conference syllabus



Canadian Pediatric Endocrine Group

Groupe canadien d'endocrinologie pédiatrique

2012 Scientific Meeting



The Fort Garry Hotel, Spa and Conference Centre

February 9 - 11, 2012

In cooperation with:



Welcome

Dear Delegates,

I would like to welcome you to the 6th 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 Winnipeg, 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,

l'aimerais vous souhaiter la bienvenue à la 6ème 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 autres et 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 Winnipeg, 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!

Mark R. Palmert, PhD, MD

President

CPEG 2012 Scientific Meeting

The Canadian Pediatric Endocrine Group acknowledges and thanks the following sponsors for their generous support in the form of an unrestricted educational grant:

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Fellowship ListingThe 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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1992 – 1993	Eli Lilly I: M. Lawson	
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1994 – 1995	Eli Lilly I: S. Taback	Eli Lilly II: A. Simone
	Novo Nordisk: S. Muirhead (Lawrence)	,
1995 – 1996	Eli Lilly I: S. Taback	Eli Lilly II: B. Cummings
	Novo Nordisk: C. Vaz (50% - One Year	
1996 – 1997	Eli Lilly I: J. Hamilton, E. Sellers	Eli Lilly II: B. Cummings
1997 – 1998	Eli Lilly I: J. Hamilton	Eli Lilly II: E. Sellers
1777 - 1770	Serono: B. Cummings	Lii Liiiy II. L. Jellers
1998 – 1999	Eli Lilly I: J. Curtis	
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1999 – 2000	Eli Lilly II: J. Curtis	
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2000 – 2001	Eli Lilly I: C. Panagiotopoulos	
	Serono: C. Huang	
2001 – 2002	Eli Lilly II: C. Panagiotopoulos	
	Hoffmann La Roche: S. Stock	
2002 – 2003	Eli Lilly I: P. Krishnamoorthy	
	Hoffmann La Roche: R. McEachern	
	Serono: P. Zimakas	
2003 – 2004	Eli Lilly II: P. Krishnamoorthy	
	Hoffman La Roche: H. Bui	
2004 – 2005	Eli Lilly I: M. Nakhla	
	Hoffmann La Roche: J. Simoneau-Roy	
2005 - 2006	Eli Lilly II: M. Nakhla	
	Hoffmann La Roche: M. Jetha	
	Serono: I. Chapados	
2006 – 2007	Eli Lilly I: BA Wicklow	
	Hoffmann La Roche: S. Amed	
2007 - 2008	Eli Lilly II: BA Wicklow	
	Hoffmann La Roche: J. Deladoey	
	Serono: B. Babic, T. Pinto	
2008 – 2009	Eli Lilly I: P. Olivier	
	Hoffmann La Roche: T. Pinto	
	Novo Nordisk: AM Sbrocchi	
2009 – 2010	Eli Lilly II: P. Olivier	
	Hoffmann La Roche: S. Runge-Wildi	
	Novo Nordisk: R. Shulman	
	Sandoz: C. Saaman	
	Serono: T. Édouard	
2010 – 2011	Eli Lilly I: J. Wasserman	
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	Novo Nordisk: E. Bassilious	
	Sandoz: S. Tsai	
2011 – 2012		
2011 - 2012	Eli Lilly II: J. Wasserman Sandoz: M. Cohen	
	Hoffman LaRoche: C. Zuijdwik	
	Nova Nordisk: M. Millette	
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Thursday, February 9, 2012

Fellows Symposium (Salon A)
Part I: Meet the Professor

Be Prepared: Transitioning from Fellow to Faculty

Dr. Heather Dean

Part II: Associate Members' Business Meeting

16:00 Registration Open (Foyer)

17:00 Welcome Reception & Exhibits Open (Crystal Ballroom)

19:00 Adjourn

19:00 Satellite Symposium

Friday, February 10, 2012

07:00 Registration Open (Foyer)

07:15 Breakfast & Exhibits (Crystal Ballroom)

08:00 Opening Remarks & Welcome (Concert Hall)

Dr. Mark Palmert, Dr. Elizabeth Sellers, and Ms. Irena Hozjan, NP-Paeds

08:15 CPEG Fellowship Awards (Concert Hall)

Presented by Dr. Mark Palmert

Theme I: Gender Dysphoria (Concert Hall)

Moderator: Dr. Danièle Pacaud

08:30 Gender Identity, Sex Hormones and the Brain (Including Q&A)

Dr. Baudewijntje PC Kreukels, Amsterdam, Netherlands

09:15 Times and Genders: They are a Changin' (Including Q&A)

Dr. Gail Knudson, Vancouver, BC

09:45 The Endocrine Management of Transgender Youth (Including Q&A)

Dr. Daniel L Metzger, Vancouver, BC

10:05 Personal Experience Navigating the Medical System and Society (Including Q&A)

Sebashtien Drake, Winnipeg, MB

10:30 Refreshment Break & Exhibits (Crystal Ballroom)

Theme IIa: Bone (Concert Hall)

Moderator: Dr. Celia Rodd

11:00 FGF23: From Phosphate Diabetes to Pediatric Bone Diseases (Including Q&A)

Dr. Michael T Collins, Bethesda, MD

12:00 Lunch & Exhibits (Crystal Ballroom)

13:00 Posters (Conference Foyer) and Desserts (served in the Crystal Ballroom)

*13:30 Split Rooms (Nurses: please see Page 5 for the program)

Theme III: Type 2 Diabetes in the Youth (Concert Hall)

Moderator: Dr. Josephine Ho

13:30 Type 2 Diabetes in Youth - Scope and Natural History of Associated Complications

(Including Q&A)

Dr. Allison Dart, Winnipeg, MB

14:00 Fatty Liver Disease a Biomarker for Type 2 Diabetes in Overweight Youth: Studies Using 1H

Magnetic Resonance Spectroscopy (Including Q&A)

Dr. Jonathan McGavock, Winnipeg, MB

14:30 Global Evidence for the Role of Fetal Programming of Obesity and Type 2 Diabetes in

Childhood (Including Q&A)

Dr. Heather Dean, Winnipeg, MB

15:00 Refreshment Break & Exhibits (Crystal Ballroom)

Abstracts (Concert Hall)

Moderators: Dr. Patricia Gallego and Dr. Rebecca Perry

15:30 Abstracts (7) (Crystal Ballroom)

17:15 Adjourn

*Nursing Promote Moderator:	ogram for Friday, February 10 (in the Gateway Room) Ms. Nicole Kirouac
13:30	Introduction to Gender Awareness and Variance for the Healthcare Provider Dr. Reece Malone, DHS, MPH, ACS, Winnipeg, MB
14:30	International Endocrine Nursing Curriculum Development Ms. Irena Hozjan, NP-Paeds, Toronto, ON
14:50	Navigating International Meetings for CPEN Nurses CPEN Members
15:00	Refreshment Break & Exhibits
15:30	ESPE 2011: Canadian Nurses Abroad CPEN Members
16:00	CPEN Business Meeting
17:00	Adjourn

Friday Night Dinner at Fort Gibraltar (866 St. Joseph Street) in The Great Hall

(Bus transportation will be provided. Please see information in your delegate package.)

18:45 Board the bus outside The Fort Garry

19:00 Reception

19:30 Dinner

Saturday, February 11, 2012

07:15 Breakfast & Exhibits (Crystal Ballroom)

08:00 Business Meeting (Concert Hall)

10:00 Refreshment Break & Exhibits (Crystal Ballroom)

**10:30 Split Rooms (Nurses: please see below for the program)

Abstracts (Concert Hall)

Moderators: Dr. Patricia Gallego and Dr. Rebecca Perry

10:30 Abstracts (6)

12:00 Lunch & Exhibits (Crystal Ballroom)

12:30 Posters (Conference Foyer) and Desserts (Crystal Ballroom)

Theme IIb: Bone (Concert Hall)

Moderator: Dr. Elizabeth Cummings

13:00 Advances in Enzyme Replacement Therapy (ERT) for Hypophosphatasia, a Disorder of Skeletal

Mineralization (Including Q&A)

Dr. Cheryl Rockman-Greenberg, Winnipeg, MB

14:00 Refreshment Break & Exhibits (Crystal Ballroom)

14:30 Presentation of Dr. John Bailey Resident Research Award

Clinical Debate (Concert Hall)

Moderator: Dr. Tracy Bridger

14:40 Debate: Prenatal Treatment in CAH

Pro: Dr. Elizabeth Sellers, Winnipeg, MB; Con: Dr. Jean-Pierre Chanoine, Vancouver, BC

Moderator: Dr. Guy Van Vliet

15:35 Debate: Need for Screening and Oral Steroid Coverage for Adrenal

Insufficiency for Patients on Inhaled Steroids

Pro: Dr. Alexandra Amhet, Ottawa, ON; Con: Dr. Jonathan Dawrant, Calgary, AB

16:25 Closing Remarks & Evaluation

Presented by Dr. Mark Palmert

16:40 Adjourn

**Nursing Program for February 11 (in the Gateway Room)

Moderator: Ms. Irena Hozjan

10:30 Education on Bullying & Harassment of Youth: Impact and Intervention Strategies

Ms. Rebecca Ulrich, BHEcol, Winnipeg, MB

11:30 CPEN Initiatives Discussion

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

Fellow Abstract Schedule

Time	Title	Presenter	Oral Abstract Number	Page
Friday,	February 10			
15:30	Numeracy in adolescents with type I diabetes: assessment and interactive gaming intervention – A Pilot Study	Bassilious	1	17
15:45	Quality of Referral of Children with Idiopathic Short Stature to the Pediatric Endocrinologist	Chiniara	2	18
16:00	Two testes and 2 X chromosomes – Why?	Al Jaser	3	19
16:15	A Child with a Disorder of Sexual Development and Early- Onset Nephrotic Syndrome	Al- Alamy	4	20
16:30	Unstimulated luteinizing hormone levels are able to predict subsequent pubertal progression in patients presenting with signs of precocious puberty	Harrington	5	21
16:45	Health-Related Quality of Life (HRQOL) and Anxiety in Adolescents with Differentiated Thyroid Cancer (DTC)	Oren	6	22
17:00	Autonomic nervous system balance in craniopharyngioma related hypothalamic obesity	Cohen	7	23
Saturda	y, February II		•	•
10:30	Congenital Blindness and Severe Juvenile Osteoporosis - Two Novel Mutations Causing a Rare Syndrome	Hursh	8	24
10:45	Serum 25-Hydroxyvitamin D Test Utilization in Calgary	Nour	9	25
11:00	The Next Generation Cohort: Childhood Outcomes of Offspring of Parents with Early Onset Type 2 Diabetes	Millar	10	26
11:15	Is the frequency of ketoacidosis at onset of type I diabetes a child health indicator that is related to income inequality?	Shulman	11	27
11:30	Foot Abnormalities in children with Type 2 Diabetes	Kojori	12	28
11:45	Clinical Management of Youth with Gender Dysphoria at BC Children's Hospital: Over 10 Years' Experience	Khatchadourian	13	29

Poster Abstract Listing

Title	Presenter	Abstract Number	Page
A search for ectopic ACTH production in a 9 year old boy	Luca	I	30
Two cases of undervirilized XY infants with unexpected diagnoses	Ens	2	31
Borderline congenital hypothyroidism (CH): clinical characteristics and natural history	Oren	3	32
Evaluation of Deprivation and Glycemic Control in Pediatric Type I Diabetes from a Large Urban Canadian Centre	Zuijdwijk	4	33
Efficacy of Short Term 30/70 Insulin in Children with Type 2 Diabetes with A1C >9% at Diagnosis	Kawchuk	5	34
Acute localized inflammatory reaction to insulin administration	Winston	6	35
Occurrence of slipped capital femoral epiphysis in children undergoing leuprolide acetate depot therapy for the treatment of central precocious puberty	Inman	7	36
Quantifying adherence to growth hormone treatment: The Canadian easypodTM connect observational study (ECOS)	Van der Meulen	8	37

Program Organizing and Scientific Committee

Robert Barnes Danièle Pacaud Elizabeth Sellers
Dardye Eugène Mark Palmert Jonathan Wasserman
Andrea Hagg Wendy Schwarz Brandy Wicklow

Nicole Kirouac

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.

Credits

Canadian Pediatric Endocrine Group Annual Conference 2012 has been approved for a maximum of 10 credit hours under Section 1 of the Maintenance of Certification (MOC) program of the Royal College of Physicians and Surgeons of Canada.

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 I: Gender Dysphoria

Gender Identity, Sex Hormones and the Brain

Baudewijntje PC Kreukels, PhD, Senior Researcher, Department of Medical Psychology, Center of Expertise on Gender Dysphoria, VU University Medical Center, Amsterdam, Netherlands

This presentation is designed to help you:

- 1. Summarize evidence for effects of sex hormones on brain and behavior
- 2. Discuss problems and limitations of studies on gender identity and the effects of sex hormones on the brain in transsexuals
- 3. Describe current research on gender identity and the effects of sex hormones on the brain in the Center of Expertise on Gender Dysphoria at the VU University Medical Center in Amsterdam, the Netherlands

Times and Genders: They are a Changin'

Gail Knudson, MD, MPE, FRCPC, Medical Director, Transgender Health Program, Vancouver Coastal Health; Clinical Associate Professor, Department of Sexual Medicine, The University of British Columbia, Vancouver, BC

- I. Summarize the changing diagnostic criteria in DSMS and ICDII with respect to adolescents with gender dysphoria
- 2. Describe the societal shifts with respect to gender non-conforming children and adolescents
- 3. Describe the evolving diagnostic evaluation based on interactions of the change in diagnostic criteria and our culture's broadening acceptance of gender variance

The Endocrine Management of Transgender Youth

Daniel L Metzger, MD, FAAP, FRCPC, Pediatric Endocrinologist, Endocrinology and Diabetes Unit, BC Children's Hospital, Vancouver, BC

- 1. Understand the role of GnRH agonists in selected youth presenting with gender dysphoria
- 2. Develop an approach to instituting cross-hormone therapy in transgender youth

Theme IIa: Bone

FGF23: From Phosphate Diabetes to Pediatric Bone Diseases

Michael T Collins, MD, Chief, Skeletal Clinical Studies Unit, National Institutes of Health, Bethesda, MD

- 1. Know that FGF23 is made by bone cells
- 2. Know that FGF23 regulates blood phosphorus and 1,25 vitamin D levels
- 3. Know how to distinguish between genetic and acquired FGF23-mediated diseases

Theme III: Type 2 Diabetes in the Youth

Type 2 Diabetes in Youth - Scope and Natural History of Associated Complications Allison Dart, MD, MSc, FRCPC, Assistant Professor, Department of Pediatrics and Child Health, University of

Manitoba, Section of Nephrology, Winnipeg, MB

- I. Review microvascular and macrovascular complications of youth onset type 2 diabetes in childhood and adolescence
- 2. Understand the natural history of renal complications associated with youth onset type 2 diabetes
- 3. Analyze literature evaluating modifiable risk factors for the development of renal complications in youth onset type 2 diabetes

Fatty Liver Disease a Biomarker for Type 2 Diabetes in Overweight Youth: Studies Using 1H Magnetic Resonance Spectroscopy

Jonathan McGavock, PhD, Robert Wallace Cameron Chair in Evidence-Based Child Health; Assistant Professor, Department of Pediatrics and Child Health, Manitoba Institute of Child Health, University of Manitoba, Winnipeg, MB

The purpose of this talk will be to review emerging data from our group and others demonstrating that excessive hepatic triglyceride content (i.e. steatosis) is an early biomarker and potentially directly involved in the loss of glucose tolerance among overweight adolescents. The talk will be organized around the following questions related to this topic:

- I. How prevalent is hepatic steatosis in youth?
- 2. Is there evidence that hepatic steatosis is involved in the pathogenesis of type 2 diabetes?
- 3. What is the evidence that steatosis is involved in obesity-related type 2 diabetes in youth?
- 4. How can we prevent or treat hepatic steatosis in overweight youth?

Global Evidence for the Role of Fetal Programming of Obesity and Type 2 Diabetes in Childhood Heather Dean, MD, FRCP(C), Assistant Dean (Academic), Faculty of Medicine; Professor, Department of Pedatrics, University of Manitoba, Winnipeg, MB

- 1. Summarize the evidence for adaptive fetal programming for obesity and type 2 diabetes in childhood
- 2. List 3 strategies for breaking the cycle of obesity and type 2 diabetes in childhood
- 3. List research priorities in Canada related to the DOHaD theory

Theme IIb: Bone

Advances in Enzyme Replacement Therapy (ERT) for Hypophosphatasia, a Disorder of Skeletal Mineralization

Cheryl Rockman-Greenberg, MD, CH, FRCP(C), Professor and Head, Department of Pediatrics & Child Health, University of Manitoba, Winnipeg, MB

At the end of this talk the listener will understand:

- 1. Three different approaches to the treatment on Inborn Errors of Metabolism (IEM)
- 2. The history of enzyme replacement therapy (ERT) for the treatment of IEM
- 3. The clinical forms of hypophosphatasia and the early results of early ERT
- 4. The challenges ahead in translating research findings into routine clinical care

Debate: Prenatal Treatment in CAH

(Pro) Elizabeth Sellers, MD MSc, Associate Professor, Department of Pediatrics and Child Health, University of Manitoba, Winnipeg, MB

(Con) Jean-Pierre Chanoine, PhD, MD, Clinical Professor and Head, Endocrinology and Diabetes Unit, BC Children's Hospital, Vancouver, BC

Common debate learning objectives:

- I. Understand the mechanisms by which prenatal dexamethasone in the pregnant mother affects fetal adrenal function
- 2. Understand the effects of dexamethasone administration to the mother of the unborn child
- Discuss the risk benefit ratio of prenatal dexamethasone treatment with respect to the prevention of excess androgens in the unborn child

Debate: Need for Screening and Oral Steroid Coverage for Adrenal Insufficiency for Patients on Inhaled Steroids

(Pro) Alexandra Ahmet, MD, FRCPC, Pediatric Endocrinologist, Children's Hospital of Eastern Ontario; Assistant Professor of Pediatrics, University of Ottawa, Ottawa, ON

- I. Review the evidence behind adrenal suppression secondary to ICS
- 2. To debate the risks and management of children with potential AS from ICS

(Con) Jonathan Dawrant, MD, Pediatric Endocrinology and Diabetes Clinic, Alberta Children's Hospital, Calgary, AB

- 1. Review selected patterns of steroid prescribing by non-endocrinology specialties
- 2. Review safety concerns around inhaled corticosteroids
- 3. Review potential safe supervision practices around patient on inhaled corticosteroids

Nursing Program

Introduction to Gender Awareness and Variance for the Healthcare Provider

Reece Malone, DHS, MPH, ACS, Education Program Coordinator, Sexuality Educator, Rainbow Resource Centre, Winnipeg, MB

- 1. Discuss the differences between gender identity, gender expression and other aspects of sexuality
- Prepare and respond to questions regarding gender identity development, gender variance and gender diversity
- Identify strategies and ways which a clinical practice can be more affirming, respectful and safe for youth, adolescents and families who are exploring or present transgender and/or gender nonconforming

International Endocrine Nursing Curriculum Development

Irena Hozjan, RN(EC), MN, NP-Paeds Nurse Practitioner, Endocrine Program, Sick Kids Toronto, Toronto, ON

- I. Discuss objectives of an international curriculum development project
- Discuss opportunities and challenges involved in developing a Specialty Nursing Curriculum in a Middle Eastern country
- 3. Review relevant population and pediatric health statistics of a Middle Eastern country

Education on Bullying & Harassment of Youth: Impact and Intervention Strategies Rebecca Ulrich, BHEcol, RespectED Education Coordinator, Canadian Red Cross, Western Zone, Winnipeg, MB

- 1. Recognize the different behaviours that constitute each type of bullying and harassment
- 2. Analyze the impact of bullying and harassment on the target, the aggressor and the bystander
- 3. Utilize prevention and intervention strategies to support individual youth and families

Biographies

Dr. Alexandra Ahmet

Alexandra Ahmet is a Pediatric Endocrinologist and Assistant Professor at the Children's Hospital of Eastern Ontario in Ottawa. She graduated from McMaster Medical School in 2000, completed her pediatric residency and endocrinology fellowship at the Hospital for Sick Children in Toronto in 2005 and has been on faculty at CHEO since. Dr. Ahmet has a clinical, educational and research focus in diseases of the adrenal glands, specifically adrenal suppression. She is the program chair for an accredited educational program about adrenal suppression in children with Asthma that has educated specialists and primary care physicians about the risk of AS across Canada. In addition to lectures and workshops about AS. Dr. Ahmet has recently published guidelines for the management of AS in children with asthma in the Journal of Allergy, Asthma and Clinical Immunology and is the PI for local studies and co-PI for a national study aimed at supporting more evidence based guidelines about AS in the future.

Dr. Jean-Pierre Chanoine

Dr. Chanoine is a Pediatrician who graduated from the Free University of Brussels, Belgium in 1982. He joined the University of British Columbia in 1998 as Clinical Professor and Head of the Endocrinology and Diabetes Unit at British Columbia's Children's Hospital. Dr. Chanoine is Director of the subspecialty research component attached to the center for Healthy Weights Program, Shapedown BC, a clinical program addressing the needs of overweight children at British Columbia Children's Hospital. He is also director of the Pediatric component of the Canadian Obesity Network.

His previous experience includes a fellowship at the University of Massachusetts Medical Center in Worcester, USA; Pediatric Endocrinologist at Hôpital des Enfants Reine Fabiola in Brussels; and Medical Director for Novo Nordisk Belgium. He is presently President of the Canadian Pediatric Endocrine Group.

Dr. Chanoine's research includes basic and clinical research on the role of hormonal and nutritional factors in the development of childhood overweight. Dr. Chanoine is also principal

investigator of a novel family-based intervention for the treatment of obesity that links health with carbon foot print and personal finances in order to increase readiness to change.

Dr. Michael T Collins

Dr. Collins is the Chief of the Skeletal Clinical Studies Unit at the National Institutes of Health, Bethesda, Maryland. He did his endocrine training at the NIH in the Interinstitute Endocrine Training Program, and his Internal Medicine and Medical School training at the University of Maryland in Baltimore. Dr. Collins has been at the NIH since completing his Fellowship training. Areas of investigation include bone biology a mineral metabolism, which are studied through clinical and translational studies. Specific areas of interest include the role of PTH, G-proteins, and cAMP in bone cell biology, and FGF23 in mineral metabolism. The primary approach is the study and treatment of patients with rare disorders of bone and mineral metabolism as models through which to understand human bone and mineral biology and physiology. Current models of focus include fibrous dysplasia bone. hypoparathyroidism, and disorders of FGF23 excess and deficiency.

Dr. Allison Dart

Allison Dart completed her medical training at the University of Manitoba, and then her pediatric residency at the University of Western Ontario and University of Ottawa. She returned to Manitoba where she completed her fellowship in Nephrology, as well as a Masters of Science in the Department of Community Health Sciences. She is now an Assistant Professor in the Department of Pediatrics and Child Health at the University of Manitoba, in the section of Nephrology. She functions both as a clinician and researcher. Her primary research interest is in the study of renal complications associated with youth onset type 2 diabetes, and she is currently involved in the iCARE study, which is a cohort study with the goal of phenotyping youth with type 2 diabetes.

Dr. Jonathan Dawrant

Jonathan Dawrant is a Pediatric Endocrinologist at Alberta Children's Hospital and a Clinical Assistant Professor at the University of Calgary. He is a

graduate of Queen's Medical School. Postgraduate training took place in Pediatrics and, subsequently, Endocrinology at the University of Calgary. His interests are broad within Pediatric Endocrinology. He works part time for Alberta Health Services as a Lead Informatics Physician for Clinical Engagement, a role that focuses and developing and optimizing computer systems and informatics tools for clinical care.

Dr. Heather Dean

Dr. Dean graduated from Medicine at Queen's University in Kingston, Ontario and pursued pediatrics and pediatric endocrinology training in Montreal, Ottawa and Winnipeg. She has been a full time academic clinician in Winnipeg since 1983. She is Assistant Dean (academic), Faculty of Medicine, Professor of Pediatrics and faculty liaison for the Interprofessional Education Initiative at the University of Manitoba. Her major research activities focus on the epidemiology, care, education, support and prevention of type 2 diabetes children. Another area of research is innovative methods for systems navigation for young adults with type I and type 2 diabetes to prevent excess preventable morbidity and mortality.

Ms. Irena Hozjan

Irena Hozjan is a Pediatric Nurse Practitioner in the ambulatory Endocrinology Clinic at SickKids. She completed her Master of Nursing and Acute Care Nurse Practitioner Program at the University of Toronto and has been a member of the Endocrinology department for over 7 years.

Irena's current clinical practice includes providing specialty ambulatory health care to individuals and families dealing with various endocrine issues and conditions including Turner Syndrome, precocious puberty, short stature and growth hormone deficiency.

Irena is one of the contributing panel members for the RNAO's revised Clinical Best Practice Guideline Enhancing Healthy Adolescent Development published 2010. She is one of the editors, chapter authors and chapter co-authors of the Turner Syndrome: Across the Lifespan book published in 2008 with a French translation published in 2009.

Irena is currently President of the Canadian Pediatric Endocrinology Nurses (CPEN), and executive member of the Canadian Pediatric Endocrine Group (CPEG). She is also a member of the Pediatric Endocrine Nursing Society (PENS), RNAO and NPAO.

Dr. Gail Knudson

Gail Knudson MD, MPE, FRCPC, is a Clinical Associate Professor at the University of British Columbia Department of Sexual Medicine, Consultant Psychiatrist at the British Columbia Centre for Sexual Medicine at Vancouver Hospital, Medical Director of the Transgender Health Program at Vancouver Coastal Health and Faculty Development Leader for the Island Medical Program, Faculty of Medicine, University of British Columbia.

Dr. Knudson is active in conducting phase III clinical trials for treating both male and female sexual dysfunction. She served as co-chair of the DSM 5 Consensus Committee for the World Professional Association for Transgender Health (WPATH) and a writing group member of the new Standards of Care for Transgender Health (version 7). Currently, she co-chairs the ICD 11 Consensus Committee for WPATH.

Dr. Knudson is the Secretary-Treasurer of WPATH and founder and former President of the Canadian Professional Association for Transgender Health (CPATH). She is the Chair of the Sexuality Special Interest Group of the American Society for Reproductive Medicine (ASRM). She is Scientific Chair for the International Society for the Study of Women's Sexual Health (ISSWSH) Annual Meeting to be held in Jerusalem, Israel, February 2012.

Dr. Baudewijntje Kreukels

Baudewijntje Kreukels is a neuropsychologist and senior researcher in the VU University Medical Center at the Department of Medical Psychology and the Amsterdam Center of Expertise on Gender Dysphoria. Since the start in 2006, she is a coordinator of the ENIGI initiative, the European Network for the Investigation of Gender Incongruence. Her primary research interests are the effects of gonadal hormones on brain and behavior.

Dr. Reece Malone

Reece Malone is the Education Program Coordinator for the Rainbow Resource Centre specializing in the development and delivery of public awareness and training on: sexual orientation, gender identity, and comprehensive sexual health and sexuality education. Reece is a graduate from the University of Winnipeg with a degree in sociology, and was awarded a Masters of Public Health and a Doctorate of Human Sexuality from the Institute for Advanced Study of Human Sexuality in San Francisco, California. Reece also holds a private consultation and clinical sexology practice in Winnipeg.

Dr. Jonathan McGavock

Jon McGavock is a CIHR New Investigator at the Manitoba Institute of Child Health. He currently is the primary investigator for 6 studies aimed at understanding the role of physical activity in the prevention and management of type 2 diabetes in youth. He works closely with the staff in the pediatric endocrinology clinic at the Winnipeg Children's Hospital to develop research studies that are relevant to the clinical management of their pediatric population.

Dr. Daniel L Metzger

Working with BC Children Hospital nursing colleagues, as well as with child mental-health professionals and the adult transgender care teams in Vancouver, we have developed an endocrine treatment center (a "clinic without walls") for transgender youth (teens to youth adults) in Vancouver. We have provided services to some 75 transgender youth.

Dr. Cheryl Rockman-Greenberg

Dr. Cheryl R. Greenberg is Professor and Head of the Department of Pediatrics and Child Health, University of Manitoba and Medical Director, Child Health Program, Winnipeg Regional Health Authority. She has held these positions since April 2004. She is the Director of the Metabolic Service and a clinical geneticist in the Program in Genetics and Metabolism since 1992 and 1979 respectively and is a Professor in the Department of Pediatrics and Child Health and the Department of Biochemistry and Medical Genetics, University of Manitoba since 1994. Dr. Greenberg obtained her MDCM from McGill in 1974. She became a Fellow

of the Royal College of Physicians and Surgeons of Canada (Pediatrics) in 1979 and in Medical Genetics in 1996. She has been a Fellow of the Canadian College of Medical Geneticists since 1982. An academic clinician, she has focused her research on the identification of the molecular basis for specific genetic disorders over-represented in Manitoba's populations with subsequent the development, in direct collaboration with the communities of interest, of diagnostic or newborn screening programmes relevant to patient care. Areas of research include Hypophosphatasia in the Mennonite population, Glutaric Acidemia Type I in the Oji-Cree, CPT I Deficiency in the Hutterites and Inuit, Bowen-Conradi Syndrome and Limb Girdle Muscular Dystrophy in the Hutterites. Since July 2008 she has been the principal investigator for industry sponsored clinical trials hypophosphatasia.

Dr. Elizabeth Sellers

Elizabeth Sellers undertook her undergraduate medical training at the University of Western Ontario in London followed by a pediatric residency at the Children's Hospital of Eastern Ontario in Ottawa and pediatric endocrine training at McGIII University in Montreal. She completed the research component of her endocrine training in Manitoba, undertaking a Masters of Science in Community Health Sciences. She is currently an Associate Professor, Department of Pediatrics and Child Health, University of Manitoba. As a clinician and clinical researcher, her primary focus has been the epidemiology, pathophysiolology, complications treatment and support of youth with type 2 diabetes with a particular interest in Indigenous populations. Liz is privileged to be part of the interprofessional team in the Section of Pediatric Endocrinology and Metabolism at the Winnipeg Children's Hospital.

Ms.Rebecca Ulrich

Rebecca Ulrich is the Provincial Manager with the Canadian Red Cross' RespectED: Violence & Abuse Prevention program in Manitoba. She is an accomplished trainer and facilitator, working with youth and adults, in education, health and social service environments. Rebecca holds a Bachelor of Human Ecology in Family Social Sciences, with a specialization in child and adolescent development and family violence.

Disclosure of Conflict of Interest

All Speakers and Planning 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. Please see below for all of our speakers, and planning committee to see their relationships, if any.

Speakers

Alexandra Ahmet

- · I have received payment from Nycomed
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- · I am a member of an advisory board or equivalent with Lilly and Serono
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- I intend to make therapeutic recommendations for medications that have not received regulatory approval

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Robert Barnes

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- I am a member of an advisory board or equivalent with Eli Lilly
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Numeracy in adolescents with type I diabetes: assessment and interactive gaming intervention – A Pilot Study

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Context: Numeracy is the ability to understand and use numbers in daily life and is key to diabetes self-management. In this study we examined the relationship between diabetes numeracy (DN), A1C, and quality of life (QL) in adolescents with type I diabetes (T1D). In addition, we studied the impact of a video game intervention designed to improve DN.

Research Design and Methods: In collaboration with diabetes team members and adolescents at our institution as well as experts in technology and education, we designed and developed a video game addressing key aspects of DN. Using the adolescent Diabetes Numeracy Test we assessed DN and we examined its relationship to AIC, QL, and demographic data. After playing the video game on 3 separate occasions we reevaluated participants' DN. To assess retention of the video game effects we re-evaluated baseline measures within I month following the last video game session and cumulative AIC over the following 6 months.

Results: We enrolled 42 adolescents (20 female, 22 male, mean age 15.5 years, 36% visible minority). A significant association between baseline A1C and numeracy scores (r=-0.43, p= 0.004) independent of ethnicity was observed. Although improvement in numeracy skills after the video game intervention did not reach statistical significance, some domains of QL measures significantly improved, including measures of emotional functioning (p=0.05). Qualitative analysis of the adolescents' experience with the video game showed that they felt challenged and remained engaged.

Conclusion: Numeracy skills are strongly correlated with diabetes control and should be targeted when educating patients with TID. We observed that video games are a feasible and developmentally appropriate method of delivering diabetes-centered experiential learning to adolescents. Larger studies are needed to further explore the role of gaming technology as a means of delivering diabetes education to adolescents with TID.

Quality of Referral of Children with Idiopathic Short Stature to the Pediatric Endocrinologist

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In 2001, our Endocrinology Service performed an audit of the quality of referral of children with short stature with regards to information provided and appropriateness of referral as judged by height velocity (HV) centiles and target height (TH). In 109 referrals (42 female/67 male), growth charts were provided for only 53%. Baseline investigations to rule out causes of short stature were not routinely performed (bone age 50%, CBC 30%, thyroid function tests 39%). Pediatricians (Ped) referred fewer children with normal height velocity than general practitioners (GP) (P=0.01), plotted more measurements on growth charts (P=0.02), and performed more baseline investigations (P=0.002); median (range): Ped 2 (2-8) vs GP 0 (0-6)). In response to these results, consultation by FAX was invited and missing information was requested by return FAX prior to the clinic visit. A second audit was performed in 2006 to assess if this improved the quality of referrals. Of 138 referrals (42 F/96 M), 69 were by FAX. 65% of referrals came from Ped (vs GP 31%). With FAX communication, growth curves were provided in 95.6% of cases (vs. 40.5% No Fax, p< 0.001 Pearson Chi-Square). More investigations were performed (Fax 6 (0-13) vs No Fax 1 (0-11), p< 0.001 Mann-Whitney). Finally, thanks to the information provided by Fax, 31 patients were not seen in clinic because of reassuring growth curve, laboratory results, and imaging.

In conclusion, with FAX communication, more baseline data were provided, and we were able to reduce the number of unnecessary visits of children with normal variants of growth. Advantages also include efficient triage, better communication between referring physicians and specialists, and an opportunity for continuing medical education. Attention must be paid to issues of confidentiality.

Two testes and 2 X chromosomes - Why?

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A 60 day old infant with genital ambiguity was assessed in the multidisciplinary urogenital clinic. The baby was born full term and weighed 3.4kg at birth. The antenatal history was non-contributory. On physical examination, there was no obvious dysmorphic features. On genitourinary exam, there was a well developed scrotum that was bifid, with rugae and pigmentation. There was penoscrotal transposition. His phallus was of a normal breadth and length with ventral curvature. In addition there was hypospadias at the base of the phallus. Both testes were palpable in the scrotum. Laboratory workup included an LH of 1.3 IU/L (0.1-4.8 IU/L), FSH 2.0 IU/L (0-15 IU/L), testosterone level was 3.7 nmol/L normal for age is < 16.0 nmol/L. Given the mild to moderately undervirilized male, the clinical impression was idiopathic isolated severe hypospadias. Surprisingly, further testing revealed a 46XX karyotype and FISH for SRY was negative. Genomic microarray analysis showed a copy number gain in chromosome region Xq27.1. This region contains the SOX3 gene. SOX3 (SRY related HMG box-containing gene 3) is a single exon gene located in a highly conserved region of the X-chromosome. It is a transcription factor, encodes a protein that is most similar to SRY. The clinical significance of duplication of the SOX3 locus in 46XX individual is currently unclear, however a recent report suggested that copy number changes within this genomic region may be associated with 46XX sex reversal. Further investigation of this copy number gain is in progress.

A Child with a Disorder of Sexual Development and Early-Onset Nephrotic Syndrome

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Denys-Drash syndrome (DDS) is a rare genetic disorder featuring the triad of congenital nephropathy, Wilms' tumor, and intersex disorder resulting from mutation in the Wilms' tumor suppressor (WTI) gene. This case illustrates the clinical course of an infant with DDS. This baby was seen for ambiguous genitalia. On physical examination there was a complete fusion of the labial scrotal folds, a phallus measuring 1.5 cm with urethra opening at the base of the shaft of the phallus. No gonads were palpable. The initial ultrasound had showed normal kidneys, normal adrenals, and no Mullerian structures. A work up of the DSS showed the karyotype was 46XY. He had a normal testosterone response to HCG stimulation test (12.6 nmol/L) and a normal testosterone to dihydrotestosterone ratio (8.5). He showed a good clinical response to testosterone. Laparoscopy revealed bilaterally undescended atrophic testicles. At 9 months of age, he presented with massive proteinuria. He underwent a renal biopsy that showed early changes of diffuse mesangial sclerosis (DMS). The baby was started on an ACE-inhibitor to reduce proteinuria and slow down the progression of deterioration. In DDS, the characteristic nephropathy is termed diffuse mesangial sclerosis. This condition clinically manifests as an early onset nephrotic syndrome and progresses to renal failure during the first 3 years of life. DDS is associated with pure XY gonadal dysgenesis. The vast majorities of these patients are destined to develop Wilms' tumor; thus require serial ultrasounds. They are at risk for the development of gonadoblastoma. Most deaths in these patients are due to end stage renal failure (ESRF). Better dialysis & transplantation programs allow improved survival in these children with DDS.

Unstimulated luteinizing hormone levels are able to predict subsequent pubertal progression in patients presenting with signs of precocious puberty

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Context: With the introduction of third generation immunochemiluminesence gonadotropin assays, there is now the ability to differentiate luteinizing hormone (LH) levels at the lower limits of detection.

Objective: To determine whether in children referred for evaluation of precocious puberty, a single basal LH level can predict subsequent pubertal progression.

Design, Setting and Participants: In this single-centre study at the Hospital for Sick Children, Toronto, we conducted a retrospective analysis of all patients (n=91, 75 females) who had been referred for assessment of early signs of puberty and who had undergone a gonadotropin releasing hormone (GnRH) stimulation test between 1st August 2007 and 31st December 2010.

Main outcome measures: The relationship of basal and stimulated gonadotropin levels to the progression in pubertal signs in the following six months.

Results: Pubertal progression occurred in 29 patients (6.3 \pm 2.4 years, 24 females), 17 patients with idiopathic and 12 with organic central precocious puberty. All patients with a basal LH level \geq 0.3 IU/L had subsequent pubertal progression, whilst in the 67 patients with a basal LH \leq 0.2 IU/L, 62 did not progress, resulting in a 100% specificity (95% CI 94.4 -100.0%) and 82.2 % sensitivity (95% CI 64.2- 94.2%). A stimulated LH level > 5 IU/L demonstrated 96.6% sensitivity (95% CI 82.2 - 99.9%), but only 85.5 % specificity (95% CI 75 - 93.4%).

Conclusion: A single basal LH level ≥ 0.3 IU/L is adequate to predict subsequent pubertal progression in patients presenting with precocious signs of puberty, eliminating the need for GnRH stimulation testing in most patients.

Health-Related Quality of Life (HRQOL) and Anxiety in Adolescents with Differentiated Thyroid Cancer (DTC)

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Background: Adolescents with DTC require lifelong monitoring with a high possibility of reoperation or radioiodine therapy. While adult DTC survivors have similar or slightly worse HRQOL this has not been evaluated in the pediatric population.

Objective: To compare HRQOL and anxiety in adolescents treated for DTC to patients with acquired autoimmune hypothyroidism.

Methods: In this cross-sectional study, 3 validated questionnaires were administered to 16 adolescents treated for DTC and 16 controls for assessment of quality of life, anxiety levels and significant life events. These included: Teen and parent PedsQL™, MASC and CLES-A. The contribution of age, time since diagnosis and biochemical variables were compared to the outcome measures.

Results: There were 16 DTC patients (7 males): 13 had papillary carcinoma, 1 had follicular carcinoma and 2 had mixed type. At diagnosis, 5 DTC patients had lymph node involvement and 2 had lung metastases, while at time of assessment only 1 DTC patient had lymph node involvement. The time since diagnosis of DTC ranged from 5 to 112 months. DTC patients were older than control subjects (p=0.004) and had lower TSH levels than control subjects at time of assessment (p=0.013). HRQOL and anxiety levels did not differ between DTC patients and control subjects. HRQOL was similar to previously reported in healthy cohort. HRQOL and anxiety level parameters were not influenced by time since diagnosis and free T4 levels measured at the time of assessment.

Conclusions: Adolescents with DTC have similar HRQOL and anxiety levels compared to healthy normative population and to those treated for autoimmune hypothyroidism.

Autonomic nervous system balance in craniopharyngioma related hypothalamic obesity

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Background: Imbalanced activity of the autonomic nervous system (ANS) is thought to be involved in craniophayngioma related hypothalamic obesity (CRHO). Increased parasympathetic activity and decreased sympathetic activity have been suggested.

Objective: To evaluate ANS activity using heart rate variability (HRV), in obese and overweight youth with craniopharyngioma (CRHO) compared to controls and to explore relationships between anthropometrics and ANS parameters.

Methods: A cross sectional study of 16 youth with CRHO and 13 controls matched for sex, age and BMI. Anthropometrics, fasting blood-work, 24hr holter monitoring for HRV and urine catecholamine collection were assesed. Questionnaires to evaluate sleepiness were filled.

Results: Craniopharyngioma patients had lower resting energy expenditures expressed as % from predicted (%REE) ($80\pm11\%$ vs. $90\pm10\%$ p=0.001), and a trend towards higher % body fat mass ($45\pm10.8\%$ vs. $38\pm10.3\%$ p= 0.086), and higher scores on the Epworth sleepiness scale (10.2 ± 3.6 vs. 7.0 ± 5.4 p=0.075) compared to controls. %REE negatively correlated with waist to height ratio in CRHO. HRV parameters of both parasympathetic activity (22.1 ± 11.4 ms2 vs. 20.0 ± 7.6 ms2 P=0.59) and sympathetic activity (1.4 ± 0.4 vs. 1.5 ± 0.3 P=0.29) did not differ between the groups. Parasympathetic activity had a negative correlation with % body fat in CRHO (p=0.029) and with waist to height ratio in both groups (p=0.025 and p=0.047). Sympathetic activity positively correlated with waist to height ratio only in CRHO (p=0.037).

Conclusions: Parasympathetic activity is negatively and sympathetic activity is positively correlated with anthropometric measures in children with craniopharyngioma. The level of activity and direction of parasympathetic correlation are similar to those seen in controls.

Congenital Blindness and Severe Juvenile Osteoporosis - Two Novel Mutations Causing a Rare Syndrome

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Osteoporosis-pseudoglioma syndrome (OPPG) is a rare autosomal recessive condition of congenital blindness and severe juvenile osteoporosis. It is caused by mutations in the low-density lipoprotein receptor-related protein 5 (LRP5) gene. We describe a case of OPPG initially presenting in infancy with blindness, and subsequently coming to attention in the second year of life with multiple fractures. This young child was noted to have blindness secondary to bilateral retinal detachment shortly after birth. At 13 months of age, she suffered a proximal femur fracture (fall from 2 feet). By 16 months of age, she was unable to stand and was having pain with movement. Her family history is notable for a sister with congenital blindness. Multiple family members have osteoporosis. On examination, she was well appearing with small orbits and no visual fixation. She had no bone tenderness to palpation. On imaging, she had multiple vertebral compression fractures (T6-T8, T10-T12, L2), in addition to buckle fractures at the left proximal tibial metaphysis. She had radiographic evidence of diffuse osteopenia. Laboratory investigations revealed normal iCa, Ca, Pi and Mg levels. Genetic testing revealed two novel mutations in the LRP5 gene (biallelic) consistent with OPPG. Given the clinical severity of her disease, zoledronic acid therapy was initiated. Since initiating bisphosphonate therapy, she has had no further clinically significant fractures, and she has had a subjective decrease in discomfort. OPPG is a rare syndrome combining congenital blindness and severe osteoporosis, of which there have been approximately 60 published cases. This patient's two novel mutations match her clinical presentation to this rare syndrome. Case reports indicate that OPPG patients may benefit from bisphosphonate therapy. Genetic testing for LRP5 gene mutations can confirm the diagnosis and may be of importance for family planning, as well as being informative to family members due to the incidence of decreased BMD in heterozygous mutation carriers.

Serum 25-Hydroxyvitamin D Test Utilization in Calgary

MUNIER NOUR, MARTHA LYON, JONATHAN DAWRANT

Background: Vitamin D deficiency is a common worldwide problem. The importance of Vitamin D in bone health is well established and emerging evidence has demonstrated a significant role in many other chronic disorders. As a result of increasing attention, ordering practices of 25-hydroxyvitamin D (25OHD) levels have changed dramatically over recent years. Serum 25OHD testing has increased exponentially and, as a result, so have costs. This has led to some regions in Canada restricting 25OHD ordering practices. We attempt to elicit emerging trends in ordering practices of 25OHD levels within Calgary over a 2 year period.

Methods: Serum 25OHD levels in Calgary were retrospectively analyzed over 24 months from January 2008 to December 2009. Ordering practices, care providers, baseline and follow-up tests were examined for emerging trends in 25OHD monitoring.

Results: Over 2 years >65,000 25-OHD levels were analyzed in 52,300 subjects. Among the data analyzed, 63% of tests were ordered by primary care physicians including pediatricians, 15% were ordered by subspecialists and 20% undocumented. Average time between repeat testing was approximately 6 months, however up to 20% of repeat tests were completed within 90 days of the previous test. Those who had repeat testing had a slightly lower 25OHD level than those who did not (64 nM vs 72 nM) with as many as 28% of repeat tests completed in individuals with baseline 25OHD values in the optimal range (75-225 nM). In subjects with low baseline 25OHD levels (<75 nM) 21% had subsequent testing.

Conclusions: Observations from this study suggest current testing strategies of 25OHD in the Calgary region do not comply with Osteoporosis Canada suggested Vitamin D monitoring guidelines. Routine testing is not recommended in low risk individuals under age 50 and repeat testing is not required following normalization of serum 25OHD. During the time period examined a fourfold increase in number of tests ordered was observed averaging from 1000 tests per month to greater than 4000 tests per month at study end. Excessive testing practices may necessitate introduction of restrictive laboratory policies to reduce health spending costs.

The Next Generation Cohort: Childhood Outcomes of Offspring of Parents with Early Onset Type 2 Diabetes

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The Next Generation cohort (NextGen) includes children born to parents with pediatric type 2 diabetes (T2D). These children exhibit classic risk factors such as unhealthy diet, lack of physical activity and obesity. They also possess novel risk factors such as a gene polymorphism, HNF-1 α G319S, and exposure to T2D in utero. When compared to children of mothers with adolescent-onset T2D, children of fathers with T2D can be used as controls to isolate the effect of the intrauterine T2D environment by controlling for HNF-1 α G319S inheritance.

Methods: Birth and family histories were collected on the children at recruitment. Annually, blood pressure, height, weight, waist circumference and the presence of acanthosis nigricans between the age 1-18 years were collected. At age 7, annual fasting blood glucose measurements and HNF-1α G319S genotyping was measured.

Results: NextGen has recruited 127 offspring of parents with youth-onset T2D since 2003. Sixty-six percent are obese at the most recent assessment with an average BMI z-score of 1.9, and 34.4% of the offspring age 10 years and older have T2DM. These children demonstrate many risk factors in prenatal and postnatal life. The cohort shows high frequencies of the hepatic nuclear factor - 1α G319S polymorphism (0.516) and the allele shows a near significant association with T2D. When comparing children of mothers and children of fathers, there was no difference in BMI z-scores or prevalence of T2DM.

<u>Conclusions:</u> The size of the cohort remains too small to make conclusions about the impact of the intrauterine environment and the HNF- 1α G319S allele on the risk of T2DM in this highly selected population.

Oral Abstract II

Is the frequency of ketoacidosis at onset of type I diabetes a child health indicator that is related to income inequality?

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Background: Income inequality, the difference in average incomes between a nation's highest and lowest income earners, has been shown to be an important determinant of health and social outcomes in advanced countries. Indicators of child well-being have been found to be less favorable where income inequality is greater.

Objective: To explore whether the frequency of diabetic ketoacidosis (DKA) at onset of type I diabetes (TID) may be an additional valuable indicator of child health outcomes, also influenced by income inequality.

Methods: We performed an environmental scan to obtain frequencies of DKA and income inequality data for all nations for which DKA frequencies were reported. We tested the relationship between income inequality and frequency of DKA at onset using simple linear regression. We also collected mean HbA1c levels in children with T1D, and assessed the relationship between income inequality and mean HbA1c.

Results: Our preliminary analysis suggests that frequency of DKA at disease onset is significantly related to income inequality in the world's wealthiest nations [figure I]. As poorer nations are added to the analysis, the significance of this relationship is lost. There is no relationship between mean HbAIc and income inequality.

Conclusion: Our findings suggest that income inequality may explain a substantial amount of variability in frequency of DKA at TID onset amongst children in wealthy nations. When poorer nations are included in the analysis, progressively less of the variability can be attributed to income inequality. This can be explained by the dilution of income inequality by severe levels of poverty: when a nation is so poor that access to food, water, insulin, and medical supplies is jeopardized, the level of income inequality becomes less relevant. Despite the lack of available, high quality and standardized data, our findings support policy initiatives aimed at reducing income inequality and preventing DKA.

Foot abnormalities in children with Type 2 Diabetes: a quality improvement project

KOJORI, F. SELLERS, E. SHAFER, L. AND HEATHER DEAN

Aims: To determine if a specialized foot care nurse integrated into a pediatric type 2 diabetes clinic would decrease the rate of foot care abnormalities in youth with type 2 diabetes.

Methods: Over a 9 month period from April 2010 to December 2010, 142 youth (less than 18 years of age) with type 2 diabetes were seen consecutively at the Winnipeg Children's Hospital. In addition to routine foot assessments provided by a physician, some of the youth were assessed by the foot care nurse. Foot abnormalities were defined as abnormal toe nail, ingrown toenail, use of antibiotics, or referral to surgery.

Results: Of the 142 youth (62% female), 76 patients had I visit, 54 had 2 visits, and 12 had 3 visits (total of 220 visits). In 116 of the 220 visits (53%), the youth was assessed by both physician and foot care nurse. Foot abnormalities occurred in 62%. Ingrown toenail was seen in (10%), antibiotics were given in (7%), and referral to surgery (2%). Of the 136 visits in which foot abnormalities were identified, the youth were seen by the foot care nurse twice as often as those who did not meet criteria for poor foot care definition (62.9% versus 30.8%). At follow up, with the foot care nurse, the rates of foot abnormalities decreased to 26.7%. Male gender was a risk factor for foot abnormalities.

Conclusions: The addition of a foot care nurse to the interprofessional pediatric type 2 diabetes team reduced foot abnormalities in youth with type 2 diabetes.

Clinical Management of Youth with Gender Dysphoria at BC Children's Hospital: Over 10 Years' Experience

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Background/Aim: To describe patient characteristics at presentation, treatment and response to treatment in youth with gender dysphoria.

Methods: A retrospective chart review of 81 youth with a diagnosis of gender dysphoria seen from 1998-2011.

Results: Of the 81 patients, 42/81 (52%) identified as female-to-male (FTM), 37/81 (46%) as male-to-female (MTF), and 2/81 (2.5%) natal males were undecided. Median age of presentation for FTM youth was 17.0 years (range 11.4–19.8 years), and median age of presentation for MTF youth was 16.6 years (range 12.3–22.5 years). GnRHa treatment was prescribed in 26/81 (32%) patients, and of these, 38.5% received their first dose on their first visit. One FTM patient developed sterile abscesses with Lupron Depot®; he was switched to Decapeptyl® CR and tolerated this well. Another FTM patient stopped Lupron Depot® 2 months after initiating treatment due to estrogen-withdrawal symptoms. Cross-sex hormones were prescribed in 61/81 patients (37/42 FTM vs. 24/37 MTF, p<0.02 by X²). Median age of initiation of testosterone injections in FTM patients was 17.8 years (range 13.7–23.7 years). Median age of initiation of estrogen therapy in MTF patients was 17.9 (range 13.3–22.3 years). Three patients stopped cross-sex hormones temporarily for reasons not related towards transitioning, but due to psychiatric co-morbidities (2 FTM) or testosterone effects causing androgenic alopecia (1 FTM). No severe complications were noted in patients treated with testosterone or estrogen.

Conclusion: Treatment with GnRHa and/or cross-sex hormones in collaboration with transgender-competent mental health care professionals is an appropriate and safe intervention in carefully selected youth with gender dysphoria.

A search for ectopic ACTH production in a 9 year old boy

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A 9 year old boy presented with 3 months of weight gain, acne and fatigue. He had a cushingoid appearance. Investigations demonstrated hypercortisolemia (cortisol 1442 nmol/L) and an elevated ACTH level (38.4 pmol/L, range 0-10.1) not responding to dexamethasone suppression. A 24 hour urine cortisol collection was >9999 mmol/day. An MRI demonstrated a 5 mm pituitary lesion and a CT demonstrated enlarged adrenal glands with a liver lesion thought to represent a hemangioma. He was referred for neurosurgical evaluation. Over several weeks the patient developed progressive Cushing's syndrome (cortisol 1124 to >2400 nmol/L), persistently elevated ACTH levels, hyperglycemia and hypokalemia. Metyrapone was initiated which led to some symptomatic relief and a reduction in cortisol from 1895 to 497 nmol/L over 24 hours. While investigations were ongoing the patient developed an adrenal hemorrhage. Repeat neuroimaging did not clearly demonstrate the pituitary lesion and inferior petrosal sinus sampling was consistent with ectopic ACTH production. The liver lesion was reevaluated. An RBC uptake scan was inconsistent with a hemangioma. An octreotide scan showed a focus of increased activity in the liver likely representing the source of ectopic ACTH. The boy underwent a wedge liver resection. Pathology demonstrated a well-differentiated neuroendocrine tumor that stained positively for ACTH by immunohistochemistry, with features suggestive of a metastatic lesion. No primary has been found. Morning cortisol levels quickly became undetectable and his energy improved. Three months after surgery, he remains on replacement doses of hydrocortisone. This boy will require ongoing monitoring for recurrence of this rare ACTH-producing tumour as well as adrenal function assessment with the goal of discontinuing hydrocortisone replacement.

Two cases of undervirilized XY infants with unexpected diagnoses

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We present two infants with XY karyotypes, mildly virilized female phenotypes and unexpected diagnoses. Parents were first cousins in both cases. Case I, a term infant, was the product of an uncomplicated pregnancy. A maternal cousin had undescended testes. At birth, the baby had mildly pigmented labioscrotal folds with palpable testes. Ultrasound confirmed bilateral testicles with no uterus or ovaries. Karyotype was XY. T (7.1nmol/L), DHEAS (2.9umol/L), LH, FSH and an ACTH stimulation test were normal. At 26 days, T (5.Inmol/L), DHT (2440pmol/L), T:DHT ratio (2.09) and androstenedione were normal. Molecular studies showed no alteration to the androgen receptor (AR) gene. Hormone binding assays confirmed normal AR activity but, unexpectedly, showed abnormal T-DHT conversion. Case 2, a term infant conceived by IVF, was born to a 29y.oGI female with PCOS and hypothyroidism. Two maternal aunts had primary amenorrhea. At birth, the baby had mild posterior fusion of the labioscrotal folds, bilateral palpable testes and mild clitoromegaly. Ultrasound showed bilateral testicles with no uterus or ovaries. Karyotype was XY. T (7.1nmol/L), DHT (5355pmol/L), T:DHT ratio (1.32), androstenedione, DHA-S, LH, FSH and an ACTH stimulation test were normal. Molecular testing of the AR gene was normal. On further genetic testing, Case I was found to have a nonsense mutation (g.51674C>T) and Case 2 had a missense mutation (g.51601G>A) of the SRD5A2 gene confirming 5-alpha reductase 2 deficiency. It is important to consider 5-alpha reductase 2 deficiency in an undervirilized XY infant, despite normal T:DHT. It is unclear why the DHT levels in both infants were robust, but it appears likely that the DHT assay in the newborn period can be falsely elevated. HCG stimulated T:DHT in undervirilized XY infants may help make an earlier diagnosis of 5-alpha reductase 2 deficiency, especially in the setting of parental consanguinity.

Borderline congenital hypothyroidism (CH): clinical characteristics and natural history

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Background: Borderline CH is characterized by an abnormal newborn screen (NBS) followed by mildly elevated TSH and normal FT4 on confirmatory tests. Literature on this condition is sparse.

Objective: To examine prevalence, characteristics & treatment outcomes of borderline CH. Methods: Retrospective study of 103 infants (60 male) with borderline CH followed in our clinic between 2000-2011. Borderline CH was defined as confirmatory TSH between 5-30 mU/L with normal FT4.

Results: Borderline CH prevalence within our CH clinic was 22.3% (103/462 patients). The incidence of borderline CH increased during the study period from 2/20 cases in 2000 to 31/74 cases in 2010. Thyroid scintigraphy results were abnormal in 77% (n=59/77) of the cases. 78% of patients were treated with levothyroxine at initial dose 8.3±2.4 mcg/kg. To normalize TSH, FT4 were often (49% of measurements) above reference during treatment. The treated group had significantly higher confirmatory TSH levels (p=0.001) and had undergone thyroid scintigraphy more often (p=0.0001) compared to the non-treated group. Among the treated infants who had reached 3 years of age, 45% underwent a trial off medication. Compared to those not trialed off therapy, these infants were less likely to have dose escalations during treatment (p=0.001).Trial off treatment was successful in 50% (n=7) of cases. FT4 prior to trial off levothyroxine were higher in children who remained off therapy (p=0.017).

Conclusions: Borderline CH is an increasingly common diagnosis in the CH clinic, now representing 42% of new patients. It is more common in males and is often a transient. Further studies are needed to determine optimum levothyroxine dosing and to determine if treatment improves neurocognitive outcomes.

Evaluation of Deprivation and Glycemic Control in Pediatric Type I Diabetes from a Large Urban Canadian Centre

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Discrepancies in the social determinants of health help to explain the health inequities that exist among individuals within a society. In patients with type I diabetes (TID), socioeconomic status and family structure have been associated with glycemic control. As the primary pediatric diabetes clinic in a large urban center, the Hospital for Sick Children (HSC) cares for a heterogeneous patient population. This evaluation sought to characterize this population through measurement of social determinants of health and to describe how these relate to diabetes metabolic outcome.

Objective: To characterize the pediatric TID patient population followed at our clinic with regards to socio-economic status and family structure (Deprivation Index), as they relate to glycemic control (defined by AIC)

Methods: De-identified patient postal code information was linked to the data set of the INSPQ Deprivation Index in order to obtain population-level measures of socioeconomic status and family structure for all TID patients followed at the HSC. The INSPQ Deprivation Index is based on 2006 Canadian Census Data and is composed of a material deprivation index (socioeconomic status) and a social deprivation index (family structure); each of which is reported as a quintile score. The deprivation index quintile scores were linked, by postal code, to mean patient AIC.

Results: The HSC TID patient population will be characterized by its distribution across quintiles for each of the deprivation indices. Mean AICs will be described by quintile for the material and social deprivation indices, for the overall patient population and by age group.

Interpretation: A better understanding of the social determinants of health and how they relate to diabetes control is of paramount importance to better address the needs of our diverse patient population.

Efficacy of Short Term 30/70 Insulin in Children with Type 2 Diabetes with AIC >9% at Diagnosis

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Children, 0-18 years of age, with type 2 diabetes in Manitoba and northwestern Ontario are treated and followed at the Diabetes Education Resource for Children & Adolescents (DER-CA). The usual practice at the DER-CA for children with an A1C >9% at diagnosis is to encourage aggressive lifestyle modification with 30/70 premix biphasic insulin twice daily. We assessed whether children with A1C >9% who were started on this regimen were able to stop insulin therapy by 3 and 12 months post diagnosis. A chart audit using the DER-CA database 2008-2010 was conducted. Only 53.3% of children with A1C >9% at diagnosis were started on insulin therapy. Of those, only 8.6% achieved a target A1C <7% or target blood glucose of 4-7 mmol/L on insulin therapy at 3 months or 12 months. The low success rate of the insulin therapy could be attributed to complex factors including a low compliance rate of ~40%.

Acute localized inflammatory reaction to insulin administration

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Objective: To present a case of localized inflammatory response to insulin and review potential treatment options.

Case: A 10 year old boy diagnosed with type I diabetes and maintained on Humulin N and Humalog (lispro, Lilly) insulins for two years suddenly developed painful, indurated, erythematous, pruritic lesions at the site of Humalog injections with associated local swelling. There were no associated systemic reactions such as rash or respiratory distress. The patient did not have a history of allergies, asthma or eczema. Aside from his type I diabetes, there were no other medical issues.

Trials of NovoRapid (aspart, Novo Nordisk) and Apidra (glulisine, Sanofi-Aventis) gave similar reactions. Skin prick and intradermal allergy testing with diluted insulin (1:10 and 1:100 respectively) failed to show an allergic reaction, suggesting localized inflammation. Subcutaneous injection challenge in the hospital showed reaction to all previously tried insulins and to previously unexposed Lantus (glargine, Sanofi-Aventis) and Levemir (detemir, Novo Nordisk) given via syringe and pen. There was a mild reaction to insulin diluent and no reaction to saline. Topical steroid cream prior to injection and the addition of dexamethasone to insulin have been proposed to minimize inflammatory reaction to insulin.

Conclusion: Inflammatory reaction is a rarely reported complication of insulin therapy with few known treatment options. We present a case of a boy with non-lgE mediated sensitivity to insulin and the outcome of anti-inflammatory interventions.

Occurrence of slipped capital femoral epiphysis in children undergoing leuprolide acetate depot therapy for the treatment of central precocious puberty

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Background: Endocrine disorders including hypothyroidism and growth hormone deficiency, but not precocious puberty, have been implicated in the occurrence of slipped capital femoral epiphysis (SCFE). SCFE is a late childhood hip disorder, peaking around age 12, which commonly presents in overweight or obese children. Interplay of mechanical and hormonal factors are thought to contribute to growth plate weakening resulting in slippage of the femoral head. There are a few reports suggesting an association between GnRHa therapy for central precocious puberty and SCFE. These reports have described SCFE in patients with CPP on active (2/6) or recently withdrawn (4/6) GnRHa therapy.

Cases: An 8.75 year old girl developed SCFE following 6.75 years of q3weeks Lupron 7.5mg injections for idiopathic CPP. At the time of event, her most recent bone age was 11 years (CA 8 years) with suppressed estradiol (33.8pmol/L, normal<100) and LH <0.2IU/L and normal IGF-I and TSH. BMI z-score was overweight at +1.75. Case two was a 10.6 years old girl who developed SCFE 3.3 years into GnRHa therapy. This patient was diagnosed with acute lymphoblastic leukemia at age 9 months, requiring radiation for bone marrow transplantation. She was also followed for short stature. CPP was diagnosed at 7.3 years and Lupron therapy varied from 7.5mg q3weeks to 22.5mg q3months due to injection anxiety. At the time of SCFE, bone age was 12 years with adequate pubertal suppression, normal TSH, and normal GH stimulation testing. BMI z-score was normal at +0.71.

Conclusion: Increasing evidence suggests an association between GnRHa therapy for CPP and the occurrence of SCFE. In four of six previously described cases, SCFE occurred shortly after withdrawal of GnRHa therapy for CPP, implicating rapid linear growth as a potential culprit for epiphyseal plate weakening. However, the other two reported cases as well as our two cases suggest that sex hormone suppression at critical times may result in inadequate epiphyseal plate maturation, thus predisposing to SCFE. Consequently, pediatric endocrinologists should be familiar with this potential adverse event during active GnRHa therapy or shortly after therapy withdrawal. Future longitudinal study is required to better describe this association.

Quantifying adherence to growth hormone treatment: The Canadian easypod™ connect observational study (ECOS)

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Background: SAIZEN®,(rh-GH Serono Canada) is indicated for pediatric patients with a variety of growth disorders. rh-GH is administered by daily sub-cutaneous injection, sometimes for many years, and adherence is challenging. Analysis of rh-GH adherence has been limited by recall bias. Complete and accurate recorded data on rh-GH use can now be collected with easypodTM. The ECOS, (clinicaltrial.gov identifier: NCT01267526) has been established to collect and analyze rh-GH dosing and clinical/auxological data from subjects prescribed SAIZEN® via easypodTM. Canada, is one of many countries currently recruiting in the ECOS, which is a multinational study.

Objectives: The primary objective of the ECOS is to assess the level of adherence of subjects receiving SAIZEN® via easypodTM.

Methods: Data will be obtained from subjects' medical notes; retrospective and prospective data will be uploaded from their easypod[™]. The proportion of subjects with ≥86% adherence at I year will be presented categorically. Data from the easypod[™] will provide a complete record of a patient's rh-GH use. Adherence data will be correlated with clinical outcomes. An adherence profile will be developed based on subjects' age, gender, indication, self-injection, and time on treatment.

Conclusions: Collecting accurate adherence data is difficult. This is true for rh-GH treatment in children with growth disorders. With data from ECOS, it will be possible for the first time to evaluate the clinical significance of rh-GH treatment adherence in various growth disorders and to explore its potential impact on growth outcome. Ultimately, drivers of and barriers to rh-GH treatment adherence will be identified, allowing appropriate support programs to be developed.

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Logos:

One to be chosen at AGM. We want your input!

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Canadian Pediatric Endocrine Group Groupe canadien d'endocrinologie pédiatrique

Mark your calendar

The next CPEG Scientific Meeting will be held in Quebec City on January 24 - 26, 2013 at the Delta Quebec.

The abstract submission is due on November 15, 2012 and the early bird deadline is November 30, 2012.