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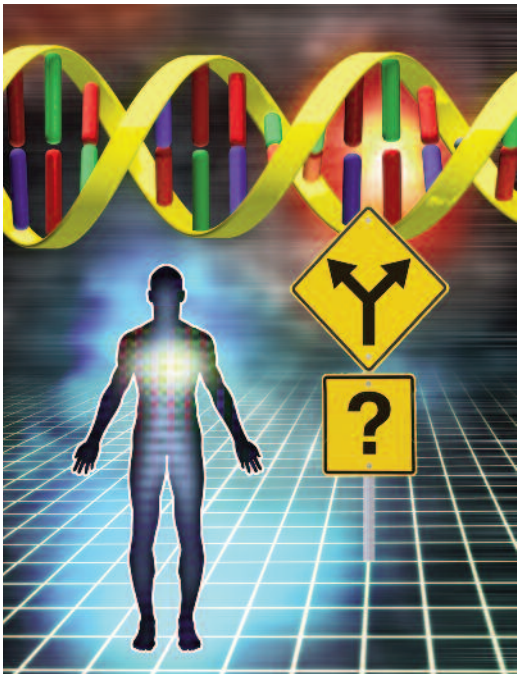
For the next 36 years, until his death, Dr. Jerome Lejeune dedicated himself to the medical care, advocacy, and research to improve the lives of persons with genetic intellectual disabilities, their families and society.

THE FORK IN THE ROAD

BY JOSEPH P. DUTKOWSKY, MD

***Editor's Note:** The following talk was delivered by Dr. Joseph P. Dutkowsky at the American Academy for Developmental Medicine and Dentistry's (AADMD) Annual Education Conference held in Princeton, NJ on June 17, 2014, the same week the 2014 Special Olympics USA Games took place.*

Raise your hand if you have ever heard the name of Jerome Lejeune. Humm, three people out of about one hundred. Now keep your hand raised if you could come up here and tell this audience what he did that matters for persons with intellectual disabilities. Down to one. This is interesting because the work of Jerome Lejeune is fundamental to the work of this academy and to the National Special Olympic Games that are happening all around us.



Two months ago I had the opportunity to be at an event in Paris that commemorated the twentieth anniversary of Jerome Lejeune's death. Interestingly, the event was not at a university or part of some academic symposium. It was in Notre Dame Cathedral. There was a formal High Mass in the great church with the Cardinal Archbishop of Paris presiding. Notre Dame was packed, standing room only, and I noted a disproportionate number of persons with Down syndrome in the congregation. After Mass there was a reception and we drank champagne on the lawn of Notre Dame celebrating the great man's legacy.

His widow then invited me and my wife to lunch at their home. We crossed The Seine to the Left Bank and went down two streets where Madam Lejeune led us to a typical row house with a shop on the first floor. She unlocked the side door and we walked down a hallway to a narrow set of stairs that led up to their home. Like many homes, children and grandchildren came and went as we talked in their modest but comfortable living room. She showed me The Kennedy Prize her husband had been awarded, the first ever given and handled to him personally by The President.

Madam Lejeune then walked me up an old wooden staircase to her husband's office which had changed little since his death. I remember there was a cup filled with sharpened and very short pencil stubs he had used to write many notes and letters. He had been a prolific academic and personal writer and all of this correspondence was there and catalogued. She even showed me and opened a suitcase where she kept the private letters he had sent her during his travels.

I guess you're wondering by now what Jerome Lejuene did that was so important to persons with intellectual developmental disabilities. Dr. Jerome Lejeune is considered to be the Father of Modern Genetics. He was the first person to look down the barrel of a microscope and count 47 chromosomes on a tissue sample from a person with Down syndrome. His additional discoveries include the identification of trisomy 8 and 9, the description of 18q syndrome, and the discovery that cri du chat syndrome is caused by a missing piece of chromosome 5.

Dr. Lejeune's name made an unexpected resurgence in the media last year when one of the co-workers in his lab claimed that this person actually was the first to count 47 chromosomes and Dr. Lejeune co-opted the credit. The response in France was intense.

A LIFE'S WORK: Madam Lejeune showing Dr. Dutkowsky her husband, Dr. Jerome Lejeune's, papers at his desk in their home in Paris.

There was a rush to rewrite medical and scientific history and correct the presumed wrong to the individual who made the claim. On this side of the Atlantic, two popular science based magazines quickly published the story and Wikipedia was even changed for a day. However, this rush to publication occurred solely on the basis of an individual's claim without any supporting evidence. Not one letter, not one lab notebook, not one collaborating witness.

Now, I already told you that I had been in Dr. Lejeune's study, that he had been a prolific writer, and that all his correspondence had been saved and catalogued. Dr. Lejeune's laboratory notebook clearly shows that he counted 47 chromosomes on a preparation from a patient with Down syndrome for the first time on May 22, 1958. In addition, a letter to Dr. Lejeune dated October 27, 1958 from the supervisor of the lab states the individual making the current claim was still counting 46 chromosomes over five months later. The evidence is definitive, in black and white from primary sources.

It's a cliche that things happen for a reason but, as cliches go, this one's more often right than not. So other than the public annoyance, editorial embarrassment, and not a small amount of stress to the Lejeune family, I do think the timing of this announcement serves a purpose.

Very, very few people will ever make a scientific discovery for the first time, especially one that directly impacts human life and society. One can only imagine the exhilaration Jerome Lejeune must have felt as he counted and recounted those 47 chromosomes, finally convinced enough to write the observation in his lab book. A pediatrician by training, he realized that for the first time, there existed a scientific basis that could be used to improve the lives of persons with Down syndrome. At the same moment, however, he said he also felt a deep horror and he knew that there were elements of society that now, because of his discovery, had an objective means to subject these persons to discrimination, segregation, and even extermination. In his words, "I see only one way left to save them, and that is to cure them. The task is immense - but so is Hope."

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For the next 36 years, until his death, Dr. Lejeune dedicated himself to the medical care, advocacy, and research to improve the lives of persons with genetic intellectual disabilities, their families and society. On his death bed, his great regret was that he had not done enough to protect his patients from a potentially menacing society. I believe Dr. Jerome Lejeune's name has resurfaced in the popular media because never more so than today are we clearly present at a fork in the road that Dr. Lejeune saw and felt viscerally in his core on that day, May 22, 1958.

With the explosion of genetic knowledge and research since the sequencing of the human genome we are closer than ever to realizing Dr. Lejeune's dream of developing genetic based treatments to alleviate pain and suffering in persons with genetic intellectual disabilities. Preventing early dementia, treating leukemia, addressing skeletal deformities, and even enhancing cognition are all within reach of this generation.

Our medical colleagues have already shown us the way in conditions such as cystic fibrosis. The discoveries turned therapies have completely rewritten the life expectancy and expectations for persons with CF and their families. Even whole medical specialties like oncology are being revolutionized by genetic based treatments. Every week we read about clinical trials of new medications that modulate the genes within a tumor leading to before unimagined tumor shrinkage on some of the most aggressive cancers known to man. These successes have come with minimal side effects compared to traditional cancer therapies. It has gotten to the point that there is consideration to have future clinical trials based not on the organ of tumor origin (such as breast, lung, or pancreatic cancer), but on the genetic signature of the malignancy. And that makes good sense.

All of this basic science research is applicable to our field of genetic intellectual disabilities. The knowledge and skill developed and being developed in our genetic

PHOTO BY MARK BRADFORD, PRESIDENT OF THE LEJEUNE FOUNDATION USA

laboratories can be transferred and adapted to our patient populations. We need only recruit and support these scientists. The day is upon us. The hour is now. It is up to us to engage the scientific community with our care givers, patients, and families to develop and employ a prudent, respectful research plan of action to use these new technologies to meet the needs of the persons with intellectual developmental disabilities we serve.

But not all in society agree. There are forces in our culture who see this as being undesirable, relatively too expensive, and frankly unnecessary. And don't kid yourself. These forces are strong, very strong. Sometimes their actions are subtle or even insidious, rarely public. We all know persons with intellectual disabilities are a medically underserved population. The data is clear; there is no question of doubt. Yet, we can't move politicians to treat these citizens with justice and equality, and our patients and families suffer. We know the majority of medical students and residents in this country do not receive even basic training in the care of persons with childhood onset disabilities. We also know that medical school deans and residency program directors when surveyed have expressed a willingness to incorporate into their curricula the care of persons with developmental disabilities. In my experience, however, those survey responses have not translated into action, and our patients and families suffer again.

DEAR FUTURE MOM: This two- minute video was produced for World Down Syndrome Day this year. It has made a difference.

Sometimes the forces are not subtle at all. There is a western European country that has said it wishes to be Downs free by the year 2030. That's only 16 years. This is a country that, only 70 years ago, had to be liberated at the cost of human life and blood from a tyrannical regime that espoused and attempted to carry out the same plan. May I suggest that International Special Olympics bypass that country when planning their future games. Think for a minute what this really means. Imagine all these playing fields in New Jersey, the gymnasiums, the pools with no athletes participating in the events. Imagine no families or friends in the stands to cheer them on. No coaches on the sidelines to instruct and encourage their athletes. Just silence, dead silence. I can only ask one question, "Have we lost our minds?!?"

So we are clearly at a fork in the road. A fork Dr. Jerome Lejeune saw and felt so clearly when he peered into that microscope on May 22, 1958. There is no mistaking where we stand. Society has to make a choice. You have to make a choice, and you can't go down both roads. I guess there is a third option. You can put on a blindfold and be led down the road of someone else's choice. Certainly, there are ample examples in history of cultures doing just that and they all have one thing in common. They all ended in catastrophe. But you've lost that choice already because you're here and your blindfold is now transparent.

So it's a choice that has to be made. You can take one road with all the rest of the perfect people. This road is wide and flat and easy. It's a comfortable walk. There's only one turn in the whole road. That's the day that you wake up and find out that the powers that be in society no longer think you're perfect anymore. There is another way. I'll warn you, though. This other road is hard. It's bumpy with steep hills and sharp turns. You can't always see what's around the next corner or over the next hill. But one thing I can tell you is what's at the end of that road. It is a place where no one's perfect, but where society is just and culture is elevated.

May there come a time when our children and our children's children look back and say this was the generation that stood up for the fundamental truth that all human life is worthy of dignity, respect, and yes, even love.

Thank you. •

ABOUT THE AUTHOR:

Joseph P. Dutkowsky, MD, Associate Medical Director of the Weinberg Family Cerebral Palsy Center, is a leading specialist in the fields of physical and intellectual disabilities and Pediatric Orthopaedic Surgeon. An activist in his field, Dr. Dutkowsky is a committed member of organizations including the American Academy of Orthopaedic Surgeons, the Pediatric Orthopaedic Society of North America, the American Academy for Developmental Medicine and Dentistry, and the American Academy for Cerebral Palsy and Developmental Medicine, for which he served as president in 2011. He serves on the Board of Directors of Cerebral Palsy Associations of New York State, and The Jerome Lejeune Foundation, USA. Additionally, he is an Attending Orthopaedic Surgeon at the Bassett Medical Center of upstate New York and an Associate Clinical Professor of Orthopaedic Surgery at the Columbia University College of Physicians and Surgeons. A pioneer in the care of adults with child-onset physical and intellectual disabilities, Dr. Dutkowsky's work was featured in an article in *The New York Times* and a television segment on PBS. Email: jd352@cumc.columbia.edu