CPEG GCEP

Canadian Pediatric Endocrine Group
Groupe canadien d'endocrinologie pédiatrique

2014 Scientific Meeting

PROGRAM

In cooperation with

a place of mind
THE UNIVERSITY OF BRITISH COLUMBIA

Interprofessional Continuing Education

February 20 - 22, 2014

Le Centre Sheraton Montréal Hotel | Montréal, QC

interprofessional.ubc.ca/CPEG2014

Welcome

Dear Delegates,

I would like to welcome you to the 8th Annual Scientific Meeting of the Canadian Pediatric Endocrine Group (CPEG). Our past meetings have been tremendously successful, and this year's meeting promises to offer our nurses, endocrinologists, and trainees another valuable educational experience. The Organizing Committee has worked hard to craft an agenda that highlights work in Montréal, includes presentations by national and international experts, and emphasizes the work of our fellows.

I would like to thank our sponsors, who make this meeting possible, and in addition thank those companies who sponsor our fellowship awards. Those awards will be announced at this meeting, and they allow us to train future endocrinologists.

I look forward, with you, to an enjoyable and collegial meeting,

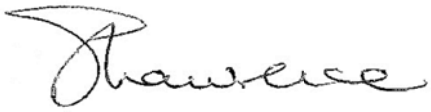
Bienvenue

Chers participants,

J'aimerais vous souhaiter la bienvenue à la 8ème réunion scientifique annuelle du Groupe Canadien d'Endocrinologie Pédiatrique (GCEP). Nos réunions précédentes ont été de belles réussites et cette année encore, notre réunion promet d'offrir aux infirmières (ers), endocrinologues, résidents et autres participants, une expérience éducationnelle de haut niveau. Le comité organisateur a travaillé ardemment pour vous offrir un programme présentant les présents travaux et réalisations du centre hôte, soit Montréal, tout en incluant des présentations d'experts nationaux et internationaux en plus de mettre à l'avant-scène le travail de nos résidents.

J'aimerais remercier nos commanditaires qui rendent cette réunion possible ainsi qu'aux compagnies pharmaceutiques qui subventionnent notre programme de bourse aux résidents permettant ainsi la formation de futurs endocrinologues pédiatres. Les récipiendaires de ces bourses seront d'ailleurs annoncés lors de cette rencontre scientifique.

Je nous souhaite donc une rencontre plaisante et empreinte de collégialité!



Sarah Lawrence, MD, FRCPC
Scientific Chair, CPEG 2014 Scientific Meeting

Table of Contents

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Fellowship Listing

The Canadian Pediatric Endocrine Group would like to acknowledge and thank the following organizations for their generous support in the form of fellowships:

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1994-1995	*Novo Nordisk* - S. Muirhead (Lawrence) *Lilly I* - S. Taback *Lilly II* - A. Simone
1995-1996	*Novo Nordisk* - C. Vaz (50%-one yr) *Lilly I* - S. Taback *Lilly II* - B. Cummings
1996-1997	*Lilly I* - J. Hamilton, E. Sellers *Lilly II* - B. Cummings
1997-1998	*Lilly I* - J. Hamilton *Lilly II* - E. Sellers *Serono* - B. Cummings
1998-1999	*Eli Lilly* - J. Curtis (YR 1) *Serono* - J. Hamilton
1999-2000	*Eli Lilly* - J. Curtis (YR 2) *Serono* - J. Hamilton
2000-2001	*Eli Lilly* - C. Panagiotopoulos (YR 1) *Serono* - C. Huang
2001-2002	*Eli Lilly* - C. Panagiotopoulos (YR 2) *Hoffmann La Roche* - S. Stock
2002-2003	*Eli Lilly* - P. Krishnamoorthy (YR 1) *Serono* - P. Zimakas *Hoffmann La Roche* - R. McEachern
2003-2004	*Eli Lilly* - P. Krishnamoorthy (YR 2) *Hoffmann La Roche* - H. Bui
2004-2005	*Eli Lilly* - M. Nakhla (YR 1) *Hoffmann La Roche* - J. Simoneau-Roy
2005-2006	*Eli Lilly* - M. Nakhla (YR 2) *Serono* - I. Chapados *Hoffmann La Roche* - M. Jetha

2006-2007	*Eli Lilly* - BA Wicklow (YR 1) *Hoffmann La Roche* - S. Amed
2007-2008	*Eli Lilly* - BA Wicklow (YR 2) *Serono* - T. Pinto, B. Babic *Hoffmann La Roche* - J. Deladoey
2008-2009	*Novo Nordisk* - AM Sbrocchi *Eli Lilly* - P. Olivier (YR 1) *Hoffmann La Roche* - T. Pinto
2009-2010	*Novo Nordisk* - R. Shulman *Eli Lilly* - P. Olivier (YR 2) *Serono* - T. Édouard *Hoffmann La Roche* - S. Runge-Wildi *Sandoz* - C. Saaman
2010-2011	*Novo Nordisk* - E. Bassilious *Eli Lilly* - J. Wasserman (YR 1) *Hoffmann La Roche* - Y. Yeshayahu *Sandoz* - S. Tsai
2011-2012	*Novo Nordisk* - M. Millete *Eli Lilly* - J. Wasserman (YR 2) *Hoffmann La Roche* - C. Zuijdwik *Sandoz* - M. Cohen
2012-2013	*Novo Nordisk* - J. Harrington *Eli Lilly* - T. Oron *Serono* - P.Luca *Hoffmann La Roche* - M. Nour *Sandoz* - D. Manouski
2013–2014	*Novo Nordisk* - K. Winston *Eli Lilly* - C. Leblicq *Hoffmann La Roche* - A. Ens *Sandoz* - B. Hursh *Pfizer* - I. Rousseau-Nepton

Program

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

Thursday, February 20, 2014

Time	Session
12:00	CPEG Executive Business Meeting (Salon Musset)
13:00	Fellows Symposium (for CPEG Fellows only) (Salon Hemon) Part I: Meet the Professor Careers and Cases in Pediatric Endocrinology: The Academic Perspective *Dr. Leanne Ward* The Community Perspective *Dr. Susan Kirsch*
16:00	Part II: Associate Members' Business Meeting
16:00	CPEG 2014 Registration Opens (Foyer)
17:00	Welcome Reception & Exhibits (Foyer)
19:00	Adjourn
19:00	Satellite Symposium (Salon Drummond)

Friday, February 21, 2014

Time	Session
07:00	Breakfast, Registration & Exhibits (Foyer)
08:00	Opening Remarks & Welcome (Salon ABC) *Dr. Danièle Pacaud, Dr. John Mitchell, and Ms. Susan Rybansky* Theme I: Fertility (Salon ABC) *Moderator: Anne Marie Sbrocchi*
08:15	Beyond Sequence Variation: Emerging Evidence that Epigenetics is an Important Regulator of Pubertal Timing *Dr. Mark Palmert*
08:55	Preserving Fertility, Preserving Hope: Options for Young Cancer Patients Facing Therapy *Dr. Aaron Jackson*

09:25 Ovarian Function in Anorexia Nervosa
Dr. Madhusmita Misra

10:10 Break & Exhibits (Foyer)

Theme II: Bone (Salon ABC)
Moderator: Nathalie Alos

10:40 Genomics of Osteoporosis
Dr. Brent Richards

11:20 Diagnosis and Treatment of Osteoporosis in Children with Chronic Illnesses
Dr. Leanne Ward

12:00 Lunch (Salon Jerry & Joyce)
Dessert & Exhibits (Foyer)

13:00 Posters (Foyer)

13:30 Split Rooms* (Nursing: see page 8)

Theme III: Diabetes (Salon ABC)
Moderator: Liz Rosolowsky

13:30 Practical Application of Continuous Glucose Monitoring in Diabetes Management
Dr. Margaret Lawson

14:15 Closed Loop Systems to Control Blood Glucose in Adult and Pediatric Patients with Type 1 Diabetes
Dr. Remi Rabasa-Lhoret

15:00 Break & Exhibits (Foyer)

Abstracts (6) (Salon ABC)
15:30 *Moderator: Elizabeth Cummings & Meranda Nakhla*

17:00 Adjourn

FRIDAY NIGHT EVENT (19:00)
Dinner & Entertainment at La Plaza at the Holiday Inn Montreal Midtown (420 Sherbrooke West)

Bus transportation will be provided starting at 18:30. Please meet in the lobby of the Sheraton.
Return buses back to the hotel will start at 21:45. The last bus will leave La Plaza at 22:45.

***Nursing Program for Friday, February 21 (Salon Hemon)**
Moderator: Anne Benzekry & Lina Moisan

Guest Speaker

13:30 Confronting Ethical Challenges in Pediatric Endocrine Nursing
Dr. Franco Carnevale

Highlights from International Meetings

14:30 Highlights from joint meeting 2013 in Milan, Italy
Ms. Susan Rybansky

14:50 Puberty, Growth and a Planning Committee in Portugal . . .
Ms. Brenda Fraser

15:10 Break & Exhibits (Foyer)

15:30 PENS 2013 Highlights

Interesting Cases

15:45 Pumping Cortisol
Ms. June Jacob

16:00 S/C Testosterone: Our Experience
Ms. Lina Moisan

16:15 Syndrome of Inappropriate Antidiuretic Hormone (SIADH) Associated with Carbamazepine Use in a Patient with Central Diabetes Insipidus
Ms. Ronelda Gillis and Ms. Lorianne Peach

16:30 Planning a Family Education Day
Ms. Irena Hozjan and Ms. Nicole Kirouac

17:00 Adjourn

FRIDAY NIGHT EVENT (19:00)
Dinner & Entertainment at La Plaza at the Holiday Inn Montreal Midtown (420 Sherbrooke West)

Bus transportation will be provided starting at 18:30. Please meet in the lobby of the Sheraton.
Return buses back to the hotel will start at 21:45. The last bus will leave La Plaza at 22:45.

***Nursing Program for Saturday, February 22 (Salon Hemon)**

13:00 CPEN AGM

14:30 Center to Center Sharing: What's New?

15:00 Re-join Salon ABC

Saturday, February 22, 2014

Time	Session
07:30	Breakfast (Foyer)
08:00	Business Meeting (Salon ABC)
10:10	CPEG Growth Charts (Salon ABC)
10:35	Break & Exhibits (Foyer)
	Theme IV: Endocrine Disruptors (Salon ABC) *Moderator: John Mitchell*
11:20	Use of Brominated Flame Retardants in our Everyday Environment: Benefits of Fire Safety vs. Risks of Endocrine Disruption *Dr. Cynthia Gates Goodyer*
12:00	Lunch (Salon Jerry & Joyce) Dessert & Exhibits (Foyer)
12:30	Posters (Foyer)
13:00	Split Rooms* (Nursing: see page 8)
13:00	Abstracts (6) (Salon ABC) *Moderator: Tracey Bridge & Robby Stein*
14:30	Break & Exhibits (Foyer)
15:00	John Bailey Award (Salon ABC) *Presented by Dr. Danièle Pacaud*
	Fellowship Awards (Salon ABC) *Presented by Dr. Elizabeth Sellers*
	Clinical Debate (Salon ABC) *Moderator: Nancy Gagné*
15:10	Use of Growth Hormone in Prader-Willi Syndrome Pro: *Dr. Jill Hamilton* Con: *Dr. Arati Mokashi*
16:10	Closing Remarks & Evaluation
16:30	Adjourn

Fellow Abstract Schedule

Time	Title	Presenter	Abstract #	Page
Friday, February 21				
15:30	An invasive prolactinoma in an 11-year-old boy with an *AIP* mutation and dramatic response to medical therapy	Naseem Y Alyahyawi	1	19
15:45	Dysglycemia among Adolescents with PCOS	Nicole Coles	2	20
16:00	Adrenal suppression in children treated with oral viscous budesonide for eosinophilic esophagitis	Shira Harel	3	21
16:15	Hypophosphatemic rickets as the heralding manifestation of cystinosis	Clare Henderson	4	22
16:30	Assessing for autonomic nervous system dysfunction in childhood obesity	Brenden Hursh	5	23
16:45	The Impact of The Treatment of Hypoparathyroidism on Renal Function in Children: long term retrospective follow up study	Isaac Levy	6	24
Saturday, February 22				
13:00	A familial case of congenital thyroid ectopy	Despoina Manousaki	7	25
13:15	Hypertension and hematuria: It's not the kidney!	Colleen A. Nugent	8	26
13:30	Development and Evaluation of Two Online Case-based Modules to Teach Medical Residents Core Pediatric Endocrinology Concepts	Heather Power	9	27
13:45	Shining some light on the powerhouse of the cell- Is there a link between vitamin D and mitochondrial function in humans?	Akash Sinha	10	28
14:00	Prevalence of Non-Alcoholic Fatty Liver Disease in Adolescents with Polycystic Ovary Syndrome	Mrouge Sobaihi	11	29
14:15	RASopathies: A Known Cause of Delayed Puberty; a Cause of Precocious Puberty Too?	Danielle C.M. Van Der Kaay	12	30

Poster Abstract Listing

Title	Presenter	Abstract #	Page
Endocrine manifestations of hepatoblastoma	Allison L Bahm	1	31
A tale of two boys: An Atypical Cause of Pubertal Precocity	Sanjukta Basak	2	32
Organizing the Quebec Interdisciplinary Pediatric Gender Variance Program: From birth through late adolescence	Karine Falardeau	3	33
The Liver That Ate Thyroxine	Chelsey S. Grimbly	4	34
A comparison of quality of sleep and quality of life in patients with glycogen storage disease on standard and modified uncooked cornstarch	Rousseau-Nepton I	5	35
Mind over muscle: Exploring the biology of fatigue in growth hormone deficiency	Akash Sinha	6	36
Is more always better? Deterioration of skeletal muscle function in a boy with Duchenne muscular dystrophy associated with the onset of precocious puberty	Akash Sinha	7	37
McCune-Albright Syndrome Presenting in Infancy	Kate Verbeenten	8	38
When Hormones Paralyze: a Rare Case of Thyrotoxic Periodic Paralysis	Welch John	9	39

Program Organizing and Scientific Committee

Allison Bahm
Anne Benzekry
Elizabeth Cummings
Nancy Gagné
Ronelda Gillis
Sarah Lawrence
John Mitchell
Lina Moisan
Meranda Nakhla
Joanne Nam
Danièle Pacaud
Constantin Polychronakos
Eileen Pyra

Credits

This event has been approved by the Canadian Paediatric Society for a maximum of 9.5 credit hours under the 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

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:

Theme 1: Fertility

Beyond Sequence Variation: Emerging Evidence that Epigenetics is an Important Regulator of Pubertal Timing
Dr. Mark Palmert, Toronto, ON
1: Recognize the limited role that common and rare genetic variants play in regulating pubertal timing within the general population
2: Assess recent findings about how the LIN28/let-7 axis regulates the HPG axis and modulates the timing of puberty
3: Recognize that alternative genetic mechanisms (such as epigenetics) are likely additional, yet largely undiscovered, determinants of pubertal timing

Preserving Fertility, Preserving Hope: Options for Young Cancer Patients Facing Therapy
Dr. Aaron Jackson, Ottawa, ON
1: Provide a summary of patient demographics and fertility preservation practices in Canada
2: Discuss the effects of cancer therapies on male and female fertility
3: Review fertility preservation options and outcomes including oocyte, embryo, sperm, and ovarian tissue freezing

Ovarian Function in Anorexia Nervosa
Dr. Madhusmita Misra, Boston, MA
1: Recognize the impact of anorexia nervosa on the reproductive axis
2: Recognize determinants and consequences of impaired ovarian function in anorexia nervosa
3: Apply available knowledge to develop appropriate therapeutic strategies

Theme II: Bone

Genomics of Osteoporosis
Dr. Brent Richards, Montreal, QC
1: Describe the methodology of a genome-wide association study
2: Recognize what makes a genetic association believable
3: Understand the relevance of modern genomics to the clinic

Diagnosis and Treatment of Osteoporosis in Children with Chronic Illnesses
Dr. Leanne M. Ward, Ottawa, ON
1: Review the current issues in the diagnosis of secondary osteoporosis in children
2: Discuss strategies for monitoring bone development in this setting
3: Summarize the current approach to treatment of osteoporosis in children with chronic illness

Theme III: Diabetes

Practical Application of Continuous Glucose Monitoring in Diabetes Management
Dr. Margaret Lawson, Ottawa, ON
1: Identify candidates that can benefit from CGM use
2: Describe strategies that help people use CGM more effectively
3: Identify information that can be gained from analysis of CGM data

Closed Loop Systems to Control Blood Glucose in Adult and Pediatric Patient with Type 1 Diabetes
Dr. Remi Rabasa-Lhoret, Montreal, QC
1: Review common barriers to optimal blood glucose control
2: Using IRCM Closed loop program describe potential benefits and pitfall of single and dual hormone closed loop glucose control in adults and pediatric patients
3: Illustrate major millstones to get a commercial closed loop device

Theme IV: Endocrine Disruptors

Use of Brominated Flame Retardants in our Everyday Environment: Benefits of Fire Safety vs. Risks of Endocrine Disruption
Dr. Cynthia Gates Goodyer, Montreal, QC
1: Explain the chemistry and function of brominated flame retardants (BFRs)
2: Address the pros and cons of BFR use in our everyday lives
3: Provide scientific data on the endocrine disrupter activities of BFRs and their potential effects on human health

Clinical Debate

Use of Growth Hormone in Prader-Willi Syndrome
Pro: Dr. Jill Hamilton, Toronto, ON
Con: Dr. Arati Mokashi, Halifax, NS
1: Describe the potential benefits and adverse effects of growth hormone therapy in Prader Willi Syndrome
2: Discuss the pros and cons of use of growth hormone in Prader Willi Syndrome
3: Discuss monitoring of children with Prader Willi Syndrome treated with growth hormone

Nursing Program

Confronting Ethical Challenges in Pediatric Endocrine Nursing
Dr. Franco Carnevale, Montreal, QC
1: Identify significant ethical concerns in pediatric endocrine nursing
2: Discuss approaches for analyzing these ethical concerns
3: Outline clinical strategies for addressing these concerns

Biographies

Dr. Franco A. Carnevale
Dr. Franco Carnevale is a nurse, psychologist and clinical ethicist. He completed his undergraduate nursing degree, and master's degrees in nursing, education, and bioethics, and a doctorate in counseling psychology at McGill University, as well a master's degree in philosophy at Université de Sherbrooke and a second doctorate in moral philosophy at Université Laval.

He has also completed graduate studies in health law, anthropology, and cultural psychiatry. Dr Carnevale's primary research interests include a wide range of concerns in pediatric ethics. His current academic appointments include (all at McGill University): Full Professor, School of Nursing; Associate Member, Faculty of Medicine (Pediatrics); Adjunct Professor, Counselling Psychology; Affiliate Member, Biomedical Ethics Unit. His clinical appointments include: Clinical Ethics Consultant, Chair of the Pediatric Ethics Committee, Nursing Consultant, and Associate Member of Pediatric Critical Care, all at the Montreal Children's Hospital-McGill University Health Centre; as well as Clinical Ethics Consultant at Le Phare, Enfants et Familles (pediatric hospice and respite care).

Dr. Cynthia Gates Goodyer
Dr. Cynthia Gates Goodyer is a co-leader (with Dr. Barbara Hales) of a CIHR-funded multidisciplinary research team from five Canadian universities and Health Canada that is using animal models as well as clinical cohorts to investigate the effects of BFRs on developmental abnormalities of reproductive systems and fertility. The team is also exploring ethical, legal and social issues surrounding BFRs since they pose a potential health risk not only to individuals but to future generations.

Her laboratory also studies human growth. They are presently working to identify regions within the human growth hormone receptor (GHR) gene that control expression of the receptor in GH target tissues such as normal bone, liver and fat as well as certain cancers. The goal of this CIHR-funded program is to create a molecular "blueprint" of the GHR gene that will allow researchers to define genetic alterations in children and adults who exhibit abnormal growth or metabolic disorders, including obesity and cancer.

Dr. Jill Hamilton
Dr. Jill Hamilton (MD, MSc, FRCPC) is a Pediatric Endocrinologist at the Hospital for Sick Children, Senior Associate Scientist at the Research Institute, and Associate Professor of Paediatrics at University of Toronto. She is Director of SickKids Team Obesity Management Program (STOMP) and the SickKids Centre for Healthy Active Kids. Her clinical and research interests focus on childhood obesity. These include: (i) understanding pathophysiologic mechanisms of metabolic risk related to weight gain in children; (ii) evaluation of hypothalamic obesity (iii) evaluating biologic and psychosocial determinants of response to obesity treatment.

Dr. Aaron Jackson
Dr. Aaron Jackson is a Gynaecologic Reproductive Endocrinology & infertility Specialist at the Ottawa Fertility Centre, where she coordinates the oncofertility program. She currently sees both male and female patients before and after cancer treatments to discuss their fertility preservation options, or to assess their fertility potential after gonadotoxic exposures. The Ottawa Fertility centre currently offers egg, embryo and sperm freezing, and Dr. Jackson is collaborating with the pediatric oncologists at the Children's Hospital of Eastern Ontario to introduce ovarian tissue freezing in the near future.

As well, she is the current President of a Canadian charity organization called Fertile Future. The mandate of this organization is to raise awareness regarding the risks of infertility secondary to cancer therapies, as well as to provide financial assistance to patients seeking fertility preserving treatments. Dr. Jackson is a member of the Ontario Regional Oncofertility Working Group, a subgroup of the Canadian Partnership Against Cancer that is working towards setting regional and national standards for fertility preservation services for adolescents and young adults with cancer.

Dr. Susan Kirsch
Dr. Susan Kirsch is a pediatric endocrinologist based in the community. She graduated from Bryn Mawr College and attended Temple School of Medicine in Philadelphia. She trained in pediatrics and pediatric endocrinology at the Hospital for Sick Children in Toronto.

Dr. Kirsch practices pediatric endocrinology and general pediatrics at the Richmond Hill Children's Clinic. Dr. Kirsch's diabetes clinic is based at Markham Stouffville Hospital. The clinic services 450 children and families.

She is also on staff at Hospital for Sick Children, Department of Endocrinology, Mackenzie Health Hospital, and Royal Victoria Hospital. She is a consultant to Eli Lilly Corporation for growth hormone related issues. Her research projects have been based both on clinical cases and on collaborative trials in the areas of diabetes and growth hormone related issues. She teaches at the University of Toronto School of Medicine.

Dr. Margaret Lawson
Margaret Lawson completed her medical degree at McMaster University, Pediatric Residency at the University of Western Ontario, and Pediatric Endocrinology Fellowship at Toronto's Hospital for Sick Children following which she joined the staff of the Children's Hospital of Eastern Ontario. She is Associate Professor of Pediatrics at the University of Ottawa and a Senior Scientist, in the Evidence to Practice Research Program of the Children's Hospital of Eastern Ontario Research Institute.

Her research interests include shared decision making, transgender health, prevention of type 1 diabetes, and the use of technology in children and youth with type 1 diabetes.Dr. Lawson is the Principal Investigator of the CGM TIME Trial – Timing of Initiation of Continuous Glucose Monitoring in Established Pediatric Diabetes. The CGM TIME Trial is a multicentre study, funded by the JDRF Canadian Clinical Trials Network, comparing simultaneous initiation of CGM and pump therapy to standard pump therapy with delayed initiation of CGM in children and adolescents with type 1 diabetes.

Dr. Madhusmita Misra
Dr. Madhusmita Misra is a pediatric endocrinologist and Associate Professor of Pediatrics at Massachusetts General Hospital (MGH) and Harvard Medical School. She directs the pediatric endocrine fellowship program at MGH, and is Associate Director of the Harvard CTSA at MGH. She is also Co-Chair of the Research Council at MGH, and a member of the Research Council of the Pediatric Endocrine Society.

Dr. Misra is an NIH funded investigator, and has worked on studies in the female athlete and in adolescents and young adults with eating disorders for over a decade. She is internationally known for her work in this domain, and has been invited to speak at numerous national and international meetings.

Dr. Misra is the recipient of multiple awards including the *John Haddad Young Investigator Award* by AIMM/ASBMR, the *Rita M. Kelly Award* and the *Claflin Distinguished Scholar Award* from Massachusetts General Hospital, and the *Janet W. McArthur Award for Excellence in Clinical Research* from Women in

Endocrinology. Dr. Misra's work in anorexia nervosa and the female athlete triad focuses on elucidating mechanisms that lead to dysregulation of neuroendocrine axes in these populations, and the impact of this dysregulation on bone metabolism, neurocognitive and other outcome.

Dr. Misra has over 90 original research papers, and over 50 reviews, chapters and clinical communications to her credit.

Dr. Arati Mokashi
Dr. Mokashi has been on staff as a Pediatric Endocrinologist at the IWK Health Centre for the past 10 years. She holds an appointment as an Assistant Professor and has a busy clinical and teaching practice. Her areas of interest include, adrenal insufficiency, diabetes and recently gender dysphoria and hormone treatment for transgender youth. She is also a busy wife and mother to 2 little boys. In her spare time she, first and foremost, spends time with her boys, but also enjoys running, biking and playing tennis.

Dr. Mark R. Palmert
Dr. Mark Palmert graduated from the Medical Scientist Training Program at Case Western Reserve University, Cleveland, Ohio, with a MD and PhD in 1992. He completed his pediatrics and pediatric endocrinology training at the Children's Hospital, Boston, where he remained on staff until 2001. He then returned to Cleveland to join the staff at the Rainbow Babies and Children's Hospital and the Departments of Pediatrics and Genetics at Case Western Reserve University. In September 2007, he moved to The Hospital for Sick Children, Toronto, where he is head of the Division of Endocrinology, a senior associate scientist in the Genetics and Genome Biology Program, and an associate professor in the Pediatrics and Physiology Departments at The University of Toronto.

Dr Palmert has extensive experience studying the regulation and disorders of pubertal timing. He has directed clinical studies of precocious and delayed puberty and in parallel has directed a laboratory-based program designed to identify genetic factors that regulate the onset of puberty.

Dr. Remi Rabasa-Lhoret
Dr. Rémi Rabasa-Lhoret is an endocrinologist, associate professor at the Nutrition Department of Université de Montréal, Faculty of Medicine, FRQS senior scholar, J-A De Sève research chair in clinical research. He is directing the metabolic laboratory and a research platform at IRCM . He has published over 160 manuscripts and book chapters and received prizes including in 2010 the young researcher award of the CSEM (Canadian Society of Endocrinology and Metabolism) and the Pierre Bois prize for excellence (2012). Main research fields of his group include external artificial pancreas, obese but metabolically healthy patients and cystic fibrosis related diabetes.

Dr. Brent Richards
Dr. Richards' has trained in genetics, medicine, endocrinology, epidemiology and biostatistics. He currently practices endocrinology and runs a research program at McGill University. His training was supported through a CIHR Fellowship and a Commonwealth Scholarship. His current research program is focused on the identification of the genetic determinants of common, aging-related diseases and the translation of these findings to improved clinical care.

Dr. Richards was a CIHR Clinical Investigator at the Lady Davis Institute of the Jewish General Hospital at McGill from 2008-2012 and currently is an FRSQ Clinician Scientist. He has published 70 peer-reviewed articles and has led several large international consortia, publishing recent findings in Nature Genetics, the Lancet and PLoS Genetics. The CIHR, CFI, FRSQ, MDEIE, CQDM, GSK, Eli Lilly and the Public Health Agency of Canada support his research.

Dr. Leanne M. Ward

Dr. Leanne Ward is an Associate Professor of Pediatrics at the University of Ottawa where she holds a Research Chair in Pediatric Bone Health. She is the Medical Director of the Pediatric Bone Health Clinical and Research Programs at the Children's Hospital of Eastern Ontario and a pediatric endocrinologist within the Division of Endocrinology and Metabolism at CHEO. Dr. Ward completed her medical degree at McMaster University and her training in Pediatrics at Dalhousie University in Halifax, Nova Scotia. She completed her pediatric endocrinology fellowship at St. Justine Hospital (University of Montreal) and trained with Dr. Francis Glorieux in pediatric metabolic bone diseases at the Shriners Hospital, McGill University before taking up her current position at the University of Ottawa in 2001.

Dr. Ward's research program is dedicated to the study of bone development among children with serious chronic illnesses, including children with neuromuscular disorders. She is 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 leukemia, rheumatic conditions and nephrotic syndrome. She has also been the recipient of funding from the Crohn's and Colitis Foundation of America to study musculoskeletal health in children with Crohn's disease. 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 (2004), a Canadian Institutes for Health Research New Investigator Award (2004) and a Canadian Child Health Clinician Scientist Career Enhancement Award (2007).

Disclosure of Conflict of Interest

All speakers 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.

Franco Carnevale
- No affiliation.

Cynthia Gates Goodyer
- No affiliation.

Jill Hamilton
- I have an involvement in research sponsored by or participation in clinical studies concerning the use of the products manufactured by Lilly, Saizen, and Omnitrope.

Aaron Jackson
- No affiliation.

Susan Kirsch
- I have an involvement in research sponsored by or participation in clinical studies concerning the use of the products manufactured by Eli Lilly.
- I have received or expect monetary support from Eli Lilly.

Margaret Lawson
- I have an involvement in research sponsored by or participation in clinical studies concerning the use of the products manufactured by Medtronic Canada.

Madhusmita Misra
- I have received or expect monetary support from Genentech.

Arati Mokashi
- I have an involvement in research sponsored by or participation in clinical studies concerning the use of the products manufactured by Eli Lilly.

Mark Palmert
- No affiliation.

Remi Rabas-Lhoret
- I have an involvement in research sponsored by or participation in clinical studies concerning the use of the products manufactured by Canadian Diabetes Association, Astra-Zeneca/BMS, CQDM, Diabète Québec, Danone, E Lilly, Fondation Fibrose Kystique, Imunotec, IRSC, Medtronic, Merck, Novo-Nordisk, Sanofi-Aventis.
- I have received or expect monetary support from Astra-Zeneca/BMS, Diabète Québec, E Lilly, Medtronic, Merck, Novo-Nordisk, Sanofi-Aventis.

Brent Richards
- No affiliation.

Leanne M Ward
- I have an involvement in research sponsored by or participation in clinical studies concerning the use of the products manufactured by being the principal investigator for an international trial of zoledronic acid in children with steroid-induced osteoporosis.
- I have received or expect monetary support from Novartis, Amgen, and Merck Frosst.

Oral Abstracts

Oral Abstract 1

An invasive prolactinoma in an 11-year-old boy with an *AIP* mutation and dramatic response to medical therapy

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An11-year 2-month-old boy presented with headaches and left anisocoria. He had no papilledema or visual field defects. He was an otherwise healthy prepubertal boy. Head CT and subsequent MRI revealed an expansive tumor that filled the sella turcica, replaced the superior portion of the clivus, sphenoid and cavernous sinuses, and enveloped the cerebral arteries bilaterally. Endocrine work-up revealed prolactin >20,000 mcg/L, FSH 1U/L; LH <0.2 U/L, testosterone <0.4 nmol/L, HCG<2 IU/L and AFP 2.1mcg/L. Additionally, IGF-1,TSH, FT4, and 1 g ACTH stimulation test were normal. Bone age was 11 years and 0 months at a chronologic age of 11 years and 2 months. With the diagnosis of invasive macroprolactinoma, his case was reviewed by the Tumor Board and he was started on bromocriptine,1.25 mg daily, incrementally increasing toward a goal of 10 mg daily. He was transitioned to cabergoline and the dose increased gradually to 0.25 mg daily. Genetic testing of the *AIP* gene revealed a mutation in p.R16H change in the AIP protein sequence caused by c.47G>A variant. Despite our initial impression that this tumor would have a poor prognosis, the patient had impressive clinical response to medical therapy with shrinkage of the tumor size by >50% in the 1st 4 months, and normalization of prolactin level within 4.5 months. In view of his clinical response, he has not yet required surgery or cranial radiation. Prolactinomas are the most common pituitary adenomas in children; however, invasive prolactinomas are much less common. *AIP* mutations are increasingly recognized in patients with early-onset somatotroph and lactotroph macroadenomas. Screening for *AIP* mutations in children should be considered in familial and early-onset or aggressive pituitary adenomas. We will present a literature review of invasive prolactinomas and a discussion of the role of *AIP* mutations in these tumors.

Oral Abstract 2
Dysglycemia among Adolescents with PCOS

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Background: Insulin resistance is known to play an integral role in the pathophysiology of PCOS and patients are at increased risk of impaired glucose tolerance. Our primary objective was to examine the prevalence of impaired glucose metabolism in a large population of adolescents with PCOS. Secondly, we wanted to identify specific clinical and biochemical risk factors associated with an increased risk of dysglycemia and determine the optimal screening method.

Methods: A retrospective chart review was performed on 360 subjects who presented to the Hospital for Sick Children between January 2004 and May 2012. Medical charts were reviewed and relevant demographic, clinical and laboratory data were extracted.

Results: A total of 300 patients fulfilled criteria for the diagnosis of PCOS and 219 of those patients had complete clinical and laboratory data. 163 patients (74.4%) had completed an OGTT, while the other 56 (25.6%) patients had completed only fasting glucose studies. Among the 163 patients with an OGTT, 26 (16.0%) had impaired glucose tolerance and 2 (1.2%) met criteria with for a provisional diagnose of type 2 diabetes. All 28 subjects with dysglycemia were identified by abnormalities on an OGTT. Conversely, the fasting glucose values only successfully detected 2 patients with dysglycemia. Screened adolescents with dysglycemia were more likely to have reported a positive family history ($p = 0.017$), have higher BMI z scores ($p < 0.01$) and higher triglyceride levels ($p < 0.01$). When patients with PCOS were divided into obese ($n = 82$), and non-obese cohorts ($n = 63$), 25.6% of the obese patients as compared to a 7.9% of non-obese patients had laboratory evidence of dysglycemia ($p = 0.008$).

Conclusions: In this largest series to date, we demonstrate that the OGTT is a more sensitive test to detect dysglycemia in adolescents with PCOS. Dysglycemia was associated with many of the known risk factors for metabolic syndrome. Although dysglycemia is more prevalent in the obese population, gysglycemia was also found in non-obese patients suggesting the need for iniversal screening in all adolescents with a diagnosis of PCOS.

Oral Abstract 3
Adrenal suppression in children treated with oral viscous budesonide for eosinophilic esophagitis

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Background: Eosinophic esophagitis (EoE) is an allergic inflammatory condition of the esophagus with increasing prevalence. Oral viscous budesonide (OVB), a topical corticosteroid, is considered first-line treatment due to its low systemic bioavailability. Recent evidence suggests that active EoE is associated with reduced elimination of budesonide. While adrenal suppression has not been reported in the short term, long-term safety of OVB treatment is unknown. Objective: Determine the prevalence of and associated risk factors for adrenal suppression in children with EoE treated with OVB for at least 3 months.

Design: Retrospective review of a quality assurance initiative since June 2012 whereby all children with EoE treated with OVB for at least 3 months were referred for Endocrine assessment including 1 μg ACTH stimulation test. Results: 11 male children (age range 3-17 years) have been assessed to date. Doses of OVB ranged from 0.5 to 2 mg/day. Five patients (45%) had suboptimal stimulated cortisol [range 279-432 nmol/L; mean (± SD) 377.8 (±29.6)], consistent with a diagnosis of adrenal suppression. Notably, none had clinical symptoms or signs of adrenal insufficiency. We found no significant association between test failure and ratio of OVB dose to body surface area or use of concomitant inhaled steroids.

Conclusions: This is the first study to show a high prevalence of asymptomatic adrenal suppression in EoE patients treated with OVB for at least 3 months. These data highlight the need for clinicians to provide families with anticipatory counseling, ensure assessment for adrenal suppression, and have a low threshold to provide stress dosing for endoscopies and intercurrent illness in these patients.

Oral Abstract 4
Hypophosphatemic rickets as the heralding manifestation of cystinosis

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A 17-month-old male was referred to the pediatric endocrinology clinic for marked short stature in the context of elevated alkaline phosphatase (ALP) and low phosphate (PO4). Height velocity had been declining over the past 11 months, with almost no linear growth for the past 6 months, and corresponding poor weight gain. History was significant for constipation, a recent mild increase in fluid intake, and a fraternal twin brother who was healthy with normal growth. On physical examination, height and weight were both well below the third percentile. Remarkable findings included an open anterior fontanel, bony prominence at the wrists, distal femurs and proximal tibias, and genu varum. Plain films confirmed bony changes consistent with rickets. ALP was elevated (1329 U/L, reference 40-390) and PO4 was low (0.96 mmol/L, reference 1.10-2.10) with normal calcium, magnesium, 25(OH) vitamin D, 1,25(OH)2 vitamin D and parathyroid hormone. Urine PO4 fractional excretion was elevated (>50%). Unexpectedly, the urinalysis revealed Fanconi syndrome with glucosuria and proteinuria, despite normal serum values of potassium, chloride, bicarbonate, glucose and albumin. Pediatric nephrology provided urgent consultation and arranged diagnostic testing for cystinosis. An elevated white blood cell cystine level confirmed the diagnosis and he was started on cysteamine. Additional urine testing revealed generalized aminoaciduria and the proteinuria to be tubular in origin, in keeping with the diagnosis. Cystinosis is the most common cause of Fanconi syndrome in children, but remains a very rare condition. The clinical presentation in this case was atypical, with hypophosphatemia causing rickets despite otherwise mild clinical and biochemical features. This case highlights the importance of a screening urinalysis to rule out Fanconi syndrome in any child presenting with hypophosphatemic rickets or growth failure.

Oral Abstract 5
Assessing for autonomic nervous system dysfunction in childhood obesity

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Background: One in four Canadian children is overweight or obese. Furthermore, medical complications related to obesity (such as type 2 diabetes), which were once the purview only of adults, are increasingly observed in children. Dysfunction of the autonomic nervous system (ANS) has been found to occur in obesity. Further studies are needed to clarify the relationship of obesity to autonomic function in children. Objective: To describe the activity of the ANS in children with obesity compared to children with normal weight, as evaluated by heart rate variability and impedance cardiography.

Methodology: Fifteen healthy normal weight children and adolescents (BMI 15 -85th percentile, 12 -18 years) and 15 children with obesity (BMI>95th percentile, 12 -18 years) have been recruited. Subjects meet specified exclusion criteria. Anthropometric data and Tanner staging are recorded. Heart rate variability measurements include resting data, as well as response to a perceived mental stress (mirror tracing task) and a physical stress (hand grip task). Results: At this time, the results from 15 obese and 10 controls have been fully assessed. The control group is older (mean: 16.49 vs. 14.79 years); however, there is similar pubertal status between groups. There is a higher proportion of females in the control group (70%) vs. the obese group (53%). There is no statistically significant difference between the two groups with regards to the balance between sympathetic and parasympathetic tone, measured during rest, as assessed by LF:HF ratio (obese mean 1.36, median 0.86, SD 1.1; control mean 1.27, median 0.96, SD 0.8; p=0.978). Additionally, there is no difference in other heart rate variability parameters at rest, including parasympathetic balance (HFnu) and sympathetic activity (PEP). Finally, there is no difference between groups in the percent change in HFnu or PEP in response to mental or physical stress.

Conclusion: In this small cross-sectional study, we were unable to detect a meaningful difference in ANS balance between obese vs. normal-weight children at rest and in response to stress. Results for all 30 participants will be available for presentation at the CPEG 2014 conference.

Oral Abstract 6

The Impact of The Treatment of Hypoparathyroidism on Renal Function in Children: long term retrospective follow up study

ISAAC LEVY, JENNIFER HARRINGTON, AND ETIENNE SOCHETT

Background: The management of patients with hypoparathyroidism is a balance between providing sufficient treatment with calcium and vitamin D analogs to avoid hypocalcemia, and overtreatment which can increase the risk for hypercalcuria and nephrocalcinosis. Studies of adults with hypoparathyroidism have shown significant rates of nephrocalcinosis and impaired renal function. Little is known about the impact of the treatment of hypoparathyroidism on renal function in children.

Objectives: To determine the prevalence and predictors for renal abnormalities (nephrocalcinosis and decreased estimated glomerular filtration rate [eGFR]) in children with treated hypoparathyroidism.

Design/Methods: A retrospective chart review of patients with permanent hypoparathyroidism, a disease duration of >1 yr, seen at the Hospital for Sick Children, Toronto, between 1995 -2012 was performed. Clinical and biochemical variables were correlated with changes in eGFR (Schwartz formula) and to the presence or absence of nephrocalcinosis (as detected on ultrasound). Time-weighted average (Avgtw) serum measurements were calculated for all biochemical variables.

Results: Data of 19 patients (11 males) with permanent hypoparathyroidism, mean duration of follow up of 8.9±4 yrs and mean age at the end of the observation of 13.3±5.1 yr were analyzed. The median dose of calcitriol was 0.015(0.003-0.044) mcg/kg/day and calcium supplements 13.9(1.87-68)mg/kg/day. The Avgtw total and ionized serum calcium (Ca) were 2.22±0.15 and 1.14±0.11 mmol/L respectively. The average urinary Ca/creatinine ratio was 0.49±0.4 and eGFR was 132.4±30.1 ml/min/1.73m2. Nephrocalcinosis was observed in 33.3% of the subjects and more frequent in patients with hypocalcemic ($p=0.04$) and hypercalcemic ($p=0.01$) episodes. A significant negative correlation was observed between eGFR and the Avgtw total Ca ($r= -0.53$, $p=0.02$), ionized Ca ($r= -0.52$, $p=0.022$) and urinary Ca/creatinine ratio($r= -0.46$, $p=0.04$). There was no association between the presence of nephrocalcinosis and eGFR.

Conclusions: The potential impact of the treatment of hypoparathyroidism on renal function begins in childhood. Even within the normal range of serum Ca, higher concentrations are associated with lower eGFR. As hypoparathyroidism is most commonly a life-long condition, careful monitoring and management of calcium abnormalities has important future implications.

Oral Abstract 7
A familial case of congenital thyroid ectopy

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Congenital hypothyroidism due to thyroid dysgenesis (CHTD) is a relatively common disorder with a prevalence of one in 4,000 live births. The most common diagnostic category is thyroid ectopy, which occurs in up to 80% of CHTD cases. CHTD is predominantly not inherited, but up to 2% cases are familial, which is 15 times more than expected by chance alone. CHTD also has a high discordance rate (92%) between monozygotic twins, and a female and ethnic (Caucasian) predominance. Germline mutations in the thyroid-related transcription factors NKX2.1, NKX2.5, FOXE1, and PAX-8 have been identified by candidate gene screening in only 3% of patients with sporadic CHTD. Linkage analysis has excluded these genes in rare multiplex families with CHTD. Moreover, evidence of non-penetrance of mutations in genes such as *NKX2.5* in close relatives of patients suggests that modifiers, possibly additional germline mutations and/or somatic mutations, are associated with CHTD. We report the case of a mother and daughter both with thyroid ectopy where whole-exome sequencing revealed transmission of rare genetic variants from both parents to the daughter as well as the occurrence of rare variants in genes involved in thyroid and parathyroid development (e.g. NKX2.1, NKX2.5 and TBX1) in both affected individuals. These results are currently being validated. In conclusion, the literature supports the existence of a familial component of CHTD involving dominant genetic predisposition factors with low penetrance. This case report emphasizes the polygenic basis of CHTD and the possible involvement of other unknown genes in the pathogenesis of the disease, which may also follow non-Mendelian pattern of inheritance. The unique combination of these genes in each individual could explain the variable penetrance of mutations of these transcription factors.This case provides a good model of a multigenic mechanism not only of CHTD, but perhaps of other congenital anomalies.

Oral Abstract 8
Hypertension and hematuria: It's not the kidney!

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Introduction: Paraganglioma (PGL) tumors, or extra-adrenal pheochromocytomas (PCC), are a rare entity in childhood, and account for up to 30% of childhood pheochromocytomas. Common gene mutations in hereditary PGL/PCCs include *VHL, RET, NF1* and *SDH*, which are important for predicting prognostic significance, and malignant potential. We present a case of a 12 year old male, with a functional bladder paraganglioma.

Clinical Report: Our patient was a previously well individual, who presented with a one week history of increased thirst, gross hematuria, nausea, anorexia and epigastric pain, but no confirmed weight loss. He had a preceding history of brief paroxysmal, bilateral frontal headaches for one year. Family history was non-contributory. Physical examination was remarkable for hypertension (150/87 mmHg), tachycardia (127 bpm), and a systolic flow murmur on cardiac exam. Bladder ultrasound showed an 8.1 x 7.5 x 6.0 cm mass, arising from the posterior bladder wall. Transurethral cystoscopy with tissue biopsy confirmed a diagnosis of bladder PGL. Bone marrow aspirate and biopsy were negative for metastatic disease. Whole body non-contrast 18-fluoro-deoxyglucose PET scan showed a large bladder mass, with no evidence of regional or distant metastasis. Genetic testing confirmed a succinate dehydrogenase B gene (*SDHB*) mutation. Parental *SDHB* mutation analysis is pending. Preoperative labs showed: normal TSH, free T4 and PTH; elevated 24 hour urine metanephrine/creatinine 0.252 mmol/mol (0.026-0.176), normetanephrine/creatinine 7.129 mmol/mol (0.059-0.254), and chromogranin A 190 U/L (<40). Pre-operative management consisted of prazosin and intravenous/oral fluids. One month later, he underwent open laparotomy, partial cystectomy, and right-sided ureteric stent insertion. Hypertension resolved immediately. Six week post-operative 24 hour urine studies showed: metanephrine 1.19 umol/d (0.20-1.23), normetanephrine 2.74 umol/d (0.29-1.59), chromogranin A 7.8 U/L (<40).

Conclusion: Even though bladder PGL tumors are exceedingly rare in the pediatric age group, this case highlights the variable presentation of PGL tumors. Genetic analysis is crucial for identifying certain mutations, such as *SDHB*, to predict malignancy risk, genetic counseling and long-term surveillance.

Oral Abstract 9
Development and Evaluation of Two Online Case-based Modules to Teach Medical Residents Core Pediatric Endocrinology Concepts

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On-line case based modules are an effective method used to teach medical learners (Brudo *et al*, 2002.). They have been shown to increase knowledge and skills, and have improved the perceived educational experience (Singh *et al*, 2011). Diabetes specific studies have shown online learning to be superior to traditional face-to-face instruction (Wiecha *et al*, 2006. Cook *et al*, 2009). However this is a paucity of data regarding the use of online modules to teach other endocrinology concepts.

Methods: Two online case-based modules on pediatric short stature were developed in conjunction with the HSIMS Department of our institution. Clinical concepts taught in the modules are as per the RCPSC Pediatric Residency Training Objectives. Modules were formatted in concordance with both resident learning preferences, as determined by a focus group on online learning, and methods known to enhance knowledge retention. Evaluation of resident learning from the modules was assessed using pre- and post-tests. A learning satisfaction survey, which gauged resident satisfaction and perceived learning, was completed by the participating residents. Main outcome measure was the difference in pre- and post-test scores. Subgroup analysis was performed, breaking down participants into residency level and endocrinology experience.

Results: Ten residents successfully completed the online modules, pre-test, post-test, and learning satisfaction survey. The overall average scores on the pre- and post-tests were 49.5% and 78.4% respectively (p value 0.001). There was a high level of learner satisfaction and perceived learning.

Conclusion: Online case-based modules are an effective method to teach core endocrinology concepts to medical trainees.

Oral Abstract 10
Shining some light on the powerhouse of the cell- Is there a link between vitamin D and mitochondrial function in humans?

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Objective: Suboptimal skeletal tissue mitochondrial function has been implicated in several disorders where fatigue is a prominent feature. Vitamin D deficiency is a well-recognised cause of fatigue and myopathy. The aim of this study was to examine the effects of cholecalciferol therapy on skeletal mitochondrial oxidative function in symptomatic, vitamin D deficient individuals.

Design: This longitudinal study assessed mitochondrial oxidative phosphorylation in the gastro-soleus compartment using Phosphorus-31 magnetic resonance spectroscopy measurements of phosphocreatine recovery kinetics in 12 symptomatic, severely vitamin D deficient subjects before and after treatment with cholecalciferol (10-12 weeks later). All subjects had serum assays before and after cholecalciferol therapy to document serum 25OHD and bone profiles. 15 healthy controls also underwent ^{31}P-MRS and serum 25OHD assessment.

Results: The phosphocreatine recovery half-time ($\tau 1/2$PCr, $\tau 1/2$ADP) was significantly reduced following cholecalciferol therapy in the subjects indicating an improvement in maximal oxidative phosphorylation ($p<0.001$, $p=0.003$). This was associated with an improvement in mean serum 25OHD levels (8.8±4.2nmol/L to 113.8±51.5nmol/L, $p<0.001$). There was no difference in phosphate metabolites at rest. A linear regression model showed that decreasing serum 25OHD levels are associated with increasing $\tau 1/2$PCr ($r=-0.41$, $p=0.009$). All patients reported an improvement in fatigue following cholecalciferol therapy.

Conclusions: Cholecalciferol therapy augments muscle mitochondrial maximal oxidative phosphorylation following exercise in symptomatic, vitamin D deficient individuals. This finding suggests that changes in mitochondrial oxidative phosphorylation in skeletal muscle could at least be partly responsible for the fatigue experienced by these patients. For the first time, we demonstrate a link between vitamin D and the mitochondria in human skeletal muscle.

Oral Abstract 11

Prevalence of Non-Alcoholic Fatty Liver Disease in Adolescents with Polycystic Ovary Syndrome

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Background: Polycystic ovary syndrome (PCOS) is a condition characterized by hyperandrogenism and oligo-anovulatory cycles. Some adult studies suggest that PCOS may be an independent risk factor for non-alcoholic fatty liver disease (NAFLD). To date there are little data on the prevalence of NAFLD in adolescents with PCOS. Because NAFLD has potentially serous complications such as progressing to cirrhosis, we aimed to review how often liver enzymes were being requested in patients with PCOS as well as estimate the frequency in which hepatic transaminases were elevated in these patients, as a marker of NAFLD.

Methods: A retrospective chart review of adolescents with PCOS was conducted. The diagnosis of PCOS was based on the Rotterdam consensus criteria. Anthropometric data and laboratory data (liver enzymes, lipid profile, glucose and insulin) were abstracted. BMI z-scores were calculated and insulin resistance was calculated using the homeostasis model assessment (HOMA-IR). Patients were considered to be obese if their BMI z-score was >2. The prevalence of NAFLD, estimated here by alanine aminotransferase (ALT) levels, was examined at three different cut-off values of ALT.

Results: 140 patients (mean age 15.96 years) with PCOS were identified. Of these, 79 subjects (51.9%) had liver enzymes measured and were included in our study. Mean BMI z-score was 1.53. Using 3 different cut-off reference values for ALT, the prevalence of NAFLD was determined. At an ALT≥22 U/L the prevalence was estimated to be 72.2%. Of these, 23 (40.4%) were obese and 34 (59.6%) were non-obese. Twenty (35.0%) had elevated HOMA-IR. There were 18 (31.6%) who were considered neither obese nor had elevated HOMA-IR. At an ALT≥37 U/L, the prevalence of NAFLD was 26.7%, and at an ALT≥45 U/L, the prevalence was 10.1%.

Conclusion: Our data suggest that a significant proportion of adolescent girls with PCOS have abnormal liver enzymes suggestive of hepatic steatosis. Furthermore, this appears to be present in girls with PCOS independent of their BMI. As this is a retrospective study, corroborating data such as imaging were not available. However, our findings suggest the need for further study of the prevalence of NAFLD in adolescents with PCOS.

Oral Abstract 12

RASopathies: A Known Cause of Delayed Puberty; a Cause of Precocious Puberty Too?

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Introduction: RASopathies are a group of developmental disorders that are caused by activating germline mutations in genes that encode regulators of the RAS/Mitogen-Activated Protein Kinase (MAPK) pathway. The RAS-MAPK pathway regulates cell differentiation, proliferation and apoptosis. RASopathies include Noonan syndrome (NS), Costello syndrome (CS), Cardio-Facio-Cutaneous syndrome (CFC) and LEOPARD syndrome. These syndromes share characteristics such as craniofacial dysmorphology, cardiac malformations, cutaneous/musculoskeletal/ocular abnormalities and neurocognitive impairment. RASopathies have been associated with short stature and, in case of NS, delayed puberty. Less appreciated is that these disorders can also be a cause of precocious puberty (PP). We report 3 cases of PP associated with CFC and CS syndromes.

Cases:
Patient 1: Male; CFC (*BRAF,* D638E); Age at onset of puberty: 4 years; Tanner P2G3 TV 8-10 ml; Post-stimulation LH 38.6 IU/l; Bone age - chronological age: 1.6 yrs; Started Lupron 1 mo after presentation; MRI: normal pituitary, nonspecific slight thinning of corpus callosum.
Patient 2: Female; CFC (*MEK1,* c.389 A>G); Age at onset of puberty: 7 years; Tanner P1B2; Basal LH 1.5 IU/l; Bone age - chronological age: 2.5 yrs; Currently monitored for pubertal progression; MRI: absence of pituitary bright spot, nonspecific slight thinning of corpus callosum.
Patient 3: Male; CS (*HRAS*, G12S); Age at onset of puberty: 7 years 9 months; Tanner P1G2 TV 6ml; Basal LH 1.3 IU/l; Bone age - chronological age: 2 yrs; Started Lupron 9 mos after presentation due to rapid progression (TV 12-15 ml 9 mos after presentation); MRI: normal pituitary.

Discussion: Proof that mutations in the RAS-MAPK pathway can cause central PP will require further studies, but our findings highlight the intriguing possibility that mutations in some genes in this pathway can lead to delayed puberty while mutations in other genes can lead to PP. Understanding the molecular basis of delayed and precocious puberty in RASopathies would inform our understanding of factors that regulate the onset of puberty.

Poster Abstracts

Poster Abstract 1
Endocrine manifestations of hepatoblastoma

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Although hepatoblastoma is the most common liver tumor in children it is relatively rare. Two recent patients highlight associated endocrine manifestations. The first, a 4y2mo old boy presented with rapidly progressive isosexual precocity over a 3-month period. Pubic hair was Tanner stage 3 and testes were 10 mL bilaterally. Labs revealed LH <0.1 IU/L, FSH <0.1 IU/L, serum testosterone 23.2 nmol/L (<0.9), DHEAS 1.7 umol/L (0.9-7.5), 17-OHP 5.9 nmol/L (0.0-0.8), androstenedione 1.7 nmol/L (0.1-0.6), βhCG 29 IU/L (<2) and AFP 164,500 mcg/L (1-4). Bone age was 8-9 years. Abdominal imaging revealed a large mass arising from left lobe of liver. Biopsy demonstrated hepatoblastoma (epithelial-type), which stained weakly for β-hCG. GnRH-independent precocity resulting from hepatoblastoma is rare, with only 20 cases reported, and is thought to result from HCG stimulation of Leydig cells. The weakly positive stain for βhCG suggests a possible alternate pathogenesis such as testosterone-producing tumor cells as reported in a previous case. The degree of testicular enlargement and elevated 17OHP in our patient are unexplained. The second boy was diagnosed with hepatoblastoma and Beckwith-Wiedemann syndrome at age 3 months. After 3 months of chemotherapy with cisplatin and doxorubicin he underwent liver transplantation. Pre-transplant imaging revealed osteopenia and compression fractures of the lower thoracic and lumbar vertebrae but no bony metastases. Bone biochemistry was normal. Pathology revealed an 1183 g mixed epithelial-mesenchymal tumor with osteoid accounting for 65% of tumor volume. Six months post transplant, bone mineralization and vertebral body height improved. Small series have reported osteopenia in absence of bone metastases in hepatoblastoma and have suggested a paraneoplastic syndrome although the mechanism has not been delineated. These two cases illustrate endocrine manifestations of a rare pediatric malignancy.

Poster Abstract 2

A tale of two boys: An Atypical Cause of Pubertal Precocity

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Testicular tumours are rare in paediatric patients, accounting for only 1% of tumours in boys. Germ-cell tumours account for 65-70% of these. Leydig cell tumours are the most common gonadal stromal tumours. Limited reports of pediatric Leydig cell tumors exist in the literature. These tumors usually present as unilateral painless testicular masses that are functionally active, secreting testosterone, and characteristically result in isosexual precocious puberty. We report the following two cases which presented within one week of each other at our institution. A 5 year 7 month old boy presented to General Endocrine clinic with a nine-month history of behavioural changes, frustration, sleep disturbance and a growth spurt. The presence of pubic hair and penile enlargement was noted more recently. His examination was unremarkable apart from penile enlargement and tumescence, scant coarse scrotal hairs and a right-sided testicular mass. Testosterone was elevated at 5.6 nmol/L and 1.1 nmol/L, with undetectable LH and FSH measuring 0.4 IU/L. Tumor markers (AFP, bHCG, LDH) were not elevated. Bone age corresponded to that of an 8 year-old boy. Ultrasound revealed an isolated 7mm hypoechoic mass in the right testicle. A second 5 year old boy presented to clinic with one-year history of pubic and facial hair, acne, growth spurt and penile enlargement. On examination, Pubic hair corresponded to Tanner stage 3 and his testicular volumes were 5 mL (left) and 6 mL (right). Laboratory investigations revealed testosterone level of 7.9 nmol/L, LH 0.5 IU/L, and FSH was undetectable. Tumor markers were not elevated. Bone age corresponded to that of a 13 year old boy. Ultrasound evaluation showed multiple bilateral hypoechoic small nodules. Both boys underwent partial (testicle sparing) orchidectomy and pathology was consistent with Leydig cell tumours. Both patients patients were offered germline testing for DICER1 mutations, which has been associated with leydig cell tumours, among other malignancies. Leydig cell tumors may present at a very young age with hormonal manifestations and may not always present as a unilateral testicular mass but occasionally may be multifocal and bilateral at diagnosis. They should be considered on the differential of boys presenting in early puberty.

Poster Abstract 3

Organizing the Quebec Interdisciplinary Pediatric Gender Variance Program: From birth through late adolescence

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Public awareness of gender variance/dysphoria is increasing dramatically in North America. However, delay in initiation of pubertal blockade from failure to seek early medical care, support, and education may result in poorer psychosocial and physiologic outcomes.

Objective: To organize the only longitudinal interdisciplinary pediatric gender variance program in Quebec to provide optimal whole-person care for families of youth experiencing gender variance.

Technique & Results: Using the foundation of a pre-existing developmental clinic to follow children with gender variance, a more comprehensive interdisciplinary program was created with a developmental-behavioural pediatrician, a pediatric endocrinologist, a child psychiatrist, an adolescent medicine specialist and educational psychologists to care for youth with gender variance. This clinical population extends beyond gender dysphoria also to families and children with disorders of sexual differentiation (DSD) where the choice of sex rearing is a challenge. Approximately 150 patients,1/3 pre-pubertal and 2/3 post Tanner Stage 2, are followed in the program. The longitudinal follow- up of pre-school and school aged children into Tanner 2 has enabled us to develop rapport with the families, minimize the discomfort of Tanner staging the genitalia, and to determine if intervention with hormonal blockade or any surgical technique may be indicated (either pre-pubertally or later). When such a recommendation is made, regardless of other specialists or clinicians who may encounter the family (particularly in the case of DSD), we find that this interdisciplinary approach helps to ensure that families and youth have received the appropriate preparation, education and counseling about the timely initiation of hormone blockade, and that they are followed closely by the members of our team.

Conclusion:The model of offering interdisciplinary care to families and youth experiencing gender variance, including those with more clearly-defined gender dysphoria and DSD, allows for longitudinal support, education and timely intervention for optimal mental and physical outcomes. Thus, by making use of the Gender Variance Program, families of gender variant youth, from birth through adolescence, are universally able to access these services for whole-person care in Montreal, Quebec, Canada

Poster Abstract 4

The Liver That Ate Thyroxine

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Background: Hepatic hemangiomas are benign tumours that commonly present in infancy and can be associated with hypothyroidism. We present a case of hepatic hemangioendothelioma causing consumptive hypothyroidism.

Case presentation: A 1 month old infant presented to medical attention with lethargy, prolonged jaundice, and hepatomegaly. Pregnancy was uneventful and family history was non contributory. On exam the baby was pale and fatigued but roused easily. Height was 52.5 cm (3rd %), weight 4.5 kg (10-25%), head circumference 37 cm (3-10%). No goiter was detected. Cardiac exam revealed a 2/6 systolic ejection murmur. There was no respiratory distress. Abdomen was markedly distended with a liver edge palpable just above the pelvic brim. There was a reducible umbilical hernia.

Results: Thyroid Profile: TSH 352 mU/L (1-10 mU/L), FT3 1.8 pmol/L (2.4-9.8 pmol/L), FT4 of 4.3 pmol/L (9-21 pmol/L), rT3 511 ng/dL (8.1-52.8 ng/dL). Other investigations: AST 50 U/L (<60), ALT 12 U/L (<50), ALP 220 U/L (110-300), GGT 225 U/L (<55), Total Bilirubin 90 umol/L (<20) , AFP 5470 ug/L (<9), Random Blood Glucose 4.5 mmol/L, Hb 59 g/L (90-140). The newborn metabolic screen was normal. MRI of the abdomen revealed multifocal extensive infiltration of the liver consistent with the diagnosis of a hemangioendothelioma.

Conclusions: The hemangioendothelioma was treated with prednisone and propranolol. The patient initially required high doses of L-Thyroxine to treat the consumptive hypothyroidism (up to 25 µg/kg/d). With involution of the tumor, repeat thyroid profile showed a TSH 1.54 with elevated FT4 of 54.3 pmol/L and FT3 of 6.5 pmol/L. Subsequently, thyroid hormone replacement dose was tapered to 5 ug/kg/d.The mechanism of consumptive hypothyroidism due to Hepatic hemangiomas and the challenges of the management of these cases will be discussed.

Poster Abstract 5

A comparison of quality of sleep and quality of life in patients with glycogen storage disease on standard and modified uncooked cornstarch

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Background: Uncooked cornstarch (UCCS) must be taken at regular intervals throughout the day and overnight to prevent hypoglycemia in patients with glycogen storage diseases (GSDs). *Glycosade*^{TM}, a new modified UCCS, has recently been shown to be an effective alternative to standard UCCS allowing a longer fasting period for patients with GSDs. Our objective is to assess the impact of this modified UCCS on quality and quantity of sleep and quality of life of children and adults with GSDs and their primary caregivers.

Methods: All patients (ages >2 years) followed in 3 centres (Montreal Children's Hospital, Hôpital St-Luc and Montreal General Hospital) were invited to take part in the study. Duration of fast without hypoglycemia, safety and metabolic control on *Glycosade*^{TM} were assessed during a 24-hour hospitalization. Glucose fluctuations were monitored using a continuous glucose monitoring device. Quality of sleep and quality of life (QoL) were examined using validated questionnaires, sleep diary and actigraphy before and 1 month after implementing this modified UCCS for both patients with GSDs and their primary caregivers. For the before-after comparison, continuous variables will be analyzed using paired t-tests to compare means or non-parametric equivalent depending on the distribution. Categorical variables will be analyzed using McNemar test. We expect to recruit 12 to 15 patients within the next year.

Results: Recruitment began in October 2013.

Conclusions: We anticipate that quality and quantity of sleep and QoL of patients and caregivers will be improved with this new therapy. *Glycosade*^{TM} is currently not covered by the Quebec Public Prescription Insurance Plan (RAMQ). This study may prove beneficial to government agencies in supporting application for reimbursement.

Poster Abstract 6

Mind over muscle: Exploring the biology of fatigue in growth hormone deficiency

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Context: Growth hormone deficient (GHD) individuals may experience fatigue which resolves with GH replacement. The precise basis of this remains unclear. Fatigue can be classified as central or peripheral. Suboptimal skeletal mitochondrial function has been implicated in several conditions where peripheral fatigue is a prominent symptom using Phosphorus-31 Magnetic resonance Spectroscopy (31P-MRS) which can measure maximal mitochondrial oxidative phosphorylation, an important parameter of mitochondrial function. We have adapted this technique to enable non-invasive measurement of muscle mitochondrial oxidative phosphorylation *in vivo* during dynamic muscle activity.

Objective: To identify the aetiology of fatigue in GHD by characterising and comparing *in vivo* skeletal muscle metabolism in age, gender and physical activity matched untreated GHD adults, treated GHD adults and healthy volunteers. We also compared the perception of fatigue with QoI-AGHDA across the 3 groups.

Design: Twenty two untreated GHD adults, 23 treated GHD adults and 20 healthy volunteers were recruited at a tertiary University centre. All patients underwent assessment of muscle mitochondrial function ($\tau 1/2$ PCr) using ^{31}P-MRS.Fasting biochemical analyses and anthropometric measurements were obtained. All patients completed questionnaires on Quality of life (QoL-AGHDA) and physical activity assessment (IPAQ).

Results: There was no difference in maximal mitochondrial function ($p=0.53$) and proton handling ($p=0.30$) of skeletal muscle between untreated GHD, treated GHD and healthy volunteers. There was no association between $\tau 1/2$ PCr and serum IGF-1 ($r=-0.13$, $p=0.32$). Untreated GHD adults complained of significantly increased fatigue and impaired QoL when compared to treated GHD adults and health. Untreated GHD patients had significantly lower IGF-1 than both treated GHD and healthy volunteers ($p<0.001$).

Conclusions: Whilst untreated GHD adults experience more fatigue when compared to treated GHD adults and normal volunteers, they do not demonstrate persistent abnormalities in peripheral skeletal tissue metabolism i.e. maximal mitochondrial oxidative function, anaerobic glycolysis nor proton clearance as assessed by 31P-MRS. This suggests a likely central component in the pathophysiology of fatigue in GH deficiency.

Poster Abstract 7

Is more always better? Deterioration of skeletal muscle function in a boy with Duchenne muscular dystrophy associated with the onset of precocious puberty

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Background: There is evidence from randomized controlled trials that glucocorticoid (GC) therapy in Duchenne muscular dystrophy improves muscle strength and function in the short-term. However, chronic GC therapy often leads to delayed puberty. Treatment of delayed puberty comprises testosterone replacement therapy which may have a significant impact on body image and bone health.

Case: We present a 7yr boy with Duchenne muscular dystrophy who presented with precocious puberty and discuss the investigational approaches to precocious puberty and the underlying diagnosis. Investigations revealed he had precocious puberty secondary to a germinoma (serum beta HcG-200 IU/L).
Importantly, a significant deterioration in skeletal muscle function was observed which coincided with the onset of puberty.

Discussion: Although the general consensus is that testosterone promotes muscle strength and bone health, could puberty and androgens have a more complex interaction with dystrophic skeletal muscle in the context of Duchenne muscular dystrophy than previously thought? Mammalian models demonstrate that height is inversely proportional to loss of muscle fibers when the dystrophin gene is absent. We discuss whether puberty and its effects may have a negative impact on skeletal muscle function in patients with DMD.

Poster Abstract 8

McCune-Albright Syndrome Presenting in Infancy

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We describe the presenting features and early clinical course of two infants, one male and one female, diagnosed in our centre with McCune-Albright Syndrome (MAS) within the first two months of life. Both children presented with nonspecific findings of failure to thrive and tachycardia, with a history of low birth weight and mild hypotonia. The diagnosis of hyperthyroidism was made at the age of five weeks (female), and 10 weeks (male). Cafe-au-lait patches were noted in both patients, prompting suspicion of MAS and investigations for other endocrinopathies. While Cushing syndrome is thought to be a less frequent feature of MAS (prevalence of 7.1% in one case series), it was diagnosed in both of our patients in early infancy. One patient has responded well to ketoconazole, while adrenal suppression has been more challenging in the other. Precocious puberty is suspected in our female patient (small breast buds, leukorrhea), while in our male patient, an early finding of hypercalcemia has raised the question of possible hyperparathyroidism. Numerous challenges exist in the management of infants with MAS. We review the existing literature on the endocrine features of MAS in infancy and discuss safety issues that may arise during the management of these patients, including the risk of opportunistic infections during the treatment of Cushing syndrome, liver dysfunction, and the need for corticosteroid stress dosing once the adrenal gland is pharmacologically suppressed.

Poster Abstract 9

When Hormones Paralyze: a Rare Case of Thyrotoxic Periodic Paralysis

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A 16-year 9-month old previously healthy Asian boy came to the Emergency Department with sudden paralysis of lower extremities. This paralysis was preceded by weakness in the middle of the night whereby the patient had to be carried to the bathroom as he was unable to walk.

On physical exam, he was alert and conscious, had heart rate of 137 beats/min, blood pressure 161/74mmHg and he had significant lower extremity weakness.

Preliminary investigations revealed a normal MRI of the spine, potassium of 1.8 mmol/L and electrocardiogram suggestive of possible right bundle branch block. Potassium supplementation was started and the paralysis quickly improved. Further investigations and close examination revealed the diagnosis of thyrotoxic periodic paralysis (TPP). The patient was found to have a goiter with TSH of 0.1 mU/L, free T4 of 55.6 pmol/L and TSH receptor antibody of 28 IU/L. Molecular genetic analysis of skeletal muscle calcium and sodium channel genes did not detect any mutations. Funding for molecular genetic analysis of the skeletal muscle potassium Kir2.6 channel has been requested.

The patient was started on propranolol and methimazole and discharged within two days. A few weeks after presentation, the patient had another episode of lower extremity paralysis. Thyroidectomy was chosen as a curative treatment and performed 5 months after diagnosis. Surgery went well, he is currently managed on replacement and has had no further episodes of paralysis.

TPP, now regarded as an endocrine channelopathy, is rare in the pediatric population. This complication of hyperthyroidism is common in Asian men and is increasingly seen in Western countries. The clinical presentation, pathophysiology and management of this condition will be reviewed.

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