


## BIOGRAPHICAL SKETCH

<b>NAME</b> <b>Johnny Y. Deladoëy</b> Email: <a href="mailto:johnny.deladoey@umontreal.ca">johnny.deladoey@umontreal.ca</a> Internet site: <a href="http://thyroid4kids.org">thyroid4kids.org</a>  CHU Sainte-Justine Endocrinology Service; Room #1722 3175, Chemin de la Côte Sainte-Catherine Montréal (Québec) H3T 1C5 Phone: (514) 345-4931 (extension 5032) FAX: (514) 345-4988	<b>POSITION TITLE</b> Clinical Associate Professor (Pediatrics) Pediatric Endocrinologist and Diabetologist (staff) Affiliated Professor (Biochemistry) 
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## EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE	YEAR(s)	FIELD OF STUDY
Collège, Abbaye de St-Maurice, Switzerland	B.Sc.	1990	Sciences & Humanities
University of Bern, Bern, Switzerland	M.D.	1996	Medicine
USMLE step1 and step 2	M.D.	1996-97	Medicine
University of Zurich, Zurich, Switzerland	Postgraduate Course	1997-98	Experimental Medicine
University of Bern, Bern, Switzerland	Ph.D.	2001	Molecular Endocrinology
University of Bern, Bern, Switzerland	Board / FMH*	2005-06	Pediatrics
University of Montreal, Montreal, QC, Canada	Board / FMH*	2007-08	Pediatric Endocrinology
University of Montreal, Montreal, QC, Canada	DES**	2008	Pediatric Endocrinology

\*FMH: Federatio Medicorum Helveticorum (Swiss Medical Association)

\*\* DÉS: Diplôme d'Études Supérieures en Pédiatrie-Endocrinologie (Postdoctoral training diploma in pediatric endocrinology)

## POSITIONS

1997-2001	<b>Research Fellow</b> , University Children's Hospital, University of Bern, Switzerland.
2002-2004	<b>Pediatric Resident</b> , University Children's Hospital, University of Bern, Switzerland.
2005-2007	Clinical and Research <b>Endocrine Fellow</b> , Sainte-Justine Hospital, University of Montréal, Montréal, QC, Canada.
2008-pres	Pediatric Endocrinologist & Diabetologist ( <b>Staff</b> ), Sainte-Justine Hospital (Centre Hospitalier Universitaire Mère-Enfant), Université de Montréal, Montréal, QC, Canada
2008-2014	<b>Clinical Assistant Professor (tenure-track)</b> , Department of Pediatrics, Medicine Faculty, Université de Montréal, Montréal, QC, Canada.
2014-pres	<b>Clinical Associate Professor</b> , Department of Pediatrics, Medicine Faculty, Université de Montréal, Montréal, QC, Canada.
2009-pres	<b>Affiliated Professor</b> , Department of Biochemistry, Science Faculty, Université de Montréal, QC, Canada.

## HONORS, SCHOLARSHIPS AND AWARDS

1998-2001	M.D.-Ph.D. Scholarship award (3136-054879) of the <b>Swiss National Science Foundation (SNF)</b> . [CHF 150,000]
2006	John Bailey Price for best presentation by a fellow, Canadian Pediatric Group Meeting, Edmonton, February 2006. [500 \$ Can]
2006	Quebec Thyroid Club; Abbott 1 <sup>st</sup> Price for the best presentation, May 2006, Lac Brome, Canada [500 \$ Can]
2006	Grand Price for the best oral presentation, Graduate Students' Annual Congress, Sainte-Justine Hospital Research Center, June 2006, Montréal, Canada. [500 \$ Can]
2007	Quebec Thyroid Club; Abbott 2 <sup>nd</sup> Price (ex aequo) for the best oral presentation, May 2007, Montréal, Canada [250 \$ Can]
2005-2007	Swiss Foundation of Medical-Biological Scholarship award (PASMA-112979) under the patronage of the SAMS and the <b>Swiss National Science Foundation (SNF)</b> . [CHF 117,000]

- 2005-2007  
2007 Scholarship Award of the Pediatric Department, University of Montreal, Canada. [CAN\$80,000]. Hoffmann La Roche / Canadian Pediatric Endocrine Fellowship Award of the Canadian Pediatric Endocrine Group (CPEG). [CAN\$30,000]
- 2008-2012 Career Development Award of the Canadian Child Health Clinician Scientist Program (**CCHSCP**) [CAN\$ 280,000]. CCHSCP is a **CIHR Strategic Training Program**. Declined upon receipt of the FRSQ scholarship (see below) and converted in Career Enhancement Program Award of the CCHSCP [CAN\$20,000].
- 2008-2012 **Research Scholar Junior 1** Award of the Fonds de Recherche du Québec-Santé (**FRQS**). [CAN\$295,500]
- 2012 **Henning Andersen Prize 2012** for the Best Clinical Abstract of the **European Society for Pediatric Endocrinology (ESPE)** [€ 1,500].
- 2014-2016 **Clinical Research Scholar Junior 2** Award of the Fonds de Recherche du Québec-Santé (**FRQS**) [CAN\$72,000].
- 2014 **International Award for Publishing Excellence in *The Journal of Clinical Endocrinology & Metabolism*** for the 2013 Article "Linear Association Between Household Income and Metabolic Control in Children With Insulin-Dependent Diabetes Mellitus Despite Free Access to Health Care"; The Endocrine Society; 2014 June.
- 2014 **Charles Hollenberg Young Investigator Award 2014** of the Canadian Society for Endocrinology and Metabolism (**CSEM**); 2014 October; [CAN\$ 20,000]
- 2015 **Mid-Career Award of the Sabbatical leave program** of the **European Society for Pediatric Endocrinology (ESPE)** for a 3-months project at Necker-Hospital / University Paris Descartes; 2015 May; [€ 14,000]

## RESEARCH SUPPORT

- 2005-2006 **PI:** Research Grant of the Endocrine Fellows Foundation (EEF), USA. [US\$7,500 (operating funds)]
- 2008-2009 **PI:** Start-up Operating Funds of the Ste-Justine Hospital Research Center [CAN\$100,000].
- 2008-2009 **PI:** Start-up Equipments Funds of the Ste-Justine Hospital Research Center [CAN\$100,000].
- 2008-2011 **PI:** Young Investigator Operating Grant of the Fonds de Recherche en Santé du Québec (**FRSQ**); [CAN\$45,500].
- 2010-2013 **PI:** Thyroid Diseases in Children. Girafonds / Ste-Justine Hospital Foundation [CAN\$ 200,000].
- 2011-2013 **PI:** Abnormal Thyroid Development: a Model Disorder for Congenital Malformations and Neurocognitive Development. **European Society for Pediatric Endocrinology (ESPE)** Research Unit Grant [€60,000 / CAN\$ 82,000]
- 2013-2016 **PI:** Genetic Determinants of Congenital Hypothyroidism. **Canadian Institutes of Health Research (CIHR)** operating grant MOP-130390 [337,338 CAN\$]
- 2010-2012 **clinical member** of the Canadian Pediatric Genetic Disorders Sequencing Consortium / Finding of Rare Disease Genes in Canada (FORGE Canada) funded by **Genome Canada, CIHR, Genome Quebec and Genome BC** [ND/yr].
- 2014-2016 **co-investigator:** Degludec use in pediatric patients (Protocol NN5401-3816). Novo Nordisk Canada Inc.
- 2010-2014 **sub-investigator:** Auto-immune Diseases and Commonly Prescribed Drugs and Vaccines : An International Multi-Centre Case-referent Epidemiological Field Study (PGRx) – Centre for Risk Research Inc, Montreal: \$58,000, 200 patients
- 2001-2015 **sub-investigator:** international study on genetics determinants of short stature funded by Eli-Lilly (Protocol B9R-EW-GDFC(b)): Eli Lilly Canada: \$2,175/patient, currently 50 patients, cumulative enrolled patients: 98.
- 2012-2013 **sub-investigator:** EMR200102\_010 – First year growth response associated with genetic markers validation Phase IV open-label study in growth hormone deficient and Turner syndrome pre-pubertal children: the PREDICT pharmacogenetics validation – Serono Laboratories Inc.; \$20,000; 6 patients
- 2011-pres **sub-investigator:** A randomised, double-blind, placebo-controlled parallel group dose-finding study of linagliptin (1 mg or 5 mg administered orally once daily) over 12 weeks in children and adolescents, from 10 to 17 years of age, with type 2 diabetes and insufficient glycemic control despite treatment with diet and exercise alone (BI protocol 1218.56) funded by Boehringer Ingelheim Canada [ND/yr]
- 2011-2015 **sub-investigator:** Easypod™ CONNECT: An International Multi-Centre Study to Monitor GH Compliance - Serono Laboratories, Inc: \$1,040/patient, 14 patients (25 planned).
- 2015-2016 **sub-investigator:** Comparison of VRS-317, a Long-acting Human Growth Hormone, to Daily rhGH in a Phase 3, Randomized, One-year, Open-label, multi-center, Non-inferiority Trial in Prepubertal

## PROFESSIONAL SOCIETIES

- Canadian Pediatric Endocrine Group (CPEG)
- European Society for Pediatric Endocrinology (ESPE)
- Collège des Médecins du Québec (CMQ)
- Fédération des Médecins Spécialistes du Québec (FMSQ)
- Endocrine Society (USA)
- Pediatric Endocrine Society (USA)
- American Thyroid Association (ATA; USA)
- Association des Médecins Endocrinologues du Québec (AMEQ)

## ACADEMIC COMMITTEES & ACTIVITIES

- 2008-pres **Thesis Committees:** Rasha Abu-Khudir (PhD; as director: 2008-2014); Sonia Cournoyer (MSc; as Jury – director: Dr H. Sartelet; 2013); Isma Benterki (PhD; as member- director: Dr. V. Poitout; 2013-pres); Samira Benhadjeba (MSc; as Jury – irector : Dr A. Trembaly; 2013).
- 2008-2009 **Member** Task Force for Renewal of the Internet Site of the Pediatrics Dpt, U of Montreal (local).
- 2009-2011 **Member**, Fonds de Recherche du Québec (**FRSQ**), PhD scholarship evaluation committee (committee FF3-5D. 2009:15 reviewed applications; 2010: 17; 2011: 13)(provincial).
- 2009-2010 **Member**, Jury, Graduate Students' Annual Congress, Sainte-Justine Hospital Research Center, U of Montreal (local).
- 2009-2010 **Member**, evaluation committee of the CHU Sainte-Justine Research Center for the following competitions (local): Grants of the Hôpital Ste-Justine Fondation / Fondation des Etoiles CHUFSJ/FDE (2009-2010 competition-7 reviewed applications; 2010-2011 competition-7 reviewed applications).
- 2009-2015 **Center Leader** of the University of Montreal and **member of the Program Advisory Committee** for the Canadian Child Health Clinician Scientist Program (**CCHSCP**). CCHSCP is a **CIHR** Strategic Training Program (national).
- 2012-pres **Member, Advisory Board**, Unité de Recherche Clinique Appliquée (**URCA**), CHU Sainte-Justine, University of Montréal. (local)
- 2013-2014 **Member**, Committee for a global consent form / broad biobank, CHU Sainte-Justine, University of Montréal.(local)
- 2013-pres **Member**, Committee of Space Management, Research Center, CHU Sainte-Justine, University of Montréal.(local)
- 2014 **Member, Canadian Institutes of Health Research (CIHR)** grant review committee (Endocrinology); Open Operating Grant Program (national)
- 2014-pres **Member, Summer School Steering Committee of the European Society for Pediatric Endocrinology (ESPE)**, an ESPE strategic initiative to train the future leader of the ESPE (international).
- 2015-pres **Member, Program Planning Committee of the Canadian Society of Endocrinology and Metabolism (CSEM)** (national)
- 2015-pres **Member**, Canadian Institutes of Health Research (CIHR) Review Panel, **Stage 2 of CIHR Foundation Scheme**.

## EDITORIAL / CONGRESSIONAL ACTIVITIES

- 2011-pres **Member of the Editorial Board:**  
2012-2015 : *Journal of Clinical Endocrinology and Metabolism* [IF 6.43]  
2011-pres. : *Frontiers in Endocrinology*  
2011-2013 : *Open Journal of Pediatrics*
- 2007-pres **ad hoc reviewer** (more than 20 reviews/yr) for the following journals [IF2012]: *Endocrinology, J Clin Endocrinol Metab, Pediatrics, Endocrine Related Cancer, Clin Endocrinol; Horm Res Ped, etc.*
- 2012-pres **Grant's reviewer for the following funding agencies:**  
*Telethon Foundation, Milan, Italy (2012-1); Canadian Institutes of Health Research (CIHR; 2014); French National Research Agency (ANR – general competition 2015); European E-Rare 3 program (2<sup>nd</sup> round- 2015).*
- 2010 **Chair, Oral Session on Thyroid Gland Development** at the 14<sup>th</sup> International Thyroid Congress, September 11-16, 2010, Paris, France (**International**).
- 2011 **Reviewer**, 2011 Pediatric Academic Societies (PAS) workshops sessions, April 30- May 3, 2011 Denver, CO (**International**).

- 2011 **Moderator**, Poster Session at the 50<sup>th</sup> Annual Meeting of the European Society for Pediatric Endocrinology (ESPE), 25-28 September 2011, Glasgow, UK (**international**).
- 2012 **Member**, Canadian Pediatric Endocrine Group (**CPEG**) abstract review committee for the 2012 CPEG meeting.
- 2012-pres **Abstracts' Reviewer** for the Annuals Meeting of the European Society for Pediatric Endocrinology (ESPE) (**international**).
- 2012 **Abstracts' Reviewer** for the 6<sup>th</sup> International Congress of the Growth Hormone Research Society (GRS), 17-20 October 2012, Munich, Germany (**international**).
- 2012-pres **Abstracts' Reviewer** for the Canadian Pediatric Endocrinology Group (CPEG) 2012, 2013 and 2014 Scientific Meeting, Canada (**national**).
- 2013 **Member, Local Organizing Committee** for the Canadian Pediatric Endocrinology Group (CPEG) 2013 Scientific Meeting, June 2013, Quebec City, QC, Canada (**national**).
- 2014 **Chair, Thyroid Session** – Guided Poster Tour. The 53<sup>rd</sup> Annual Meeting, European Society for Pediatric Society (ESPE), 18-21 September 2014, Dublin, Ireland (**INTERNATIONAL**).

## EDUCATIONAL CONTRIBUTIONS

### Clinical teaching

2008-pres Attending Physician, Pediatrics, Pediatric and Adult Endocrinology, CHU-Sainte-Justine.

### Formal Teaching

- 2008-pres Clinical Endocrinology, University of Montreal (graduate course **END8002**, 5 to 10 students, 1 session of 1h about Congenital Hypothyroidism, yearly).
- 2009 Epigenetic in Human Disease, April 2009, University of Montreal, (graduate course **SMC6061**, 20 students, 1h in 2009)
- 2010-2014 Metabolism 1, University of Montreal, (graduate course **BCM1502**, ca. 120-140 students, 8 to 10h, yearly)

### Graduate

- 2008-2014 Thesis Director, PhD Thesis Committee, Rasha Abu-Khudir.
- 2011-2015 Thesis Director, PhD Thesis Committee, Fabien Magne.
- 2012 Jury, Master thesis, Sonia Cournoyer (Dir.: Dr Hervé Sartelet).
- 2013 Jury, Master thesis, Samira Benhadjeba (Dir.: Dr André Tremblay).
- 2014-pres Member, PhD Thesis Committee, Samira Benhadjeba (Dir.: Dr André Tremblay).

### Trainees (supervision<sup>1</sup> or co-supervision<sup>2</sup>; only MD fellows with scientific supervision are listed)

Period	Name	Present Position
2005-2006	Claire Perruisseau-Carrier M. Sc. <sup>2</sup>	PhD student – Merck Serono (Switzerland)
2008-2009	Janette Saavedra, MD; Fellow <sup>2</sup>	Consulting Physician, (Equator)
2007-2014	Rasha Abu-Khudir M. Sc.; PhD <sup>1</sup>	Lecturer, U of Tantra, Egypt
2007-2010	Sophie Stoppa-Vaucher MD; Fellow <sup>1</sup>	Attending Physician, U of Lausanne (Switzerland)
2009-2012	Coralie Leblicq MD; Fellow <sup>2</sup>	MSc, McGill University, Montreal
2009-2011	Steffi Wildi-Runge MD; Fellow <sup>2</sup>	Fellow, U of Montreal
2010-2007	Stéphanie Michaud MD; Fellow <sup>2</sup>	Fellow, McGill University
2010-2011	Isabelle Vandernoot MD; PhD student <sup>1,2</sup>	PhD student, Free University of Brussels (Belgium)
2011-2012	Caroline Hasselmann MD; Fellow <sup>2</sup>	Consulting Physician, CHU Tours, France
2011-pres	Fabien Magne; PhD student <sup>1</sup>	PhD student, U of Montreal
2012-pres	Despoina Manousaki, MD, Fellow <sup>2</sup>	Fellow, U of Montreal

### Awards to trainees

- 2009 Canadian Pediatric Endocrine Group Award declined upon receipt of Eugène Litta Foundation Award (Geneva) to Sophie Stoppa-Vaucher. [CHF 50,000] (**national**)
- 2009 Quebec Thyroid Club; Abbott 3<sup>rd</sup> Price for the best oral presentation, May 2009, Montréal, Canada to Rasha Abu-Khudir. [200 \$ Can] (**local**)
- 2009 Prize for the best oral presentation by a resident at the Annual Meeting of the Canadian Society of Endocrinology and Metabolism, November 2009, Montréal, Canada to Sophie Stoppa-Vaucher. [500 \$ Can] (**national**)
- 2009 Excellence PhD Award of the Faculté des Études Supérieures and the Biochemistry Department, University of Montréal, November 2009, to Rasha Abu-Khudir. [3,000 \$ Can] (**local**)
- 2010 Excellence PhD Award of the Faculté des Études Supérieures and the Biochemistry Department, University of Montréal, November 2010, to Rasha Abu-Khudir. [4,500 \$ Can] (**local**)

- 2011-2013 PhD Award of Fondation des Étoiles / Fondation CHU Sainte-Justine, December 2010, for Rasha Abu-Khudir. [17,500\$/yr Can; acceptance rate 9/31] **(local)**
- 2011 Presidential Poster Competition Award for best poster presentation at The Endocrine Society's 93<sup>rd</sup> Annual Meeting, June 4-7 2011, Boston, MA; for Steffi Wildi-Runge MD; Fellow. **(international)**
- 2013 Award for the best oral presentation by a PhD student at the 28<sup>th</sup> Graduate Student Congress of the Research Center of the CHU Sainte-Justine, 30 May 2013, for Rasha Abu-Khudir [250\$ Can] **(local)**
- 2014 Prix Réseau de recherche en santé cardiométabolique, diabète et obésité (CMDO) for the best presentation by a PhD student at the 29<sup>th</sup> Graduate Student Congress of the Research Center of the CHU Sainte-Justine, 29 May 2014, for Fabien Magne [500\$ Can] **(local)**

#### **PEER-REVIEWED PUBLICATIONS in chronological order (Trainees)**

1. Flück C, **Deladoëy J**, Rutishauser K, Eblé A, Marti U, Wu W, Mullis PE. Phenotypic variability in familial combined pituitary hormone deficiency caused by a PROP1 gene mutation resulting in the substitution of Arg->Cys at codon 120 (R120C). *J Clin Endocrinol Metab* 1998; 83: 3727-3734.
2. **Deladoëy J**, Flück C, Buyukgebiz A, Kuhlmann BV, Eblé A, Hindmarsh PC, Wu W, Mullis PE. „Hot spot“ in the PROP1 gene responsible for combined pituitary hormone deficiency. *J Clin Endocrinol Metab* 1999; 84: 1645-1650.
3. **Deladoëy J**, Flück C, Bex M, Yoshimura N, Harada N, Mullis PE. Aromatase deficiency caused by a novel P450arom gene mutation: impact of absent estrogen production on serum gonadotropin concentration in a boy. *J Clin Endocrinol Metab* 1999 ; 84: 4050-4054.
4. Nuoffer JM, Flück C, **Deladoëy J**, Eble A, Dattani MT, Mullis PE. Regulation of human GH receptor gene transcription by 20 and 22 kDa GH in a human hepatoma cell line. *J Endocrinol* 2000; 165:313-20.
5. Wajnrajch MP, Gertner JM, Mullis PE, **Deladoëy J**, Cogan JD, Lekhakula S, Kim S, Dannies PS, Saenger P, Moshang T, Phillips III JA, Leibel RL. Arg183His, a new mutational „hot spot“ in the growth hormone (GH) gene causing isolated GH deficiency type II. *J Endocrine Genetics* 2000; 1:125-135.
6. **Deladoëy J**, Stocker P, Mullis PE. Autosomal dominant GH deficiency due to an Arg183His GH-1 gene mutation: clinical and molecular evidence of impaired regulated GH secretion. *J Clin Endocrinol Metab* 2001; 86:3941-3947.
7. Vuissoz JM\*, **Deladoëy J\***, Buyukgebiz A, Cemeroglu P, Gex G, Gallati S, Mullis PE. New autosomal recessive mutation of the TSH-beta subunit gene causing central isolated hypothyroidism. *J Clin Endocrinol Metab* 2001; 86:4468-4471 **(\*equal contributors)**.
8. Flück CE, **Deladoëy J**, Nayak S, Zeller O, Kopp P, Mullis PE. Autosomal dominant neurohypophyseal diabetes insipidus in a Swiss family, caused by a novel mutation (C59Delta/A60W) in the neurophysin moiety of prepro-vasopressin-neurophysin II (AVP-NP II). *Eur J Endocrinol* 2001; 145:439-444.
9. Mullis PE, **Deladoëy J**, Dannies PS. New GH-1 Gene mutations: expanding the spectrum of causes of isolated growth hormone deficiency. *J Pediatr Endocrinol Metab* 2002; 15: 1301-1310.
10. Mullis PE, **Deladoëy J**, Dannies PS. Molecular and cellular basis of isolated dominant growth hormone deficiency, IGHD type II: insights on the secretory pathway of peptides hormones. *Horm Res* 2002; 58: 53-66.
11. **Deladoëy J**, Gex G, Vuissoz JM, Strasburger CJ, Wajnrajch MP, Mullis PE. Effect of different growth hormone (GH) mutants on the GH-receptor gene transcription in a human hepatoma cell line: a comparative study with 22-kDa normal GH. *Eur J Endocrinol* 2002; 146: 573-581.
12. **Deladoëy J\***, Vuissoz JM\*, Doméné HM\*, Malik N, Grunerio-Papendieck L, Chiesa A, Heinrich JJ, Mullis PE. Congenital secondary hypothyroidism due to C105V (C105 fr sh, 114X) TSH-b mutation: genetic study of five unrelated families from Switzerland and Argentina. *Thyroid* 2003; 13: 553-559 **(\*equal contributors)**.
13. Mullis PE, Robinson IC, Salemi S, Eble A, Besson A, Vuissoz JM, **Deladoëy J**, Simon D, Czernichow P, Binder G. Isolated autosomal dominant growth hormone deficiency (IGHD II): An evolving pituitary deficit? A multi-center follow-up study. *J Clin Endocrinol Metab* 2005; 90: 2089-2096.
14. Besson A, Salemi S, **Deladoëy J**, Vuissoz JM, Eble A, Fluck C, Mullis PE. Short stature caused by a biologically inactive mutant growth hormone (GH-C53S). *J Clin Endocrinol Metab* 2005; 90: 2493-2499.
  - Cited in the *Yearbook of Pediatric Endocrinology 2005* (Karger).
15. Salemi S, Yousefi S, Eblé A, **Deladoëy J**, Mullis PE. Impact of del32-71-GH (exon 3 skipped GH) on intracellular GH distribution, secretion and cell viability: a quantitative confocal microscopy analysis. *Horm Res* 2006; 65: 132-141.
16. Salemi S, Yousefi S, Eblé A, Lochmatter D, **Deladoëy J**, Robinson ICAF, Simon HU, Mullis PE. Isolated Autosomal Dominant Growth Hormone Deficiency (IGHD II): Stimulating mutant GH-1 gene expression drives GH-1 splice-site selection, cell proliferation and apoptosis. *Endocrinology* 2007; 148: 45-53.

17. **Deladoëy J**, Bélanger N, Van Vliet G. Random Variability in Congenital Hypothyroidism from Thyroid Dysgenesis over 16 years in Quebec. *J Clin Endocrinol Metab* 2007; 92: 3158-3161.
  - Cited in the *Yearbook of Pediatric Endocrinology 2008* (Karger).
18. **Deladoëy J**, Pfarr N, Vuissoz JM, Parma J, Vassart G, Biesterfeld S, Pohlenz J, Van Vliet G. Pseudodominant inheritance of goitrous congenital hypothyroidism caused by TPO mutations: molecular and in silico studies. *J Clin Endocrinol Metab* 2008; 93: 627-633.
  - Cited and evaluated by the *Faculty of 1000 Medicine* (<http://f1000.com/1116848>)
19. Maquet E, Costagliola S, Parma J, Christophe-Hobertus C, Oligny LL, Fournet JC, Robitaille Y, Vuissoz JM, Payot A, Laberge S, Vassart G, Van Vliet G, **Deladoëy J**. Lethal respiratory failure and mild primary hypothyroidism in a term girl with a de novo heterozygous mutation in the TITF1/NKX2.1 gene. *J Clin Endocrinol Metab* 2009; 94: 197-203.
20. Shenoy A, Esquibies AE, Dunbar N, Dishop MK, Reyes-Mugica M, Langston C, **Deladoëy J**, Abu-Khudir R, Carpenter T, Bazy-Asaad A. A novel presentation of diffuse lung disease secondary to congenital hypothyroidism. *J Pediatr* 2009; 155:593-5.
  - Highlighted by an editorial (*J Pediatr* 2009; 155: A1)
21. Stoppa-Vaucher S, Francoeur D, Grignon A, Alos N, Pohlenz J, Van Vliet G, **Deladoëy J**. A 19-weeks fetus with non-immune hypothyroidism and goiter: a plea for conservative management. *J Pediatr*; 2010; 156:1026-9.
  - Cited in the *Yearbook of Pediatric Endocrinology 2010* (Karger).
22. Stoppa-Vaucher S, Lapointe A, Turpin S, Rydlewski C, Vassart G, **Deladoëy J**. Ectopic Thyroid Gland causing Dysphonia: Imaging and Molecular Studies. *J Clin Endocrinol Metab* 2010; 95: 4509-4510.
  - Selected case report for the book *Diagnostic Dilemmas* published in 2011 by the Endocrine Society.
23. Abu-Khudir R, Paquette J, Lefort A, Libert F, Chanoine JP, Vassart G, **Deladoëy J**. Transcriptome, Methylome and Genomic Variations Analysis of Ectopic Thyroid Glands. *PLoS ONE* 2010; 5(10): e13420
  - Highlighted by the Target Intelligence Service (TIS), a database used by pharmaceutical companies to identify groundbreaking research of relevance for their own work on potential drug target.
  - Cited and evaluated by the *Faculty of 1000 Medicine* (<http://f1000.com/8843956>)
  - Cited in the *Yearbook of Pediatric Endocrinology 2011* (Karger).
  - More than 5,000 views with 1,000 downloads on PLoS One and PMC web site (February 8<sup>th</sup>, 2015).
24. Leblicq C, Rottembourg D, **Deladoëy J**, Van Vliet G, Deal C. Are Guidelines for Glucocorticoid Coverage in Adrenal Insufficiency Currently Followed? *J Pediatr* 2010 158:492-498.
  - Highlighted by an editorial (*J Pediatr* 2009; 158: A1)
  - Highlighted by an editorial in *Endocrine Today*
25. Stoppa-Vaucher S, Van Vliet G, **Deladoëy J**. Variation by Ethnicity in the Prevalence of Congenital Hypothyroidism due to Thyroid Dysgenesis. *Thyroid* 2011; 21(1):13-18.
26. Saavedra J\*, **Deladoëy J\***, Saint-Vil D, Boivin Y, Alos N, Deal C, Van Vliet G, Huot C. Is Ultrasonography Useful for Predicting Thyroid Cancer in Children with Apparently Benign Cytopathologic Features? (\*co-1<sup>st</sup> author). *Horm Res Paediatr* 2011; 75:269-75.
27. **Deladoëy J**, Ruel J, Giguère Y, Van Vliet G. Is the incidence of congenital hypothyroidism really increasing? A 20-year retrospective population-based study in Québec. *J Clin Endocrinol Metab* 2011; 96: 2422-2429.
  - Highlighted by an editorial (*J Clin Endocrinol Metab* 2011; 96: 2395-2397)
  - Induced a change in screening protocol in Quebec (higher threshold for 2<sup>nd</sup> test – January 2012)
  - Highlighted in 'the 2011 Year in Review' of the Lawson Wilkins Pediatric Endocrine Society (USA)
  - Cited in the *Yearbook of Pediatric Endocrinology 2012* (Karger).
28. Stoppa-Vaucher S, Van Vliet G, **Deladoëy J**. Discovery of a fetal goiter on prenatal ultrasound in women treated for Graves' disease: First, do not harm. *Thyroid* 2011; 21: 931.
29. Vandernoot I, Sartelet H, Abu-Khudir R, Chanoine JP, **Deladoëy J**. Evidence for calcitonin-producing cells in human lingual thyroids. *J Clin Endocrinol Metab* 2012; 97: 951-956.
30. Stoppa-Vaucher S, Aybe T, Paquette J, Patey N, Francoeur D, Vuissoz JM, **Deladoëy J**, Ogata T, Deal C. 46, XY Gonadal Dysgenesis: New Point Mutation in Two Siblings and a Germ Line Mosaicism in Their Father. *Clinical Genetics* 2012; 82:505-513.
31. Wildi-Runge S, Stoppa-Vaucher S; Lambert R, Turpin S, Van Vliet G, **Deladoëy J**. A high prevalence of dual thyroid ectopy in congenital hypothyroidism: evidence for insufficient signaling gradients during embryonic thyroid migration or for the polyclonal nature of the thyroid gland? *J Clin Endocrinol Metab* 2012; 97: E978-81.
  - Highlighted by an editorial in *Endocrine News* (May 2012), the monthly magazine for clinician endocrinologists, a publication of The Endocrine Society.
  - Cited in the *Yearbook of Pediatric Endocrinology 2013* (Karger).



32. Samuels ME, Gallo-Payet N, Pinard S, Hasselmann C, Magne F, Patry L, Chouinard L, Schwartzentruber J, René P, Sawyer N, Bouvier M, Djemli A, Delvin E, Huot C, Eugene D, Deal CL, Van Vliet G, Majewski J, **Deladoëy J**. Bioinactive ACTH Causing Glucocorticoid Deficiency. *J Clin Endocrinol Metab* 2013; 98: 736-742.
- Highlighted by an article in lay journal *La Presse* (31 January 2013), the main French daily journal of the city of Montreal.
  - Interview for Radio-Canada International (6 February 2013).
  - Highlighted by an article on the web site of the Société Française d'Endocrinologie (<http://www.s fendocrino.org>; Newsletter Recherche no. 6 – Mars 2013).
  - Cited in the *Yearbook of Pediatric Endocrinology 2013* (Karger).
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  - Highlighted by an article in the blog of **kpcc** operated by **Southern California Public Radio** (15 April 2013). The station has among the widest-reaching broadcast areas of all public radio stations in Southern California, with a signal that reaches through most of [Los Angeles](#) and [Orange County](#). The station is listened to by approximately 600,000 listeners each week.
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38. Samuels M, Hasselmann C, Deal C, **Deladoëy J**, Van Vliet G. Whole-exome sequencing: opportunities in pediatric endocrinology. *Pers Med* 2014; 11(1):63-78.
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### Publications submitted or in preparation

1. Larrivée-Vanier S, Magne F, Patey N, Chanoine JP, Vuissoz JM, Van Vliet G, **Deladoëy J**. Conserved telomere length in human ectopic thyroids: an argument against premature differentiation causing arrested migration (submitted to *Thyroid* – in revision)
2. Henderson M, Benedetti A, Barnett T, Mathieu ME, **Deladoëy J**, Gray-Donald K. Adiposity, physical activity, fitness and screen time: how do they influence insulin dynamics over 2 years in youth? (submitted to *JAMA pediatrics*)
3. Perlsteyn M, **Deladoëy J**, Van Vliet G. Similar levothyroxine requirements of school-age children with congenital or acquired hypothyroidism (submitted to *J Pediatr*)
4. Sawyer SL, Hartley T, Dymont DA, Beaulieu CL, Schwartzentruber J, Smith A, Bedford M, Bernard G, Bernier F, Brais B, Bulman DE, Warman Chardon J, Chitayat D, **Deladoëy J**, Fernandez B, Frosk P, Geraghty M, Gerull B, Gibson W, Gow RM, Graham G, Green JS, Heon E, Horvath G, Innes AM, Jabado N, Kim RH, Koenekoop RK, Khan A, Lehmann OJ, Mendoza-Londono R, Michaud JL, Nikkel S, Penney LS, Polychronakos C, Richer J, Rouleau GA, Samuels ME, Siu V, Suchowersky O, Tarnopolsky MA, Yoon G, Zahir F, FORGE Canada Consortium, Care4Rare Canada Consortium, Majewski J, Boycott KM. Utility of whole-exome sequencing toward the end of the diagnostic odyssey: time to address gaps in care (submitted to *Clin Genetics*)
5. Fernandez I, Khodja Y, Capo-chichi J, Schwartzentruber J, Hasselmann C, Nassif C, **Deladoëy J**, Haddad E, Majewski J, Van Vliet G, Le Deist F, Samuels ME. Mutation in IKBKE in a patient with immunodeficiency and pituitary hormone deficiencies: GOLIATH, a variant of DAVID syndrome (submitted to *Blood*)
6. Manousaki D, **Deladoëy J**, Geoffroy L, Olivier P. Continuous Subcutaneous Insulin Infusion in Children: a Protocol Validated to Avoid Hypoglycemia at Initiation (in preparation)
7. Magne F, Larrivée-Vanier S, Carré A, Abu-Khudir R, Van Vliet G, Samuels M, Polak M, **Deladoëy J**. Recurrent *de novo* functional variant in the tissue-specific differentially methylated region of the *FOXE1* promoter is associated with congenital thyroid ectopy (in preparation)

### Book Chapters

1. **Deladoëy J**, Vassart G, Van Vliet G. Possible non-Mendelian mechanisms of thyroid dysgenesis. In: Guy Van Vliet, Michel Polak, editors. *Pediatric Thyroid Disorders: Advances in Developmental Biology, Physiology and Clinical Management*. Basel: Karger; Endocr Dev 2007; 10: 29-42.
2. **Deladoëy J**. An 8-year old girl with a lingual mass. In: Leonard Wartofsky, Editor. *Diagnostic Dilemmas: Images in Endocrinology*. The Endocrine Society, September 2011. ISBN 1-879225-87-5
3. **Deladoëy J**. Congenital Hypothyroidism due to Thyroid Dysgenesis: from Epidemiology to Molecular Mechanisms. In: Draha Springer, editor. *A New Look at Hypothyroidism*. InTech edition, February 2012. ISBN 978-953-51-0020-1.
4. Van Vliet G, **Deladoëy J**. Hypothyroidism in infants, children and adolescents. In: Lewis E. Bravermann, editor. *Werner and Ingbar's The Thyroid: A Fundamental and Clinical Text*, 10<sup>th</sup> Edition, Chapter 54, pp. 787-802. Lippincott Williams & Wilkins, New York, 2013. ISBN 978-1-4511-2063-9.
5. Van Vliet G, **Deladoëy J**. Disorders of the Thyroid in the Newborn and Infant. In: Mark A. Sperling, editor. *Pediatric Endocrinology*, 4<sup>th</sup> Edition. Elsevier, Philadelphia, 2014. ISBN 9781455748587.
6. **Deladoëy J**. Goitre, hypothyroïdie et hyperthyroïdie. In: Marie Gauthier, editor. *Dictionnaire de Thérapeutique Pédiatrique Weber*, 3<sup>rd</sup> Edition, chapitre 103, pp. 499-504. Chenelière Éducation, Montréal, 2015. ISBN 978-2-7650-4746-9.
7. Luu T.M., **Deladoëy J**, Lapointe A, Samson Y. Masses Cervicales. In: Marie Gauthier, editor. *Dictionnaire de Thérapeutique Pédiatrique Weber*, 3<sup>rd</sup> Edition, chapitre 161, pp. 785-787. Chenelière Éducation, Montréal, 2015. ISBN 978-2-7650-4746-9.
8. Van Vliet G, **Deladoëy J**. Congenital Hypothyroidism: diagnosis, treatment and outcome. In: Gabor Szinnai, editor. *Endocrine Development: Pediatric Thyroidology*. S.Karger Publishers, Basel, Switzerland, Endocr Dev 2014; 26: 50-59.
9. Szinnai G, Leger J, Bauer A, Pearce E, Ramos HE, Canalli MH, Onigata K, Elisei R, Radetti G, Polak M, Van Vliet G, **Deladoëy J**. Clinical case seminar in pediatric thyroid disease. In: Gabor Szinnai, editor. *Endocrine Development: Pediatric Thyroidology*. S.Karger Publishers, Basel, Switzerland, Endocr Dev 2014; 26: 214-244.
10. Pohlenz J, Van Vliet G, **Deladoëy J**. Developmental abnormalities of the thyroid. In: Roy E. Weiss and



Samuel Refetoff, editors. Genetic Diagnosis of Endocrine Disorders, 2<sup>nd</sup> edition. Academic Press / Elsevier, San Diego, CA, 2015 (in press)

11. **Deladoëy J**, Van Vliet G, Giguère Y. Neonatal screening for congenital hypothyroidism. In: Bona, De Luca, Monzani (eds). Thyroid Diseases in Childhood. Springer Health Care Italia S.r.l., Milan, Italy, 2015 (in press)
12. Van Vliet G, **Deladoëy J**. Interpreting hyperthyrotropinemia in children. In: Daneman D and Palmert M, editors. Issue on Pediatric Endocrinology & Diabetes. *Pediatr Clin North Am* 2015 (in press)

#### **ABSTRACTS / Trainees**

**(if presented at several meetings, only listed one time)**

1. **Deladoëy J**, Mullis PE. Effect of the dominant negative human growth hormone (hGH) gene mutations on the regulated secretory pathway: why the wild type hGH is losing its way. Swiss society of Endocrinology and Diabetology, Meeting 5-6 November 1999, Bern, Switzerland. (oral presentation / national).
2. **Deladoëy J**, Mullis PE. Effect of the dominant negative human growth hormone (hGH) gene mutation Arg183His on the regulated secretory pathway. Growth Hormone Research Society International Meeting, 7-9 September 2000, Gothenburg, Sweden; Abstract O64, **GH & IGF Res. (2000) 10: 129-186**. (oral presentation / international).
3. Vuissoz JM, **Deladoëy J**, Buyukgebiz A, Cemeruglu P, Gex G, Gallati S, Mullis PE. New autosomal recessive mutation of the TSH- $\beta$  subunit gene causing central isolated hypothyroidism. Swiss Society of Pediatrics, Annual Meeting, 14-16 June 2001, Luzern, Switzerland. (oral presentation / national).
4. Vuissoz JM, **Deladoëy J**, Eblé A, Janner M, Gex G, Nuoffer JM, Gallati S, Mullis PE. Mutational "hot spot" in the TSH- $\beta$  subunit gene causing isolated thyroid-stimulating hormone (TSH) deficiency. Swiss Society of Endocrinology and Diabetology, Meeting 9 November 2001, Bern, Switzerland. (oral presentation / national).
5. Salemi S, Besson A, Baltensperger K, Eblé A, **Deladoëy J**, Mullis PE. Autosomal dominant GH-deficiency (IGHD Type II) caused by different GH-1 gene mutations: alteration of the GH secretory pathway analysed by quantitative confocal microscopy. Swiss Society of Endocrinology and Diabetology Meeting, November 14<sup>th</sup> 2003, Bern, Switzerland. (oral presentation / national)
6. Salemi S, Besson A, Baltensperger K, Eblé A, **Deladoëy J**, Mullis PE. Autosomal dominant GH-deficiency (IGHD Type II) caused by different GH-1 gene mutations: alteration of the GH secretory pathway analysed by quantitative confocal microscopy. Growth hormone Research Society International Meeting, April 18<sup>th</sup>-22<sup>th</sup> 2004, Cairns, Australia. Abstract 142. **Growth Horm IGF Res (2004) 14: 129-171**. (oral presentation / international)
7. **Deladoëy J**, Deal C, Van Vliet G. Molecular mechanisms of thyroid dysgenesis. CCHCSP (Canadian Child Health Clinician Scientist Program), Third Annual National Symposium, October 14<sup>th</sup>-16<sup>th</sup> 2005, St. John's, Newfoundland; and the 18th Annual Meeting of the Canadian Pediatric Endocrine Group, February 3<sup>rd</sup>-5<sup>th</sup> 2006, Edmonton, Canada. (awarded). (oral presentation / national)
8. **Deladoëy J**, Deal C, Van Vliet G, Vuissoz JM. Comment fonctionne une désiodinase ? Une étude *in silico*. Thyroid Club of Quebec, May 17<sup>th</sup> 2006, Lac Brome, Canada (awarded); and XXith Annual Congress of the Research Center-CHU Sainte-Justine, June 2<sup>nd</sup> 2006, Montreal, Canada (awarded). (oral presentation / local)
9. **Deladoëy J**, Oligny LL, Payot A, Laberge S, Parma J, Vassart G, Van Vliet G. Lethal respiratory failure and mild primary hypothyroidism in a term girl with a *de novo* heterozygous mutation in the *TTF1* gene. Presented at the Pediatric Academic Societies Meeting, May 5<sup>th</sup>-8<sup>th</sup> 2007, Toronto, Canada. (poster / international)
10. **Deladoëy J**, Vuissoz JM. *In silico* analysis of the growth hormone – growth hormone receptor interaction (P2-387). Presented at the Endocrine Society's 89<sup>th</sup> Annual Meeting (Endo 07), June 2<sup>nd</sup>-5<sup>th</sup> 2007, Toronto, Canada; and presented at the 5<sup>th</sup> Annual National Symposium of the CCHCSP (Canadian Child Health Clinician Scientist Program), October 12<sup>th</sup>-16<sup>th</sup> 2007, Montreal, Canada. (posters / international)
11. **Deladoëy J**, Pfarr N, Vuissoz JM, Parma J, Vassart G, Pohlenz J, Van Vliet G. Pseudodominant inheritance of congenital thyroid dysmorphogenesis from TPO mutations in a non-consanguineous family: clinical, genetic and *in silico* studies. *Oral presentation* at 19th Annual Meeting of the Canadian Pediatric Endocrine Group, April 19<sup>th</sup>-22<sup>nd</sup>, Hamilton, Canada; And presented and nominated for a President's Poster Award at the 46th Annual Meeting of the European Society for Paediatric Endocrinology (ESPE), June 27<sup>th</sup>-30<sup>th</sup> 2007, Helsinki, Finland. (poster / international)
12. **Deladoëy J**. Non-Mendelian mechanisms of congenital hypothyroidism from thyroid dysgenesis. Presented at the Annual Meeting of the AMSPDC / PSDP (Pediatric Scientist Development Program), March 6<sup>th</sup> -9<sup>th</sup>, 2008, Santa-Fe, New Mexico, USA. (poster / international)
13. Abu-Khudir R, Paquette J, Deal C, Van Vliet G, **Deladoëy J**. Non-Mendelian mechanisms of congenital hypothyroidism from thyroid dysgenesis. Presented at CCHCSP (Canadian Child Health Clinician Scientist

- Program), 6<sup>th</sup> Annual Meeting, October 17<sup>th</sup> -19<sup>th</sup>, 2008, Edmonton, Canada. (oral presentation / national)
14. Stoppa-Vaucher S, Francoeur D, Grignon A, Van Vliet G, **Deladoëy J**. A 19-weeks fetus with non-immune hypothyroidism and goiter: treatment or observation? Presented at the 2009 CPEG (Canadian Pediatric Endocrine Group) Scientific Meeting, February 26<sup>th</sup>-28<sup>th</sup>, 2009, Ottawa, Canada. (oral presentation /national); and 8<sup>th</sup> Joint Meeting of the LWPES/ESPE, September 9-12, 2009 , New York City, NY (poster P03-343 /international)
  15. Abu-Khudir R, Paquette J, Libert F, Chanoine JP, Vassart G, Van Vliet G, **Deladoëy J**. Role of Genetic and Epigenetic Alterations in Thyroid Dysgenesis: A Combined Analysis of Transcriptome, Methylome and Structural Genome Variants in Ectopic versus Orthotopic Thyroid Tissues. New Inroads to Child Health (NICHe) Conference 2009, Child Health and Epigenetics, Developmental Plasticity and Programming (Sponsors: ESPE, Växhuset Foundation for Children, Nature Publishing Group), May 15<sup>th</sup>-17<sup>th</sup>, 2009, Göteborg, Sweden; Poster Presentation at ENDO 09 (P1-80), The Endocrine Society's 91st Annual Meeting, June 10-13 in Washington, DC. (*Invited to the ENDO 09 Presidential Poster Competition*); Gordon Research Conference on Cell Contact & Adhesion, June 28<sup>th</sup>-July 3<sup>rd</sup>, 2009, Waterville Valley, NH, USA. (3x posters / international)
  16. Leblicq C, Rottembourg D, **Deladoëy J**, Van Vliet G, Deal C. Are Guidelines for Glucocorticoid Coverage in Adrenal Insufficiency Being Followed? 8<sup>th</sup> Joint Meeting of the LWPES/ESPE, September 9-12, 2009, New York City, NY (poster P01-019, international)
  17. Stoppa-Vaucher S, Paquette J, Vuissoz JM, **Deladoëy J**, Francoeur D, Deal C. 46XY Gonadal Dysgenesis: Y sperm are not created equal! Canadian Society of Endocrinology and Metabolism (CSEM) Annual Meeting, October 17, 2009, Montreal, Canada. (oral/national)
  18. Abu-Khudir R, Paquette J, Lefort A, Libert F, Chanoine JP, Vassart G, Van Vliet G, **Deladoëy J**. Identification of new candidate genes associated with congenital hypothyroidism from thyroid ectopy by integrated molecular profiling (miRNA profile added). Presented at CCHCSP (Canadian Child Health Clinician Scientist program) 7th Annual Meeting, October 16th -18th, 2009, Halifax, Canada. (poster/national)
  19. Abu-Khudir R, Paquette J, Libert F, Chanoine JP, Vassart G, **Deladoëy J**. Identification of new candidate genes associated with congenital hypothyroidism from thyroid ectopy by integrated molecular profiling. Invited to the Eighth Annual New Principal Investigators Meeting / CIHR, November 6-8, 2009 Jackson's Point, ON (accepted poster, national)
  20. Grignon A, Rypens F, Francoeur D, Van Vliet G, Stoppa-Vaucher S, **Deladoëy J**. Le goître foetal, une question de taille. Société Canadienne Française de Radiologie, Annual Meeting, November 13-15, 2009, Montreal , Canada. (oral/local)
  21. Stoppa-Vaucher S, Francoeur D, Grignon A, Alos N, Pohlenz J, Van Vliet G, **Deladoëy J**. Un foetus de 19 semaines avec hypothyroïdie non-immune et un goiter: doit-on traiter ou observer? Resident Annual Meeting / CHU Sainte-Justine, February 24<sup>th</sup>, 2010, Montreal, Canada. (oral/local)
  22. Leblicq C, Rottembourg D, **Deladoëy J**, Van Vliet G, Deal C. Les protocoles d'adaptation des glucocorticoïdes en situation de stress dans l'insuffisance surrénalienne sont-ils appliqués. Resident Annual Meeting / CHU Sainte-Justine, February 24<sup>th</sup>, 2010, Montreal, Canada. (oral/local)
  23. Abu-Khudir R, Paquette J, Lefort A, Libert F, Chanoine JP, Vassart G, **Deladoëy J**. Identification of new candidate genes associated with defective thyroid migration by expression profiling. Annual Meeting of the Students in Biochemistry, March 12, 2010, University of Montreal, Montreal, Canada. (poster/local)
  24. Abu-Khudir R, Paquette J, Lefort A, Libert F, Chanoine JP, Vassart G, **Deladoëy J**. Identification of new candidate genes associated with defective thyroid migration by expression profiling. Annual Meeting of the Students of the Research Center of the CHU Sainte-Justine, May 26, 2010, CHU Sainte-Justine, Montreal, Canada. (poster/local)
  25. Decaussin-Petrucci M, Sassolas G, Hafdi-Nejjari Z, Berger N, Sturm N, Laverriere MH, Dumollard JM, Scoazec JY, Poulet A, **Deladoëy J**, Borson-Chazot F, Sartelet H. Expression of CD 133+, a cancer stem cell marker, in differentiated thyroid cancers of children and young adults. 14<sup>th</sup> International Thyroid Congress, September 11-16, 2010, Paris, France. (poster-discussion workshop / international).
  26. Stoppa-Vaucher S, Van Vliet G, **Deladoëy J**. Variation by Ethnicity in the Prevalence of Congenital Hypothyroidism due to Thyroid Dysgenesis. European Society for Pediatric Endocrinology (ESPE), 49<sup>th</sup> Annual Meeting, September 22-24, 2010, Prague, Czech Republic. (poster/international)
  27. Stoppa-Vaucher S, Paquette J, Vuissoz JM, **Deladoëy J**, Francoeur D, Deal C. Familial 46XY Complete Gonadal Dysgenesis – Mosaic in Sperm. European Society for Pediatric Endocrinology (ESPE), 49<sup>th</sup> Annual Meeting, September 22-24, 2010, Prague, Czech Republic. (poster/international)
  28. Saint-Vil D, Saavedra J, Huot C, Boivin Y, Van Vliet G, Deal C, Alos N, **Deladoëy J**. Ultrasonography(US) Is Useful In Predicting Thyroid Cancer In Children With Thyroid Nodules And Apparently Benign

- Cytopathologic Features. Canadian Association of Pediatric Surgeons, Annual Meeting, 23-26 September, 2010, Saskatoon, Canada. (poster/national)
29. Hitz MP, Opitz R, Abu-Kuhdir R, Vandernoot I, Paquette J, Desilets V, Costagliola S, Andelfinger G, **Deladoëy J**. Copy number variation in patients with combined congenital thyroid and heart defects. German Society of Human Genetics, 22<sup>th</sup> Annual Meeting, 16-18 March, 2011, Regensburg, Germany (poster/national); European Society for Pediatric Endocrinology (ESPE), 50<sup>th</sup> Annual Meeting, 25-28 September 2011, Glasgow, UK (poster, international)
    - Nominated for the President Poster Award at the ESPE 50<sup>th</sup> Annual Meeting, 25-28 September 2011, Glasgow, UK.
  30. **Deladoëy J**, Ruel J, Giguère Y, Van Vliet G. Lowering thyrotropin cutoff for newborn screening: Additional cases of congenital hypothyroidism are identified, but what do they really have? Canadian Pediatric Society, 88<sup>th</sup> Annual Meeting, 15-18 June 2011, Québec City, Canada (**ORAL/national**); The Endocrine Society's 93<sup>rd</sup> Annual Meeting, June 4-7 2011 in Boston (poster/international); AND Canadian Pediatric Endocrine Group (CPEG) Annual Meeting, 10-12 February 2011, Toronto, Canada (poster/national)
  31. Wildi-Runge S, **Deladoëy J**, Bélanger C, Deal C, Van Vliet G, Alos N, Huot C. Can recovery from hypopituitary-adrenal (HPA) axis suppression following supraphysiological doses of glucocorticoids be predicted? Canadian Pediatric Society, 88<sup>th</sup> Annual Meeting, 15-18 June 2010, Québec City, Canada (**ORAL/national**); The Endocrine Society's 93<sup>rd</sup> Annual Meeting, June 4-7 in Boston (poster/international\*\*); AND Canadian Pediatric Endocrine Group (CPEG) Annual Meeting, 10-12 February 2011, Toronto, Canada (poster/national)
    - Highlighted by an editorial in Endocrine Today
    - \*\*Presidential Poster Competition Award for best poster presentation at The Endocrine Society's 93<sup>rd</sup> Annual Meeting, June 4-7 2011, Boston, MA.
  32. Abu-Khudir R, Paquette J, **Deladoëy J**. Epigenetic regulation of the human transcription factor-2 *TTF2/FOXE1* gene. Annual Meeting of the Students of the Research Center of the CHU Sainte-Justine, May, 2011, CHU Sainte-Justine, Montreal, Canada. (poster/local)
  33. Vandernoot I, Hitz MP, Van Vliet G, Costagliola S, Andelfinger G, **Deladoëy J**. Copy number variants associated with congenital hypothyroidism from thyroid ectopy. Annual Meeting of the Students of the Research Center of the CHU Sainte-Justine, May, 2011, CHU Sainte-Justine, Montreal, Canada. (poster/local).
  34. Abu-Khudir R, Magne F, **Deladoëy J**. Epigenetic control of *FOXE1* expression in humans. The Endocrine Society's 94<sup>th</sup> Annual Meeting, 23-26 June 2012, Houston TX-USA (ENDO 2012) (poster / international).
    - Nominated for Presidential Poster Competition Award for best poster presentation.
  35. Samuels ME, Gallo-Payet N, Chouinard L, Hasselmann C, Magne F, Patry L, Pinard S, Jeremy Schwartzentruber J, Djemli A, Delvin E, Deal CL, Van Vliet G, Majewski J, **Deladoëy J**. Isolated glucocorticoid deficiency caused by immunoreactive but biologically inactive ACTH. The 51<sup>st</sup> Annual Meeting, European Society for Pediatric Endocrinology (ESPE), 20-23 September 2012, Leipzig, Germany (oral - PLENARY SESSION / international).
    - 2012 Henning Andersen Prize of Best Clinical Abstract of the **European Society for Pediatric Endocrinology (ESPE)**.
  36. Michaud S, Sharkia M, Berthier MT, Giguère Y, Metzger D, Stewart L, Deal C, **Deladoëy J**, Chanoine JP, Van Vliet G. No evidence for Central Hypothyroidism at Birth in Prader-Willi Syndrome (PWS). The 51<sup>st</sup> Annual Meeting, European Society for Pediatric Endocrinology (ESPE), 20-23 September 2012, Leipzig, Germany (poster / international).
  37. Samuels ME, Gallo-Payet N, Pinard S, Hasselmann C, Magne F, Patry L, Chouinard L, Schwartzentruber J, Djemli A, Delvin E, René P, Bouvier M, Metherell LA, Clark AJL, Huot C, Deal CL, Van Vliet G, Majewski J, **Deladoëy J**. Clinical diagnosis and revised treatment of a patient with *POMC* mutations and atypical presentation by whole exome sequencing. Journées Génétiques du Réseau de Médecine Génétique Appliquée, 22-24 May 2012, Montréal, Canada (ORAL PRESENTATION / local).
  38. **Deladoëy J**, Gallo-Payet N, Pinard S, Djemli A, René P, Bouvier M, Deal CL, Van Vliet G, Majewski J, Samuels ME. Déficience en glucocorticoïdes cause par une ACTH bioinactive mais immunoréactive. The 54<sup>th</sup> Annual Meeting of the Club de Recherche Clinique du Québec (CRCQ), 11-13 October 2012, Orford QC, Canada (poster / provincial).
  39. Samuels M, Hasselmann C, Majewski, Deal C, Huot C, Van Vliet G, **Deladoëy J**. Exome sequencing in a pediatric endocrinology service. The 12<sup>th</sup> International Symposium on Mutation in the Genome, 22-26 April 2013, Lake Louise AB, Canada (ORAL PRESENTATION / international).
  40. Abu-Khudir R, Magne F, **Deladoëy J**. Epigenetic control of *FOXE1* expression in humans. XXVIII<sup>e</sup> Graduate Student Congress of the Research Center of the CHU Sainte-Justine, 30 May 2013 (ORAL PRESENTATION / local).
    - Award for the best oral presentation by a PhD student.

41. **Deladoëy J**, Henderson M, Geoffroy L. Linear association between household income and metabolic control in children with insulin-dependent diabetes mellitus in spite of free access to health care .The 9<sup>th</sup> Joint Meeting of European Society for Pediatric Society (ESPE), the Pediatric Endocrine Society (PES), the Australasian Pediatric Endocrine Group (APEG), the Asia Pacific Endocrine Society (APPES), the Japanese Society for Pediatric Endocrinology (JSPE) and the Latin American Society of Pediatric Endocrinology (SLEP), 19-22 September 2013, Milan, Italy (ORAL PRESENTATION / international)
42. Hasselmann C, Samuels M, Majewski J, Deal C, Huot C, Van Vliet G, **Deladoëy J**. Diagnostic mysteries solved by exome sequencing. The 9<sup>th</sup> Joint Meeting of European Society for Pediatric Society (ESPE), the Pediatric Endocrine Society (PES), the Australasian Pediatric Endocrine Group (APEG), the Asia Pacific Endocrine Society (APPES), the Japanese Society for Pediatric Endocrinology (JSPE) and the Latin American Society of Pediatric Endocrinology (SLEP), 19-22 September 2013, Milan, Italy (Poster / international)
  - Nominated for Presidential Poster Competition Award for best poster presentation.
43. Manousaki D, Magne F, Van Vliet G, Samuels ME, Bui H, **Deladoëy J**. Thyroid ectopy in mother and daughter. Canadian Pediatric Endocrine Group (CPEG) Annual Meeting, 20-22 February 2013, Montreal, Canada (ORAL PRESENTATION / national).
44. Magne F, Abu-Khudir R, Carré A, Larrivée-Vanier S, Van Vliet G, Samuels ME, Polak M, **Deladoëy J**. Un polymorphisme dans la region promotrice de *FOXE1* est fonctionnellement associé à l'hypothyroïdie congénitale par ectopie thyroïdienne. The 29<sup>th</sup> Graduate Student Congress of the Research Center of the CHU Sainte-Justine, 29 May 2014, Montreal, Canada (Poster / AWARDED / local)
45. Meijer IA, Michaud J, Luan T, Rossignol E, Van Vliet G, **Deladoëy J**, Chouinard S, Bernard G. Novel mutations in TITF1 cause benign hereditary chorea, hypothyroidism and neonatal respiratory syndrome. 2014 Child Neurology Society Annual Meeting, 22-25 October 2014, Columbus, OH (Poster / international) Ann Neurol 2014; 76 (Supp): S223-S224.
46. Magne F, Larrivée-Vanier S, Carré A, Abu-Khudir R, Van Vliet G, Samuels M, Polak M, **Deladoëy J**. Genetic evidence for deficient cell directed migration in children with sublingual thyroid ectopy, the commonest cause of congenital hypothyroidism. Gordon Conference, Cell directed migration, 25-30 January 2015, Galveston TX. (Poster / international)
47. Perlstyn M, **Deladoëy J**, Van Vliet G. Is the dose of levothyroxine required to reach a euthyroid state really higher in school-aged children with congenital vs acquired hypothyroidism. 2015 Pediatric Academic Societies' (PAS) Meeting 2015 (Poster / international).
48. Capo-chichi J, Hasselmann C, **Deladoëy J**, Schwartzentruber J, Majewski J, Van Vliet G, Samuels M. GOLIATH, a variant of DAVID syndrome, is associated with an intronic variant in IKBKE. 2015 Pediatric Academic Societies' (PAS) Meeting 2015 (Poster / international).
49. Bourdet K, Valette S, **Deladoëy J**, Van Vliet G. Contrasting central diabetes insipidus due to preproAVP mutations: earlier onset of symptoms in recessive than in dominant forms. The 54<sup>th</sup> Annual Meeting, European Society for Pediatric Endocrinology (ESPE), 1-3 October 2015, Barcelona, Spain.

#### INVITED PRESENTATIONS / WORKSHOPS / CME

1. Congenital Hypothyroidism from Thyroid Dysgenesis (CHTD): a non-Mendelian Condition? Part II: Molecular Approaches. Second ESPE Workshop on Thyroid Development and its Disorder, 24-25 April 2008, Necker-Enfants Malades Hospital, Paris, France. (**INTERNATIONAL**)
2. Non-Mendelian mechanisms of congenital hypothyroidism from thyroid dysgenesis: role for epigenetics and for de novo copy number variants? Annual Scientific Meeting of the CHU Sainte-Justine Research Center, 4-5 June 2008, Saint-Adèle, Québec, Canada. (**local**)
3. Des mécanismes non-Mendéliens sont-ils impliqués dans les malformations congénitales de la glande thyroïde? Seminar, Department of Biochemistry, Université de Montréal, 6 March 2009, Montréal, Québec, Canada. (**local**)
4. Prise en charge de l'hypothyroïdie congénitale. Club de la Thyroïde Provence 2009 / Grand Sud, 12-13 March 2009, CHU de la Timone, Marseille, France. (**INTERNATIONAL**)
5. Symposium - Non-Mendelian Mechanisms of congenital hypothyroidism. The Endocrine Society's 91<sup>th</sup> Annual Meeting 2009 (ENDO 09), 10-13 June 2009, Washington DC, USA. (**INTERNATIONAL**)
6. Développement de la thyroïde: de l'épidémiologie à la découverte de nouveaux gènes. Réunion scientifique de l'axe de Santé Métabolique / Service de Génétique du CHU Sainte-Justine, 24 February 2010, Montréal, Canada. (**local**)
7. Petite taille chez l'enfant. Formation continue organisée par CSSS de la Haute-Yamaska. Jeudi 13 mai 2010, Grandby, Canada. (**local**)
8. Pediatric Grand Round: Normal values for thyroid functions. 29 February 2012, CHU Sainte-Justine, Montréal, Canada (**local**)

9. Plenary Session – 2012 Henning Andersen Prize of Best Clinical Abstract, for the first description of a biologically inactive ACTH in human. The 51<sup>st</sup> Annual Meeting, European Society for Pediatric Endocrinology (ESPE), 20-23 September 2012, Leipzig, Germany (**INTERNATIONAL**)
10. Debate – Thyroid Nodules: To remove or not remove? That is the question (Con side). Canadian Pediatric Endocrinology Group (CPEG) 2013 Scientific Meeting, 24-26 January 2013, Quebec City, Canada (**NATIONAL**)
11. Scientific Conference of the CHU Sainte-Justine Research Center. Congenital Hypothyroidism: from bedside to bench, and back. CHU Sainte-Justine, 21 March 2013, Montreal, Canada (**local**).
12. Workshop for Residents in Clinical Biochemistry of CHUQ. Thyroid Hormones: norms, variations and confusions. CHUQ – Laval University, 25 March 2013, Québec, Canada (**provincial**).
13. Session – Diagnostic Solved by Next Generation Sequencing: New Causes of Primary Adrenal Failure. Bioinactive ACTH. The Endocrine Society's 95<sup>th</sup> Annual Meeting 2013 (ENDO 13), 15-18 June 2013, San Francisco CA, USA. (**INTERNATIONAL**)
14. Symposium - Congenital Hypothyroidism Revisted, Lecture on The Screening Programs and Epidemiology. The 9<sup>th</sup> Joint Meeting of European Society for Pediatric Society (ESPE), the Pediatric Endocrine Society (PES), the Australasian Pediatric Endocrine Group (APEG), the Asia Pacific Endocrine Society (APES), the Japanese Society for Pediatric Endocrinology (JSPE) and the Latin American Society of Pediatric Endocrinology (SLEP), 19-22 September 2013, Milan, Italy (**INTERNATIONAL**)
15. Workshop for Pediatricians. Interpreting laboratory results: thyroid function test in children. Société Valaisanne de Médecine (CME accredited by the Swiss Society for Pediatrics - SSP), 23 September 2013, Clinique Vigemed, Martigny, Switzerland (**INTERNATIONAL**).
16. Pediatric Grand Round. The use of next generation sequencing in a clinical context: is it that simple? University Children' Hospital (UKBB), University of Basel, 24 September 2013, Basel, Switzerland (**INTERNATIONAL**)
17. Canadian Endocrine Resident's Review Course 2013: Congenital Adrenal Hyperplasia. The 6<sup>th</sup> Annual Canadian Endocrine Residents' Review Course, organized by the Canadian Diabetes Association/Canadian Society for Endocrinology and Metabolism as part of the Vascular 2013 Meeting, 16 October 2013, Montréal, Canada (**NATIONAL**)
18. Endocrine Rounds: Congenital Hypothyroidism: from bedside to bench, and back. Montreal Children Hospital, McGill University, 11 December 2013, Montreal, Canada (**local**)
19. Symposium: Update on the Genetics of Congenital Adrenal Failure. The 17<sup>th</sup> Annual Meeting of the Canadian Diabetes Association – Canadian Society of Endocrinology and Metabolism (CDA/CSEM), 22-25 October 2014, Winnipeg, Manitoba, Canada. (**NATIONAL**)
20. Plenary Session: Charles Hollenberg Young Investigator Award's Lecture. The 17<sup>th</sup> Annual Meeting of the Canadian Diabetes Association – Canadian Society of Endocrinology and Metabolism (CDA/CSEM), 22-25 October 2014, Winnipeg, Manitoba, Canada. (**NATIONAL**)
21. Session: Recent Advances in Genetics of Congenital Hypothyroidism. Canadian Pediatric Endocrinology Group (CPEG) 2015 Scientific Meeting, 19-21 February 2015, Halifax, Canada (**NATIONAL**)
22. Summer School: Genetics of Multiple Pituitary Hormone Deficiency. The 29<sup>th</sup> Summer School of the European Society for Pediatric Endocrinology (ESPE), 28-30 September 2015, Poblet Monastery, Catalonia – Spain. (**INTERNATIONAL**)