2018 SCIENTIFIC MEETING

PROGRAM

February 22 – 24, 2018
Sheraton Vancouver Wall Centre Vancouver, BC

In cooperation with
Welcome
Dear Delegates,

I would like to extend to you a warm welcome to the 12th Annual Scientific Meeting of the Canadian Pediatric Endocrine Group (CPEG). Our past meetings have provided a wonderful opportunity for the Canadian pediatric endocrine community to come together to learn, network and share ideas. The organizing committee has worked hard to craft a program that highlights local Vancouver work and includes presentations by national and international experts. It also provides a forum for our fellows to present their work. We have an exciting program that we hope will meet the educational needs of our nurses, scientists, endocrinologists and trainees.

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

I wish you a stimulating and collegial meeting.

Sincerely,

Beth Cummings, MD, FRCPC
Scientific Chair, CPEG 2018 Scientific Meeting
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Financial Contributors

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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Further, Together
# Fellowship Listing

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<tr>
<th>Year</th>
<th>Fellows</th>
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<tbody>
<tr>
<td>1998-1999</td>
<td>J. Curtis, J. Hamilton</td>
<td></td>
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<tr>
<td>2000-2001</td>
<td>C. Panagiotopoulos, C. Huang</td>
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<tr>
<td>2002-2003</td>
<td>P. Krishnamoorthy, P. Zimakas, R. McEachern</td>
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The CPEG Fellowship Program was and/or is supported by the following: Eli Lilly, EMD Serono, Hoffmann La Roche, Novo Nordisk, Pfizer, and Sandoz.
Program

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

Thursday, February 22, 2018

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
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<tbody>
<tr>
<td>12:00</td>
<td>CPEG Executive Business Meeting (Chartroom, 3rd floor)</td>
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<tr>
<td>13:00</td>
<td>Fellow Symposium (for current/incoming CPEG Fellows only) (Port McNeill)</td>
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<tr>
<td>13:20</td>
<td>Welcome &amp; Lunch</td>
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<tr>
<td>14:20</td>
<td>Refreshment Break</td>
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<tr>
<td>14:50</td>
<td>Endocrine Care of Trans Youth</td>
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<tr>
<td></td>
<td>Dr. Dan Metzger</td>
</tr>
<tr>
<td>15:50</td>
<td>Conclusion</td>
</tr>
<tr>
<td>16:00</td>
<td>CPEG 2018 Registration Opens (Grand Ballroom CD &amp; Foyer)</td>
</tr>
<tr>
<td>17:00</td>
<td>Welcome Reception &amp; Exhibits (Grand Ballroom CD)</td>
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<tr>
<td>19:00</td>
<td>Adjourn</td>
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Friday, February 23, 2018

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
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<tbody>
<tr>
<td>07:00</td>
<td>Registration (Grand Ballroom CD &amp; Foyer)</td>
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<tr>
<td></td>
<td>Breakfast &amp; Exhibits (Grand Ballroom CD &amp; Foyer)</td>
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<tr>
<td>08:00</td>
<td>Opening Remarks &amp; Welcome (Grand Ballroom AB)</td>
</tr>
<tr>
<td></td>
<td>Dr. Beth Cummings, Ms. Susan Murphy, and Dr. Brenden E. Hursh</td>
</tr>
<tr>
<td></td>
<td>Poster Highlights</td>
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<tr>
<td></td>
<td>Each poster presenter will give a 1-minute &amp; 1-slide presentation</td>
</tr>
</tbody>
</table>
Theme I: Obesity (Grand Ballroom AB)  
Moderator: Dr. Melanie Henderson

08:30  Bugs at the Dinner Table: The Role of the Gut Microbiome in Obesity and Metabolic Syndrome  
Dr. Nikhil Pai

09:15  Severe Early-Onset Obesity: Genetic Causes and How to Diagnose Them  
Dr. William T. Gibson

10:00  Break & Exhibits (Grand Ballroom CD & Foyer)

Theme II: Cancer (Grand Ballroom AB)  
Moderator: Dr. Paola Luca

10:30  Fertility After Childhood Cancer: Possibilities for Improving Success  
Dr. Sheila Pritchard

11:15  Endocrine Late Effects of Childhood Cancer Therapy: What We Know and What We Still Need To Learn  
Dr. Stacey Urbach

11:40  Poster Walks

Bone/Calcium (Foyer)—Moderator: Dr. Natalie Alos  
Diabetes (Grand Ballroom AB)—Moderator: Dr. Robby Stein

12:15  Lunch & Exhibits (Grand Ballroom CD & Foyer)

* Nurses split, see page 7

THEME III: Diabetes (Grand Ballroom AB)  
Moderator: Dr. Susan Sanderson

13:30  Cystic Fibrosis Related Diabetes Mellitus (CFRD): A Common Rare Disease  
Dr. Katie Larson Ode

14:15  Metabolic Effects of Antipsychotic Treatment in Children with Mental Health Conditions  
Dr. Dina Panagiotopoulos

15:00  Break & Exhibits (Grand Ballroom CD & Foyer)

15:30  Oral Abstract Presentations (6) (Grand Ballroom AB)  
Moderators: Dr. Shazhan Amed & Dr. Ellen Goldbloom

17:00  Adjourn

FRIDAY NIGHT EVENT

Dinner & Entertainment at The Boathouse (1305 Arbutus St, Vancouver, BC)

Transportation will be provided from the Wall Centre at 17:30 and 17:45. Return bus will be provided from 20:30 – 21:25.

18:00  Cocktail Reception

19:00  Dinner and Professional Improv Show by Vancouver TheatreSports
*Nursing Program for Friday, February 23 & Saturday, February 24  
(Port McNeill)

Moderators: Ms. Susan Murphy, Ms. Barbara Butler

*Nursing Program for Friday, February 23

13:30  Mindful Awareness and Resilience Skills for Adolescents  
Sabrina Gill, Joanna McDermid, and a patient

15:00  Break & Exhibits (Grand Ballroom CD & Foyer)

15:30  Alternative and Emerging Therapies Promoted for the Treatment of Prader-Willi Syndrome: Clinical Necessity, Economic Burden, Hope or Hype?  
Irena Hozjan

16:15  Group Discussion: Challenging Cases (with a focus on Prader-Willi Syndrome)

16:45  Wrap-up and Evaluations

17:00  Adjourn

18:00  FRIDAY NIGHT EVENT  
See page 6 for more information

*Nursing Program for Saturday, February 24

13:30  CPEN AGM

15:00  Break and Exhibits (Grand Ballroom CD & Foyer)

15:30  Re-join CPEG group
## Saturday, February 24, 2018

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
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<tbody>
<tr>
<td>07:30</td>
<td>Breakfast (Grand Ballroom CD)</td>
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</table>
| 08:00 | Business Meeting (Room: Grand Ballroom AB)  
   Fellowship Awards  
   *Presented by Dr. Celia Rodd* |
| 10:00 | Break & Exhibits (Grand Ballroom CD & Foyer) |
| 10:30 | **THEME IV: Laboratory Conundrums (Grand Ballroom AB)**  
   *Moderator: Dr. Sarah Lawrence*  
   Featured Presentation  
   *Dr. Dan Holmes* |
| 12:00 | Lunch & Exhibits (Grand Ballroom CD & Foyer)  
   *Poster Walks*  
   General Endocrine (Foyer)—*Moderator: Dr. Alexandra Ahmet*  
   Transhealth/Genetics (Grand Ballroom AB)—*Moderator: Dr. Dan Metzger* |
| 13:00 |  
   *Oral Abstract Presentations (6) (Grand Ballroom AB)*  
   *Moderators: Dr. Celine Huot & Dr. Brenden Hursh* |
| 15:00 | Break & Exhibits (Grand Ballroom CD & Foyer) |
| 15:30 | John Bailey Award (Webster BC)  
   *Presented by Dr. Elizabeth Sellers*  
   Debate: Be It Resolved That Growth Hormone Should Be Used in the Treatment of Children with Idiopathic Short Stature (ISS) (Grand Ballroom AB)  
   *Moderator: Dr. Ereny Bassilious*  
   *Pro: Dr. John VanderMeulen*  
   *Con: Dr. Mark Palmert* |
| 16:35 | Closing Remarks & Evaluation |
| 16:45 | Adjourn |
## Fellow (Oral) Abstract Schedule

<table>
<thead>
<tr>
<th>Time</th>
<th>Title</th>
<th>Presenter</th>
<th>Abstract #</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>15:30</td>
<td>Diabetes Appointment Attendance Around Transition to Adult Care: Type 2 vs Type 1 Diabetes</td>
<td>Katherine Pundyk</td>
<td>1</td>
<td>20</td>
</tr>
<tr>
<td>15:45</td>
<td>Adiposity is Associated with Early Vascular Damage in Children with Type 1 Diabetes</td>
<td>Sinead Glackin</td>
<td>2</td>
<td>21</td>
</tr>
<tr>
<td>16:00</td>
<td>Evaluating the Low-Dose ACTH Stimulation Test: Ideal Timings for Cortisol Measurement</td>
<td>Harpreet Gill</td>
<td>3</td>
<td>22</td>
</tr>
<tr>
<td>16:15</td>
<td>Evaluation of a 24-Hour Telephone Paging Service: Are We Preventing Emergency Department Visits for Pediatric Diabetes and Endocrine Patients?</td>
<td>Colleen Nugent</td>
<td>4</td>
<td>23</td>
</tr>
<tr>
<td>16:30</td>
<td>The Role GLP-2 In Development of Experimental Non-Alcoholic Fatty Liver Disease</td>
<td>Shai Fuchs</td>
<td>5</td>
<td>24</td>
</tr>
<tr>
<td>16:45</td>
<td>Chimeric 46XX/46XY Causes Ovotesticular Disorder of Sexual Development</td>
<td>Trisha Patel</td>
<td>6</td>
<td>25</td>
</tr>
</tbody>
</table>

### Saturday, February 24

<table>
<thead>
<tr>
<th>Time</th>
<th>Title</th>
<th>Presenter</th>
<th>Abstract #</th>
<th>Page</th>
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</thead>
<tbody>
<tr>
<td>13:30</td>
<td>A Rare Cause of Gross Hematuria</td>
<td>Stacy Zahanova</td>
<td>7</td>
<td>26</td>
</tr>
<tr>
<td>13:45</td>
<td>PMM2 Gene Mutation Found in a Child with Congenital Hyperinsulinism and Polycystic Kidney Disease</td>
<td>Helen Paciocco</td>
<td>8</td>
<td>27</td>
</tr>
<tr>
<td>14:00</td>
<td>Increasing Trends of Diabetic Ketoacidosis in Quebec, a Population-Based Study</td>
<td>Marie-Eve Robinson</td>
<td>9</td>
<td>28</td>
</tr>
<tr>
<td>14:15</td>
<td>Bone Health and Vitamin D Status in Transgender Adolescents Starting GnRH Agonists</td>
<td>Behdad Navabi</td>
<td>10</td>
<td>29</td>
</tr>
<tr>
<td>14:30</td>
<td>Type 1 Diabetes Outcomes: Does Distance to Clinic Matter?</td>
<td>Danya Fox</td>
<td>11</td>
<td>30</td>
</tr>
<tr>
<td>14:45</td>
<td>The Identification of Early Childhood Cardiometabolic Risk in Infants of Mothers with Gestational Diabetes</td>
<td>Nicole Coles</td>
<td>12</td>
<td>31</td>
</tr>
</tbody>
</table>
## Poster Abstract Listing

### Bone/Calcium | Friday, February 23, 2018, 11:40 (Foyer)

<table>
<thead>
<tr>
<th>Title</th>
<th>Presenter</th>
<th>Abstract #</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genotype Phenotype Correlation in Children and Adolescents with Osteogenesis Imperfecta: A Retrospective Cohort Study</td>
<td>Abdulmajeed AlSubaihin</td>
<td>1</td>
<td>32</td>
</tr>
<tr>
<td>Pamidronate First-Line Treatment of Hypercalcaemia in Neonatal Subcutaneous Fat Necrosis</td>
<td>Alexander Chesover</td>
<td>2</td>
<td>33</td>
</tr>
<tr>
<td>Evaluation of Obesity and Body Composition in a Genetically Diverse Cohort of Children with Osteogenesis Imperfecta</td>
<td>Nicole Coles</td>
<td>3</td>
<td>34</td>
</tr>
<tr>
<td>Psychostimulants and Calcitriol Interactions: Prescribers Beware!</td>
<td>Marie-Béatrice Saade*</td>
<td>4</td>
<td>35</td>
</tr>
<tr>
<td>Prevention of Post-Thyroidectomy Hypocalcaemia In Children and Adolescents</td>
<td>Kung-Ting (Jeff) Kao</td>
<td>5</td>
<td>36</td>
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*To be presented by a co-author.*

### Diabetes | Friday, February 23, 2018, 11:40 (Grand Ballroom AB)

<table>
<thead>
<tr>
<th>Title</th>
<th>Presenter</th>
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</thead>
<tbody>
<tr>
<td>Variability in Management of Diabetic Ketoacidosis in the Pediatric Population</td>
<td>Zoyah Thawer</td>
<td>6</td>
<td>37</td>
</tr>
<tr>
<td>Implementation and Evaluation of a DKA Order Set in a Pediatric Tertiary Care Hospital: A Quality Improvement Initiative</td>
<td>Kayla Flood</td>
<td>7</td>
<td>38</td>
</tr>
<tr>
<td>Living with Type 1 Diabetes, Youth and Parent Perceptions</td>
<td>Alex Fung</td>
<td>8</td>
<td>39</td>
</tr>
<tr>
<td>Seeing Eye-to-Eye: A Single Center Quality Improvement Experience in Retinopathy Screening and Reporting in Pediatric Type 1 Diabetes Patients</td>
<td>Carol Lam</td>
<td>9</td>
<td>40</td>
</tr>
<tr>
<td>Adherence to a Pediatric Diabetic Ketoacidosis Protocol in Children Presenting to a Tertiary Care Hospital</td>
<td>Rebecca Ronsley</td>
<td>10</td>
<td>41</td>
</tr>
<tr>
<td>An Observational Study of Canadian Children And Youth with Type 1 Diabetes Following Initiation of Continuous Subcutaneous Insulin Infusion (CSII)</td>
<td>Stacy Zahanova</td>
<td>11</td>
<td>42</td>
</tr>
</tbody>
</table>

### General Endocrinology | Saturday, February 24, 2018, 13:00 (Foyer)

<table>
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<tr>
<th>Title</th>
<th>Presenter</th>
<th>Abstract #</th>
<th>Page</th>
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<tbody>
<tr>
<td>An Under-Recognized Cause of False Positive Newborn Screening for Congenital Adrenal Hyperplasia</td>
<td>Paul Kahlke</td>
<td>12</td>
<td>43</td>
</tr>
<tr>
<td>Timing Of Peak Serum Cortisol Levels Following Low-Dose ACTH Stimulation In Neonates</td>
<td>Robyn LeDrew</td>
<td>13</td>
<td>44</td>
</tr>
<tr>
<td>Are We Doing Enough? Assessing Practice Patterns and Identifying Challenges to Providing Healthy Living Counselling at A Regional Tertiary Level Care Pediatric Hospital</td>
<td>Colleen Nugent</td>
<td>14</td>
<td>45</td>
</tr>
<tr>
<td>Self-Perception of Training In Core Pediatric Endocrinology Topics among General Pediatrics Residents: A Learning Needs Assessment</td>
<td>Paul Kahlke</td>
<td>15</td>
<td>46</td>
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</table>

### Transhealth/Genetics | Saturday, February 24, 2018, 13:00 (Grand Ballroom AB)

<table>
<thead>
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<th>Title</th>
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<tbody>
<tr>
<td>Then versus Now: Changing Characteristics in Adolescents Referred to a Gender Clinic</td>
<td>Lyne Chiniara</td>
<td>16</td>
<td>47</td>
</tr>
<tr>
<td>Perspectives of Transgender Youth and their Parents Regarding Fertility Preservation</td>
<td>Lyne Chiniara</td>
<td>17</td>
<td>48</td>
</tr>
<tr>
<td>Like Mother Like Daughter: Evolving Central Diabetes Insipidus Secondary to an AVP Gene Mutation</td>
<td>Julia Sorbara</td>
<td>18</td>
<td>49</td>
</tr>
</tbody>
</table>
Program Organizing and Scientific Committee

Ereny Bassilious  
Barbara Butler  
Nicole Coles  
Beth Cummings  

Danya Fox  
Brenden Hursh  
Laurent Legault  
Susan Murphy  

Jo Nam  
Dina Panagiotopoulos  
Caroline Zuijdijk  

Credits

This event has been approved by the Canadian Paediatric Society for a maximum of 8.75 credit hours as an Accredited Group Learning Activity (Section 1) as defined by the Maintenance of Certification program of The Royal College of Physicians and Surgeons of Canada. The specific opinions and content of this event are not necessarily those of the CPS, and are the responsibility of the organizer(s) alone.

Dr. John Bailey Resident Research Award

The John Bailey Award is named after Dr. John D. Bailey. Dr. Bailey graduated from the University of Toronto Medical School in 1949, did his training in pediatrics at the Hospital for Sick Children, Toronto and the University of Manitoba in Winnipeg. By the mid-1950s he was firmly established as a major contributor to pediatric care and education at the Hospital for Sick Children and University of Toronto. His major area of interest was in understanding normal and abnormal growth patterns in childhood and adolescence. He was the first chief of the Endocrine Division at the Hospital for Sick Children and an important contributor to the development of the Canadian national growth hormone treatment program. He was the consummate academic: clinically fastidious, intellectually challenging and constantly learning and sharing.

Below is a list of the recipients of the Dr. John Bailey Resident Research Award:

- 2007 Meranda Nakhla  
- 2008 Meranda Nakhla  
- 2009 David Saleh  
- 2010 Brandy Wicklow  
- 2011 Jonathan Wasserman  
- 2012 Jennifer Harrington  
- 2013 Karine Khatchadourian  
- 2014 Akash Sinha  
- 2015 Rayzel Shulman  
- 2016 Sanjukta Basak  
- 2017 Stephen Zborovski
Learning Objectives

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

Session Learning Objectives:

Fellows Symposium

Finishing Fellowship: Tips for Transition to Practice
Mark Inman, MD, FRCPC, Pediatric Endocrinologist, Department of Pediatrics, Royal University Hospital; Clinical Assistant Professor, University of Saskatchewan, Saskatoon, SK, Canada
1. List important steps to consider during the completion of your fellowship to pave the way for a successful transition to practice
2. Be able to apply various strategies for successful employment in pediatric endocrinology
3. Compare various practice options and be able to identify key factors that may influence your decision-making process when considering job opportunities

Endocrine Care of Trans Youth
Dan Metzger, MD, Investigator, BC Children’s Hospital; Pediatric Endocrinologist, BC Children’s Hospital; Clinical Professor, Division of Endocrinology, Department of Pediatrics, University of British Columbia, Vancouver, BC, Canada
1. Describe the spectrum and variety of gender identity and expression you will encounter in your training
2. Practice care in accordance with the 2017 Endocrine Society guidelines for treating gender dysphoria in adolescents
3. Recognize the importance of a trans-friendly approach in all aspects of medical care

Theme I: Obesity

Bugs at the Dinner Table: The Role of the Gut Microbiome in Obesity and Metabolic Syndrome
Nikhil Pai, MD, Assistant Professor, Department of Pediatrics, Division of Gastroenterology and Nutrition, McMaster University, Hamilton, ON, Canada
1. Briefly review the relationship between the intestinal microbiome and obesity
2. Discuss animal and clinical evidence supporting the role of gut bacteria in the development of Type II Diabetes
3. Describe the interaction between intestinal microbiota and nonalcoholic fatty liver disease

Severe Early-Onset Obesity: Genetic Causes and How to Diagnose Them
William T. Gibson, MD, PhD, FRCPC, FCCMG, Associate Professor, Department of Medical Genetics and Genomics, University of British Columbia; Senior Clinician Scientist, BC Children’s Hospital, Vancouver, BC, Canada
1. List five genetic tests useful in the workup of a suspected monogenic/oligogenic obesity disorder
2. Discuss signs in the physical examination that would indicate a monogenic obesity disorder is likely
3. List five paediatric-onset obesity disorders for which specific treatments are available
Theme II: Cancer

Fertility After Childhood Cancer: Possibilities for Improving Success
Sheila Pritchard, MD, FRCPC, Clinical Associate Professor, Division of Hematology and Oncology, Department of Pediatrics, University of British Columbia; Pediatric Oncologist/Hematologist, BC Children’s Hospital, Vancouver, BC, Canada
1. Review mechanisms that can adversely affect fertility in survivors of childhood cancer
2. Review methods of fertility prediction
3. Review available and potential future technologies for fertility preservation in this population

Endocrine Late Effects of Childhood Cancer Therapy: What We Know and What We Still Need To Learn
Stacey Urbach, MD, MPH, FRCPC, Division of Endocrinology, The Hospital for Sick Children; Assistant Professor, Department of Paediatrics, University of Toronto, Toronto, ON, Canada
1. Describe the endocrine late effects experienced by childhood cancer survivors
2. Utilize an exposure-based approach to the management of endocrine disorders in survivors of childhood cancer
3. Apply this approach to exposures for which we have limited late effects information

Theme III: Diabetes

Cystic Fibrosis Related Diabetes Mellitus (CFRD): A Common Rare Disease
Katie Larson Ode, MD, Clinical Associate Professor, Pediatric Endocrinology and Diabetes, University of Iowa Stead Family Children’s Hospital, Iowa City, IA, USA
1. Appreciate that diabetes is very common in people with cystic fibrosis
2. Understand how CFRD is unique from Type 1 and Type 2 diabetes mellitus
3. Practice CFRD care that is consistent with official Cystic Fibrosis Foundation (CFF) guidelines

Metabolic Effects of Antipsychotic Treatment in Children with Mental Health Conditions
Dina Panagiotopoulos, MD, FRCPC, Pediatric Endocrinologist; Investigator, Canucks for Kids Fund Childhood Diabetes Laboratories, BC Children’s Hospital; Clinical Professor, Division of Endocrinology, Department of Pediatrics, University of British Columbia, Vancouver, BC, Canada
1. Briefly describe the epidemiology of second-generation antipsychotic (SGA) prescribing practices in North America
2. Summarize the literature on the metabolic effects of SGAs
3. Describe a multi-disciplinary approach including evidence-based guidelines and resources for monitoring and management of these metabolic complications while recognizing the complex inter-relationship between mental health and metabolic risk

Theme IV: Laboratory Conundrums

Dan Holmes, MD, FRCPC, Clinical Professor, Pathology and Laboratory Medicine, University of British Columbia; Division Head, Clinical Chemistry, St. Paul’s Hospital, Vancouver, BC, Canada
1. Learn how mass spectrometry has affected endocrine practice - applications to thyroglobulin, steroids and insulin
2. Discuss challenges in the use of common endocrine tests including cortisol and thyroid function tests
3. Describe the nuances in the laboratory work-up to determine the cause of insulin-induced hypoglycemia
Clinical Debate

Debate: Be It Resolved That Growth Hormone Should Be Used in the Treatment of Children with Idiopathic Short Stature (ISS)

Pro: John VanderMeulen, MD, FRCPC, Division Head, Professor, Department of Pediatrics, Division of Endocrinology, McMaster University, Hamilton, ON, Canada

Con: Mark Palmert, MD, PhD, Associate Chair of Pediatrics (Ambulatory Care), Head, Division of Endocrinology, The Hospital for Sick Children, Toronto, ON, Canada

1. Define idiopathic short stature
2. List the pros of treating individuals with idiopathic short stature with growth hormone
3. List the cons of treating individuals with idiopathic short stature with growth hormone
Biographies

William T. Gibson
William Gibson MD PhD is a Senior Clinician Scientist at the BC Children’s Hospital Research Institute, affiliated with UBC. He is best known for his team’s discovery of the gene for Weaver syndrome in late 2011, and for their more recent discovery of EED-related overgrowth (Cohen-Gibson syndrome) in 2015-2016. Dr. Gibson did his MD at UWO, residency in Medical Genetics in Calgary and PhD at the University of Cambridge in the O’Rahilly lab.

As an Associate Professor in the Department of Medical Genetics at UBC, his team solves genetic disorders, with a focus on rare obesity and overgrowth syndromes. With funding from CIHR, his group is actively recruiting patients with severe, early-onset obesity, macrocephaly and/or tall stature. They are also actively recruiting patients with familial brain aneurysms (funded by the HSFC and the Brain Aneurysm Foundation). Dr. Gibson’s team welcomes contact and referrals from colleagues and trainees at every stage of their careers.

Sabrina Gill
Sabrina graduated with a Bachelor of Science in Nursing from the University College of the Cariboo. She also holds certificates from the Mental Health Specialty Program and Practice Education for Health and Human Services. Sabrina has gained extensive experience working with the Adolescent population as an RN and has a special interest in educating adolescents on how to build resilience and strength. She has the opportunity to work in a diverse role as a clinician, educator, and resource for staff, patients and the community. Her main goal is to advocate and encourage “youth friendly care” for the adolescent patients at BCCH.

Daniel Holmes
Daniel Holmes earned his undergraduate degree in Chemical Physics from the University of Toronto. He went to medical school at the University of British Columbia (UBC) where he also did his residency in Medical Biochemistry. He is a Clinical Professor of Pathology and Laboratory Medicine at UBC and Division Head of Clinical Chemistry at St. Paul’s Hospital in Vancouver. Interests include laboratory medicine statistics, clinical endocrinology with a focus on secondary hypertension, clinical lipidology and clinical mass spectrometry. Assay development efforts in the last five years have focused on novel use of mass spectrometry for assays directed at specialized endocrine testing.

Irena Hozjan
Irena Hozjan is a pediatric Nurse Practitioner in the ambulatory endocrine program at SickKids in Toronto, Ontario and is an Adjunct Lecturer at the Lawrence S. Bloomberg Faculty of Nursing, University of Toronto. She is a graduate of the Master of Nursing and Acute Care Nurse Practitioner Programs at the University of Toronto.

Ms. Hozjan is Past-President of the Canadian Pediatric Endocrinology Nurses and past executive member of the Canadian Pediatric Endocrine Group. She is a regional, national and international speaker on pediatric nursing and pediatric endocrine conditions and has been a consultant to College of Nurses of Ontario (CON), Registered Nurses of Ontario (RNAO), and SickKids International. Irena is a recipient of the Pediatric Endocrinology Nursing Society (PENS) Nursing Excellence Award for Advanced Clinical Practice.
Mark Inman
Mark Inman is a pediatric endocrinologist at the Royal University Hospital in Saskatoon and a clinical assistant professor at the University of Saskatchewan. He obtained his BSc at Acadia University in Wolfville NS (2005), MD at the University of Toronto in Toronto ON (2009), Pediatric Residency at Dalhousie University and IWK Health Center in Halifax NS (2013) and Pediatric Endocrine Fellowship at the University of Toronto and the Hospital for Sick Children (2015). He currently serves as the coordinator of the undergraduate medical education endocrine module at the University of Saskatchewan. His research interests include social determinants of health within pediatric diabetes care as well as quality improvement initiatives enhancing pediatric care.

Katie Larson Ode
Dr. Larson Ode obtained her MD from the University of Wisconsin and her residency in categorical pediatrics at Rainbow Babies and Children’s Hospital, Case Western Reserve University. She completed fellowship in Pediatric Endocrinology at the University of Minnesota, where she also completed a Masters in clinical research. Dr. Larson Ode is currently faculty at the University of Iowa. She has an active research program with ongoing NIH funding investigating the early pathogenesis of cystic fibrosis related diabetes mellitus (CFRD) in young children with CF. The University of Iowa has a rich environment of excellent basic science models for studying Cystic Fibrosis and CFRD; Dr. Larson Ode is a close collaborator with the CFRD teams which have developed two novel animal models (the CF ferret and CF pig), allowing, for the first time ever, animal models of CFRD where diabetes arises spontaneously in the context of the natural history of the disease. Dr. Larson Ode has also been recognized by the Cystic Fibrosis Foundation (CFF) as an emerging leader in CF endocrinology and has received grant support from the CFF for her clinical work in CFRD at the University of Iowa.

Joanna McDermid
Joanna McDermid is a Consulting Psychiatrist with BC Children’s Hospital and Clinical Instructor with Department of Psychiatry UBC Faculty of Medicine. Her work is centered around mindfulness and resilience and the creation of mindful organizations.

Dan Metzger
Dr. Dan Metzger is a Clinical Professor of Pediatrics at the University of British Columbia, and a Pediatric Endocrinologist working on the Endocrinology & Diabetes Unit of BC Children’s Hospital. Dr. Metzger and his team, working with mental-health colleagues at BCCH and in the community as a “clinic without walls”, began seeing transgender kids in 1998. They have now seen nearly 400 kids, with one of the busiest clinics in Canada. In 2014, they published one of the first North American reports on the results of their experience caring for trans and gender-questioning children and youth.

Nikhil Pai
Nikhil Pai is an Assistant Professor in the Division of Pediatric Gastroenterology at McMaster Children’s Hospital. He completed his training in Pediatric Gastroenterology and Nutrition through Harvard Medical School and Boston Children’s Hospital. His postdoctoral research was through the Obesity, Metabolism and Nutrition Institute at Massachusetts General Hospital under Dr Lee Kaplan and Peter Turnbaugh, where he studied therapeutic manipulation of gut bacteria in obesity, and post-bariatric surgery mouse models. He is the Medical Lead of the Clinic Nutrition Service at McMaster Children’s Hospital and his research focuses on the role of fecal microbiota transplant for pediatric ulcerative colitis and Crohn’s disease. He loves talking to patients and colleagues about why poop is so fascinating, and doesn’t find it the least bit gross.
Mark R. Palmert  
Dr. Palmert is a Professor of Pediatrics and Physiology at the University of Toronto. Mark graduated from the Medical Scientist Training Program at Case Western Reserve University, Cleveland, Ohio, with a MD and PhD in 1992 and then completed his pediatrics and pediatric endocrinology training at the Children’s Hospital, Boston. Prior to moving to Toronto in 2007 to become Head of the Division of Endocrinology, Mark held staff positions in Boston and at the Rainbow Babies and Children’s Hospital in Cleveland. In 2017 Mark began a new role as Associate Chair of Pediatrics (Ambulatory Care) at SickKids.

Dina Panagiotopoulos  
Dr. Dina Panagiotopoulos is a Clinical Professor in the Department of Pediatrics, University of British Columbia and an endocrinologist at B.C. Children’s Hospital where she also serves as the Medical Director for both the Type 2 Diabetes/Insulin Resistance Clinic and the Provincial Mental Health Metabolic Program. Her research spans both forms (type 1 and type 2) of childhood diabetes. She is the clinical core leader of a research team in childhood autoimmunity at B.C. Children’s Hospital, where she has developed a provincial biobank/clinical registry for childhood type 1 diabetes. She also serves as the Vancouver affiliate principal investigator for TrialNet, a multicentre international research effort to understand the natural history of type 1 diabetes. In the area of type 2 diabetes, her research focus is on vulnerable and “at-risk” populations – including Aboriginal youth, and children with mental health challenges.

Sheila Pritchard  
Dr. Sheila Pritchard is a pediatric oncologist at BC Children’s Hospital in Vancouver and is the Director of the Late Effects program for BC. Sheila has a particular interest in the late effects of cancer treatment on fertility and the possibilities for fertility preservation for young oncology patients.

Stacey Urbach  
Dr. Stacey Urbach is a staff physician in the Division of Endocrinology at The Hospital for Sick Children in Toronto. She completed medical school at The University of Toronto and went on to do her pediatric residency and endocrine fellowship at SickKids. Dr. Urbach spent part of her fellowship training in Portland, Oregon where she completed a Master’s in Public Health. Her thesis examined the predictors of glucose control in children with Type 1 Diabetes. During her clinical training, Dr. Urbach had the opportunity to work with the members of the long term follow-up cancer care team at SickKids. Her area of interest lies is in the endocrine care of childhood cancer survivors and she has been instrumental in building the long term follow-up program which provides coordinated care for this vulnerable population.

John VanderMeulen  
Dr. VanderMeulen is a professor of pediatrics, McMaster University, and has been the division head of pediatric endocrinology at McMaster Children’s Hospital since 1989. He obtained his MD from the University of Toronto in 1984 and his PhD in 1985 from the department of medical biophysics, division of cell biology, at the University of Toronto. His doctoral research focus was in the field of molecular mechanisms underlying secretion by exocytosis. He trained in pediatric endocrinology at the Hospital for Sick Children in Toronto prior to undertaking post-doctoral training at Mcmaster University in the area of G-protein coupled signal transduction. His early research career was focused in the same area, receiving research funding from both the Medical Research Council of Canada and the Heart and Stroke Foundation of Canada. From a one-person division in 1989, the division of pediatric endocrinology has grown to 5 full-time and 1 part-time faculty. Among numerous other benefits, this has resulted in major improvements to his call schedule.
Conflict of Interest Disclosures

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

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- I hold investments in Amgen and Gilead Sciences
- I have an involvement in research sponsored by or participation in clinical studies concerning the use of the products from OPKO, Pfizer, and EMD Serono.
Oral Abstract 1
Diabetes Appointment Attendance around Transition to Adult Care: Type 2 vs Type 1 Diabetes

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Objective:
Youth-onset type 2 diabetes mellitus (T2D) carries a high burden of illness in early adulthood, yet there is a paucity of studies regarding transition in this population. The study’s aim is to describe the healthcare interactions 2 years before and 2 years after transition in those with T2D and type 1 diabetes (T1D).

Methods:
It is a retrospective study using clinical and administrative data from the Manitoba Centre for Health Policy, which enables linking of pediatric and adult data. The patient population includes Manitoban residents diagnosed with youth onset T2D (196 youth) and T1D (456) who transitioned to adult health care services between 2005 and 2013, with data collected 2 years before and 2 years after transition.

Results:
When compared directly, those with T2D were less likely to attend diabetes appointments compared to those with T1D both pre (p<0.01) and post (p<0.01) transition. A regression analysis revealed that those with T2D (OR:0.78 95%CI 0.69-0.89), less visits pre-transition (OR:1.1 95%CI 1.06-1.17), and males (OR:0.83 95%CI 0.74-0.92) were less likely to attend diabetes appointments after transition; age at diagnosis and age at transition were not significant variables. At the first diabetes appointment post-transition, those with T2D were less likely to see an internal medicine physician (OR:0.45 95%CI 0.27-0.74) and more likely to see a rural family physician (OR:3.40 95%CI 1.94-5.96) than an urban family physician, compared to those with T1D. Hospital admission rates were higher both pre (p<0.01) and post (p<0.01) transition in those with T2D vs those with T1D. Those with T2D were more likely to be admitted for a reason other than diabetes both pre (OR:18.30 95%CI 8.22-40.53) and post (OR:5.61 95%CI 3.04-10.34) than those with T1D.

Conclusion:
This study revealed that youth with T2D attend less diabetes appointments post compared to pre transition, and less appointments both pre and post transition than youth with T1D. In addition, males are at highest risk of poor attendance post transition. Those with T2D are more likely to be seen in a rural setting and by general practitioners, and have higher rates of hospital admissions. Understanding the characteristics of the population is key component to developing a successful transition service for youth with T2D.
Oral Abstract 2
Adiposity is Associated with Early Vascular Damage in Children with Type 1 Diabetes

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Objectives:
Cardiovascular disease is a major complication of type 1 diabetes (T1D) traditionally thought to manifest in adulthood. Recent evidence suggests that cardiovascular damage is present in children with T1D. The objectives of this study were to investigate the effect of T1D duration and adiposity on carotid intima media thickness (cIMT), a subclinical indicator of atherosclerosis, in children with T1D.

Methods:
A cross-sectional pilot study of children with T1D aged 9 to 15 years (n=45) were recruited into 2 groups; short duration (6 months-2 years); and long duration (>5 years). Glycemic control (A1C), serum HDL cholesterol (HDL), z-scores for adiposity markers (zBMI; waist circumference, zWC; and waist/height ratio, zWHtR), systolic blood pressure (zSBP) and diastolic (zDBP) were collected and relationships to T1D duration and cIMT were determined. A subset of children (n=27), also had 24 hour Ambulatory blood pressure monitoring (ABPM) performed.

Results:
Of the 45 children recruited, 60% were female and 49% had T1D for short duration. The mean age, mean A1C, mean zBMI was 13.97 years, 7.8%, 0.18 in the short duration group and 13.84 years, 7.67% and 0.65 in the long duration group. Markers of adiposity were all associated with cIMT in unadjusted models [zWC (p=0.009), zWHtR (p=0.024), zBMI (p=0.007)] and remained when adjusted for disease duration and A1C [zWC (p=0.009), zWHtR (p=0.036), zBMI (p=0.008)]. Disease duration and A1C were not associated with cIMT. Long duration of disease was associated with increased zBMI (p=0.059). There was no effect of disease duration on zWC, zWHtR, A1C, HDL, zSBP, and zDBP. Abnormalities in the ABPM analysis were observed in 65% of the subjects (n=17); 8 in the short duration group and 9 in the long duration group; but only 3 of these had an abnormal BP on once off clinic measurement. Abnormal ABPM was not found to be associated with zBMI, zWC or zWHtR and only 3 subjects with abnormal ABPM were overweight or obese.

Conclusion:
Adiposity is associated with cIMT in children and adolescents with T1D, independent of disease duration and A1C. These data highlight the importance of addressing adiposity in children with T1D to prevent cardiovascular complications.
Oral Abstract 3
Evaluating the Low-Dose ACTH Stimulation Test: Ideal Timings for Cortisol Measurement

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Background:
The low-dose ACTH stimulation test (LDST) is used for the diagnosis of central adrenal insufficiency (AI). However, protocols vary significantly between institutions, with some testing only 30 minutes post administration of Cosyntropin and others, multiple times between 0 and 60 minutes.

Objective:
The primary objective of this study is to determine the optimal times to draw serum cortisol levels in children undergoing LDST. The secondary objective is to determine if baseline cortisol levels drawn between 0800-0900 accurately predict the LDST result.

Methods:
A retrospective chart review was performed. All patients aged 3 months to 18 years who had undergone a LDST at the Children’s Hospital of Eastern Ontario between February 1, 2014 and September 30, 2017 were included. As per hospital protocol, cortisol levels were drawn at 0, 15, 30, and 60 minutes post administration of Cosyntropin 1 mcg. A cortisol value > 499 nmol/L was used to define adrenal sufficiency.

Results:
221 patients met inclusion criteria. The mean age was 9.7 years and 32% were female. 19% of patients had a peak cortisol level at 15 minutes, 67% peaked at 30 minutes and 14% at 60 minutes. Of the 123 patients who passed the LDST, 9 patients (7.3%) would have been misdiagnosed as having AI if the 15 minute cortisol test was discontinued. Discontinuing the 60 minute test would have misdiagnosed 3 patients (2.4%). Of the 125 patients who had normal baseline cortisol levels (>184 nmol/L) drawn between 0800-0900, only 57.6% had a peak level > 499 nmol/L. However, 91.2% of patients with a baseline cortisol level >184 nmol/L had a peak level > 399 nmol/L.

Conclusion:
Our results suggest that while the majority of patients peak at 30 minutes post administration of Cosyntropin, cortisol levels drawn at 15 and 60 minutes may reduce the risk of a false positive test. Misdiagnosis of AI can be associated with a significant burden for patients, families and the healthcare system. Therefore, consideration for testing cortisol levels at 15 and 60 minutes during a LDST should be made. Further, the immunoassay-specific threshold of 185 nmol/L for morning cortisol does not predict adrenal sufficiency, although it may indicate milder AI.
Oral Abstract 4
Evaluation of a 24-Hour Telephone Paging Service: Are We Preventing Emergency Department Visits for Pediatric Diabetes and Endocrine Patients?

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Introduction:
Studies have shown that 24-hour paging services can prevent emergency department (ED) visits for pediatric diabetes patients, but no studies have evaluated outcomes of pediatric endocrine patients. The Endocrinology and Diabetes Unit (EDU) at British Columbia Children’s Hospital (BCCH) is the only tertiary level pediatric center in British Columbia (BC) and provides a 24-hour paging service for parents and patients.

Objective:
To determine if a physician staffed 24-hour paging service prevents ED visits for pediatric endocrine and diabetes patients.

Methods:
In this 12-month prospective observational study, we recruited endocrine and diabetes patients whose caregivers used the EDU call service and met pre-defined set of criteria for a ‘preventable’ ED visit in the absence of access to an on-call line (i.e. diabetes patients with persistent hypoglycemia, or endocrine patients requiring stress dose steroids). Callers that met criteria were invited to complete a telephone survey that collected demographic data and ED visits within 72 hours of the call. Data were analyzed using descriptive statistics.

Results:
Of the 1238 calls made to the EDU paging service during the study period, 199 met study inclusion criteria (83% diabetes, 17% endocrine). Recruitment rate was 33% (n=66), of which 44 callers completed the survey. Most calls were from diabetes patients (84%) who needed advice for non-routine insulin dose adjustment, illness management, or persistent/severe hypoglycemia (69%); endocrine callers were patients with adrenal insufficiency, hypoglycemia disorders or hypocalcemia, needing urgent illness management advice (57%) or had an adverse drug reaction (43%). The majority of callers were located within a one hour drive of BCCH (61%), Canadian born (81%) and had some post-secondary level education (84%). Nearly 100% of callers were satisfied/very satisfied with the paging service, and only 2/44 (4.5%) patients visited an ED within 72 hours of making the call.

Conclusions:
This is the first Canadian study showing that a 24-hour paging service used by pediatric endocrine and diabetes patients with an urgent/emergent medical issue prevented 95.5% of potential ED visits. Extrapolating this to all calls that met study criteria (N=199), 190 ED visits may have been prevented over the 12-month period.
Oral Abstract 5
The Role GLP-2 in Development of Experimental Non-Alcoholic Fatty Liver Disease

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Non-alcoholic fatty liver disease (NAFLD) is a common complication of obesity with potential to evolve to liver inflammation, cirrhosis and malignancy. Hepatic lipotoxicity is also a driving force in progression from obesity to the metabolic syndrome. Glucagon-like peptides (GLP-1 and GLP-2) secreted from entero-endocrine L-cells control prandial fat absorption, potentially modifying liver fat and inflammation. Although GLP-1 has been extensively studied, little is known about how or whether GLP-2 action modifies the development of NAFLD.

Methods:
We study whether sustained activation of GLP-2R signaling in mice fed a high fat, high fructose cholesterol-enriched diet (HFCD) modifies the development of steatohepatitis. Forty wildtype mice were placed on either a HFCD or a control diet for 16 weeks, then treated with the GLP-2 analog (h[Gly(2)]GLP-2) or Saline, for 11 days. Experimental endpoints included hepatic and plasma profiles of inflammation, glucose tolerance and histological, gene expression and biochemical analyses of liver samples. We studied GLP-2R expression in cell subsets ex-vivo through fractionation and differential centrifugation of hepatic cell types, to localize GLP-2R expression within the liver.

Results:
Mice treated with h[Gly(2)]GLP-2 had significant increases in intestinal mass and gallbladder volume, both known effects of GLP-2 activity, confirming efficacy of treatment. Mice on HFCD had significantly higher weight gain, glucose intolerance, hypertransaminasemia, liver adiposity, and total cholesterol. HFCD-fed mice also exhibited elevated hepatic expression of mRNA transcripts encoding inflammatory genes and cytokines. No differences in body weight, plasma lipids, hepatic lipid content and transcript levels of key genes controlling hepatic lipid metabolism were detected following treatment with h[Gly(2)]GLP-2. Increased cholesterol content in HDL particles from treated mice was observed by FPLC analysis. Furthermore, an extensive profile of plasma and liver cytokines did not reveal any modulation of hepatic or systemic inflammation as a result of treatment. In contrast to previously published localization of GLP-2R expression, ex-vivo liver fractionation studies revealed that the canonical GLP-2 receptor is not expressed in primary hepatocytes, but localized to the fraction of non-parenchymal liver cells.

Conclusion:
In a mouse model of diet-induced NAFLD, treatment with h[Gly(2)]GLP-2 does not alter disease manifestations, likely reflecting the absence of hepatocyte GLP-2R expression.
Oral Abstract 6
Chimeric 46XX/46XY Causes Ovotesticular Disorder of Sexual Development

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Introduction:
Ovotesticular disorder of sexual development (OT-DSD) is a rare cause of DSD. Most cases have a 46XX karyotype whereas 46XY karyotype is isolated in 15% of cases. The remainder of cases, 10-20%, result from mosaics and chimeras. We present a case of a 16 year old male, healthy and developmentally normal, who has chimeric karyotype 46XX/46XY and OT-DSD.

Case Presentation:
Our patient was born at term after an uncomplicated pregnancy. Examination at birth revealed ambiguous genitalia, confirmed by Endocrinology at 2 weeks of age. The infant had a 2.8 cm phallus, hypospadias with perineal urethra, significant chordee, posterior labioscrotal fusion, a palpable right inguinal gonad, and no palpable left gonad. Hormonal testing included: LH 3.4 U/L, FSH 3.3 U/L, testosterone 3 nmol/L, and 17-OHP 4.3 nmol/L. There was chimerism in all samples: right buccal smear 19% 46XY/81% 46XX, left buccal smear 4% 46XY/96% 46XX, and lymphocytes 11% 46XY/89% 46XX. Fertilization of the second polar body is the proposed mechanism of chimerism. In consultation with Endocrinology, Urology, and Medical Genetics, our patient was reared as a male. His right gonad was grossly abnormal, but biopsy demonstrated normal testis. Laparotomy at seven months resulted in left gonadectomy and removal of Mullerian structures. Cytogenetics of the left gonad revealed two cells lines (46XX/46XY) and pathology confirmed ovotestis. Two staged hypospadias repair resulted in good cosmetic result and function. Puberty began spontaneously and initially progressed normally. He felt comfortable with the male gender assignment. At 13 9/12-years, there was evidence of gonadal dysfunction with LH 5.4 IU/L, FSH 10.2 IU/L, and testosterone 1.52 nmol/L; he was started on Delatestryl 80 mg IM every 3 weeks. Initial semen analysis showed no sperm. Delatestryl was discontinued and he began hCG injections. Sperm retrieval was unsuccessful. He required surgery for bilateral gynecomastia. At 15 4/12-years, he was started on Delatestryl 100 mg IM every three weeks, given low testosterone 0.9 nmol/L. Testicular ultrasounds have been unremarkable. Presently, he continues with testosterone replacement therapy, titrated based on interval hormonal testing.

Conclusion:
Individuals with OT-DSD require lifelong multidisciplinary care in regards to puberty, fertility, and screening for tumors.
Oral Abstract 7
A Rare Cause of Gross Hematuria

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Background:
Isolated gross hematuria is a rare side effect of arginine with unknown mechanism of action, and has been previously reported in four children. We add to this small body of literature with four additional cases of arginine-related hematuria following combined clonidine-arginine growth hormone stimulation testing.

Cases:
Four pediatric patients presented with a first episode of transient asymptomatic gross hematuria following combined clonidine and arginine growth hormone stimulation testing. All four patients were males, with ages ranging from 11 to 15 years. Common causes of gross hematuria were excluded, such as post-infectious glomerulonephritis, urinary tract infections, and nephrolithiasis. Etiology was attributed to arginine used during clinical testing. Mild hypotension during stimulation testing was present in all of our patients, as well as the previously reported cases, but this is a common side effect of clonidine. Hematuria was unrelated to growth hormone deficiency, as three of our patients had normal growth hormone peaks. All cases of arginine-related hematuria resolved spontaneously within one month. The four cases that we describe presented within a two-year window at our institution. All children were treated with medications of different lot numbers, and there were no recent changes to medication brand, product formulation or drug constitution at our institution. Hematuria has not been reported in patients receiving arginine for other indications, such as metabolic disorders.

Conclusions:
Arginine should be suspected as the etiology for transient gross hematuria in the context of recent exposure, and clinicians may consider minimal additional investigations in these patients. Further studies are required to define the prevalence and mechanism of action of arginine-related hematuria.
Oral Abstract 8
PMM2 Gene Mutation Found in a Child with Congenital Hyperinsulinism and Polycystic Kidney Disease

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The co-occurrence of hyperinsulinism (HI) and polycystic kidney disease, two rare diseases, was previously thought to have no genetic link. We present the case of a 2-year-old male who was diagnosed with hyperinsulinism (HI) and polycystic kidney disease. HI was diagnosed in the neonatal period on day of life one with severe hypoglycemia, which was responsive to Diazoxide therapy. Patient required up to 14.5 mg/kg/day of Diazoxide to achieve normoglycemia. Testing for common HI gene mutations (ABCC8, KCNJ11) was negative. Polycystic kidney disease was diagnosed on antenatal ultrasound. Genetic testing for ADPKD and ARPKD was negative. Patient’s course was later complicated by significant progression of his renal disease with stage 4 chronic kidney disease (CKD) and renal cystic enlargement, measuring up to 19 x 12 cm. Due to these complications, he required bilateral nephrectomy and is undergoing treatment with dialysis. Following treatment with nephrectomy, his Diazoxide requirement decreased to 8 mg/kg/day. Prior to 2017, there was no known genetic link between the two rare disorders of HI and polycystic kidney disease (HIPKD). In 2017 Cabezas et al., published a report of 17 patients with the co-occurrence of hyperinsulinism and polycystic kidney disease. Genetic testing in these patients for ABCC8, KCNJ11 and ARPKD were negative. All patients in this study were found to have a promoter mutation in the novel gene Phosphomannomutase (PMM2), with either a homozygous or compound heterozygous state. The PMM2 gene encodes a key enzyme involved in N-glycosylation. Mutation in this gene has shown to cause abnormal glycosylation, which has been associated with polycystic kidney disease. The effect of PMM2 mutation on insulin secretion was studied in murine models, which has shown to result in an increase in pancreatic B-cell insulin secretion (Cabezas et al., 2017). After contacting a research team in Exeter in the United Kingdom, we submitted our patient’s blood work for genetic testing. Our patient was found to be compound heterozygous for the PMM2 missense mutation (p.Arg141His) and the PMM2 promoter mutation (c.-167G>T). This brings to light a novel and previously unidentified mechanism that PMM2 gene mutations can lead to both HI and polycystic kidney disease as seen in our patient.
Oral Abstract 9

Increasing Trends of Diabetic Ketoacidosis in Quebec: A Population-Based Study

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Background:
Diabetic ketoacidosis (DKA) at type 1 diabetes (T1D) diagnosis is a largely preventable acute complication. The frequency of DKA at diabetes onset varies significantly worldwide (12.8-80%)(1). Recent Canadian data on the trends of DKA prevalence at diabetes onset is unknown.

Objectives and Methods:
We aimed to determine the temporal changes in DKA prevalence at diabetes diagnosis in Quebec. We conducted a population-based cohort study of children (1–17 years) living in Quebec, diagnosed with diabetes between 2001 and 2014. We used multiple health administrative data of individuals linked by a unique encoded identifier across databases that contains information on patient demographics, hospitalizations (MED-ECHO) and physician remunerated services (RAMQ). We identified our cohort within administrative data using a definition validated in Canadian children(2). We used multivariate Poisson regression analysis with robust error variance to determine the trend in DKA prevalence over time.

Results:
DKA was present in 1471 of 5741 new cases of diabetes during the study period (26%). Children ages 5–11 years had the highest frequency of DKA (30%), followed by those 1–4 years (29%) and 12–17 years (20%) over the study period. Overall, the age- and sex-standardized rate of DKA increased from 22% (95% confidence interval (CI): 17%, 26%) in 2001 to 30% (95% CI: 24%, 36%) in 2014. In the multivariate Poisson regression analysis, there was an increased trend in the rates of DKA at diagnosis by 2.0 % per year (95% CI 0.8%, 3.1%). Increase in DKA trends was the most significant for the 5–11 y.o age-group with an increase of 2.7% per year (95% CI: 1.0%,4.5%), followed by the 12–17 y.o. with an increase of 2.2% per year (CI: 0.2%, 5.1%).

Discussion:
Despite an increasing incidence of T1D, the prevalence of DKA at diabetes onset in Quebec has increased between 2001 and 2014. This differs from other countries with universal health care systems, which have reported either decreasing or stable DKA trends. Our findings are concerning and warrants future research in understanding the drivers of this increasing DKA risk both at a patient level as well as a health systems level. REFERENCES: 1. Usher-Smith JA, Thompson M, Ercole A, Walter FM. Variation between countries in the frequency of diabetic ketoacidosis at first presentation of type 1 diabetes in children: a systematic review. Diabetologia. 2012;55(11):2878-2894. 2. Dart AB, Martens PJ, Sellers EA, Brownell MD, Rigatto C, Dean HJ. Validation of a Pediatric Diabetes Case Definition Using Administrative Health Data in Manitoba, Canada. Diabetes Care. 2011;34(4).
Oral Abstract 10
Bone Health and Vitamin D Status in Transgender Adolescents Starting GnRH Agonists

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Secondary sex characteristics are distressing for adolescents with gender dysphoria. Puberty suppressing therapies can be offered as of Tanner 2 (World Professional Association for Transgender Health). However, puberty is important for bone health as sex steroids augment bone mass. In young transgender adults, gonadotrophin-releasing hormone agonists (GnRHa) were shown to decrease Bone Mineral Density (BMD) z-scores. Evidence in adolescents is scant.

Methods:
A retrospective chart review (January 1, 2006 to April 30, 2017) examined bone health in youth with gender dysphoria followed in CHEO’s Diversity Clinic. We collected data on natal and affirmed sex, pre and post GnRHa BMD profiles (Lumbar Spine (LS), Left Total Hip (Lt-TH), and Total Body excluding Head (TB-H) z-scores), and 25OHD levels.

Results:
The study included 173 youth (mean age 15.7±1.9 (SD) years): 120 (69.4%) trans males, 51 (29.5%) trans females, and 2 (1.2%) non-binary. Baseline BMD z-scores were LS: -0.2±1.2, 6% <-2, Lt-TH: -0.1±1.2, 4.8% <-2, and TB-H: 0.3±1.2, 2.4% <-2. Natal male and females had different baseline BMD z-scores at Lt-TH: -0.4±1.4, 0.1±1.1 (P= 0.006) and TB-H: 0.0±1.4, TB-H: 0.4±1.1 (P=0.02). Baseline 25OHD level was 49.4±20.3 nmol/L, 9.5% vitamin D deficient (< 25 nmol/L), and 81.6% insufficient (25-74 nmol/L). All youth were advised to start vitamin D 1000-2000 IU daily. There was no association between baseline 25OHD levels and BMD. Mean difference between pre- and post-GnRHa BMD LS z-scores was -0.48 [95% CI: -0.63, -0.33] (P < 0.001) (n=142). BMD 1 year ± 90 days on GnRHa, not yet on CSH, was -0.5 ± 1.4, 16% <-2 (n=31); BMD was not associated with 25OHD levels in the subgroup (n=18) with available 25OHD.

Conclusion:
Trans female youth had significantly lower BMD z-scores than trans males. 25OHD levels were low in most youth with the majority having vitamin D insufficiency. GnRHa therapy was associated with decreased BMD z-score at LS level. Longer follow-up will allow examination of age and Tanner stage at initiation of GnRHa and of cross-sex hormones on bone health. It seems prudent to include baseline and follow up BMDs and 25OHD monitoring in the routine care of transgender adolescents on GnRHa therapy.
Oral Abstract 11
Type 1 Diabetes Outcomes: Does Distance to Clinic Matter?

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Objectives:
1. To compare characteristics of patients travelling varying durations to a tertiary type 1 diabetes (T1D) clinic at British Columbia Children's Hospital (BCCH) and patients receiving community-based care. 2. To describe the relationship between travel time to clinic and clinical and patient-reported outcomes. 3. To describe the relationship between tertiary vs. community-based care and key clinical and patient-reported outcomes.

Methods:
Participants were recruited from T1D clinics across British Columbia. Clinical data were collected by chart review and parent and patient reported outcomes were collected by online surveys (age-permitting), including the Adherence in Diabetes Questionnaire (ADQ). Clinic type was categorized based on self-reported primary diabetes physician. For patients attending clinic at BCCH, travel time to clinic was categorized as <1 hour (h), 1-2h, or >2h, based on parent report. Descriptive statistics, linear and logistic regression were used, as applicable.

Results:
There were 189 participants (BCCH <1h n=71; BCCH 1-2h n=25; BCCH >2h n=20, community n=73). Baseline characteristics (age, duration of T1D, household income) were similar across all groups. In the BCCH groups, mean number of visits per year ranged from 2.05-2.23, compared to 3.26 visits/year for the community group. Insulin regimens were similar across the BCCH groups, whereas in the community, a higher proportion of children were using an insulin pump. ADQ scores were similar among all groups. Mean (SD) A1Cs were as follows: BCCH <1h 7.88% (1.01), BCCH 1-2h 8.16% (1.13), BCCH >2h 8.64% (1.18), community 8.15% (0.86). Adjusted mean difference in A1C was +0.65% (95% CI 0.15, 1.15) and +0.52% (95% CI 0.02, 1.02) for the BCCH >2h group compared to BCCH <1h group and community group, respectively. Participants traveling >1h to BCCH were more likely to report barriers to attending clinic, most commonly that the clinic was too far away.

Conclusions:
Despite similar patient characteristics and self-reported treatment adherence, children travelling >2h to attend T1D clinic at BCCH had significantly higher mean A1Cs compared to those travelling <1h to BCCH and those receiving community care. Therefore, access to care closer to home may positively benefit glycemic control in children with T1D and reduce barriers to attending clinic.
Oral Abstract 12
The Identification of Early Childhood Cardiometabolic Risk in Infants of Mothers with Gestational Diabetes

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Background:
Gestational diabetes (GDM) results in exposure to hyperglycemia in utero which has been associated with increased adiposity and diabetes in the offspring later in life. The timing and evolution of these changes during early childhood are not well understood.

Objective:
To evaluate the impact of exposure to GDM in utero on early signs of cardio-metabolic risk (CMR) in childhood

Methods:
A prospective, observational cohort of infants born to mothers with and without GDM underwent longitudinal anthropometric and metabolic characterization between birth and 3 years of age. Associations between anthropometric and biochemical measures of CMR including insulin resistance (HOMA-IR) were evaluated over time.

Results:
A total of 91 infants (40 GDM; 51 non-GDM) had fasting blood work and anthropometrics at 3 years of age. At baseline, infants born to mothers with GDM were born earlier (38.4±1.3 vs. 38.95±1.3 weeks gestation, p<0.0001), at a lower birth weight (3302±389 vs. 3464±441grams, p<0.0001) and were less likely to be exclusively breastfed (39.73% vs. 51.95%, p=0.006). After early infancy, there were no differences in anthropometric or metabolic measures between the two groups to age 3 years. On univariate regression, HOMA-IR at 3 years was positively correlated with weight for length z score, waist circumference and weight change in non-GDM exposed infants only (p<0.05). In contrast, maternal insulin sensitivity and adiponectin levels were inversely correlated with offspring HOMA-IR only in infants exposed to GDM (p<0.005). On multiple regression analysis, adjusting for maternal body mass index, 1 kg of offspring weight gain in early childhood predicted a 26% higher HOMA-IR (p=0.002) and a 1 unit increase in maternal insulin sensitivity predicted 7% lower HOMA-IR(p=0.02) in the overall group of infants (model r2=0.25).

Conclusions:
At 3 years of age, insulin resistance in the overall cohort is determined by post-natal weight gain in the offspring and intrapartum maternal insulin sensitivity. Future analysis will evaluate the determinants of HOMA-IR in both the GDM and non-GDM groups separately.
Poster Abstract 1
Genotype Phenotype Correlation in Children and Adolescents with Osteogenesis Imperfecta: A Retrospective Cohort Study

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Background:
Osteogenesis Imperfecta (OI) is a genetic bone condition characterized by varying degrees of bone fragility and deformity caused by mutations in genes involved in type 1 collagen production, function and post translational modification; as well as osteoblast differentiation and function. This study aims to characterize OI phenotype and outcomes by genotype grouping according to gene function.

Methods:
Patients with OI confirmed by genetic diagnosis and followed at the Hospital of Sick Children between 1998 and 2017 were retrospectively analyzed. Patients were classified according to their genotype into 4 groups; G1: quantitative COL1A1 mutations; G2: qualitative COL1A1 and COL1A2 mutations; G3: Collagen post translational defects (BMP1, FKBP10, and LEPRE1); G4: altered osteoblast differentiation and function (IFITM5, PLS3, SERPINF1, WNT1 and XYLIT2). Data were collected at presentation and during follow up for clinical and biochemical features to examine phenotypic variation among groups.

Results:
A preliminary analysis was undertaken of 40 patients, with 30% of the cohort in G1, 19% in G2, 7% in G3 and 14% in G4. The median duration of follow up was 7 years (range 1-17 years). While age, sex and BMI z-score at presentation were not significantly different between groups, height (p=0.008) and weight z-scores (p=0.045) differed by genotype, as did the presence of blue sclera (p=0.003) and vertebral fractures (p = 0.015). Bone specific alkaline phosphatase z-scores were significantly higher in G1 compared to the other groups (p=0.032), but C-telopeptide z-score levels were similar. On follow up, groups differed by severity of scoliosis. There was also a trend of difference in number of vertebral body fractures.

Conclusions:
In this preliminary analysis, while there is a significant phenotypic overlap by OI genotype, there are some observable differences in clinical presentation and evolving phenotypic features. Extending the analysis to a larger number of patients and treatment data may further characterize the impact of treatment on the evolving phenotypic features.
Poster Abstract 2
Pamidronate First-Line Treatment of Hypercalcaemia in Neonatal Subcutaneous Fat Necrosis

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Introduction:
Subcutaneous fat necrosis (SCFN) is a granulomatous panniculitis precipitated by an ischaemic, hypoxic or hypothermic insult to adipose tissue after fetal or perinatal distress or therapeutic hypothermia. Hypercalcaemia can complicate SCFN which may be associated with persistent nephrocalcinosis. Intravenous bisphosphonates, which reduce osteoclast activity and thus bone resorption, are described to successfully treat hypercalcaemia in neonatal SCFN but only once established treatments fail. Two cases are presented demonstrating the novel, successful first-line use of hydration and intravenous pamidronate for this indication.

Literature Review:
Nine cases of hypercalcaemia secondary to SCFN treated with intravenous pamidronate were identified. Seven had nephrocalcinosis. First line treatment included intravenous fluid, low calcium and vitamin D diet, use of loop diuretics (Furosemide) and glucocorticoids. Subsequent use of pamidronate, ranging from 0.25-1 mg/kg/day for 1-4 consecutive days, resulted in safe and effective resolution of hypercalcaemia in 1-7 days.

Cases:
Female born at term, 4.1 kg, by vaginal delivery complicated by meconium stained liquor and shoulder dystocia. After a characteristic history and examination, SCFN and hypercalcaemia were diagnosed at 28 days with calcium 3.47, phosphate 2.25, and PTH <5 ng/L (12-78). Saline hyperhydration from day one of admission, followed by intravenous pamidronate (0.5 mg/kg) resulted in a response within 12 hours and normalization of calcium on day 3 of admission. The renal ultrasound is pending. Male born at term, 4.73 kg, by emergency caesarean section, with poor tone and developed seizures. SCFN was diagnosed at 2 weeks and admitted at 19 days with hypercalcaemia. Calcium 3.02 and PTH <5 ng/L. Saline hyperhydration from day one of admission. Pamidronate 0.25 mg/kg on day two and calcium normalised on day three. The renal ultrasound was normal.

Conclusion:
Previous evidence supports a multi-factorial aetiology of the hypercalcemia including increased expression of 1-alpha-hydroxylase from activated macrophages and increased bone turnover. Previous paediatric case studies describe use of hydration followed by combination therapy, including furosemide and glucocorticoids, however these agents may exacerbate the risk of nephrocalcinosis. Pamidronate treatment is without this risk and more sustained benefit. These cases demonstrate safe and successful first-line use of pamidronate for hypercalcaemia in neonatal SCFN.
**Poster Abstract 3**  
**Evaluation of Obesity and Body Composition in a Genetically Diverse Cohort of Children with Osteogenesis Imperfecta**

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**Background:**  
Osteogenesis imperfecta (OI) is a condition characterized by bone fragility and short stature arising from mutations in genes involved in type 1 collagen synthesis, metabolism or altered osteoblast function. A small number of studies have described increased prevalence of obesity in OI, particularly among those with OI Type-III. Little is known, however, about the longitudinal changes in body composition in children with OI, in particular those with rare, autosomal recessive forms.

**Objectives:**  
To describe the prevalence of obesity in a genetically diverse cohort of children with OI and to understand the differences in body composition according to disease severity, mobility and genotype.

**Methods:** Retrospective review of 57 patients with OI followed at the Hospital for Sick Children from 1998 to 2017. Anthropometric and body composition data using dual energy X-ray absorptiometry were collected from the most recent clinic visit and analyzed cross-sectionally. Patients were classified according to disease severity, ambulation status and genetic mutation.

**Results:**  
The prevalence of overweight and obesity was 43.9% in children 5 to 18 years. Increasing age correlated with higher BMI and percent fat mass z scores (PFM) (r²=0.05, r²=0.17; p<0.05). Height, weight and lean mass index (LMI) z scores in the cohort were lower compared to the normal population (-1.95±3.02, -0.73±1.43 and -1.09±1.31, p<0.01). Although PFM z scores were higher (0.43±1.17, p=0.02) there was no difference in BMI z scores (0.33±1.40 p=0.08). Within the OI cohort, PFM, LMI and BMI z score were higher among those patients with more significant disease severity and non-ambulatory status (p<0.05) but no differences were observed between genotypes. While, patients with non-COL1A1/COL1A2 mutations had lower height and weight z scores (p<0.05), there was no differences in their body composition measures when compared with patients with COL1A1/COL1A2 mutations.

**Conclusions:**  
Overweight and obesity is more prevalent among children with OI compared to the Canadian population, with increasing BMI and PFM z scores over time. Higher disease severity and physical impairment are more significantly associated with adiposity than genotype. Percent fat mass z scores, rather than BMI, may be a more accurate measure of adiposity in children with OI.
Poster Abstract 4
Psychostimulants and Calcitriol Interactions: Prescribers Beware!

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We report two cases of hypercalcemia related to the combination of calcitriol and psychostimulants. No interaction between these two medications has yet been reported. Patient 1 was a 7.8 years old girl when diagnosed with hypothyroidism (negative thyroid antibodies) and treated with levothyroxine. She started methylphenidate at age 8.7 years for attention disorder (taken only during school days). At age 11.7y, she was admitted for hypocalcemic seizures (tCa: 1.48 mmol/l, iCa 0.70 mmol/l), hyperphosphatemia (2.9 mmol/l) and elevated PTH (45.7 pmol/l) leading to PTH resistance diagnosis. She was started on calcitriol therapy. At age 12.3y, two months after starting the school year and the medication with methylphenidate, she presented with asthenia, polyuria, and vomiting. Lab-tests revealed a high serum calcium (3.16 mmol/l) associated with an acute renal failure (serum creatinine:150 µmol/l), normophosphatemia (1.52 mmol/l), suppressed PTH (0.9 pmol/l) and low urinary calcium/creatinine ratio (0.45 mmol/mmol). 25(OH) vitamin D and TSH levels were normal, as well as 1.25OHD level (136 nmol/l). Calcitriol and methylphenidate were stopped. She was treated with intravenous hyperhydration, which gradually normalized calcium balance and renal function.

Patient 2 was a 11 years old girl known for a thyroid papillary carcinoma. She was substituted with levothyroxine after total thyroidectomy and with calcitriol for postoperative hypoparathyroidism. At age 14.8y, she started lisdexamphetamine 5 days a week for attention disorder. Six months after initiation she presented with vomiting, asthenia and tachycardia. Lab-tests showed hypercalcemia (tCa: 2.94 mmol/l) without renal failure. Calcitriol and psychostimulant were stopped to normalize serum calcium levels. In both cases, reinstatement of previous doses of calcitriol without psychostimulant did not reveal recurrence of hypercalcemia. Potential pathophysiological mechanism of this adverse drug reaction: Calcitriol is a potent hypercalcemic hormone increasing the calcium renal reabsorption. Psychostimulants are amphetamine derivatives. Extracellular volume being a direct determinant of calcium renal tubular reabsorption, their vasoconstrictor effect on the renal afferent artery may decrease glomerular filtration and increase calcium tubular reabsorption leading to hypercalcemia. These are the first case reports of adverse drug reactions with this drug combination. Prescribers may need to monitor serum calcium levels while giving the psychostimulants-calcitriol combination to their patients.
Poster Abstract 5
Prevention of Post-Thyroidectomy Hypocalcaemia in Children and Adolescents

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Background:
Hypocalcemia due to transient hypoparathyroidism from surgical manipulation is the most common complication of total thyroidectomy. The incidence of postoperative hypocalcemia is higher in the pediatric population than in adults, with potential for increased morbidity and treatment cost. The objective of this study was to review the pediatric literature to develop evidence-based guidance on monitoring and prevention of this complication.

Methods:
Literature search focused on the English pediatric (≤18 years of age) published articles using PubMed and Medline. Evidence relating to the following issues was reviewed: 1. Incidence of hypocalcemia secondary to thyroidectomy 2. Risk factors for developing hypocalcemia and 3. Strategies to prevent hypocalcemia. Adult literature was reviewed if no pediatric studies were found.

Results:
20 pediatric studies reported on the prevalence and predictive factors of hypocalcemia. On average, 30 to 40% of children developed parathyroid dysfunction and hypocalcemia after total thyroidectomy, the majority being transient. Risk factors for hypocalcemia included: manipulation of parathyroid glands, lymph node or central dissection, hyperthyroidism, young age, surgical inexperience and low preoperative serum 25-hydroxyvitamin D. Only one pediatric study examined the role of perioperative intact PTH (iPTH) in predicting postoperative hypocalcemia. The sensitivity and specificity of using serum iPTH cut-off of 14-16 pg/mL (1.48-1.70 pmol/L) to predict hypocalcemia was 80% and 100% respectively at 5-minutes postoperatively. Other strategies reported only in the adult literature included: prediction of hypocalcemia through serial calcium measurements; and prevention of hypocalcemia through prophylactic pre- and/or postoperative 1,25(OH)2-hydroxyvitamin D replacement.

Conclusion:
There currently exists a dearth of pediatric-specific research studies to guide prediction and prevention of hypocalcemia secondary to thyroidectomy. Treatment and monitoring strategies vary widely and appears largely across pediatric endocrinologists and surgeons. Supported by this literature review, we have developed a pathway using preventative administration of 1,25(OH)2-hydroxyvitamin D in order to prevent hypocalcemia while minimizing the need for testing, treatment and follow-up in children undergoing thyroidectomy.
Poster Abstract 6
Variability in Management of Diabetic Ketoacidosis in the Pediatric Population

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Although many pediatric Diabetic Ketoacidosis (DKA) management guidelines are available at tertiary care centres, there is often a lack of familiarity with them in some community hospitals, leading to management inconsistent with evidence-based clinical practice. Our objectives are to review the management of pediatric patients presenting in DKA, identify those that received initial treatment not consistent with current guidelines, and track associated complications. In this retrospective chart review, 47 charts of pediatric patients admitted to our institution (including patients transferred from peripheral hospitals) from January, 2012 to July, 2017 were identified by ICD-10 codes containing “ketoacidosis”. Primary outcome measures for inadequate management included major risk factors for cerebral edema - IV insulin boluses, IV bicarbonate, hypo-osmolar IV fluid administration and non-NPO status - as well as inappropriate IV fluid boluses >10mL/kg, insulin infusion initiated within the first hour after IV fluids were initiated, and inadequate potassium replacement. At least one area of inadequate management occurred at a rate of 74.5% with no significant difference between community hospitals and our tertiary centre. The most common parameter was inadequate replacement of potassium (44.7%), followed by receiving an IV fluid bolus >10mL/kg (40.4%). Twenty-three percent of patients received inadequate management that included a treatment-related major risk factor for cerebral edema, with the risk being significantly higher at outside centres (12.0% compared to 36.4% at outside centres; p=0.049). The most common complication was hypoglycemia (BG<4.0mM; 14.9%) and there were no cases of cerebral edema or patients that received a CT head. Though the mean time to insulin infusion initiation was not significantly different between our and outside centres, there was a statistically significant difference in time to correction of acidosis, with correction of DKA happening more quickly at our tertiary centre (11.0h compared to 13.8h at outside centres; p=0.04). Optimal pediatric DKA management continues to be a challenge despite the presence of multiple, evidence-based guidelines. Gradual resuscitation with appropriate fluids appears to be the most common area in which variability exists in community and tertiary care centres.
Poster Abstract 7
Implementation and Evaluation of a DKA Order Set in a Pediatric Tertiary Care Hospital: A Quality Improvement Initiative

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Background:
Diabetic ketoacidosis (DKA) is a common clinical presentation in new and previously diagnosed pediatric patients with type 1 diabetes. In contrast to other Canadian tertiary pediatric hospitals, our center lacked a physician-endorsed evidence-informed care pathway for management of DKA, resulting in variability in patient management and outcomes. This project was a quality improvement initiative which sought to develop and pilot a pediatric DKA order set.

Aims: To attain broad clinical uptake of the order set at our tertiary care center over a 12-month period. Secondary aims included improved standard-of-care DKA management of fluid, potassium, and glucose administration.

Methods:
A pediatric multidisciplinary collaborative was created to examine evidence for the development and implementation of a DKA order set. Implementation of the order set involved department wide education, targeted end-user education, and quarterly end-user review. A modified plan-do-study-act (PDSA) cycle guided by end-user feedback and early clinical outcomes allowed progressive order set modifications.

Results:
A retrospective chart review of fifty pediatric patients presenting to our center (April 2014 – September 2016) was compared to thirty patients presenting during the first-year post-implementation phase (September 2016 – September 2017). There were no statistically significant differences in demographic or clinical characteristics between groups. We achieved 83% uptake of the order set for patients presenting to our tertiary center and 67% uptake for patients transferred from peripheral centers. DKA management improvements included: appropriate intravenous (IV) maintenance fluid rates (20% vs. 48.3%, p=0.008), earlier administration of potassium to IV fluids (66% vs. 93.1%, p=0.006); appropriate IV potassium (40 mmol/L) dosing (40% vs. 79.3%, p=0.0007) and earlier addition of IV dextrose (67.4% vs. 93.1%, p=0.009). No differences in moderate to severe hypokalemia (< 3.0 mmol/L), hypoglycemia (<4.0 mmol/L) or clinically suspected cerebral edema occurred.

Conclusions:
Implementation of a DKA order set in a tertiary hospital required identification of key stakeholders, formation of a multidisciplinary team, and a rigorous evaluation process. There was an observed increase in physician order set uptake and DKA management practice improvements. Future goals involve expanding the implementation and evaluation process to provincial regional and remote centers and analyzing the impact on resource utilization.
Poster Abstract 8
Living with Type 1 Diabetes, Youth and Parent Perceptions

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Background:
Type 1 diabetes (T1D) is a lifelong condition that requires constant self-management, family support and ongoing learning. There are metrics available to assess parent and child perceptions of a youth’s quality of life (QoL), self-management, and family conflict. We aimed to gain insight into these perceptions.

Methods:
This is a cross-sectional observational study of youth with T1D and their parents. Both youth and a parent completed the Diabetes Family Conflict Scale (DFCS), Adherence in Diabetes Questionnaire (ADQ), and the PedsQL [child (8-12 years), adolescent (13-18 years); generic/diabetes modules]. Demographic and diabetes history were recorded. PedsQL total and subset scores (physical, emotional, social, school, and psychological functioning), and responses to individual questions from each questionnaire were compared between youth and adults. Data were analyzed using t-test and Pearson’s chi-squared test with P<0.05 considered significant.

Results:
52 families completed the questionnaires (Patients: 47.2% female, age 11.5±2.0 years (mean, SD), duration of diabetes 5.9±3.1 years; parents: 86.5% female, age 43.5±5.2 years). While no differences were identified in total or subset QoL scores, youth and parents did diverge in regards to specific questions on all three surveys. Compared to the adolescents themselves, parents had statistically significant heightened concern for the adolescents’ wellbeing in the following areas (p<0.001): feeling sad or blue, feeling angry, worrying about what will happen to them, having trouble getting along with other teens, experiencing other teens teasing them, feeling tired, worrying about themselves going “low”, and experiencing difficulty asking doctors and nurses questions. Compared to both adolescents and children, parents reported significantly more frequent occurrences of feeling embarrassed about having diabetes and diabetes treatment and arguing with parents about diabetes care. Finally, young people perceive more challenges keeping the agreement with the diabetes care team regarding treatment than parents.

Conclusion:
This study identifies differing perceptions between youth and their parents regarding the youth’s experience living with diabetes. Generally, parents seem to perceive a higher level of distress in their youth, especially during adolescence, than acknowledged by the youth themselves. This data provides insight for medical professionals to support youth and their families in diabetes care.
Poster Abstract 9
Seeing Eye-to-Eye: A Single Center Quality Improvement Experience in Retinopathy Screening and Reporting in Pediatric Type 1 Diabetes Patients

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Introduction:
Annual retinopathy screening is recommended for individuals with type 1 diabetes who are 15 years or older and have been diagnosed for five years or longer. Yet, literature shows that rates of retinopathy screening are low, and interventions to increase uptake are minimally effective.

Objective:
This quality improvement (QI) project aimed to evaluate whether implementation of novel strategies at a pediatrics diabetes centre can enhance retinopathy screening by (1) increasing rates of screening and (2) improving communication of screening results from the eyecare professional to the diabetes team.

Methods:
Between January and November 2017, a QI study incorporating several Plan, Do Study, Act (PDSA) cycles was undertaken. Examples of interventions included patient awareness campaigns, information packages distributed to patients in clinic, availability of same-day eye exams at a nearby optometrist, and mailed out packages to eligible patients including a templated report for eyecare professionals to return to the diabetes clinic. Outcome measures included the rate of retinopathy screening by patient report in the past 12 months, and the presence of a retinopathy examination report from an eyecare professional in the patients’ electronic medical record.

Results:
The baseline retinopathy screening rate in our clinic was 71%. Analysis per month according to established run chart rules revealed no sustainable change in the rate of retinopathy screening. Excluding patients who were enrolled in local research studies that included retinopathy screens, we were, however, able to demonstrate a significant increase in the percentage of eligible patients with retinopathy reports from eyecare professionals (13.2% in 2017 compared to 2.5% in 2015 and 6.7% in 2016, p<0.001), with the majority being in the latter half of the year after the mail-out occurred.

Conclusion:
Previous attempts by others to improve rates of retinopathy screening have had limited impact. We too did not alter screening rates, with improvement possibly more difficult in our clinic due to the high baseline rates. However, our interventions, especially mail-out of information packages to eligible patients did improve communication of results from the eyecare professionals to the diabetes team.
Poster Abstract 10
Adherence to a Pediatric Diabetic Ketoacidosis Protocol in Children Presenting to a Tertiary Care Hospital

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Objective:
To review adherence to a provincial DKA protocol and to assess factors associated with intravenous fluid administration and the length time on an insulin infusion.

Methods:
A retrospective chart review was conducted of all DKA admissions to British Columbia Children’s Hospital (BCCH) during September 2008 to December 2013. Data collection included diabetes history, estimation of dehydration, insulin and fluid infusion rates, and frequency of laboratory investigations. Markers of adherence included appropriate use of a fluid bolus, normal saline and insulin infusion time, fluid intake and outputs and the frequency of blood work during the insulin infusion. A log-linear regression model was fitted to assess the factors associated with insulin infusion duration.

Results:
Of 157 children [median (IQR) age: 10.6 years (5.0, 13.8)] hospitalized for DKA, 45% (n=70) were male, 55% (n=86) were transferred from other hospitals, and 26% (n=40) were admitted to intensive care unit (ICU). Thirty-five percent of subjects estimated to have mild or moderate dehydration received fluid boluses. In the adjusted analysis, the average duration on DKA protocol was 39% (95% CI: 12%, 67%) longer for children admitted with severe dehydration (compared to those with mild dehydration).

Conclusions:
Health care providers’ adherence to the BCCH DKA protocol is poor. More severe dehydration at presentation is associated with longer duration of insulin infusion. Further knowledge translation initiatives focused on accurate estimation of volume depletion to ensure appropriate initial fluid resuscitation—as well as careful monitoring during DKA hospitalization—are important, especially in community centers.
An Observational Study of Canadian Children and Youth with Type 1 Diabetes Following Initiation of Continuous Subcutaneous Insulin Infusion (CSII)

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Background:
Continuous subcutaneous insulin infusion (CSII), or insulin pump therapy, is associated with improved HbA1c, patient satisfaction and quality of life. Long-term pediatric studies on this topic are limited. We report our long-term experiences with CSII initiation in a tertiary pediatric centre through a large observational cohort study.

Methods:
Retrospective chart review was completed for all children and youth with type 1 diabetes initiating CSII at CHEO between 2001 and 2015. Inclusion criteria were: age less than 18, at least 1 year of follow-up prior to and after CSII initiation, and no extended periods of inactivity (or removal) from CSII. Outcomes included HbA1c, proportion of patients with change in HbA1c of 0.5% or more, and proportion within HbA1c target of <7.5%. Patients were divided into subgroups by age category, gender, and duration of diabetes.

Results:
Of 273 pump starts, 198 patients met inclusion criteria (96 males and 102 females). Mean age at diabetes diagnosis was 6 years (+/- 3.8). At pump initiation, mean duration of diabetes was 3.8 years (+/- 2.8). Mean follow-up time on CSII was 4.7 years (+/- 2.7, range 1 to 12). Analysis was truncated at 9 years due to small sample size beyond this. There were no significant differences in outcome measures between males and females. HbA1c in the first year following initiation of CSII declined from 7.9% to 7.5%. However, this change was not sustained, and a gradual return to baseline HbA1c was observed over the subsequent 2-3 years. The proportion of patients meeting HbA1c target (<7.5%) remained higher than baseline through 6 years of follow up. Upon subgroup analysis, HbA1c improvement was sustained in children younger than 6 years, and transient in children 6-12 years of age; there was relative HbA1c stability in adolescents.

Conclusions:
We observed an overall improvement in HbA1c in children and youth initiated on CSII, in keeping with other reports in the literature. While long-term follow-up demonstrated that this HbA1c benefit was not sustained over time, the proportion of patients meeting target HbA1c remained higher than baseline. The most sustained HbA1c reduction was observed in children <6 years.
Poster Abstract 12
An Under-Recognized Cause of False Positive Newborn Screening for Congenital Adrenal Hyperplasia

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Objective:
To describe situations where sample contamination with EDTA can lead to false positive results in the newborn screen (NBS) for congenital adrenal hyperplasia (CAH).

Methods:
A retrospective case series from a tertiary care pediatric endocrinology service (Alberta Children’s Hospital, Calgary, Alberta) between 2016 and 2017.

Case 1:
A healthy-appearing newborn female had a critical NBS for CAH, with a 17-OHP level >735.5 nmol/L. Initial testing showed normal electrolytes and a low random cortisol. The patient was placed on glucocorticoid and mineralocorticoid coverage pending further results. Follow-up testing showed a normal stimulated cortisol and normal serum 17-OHP. Retesting of the initial blood spot revealed inconsistent values for 17-OHP (range 45 to >735.3 nmol/L). The patient’s mother later recalled that the laboratory assistant had taken the NBS sample from another sample tube, rather than directly from the patient.

Case 2 & 3:
A critical NBS for CAH was reported on a 34-week gestational age twin ‘A’ who was generally well and being monitored for prematurity. Twin A’s NBS 17-OHP level was 162 nmol/L, while twin B’s 17-OHP level was 136 nmol/L. Both infants were monitored in the NICU while workup was pending. Serum and repeat NBS 17-OHP levels were normal for both infants. The initial NBSs were taken from a specimen collection tube rather than directly from the patient.

Discussion:
These cases of false-positive NBS for CAH screening are consistent with previously reported experimental and observational studies of EDTA contamination of NBSs. This occurs when blood for NBS is obtained from an EDTA-containing vessel rather than directly from the patient. Potential error reduction strategies include better standardization of collection methods and adoption of a two-step screening protocol that utilizes a second analytic method to test for presence of CAH.

Conclusions:
1. EDTA contamination at the time of collection can cause false positive results on CAH NBS and result in prolonged hospital admission, unnecessary additional testing, and increase stress for families.
2. Pre-analytical errors should be considered when clinical presentation and laboratory testing do not match.
Poster Abstract 13
Timing of Peak Serum Cortisol Levels Following Low-Dose ACTH Stimulation in Neonates

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Background:
Low-dose ACTH stimulation testing (LDST) is the preferred method of diagnosing central adrenal insufficiency in neonates. However, protocols vary significantly between institutions, with some testing only at 30 minutes and others testing multiple times between 15 and 60 minutes. A recent study of neonates concluded that the majority of LDSTs peak at 40 minutes, however testing was not performed beyond 40 minutes (Sari 2012). The importance of 60-minute cortisol levels remains unknown. The purpose of this study is to determine the optimal times to draw serum cortisol levels in neonates undergoing LDST.

Methods:
A single center retrospective chart review was performed. All neonatal patients (less than 28 days corrected gestational age) who underwent LDST at the Children’s Hospital of Eastern Ontario between February 1, 2014 and September 30, 2017 were included. As per hospital protocol, cortisol levels were drawn at 0, 15, 30, and 60 minutes post administration of cosyntropin 1 mcg/kg (max 1 mcg). A cortisol value > 499 nmol/L was used to define adrenal sufficiency.

Results:
Of 20 patients, 1 patient (5 %) had a peak cortisol level at 15 minutes, 3 patients (15 %) had peak cortisol levels at 30 minutes, and 16 patients (80%) had peak cortisol levels at 60 minutes. The number of patients who would have been misdiagnosed as adrenally insufficient based on alternative sampling plans are as follows: 0 if testing was performed only at 30 and 60 minutes; 0 if testing was performed only at 15 and 60 minutes; 2 (10%) if testing was performed only at 15 and 30 minutes; 2 (10%) if testing was performed only at 30 minutes; and 0 if testing was performed only at 60 minutes.

Conclusions:
Our findings suggest that the majority of neonates undergoing LDST have peak cortisol levels at 60 minutes. If testing was performed only at 30 minutes, 10% of patients would have been misdiagnosed as adrenally insufficient. Testing at 15 minutes did not provide additional information. Based on these findings, we recommend that when performing LDST in neonates, cortisol levels should be drawn at least at 0, 30, and 60 minutes.
Poster Abstract 14
Are We Doing Enough? Assessing Practice Patterns and Identifying Challenges to Providing Healthy Living Counselling at a Regional Tertiary Level Care Pediatric Hospital

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Objective:
Childhood overweight/obesity rates remain high and treatment has been largely ineffective. Pediatric healthcare providers (HCP) have a key role to play in prevention. Study objective was to gather pediatric HCP views on: (i) barriers/facilitators to providing healthy lifestyle counselling (HLC) during clinic visits, (ii) current practice patterns and knowledge of healthy living recommendations and (iii) preferred methods for improving knowledge and skills related to HLC.

Methods:
A needs assessment survey, informed by literature review and input from key stakeholders, was distributed via email to 705 HCPs at BC Children’s Hospital. Descriptive statistics and thematic analysis were performed. Survey respondents were invited to participate in a follow-up focus group to further explore HLC barriers and facilitators, and HCP educational needs.

Results:
Survey response rate was 31.2% (40% physicians, 17% nurses, 25% allied health, 16% social workers/psychologists, 2% other). Most HCPs self-reported assessing height (82%) and weight (89%) at least half of the time; only 56% calculated body mass index (BMI) half of the time and 26% reported that BMI was only done if patients appear overweight/obese. Physical activity (69%) and sugary drink intake (80%) were assessed more frequently than screen time (52%), junk food (44%), and fruit/vegetable intake (51%). Although 88% of participants provided HLC, only 46% believed they were meeting patients’ needs, and only 40% reported moderate/high levels of confidence discussing weight issues. Barriers to HLC included: time constraints, finding the “right time,” poor self-efficacy, and having unhealthy personal lifestyle habits. Factors facilitating HLC included: additional human resources, a hospital environment that supports healthy habits, and the ability to refer patients to community-based programs. Most (90%) respondents believed they could improve their HLC skills, and 81% were interested in additional training. Nineteen HCPs participated in two focus groups. HLC barrier and facilitator themes were similar to the survey, but having a hospital environment that endorses healthy habits (e.g. healthy food vendors, play areas) was identified as being critically important.

Conclusions:
We report a gap in pediatric HCP practice and challenges with HLC. Next step is development of a toolkit addressing these barriers and knowledge gaps, with the aim of increasing HCP capacity to integrate HLC into routine practice.
Poster Abstract 15
Self-Perception of Training in Core Pediatric Endocrinology Topics Among General Pediatrics Residents: a Learning Needs Assessment

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Introduction:
A learning needs assessment was undertaken as a first step in designing new formal Pediatric Endocrinology teaching curriculum for General Pediatrics residents at the University of Calgary (UofC) Methods: A cross-sectional quality improvement project. A survey was developed based on fourteen core subjects in Pediatric Endocrinology modified from the College of Physicians and Surgeons of Canada Objectives of Training in Pediatrics (2008). Surveys were delivered to residents currently training in the General Pediatrics training program at UofC. Respondents answered their agreement on a 5-point Likert scale regarding three statements for each core subject: (1) “subject is important to my future clinical practice,” (2) “my current knowledge/skill is adequate for my future clinical practice,” (3) “my residency training in this field was adequate.”

Results:
A total of 24 of 44 (55%) residents have so far completed the survey, including 10 who have completed a core residency rotation in Pediatric Endocrinology. Residents who had not yet completed a core rotation in Pediatric Endocrinology felt they had adequate knowledge/skill (ie. clinical competence) in an average of 2.1 of 14 (15%) core subjects, while residents who had finished a rotation felt clinically competent in an average of 7.3 of 14 (52%) core subjects (p for difference = 0.0001). Residents generally regarded all subjects as important. Residents were most likely to agree with the importance to future clinical practice of hypoglycemia (96%) and adrenal insufficiency (91%) and least likely to agree with the importance of bone fragility disorders (65%) and transgender medicine (70%). Among those who had finished a rotation, very few (30% or less) felt clinically competent in lipid disorders, calcium disorders, puberty disorders, bone fragility, and transgender medicine. Residents tended to feel they had received inadequate training in these same subjects.

Conclusions:
1. General Pediatric residents generally regard all Pediatric Endocrinology subjects as important to their future clinical practice.
2. The current curriculum achieves self-reported competence in approximately one half of endocrinology topics.
3. Future curriculum design efforts should focus on improving teaching in lipid disorders, calcium disorders, puberty disorders, bone fragility, and transgender medicine.
Poster Abstract 16
Then versus Now: Changing Characteristics in Adolescents Referred to a Gender Clinic

Chiniara LN1, Bonifacio HJ1, Palmert MR1
1) Hospital for Sick Children, University of Toronto, Toronto, ON

Purpose:
The aim of this study was to explore recent characteristics, including mental health comorbidities, in transgender adolescents, and to compare these data to previous published cohorts.

Methods:
A retrospective chart review was conducted among adolescents (aged 12-18 years) assessed in the gender variance clinic at the Hospital for Sick Children between January 2014 and June 2016. Demographic data, clinical characteristics, mental health comorbidities (history of diagnosis, use of medications, self-harm behaviors and ideation, as well as questionnaire data) were obtained. Baseline and repeat blood work, when available, were also reviewed.

Results:
Charts from 203 adolescents (156 assigned female at birth (AFAB) (77%) aged 16.3 ±1.63 years, 47 assigned male at birth (AMAB) aged 16.1 ±1.70 years were reviewed. Individuals from minority racial/ethnic populations were under-represented as 73% were of caucasian descent. There was no statistically significant difference between gender groups except for Tanner stage (AFAB: mean 4.42 ± 0.8 and AMAB mean 4.03 ± 1.1, p=.040) and mental health comorbidities (higher levels, most notably of anxiety, among AFAB, p=0.03). Among all youth, self-report data and baseline psychological questionnaires showed high levels of gender dysphoria (100%), mood disorders (37.4% with depression, 28.1% with anxiety) and suicidal ideation (33%). On cross-sex hormones, hemoglobin levels increased slightly in AFAB (p=.002, highest=166 g/L); hemoglobin levels decreased in AMAB (p=.019, lowest=132g/L).

Conclusion:
Our study supports an evolving demographic trend among transgender adolescents with more AFAB than AMAB youth now presenting to clinics. The data also corroborate studies indicating that extensive laboratory testing is not a necessary part of caring for these youth. Why more AFAB are now presenting to clinic and racial/ethnic minorities are so underrepresented is not clear, but may impact clinical care as mental health comorbidities were common among AFAB youth.
Poster Abstract 17
Perspectives of Transgender Youth and their Parents Regarding Fertility Preservation

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Objective:
The aim of this study was to investigate the perspectives and values of youth with gender dysphoria and their parents concerning fertility preservation and future parenthood.

Methods:
A cross-sectional questionnaire-based study among transgender youth and their parents was designed and administered to assess knowledge of potential effects of treatments for gender dysphoria on fertility, current and future life priorities, and preferences regarding future fertility/parenting options.

Results:
A total of 79 youth (81% assigned females at birth (AFAB), 19% assigned males at birth (AMAB), age range 12 to 20, with 68% between ages 16-20 years) and 73 parents participated. The top current life priority for youth was being in good health, and the least important priority was Having children. Anticipated life priorities 10 years from now were ranked similarly. Parents’ rankings paralleled the youth responses; however, parents ranked having children as a significantly higher priority for AFAB compared to AMAB youth in 10 years. The majority of youth (66% AFAB, 67% AMAB) want to be a parent in the future. However, most do not envision having a biological child. A large majority (72% AF, 80% AM) were open to adoption. All youth knew treatment with cross-sex hormones could alter future fertility but none of the youth surveyed pursued fertility preservation.

Conclusion:
Fertility is a low current and future life priority for transgender youth. The majority of youth wish to become parents but are open to alternative strategies for building a family. These data may help explain the reported low rates of accessing fertility preservation among this population. Further studies are needed to assess if youths’ life priorities and values change over time.
Poster Abstract 18
Like Mother Like Daughter: Evolving Central Diabetes Insipidus Secondary to an AVP Gene Mutation

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We present a case of a 5-year-old previously well girl with a 3-year history of polydipsia, water-seeking behaviours, and nocturia. Reported water intake was 2-3 litres per day. There were no growth or neurologic concerns. There was a maternal history of idiopathic central diabetes insipidus (DI), diagnosed at age 9 and managed with desmopressin. Physical exam was within normal limits. Initial investigations performed after four hours of fluid restriction revealed a mildly elevated Na of 146mmol/L and normal serum osmolarity (sOsm) of 297mmol/kgH2O. Urine was dilute with an osmolarity (uOsm) of 187mmol/kgH2O. Serum glucose, calcium, and HbA1C were normal and there was no glucosuria. Pituitary screen was unremarkable. Alpha-fetoprotein was normal. Water deprivation test was suggestive of partial diabetes insipidus. After 7 hours of water restriction, Na peaked at 148mmol/L and sOsm peaked at 302mmol/kgH2O. uOsm at this time was 314mmol/kgH2O. The test was terminated after 9 hours of water restriction as there had been minimal urine output. Final values were as follows: Na 145 mmol/L, sOsm 294mmol/kgH2O, uOsm 324mmol/kgH2O, and ADH<0.6 pmol/L (0.80-3.50). Desmopressin was not administered. Polyuria, polydipsia and nocturia persisted. A trial of once daily desmopressin was initiated with marked improvement in symptoms. MRI at this time revealed a normal pituitary stalk and a smaller than normal posterior pituitary bright spot. Twice daily desmopressin was eventually required for symptom control. Arginine vasopressin (AVP) gene sequencing revealed a likely pathogenic heterozygous mutation (c.133G>T), resulting in the amino acid substitution p.Gly45Cys. Familial central (neurohypophyseal) diabetes insipidus (FNDI) is a rare hereditary disorder with the majority of cases caused by a heterozygous mutation of the AVP gene. FNDI is often autosomal dominant. Progression to complete DI over time is common and thought to be related to accumulation of cytotoxic AVP precursors. The c.133G>T mutation identified here was first described in a Turkish family in 2015 and has not been reported again since. In this case, genetic testing was used to confirm a diagnosis of FNDI despite an inconclusive water deprivation test. Identifying a genetic change eliminates the need for ongoing surveillance of evolving neuropathology that may underlie central DI.
Severe Early-Onset Obesity: Genetic Causes and How to Diagnose Them

Dr. William T. Gibson

CME Objectives

After this presentation, participants will be able to:

1. List five genetic tests useful in the workup of a suspected monogenic/oligogenic obesity disorder

Key Clinical Messages

- Feeding behaviour can be “hardwired,” thereby placing it effectively outside of conscious control
- The risk of developing obesity can be genetically programmed
- BMI Z-score can help you decide (if >3.0)

 Syndromic Obesity Disorders

- Prader-Willi Syndrome
- Bardet-Biedl Syndrome
- Alstrom Syndrome
- Cohen Syndrome
- Pseudohypoparathyroidism
- Familial Partial Lipodystrophy

Rare Obesity Disorders Relatively New to Clinical Medicine

- Congenital Leptin Deficiency (1997)
- Proprotein Convertase I Deficiency (1997)
- Pro-opiomelanocortin Deficiency (1998)
- Congenital Leptin Receptor

What is the “Architecture of Our Remaining Ignorance?”

How many common obesity genes are in the genome? How many rare obesity genes are in the human genome?
When to think of a Major Genetic Contribution to Obesity?

- Very early-onset of disease and/or very severe disease
- Intellectual disability and/or visual loss
- One or more birth defects
- Unusual facial appearance unlike other family members

Key Message

- Reduced signalling through the leptin-melanocortin axis (Lep, LepR, POMC, MC4R, Gsα) has measurable effects on energy balance, and can have a genetic basis

Disruption of the Hypothalamic Leptin-Melanocortin Signalling Pathway

Melanocortin-4 Receptor Deficiency

- The most common cause of severe, early-onset obesity
- 5%-8% of cases
- Tall stature, hyperinsulinemia, relatively preserved pancreatic function
Leptin Receptor Deficiency

- Accounts for 3% of severe, early-onset obesity (severe = BMI SDS >3)
- In general, features are less severe than those of subjects with congenital leptin deficiency
- Affected subjects characterized by hyperphagia, severe obesity, and abnormal pubertal growth

Leptin Receptor Deficiency

- Adjusted basal metabolic rate per kilogram of lean mass was unaffected by LEPR mutations (no evidence of a major deficit in basal energy expenditure)
- Childhood stature normal, but final adult height reduced (no pubertal growth spurt)

Heterozygote Effects

- Leptin deficiency, leptin receptor deficiency, POMC-deficiency and MC4R deficiency all have measurable effects in heterozygote carriers
  - LEP carriers: 5% increase in body fat
  - LEPR carriers: 5-8% body fat increased, BMI no change
  - POMC carriers: BMI increased BMI SDS 1.7±0.5
  - MC4R carriers: varies w/ severity of mutation
  - Sophisticated body composition analysis is often required for the effect to be identified

Somatic Mosaicism - PHP

Mother's

23 year-old Brother

25 year-old Sister

Alstrom Syndrome

Obesity Genotyping: Beyond BWS and PWS

- Chromosome Microarray Analysis (should be available locally)
- Next-Generation Sequencing Panels

Why should we do Microarray Studies in Obesity?
- Best for patients with “Obesity plus something else”
- Will find a lot of cases of PWS (but not all)
- Will find 16p11.2 microdeletions
- Will occasionally find a specific

Gene-Specific and Sequencing Studies in Patients with Severe Obesity
- Gene-Specific Studies
  - Leptin deficiency and Pseudohypoparathyroidism are treatable
- Chromosome Microarray Analysis (CMA)
  - Microarrays find deletions in the Wilms’ tumour region that require surveillance for tumours

Microarray Results in Obesity

![Microarray Results in Obesity](image)

DNA Diagnostic Companies that offer “Obesity Panels”

- Agilent Genetics
- Alphagenetics
- Instituto de Medicina Genómica
- University of Chicago Genetics
- University of Connecticut Health
- University of Cincinnati

Summary of results [NGS Panel]

<table>
<thead>
<tr>
<th>Test results</th>
<th>Negative</th>
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<tbody>
<tr>
<td>Sequencing performance metrics O6-Seq</td>
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</tr>
<tr>
<td>Panel</td>
<td>Congenital and familial Lipodystrophy Panel</td>
</tr>
<tr>
<td>Genes</td>
<td>Exons</td>
</tr>
<tr>
<td>Congenital and familial Lipodystrophy Panel</td>
<td>11</td>
</tr>
</tbody>
</table>

Target region and gene list

-卜春杰 randomly selected from the following regions and included in the following regions:
- Ones that are randomly selected from the other regions and included in the other regions:
- Ones that are randomly selected from the same regions and included in the same regions:
- Ones that are randomly selected from the different regions and included in the different regions:
- Ones that are randomly selected from the same regions and included in the different regions:
- Ones that are randomly selected from the same regions and included in the same regions:
Obesity Phenotyping: Beyond Body-Mass Index

- Biochemical Markers (Fasting Glucose, lipids, transaminases, leptin, adiponectin)
- Bone Age
- Food intake (questionnaire, test meal)
- Activity level (screen time, sports, accelerometry)

Key Point from PTEN Article

- Patients with PTEN mutations (and tumour susceptibility due to Cowden disease) have higher risk for obesity but lower risk for type 2 diabetes
- Enhanced glucose disposal proven with hyperinsulinemic-euglycemic clamp studies

Prader-Willi Syndrome (A070 - no GH)

<table>
<thead>
<tr>
<th>Metabolic Parameter</th>
<th>Value</th>
</tr>
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<tbody>
<tr>
<td>Respiratory Quotient</td>
<td>0.68</td>
</tr>
<tr>
<td>Body Mass Index</td>
<td>36.2 kg/m²</td>
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<tr>
<td>Test Meal Eaten</td>
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<tr>
<td>Resting Energy Expenditure</td>
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<tr>
<td>Lean Body Mass</td>
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<tr>
<td>REE/LBM</td>
<td>32.2 kcal/kg/d</td>
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<tr>
<td>REE/LBM Reference Range</td>
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</tr>
</tbody>
</table>

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