C showed a 1% reduction.

Conclusion: Hypothyroidism can be challenging to diagnose clinically. When severe, hypothyroidism can lead to reduced glomerular filtration, masking concurrent onset of classic symptoms of T1D. Evaluation for T1D may warranted in children presenting with severe hypothyroidism

P26

Not all salt wasting is Congenital Adrenal HYPERplasia: Case report of delayed presentation with NR0B1 mutation

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Background: NR0B1/DAX1 mutations have been described to cause X-linked adrenal hypoplasia congenita (AHC). With increasing recognition for genetic testing in endocrinology, we can now distinguish different causes of adrenal insufficiency, some of which are not identified on newborn screening panels.

Case presentation: This case report describes a 19-month-old child patient who presented with a 1-week history of vomiting and loose stools, weight loss of 1 kg, and was incidentally found to be hyponatremic at 112 mmol/L. On exam, he had hyperpigmentation of his axilla and bilateral inguinal canal with normal male genitourinary anatomy. The family history was significant for two maternal uncles with presumed salt-wasting congenital adrenal hyperplasia and maternal female relatives with premature ovarian insufficiency. The endocrinology panel identified an elevated renin of > 30,000 ng/L and inappropriately normal aldosterone of 218 pmol/L. He had a robust random cortisol at the time of presentation but developed cortisol deficiency 8 months later. Due to strong family history of male primary adrenal insufficiency, X-linked congenital adrenal hypoplasia was suspected. Genetic testing revealed maternally inherited nonsense mutation (c.670C>T) in the NR0B1 gene.

Conclusion: Newborn screens, particularly in Ontario do not capture all causes of adrenal insufficiency. This case report highlights the importance of genetic testing to identify rare causes of adrenal insufficiency including NR0B1/DAX1 mutation.

P27

Prevalence and Risk Factors for Bladder and Bowel Dysfunction in Children with Type 1 Diabetes

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Background: Long-term DCCT follow up has demonstrated that urinary incontinence is associated with higher HbA1c levels in women with type 1 diabetes, independent of other recognized risk factors. A Duke University pilot study revealed 33% of children (11-17 years) followed for either type 1 or 2 diabetes had bladder and bowel dysfunction (BBD); however, specific prevalence data in children with type 1 diabetes (T1D) is not available. BBD is a common but underdiagnosed condition that can cause considerable physical and psychosocial burden for children and their families.

Objectives: This study aimed to 1) determine the prevalence of BBD in children with T1D and healthy controls, and 2) explore clinical factors associated with childhood BBD.

Methods: This was a cross-sectional study of children aged 5-16 years with T1D and healthy controls, recruited from five endocrinology and primary care clinics in Canada and the US. Participants completed both the Vancouver Symptom Score (VSS) survey, a validated 13-item Likert scale questionnaire for non-neurogenic lower urinary tract dysfunction/dysfunctional elimination syndrome, and a 4-point Likert scale to rate bother of symptoms. Clinical information including age, sex, BMI, and HbA1c was collected. Binary logistic regression analyses were used to explore clinical factors associated with BBD.

Results: 242 children with T1D and 86 healthy controls were recruited. The overall BBD prevalence in children with T1D was 21.5% and 10.5% in the control group. The odds of BBD increased 2.5-fold (95% CI 1.07-5.82) for children with T1D. Younger age was also significantly associated with BBD (OR: 0.90; p=0.017). In children with T1D, A1c of 7.9 to <8.7% and >8.8% increased odds of BBD by 4.1-fold (95% CI 1.45-11.48) and 4.8-fold (95% CI 1.70-13.73) respectively. Urinary incontinence, male sex and BDD diagnosis were associated with higher odds of reporting bother symptoms in T1D patients with odds ratios of 1.96 (95% CI 1.26-3.04), 8.33 (95% CI 1.91-36.31) and 8.19 (95% CI 1.07-62.98), respectively.

Conclusions: Routine screening for BBD should be incorporated into standard T1D care given the high prevalence of BBD in this population. Poor glycemic control further increased BBD risk, emphasizing the importance of intensive glucose management.

P28

A Description of ACTH Stimulation Testing Results in Term Neonates

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Introduction: Adrenal insufficiency (AI) in the neonate is associated with significant morbidity and mortality. The clinical features of AI are non-specific, and so biochemical confirmation is necessary. The normal cortisol response after ACTH stimulation has not been clearly defined in healthy newborns and young infants.

Some data shows that neonates may have lower cortisol levels than older children and adults. One proposed explanation is that neonates and young infants have half the level of cortisol binding globulin (CBG) compared to older children. Thus, the lower CBG level may translate to lower total cortisol levels with ACTH stimulation.

Objective: To describe the clinical features and stimulated cortisol response of corrected term newborns and young infants (0 to 100 days of age) who underwent ACTH stimulation testing.

Methods: This is a single center, retrospective cohort study reviewing the medical records of children under 100 days of life, who underwent ACTH stimulation testing between 2014 and 2021. This study focused on a stimulation test result between 250 and 500 nmol/L.

Results: 14 patients were identified of whom, 12 were born at term and 2 were preterm. The most common cause for screening was hypoglycemia with 7 out of the 14 patients. Other causes included positive newborn screening for congenital adrenal hyperplasia (CAH), genetic abnormalities at risk for hypopituitarism, differences in external genitalia on exam, and electrolyte abnormalities. The results ranged from 319 to 495 nmol/L with a median of 459 nmol/L (IQR 410.5 – 470.25 nmol/L). Two patients were started on hydrocortisone therapy before the stimulation test, but both were taken off after the test. 13 of the 14 patients did not require any endocrine follow up. One patient was started on physiological glucocorticoid medication for non-classical CAH management.

Conclusions: In this study, in term newborns, hypoglycemia was the most common cause for ACTH stimulation testing and results over 410 nmol/L seem associated with no further need for ongoing steroids or endocrine follow-up.

P29

Echoes of the Past: A Case of Multiple Endocrinopathies due to Iron Overload

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Background: Thalassemia major is a transfusion-dependent hemoglobinopathy that often leads to iron overload, as repeated transfusions deposit excess iron in various organs, including the heart, liver, pituitary, pancreas, and thyroid. This iron accumulation can damage organ tissue through oxidative stress, harming DNA, proteins, and lipids. While the blood-brain barrier (BBB) protects brain cells from iron deposition, the pituitary gland lies outside the BBB and is thus susceptible to iron toxicity. This vulnerability often results in hypogonadism, hypoparathyroidism, hypothyroidism, growth hormone deficiency, and delayed puberty.

Case Presentation: A 15-year-old Afghan girl with transfusion-dependent thalassemia major presented to our Pediatric Endocrinology Clinic after asymptomatic hypocalcemia was detected during routine screening at her first hematology visit in Canada. She had been receiving monthly transfusions in Afghanistan since the age of two months without effective iron chelation therapy. Iron overload symptoms included liver cirrhosis, growth delay, and delayed puberty.

Initial evaluations indicated primary hypoparathyroidism, evidenced by hypocalcemia and hyperphosphatemia with normal PTH levels, and primary hypothyroidism, with elevated TSH and borderline free T4 levels. Physical examination showed short stature (height Z-score: -3) and Tanner stage I breast development. She was started on calcium carbonate, calcitriol, and vitamin D for hypoparathyroidism and levothyroxine for hypothyroidism.

Further assessments revealed low IGF-1 and gonadotropin levels and reduced bone mineral density. A normal ACTH stimulation test ruled out cortisol insufficiency.

Additional testing for growth hormone deficiency and potential pubertal induction is planned. A multidisciplinary team, including endocrinology and hematology, is managing her iron overload and endocrine deficiencies while optimizing skeletal health.

Discussion: This case highlights the importance of thorough endocrine evaluations in patients undergoing regular transfusion therapy. While current chelation treatments can reduce the risk of iron overload, access to these therapies may be limited for some patients, and in certain cases, they may not be entirely effective in preventing iron accumulation. The pituitary gland is especially susceptible to iron deposition, making continuous monitoring and individualized management essential to address the complex endocrine disorders that can arise from iron-related damage in transfusion- dependent patients.

P30

The genetics of mild isolated neonatal hyperthyrotropinemia – an additional tool to help predict transient vs. permanent congenital hypothyroidism?

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Background: The incidence of congenital hypothyroidism (CH) has increased over time and is commonly correlated with increased detection of transient disease. The distinction between transient CH (T-CH) and permanent CH (P-CH) remains poorly defined during the first few years of life, with limited ability to anticipate which children may successfully discontinue L-thyroxine therapy. Patients with mild isolated neonatal hyperthyrotropinemia (MINH, defined as TSH 17-40 mIU/L, normal FT4, and gland in situ) commonly fall into the category of T-CH. Recent literature supports that up to one third of patients with MINH carry causative variants in the TSH-R gene. We sought to evaluate the germline status of patients with MINH to better understand the spectrum of genetic findings in this patient population, and if genetic variants can be used as a predictor for transient disease.

Methods: Next generation sequencing was performed using a targeted panel of 44- genes associated with TSH signaling or thyroid gland development. Patients with MINH were identified through Newborn Screening Ontario (HSC and CHEO retrieval centres) between April 2006 and August 2015 (n= 89). Clinical data were collected from birth and up to 4 years of age, when available.

Results: 24% had a pathogenic or likely pathogenic variant(s) consistent with the inheritance pattern of the disease; 11% had a probable diagnosis (one pathogenic or likely pathogenic variant and a second variant of uncertain significance (VUS) in a recessive gene); 13% had one or more VUS of interest consistent with the inheritance pattern of the gene; 33% had an inconclusive result (either a single pathogenic or likely pathogenic variant detected in a gene associated with recessive inheritance or VUS that were considered weak candidates); the remaining 16% had a negative result with no variants of interest detected. Correlation between germline status and outcome will be presented in the context of differentiating T-CH from P- CH. Further study may help determine whether germline testing would be a cost- effective approach to reducing the burden of care for children with CH identified by NBS.

P31

Unrecognized social needs in a pediatric diabetes clinic: insights from Navigating Social Resources for Children's Health (NSRCH) study

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Background: Unmet social needs negatively impact health outcomes in pediatric diabetes. Although standard care models include social workers (SW) as part of the diabetes interdisciplinary team, access to SWs is limited and models that systematically screen and address social needs in this population are lacking. To address this gap, we established the Navigating Social Resources for Children's Health (NSRCH) study at the Children's Hospital of Eastern Ontario (CHEO) with an overall goal of screening all families of children with diabetes for unmet social needs (Phase 1) and then inviting those with identified social needs (and not already followed by SW) to be randomized to receive support from either a lay social navigator (intervention) or SW (standard care) (Phase 2).

Objective: To report on the results of the screening phase of the NSRCH study, including response rate to the screening questionnaire, positive screen rate, and identified social needs.

Methods: Parents/guardians of all children 0-17 years with type 1 or 2 diabetes followed in CHEO's diabetes clinics were invited to complete a 10-item screening questionnaire to identify unmet social needs. A positive screen was defined as one or more positive response(s).

Results: From July 2023 to October 2024, 785 families were invited to complete the NSRCH Phase 1 screening questionnaire, with 412 questionnaires completed. Of these, 49% (n=203) identified at least one unmet social need with 60% (n=122) of positive screens having two or more unmet needs. Only 23% (n=48) of respondents who screened positively were already followed by SW. Insufficient income was the most common need identified (n=121;59%), followed by food insecurity (n=101;49%) and access to diabetes technology (n=50;25%). Additional needs included finding employment (n=37;18%), transportation (n=32;16%), housing (n=28;14%), and access to technology for healthcare (n=20;10%).

Conclusion: Approximately half of the families screened had unmet social needs and most were not connected with SW support, highlighting a gap in our current diabetes standard of care model and the importance of routine screening in this population. Phase 2 of the NSRCH study will provide insight into the use of a social navigator as a means to address these identified social needs.

P32

The causal role of endocrine disrupting chemicals in pubertal timing: a Mendelian randomization study

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Introduction: Endocrine disrupting chemicals (EDCs) interfere with hormonal homeostasis, and have been linked to altered pubertal timing, defined by the age of menarche (AAM) in girls and the age at voice change (AVC) in boys. However, the causal nature of these associations remains unclear. We used Mendelian randomization (MR) to investigate if genetically altered serum levels of EDCs affect pubertal timing.

Methods: We performed univariate MR to assess the causal role of various EDCs on AAM and AVC. This analysis used the largest available GWAS for 22 EDCs as well as , and European and multi-ethnic GWAS data on AAM in girls and AVC in boys from the ReproGen consortium. Multivariate MR (MVMR) was then conducted to examine the mediating effect of body mass index (BMI).

Results: Our analysis revealed three MR associations with AAM: PCB 74 (βIVW: - 0.015, 95% CI [-0.028, -0.003], p= 0.014), DBP (βIVW: 0.006, 95% CI [0.001, 0.010], p= 0.013) and PCB 206 (βIVW: -0.024, 95% CI [-0.041, -0.006],p=0.0068). Analyses in boys are ongoing. MVMR suggested BMI's mediating role in associations with AAM.

Conclusion: Our MR findings indicate that exposure to PCB 74 and PCB 206 leads to earlier AAM in girls, and exposure to DBP leads to delayed AAM, with a BMI potentially mediating these effects. Our findings contribute to the current knowledge on the effects of EDCs on human pubertal timing.

Optional Non-Accredited Industry Symposia (All times are listed in local time)

Thursday, February 6, 2025 (Room: Salon D)

Lunch Session: 1145-1215

Recent Updates in the Diagnosis and Management of XLH

An update on recent data and publications in the diagnosis and management of XLH.

Dr Leanne Ward MD FRCPC is a world-renowned Pediatric Endocrinologist and Researcher with a special interest in rare metabolic bone disease. She is currently a Professor at the Department of Pediatrics with cross-appointment to the Department of Surgery, Faculty of Medicine, University of Ottawa.

Dr. Leanne M. Ward
MD FRCPC
Professor of Pediatrics,
Senior Research Chair in Pediatric Bone Disorders,
University of Ottawa
Medical Director, Pediatric Genetic and Metabolic Bone Disease Program

Friday, February 7, 2025 (Room: Salon D)

Breakfast Session: 0745-0815

Should We Be Testing for and Encouraging Adult GH Replacement Prior to Transition?

In this session, participants will:

- Discuss the potential benefits of treatment of childhood-onset adult growth hormone deficiency
- Clarify how diagnosis of adult growth hormone deficiency is made after cessation of childhood growth hormone treatment
- Review existing guidelines regarding transition of pediatric growth hormone deficiency patients and discuss our role as paediatric endocrinologists

Dr. Preetha Krishnamoorthy
MDCM FRCPC
Associate Professor,
McGill University
Pediatric Endocrinologist,
The Montreal Children's Hospital

Optional Non-Accredited Industry Symposia (All times are listed in local time)

Friday, February 7, 2025 (Room: Salon B1)
Lunch Session: 1315-1345

Sharing Personalized Strategies to Advance Pediatric Obesity Care

Join us in an ongoing, inspiring journey to enable pediatric obesity care. This session explores the HEAL program's journey in pediatric weight management, the complexities of obesity as a chronic, relapsing and multifactorial disease, and the power of personalized care with an openness for audience interaction. Learn about how you can use genetic testing, multidisciplinary care, and advanced therapies, to empower and bring hope to families navigating the challenges of obesity with tailored solutions.

In this session, participants will:

- Share the HEAL (Healthy Eating, Activity, Lifestyle) Program's evolving journey in pediatric weight management
- Look at personalized care strategies for diagnosing and treating rare genetic obesity
- Explore evidence-based approaches aimed at enhancing care for patients and families

Dr. Marina Ybarra
MD MSc
Director, Pediatric Weight Management Program
London Health Sciences Centre - Children's Hospital
Assistant Professor, Dept. of Pediatrics, Western University

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