



L'Institut de recherche  
du Centre universitaire de santé McGill  
à L'Hôpital de Montréal pour enfants  
The Research Institute  
of the McGill University Health Centre  
at The Montreal Children's Hospital



Child health  
research:

## **BUILDING** ON OUR **STRENGTHS**

2010–2011  
ANNUAL REPORT



Photo: Robert Derval

In September of 2010, over 100 children helped break ground for the new Montreal Children's Hospital, scheduled to open in 2015 among the first McGill University Health Centre (MUHC) facilities on the Glen Campus.

Within a year of that ceremony, the atrium and laboratories of the new Research Institute of the MUHC have risen quickly.



Photo: Eric Simard

This is the future look of the Glen site.



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“WE ARE NOT BUILDING A WORLD-CLASS RESEARCH FACILITY FROM THE GROUND UP. WE ARE BUILDING IT, RATHER, FROM THE INSIDE OUT.”



### Jacquetta Trasler, MD, PhD

Scientific Director, Child Health Research  
The Montreal Children's Hospital (MCH)  
of the McGill University Health Centre (MUHC)

## Message from the Scientific Director Child Health Research, MUHC

The inside cover of this year's report tells a tale of transformation. Yet dramatic as the external evidence is, I must stress that we are not building a world-class research facility from the ground up. We are building it, rather, from the inside out: building on the strengths of our research community.

Our strengths begin with individual child health research programs at The Montreal Children's Hospital (MCH), like the five profiled in this report. These strengths are tested and consolidated within our network of peers, by colleagues who make the time to contribute to scientific review and multidisciplinary collaborations. This year, we laid the groundwork to group our investigators into “research neighbourhoods” in which many will be close to their counterparts in adult medicine on the new site. I can only marvel at how this compounds the possibilities, both for research and for better health care for children.

Closer collaboration with our colleagues across the Research Institute of the McGill University Health Centre (RI-MUHC) will permit the mapping of disease trajectories from fetus to child, and child to adult. It will lead to the development of screening tests and preventive therapies for birth defects and adult diseases originating in early life. It will increase the critical mass of expertise forming around the themes in our Prenatal and Childhood Origins of Disease program.

We are now at the peak of a ten-year planning period for redevelopment. Thanks to the steadfast support of our Advisory Board and funding partners, including the FRQS, CIHR, CFI, MCH Foundation and Foundation of Stars, we are approaching the time and the place in which we can fully deploy the strengths of our investigators, staff and trainees in a superlative child health research environment.

“WITH THE GLEN CAMPUS UNDER CONSTRUCTION,  
A NEW PARADIGM FOR HEALTH DISCOVERY IS BEING CREATED.”

## Message

### from the Executive Director and Chief Scientific Officer Research Institute of the MUHC

While I am grateful for the opportunity to recognize recent research accomplishments, in this year’s report I feel compelled to reflect at once further backward in time, and forward.

For over a century, The Montreal Children’s Hospital has made significant contributions to the care of children. Its researchers have been prolific in a number of fields, clearly driven to pursue continuous improvements and excellence. To advance further, however, new platforms are required to speed innovation, accelerate the translation of basic discoveries to public uses and provide the underpinnings for novel health policies. That’s why the future holds such promise.

With the Glen Campus under construction, a new paradigm for health discovery is being created. The Montreal Children’s Hospital is coming together with a number of MUHC adult hospitals, a Cancer Centre and some of the world’s finest research facilities. Complete with robust informatics and technologies, the RI-MUHC’s Centre for Innovative Medicine and Centre for Translational Biology will integrate fundamental, clinical and evaluative research. Professionals will thus be able to bridge the entire life span, strengthening synergies with each other and among their various research themes. As the volume and intensity of their research collaborations increase, the world’s understanding of pediatric diseases—and how best to prevent, diagnose, treat and/or cure them—will be enriched dramatically.

Ultimately, we will be building on our strengths so that we continue to improve people’s health and well-being, from birth through all stages of life.



**Vassilios Papadopoulos,**  
DPharm, PhD

Executive Director and Chief Scientific Officer  
Research Institute of the McGill University  
Health Centre (RI-MUHC)

Associate Executive Director  
for Research, MUHC



“THE PRENATAL AND CHILDHOOD ORIGINS OF DISEASE PROGRAM IS ONE OF THE GREAT STRENGTHS OF THE RI-MUHC AND MCGILL.”



**Gretta Taylor Chambers,  
CC, OQ BA, DLitt**

Chair of the Advisory Group  
on Research to the CSCA

Chancellor Emerita, McGill University

## Message

### from the Chair of the Advisory Group on Research to the Council for Services to Children and Adolescents (CSCA)

The Advisory Group would like to congratulate the research community at The Montreal Children’s Hospital, from its scientists to its administrators, on having completed an exceptionally productive year. Great emphasis has been placed on the examination of prenatal and childhood origins of disease as providing the best possible defence against disease by getting to its root causes as early as possible. Much research has gone into the discovering and understanding of birth defects, developmental disorders and diseases in children.

As the community prepares to regroup around the Glen and Mountain campuses of the new MUHC, a concentration of expertise is coalescing around these key areas. The Prenatal and Childhood Origins of Disease Program is one of the great strengths of the RI-MUHC and McGill, and MCH investigators excel in each of the three main research themes that it comprises. All have direct bearing on lifelong health care.

One group is responsible for breakthroughs in such areas as birth defects, diabetes and brain tumours. A second leads the way in fields ranging from reproductive/perinatal epidemiology to environmental contaminants, effects of prescription drugs and high-risk children. A third conducts important studies in prenatal asphyxia, autism and attention deficit disorder.

It will be obvious to readers of this report that we are blessed with a team of remarkable investigators and research recruits. The Advisory Group is proud to be associated with them and very grateful to be part of their child health outreach endeavour.

“WE NOW HAVE VISIBLE EVIDENCE OF A LONG SOUGHT GOAL:  
A NEW MONTREAL CHILDREN’S HOSPITAL WITHIN THE MUHC CAMPUS.”

## Message

### from the Associate Executive Director of The Montreal Children’s Hospital

The excitement is palpable! We now have visible evidence of a long sought goal: a new Montreal Children’s Hospital within the MUHC campus emerging at the Glen. Indeed, the new biomedical research building of the RI-MUHC is rising from the ground. In addition, the Centre for Innovative Medicine has been strategically located between the pediatric and adult hospitals—recognition of the merits of research colleagues working close together, and of the importance of clinical research within a university teaching hospital. We are clearly going to have world-class research facilities!

However, it is not only bricks and mortar that define a world-class research facility. It is the very special people working within these confines—the investigators, their research assistants, trainees and dedicated support staff—who define us. As described within this annual report, we have exceptional talent under the outstanding leadership of Dr. Jacquetta Trasler to carry us forward into the new site.

With the strong support of Dr. Vassili Papadopoulos, Dr. Trasler has helped shape our future research direction for the next decade. Our research community is vibrant and visionary; its future, and ours, is very bright. Our patients and their families will undoubtedly benefit from innovative research programs.



Harvey J. Guyda, MD, FRCP(C)

Associate Executive Director  
The Montreal Children’s Hospital  
of the MUHC

# Our Researchers

## By Axis of the Research Institute of the MUHC

### Cancer Axis

Sharon Abish  
David Mitchell  
Janusz Rak

### Cardiovascular Diseases and Critical Care Axis

Marie Josée Béland  
Dominic Chalut  
Adrian Dancea  
Ronald Gottesman  
Sam Shemie  
Dominique Shum-Tim  
Davinia Withington

### Endocrinology, Diabetes, Nutrition and Kidney Diseases Axis

Najma Ahmed  
Lorraine Bell  
Preetha Krishnamoorthy  
Laurent Legault  
Véronique Morinville  
Constantin Polychronakos  
Gloria S. Tannenbaum  
Michele Zappitelli

### Health Outcomes Axis

Maala Bhatt  
Franco Carnevale  
Evelyn Constantin

Geoffrey E. Dougherty  
Ciarán Duffy  
Mohamed El-Sherbiny  
Sylviane Forget  
Bethany Foster  
John Richard Hamilton  
Michael S. Kramer  
Lucyna Lach  
Stephen Liben  
Mary Ellen Macdonald  
Romain Mandel  
John J. Manoukian  
David McGillivray  
Meranda Nakhla  
Hema Patel  
Robert William Platt  
I. Barry Pless  
Caroline Quach  
Saleem Razack  
Janet Elizabeth Rennick

### Human Reproduction and Development Axis

Sam Joseph Daniel  
Cynthia Gates Goodyer  
Paul R. Goodyer  
Indra Gupta  
Roman Jednak  
Loydie Jerome-Majewska  
Céleste Johnston  
Jean-Martin Laberge  
Annette Majnemer  
Aimée Ryan  
Michael Shevell  
Laurie Snider

### Infection and Immunity Axis

Reza Alizadehfar  
Martin Bitzan  
Bruce Mazer  
Christine McCusker  
Jane McDonald  
Dorothy L. Moore  
Marie-Noël Primeau  
Earl Rubin  
Ernest G. Seidman

### Medical Genetics and Genomics Axis

Nancy Braverman  
Kathleen Glass  
Nada Jabado  
Feige Kaplan  
Robert K. Koenekoop  
John Mitchell  
Rima Rozen  
Charles R. Scriver  
Jacquetta Trasler

### Mental Illness and Addiction Axis

Eric Fombonne  
Brian Greenfield  
Lily Hechtman  
Cécile Rousseau

### Musculoskeletal Disorders Axis

Gaëlle Chédeville  
Reggie Hamdy  
Jean A. Ouellet  
Frank Rauch  
Celia Rodd  
Rosie Scuccimarrì

### Neurosciences Axis

Jeffrey Atkinson  
Marie-Emmanuelle Dilenge  
Isabelle Gagnon  
Shuvo Ghosh  
Krista L. Hyde  
Pierre Lachapelle  
Catherine Limperopoulos  
Bernard Rosenblatt  
Teresa Valois Gomez  
Pia Wintermark

### Respiratory Health Axis

Robert Brouillette  
Larry C. Lands  
Johanne Morel  
Francisco Noya  
Pramod Puligandla  
Charles Rohlicek  
Guilherme Sant'Anna

## Researchers Affiliated with MCH

Robert Barnes  
Farhan Bhanji  
Claudette Bardin  
Louis Beaumier  
Margaret Berry  
Karen A. Brown  
Natalie Buu  
Sarah Campillo

John Paul Capolicchio  
Aurore Côté  
Joëlle Desparmet  
Alessandra Duncan  
Giosi Di Meglio  
Sherif Emil  
Ricardo Faingold  
Jean-Pierre Farmer

Chantal Frigon  
Josée Lavoie  
Serge Melançon  
Klaus Minde  
José Luis Montes  
Thérèse Perreault  
Maria Ramsay  
Patricia Riley

Melvin Schloss  
Christo I. Tchervenkov  
Ted Tewfik  
Blair Newell Whittemore  
H. Bruce Williams



# New Recruits



## Krista L. Hyde, PhD

Assistant Professor, Departments of Psychiatry and Neurology, McGill University  
Medical Scientist, Division of Psychiatry  
The Montreal Children's Hospital of the MUHC

**Dr. Krista L. Hyde, Neurosciences Axis, received her PhD in cognitive neuroscience from the Université de Montréal. She completed post-doctoral training at the Montreal Neurological Institute in collaboration with researchers at Harvard Medical School.**

Dr. Hyde's multi-disciplinary research program uses magnetic resonance imaging (MRI) techniques to measure brain structure and function in combination with behaviour. By looking at brain plasticity as a function of such specialized training as music, for instance, she is investigating whether auditory research can make it possible to pick up early brain changes in children with autism spectrum disorder (ASD). Another study focus is the correlation of brain and behavioural measures with genetic profiles in developmental disorders.

Together, these programs open prospects for earlier and more effective biologically based treatments of ASD and other developmental disorders.



## Pia Wintermark, MD

Assistant Professor of Pediatrics  
McGill University  
Neonatology, The Montreal Children's Hospital of the MUHC

**Dr. Pia Wintermark, in both the Neurosciences and the Cardiovascular and Critical Care axes, received her MD from the University of Lausanne in Switzerland. She completed her training in pediatrics and neonatology at the University of Lausanne and in the Harvard Neonatal-Perinatal Program at the Children's Hospital in Boston.**

Dr. Wintermark seeks to understand the causes and consequences of brain injuries in very sick newborns. She aims to develop innovative strategies to prevent or repair these injuries and improve the future neurodevelopmental outcome of these newborns. Her laboratory uses both clinical research (including advanced neuroimaging techniques and bedside monitoring) and basic science techniques to understand the mechanisms underlying brain injuries.



# Our Strengths:

## Prenatal and Childhood Origins of Disease Program

### Themes of the Prenatal and Childhood Origins of Disease Program

#### 1. Genetics and Genomics of Rare Disease, Common Disease and Cancer

*Examples:*

research into birth defects, diabetes, brain tumours

*Researcher profiles in this report:*

Drs. Paul Goodyer and Rima Rozen

#### 2. Prenatal and Childhood Environmental Origins of Disease

*Examples:*

research in the fields of reproductive/perinatal epidemiology, environmental contaminants, assisted reproduction, effects of prescription drugs, high-risk children

*Researcher profiles in this report:*

Drs. Sam Daniel, Robert Platt and Caroline Quach

#### 3. Brain, Behaviour and Development

*Examples:*

studies in autism, prenatal asphyxia, attention deficit disorder

*Recruit profiles in this report:*

Drs. Krista Hyde and Pia Wintermark



Every child deserves the best possible chance to grow into a healthy adult. Detecting disease early is the best way to prevent or decrease lifelong illness and suffering, and this is our strategy in building research strengths at The Montreal Children's Hospital (MCH). To deliver better health care, we must first discover and better understand the causes of birth defects, developmental disorders and diseases in children.

Strengths in these areas are the backbone of the Prenatal and Childhood Origins of Disease program, our blueprint for child health research as we regroup for the move to new facilities. As may be seen in the profiles of investigators and research recruits presented in this report, a concentration of expertise has emerged around each of the three themes in this program, shown opposite.

MCH investigators' advances in these key areas are in step with new research priorities identified by the federal and provincial governments, and by the Canadian Institutes of Health Research (CIHR). Moreover, our Prenatal and Childhood Origins of Disease program is enriched by its placement on the continuum of Translational Research and Intervention Across the Lifespan—a concept that won the RI-MUHC a \$100 million grant from the Canada Foundation for Innovation to build a cutting-edge research centre on the Glen Campus.

With the collaboration of colleagues across the RI-MUHC and McGill, we are ready to build on established strengths and become a national and international leader for the reproductive and child health priorities of the twenty-first century.

# Honours and Distinctions

## 2010

**Dr. Nada Jabado's** recent research breakthrough with Dr. Jacek Majewski was identified in the December 15 issue of *L'actualité* as one of 35 inventions that will "change everything." This research, published in *Human Mutation* in July, demonstrated that the sequencing of one person's exome can permit effective research into mutations indicating a genetic disease, without the need to sequence an entire genome.

**Dr. Michael S. Kramer** was the 2010 recipient of the Paediatric Academic Leadership—Clinical Investigator Award from the Pediatric Chairs of Canada in October. The award recognized his administrative leadership in the child health research community in Canada and his prominent role in influencing child health globally.

**Mr. Brian Meehan**, a research assistant in Dr. Janusz Rak's cancer and angiogenesis laboratory, received the first MUHC Director General's Award in Pediatric Research.

**Dr. Constantin Polychronakos** was named among the top ten scientists of Greek origin in 2010 by *Status* magazine (Athens).

**Dr. Gary Pেকেles** was invited to participate in the Scientific Committee for the 26th International Pediatric Association (IPA) Congress of Pediatrics, Johannesburg, South Africa, in August 2010.

**Dr. Rima Rozen** was elected Fellow of the Royal Society of Canada. Dr. Rozen is internationally recognized for her work on the genetics of several disorders, including birth defects, heart disease and inborn errors of metabolism.

## 2011

**Dr. Kathleen Glass** was awarded a Lifetime Achievement Award from the Canadian Bioethics Society.

**Dr. Dorothy Moore** was honoured as this year's recipient of the Canadian Paediatric Society (CPS) Member Recognition Award. The CPS gives the award annually to recognize outstanding contributions of its members as spokespeople, peer reviewers and liaisons with other organizations, and as participants in committees or on the Board of Directors.

**Dr. Nada Jabado** was among the top researchers from across Canada chosen to participate in two initiatives supported by the Canadian Government. The Canadian Pediatric Cancer Genome Consortium and the Finding of Rare Disease Genes in Canada (FORGE Canada) team both aim to identify the genes that cause the most challenging types of cancer and rare disease in children, and find new treatments.

**Dr. Nada Jabado** received the 2011 Aldo Award of Excellence in Research from The Montreal Children's Hospital Foundation, presented to a researcher whose initiatives have made unique and significant contributions to paediatric care.

**Dr. Thérèse Perreault** received the 2011 Jean Coutu Medical Award of Excellence from The Montreal Children's Hospital Foundation.

**Dr. Constantin Polychronakos** received an honorary doctorate from the Medical Faculty of the Aristotelean University in Greece.



# Investigator and Trainee Awards



## Investigators

### Canada Research Chair

#### Tier 1

- Eric Fombonne
- Ernest Seidman

#### Tier 2

- Catherine Limperopoulos

### Fonds de recherche du Québec—Santé

#### National researcher

- Bruce Mazer

#### Research Scholar—Senior

- Robert Platt

#### Clinical Research Scholar—Senior

- Indra Gupta

#### Research Scholar—Junior 2

- Nada Jabado

#### Clinical Research Scholar—Junior 2

- Sam Daniel
- Bethany Foster
- Jean A. Ouellet

- Caroline Quach
- Frank Rauch

#### Clinical Research Scholar—Junior 1

- Evelyn Constantin
- Isabelle Gagnon
- Janet Rennick
- Michele Zappitelli

### Kidney Foundation of Canada

#### Krescent New Investigator Award

- Michele Zappitelli

## Post-doctoral Fellowships

### Canadian Institutes of Health Research

- Valerie Marcil

### Fonds de recherche du Québec—Santé

- Hugues Beauchemin
- Cédric Clouchoux
- Flavia Lombardi Lopes
- Serge McGraw
- Julien St-Jean
- Yebeben Ruiz-Casares

### Kidney Foundation of Canada

- Reyhan El Kares

## Doctoral Research Awards

### Canadian Institutes of Health Research

- Donovan Chan
- Adam Fontebasso
- Nafisa Jadavji
- Xiaoyang Liu
- Kirsten Niles
- Denise Keiko Shikako Thomas

### Fonds de recherche du Québec—Santé

- Marie Brossard-Racine
- Michelle Collins
- Noémi Dahan-Oliel
- Patricia Fontela
- Sina Gallo
- Grzegorz Sobieraj
- Hana Zouk

## Master's Research Awards

### Canadian Institutes of Health Research

- Justine Lee Garner
- Halim Khairallah
- Lynne Li
- Mallory Owen

### Fonds de recherche du Québec—Santé

- Vasiliki Darsaklis
- Tonje Persson

# Researcher Profiles

Step into an  
MCH laboratory.

BUILD A WORLD  
OF HOPE.



# Researcher Profiles

## Preventing Hearing Loss after Chemotherapy

Eighty per cent of kids diagnosed with cancer now survive their disease. Unfortunately, most young survivors exposed to platinum chemotherapy end up with an irreversible hearing loss. This side effect is particularly important in children, as hearing loss affects the development of their language and social skills.



**Sam Daniel, MDCM, MSc, FRCS(C)**  
Human Reproduction and Development Axis

Associate Professor, Department of Otolaryngology–  
Head and Neck Surgery, McGill University

Director, Otolaryngology–Head and Neck Surgery  
The Montreal Children’s Hospital of the MUHC

Dr. Sam Daniel has established the McGill Auditory Sciences Laboratory, where his research team is uniquely equipped to develop models to further the diagnosis and treatment of drug-induced hearing loss. A clinician-scientist with expertise in ototoxicity and middle ear mechanics research, Dr. Daniel is now exploring ways to prevent platinum ototoxicity, the form of hearing loss experienced by cancer patients. The loss is caused by toxic compounds released after exposure to the chemotherapeutic agent.

Dr. Daniel’s research team has developed a novel strategy using protective agents to curb ototoxicity linked with cisplatin, the most commonly used chemotherapeutic agent. The team is also testing innovative delivery techniques to carry these protective drugs to the inner ear.

This research will not only improve the quality of life for cancer survivors; it will also reduce restrictions on some of the most powerful agents used to fight cancer. Controlling the adverse side effect of ototoxicity may allow higher treatment doses of chemotherapy, and consequently better patient survival.



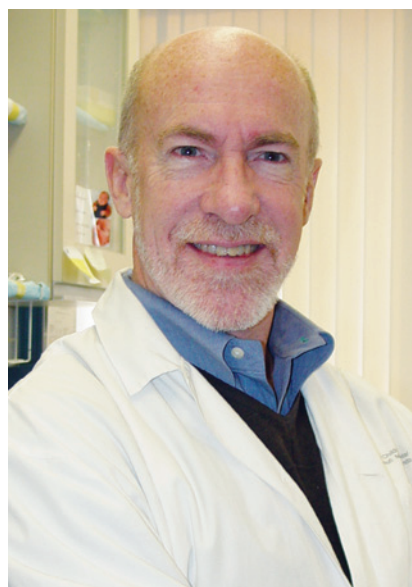
## New Therapies for Kids with Rare Diseases

The Renal-Genetics Clinic at The Montreal Children's Hospital offers specialized care to kids with "orphan" diseases of the kidney that often fall below the radar at large research funding agencies. Since 1980, Dr. Paul R. Goodyer has met the dual challenges of directing this clinic and building a research program closely tied to its patients' needs. His laboratory aims to unravel the molecular basis for these diseases and translate the information into new therapies.

Recently, Dr. Goodyer's group discovered dysfunctional variants of three genes that set congenital nephron number during fetal life. Studies are underway with colleagues in India to determine whether the impact of these genes can be offset by vitamin A supplementation during pregnancy.

A second important discovery is that microvesicles shed by stem cells can reverse the pathologic accumulation of cystine in tissues from children with cystinosis. Dr. Goodyer leads an international consortium committed to developing stem cell therapy for this rare but devastating disease.

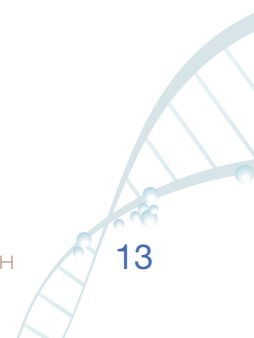
Dr. Goodyer's group has also found that a non-canonical WNT signalling pathway drives cyst formation during kidney development in children with polycystic kidney disease. Studies are underway to establish that cyst formation can be reversed by augmenting the non-canonical WNT signalling pathway.



**Paul R. Goodyer, MD**  
**Human Reproduction  
and Development Axis**

Professor of Pediatrics  
McGill University

Pediatric Nephrology  
The Montreal Children's Hospital of the MUHC



# Researcher Profiles

## Getting Better Answers in Child Health Research

Whether the subject is fetal growth, kidney transplant management or factors affecting preterm birth, well-designed studies always get the best answers. Dr. Robert Platt, a biostatistician, develops statistical methods for health research and maps out better ways of studying and interpreting pediatric outcomes.



**Robert Platt, PhD**  
Health Outcomes Axis

Professor of Pediatrics and of Epidemiology, Biostatistics  
and Occupational Health, McGill University

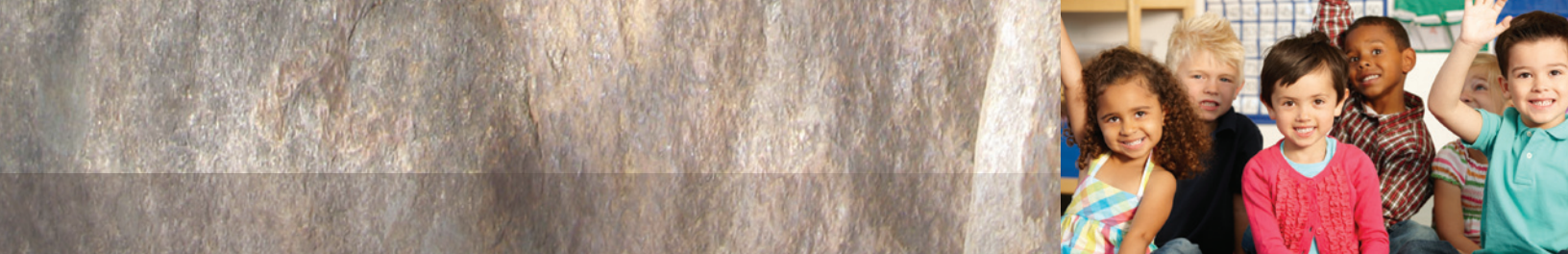
Pediatrics/Epidemiology and Biostatistics  
The Montreal Children's Hospital of the MUHC

Dr. Platt's research group has shown that some ways of studying fetal growth give incorrect results, often missing the important effects of exposures during pregnancy. His group is developing statistical tools that can identify small fetuses at risk for later health problems and allow for the very different growth patterns of singletons and twins.

Dr. Platt also focuses on causal inference methods, using statistical tools to establish causation. Typical statistical methods show only associations, but Dr. Platt and his team are interested in whether an exposure to a particular drug, food or chemical causes an outcome. They are examining the assumptions and methods that allow us to use statistics to interpret causation.

In child health and perinatal research, Dr. Platt has worked with colleagues to improve research methods in a variety of studies. Topics include breastfeeding and long term outcomes, how to manage care for kidney transplant recipients, how social factors affect preterm birth, and whether pacifier use affects weaning.





## Researching the Risks of Infection

Infections are part of every child's—and parent's—life. A child has, on average, eight to ten colds or episodes of gastroenteritis per year, mainly during winter and in the first year of attending daycare. Hospitalized children run the same risks but with the threat of more serious infections, often following treatments and interventions. Blood infections may result from central lines or surgical site infections, for instance. While treatments with antimicrobials can work wonders, Dr. Caroline Quach's research team is investigating how best to prevent infections.

Dr. Quach conducts epidemiological studies of risk factors leading to infections. Her work to date on bloodstream infections shows that having to report and compare infection rates makes healthcare units realize that they have a problem, and enables them to fix it.

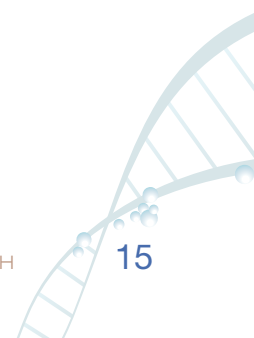
Dr. Quach has also studied risks of infections associated with Emergency Department (ED) visits. Findings show that children do not have more colds or episodes of gastroenteritis after visiting an ED. However, studies of the elderly suggest that ED visits are associated with a risk of infection in people who have few daily contacts with others. Dr. Quach's team is now working on the prevention of gastroenteritis and on effects of the new rotavirus vaccine.



**Caroline Quach, MD, MSc**  
Health Outcomes Axis

Associate Professor of Pediatrics  
Associate member, Department of Epidemiology,  
Biostatistics and Occupational Health  
McGill University

Pediatric Infectious Diseases  
The Montreal Children's Hospital of the MUHC



# Researcher Profiles

## Preventing Birth Defects and Heart Disease

A geneticist with expertise in cellular and molecular research, Dr. Rima Rozen studies genes and nutrients that have an important role in birth defects and heart disease. Central to this research is a pathway involving the metabolism of a critical B vitamin, folic acid.



**Rima Rozen, PhD, FRSC, FCCMG**  
Medical Genetics and Genomics Axis

Associate Vice-Principal (Research and International Relations)  
and James McGill Professor  
Departments of Human Genetics and of Pediatrics  
McGill University

Medical Genetics  
The Montreal Children's Hospital of the MUHC

In landmark studies, Dr. Rozen isolated an important gene in folate metabolism called MTHFR and identified a common genetic mutation that is the first known genetic risk factor for spina bifida. This mutation also increases the risk for coronary heart disease and stroke, due to an elevation of the toxic compound, homocysteine. Dr. Rozen showed that individuals with this mutation require additional folate in their diet to overcome the effect of the mutation, reduce their homocysteine levels and decrease their risk of having children with spina bifida.

In other studies, Dr. Rozen found that adequate intake of folate, choline and riboflavin may be important for prevention of congenital heart defects. Her laboratory has also characterized genetic mutations in homocystinuria, an inherited metabolic disorder, and developed prenatal diagnosis using molecular methods for this disorder. The connection between low dietary folate and the development of cancer as well as birth defects is a current focus. Her work has led to new genetic tests and to dietary recommendations.



# Selected Publications (from nearly 300)

## 2010

Akoume MY, Azeddine B, Turgeon I, Franco A, Labelle H, Poitras B, Rivard CH, Grimard G, Ouellet J, Parent S, Moreau A. **Cell-based screening test for idiopathic scoliosis using cellular dielectric spectroscopy.** *Spine (Phila Pa 1976)* 35(13):E601-8, 2010.

Anastasio N, Ben-Omran T, Teebi A, Ha KC, Lalonde E, Ali R, Almureikhi M, Der Kaloustian VM, Liu J, Rosenblatt DS, Majewski J, Jerome-Majewska LA. **Mutations in SCARF2 are responsible for Van Den Ende-Gupta syndrome.** *Am J Hum Genet* 87(4):553-9, 2010.

Bhanji F, Mancini ME, Sinz E, Rodgers DL, McNeil MA, Hoadley TA, Meeks RA, Hamilton MF, Meaney PA, Hunt EA, Nadkarni VM, Hazinski MF. **Paediatric Resuscitation Training – Do medical students believe it should be a mandatory component of the curriculum?** *Resuscitation* 82(5):584-7, 2010.

Bin Salleh H, McGillivray D, Martin M, Patel H. **Duration of fever affects the likelihood of a positive bag urinalysis or catheter culture in young children.** *J Pediatr* 156(4):629-33, 2010.

Bitzan M, Schaefer F, Reymond D. **Treatment of typical (enteropathic) hemolytic uremic syndrome.** *Semin Thromb Hemost* 36(6):594-610, 2010.

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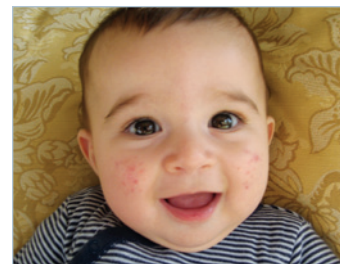
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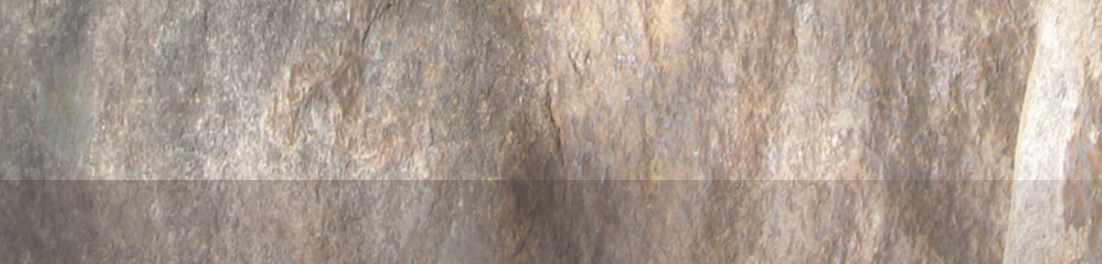
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# Funding (01/04/2010–31/03/2011)



## Major Benefactors of the Research Program at The Montreal Children's Hospital of the MUHC

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The generosity of visionary donors enabled The Montreal Children's Hospital Foundation to make disbursements of \$2 million in support of child health research in 2010–2011.

Among the major contributors were the Cole Foundation, Shire Canada, the Children's Leukemia Research Association, Leucan, the Foundation for Fighting Blindness—Canada, and the Foundation for Retinal Research, as well as a number of loyal individuals. Their gifts led to significant research advances in areas such as oncology, ophthalmology, the study of allergies, and the treatment of attention deficit hyperactivity disorder.

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1,209,625	Fonds de recherche du Québec—Santé
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59,410	Dairy Farmers of Canada
50,694	Autism Speaks (USA)
50,319	Heart & Stroke Foundation of Canada
31,318	Social Sciences and Humanities Research Council of Canada
6,282	Institut national de la santé et de la recherche médicale (France)
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3,360	Sick Kids Foundation
1,375	Canadian Arthritis Network
707	Fonds de recherche du Québec—Société et culture
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<b>\$8,876,306</b>	<b>TOTAL</b>

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**\$571,777**      **TOTAL**



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## Our Research Community

The Research Institute of the McGill University Health Centre at The Montreal Children's Hospital is composed of over 100 researchers and over 100 graduate students and post-doctoral fellows engaged in a broad spectrum of basic and clinical research. It also comprises more than 100 technicians, coordinators, nurses and administrative personnel.

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