Canadian Pediatric Endocrine Group / Groupe canadien d'endocrinologie pédiatrique

2010

Scientific Meeting

March 4 - 6 mars 2010

Sheraton Suites Calgary Eau Claire

Calgary, AB

CPEG
GCEP

In cooperation with: UBC

Welcome

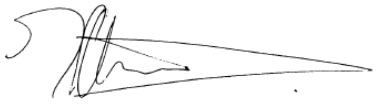
Dear Delegates,

Welcome to this 4th independent meeting of the Canadian Pediatric Endocrine Group (CPEG)/ Groupe canadien d'endocrinologie pédiatrique (GCEP). You may remember that CPEG was established in 1996 and originally met as a group of investigators who were part of an industry-sponsored randomized clinical study. In 2006, it was decided that CPEG meetings would be organized independently, starting with London, ON, in 2007. We have since then visited Vancouver (2008) and Ottawa (2009) and it is my pleasure to welcome you in 2010 in Calgary.

Over the last 4 years, our organization has thrived in many ways. Of course, CPEG remains an opportunity for all Canadian pediatric endocrinologists to attend a 2-day, high quality scientific program. Of course we remain a convivial group of friendly health professionals who enjoy meeting each other. But there have also been some recent developments that make the meeting experience each year more enjoyable. In 2008, Canadian endocrine nurses have started to organize their own sessions that are held in parallel to the main CPEG program. In 2009, thanks to istcl, CPEG has launched its own portal that will prompt us to communicate more often. This year, two industry-sponsored satellite symposia have been organized just before and after the scientific meeting.

I want to thank all our sponsors for supporting our meeting, the organizing committee for the many hours spent preparing the program and, last but not least, our fellows and residents for their stimulating presence.

Enjoy the meeting!



Jean-Pierre Chanoine, M.D.
President, CPEG, 2009-2011

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Thursday, March 4, 2010

16:00	Registration Open (Main Foyer)
17:00	Welcome Reception and Exhibits Open Hors d'oeuvre served (Wildrose South)
19:00	Adjourn
19:00	Satellite Symposium (please see the information in your kits)

Friday, March 5, 2010

07:45	Registration Open
07:45	Breakfast (Served in the Wildrose Centre)
08:30	Opening Remarks & Welcome (Wildrose North) *Dr. Jean-Pierre Chanoine,* *Dr. Danièle Pacaud and Lina Moisan*

Theme I: Disorders of Sex Development

Moderator: Dr. Diane Wherrett

08:45	Initiatives to Improve Outcomes in Surgical Management of DSDs *Dr. Pippi Salle, Toronto, ON*
09:30	Care for Disorders of Sex Development: Ongoing Challenges Dr. Katrina Karkazis, Mill Valley, CA
10:15	Refreshment break, exhibits open
**10:45	Split rooms*, MD's: Abstracts from CPEG Members and Fellow Presentations Nurses to meet in the Willow Room (please refer to Page 4 of the program)

Theme II: Abstracts

Moderator: Dr. Josephine Ho

10:45	MD's: Abstracts from CPEG Members and Fellow Presentations (See Page 6 for detailed schedule)
12:00	Lunch (Provided in Wildrose Centre)
12:45	Coffee and Dessert in the Exhibit Area & Poster Session: *Poster presenters will be available for questions from 12:45 – 13:30.*

13:30 MD's: Fellow Presentations Cont'd

15:00 Nutrition break, exhibits open

Theme III: Beta Cell
Moderator: Dr. David Hill

15:30 The Role of Growth Factors in Regulation of Pancreatic Beta Cell Mass
Dr. Carol Huang, Calgary, AB

16:15 Role of Immune Modulation in Beta Cell Preservation: Updates
Dr. Pere Santamaria, Calgary, AB

17:00 Adjourn

****Nursing Program (in the Willow Room)**
Moderator for Presentation: Ms. Wendy Schwarz
Moderator for 2009 Updates: Ms. Eileen Pyra

10:45 Presentation: Engaging Your Multi-Generational Workforce
(including 15-minutes Q&A)
Ms. Glenna Cross, Calgary, AB

12:00 Lunch (Provided in Wildrose Centre)

12:45 Coffee and Dessert in the Exhibit Area
Poster Session: *Poster presenters will be available for questions from 12:45 – 13:30.*

13:30 Round Table

14:30 CPEN/ICEP AGM

15:30 Coffee Break (in the meeting room)

15:45 2009 Updates:
- Web Portal Use, Robert Preston, istcl
- 2009 PENS Conference, Susan Rybansky
- Disorders of Sexual Development Conference, 2009, Peggy Kalancha

17:00 Adjourn

19:00 Reception, in the exhibit area, exhibits open

19:30 Dinner

Saturday, March 6, 2010

07:00 Breakfast Served in the exhibit area - exhibits open

07:45 Business Meeting

09:45 Refreshment break, exhibits open

10:15 CPEG Fellowship Award

Presentation of Dr. John Bailey Resident Research Award
Presented by Dr. Jean-Pierre Chanoine

Theme IV: Relationships with Industry
Moderator: Dr. Danièle Pacaud

10:30 Considerations for the Clinical Use of Subsequent-Entry Biologics in Canada
Dr. Philip Schwab, Ottawa, ON

11:15 Physicians and the Pharmaceutical Industry: Contemporary Thinking about Conflicts of Interest
Dr. Jocelyn Lockyer, Calgary, AB

12:00 Lunch and dessert, exhibits open

Theme V: Adrenal
Moderator: Dr. David Stephure

13:00 Adrenal Gland Function and Adrenal Insufficiency in Neonates
Dr. Susan M. Scott, Albuquerque, NM

13:45 Clinical and Molecular Genetics of Adrenal Hyperplasias and Other Tumours
Dr. Constantine A. Stratakis, Bethesda, MD

14:30 Nutrition break, exhibits open

Theme VI: Growth Hormone in PWS Patients
Moderator: Dr. Danièle Pacaud

15:00 Debate: Growth Hormone in PWS Patients
Pro: *Dr. Jean-Pierre Chanoine, Vancouver, BC* vs.
Con: *Dr. Guy Van Vliet, Montréal, QC*

16:00 A Protocol for a Pan-Canadian PWS Study
Dr. Cheri L. Deal, Montréal, QC

16:10 Evaluations & Closing Remarks

16:15 Adjourn

18:00 Satellite Symposium (please see the information in your kits)
Have a safe journey home!

Fellow Presentation Schedule: Friday, March 5, 2010

10:45 Effect of Intravenous Bisphosphonate Therapy among Boys with Duchenne Muscular Dystrophy and Osteoporosis: Clinical Outcomes
(Anne Marie Sbrocchi, Frank Rauch, Victor Konji, Monica Tomiak, M.Math, Pierre Jacob, Leanne Marie Ward)

11:00 Treatment of Symptomatic Osteoporosis with One-day Intravenous Pamidronate
(Tania Martinez-Soto, Danièle Pacaud, David Stephure, Rebecca Trussell, Carol Huang)

11:15 Macrophage-muscle Crosstalk in Obesogenic Environment: Some Cells Talk and Others Listen
(M. Constantine Samaan, Philip J. Bilan, Amira Klip)

11:30 Undetectable AMH Levels Allow Earlier Diagnosis of Anorchia than FSH
(Sophie Stoppa-Vaucher, Anissa Djemli, Guy Van Vliet)

11:45 Increased Hepatic and Skeletal Muscle TriglyCeride Content is Associated with Insulin Resistance in Adolescents
(Brandy Wicklow, Kristy Wittmeier, Andrea Macintosh, Elizabeth Sellers, Heather Dean, Jonathan McGavock)

13:30 Long-term Outcome of Paediatric Patients with Pseudo Vitamin D Deficiency Rickets
(Thomas Edouard, Nathalie Alos, Francis Glorieux, Franck Rauch)

13:45 Hyponatremic, Hyperkalemic Dehydration in a Female Infant
(Jonathan D. Wasserman, and Farid Mahmud)

14:00 Primary Adrenocortical Insufficiency in the Pediatric Population of Newfoundland and Labrador
(Sarah Tsai, Joseph Curtis, Bridget Fernandez, Ara Healey, Fiona Curtis)

14:15 When Can 'Normal' Thyroid and Gonadal Function Not Be Normal?
(Stefanie Wildi-Runge, Yves Robitaille, Cheri L. Deal)

14:30 Are Guidelines for Glucocorticoid Coverage in Adrenal Insufficiency Currently Followed?
(Coralie Leblicq, Diane Rottembourg, Johnny Deladoëy, Guy Van Vliet, Cheri L. Deal)

14:45 Effectiveness of a Multidisciplinary, Family-Centered Weight Management Program for Children and Adolescents
(Mohammed Al-Dhubaiei, Mary Hinchliffe, Louise Masse, Jean-Pierre Chanoine, Constadina Panagiotopoulos)

Program Organizing and Scientific Committee

Alex Ahmet	Robert Barnes	Jean-Pierre Chanoine
Cheril Clarson	Brenda Fraser	Josephine Ho
Danièle Pacaud	Zubin Punthakee	Eileen Pyra
Wendy Schwarz	Diane Wherrett	

Credits

This event is an Accredited Group Learning Activity as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada and approved by the Canadian Paediatric Society (16.75 credit hours).

Learning Objectives

Theme I: Disorders of Sex Development

Initiatives to Improve Outcomes in Surgical Management of DSDs
Dr. Joao Luiz Pippi Salle, Toronto, ON

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

1. describe the historical evolution of surgical techniques designed for genital reconstruction in DSD.
2. explore the surgical thinking behind the innovative techniques for DSD.
3. discuss methods used to measure outcomes of surgery for DSD and results.
4. elaborate some of the difficulties encountered when working in intraprofessional teams.

Care for Disorders of Sex Development: Ongoing Challenges
Dr. Katrina Karkazis, Mill Valley, CA

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

1. recognize events leading to a revised consensus statement on the management of DSD and features of the new guidelines regarding clinical care.
2. identify the range of psychosocial issues for those affected by a DSD and their families.
3. identify methods to improve care for those with DSD.

Theme III: Beta Cell

The Role of Growth Factors in Regulation of Pancreatic Beta Cell Mass
Dr. Carol Huang, Calgary, AB

At the end of the session the participants will be able to have an:

1. overview of molecular mechanisms that regulate beta cell mass.
2. overview of how growth factor(s) regulate signaling molecules that determines beta cell mass.
3. overview of clinical trials that use growth factors to enhance beta cell function.

Role of Immune Modulation in Beta Cell Preservation: Updates
Dr. Pere Santamaria, Calgary, AB

At the end of the session the participants will be able to answer these questions:

1. what are the fundamental immunological mechanisms underlying T1D?
2. what strategies are being considered for therapeutic intervention?
3. what qualifies as an "antigen-specific" immunointerventional approach in autoimmunity?

Theme IV: Relationship with Industry

Considerations for the Clinical Use of Subsequent-Entry Biologics in Canada
Dr. Philip Schwab, Ottawa, ON

In this session:

1. delegates will be introduced to the concept of Subsequent-Entry Biologics and the regulatory process that Health Canada is developing for their approval.
2. delegates will be introduced to the unique pharmacovigilance and patient safety aspects related to subsequent-entry biologics.
3. delegates will gain an understanding of how jurisdictions around the world are handling subsequent-entry biologics.

Physicians and the Pharmaceutical Industry: Contemporary Thinking about Conflicts of Interest
Dr. Jocelyn Lockyer, Calgary, AB

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

1. describe how 'gifts' (even modest gifts) create expectations for reciprocation.
2. discuss why medical leaders are concerned about the influence that industry (pharmaceutical and medical device manufacturers) has had on medical education and medical research.
3. identify how the influence of industry can be managed by teachers, researchers and medical leaders.

Theme V: Adrenal

Adrenal Gland Function and Adrenal Insufficiency in Neonates
Dr. Susan M. Scott, Albuquerque, NM

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

1. to evaluate the evidence for adrenal insufficiency in the newborn infant.
2. to understand etiology of adrenal insufficiency in the newborn infant.
3. to discuss the outcome of infants with adrenal insufficiency.

Clinical and Molecular Genetics of Adrenal Hyperplasias and Other Tumours
Dr. Constantine A Stratakis, Bethesda, MD

At the end of the session the participants will be able to understand:

1. the diagnosis of treatment of Cushing syndrome.
2. the genetic testing for adrenal tumor predisposition.
3. the molecular understanding of endocrine tumorigen.

Theme VI: Growth Hormone in PWS Patients

Debate: Growth Hormone in PWS Patients
Pro: Dr. Jean-Pierre Chanoine, Vancouver, BC vs.
Con: Dr. Guy Van Vliet, Montréal, QC

At the end of the session the participants will be able to understand:

1. to review the pathophysiology of Prader Willi Syndrome (PWS).
2. to understand the role of human growth hormone (hGH) on body composition and weight in PWS.
3. to discuss the overall benefit of hGH in PWS.

Nursing Program

Engaging Your Multi-Generational Workforce
Ms. Glenna Cross, Calgary, AB

This session's objectives are:

1. to increase participants' understanding of the influences and motivations of all generations.
2. to increase participant's ability to communicate more effectively with all generations.
3. to encourage valuing of the unique strengths and capacities of each generation.

Biographies

Dr. Joao Luiz Pippi Salle

Dr. Joao Pippi Salle is a consulting pediatric urologist at Hospital for Sick Children. After completing his medical degree in 1972, at the Federal University of Rio Grande do Sul, Brazil, Dr. Pippi Salle underwent his 7 years surgical training in General Surgery, Pediatric Surgery and Pediatric Urology in the USA, South Africa and Canada. He completed his PhD studies while in Brazil, developing a surgical technique for the treatment of refractory urinary incontinence in children.

Dr. Pippi Salle was an Associate Professor of Surgery (Urology) at McGill University from 1996 to 2003. During this time he was the Chief of the Division of Urology at The Montreal Children's Hospital. He moved to Toronto in 2003 as an Associate Professor of the Dept. of Surgery.

Dr. Pippi Salle has been involved in several educational activities since early in his career. He has chaired several pediatric urological events and workshops. He has always been involved in post-graduate teaching and received the Surgical Teaching Award of the Montreal Children's Hospital and the McGill Urology Teaching Award in 1997.
Over the last year Dr. Pippi Salle has been developing an Interactive Multimedia CD-ROM for Teaching Pediatric Urology which was supported by a successful application for the ITDF competition in 2004-2005.

Dr. Pippi Salle has 51 published papers in peer reviewed journals and is the author of 25 book chapters. He has been invited by several institutions around the world where he delivered more than 150 lectures.

His wife Nicola and 4 children Michelle, Alexandre, Gabriela and Ana Claudia have been supportive companions on his ventures.

Dr. Katrina Karkazis

Katrina Karkazis received a PhD in medical and cultural anthropology from Columbia University, where she also received a Masters in Public Health in maternal and child health. She subsequently completed postdoctoral training in empirical bioethics at the Center for Biomedical Ethics at Stanford University, where she currently holds the position of Senior Research Scholar. She is a member of the Stanford Hospital ethics committee and regularly performs ethics consults.

She recently completed a book on contemporary controversies over medical treatment for infants with disorders of sex development in the United States entitled Fixing Sex, Intersex, Medical Authority, and Lived Experience (Duke 2008), which has garnered positive reviews in the New England Journal of Medicine, Medical Genetics, American Journal of Human Genetics, American Journal of Bioethics, Choice, and Journal of Health Psychology.

Her work has appeared in the Lancet, Pediatrics, American Journal of Bioethics, and Hastings Center Report, among others. She frequently speaks to clinical and lay audiences about improving care for children with DSD and has been interviewed by numerous media outlets about DSD.

Dr. Carol Huang

Dr. Huang obtained her MD from University of Toronto in 1996. She completed her pediatric residency training, followed by a fellowship in pediatric endocrinology, both at the Hospital for Sick Children, Toronto. Between 2000 and 2004, she undertook her PhD studies under the supervision of Dr. Amira Klip at the University of Toronto, focusing on understanding how insulin signaling pathways regulate activity of glucose transporters in muscle cells.

In 2005, she joined the Alberta Children's Hospital as a pediatric endocrinologist. Currently, she is an Assistant Professor at University of Calgary.

Dr. Huang's research interest is on the role of growth factors in regulating beta cell mass. Specifically, her lab is looking at how placental hormones enhance beta cell proliferation, and the signaling pathways involved. Another area of research is aimed at understanding how adipokines regulates beta cell proliferation.

Dr. Pere Santa Maria

Dr. Santamaria is Professor in the Department of Microbiology and Infectious Diseases, and also Director and Chair of Julia McFarlane Diabetes Research Centre in the Faculty of Medicine at the University of Calgary, Alberta, Canada. He earned M.D. and Ph.D. degrees from the University of Barcelona, Spain, in 1983 and 1987, respectively, and completed his medical specialty training in Clinical Immunology in 1987, also in Barcelona. From 1988 – 1992, he pursued postdoctoral research training in the Department of Medicine and Institute of Human Genetics at the University of Minnesota in Minneapolis.

He joined the Faculty of Medicine at the University of Calgary in 1992. Dr. Santamaria has been the recipient of several honors and awards, including the Alberta Heritage Foundation for Medical Research Senior Scholar, Scientist and Senior Scientist awards, the Canadian Diabetes Association Young Investigator award and the Juvenile Diabetes Research Foundation Scholar award. Dr. Santamaria's research focuses on the pathogenesis and immunogenetics of autoimmune type 1 diabetes

Ms. Glenna Cross

Glenna is a communications consultant, with nearly 30 years experience in strategic communications, facilitation, communications teaching/training, employee engagement, business management, stakeholder consultation and leadership.

Glenna's business model as a consultant is all about building capacity within communications teams, as well as with the communication skills of managers and front line employees.

Her teaching and training focuses on employee communications including *"Engaging Your Multi-Generational Workforce"* and "*Face-to-Face Communications: A Tool for Employee Engagement"*.

Glenna's educational credentials include a Bachelor of Commerce in Marketing and the first-ever Master of Communications Studies from the University of Calgary. In addition, Glenna earned her accreditation (ABC) and the honorary Master Communicator designation from her professional association -- the International Association of Business Communicators (IABC).

Glenna is a proud Baby Boomer with a soft spot for hard core Gen Xer's and confidence in the future for the Millennials.

Dr. Philip Schwab

As Vice President for Industry Relations at BIOTECanada, Phil works with member companies in the health, vaccine, agriculture and industrial biotechnology sectors to develop policy positions and to communicate industry priorities to government and industry decision makers. Phil also serves as a member of the Multi Sector Advisory Committee for the Pan Provincial Vaccine Enterprise (PREVENT) and as a director of the Pharmaceutical Advertising Advisory Board on behalf of BIOTECanada.

Prior to joining BIOTECanada, Phil served as Director of Programs at Genome Canada, where he coordinated the scientific review processes for competitive programs across the spectrum of genomics and proteomics research. For over ten years Phil served as a science policy and legislative affairs advisory at the United States Department of Agriculture in Washington, DC and as a professional staff member on Capitol Hill for Senator Tom Harkin of Iowa, Representative Earl Pomeroy of North Dakota and Senator Tom Daschle of South Dakota. Dr. Schwab holds a Ph.D. and M.S. in Plant Breeding and Genetics with a minor in conservation biology from the University of Minnesota and a B.S. degree from Michigan State University.

Dr. Jocelyn Lockyer

Jocelyn obtained a Masters in Health Administration from the University of Ottawa and a PhD in Adult, Continuing and Higher Education from the University of Calgary.

Jocelyn has been actively engaged as a medical educator at the University of Calgary where she teaches MSc and PhD students in the Medical Education Research Unit. Her students have done theses and published in all aspects of medical education from admissions, through undergraduate education, to residency education.

As an Associate Dean, she is responsible for the development and evaluation of short courses for practicing physicians. Recent funding from the Alberta Government through the Alberta Medical Association has led to the development of the Physician Learning Program. This program will provide physicians with data about their practices and be used in aggregate to guide new educational products.

Jocelyn is actively engaged in several research projects including the development and assessment of on multi source feedback, assessing the impact of short courses for practicing physicians, conducting needs assessment, and understanding the learning that occurs when physicians make geographic transitions in practice.

She recently chaired her Faculty of Medicine's Task Force to develop recommendations to manage the influence of industry on medical education. She has also served on a national task force of CME Associate Deans examining the influence of industry on CME.

Dr. Susan M. Scott

Susan Scott, MD is a Professor Emerita at the University of New Mexico School of Medicine. Dr. Scott completed a residency in Pediatrics and a fellowship in Neonatology at Washington University. She followed this with a fellowship in Pediatric Endocrinology at Harbor-UCLA within interest in the relationship between growth factors and the endocrine systems. While working with Del Fisher, at Harbor, she began exploring the relationship between thyroid hormone and both epidermal and nerve growth factors in both the newborn infant and a mouse model.

In 1984, beginning a 25 year faculty career at the University of New Mexico, she began working on the relationship of cortisol as well as thyroid hormone on EGF. EGF values were low and then increased with dexamethasone therapy in preterm infants with severe lung disease. In those who recovered, the EGF values stayed higher than in those infants who died after tapering suggesting a possible relationship between EGF and steroids.

Little information on normal cortisol values in the preterm infant was available and an exploration of this area lead to a shift away from growth factors and into studies of the relationship of cortisol to newborn outcome. Dr. Scott and colleagues reported relationships between gestational age, illness and requirements for support correlated to cortisol values. Further, when thyroid hormone and cortisol were studied for their relationship with survival, only cortisol values correlated with survival.

Dr. Scott began her service in the Dean's office in 1998while continuing to participate in research through mentoring junior faculty. She became senior associate dean for academic affairs, dealing with the hiring, separations and faculty development for a 800-member faculty. In July, 2009, Dr. Scott retired from the UNM School of Medicine and began as a 1L (first year student) at the UNM School of Law with the intention to work in child health law policy.

Dr. Constantine A. Stratakis

Dr. Stratakis received his medical and post-doctoral training in Athens, Greece, and National Institute of Child Health & Human Development (NICHD), Bethesda, MD, respectively. After residency and fellowships in Pediatrics, Pediatric Endocrinology and Medical Genetics at Georgetown University, Washington, DC, he went back to NICHD, where he is Head of the Program on Developmental Endocrinology and Genetics, and Director of the Pediatric Endocrinology Program.

Since July 1st, 2009, Dr. Stratakis has been serving as the acting Scientific Director of NICHD, NIH. Dr. Stratakis serves in the Editorial Boards of many journals and as of January 1st, 2010, is the Deputy Editor of the Journal of Clinical Endocrinology & Metabolism. Dr. Stratakis identified genes for Carney complex, bilateral adrenal hyperplasias and other endocrine disorders.

He is the author of more than 300 publications. His current work focuses on the genetic and molecular mechanisms leading to disorders that affect the adrenal, multiple endorine tumors and developmental abnormalities. His laboratory studies the function of the regulatory subunit type 1-A of protein kinase A (PKA) (PRKAR1A), phosphodiesterase-11A and 8B (PDE11A and PDE8B) and created mouse models in which the respective genes have been knocked out. Genome-wide searches for related genes are ongoing. More recently a new disease was described (Carney-Stratakis syndrome-CSS) in which his laboratory identified mutations in the succinate dehydrogenase subunits B, C and D (SDHB, SDHC and SDHD) genes. A search is ongoing for genetic defects leading to gastrointestinal stronal tumors that are associated with endocrine neoplasms in collaboration with other investigators.

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 care component attached to Shapedown BC, a clinical program addressing the needs of overweight children at BC's 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.

Dr Chanoine's research is focusing on the role of hormonal and nutritional factors in the development of childhood overweight. The main line of research includes laboratory-based projects aimed at understanding the physiological role of ghrelin, a potent orexigenic peptide, during the fetal and early postnatal period and clinical projects looking at the role of appetite-regulating hormones in the development and treatment of childhood and adolescent overweight. Dr Chanoine is also principal investigator of "Healthy Buddies", a peer-led curriculum aiming at preventing the development of overweight in elementary school children.

Dr. Guy Van Vliet

After college and medical school (1969-1976) and pediatric residency (1976-1980) at the Université Libre de Bruxelles (ULB) and pediatric endocrinology training at the University of California, San Francisco (1980-1983), Dr Guy Van Vliet was on staff at the ULB for six years before moving to the Hôpital Sainte-Justine/Université de Montréal in 1989, where he has been Chief of the Endocrinology Service since 1991 and a Professor of Pediatrics since 1995. In 2001-2002, he spent a sabbatical year at the Hôpital Robert-Debré in Paris. Congenital hypothyroidism has been a major focus of his research for the past two decades, with studies ranging from molecular pathophysiology to public health aspects such as population screening. On the other hand, he has remained involved in studies on the diagnosis and treatment of various other pediatric endocrine disorders, including on the clinical use of growth hormone for children with Prader-Willi syndrome.

Disclosure of Conflict of Interest

All Speakers and Planning Committee members have been asked to 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 a list of the speakers, moderators, and planning committee and their relationships, if any.

Alex Ahmet
- Has received a grant or an honorarium from Nycomed and MD Brief

Robert Barnes
- No relationship

Jean-Pierre Chanoine
- Is a member of an Advisory Board or equivalent with Eli Lilly, Serono, Roche
- Has received payment from commercial organizations: Eli Lilly, Serono, Roche

Cheril Clarson
- Is a member of an Advisory Board or equivalent with Eli Lilly
- Has received a grant or an honorarium from Serono

Glenna Cross
- Has received payment from commercial organizations (for extensive client list, please see www.cross-wise.ca)
- Hold investments in Cross Wise Communications

Cheri Deal
- Is a member of an Advisory Board or equivalent with GeNeSIS Scientic Advisory Board, and Eli Lilly
- Has received payment from commercial organizations: Eli Lilly, Serono
- Has received a grant or an honorarium from Eli Lilly, Pfizer, Serono, Novo Nordisk, Hoffman Canada
- Am currently participating in or have participated in a clinical trial with Eli Lilly and Serono

Brenda Fraser
- Is a member of an Advisory Board or equivalent with Eli Lilly, Serono, and Sandoz, as well as on the nursing advisory board
- Has received a grant or an honorarium from advisory boards

David Hill
- Has received a grant or an honorarium from Merck & Co.

Josephine Ho
- Am currently participating in or have participated in a clinical trial within the past two years with Macrogenics and Eli Lilly

Carol Huang
- No relationship

Katrina Karkazis
- No relationship

Jocelyn Lockyer
- No relationship

Danièle Pacaud
- Is a member of an Advisory Board or equivalent with Novo Nordisk
- Has received a grant or an honorarium from Pfizer, Eli Lilly, Serono and Sanofi Aventis
- Am currently participating in or have participated in a clinical trial within the past two years with Eli Lilly, Serono, and Macrogenics/Parexel

Eileen Pyra
- Has received a grant or an honorarium from Eli Lilly, Sandoz and Serono

Zubin Punthakee
- Is a member of an Advisory Board or equivalent with Serono, Eli Lilly
- Am a member of a speakers bureau for Serono, Eli Lilly, Novo Nordisk, Sanofi, Merck, GSK and Nycomed
- Has received a grant or an honorarium from Sanofi
- Am currently participating in or have participated in a clinical trial within the past two years with Boehringer Ingelheim, Roche

Joao Pippi Salle
- Holds a patent for a product referred to in the CME/CPD program or that is marketed by a commercial organization: Cook Urological

Pere Santamaria
- No relationship

Philip Schwab
- Employed by BIOTECanada who is an industry sponsored trade association. A member list can be found at http://biotech.ca/en/who-we-are/members.aspx

Wendy Schwarz
- Is a member of an Advisory Board or equivalent with EMD Serono Canada, Eli Lilly Canada
- Has received a grant or an honorarium from EMD Serono Canada, Eli Lilly Canada and nursing advisory board meetings

Susan M. Scott
- No relationship

David K. Stephure
- Has received a grant or an honorarium from Novo Nordisk Canada
- Am currently participating in or have participated in a clinical trial within the past two years with Novartis Pharmaceuticals and GeNeSIS Post Marketing Surveillance Study (Eli Lilly Canada)

Constantine A. Stratakis
- No relationship

Guy Van Vliet
- No relationship

Diane Wherrett
- Is a member of an Advisory Board or equivalent with Eli Lilly

Oral Abstracts

Effect of Intravenous Bisphosphonate Therapy among Boys with Duchenne Muscular Dystrophy and Osteoporosis: Clinical Outcomes

Anne Marie Sbrocchi, MD[1]; Frank Rauch, MD[2], Victor Konji PhD[1]; Monica Tomiak, M.Math[1]; Pierre Jacob, MD[3]; Leanne Marie Ward, MD[1]

[1]Division of Endocrinology, Children's Hospital of Eastern Ontario, Ottawa, ON, Canada

[2]Genetics Unit, Shriners Hospital for Children, Montreal, QC, Canada

[3]Division of Neurology, Children's Hospital of Eastern Ontario, Ottawa, ON, Canada

Introduction: Boys with Duchenne Muscular Dystrophy (DMD) may develop symptomatic vertebral fractures. Bisphosphonates have been used to treat the spine fragility; however, detailed analyses of the response to therapy are lacking. The objective of this study was to assess the efficacy and safety of IV bisphosphonate treatment in boys with spinal osteoporosis due to DMD.

Methods: This was a one-year, retrospective observational study of 7 boys (age 8.5-14.3 years) with DMD who had received either IV pamidronate (9 mg/kg/year) or zoledronate (0.1 mg/kg/year) to treat painful vertebral fractures. The co-primary outcomes were change in vertebral morphometry and back pain status at 12 months post-bisphosphonate initiation. Secondary outcomes included changes in lumbar spine volumetric bone mineral density (vBMD) and adverse events.

Results: A description of the cohort at baseline and 12 months is presented in the Table. All boys had at least one painful vertebral fracture. All but one had received glucocorticoid therapy prior to treatment initiation. Only one boy was pubertal and only one was fully ambulatory. Grade 2 (moderate) and Grade 3 (severe) vertebral fractures improved at 12 months, and this was associated with improvement of back pain. The median spine vBMD also improved. First-dose side effects were present in 4 patients and included fever (N=2), nausea (N=2), myalgias (N=3) and hypocalcemia (N=2).

Conclusion: In boys with spinal osteoporosis and DMD, IV bisphosponate therapy administered over 12 months was associated with improved vertebral morphometry; similarly, there was amelioration in back pain. The therapy was generally well-tolerated.

Table: Clinical Parameters Pre- and 12 Months Post-Treatment

Clinical Characteristics	Results 12 Months Post Bisphosphonate Initiation (N=7)	
	Pre-Treatment	12 Months Post
Anthropometry		
Height Z-score	-1.7 (-4.2, -0.5)	-2.0 (-3.5, -0.1)
Weight Z-score	0.4 (-2.4, 1.8)	-1.7 (-1.9, 1.9)
Back Pain, N	7	3
Vertebral Morphometry		
# of Vertebral Fracture (VF) Events	27	27
Genant Grade for VF Events, N (%)		
Grade 0.5 = 15-19.9% loss in VH	5 (18%)	11 (41%)
Grade 1 = 20-25% loss in VH	4 (15%)	7 (26%)
Grade 2 = 25.1-40% loss in VH	14 (52%)	9 (33%)
Grade 3 = >40% loss in VH	4 (15%)	0 (0%)
Lumbar Spine Volumetric BMD (Z-score)	-1.0 (-3.0, 0.9)	-0.1 (-2.6, 1.4)

Values reported are median (min, max) unless otherwise specified; VH=Vertebral height, BMD= Bone mineral density.

Treatment of Symptomatic Osteoporosis with One-day Intravenous Pamidronate

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The improved survival of children with chronic conditions has led to an increase in the prevalence of bone fragility in children. Poor bone health may be associated with bone pain, fragility fractures, and immobilization. Bisphosphonates have been used to improve bone strength in this group of patients. However, efficacy and optimal dosing in non-Osteogenesis Imperfecta (OI) patients is not established. We conducted a retrospective study of patients with non-OI-related bone fragility, with significant fracture history or reduced BMD as determined by dual x-ray absorptiometry, treated with either 9mg/kg/yr or 4mg/kg/yr of intravenous pamidronate. Our primary and secondary objectives were to compare the effects of 1-year pamidronate on changes in height-adjusted lumbar spine BMD Z score and fracture rate at these two doses.

Between 2000 and 2009, 15 patients received IV pamidronate at our clinic. Eight patients (group 1), age 10 ± 2.6 years (range 8-15), were treated with 4mg/kg/year of pamidronate while 7 patients (group 2), age13 ± 2.1 years (range 11-16,), were treated with 9mg/kg/year. Two of 8 patients in group 1 and 5 of 7 patients in group 2 were receiving corticosteroids. In the year preceeding biphosphonate, 10 of 15 patients had at least one fracture. After starting treatment, none of the patients experienced fractures. After one year, there was no significant difference in change in height adjusted lumbar spine BMD between the two groups (point estimate 0.300; 95%CI (-0.600, 0.850); P= 0.4863). No serious adverse effects or hypocalcemia were observed. In conclusion, in this retrospective study, iv pamidronate at a dosage of 4mg/kg/yr had an equivalent effect on height adjusted lumbar spine BMD z-score compared to 9mg/kg/yr. These results require substantiation with larger controlled clinical trials, to delineate the minimal effective dose in these patients and to address the long term safety of the treatment.

	Patient	Diagnosis	Sex	Age	Steroids	Fractures prior treatment	Fractures post treatment	Height z-score	DXA 1	DXA 2	Change in BMD
GROUP 1	1	IJD	M	15	No	Yes	No	1.256	-3.75	-3.6	0.15
	2	JRA	M	8	Yes	Yes	No	0.54	-2.8	-1.4	1.4
	3	DMD	M	13	Yes	Yes	No	-2.2	-1.5	-2	-0.5
	4	CP	F	9	No	Yes	No	-5.66	-1.8	-0.5	1.3
	5	DMD	F	8	Yes	No	No	0.14	-2.9	-1.6	1.3
	6	IJO	F	10	No	Yes	No	0.3	-2.4	-1.3	1.1
	7	IJO	M	9	No	Yes	No	0.43	-2.1	-1.7	0.4
	8	Beals Syndrome	F	8	No	Yes	No	0,2	-3.3	-2.4	0.9
GROUP 2	9	Nephrotic Syndrome	F	11	Yes	Yes	No	-2.7	-2.5	-1.5	1
	10	Nephrotic Syndrome	M	11	Yes	Yes	No	0.349	-3	-2.5	0.5
	11	Poor nutrition	M	16	No	No	No	-2.971	-3	-2.5	0.5
	12	DMD	M	14	Yes	No	No	0.12	-3.51	-3.3	0.21
	13	Crohn's	F	16	Yes	Yes	No	0	-2.4	-1.3	1.1
	14	IJO	F	14	No	Yes	No	0.328	-2.95	-2.4	0.55
	15	Chron	F	13	Yes	Yes	No	-2.8	-0.4	-0.239	0.165

Macrophage-muscle Crosstalk in Obesogenic Environment: Some Cells Talk and Others Listen

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Obesity has reached epidemic levels worldwide, and there is urgent need to understand its principal mechanisms. Obesity is part of a cluster of features termed metabolic syndrome, which in addition includes hyperlipidemia, insulin resistance and hypertension; this diagnosis carries significant health risks to patients including cardiovascular disease and type 2 diabetes. It has been demonstrated that obesity is associated with chronic low grade inflammation, and more recently the role of the immune system has been highlighted in this inflammatory response, with evidence of macrophage infiltration of adipose tissue. It is believed that both inflammation and hyperlipidemia [lipotoxicity] result in insulin resistance in metabolic organs including skeletal muscle, adipose tissue and liver; while this is well studied in adipose tissue, little information is available regarding mechanisms by which immune system- muscle interaction contributes to skeletal muscle insulin resistance and inflammation, despite the fact that muscle is the main organ for post-prandial glucose disposal.

Our work aimed to evaluate effect of obesogenic environment [using saturated fatty acid palmitate] on macrophage-muscle crosstalk; we used cell culture systems of rat skeletal muscle cell line [L6GLUT4myc] and both primary rat macrophages and macrophage cell lines. We found that treating macrophages with palmitate results in production of factors that render the muscle insulin resistant, production of chemoattractants that enhance migration of other macrophages, and activation of inflammatory pathways in muscle. On the other hand, treating muscle with palmitate results in production of factors that attract macrophages and activate inflammatory cytokine and chemokine gene expression in muscle. Our work highlights skeletal muscle-macrophage interaction in obesogenic environment and its role in development of inflammation and insulin resistance.

Undetectable AMH Levels Allow Earlier Diagnosis of Anorchia than FSH

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A newborn boy was referred to investigate micropenis and bilateral cryptorchidism. He was born at 40 gestational weeks to non-consanguineous Caucasian parents. The family history was negative for endocrinopathy and two brothers aged 8 and 11 years were healthy. On the first day of life, the pediatrician could not palpate the testes and noted a small penis. At 3 days of life, we could palpate a structure of about 5 to 8 mm in diameter in the inguinal canal bilaterally. Stretched penile length was 2 cm and diameter was 0.5 cm. No other anomalies were found apart from a right pre-auricular tag. On ultrasound examination, the two inguinal masses measured 5 mm (left) and 6 mm (right); they were hyperechogenic and calcified; no Müllerian structures were identified. Plasma FSH, LH and testosterone levels were not informative (13.42 UI/L, 4.91 UI/L, 2.13 nmol/l, respectively) but AMH was undetectable (< 0.1 ng/ml, normal range 15.5-48.7), allowing us to make the diagnosis of bilateral anorchia. At 6 weeks of age, the diagnosis was confirmed by elevated levels of FSH (84.65 UI/L) and LH (31.25 UI/L) and a low level of Testosterone (0.63 nmol/l). Three injections of testosterone enanthate (25 mg) were given IM at 6, 10 and 14 weeks, which resulted in doubling of penile size. We conclude that the level of AMH allows making the diagnosis of bilateral vanishing testis syndrome earlier than that of FSH. Immediately after birth, plasma gonadotrophins are still likely inhibited by maternal estrogens in the neonate even in the absence of functional gonads. This is reminiscent of the observations in newborn girls with Turner syndrome, who have normal FSH in spite of later evidence of ovarian insufficiency. An early diagnosis of bilateral anorchia avoids more invasive investigations such as hCG stimulation or laparoscopy. (296)

Increased Hepatic and Skeletal Muscle TriglyCeride Content is Associated with Insulin Resistance in Adolescents

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BACKGROUND: Excessive triglyceride accumulation in non-adipocytes is being elucidated as an important potent biomarker of glucose intolerance. Little information exists describing determinants of hepatic and muscle triglyceride in youth. This was an observational study of 78 normoglycemic adolescents classified as either healthy weight (n=15) or overweight (n=63).

METHODS: Hepatic and muscle triglyceride content was determined non-invasively with ^{1}H-magnetic resonance spectroscopy, allowing for discrimination of intracellular lipid accumulation, thought to be an integral component of insulin resistance at the tissue level and glucose intolerance. Percent body fat (DEXA), height, weight, and waist circumference were measured to assess anthropometric variables. Serum triglycerides and free-fatty acids during an oral glucose tolerance test were also measured to investigate the association between level of lipemia and organ triglyceride content.

RESULTS: In a multiple linear regression hepatic lipid content was positively associated with waist circumference ($p < 0.05$) and serum triglyceride levels (p 0.001) and insulin sensitivity. Muscle triglyceride content was not significantly associated with either anthropometric variables or serum triglyceride concentrations. Interestingly, hepatic and muscle triglyceride content were only modestly associated with one another, suggesting the potential for unique pathogenesis for steatosis in these organs.

CONCLUSIONS: Data suggest serum triglyceride levels, central adiposity, and adipocyte insulin sensitivity are critical determinants of hepatic steatosis in adolescents. 1H-magnetic resonance spectroscopy is a reliable method to non-invasively measure intracellular lipid content which will help aid in the advancement of the study of tissue steatosis in the future. Further study into interventions reducing serum lipid, and thus potentially tissue lipid content, are needed to target prevention of steatosis and progression of glucose intolerance in youth.

Long-term Outcome of Paediatric Patients with Pseudo Vitamin D Deficiency Rickets

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Background: Pseudo-vitamin D Deficiency Rickets (PDDR) is a rare autosomal recessive disease associated with mutations in the 25-hydroxyvitamin D-1α-hydroxylase gene (CYP27B1). It results in symptoms of abnormal ion homeostasis, secondary hyperparathyroidism, growth retardation, hypotonia, rickets, and osteomalacia. The treatment of choice is oral $1,25(OH)_2D_3$ (calcitriol). In children, it rapidly corrects the abnormal phenotype, restoring normocalcemia, eliminating secondary hyperparathyroidism and features of rickets. However, the long-term outcome of children with PDDR has never been documented.

Materials and Methods: This is a retrospective, clinical study evaluating the outcome of children with PDDR followed at the Shriners and CHU-Sainte-Justine in Montreal. All patients age $\geq$17y with PDDR were included and auxological, biochemical data and bone status were recorded.

Results: 25 patients (14 F, 11 M), with a median age of 26y (range 17 to 45) were studied. All received oral calcitriol at a median dose of 0.773 μg per day (range 0.5 to 1.5). Median height was -1.4 SDS (range -5.5 to 1) with short stature (height below -2 SDS) in 9 patients. Median weight was -0.3 SDS (range -3 to 2.2). All phosphocalcic parameters were normal with a median calcemia of 2.3 mmol/l (range 2.1 to 2.5), ionized calcemia of 1.3 mmol/l (range 1.2 to 1.3), phosphoremia of 1 mmol/l (range 0.7 to 1.4) and urinary calcium/creatinine of 0.45 (range 0.1 to 1.1). PTH levels were above the normal maximal value in 5 patients. None of the patients had nephrocalcinosis. Median area bone mineral densitometry (aBMD) of the lumbar spine was -0.4 Zs (range -2.3 to 2.6) with low aBMD (below -2 Zs) in 2 patients (8%).

Conclusion: Replacement therapy with 1,25(OH)2D3 results in normalization of biochemical parameters in adults with PDDR. However, short stature is found in one third of patients. BMD in these patients is in accordance with their height.

Hyponatremic, Hyperkalemic Dehydration in a Female Infant

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A 2 week-old infant was referred to our facility after presenting with emesis, lethargy and hemodynamic compromise. The child was afebrile and had been well until the day of presentation. Initial bloodwork revealed hyponatremia (Na=123) and severe hypokalemia (K=11.0) Newborn screen for CAH was normal.

The infant was the full-term product of a normal pregnancy, born to healthy non-consanguineous parents. There was no significant family history of early childhood death, metabolic, genetic or endocrine disorders.

On examination, the general appearance was that of a non-dysmorphic, phenotypic female without virilisation or hyperpigmentation.

Initial treatment included Hydrocortisone 100 mg/m^2/day. Sodium supplementation was introduced and measures were taken to treat the hyperkalemia including binding resins, inhaled beta-agonists and insulin-glucose infusion. Despite these interventions over the first 6 days of hospitalization, the electrolyte imbalance persisted, including symptomatic hyperkalemia with EKG changes. An empiric trial of mineralocorticoid was initiated but showed little benefit.

Investigations included baseline cortisol: 86 nmol/L, ACTH-stimulated cortisol: 996 nmol/L. 17-Hydroxyprogesterone: 2.1 nmol/L. Urine was sterile. Abdominal ultrasound revealed normal adrenal glands, kidneys, collecting system and uterus. The karyotype was 46, XX.

Prior to initiation of steroids, Plasma Renin Activity was 64 ng/L/s [0-13.9] and Aldosterone >57,780 pmol/L [<444]. Based on these data, a diagnosis of **Pseudohypoaldosteronism Type I** was established. Subsequent genetic testing revealed a novel mutation in the epithelial sodium channel (ENaC).

At 9 months, the infant remains on sodium supplementation and potassium-binding resin. Weight is at the 95th percentile for age and length at the 50th percentile. Developmental milestones have been met age-appropriately.

Summary: While CAH remains at the forefront of consideration when confronted with an infant with hyponatremic dehydration and hyperkalemia, other etiologies should be considered including obstructive and infectious uropathy, as well as specific lesions in mineralocorticoid processing and signalling, as illustrated by this case.

Primary Adrenocortical Insufficiency in the Pediatric Population of Newfoundland and Labrador

Sarah Tsai, Joseph Curtis, Bridget Fernandez, Ara Healey, Fiona Curtis

Autoimmune destruction of the adrenal gland is one of the most common causes of primary adrenal insufficiency in the pediatric population. Many patients with non-CAH adrenal gland failure are presumed to have Addison's disease. However, in the past decade, there has been a significant increase in the understanding of the molecular pathogenesis of congenital and acquired adrenocortical failure. Mutations in several genes have been described in recent literature. Such advances may have implications for the management of patients.

The objectives of our study were: (1) to describe the clinical presentation of children with new onset primary adrenal insufficiency that are presumed to be caused by an autoimmune process (2) to identify genetic mutations in patients with adrenocortical failure (3) to compare the clinical presentation of patients with an identified genetic mutation to those without a known genetic cause for adrenocortical failure.

Our study cohort consisted of thirteen patients followed for adrenocortical failure by the Pediatric Endocrinology service at the Janeway Children's Health and Rehabilitation Centre between January, 1980 and February, 2008. We reviewed the chart of each study participant for initial clinical presentation and for course of illness. Patients with negative anti-adrenal antibodies and no history of salt-wasting were selected to have testing for several gene mutations that are known to be involved in the development of primary adrenal insufficiency. One patient has a rare missense abnormality in the ACTH receptor gene and was clinically similar to other patients with isolated glucocorticoid deficiency. Two siblings, with isolated glucocorticoid deficiency and lack of hypogonadism, have a rare mutation in the steriodogenic acute regulatory (STAR) protein gene, which has been described in three other families (one of which was from Newfoundland). These findings underscore the need to consider diagnoses other than autoimmune adrenal dysfunction in patients with negative anti-adrenal antibodies and no clinical evidence of mineralocorticoid deficiency.

When Can 'Normal' Thyroid and Gonadal Function Not Be Normal?

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Background: Primary intracranial germ cell tumours usually present in the first two decades of life, often with precocious puberty. The most common location is in pineal gland; suprasellar germ cell tumours are rare.

Case: We report a French Canadian, 17 y-old male who presented to the ER with a history of mild weight loss and an episode of syncope while hiking in Mexico, but with no other neurological symptoms. Puberty began at age 13 y (growth spurt: 14-15 y), and he attained an adult height within genetic target by age 16 y. The previous medical history was negative. Systems review revealed increased thirst, nocturia, and myopia followed since childhood. The mother was treated for an oligo-astrocytoma in 2007. Clinical exam showed a euthyroid, well-looking young man with 20 cc testicles. Endocrine evaluation revealed elevated testosterone, mildly elevated PRL, borderline normal FT4, and reduced IGF-I, morning cortisol and urine osmolality; tumour markers were positive in serum and CSF (hCG, AFP). A transphenoidal biopsy of a 4.5 cm, homogeneous, non-calcified, suprasellar mass confirmed the diagnosis of choriocarcinoma and stained intensely for hGC and hGH, presumably the placental variant (GHv) as previously found *in vitro* in choriocarcinoma cell lines. Combined chemotherapy and irradiation lead to tumour regression and undetectable serum hCG to 24 months. He is doing well on complete hormone replacement therapy.

Conclusions and Teaching Points: Choriocarcinomas can have a hormonal profile that delays the development of symptoms, due to hCG stimulation of both the gonadal and thyroid axes. To what extent the patient's tumour GH contributed to his normal growth is not known. Caution is necessary during surgery since there is a very high risk of haemorrhage. Prognosis for this intracranial neoplasm is very reserved, although combined radiotherapy and chemotherapy has been successful to date in our patient.

Are Guidelines for Glucocorticoid Coverage in Adrenal Insufficiency Currently Followed?

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OBJECTIVES

To evaluate whether stress management protocols were followed in children with adrenal failure and to search for evidence of acute adrenal failure (AAF) linked to inappropriate care.

PATIENTS AND METHODS

Patients followed for primary adrenal insufficiency (PAI: n=102) or secondary adrenal insufficiency (SAI: n=34) by the Endocrinology Service between 1973 and 2007 were included. All hospitalizations subsequent to the initiation of glucocorticoid treatment, both urgent (n=157, 73% for infections) and elective (n=90) were examined. We recorded clinical evidence of AAF, glucocorticoid management prior to admission and details of glucocorticoid prescription and administration by the emergency department and ward medical teams. Data were analyzed over three time periods because of significant changes in health care personnel/delivery: <1990, 1990-1997 and 1998-2007.

RESULTS

For urgent hospitalizations, subgroup and time period did not influence the proportion of patients hospitalized. 45% of PAI and 38% of SAI ($p=0.55$) patients had at least 1 hospitalization. The use of stress glucocorticoid doses by parents increased significantly after 1997 ($p<0.05$), although still only 47% had increased glucocorticoids prior to hospitalization. Similarly, glucocorticoid stress doses were more frequently administered in the emergency department after 1990 ($p<0.05$), reaching 65% of urgent hospitalizations. Upon arrival, patients with signs and/or symptoms of AAF (58 urgent hospitalizations, PAI=51; SAI=7) decreased from 56% in 1990-1997 to 27% after 1997 ($p<0.01$). 24% of all hospitalizations were marked by sub-optimal adherence to glucocorticoid stress protocols, with rare but significant clinical consequences (2 cases of AAF with no long-term morbidity and 1 death in a child with septo-optic dysplasia).

CONCLUSIONS

Despite an increased use of glucocorticoid stress dose protocols by parents and physicians, patients remain at risk of morbidity and mortality from AAF. This risk may be minimized with a more conscientious application of glucocorticoid stress doses, but other patient-specific risk factors may also be implicated.

Effectiveness of a Multidisciplinary, Family-Centered Weight Management Program for Children and Adolescents

Mohammed Al-Dhubaiei, MD, MHSc, Mary Hinchliffe, MD, Louise Masse, PhD, Jean-Pierre Chanoine, MD, and Constadina Panagiotopoulos, MD, FRCPC

Purpose: Obesity places youth at both an increased risk of health problems such as diabetes as well as psychological distress. A recent environmental scan of Canadian obesity treatment centers showed that these programs had yet to be evaluated. Shapedown BC is a family-oriented, behavioral weight-management program that provides education and support to youth and their parents, and targets changes in physical activity, nutrition and psychosocial functioning. The group program is co-facilitated by a dietitian, psychologist and physical activity coach over 10 weekly sessions. In January 2007, an 18-month prospective program evaluation was initiated to investigate program effectiveness.

Methods: 119 obese youth aged 6-17 years (BMI ≥95th percentile for age and sex) and their families were enrolled. Evaluation at baseline and 10 weeks (program completion) included: height, weight, waist circumference, fasting glucose and insulin, and psychological assessment (Beck Youth Inventory (BYI) scale for self-concept, anxiety and depression, and Family Environment Scale (FES) to measure quality of family relationships). The Child Behaviour Checklist (CBCL) was completed by parents at baseline as a measure of behavioral and emotional problems. Statistics: paired samples t-test, $p < 0.05$.

Results: 88 participants attended 7 or more sessions. There was a significant decrease in BMI z-score (mean change -0.05 ±0.097; 95% CI -0.033, -0.07; $p < 0.0005$), waist circumference (-2.2 ± 4.24 cm; 95% CI -1.28, -3.12; $p<0.0005$), fasting insulin (-16 ± 69 pmol/L; $p<0.0005$) and fasting glucose (-0.02 ± 0.39 mmol/L; $p < 0.0005$). On CBCL, there was a high prevalence of internalizing (49.1%), and externalizing problems (19%) at baseline. There were significant improvements in self-concept (mean change 5.0 ± 8.0; 95% CI 3.3, 6.8; $p<0.0005$) and anxiety scores (-4.0± 10.6; 95% CI -6.3, -1.6; $p=0.001$).

Conclusions: Shapedown BC is effective in addressing medical and psychological concerns. Long-term follow-up is required to determine whether these effects can be sustained.

**This oral abstract will be presented in the nursing program.*

Childhood Osteoporosis: Screening, Treatment and Safe Handling Practices: A Child Health Project

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Osteoporosis is a challenge facing children of all ages with multiple different health conditions and physical abilities. The reality of this challenge stemmed the development of our Child Health Program's interdisciplinary project team here at the Winnipeg Regional Health Authority in Manitoba, Canada. Our goal is to develop protocols and tools to help identify at risk children and prevent fractures in these children. We have developed an evidence-based screening tool to allow primary caregivers to quickly recognize the "At Risk for Osteoporosis" child and determine their next level of care related to bone health. The use of our standardized evidence-based diagnosis, treatment and prevention protocols should empower all care providers to make bone health a priority for their patients. A "Handle with Care" protocol along with identifiable signage will give caregivers and others who may handle the child the ability to do so safely with adequate knowledge of fracture prevention strategies. A resource for staff and families which includes definitions of osteoporosis, diagnostic criteria, treatment plans as well as prevention strategies will be developed. Nutrition and lifestyle recommendations including activities of daily living, safe handling practices, and tips to prevent injury will also be explained. A key component is to expand fracture prevention in children with osteoporosis in the community through education of professionals and families.

Poster Abstract

An Unusual Cause of Hyperthyroidism – A Case Report

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A fifteen year old girl with a 2 month history of tremor, diaphoresis, weight loss, headaches and secondary amenorrhea presented with an elevated free T4 level of >65pmol/L and a TSH level of 10.5mIU/L (confirmed on repeated measurements). She was started on Tapazole and Propranolol treatment and referred to our centre for further investigation. The TSH level remained elevated and showed a blunted response to TRH stimulation. The remainder of pituitary labs were normal, as was IGF-1, TSH antibodies were negative, and alpha subunit level was elevated. On physical examination the patient was tachycardic, hyperreflexic, tremulous, diaphoretic and had a large and firm thyroid gland, which on ultrasound was inhomogeneous. An MRI of the brain was performed and revealed a 1.0 x 0.8cm lesion in the anterior pituitary gland. The patient was diagnosed with a TSH secreting adenoma and was started on subcutaneous Octreotide therapy in conjunction with Tapazole. She required large doses, up to 300mcg three times daily, in order to achieve euthyroid state. Once her thyroid function normalized, she underwent transphenoidal resection of the adenoma. On pathological examination the adenoma had an unusual appearance, staining for multiple hormones including TSH and Prolactin. One year after surgery our patient remains euthyroid on no medications with no evidence of recurrence of the adenoma.

TSH secreting pituitary adenomas, a rare cause of secondary hyperthyroidism, represent <1% of all pituitary adenomas and have been rarely described in the pediatric literature. Timely recognition and appropriate therapy is important to prevent potential complications of the growing lesion. The diagnosis should be considered when TSH levels are inappropriately nonsuppressed in the presence of high levels of free thyroid hormones, and can be confirmed by the presence of a pituitary lesion on MRI as well as supportive TRH stimulation test and elevated levels of alpha subunit. Important differential diagnoses include thyroid hormone receptor gene defects or assay problems. The treatment of choice for TSH secreting adenoma is surgical excision.

Table of laboratory values at baseline and TRH stimulation:

TRH stimulation test:

	Baseline	+20	+30	+60
TSH	9.43	12.4	12.9	11.0
FreeT4	>77.2 pmol/L			
T3	8.1 pmol/L			
TBII	<1.0 U/L			
Anti-TPO	<10 IU/ml			
α Subunit	5.0ng/ml (0.04-0.38)			

Poster Abstract

Allan-Herndon-Dudley Syndrome – A Case Presentation

Michal Cohen, Annette Feigenbaum and Jill Hamilton, Divisions of Endocrinology and Genetics/Metabolics, Hospital for Sick Children & University of Toronto

A 21 month old boy was referred to the Metabolic service with a history of significant hypotonia and failure to thrive. Work up revealed abnormal thyroid functions with increased serum triiodothyronine (T3) (5.8 pmol/L), low free thyroxine (8.9 pmol/L) and normal TSH level (3.8 mU/L). This combination is suggestive of Allan-Herndon-Dudley syndrome (AHDS). Genetic testing confirmed a mutation in the monocarboxylate transporter 8 (MCT8) gene.

AHDS, first described in 1944, is one of the first x-linked mental retardation syndromes reported. It is a rare syndrome characterized by a unique combination of mental retardation, hypotonia and high circulating T3. MCT8 facilitates cellular transport of thyroid hormone into several tissues, including the brain. Interestingly, mutations in this transporter protein may lead to tissue-specific resistance. In several reported cases of AHDS, transport of T3 into the central nervous system is decreased and peripheral tissues are exposed to high levels of T3 leading to symptoms of thyrotoxicosis such as tachycardia and poor weight gain. MCT8 is encoded by a gene located on chromosome X and in 2004 mutations were found to be associated with AHDS.

To date, no curative therapy is available for these patients. Treatment with a combination of propylthiouracil (PTU) and levo-thyroxine has been reported to demonstrate a beneficial effect on the nutritional status and heart rate; however with no influence on the neurologic condition.

Treatment with this combination was administered to our patient with initial beneficial effects on his general well being. Unfortunately, due to neutropenia side effect of PTU, medications had to be discontinued after only 3 weeks of therapy. 3,5-diiodothyropropionic acid (DITPA), a thyroid hormone analogue that is less dependent on MCT8 for transfer into tissues could potentially be of benefit but is not available as a registered medicine.

Poster Abstract

Case Presentation: Bone Deformities in a Previously Healthy Adolescent Female

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We report a case of a previously healthy 17-year-old girl who presented to the Children's Hospital of Eastern Ontario with a history of seizures and severe limb deformities. Seizure onset was approximately 3 years ago; however, despite this, our patient did not seek medical attention. Coinciding with a worsening of her seizures over the last year, our patient had become immobile and housebound. On exam, she appeared malnourished with a weight of 29 kg. Furthermore, she was noted to have severe short stature, scoliosis and multiple severe limb deformities. Initial blood work revealed severe hypocalcemia, an elevated alkaline phosphatase, and an elevated parathyroid hormone level. Phosphate and magnesium levels were normal. X-rays revealed multiple bony deformities secondary to microfractures and pseudarthrosis. Our patient was assessed by many services including neurology, gastroenterology, cardiology, psychiatry, adolescent medicine, and bioethics. Her course was further complicated by a complex psychosocial situation that involved a resistance to medical assessment and treatment by our patient and her caregivers: her mother and sister. Further evaluation revealed the etiology of our patient's clinical presentation.

Poster Abstract

A Case of Extreme Infantile Obesity and Hyperphagia

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Case Report: An 11 month old boy presented with a history of excessive eating and early onset weight gain. The infant was born at term with a birth weight of 3.810 kg, and an uneventful neonatal course. He was initially breast fed and demanded frequent feedings (hourly); at 4.5 months his weight was 13.8 kg (+6 Weight SDS) and his intake comprised 2100 ml of formula daily (1400 Kcal/d, Age appropriate energy need is 650 Kcal/d). The patient was the first child of consanguineous parents of Turkish descent, who were otherwise healthy, and non-obese. On physical examination, his weight was 18 kg (+ 4.9 Weight SDS), and his length was 74cm (-0.3 Height SDS). No dysmorphism was noted, and the examination was unremarkable aside from obesity.

Investigations: Endocrine testing (TFT's, cortisol, ACTH, IGF1 and insulin levels) were all within the normal range as were tests of lipid, liver and renal testing. Additonal testing revealed an elevated Leptin level of 86.6 ng/ml (1-35). Genetic evaluation showed normal male karyotype (46XY) and negative tests for Prader-Willi and Beckwith-Wiedman syndromes. Sequencing of the Leptin receptor revealed 2 homozygous missense mutations in the *LEPR* gene, not reported previously. The mutations were located in the fibronectin 3 domain where previously described mutations have caused complete or partial loss of function of the receptor. Sequencing of the MC4R was normal.

Discussion: We present a case of early onset extreme obesity and hyperphagia, with a family history of early onset obesity and parental consanguinity. The elevated leptin level excluded leptin deficiency, and further testing confirmed a novel LEPR mutation as the diagnosis. Research comparing children with genetic obesities shows that leptin deficient patients are more obese than LEPR deficient patients, who are clinically similar in obese phenotype to MC4R mutation patients. In this case, the combination of a family history of early onset obesity, consanguinity, and severe hyperphagia, led to the consideration of more comprehensive genetic testing.

Poster Abstract

Gigantism Due to Somatotroph Hyperplasia in a 4-Year-old Boy: A Genetic Mystery

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Rare in early childhood, gigantism is usually due to a GH-secreting pituitary adenoma. We describe a 4-year-old boy with growth hormone and prolactin excess as a result of hyperplasia.

From birth to 2 years of age, the boy grew along the 90th percentile. Then, his growth velocity increased gradually to 15 cm/year. At 4 y 8 m, his height reached 129.7 cm (+ 5.2 SD; height age, 8.5 y) and his weight, 35.5 kg (+ 4.0 SD). His family history was non contributory. On examination, he had coarse facial features, no cutaneous signs and Tanner I genitalia. Bone age was 5 years. Fasting serum GH level was high (> 35 mcg/L) and was not suppressed by an oral glucose load. Serum prolactin and IGF1 concentrations were elevated; 185.2 mcg/L (normal range, 0-18) and 74.5 nmol/L (normal range, 6.4-36), respectively. Brain MRI showed a 1.5 X 1.8 X 1.3 cm intrasellar solid mass. The surgically-removed pituitary tissue revealed somatotroph and lactotroph hyperplasia, without adenoma. Post operatively, GH and prolactin levels remain high and the patient requires medical management.

GH cell hyperplasia is rare in adults. In children, only four cases causing gigantism have been described, however two of which demonstrated pure hyperplasia. Etiologies for all age groups were attributed to McCune-Albright syndrome (MAS), GHRH-producing tumors, Carney complex and idiopathic congenital causes. Moreover, germline mutations in the aryl hydrocarbon receptor-interacting protein (AIP) gene have recently been associated with pituitary GH-secreting adenomas, but have never been related to somatotroph hyperplasia. We have excluded these diagnoses: negative GNAS1 (on hyperplastic pituitary DNA), and negative AIP gene mutations; normal GHRH level, 24-h 5HIAA urine collection and chromogranin A. To date, the etiology of gigantism in our patient remains undetermined. Additional genetic studies are underway to better understand molecular cause of somatotroph hyperplasia.

Poster Abstract

Rituximab Treatment in a Patient with Severe Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy

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Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED) is a rare monogenic autosomal recessive syndrome caused by mutation in the AIRE gene. The AIRE protein plays a role in transcriptional regulation of self-antigens, and is important in the induction and maintenance of central self-tolerance. In AIRE deficient mice and humans, tolerance to self-antigens is compromised and autoantibodies directed at multiple organs are generated by B lymphocytes. Rituximab, an anti-CD20 monoclonal antibody, causes depletion in B cells. B cell-directed therapies have shown promising results in autoimmune diseases such as rheumatoid arthritis and lupus, however clinical data in endocrine diseases is limited.

We report the use of Rituximab in a patient with severe APECED. In addition to the classic features of the disorder (adrenal insufficiency, hypoparathyroidism, candidiasis, keratoconjunctivitis with alopecia universalis), this patient also manifested signs of exocrine pancreatic insufficiency, diabetes mellitus and hepatic inflammation.

Initially, the patient was placed on oral CyA (5mg/kg/d) at age 13y, with remarkable improvement in gastrointestinal and endocrine function, but with renal side effects necessitating cessation of therapy at age 16y. Thereafter, a 5-year trial of thioguanine only slightly ameliorated her abdominal symptoms. Rituximab therapy (4x 375 mg/m^2/wk) was initiated at age 21y. Our patient showed remarkable improvement in energy levels, calcium balance, glycemic values and hair regrowth. Positive changes were temporary and by 6m post treatment, symptoms of, hypocalcemia, intermittent adrenal crises and even hair loss reemerged. After 5y of Rituximab therapy, it is evident that immunosuppressive therapy is needed in a cyclic pattern every 5-6m in order to stabilize calcium values, maintain proper energy levels and to prevent hospitalizations for adrenal insufficiency.

Conclusion: Rituximab may be an effective treatment for patients with severe APECED. Positive outcome seems to be transient with Rituximab maintenance therapy required biannually in order to prevent relapses, although vigilant attention to potential side effects is needed.

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Mark your calendar

The next CPEG Scientific Meeting will be held in Toronto at Le Méridien King Edward on February 10 – 12, 2011