

Doctor Jérôme Lejeune's Gaze at the University of Ottawa

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"Dad has, above all, a gaze. His blue eyes sparkle with intelligence and humor and they look at you with infinite tenderness. Still they are demanding because they love the truth. They are tirelessly seeking the 'why' and the 'how' of what they see."

CLARA LEJEUNE

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ABSTRACT

From January 29th to February 1st, 2015, a group of University of Ottawa undergraduate students and I hosted an exhibit about the life and research of French physician Dr. Jérôme Lejeune, who in 1959 discovered Trisomy 21, the genetic origin of Down syndrome (DS). Considered the father of modern genetics, Dr. Lejeune believed medicine should serve the patient and not the disease. The exhibit focused on Lejeune's profound humanity and compassion that accompanied his commitment to scientific truth. It also showed how this great scientist maintained an unshakeable adherence to his faith and conscience despite challenges and adversity.

RÉSUMÉ

Du 29 janvier au 1er février 2015, un groupe d'étudiants au premier cycle universitaire ont organisé une exposition portant sur la vie et les travaux de recherche du médecin français Dr Jérôme Lejeune, qui en 1959 a découvert la trisomie 21, l'origine génétique du syndrome de Down. Dr Lejeune est considéré comme le père de la génétique moderne. Il croyait que la médecine devait se concentrer sur le patient et non seulement sur sa maladie. L'exposition a mis en valeur l'humanité et la compassion du Dr Lejeune en illustrant comment ce grand scientifique a su maintenir sa foi et ses croyances malgré les défis à relever.

HOW IT STARTED

My first encounter with Dr. Jérôme Lejeune occurred in the late 1980s when I was a practising pediatrician in Italy. Although I had never heard of him, I attended a lecture he gave in Milan. His smile and gaze, and the way he deeply communicated with the 3,500 people in attendance that day, impressed me. He appeared to direct his loving attention to the children listening to him in the front rows. Though his topic was challenging, I was surprised to see how well they seemed to follow his words. Then suddenly, I realized that I was surrounded by many children affected by Trisomy 21, all of whom were listening in silence and in awe. Right next to me, a father, whose son was affected by Down syndrome (DS), turned to me with tears in his eyes and said, "I am coming from France to listen to the Professor, he helped my whole family and my son like nobody else ever did!"

I remembered Dr. Lejeune discussing the difficulty of doing research when faced with the complexity of the human organism

and all the molecular processes involved (Figure 1). Using music as an analogy, he explained, "The orchestra of life has about 50,000 musicians... to have a tiny extra piece of chromosome is like having one additional musician in an orchestra who is playing faster or slower than everyone else. It would cause cacophony," he said, and then added with a smile, "I am trying to find the discordant musician!" At that very moment I realized that Dr. Lejeune was like a father to the children with DS, and that what was transpiring between him and his audience was unique. Throughout my academic career (in both Europe and Canada) I have yet to attend to such a riveting and inspirational lecture.

Fast-forwarding to about three years ago, I received a book entitled "Life is a Blessing" [1] as a Christmas gift. It was the biography of Dr. Lejeune as told by his daughter, Clara. On the first page, to my surprise, a dedication: "À Emanuela en souvenir de Jérôme Lejeune qui nous a encouragés à avancer sur le chemin de l'espérance. New York, 14/1/2011 Birthe Lejeune" [To Emanuela in remembrance of Jérôme Lejeune who encour-

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La Machine de Lejeune

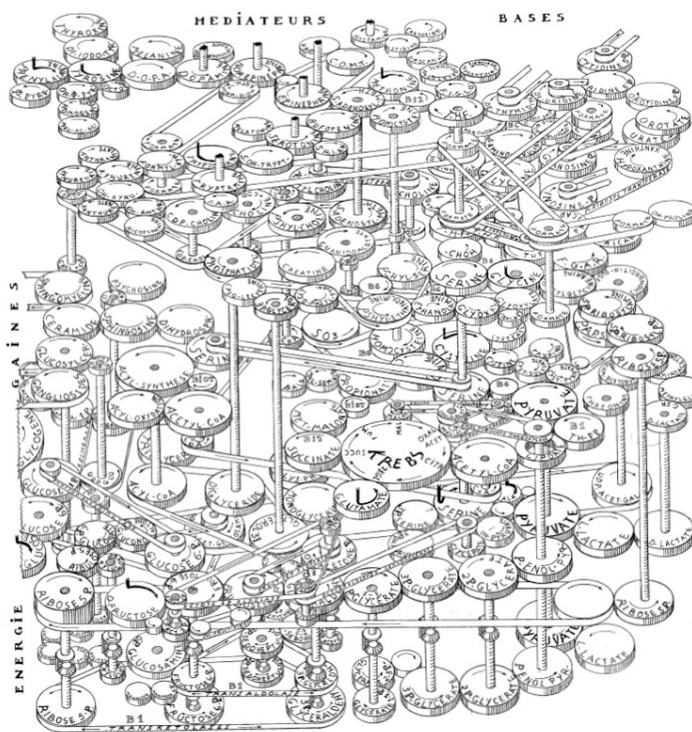


Figure 1. Lejeune's machine (reprinted with kind permission of the Jérôme Lejeune Foundation). According to a recent review by Strippoli et al. (2013) [9], Dr. Lejeune anticipated the field of systems biology decades before it became popularized. In this diagram he conceptualizes complex interrelated molecular pathways as analogous to a system of mechanical gears. He attempted to understand how a central defect—Trisomy 21—can have such diverse effects.

aged us to advance on the path of hope. New York, 14/1/2011. *Birthe Lejeune*]. This book became my companion until the day I entered the University of Ottawa's Faculty Development office and asked if I could organize an exhibit about Dr. Lejeune's life. When asked why I wanted to take on such a laborious project, I told them of how I had come to meet Dr. Lejeune, and how I wanted to share his unique perspective on patient care with my peers. I explained that this three-day exhibit, which I had seen in Italy, and later, at McGill University in Montreal, would ignite a rich discussion amongst undergraduate medical students as well as faculty in regard of what it meant to be human. I wanted my students and colleagues to learn how his passion for life—that he defended from the very beginning—had been the centre of an unprecedented scientific, ethical, societal, political, and religious debate. Furthermore, I wanted them to see a glimpse of Dr. Lejeune's gaze!

THE PREPARATION

What followed was almost a year of preparation, during which I joined with a group of seven undergraduate students, four colleagues, and two mothers of children affected by Trisomy 21, to learn how to guide the exhibit. We spent countless hours striving to learn the nature of Dr. Lejeune's work and of his monumental accomplishments, as well as how he had suffered in order to advocate for his patients. But who was he?

After completing his medical training in 1951, a young Jérôme Lejeune joined Professor Raymond Turpin's team at the Centre National de la Recherche Scientifique in Paris as a research assistant, where he was instructed to study the causes of DS. At the time, no one knew the cause of this condition. Some scientists suspected it was the result of a sexually transmitted disease like syphilis; others thought it represented a form of racial evolution. For these reasons, people affected by the syndrome faced terrible discrimination.

In the 1950s, when scientists had just determined that each human being had 46 chromosomes in 23 pairs, Dr. Lejeune began to look at the chromosomes of patients with DS and discovered an extra chromosome in the 21st pair, ergo the name Trisomy 21 [2]. By identifying the genetic cause, Dr. Lejeune helped restore dignity to those affected by DS; for the first time in the history of medicine distinct symptoms were connected to a specific alteration of the genetic material. In addition, this opened the door for further research into the genetic basis of disease, and led to the discovery of many other conditions such as Cri du Chat, Fragile X, Turner, and Klinefelter syndromes [3].

As a result of his work, Dr. Lejeune was the recipient of many prestigious awards. In 1962, he was awarded the Kennedy Prize by United States President, John F. Kennedy, for his discovery of Trisomy 21. He later went on to receive the William Allan Memorial Award in 1969, which was the highest accolade from the American Society of Human Genetics. During his acceptance speech, Dr. Lejeune summoned geneticists and left them speechless by asking, "What is man, and when does a human being begin?" [4].

The purpose of Lejeune's scientific work was to learn passionately about a disease in order to develop a cure. To him, however, providing his vulnerable patients with appropriate care and hope was of equal importance. In addition to his brilliant discoveries, Dr. Lejeune was a caring clinician who established a "legendary" relationship with his 5,000 patients in the DS clinic; knowing each one of them by name [8]. One couple described going to an appointment with their son. He sat their child on his lap, asked him questions and treated him with such love that the couple said afterwards, "He was not examining a sick individual, but our child.



Figure 2. The speakers. From left: Dr. Lise Poirier-Groulx (University of Ottawa), Dr. Emanuela Ferretti (University of Ottawa), and Dr. Mark Basik (McGill University).

He explained everything to us, the nature of the illness, what future our child could expect and we left with our child and with peace in our heart. He made us discover the love of parents.” [1].

In the aftermath of his discovery, Dr. Lejeune was faced with the reality that its clinical impact was to enable antenatal detection and termination of affected individuals. A mechanistic cure for those affected, as he had initially hoped for, was proving elusive. Here was a single individual personally facing what has now become one of the most complex issues of modern society. Ultimately Lejeune, a man of faith, was guided by his faith and took a public stance against termination of fetuses with Trisomy 21. Lejeune paid a huge price for this. One of the most listened to scientists in the world, and the first in France to be appointed to the Chair of Fundamental Genetics, became ignored and ostracized by the medical, academic, and political elite of France. He lost research funding and faced harassment. Despite all this, Lejeune remained steadfast, stating that, “It cannot be denied that the price of these diseases is high—in suffering for the individual and in burden for society. Not to mention what parents suffer! But we can assign a value to that price: it is precisely what a society must pay to remain fully human” [5,6].

THE OPENING

Finally, on January 29th, the opening evening of the exhibit had arrived (Figure 2). At a panel discussion, Dr. Mark Basik from McGill University described how Dr. Lejeune’s example guided his work researching the genetic causes of metastatic breast cancer. Dr. Basik said, “He inspires me in so many ways, especially the way his research was driven by the love for his patients; he was not afraid of being scooped—he just wanted to spread the news. His motivation was to find a cure, and do something to help his patients.”

Then, family physician Dr. Lise Poirier-Groulx, a lecturer for the University of Ottawa Faculty of Medicine, shared her personal journey as a mother of a now 15-year-old boy with DS, describing the isolation and brokenness she and her family experienced. “This didn’t happen to me; it usually happened to my patients...,” she said. Yet her experience transformed her into an advocate for those with disabilities. “When I prepare the first year medical students on coping and adapting to disability, I tell them that it’s not about who is disabled but it is much more about when we become disabled,” said Dr. Poirier-Groulx. “Without being pessimistic and unless we exit this life suddenly, we all be incapacitated or become disabled at some point.”

THE EXHIBIT – (Figures 3, 4)

During the guided tour of the exhibit, we all experienced how Dr. Lejeune still touches the hearts of different people. On the first day, I was explaining to a group of people of how Dr. Lejeune had testified in front of the judges of the Supreme Court of Canada in 1983 on the nature of the unborn child. On one of the panels we read Lejeune’s message, “Our duty has always been not to inflict the sentence but to try to commute the penalty. In any foreseeable genetic trial, I do not know enough to judge, but I feel enough to advocate” [4]. A man in the crowd identified himself as Dr. Lejeune’s driver throughout the duration of the trial, and told us of how charming and resolute Dr. Lejeune had been at the time, and how seeing Dr. Lejeune’s words here had brought him back.

On the second day, while I was guiding one of my colleagues through the 36 panels of the exhibit, he reflected, “When I see the way Lejeune has lost fame, power, and money to fight for the vulnerable, I need to reflect on my own... the way I advocate pales in front of him; we do not understand what advocacy for our patients truly means!” Even when Lejeune was seriously ill, he worried for his patients, saying “I was the doctor who was supposed to cure them, as I leave; I feel I am abandoning them” [1].

By the third day into the exhibit, I noticed that my students seemed to have found inspiration in learning about Dr. Jérôme Lejeune’s life. One of my student-guides, Stephan, while pointing at a poster displaying the gigantic smiling face of Dr. Lejeune said, “He is the doctor I want to be!” I too grew to love Dr. Lejeune’s humanity and personality. His straightforward way to live his faith has undoubtedly impacted my personal and professional life.

THE NEXT STEP

The next stop for Dr. Lejeune’s exhibit will be the Mayo Medical School Campus, after which it will travel to several other uni-

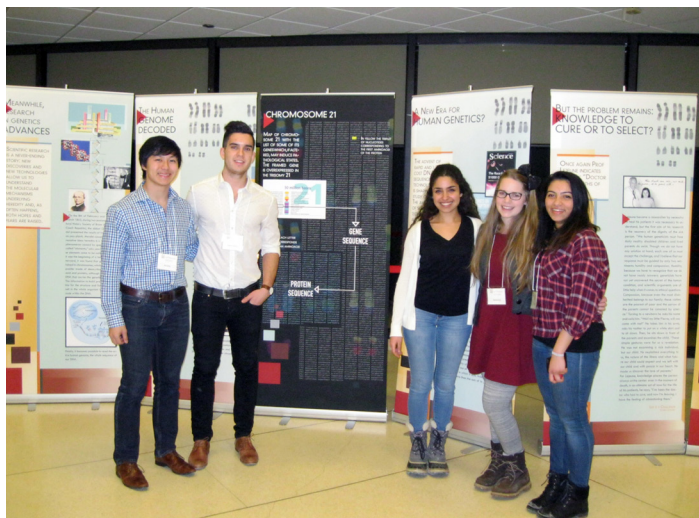


Figure 3. The protagonists. Five of the seven student guides of the Jérôme Lejeune exhibit. From left: Brian, Stephan, Caroline, Stephanie and Batoul (Alex and Nohémie are missing.)

versities within the United States. Meanwhile, a professor from the Department of Innovation in Medical Education who saw the exhibit emailed me a brilliant idea: “What a remarkable display... I wonder if some of this material could be worked into an on-line module as Case Based Learning for the first year medical students... since it contains the elements of history of Medicine, medical ethics, professionalism...”.

In our time the prevailing view of illness has taken a stance of distance from the diseased. The ultra-specialization of medicine has fostered in medical students an emphasis on knowledge and technical skills, with a neglect of the Hippocratic aspect of the practice of medicine. Dr. Lejeune was a near-perfect clinician-researcher whose thinking was ahead of his time. His teaching could offer, to our medical students, a vision that uses science to serve the patient, not the disease. His outlook has taught me that, in a field that is constantly moving and changing, the only trend worth following is the search for truth. Clearly, truth for him was no more and no less than what he was seeing through his microscope. But to honestly love the truth means to make all of your practical professional endeavors revolve around it, without compromise. As the world-renowned ethicist Dr. Edmund Pellegrino once stated, a doctor “binds himself to competence as a moral obligation” and “places the well-being of those he presumes to help above his own personal gain. If these two considerations do not shape every medical act and every encounter with the patient, the profession becomes a lie: the physician is a fraud and his whole enterprise undiluted hypocrisy.” [7].

My students prepared to become exhibit guides with great passion and enthusiasm, and I am sure that they have found in Dr. Lejeune a role model who put truth at the core of our medical profession and research. While we were dismantling the exhib-



Figure 4. Alex leading an interactive tour of the exhibit at the University of Ottawa (Roger Guindon Hall atrium).

it, we were talking about Lejeune’s relentless attitude to follow what is real. Alex—one of the undergrad student-guides—said, “He had always a central question in his mind: What is man? (He meant in the ontogenetic and metaphysical sense.) And from that fundamental question he found all answers.” Dr. Lejeune’s words then resounded in my mind: “A physician that doesn’t have hope will never find the solution.” Lejeune’s ultimate judgment of humanity, despite all he went through, was optimistic and he exhorts this optimism—to the physician—as a moral obligation.

I have slowly started to grasp what “to advance on the path of hope” means. As I pass through the atrium of the university, I recall how my students had truly been the heart and soul of the exhibit. I wish for them to be inspired and to consider different ways to be physicians, or researchers, if that is to be their path. I have confidence that they too will learn that their future happiness will lie not only in their capabilities, success, and accomplishments, but foremost in their deep affection combined with devotion to their patients (what Dr. Lejeune used to call “compassion”), and their awareness of their own limitations (Dr. Lejeune’s definition of “humility”).

Finally, I hope that the exhibit will come again to Ottawa and continue to inspire future generations of students and physicians in training.

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