NATURE | NEWS

Down's syndrome discovery dispute resurfaces in France

Debate flares over who discovered extra chromosome that causes the disease.

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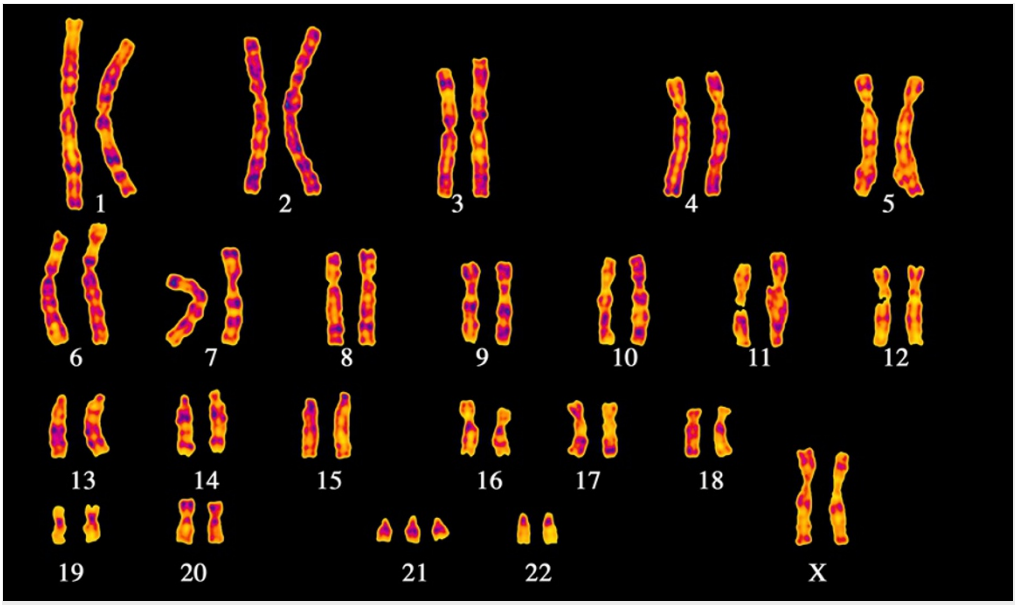

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An extra copy of chromosome 21 causes Down's syndrome — but there is a dispute over who discovered it.

A disagreement over the discovery of the cause of Down's syndrome has resurfaced in France more than 50 years after the findings were published.

The dispute erupted again at the French Federation of Human Genetics' (FFGH) seventh biennial congress on human and medical genetics in Bordeaux at the end of last month.

Paediatric cardiologist Marthe Gautier, who was involved in the experiments that led to the identification of the extra copy of chromosome 21 — the cause of the syndrome — was due to relate her role in the discovery when two bailiffs arrived with a court authorization to record the session. The FFGH then decided at the last minute to cancel Gautier's presentation.

The bailiffs were representing the Paris-based Jérôme Lejeune Foundation, which finances a large proportion of current Down's syndrome research in France. The foundation does not deny Gautier's contribution to the work leading to the discovery, but it credits the late Lejeune for the discovery itself.

Lejeune, a geneticist, was first author of the key paper reporting[1] the finding, published by the French Academy of Sciences in January 1959. Gautier was listed as second author, and Raymond Turpin, a paediatric geneticist and Gautier's and Lejeune's boss at the Trousseau Hospital in Paris, was listed as third author.

Recognizing roles

The FFGH says that it wanted to honour Gautier's role in the discovery by giving her the floor and awarding her the federation's grand prize. "Without questioning Jérôme Lejeune's very important contribution to French genetics through the article on trisomy 21 and other work, we simply wanted to make a gesture in recognition of the determinant character of Marthe Gautier's contribution," the federation said in a statement.

But when the bailiffs walked in, “we realized the recording might be used in a court case”, FFGH treasurer and former president Dominique Bonneau told *Nature*. “Not only do we not have the funds to fight a libel suit, but we felt it was inappropriate to hold the presentation under such strong legal pressure.” Gautier received her prize discreetly and nine eminent geneticists signed a statement endorsing the decision to cancel the presentation.

Jean-Marie Le Méné, president of the Jérôme Lejeune Foundation, says that the bailiffs were sent because the foundation wanted an official recording of the talk so that there could be no dispute over what was said. “We needed to know what was said in case Jérôme Lejeune’s memory was smeared,” he says.

On the basis of a letter from Turpin to Lejeune in October 1958, Le Méné asserts that Lejeune, and not Gautier, identified the key forty-seventh chromosome — the third copy of chromosome 21. “The letter said that Gautier was still at 46 chromosomes, even though she now claims to have made the discovery six months earlier,” says Le Méné.

Lejeune had hoped to win a Nobel prize for the discovery, and is quoted as saying that he was passed over because of his anti-abortion views.

Different stories

In a press statement posted on its website, the Jérôme Lejeune Foundation says that the media has been unfair to Lejeune. It singles out two French newspapers: *Le Monde*, with its article ‘Trisomy: a pioneer intimidated’, and *Libération*, and suggests that some media are using Gautier to discredit the Catholic, pro-life Lejeune as the discoverer.

Gautier says that she was the first to pinpoint a forty-seventh chromosome, but could not identify it because her microscope was not powerful enough. She handed her slides over to Lejeune, who photographed them at a laboratory with better equipment. She adds that Lejeune took full credit for the results at a congress in August 1958 in Montreal, Canada, and that she was told about the 1959 paper[1] only shortly before it was submitted.

Le Méné, however, maintains that there is no evidence that Gautier made the key discovery. “The scientific community has never disputed the fact that Lejeune was responsible for this remarkable breakthrough, and Gautier has not provided the least proof that this was not the case.”

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References

1. Lejeune, J., Gauthier, M. & Turpin, R. *C. R. Acad. Sci.* **248**, 602–603 (1959).