

Annual Report

CHU Sainte-Justine Research Centre



2019
2020



Research Centre
**CHU
Sainte-Justine**
Mother and Child
University Hospital Centre

Université 
de Montréal

This **2019–2020 annual report** outlines the operational and financial performance for the period from **April 1, 2019, to March 31, 2020.**

Summary

“Towards Precision Health - Healthy Children and Mothers for a Healthy Future”

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A Letter from the Director

Dr. Jacques L. Michaud



Dear Colleagues and Friends,

This past year was one of a transition to a new development plan that will guide our efforts for the next five years. This plan involves a major initiative aimed at unifying and engaging everyone within our institution around a common vision: **“Toward Precision Health: Healthy Children and Mothers for a Healthy Future.”** It leverages the synergy generated by more than **200 research teams** committed to basic, clinical and translational research and an outstanding group of career scientists and clinician-researchers supported by the leadership of the

Sainte-Justine University Hospital Centre, the CHU Sainte-Justine Foundation, the Université de Montréal and other institutional partners.

This plan is consistent with the strategic choices made by the **CHU Sainte-Justine Research Centre** and the care-related priorities of the hospital and capitalizes on the many significant achievements made in the past five years.

This **2019–2020 annual report** reflects what we have accomplished in the past year and shines the spotlight on certain strategic efforts that we aim to undertake in the fields of precision medicine and prevention science going forward in order to maintain our leadership position in mother-and-child research, both here at home and around the world.

Dr. Jacques L. Michaud, MD

Director of Research, Sainte-Justine University Hospital Centre



Overview of the Research Centre

Healthy children and mothers for a healthy future

The Sainte-Justine University Hospital Centre (Sainte-Justine) is an academic hospital with a multipronged mission revolving around **health care, research, teaching, and health technology and intervention assessment**, in addition to cross-disciplinary **prevention and promotion initiatives** aimed at the wellness of mothers-to-be, children and youth in Quebec.

The **Sainte-Justine University Hospital Research Centre** (Research Centre) is committed to delivering true “**precision health**,” which will not only affect diagnosis and case management but will also impact health trajectories, thereby helping to build a better future for families.

The **Research Centre**’s strategic priorities are rooted in the integration of **basic, clinical and translational research** and focus on four main objectives:

1. Gain a better understanding of the causes of disease, develop diagnostic tools, enable faster discovery of more effective therapies and help improve the quality of patient care.
2. Develop screening and prevention tools.
3. Groom the next generation of scientists through multi-, inter- and transdisciplinary research training programs with a patient-oriented approach.
4. Remain a leader in mother-and-child research across Canada and worldwide by forging ties with strategic internal and external partners.



2 business licenses

17 inventions and disclosures

29 patents

210 researchers, including **110** clinicians

621 research contracts and agreements

+500 students

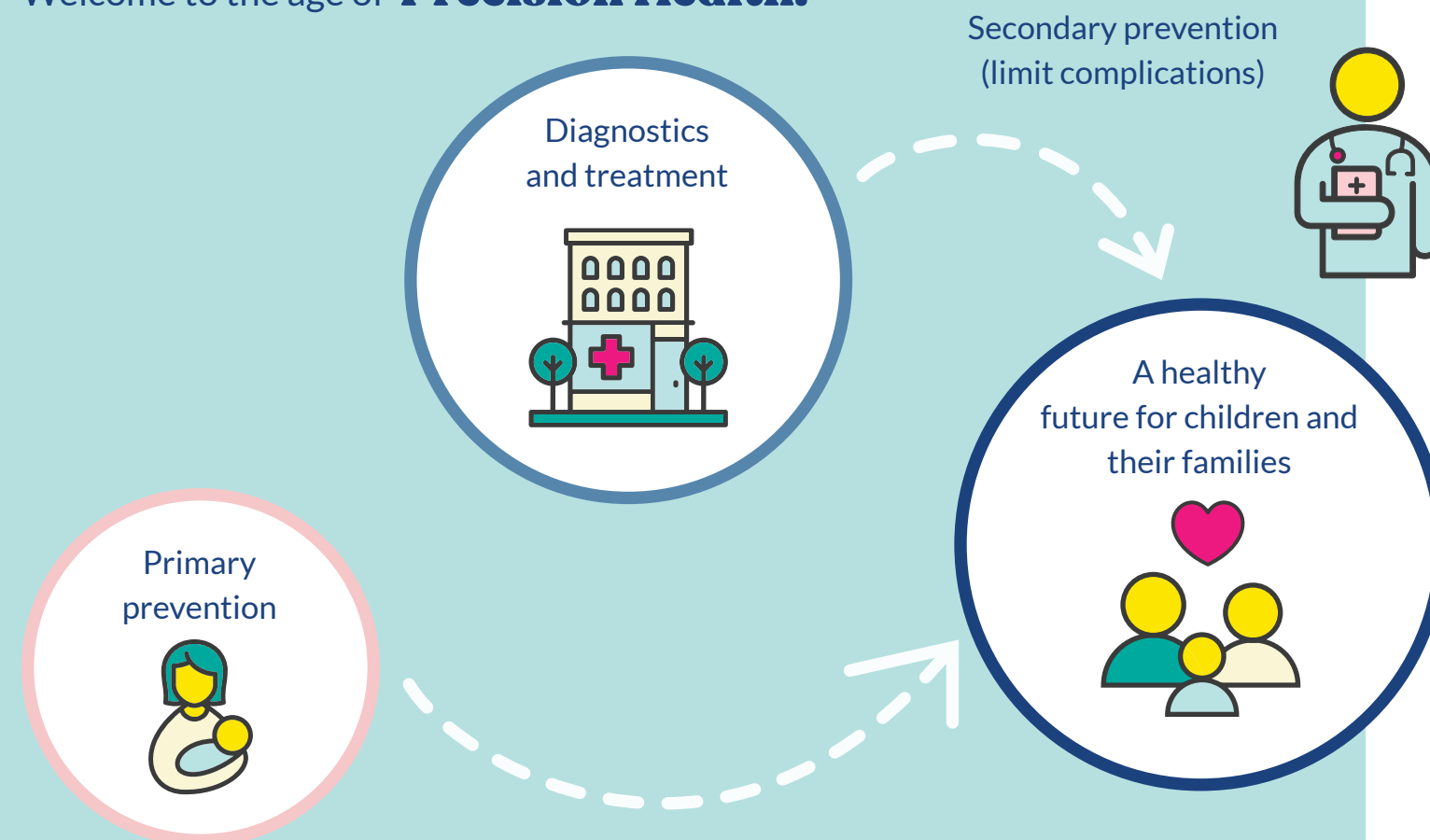
+600 publications

+1 200 individuals

A Patient-Centric Focus

No two children have the same **genetic makeup**, the same **environmental history**, the same **developmental trajectory** or the same **diagnosis**. As such, it is important to **adapt every intervention plan** to a child's individual profile.

Welcome to the age of **Precision Health**.



The aim of precision health is to develop **diagnostics that are more accurate and more personalized within a shorter timeframe**. In precision health, there's no such thing as an "average" patient. Each individual is unique, and each approach is tailored to the variables that make them who they are.

Precision health also applies to **promotional initiatives**, with the development of **preventive intervention programs** in neuropsychiatry, cardiovascular health, infectious disease and more, with the overarching goal of improving the health of the Quebec population as a whole.

Childhood is a window of opportunity, where intervention has a greater impact than at any other time in life.

Developing effective early treatment and intervention programs is the **Research Centre's** main mission.





Diagnosics and treatment: Precision medicine

Genomics, bioinformatics and omics

Genomics is one of the main thrusts of our **precision medicine** development strategy. Nearly 40% of patients with a rare genetic disorder do not have a confirmed diagnosis even when their genome has been partially sequenced because our knowledge of the corresponding genes is incomplete and the methods we use to analyze them have yet to be perfected.

We have therefore endeavoured to **recruit high-level researchers** in **genomics** as well as **bioinformatics** to optimize the use of **genomic data**. In addition, we have made a point of **acquiring various omics platforms**.

We welcomed the following professionals in 2019–2020:



Vincent-Philippe Lavallée, MD, a clinician-researcher with the Department of Pediatrics, who is working to gain a clearer understanding of how **recurrent mutations** alter the transcriptome and shape the hierarchy and plasticity of tumour and immune cells in **acute leukemia** to develop targeted approaches to treating pediatric cancers.

Despoina Manousaki, MD, PhD, a clinician-researcher with the Department of Pediatrics. Her team studies **genetics, bioinformatics, biomarkers and genetic epidemiology** to better understand the genetic architecture of **complex diseases in childhood**, including obesity and endocrine disorders, and to **leverage genomic data** to generate relevant findings for clinicians.



Martin Smith, PhD, a researcher with the Department of Biochemistry and Molecular Medicine. Dr. Smith is a computational biologist with broad experience in **genomics, infectious disease and immunology**. He is interested in **genome and transcriptome annotation** using **comparative genomics and machine learning**. Through his work, he seeks to utilize real-time nanopore sequencing to develop and implement new immune disease and cancer applications.

We have also actively worked to **develop our infrastructure and initiate major projects** to fully harness the potential of this technology.

Funding obtained by **Étienne Caron, PhD**, from the Canadian Foundation for Innovation (CFI)'s **John R. Evans Leaders Fund**, in conjunction with the CHU Sainte-Justine Foundation and the Fondation Charles-Bruneau, will help develop a **mass spectrometry platform** focusing on cancer immunotherapy to gain a fuller understanding of the mechanisms of tumour resistance in certain patients in response to this type of therapy.

Gene, cell and tissue therapy

The advent of new technologies such as the production of **induced pluripotent stem cell (iPSCs), CRISPR gene editing, organoid culture and safer new-generation viral vectors** augurs well for advances in the clinical applications of **gene, cell and tissue therapy**.

The **Research Centre** is well positioned to contribute to the development of these therapies, backed by Sainte-Justine's clinical cell therapy program, research platforms and a critical mass of experienced investigators.

We recently welcomed **Houman Savoji, PhD**, to the Department of Pharmacology and Physiology, in collaboration with the TransMedTech Institute. His primary research interests lie in advanced manufacturing with particular emphasis on **micro- and nano-fabrication of functional biomaterials for tissue engineering and regenerative medicine applications**. His focus is on designing, developing, optimizing, implementing and characterizing novel functional biomaterials using emerging engineering technologies including 3D bioprinting, microfluidics and electrospinning.



Neurodevelopment

Study of neural circuits



Roberto Araya, PhD, is a researcher with the Department of Neurosciences. His research interests include **synaptic integration of neocortical pyramidal neurons, and the structure and function of dendritic spines** and their role in the processing of information and the plasticity of cortical circuits.

The research focus for **Jannic Boehm, PhD**, also with the Department of Neurosciences, is **changes in synaptic plasticity, regulation of mRNA transport and protein translation during synaptic plasticity**.



Neuroimaging



Benjamin De Leener, PhD, a researcher with the Department of Computer and Software Engineering, in collaboration with the TransMedTech Institute, is an expert in computer engineering, whose main research interests pertain to the **development of new methods to analyze and process medical data**, specifically as it applies to magnetic resonance imaging and neuroscience. One of his major contributions is the development of the **Spinal Cord Toolbox** open-source software for analyzing spinal cord imaging.

Alexander Weil, MD, a clinician-researcher with the Surgery Department, is conducting research focusing on **molecular biology and pediatric epilepsy, the identification of predictive factors of surgical outcome in epilepsy, the discovery of new treatments and the application of new technologies in neurosurgery** to improve the safety and efficacy of surgery and increase patient survival rates.





Primary prevention

Early childhood offers a unique window of opportunity for breaking the intergenerational cycle linking socioeconomic adversity and health problems such as **mental disorders and obesity as well as academic failure. Intervention very early in life** targeting expectant mothers and young children is the best investment society can make in improving human capital.



During the year, we welcomed **Nicholas Chadi, MD**, a clinician-researcher with the Department of Pediatrics. His research interests lie in the **prevention and treatment of adolescent substance abuse disorders** and specifically on problems related to **tobacco, e-cigarette and marijuana addiction**. His work also seeks to improve the understanding of the impacts of public policy, risk perceptions and personal beliefs on alcohol and drug use among teenagers.

Artificial intelligence and decision support systems

Research revolving around **deep learning and big data** represents one of our strategic priorities for developing decision-making tools used in managing various pediatric conditions.

Notable hires in the past year include:

Morgan Craig, PhD, a researcher with the Department of Mathematics and Statistics, who uses **quantitative and translational medicine** to study biologically relevant questions of significant medical importance using quantitative approaches, particularly computational biology, with emphasis on the **optimization of treatment strategies for a variety of diseases by unravelling their pathophysiological mechanisms**.



Simon de Montigny, PhD, a researcher with the School of Public Health, who is working on the **development of decision support systems using artificial intelligence and deep learning** in the fields of cancer, infectious disease and intensive care.

Clinical research and patient partners

The environment at Sainte-Justine is particularly conducive to research, given the high volume of clinical activity, the breadth and depth of staff expertise, and the hospital's state-of-the-art infrastructure.

One of the main objectives of our plan is to further pursue **excellence in clinical research** at the **Research Centre** and **accelerate medical innovation to respond more effectively to the needs of the people we serve**. We work closely with patient partners in an effort to improve the quality of care and services, drawing on their own experience with our healthcare professionals.

New staff members who joined us during the year include **Melissa Fiscaletti, MD**, a clinician-researcher with the Department of Pediatrics. She studies **juvenile osteoporosis**, with particular emphasis on *osteogenesis imperfecta*, using whole exome and whole genome sequencing to identify novel mutations and understand the underlying molecular mechanisms.





Secondary prevention

Technology Centre for Pediatric Rehabilitation

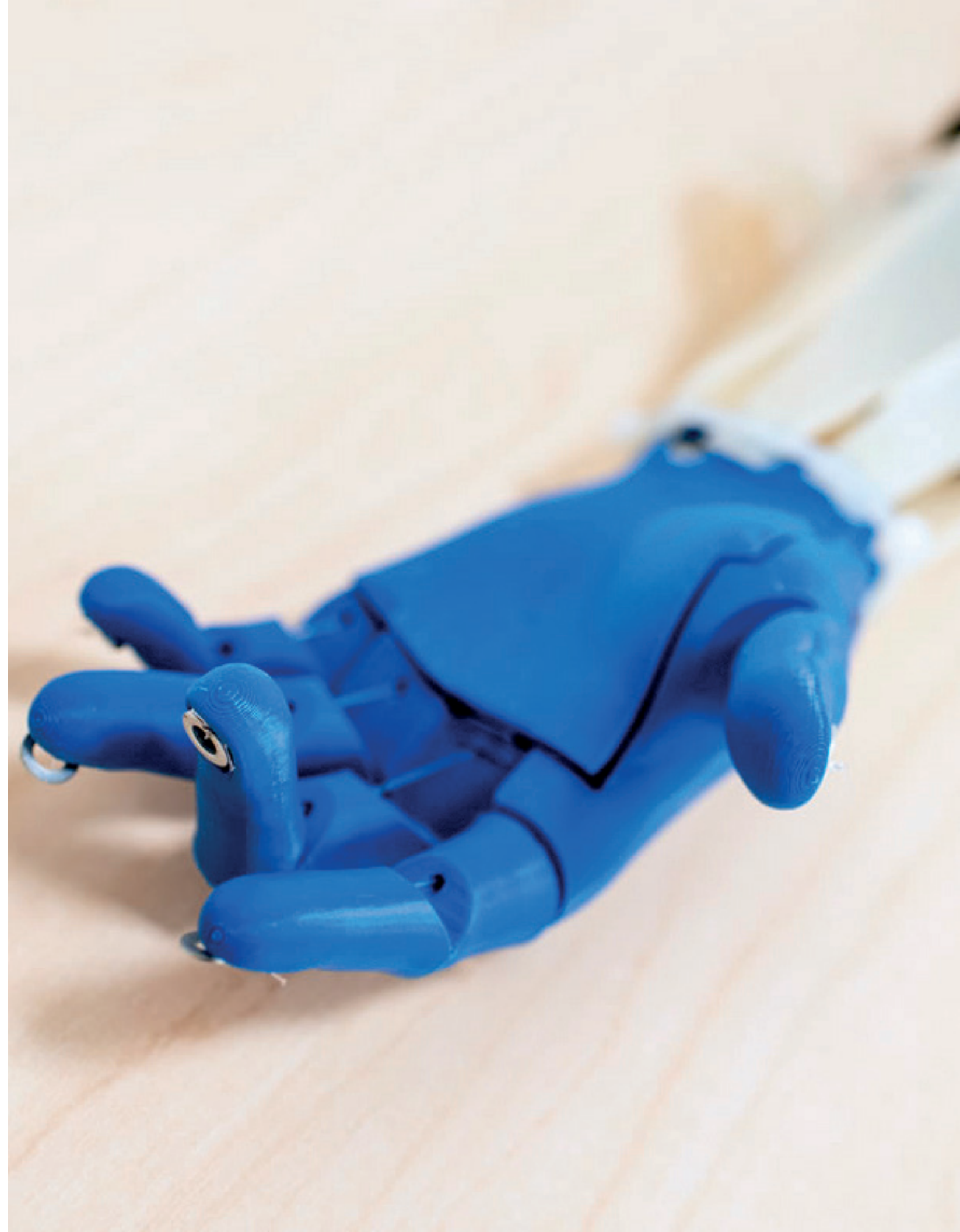
This major initiative brings healthcare professionals, researchers and industry partners together under the same roof to develop **forward-thinking rehabilitation approaches** in line with a precision medicine vision. We have taken advantage of the Centre's innovative platforms to attract several leaders in this field, including:



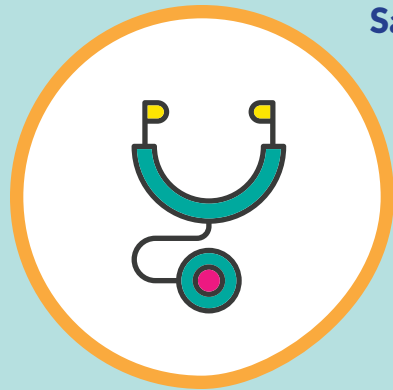
Philippe Dixon, PhD, a researcher with the School of Kinesiology and Physical Education Sciences, whose expertise is in the analysis of human movements. His research embraces a **biomechanical**

approach to understanding movements in populations with musculoskeletal disorders. The aim of his work is to help improve mobility and quality of life, inform clinical interventions and advance kinesiological science through the use of a rigorous, innovative methodology.

Abolfazl Mohebbi, PhD, a researcher with the Department of Mechanical Engineering in collaboration with the TransMedTech Institute, whose research interests include **neuromuscular control and rehabilitation, biomechanics, medical robotics and robotic surgery, applied artificial intelligence and multi-criteria decision analysis in medicine, and virtual reality and haptics.**



Clinical Research



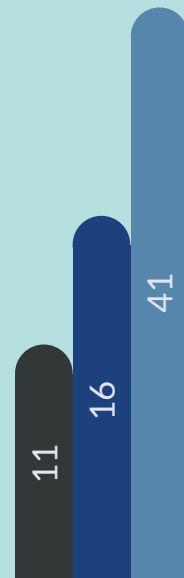
Sainte-Justine’s Research Directorate and Executive Management Office have made it a priority to set up a new governance and centralized management structure for clinical research.

This new structure is aligned with Sainte-Justine’s commitment to continuous quality improvement and integrated risk management. It has enabled the **development of key processes for standardizing best practices in clinical research** across the institution.

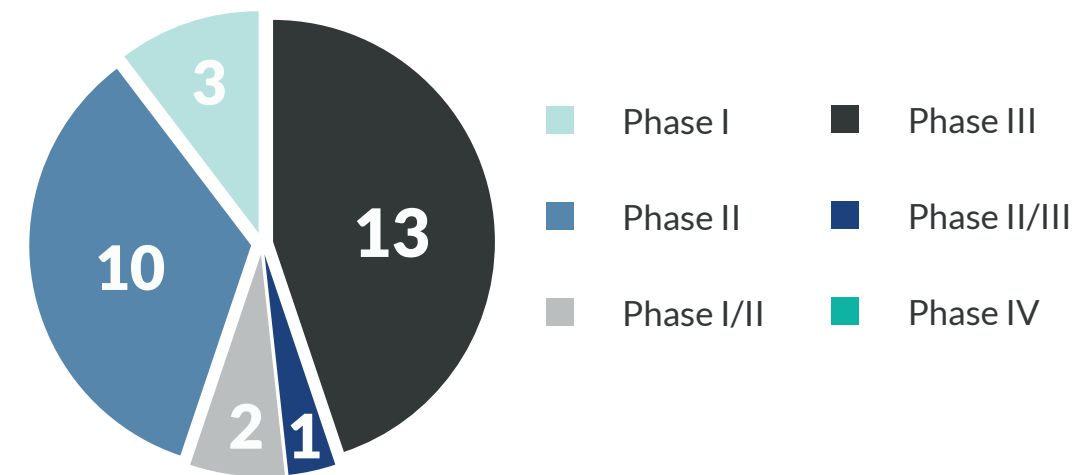
Clinical Research by the Numbers

New clinical trial projects submitted

- Single-center projects
- Multicenter projects with internal evaluation
- Multicenter projects with external evaluation

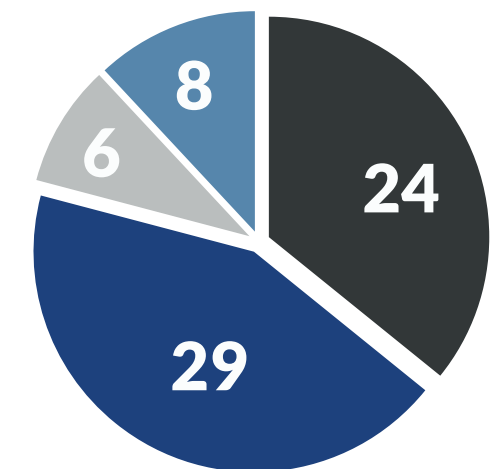


Number of clinical trial projects by experimental research phase

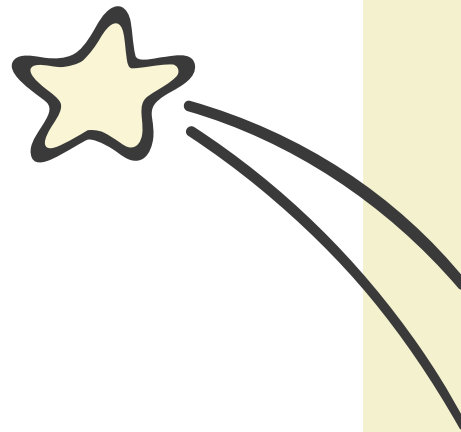


Number of clinical trial projects by type of funding

- Industry initiated and / or funded
- Academic funding
- No funding
- Other



Outreach



A year filled with exciting new discoveries



Major breakthroughs in congenital heart disease

The research team of **Gregor Andelfinger, MD**, discovered a new mechanism involved in a common congenital disease of the aortic valve. The culprit is the *ADAMTS19* gene, the mutation in which changes the regulatory mechanism in the valves. The findings of the study, which was carried out in conjunction with the Hadassah Hebrew University Medical Center, were published in the esteemed *Nature Genetics* journal.

His team also discovered a promising new treatment for infants with Noonan syndrome, a rare genetic condition that generally presents at birth and is often linked to early-onset severe heart disease. These research findings can be found in the *Journal of the American College of Cardiology*.

In addition, Dr. Andelfinger was singled out as a **“Personality of the Week”** by *La Presse* in recognition of this groundbreaking work.

Down syndrome: Insights into the role of the RCAN1 gene in memory and learning

The findings of a study conducted by **Jannic Boehm, PhD**, published in *Current Biology*, shed light on a new mechanism involved in the expression of Down syndrome, one of the main causes of intellectual disability and congenital heart defects in children. Dr. Boehm’s team investigated the *RCAN1* gene, which is overexpressed in the brains of fetuses with Down syndrome. Their work helps explain how *RCAN1* influences the way the condition manifests itself.

Understanding the mechanisms underlying chronic granulomatous disease

What are the underlying mechanisms of the rare genetic condition known as chronic granulomatous disease (CGD)? That is what a team of investigators led by **Fabien Touzot, MD, PhD**, determined when they created the first cellular model of CGD, a hereditary illness caused by mutations in the NADPH oxidase enzyme. The *Journal of Allergy and Clinical Immunology* published the study.





Teen depression linked to social media and television consumption

Using social media and watching TV are linked to an increase in depression in teenagers, according to a study by **Patricia Conrod, PhD**, published in the scientific journal *JAMA Pediatrics*. The study showed that when teens increased their use of social media and watched more TV than usual in a given year, their symptoms of depression also increased in that year. More frequent users of social media and TV had more severe symptoms of depression.

The study was the **second-most popular article** in *JAMA Pediatrics* in 2019, according to the Altmetric attention score, which collates mentions of scholarly articles across traditional and social media.

Connections between vaping and marijuana use

Research led by **Nicholas Chadi, MD**, on the association between electronic cigarette and marijuana use among adolescents and young adults was featured in *JAMA Pediatrics*. The extensive meta-analysis found that 12-to-17-year-olds who vaped were more likely to use marijuana than 18-to-24-year-olds who vaped.

Transfusions using fresh red blood cell units are not more beneficial to children in intensive care

A joint study led by **Marisa Tucci, MD**, and investigators at the Washington University School of Medicine in St. Louis showed that children with anemia who were given a transfusion in intensive care experienced no benefits from receiving fresher red blood cell units, i.e., those stored for seven or days or less, compared with those who received older units. Furthermore, the frequency and severity of organ failure or death in critically ill children were not reduced when fresher units were used. These findings appeared in the *Journal of the American Medical Association*.




Speeding up the drug discovery process to help patients

A new strategy that can predict the potential clinical implications of new therapeutic compounds based on simple cellular responses was developed by an international research team co-led by **Graciela Piñeyro, PhD**. This represents a major step forward in developing more effective drugs with fewer side effects, much faster than before. The study was published in *Nature Communications*.



Awards and Distinctions



Fernando Alvarez, MD, received the prestigious **Sass-Kortsak Award** at the Canadian Liver Meeting for his noteworthy contributions to the advancement of knowledge of liver disease in children. This is one of the most coveted honours in the world of pediatric hepatology.

Miriam Beauchamp, PhD, was named to the **Royal Society of Canada's College of New Scholars, Artists and Scientists**. The College recognizes individuals who have begun demonstrating leading scholarly, research or artistic excellence within 15 years of having completed their postdoctoral program or its equivalent.

Sylvain Chemtob, MD, PhD, was awarded the **2019 Acfas Léo-Pariseau Prize for biological sciences and health sciences** in recognition of the excellence and impact of his work and his contributions. Dr. Chemtob also received the **Michel-Sarrazin Prize** at the 61st congress of the Club de recherches cliniques du Québec. The annual career achievement award is given to a Quebec-based scientist for outstanding research contributions. Dr. Chemtob is one of the world's leading experts in retinopathy of prematurity.



Jacques Lacroix, MD, was presented with the **2019 Distinguished Lecturer Award in Critical Care Sciences** from the Institute of Circulatory and Respiratory Health of the Canadian Institutes of Health Research (CIHR) and the Canadian Critical Care Society in recognition of his extraordinary contributions to the advancement of critical care sciences in Canada.

Anne Monique Nuyt, MD, became the **Sainte-Justine Circle Research Chair of Developmental Origins of Health and Disease (DOHaD)**. The initiative was made possible through the exceptional commitment of the Sainte-Justine Circle to the Centre of Excellence in Neonatology. The purpose of this research chair is to advance knowledge and best practices related to the care of preterm babies.

Caroline Quach, MD, was inducted as a **Fellow of the Canadian Academy of Health Sciences** in recognition for her substantial contributions to the field as well as for the excellence and innovative nature of her work. She was also named one of the **Women's Executive Network's Top 100 Most Powerful Women in Canada**, an award that recognizes the incredible accomplishments of Canadian women and increases the visibility of strong leaders in order to inspire future generations.



Partnerships

A collaborative undertaking involving 1,000 families to transform autism care

A donation of close of \$10 million from the **Marcelle and Jean Coutu Foundation** was announced to enable Sainte-Justine, the Hôpital en santé mentale Rivière-des-Prairies du CIUSSS du Nord-de-l'Île-de-Montréal, the Neuro – McGill University and McGill University Health Centre (MUHC), the Douglas Mental Health University Institute and the Montreal West Island Integrated University Health and Social Services Centre (CIUSSS-ODIM), all partner institutions of the **Transforming Autism Care Consortium (TACC)**, to establish **Quebec 1,000 (Q1K)**, an innovative, multidisciplinary and collaborative initiative designed to have an unprecedented impact on autism research.

Institutional Events

Big data and artificial intelligence: Strategic brainstorming day

Sainte-Justine held its first **strategic brainstorming day** in October 2019, exploring various facets of **big data and artificial intelligence**.

The event enabled participants to:

- Discuss current and future applications of big data and AI in health care, teaching and research.
- Address the technological considerations involved in data capture, data warehouses and other aspects.
- Contemplate ethical and legal issues.
- Advance a coherent big data and AI development strategy for Sainte-Justine.





Workshop in partnership with NRC

In 2019, our partnership with the National Research Council (NRC) entered a new phase with the **signing of an agreement setting out the legal framework for collaborative initiatives**. The new organizational structure known as the **Collaborative Unit for Translational Research** will focus on five priority areas: immunology, rare diseases, perinatology, therapeutics and micro-nano biodevices. These areas will be respectively overseen by Drs. Elie Haddad, Gregor Andelfinger and Sylvain Chemtob, from Sainte-Justine, and Drs. Sue Twine and Teodor Veres, from NRC.

To launch this new phase, a **kick-off workshop was held on October 30, 2019**, at Sainte-Justine, with more than 100 researchers, students and postdoctoral fellows in attendance. A series of presentations highlighted the strengths of each area, spurring a number of lively discussions, which carried over to the dynamic networking session that closed out the day's activities. The event was a resounding success, leading to the emergence of a dozen new project ideas for a subsequent call for proposals.

RLet's start again from the beginning: Why and how should we support maternal mental health in the perinatal period?

The **transition to parenthood can be both rewarding and demanding**. Many new parents experience moments of vulnerability, where their mental health is put to the test by the new challenges they are up against. This is even truer for families who are socially isolated, experiencing financial hardship or dealing with perinatal complications. Resources to help these families are currently lacking or ill suited to their needs.

During this seminar spearheaded by **Sylvana Côté, PhD**, and **Anne Monique Nuyt, MD**, three experts in economic and psychiatric matters outlined ways that can be used to promote mothers' mental health and foster an optimal environment for children.



Parliamentary committee on preschool education for 4-year-olds

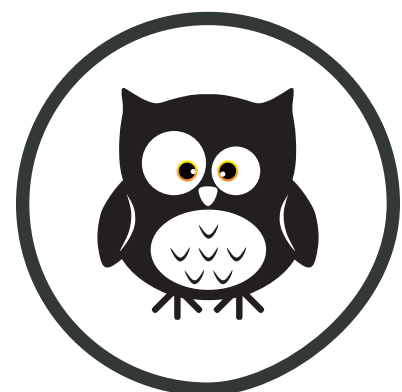
Sylvana Côté, PhD, submitted a brief to the Quebec National Assembly as part of the parliamentary committee on Bill 5, *An Act to amend the Education Act and other provisions regarding preschool education services for students 4 years of age*. The title of the brief was **Les maternelles 4 ans et les services d'éducation préscolaire pour les enfants de familles défavorisées : les recherches québécoises en contexte**

international (Kindergarten for 4-year-olds and preschool education services for children from underprivileged families: Quebec studies in an international context). Its aim was to shed light on the impact this choice could have on the development of children from low-income communities and summarize the findings of Quebec studies on the connections between daycare services, junior kindergarten and child development in socioeconomically insecure families.

Night Lab – Forest of Mysteries

The **fourth annual Night Lab – Forest of Mysteries** took place on February 29, 2020, as part of the Nuit blanche à Montréal event organized by the MONTRÉAL EN LUMIÈRE festival.

More than 1,200 visitors took advantage of this opportunity to make friends with **Scientix the Owl** and take part in some fascinating hands-on activities and experiments, where they could get a closer look at various lab instruments, participate in a scientific photo contest, watch lab teams at work and talk to experts in maternal and pediatric health.



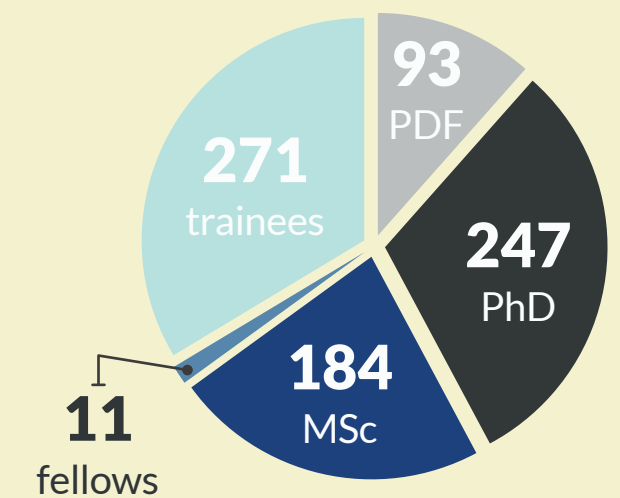
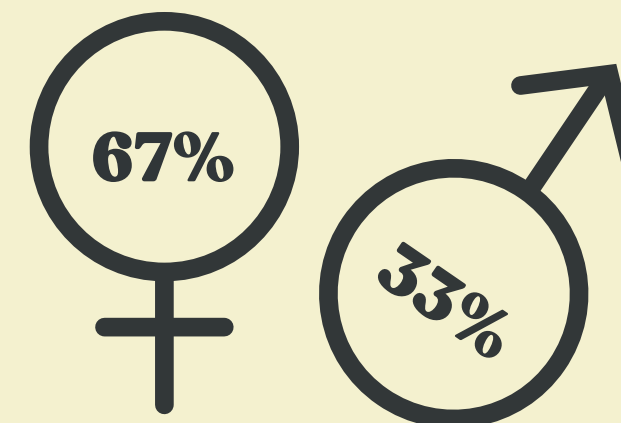
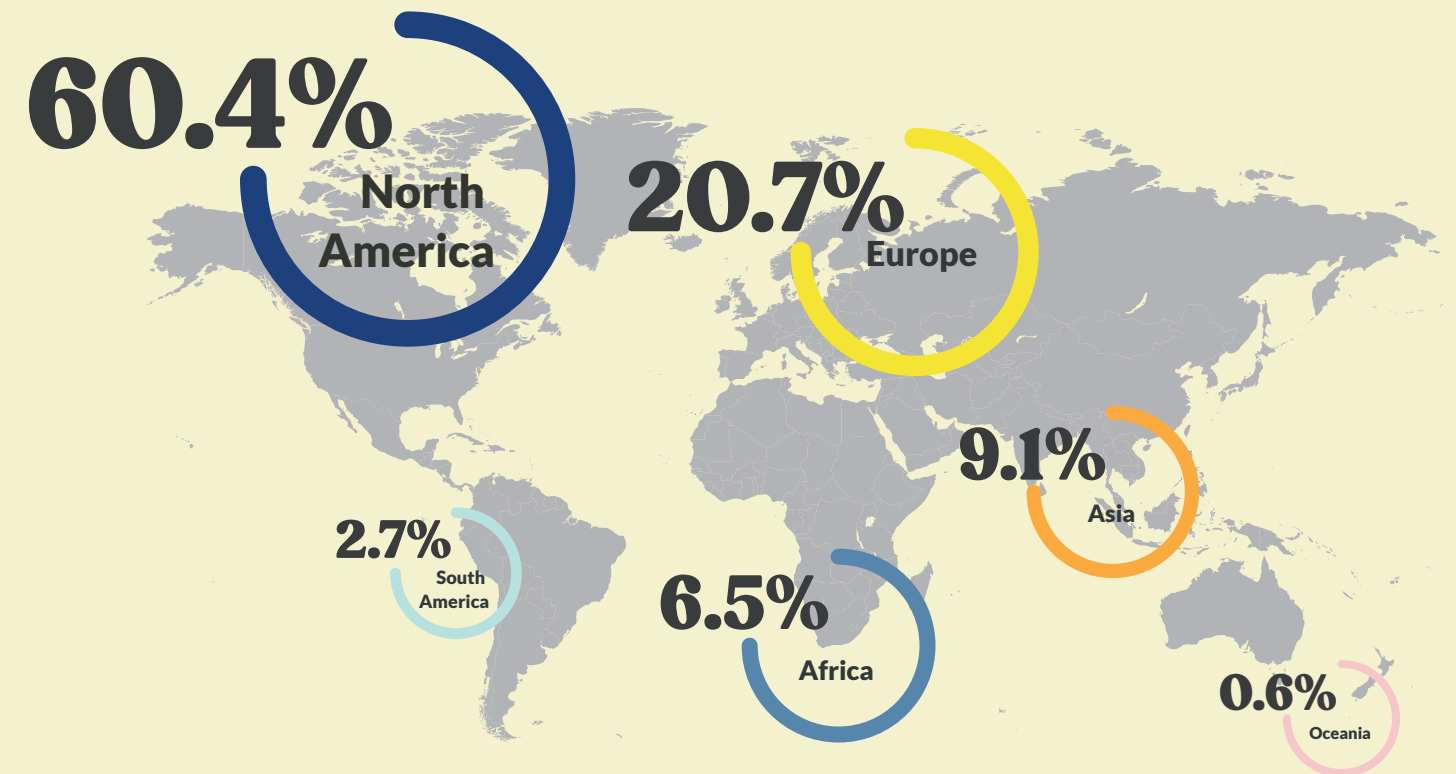
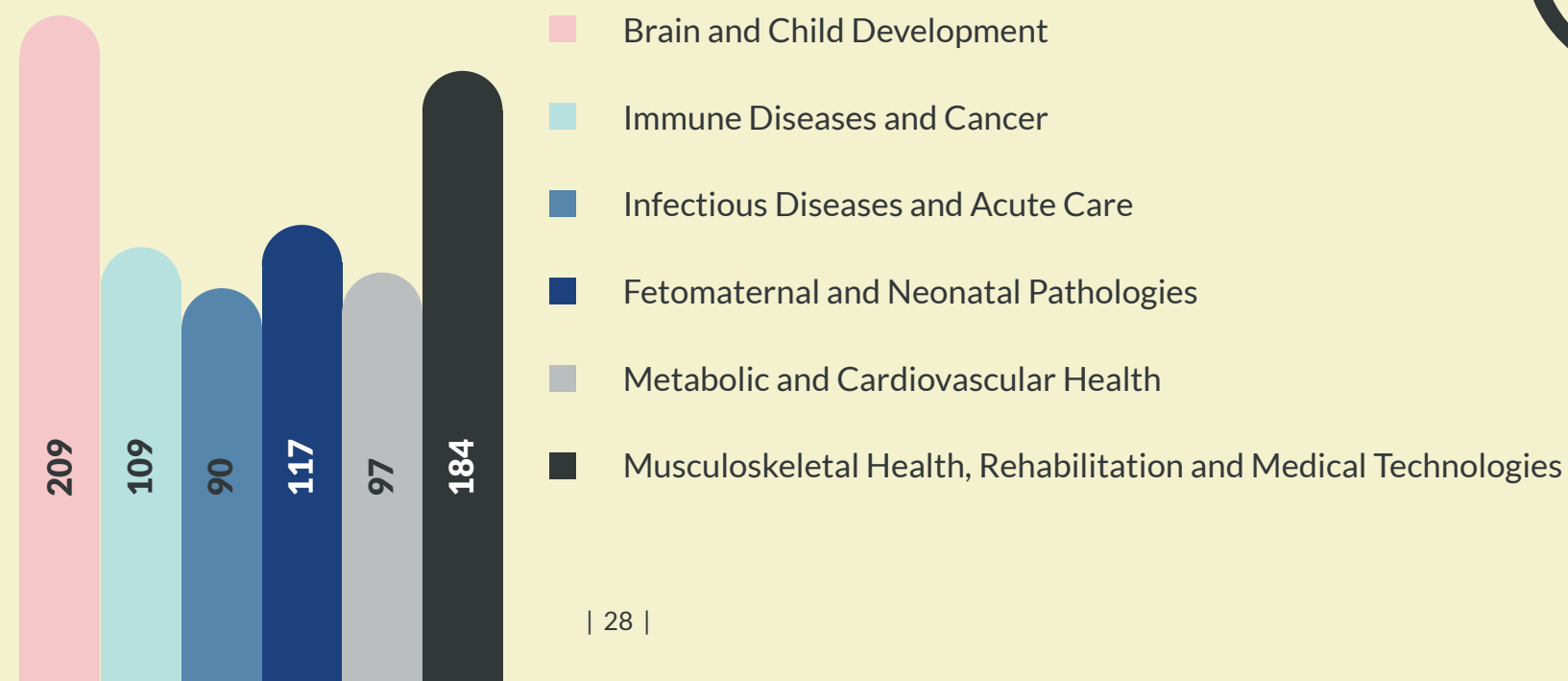
Teaching the Scientists of Tomorrow

Our student community at a glance

By grooming the next generation of scientists through **multi-, inter- and transdisciplinary research training programs** with a patient-oriented approach, we will be able to ensure long-term continuity and excellence in mother-and-child health.

Under the guidance of **world-class researchers**, our students learn the tools of the trade in state-of-the-art laboratory facilities alongside colleagues from around the world. **Nearly 40%** of our graduate and postgraduate students come to us from **one of 50 countries**.

Number of students, interns and fellow per research axis



Student success



Fonds de recherche du Québec

A total of **30 students and postdoctoral fellows** were awarded a scholarship during the 2019–2020 FRQ competitions.

Safari Joseph Balegamire, a PhD student working under the supervision of **Benoît Mâsse, PhD**, and **Isabelle Boucoiran, MD**, finished first in the doctoral training scholarship competition for his project on the prevalence, risk factors and impacts on pregnancy and newborns with regard to non-primary cytomegalovirus infection during pregnancy in Quebec.

Fonds de recherche du Québec – Santé

Fanny Dégeilh, a postdoctoral fellow working under the supervision of **Miriam Beauchamp, PhD**, won the **Relève étoile Jacques-Genest award** from the FRQS for her paper, *Quality of maternal behaviour during infancy predicts functional connectivity between default mode network and salience network 9 years later*, published in *Developmental Cognitive Neuroscience*. The objective of this award is to recognize exceptional research contributions of university students and postdoctoral fellows.

CHU Sainte-Justine Foundation

Of the 73 students who applied to the **CHU Sainte-Justine Foundation Scholarship competition**, 22 were awarded funding. The winners were selected by an evaluation committee based on academic performance, previous awards and scholarships, publishing activity and the promising nature of their research project.



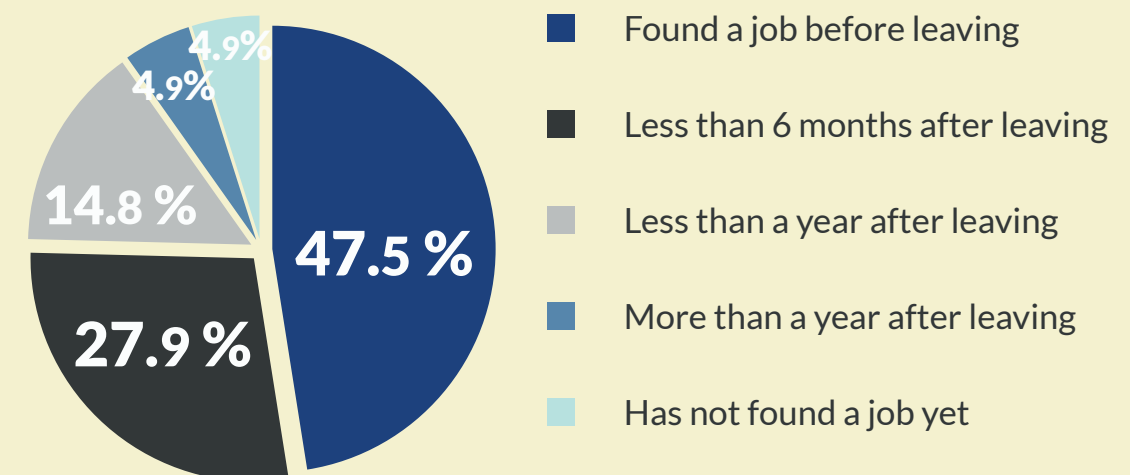
Scholarship granting organizations

- Basque Government
- BoBeau Coeur Fund
- Brain Canada Foundation
- Canadian Francophonie Scholarship Program (CFSP)
- Canadian Institutes of Health Research (CIHR)
- Canadian Vascular Network (CVN)
- CHU Sainte-Justine Foundation
- Cole Foundation
- Fonds de recherche du Québec – Nature et technologies (FRQNT)
- Fonds de recherche du Québec – Santé (FRQS)
- Fonds de recherche du Québec – Société et culture (FRQSC)
- Grand défi Pierre Lavoie
- International Society for Pediatric and Adolescent Diabetes (ISPAD)
- Ministère de l'Éducation et de l'Enseignement supérieur du Québec (MEES)
- Mitacs
- Natural Sciences and Engineering Research Council of Canada (NSERC)
- Savoy Foundation
- Social Sciences and Humanities Research Council of Canada (SSHRC)

Survey of former students

Survey of students who left the **Research Centre** between 2010 and 2020.

Period of time before finding a job:

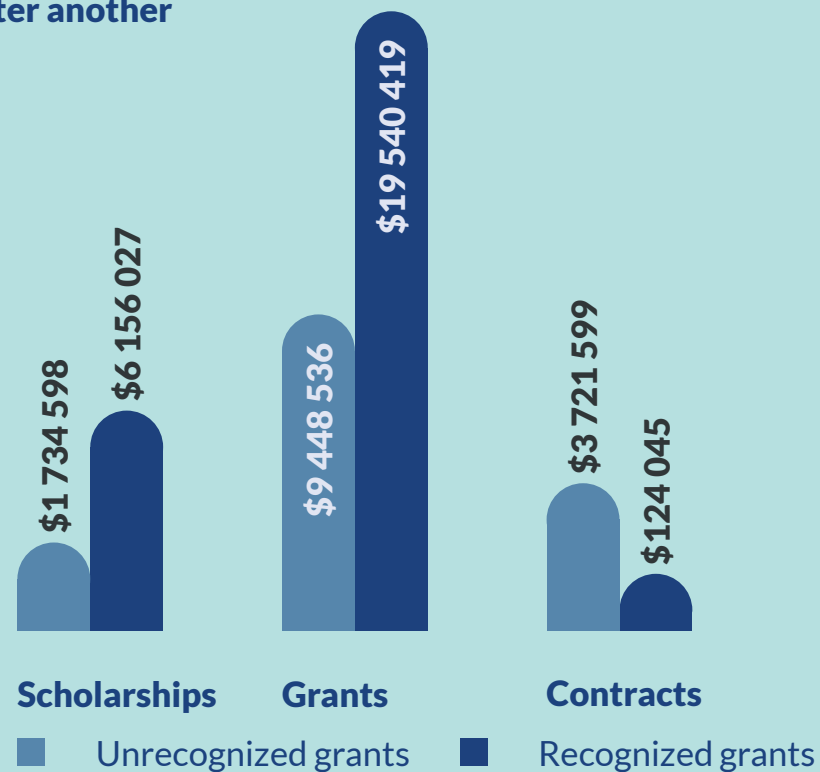


Funding

Another banner year for the Research Centre

Pediatric research is a very competitive field. Given our **reputation for excellence both domestically and internationally**, our **teams have a head start when it comes to winning the race** for research funds and scholarships and getting major strategic initiatives off the ground.

Once again this year, our **Research Centre** professionals managed to set themselves apart in a crowded playing field, **walking away with one competition after another at the provincial, national and international level.**





Multiple distinctions

Bénédicte Amilhon, PhD, and **Étienne Caron, PhD**, were selected for the Canada Foundation for Innovation's **John R. Evans Leaders Fund** to develop a platform to analyze neural circuits and a mass spectrometry platform for cancer immunotherapy.

Philippe Bégin, MD, PhD, was the recipient of a **CIHR grant** to develop oral immunotherapy, a new treatment for food allergies. **Sébastien Perreault, MD**, was also awarded **CIHR funding** to conduct a pan-Canadian, multicentre phase II study of the use of trametinib to treat refractory low-grade gliomas (the most common brain tumour in children) and plexiform neurofibromas (a rare benign nerve tumour).

Anick Bérard, PhD, saw her **CAN-AIM team funding** from **CIHR** renewed to continue her study on drug safety during pregnancy. She was also awarded a **CIHR grant** for the **Canadian Mother-Child Cohort Active Surveillance Initiative**.



Michel Duval, MD, obtained financial support as part of the **LeadAction-Onco competition** created by Oncopole and IRICoR for his project on the development of a novel therapeutic approach to maximize the effect of the immune system against leukemia.

Sébastien Jacquemont, MD, and his research team will be part of the **Genome to Mental Health (GMH) consortium**, a new initiative with \$6 million in funding under the Rare Genetic Disorders as a Window into the Genetic Architecture of Mental Disorders by the National Institute of Mental Health (NIMH) and the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD).

Marie Laberge, PhD, obtained a grant for her project to develop an integrated model for the management of occupational injury prevention supported by digital technologies for the Work-Oriented Training Path (WOTP) from the **Institut de recherche Robert-Sauvé en santé et en sécurité du travail**.

Major Publications



Brain and Child Development

Researcher	Journal	Paper title
Boehm, Jannic	<i>Current Biology</i>	RCAN1 Regulates Bidirectional Synaptic Plasticity
Chadi, Nicholas	<i>JAMA Pediatrics</i>	Treatment for Nicotine Use Disorder Among Medicaid-enrolled Adolescents and Young Adults
Conrod, Patricia	<i>JAMA Pediatrics</i>	Association of Screen Time and Depression in Adolescence
Conrod, Patricia	<i>Neuron</i>	A Neuroethics Backbone for the Evolving Canadian Brain Research Strategy
Di Cristo, Graziella	<i>Journal of Neuroscience</i>	p75 Neurotrophin Receptor Activation Regulates the Timing of the Maturation of Cortical Parvalbumin Interneuron Connectivity and Promotes Juvenile-like Plasticity in Adult Visual Cortex
Jacquemont, Sébastien Forgeot d'Arc, B.	<i>Science Translational Medicine</i>	Tinkering with the Vasopressin Pathway in Autism
Lippé, Sarah Krajcinovic, M.; Laverdière C.; Sinnett, D.	<i>Cancer</i>	Visual Short-Term Memory Activation Patterns in Adult Survivors of Childhood Acute Lymphoblastic Leukemia
Piñeyro, Graciela	<i>Nature Communications</i>	Exploring Use of Unsupervised Clustering to Associate Signaling Profiles of GPCR Ligands to Clinical Response
Théorêt, Hugo Lepore, F.	<i>Annals of Neurology</i>	Neural Function in DCC Mutation Carriers with and without Mirror Movements
Weil, Alexander	<i>Nature Genetics</i>	Stalled Developmental Programs at the Root of Pediatric Brain Tumors

Immune Diseases and Cancer

Researcher	Journal	Paper title
Barreiro, Luis	<i>Nature</i>	IL-15, Gluten and HLA-DQ8 Drive Tissue Destruction in Coeliac Disease
Beauséjour, Christian Decaluwe, H.	<i>Aging Cell</i>	Restored Immune Cell Functions Upon Clearance of Senescence in the Irradiated Splenic Environment
Beauséjour, Christian	<i>Nature</i>	L1 Drives IFN in Senescent Cells and Promotes Age-associated Inflammation
Bégin, Philippe	<i>Journal of Clinical Immunology</i>	TREX-1-Related Disease Associated with the Presence of Cryofibrinogenemi
Bouron-Dal Soglio, Dorothee	<i>New England Journal of Medicine</i>	Treatment for Nicotine Use Disorder Among Medicaid-enrolled Adolescents and Young Adults
Haddad, Elie Touzot, F.	<i>Journal of Clinical Immunology</i>	Neuroinflammatory Disease as an Isolated Manifestation of Hemophagocytic Lymphohistiocytosis
Ferretti, Vincent	<i>Nature Communications</i>	A User Guide for the Online Exploration and Visualization of PCAWG Data
Raynal, Noël Sinnett, D.; Beauséjour, C.; McGraw, S.	<i>Journal of Experimental & Clinical Cancer Research</i>	Heart Failure Drug Proscillaridin A Targets MYC Overexpressing Leukemia through Global Loss of Lysine Acetylation
Sinnett, Daniel Bouron-Dal Soglio, D.; Duval, M.; Leclerc J.M.; Laverdière, C.; Tran, T.H.; Perreault, S.; Teira, P.; Cellot, S.	<i>JAMA Network Open</i>	Molecular Profiling of Hard-to-treat Childhood and Adolescent Cancers
Sinnett, Daniel Perreault, S.; Cellot, S.	<i>Oncogene</i>	Recurrent Somatic BRAF Insertion (p.V504_R506dup): A Tumor Marker and a Potential Therapeutic Target in Pilocytic Astrocytoma

Infectious Diseases and Acute Care

Researcher	Journal	Paper title
Boucoiran, Isabelle Kakkar, F.	<i>Clinical Infectious Diseases</i>	The Women and Children's Infectious Diseases Center: An Integrated Approach to Congenital Infectious Diseases
Emeriaud, Guillaume Gauvin, F.; Lacroix, J.	<i>Critical Care Medicine</i>	Transfusion-associated Circulatory Overload in ICUs: A Scoping Review of Incidence, Risk Factors, and Outcomes
Gravel, Jocelyn	<i>JAMA Pediatrics</i>	Pediatric Emergency Research Canada (PERC) Concussion Team. Natural Progression of Symptom Change and Recovery from Concussion in a Pediatric Population
Jouvet, Philippe	<i>Lancet Respiratory Medicine</i>	Paediatric Acute Respiratory Distress Syndrome Incidence and Epidemiology (PARDIE): An International, Observational Study
Jouvet, Philippe	<i>Nature Reviews Nephrology</i>	Consensus Guidelines for Management of Hyperammonaemia in Paediatric Patients Receiving Continuous Kidney Replacement Therapy
Kleiber, Niina	<i>Critical Care Medicine</i>	Enteral Acetaminophen Bioavailability in Pediatric Intensive Care Patients Determined With an Oral Microtracer and Pharmacokinetic Modeling to Optimize Dosing
Laberge, Sophie	<i>JAMA Pediatrics</i>	Effectiveness of Intrapleural Tissue Plasminogen Activator and Dornase Alfa vs Tissue Plasminogen Activator Alone in Children with Pleural Empyema: A Randomized Clinical Trial
Lacroix, Jacques	<i>Journal of the American Medical Association</i>	Effect of Fresh vs Standard-issue Red Blood Cell Transfusions on Multiple Organ Dysfunction Syndrome in Critically Ill Pediatric Patients: A Randomized Clinical Trial
Mâsse, Benoît	<i>Journal of the American Medical Association</i>	Effect of Maternal Docosahexaenoic Acid Supplementation on Bronchopulmonary Dysplasia-Free Survival in Breastfed Preterm Infants: A Randomized Clinical Trial
Soudeyns, Hugo Kakkar, F.	<i>Clinical Infectious Diseases</i>	Clinical Correlates of HIV-1 DNA and Inducible HIV-1 RNA Reservoirs in Peripheral Blood in Children with Perinatally Acquired HIV-1 Infection with Sustained Virologic Suppression for at Least 5 Years

Fetomaternal and Neonatal Pathologies

Researcher	Journal	Paper title
Andelfinger, Gregor	<i>Nature Genetics</i>	Loss of ADAMTS19 Causes Progressive Non-syndromic Heart Valve Disease
Bérard, Anick	<i>JAMA Psychiatry</i>	Association Between Incident Exposure to Benzodiazepines in Early Pregnancy and Risk of Spontaneous Abortion
Dubrac, Alexandre	<i>Nature Communications</i>	Endophilin-A2 Dependent VEGFR2 Endocytosis Promotes Sprouting Angiogenesis
Janvier, Annie	<i>American Journal of Bioethics</i>	Empirical Over Theoretical Ethics: Choosing what Matters to Patients and Families
Lodygensky, Gregory Chemtob, S.	<i>Brain, Behavior, and Immunity</i>	Assessing Therapeutic Response Non-invasively in a Neonatal Rat Model of Acute Inflammatory White Matter Injury Using High-field MRI
Lodygensky, Gregory McGraw, S.	<i>FASEB Journal</i>	Alteration of the Brain Methylation Landscape Following Postnatal Inflammatory Injury in Rat Pups
Luu, Thuy Mai	<i>Canadian Medical Association Journal</i>	Maternal Prepregnancy Surgery and Risk of Neonatal Abstinence Syndrome in Future Newborns: A Longitudinal Cohort Study
Luu, Thuy Mai Laverdière, C.	<i>International Journal of Cancer</i>	Neonatal Phototherapy and Future Risk of Childhood Cancer
Nuyt, Anne Monique Luu, T.M.	<i>JAMA Pediatrics</i>	Cardiovascular Risk in Adults Born Preterm: Time to Act
Van Vliet, Guy	<i>Pediatrics</i>	Avoiding the Overdiagnosis of Congenital Hypothyroidism in Preterm Newborns

Metabolic and Cardiovascular Health

Researcher	Journal	Paper title
Deladoëy, Johnny Van Vliet, G.	<i>Thyroid</i>	Wide Spectrum of DUOX2 Deficiency: From Life-threatening Compressive Goiter in Infancy to Lifelong Euthyroidism
Fournier, Anne Dahdah, N.S.	<i>Pediatrics</i>	Treatment Intensification in Patients with Kawasaki Disease and Coronary Aneurysm at Diagnosis.
Henderson, Mélanie	<i>New England Journal of Medicine</i>	Persistence of Obesity from Early Childhood Onward
Jantchou, Prévost Deslandres, C.	<i>Journal of Crohn's and Colitis</i>	Management of Paediatric Patients with Medically Refractory Crohn's Disease Using Ustekinumab: A Multi-centred Cohort Study
Joyal, Jean-Sébastien	<i>Proceedings of the National Academy of Sciences</i>	NOTCH1 Signaling Induces Pathological Vascular Permeability in Diabetic Retinopathy
Laberge, Anne-Marie	<i>American Journal of Bioethics</i>	Toward Broader Genetic Contextualism: Genetic Testing Enters the Age of Evidence-based Medicine
Lavoie, Jean-Claude	<i>Nutrients</i>	Glutathione Supplementation of Parenteral Nutrition Prevents Oxidative Stress and Sustains Protein Synthesis in Guinea Pig Model
Levy, Emile Marcil, V.	<i>Scientific Reports</i>	Altered Proteome of High-density Lipoproteins from Paediatric Acute Lymphoblastic Leukemia Survivors
Pshezhetsky, Alexey V.	<i>Journal of Clinical Medicine</i>	Molecular Bases of Neurodegeneration and Cognitive Decline, the Major Burden of Sanfilippo Disease
St-Pierre, David Lavoie, J.-C.	<i>Frontiers in Physiology</i>	A Short-term High-fat Diet Alters Glutathione Levels and IL-6 Gene Expression in Oxidative Skeletal Muscles of Young Rats

Musculoskeletal Health, Rehabilitation and Medical Technologies

Researcher	Journal	Paper title
Begon, Mickaël Ballaz, L.	<i>Clinical Neurophysiology</i>	Reliability of Maximum Isometric Hip and Knee Torque Measurements in Children with Cerebral Palsy Using a Paediatric Exoskeleton - Lokomat
Campeau, Philippe	<i>American Journal of Human Genetics</i>	Gain-of-function Mutations in KCNN3 Encoding the Small-conductance Ca ²⁺ -activated K ⁺ channel SK3 Cause Zimmermann-Laband Syndrome
Campeau, Philippe Carmant, L.; Rossignol, E.; Michaud, J.L.	<i>American Journal of Human Genetics</i>	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons
Campeau, Philippe	<i>American Journal of Human Genetics</i>	Mutations in ANAPC1, Encoding a Scaffold Subunit of the Anaphase-promoting Complex, Cause Rothmund-Thomson Syndrome Type 1
Dumont, Nicolas Michaud, J.L.; Campeau, P.	<i>Genetics in Medicine</i>	Biallelic Variants in the Transcription Factor PAX7 are a New Genetic Cause of Myopathy
Dumont, Nicolas	<i>Stem Cells</i>	Macrophages are Key Regulators of Stem Cells During Skeletal Muscle Regeneration and Diseases
Mailhot, Geneviève	<i>Transplantation</i>	Trends, Determinants, and Impact on Survival of Post-Lung Transplant Weight Changes: A Single-center Longitudinal Retrospective Study
Moreau, Alain Labelle, H.; Mac-Thiong, J.-M.; Parent, S.	<i>Scientific Reports</i>	A Differential Hypofunctionality of Gαi Proteins Occurs in Adolescent Idiopathic Scoliosis and Correlates with the Risk of Disease Progression
Rauch, Frank	<i>Bone</i>	Novel ActRIIB Ligand Trap Increases Muscle Mass and Improves Bone Geometry in a Mouse Model of Severe Osteogenesis Imperfecta
Rauch, Frank	<i>Genetics in Medicine</i>	Mobility in Osteogenesis Imperfecta: A Multicenter North American Study



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