Chez nous

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Living at home with a rare disease

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Hôpital de Montréal pour enfants
Centre universitaire de santé McGill

Montreal Children's Hospital
McGill University Health Centre

ALSO IN THIS ISSUE:

Maxime:

Living at home with a rare disease

By Maureen McCarthy

For those who meet Maxime for the first time, there's little that would tell them he lives with an extremely rare genetic disease. "He's always happy," says his dad Jasmin. "He's the easiest baby—so curious and always wanting to look at everything."

Maxime was born in October 2017 at LaSalle Hospital, and within hours of his birth, it was clear that something was wrong. He had continuous diarrhea and was becoming seriously dehydrated, causing him to quickly lose weight. Within three days, he was transferred to the neonatal intensive care unit (NICU) at the Montreal Children's Hospital and was immediately put on an IV for rehydration. He was given a number of different feeding formulas to address the problem but his diarrhea continued. After several days, Jasmin and

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▶ Top: Jasmin, Maxime and Vanessa

his wife, Vanessa, met with gastroenterologist Dr. Ana Sant'Anna, who suspected Maxime might have microvillus inclusion disease (MVID), a condition she had seen while working at McMaster University Medical Centre in Hamilton. A biopsy was requested, and the results confirmed that her hunch was correct.

MVID is an extremely rare disease, with estimates varying between 100 and 200 known cases around the world. The condition is characterized by chronic, watery, life-threatening diarrhea that usually starts in the first hours of life. MVID is usually caused by a mutation in a gene which is responsible for making the myosin Vb protein. An absence of myosin Vb inhibits certain cells in the small intestine from producing microvilli, which absorb nutrients and fluids from food as it goes through the intestine. The key problem for children with MVID is they can develop malnutrition and dehydration, and in turn, other problems related to growth and development. Their nutritional support must come from intravenous feeding, also known as total parenteral nutrition (TPN).

A longer than expected stay

Maxime spent about five weeks in the NICU so he could gain weight and grow a bit, and when he was stable enough, he was moved to the hospital's B09 inpatient unit. The family didn't expect that this would be home for the next seven months. Dr. Mylène Dandavino, Program Head of Medical Inpatient Services, explains that many families who transition through the B09 unit have children who will need technological support at home. "The diagnoses we see can vary greatly, and families are sometimes here for long periods before their child can safely be cared for at home," she says. "They have to learn a lot in order to care for their child at home. They meet many physicians, nurses and other healthcare providers, and given that we're a teaching hospital, they also meet many residents and students. Maxime's family, like many others, was very resilient in adapting to this staff turnover and to life on the unit. They are one of many amazing families we've had the chance to meet and work with."

▶ Annie Chaput, Nurse Clinician in the hospital's Complex Care service, is part of the team that sees Maxime regularly for check-ups now that he is at home.

Maxime was put on TPN which is a feeding method that bypasses the gastrointestinal tract by giving fluids into a vein to provide nutrients the body needs. The feeding is done through a peripherally inserted central catheter (PICC) inserted into the patient's arm, and it provides them with amino acids, vitamins, minerals, and lipids. Regular blood and urine tests help the health care team know what adjustments are needed.

Going home with 24/7 TPN —a first at the Children's

Annie Chaput, Nurse Clinician in the hospital's Complex Care service, met Jasmin and Vanessa shortly after they arrived on B09. "We started teaching them how to do the TPN because we thought they'd

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On the cover: Maxime and his dad, Jasmin

Cover photo: Owen Egan

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be able to go home fairly soon," says Annie. "Normally our patients don't go home until they can be off TPN for six hours a day." Maxime was on TPN around the clock, and when they first tried to take him off the feeding for just a few minutes, he immediately developed hypoglycemia, a potentially life-threatening condition for him. "Early on, we considered the possibility of letting Maxime go home on 24/7 TPN, but his parents wanted to wait a bit to see if things improved." By February, Maxime still needed TPN around the clock, but since he was stable and had no other health problems, Jasmin and Vanessa made the decision with the team to start the process of going home, which involved coordinating with the CLSC for nursing and other support. On June 21, Maxime went home for the first time.

A new life at home

Maxime now has regular appointments in Complex Care where his team includes Annie, nutritionist Marie-Josée Trempe, Dr. Sant'Anna, and pediatrician Dr. Hema Patel. In July, the Complex Care team decided to try taking him off TPN again, and this time it was successful. The pump Maxime has at home can taper down the infusion so that his body slowly adjusts and he doesn't develop hypoglycemia. "The first time we tried it and stopped the infusion through Maxime's PICC, his glucose levels were good for a full hour," says Annie. They experimented further, and now Maxime can go four hours a day without TPN.

One of the considerations about being at home is the risk of an abrupt change such as a problem with the PICC, or if the pump stops working all of a sudden. There isn't much time to react. Jasmin and Vanessa have a Glucagon injection at home which they can give to Maxime if that ever happens. Eventually, the Complex Care team will reintroduce the challenge of these sudden changes to see if things have improved and if Maxime's body is working it out by himself.

Maxime's parents are also introducing small amounts of food to him, which helps him develop his ability to chew, as well as contribute to his speech development.

> **"Early on, we considered the possibility of letting Maxime go home on 24/7 TPN..."**

Sharing knowledge and experience

Recently, Annie, Marie-Josée and Dr. Sant'Anna spoke to Dr. Bram Raphael at Boston Children's Hospital, who has worked with children with MVID. "We found nothing in terms of research in the past decade, so it was really helpful to talk to someone who agreed with our diagnosis and treatment approach with Maxime," says Annie. At the end of September, they'll attend a conference on intestinal failure in pediatrics. "We're hoping to be able to discuss Maxime's case with others and learn more about different issues surrounding intestinal failure," says Annie. "And mostly, we're hoping to be able to speak with other people who have encountered MVID in their practice." ●

▶ Jasmin and Vanessa talk to Dr. Mylène Dandavino, Program Head of Medical Inpatient Services, who got to know the family well during their seven-month stay on the B09 unit.

A day in the life of...

Three medical technologists in the MUHC Central Lab

By Stephanie Tsirgiotis

There's a good chance you've never met medical technologists Jessica Driscoll, Annie Malenfant and Rabii El Ouarari. "We definitely work behind the scenes," says Jessica. The Research Institute of the McGill University Health Centre houses the MUHC's Central Lab, named for the Marcelle and Jean Coutu Foundation, where the trio work alongside each other to identify, confirm and categorize different types of leukemia in pediatric and adult patients.

Step 1: Reviewing abnormal test results

Childhood leukemia is the most common form of cancer in children and adolescents. It affects a child's white blood cells and can be detected through a series of tests and careful analysis. Jessica Driscoll's work involves reviewing abnormal blood results obtained during a complete blood count (CBC) to help identify potential leukemia cases.

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▶ Top: (l. to r.) Rabii El Ouarari, Annie Malenfant, and Jessica Driscoll.

▶ Jessica and her colleagues review 150 pediatric CBC results a day, but she has never met a patient in person. "Patients don't know we exist, but we follow their progress very closely. When I read *Chez nous*, I'm always happy to come across a story about a child we know by name. It's great to see what they look like!"

A CBC is a blood test that evaluates a person's overall health and it can detect a wide range of disorders and diseases, including leukemia. Jessica reviews the patient's red and white blood cell counts, as well as their platelets and hemoglobins. "I look at the scattergram first, which is a graph that differentiates each white blood cell by cell type and size, and then I confirm my suspicion with the numeric results," she explains.

Jessica and her colleagues review 150 pediatric CBC results a day. "Thankfully, most of these results end up being non-leukemic diseases or disorders," she says. Besides helping physicians make a diagnosis, she also reviews blood tests of oncology inpatients and outpatients currently undergoing treatments such as chemotherapy, and patients in the Neonatal Intensive Care Unit and Pediatric Intensive Care Unit who are battling various diseases and disorders.

When searching for signs of leukemia, some of the red flags are very high or very low white blood cell counts, as well as low hemoglobin and platelet levels. "The moment I see a red flag, I immediately get on the phone with the physician who requested the CBC," she says. "It's a hard phone call to make, because I know I'm about to completely change someone's life."

After informing the physician of her findings, she quickly heads over to the morphology room with a microscope slide in hand. "This is where Annie comes into play," she says. "She takes a look at the smear and if she sees any suspicious cells, she calls the hematologist, who then comes down to the lab to take a closer look and ask for further testing. And of course, the faster we get this done, the better."

Step 2: Analyzing a patient's cells

Annie Malenfant spends her days looking at cells through a microscope. She specializes in cellular morphology, which involves identifying the shape, structure, form and size of cells. "I can look at a cell and tell whether or not it's a cancer cell," she says. "When reviewing someone's

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blood counts, it's difficult to differentiate between an infection and leukemia, because in both instances, the patient's white blood cell count is abnormal. But I can tell the difference by looking at the cells."

When a patient is battling an infection, the body produces a type of infection-fighting white blood cell, called a lymphocyte; and when a patient is battling leukemia, the bone marrow begins to rapidly produce abnormal white blood cells. "Because these white blood cells are produced so rapidly, they tend to look very immature," she explains. "That's when I know it could be leukemia."

Immature cells, also known as blast cells, tend to have a smoother-looking nucleus and their surface is more textured. They also have a different colour. "Cells battling an infection tend to have a darker blue cytoplasm, and they're reactive. You can actually see them reacting to the environment around them," says Annie.

"It's a hard phone call to make, because I'm about to completely change someone's life."

On any given day, Annie will analyze 40 to 60 slides, which include blood specimens, bone marrow, as well as cerebral and body fluids. She is also sometimes called onto the unit to help retrieve cells during a bone marrow aspiration. "To get the best quality result, I need to be present in order to make a slide immediately at the child's bedside," she says. "At the end of the day, there is always a patient behind each cell, each slide, so we do whatever we can, as quickly as we can, to get a diagnosis."

Diagnosing leukemia in young children, especially newborns, is especially challenging. "Babies are the most difficult to diagnose, because all of their cells look immature and young. It's hard to distinguish between a blast cell and a normal, newborn cell. This is why Rabii's work is so important. He not only helps narrow down the exact type of leukemia, but he can confirm the cases which are difficult to detect with a microscope."

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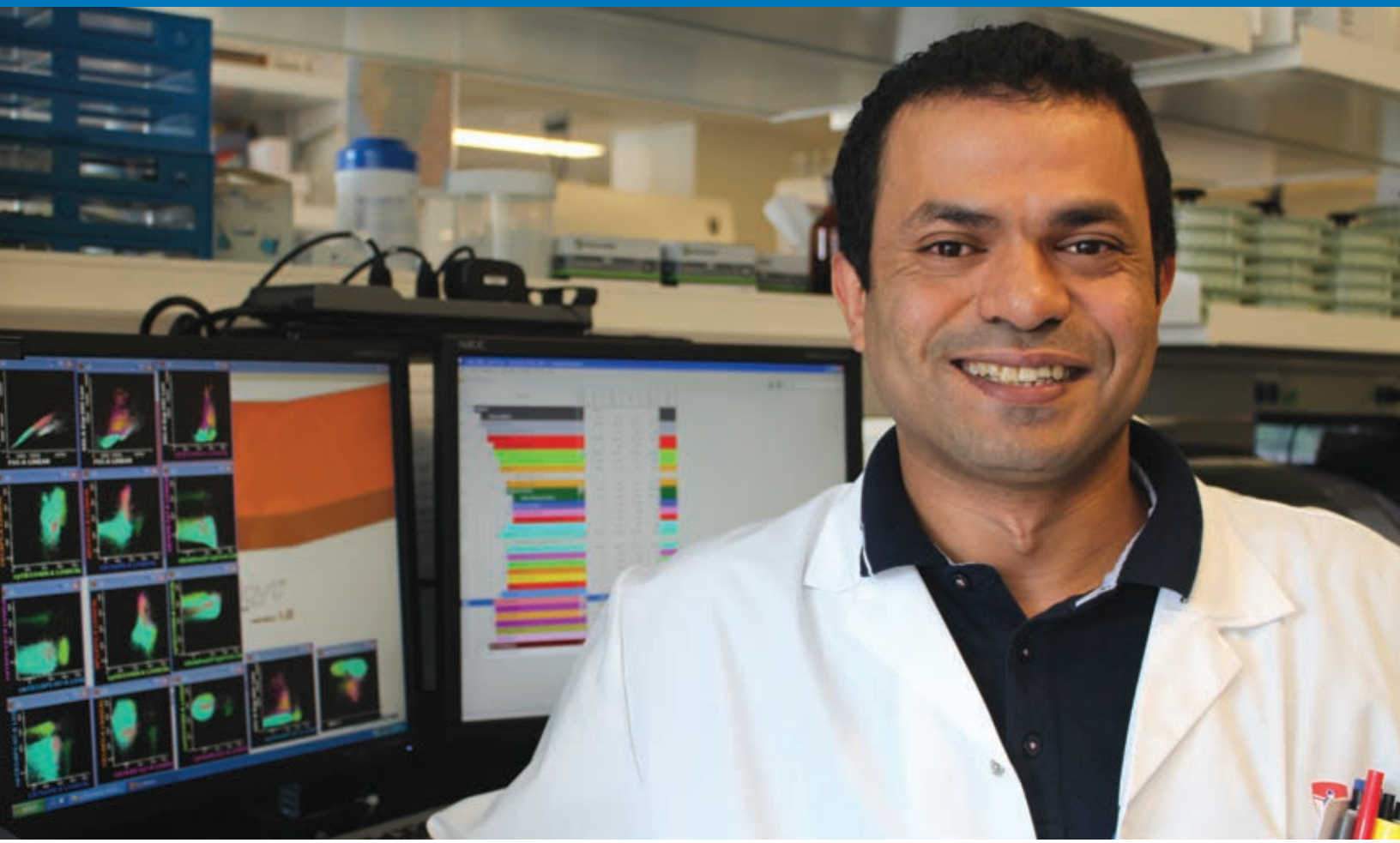
▶ Annie Malenfant specializes in cellular morphology and is able to identify the shape, structure, form and size of cells.



▶ Rabii El Ouarari can detect more than 20 different types of leukemia using flow cytometry and immunophenotyping.

Step 3: Narrowing down the diagnosis

Rabii El Ouarari, a former teacher from Morocco, always loved the idea of working in a laboratory and decided to make his dream a reality after immigrating to Quebec. After completing his degree in medical technology, Rabii started working at the MUHC, and quickly discovered he had an interest and a particular skill for working in hematology.

Rabii specializes in flow cytometry, a laser-based technology used for cell analysis. It allows Rabii to rapidly and accurately profile over 100,000 cells at a time. "I can collect data quickly from thousands of cells, because flow cytometry allows me to view many antibodies at the same time," he says. "This process can lead to the diagnosis, prognosis and classification of various forms of leukemia."

In fact, Rabii can detect more than 20 different types of leukemia, which allows physicians to treat their patients with specific therapies targeted to their type of cancer. "There are many different types of leukemia and they all have different characteristics," he says. "I'm able to distinguish between lymphoid and myeloid leukemias by looking at their distinct cellular features, including their antigens [proteins] and antibodies."

He is able to make this distinction thanks to a technique called immunophenotyping. This test allows Rabii to identify specific antigens found only in cancerous cells. "The antigens are stained with a fluorescent marker and this marker only binds to specific antigens," he explains. "This allows me to see the leukemia cells, because they are a different colour from normal cells."

In 2016, the MUHC became part of EuroFlow, a consortium composed of research centres with expertise in flow cytometry and immunophenotying. "We are now leaders in Canada and are performing at the highest standard," says Rabii. "We can diagnose different subtypes of leukemia even more precisely than before. It has completely changed our lives and the lives of our patients." ●

News from our patients

Dave and Liam, familiar faces to many Children's staff, both received long awaited organ transplants this summer.

Dave Laplante, a cystic fibrosis patient

followed at the Children's, spent a significant amount of time at the hospital. In 2017, he alternated between the hospital and home spending two weeks at a time at each place until his condition became critical; in January 2018 he was admitted onto B09 indefinitely while waiting for a lung donor to become available. With his mother by his side the entire time, Dave was transferred to the CHUM on May 31 in preparation for admission to the intensive care unit. Fortunately, just four days later, he and his family received the long-awaited news that a pair of lungs had become available and that he would be undergoing a lung transplant.

Dave spent almost four more weeks in hospital after his surgery, but is now home and doing well. Although he still has some physical conditioning to do, he feels much better and is not out of breath as easily. The 18-year-old plans on completing his secondary V studies in January and then taking auto mechanics so he can follow his passion for cars.

A successful heart transplant allows Liam to go home

July 27 was a very special day for eight-year-old Liam, a patient at the Montreal Children's Hospital who was admitted to the hospital in the summer of 2017 with a heart infection. In the first few weeks of his hospitalization, his condition deteriorated and eventually, he had to be put on a Berlin heart device, which was used as a bridge to transplant for almost a year while he waited for a new heart. The Berlin heart allowed Liam to walk around and leave his room, and he even participated, along with his mother, in this year's ***Caring for Kids*** Radiothon held in the hospital atrium.

After such a long stay at the hospital, Liam's mom Kim Morris couldn't believe the news when they learned that a donor heart was available. The successful heart transplant surgery lasted 12 hours, and Liam spent another two weeks at the Children's before finally going home.

Liam now comes back to the Children's for check-ups. Life at home is going well and he's starting to enjoy many of the things he missed while in the hospital. "We are forever grateful to the family who made the decision that allowed Liam to have the transplant," says Kim. "Liam's new heart is the gift of a new life for him."

Small Miracles

A series of photos unveils unexpected surprises and renews lost connections

By Sandra Sciangula

On the wall along the hall connecting the Montreal Children's Hospital's (MCH) Neonatal Intensive Care Unit (NICU) to the Royal Victoria Hospital's (RVH) Birthing Centre, a series of 15 photos entitled Small Miracles is on display. The photos, taken in 1997 by photographer Ewa Zebrowski, whose work can be found in collections across the globe, depict what a typical day in the NICU was like more than 20 years ago. The inspiration for the series was Ewa's own dramatic deliveries: her two sons were born prematurely in 1983 and 1985 and were cared for in the RVH NICU. Ewa returned to the unit 11 years after the birth of her second son because she wanted to capture the experience from behind the camera.

With the help of Dr. Robert Usher, the medical director of the unit at the time, Ewa was introduced to a family who gave her permission to photograph their son, Stephane Lemoing, who was born at 27 weeks. "My intention was to celebrate the doctors and nurses who care for these infants and to convey what it means to have a child who is born prematurely," says Ewa. The Small Miracles installation was inaugurated with

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▶ Top: (l. to r.) Dr. Daniel Faucher, nurses Jennifer Guerrero and Hélène Caron, and photographer Ewa Zebrowski attend the vernissage for "Small Miracles".

a vernissage on August 23, and was attended by physicians, staff and members of the community.

A surprise discovery

While looking at one of the photos, neonatologist Dr. Daniel Faucher got a pleasant surprise. He recognized himself captured at a distance speaking to a group of people on the unit. "I had no idea I was photographed for this!" he says. There's a clock in the photo that reads 10:25 which led Dr. Faucher to assume the photo was taken during rounds. He recognized some faces in the photos and with nurses Jennifer Guerrero and Hélène Caron, who also worked on the old unit, he reminisced about colleagues and life on the former unit.

On the eve of the vernissage—after years of not being in touch—Ewa managed to reconnect with the family photographed for her series and asked them to attend the event. Denis Lemoing and Lisa Gillis, Stephane's parents, were able to attend the vernissage and spoke with pride about their son who is now 21 years old and described as tenacious by his father.

▶ One of the photos taken by Ewa Zebrowski which is part of the Small Miracles exhibit shows Lisa Gillis (r.) and her older son Eric visiting Stephane in the NICU.

A small miracle... 21 years later

Although Stephane has faced a fair share of adversity throughout his life, he is an accomplished young man. At the age of five he was diagnosed with Asperger's and autism spectrum disorder. "Throughout high school I felt isolated and school was hard for me," he says. However, he refused to give up. In grade eight, Stephane was recognized by his school for his high grades. In grade 10, he was recognized for his perseverance, and in grade 11, was invited to Montreal City Hall to sign the Golden Book, to acknowledge his perseverance.

After high school, Stephane studied two trades, one in general building maintenance and the other in hygiene and sanitation; he received an award of distinction from the city of Lachine for dedication to his work. Stephane now works at Saputo and Home Depot. He keeps busy by cycling, working out, and going out with friends. He hasn't seen the photo series yet but he recalls a photo of his mother and older brother, Eric, visiting him in the NICU, a souvenir which is safely kept in one of his mother's photo albums.

Small Miracles is a new acquisition for the hospital and will be on permanent display. "The goal of art in the hospital is to bring a human element to what can feel like an institutional space," says Alexandra Kirsh, Curator of the RBC Art and Heritage Centre of the MUHC. "This series is meant to remind us of where we came from and that even though technology has changed drastically over the years, the love and care provided to patients has remained the same." ●

▶ Stephane (middle) and his mom, Lisa (2nd from right), are seen with officials from the city of Lachine after Stephane received his award of distinction from the city.

Quebec announces newborn screening for cystic fibrosis

Dr. Larry Lands, Director of Respiratory Medicine at the Montreal Children's Hospital, is one of many people who welcomed the news that Quebec would be introducing newborn screening for cystic fibrosis (CF) on September 17. "Quebec is the last jurisdiction in North America to start newborn screening for CF," says Dr. Lands. "It's an important milestone and represents work done by many people for more than a decade."

In 2016, the Journal of Cystic Fibrosis published a study led by Dr. Denise Mak of Cystic Fibrosis (CF) Canada and co-authored by Dr. Lands, which looked at data from the Canadian CF Registry on children in Alberta, Ontario and Quebec. The study analyzed factors such as growth, number of hospitalizations, and rates of infection from two bacteria often associated with CF. Children screened at birth who were then diagnosed with CF showed better outcomes across these measures than children who were diagnosed later without the benefit of newborn screening.

"Despite access to top-notch therapy, the Quebec patients did not do as well on these measures of growth and development," says Dr. Lands. "We concluded that newborn screening would lead to better long-term health outcomes."

Identifying a key enzyme

All newborns in Quebec have a small blood test to check for certain diseases, and screening for CF will now be added to the testing. The CF test looks at the baby's level of immune reactive trypsinogen (IRT), an enzyme produced by the pancreas. "If IRT is high, then the baby will be recommended for genetic testing," says Dr. Lands. The genetic testing then determines if the child has the cystic fibrosis transmembrane conductance regulator (CFTR) gene.

Making new therapies available to more patients

Dr. Lands points out that it's an exciting time in the management of cystic fibrosis because there are new therapies that correct the fundamental defect in the CFTR gene. Pharmaceutical companies which are making the first generation of correctors have already begun to conduct studies in children under two years old. "Our team members have been early adapters for these new correctors, and we've taken the approach of letting our families know what's available and enrolling them in research trials," says Dr. Lands.

The McGill University Health Centre (MUHC) is part of a national clinical trials network started by CF Canada, and Dr. Lands is heading up the MUHC site for the network. "This initiative will allow more patients to be involved in new therapies, and we'll have the opportunity to bring in patients from across eastern Quebec," he says. "There are a variety of pediatric studies going on now, and we'll soon be starting a Phase I trial in adult CF patients."

▶ The Children's Cystic Fibrosis Clinic team (l. to r.) Dr. Jocelyn Lavigne, Émilie Cadorette, Nancy Alarie, Dr. Karine Gauthier, Dr. Larry Lands, Sophie Vallée-Smejda, Dr. Adam Shapiro, and Debbie Fertuck. Absent from photo: Christina Gallagher and Lianne Kopel.

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