CPEG GCEP

Canadian Pediatric Endocrine Group
Groupe canadien d'endocrinologie pédiatrique

2019

SCIENTIFIC MEETING

PROGRAM

FEBRUARY 21 – 23, 2019

The Westin Ottawa
Ottawa, ON

In cooperation with

THE UNIVERSITY OF BRITISH COLUMBIA
Interprofessional Continuing Education

Welcome

Dear Delegates,

I would like to extend to you a warm welcome to the 13th Annual Scientific Meeting of the Canadian Pediatric Endocrine Group (CPEG). Our past meetings have provided a wonderful opportunity for the Canadian pediatric endocrine community to come together to learn, network and share ideas. The organizing committee has worked hard to craft a program that highlights local Ottawa work and includes presentations by national and international experts. It also provides a forum for our fellows to present their work. We have an exciting program that we hope will meet the educational needs of our nurses, scientists, endocrinologists and trainees.

I would like to thank our sponsors, who make this meeting possible. I would also like to thank those companies who sponsor our CPEG Fellowship Awards and allow us to train endocrinologists for the future. We look forward to the award announcements at this meeting.

I wish you a stimulating and collegial meeting.

Sincerely,

Bienvenue

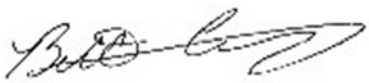
Chers délégués,

Je tiens à vous accueillir chaleureusement à la 13ème réunion scientifique annuelle du Groupe canadien d'endocrinologie pédiatrique (GCEP). Nos dernières réunions ont été d'excellentes occasions, pour la communauté canadienne d'endocrinologie pédiatrique, pour se réunir afin d'apprendre, de réseauter et de partager nos idées. Le comité organisateur a travaillé fort pour concevoir un programme qui met en lumière les travaux des gens de Ottawa ainsi que ceux d'experts nationaux et internationaux. Il fournit également un forum pour que nos « fellows » aient l'occasion de présenter leurs travaux. Nous avons un programme captivant qui, nous l'espérons, répondra aux besoins éducatifs du personnel infirmier, des chercheurs, des endocrinologues et des étudiants du domaine de l'endocrinologie.

Je tiens à remercier nos commanditaires, qui rendent cette rencontre possible. Je tiens aussi à les remercier pour le soutien financier qu'ils offrent à notre programme de bourses CPEG; un programme qui nous permet de former les endocrinologues de demain. Nous attendons d'ailleurs avec impatience l'annonce des récipiendaires de cette année lors de ce congrès.

Je vous souhaite une réunion agréable et stimulante.

Bien cordialement,



Beth Cummings, MD, FRCPC
Scientific Chair, CPEG 2019 Scientific Meeting

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Financial Contributors

We would like to acknowledge with great appreciation the financial contributions through unrestricted educational grants from the following organizations:

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ALEXION

omnipod INSULIN MANAGEMENT SYSTEM

SANDOZ A Novartis Division

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
2006-2007	B. Wicklow S. Amed
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. Édouard 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

The CPEG Fellowship Program was and/or is supported by the following:
Eli Lilly, EMD Serono, Hoffmann La Roche, Novo Nordisk, Pfizer, and Sandoz

Program

Please note: 25% of the scientific program will be interactive.

Thursday, February 21, 2019

Time	Session
08:30	CPEN Executive Business Meeting (ROOM: Alberta)
12:00	CPEG Executive Business Meeting (ROOM: New Brunswick)
	Fellows Symposium (for CPEG Fellows only)
13:00	Welcome
13:20	Going MENtal: Overcoming your fear of Hereditary Endocrine Neoplasia Syndromes *Jonathan Wasserman*
14:30	Refreshment Break
14:50	Transition to Practice and Navigating the Job Market – Advice From A New Staff *Scott Somerville*
15:50	Conclusion
16:00	CPEG 2019 Registration Opens
17:00	Welcome Reception & Exhibits
19:00	Adjourn

Friday, February 22, 2019

Time	Session
07:00	Registration (Room: Provinces/Confederation I/II Foyer) Breakfast & Exhibits (Room: Confederation I)
08:00	Opening Remarks & Welcome (Room: Confederation II) *Beth Cummings, Caroline Zuijdwijk, Alice Boland* Friday Poster Highlights *Each poster presenter will give a 1-minute & 1-slide presentation.*

08:30 THEME I: New Horizons in Pediatric Bone Diseases (Room: Confederation II)
Moderator: Jennifer Harrington

FGF23-Mediated Hypophosphatemia: From Discovery to Therapy
Leanne M. Ward

Diagnosing and Treating Osteoporosis in Children
Frank Rauch

10:00 Break and Exhibits (Room: Confederation I)

10:30 THEME II: Adverse Childhood Experiences and Endocrine Disease—Evidence and Opportunities (Room: Confederation II)
Moderator: Daniele Pacaud

Adverse Childhood Experiences: Key determinants of metabolic health and treatment response among adolescents
Jon McGavock

CPS and CPEG Working Together for Policy Change
Marie Adèle Davis

12:00 Poster Viewing (Room: Provinces II)

12:30 Lunch & Exhibits (Room: Confederation I)

13:30 Nurses split, see page 7

THEME III: Complications of Diabetes (Room: Confederation II)
Moderator: Judith Simoneau

Early Treatment of Adolescents using ACE-I and Statins: Insights from the Adolescent Diabetes Cardiorenal Intervention Trial (AdDIT)
Farid H. Mahmud

Time to Shift Fluid Management in DKA
Sarah Lawrence, Mona Jabbour

15:00 Break & Exhibits (Room: Confederation I) & Poster Viewing (Room: Provinces II)

15:30 Oral Abstract Presentations (6) (Room: Confederation II)

17:00 Adjourn

FRIDAY NIGHT EVENT
Dinner and Entertainment at the National Arts Centre (1 Elgin St, Ottawa, ON K1P 5W1)
(500m walk from the hotel)
18:00 – 18:30 Cocktail Reception
18:30 – 19:15 Entertainment: Escape Manor
19:15 Dinner & Dessert

*Nursing Program for Friday, February 22 & Saturday, February 23 (Provinces I)

Moderators: Ms. Sara Chang & Ms. Alice Boland

*Nursing Program for Friday, February 22

13:30	Roundtable Discussion for Transgender Patients
14:15	Approach to Transgender Patients *Sebastien Pangallo, Debbie Turner*
15:00	Break & Exhibits (Room: Confederation I)
15:30	Motivational Interviewing with Adolescent Patients *Katie Frost*
17:00	Adjourn
18:00	FRIDAY NIGHT EVENT Please see above

*Nursing Program for Saturday, February 23

13:30	CPEN AGM
15:00	Break and Exhibits (Room: Confederation I)
15:35	Re-join CPEG group

Saturday, February 23, 2019

Time	Session
07:30	Breakfast (Room: Confederation I)
08:00	Business Meeting (Room: Confederation II)
09:55	Fellowship Awards *Presented by Dr. Carol Huang*
10:00	Break and Exhibits (Room: Confederation I)
10:30	THEME IV: Congenital Hypopituitarism and Septo-optic Dysplasia – The who, what, when and how (Room: Confederation II) *Moderator: Seth Marks* *Presented by Sally Radovick, Shazhan Amed, Brandy Wicklow*
12:00	Saturday Poster Highlights (Room: Confederation II) Each poster presenter will give a 1-minute & 1-slide presentation.
12:15	Poster Viewing (Room: Provinces II)
12:45	Lunch & Exhibits (Room: Confederation I)
13:30	Oral Abstract Presentations (6) (Room: Confederation II) Nurses split, see page 7
15:00	Break & Exhibits (Room: Confederation I) & Poster Viewing (Room: Provinces II)
15:30	John Bailey Award (Room: Confederation II) *Presented by Laurent Legault*
15:35	Debate: Be it resolved that the ketogenic diet is safe and effective in the management of pediatric type 1 diabetes (Room: Confederation II) *Moderator: Jill Hamilton* *PRO: Caroline Zuijdwijk* *CON: Julia von Oettingen*
16:35	Closing Remarks & Evaluation (Room: Confederation II)
16:45	Adjourn

Fellow (Oral) Abstract Schedule

Time	Title	Presenter	Abstract #	Page
Friday, February 22 *Moderators: Sue Stock & Rose Girgis*				
15:30	Are intra-articular injections safe?	Marc-Antoine Bédard	1	22
15:45	Re-Evaluation of the 17-Hydroxyprogesterone (17-OHP) Screening Threshold for Diagnosing Non-Classic Congenital Adrenal Hyperplasia (NCCAH) in the Era of Liquid Chromatography Tandem-Mass Spectrometry (LC-MS/MS)	Alexander D. Chesover	2	23
16:00	The GLP-2 receptor modulates the inflammatory response to diet-induced liver injury	Shai Z. Fuchs	3	24
16:15	Monogenic Diabetes Misdiagnosed as Type 1 Diabetes	Claire Lange	4	25
16:30	Interaction between the development of the thyroid and of pharyngeal vessels: from exome sequencing in humans to validation of candidate genes in zebrafish	Stéphanie Larrivée-Vanier	5	26
16:45	Gender Diversity Training in Canadian Pediatric Postgraduate Medical Education: A Needs Assessment Survey	Alexa Marr	6	27

Saturday, February 23 *Moderators: Rachel Scott & Andrea Ens*				
13:30	Effects of Pubertal Suppression with GnRH Analogs on Body Composition and Body Mass Index of Transgender Adolescents	Behdad Navabi	7	28
13:45	Residence in Haiti vs. Quebec impacts phenotypes of diabetes and glycemic control in Haitian youth	Phung K, Sainvil	8	29
14:00	Osteogenesis Imperfecta: Skeletal Outcomes After Bisphosphonate Discontinuation at Final Height	Marie-Eve Robinson	9	30
14:15	Does Age Matter? Mental Health Implications and Determinants of When Youth Present to a Gender Clinic	Julia C. Sorbara	10	31
14:30	Effect of Sensor-Augmented Pump Therapy on Fear of Hypoglycemia in Children with Type 1 Diabetes and their Parents in the CGM TIME Trial	Kate C. Verbeeten	11	32
14:45	Outcomes and experiences of young adults with type 1 diabetes transitioned from pediatric to adult care: a cross sectional assessment	Annie Lu	12	33

Poster Abstract Listing

Please note that abstract poster presenters for #1-15 will be standing by their poster on Friday, February 22 at 12:00 – 12:30 and #16-28 will be on Saturday at 12:15-12:45

Program Organizing and Scientific Committee

Ereny Bassilious
Alice Boland
Sara Chang
Beth Cummings
Ellen Goldbloom
Brenden Hursh
Karine Khatchadourian
Laurent Legault
Seth Marks
Susan Murphy
Jo Nam
Julia Sorbara
Caroline Zuijdwijk

Credits

This event has been approved by the Canadian Paediatric Society for a **maximum of 10.5 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.

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
- 2008 Meranda Nakhla
- 2009 David Saleh
- 2010 Brandy Wicklow
- 2011 Jonathan Wasserman
- 2012 Jennifer Harrington
- 2013 Karine Khatchadourian
- 2014 Akash Sinha
- 2015 Rayzel Shulman
- 2016 Sanjukta Basak
- 2017 Stephen Zborovski
- 2018 Marie Eve-Robinson

CPEG Distinguished Service Award

The CPEG Distinguished Service Award will be awarded periodically (not annually) to a member who has shown exemplary service to the organization or to the discipline of pediatric endocrinology in Canada. The award will be focused on work that furthers the aims of CPEG and can be in one or more of the following areas: administration, teaching, research, clinical service. Nominations will be solicited by the CPEG Executive Committee every 2–4 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 $500 donation to a charity of their choice.

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

- 2017 – Daniel Metzger

Learning Objectives

The overall learning objective of this meeting is to present the current state of knowledge of topics in pediatric endocrinology and diabetes.

Session Learning Objectives:

Fellows Symposium

Going MENtal: Overcoming your fear of Hereditary Endocrine Neoplasia Syndromes

***Jonathan Wasserman**, MD, PhD, Staff Endocrinologist, The Hospital for Sick Children; Assistant Professor, Department of Paediatrics, The University of Toronto, Toronto, Ontario*

1. Recognize specific diagnoses or patterns that raise concern for hereditary endocrine tumour syndromes
2. Identify and utilize available resources to guide pre-symptomatic surveillance and management of affected patients
3. Interpret genetic testing and apply genotype-phenotype relationships to guide care of affected individuals
4. Appreciate risks and ethical challenges associated with genetic testing and tumour surveillance as applied to these conditions

Transition to Practice and Navigating the Job Market – Advice From A New Staff

***Scott Somerville,** MD, FRCPC, Pediatric Endocrinology Group, Children's Hospital of Eastern Ontario; Lecturer, Department of Pediatrics, The University of Ottawa, Ottawa, ON*

1. Describe classic and alternative routes to an academic position in endocrinology
2. Discuss additional strategies to increase value to a program
3. Explain new financial and management issues associated with becoming staff

Theme I: New Horizons in Pediatric Bone Diseases

FGF23-Mediated Hypophosphatemia: From Discovery to Therapy

***Leanne M. Ward,** MD, FRCPC, Professor of Pediatrics, Research Chair in Pediatric Bone Health, University of Ottawa; Director, Pediatric Bone Health and Rare Bone Disease Clinical and Research Programs, Division of Endocrinology and Metabolism, Children's Hospital of Eastern Ontario, Ottawa, ON*

1. Review the discovery of FGF23 as a phosphaturic hormone, and the mechanisms by which FGF23 regulates phosphate metabolism
2. Discuss the approach to the diagnosis of FGF23-mediated hypophosphatemic disorders
3. Summarize the latest research on conventional and novel FGF23-neutralizing antibody therapy for the treatment of x-linked hypophosphatemia.

Diagnosing and Treating Osteoporosis in Children

***Frank Rauch,** MD, Shriners Hospital for Children, Professor of Pediatrics, McGill University, Montreal, QC*

1. Discuss the use of bone densitometry for the assessment of pediatric osteoporosis
2. Recognize the role of molecular genetic testing in the diagnosis of pediatric osteoporosis
3. Compare the medical treatment options for pediatric osteoporosis

Theme II: Adverse Childhood Experiences and Endocrine Disease—Evidence and Opportunities

Adverse Childhood Experiences: Key determinants of metabolic health and treatment response among adolescents

Jon McGavock, PhD, CIHR Applied Public Health Chair, Associate Professor, Department of Pediatrics, Faculty of Health Sciences, University of Manitoba, Winnipeg, MB

1. Summarize the ACEs study and systematic reviews of adverse experiences in childhood and metabolic health later in life
2. Review the bio-psycho-social factors linking adverse experiences in childhood and metabolic disease
3. Review trauma-informed approaches to clinical management of chronic endocrine disorders in adolescence and young adulthood.

CPS and CPEG Working Together for Policy Change

Marie Adèle Davis, Executive Director, Canadian Paediatric Society, Ottawa, ON

1. Understand how CPS and CPEG can use ACES evidence to inform our joint advocacy efforts.
2. Discuss how to best engage policy makers to work for change that impacts children and youth.
3. Understand how to leverage partnerships to best influence governments and policy makers for positive change.

Theme III: Complications of Diabetes

Early Treatment of Adolescents using ACE-I and Statins: Insights from the Adolescent Diabetes Cardiorenal Intervention Trial (AdDIT)

Farid H. Mahmud, MD, FRCPC, Associate Professor, Division of Endocrinology, Hospital for Sick Children and University of Toronto, Toronto, ON

1. Describe the onset and progression of diabetes related kidney disease during adolescence
2. Share the rationale and design of the AdDIT study
3. Discuss the results of the AdDIT trial
4. Reflect on the context of these data and future perspectives

Time to Shift Fluid Management in DKA

Sarah Lawrence, MD, FRCPC, Chief, Division of Endocrinology and Metabolism, Children's Hospital of Eastern Ontario; Associate Professor, Department of Pediatrics, University of Ottawa, Ottawa, ON

Mona Jabbour, MD, FRCPC, Co-director of Implementation for TREKK, Vice Chair, Department of Pediatrics, Children's Hospital of Eastern Ontario; Associate Professor, Department of Pediatrics, University of Ottawa, Ottawa, ON

1. Review the pathophysiology of pediatric DKA and cerebral injury
2. Describe new evidence from the PECARN FLUID trial supporting more aggressive fluid management in pediatric DKA
3. Demonstrate how this practice-changing evidence is being translated to emergency departments and pediatricians across the country

Theme IV: Congenital Hypopituitarism and Septo-optic Dysplasia – The who, what, when and how

Sally Radovick, MD, Professor and Henry Rutgers Term Chair, Department of Pediatrics, Senior Associate Dean for Clinical and Translational Research, Robert Wood Johnson Medical School; Physician-in-Chief, Bristol Myers-Squibb Children's Hospital Chancellor's Scholar, Rutgers Biomedical and Health Sciences, Child Health Institute of New Jersey-Rutgers University, New Brunswick, NJ

Shazhan Amed, MD, Investigator, BC Children's Hospital Affiliate of the Centre for Health Services and Policy Research, University of British Columbia, Vancouver, BC

Brandy Wicklow, MD, Pediatric Endocrinologist at HSC Children's; Assistant Professor, Department of Pediatrics and Child Health, College of Medicine, University of Manitoba, Winnipeg, MB

1. Outline the genetics involved in congenital hypopituitarism.
2. Review the risk factors associated with the development of pituitary dysfunction in children with septo-optic dysplasia.
3. Describe the characteristics of children diagnosed with septo-optic dysplasia.

Debate:

Be it resolved that the ketogenic diet is safe and effective in the management of pediatric type 1 diabetes

PRO: Caroline Zuijdwijk, MD, FRCPC, Pediatric Endocrinologist, Division of Endocrinology & Metabolism, CHEO; Assistant Professor, Department of Pediatrics, University of Ottawa, Ottawa, ON
CON: Julia von Oettingen, MD PhD MMSc FRCP, Pediatric Endocrinologist, Montreal Children's Hospital; Assistant Professor, McGill University

1. List the potential benefits of the ketogenic diet in the management of diabetes
2. List the potential risks of the ketogenic diet in the management of diabetes
3. Discuss how this can be applied to our management of children with type 1 diabetes

Biographies

Shazhan Amed

Dr. Amed is a pediatric endocrinologist at BC Children's Hospital, a Clinical Associate Professor at UBC, an Associate Clinician Scientist at the BC Children's Hospital Research Institute, and Acting Head of the Division of Endocrinology at BC Children's Hospital.

Dr. Amed completed medical school at the University of Calgary (Calgary, Alberta), Pediatric Residency at the University of Manitoba (Winnipeg, Manitoba), and her Endocrinology Fellowship at The Hospital for Sick Children (Toronto, Ontario). She has also completed a Masters of Science in Public Health at the London School of Hygiene and Tropical Medicine (London, England, U.K). Dr. Amed is a health services and population health researcher. She is the Founder and Lead of Live 5-2-1-0 – an initiative that works with communities to prevent childhood obesity and youth-onset type 2 diabetes. For this work, she recently received annual funding from the BC Ministry of Health. She also leads a childhood diabetes research program that uses linked administrative data combined with clinical and patient-level data to conduct population-level surveillance of childhood type 1 and type 2 diabetes and implement and evaluate quality improvement initiatives related to health service delivery for children and youth with diabetes. Over the last 5 years, she has published over 25 articles in high impact diabetes and pediatrics journals and has been successful in securing almost 2.5 million dollars in grant funding from national granting agencies such as the CIHR, Diabetes Canada, Lawson Foundation, and Public Health Agency of Canada.

Marie Davis

Marie Adèle Davis is the Executive Director at the Canadian Paediatric Society. She received a B.Sc. in1986 from McGill University. Upon finishing at McGill, Marie Adèle successfully completed her Masters of Business Administration at INSEAD in Fontainebleau, France, in December 1987. She spent time as a management consultant with Monitor Company in Toronto and Boston. An interest in not-for-profit work took Marie Adèle to Ottawa in 1989 where she worked for the Ottawa General in a number of roles including Associate Vice President of Medical Affairs for the Ottawa Hospital from 1998-99.

Marie Adèle has been the Executive Director of the Canadian Paediatric Society since 1999. The Canadian Paediatric Society is a national advocacy organization committed to the health needs of children and youth. Marie Adèle is a member of the Strategy & Connected Care Committee of the Children's Hospital of Eastern Ontario and the Advisory Committee for the Ontario Centre of Excellence for Child and Youth Mental Health. Once a week, she volunteers at the Montfort Hospital in the Mental Health Unit.

Katie Frost

Katie is a professional coach and workshop facilitator. She specialises in working with young people, supporting them to find their unique strengths, confidence, direction and motivation. Taking a psychologically informed approach, she enjoys working with hard to reach young people to support them to achieve their goals and potential. Katie is interested in addressing the emotional barriers that can get in the way of people making positive changes in their lives.

Katie is passionate about learning, is a member of the Association of Coaching and holds a Diploma in Coaching from the University of Cambridge. Trained in motivational interviewing she practices these techniques within her coaching practice and is qualified in a range of psychometric tools.

Mona Jabbour

Dr. Mona Jabbour earned her MD at McMaster University in Hamilton, followed by training in pediatrics at the University of Ottawa in 1992. She completed her Master's of Higher Education with specialization in Health Professions Education at the University of Toronto in 1997. Dr. Jabbour's interests in education revolve around development and evaluation of iLearn-Peds, a pediatric curriculum based on e-learning modules, simulation training and outreach education. Her research interests include: improving systems of care, mobilizing best evidence to practice settings and emergency department management of children and youth presenting with mental health concerns. Dr. Jabbour is involved in TRanslating Emergency Knowledge for Kids (TREKK), a national network of pediatric emergency and community emergency department sites that have been created to mobilize knowledge on best pediatric emergency care. She is also involved in a study to implement clinical pathways in community emergency department settings.

Sarah Lawrence

Sarah Lawrence is the Division Head for Pediatric Endocrinology at CHEO where she has practiced since 1995. She completed medical school at Dalhousie University, pediatrics residency at the University of Ottawa and endocrinology training at the Montreal Children's Hospital, McGill University. She collaborated with Beth Cummings and Denis Daneman on a CPSP surveillance study of risk factors for cerebral edema in DKA in 2005. More recently she has collaborated with colleagues in CPEG and the TREKK group on national recommendations for DKA management based on recent evidence.

Jon McGavock

Jon McGavock is a settler scientist from Treaty 1 territory. He is the co-lead of the DREAM theme, the Indigenous Patient Goal Group within Diabetes Action Canada and holds a CIHR Applied Health Chair in Resilience and Obesity in Youth. His work revolves around developing novel approaches to improving health outcomes among youth, particularly Indigenous youth, living with obesity and type 2 diabetes. His lab is funded by CIHR, Diabetes Action Canada, Diabetes Canada, the Heart and Stroke Foundation of Canada and the Lawson Foundation.

Farid Muhmud

Farid Mahmud MD FRCPC is currently Staff Physician in the Division of Endocrinology, Department of Paediatrics and Associate Professor at the University of Toronto and Associate Scientist at The Hospital for Sick Children Research Institute. Dr. Mahmud's overall research focus is diabetes, clinical and translational research, relating to co-morbid autoimmune conditions and early evaluation and prevention of related complications. His research interests include the evaluation of interventions in high risk pediatric groups and the evaluation of impact of the Social Determinants of Health in chronic disease populations. Dr. Mahmud serves as Principal Investigator of the Adolescent Diabetes Cardio-Renal Intervention Trial (AdDIT) in Canada and as PI of the Celiac Disease and Diabetes - Dietary Intervention and Evaluation Trial (CD-DIET).

Sebastien Pangallo

Graduated from the University of Ottawa with a Master of Social Work in 2012. Has been working at CHEO since 2012 and worked in various clinics until becoming the Adolescent Health social worker in 2013. Primary focus has been on adolescence living with chronic illness, more specifically visually impaired adolescence. In the past years has had more of a focus on supporting gender diverse youth and their families.

Sally Radovick

Sally Radovick, MD, received her medical degree from Northeastern Ohio Universities College of Medicine. She then completed her residency in Pediatrics at Case Western Reserve University and her fellowship in Pediatric Endocrinology at the National Institutes of Health (NIH). She is currently the Chair of Pediatrics and Senior Associate Dean for Clinical and Translational Research at Rutgers-Robert Wood Johnson Medical School. Prior to this position, she was the Division Director of Pediatric Endocrinology and the Vice Chair for Research in the Department of Pediatrics at Johns Hopkins University School of Medicine.

Frank Rauch

Frank Rauch, MD, is a Professor of Pediatrics and clinician-scientist at the Shriners Hospital for Children and at McGill University. He obtained his MD degree from the Technical University of Munich, and trained as a pediatrician at the Children's Hospital of Cologne University, Germany. His clinical activities and research program concentrate on improving bone health in children, with a special focus on genetic conditions leading to fractures and on the role of the muscle system in bone diseases. In his recent work, Dr. Rauch has identified new genetic causes of brittle bone disorders and has assessed the long-term effects of bisphosphonate treatment in children with osteogenesis imperfecta. He is also collaborating with Statistics Canada in a study that assesses muscle and bone health in Canadians. Dr. Rauch has authored or coauthored more than 200 original publications.

Scott Somerville

Scott Somerville is currently a new pediatric endocrinology staff at the Children's Hospital of Eastern Ontario. He completed his an Honours Specialization in Physiology and his Doctor of Medicine at the University of Ottawa. He completed a 3 year pediatric residency program at CHEO, 2 years of Endocrinology fellowship at The University of Alberta, and returned to CHEO in 2016. His research interests include Turner Syndrome, Transgender Medicine, and Oncologic-Endocrinology. His academic interests include a strong basis in teaching and quality improvement.

Debbie Turner

Debbie Turner is a registered nurse with over 28 years of nursing experience. Started out with the adult population at the Ottawa hospital and switched to Pediatrics at the Children's Hospital of Eastern Ontario in 1992. Worked at the bedside in acute care medicine for over 23 years. A recent switch to the Adolescent Health Clinic in 2014 has brought a new focus on outpatient case management. This most recent position brings with it a focus on youth including those exploring gender and diversity.

Julia von Oettingen

Julia von Oettingen is a pediatric endocrinologist at the Montreal Children's Hospital, Assistant Professor at McGill University, and a Fonds de Recherche du Quebec Sante supported Clinician-Scientist at the McGill University Health Center Research Institute. Originally from Germany, she completed her MD-PhD at Leipzig University, her pediatric residency at the Massachusetts General Hospital in Boston, and her endocrine fellowship training at the Boston Children's Hospital. She obtained her master's in clinical and translational investigation from Harvard University. Dr. von Oettingen's research program focuses on global health in pediatric endocrinology, including epidemiology and phenotypes of diabetes in non-Caucasian populations, and care delivery innovation in low-resource settings. She is the founding medical director of Kay Mackenson Clinic, a center for children with diabetes and other chronic diseases in Haiti that collaborates with the Haitian Diabetes Association to provide access to quality diabetes care countrywide. Julia is a site visitor and serves on the steering committee of the Life for a Child program, and is an executive committee member of Global Pediatric Endocrinology and Diabetes. She is a technical adviser to Partners in Health in Haiti,

consultant to UNICEF and the Ministry of Health in Haiti, and pediatric endocrinology consultant to Medecins Sans Frontieres.

Leanne Ward

Dr. Leanne Ward is a Professor of Pediatrics at the University of Ottawa where she has held a Research Chair in Pediatric Bone Health since 2010. She is the Medical Director of the Pediatric Bone Health and Rare Bone Disease Clinical and Research Programs at the Children's Hospital of Eastern Ontario, and a pediatric endocrinologist in the Division of Endocrinology and Metabolism. Dr. Ward's research program is dedicated to the study of bone development and the diagnosis and treatment of bone disorders in children. She has been the principal investigator of the "STOPP" research program (STeroid-induced Osteoporosis in the Pediatric Population), a pan-Canadian project funded by the Canadian Institutes of Health Research to evaluate the effect of glucocorticoids on bone health in children with chronic illnesses. Dr. Ward actively leads and collaborates on a number of clinical trials for children with osteogenesis imperfecta, rickets and chronic illness osteoporosis. She has served as an endocrinology and bone health advisor to various international organizations on skeletal health in children, including the Centres for Disease Control Clinical Care Guidelines for Duchenne Muscular Dystrophy, the International Late Effects of Childhood Cancer Guideline Harmonization Group and the International Conference on Children's Bone Health. Dr. Ward has received a number of awards for her work in pediatric bone health, including a Canadian Child Health Clinician Scientist Career Development Award, a Canadian Institutes for Health Research New Investigator Award, a Canadian Child Health Clinician Scientist Career Enhancement Award, and two, five-year Research Chairs in Pediatric Bone Health (University of Ottawa, 2010 and 2015).

Jonathan Wasserman

Dr. Wasserman obtained his BSc in Physiology from McGill University and earned a PhD in genetics from Cambridge University. 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 then served as a Paediatric Hospitalist. He returned to Toronto in 2008 to pursue a fellowship in Pediatric Endocrinology and has been on staff in the Endocrine division at SickKids since July 2012.

Dr. Wasserman is a Clinician-Investigator with an academic focus on pediatric endocrine malignancies. He uses basic and translational approaches, as well as outcomes analysis and health services research to explore this field. His group is working to define the molecular alterations underlying pediatric thyroid neoplasms as applied to both diagnostics and therapeutics. They have also been looking at test characteristics of diagnostic modalities applied to pediatric thyroid nodules and have demonstrated that adult classifications underestimate the likelihood of malignancy in children. Finally, they are leveraging administrative health databases to extrapolate health care utilization patterns and outcomes for individuals treated for thyroid carcinoma in childhood.

In addition to his endocrine practice, he sees patients as part of SickKids' Cancer Genetics Program and has co-authored several recent consensus guidelines regarding pre-symptomatic surveillance for children with hereditary cancer syndromes.

Brandy Wicklow

Brandy Wicklow is a Pediatric Endocrinologist at the Winnipeg Children's Hospital, Associate Professor at the University of Manitoba, and Clinician Scientist at the Children's Hospital Research Institute of Manitoba (CHRIM). Her research is focused on the determinants of type 2 diabetes (T2D) in children, with a particular interest in the Indigenous population of Northern Manitoba, Canada with whom she works closely both in clinical care and research. She is the Principle Investigator of a birth cohort of children born to mothers and fathers diagnosed with childhood T2D (The Next Generation Cohort)

examining the effects of in utero T2D exposure on growth, development and the natural history of T2D in offspring. She is the co-lead of the iCARE (Improving Renal Complications in Adolescents with Type 2 Diabetes through Research) with Dr Allison Dart; a cohort study which aims to determine modifiable risk factors in the natural history of diabetes related renal disease.

Caroline Zuijdwijk

Dr. Zuijdwijk received her medical degree from McMaster University and completed her pediatric residency at Memorial University. She then completed her Pediatric Endocrinology fellowship at CHEO (University of Ottawa), following which she pursued a research fellowship at the Hospital for Sick Children (University of Toronto). She joined the Division of Endocrinology & Metabolism at CHEO and became an Assistant Professor of Paediatrics at the University of Ottawa in 2012. Her principal research interest is in quality improvement in pediatric type 1 diabetes, both locally and abroad.

Conflict of Interest Disclosures

All speakers and committee members must disclose whether they do or do not have a relationship with a commercial entity such as a pharmaceutical organization, medical device company or a communications firm.

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- I am a member of the advisory board for Canada Medtronic.

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- I have received an honorarium for my participation in the advisory board for Genotropin Pediatric Nurse Consultant Meeting.

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- I have received a grant or an honorarium from Novo Nordisk.

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- I have received payment in the form of travel support from Novo Nordisk.
- I received a speaker honorarium from Eli Lilly.
- I hold a patent for a product that is marketed by a commercial organization.
- My institution received payment for my participation in a clinical train with Merck, Asta Zeneca, Sanofi, Novo Nordisk and Verastis.

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- My institution has received payment for bone health imaging services for a regulated clinical trial from Raveragen and Catabasis.
- My institution has received an unrestricted educational grant from Ultragenyx.
- My institution has received an honorarium for my participation on an Advisory Board with Novartis, Amgen, Alexion and Ultragenyx.
- My institution has received payment for current participation in bone health clinical trials with Novartis, Ultragenyx and Amgen.

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Caroline Zuijdwijk
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- I am currently participating in a clinical trial for Medtronic. I am a CHEO co-investor to Multi-Centre RCT: Home use Hybrid Closed Loop System.

Oral Abstracts

Oral Abstract 1
Are intra-articular injections safe?

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Background: Intra-articular corticosteroid injection (IACI) is a common procedure in the care of children with juvenile idiopathic arthritis. The main objective of this procedure is to relieve local inflammation while minimizing systemic absorption. Despite adult studies showing a significant risk of adrenal insufficiency (AI), no monitoring is recommended. Data in children is very limited. The main objective of this study is to evaluate if IACI causes AI at two weeks in children with inflammatory arthritis. Secondary endpoints are risk factors, duration, symptoms and complications associated with AI.

Methods: We conducted a retrospective study including children 0-18 years who had an IACI at the CHU de Québec (Université Laval) since June 2017. A 0800-hour am cortisol (8HC) was performed two weeks after the injection and ACTH 1ug stimulation test was performed if 8HC level was low as per the endocrinologist. AI was defined as an 8HC under 50 nmol/L or an abnormal ACTH testing. Exclusion criteria were: children who received corticosteroids in the past three months (IACI, IV, IM, intranasal, ocular or inhaled), congenital disorders of steroidogenesis and patient with inadequate 8HC sampling and no confirmatory ACTH stimulation test.

Results: 53 children have received IACI at this point. 43 of these patients are included for analysis (10 patients are excluded or have pending results). There are 28 girls (65,1%) and 15 boys (34,9%), with a mean age of 10,2 years. The vast majority (37/43; 86%) received an injection of triamcinolone hexacetonide. The principal diagnostic of this population was idiopathic juvenile arthritis (41/43; 95,3%). 18 of the 43 patients (41,9%) developed AI. Mean duration of AI in patients with complete follow-up (6/18) was 111 days, with a range between 28 and 190 days. 4 patients had symptom of AI, namely fatigue (2/4), nausea (2/4) and abdominal pain (3/4). None were hospitalized. Further analysis are in progress to evaluate other secondary outcomes.

Conclusions: In this cohort of children who had an IACI, we found a high prevalence of AI. Monitoring of these patients seems warranted until further evidence is available.

Oral Abstract 2

Re-Evaluation of the 17-Hydroxyprogesterone (17-OHP) Screening Threshold for Diagnosing Non-Classic Congenital Adrenal Hyperplasia (NCCAH) in the Era of Liquid Chromatography Tandem-Mass Spectrometry (LC-MS/MS)

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Background: NCCAH is characterized by reduced 21-hydroxylase activity leading to elevations in adrenal androgens. It may be associated with an inadequate cortisol response to ACTH stimulation. A screening 17-OHP >6 nmol/L based on immunoassay prompts NCCAH diagnostic testing with an ACTH stimulation test. Peak 17-OHP <30 nmol/L post-ACTH excludes NCCAH. In 2011, we replaced immunoassay with LC-MS/MS for 17-OHP quantification which has greater specificity. A method comparison revealed a negative bias between LC-MS/MS and immunoassay measurements.

Aims: To evaluate the 17-OHP threshold that predicts genetically-confirmed NCCAH. A secondary objective was to determine the prevalence of adrenal insufficiency (AI) in this population.

Methods: A retrospective chart review was performed of clinical and genetic data for patients <18 years who underwent ACTH stimulation tests with cortisol and 17-OHP measurements from 2011 to 2018. NCCAH was genetically confirmed; other adrenal pathologies were excluded. A peak cortisol < 500 nmol/L, measured by immunoassay, defined AI. Using correlation data between immunoassay and LC-MS/MS, a new 17-OHP threshold of 3.3 nmol/L for the LC-MS/MS method was considered equivalent to 6 nmol/L by immunoassay; similarly, 20 nmol/L was equivalent to 30 nmol/L.

Results: 188 patients met inclusion criteria. 23 (12%) had NCCAH of which 21/23 had genetic confirmation; the remaining 2 had peak 17-OHP >30 nmol/L by LC-MS/MS. Baseline 17-OHP >6 nmol/L most accurately diagnosed NCCAH (sensitivity and specificity 96%) with no improvement using 17-OHP >3.3 nmol/L. 20/21 genetically confirmed NCCAH had peak 17-OHP >30 nmol/L. Of three with 17-OHP peak 20-30 nmol/L, one was a CAH carrier, one had other adrenal pathology, and another with unknown diagnosis. 87% with NCCAH had biochemical AI. Baseline 17-OHP >6 nmol/L predicted all with AI.

Conclusion: Despite increased specificity of LC-MS/MS for steroid measurements, a baseline 17-OHP >6 nmol/L remained the most accurate predictor of NCCAH. This threshold also predicts all with AI, notably prevalent in our cohort. Analysis is limited by a small cohort, low NCCAH incidence and limited genetic data in those presumed without NCCAH. However, this re-evaluation of screening and diagnostic thresholds is important in the era of evolving assays and these results support current practice guidelines.

Oral Abstract 3
The GLP-2 receptor modulates the inflammatory response to diet-induced liver injury

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Background: Non-alcoholic fatty liver disease (NAFLD) is implicated as an early and key driver of the metabolic syndrome through lipotoxicity. The progression from simple steatosis to nonalcoholic steatohepatitis is correlated with progressive development of the metabolic syndrome. My research looks at whether meal-derived hormonal signaling impacts, directly or indirectly, the development of NAFLD. The proglucagon-derived peptide GLP-2, secreted from the gut basally and at enhanced levels during meals, has a trophic effect on the intestinal lining. GLP-2 signaling leads to increased absorption of dietary fat, carbohydrates and protein. The proabsorptive actions of GLP-2 have led to its use in the treatment of parenteral nutrition-dependent subjects with short bowel syndrome. We hypothesize that enhanced or attenuated GLP-2R signaling modifies the development of experimental hepatic steatosis.

Methods: Young (7-8wo) Glp2r-/- mice (n=20) and control Glp2r+/+ (n=20) were randomized to high fat, high fructose, cholesterol enriched (HFHF) diet or a control diet for 17 weeks. To study the impact of GLP-2 gain of function in fully developed NASH, Young (7wo) WT mice placed on a HFHF or control diet. During weeks 16-17 mice were randomized to treatment with a GLP-2 analog or saline. Extensive metabolic, histological, molecular and inflammatory phenotyping was obtained.

Results: Mice treated with GLP-2 did not exhibit enhanced hepatic fat accumulation, dyslipidemia, or changes in hepatic or circulating markers of inflammation. In contrast, despite comparable weight gain, Glp2r-/- mice exhibited enhanced steatosis and impaired glucose excursion on glucose tolerance test. Levels of hepatic pro inflammatory cytokines and transaminase levels were also increased. The GLP-2R was localized, using FACS and cell fractionation, to the non-hepatocyte fraction of isolated liver cells.

Discussion and impact: Despite reports that GLP-2 enhances lipid absorption, GLP-2 did not exacerbate the development of experimental high fat diet-induced liver injury. In contrast, our findings reveal an essential role for the endogenous GLP-2 receptor in modulating the adaptive hepatic response to nutrient-induced cellular stress and the control of liver inflammation.

Oral Abstract 4
Monogenic Diabetes Misdiagnosed as Type 1 Diabetes

Claire Lange(1), Coralie Leblicq(1), Laura Rendon(1), Tedi Qedro(1), Tugba Demirci(1), Luc Marchand(1), Constantin Polychronakos(1,2)*
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Background: According to the Search for Diabetes in Youth study and the Norwegian T1D registry, approximately 1% of individuals diagnosed as T1D have monogenic diabetes (MD). This percentage rises to 4% in the autoantibody-negative (aAb-) cases. Incorrect diagnosis has significant consequences since many MD cases can be treated more effectively with sulfonylureas and, potentially, incretin-based therapies. In addition to the 14 MODY genes, several neonatal and syndromic diabetes genes must be tested for precise diagnosis, which makes genetic testing expensive and impractical. Better algorithms for selecting cases to sequence are needed.
Hypothesis & Objective: We hypothesize that selecting cases with low genetic risk for autoimmune T1D will make case selection much more powerful. We combined autoantibody negativity with low genetic risk to select sibling pairs from the T1D Genetics consortium for whole-exome sequencing (WES).

Methods: The 2,345 sibling pairs included 167 families were both siblings were aAb-. Of these, 52 families were also negative for high-risk HLA. Twelve had a diabetic parent and, of these, 8 were available for targeted testing for MODY3 (the most common MD). One proband from each of 38 of the remaining families was examined by WES, looking for mutations in known MODY, neonatal and syndromic diabetes genes.
Results: Three of the eight (37.5%) sibling pairs with a diabetic parent had mutations in HNF1α, the MODY3 gene. HNF1α was also mutated in 4 of the 38 sibling pairs (10.5%) with no family history of diabetes. WFS1 (the DIDMOAD gene) had compound heterozygous mutations in 5/38 (13.1%), in the absence of syndromic features. We also found one of each with mutations in KCNJ11, ABCC8 and INS.

Conclusions: 1) Almost one-third of patients diagnosed as T1D by strict clinical criteria but aAb- and low genetic risk have monogenic diabetes. 2) Many of these are late-onset neonatal or have DIDMOAD (Wolfram syndrome) with diabetes as the only manifestation.

Oral Abstract 5
Interaction between the development of the thyroid and of pharyngeal vessels: from exome sequencing in humans to validation of candidate genes in zebrafish

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Congenital hypothyroidism due to thyroid dysgenesis (CHTD) is mainly a sporadic and non-syndromic condition occurring in 1:4,500 live births. Non-syndromic (NS) CHTD shows ethnic predominance but low familial recurrence risk (~2%) and low concordance rate between monozygotic twins, suggesting a two-hit scenario combining post-zygotic events with either a de novo monogenic mutation or incomplete penetrance of polygenic inherited variants. As this latter possibility was recently proven right in cases of non-syndromic congenital heart defects, we analysed the burden of rare non-synonymous variants in the exome of 37 cases of NS-CHTD compared to that of 495 controls sequenced and analysed on the same platform. Gene-based burden analysis identified several genes enriched in NS-CHTD, including IKBKE. IKBKE is a member of the inhibitor of κB kinase (IKK) family. It is implicated in the non-canonical pathway of NF-κB and interferon regulatory factor signalling. Furthermore, it has been associated with inflammation, cell transformation, and progression of many cancers. Functional assays showed that IKBKE depletion decreases migration of Nthy-Ori cells (a human thyroid cell line) in vitro. Moreover, ikbke depletion in zebrafish caused defective aortic arch artery formation and abnormal thyroid morphogenesis. The thyroid phenotype was partially rescued by injection of human IKBKE RNA in ikbke morphants. Our results further expand the growing list of predisposing genes for CHTD and confirm the association between vasculogenesis and congenital thyroid malformations.

Oral Abstract 6
Gender Diversity Training in Canadian Pediatric Postgraduate Medical Education: A Needs Assessment Survey

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Objective: To examine resident and program director perspectives on current state of postgraduate medical education on gender diversity in Canadian pediatric residency programs.

Background: Primary care providers are seeing more gender diverse children and youth in their offices with an exponential growth in referrals to Canadian specialty clinics for these children and youth, and potential for significant mental health comorbidities. Gender-affirming support and management have been shown to improve overall outcomes. There is currently no mandatory curriculum on gender diversity for Canadian pediatric residency programs.

Methods: Cross-sectional online surveys in English and French distributed to program directors and pediatric residents in the 17 Canadian pediatric residency programs. Data were analyzed by descriptive statistics with 95% confidence intervals.

Results: Response rate was 88.2% from program directors (PDs) and 24.5% from pediatric residents. Amongst PDs, 14.3% [95%CI: 6.3, 22.3] reported a formal curriculum for gender diversity teaching whereas 66.7% [58.2, 75.1] had diversity care for children and youth in their hospital/community, with 80.0% from a multidisciplinary team. Sixty-four percent [53.3, 75.2] of PDs estimated their residents received ≤ two hours teaching on gender diversity, with 53.3% [44.4, 62.3] reporting specific teaching on diagnosis of gender dysphoria in adolescents and 40.0% [31.2, 48.8] on diagnosis in children, social transitioning, and mental health. Residents reported comfort levels ≤ 50% on specific gender diversity topics. Amongst residents, 73.8% [67.9, 79.6] reported that mandatory time in a gender diversity clinic would be the most effective teaching tool with 61.9% [55.4, 68.3] favouring academic half day. PDs favoured an online module as the most effective teaching tool (66.7% [58.2, 75.1]), followed by mandatory time in a gender diversity clinic (60.0% [51.2, 68.8]). Barriers to more teaching included lack of time and space in a busy curriculum. Over 90% of resident respondents indicated that more teaching on gender diversity is required.

Conclusions: Program directors and residents in Canadian pediatric residency programs report significant variability in education about gender diversity in children and youth. Residents reported low comfort levels with this topic and need for further education. A formal pediatric postgraduate curriculum on gender diversity is recommended for Canada

Oral Abstract 7
Effects of Pubertal Suppression with GnRH Analogs on Body Composition and Body Mass Index of Transgender Adolescents

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Background: Adolescents with gender dysphoria are offered GnRH analogs (GnRHa) to delay the onset and progression of puberty. Pubertal suppression with GnRHa is a reversible intervention that can result in more satisfactory physical and psychological outcomes. GnRHa are also used in central precocious puberty; however, there are controversial effects on their BMI, body composition and fat mass.

Methods: A retrospective chart review was conducted for youth with gender dysphoria followed in CHEO's Diversity Clinic who had whole-body dual-energy x-ray absorptiometry (DEXA) between January 1, 2006 and April 30, 2017. Those with a baseline DEXA within 90 days of GnRHa and post GnRHa DEXA were included in the analysis. Data on natal and affirmed gender, Tanner stage, height, weight, BMI, initiation of GnRHa and cross-sex hormones (CSH), fat percentage, blood pressure and lipid profiles were retrieved.

Results: The study included 46 trans youth: 14 transfemales with mean age of 15.02 ± 1.45 (SD) years, and 32 transmales 15.83 ± 1.48 years, with 84.3% of transmales and 50% of transfemales at Tanner stage 4-5. Baseline BMI was 24.00 ± 5.26 in transmales and 21.86 ± 3.45 in transfemales (ns). Baseline body fat (%) was 37.77 ± 10.62 in transmales, and 27.25 ± 12.89 in transfemales ($p < 0.001$). Pre-post GnRHa assessments (311.1 ± 80.8 days interval) showed statistically significant increase in BMI among transmales 2.17 (95%CI 1.00, 3.33, $p<0.001$), but not in transfemales 0.41 (-0.88, 1.71), and increase in fat (%) in both transmales 4.04 (2.23, 5.84, $p<0.001$) and transfemales 3.87 (0.04, 7.70, $p < 0.048$). Ongoing analyses will examine their blood pressure and lipid profiles.

Conclusion: This study examined body composition in a large cohort of adolescent trans youth who started pubertal suppression in mid teen years with pre and post GnRHa data. Pubertal suppression with GnRHa was associated with increase in BMI in transmales but not transfemales and increase in fat percentage in both transfemales and transmales. Clinicians involved in the care of trans youth should counsel them around these potential changes and proactively encourage principles of healthy active living.

Oral Abstract 8

Residence in Haiti vs. Quebec impacts phenotypes of diabetes and glycemic control in Haitian youth

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Background: In Haitian youth residing in Haiti, atypical diabetes, longer partial remission, and increased rates of premature complications are more common. The underlying causes are unclear.

Objectives: To characterize clinical presentation and glycemic control in Haitian youth with diabetes residing in Quebec vs. Haiti.

Methods: Retrospective review of Haitian youth with diabetes diagnosed at 0-25 years at a diabetes clinic in Haiti between 01/2013-03/2018, and at the Montreal Children's Hospital between 09/2001-09/2018. We documented age and diabetic ketoacidosis (DKA) in Quebec or coma (presumed DKA) in Haiti, diabetes type, and yearly anthropometric data, total daily insulin dose (TDD), and hemoglobin A1c (A1c). We used univariate and multivariate regression analyses to determine predictors of most recent A1c.

Results: 18 and 90 patients were included in Quebec and Haiti, respectively, of whom 3 (17%) and 1 (1%) had type 2 diabetes and were excluded from further analysis. Moderate-severe DKA and coma, respectively, were present in 3 (20%) in Quebec and 17 (19%) in Haiti. In Quebec vs. Haiti, sex was female in 67 vs. 60% ($p=0.62$), mean age at diagnosis 6.5±4.3 vs. 14.1 ± 4.6 years ($p<.0001$), mean diabetes duration 7.0±4.3 vs. 4.0±3.5 years ($p<.0001$), mean TDD 1.0 ± 0.4 vs. 0.5±0.3 units/kg/day ($p<.0001$), mean BMI z-score 1.2±1.0 vs. -0.8±0.9 ($p<.0001$), and most recent A1c was 9.5±2.3 vs. 11.4±2.6 ($p<.0001$). In univariate analyses, residence in Haiti vs. Quebec ($p=0.009$) and lower BMI z-score ($p=0.004$) predicted higher A1c. Shorter diabetes duration was marginally predictive ($p=0.07$), and age, TDD and sex were not. In an adjusted model, residence in Haiti vs. Quebec ($p=0.03$) remained predictive of higher A1c, as were higher TDD ($p=0.0006$), lower BMI z-score ($p=0.02$), and female sex ($p=0.04$), but not age ($p=0.15$) or diabetes duration ($p=0.71$).

Conclusions: Haitian youth in Quebec vs. Haiti are younger at diagnosis and have higher insulin requirements, similar to Caucasian youth. Glycemic control is poor in both groups but worse in Haiti, even when adjusting for markers of poor insulin adherence (high TDD and low BMI). Environmental factors may underlie a later presentation and longer endogenous insulin production in Haitian youth residing in Haiti (hygiene hypothesis).

Oral Abstract 9
Osteogenesis Imperfecta:
Skeletal Outcomes After Bisphosphonate Discontinuation at Final Height

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Background: Intravenous cyclical bisphosphonates are widely used to treat children with moderate to severe osteogenesis imperfecta (OI). They increase bone mineral density, diminish fracture rates and improve mobility. Bisphosphonates are often discontinued when growth is completed, as the treatment effect is smaller in the mature skeleton and there are concerns about adverse events when bisphosphonates accumulate in the skeleton. It remains unclear for what period of time areal bone mineral density (aBMD) gains persist after bisphosphonate discontinuation.

Methods: We assessed patients with OI who had started intravenous bisphosphonates (either pamidronate or zoledronate) before 13 years of age, were treated for at least two years and discontinued treatment after completion of growth. Lumbar spine densitometry by dual-energy x-ray absorptiometry and spine radiographs were performed at treatment discontinuation as well as at 24 and 48 months thereafter.

Results: Thirty-three patients (23 females) were followed for at least four years after bisphosphonate discontinuation. Patients had started treatment between ages 0.1-12.6 years and had stopped 4.7-15.7 years later, when their age ranged between 13.1-20.0 years (mean: 16.2 years (SD: 1.90)). Lumbar spine aBMD increased by 3.8% two years after treatment discontinuation ($P < 0.05$) and remained stable afterwards (0.2% increase from two to four years, $P = 0.86$). Lumbar spine aBMD z-score remained stable over time, with mean (SD) values of -1.8 (1.2) at the last treatment, -1.9 (1.1) after two years, and -2.0 (1.2) after 4 years post bisphosphonate discontinuation ($P = 0.53$). In three patients, the lumbar spine aBMD z-score declined by more than 1 SD in the four years following treatment discontinuation (range: -1.16 to -1.28). The proportion of patients with new long-bone fractures in the two years before treatment discontinuation decreased compared to the last 2 years of follow-up (42% and 15%, respectively; $P < 0.05$). No patient sustained an incident vertebral compression fracture at four years of follow-up.

Conclusion: Lumbar spine aBMD z-scores remained stable four years after bisphosphonate discontinuation in adolescents and young adults with OI. Patients did not have an increased number of vertebral or long-bone fractures. However, a minority of patients had significant decline four years after treatment discontinuation.

Oral Abstract 10
Does Age Matter? Mental Health Implications and Determinants of When Youth Present to a Gender Clinic

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Background/Aims: Gender incongruent youth experience high rates of mental health comorbidities. We hypothesized that youth presenting to our clinic later in age (and who may have gone longer without care) would have more mental health comorbidities than their younger peers and that identifiable barriers may have delayed their clinic attendance.

Methods: Charts of new patients to the SickKids Transgender Youth Clinic (TYC) with a diagnosis of gender dysphoria were reviewed for demographic information, self-reported markers of psychological distress, and validated mental health questionnaire scores. Subjects were classified as younger presenting youth (YPY): <15 years of age at presentation or older presenting youth (OPY): > 15 years of age; data from OPY and YPY were compared. Factors influencing age at presentation to care were explored through 24 semi-structured interviews with OPY, YPY, and their caregivers. Interview transcripts underwent thematic content analysis by three independent coders.

Results: Of 300 patients, 184 (61%) were OPY (mean age: 16.23 + 0.70 years, 77% assigned female at birth [AFAB], mean Tanner stage [TS]: 4.47 + 0.52) while 116 (39%) were YPY (mean age: 13.59 + 1.05 years, 74% AFAB, mean TS: 3.88 + 0.89). Upon TYC presentation, significantly more OPY than YPY had a diagnosis of depression (46% vs 30% p=0.006), were using psychoactive medications (36% vs 23%, p =0.017), reported past suicidal ideation (52% vs 40%, p=0.034), had self-harmed (40% vs 28%, p=0.022) and had attempted suicide (17% vs 9%, p=0.033). 11 themes were identified from interviews that influenced age at first clinic visit. OPY tended to see multiple physicians or only allied health providers prior to TYC referral while more YPY were referred after seeing a single primary care provider. OPY/caregivers described more religious, family, and peer group tensions related to gender nonconformity while YPY/caregivers described a sense of urgency for medical treatment, supportive school environments, and involvement with LGBTQ+ support groups.

Conclusions: Upon presentation to a gender clinic, older youth have higher rates of mental health distress than younger youth. Further study of factors that influence age at first visit is important and may identify modifiable factors that impact care and outcomes.

Oral Abstract 11
Effect of Sensor-Augmented Pump Therapy on Fear of Hypoglycemia in Children with Type 1 Diabetes and their Parents in the CGM TIME Trial

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OBJECTIVE: To determine if changing from insulin injections to sensor-augmented pump therapy (SAPT) decreases fear of hypoglycemia among children with type 1 diabetes and their parents.

METHODS: The CGM TIME Trial is a multicentre randomized controlled trial that enrolled 144 children with type 1 diabetes on insulin injections who were changing to pump therapy (MiniMed™ Veo™, Medtronic Canada), with CGM (MiniMed™ Enlite™ sensor) introduced simultaneously or 6 months later. The previously validated Hypoglycemia Fear Scale (HFS) questionnaire was completed by children ≥ 10 years old, and all parents, at study entry and 12 months later. Differences in HFS scores between the two time points were compared with paired t-tests and individual-item responses with paired Wilcoxon tests. Participants in the Simultaneous and Delayed Groups were combined for all analyses.

RESULTS: 121 parents of 140 children and 91/99 children ≥10 had complete data and were included in the analysis. Mean score for the Behaviour subscale for children decreased from 2.11 (SD 0.59) to 1.72 (SD 0.61) ($p<0.001$) and for parents from 2.07 (SD 0.75) to 1.74 (0.74) ($p<0.001$). For the Worry subscale, mean scores decreased from 1.19 (SD 0.79) to 0.79 (SD 0.76) ($p<0.001$) for children and from 1.54 (SD 0.88) to 1.17 (SD 0.69) ($p<0.001$) for parents. Analysis of additional HFS subscales revealed significant decreases in scores on the Maintain High Blood Glucose subscale ($p<0.001$ for children and parents); the Avoidance subscale ($p<0.001$ for children and parents); the Helplessness subscale ($p<0.001$ for children and parents); and the Social subscale ($p<0.001$ for children and $p= 0.02$ for parents). Median scores for individual questions were lower at 12 months compared to study entry for 25/25 child questions and 23/25 parent questions. 10/25 child items and 12/25 parent items were significantly lower at 12 months with p-values <0.001.

CONCLUSIONS: Sensor-augmented pump therapy significantly reduced fear of hypoglycemia in children with type 1 diabetes and their parents, and in particular, certain aspects of fear related to living with type 1 diabetes. Future analyses will use linear regression to determine if the degree of adherence to CGM is associated with greater change in HFS scores.

Oral Abstract 12

Outcomes and experiences of young adults with type 1 diabetes transitioned from pediatric to adult care: a cross sectional assessment

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Introduction: Transition from pediatric to adult care is a high risk time for youth with type 1 diabetes. Canadian studies have demonstrated that 14-24% of young adults had gaps of more than one year between their last pediatric visit and their first adult appointment. Irregular follow-up has been associated with worse glycemic control and diabetes complications. To identify opportunities to improve transition care, we aimed to assess outcomes and experiences of youth graduating from our clinic and to identify factors associated with timely follow up (adult care within 6 months) and satisfaction with the transition process.

Methods: We mailed surveys to all young adults with type 1 diabetes and their parents who transferred to adult care from 2011-2017. Surveys asked patients about elements of current and past care, perceived gaps in the transition process and factors that may predict successful transition, such as hemoglobin A1C (HbA1C), duration of diabetes, and referral to a young adult diabetes program. Additional clinical and demographic data were obtained by chart review.

Results: We received 64 responses, of 373 surveys mailed. The majority (88.3%) of respondents had their first adult diabetes physician visit within 6 months. Only 1.7% had not established care by 12 months. Time to first adult visit was not associated with hemoglobin A1C or other risk factors. However, patients and parents who reported they did not discuss transition with their pediatric team in the year prior to transfer had a higher mean HbA1C than those who reported they had such discussions (8.2±1.2%, and 10.1±2.0% respectively, p=0.005). Higher HbA1C was also associated with patients and parents not feeling prepared to leave pediatric care (p=0.002 for parents, p=0.01 for patients), with switching to a different adult provider after their initial visit with the provider to whom they were referred (p=0.009), and overall parent dissatisfaction with the transition process (p=0.002).

Conclusion: While most of our patients transitioned to adult diabetes care within 6 months, not all experiences were ideal. There remains room for improving the experiences of patients with higher HbA1C and their parents, who reported being less prepared and satisfied with the transition process.

Poster Abstracts

Poster Abstract 1
When Low Blood Sugars Cause High Anxiety: Assessing Fear of Hypoglycemia in Parents of Children with Type 1 Diabetes Mellitus

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Background: Hypoglycemia is the most common acute complication of type 1 diabetes mellitus (T1DM). The discomfort associated with hypoglycemia and the difficulty in predicting episodes combined with the chance for long-term harm can cause children and their parents to develop significant fear of hypoglycemia (FOH). This fear and associated anxiety can be disruptive to activities of daily living and lead to reduced quality of life. The objective of this study was to determine the prevalence of FOH in parents of children with T1DM at our center and to identify factors associated with higher FOH.

Methods: A total of 264 parents of children/youth (2 to 18 years of age, mean 12.4 ± 3.5) with T1DM completed an in-clinic survey that included demographic and disease-specific questions as well as the Speilberger State-Trait Anxiety Inventory and the Hypoglycemia Fear Survey – Parent version (HFS-P) (worry and behaviour subscales).

Results: Mean parental HFS-P score was 67 ± 19 (range 31-119, possible range 25-125). The most frequent worries were related to the child being hypoglycemic while alone or asleep. A higher HFS-P score was associated with more frequent and severe hypoglycemic episodes, younger age of the child and higher state-trait anxiety scores as well as current use of a continuous glucose monitor and more frequent blood glucose checks. Higher HFS-P was also associated with parents reporting more reluctance to make insulin dose adjustments recommended by their diabetes team, worse parental sleep quality and more daytime fatigue. HFS-P scores did not differ between those using insulin injections and those using insulin pumps and was also not associated with HbA1C values.

Conclusions: Parents of children with T1DM experience fear of hypoglycemia, especially during times when children are vulnerable (asleep or alone). Moreover, FOH can have an impact on clinical care (with parents being reluctant to administer suggested insulin doses) and quality of life (due to parental/child sleep disruption). Our results highlight the need for the development, implementation and evaluation of interventions aimed at reducing FOH in parents of children/youth with T1DM.

Poster Abstract 2
A Closer Look at the Water Deprivation Test

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Introduction: The water deprivation test is used to differentiate between different causes of polyuria (primary polydipsia, central diabetes insipidus (DI) and nephrogenic DI). As the name implies, the patient is deprived of water with regular monitoring of urine and serum osmolality at baseline and specific time intervals.

Objectives: 1. Identify baseline characteristics that may predict the diagnosis of primary polydipsia. 2. Identify number of hours and the frequency of sampling required to establish the diagnosis. 3. Identify sensitivity and specificity of urine osmolality/serum osmolality ratio as a diagnostic method compared to absolute urine osmolality.

Study design: Retrospective chart review looking at water deprivation tests conducted at McMaster's Children Hospital from 1/1/2007 to 2/3/2018. The following variables were collected: age, sex, primary diagnosis, pretest diagnosis, definitive diagnosis, number of hours required, serum osmolality, urine osmolality, urine osmolality/serum osmolality ratio at baseline and every hour.

Results: Total of 21 patients were included. Average age of presentation was 6.63 years (SD=4.5). Male to female ratio was 4:3. The majority of patients had no underlying medical condition at the time of the test (n=10). Diagnosis of diabetes insipidus was suspected in 12 out of 21 patients based on history, physical examination and basic laboratory investigations. Only 7 out of 21 patients proved to have central DI based on the water deprivation test. Of note, 4 out of 21 patients (19%) found to have underlying CNS pathology had polyuria as the only presenting symptom. 14 out of 21 patients were diagnosed with primary polydipsia. A logistic regression analysis was performed to ascertain the effects of age, gender, time of diagnosis, primary diagnosis and baseline serum sodium140 as predictors for the diagnosis of primary polydipsia. No variable was associated with primary polydipsia diagnosis (all $P<0.05$). 71% of patients had a diagnosis established by 4 hours. The calculations of the specificity, sensitivity, positive and negative predictive values for serum osmolality to urine osmolality ratio are in progress and will be available shortly.

Conclusions: Polyuria may be the only presenting symptom of underlying CNS disease. Baseline serum sodium levels does not predict the presence or absence of DI.

Poster Abstract 3
Assessment of the quality and consistency of in-hospital monitoring of blood glucose during pediatric cystic fibrosis exacerbations

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Prompt diagnosis and intervention for cystic fibrosis-related diabetes (CFRD) preserves nutritional status and respiratory function. Increased insulin resistance during intercurrent illness may facilitate early detection of dysglycemia. The International Society for Pediatric and Adolescent Diabetes specifies that assessment of fasting and 2-hour post-prandial blood glucoses during the first 48 hours of admission for pulmonary exacerbations is an accepted screening tool for CFRD. We conducted a retrospective study over 12 months to identify how consistently screening is ordered, implemented, and interpreted for pediatric patients with CF hospitalized for pulmonary exacerbations. Eighteen patients aged 10 to 18 years (mean 13.4 ± 2.0 years) fit inclusion and exclusion criteria. Two patients had a known diagnosis of impaired glucose tolerance (IGT). Three patients received oral or IV steroids during their admission. Glycemic monitoring was ordered in 14/16 (88%) of patients with normal glucose tolerance but not for the 2 patients with IGT. Orders were inconsistent between patients. No patients had a full 48 hours of consistent pre- and 2-h post meal blood glucose checks. Post-prandial checks were performed between 0.5 and 4 hours post-meal (average 2.4 ± 0.6 hours). Post-prandial glucose levels were between 7.8 and 11.0 mmol/L in 2/14 of patients and greater than 11 mmol/L in 1 patient. One additional patient had no blood glucose checks for the first 48 hours but had a post-prandial glucose of 11.2 mmol/L on the fourth day of admission. Serum glucose was not ordered in patients with post-prandial glucose values in the diabetic range. Glycemic monitoring was not continued after 48 hours in patients with post-prandial glucose values in the IGT or diabetic range. OGTT and HbA1c were not performed for patients who had screened positively for diabetes. These data suggest missed opportunities to confirm positive screens for dysglycemia and for follow up testing of dysglycemia in hospital.

Conclusions: Glycemic monitoring during CF exacerbations may improve early detection of CFRD Optimization of in-hospital order sets, consistency of 2-hour post prandial blood glucose checks, and interpretation and communications of glycemic data will improve early identification of patients with CFRD.

Poster Abstract 4
An unusual presentation of Type 1 Diabetes

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Introduction: Hyperglycemic hyperosmolar state (HHS) and diabetic ketoacidosis (DKA) are life-threatening emergencies in diabetic patients. While DKA at presentation of T1D in children represents about 25% of cases, HHS is very unusual as a first presentation.

Case: A 10 year old Haitian-Sudanese boy presented to our ER for obtundation. He had a 10 days history of polydipsia and polyuria followed later by vomiting, for which he drank mainly juice and soft drinks. On the day of presentation, he had difficulty rousing. In ER, he was tachycardic and hypotensive with a GCS of 8. Initial labs showed pH 7.1, bicarb 9.2, glucose 130mmol/l, Na 125mmol/l, K 4.0mmol/l, creatinine 352umol/l, urea 25.2mmol/l, and mild ketonuria. He was started on our DKA protocol after receiving three fluid blouses for hypovolemic shock. Urgent CT and MRI head were normal. The patient was admitted to ICU for management of profound dehydration, requiring intubation and inotropes. HbA1c was found to be 11%. Repeat MRI brain on day 3 due to agitation revealed a superior sagittal and straight sinus thrombosis. Other complications during his hospitalization included unilateral vocal cord paralysis without a clear etiology and non-pressure ulcers over the ischia region and in the left gluteal fold. The patient was admitted for 34 days, the latter weeks dedicated mainly to wound care. He was discharged with only a residual vocal cord paralysis but a grossly normal neurological exam. His BG were well controlled on 1.2unit/kg/day of insulin.

Discussion: The classic presentation of HHS in children is usually in obese adolescents with T2D. The symptoms tend to occur more gradually, and the mortality rate from HHS is higher than that from DKA. Their fluid deficit is estimated to be double that associated with DKA alone, and as such tends to be grossly underestimated, especially by treating physicians who rarely encounter this condition. Our patient presented with both HHS and DKA and fortunately survived with few sequelae. At follow-up he had progressed to have remission (honeymoon period) lasting five months and continues to have well-controlled diabetes with the most recent HbA1c of 7.1% on 0.75unit/kg/day of insulin.

Poster Abstract 5
A Challenging Case of Radioinsensitive Papillary Thyroid Cancer

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Introduction: Papillary thyroid cancer (PTC) accounts for most of pediatric thyroid cancer cases. PTC may present as multifocal, bilateral, and metastatic disease. We present a case of a 16-year-old girl with PTC and radioinsensitive disease recurrence.

Case Presentation: Our patient was previously healthy with no prior radiation exposure or family history of thyroid cancer. She presented with a painless right anterior cervical lymph node. Ultrasound guided FNA confirmed PTC. Baseline contrast CT of the neck, chest, and abdomen demonstrated multiple abnormal right cervical and superior mediastinal level II-VII lymph nodes, with the largest measuring 5.2x2.5x2.6 cm. A complete thyroidectomy and modified radical right and central neck dissection was performed. L-thyroxine was titrated to TSH suppression of < 0.1 mU/L to reduce the risk of recurrence. Pathology confirmed PTC, diffuse sclerosing variant. Although angioinvasion was not present, there was lymphatic invasion, perineural invasion, extranodal extension, and positive margins. Lymph nodes dissected from level I-V showed 14/33 were positive for metastatic disease. Diagnostic 123I whole-body scan (WBS) at 24 hours was 0.17% with no significant focal activity in the neck. Nonetheless, given her risk of microscopic residual disease, she received radioactive iodine (RAI) ablation 125 mCi post thyroidectomy. Post RAI ablation, WBS revealed a very small area of increased activity in the left neck. Approximately four months later, she presented with a palpable right neck mass, confirmed to be PTC recurrence. There were four abnormal nodules in the right neck seen on ultrasound, but 123I-WBS measured 0.1% at 24 hours and there was no focal activity seen. She had level II right neck dissection. Surveillance imaging two months later demonstrated possible disease recurrence vs. residual disease along the right thyroid bed and paratracheal region. Thyroglobulin levels (measured by LC-MS) have remained undetectable. She was referred to Oncology for possible targeted therapy. A RET fusion was found in the tumor on a research fusion panel.

Conclusions: Activation of the MAPK pathway is critical for PTC development and new, targeted therapy may play a pivotal role in the treatment of radioinsensitive disease.

Poster Abstract 6
Risk of psychiatric disorders and suicide attempts in children and emerging adults with diabetes

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Background: The prevalence of type 1 diabetes (T1D) is increasing, thereby leading to an increasing number of adolescents with T1D transferring from paediatric to adult care. During this crucial transition period, the adolescent with T1D faces emotional, occupational, social, and financial changes which may increase his/her risk of mental health disorders. However, the risk of mental health disorders during late adolescence and emerging adulthood in individuals with diabetes has not been well studied.

Objectives: We determined the risk of psychiatric disorders, hospitalization for suicide attempts and death by suicide in adolescents and emerging adults with diabetes compared to their same-aged peers without diabetes.

Research Design and Method: We conducted a retrospective cohort study of children living in Québec, Canada, using linked health administrative databases. Children diagnosed with diabetes between ages 1 to 15 years from April 1, 1997, and December 31, 2013, and all children without diabetes in the same age group were included. Follow-up started at age 15 years and ended at age 25 years. Individuals with a code for psychiatric disorders in the two years preceding the date of entry in the cohort were excluded. Separate multivariate Cox proportional hazard models were used for each mental health outcomes.

Results: The cohort included 3,544 individuals with diabetes and 1,388,397 without diabetes. Adolescents and emerging adults with diabetes were 33% more likely to experience a severe mood disorder (adjusted hazard ratio (HR) 1.33, 95% confidence interval (CI) 1.19-1.50), 325% more likely to be hospitalized for a suicide attempt (HR 3.25, 95% CI 1.79-5.88), 82% more likely to visit a psychiatrist at least once (HR 1.82, 95% CI 1.67-1.98) and 29% more likely to have any psychiatric disorder (HR 1.29, 95% CI 1.21-1.37), compared to their peers without diabetes.

Conclusions: The risk over time of severe mood disorders, hospitalizations for suicide attempts, any visit to a psychiatrist, and any psychiatric disorders was higher in adolescents and young adults 15-25 years old with diabetes compared to their peers without diabetes.

Poster Abstract 7

Low Frequency Synonymous Coding Variation in CYP2R1 has Large Effects on Vitamin D Level and Risk of Multiple Sclerosis.

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Introduction: Vitamin D insufficiency is common, correctable and influenced by genetic factors, and it has been associated to risk of several diseases. We sought to identify low-frequency genetic variants that strongly increased the risk of vitamin D insufficiency and tested their effect on risk of multiple sclerosis, a disease influenced by low vitamin D concentrations.

Methods/Results: We used whole-genome sequencing data from 2,619 individuals through the UK10K program and deep imputation data from 39,655 genome-wide genotyped individuals. Meta-analysis of the summary statistics from 19 cohorts identified a low-frequency synonymous coding variant (rs117913124[A], minor allele frequency=2.5%) in the CYP2R1 gene which conferred a large effect on 25-hydroxyvitamin D (25OHD) levels (-0.43 standard deviations of natural log-transformed 25OHD, per A allele, P-value = 1.5 x10-88). The effect on 25OHD was four-times larger and independent of the effect of a previously described common variant near CYP2R1. By analyzing 8,711 individuals we showed that heterozygote carriers of this low-frequency variant have an increased risk of vitamin D insufficiency (OR=2.2, 95% CI 1.78-2.78, P=1.26 x 10-12). Individuals carrying one copy of this variant had also an increased odds of multiple sclerosis (OR=1.4, 95%CI 1.19-1.64, P=2.63 x 10-5) in a sample of 5,927 cases and 5,599 controls.

Conclusions: We describe a novel low-frequency coding variant in the CYP2R1 gene, which exerts the largest effect upon 25OHD levels identified to date in the general European population. Since CYP2R1 is known to encode a critical enzyme in the production of the active form of vitamin D, these findings implicate vitamin D in the etiology of multiple sclerosis.

Poster Abstract 8
Empowering families to upload insulin pump and glucose sensor data to improve paediatric type 1 diabetes self-management and clinic flow

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Introduction: Insulin pumps and glucose sensors provide data that assists with glycemic pattern recognition and insulin dose adjustment to help improve metabolic control. Fundamental to utilizing this technology is families' ability to upload, review and share device data. Literature describing the frequency of this practice is scarce. Many families are not routinely uploading and reviewing device data between visits, resulting in lost opportunities to improve glycemic control and extra time spent uploading devices in clinic. Within the framework of a quality improvement initiative, we aimed to quantify rates of uploading devices, understand barriers and measure the impact of uploading devices on clinic flow.

Methods: To collect baseline data, patients <18 years with type 1 diabetes using a pump or sensor were given a survey to determine their barriers to, routines, and understanding of uploading data. Weekly process measures, including clinic time spent uploading, clinic flow analysis, and the proportion of patients uploading between clinics were also collected.

Results: The 52 patients who completed the survey had a median age of 13.5 (4-17) years, median diabetes duration of 5.0 (0.4-14) years, and 98% used an insulin pump, of which 53% used a glucose sensor. While most (73% of pump, 82% of sensor users) knew how to upload their devices at home, only 44% of pump and 27% of sensor users did this prior to their appointment on the day surveyed, and 23% of pump and 31% of sensor users had never uploaded. The most common barriers were lack of time and that they hadn't thought to try. In addition, 40% reported technical difficulties with uploading. Median upload time per device was 8.0 (3-30) minutes and median clinic uploading time was 42.5 (11.0-87.0) minutes.

Conclusion: Baseline survey data demonstrates an opportunity for improvement. In the next phase of the initiative, educational resources for uploading and use of reports are being provided. Follow-up surveys at 3 and 6 months, as well as ongoing collection of process measures, will inform us of the effectiveness of our interventions.

Poster Abstract 9
Managing Type 1 Diabetes Mellitus in Infancy: A Comparison of Two Cases

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Background: Type 1 diabetes mellitus (T1DM) is uncommon in the first year of life. Management of these patients shares similarities with the neonatal diabetes population including almost continuous carbohydrate intake, variable physical activity, and the need for insulin micro dosing. There are no guidelines to direct the management of these patients, although case reports have demonstrated the feasibility of continuous subcutaneous insulin infusion. However, pumps are labour intensive and may not be possible for all families. For patients diagnosed with T1DM in infancy, it is important there are options which are safe, feasible and achieve similar glycemic control. We review the literature on this topic and describe our centre's experience managing two such infants.

The Cases: ES and AT presented in DKA at eight months of age, in August and September of 2018 respectively. HbA1c at diagnosis was 9.1% and 6.8%. Autoantibodies were positive. Both infants were breastfed frequently with inconsistent intake of solids. Both infants were discharged from hospital after 2-3 days on insulin detemir BID (1.5 – 2.5units), with persistent hyperglycemia and/or ketosis managed with rapid insulin. Both infants were started on continuous glucose monitors within one month of diagnosis. Soon after, AT was started on Medtronic pump. Challenges included estimating boluses for breastfeeding, and accidental double priming of pump leading to moderate hypoglycemia and inpatient admission. ES was transitioned to daily insulin glargine within two months of diagnosis, and subsequently to multiple daily injections with insulin glargine and diluted insulin aspart. Challenges included extreme glycemic variability, and unpredictability. At 2 months post diagnosis, percent time in target and frequency of hypoglycemia were superior with pump therapy. Coefficient of variability was equivalent. Objective measures will be compared at 3 and 6 months. Key

Learnings: • Pump therapy is feasible and safe in the child with T1DM under one year. It may provide superior outcomes (% time in target, frequency of hypoglycemia) compared to MDI. Trade offs include burden of care, risk of DKA. • It is valuable to consider elective hospital admission at pump start. • Multiple insulin regimens may be reasonable in this population. Management should depend on family preferences.

Poster Abstract 10
Clinical and Endocrinological Manifestations of Partial Ectopic Posterior Pituitary: A New Imaging Entity

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Background: Abnormal posterior pituitary development can be associated with endocrine manifestations. Partial ectopic posterior pituitary (PEPP) has not been reported and associated endocrinological consequences are unknown.

Objective: To describe clinical and endocrinological manifestations associated with PEPP seen on head magnetic resonance imaging (MRI). Methods: This is a single-center case series, from a tertiary public university health center in Montreal, Canada. Cases of children with possible PEPP were selected prospectively from 2005 to 2017, based on head MRI findings. History, exam findings and hormonal evaluation were extracted from the medical record, and images were reviewed and interpreted by an experienced pediatric neuro-radiologist.

Results: All the cases, two boys and four girls between 8 days and 14 years old, were characterized by the presence of two midline bright spots on the thin focused T1 weighted sequences obtained with fat suppression technique. While one bright spot was located at the normal expected site of the neurohypophysis in the posterior sella, another was in the midline median eminence or along the normal appearing pituitary stalk above the sella, most likely corresponding to a partial presentation of an ectopic posterior pituitary gland. The possible PEEP was associated with different clinical phenotypes. One patient had isolated growth hormone deficiency, another had combined thyroid stimulating hormone and growth hormone deficiency, while the others had intact pituitary function at their last follow up. Of the remaining four patients, one had CHARGE syndrome, another one had motor developmental delay and one had septo-optic dysplasia without evidence of endocrinopathies to date.

Conclusions: Anterior pituitary hormone deficiencies may be detected in patients with PEPP found on head MRI. Long-term follow-up may provide additional information on the development of other pituitary hormone deficiencies.

Poster Abstract 11

A case of hypothyroidism following enteral iodinated contrast media: Is it time to consider routine screening for thyroid dysfunction in infants following iodine exposure?

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Case: On day 29 of life, an ex-28 week male had an elevated TSH of 23.28 mIU/L (0.8-9.0)] and low free-T4 of 8 pmol/L(9.8-23.0 pmol/L) during assessment of enteroparesis. Pertinent history included negative newborn screen, and iodine exposure during barium enema on DOL 15 and barium swallow on DOL 23. On examination, there was no goitre or hemangioma. Repeat thyroid function testing (TFT) on DOL 32 showed TSH 40.59 mIU/Land free-T4 6.7 pmol/L. Thyroid scan on DOL 34 was normal. Urine iodine on DOL 37 was markedly elevated [3188 μg/L (42-350)]. A provisional diagnosis of primary hypothyroidism secondary to iodine exposure was made and levothyroxine 25 mcg was started on DOL 34. Based on of TFT trends, levothyroxine was weaned on DOL 91 (12.5 mcg) and a successfully discontinued on DOL 165. Background Iodinated Contrast Media (ICM) is a source of excess iodine and has been described as a risk factor for the development of hypothyroidism in both term and preterm infants. The US Food and Drug Administration and Health Canada have both issued safety alerts on risk of hypothyroidism in infants exposed to ICM. Most literature is related to IV exposure. We report the second case of ICM-induced hypothyroidism following enema in a premature infant. In the previously reported case, TSH was normal 6 days after exposure but rose to 368 mIU/mL 61 days after ICM exposure. In both cases there was a history of dysmotility, possibly leading to extended exposure to high iodine load and in turn increased risk of development of hypothyroidism and delayed onset of the abnormality.

Conclusion: Given the potential impact of hypothyroidism during the neonatal period, we recommend: • An awareness of the potential risk of hypothyroidism in neonates following exposure to ICM, including enteral exposure. • Use of non-iodinated contrast media when possible. • Consideration of screening TFTs in neonates exposed to ICM, particularly those with additional risk factors including gastric dysmotility when exposed enterally. • Reporting of cases of hypothyroidism secondary to ICM to support the development of future guidelines.

Poster Abstract 12
An Allergy to a Life-Saving Medication?

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Case: A nine-year-old female with celiac disease was diagnosed with type one diabetes after presenting in severe diabetic ketoacidosis. She was initially started on three times daily insulin injections, which were well tolerated. Ten months after diagnosis, the patient and her family elected to start pump therapy with OmniPod®. After two years and five months on OmniPod® with Novorapid®, the patient developed impressive lipoatrophy at her pod sites. Dermatology was consulted to evaluate for the possibility of steroids, but did not recommend topical or local intervention. Other brands of fast-acting insulin were then tried (including Humalog® and Apidra®), but the patient developed erythematous, indurated lesions at this site of pod insertion. Different adhesives and skin protectors were used without improvement. To rule out an allergy to the catheter or the pod itself, two pods were placed – one with an insulin infusion and one with only saline. No reaction developed at the site of the pod infusing saline. Therefore, an insulin allergy was suspected, worryingly to multiple types of fasting-acting insulin. Systemic antihistamine medication was then added without success. At this point, Allergy/Immunology was consulted for possible desensitization to insulin, but a trial off the pod was first recommended. Interestingly, for the first two weeks on multiple daily injections, the patient did not have any cutaneous reactions. However, she then again developed localized erythema to Humalog® injections, although not to Lantus® injections. These reactions lessened with topical Benadryl® cream and a longer pen needle. At the present time, the patient and her family are eager to restart pump therapy, this time using a Medtronic® pump with a longer catheter for infusion.

Discussion: Insulin allergy is rare, but when present, creates great difficulty for clinical management. More than two years after diagnosis, this patient developed a localized cutaneous reaction to most of the available rapid preparations of insulin. It currently remains to be seen whether a different continuous delivery mechanism, with continued topical antihistamines, may be helpful in this patient.

Poster Abstract 13
Toddler Thyroid Troubles

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Introduction: A 23-month-old male was admitted for persistent rash, periorbital edema, and an enlarging neck mass. The pustular and petechial rash was primarily on his scalp and had been unresponsive to multiple treatment modalities. Initial work-up for his neck mass revealed a TSH of 19.7mU/L. Pediatric Endocrinology was consulted to initiate treatment for hypothyroidism.

Case Description: The toddler had a strikingly large goiter which had rapidly progressed over 1 month. It was prudent to determine the etiology prior to proceeding with treatment. His age of presentation was perplexing, as this would be a late presentation for congenital hypothyroidism, but early for acquired autoimmune etiologies. His TSH on newborn metabolic screening was normal. He consumed an iodine replete diet. Anti-thyroglobulin and anti-thyroperoxidase antibodies were negative. A repeat TSH was 28.6 mU/L and free T4 was 6.8 pmol/L. Thyroid ultrasound demonstrated a diffusely enlarged and hypervascular gland, indicating high metabolic activity which was atypical in a biochemically hypothyroid patient. An initial skin biopsy was suboptimal for evaluation, though did not show any obvious inflammatory infiltrate. There was no suggestion of infiltrative disease based on liver enzymes and peripheral blood smear. Lymphopenia was identified and the working diagnosis shifted towards an immunodeficiency. The patient was scheduled for additional biopsies (skin and ophthalmologic). Given the unclear etiology of his goiter, a thyroid biopsy was suggested and ultimately yielded a diagnosis of langerhans cell histiocytosis (LCH). Cytology revealed atypical histiocytic infiltrate. No normal thyroid elements were identified. A CT chest and neck demonstrated significant mass effect on the subglottic airway with the narrowest tracheal segment measuring 6mm anterior to posterior.

Discussion: The patient was diagnosed with multisystem LCH. Sites of involvement included scalp, maxilla, neck, mediastinum, right axilla and pleura. Corticosteroids were initiated urgently to reduce airway compromising mass effect. Rarely do goitres in children become large enough to compress the airway; however, this case highlights the necessity to consider critical neck structures when a large goiter is present. The patient's hypothyroidism and goiter was secondary to LCH infiltration which is a rare etiology of primary acquired hypothyroidism.

Poster Abstract 14
Evaluating the natural history of subcutaneous fat necrosis

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Background: Subcutaneous fat necrosis (SCFN) of the newborn is an uncommon condition most commonly seen in term infants who have experienced perinatal stress presenting as nodules or plaques on the face, back or upper extremities. The associated complication of hypercalcemia is thought to be secondary to elevated 1,25-OH vitamin D from increased expression of 1-alpha hydroxylase from inflammatory granulomatous cells. However the natural history of SCFN, associated hypercalcemia and its possible long term effects has not been well described. Clinical observation at the Hospital for Sick Children (HSC) suggests that ongoing hypercalcemia, albeit less severe than at presentation, persists for a number of months beyond the resolution of palpable nodules. Given this, our objectives were to evaluate the natural history of SCFN, associated hypercalcemia and serial laboratory values.

Cases: We reviewed the cases of 8 children diagnosed with SCFN and associated hypercalcemia followed in the HSC Calcium Clinic; 7 of 8 followed to resolution. Of the 8 children; 5 presented following hypoxic ischemic encephalopathy and cooling, 1 following traumatic vacuum assisted delivery and 2 were found to have incidental skin lesions. On average hypercalcemia was detected at 1 month of age, with a peak calcium value of 3.42± 0.56 mmol/L. The children were typically seen in follow up every 4 months, and after initial hospital treatment, were managed on a restricted calcium intake. Based on serial clinical exams, palpable subcutaneous nodules resolved on average after 10.5±7.6 months. The resolution of hypercalcemia lagged behind, with a normalization of serum calcium occurring at 25.3±9.5 months. Four of the children developed nephrocalcinosis. These children had the highest peak serum calcium values at diagnosis of the group.

Discussion: Given hypercalcemia can persist well beyond the clinical resolution of nodules, there is risk that these children may be discharged from follow-up prematurely with possible persistent urinary calcium excretion and risk of nephrocalcinosis. A closer look at other, possibly predictive factors, including 1,25 vitamin D levels and duration of dietary restriction may be useful in guiding our future monitoring and management of these patients.

Poster Abstract 15
Vaginal Bleeding in a Premature Neonate—An Interesting Case

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A female infant born via spontaneous vaginal delivery at 23+2 weeks of gestation, birth weight 540 grams was admitted to the Neonatal Intensive Care Unit due to respiratory distress syndrome and followed for comorbidities associated with prematurity. L-thyroxin for hypothyroidism was started at 31+2 weeks corrected gestational age (CGA) for a persistent elevation of TSH. At 51+5 weeks CGA she experienced a vaginal bleeding that lasted for 7 days associated with Tanner II breasts. Other clinical findings included: swelling of the clitoral hood and labia majora, and one café-au-lait spot 2 cm in diameter in the left axilla. Investigations showed elevated estradiol (329 pmol/L), FSH 3.2 IU/L and LH 2.4 IU/L, normal random cortisol and normal thyroid function on treatment. Bone age was not advanced. Pelvic ultrasound revealed a prominent uterus for patient's age and a dominant left ovarian follicle 8 x 7 mm, and no ovarian mass. Alpha-fetoprotein and total HCG were negative. Skin pigmentation was not characteristic of McCune Albright Syndrome. A brain MRI did not show pituitary or hypothalamic lesions. Further history revealed maternal use of galactagogues four weeks prior to the onset of vaginal bleeding, including 6 different products containing 9 herbal ingredients with phytoestrogens properties. No treatment was initiated for the vaginal bleeding at this time. Given the lack of literature on the pharmacokinetics of these products, breast milk was discontinued. Three monthly vaginal bleeding occurred, after which time they ceased. Breast enlargement regressed. Gonadotropins and estradiol levels decreased progressively and pelvic ultrasound was unremarkable. Her growth velocity continues to be normal.

Our case highlights: 1) the uncertainty of the dose at which phytoestrogens used as galactagogues can stimulate the hypothalamic-pituitary-gonadal (HPG) axis in young infants; 2) challenges in differentiating between a primed axis leading to central precocious puberty and an atypical physiologic post-natal gonadotropin surge (mini-puberty); 3) lack of consensus on use of GnRH agonists versus a watch and wait approach in young infants.

Poster Abstract 16
Hypercalcemia in a mother and her infant: Expanding clinical presentation in heterozygous CYP24A1 mutation (c.1186C>T) in infancy

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Background: Idiopathic infantile Hypercalcemia (IIH) is an uncommon disorder with a variable clinical presentation. It is characterized by increased serum and/or urinary calcium, elevated 1,25 dihydroxyvitamin D (1,25(OH)2D), and suppressed PTH. Current treatment includes a diet low in calcium and vitamin D. Biallelic mutations in the CYP24A1 gene, which encodes the principle enzyme that inactivates vitamin D metabolites, cause most cases of IIH. However, the clinical significance of heterozygous CYP24A1 mutations is less clear. We present a case of a newborn, who presented with the characteristic biochemical profile of IIH and was found to have a heterozygous pathogenic mutation in the CYP24A1 gene (c.1186C>T).

Case: The infant's mother (G1P1) had a history of nephrolithiasis and chronic hypercalcemia. Her albumin-corrected total serum calcium at delivery was 2.9 mmol/L. Her female infant was asymptomatic and hypercalcemia (2.8 mmol/L) was discovered when she was assessed at 3 weeks of age. In addition, she had elevated 1,25(OH)2D (477 pmol/L, ULN 190), increased urinary calcium:creatinine ratio (1.12) and PTH in the lower normal range (21ng/L, normal 12-78), consistent with IIH. Vitamin D supplementation was stopped and she was started on a low-calcium/vitamin D-free formula (Calcilo XD) at the age of 3 weeks. This treatment resulted in stable blood calcium slightly above normal range. DNA analysis for the CYP24A1 gene confirmed a pathogenic heterozygous mutation in exon 9 [(c.1186C>T; p.Arg396Trp)] inherited from her mother, who was found to be homozygous for the same mutation. Sanger sequencing as well as deletion/duplication analyses, were normal for the paternal CYP24A1 allele. The clinically asymptomatic maternal grandparents were not related yet carried the same heterozygous CYP24A1 mutation.

Discussion: Only a few patients with nephrolithiasis and/or nephrocalcinosis are found in literature to have heterozygous CYP24A1 mutations, none of them were described to have IIH. Thus far the Arg396Trp mutation has been associated with IIH only in homozygous or compound heterozygous individuals. We present an infant with the characteristic profile of IIH who was found to be heterozygous for this mutation.

Conclusion: The clinical presentation in heterozygous mutations of the CYP24A1 gene such as c.1186C>T, may be broader than previously appreciated.

Poster Abstract 17
Genome-Wide Meta-Analysis identifies a novel low frequency STK39 variant of large effect on risk of Type 1 Diabetes

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The genetic etiology of type 1 diabetes (T1D) has been extensively studied, with 57 loci identified to date, mainly through genome-wide association studies (GWAS). Most of these known genetic associations involve common variants, while a sizable portion of the missing heritability of T1D may be attributed to unidentified rare single nucleotide polymorphisms (SNPs) (minor allele frequency (MAF) < 5%). The recent availability of large human whole genome sequencing datasets enables the interrogation of rare genetic variation by imputation from directly genotyped data, in search of large genetic effects by rare alleles. Here, through deep imputation, GWAS, and meta-analysis of 12 European cohorts totaling of 9,684 T1D cases and 15,743 controls, we identified 25 independent genome-wide significant variants outside the major histocompatibility complex, among which 4 had MAF in controls < 5%. All 4 rare SNPs had large effects (ORdiscovery>1.5). Three rare variants successfully replicated at p<0.05 and with pcombined < pdiscovery in a separate cohort (4,329 T1D cases from the T1DGC sibling pairs/9,543 controls from UK Biobank), all of which were novel. In silico analysis using topological domains and proximity to functional elements prioritized a low-frequency variant at 2q24.3 (rs60587303 (C), MAF controls 0.5%) within the first intron of STK39, with a ORcombined of 1.97 (95% CI 1.58-2.47, Pcombined 2.9 x 10-9), an effect comparable to those of the insulin (INS) and PTPN22 loci. Additional analysis showed that admixture did not unduly bias results for this SNP. This variant overlaps a DNase I hypersensitivity cluster in 68 cell types and a cluster of transcription factor bindings sites including FOS, JUN and STAT3. Pharmacological blocking of Stk39 in murine T-cells lines increased activation and proliferation of effector T cells and secretion of interferon gamma from CD4+ T cells. Thus, we identified 3 novel rare T1D variants, which have effect sizes larger than most previously described common SNPs. Our findings highlight STK39 as a protein, with no previously known role in T1D, which appears to influence T-cell activation and effector functions. These findings add to the knowledge of the genetic architecture of T1D and demonstrate STK39 as a new T1D gene.

Poster Abstract 18
Tuberculosis as a Rare Cause of Pituitary Dysfunction

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Introduction: Central nervous system tuberculosis (TB) is an uncommon presentation, particularly in the developed world. We present a case of a 12-year-old female who develops hypopituitarism secondary to intracranial tuberculomas.

Case Presentation: Initially, our patient presented with fever, headache, neck pain, diplopia, and emesis. There were no TB exposures, but travel history revealed previous visits to an endemic TB area. CSF culture and PCR were positive for Mycobacterium tuberculosis. MRI brain demonstrated TB meningitis. There was no evidence of disseminated TB. Standard TB treatment was started, including high dose glucocorticoids and RIPE (rifampin, isoniazid, pyrazinamide, ethambutol). Her course was complicated by hydrocephalus requiring temporary EVD, possible seizure requiring prophylactic antiepileptic medications, and acute left sided hemiparesis and facial droop secondary to cerebral vasculitis. Per treatment guidelines, 4-drug therapy was transitioned to 2-drug therapy and glucocorticoids were weaned off. Shortly after, she had acute loss of consciousness associated with worsening left hemiparesis and behavioral changes. Repeat MRI brain revealed new tuberculomas. It was felt this event represented a paradoxical immune reaction. However, TB reactivation could not be ruled out so RIPE and high dose glucocorticoids were restarted. Her clinical status improved over time. After 24 months of total TB therapy and 9 months of glucocorticoids, MRI brain revealed improving tuberculomas and stable vasculopathy. Four months after treatment completion, she presented with polydipsia, polyuria, and nocturnal enuresis. Extensive pituitary hormone testing revealed ADH, ACTH, TSH, GH, and probable LH/FSH deficiencies. She was started on hormone replacement therapy. Follow up MRI brain revealed new tuberculomas and new abnormal thickening and enhancement of the pituitary infundibulum. It was unclear if her disease progression was due to active TB, reactivation of TB, a paradoxical immune reaction, or incomplete TB treatment. After 2 months of standard TB treatment, there was interval worsening. PCR and culture for TB were negative. Immunological work up is pending. Presently, our patient is awaiting novel treatment with infliximab, based on some success seen in adult case reports.

Conclusion: Pituitary tuberculosis is rare and difficult to diagnose radiologically but must be considered in cases of pituitary stalk thickening.

Poster Abstract 19
Use of flash glucose monitoring to screen for dysglycemia in pediatric patients with cystic fibrosis: a Feasibility Study

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Early intervention in cystic fibrosis-related diabetes (CFRD) can limit adverse impacts on nutrition, respiratory function, and longevity. Diagnosis is challenging in asymptomatic patients since oral glucose tolerance testing (OGTT) is inconvenient and subject to high intra-individual variability. We assessed the feasibility of flash glucose monitoring to screen for dysglycemia in pediatric patients with cystic fibrosis (CF). We recruited CF patients aged 10 to 18 years without pre-existing diabetes. Study participants underwent OGTT with measurement of glucose, insulin, and C-peptide at 0, 30, 60 and 120 minutes. Serum was collected for biomarker analysis. Participants wore a Freestyle Libre sensor for up to 14 days following OGTT. Of 41 patients contacted, 10 (24%) underwent OGTT. Seven of 10 participants (70%) wore a sensor. The mean age was 12.1 ± 0.9 years and 43% were female. The average BMI z-score was 0.8 ± 1.2 and the average FEV1 was 85.6 ± 7.9%. Fasting blood glucose levels were normal in 6/7 patients (mean 5.4 ± 0.2 mmol/L). One patient had impaired fasting glucose. Dysglycemia was identified by OGTT in 4/7 subjects; indeterminate glycemia (n=1), impaired glucose tolerance (n=2), and diabetes (n=1). The participant who screened positive for CFRD was not on insulin or oral or IV corticosteroids during the study. The change in BMI z-score over the preceding 6 months was -0.04 ± 0.05 (normal glucose tolerance) and -0.34 ± 0.13 (indeterminate/impaired glucose tolerance). Participants wore sensors for 9.8 ± 1.7 days and performed 6.8±0.9 scans/day. Mean sensor glucose was 5.3 ± 0.5, 6.3 ± 0.2, and 6.9 mmol/L for normoglycemic (n=3), indeterminate/impaired fasting glucose (n=3), and diabetes participants (n=1), respectively. The proportion of time above 8 mmol/L was: 7.0 ± 3.2%, 12.7± 0.5%, and 18%, respectively.

Conclusions: Increased mean glucose or time above range detected by flash glucose monitoring may be a useful screening tool to detect early dysglycemia in children with CF. Its use may be limited by cost and patient acceptability.

Poster Abstract 20

Avoidant/Restrictive Food Intake Disorder in Children and Adolescents: An Important Consideration in Patients Referred to Endocrinology for Short Stature and/or Poor Linear Growth

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Background: Avoidant restrictive food intake disorder (ARFID) is a newly classified disorder in the Diagnostic and Statistical Manual of Mental Disorders (fifth edition). ARFID encompasses heterogeneous clinical presentations that result in either significant weight loss, nutritional deficiency, dependence on enteral feeding or nutritional supplements, and/or marked interference in psychosocial functioning.

Objective: The present study sought to examine the number of referrals, assessment findings, and treatment response of youth referred to an ARFID subspecialty clinic between December 2014 and May 2017 by pediatric endocrinologists. Method: A retrospective study was completed of consecutive referrals received over a 30 month period.

Results: A total of 30 patient referrals were accepted and assessed over the study time frame. Of these, 17% of referrals (n=5; 3 males and 2 females) were referred by a pediatric endocrinologist. In all cases, the presenting concern was short stature and/or poor linear growth secondary to poor weight gain. All youth had a history of poor nutritional intake but did not have features of an anorexia nervosa, bulimia nervosa or any other comorbid eating disorders. In all 5 cases, weight was more affected than height. Three patients were being treated concurrently with growth hormone (GH). One additional patient was diagnosed with ARFID after reaching a sub-optimal final adult height following GH therapy. Patients were followed for ARFID for a mean duration of 239 days (range 32-461). All 5 patients had improvement in BMI SDS from -2.51 (-3.60 to-1.72) at presentation to -1.75 (02.93 to -0.93) at final assessment. All 4 patients with remaining growth potential at presentation had improvement in height with mean height SDS improving from -2.84 to -2.14. Final adult height had not been reached at the time of final assessment.

Discussion: ARFID is heterogeneous disorder that can manifest in a number of different ways and with a variety of different symptoms including poor growth. It is imperative that pediatric endocrinologists consider ARFID during their evaluation of short stature to ensure that treatment approaches address nutritional challenges when indicated in order to optimize patient's ability for ongoing growth.

Poster Abstract 21
Diagnostic Dilemma in a 46-XY Female

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Introduction: Disorders of sex development (DSD) are conditions with discrepancies between the chromosomal, gonadal, and phenotypic sex. A 17-year-old female was investigated for primary amenorrhea and found to have bilateral adnexal masses. The masses showed significant interval growth and she was referred for evaluation prior to surgical removal.

Case Description: The previously healthy patient presented with primary amenorrhea at 17 years old. She had undergone female puberty with thelarche developing to Tanner stage V. There were no signs of virilization or adrenal insufficiency and she was non-dysmorphic. The patient's body habitus was tall and lean and she had broad hands and long fingers. Breasts and pubic hair were Tanner stage V. A positive feature on exam was an elongated clitoral length (2cm). Blood pressure was normal. Laboratory investigations included the following: an elevated beta-HCG (93 IU/L), evidence of gonadal insufficiency with elevated FSH (43 U/L), estradiol of 145 pmol/L, and testosterone of 3.8 nmol/L. Pelvic MRI showed a 9.4 cm left mass and 8.3 cm right mass and normal uterus. Karyotype showed a 46 XY complement and a DSD panel showed a heterozygous pathogenic variant in POR gene responsible for P450 oxidoreductase deficiency (PORD). Normal appearing vagina, cervix, uterus and Fallopian tubes were confirmed intra-operatively. Pathology identified bilateral dysgerminomas arising in gonadoblastomas with no underlying gonadal tissue.

Discussion: PORD is a unique congenital adrenal hyperplasia variant with variable phenotypic presentation. Manifestations include DSD, glucocorticoid deficiency, and skeletal dysplasia. In retrospect, the patient's PORD malformation score was 0. She tolerated surgery without steroid supplementation but it will be prudent to formally assess the functionality of her adrenal axis. Glucocorticoid requirements described in PORD can be permanent or only with physiological stress. After resection, beta-HCG, testosterone, and estradiol were undetectable. Testosterone and estrogen production were attributed to the gonadoblastomas, with testosterone formation likely occurred later to account for minimal virilization. After resection, beta-HCG, testosterone, and estradiol were undetectable. This case highlights ongoing difficulty in diagnosing individuals with DSD despite progressive understanding of some etiologies, and emphasizes the need for complementary investigations, both biochemical and genetics.

Poster Abstract 22
Conjugated Hyperbilirubinemia Among Infants with Hyperinsulinemic Hypoglycemia

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Objectives: A subset of neonates presenting with hyperinsulinism develop conjugated hyperbilirubinemia. The relationship between these two conditions has not been previously described. We aimed to characterize the subset of hyperinsulinism patients who develop neonatal cholestasis and to assess how these patients differ from their non-cholestatic counterparts.

Methods: A retrospective chart review was performed. Within a larger dataset of 98 patients with hyperinsulinism, 20 patients were flagged as having conjugated hyperbilirubinemia. Data was collected regarding their demographics, hyperbilirubinemia work-up and natural history of the conjugated hyperbilirubinemia. Characteristics of the hyperbilirubinemic and non-hyperbilirubinemic subgroups were then compared.

Results: The 20 patients with conjugated hyperbilirubinemia and hyperinsulinism had a mean gestational age of 36 weeks and a mean maximum conjugated bilirubin of 104 umol/L. No underlying etiology for the hyperbilirubinemia was found in any of the 20 cases. Hypopituitarism was excluded in all patients. No infant was found to have biliary atresia, though 11 patients had HIDA scans and 3 had cholangiograms. Liver biopsies were performed on 5 patients, all with non-specific cholestatic and inflammatory findings. All 5 of these patients had complete spontaneous resolution of their conjugated hyperbilirubinemia. Of all 20 infants, 18 (90%) had documented resolution. The mean time to resolution was 86 days. When compared to the other children with hyperinsulinism, the conjugated hyperbilirubinemia patients had lower mean birth weights (z-score -0.94 vs -0.11, $p=0.04$), were more likely to have experienced fetal distress (75% vs 42%, $p<0.01$), and presented earlier with hypoglycemia (day 1 of life vs day 67, $p=0.01$). There was no difference in gestational age, or percentage of infants with congenital versus transient forms of hyperinsulinism.

Conclusions: A significant percentage of infants with hyperinsulinism present with conjugated hyperbilirubinemia from an unknown etiology. The association with lower birth weight and history of fetal distress potentially suggests that intrauterine factors leading to hyperinsulinism in these infants may also predispose towards conjugated hyperbilirubinemia. While further investigation is needed, the association of conjugated hyperbilirubinemia and hyperinsulinism calls into question the need for invasive investigations for the conjugated hyperbilirubinemia in this patient group.

Poster Abstract 23
Central Venous Cathether-Associated Thrombosis in Children with Congenital Hyperinsulinism

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Background & Objectives: Congenital Hyperinsulinism (CHI) is the most common cause of severe and persistent hypoglycemia in infancy, often requiring high concentrations of dextrose through a central venous catheter (CVC) to achieve euglycemia and prevent permanent neurologic injury. However, CVCs are a significant risk factor for thrombosis in the pediatric population. We therefore aimed to investigate the incidence and risk factors for CVC-associated thrombosis in patients with CHI and the outcomes of enoxaparin prophylaxis.

Methods: 3-year (2014-2017) retrospective study of patients with CHI requiring CVC (n=33) not on enoxaparin, and a separate cohort (n=7) receiving enoxaparin prophylaxis. Thrombosis incidence, and association with CHI and CVC-related factors were analysed.

Results: Six (18%) patients developed CVC-associated thrombosis (4.2 thromboses/1000 CVC days). There was no difference in the frequency of genetic mutations or focal CHI. However, the presence of compound heterozygous/homozygous ABCC8 or KCNJ11 mutations correlated with thrombosis in a stepwise regression model [R2=0.40, p=0.001]. No difference was observed in CVC duration, CVC features, high concentration dextrose or glucagon infused through the CVC. None of the patients on enoxaparin prophylaxis developed thrombosis or bleeding complications, and their characteristics did not significantly differ from those with thrombosis.

Conclusions: CVC-associated thrombosis can occur in a significant proportion of patients with CHI. Intrinsic factors determining CHI severity may increase this risk, for which anticoagulant prophylaxis may be indicated.

Poster Abstract 24
Determinants of successful weight loss in a pediatric behavior modification program: the CIRCUIT Program Experience

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Background: The CIRCUIT program is a lifestyle intervention using personalized strategies to increase physical activity (PA) among youth aged 5-18 years at risk for cardiovascular disease. The program has previously been shown to be effective at improving cardiometabolic health outcomes including body mass index, diastolic blood pressure, as well as aerobic and anaerobic fitness levels. Why the program succeeds in some but not others remains unclear.

Objective: To identify potential determinants of successful weight loss i.e. baseline characteristics associated with a reduction in body mass index z-score (zBMI) among youth enrolled in the CIRCUIT program.

Methods: Baseline characteristics included sociodemographics (age, sex, ethnicity, maternal education, family structure), psychosocial factors (multiple items assessing maternal support to be active), and zBMI. The program was considered to be successful if participants had a reduction in zBMI of at least 0.2 SD over a period of one year. Among the final study sample of 170 participants, 46% were girls, 64% were Caucasians, and 69% lived in 2-parent households. Mean baseline zBMI was 3.3. Differences in baseline characteristics between the success and non-success groups were compared using chi-squared- and t-tests.

Results: Participants in the successful group (n=79), on average, were younger (10.0 vs 12.2 years, $p<0.01$) and had a higher baseline zBMI (3.5 vs 3.2, $p=0.07$), compared to participants in the non-successful group (n=91). They also were more likely to report having mothers who enjoyed doing PA (54% vs 27%, $p=0.02$) and would plan PA for outings (48% vs 22%, $p=0.01$). No other group differences were observed.

Conclusion: The CIRCUIT program was more successful among participants who were younger, had a higher zBMI and had greater maternal support for PA. In order to optimize success in lifestyle interventions for youth, future research should investigate the underlying mechanisms of the observed associations.

Poster Abstract 25
Not a Simple Febrile Seizure: Thyroid Storm in a Toddler

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Introduction: Thyroid storm is an uncommon complication of hyperthyroidism and is even less common in young children. We present a case of thyroid storm in a toddler, initially diagnosed as a febrile seizure.

Case: A two-year-nine-month-old female presented to the emergency department due to seizure-like activity at home. She had been ill for two days with fever, emesis, and diarrhea. Initial vitals showed fever to 39.1, tachycardia to 202, hypertension to 129/61, and weight of 14.4 kilograms. She was alert but irritable. Laboratory investigation revealed metabolic acidosis. She received fluid resuscitation and was admitted to the hospital for further management of a possible febrile seizure. Several hours later, a goiter was noted. Further history revealed sweating and poor weight gain for several months as well as hyperthyroidism in the mother and several maternal aunts. Repeat investigations showed TSH <0.02mIU/L (reference range 0.34 – 5.6) and FT4 60.30pmol/L (reference range 8 – 18). Both anti-TSH receptor antibodies and anti-thyroid peroxidase antibodies were markedly positive. Criteria for thyroid storm were met given hyperpyrexia, tachycardia, gastrointestinal dysfunction, and likely seizure. The patient was transferred to the pediatric intensive care unit and started on methimazole 5mg PO q6hrs, propranolol 1mg/kg PO q12hrs, and hydrocortisone 50mg/m2 IV q8hrs. Lugol's iodine solution four drops q8hrs was added within several hours given continued tachycardia, hypertension, and rising FT4 to 70.4pmol/L. Vital signs improved over the next 48 hours. Lugol's iodine solution was continued for six days, and hydrocortisone was weaned over one week. The patient was discharged thirteen days after admission on methimazole 5mg PO q6hrs and propranolol 15mg PO q8hrs; FT4 was 31.3pmol/L. Two days after discharge, methimazole was increased to 10mg PO q8hrs due to non-declining FT4. Now four months after presentation, the patient remains clinically well. TSH is now detectable and FT4 is declining, necessitating careful weaning of methimazole.

Discussion: This is one of the youngest reported cases of thyroid storm. Thyrotoxic crisis should be considered in the differential diagnosis for toddlers with febrile illness and seizure. Medical treatment with methimazole, propranolol, hydrocortisone, and iodine was safe and effective in this case of pediatric thyroid storm.

Poster Abstract 26
A Unique Form of Rickets

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Background: Vitamin D is a vital hormone in preserving calcium and phosphorus homeostasis in the body and maintaining normal growth and mineralization of bones.

Case Presentation: We encountered a patient with signs and symptoms suggestive of rickets. This patient had rachitic skeletal manifestations, macrocephaly, retarded motor development, hypocalcemia, hypophosphatemia, markedly elevated alkaline phosphatase, and secondary hyperparathyroidism. She was initially treated with cholecalciferol as a classic case of vitamin D deficiency. However, she exhibited no clinical improvement. Further investigations showed normal 25 hydroxyvitamin D [25(OH)D] and low 1,25-dihydroxyvitamin D [1,25(OH)2D]. These biochemical abnormalities are consistent with the diagnosis of vitamin D dependent rickets type 1A (VDDR1A). Molecular analysis of CYP27B1 gene revealed novel homozygous mutation "Gly125Arg". Unaffected parents were heterozygous carriers.

Conclusion: It is highly suggestive that the Gly125Arg mutation in the CYP27B1 gene is the cause of this disease in our patient. This mutation is likely to be a pathogenic mutation that leads to VDDR1A.

Poster Abstract 27
Bisphosphonate-related osteonecrosis of the jaw in a pediatric patient

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Introduction: Bisphosphonates are used to treat osteoclast-mediated bone resorption, including osteogenesis imperfecta, osteoporosis, cancer-related osteolysis, and malignant hypercalcemia. Bisphosphonate-related osteonecrosis of the jaw (BRONJ) is a serious side effect in adult patients, defined by exposed, avascular, non-healing bone in either the mandible or maxilla in association with current or previous intravenous or oral bisphosphonate therapy. We herein present a case of BRONJ in a pediatric patient who has received longstanding intravenous bisphosphonate treatment.

Case Description: A 14-year-old girl with a severe form of deforming osteogenesis imperfecta (COL1A1 gene mutation) that was diagnosed soon after birth. She had sustained numerous bone fractures and bony deformities and had been managed on intravenous bisphosphonate therapy since 6 months of life. She was initially treated with pamidronic acid then switched to zoledronic acid two years ago. She had tolerated bisphosphonate therapy with minimal side effects. Over the past 6 months, however, she experienced loosening of her right upper molars, which led to visits to various dental professionals. Investigations, including nuclear white cell scan and assessment by an oral dental surgeon, confirmed a diagnosis of osteonecrosis of the jaw.

Discussion: BRONJ is well described in the adult literature. It is previously believed that BRONJ is not an entity seen in children. We present a case of osteonecrosis of the jaw in a pediatric patient who has been managed on long-term bisphosphonate treatment. To our knowledge, this is the first case that has been reported to date. This raises the question regarding whether recommendations for use of bisphosphonate and dental screening should be modified in this population.

Poster Abstract 28
Predictors of Lung Metastasis in Pediatric Differentiated Thyroid Carcinoma: A Case Series

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Background: The incidence of differentiated thyroid cancer (DTC) in the paediatric population is lower than adults and, although it frequently presents at a more advanced stage, it is generally associated with an excellent long-term prognosis. Nevertheless, the prevalence of lung metastasis at diagnosis in children is estimated at roughly 25%, 3-4 times higher than in adults. Our understanding of the factors predicting lung metastasis in children lack paediatric-specific data. Beyond a known association between lateral cervical lymph node disease and lung metastases, little is known about factors that predict or predispose to lung metastases in children.

Aims: 1) To characterise a single tertiary paediatric centre's cohort of patients with DTC complicated by lung metastasis; 2) to identify predictive factors for lung metastasis in paediatric DTC.

Methods: A retrospective review of electronic medical records from 1998 to 2017 at a single tertiary paediatric centre to gather age and clinical features at presentation, imaging results, radioactive iodine dose and short-term response to treatment. In addition, cytology and histology results were ascertained.

Results: 19/93 (20%) patients treated for DTC had lung metastasis. Mean age at DTC diagnosis was 11.1 ± 2.4 years and 13.9 ± 2.3 years for those with and without lung metastasis, respectively ($p<0.001$). Lung metastasis were diagnosed at or within 6 months of DTC diagnosis in 13/19 (76%) patients. No patients with microcarcinoma had lung metastasis and all with T4 disease had lung metastasis. All with lung metastasis had papillary thyroid carcinoma while 0/2 with follicular carcinoma had lung metastases. Presence of palpable lymphadenopathy (LN), suspicious LN on ultrasound and pathological LN disease were all associated with pulmonary metastasis ($p\leq0.01$). Post-operative stimulated thyroglobulin >2 μg/L was also associated with lung metastasis ($p=0.0497$). The negative predictive value of post-operative stimulated thyroglobulin <2 μg/L was 93%.

Conclusions: Lung metastasis were present in 20% of this paediatric population and were more common in younger children. Most lung metastases were identified within 6 months of initial diagnosis. There is a positive association of lung metastasis with more advanced tumour stage, clinical LN disease and raised post-operative stimulated thyroglobulin, all with negative predictive values ≥90%.

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