Jérôme Lejeune (1926–1994) [1]

By: Fitzgerald, Grace Arslan, Maeen Keywords: Chromosomal Abnormalities [2] History of Down Syndrome [3]

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 [4] to discuss ethical dilemmas such as abortion [5] 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 [6] 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 [7]. Down Syndrome refers to the physical anomalies present as a result of Trisomy 21. Trisomy 21 can be detected using a *karyotype* [8], 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* [8] have a slightly different size and shape than the other pairs. Karyotyping can be used during pregnancy [7] 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 [9] 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 [10] and effects on humans [6] through the National Center for Scientific Research, or CNRS, where he was a student researcher and later became an international expert on atomic radiation [10]. 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 [10] causing genetic damage. In 1955, the United Nations formed a scientific committee to study the effects of atomic radiation [10] on humans [6], 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 [10], 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 [11], 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* [8]. 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* [8]. On 22 May 1958, while observing the *karyotype* [8] 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
Sources


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