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General information

Education

1990	M.D. (Federal Diploma of Medicine), University of Geneva Medical School, Geneva, Switzerland

Postdoctoral Training

10/90-09/92	Resident , Internal Medicine, Fribourg Hospital (Switzerland)
10/92-01/94	Resident , Internal Medicine, Geneva University Hospital (Switzerland)
01/94-12/94	Resident Internal Medicine, Diabetes and Endocrinology, Metabolic Unit, Guys Hospital, London (UK)
01/95-09/96	Chief Resident , Internal Medicine, Geneva University Hospital (Switzerland)
10/96-10/97	Clinical Fellow, Endocrinology , Geneva University Hospital (Switzerland)
10/97-06/98	Chief Resident , Diabetes, Geneva University Hospital (Switzerland)
07/98-07/01	Clinical and Research Endocrine Fellow, Endocrinology and Diabetes Mellitus, Massachusetts General Hospital, Boston, MA

Licensure and Certification

1990	Foreign Medical Graduates Examination (USA)
1990	Full Medical Licensure (Switzerland)
1996	Federal Internal Medicine Board (Switzerland)
1997	Thesis
1998	United States Medical Licensing Examination, Steps I and II
1998	Limited Medical Licensure, Massachusetts
2001	Clinical Effectiveness Program, Harvard School of Public Health
2002	O Visa
2003	United States Medical Licensing Examination, Step III
2003	Temporary Medical Licensure, Massachusetts

2005	Full Medical Licensure, Massachusetts

Academic Appointments

1998-2002	Clinical and Research Fellow in Medicine, Harvard Medical School, Boston, MA
2002-2005	Instructor in Medicine, Harvard Medical School, Boston, MA
2005- 2010	Assistant Professor in Medicine, Massachusetts General Hospital, Boston, MA
2007-	Associate Faculty, Center for Human Genetic Research (CHGR), Massachusetts General Hospital, Boston, MA
2010-	Associate Professor in Medicine, Massachusetts General Hospital, Boston, MA
2010-	**Professor in Medicine, University of Lausanne, Switzerland, Faculty of Biology & Medicine, Lausanne, Switzerland**

Hospital or Affiliated Institution Appointments

07/98-01/02	Clinical Fellow in Endocrinology, Massachusetts General Hospital, Boston, MA
02/02-	Clinical assistant in Medicine, Massachusetts General Hospital, Boston, MA
09/10	**Co-Chief of Endocrinology, Diabetes and Metabolism, University Hospital CHUV, Lausanne, Switzerland**
09/10	**Chief of the Unit of Pediatric Endocrinology, Dept of Pediatric, University Hospital, CHUV, Lausanne, Switzerland**

Hospital and Health Care Organization Clinical Service Responsibilities

2003-	Reproductive Endocrine Associates, Massachusetts General Hospital
2003-	Attending Physician, Endocrine unit, Massachusetts General Hospital

Major Administrative Responsibilities

1995-1996	Coordinator, Clinical Conference Series, Internal Medicine Department, University Hospital Geneva
1996-1998	Coordinator, Clinical Conference Series, Division of Diabetes and Endocrinology, University Hospital Geneva
1996-1998	Coordinator, Journal Club, Division of Diabetes and Endocrinology, University Hospital Geneva
2002-	Coordinator, Clinical Conference Series, Reproductive Endocrine Unit, Massachusetts General Hospital
2005-	Chief head of FGFRE, Massachusetts General Hospital
2007-	Coordinator, U54 Speaker Series, Reproductive Endocrine Unit, Massachusetts General Hospital
2010-	**Co-Chief of Endocrinology, Diabetes and Metabolism, University Hospital CHUV, Lausanne, Switzerland**
2010-	**Chief of the Unit of Pediatric Endocrinology, Dept of Pediatric, University Hospital, CHUV, Lausanne, Switzerland**

Major Committee Assignments

International

2002-2004	Associates Council, Endocrine Society
2012	Membre du Comité d'organisation du Symposium Francophone, Paris, France
2012	Présidente de l'Action Européenne COST « Human GnRH Deficiency »

Professional Societies

1990-	College of Physicians of Switzerland, Member
1990-	Swiss Internal Medicine Society, Member
1996-	Swiss Endocrine Society, Member
1998-	Massachusetts Medical Society, Member
2001-	American Endocrine Society, Member
2002-	American Diabetes Association, Member
2011-	Swiss Scientific National Funds, Member of the Evaluation Committee
2011-	European Society of Endocrinology
2012-	Société Suisse d'Endocrinologie et Diabétologie
2012-	Académie Suisse des Sciences Médicales

Editorial Boards

2001-	Reviewer, Molecular Endocrinology
2005-	Reviewer, Endocrinology
2005-	Reviewer, Clinical Endocrinology
2007-	Reviewer, International Journal of Andrology
2009-	Reviewer, Journal of Clinical Investigation
2009-	Reviewer, European Journal of Human Genetics
2009-	Reviewer, American Journal of Human Genetics

Awards and Honors

1998	Denber Pinard Award for the project "Ketoacidosis in Type II Diabetes", University of Geneva Hospital, Geneva, Switzerland
2000	Endocrine Fellows Foundation Award for the project "The role of FSH in human gonadal development"
2001	Clinical Research Merck Awardee, Endocrine Society
2001	MCR Travel Award, Endocrine Society
2002	Award for the project: "The role of FSH in human gonadal development", Endocrine Fellows Foundation
2004	Claflin Award for Clinical Research :"Spectrum of FGFR1 mutations in Kallmann Syndrome", Massachusetts General Hospital, Boston, MA
2005	Poster of Distinction (Scientific Advisory Committee Meeting), Massachusetts General Hospital, Boston, MA
2006	Poster of Distinction (Scientific Advisory Committee Meeting), Massachusetts General Hospital, Boston, MA

2007	Poster of Distinction, Scientific Advisory Committee Meeting , Massachusetts General Hospital, Boston, MA
2007	Best Abstract, European Pediatric Endocrine Society, Helsinki
2009	Krane Award, Best Translational Research at MGH, Boston, MA

Research, Teaching, and Clinical Contributions

A. Narrative report of Research, Teaching, and Clinical Contributions

I am a translational researcher focused on the neuroendocrine and gonadal control of human reproduction. My approach is collaborative and multidisciplinary and span the fields of human genetics, molecular biology, and crystallography. I also have a commitment to teaching and maintaining a clinical practice in reproductive endocrinology.

Research

One of my areas of research is to delineate the role of FSH in maximizing the biologic potential of the Sertoli cell compartment in immature human testes. The central hypothesis is that optimizing the Sertoli cell compartment is critical for optimal testicular size and maximal spermatogenesis. The Reproductive Endocrine Unit has a long history of clinical experience with patients with idiopathic hypogonadotropic hypogonadism (IHH). This patient population provides a unique disease model to examine whether FSH followed by GnRH therapy is more effective than GnRH alone in producing spermatogenesis in the most severely affected IHH men.

My current NIH funded studies focus on the role of FGF signaling in GnRH neuron development using the human disease model of IHH. Using human genetics, functional and structural studies, my group was first to demonstrate that FGF8 is a critical ligand for FGFR1 in the development of GnRH neurons. This discovery opened the door to explore other components in this pathway as modulators and candidates gene for IHH. I am also studying the role of these rare variants in milder reproductive phenotypes like constitutional delayed of puberty or hypothalamus amenorrhea as well as congenital panhypopituitarism.

Teaching

I am involved in teaching research at several levels including close supervision of undergraduate students, medical students, and endocrine fellows. In addition, I teach clinical endocrinology to clinical fellows and serve as an Endocrine Consultant at MGH. I also lecture in the Harvard Clinical Endocrinology Postgraduate Course.

Clinical

I have an active clinical practice in the Reproductive Endocrine Associates at the Massachusetts General Hospital. My focus is on patients with reproductive disorders including primary and secondary amenorrhea, polycystic ovarian syndrome, menopause, and infertility. The male patients in my practice include men with hypogonadism and/or infertility.

B. Funding Information

1999-2000	P.I., Private Grant/ Roche Research Foundation, Role of FSH in gonadal development
2000-2001	P.I., Private Grant/Eugenio Litta Foundation, Role of FSH in gonadal development
2000-2001	P.I., Private Grant/Novartis Research Grant, Role of FSH in gonadal development
2000-2001	P.I., Endocrine Society Fellowship, Role of FSH in gonadal development
2002-2007	Investigator, N.I.H., R01-HD15788, Male Reproductive Physiology in the Human (PI Crowley, WF, Jr)
2004-2006	P.I., Private Grant, Claflin Distinguished Scholar Award
2005-2010	Key investigator, N.I.H., U54 HD28138, Neuroendocrine and Gonadal Control of Reproduction (Center Director Crowley WF, Jr)
2006-2011	NIH/NIDDK, RO1 DK 071168 (PI : Hayes/Pitteloud), Modulation of Insulin Action by Testosterone in Men
2008-2013	P.I., N.I.H., R01 HD056264, Role of fibroblast growth factor signaling in the neuroendocrine control of reproduction
2011-2014	SNF, 31003A 135648 (PI : Pitteloud), The neuroendocrine control of human reproduction
2012-2015	ERC European Collaboration in Science and Technology (C.O.S.T.). BM1105 (Proposer: Pitteloud), GnRH deficiency: Elucidation of the neuroendocrine control of human reproduction
2012-2015	Swiss-COST SER C12.0106 (PI: Pitteloud), Enhancing bioinformatic approaches for analysis of exome sequencing data generated form GnRH deficient patients
2012-2015	CRSII3-141960 Sinergia FNS (Proposer: Pitteloud), FGF21 is a link between reproduction and energy balance

C. Report of Teaching

1. Local contributions

a. Medical School Courses

1996-1998	Clinical tutorials			
	Tutor	8 Medical Students	*contact time* 46 hours/year for 1 year(s)	*prep time* 46 hours/year for 1 year(s)

1997-1998	Endocrinology and Diabetes Mellitus tutorials			
	Site Director	8 Medical Students	*contact time* 6 hours/week for 12 week(s)	*prep time* 6 hours/week for 12 week(s)

10.03.2011	Partial androgen insensitivity syndrome. Cours à option Nr. 27 « Troubles sexuels », Unil, Lausanne, Suisse
24.03.2011	Diabète. Cours à Option Nr. 25 « La Physiopathologie et la pathologie à partir de cas ». Epalinges, Suisse

11.04.2011	Insuline. Cours Bachelor Biologie 3ème année « Physiology of complex systems ». Epalinges, Suisse
12.04.2011	Hydrates de carbone. Cours Bachelor Biologie 3ème année « Physiology of complex systems ». Epalinges, Suisse
13.04.2011	Reproduction. Cours Bachelor Biologie 3ème année « Physiology of complex systems ». Epalinges, Suisse
27.05.2011	Endocrinologie du cycle ovarien. Cours B2.6 "Urogénital et homéostasie – Endocrinologie", Epalinges Suisse
30.05.2011	Endocrinologie masculin, andrologie / Les hypogonadismes / Retour du cas clinique d'endocrinologie. Cours B2.6 "Urogénital et homéostasie – Endocrinologie", Epalinges Suisse
18.10.2011	Diabète : investigations, complications. Cours M2.2 « Maladies chroniques complexes – Endocrinologie », Lausanne CHUV, Suisse
23.04.2012	Métabolisme glucidique (des hydrates de carbone au glucose). Cours Bachelor Biologie 3ème année « Physiology of complex systems ». Epalinges, Suisse
24.04.2012	Rôle de l'insuline. Cours Bachelor Biologie 3ème année « Physiology of complex systems ». Epalinges, Suisse
25.04.2012	Hormones et reproduction. Cours Bachelor Biologie 3ème année « Physiology of complex systems ». Epalinges, Suisse
29.05.2012	Endocrinologie du cycle ovarien. Cours B2.6 "Urogénital et homéostasie – Endocrinologie", Epalinges, Suisse
30.05.2012	Endocrinologie masculine, andrologie / Les hypogonadismes / Retour du cas clinique d'endocrinologie. Cours B2.6 "Urogénital et homéostasie – Endocrinologie", Epalinges, Suisse
27.11.2012	Cours puberté. Etudiants 2ème année. Unité de reproduction, Dept of Genetic Medicine and Development Laboratory, HUG, Genève
07.03.2013	Endocrinologie/Néonatalogie. Cours à option – Chirurgie pédiatrique – B3.9 (OPT 27), 3ème année, Lausanne
21.03.2013	Endocrinologie Diabète. Cours à option – Physiopathologie et pathologie – B3.9 (OPT 25), 3ème année, Lausanne
11.04.2013	Hormones thyroïdiennes. Cours Bachelor Biologie 3ème année "Physiology of complex systems", Epalinges, Suisse
16.04.2013	Rôle de l'insuline. Cours Bachelor Biologie 3ème année "Physiology of complex systems", Epalinges, Suisse
18.04.2013	Hormones et reproduction. Cours Bachelor Biologie 3ème année "Physiology of complex systems", Epalinges, Suisse
29.05.2013	Endocrinologie masculine, andrologie / Les hypogonadismes / Retour du cas clinique d'endocrinologie. Cours B2.6 "Urogénital et homéostasie – Endocrinologie", Epalinges, Suisse

b. Graduate Medical Courses

1995-1996	Internal Medicine Clinical Conference Series			
	Lecturer	15 Graduate Students	*Contact time :* 4 hours/month for 9 month(s)	*Prep. Time :* 8 hours/month for 9 month(s)

1996-1998	Diabetes Clinical Conference Series			
	Lecturer	2 Medical Students 5 Graduate Students 4 Residents	*Contact time :* 2 hours/month for 9 month(s)	*Prep. Time :* 8 hours/month for 9 month(s)

2001-	Reproductive Endocrine Unit Clinical Conferences Series			
	Lecturer	3 Medical Students 25 Post-doc Students 5 Other Students	*Contact time :* 4 hours/year for 1 year(s)	*Prep. Time :* 12 hours/year for 1 year(s)

2001-	Reproductive Endocrine Unit Journal Club			
	Lecturer	2 Medical Students 6 Graduate Students 6 Post-doc Students	*contact time :* 3 hours/year for 1 year(s)	*prep time :* 9 hours/year for 1 year(s)

2003-	Harvard Internal Medicine Postgraduate Course			
	Lecturer	200 Graduate Students 50 Other Students	*contact time :* 1 hours/year for 1 year(s)	*prep time :* 4 hours/year for 1 year(s)

2006-	Genetics 228			
	Lecturer	25 Graduate Students	*prep time :* 1 hours/year for 1 year(s)	*prep time :* 4 hours/year for 1 year(s)

20.01.2011	Prolactinome. Colloque cas clinique. CHUV, Lausanne, Suisse
15.02.2011	Ménopause. Colloque cas clinique. CHUV, Lausanne, Suisse
18.04.2011	Hyperfonction et hypofonction surrénaliennes. Colloque de formation postgraduée et continue du DMI, CHUV, Lausanne, Suisse
21.06.2011	Gynécomastie. Colloque cas clinique EDM, CHUV, Lausanne
07.07.2011	Les diabètes avec accent sur MODY. Colloque cas clinique EDM, CHUV, Lausanne, Suisse
07.06.2012	Registre européen – déficience en GnRH. Colloque cas clinique EDM, CHUV, Lausanne
30.07.2012	Pathologies hypophysaires et médecine interne. Colloque de formation postgraduée et continue du DMI, CHUV, Lausanne

31.10.2012	Acromégalie. Colloque hebdomadaire du Service de neurologie, CHUV, Lausanne, Suisse

c. Local Invited Presentations

Endocrine Grand Rounds	
2006-	FGFR1 mutations underlying human GnRH deficiency, Massachusetts General Hospital Lecturer: 25 participants, 1 hour contact time per year, 6 hours prep time per year
Grand Rounds	
2001-	The Role of FSH in Gonadal Development, New England Medical Center, Boston Lecturer: 32 participants, 1 hour contact time per year, 6 hours prep time per year
2008-	Gonadotrophin releasing hormone deficiency in human: new molecular and clinical aspects, Brigham and Women's Hospital
2009-	Role of FGF signaling in the neuroendocrine control of reproduction, Massachusetts General Hospital
Paediatric Endocrine Grand Rounds	
2008-	The role of FGF signaling in the neuroendocrine control of human reproduction, Massachusetts General Hospital Lecturer: 75 participants, 1 hour contact time per year, 8 hours prep time per year
Seminar	
2002-	The Role of FSH in Gonadal Development, Children's Hospital Lecturer: 47 participants, 1 hour contact time per year, 6 hours prep time per year
2005-	Role of FGFR1 in neuroendocrine control of reproduction, Brigham and Women's Hospital

d. Continuing Medical Education Courses

2002-	Harvard Clinical Endocrinology Postgraduate Course Lecturer: 30 participants, 1 hour contact time per year, 8 hours prep time per year
2007-	Harvard Clinical Endocrinology Postgraduate Course Lecturer: 225 participants, 1 hour contact time per year, 10 hours prep time per year
2009-	Update in Internal Medicine, American College of Physicians Lecturer: 250 participants, 2 hours contact time per year, 12 hours prep time per year

e. Advisory and Supervisory Responsibilities in Clinical or Laboratory Setting

1995-	4 Residents for 200 hrs/year, Supervising in patient and ambulatory clinic, Geneva University, Switzerland
1997-	2 Diabetic Fellows for 200 hrs/year, Supervising in patient and ambulatory clinic, Geneva University Hospital, Switzerland
2002-	4 Endocrine Fellows for 70 hrs/year, Attending in Endocrinology, Massachusetts General Hospital
2003	1 Undergranduate Student for 80 hrs/year, Precepting in a lab, Massachusetts General Hospital

2004-2005	1 Medical Student for 100 hrs/year, Supervisor of research project, Massachusetts General Hospital
2004-2006	1 Undergraduate Student for 100 hrs/year, Supervisor Research Project, Massachusetts General Hospital
2005-2007	2 Research Endocrine Fellows for 300 hrs/year, Supervisor of reseach project, Massachusetts General Hospital
2006-2007	1 Premedical Student for 300 hrs/year, Supervision of research project, Massachusetts General Hospital
2009-	4 Fellows for 300 hrs/year, Precepting in a lab and in clinical research, Harvard Medical School

f. Leadership Roles

1995-1996	Chief Resident in Internal Medicine, Geneva University Hospital, Switzerland Responsibility: Oversee Internal Medicine residents
1997-1998	Chief Resident Internal Medicine, Diabetes Unit, Geneva University Hospital Responsibility: Oversee Clinical Diabetic Fellows
2001- 2008	Coordinator, Reproductive Endocrine Unit Clinical Conference Series, Massachusetts General Hospital Responsibility: Organize series and identify and invite speakers
2007- 2010	Coordinator, U54 Research Seminar Series, Reproductive Endocrine Unit, Massachusetts General Hospital Responsibility: Organize, identify, and invite guest speakers
2007- 2010	U54 Translational Research Training Program, Massachusetts General Hospital Responsibility: Develop and implement comprehensive educational program for translational research

g. Advisees / Trainees

Training duration	*Name*	*Current position*
2003	Marina Stakes	Dentist student
2003-2004	Astrid Meysing	Assistant ophtalmologist
2004-2010	Lacey Plummer	Assistant Researcher
2004-2005	Marissa Caudill	Psychiatrist Resident, UCLA
2004-2007	John Falardeau	Medical Resident, Mt-Auburn Hospital, Boston, MA
2005	Marina Stakes	Tufts University School of Dental Medicine
2005-2007	Taneli Raivio	Pediatrician, Institute for Biomedicine, Helsinki, Finland
2005-2007	Elka Jacobson-Dickman	Endocrine Pediatric Attending, Maimonides Medical Center, Brooklyn New York
2006-2007	Lindsay Cole	Medical Student, Harvard Medical School, Boston, MA
2008-2010	Cecilia Martin	Research Fellow, Brigham & Womens Hospital, Boston, MA
2008-2010	Bihwa Feng	Research Fellow, Brandeis University, Waltham, MA
2008-2010	Xuan Hoang	Medical Student, University of California San Francisco, San Francisco, CA

2008-2009	Lisa Carona	Medical Student, Ohio State University, Columbus, OH
2008-2009	Sadia Durrani	Medical Student, University of Medicine & Dentistry of New Jersey
2008-2010	Madgdalena Avbelj	Pediatrician, University Children's Hospital, Ljubljana, Slovenia
2009-2010	Sykiotis Gerry	Research Fellow, MGH, accepted in the fall of 2010 as a Endocrine Fellow, University of Patras, Greece
2008-2009	Lisa Carona	Medical Student, Ohio State University, Columbus, OH
2009-	Hichem Miraoui	Research Fellow, CHUV, Lausanne

2. Regional, national or international contributions

a. National

2005-	Phenotypic Spectrum of FGFR1 mutations in human, NIH, Houston, Texas*[Seminar]*
2007-	Role of FSH in gonadal development, Neuroendocrine Unit, Pittsburg University, Pittsburg, PA*[Visiting Professorship]*
2007-	FGF8 is a key ligand for FGFR1 in GnRH ontogeny, Albert Einstein College of Medicine, Bronx, NY*[Invited Lecture]*
2007-	Reproductive Manifestations of Medical and Pediatric Endocrine Diseases, American Society of Reproductive Medicine, Washington DC*[Symposium]*
2008-	Genetics of Hypogonadotropic Hypogonadism, UCSF*[Visiting Professorship]*

b. National from August 2010

30.09.2010	Diagnosis and genetic of hypogonadism. 4ème Journée d'Endocrinologie Vaud-Genève, Morges, Switzerland (invited lecture)
16.11.2010	What is new in GnRH. Visiting Professor. Service Endocrinology & Pediatric Endocrinology, University Inselspital. Bern, Suisse (invited lecture)
09.12.2010	Percer les mystères de la puberté. Leçon inaugurale. CHUV, Lausanne, Suisse
17.12.2010	Des nouveaux gènes contrôlant la reproduction humaine et leur identification. Soirée scientifique de l'Unité de médecine de la reproduction, CHUV, Lausanne (invited lecture)
14.01.2011	Role of FGF signaling in the neuroendocrine control of reproduction. Faber Foundation. Lausanne, Suisse (invited lecture)
31.01.2011	Role of FGF signaling in the neuroendocrine control of reproduction. Visiting professor seminar. Center for Integrative Genomics. Lausanne, Suisse (invited lecture)
19.03.2011	La transition de la pédiatrie vers les adultes. Conférence Turner Syndrome, Bussigny-sur-Lausanne, Suisse (co-invited lecture)
01.04.2011	Diabétologie : Traitement basé sur les valeurs de laboratoire. Primary Care Academy. Montreux, Suisse (invited lecture)
14.04.2011	Puberté tardive : clinique et génétique. Colloque d'endocrinologie pédiatrique. Lausanne, Suisse (invited lecture)

19.05.2011	Genetics basis of hypogonadotrophic hypogonadism. Visiting professor. Service d'endocrinologie, Hôpitaux Universitaires de Genève. Genève, Suisse (invited lecture)
26.05.2011	Le principe d'action des incrétines et leur rôle dans le traitement du diabète de type 2. Symposia Novo Nordisk. Lausanne, Suisse (invited lecture and moderator)
06.10.2011	Endocrinologie de la grossesse. Endocrinologie et grossesse. 5ème Réunion Vaud-Genève d'endocrinologie, Morges, Suisse (invited lecture and co-organizer)
22.09.2011	L'obésité chez l'enfant et adolescent. Colloque d'endocrinologie pédiatrique, Lausanne, CHUV (co-organizer and moderator)
13.10.2011	Traitement des aspects métaboliques du syndrome des ovaires polykystiques. Polycystic Ovarian Syndrome (PCOS) Symposia. CHUV, Lausanne, Suisse (invited lecture, moderator and organizer)
25.10.2011	Human GnRH deficiency: Deciphering molecular pathogenesis. Advanced cell culture techniques for tissue engineering and drug discovery and development. XXII. BD-STCS Workshop 2011, Bâle, Suisse (invited lecture)
03.11.2011	Syndrome des ovaires polykystiques : identification, investigation et suivi. Colloques de l'Unité Multidisciplinaire de Santé des Adolescents (UMSA), CHUV, Lausanne (invited lecture)
08.11.2011	Discussion: importance and implications for daily practice. Advisory Brd Meeting "Degludec The future of insulin therapy, Morges, Suisse
01.12.2011	Aspects cliniques du syndrome des ovaires polykystiques. Les rencontres de Prangins, Prangins, Suisse (invited lecture)
02.12.2011	Male hypogonadism. Session: Continuous Medical Education. Annual SGED Meeting, Berne, Suisse (invited lecture)
06.12.2011	Retard pubertaire et prise en charge. Vidéoconférence des hôpitaux de Suisse romande. Colloque de pédiatrie, CHUV, Lausanne, Suisse (invited lecture)
19.01.2012	Puberté tardive – Kallmann – Projet Européen, Réunion annuelle SSEDP/SGPED, St-Sulpice, Suisse
08.03.2012	Troubles du cycle chez la jeune femme. Place et importance du syndrome des ovaires polykystiques. Colloque de formation de la Sté Neuchâteloise de Médecine, Hôpital de Neuchâtel, Suisse
08.03.2012	Diabète et hypogonadisme : prise en charge. Journée Vaud-Genève de diabétologie 2012, Morges, Suisse (invited lecture and co-organizer)
29.03.2012	Hypogonadisme chez l'homme. Sessions interactives avec cas pratiques. Symposium Médical de l'Institut Central (ICHV) 2012 : L'endocrinologie dans tous ses états. Clinique Romande de Réadaptation SUVA, Sion (invited lecture)
03.05.2012	Progrès récents dans la recherche des incrétines. Nouveautés dans le traitement du diabète de type 2, CHUV, Lausanne, Suisse

18.05.2012	FGF21 a link between metabolism and reproduction. Session: Human genetics and models. FGF Gordon Conference 2012, Les Diablerets, Suisse
07.06.2012	Symposium « Thyroïde : aspects pédiatriques et adultes », CHUV, Lausanne, Suisse (organizer)
14.06.2012	Altérations de la fonction thyroïdienne chez l'enfant et l'adolescent. Colloque d'endocrinologie pédiatrique, CHUV, Lausanne, Suisse (organizer)
30.08.2012	Pathologies hypophysaires. Premières rencontres Endorinologie-Neurochirurgie », CHUV, Lausanne, Suisse (organizer)
01.11.2012	Que proposer à un enfant obèse et sa famille ? Symposium de l'Unité d'obésité pédiatrique CHUV, Hôpital de l'Enfance, Lausanne, Suisse (organizer)
07.11.2012	Morbus Cushing : Statu quo et prise en charge. Atelier Novartis Consultant Network, Lausanne (chairwoman)
16.11.2012	PCO et syndrome métabolique. Les journées NETTER de la Société Européenne de Gynécologie, Montreux, Suisse
13.12.2012	Apport du diagnostic génétique pour les pathologies surrénaliennes. 6éme Journée VD-GE d'endocrinologie « Pathologies surrénaliennes ». Centre de congrès La Longeraie, Morges, Suisse (invited lecture, co-organizer)
04.09.2013	Role of FGF signaling in the neuroendocrine control of reproduction. Séminaire endocrinologie et Diabète Hôpital Universitaire de Bâle (visiting professor et invited lecture)
21.11.2013	Redefining syndromes in the era of exomes. Annual Symposium of the Swiss Endocrine society, Bern
12.12.2013	Fertilité : Evaluation et traitement de l'infertilité chez l'homme : le point de vue de l'endocrinologue. 7ème Journée VD-GE Endocrinologie , La Longeraie, Morges (invited lecture et co-organizer)
23.04.2015	Hirsutismus. Formation pour spécialistes FMH Endocrinologie / Diabétologie (FOSPED), Bienne, Switzerland
04.06.2015	Nouvelles stratégies thérapeutiques dans l'œdème maculaire diabétique. Hôpital ophtalmique Jules-Gonin, Lausanne (invited lecture)
23.06.2015	Traitement de fertilité dans l'hypogonadisme hypogonadotrope masculin. Séminaires hebdomadaires du Service d'Endocrinologie, Diabétologie et Métabolisme, CHUV
27.10.2015	Déficit en testostérone et troubles métaboliques**.** Séminaires hebdomadaires du Service d'Endocrinologie, Diabétologie et Métabolisme, CHUV
29.10.2015	Syndrome des ovaires polykystiques, entrave à la fertilité et haut risque cardiovasculaire sur la durée. 6ème Journée jurassienne d'endocrinologie et de diabétologie, Delémont
03.12.2015	Variations de la différenciation sexuelle (VDS) / Disorders of Sex Development (DSD): un challenge pour la transition (symposium, invited lecture and co-organizer)

14.01.2016	Diagnosis and treatment of hypogonadotropic hypogonadism. Women's Health Congress 2016, University Hospital, Basel (invited lecture)
02.02.2016	Acromégalie : nouveautés. Séminaires hebdomadaires du Service d'Endocrinologie, Diabétologie et Métabolisme, CHUV
12.01.2017	Detection of polycystic ovaries early in puberty - does it make any difference? Women's Health Congress 2017, CHUV (Symposium)
26.01.2017	Endocrinologie de la reproduction. The Year in Endocrinology / Publications incontournables de l'année 2016 (symposium, invited lecture and co-organizer)
16.02.2017	Hypogonadisme chez l'homme (quand faut-il le chercher et qui substituer en testostérone?) Société vaudoise de médecine (Symposium)
02.03.2017	The Year in Diabetes / Publications incontournables de l'année 2016 (symposium, co-organizer)
06.04.2017	The Year in Obesity / Publications incontournables de l'année 2016 (symposium, co-organizer)
07.12.2017	Prolactinome: quand arrêter le traitement d'agonistes dopaminergiques. Rencontre endocrinologie-neurochirurgie (symposium, invited lecture, co-organizer)
18.01.2018	Reproduction. The Year in Endocrinology / Publications incontournables de l'année 2017 (symposium, invited lecture and co-organizer)
15.02.2018	The Year in Diabetes / Publications incontournables de l'année 2017 (symposium, co-organizer)
15.03.2018	The Year in Obesity / Publications incontournables de l'année 2017 (symposium, co-organizer)

c. International

2004-	GPR54 as a regulator of puberty: idiopathic hypogonadotropic hypogonadism, the disease model, 36th International Symposium on GH and Growth Factors*[Invited Lecture, Geneva, Switzerland]*
2005-	IHH as a Digenic Disorder, Genetic Gordon Conference, Newport, RI *[Invited Lecture]*
2005-	Mutations in FGFR1: Wide spectrum of pubertal development., Puberty Conference*[Invited Lecture]*
2007-	Novel insight into the genetics of idiopathic hypogonadotropic hypogonadism, Endocrine Society Meeting, Toronto, CA*[Symposium]*
2009-	Hormone replacement therapy: update, American College of Physicians *[Invited Lecture]*
2009-	PCOS: the metabolic aspects, American College of Physicians *[Invited Lecture]*
2009-	Genetics of idiopathic hypogonadotropic hypogonadism, International Andrology Society, Barcelona Spain *[Invited Lecture]*

2009-	Reversal of Hypogonadotropic Hypogonadism, 11th European Congress of Endocrinology *[Seminar]*
2009-	Gonadotrophin releasing hormone deficiency in human: new molecular and clinical aspects, Conference in Pediatric Endocrinology, Paris *[Invited Lecture]*
2010	Role of FGF signaling in the neuroendocrine control of reproduction. Symposium Gordon Conference "FGF signaling and development", Ventura, CA, USA (invited lecture)
2010	Genetic basis of hypogaondotropic hypogonadism, 42th International Symposium on Endocrinology and Metabolism, Vienna, Austria
2010	Meet the Professor: Klinefelter Syndrome, 92nd Annual Meeting of the Endocrine Society, San Diego, CA, USA

d. International from August 2010

22.09.2010	Genetics of idiopathic hypogonadotropic hypogonadism, 49th Annual Meeting of the European Society for Pediatric Endocrinology (ESPE), Prague, Czeck Republic (invited lecture)
03.02.2011	De l'enfance à l'âge adulte (invited lecture). Conséquences des cancers de l'enfance (moderator). 4ème Symposium Francophone Novo Nordisk. Gand, Belgique
01.03.2011	Role of FSH in human gonadal development: clinical implications for male fertility. Gonal-F Male hypogonadotropic hypogonadism Symposia Tokyo, Japan (invited lecture)
20.05.2011	Gonadotropin sensitivity. 43rd International Symposia on GH and Growth Factors in Endocrinology and Metabolism (KICGS). Rome, Italy (invited lecture)
4-7.06.2011	Management of hypogonadism through puberty. Meet the Professor Lecture. 93rd Annual Meeting of the Endocrine Society. Boston, USA (invited lecture)
4-7.06.2011	Genetic susceptibility to hypothalamic amenorrhea. 93rd Annual Meeting of the Endocrine Society. Boston, USA (invited lecture)
12.09.2011	47th EASD Annual Meeting, Lisbonne
27.09.2011	Monogenic disorders behind hypogonadotropic hypogonadism. Sandoz International Satellite Symposium, ESPE 2011, Glasgow, UK
20.10.2011	Complex genetics of human GnRH deficiency. Institute of Metabolic Science Seminar Series. (Visiting Professor), Cambridge, UK
29.10.2011	Session 3: Hypogonadotropic hypogonadism. Panel discussion: Treatment of hypogonadotropic hypogonadism in male from childhood. 43rd International Symposium on Endocrinology and Metabolism, Tokyo, Japan (invited lecture)
10.11.2011	Novel insights into hypogonadotrophic hypogonadism. 39th Meeting of the British Society for Pediatric Endocrinology and Diabetes. London, England (invited lecture)
25.02.2012	GnRH deficiency: clinical and genetic aspects. Visiting Professor, Assir Central Hospital, Abha, Arabia Saudia

22.03.2012	Novel mutations causing infertility. Session: Clinical Management Workshop 3: Advances in assessment and management of infertility. Society for Endocrinology BES 2012, Harrogate, UK (invited lecture)
27.04.2012	Chez qui dépister un diabète ? 10th Swiss Russian General Practioners Conference 2012, St-Petersbourg, Russie (invited lecture)
05.05.2012	New mechanisms of hypothalamic amenorrhea. Symposium 49: Genetic breakthroughs in reproductive pathology and physiology. 15th International Congress of Endocrinology and 14th European Congress of Endocrinology. Florence, Italy (invited lecture)
20.05.2012	Genetics of idiopathic hypogonadotropic hypogonadism, Neuroendocrine symposium, Shangaï, China
22-26.06.12	Annual Meeting of the Endocrine Society, Houston, USA
12.09.2012	Functional Hypothalamic Amenorrhea: genetic basis. European Neuroendocrine Association (ENEA). Vienna, Austria (invited lecture)
28.09.2012	"Clinical and genetic aspects of hypogonadotropic hypogonadism". 46th Annual Scientific Meeting of the Japanese Society for Pediatric Endocrinology, Osaka, Japan
13.10.2012	Symposium on 'New Genetics of Familial Endocrinopathies' and Symposium on "Male infertility". Annual meeting of the Canadian Society of Endocrinology and Metabolism & the Canadian Diabetes Association, Vancouver, CAN
22.11.2012	SciTech EU 2012 Event, Bruxelles, Belgium
28.12.2012	Delayed Puberty. Session: Normal and abnormal development of the male reproductive tract. 7th European Congress of Andrology 2012, Berlin, Germany.
19.01.2013	Hypogonadisme et diabète. Session on Diabetes and sexuality. Les XIV Rencontres Franco-Suisse (Sanofi-Aventis), Evian France (symposium)
24.04.2013	Genetic basis of congenital hypogonadotropic hypogonadism. German Annual Meeting for Pediatric Endocrinology. Muenster, Germany (symposium)
25.05.2015	Hypogonadisme à l'âge adulte: Flashback sur la puberté. 5ème Symposium francophone (Novo Nordisk). Marseille, France (symposium)
30.08.2013	Oligogenicity in Congenital Hypogonadotropic Hypogonadism. European Joint Scientific Training School COST-FENS-IBRO, Prato, Italy (symposium)
21.09.2013	Genetic aspects of central hypogonadism in the male: The missing genotype-phenotype correlation. 5th International Symposium on Genetics of Male Infertility, Florence, Italy (symposium)
22.09.2013	Redefining syndromes in the era of exomes. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy (symposium)
06.10.2013	Congenital hypogonadotropic hypogonadism. British Society for Endocrinology Clinical Updates Course, Bristol, UK
22.11.2013	Histoire naturelle de pubertés retardées. 34èmes Journées Guéritée, Paris, France (symposium)

06.06.2014	Hormonal control of spermatogenesis in men: Therapeutic aspects in hypogonadotropic hypogonadism. Les 57èmes Journées Internationales d'Endocrinologie Clinique H.P. Klotz, Paris, France (symposium)
08.07.2014	Genetics of congenital hypogonadotropic hypogonadism. 9th Annuel Meeting of the European Neuroscience, Milan, Italy (symposium)
20.09.2014	Lessons drawn from gynecological disorders in relation with hypothalamic-pituitary malfunction. 53rd Annual Meeting of the Society of Pediatric Endocrinology, Dublin, Ireland (symposium)
08.10.2014	FGF21: un lien entre reproduction et métabolisme. 31ème Congrès de la Société Française d'Endocrinologie, Lyon, France (symposium)
22.11.2014	Male hypogonadism: Pubertal development & fertility. 1st Sandoz Endocrinology Debate and Global Exchange, Vienna, Austria (symposium)
12.12.2014	Induction of spermatogenesis in men with hypogonadism. 9th Hammersmith Multidisciplinary Endocrinology Symposium. Hammersmith, UK (symposium)
18.02.2015	The genetics of hypogonadotropic hypogonadism in males and females. ESE Basic Endocrinology Course in Reproductive Endocrinology 2015. Edinburgh, UK.
03.02.2015	Use of Genomics in Elucidating Puberty. 14th International Pituitary Congress, San Diego, USA (symposium)
05.03.2015	FGF21: Metabolism and Reproduction. The Endocrine Society's 97th Annual Meeting and Expo (Endo 2015), San Diego, USA (symposium)
27.04.2015	NGS in the molecular diagnostic of CHH - exome vs targeted genes: What is the best strategy? COST Action BM1105 Joint Scientific Meeting & Training School, Monash University Prato Center, Prato, Italy (symposium)
02.10.2015	Pubertal Induction and Fertility Prospects in Males. 54th Annual Meeting of European Society for Pediatric Endocrinology, Barcelona, Spain (symposium)
03.11.2015	FGF's and Hypogonadism. Society for Endocrinology British Endocrine Society 2015, Edinburgh, UK (symposium)
11.11.2015	Hypogonadotropic hypogonadism: Etiology and treatment. Global Excellence Symposium in Growth and Reproduction, Copenhagen, Denmark (symposium)
27.01.2016	GnRH: An overview of its critical role in puberty and reproductive development. Royal Society of Medicine, London, UK (symposium)
24.09.2016	FGFR mutations in congenital hypogonadotropic hypogonadism. Joint meeting of the International Congress of Endocrinology (ICE) and the China Society of Endocrinology (CSE), Beijing, China (symposium)
23.09.2016	Gonadotrophin replacement for induction of fertility in hypogonadal men. 9th European Congress of Andrology (ECA2016) (Symposium)
28.01.2017	FGF21: a link between metabolism and reproduction. Matajc Day International Second Annual Meeting in Pediatric endocrinology, Bled, Slovenia (Symposium)

17.03.2017	Lecture 1: Hypogonadotropic hypogonadism. Finnish society for pediatric Endocrinology Lecture 2: Transition Clinic – Our experience. Finnish society for pediatric Endocrinology, Rovaniemi, Finland (Symposium)
21.05.2017	Pubertal induction and hormone replacement in young males. 19th European Congress of Endocrinology, Lisbon, Portugal (Symposium)
21.09.2017	FGF21: a peripheral hormone linking metabolism with reproduction in human? 42ème Colloque de la Société de Neuroendocrinologie (SNE), Dijon, France
25.11.2017	Fertility and pregnancy in relation to hypogonadotropic hypogonadism. European Society of Endocrinology (ESE), Krakow, Poland
14.12.2017	Treatment of congenital hypogonadotropic hypogonadism. European Masterclass on fertility and pediatric endocrinology, Utrecht, The Netherlands
12.06.2018	Hypogonadotropic hypogonadism in adolescents. European masterclass on hypogonadotropic hypogonadism, Hoofddorp, The Netherlands

3. Report of Clinical Activities

1998-	Endocrinology, Reproductive Endocrinology Massachusetts General Hospital Clinical Activity Description: My clinical practice is conducted in Reproductive Endocrine Associates at the Massachusetts General Hospital and includes evaluation and treatment of patients with polycystic ovarian syndrome, hypothalamic amenorrhea, hirsutism, male and female infertility, menopause and osteoporosis, delayed puberty, and patients with idiopathic hypogonadotropic hypogonadism and Kallmann syndrome. I also serve as an Attending Physician in the Endocrine Unit. Patient Load: Patients: 400/year; high level of complexity, large number of secondary or tertiary referrals Clinical Contributions: I have expertise in the use of clomiphene, exogenous gonadotropins and pulsatile GnRH for women with anovulatory or idiopathic infertility. Our ovulation induction program is distinguished by high success rates and low rates of multiple gestation. I have elucidated the clinical predictors of outcome on pulsatile GnRH therapy in men with idiopathic hypogonadotropic hypogonadism (IHH) and developed a study of the effect of recombinant FSH prior to pulsatile GnRH therapy for men with IHH, a therapy that could potentially maximizes fertility.

Bibliography

Original articles

1	**Pitteloud N**, Binz K, Caulfield A, Philippe J. Ketoacidosis during gestational diabetes. Case report. Diabetes Care. 1998;21(6):1031-2.
2	**Pitteloud N**, Philippe J. Characteristics of Caucasian type 2 diabetic patients during ketoacidosis and at follow-up. Schweiz Med Wochenschr. 2000;130(16):576-82.
3	Beranova M, Oliveira LM, Bédécarrats GY, Schipani E, Vallejo M, Ammini AC, Quintos JB, Hall JE, Martin KA, Hayes FJ, **Pitteloud N**, Kaiser UB, Crowley WF, Seminara SB. Prevalence, phenotypic spectrum, and modes of inheritance of gonadotropin-releasing hormone receptor mutations in idiopathic hypogonadotropic hypogonadism. J Clin Endocrinol Metab. 2001;86(4):1580-8.
4	**Pitteloud N**, Boepple PA, DeCruz S, Valkenburgh SB, Crowley WF, Hayes FJ. The fertile eunuch variant of idiopathic hypogonadotropic hypogonadism: spontaneous reversal associated with a homozygous mutation in the gonadotropin-releasing hormone receptor. J Clin Endocrinol Metab. 2001;86(6):2470-5.
5	Hayes FJ, **Pitteloud N**, DeCruz S, Crowley WF, Boepple PA. Importance of inhibin B in the regulation of FSH secretion in the human male. J Clin Endocrinol Metab. 2001;86(11):5541-6.
6	**Pitteloud N**, Hayes FJ, Boepple PA, DeCruz S, Seminara SB, MacLaughlin DT, Crowley WF. The role of prior pubertal development, biochemical markers of testicular maturation, and genetics in elucidating the phenotypic heterogeneity of idiopathic hypogonadotropic hypogonadism. J Clin Endocrinol Metab. 2002;87(1):152-60.
7	Pitteloud N, Hayes FJ, Dwyer A, Boepple PA, Lee H, Crowley WF. Predictors of outcome of long-term GnRH therapy in men with idiopathic hypogonadotropic hypogonadism. J Clin Endocrinol Metab. 2002;87:4128-36.
8	**Pitteloud N**, Villegas J, Dwyer AA, Crowley WF, McPhaul MJ, Hayes FJ. Acute stress masking the biochemical phenotype of partial androgen insensitivity syndrome in a patient with a novel mutation in the androgen receptor. J Clin Endocrinol Metab. 2004;89(3):1053-8.
9	**Pitteloud N**, Acierno JS, Meysing AU, Dwyer AA, Hayes FJ, Crowley WF. Reversible Kallmann Syndrome, delayed puberty, and isolated anosmia occurring in a single family with a mutation in the FGFR1 gene. J Clin Endocrinol Metab. 2005;90(3):1317-22.
10	**Pitteloud N**, Hardin M, Dwyer AA, Valassi E, Yialamas M, Elahi D, Hayes FJ. Increasing insulin resistance is associated with a decrease in Leydig cell testosterone secretion in men. J Clin Endocrinol Metab. 2005;90(5):2636-41.
11	**Pitteloud N**, Mootha VK, Dwyer AA, Hardin M, Lee H, Eriksson K-F, Tripathy D, Yialamas M, Groop L, Elahi D, Hayes FJ. Relationship between testosterone levels, insulin sensitivity and mitochondrial function in men. Diabetes Care, 2005;28(7):1636-42.
12	Kumar, PA, **Pitteloud N** (first Co-author), Andrews PA, Dwyer AA, Hayes F, Crowley WF, Dym M. Testis morphology in patients with idiopathic hypogonadotropic hypogonadism. Hum Reprod. 2006;21:1033-40.

13	**Pitteloud N**, Acierno J.S, jr., Meysing A., Eliseenkova A.V., Ma J., Ibrahimi O.A., Metzger D., Hayes F.J., Dwyer A.A., Hughes V.A., Yialamas, M., Hall J.E., Grant E., Mohammadi M., Crowley W.F.Jr. 2006. Mutations in FGFR1 cause both Kallmann syndrome and normosmic idiopathic hypogonadotropic hypogonadism. PNAS. 2006;103:6281-6.
14	**Pitteloud N**, Meysing A, Quinton R, Acierno JS, Dwyer AA, Plummer L, Fliers E, Boepple PA, Hayes FJ, Seminara SB, Bouloux P, Mohammadi M, Crowley WF, Jr. . Mutations in Fibroblast Growth Factor Receptor 1 cause Kallmann syndrome with a wide spectrum of reproductive phenotypes. Mol Cell Endocrinol. 2006;254-255:60-9.
15	Pallais JC, Bo-Abbas Y, **Pitteloud N**, Crowley WF Jr, Seminara SB. . Neuroendocrine, gonadal, placental, and obstetric phenotypes in patients with IHH and mutations in the G-protein coupled receptor, GPR54. Mol Cell Endocrinol. 2006;254-255:70-7.
16	Cerrato F, Shagoury J, Kralickova M, Dwyer A, Falardeau J, Ozata M, Van Vliet G, Bouloux P, Hall JE, Hayes FJ, **Pitteloud N**, Martin KA, Welt C, Seminara SB. Coding sequence analysis of GNRHR and GPR54 in patients with congenital and adult-onset forms of hypogonadotropic hypogonadism. Eur J Endocrinol. 2006;155:Suppl 1:S3-S10.
17	**Pitteloud N**, Quinton R, Pearce S, Raivio T, Acierno J, Dwyer A, Plummer L, Hughes V, Seminara S, Cheng YZ, Li WP, Maccoll G, Eliseenkova AV, Olsen SK, Ibrahimi OA, Hayes FJ, Boepple P, Hall JE, Bouloux P, Mohammadi M, Crowley W. Digenic mutations account for variable phenotypes in idiopathic hypogonadotropic hypogonadism. J Clin Invest. 2007;117 :457-63.
18	Yialamas MA, Dwyer AA, Hanley E, Lee H, **Pitteloud N**, Hayes FJ. Acute sex steroid withdrawal reduces insulin sensitivity in healthy men with idiopathic hypogonadotropic hypogonadism. J Clin Endocrinol Metab. 2007;92:4254-4259.
19	Raivio T, Falardeau J, Dwyer A, Quinton R, Hayes FJ, Hughes VA, Cole LW, Pearce SH, Lee H, Boepple P, Crowley WF, **Pitteloud N**. Reversal of idiopathic hypogonadotropic hypogonadism. N Engl J Med. 2007;357 :863-73.
20	**Pitteloud N**, Zhang C, Pignatelli D, Li JD, Raivio T, Cole LW, Plummer L, Jacobson-Dickman EE, Mellon PL, Zhou QY, Crowley WF Jr. Loss-of-function mutation in the prokineticin 2 gene causes Kallmann syndrome and normosmic idiopathic hypogonadotropic hypogonadism. Proc Natl Acad Sci U S A. 2007;104 :17447-52.
21	**Pitteloud N**, Dwyer AA, Decruz S, Lee H, Boepple PA, Crowley WF Jr, Hayes FJ. Inhibition of LH Secretion by Testosterone in Men Requires Aromatization for its Pituitary but not its Hypothalamic Effects: Evidence from the Tandem Study of Normal and Gonadotropin-Releasing Hormone-Deficient Men. Journal of Clin Endocrinol Metab. 2008;(93):784-91.
22	**Pitteloud N**, Dwyer AA, Decruz S, Lee H, Boepple PA, Crowley WF Jr, Hayes FJ. The Relative Role of Gonadal Sex Steroids and Gonadotropin-Releasing Hormone Pulse Frequency in the Regulation of FSH Secretion in Men. Journal of Clin Endocrinol Metab. 2008;93:2686-92.
23	Boepple PA, Hayes FJ, Dwyer AA, Raivio T, Lee H, Crowley WF Jr, **Pitteloud N**. Relative roles of inhibin B and sex steroids in the negative feedback regulation of follicle-stimulating hormone in men across the full spectrum of seminiferous epithelium function. Journal of Endocrinology and Metabolism. 2008;93:1809-14.

24	Cole LW, Sidis Y, Zhang C, Quinton R, Plummer L, Pignatelli D, Hughes VA, Dwyer AA, Raivio T, Hayes FJ, Seminara SB, Huot C, Alos N, Speiser P, Takeshita A, Van Vliet G, Pearce S, Crowley WF Jr, Zhou QY, **Pitteloud N**. Mutations in Prokineticin 2 (PROK2) and PROK2 Receptor (PROKR2) in Human Gonadotrophin-Releasing Hormone Deficiency: Molecular Genetics and Clinical Spectrum. Journal of Endocrinology and Metabolism. 2008;93:3551-59.
25	Falardeau J, Chung WC, Beenken A, Raivio T, Plummer L, Sidis Y, Jacobson-Dickman EE, Eliseenkova AV, Ma J, Dwyer A, Quinton R, Na S, Hall JE, Huot C, Alois N, Pearce SH, Cole LW, Hughes V, Mohammadi M, Tsai P, **Pitteloud N**. Decreased FGF8 signaling causes deficiency of gonadotropin-releasing hormone in humans and mice. J. Clin. Invest. 2008; 118 :2822-31.
26	Chen H, Xu CF, Ma J, Eliseenkova AV, Li W, Pollock PM, **Pitteloud N**, Miller WT, Neubert TA, Mohammadi M. Proc Natl Acad Sci U S A. 2008 Dec 16;105(50):19660-5. Epub 2008 Dec 5 . A crystallographic snapshot of tyrosine trans-phosphorylation in action. Proc Natl Acad Sci U S A. 2008;105:19660-5.
28	Jongmans MC, van Ravenswaaij-Arts CM, **Pitteloud N**, Ogata T, Sato N, Claahsen-van der Grinten HL, van der Donk K, Seminara S, Bergman JE, Brunner HG, Crowley WF Jr, Hoefsloot LH. . CHD7 mutations in patients initially diagnosed with Kallmann syndrome--the clinical overlap with CHARGE syndrome. Clin Genet. 2009;75:65-71.
29	**Pitteloud N**, Thambundit A, Dwyer AA, Falardeau JL, Plummer L, Caronia LM, Hayes FJ, Lee H, Boepple PA, Crowley WF Jr. Role of seminiferous tubular development in determining the FSH versus LH responsiveness to GnRH in early sexual maturation. Neuroendocrinology. 2009;90(3):260-8.
30	Raivio T, Sidis Y, Plummer L, Chen H, Ma J, Mukherjee A, Jacobson-Dickman E, Quinton R, Van Vliet G, Lavoie H, Hughes VA, Dwyer A, Hayes FJ, Xu S, Sparks S, Kaiser UB, Mohammadi M, **Pitteloud N**. Impaired fibroblast growth factor receptor 1 signaling as a cause of normosmic idiopathic hypogonadotropic hypogonadism. J Clin Endocrinol Metab. 2009 Nov;94(11):4380-90.
31	Chan YM, de Guillebon A, Lang-Muritano M, Plummer L, Cerrato F, Tsiaras S, Gaspert A, Lavoie HB, Wu CH, Crowley WF Jr, Amory JK, **Pitteloud N**, Seminara SB. GNRH1 mutations in patients with idiopathic hypogonadotropic hypogonadism. Proc Natl Acad Sci U S A. 2009 Jul 14;106(28):11703-8.
32	Sykiotis GP, Plummer L, Hughes VA, Au M, Durrani S, Nayak-Young S, Dwyer AA, Quinton R, Hall JE, Gusella JF, Seminara SB, Crowley WF Jr, **Pitteloud N**. Oligogenic basis of isolated gonadotropin-releasing hormone deficiency. Proc Natl Acad Sci U S A. 2010 Aug 24;107(34):15140-4.
33	Dwyer AA, Hayes FJ, Plummer L, **Pitteloud N**, Crowley WF Jr. The long-term clinical follow-up and natural history of men with adult-onset idiopathic hypogonadotropic hypogonadism. J Clin Endocrinol Metab. 2010 Sep;95(9):4235-43.
34	Trarbach EB, Abreu AP, Silveira LF, Garmes HM, Baptista MT, Teles MG, Costa EM, Mohammadi M, **Pitteloud N**, Mendonca BB, Latronico AC. Nonsense mutations in FGF8 gene causing different degrees of human gonadotropin-releasing deficiency. J Clin Endocrinol Metab. 2010 Jul;95(7):3491-6.

35	Sykiotis GP, Hoang XH, Avbelj M, Hayes FJ, Thambundit A, Dwyer A, Au M, Plummer L, Crowley WF Jr, **Pitteloud N**. Congenital idiopathic hypogonadotropic hypogonadism: evidence of defects in the hypothalamus, pituitary, and testes. J Clin Endocrinol Metab. 2010 Jun;95(6):3019-27.
36	Caronia LM, Martin C, Welt CK, Sykiotis GP, Quinton R, Thambundit A, Avbelj M, Dhruvakumar S, Plummer L, Hughes VA, Seminara SB, Boepple PA, Sidis A, Crowley Jr WF, Martin KA, Hall JE, **Pitteloud N**. A genetic basis for functional hypothalamic amenorrhea. N Engl J Med. 2011 Jan:364(3):215-25
37	Martin C, Balasubramanian R, Dwyer AA, Au MG, Sidis Y, Kaiser UB, Seminara SB, **Pitteloud N**, Zhou QY, Crowley WF Jr. The role of the prokineticin 2 pathway in human reproduction: evidence from the study of human and murine gene mutations. Endocr Rev 2011 Apr:32(2):225-46
38	Shaw ND, Seminara SB, Welt CK, Au MG, Plummer L, Hughes VA, Dwyer AA, Martin KA, Quinton R, Mericq V, Merino PM, Gusella JF, Crowley WF Jr, **Pitteloud N**, Hall JE. Expanding the phenotype and genotype of female GnRH deficiency. J Clin Endocrinol Metab 2011 Mar:96(3):E566-76
39	Trabado S, Maione L, Salenave S, Baron S, Galland F, Bry-Gauillard H, Guiochon-Mantel A, Chanson P, **Pitteloud N**, Sinisi AA, Brailly-Tabard S, Young J. Estradiol levels in men with congenital hypogonadotropic hypogonadism and the effects of different modalities of hormonal treatment. Fertil Steril 2011 Jun;95(7):2324-9, 2329.e1-3. Epub 2011 May 4. PMID: 21536274
40	Tornberg J, Sykiotis GP, Keefe K, Plummer L, Hoang X, Hall JE, Quinton R, Seminara SB, Hughes V, Van Vliet G, Van Uum S, Crowley WF, Habuchi H, Kimata K, **Pitteloud N**, Bülow HE. Heparan sulfate 6-O-sulfotransferase 1, a gene involved in extracellular sugar modifications, is mutated in patients with idiopathic hypogonadotropic hypogonadism. Proc Natl Acad Sci U S A. 2011 Jul 12;108(28):11524-9. Epub 2011 Jun 23. PMID: 21700882
41	McCabe MJ, Gaston-Massuet C, Tziaferi V, Gregory LC, Alatzoglou KS, Signore M, Puelles E, Gerrelli D, Farooqi IS, Raza J, Walker J, Kavanaugh SI, Tsai PS, **Pitteloud N**, Martinez-Barbera JP, Dattani MT. Novel FGF8 mutations associated with recessive holoprosencephaly, craniofacial defects, and hypothalamo-pituitary dysfunction. J Clin Endocrinol Metab. 2011 Oct;96(10):E1709-18. Epub 2011 Aug 10. PMID: 21832120
42	Chan YM, Broder-Fingert S, Paraschos S, Lapatto R, Au M, Hughes V, Bianco SD, Min L, Plummer L, Cerrato F, De Guillebon A, Wu IH, Wahab F, Dwyer A, Kirsch S, Quinton R, Cheetham T, Ozata M, Ten S, Chanoine JP, **Pitteloud N**, Martin KA, Schiffmann R, Van der Kamp HJ, Nader S, Hall JE, Kaiser UB, Seminara SB. GnRH-deficient phenotypes in humans and mice with heterozygous variants in KISS1/Kiss1. J Clin Endocrinol Metab. 2011 Nov;96(11):E1771-81. Epub 2011 Aug 31. PMID: 21880801
43	Lewkowitz-Shpuntoff HM, Hughes VA, Plummer L, Au MG, Doty RL, Seminara SB, Chan YM, **Pitteloud N**, Crowley WF Jr, Balasubramanian R. Olfactory phenotypic spectrum in idiopathic hypogonadotropic hypogonadism: Pathophysiological and genetic implications. J Clin Endocrinol Metab 2012 Apr 97(1):E136-44. PMID: 22072740

44	Raivio T, Avbelj M, McCabe MJ, Romero CJ, Dwyer AA, Tommiska J, Sykiotis GP, Gregory LC, Diaczok D, Tziaferi V, Elting MW, Padidela R, Plummer L, Martin C, Feng B, Zhang C, Zhou QY, Quinton R, Sidis Y, Radovick S, Dattani MT, **Pitteloud N**. Genetic overlap in Kallmann syndrome, combined pituitary hormone deficiency, and septo-optic dysplasia. J Clin Endocrinol Metab 2012 Apr 97(4):E694-9. PMID: 22319038
45	Goetz R, Ohnishi M, Ding X, Kurosu H, Wang L, Akiyoshi J, Ma J, Gai W, Sidis Y, **Pitteloud N**, Kuro-O M, Razzaque MS, Mohammadi M. Klotho coreceptors inhibit signaling by paracine fibroblast growth factor 8 subfamily ligands. Mol Cell Biol 2012 May 32(10):1944-54. PMID: 22451487
46	Gianetti E, Hall JE, Au MG, Kaiser UB, Quinton R, Stewart JA, Metzger DL, **Pitteloud N**, Mericq V, Merino PM, Levitsky LL, Izatt L, Lang-Muritano M, Fujimoto VY, Dluhy RG, Chase ML, Crowley WFJr, Plummer L, Seminara SB. When genetic load does not correlate with phenotypic spectrum: lessons from the GnRH receptor (GNRHR). J Clin Endocirnol Metab 2012 Sep 97(9):E1798-807. PMID:22745237
47	Avbelj Stefanija M, Jeanpierre M, Sykiotis GP, Young J, Quinton R,Abreu AP, Plummer L, Au MG, Balasubramanian R, Dwyer AA,Florez JC, Cheetham T, Pearce SH, Purushothaman R, Schinzel A, Pugeat M, Jacobson-Dickman EE, Ten S, Latronico AC, Gusella JF, Dode C, Crowley WF Jr, **Pitteloud N**. Hum Mol Genet 2012 Oct 21(19): 4314-24. PMID 22773735
48	Caronia LM, Dwyer AA, Hayden D, Amati F, **Pitteloud N**, Hayes FJ. Abrupt decrease in serum testosterone levels after an oral glucose load in men: implications for screening for hypogonadism. Clin Endocrinol (Oxf). 2012 (78 (2): 291-296. PMID 22804876
49	Koika V, Varnavas P, Valavani H, Sidis Y, Plummer L, Dwyer A, Quinton R, Kanaka-Gantenbein C, **Pitteloud N**, Sertedaki A, Dacou-Voutetakis C, Georgopoulos NA. Comparative functional analysis of two fibroblast growth factor receptor 1 (FGFR1) mutations affecting the same residue (R254W and R254Q) in isolated hypogonadotropic hypogonadism (IHH). Gene. 2013; 516(1):146-51. PMID:23276709
50	Abel BS, Shaw ND, Brown JM, Adams JM, Alati T, Martin KA, **Pitteloud N**, Seminara SB, Plummer L, Pignatelli D, Crowley WF Jr, Welt CK, Hall JE. Responsiveness to a physiological regimen of GnRH therapy and relation to genotype in women with isolated hypogonadotropic hypogonadism. J Clin Endocrinol Metab 98:E206-16, 2013 J Clin Endocrinol Metab. 2013; 98(2):E206-16. PMID:23341491
51	McCabe MJ, Gaston-Massuet C, Gregory LC, Alatzoglou KS, Tziaferi V, Sbai O, Rondard P, Masumoto KH, Nagano M, Shigeyoshi Y, Pfeifer M, Hulse T, Buchanan CR, **Pitteloud N**, Martinez-Barbera JP, Dattani MT. Variations in PROKR2, but not PROK2, are associated with hypopituitarism and septo-optic dysplasia. J Clin Endocrinol Metab 2013; 98(3):E547-57. PMID:23386640
52	Costa-Barbosa FA, Balasubramanian R, Keefe KW, Shaw ND, Al-Tassan N, Plummer L, Dwyer AA, Buck CL, Choi JH, Seminara SB, Quinton R, Monies D, Meyer B, Hall JE, **Pitteloud N**, Crowley WF Jr. Prioritizing genetic testing in patients with Kallmann syndrome using clinical phenotypes. J Clin Endocrinol Metab. 2013; 98(5):E943-53. PMID:23533228

53	Miraoui H, Dwyer AA, Sykiotis GP, Plummer L, Chung W, Feng B, Beenken A, Clarke J, Pers TH, Dworzynski P, Keefe K, Niedziela M, Raivio T, Crowley WF Jr, Seminara SB, Quinton R, Hughes VA, Kumanov P, Young J, Yialamas MA, Hall JE, Van Vliet G, Chanoine JP, Rubenstein J, Mohammadi M, Tsai PS, Sidis Y, Lage K, **Pitteloud N**. Mutations in FGF17, IL17RD, DUSP6, SPRY4, and FLRT3 are identified in individuals with congenital hypogonadotropic hypogonadism. Am J Hum Genet. 2013; 92(5):725-43. PMID:23643382
54	Wiehle R, Cunningham GR, **Pitteloud N**, Wike J, Hsu K, Fontenot GK, Rosner M, Dwyer A, Podolski J. Testosterone Restoration by Enclomiphene Citrate in Men with Secondary Hypogonadism: Pharmacodynamics and Pharmacokinetics. BJU Int. 2013 Jul 12. doi: 10.1111/bju.12363. PMID: 23875626
55	Dwyer AA, Sykiotis GP, Hayes FJ, Boepple PA, Lee H, Loughlin KR, Dym M, Sluss PM, Crowley WF Jr, **Pitteloud N**. Trial of recombinant follicle-stimulating hormone pretreatment for GnRH-induced fertility in patients with congenital hypogonadotropic hypogonadism. J Clin Endocrinol Metab. 2013; 98(11):E1790-5. PMID: 24037890
56	Sidhoum VF, Chan YM, Lippincott MF, Balasubramanian R, Quinton R, Plummer L, Dwyer A, **Pitteloud N**, Hayes FJ, Hall JE, Martin KA, Boepple PA, Seminara SB. Reversal and relapse of hypogonadotropic hypogonadism: resilience and fragility of the reproductive neuroendocrine system. J Clin Endocrinol Metab. 2014; 99(3):861-70. PMID: 24423288
57	Balasubramanian R, Cohen DA, Klerman EB, Pignatelli D, Hall JE, Dwyer AA, Czeisler CA, **Pitteloud N**, Crowley WF. Absence of central circadian pacemaker abnormalities in humans with loss of function mutation in prokineticin 2. J Clin Endocrinol Metab. 2014; 99(3):E561-6. PMID: 24423319
58	Känsäkoski J, Fagerholm R, Laitinen EM, Vaaralahti K, Hackman P, **Pitteloud N**, Raivio T, Tommiska J. Mutation screening of SEMA3A and SEMA7A in patients with congenital hypogonadotropic hypogonadism. Pediatr Res. 2014; 75(5):641-4. PMID: 2452209
59	Dwyer AA, Quinton R, Morin D, **Pitteloud N**. Identifying the unmet health needs of patients with congenital hypogonadotropic hypogonadism using a web-based needs assessment: implications for online interventions and peer-to-peer support. Orphanet J Rare Dis. 2014 Jun 11;9 (1):83. PMID:24915927
60	Villanueva C*, Jacobson-Dickman E*, Cheng Xu C, Manouvrier S, Dwyer AA, Sykiotis GP, Beenken A, Liu Y, Tommiska J, Hu Y, Tiosano D, Gerard M, Leger J, Drouin-Garraud V, Lefebvre H, Polak M, Carel JC, Phan-Hug F, Hauschild M, Plummer L, Rey JP, Raivio TBouloux P, Sidis Y,Mohammadi M, de Roux N, **Pitteloud N**. Congenital hypogonadotropic hypogonadism with split hand/foot malformation: a clinical entity with a high frequency of FGFR1 mutations. Genet Med. 2014 Aug;17(8):651-9. (* denotes co-first authorship) PMID:25394172
61	Deillon E, Hauschild M, Faouzi M, Stoppa-Vaucher S, Elowe-Gruau E, Dwyer A, Theintz GE, Dubuis JM, Mullis PE, **Pitteloud N**, Phan-Hug F. Natural History of Growth Hormone Deficiency in a Pediatric Cohort. Horm Res Paediatr. 2015;83(4):252-61. PMID: 25676059
62	Correa FA, Trarbach EB, Tusset C, Latronico AC, Montenegro LR, Carvalho LR, Franca MM, Otto AP, Costalonga EF, Brito VN, Abreu AP, Nishi MY, Jorge AA, Arnhold IJ, Sidis Y, **Pitteloud N**, Mendonca BB. FGFR1 and PROKR2 rare variants found in patients with combined pituitary hormone deficiencies. Endocr Connect. 2015 Jun;4(2):100-7. PMID: 25759380

63	Dwyer AA, Quinton R, **Pitteloud N**, Morin D. Psychosexual development in men with congential hypogonadotropic hypogonadism on long-term treatment: A mixed-methods study. Sex Med. Mar; 3(1):32-41. PMID: 25844173
64	Ducry J, Gomez F, Prior JO, Boubaker A, Matter M, Monti M, Pu Y, **Pitteloud N**, Portmann L. Mid-gut ACTH-secreting neuroendocrine tumor unmasked with (18)F-dihydroxyphenylalanine-positron emission tomography. Endocrinol Diabetes Metab Case Rep. 2015;2015:140104. PMID: 25861450
65	Tommiska J, Känsäkoski J, **Pitteloud N**, Wu F, Raivio T. Gonadotropin-releasing hormone receptor mutations in ageing men. Clin Endocrinol (Oxf). 2016 Jan;84(1):150-1. doi: 10.1111/cen.12833. Epub 2015 Jul 1. No abstract available. PMID: 26044071
66	Lamine F, Lalubin F, **Pitteloud N**, Burnier M, Zanchi A. Chronic kidney disease in type 2 diabetic patients followed-up by primary care physicians in Switzerland: prevalence and prescription of antidiabetic drugs. Swiss Med Wkly. 146:w14282. 2016. PMID: 26922155
67	Starnoni D, Daniel RT, Marino L, **Pitteloud N**, Levivier M, Messerer M. Surgical treatment of acromegaly according to the 2010 remission criteria: systematic review and meta-analysis. Acta Neurochir (Wien). 2016 Nov;158(11):2109-2121. Epub 2016 Sep 2. PMID: 27586125
68	Dwyer AA, Tiemensma J, Quinton R, **Pitteloud N**, Morin D. Adherence to treatment in men with hypogonadotrophic hypogonadism. Clin Endocrinol (Oxf). 2017 Mar;86(3):377-383. doi: 10.1111/cen.13236. Epub 2017 Jan 11. PMID: 27647266
69	**Pitteloud N**, Gamulin A, Barea C, Damet J, Racloz G, Sans-Merce M. Radiation exposure using the O-arm® surgical imaging system. Eur Spine J. 2017 Mar;26(3):651-657. doi: 10.1007/s00586-016-4773-0. Epub 2016 Sep 21. PMID: 27652675
70	Xu C, Lang-Muritano M, Phan-Hug F, Dwyer AA, Sykiotis GP, Cassatella D, Acierno J Jr, Mohammadi M, **Pitteloud N**. Genetic testing facilitates prepubertal diagnosis of congenital hypogonadotropic hypogonadism. Clin Genet. 2017 Feb 14. doi: 10.1111/cge.12996. [Epub ahead of print] PMID: 28195315
71	Christou F, **Pitteloud N**, Gomez F. The induction of ovulation by pulsatile administration of GnRH: an appropriate method in hypothalamic amenorrhea. Gynecol Endocrinol. 2017 Mar 6:1-4. doi: 10.1080/09513590.2017.1296948. [Epub ahead of print] PMID: 28277105
72	Marcos S, Monnier C, Rovira Algans X, Fouveaut C, **Pitteloud N**, Ango F, Dodé C, Hardelin JP. Defective signaling through plexin-A1 compromises the development of the peripheral olfactory system and neuroendocrine reproductive axis in mice. Hum Mol Genet. 2017 Mar 2. doi: 10.1093/hmg/ddx080. [Epub ahead of print] PMID: 28334861
73	Phan-Hug F, Kraus C, Paoloni-Giacobino A, Fellmann F, Typaldou SA, Ansermet F, Alamo L, Eggert N, Pelet O, Vial Y, Muehlethaler V, Birraux J, Ramseyer P, Renteria SC, Dwyer A, **Pitteloud N**, Meyrat BJ. [Patients with variations of sex development: an example of interdisciplinary care]. Rev Med Suisse. 2016 Nov 9;12(538):1923-1929. French. PMID: 28696598
74	Dzemaili S, Tiemensma J, Quinton R, **Pitteloud N**, Morin D, Dwyer A. Beyond hormone replacement: quality of life in women with congenital hypogonadotropic hypogonadism. Endocr Connect. 2017 Jul 11. pii: EC-17-0095. doi: 10.1530/EC-17-0095. [Epub ahead of print] PMID: 28698240

75	Xu C, Messina A, Somm E, Miraoui H, Kinnunen T, Acierno J Jr, Niederländer NJ, Bouilly J, Dwyer AA, Sidis Y, Cassatella D, Sykiotis GP, Quinton R, De Geyter C, Dirlewanger M, Schwitzgebel V, Cole TR, Toogood AA, Kirk JM, Plummer L, Albrecht U, Crowley WF Jr, Mohammadi M, Tena-Sempere M, Prevot V, **Pitteloud N**. 2017. KLB, encoding β-Klotho, is mutated in patients with congenital hypogonadotropic hypogonadism. EMBO Mol Med. Oct;9(10):1379-1397. PMID: 28754744
76	Tommiska J, Känsäkoski J, Skibsbye L, Vaaralahti K, Liu X, Lodge EJ, Tang C, Yuan L, Fagerholm R, Kanters JK, Lahermo P, Kaunisto M, Keski-Filppula R, Vuoristo S, Pulli K, Ebeling T, Valanne L, Sankila EM, Kivirikko S, Lääperi M, Casoni F, Giacobini P, Phan-Hug F, Buki T, Tena-Sempere M, **Pitteloud N**, Veijola R, Lipsanen-Nyman M, Kaunisto K, Mollard P, Andoniadou CL, Hirsch JA, Varjosalo M, Jespersen T, Raivio T. 2017. Two missense mutations in KCNQ1 cause pituitary hormone deficiency and maternally inherited gingival fibromatosis. Nat Commun. Nov 3;8(1):1289. PMID: 2909770
77	Xu C, Cassatella D, van der Sloot AM, Quinton R, Hauschild M, De Geyter C, Flück C, Feller K, Bartholdi D, Nemeth A, Halperin I, Pekic Djurdjevic S, Maeder P, Papadakis G, Dwyer AA, Marino L, Favre L, Pignatelli D, Niederländer NJ, Acierno J Jr, **Pitteloud N**. 2017. Evaluating CHARGE syndrome in congenital hypogonadotropic hypogonadism patients harboring CHD7 variants. Genet Med. Nov 16. doi: 10.1038/gim.2017.197. PMID: 29144511
78	Bouilly J, Messina A, Papadakis G, Cassatella D, Xu C, Acierno JS, Tata B, Sykiotis G, Santini S, Sidis Y, Elowe-Gruau E, Phan-Hug F, Hauschild M, Bouloux PM, Quinton R, Lang-Muritano M, Favre L, Marino L, Giacobini P, Dwyer AA, Niederländer NJ, **Pitteloud N**. 2018. DCC/NTN1 complex mutations in patients with congenital hypogonadotropic hypogonadism impair GnRH neuron development. Hum Mol Genet. Jan 15;27(2):359-372. PMID: 29202173
79	Cassatella D, Howard SR, Acierno JS, Xu C, Papadakis GE, Santoni FA, Dwyer AA, Santini S, Sykiotis GP, Chambion C, Meylan J, Marino L, Favre L, Li J, Liu X, Zhang J, Bouloux PM, Geyter C, Paepe A, Dhillo WS, Ferrara JM, Hauschild M, Lang-Muritano M, Lemke JR, Flück C, Nemeth A, Phan-Hug F, Pignatelli D, Popovic V, Pekic S, Quinton R, Szinnai G, l'Allemand D, Konrad D, Sharif S, Iyidir ÖT, Stevenson BJ, Yang H, Dunkel L, **Pitteloud N**. Congenital hypogonadotropic hypogonadism and constitutional delay of growth and puberty have distinct genetic architectures. Eur J Endocrinol. 2018. Apr;178(4):377-388. PMID: 29419413
80	Somm E, Henry H, Bruce S, Bonnet N, Niederländer NJ, Messina A, Aeby S, Rosikiewicz M, Fajas L, Sempoux C, Ferrari S, Greub G, and **Pitteloud N**. 2018. Beta-Klotho deficiency shifts the gut-liver bile acid axis and induces hepatic alterations in mice. Am J Physiol Endocrinol Metab (in revision).

Proceedings of Meetings

1	Nelly Pitteloud. GPR54 gene as a regulator of puberty: idiopathic hypogonadotropic hypogonadism, the disease model. In: A. Beckers, P.Czernichow, E. Reiter, M.L. Vance. Proceedings of the 36th International symposium on Growth Hormone and Growth Factors in Endocrinology and Metabolism; 5/14-15/2004; Geneva, Switzerland. 2004. p. 43-45.

Reviews/Chapters/Editorials

1	**Nelly Pitteloud**, Jacques Philippe. Risque Coronarien chez les Diabétiques: effets préventifs des statines. Médicine et Hygiène. 1998;56:529-36.
2	Hayes FJ, **Pitteloud N**. Hypogonadotropic Hypogonadism and Gonadotropin Therapy. In: The Endocrinology of the Male (McLachlan R ed.). www@endotext.org;2001.
3	**Nelly Pitteloud**, William Crowley, Jr. Congenital Hypogonadotropic Hypogonadism: Clinical Features and Pathophysiology. Endocrinology Series: Hypogonadism: Basic, Clinical, and Therapeutic Principles. 2005.
4	Pallais JC, Au M, **Pitteloud N**, Seminara S, Crowley WF Jr. Kallmann Syndrome (online chapter). In: Pagon RA, Bird TC, Dolan CR, Stephens K, editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-.2007 May 23 [updated 2010 Apr 8].PMID: 20301509
5	Pallais JC, Caudill M, **Pitteloud N**, Seminara S, Crowley, Jr WF. Hypogonadotropic Hypogonadism Overview (online chapter). In: Pagon RA, Bird TC, Dolan CR, Stephens K, editors. Gene Reviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-.2007 May 23.PMID: 20301455
6	Crowley WF Jr, **Pitteloud N**, Seminara S. New genes controlling human reproduction and how you find them. Trans Am Clin Climatol Assoc. 2008;119:29-37.
7	Balasubramanian R, Dwyer A, Seminara SB, **Pitteloud N**, Kaiser UB, Crowley WF Jr. Human GnRH deficiency: a unique disease model to unravel the ontogeny of GnRH neurons. Neuroendocrinology. 2010;92(2):81-99.
8	Sykiotis GP, **Pitteloud N**, Seminara SB, Kaiser UB, Crowley WF Jr. Deciphering genetic disease in the genomic era: the model of GnRH deficiency. Sci Transl Med. 2010 May 19;2(32):32rv2.
9	Pitteloud N, Durrani S, Raivio T, Sykiotis GP Complex genetics in idiopathic hypogonadotropic hypogonadism.. Front Horm Res. 2010;39:142-53.
10	Mitchell AL, Dwyer A, **Pitteloud N**, Quinton R. Genetic basis and variable phenotypic expression of Kallmann syndrome: towards a unifying theory. Trends Endocrinol Metab 2011 Jul;22(7):249-58. Epub 2011 Apr 20. PMID: 21511493
11	Miraoui H, Dwyer A, **Pitteloud N**. Role of fibroblast growth factor (FGF) signaling in the neuroendocrine control of human reproduction. Mol Cell Endocrinol. 2011 346(1-2) :37-43. PMID: 21664428
12	Balasubramanian R, Plummer L, Sidis Y, **Pitteloud N**, Martin C, Zhou QY, Crowley WF Jr. The puzzles of the prokineticin 2 pathway in human reproduction. Mol Cell Endocrinol. 2011 Oct 22;346(1-2) :44-50. Epub 2011Jun 1. PMID: 21664414
13	Phan-Hug F, Hauschild M, Dwyer A, **Pitteloud N**. [Caring for patients with pediatric endocrinopathies and diabetes into adulthood: challenges of an often difficult transition]. Rev Med Suisse. 2012 Nov 14;8(362):2170-2. PMID:23240318
14	**Pitteloud N**. Managing delayed or altered puberty in boys. BMJ (clinical Research Ed.) 2012; 345 pp. e7913. PMID:23207503
15	Virgini V, Baumgartner C, Bischoff T, Haller DM, Frey P, Rosemann T, Collet TH, Sykiotis G, **Pitteloud N**, Rodondi N. [How do Swiss family physicians treat subclinical hypothyroidism?]. Rev Med Suisse. 2014; 10(420):526-9. PMID: 24701670
16	Elowe-Gruau E, Phan-Hug F, Stoppa-Vaucher S, **Pitteloud N**, Hauschild M. [Follow-up and management of children born small for gestational age a endocrine and metabolic aspects].Rev Med Suisse. 2014;10(418):426-9. PMID: 24640277

17	**Pitteloud N**, Dwyer A. Hormonal control of spermatogenesis in men: Therapeutic aspects in hypogonadotropic hypogonadism. Ann Endocrinol (Paris). 2014; 75(2):98-100. PMID: 24793994
18	Dwyer AA, Riavio T, **Pitteloud N**. Gonadotropin replacement for induction of fertility in hypogonadal men. Best Pract Res Clin Endocrinol Metab 2015; 29(1):91-103. PMID: 25617175
19	Dwyer AA, Phan-Hug F, Hauschild M, Elowe-Gruau E, **Pitteloud N**. Transition in endocrinology: Hypogonadism in adolescence. Eur J Endocrinol. 2015; 173(1):R15-R24. PMID:2565325
20	Hayes F, Dwyer A, **Pitteloud N.** Hypogonadotropic Hypogonadism (Hh) and Gonadotropin Therapy. In: De Groot LJ, Beck-Peccoz P, Chrousos G, Dungan K, Grossman A, Hershman JM, Koch C, McLachlan R, New M, Rebar R, Singer F, Vinik A, Weickert MO, editors. Endotext [Internet]. South Dartmouth (MA): MDText.com, Inc.; 2000-.2013 Nov 25. PMID: 25905304
21	Tran C, Barbey F, **Pitteloud N**, Philippe J, Kern I, Bonafé L. [Inborn errors of metabolism: transition from childhood to adulthood]. Rev Med Suisse. 2015 Feb 18;11(462):445-9. PMID: 25915985
22	Boehm U, Bouloux PM, Dattani M, de Roux N, Dodé C, Dunkel L, Dwyer AA, Giacobini P, Hardelin J-P, Juul A, Maghnie M, **Pitteloud N**, Prevot V, Quinton R, Raivio T, Tena-Sempere M, Young J. European consensus statement on congenital hypogonadotrophic hypogonadism - pathogenesis, diagnosis, and treatment. Nat Rev Endocrinol. 2015; 11(9):547-64.PMID: 26194704
23	Dwyer AA, Raivio T, **Pitteloud N**. MANAGEMENT OF ENDOCRINE DISEASE: Reversible hypogonadotropic hypogonadism. Eur J Endocrinol. 2016 Jun;174(6):R267-74. doi: 10.1530/EJE-15-1033. Epub 2016 Jan 20. Review. PMID: 26792935
24	Lamine F, Lalubin F, **Pitteloud N**, Burnier M, Zanchi A. Chronic kidney disease in type 2 diabetic patients followed-up by primary care physicians in Switzerland: prevalence and prescription of antidiabetic drugs. Swiss Med Wkly. 2016 Feb 28;146:w14282. doi: 10.4414/smw.2016.14282. eCollection 2016. PMID: 26922155
25	Chavan R, Preitner N, Okabe T, Strittmatter LM, Xu C, Ripperger JA, **Pitteloud N**, Albrecht U. REV-ERBα regulates Fgf21 expression in the liver via hepatic nuclear factor 6. Biol Open. 2017 Jan 15;6(1):1-7. doi: 10.1242/bio.021519. PMID: 27875243
26	COST Action BM1105., Badiu C, Bonomi M, Borshchevsky I, Cools M, Craen M, Ghervan C, Hauschild M, Hershkovitz E, Hrabovszky E, Juul A, Kim SH, Kumanov P, Lecumberri B, Lemos MC, Neocleous V, Niedziela M, Djurdjevic SP, Persani L, Phan-Hug F, Pignatelli D, **Pitteloud N**, Popovic V, Quinton R, Skordis N, Smith N, Stefanija MA, Xu C, Young J, Dwyer AA. Developing and evaluating rare disease educational materials co-created by expert clinicians and patients: the paradigm of congenital hypogonadotropic hypogonadism. Orphanet J Rare Dis. 2017 Mar 20;12(1):57. doi: 10.1186/s13023-017-0608-2. PMID: 28320476
27	Somm E, Henry H, Bruce SJ, Aeby S, Rosikiewicz M, Sykiotis GP, Asrih M, Jornayvaz FR, Denechaud PD, Albrecht U, Mohammadi M, Dwyer A, Acierno JS Jr, Schoonjans K, Fajas L, Greub G, **Pitteloud N**. β-Klotho deficiency protects against obesity through a crosstalk between liver, microbiota, and brown adipose tissue. JCI Insight. 2017 Apr 20;2(8). pii: 91809. doi: 10.1172/jci.insight.91809. [Epub ahead of print] PMID: 28422755

28	Corcillo A, Pivin E, Lalubin F, **Pitteloud N**, Burnier M, Zanchi A. Glycaemic, blood pressure and lipid goal attainment and chronic kidney disease stage of type 2 diabetic patients treated in primary care practices. Swiss Med Wkly. 2017 Jul 11;147:w14459. doi: smw.2017.14459. eCollection 2017 Jul 11. PMID: 28695552

Thesis

1	Pitteloud, N. Acidocétose dans le diabète de type 2. Switzerland: University of Geneva;1997

Abstracts – Oral Presentations

1	Pitteloud N, Dwyer A, Hayes FJ. Impact of insulin resistance on testosterone secretion in the male: evidence of a dual defect in the HPG axis. Abstract OR26-3, Program of the 84th Annual Meeting of the Endocrine Society, San Francisco. 2002.
2	Pitteloud N, Hayes FJ, Dwyer A, Lee H, Crowley WF, Jr. Inhibin B (IB) and FSH have a logarithmic rather than linear relationship in the human male: Evidence from several human models. Abstract OR28-3, Program of the 85th Annual Meeting of the Endocrine Society, Philadelphia. 2003.
3	Pitteloud N, Quinton R, Pearce S, Acierno J, Dwyer A, Plummer L, Hughes V, Yu-Zhu C, Wei-Ping L, MacColl G, Hayes FJ, Seminara S, Hall J, Bouloux P, Mohammadi M, Crowley WF, Jr. Idiopathic hypogonadotropic hypogonadism and Kallmann syndrome associated with mutations in more than 1 gene: new evidence for a digenic mode of inheritance. Abstract OR52-3, p153, Program of the 88th Annual Meeting of the Endocrine Society, Boston. 2006.
4	Yialamas M, Dwyer AA, Falardeau J, Pitteloud N, Hayes FJ. Effect of acute sex steroid withdrawal on insulin sensitivity in men. Abstract OR22-1, p103, Program of the 88th Annual Meeting of the Endocrine Society, Boston. 2006.
5	Raivio T, Plummer L, Dwyer A, Hughes V, Hayes FJ, Crowley WF Jr, Pitteloud N. Mutations in Fibroblast Growth Factor-1 gene (FGFR1) occur in 8% of cases with normosmic idiopathic hypogonadotropic hypogonadism (nIHH). Abstract OR52-4, p154, Program of the 88th Annual Meeting of the Endocrine Society, Boston. 2006.
6	Cole LW, Plummer L, Quinton R, Jacobson-Dickman E, Raivio T, Seminara SB, Hughes V, Dwyer A, Hayes FJ, Pearce S, Mohammadi M, Bouloux P, Crowley WF Jr, Pitteloud N. An increasing role for oligogenicity in normosmic idiopathic hypogonadotropic hypogonadism (nIKK) and Kallmann Syndrome (KS). Abstract p2-274, p396, Program of the 89th Annual Meeting of the Endocrine Society, Toronto. 2007.
7	Raivio T, Cole LW, Hayes FJ, Seminara SB, Hughes VA, Dwyer A, Quinton R, Pearce SH, Hall JE, Crowley WF Jr, Mohammadi M, Pitteloud N . FGF8 is a key ligand for FGFR1 in GnRH ontogeny: evidence from a human disease model. Abstract P4-254, p 733, Program of the 89th Annual Meeting of the Endocrine Society, Toronto, 2007.
8	Pitteloud N, Cole LW, Sidis Y, Plummer L, Hughes VA, Quinton R, Seminara SB, Hayes FJ, Huot C, Alos N, Lavoie HB, Speiser PW, Takeshita A, Van Vliet G, Pearce S, Dwyer AA, Qun-Yong Z, Crowley WF Jr. Mutations in the gene for prokineticin 2 receptor (PROKR2) cause both Kallmann syndrome (KS) and normosmic idiopathic hypogonadotropic hypogonadism (nIHH). Abstract OR8-2, p84, Program of the 89th Annual Meeting of the Endocrine Society, Toronto. 2007.

9	Pitteloud N, Dwyer A, Falardeau J, Hayes FJ, Seminara SB, Raivio T, Cole L, Crowley WF Jr. Physiologic and genetic insights into adult onset hypogonadotropic hypogonadism (AHH). P3-406, p601, Program of the 89th Annual Meeting of the Endocrine Society, Toronto. 2007.
10	Pitteloud N, Thambundit A, Dwyer A, Hayes FJ, Hughes V, Seminara S, Hughes V, Crowley WF Jr. Previously underappreciated pituitary and gonadal defects in IHH/KS men. Abstract P3-407, p601, Program of the 89th Annual Meeting of the Endocrine Society, Toronto. 2007.
11	Quinton R, Hughes V, Hughes V, Dwyer A, Plummer L, Seminara S, Jacobson-Dickman E, Hayes FJ, Pearce S, Mohammadi M, Bouloux P, Crowley WF Jr, Pitteloud N. Variable reproductive and non-reproductive phenotypes within and across kindreds harboring identical mutations of FGFR1. Abstract P4-253, p733, Program of the 89th Annual Meeting of the Endocrine Society, Toronto. 2007.
12	Dwyer A, Raivio T, Falardeau J, Hayes FJ, Hughes V, Crowley WF Jr, Pitteloud N. Role of FSH in enhancing LH-stimulated testosterone secretion: use of the human model of idiopathic hypogonadotropic hypogonadism (IHH). Abstract P3-568, p781, Program of the 88th Annual Meeting of the Endocrine Society, Boston. 2007.
13	Mutations in the FGF8 genetic network underlie a large proportion of isolated human GnRH deficiency. H Miraoui, B Feng, G Sykiotis, L Plummer, R Quinton, V Hughes, WF Crowley, A Dwyer, Y Sidis, M Mohammadi, N Pitteloud. 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011
14	Role of FSH prior to LH + FSH on testes development in humans: Effects on histologic, biochemical, and fertility parameters in men with isolated gonadotropin-releasing hormone (GnRH) deficiency and prepubertal testes. A Dwyer, G Sykiotis, FJ Hayes, PA Boepple, KR Loughlin, M Dym, WF Crowley, N Pitteloud. 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011
15	Responsiveness to Pulsatile GnRH in Women with Isolated GnRH Deficiency Identifies Additional Pituitary and Ovarian Defects. JM Brown, B Abel, JM Adams, T Alati, ND Shaw, KA Martin, N Pitteloud, SB Seminara, WF Crowley, CK Welt, JE Hall. 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011
16	A New Untragenic Micro Deletion of the IGF-1 Receptor Gene in an Italian Family with Developmental and Reproductive Defects. F Phan-Hug, F Zufferey, M Bellavia, D Martinet, FN Butschi, GP Sykiotis, PE Mullis, N Pitteloud, S Jacquemont. 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011
17	An Interdisciplinary Education Program Training Health Professionals to Encourage Physical Activity in Patients with Type 2 Diabetes. O Giet, D Sofra, T Cancelli, L Allet, P Marques-Vidal, J Ruiz, N Pitteloud, H Delgado, M Castellsague, C Negre, S Beer, J Puder. 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011
18	Spectrum of Phenotypes Associated with Various Levels of Mutational Burden in Humans with Isolated GnRH Deficiency Due to Defects in the GNRHR Gene. E Gianetti, JE Hall, MG Au, L Plummer, R Quinton, JA Stewart, DL Metzger, N Pitteloud, V Mericq, PM Merino, LL Levitsky L Izatt, M Lang-Muritano, RG Dluhy, WF Crowley, SB seminara. 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011
19	Olfactory Function in GnRH Deficiency Displays a Phenotypic Spectrum: Potential Implications for Genetic Screening and Pathophysiology. HM Lewkowitz-Shpuntoff, R Balasubramanian, V Hughes, L Plummer, MG Au, SB Seminara, N Pitteloud, R Doty, WF Crowley. 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011

20	Rare Nucleotide Variants in KISS1 in Patients with GnRH-Deficient Phenotypes. YM Chan, R Lapatto, M Au, V Hughes, SDC Bianco, L Min, L Plummer, F Cerrato, A de Guillebon, F Wahab, A Dwyer, SE Krisch, R Quinton, TD Cheetham, M Ozata, SB Ten, JP Chanoine, N Pitteloud, WF Crowley, KA Martin, R Schiffmann, JE Hall, UB Kaiser, SB seminara. 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011
21	Gain-of-function mutations in FGFR1 in human GnRH deficiency. H Miraoui, KW Keefe, G Sykiotis, L Plummer, T Raivio, A Dwyer, Y Sidis, P Tsai, M Mohammadi, N Pitteloud. Oral presentation 93rd Annual Meeting of the Endocrine Society, Boston, MA, June 2011
22	Role of FRS2/FRS3 in GnRH neuron ontogeny. D Cassatella, E Jacobson-Dickman, G Sykiotis, A Beenken, H Chen, T Raivio, L Plummer,J-P Rey, A Dwyer, AV Eliseenkova, P Speiser, Y Sidis, M Mohammadi, N Pitteloud. Gordon Research Conference "Fibroblast Growth Factors in Development & Disease". Les Diablerets, Switzerland, May 2012
23	Identifying new disease-associated genes in Kallmann Syndrome using whole-exome sequencing. D. Cassatella, J Liang, A Dwyer, JP Rey, VH Hughes, R Quinton, P Bouloux, M Lang-Muritano, Y Sidis, N Pitteloud. *Received Best Poster Award. University of Lausanne Faculty of Biology and Medicine Research Day- "The World of Omics". Lausanne, Switzerland, June 2012
24	Creating a European consortium to study GnRH deficiency (COST Action BM1105). N Pitteloud & **A** Dwyer. *Received the ESPE Presidential Poster Award. 51st Annual Meeting of the European Society for Pediatric Endocrinology, Leipzig Germany, September 2012.

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