

# Chez nous

MCH EMPLOYEE NEWSLETTER | Published by Public Relations and Communications | [www.thechildren.com](http://www.thechildren.com)

MARCH 2019



## Standing taller: young boy tackles ultra-rare disease — page 2

Hôpital de Montréal  
pour enfants  
Centre universitaire  
de santé McGill



Montreal Children's  
Hospital  
McGill University  
Health Centre

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# Bravo Lorenzo!

## Young boy tackles ultra-rare genetic disorder

By Stephanie Tsirgiotis

Seven-year-old Lorenzo Bravo is standing a lot taller these days. In the last year and a half, he's not only grown four inches, but he's also learned how to fasten his own seatbelt, open doors and climb a full flight of stairs. "These are all things he couldn't do just a little while ago,"

says Lorenzo's father, Jorge. "It's amazing how far he's come in such a short period of time."

### Falling off the growth curve

Lorenzo and his family immigrated to Canada in June 2012 from Lima, Peru. Jorge and his [continued >](#)

► (Top: l. to r.) Lorenzo's mother, Mariela, and his grandmother, Mary. (Bottom: l. to r.) Lorenzo's aunt, Maria Fernanda, and Lorenzo.

wife, Mariela Paredes, decided to move to Drummondville, Quebec, and quickly started a new life for themselves.

As a baby, Lorenzo seemed perfectly normal other than a very rigid walk. But then at the age of four, he stopped growing. “Everyone in my wife’s family is short, so we thought he was just following in their footsteps,” Jorge recalls.

But over the next year, Lorenzo’s fine motor skills stopped developing properly and he became less mobile. The nurse at his school also noticed that something was wrong with his eyes. They looked cloudy. Lorenzo was referred to Dr. Louise Auger, director of the Multicultural clinic at the Montreal Children’s Hospital (MCH). After his first visit, Dr. Auger thought he might be suffering from a genetic condition and sent him for further tests. Her hunch was right.

### An ultra-rare disease

Lorenzo was diagnosed with an ultra-rare genetic disorder called mucopolysaccharidosis type VI (MPS VI), also known as Maroteaux-Lamy syndrome. He began seeing pediatric endocrinologist, Dr. John Mitchell, director of the



► Dr. John Mitchell and Lorenzo share a good laugh.

MCH’s Endocrinology division and an expert in rare metabolic diseases. “There are only two patients in Quebec with this disease, and 10 in Canada,” he says.

MPS VI is caused by a mutation in the ARSB gene, which in these cases, stops the body from producing an enzyme needed to break down large sugar molecules, called glycosaminoglycans (GAGs). “These molecules build up inside cells, which inevitably leads to the

inflammation of various tissues and organs, especially those in the musculoskeletal system,” explains Dr. Mitchell. “Without proper treatment, the accumulation of these GAGs can cause cell death.”

Because the disease was affecting different organs and systems in his body, Lorenzo began being followed by a number of different specialties.

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**Chez nous** is published by the MCH Public Relations and Communications office.

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Production of *Chez nous* is made possible thanks to funding from the Montreal Children’s Hospital Foundation.

*On the cover:*  
Lorenzo Bravo

*Cover photo:* Owen Egan

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► (Top: l. to r.) Maria Fernanda, Lina Moisan, Lorenzo’s nurse, Mary, and Dr. John Mitchell. (Bottom: l. to r.) Mariela and Lorenzo.

“My wife and I had to close our company, because we were in the hospital at least three times a week,” says Jorge. “We both wanted to be there for our son.” Jorge’s mother, Mary, had a hard time watching her family struggle from a distance and decided to move to Canada from Chile to help with Lorenzo. “That decision really changed our lives. It allowed my wife and I to go back to school and better our lives,” he says. “We couldn’t do this on our own.”

### **Not a cure, but close**

In 2018, Dr. Mitchell started an enzyme replacement therapy for Lorenzo. Every

Monday, a nurse comes to the family’s home and gives Lorenzo a four-hour intravenous infusion. “We’re able to deliver the missing enzyme directly into his body and it begins clearing out the GAGs,” says Dr. Mitchell. “It works best with organs that have a higher blood flow, like the liver.” The treatment doesn’t work as well for organs such as the cornea, which have a slower blood flow. “This therapy is not a cure, but it does change the course of the disease. It affects different organs at different rates, but doesn’t reverse damage already done,” he adds.

For example, Lorenzo had to undergo hernia surgery in 2017, and he still suffers from carpal tunnel in both his hands, for which he’ll undergo surgery in late March and July. Lorenzo did manage to grow two to three inches in six months however, because the therapy helped to reduce the inflammation in his body. It has also greatly improved his mobility, endurance and fine motor skills. “Lorenzo no longer needs a full-time aide at school and his confidence has grown tremendously. He takes more time to get ready than other children, but he’s able to do everything by himself now,” says his father. “My wife gives us the strength to continue. She is an exceptional mother.”

Even though MPS VI is a genetic disorder, nobody else in Lorenzo’s family has the disease, and that includes his younger brother, Leonardo. Both his parents, however, are carriers. It’s a surprising set of odds given his parents’

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diverse backgrounds: Jorge's ancestors are from Germany and Mariela's can be traced back to China. "Lorenzo's disease changed our lives forever," says Jorge. "But it has also brought our family so much closer." ●



► Lorenzo can now climb a full flight of stairs without getting tired.

## Seizing the moment: Sarah's journey

March 26 is Purple Day, dedicated to increasing awareness about epilepsy worldwide

"Despite everything that she's gone through, Sarah is a happy, positive and constantly smiling seven year old. She even comes out of seizures laughing and telling jokes," says Jessica Lowe, Sarah's mom. Sarah was diagnosed with epilepsy in 2017, when uncontrollable seizures began taking over her daily life. She was placed on a variety of different medications to try and better control her illness, but the seizures were unrelenting and varied between convulsions and falling episodes with no abating. After much trial and error with medication, pediatric neurologist Dr. Kenneth Myers and pediatric neurosurgeon, Dr. Jean-Pierre Farmer, recently reviewed the results of lengthy testing conducted while trying to provoke Sarah's seizures for analysis.

"Dr. Farmer explained that Sarah's abnormal brain activity was coming from the right side of the brain in two separate points — the same side as where she had a tumour removed at three weeks old," says Jessica. "He recommended Sarah undergo a hemispherectomy to cut the connections between her left and right brain hemispheres with the goal of completely eliminating her seizures."

The decision to undergo such major surgery was by no means an easy one. Jessica says that while the family was informed that the surgery has a 90 per cent success rate of completely eliminating seizures, there were side effects to consider as well. "Despite the risks, these were all side effects that Sarah mostly deals with already," says Jessica. "On her worst day, Sarah can have upwards of 200 falling



seizures within a 24-hour period. And while she is developing well, and can walk, talk and goes to regular school, she does have issues with mobility and speech delays. The team explained to us that having the surgery would greatly improve Sarah's development. It was a decision that we thought long and hard about."

After speaking with several other families whose children had undergone the surgery with very positive results, the family decided to go forward with it, and Sarah's surgery is planned for some time at the end of this year. "We know it will be a major operation and we are prepared for the things that come with it," says Jessica. "What reassures us the most is that Dr. Farmer will be performing it. He's already helped Sarah before and we couldn't trust anyone more. We have a truly wonderful team surrounding us and they've made all the difference for Sarah and our family." ●

# Accreditation Canada

Over the next two issues, *Chez nous* will break down what you need to know and how to prepare for the upcoming visit by Accreditation Canada.

Health care accreditation is an ongoing process of assessing health care organizations against standards of excellence to identify what is being done well and what needs to be improved. Representatives from Accreditation Canada will be visiting the McGill University Health Centre (MUHC) between **May 27 and 30, 2019**.

## Why does everyone need to be prepared?

Specific teams throughout the MUHC will be targeted for assessment during this visit. At the Montreal Children's Hospital, this includes Mental Health.

But even if you don't work in Mental Health, all staff members and physicians at the MCH must be prepared to answer questions by the surveyors conducting the accreditation. This includes being informed about key transversal topics, such as: the MUHC's organizational

activities, patient partnership activities, staff roles and responsibilities, staff training, quality and risk management, medication management, infection prevention and control, and reprocessing of reusable medical devices.

Another important aspect is knowing and understanding the Required Organizational Practices (ROPs), especially if they relate to your field of work. The ROPs, which are part of Accreditation Canada's Qmentum program, are defined as essential practices that health service providers must have in place to enhance safety and minimize risk.

## What are the ROPs?

All of the ROPs fall into six safety categories: safety culture, communication, medication use, worklife/workforce, infection prevention, and risk assessment. Here are the 29 ROPs which have been established for the MUHC:

### RISK ASSESSMENT

- A documented and coordinated fall prevention program is in place and is evaluated.
- Using a standardized tool, you assess the risk for each patient of developing pressure sores and intervene to prevent their occurrence.
- The MUHC assesses and monitors patients that are at risk of suicide.
- You identify patients that are at risk of developing a venous thromboembolism and provide appropriate thromboprophylaxis to prevent it from happening.

### SAFETY CULTURE

- The MUHC Board of Directors is fully committed to quality and safety.
- The MUHC has a formal process for disclosure of accidents that promotes communication and support to patients and families. You must document the disclosure in the patient record.
- The MUHC has a formal mechanism for managing incidents and accidents that allows us to learn from them.
- Regular reports about incidents/accidents and the improvements that were made are given to managers.

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# is coming your way!

## COMMUNICATION

- Before you provide any service or procedure or administer medications, you use two ways to identify the patient, in partnership with the patient.
- You never use dangerous abbreviations like “QD” or “IU” on prescriptions, order forms or medication labels.
- You use effective data recording tools and communication strategies to standardize the transfer of patient information at all instances where the patient transitions from one care team to another: admission, transfer between units, or discharge.
- Medication Reconciliation is an organizational priority for the MUHC.
- At the beginning of any service, be it at admissions, during transfers to other units or at discharge, a complete list of all medications taken by the patient (Medication Reconciliation) is produced in partnership (collaboration) with the patient and family and others involved; any discrepancies between the information sources are investigated and resolved, and an updated medication list is given to healthcare providers and to the patient and family.
- Before every surgical procedure, your team completes the Time Out Surgical Checklist, and the checklist’s use is monitored.

## INFECTION PREVENTION

- Hand hygiene compliance is assessed by audits, you are informed of the results and you make improvements to your practices.
- You have received education/training on hand hygiene.
- The rates of nosocomial infections are monitored and they are shared with you.
- A reprocessing program (cleaning, disinfection and sterilization) of medical devices and equipment is in place, and is evaluated and improved as necessary.

## MEDICATION USE

- An antimicrobial management program is in place to achieve the best patient outcomes, reduce risks of infections, reduce or stabilize antibiotic resistance and promote patient safety.
- Storage of concentrated electrolytes (e.g. potassium chloride) must be avoided in patient care areas.
- The availability of heparin is limited and high-dose formats have been removed from patient care areas.
- A program to manage high alert medications (e.g. insulin) is in place to improve patient safety and reduce the possibility of harm.
- You have received training on the safe use of infusion pumps, the way in which you use them was evaluated, and you report any problems related to the pumps to those responsible.
- Narcotics are limited in quantity, are properly controlled and high concentration formats are have been removed from patient care areas.

## WORKLIFE/WORKFORCE

- Data on patient flow (e.g. length of stay, admission delays) are used to identify barriers to optimal service delivery in the Emergency Department.
- Patient safety training is provided annually.
- A Patient Safety Plan has been developed and improvements are implemented.
- A preventive maintenance program is implemented for all medical devices, equipment, and technology.
- The MUHC has a strategy to prevent workplace violence.

Over the next two months, pay close attention to weekly updates on the Intranet/MyMUHC. Your contribution is absolutely vital to the success of the accreditation process. Make sure to also pick up the April issue of *Chez nous* to discover **What to expect during the visit.**



**March is National  
Social Work Month**

## **A day in the life of a... critical care social worker**

**By Pamela Toman**

The arrival of a new baby is supposed to be the happiest time in a family's life. But for parents of babies born extremely premature or sick, the excitement of that moment is often overshadowed by a sudden and unexpected admission to the Neonatal Intensive Care Unit (NICU).

"In that moment, everything stops for the family," explains Angela Heguy-Johnston, one of three social workers covering the unit and dedicated to providing essential psycho-social support to a baby's parents, who find themselves in a life-altering situation with little preparation. "It's my role to give these families power when they feel powerless."

[continued >](#)

► Top: Angela Heguy-Johnston



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#### **Helping parents find their way**

Angela’s day typically begins by dividing the patient roster with the other social workers on the unit and introducing herself to parents for the first time. Unlike her critical care colleagues who put all their energy into treating and

caring for a sick and fragile neonate, Angela’s role is uniquely focused on accompanying parents and arming them with the tools they need to stay afloat, both emotionally and functionally. “This couple didn’t know they would end up here and don’t know where this journey will take them,” she explains. “They envisioned having a baby shower, preparing their baby room, and yet they find themselves here. It’s a very big deal.”

In her initial meetings with families, Angela tries to focus on procedural steps that can ease the financial burden a family can experience due to a prolonged hospitalization after a child’s birth. Some of these benefits can include employment insurance, parental leave or other governmental programs for which they might qualify. Parking passes, meal tickets, and gift cards or vouchers are given to families requiring additional instrumental support. “Some

of these benefits can seem small,” says Angela, but every bit of assistance is appreciated when a family is in need.

#### **Providing ongoing support to families**

Over the days and weeks that follow, Angela says a family’s needs naturally change from procedural to emotional due to the sometimes difficult news they’ve been given about their child’s health or prognosis. “As I get to know a family, my role shifts to provide a safe space for them to express their feelings and to make suggestions that can help a parent better cope with their experience,” she explains. It’s not uncommon for a parent to experience feelings of stress, isolation and grief that can remain, even when their child is doing much better. “Sometimes this takes the form of referring them to their family doctor if we notice signs that things

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► Angela consults with her colleagues in the NICU, social workers Melinda Di Minno (left) and Nancy Lee Saul (right).

aren't going well," she explains. "At other times, we'll refer a parent to the psychologist on our team." The goal is always to try and help a parent make sense of what has just happened and feel that their needs are addressed, along with their child's.

### Preparing for life outside the hospital

It's a bittersweet moment when a family who has spent such a long time in hospital prepares for a long-awaited transition home, says Angela. "These parents spend weeks or months on the unit and it becomes a part of their life," she explains. "Therefore going home can sometimes feel like a different kind of loss."

Angela says she remains very cognizant of the fact that parents of NICU babies

are not going home like they would in the context of a regular birth, and that they will continue to need psycho-social support as they adapt to a life outside the hospital. "My relationship with a family doesn't end upon discharge," she says. "I do follow-ups by phone to ensure that their needs are being met outside of the hospital, either by transferring a file to a social worker at the CLSC or to a colleague if they will continue to be followed as outpatients."

One of the most validating experiences is when families who have been discharged return to the hospital for a follow-up and decide to pay her a visit several months or even years later. "To see a family with a now two- or three-year-old, and see how far they've all come...is a really great day."

### Destined to be part of the Children's family

Fulfilling the role of a social worker at the Children's is a journey you could say Angela has been preparing for her entire life. Upon graduating from McGill's Social Work program, she began working at the Children's in June 2015. Having grown up with two parents who built long careers at the Children's, as well as two brothers whose career paths have also intersected with the institution, Angela always knew she wanted to be part of the MCH family in some way. Having completed a DEC in Recreation Studies earlier in her academic journey, Angela says an internship in the Psychiatry Unit many years ago made her shift her career goals. "I realized social work was more the direction I wanted to go, and luckily it wasn't too late." ●



► Angela meets weekly with the NICU's psycho-social team.

# International team of scientists detect cause of rare pediatric brain disorder

By Stephanie Tsirgiotis and Susan Usher

Mathilde Poliquin came into the world with chubby cheeks and a full head of auburn hair. But she was a very sick baby, and on January 19, 2013, she was transferred to the Neonatal Intensive Care Unit at the Montreal Children's Hospital. By the time she arrived, she was sicker than initially expected. Mathilde's small head was of particular concern to doctors.

She underwent neurological tests, and sadly, they came back abnormal: her brain hadn't developed properly and the white matter (or myelin) was atypical. Doctors confirmed she was suffering from an unidentified form of genetic leukoencephalopathy, a family of diseases affecting both the nerve cells and the white matter.

Over the next two months, Mathilde became very ill. Doctors informed her family that she didn't have long to live. Her parents, Emily and Gabriel, along with her medical team, decided to follow a palliative approach to keep her as comfortable as possible. She passed away at two-and-a-half months old surrounded by the people who loved her most.

Seven years later, thanks to an international effort led by physician-scientists at Rady Children's Institute for Genomic Medicine (RCIGM)-San Diego in California, Dr. Geneviève Bernard's team at the Research Institute of the McGill University Health Centre (RI-MUHC) was able to confirm the diagnosis for Mathilde: she died from VARS-related disorder, an extremely rare neurodevelopmental condition.

Their findings, which were recently published in *Nature Communications*, are paving the way for the first step in developing potential therapies for this neurodegenerative disease. By performing advanced genetic tests on blood samples from seven children with neuro-developmental disabilities, scientists in San Diego, Montreal and Cairo discovered mutations in the VARS gene.

These mutations led to a defect in the enzyme responsible for generating proteins containing valine, an amino acid necessary for cellular health. They also discovered that enzyme activity in the cells of these young patients was also significantly reduced. The findings suggest that children with this disorder may benefit from treatments that produce new valine-containing proteins in the brain.

By conducting whole genome and exome tests on these children, scientists were able to search their entire genetic code for imperfections and were able to identify the source of the disease, which in turn, can help doctors develop precise treatment plans in the future.

"Trying new approaches to understand these diseases is important. Having an answer about what's making their child so sick helps families grieve," adds Dr. Bernard, a pediatric neurologist at the Montreal Children's Hospital and a researcher with the Child Health and Human Development Program of the RI-MUHC. ●



► Mathilde Poliquin

# Getting the big picture

## New MCH dashboard portal is one-stop data shop for clinical sector managers

By Maureen McCarthy

The MCH Bureau de la qualité et de l'amélioration continue (BQAC) recently launched a web-based dashboard to group together a variety of data sources to support continuous improvement across the hospital.

While many dashboards were previously available, they were all Excel-based which limited their usability. Last year, the MCH Executive Committee (EXCO) met with managers to discuss the issue and what measures could be taken to create a better, more cohesive data infrastructure. The BQAC, led by Frédéric De Civita, was asked to manage the project, and part of the project strategy was to have someone responsible for data maintenance. Simon Gignac was hired to fill that role.

Frédéric and Simon developed a concept for the new portal, one which could provide access to 10 different databases. They used an existing software with extensive customization capabilities which allowed them to look at all the data sources, measures and indicators available within the hospital and establish an optimal way to display the information. According to Frédéric, a key element of the new dashboard is that it's web-based and is connected to more data sources than ever before.

The dashboard covers a range of areas and topics from Emergency to Hand Hygiene, to Inpatient Activity and Patient Satisfaction. Some of the data is updated in real time.

The new dashboard is a dynamic and powerful interactive tool. "Essentially, it's a one-stop shop for clinical sector managers to access the data they need when they need it," says Frédéric. "For example, if you go to the Emergency tab, you can immediately see the average length of stay for a given time period. A user can then click on the drop-down menu and narrow that down to Level 4 and 5 length of stay, or see the length of stay for children who were eventually admitted. The possibilities are endless." The dashboard also has tools to allow each user to personalize their experience.

In its initial launch phase, the dashboard is being made available to the hospital's clinical sector managers as well as those responsible for quality improvement in their sectors. The next phase (in 2019-2020) will include adding new data sources as needed, increasing access for other users in the hospital, and working with managers individually to customize their own dashboards. ●

