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Dr. Geneviève Bernard: “My illness, my fight”

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Dr. Geneviève Bernard (Photo: Courtesy)

A pediatric neurologist, Dr. Geneviève Bernard treats children with rare neurodegenerative diseases for which there are few or no treatments. While she does everything possible to improve the quality of life of her patients and their families, the doctor says she learns a lot from them.

In her personal life, Dr. Geneviève Bernard readily admits that she has a romantic side and can cry easily: "When I watch the Telethon, for example, I can't help but cry." In her practice, "it's different," assures the pediatric neurologist who works at the Montreal Children's Hospital (MCH) of the McGill University Health Centre (MUHC). "I have power over what's going to happen. Even if I don't necessarily know the end result, I know the path to take to support parents through difficulties. That helps me do my job," she explains.

Her daily life in the medical genetics department at the MCH is far from rosy. "It's often even sad," admits the clinician-researcher. A specialist in neurodegenerative diseases, she takes care of children with increasing disabilities, for whom treatment options are limited or non-existent and whose life prognosis may be engaged in the more or less long term. Despite this adversity, "I could not do without the intense relationships that I develop with 'my' children and their families," admits the doctor.

She spontaneously evokes the aura of children with a rare disease and their communicative radiance. “They have enormous resilience. Even if most of them have short lives, they live more intensely. Every day, every moment is important. With their family, they manage to put priorities in the right place. They have this ability to enjoy seemingly trivial things that bring them happiness,” emphasizes Dr. Bernard, convinced that her patients are extraordinary and that they change the world: “They teach us life lessons that make the people they meet better.” And she knows what she’s talking about.

She herself is the mother of a 12-year-old boy who suffers from a rare genetic disease that was not detected at the time of his adoption and for which there is no precise diagnosis. "I have seen many parents go through something similar, who do not know what their child is suffering from, how his condition will evolve and what the outcome will be. They have helped me a lot in my practice and in my life, because they have given me tools to manage the unknown and overcome these difficulties," says the doctor.

Her two other children, also adopted from Vietnam, also have health problems. “Between the three of them, my children have used almost all of the MUHC’s services. I also know which CLSC or community organization offers which service and under what conditions. This allows me to help my patients’ parents by sharing my own experience with them.”

From the patient

The neuropediatrician has applied this principle of communicating vessels to her research for 13 years. Her motto: "From the patient's bedside to the laboratory and from the laboratory to the patient's bedside." A world-renowned expert on leukodystrophies, she receives families from as far away as Argentina and Israel in Montreal to consult her, in the hope that she can diagnose their child's disorder and improve their quality of life.

Dr. Bernard studies each of them in the clinic and in her laboratory with the aim of perfecting the characterization of these diseases that affect the "white matter" of the brain. She thus applies herself to describing their evolution and their impacts, to identifying new causal genes and to defining their pathophysiology. Her underlying objective is to improve the medical care offered to these patients, to find avenues of treatment, to prepare the ground for future therapeutic trials and to develop treatments.

She is more specifically interested in RNA polymerase III-related leukodystrophy or 4H, which is a common form of leukodystrophy characterized by insufficient myelin. "It's my disease, my fight," claims the researcher, specifying that with her team, they have identified four of the five genes causing the disease and produced clinical descriptions of the widest range of patients in the world. The culmination of a decade of hard work, they managed last year to create the first animal model representative of 4H leukodystrophy. "By developing this model, we have answered fundamental questions about the pathogenesis of the disease that will allow us to develop new therapeutic approaches and test them," says Dr. Bernard, who is also responsible for international clinical trials.

During her studies in medicine and neuroscience, Dr. Bernard told herself that research was not for her. “My career path is the result of a series of coincidences that gave me the desire to do it,” she explains. This sequence began with a presentation given at the beginning of her residency in pediatric neurology on leukodystrophies, “such a difficult subject” that she sought to better understand it. Then, during her fellowship at the Université de Montréal in neurogenetics and

movement disorders, under the direction of Dr. Bernard Brais, he offered her the opportunity to collaborate on a new research project on leukodystrophies, which led her to discover the first gene responsible for 4H. Then, “it snowballed,” observes the neuropediatrician, admitting that she had to confront her imposter syndrome at each stage of the development of her research program.

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Research in synergy

In addition to her clinic and laboratory, Dr. Bernard is also co-director of the new Network to Advance Research on Rare Diseases in Quebec (RARE.Qc), funded by the Fonds de recherche du Québec – Santé (FRQS). RARE.Qc aims to foster collaborations between researchers working on rare and undiagnosed diseases, by adopting a translational and reverse translational approach to involve patients and their families in scientific progress. The pediatric neurologist believes that the creation of such a network, announced last winter, brings hope to patients. “Research on rare diseases must rely on collaborations to progress,” she says, noting that interest in these diseases is growing significantly, both at the government and pharmaceutical laboratory levels.

A major challenge regarding research on rare diseases remains, however, obtaining research funding. "Some subjects are less difficult to finance, such as basic research or the exploration of new therapies. On the other hand, it is more difficult for natural history studies, retrospective and prospective observational studies or those that focus on the impacts of rare diseases on patients and their families," regrets Dr. Bernard, emphasizing that these studies could nevertheless "have a positive influence on care and improve the quality of life of patients, starting now."