

Jérôme Lejeune (1926–1994)

Jérôme Lejeune was a French physician and researcher who studied genetics and developmental disorders. According to the Jérôme Lejeune Foundation, in 1958, Lejeune discovered that the existence of an extra twenty-first chromosome, a condition called Trisomy 21, causes Down Syndrome. Down Syndrome is a condition present in an individual since birth and is characterized by physical and developmental anomalies such as small ears, a short neck, heart defects, and short height as children and adults. Throughout his career, Lejeune also discovered that other developmental disorders, such as cri du chat (cry of the cat) syndrome, were caused by chromosomal abnormalities. Lejeune also used his influence in the scientific community to promote pro-life beliefs, and often met with Pope John Paul II to discuss ethical dilemmas such as abortion of fetuses after detection of chromosomal abnormalities. Lejeune was one of the first researchers to link chromosomal abnormalities to developmental disorders with his discovery of Trisomy 21, leading future researchers to identify more links between the two.

Lejeune studied chromosomal abnormalities. Chromosomes are made of condensed DNA and carry genetic information necessary for proper development and growth. Typically, humans with normal development have twenty-three pairs of chromosomes for a total of forty-six chromosomes. Humans with chromosomal abnormalities may have extra or missing chromosomes. Trisomy refers to the presence of three copies of a chromosome instead of two. Humans with Trisomy 21 often have an extra copy of chromosome twenty-one, the chromosome that affects normal development during pregnancy. Down Syndrome refers to the physical anomalies present as a result of Trisomy 21. Trisomy 21 can be detected using a karyotype, which is a laboratory technique where researchers can use microscopes to produce images of an individual's collection of chromosomes. Each pair of chromosomes on the karyotype have a slightly different size and shape than the other pairs. Karyotyping can be used during pregnancy to detect chromosomal abnormalities such as Trisomy 21.

Lejeune was born on 13 June 1926 in Montrouge, France. At the age of fifteen, Lejeune received his baccalaureate, which is equivalent to a high school degree in the US, in 1941 and went on to study medicine at what is now known as the University of Paris in Paris, France. Lejeune defended his doctoral thesis in medicine on 15 June 1951 and received his doctorate in medicine promptly after. Soon after, Lejeune married a Danish woman named Birthe Bringsted on 1 May 1952. The two had five children together.

After finishing his medical studies, Lejeune studied atomic radiation and effects on humans through the National Center for Scientific Research, or CNRS, where he was a student researcher and later became an international expert on atomic radiation. In 1955, Lejeune and Raymond Turpin published a research paper on the possible effects atomic energy could have on the stability of human heredity. Turpin was the head of the Pediatrics Unit at Armand-Trousseau Hospital in Paris where Lejeune worked under Turpin while they studied genetics. They conducted the research on atomic energy while there was a rise in the emphasis on genetics and an awareness on radiation causing genetic damage. In 1955, the United Nations formed a scientific committee to study the effects of atomic radiation on humans, and in 1957 they asked Lejeune to serve as the French genetics expert on the committee. Through his work with the committee in atomic radiation, Lejeune was able to meet and make connections with leading experts in medical and physical sciences of that time and pursue genetics research.

Through the same organization, CNRS, Lejeune also began an assistantship taking consultations for children with Down Syndrome with Turpin. According to Marianna Karamanou, an associate

professor in the History of Medicine Department at the University of Athens, Greece, the scientific community at the time knew little about the cause of Down Syndrome and held the belief that it was a racial defect. The general public also often held the parents of those with Down Syndrome responsible for their children's condition and accused them of being alcoholics. According to The Association of Friends of Professor Jérôme Lejeune, Lejeune felt compassion towards children with Down Syndrome and wanted to find a successful treatment for the condition. He was able to pursue that while working under Turpin in the Pediatrics Unit of the Armand Trousseau Hospital in the 1950s. According to the Lejeune Foundation, Lejeune was also continuing his studies while working with Turpin and received his degree in genetics in 1954 and his degree in biochemistry in 1955 and continued to study the genetic cause for Down Syndrome in Turpin's lab.

While working in Turpin's lab, Lejeune searched for a genetic cause of Down Syndrome. He worked alongside Marthe Gautier, an American physician who joined Turpin's lab at the Armand Trousseau Hospital in 1956. In the lab, Gautier taught Lejeune how to observe a person's karyotype. Karyotyping was one of many techniques being used increasingly as the field of genetics expanded. The twenty-first pair of chromosomes are the smallest chromosomes in the karyotype. On 22 May 1958, while observing the karyotype of a person with Down Syndrome, Lejeune discovered the presence of an extra copy of chromosome twenty-one, resulting in a total of forty-seven chromosomes. He continued to observe the karyotypes of people with Down Syndrome and found an extra copy of chromosome twenty-one in all people he observed with Down Syndrome. Lejeune and his colleagues, Turpin and Gautier, published their discovery of that link in the journal of the Académie des Sciences (Academy of Sciences) on 26 January 1959. That organization is committed to the promotion of science. The published article, titled "Human Chromosomes in Tissue Culture," listed Lejeune as the first author, Gautier as the co-author and Turpin as the senior author. The article referred to that genetic find as Trisomy 21.

After the discovery of Trisomy 21, Lejeune discovered other chromosomal abnormalities linked to developmental disorders. In 1963 he discovered cri du chat (cry of the cat) syndrome, a condition that arises when chromosome five is missing a segment. People with cri du chat syndrome often have a high-pitched cry that researchers perceived similar to a cat alongside a small head size, wideset eyes, and other developmental anomalies. As of 2021, that abnormality is estimated to be present in one in 50,000 newborns worldwide. In 1966, Lejeune described 18q-Syndrome, which is the loss of a portion of chromosome eighteen and results in delayed development and seizure disorders. As of 2021, 18q-Syndrome is estimated to be present in one in 55,000 newborns worldwide.

In the 1960s, following his discoveries, Lejeune began holding higher level positions within scientific institutions in France. In 1964, Lejeune became the Professorial Chair of fundamental genetics at the Paris Faculty of Medicine in Paris and was the first person to hold this title. While holding this position, Lejeune continued his genetic research while speaking at conferences around the world about his discovery of Trisomy 21. In 1965, Lejeune became the head of the Cytogenetics Unit at the Necker Children's Hospital in Paris. Cytogenetics is the study of chromosomes, and during his time there, Lejeune and his coworkers investigated more than 30,000 cases of patients with chromosomal abnormalities and helped treat more than 9,000 people with mental disabilities.

Throughout his career, Lejeune was also an active Catholic, and he held membership within institutions at the intersection of religion and science. In 1974, Lejeune became a member of the Pontifical Academy of Sciences, which is a science academy of the Vatican City and claims to promote the progress of math, physical science, and natural sciences. According to the Jerome Lejeune Foundation, Lejeune was a close friend of Pope John Paul II and met with him often to discuss moral issues such as the pro-life movement. According to the Lejeune Clinic, after the discovery of Trisomy 21, other researchers began using Lejeune's techniques to detect Trisomy 21 in fetuses during pregnancy. However, many pregnant people would choose to abort their fetus after their physician detected Trisomy 21, a choice that Lejeune advocated against. Lejeune spoke at multiple conferences about his belief that fetuses should not be terminated based on positive screening for Trisomy 21.

According to Karamanou, Lejeune's beliefs about abortion did not align with the rest of the scientific community at the time. Lejeune held the traditional Catholic conviction that life begins at the moment of conception and partook in the pro-life movement, unlike most of the scientific commu-

nity at the time, Karamanou states. His strong pro-life convictions and commitment to Pope John Paul II led Lejeune to become the president of the Pontifical Academy for Life in 1981. According to Elizabeth Pain, an editor for American Association for the Advancement of Science, Lejeune's work in the pro-life movement was highly contested by scientists who were a part of the left wing. However, Karamanou claims that at least 20,000 supported his message that doctors should not perform abortions since the performing one would entail the doctor resolving the moral, economic, or social issues surrounding abortion.

In 2009, Lejeune was also involved in controversy over the discovery of Trisomy 21 many years later. Gautier, Lejeune's former co-author and fellow researcher in Turpin's lab, published a controversial account in the journal *Human Genetics* of the events that took place in the lab around the time of Lejeune's discovery. In that account, Gautier claims that she was the one who observed the karyotype and made the discovery of Trisomy 21, but Lejeune took pictures of the slides without her knowledge and used them to make it seem as though it was his discovery. In 1958, Lejeune spoke about the discovery at a seminar at McGill University, which is located in Montreal, Canada, without mentioning the talk to Turpin or Gautier ahead of time. She claims Lejeune presented the findings as his own. That account of events contradicts the accounts found in Lejeune's biography and the ones told by The Jerome Lejeune Foundation. As of 2021, Gautier and The Jerome Lejeune Foundation both maintain their position.

According to Karamanou, Lejeune's discoveries of chromosomal abnormalities laid foundations for medical genetics and opened questions regarding ethics surrounding science, specifically within the realm of abortion and the pro-life movement. Lejeune helped discover the cause of various developmental abnormalities and introduced techniques to detect those abnormalities as early as in pregnancy. Soon after the publication of the discovery of Trisomy 21, Lejeune received international recognition for his findings. In 1962, Lejeune received the Kennedy Prize from then US President John F. Kennedy for his work and discovery in the field of genetics. In 1969, Lejeune received the William Allen Memorial Award, which is an award to recognize scientific contributions in the field of genetics, San Francisco, California, for his work with chromosomal pathologies.

Lejeune died of lung cancer on 3 April 1994 in Paris.

Sources

1. Boston Children's Hospital. "Down Syndrome | Diagnosis & Treatment" Boston Children's Hospital. <https://www.childrenshospital.org/conditions-and-treatments/conditions/d/down-syndrome/diagnosis-and-treatment> (Accessed June 30, 2021).
2. Centers for Disease Control and Prevention. "Facts about Down Syndrome | CDC." Centers for Disease Control and Prevention, April 6, 2021. <https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html> (Accessed June 30, 2021)
3. Creager, Angela. "Radiation, Cancer, and Mutation in the Atomic Age." *Historical Studies in the Natural Sciences* 45 (2015): 14-48.
4. Disabled World. "Cri Du Chat Syndrome: Causes, Symptoms and Treatment." Disabled World. <https://www.disabled-world.com/disability/types/cri-du-chat.php> (Accessed June 30, 2021).
5. Engel, Randy. "Trisomy 21." *People Concerned for the Unborn Child*. <https://www.pcuc.org/trisomy-21/> (Accessed June 30, 2021).
6. Gautier, Marthe, and Peter S. Harper. "Fiftieth Anniversary of Trisomy 21: Returning to a Discovery." *Human Genetics* 126 (2009): 317-24. <https://doi.org/10.1007/s00439-009-0690-1> (Accessed June 30, 2021).
7. Global Down Syndrome Foundation. "Down Syndrome Research and Medical Care Timeline." Global Down Syndrome Foundation. <https://www.globaldownsyndrome.org/about-down-syndrome/history-of-down-syndrome/research-and-medical-care-timeline/> Accessed June 30, 2021).
8. Institut de France Académie Des Sciences "Académie Des Sciences - Home." Institut de France Académie Des Sciences. <https://www.academie-sciences.fr/en/> (Accessed June 30, 2021).
9. Institut Lejeune. "Who Was Professor Jérôme Lejeune?" Institut Lejeune. <https://www.institut-lejeune.org/en/who-are-we/who-was-professor-jerome-lejeune.html> (Accessed June 30, 2021).

10. Karamanou, Marianna, Emmanouil Kanavakis, Ariadni Mavrou, Eleni Petridou, and George Androutsos. "Jérôme Lejeune (1926-1994): Father of Modern Genetics." *Acta Medico-Historica Adriatica: AMHA* 10 (2012): 311-6.
11. Kunze, J., E. Stephan, and M. Tolksdorf. "Ring-chromosom 18." *Humangenetik* 15 (1972): 289-318. <https://doi.org/10.1007/BF00281730> (Accessed June 30, 2021).
12. Medline Plus. "Distal 18q Deletion Syndrome: MedlinePlus Genetics." Medline Plus. <https://medlineplus.gov/genetics/condition/distal-18q-deletion-syndrome/> (Accessed June 30, 2021).
13. Mégarbané, André, Aimé Ravel, Clotilde Mircher, Franck Sturtz, Yann Grattau, Marie-Odile Rethoré, Jean-Maurice Delabar, and William C. Mobley. "The 50th Anniversary of the Discovery of Trisomy 21: The Past, Present, and Future of Research and Treatment of Down Syndrome." *Genetics in Medicine* 11 (2009): 611-6. <https://doi.org/10.1097/GIM.0b013e3181b2e34c> (Accessed June 30, 2021).
14. Migeon, Barbara Ruben. "Les Chromosomes Humains (Caryotype Normal et Variations Pathologiques) by Raymond Turpin and Jerome Lejeune. Paris: Gautier-Villars, 1965, 535 Pp. 100 Figures, 54 F." *Pediatrics* 36 (1965): 965-6. <https://pediatrics.aappublications.org/content/36/6/965.2> (Accessed June 30, 2021).
15. National Organization for Rare Disorders. "Cri Du Chat Syndrome." National Organization for Rare Disorders. <https://rarediseases.org/rare-diseases/cri-du-chat-syndrome/> (Accessed June 30, 2021).
16. Pace, Eric. "Dr. Jerome Lejeune Dies at 67; Found Cause of Down Syndrome." *The New York Times*, April 12, 1994, sec. Obituaries. <https://www.nytimes.com/1994/04/12/obituaries/dr-jerome-lejeune-dies-at-67-found-cause-of-down-syndrome.html> (Accessed June 30, 2021).
17. Pain, Elisabeth. "After More Than 50 Years, a Dispute Over Down Syndrome Discovery." *Science | American Association for the Advancement of Science*, February 11, 2014. <https://www.sciencemag.org/news/2014/02/after-more-50-years-dispute-over-down-syndrome-discovery> (Accessed June 30, 2021).
18. Roizen, Nancy J., and David Patterson. "Down's Syndrome." *The Lancet* 361 (2003): 1281-89.
19. The Association of Friends of Professor Jérôme Lejeune. "HOME." The Association of Friends of Professor Jérôme Lejeune. <http://www.amislejeune.org/index.php/en/> (Accessed June 30, 2021).
20. The Lejeune Clinic. "Jerome Lejeune." The Lejeune Clinic. <https://www.lejeuneclinic.com/about-us/jerome-lejeune> (Accessed June 30, 2021).
21. The National Catholic Bioethics Center. "A Model of Catholic Bioethics: Venerable Jérôme Lejeune." The National Catholic Bioethics Center. <https://www.ncbcenter.org/messages-from-presidents/lejeune> (Accessed June 30, 2021).