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Jérôme Lejeune pioneer in the **discovery** **of Trisomy 21**

A whole life dedicated to the care of his patients and to the search for a treatment

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Introduction

On January 26, 1959, and then on March 16, 1959, two communications to the Academy of Sciences, establishing the presence of 47 chromosomes in the so-called mongolian children, were published under the signatures Lejeune, Gautier, Turpin, and in this order indicating that the first one is the discoverer, that the last one is the team leader and that between the two is the contributor to the discovery.

For 50 years, nothing happened. But, since 2009, the memory of Jérôme Lejeune has been attacked by Marthe Gautier, born in 1925, the only survivor of the team, who claims today to have discovered trisomy 21 by herself. Jérôme Lejeune would have been a usurper and Marthe Gautier would have been defrauded.

Some of these claims had been taken up by an opinion of the INSERM ethics committee made public on September 14, 2014. It stated that Jérôme Lejeune's share "was unlikely to have been preponderant" and confined his contribution solely to the valorization of the discovery.

Faced with these accusations and this denigration of Jérôme Lejeune's role, the Jerome Lejeune Foundation decided to react. In 2009, it had remained silent, noting that Marthe Gautier's personal declarations had only limited circulation. Nevertheless, in February 2014, when it learned that Marthe Gautier was about to speak on the discovery of trisomy 21 before the Assises de Génétique Humaine et Médicale in Bordeaux, the Foundation had decided to have her words recorded by a bailiff, by virtue of an authorization granted by the courts, in order to defend the reputation of Jérôme Lejeune if he were to be defamed. The Foundation's intuition was right. This opinion of the INSERM ethics committee rewrote the history of the discovery following Marthe Gautier. The Foundation was therefore led to communicate several elements and documents that shed light on a controversy that has little to do with science.

It was Jérôme Lejeune, and not Marthe Gautier, who was asked by Prof. Raymond Turpin, their head of department at Trousseau, to be the first to sign the January 1959 publication, thus designating himself, in the eyes of history, as the one who would remain the discoverer of the cause of mongolism.

And it is in fact Jérôme Lejeune who, following this discovery, will devote his life to seeing patients with Down syndrome, to seeking a treatment to cure them and to defending their lives, which are so precious to him. Jérôme Lejeune, for his part, never ceased to praise Marthe Gautier's contribution to the discovery. In the epistolary exchanges between the two colleagues, there is no trace of polemic, on the contrary.

The rest is a rereading of a past repainted in the colors of contemporary ideologies to create and feed a pseudo polemic. Marthe Gautier, like the young women scientists of the time, would have been the victim of a mandarin machismo on the part of Lejeune. The argument is ridiculous since Lejeune was Marthe Gautier's youngest son and he was not the head of the department.

I. Summary – Main points of the controversy

In 2009, on the occasion of the 50th anniversary of the discovery of trisomy 21, Doctor Marthe Gautier made polemical statements in an article in *Médecine/Sciences* (Volume 25, Number 3, March 2009), challenging the role of Professor Lejeune. She states in substance:

- that she was the first to count 47 chromosomes, and that she did it all by herself,
- that "a newcomer", a "trainee" named Lejeune, then simply stole the discovery from her,
- that Lejeune attributed the discovery to himself and thus reaped all the glory afterwards.

These statements were repeated in other interviews (including the main one to Randy Engel on March 6, 2013), with variations, and repeated in early 2014 without any article offering objective evidence or providing testimony to support Marthe Gautier's thesis.

However, the facts and archival documents show a completely different reality:

- **The discovery was the result of a long and patient process of collective research on Down syndrome.** It is no exception to the rule that the first hypotheses had to be tested and confirmed for months.
- **Handwritten notes in the analysis notebook in May 1958 show that Jérôme Lejeune was the first to count 47 chromosomes** and that he was the driving force of the team, without whom the work would hardly progress.
- **Jérôme Lejeune** publicly honored each member of Turpin's team, including (but not limited to) Marthe Gautier, notably during his inaugural lecture in 1965. The correspondence between Lejeune and Gautier shows that they had an active and trusting working relationship. This relationship lasted at least until 1962, three years after the publication of the discovery (Lejeune, Gautier and Turpin).

In view of these facts, Marthe Gautier's accusations appear unfounded and raise many unanswered questions. In particular:

- **Marthe Gautier** speaks out 50 years after the 1959 discovery and after the death of all the other protagonists (including Jérôme Lejeune, more than 25 years ago) - and without any questioning having been issued before. Why is this?
- **There is no evidence to support the accusations against Lejeune.** The only document Marthe Gautier has put forward is a letter Lejeune sent to her in the fall of 1958 praising his preparations of cellular techniques. To his credit, Lejeune did so again during his inaugural lecture. Recognizing the work of Marthe Gautier does not mean attributing the discovery to her.
- **Since 2009, the accusations have changed in nature and tone as Marthe Gautier's statements have been made. There are many inconsistencies.**

II. Questions / Answers

A. Who discovered the cause of Down syndrome in the end?

- The discovery of the cause of Down syndrome was first the result of a long, patient and progressive team work, including Marthe Gautier. Marthe Gautier claims that she discovered the cause "alone, with her laboratory assistants", which she immediately contradicts by admitting that she did not have the necessary material to prove "her discovery" (see following questions).
- Jérôme Lejeune clearly had the role of driving force and coordinator of the research, under the impulse of the head of the laboratory Raymond Turpin. On May 22, 1958, he identified for the first time a "mongoloid" patient who had a supernumerary chromosome.
- Notes from the laboratory notebook, kept day to day by Jérôme Lejeune from July 10, 1957, indicate very precisely that he counted on May 22, 1958 on the notebook of 47 chromosomes. In December 1958, he identified two new cases of 47 chromosomes linked to mongolism which confirmed the first observation.

References:

- Cover of the laboratory notebook kept by Jérôme Lejeune (Appendix 1): <https://www.flickr.com/photos/125139824@N04/14335829151/>
- Page of the laboratory notebook which indicates the first dated observations (Appendix 2): <https://www.flickr.com/photos/125139824@N04/15164085700/>

B. Was Marthe Gautier, as she claims, the first to count 47 chromosomes?

First of all, the dates cited by Marthe Gautier to justify her version and to explain how she would have been the first to count 47 chromosomes are confusing or later than Jérôme Lejeune's own observations.

Then Marthe Gautier's statements to have been able to "succeed alone with (her) laboratory assistants (...) in highlighting an anomaly" are contradictory. It is impossible that Marthe Gautier could have established the chromosome count alone. She says it herself in her different declarations, since she affirms that "The extra chromosome is small, the lab does not have a photomicroscope that would allow to attest its presence and to establish the karyotype" then admits that, without a photo, it is impossible to attest the presence of an extra chromosome...

References:

- The articles that show the imprecise, changing and contradictory accusations of Marthe Gautier on this point:

https://www.medecinesciences.org/en/articles/medsci/full_html/2009/04/medsci2009253p311/medsci2009253p311.html

<http://www.newengelpublishing.com/randy-engel-interview-with-marthe-gautier-french-translation>:

- Page from the laboratory notebook that indicates the first dated observations (Appendix 2): <https://www.flickr.com/photos/125139824@N04/15164085700/>
- Letter from Raymond Turpin to Jérôme Lejeune dated 10/27/1958 (Appendix 3): <https://www.flickr.com/photos/125139824@N04/15351427675/>

C. Did Jérôme Lejeune "sequester" Gautier's preparations? And therefore stole the discovery from Marthe Gautier?

- Once again, the notes in the laboratory notebook contradict Marthe Gautier's version that the slides containing her preparations were "sequestered" (article in Médecine / Sciences, March 2009) by Lejeune from June 1958 onwards, after he offered to help her because he had access to a microscope: the notes in the laboratory notebook where the observations are recorded indicate that Jérôme Lejeune counted 47 chromosomes in May 1958.
- As indicated below, the correspondence between Gautier and Lejeune in the months and years following the "sequestration" testify to a close, cordial and efficient collaboration between them. It is difficult to see how this would have been possible after the sequestration and the supposed silence of Jérôme Lejeune.
- Moreover, if Marthe Gautier had had the leading role, why did she not launch the publication without waiting for Lejeune's return from his trip in the fall of 1958? The argument of the photo does not hold since she did not need the photo to publish. The publication does not include a photo and in any case what was stopping her from redoing the photo?

References:

- Page of the laboratory notebook which indicates the first dated observations (Appendix 2): <https://www.flickr.com/photos/125139824@N04/15164085700/>
- Letter from Marthe Gautier to Lejeune dated 20/10/1958 showing their close collaboration (Appendix 4): <https://www.flickr.com/photos/125139824@N04/15164660018/>

D. Did Marthe Gautier, as she claims, not know Jérôme Lejeune before the discovery? Was she working alone? Were the relationships between the team members distant or strained?

On the contrary, the archives show a team working in harmony and in a complementary way throughout the process.

- A letter from Marthe Gautier in June 1957 discredits her own accusations that she did not know Jérôme Lejeune, "a newcomer," before her cell culture work, which she began in July 1957. Yet she begins her letter with "Dear friend, very happy to hear from you," and keeps him informed of their work.
- Marthe Gautier was certainly working in a laboratory set up at the request of Professor Turpin, but in close collaboration with the team and in particular with Jérôme Lejeune. She kept up a regular correspondence with him whenever he traveled (at least three letters attest to this) between 1957 and 1958.
- Professor Turpin, head of the department, was obviously well aware of Marthe Gautier's work, as can be seen in his letters of 27/6/1957 and then of 12 and 27/10/1958, in which he informed Lejeune of her work on each occasion. He says in particular on October 12 "Mademoiselle Gautier always writes to me regularly".
- Finally, the professional relationship of trust between Lejeune and Gautier lasted at least three more years after the first publication on the discovery. In his correspondence to his wife, Jérôme Lejeune quotes Marthe Gautier regularly and in positive terms that prove that their collaboration is good and continuous. In a letter dated July 12, 1962, Lejeune wrote "Gautier who had arrived showed herself absolutely charming (...) She promised to help.
- The content of all the letters clearly shows a cordial and active collaboration, not distant and tense, even over time. Marthe Gautier begins all her letters to Jérôme Lejeune with "Dear friend" and even asks him in her letter of 20/10/1958: "Pick up all the tips you can about tissue cultures and all the offprints that are given at random during your visits."

References:

- Letter from Marthe Gautier to Lejeune dated 20/10/1958 showing their close collaboration (Appendix4)
<https://www.flickr.com/photos/125139824@N04/15164660018/>
- Letter from Raymond Turpin to Jérôme Lejeune dated 27/10/1958 (Appendix 3):
<https://www.flickr.com/photos/125139824@N04/15351427675/>
- Letter from Lejeune to Gautier of 5/11/1958 shown by Gautier in his article:
http://www.medecinesciences.org/articles/medsci/full_html/2009/04/medsci2009253p311/medsci2009253p311.html
- Letters from Lejeune to his wife Birthe of 2/7/1961, 16/7/1962 and 30/7/1962.

E. Was Lejeune a mere trainee at the time, and inferior in hierarchy to Marthe Gautier? Did Marthe Gautier have the leading role in the discovery she describes?

- Dr. Lejeune was not a research trainee in 1958, contrary to Marthe Gautier's assertion, but a research fellow (at the CNRS), which in the French university hierarchy means two higher ranks and the responsibilities that go with them.

When Marthe Gautier joined the team, Jérôme Lejeune already had an important place in Professor Turpin's team, which he kept throughout the period of the discovery. Turpin sent him to the UN in 1957 and to the USA in 1958 to "defend the position of French genetics" at the prestigious scientific congress in Montreal. He entrusted him with the writing of articles and especially the writing of the book on chromosomes, and finally he supported him in the project of creation of the Institute of Progenesis... Marthe Gautier is not mentioned in any of these 'files' (while Marie-Odile Rethoré, another member of the team is, which leads us to think that Turpin would have also mentioned Marthe Gautier if she had collaborated).

- Lejeune has a driving role in the team. As proof: when he was away, Marthe Gautier herself admitted that the work was not progressing. The notes from the laboratory, Lejeune's diary, the multiple correspondences of the team, and the private exchanges point to Jérôme Lejeune as the central figure towards whom the reports converge and from whom the research hypotheses start.

Conversely, a multitude of letters make it clear that Marthe Gautier is not the central figure she claims to be and Jérôme Lejeune is not the young "intern", "newcomer to the laboratory" she describes. Although she initiated a heart cell culture in June 1957, Marthe Gautier is a person who is not very present because she has other hospital activities in cardiology: "I have not seen Mademoiselle Gautier (assistant at Bicêtre) because she only has to come in the afternoon."

References:

- Dated summary of the titles and research positions of J. Lejeune in an official publication(Appendix5):
<https://www.flickr.com/photos/125139824@N04/14152627809/>
- Correspondence from Raymond Turpin to Jérôme Lejeune of 29/6/57, 5/9/1958, 12/10/1958, 20/10/1958 etc.
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- Correspondence between Jérôme Lejeune and his wife and his personal diary, notably from 16/1/1959 (Appendix 6)
<https://www.flickr.com/photos/125139824@N04/15351221892/>

F. Is Jérôme Lejeune this character with a "not very brilliant" and opportunistic career that Marthe Gautier describes? Did he publish the first article on the discovery "in a hurry" to overtake other international research teams?

- Professor Lejeune's interest and expertise, especially in genetics, which Marthe Gautier contested, were proven very early on and will never be denied. This dismisses accusations of opportunism. **By the beginning of 1958, Lejeune had already co-authored 7 scientific publications specifically on "mongolism", in contrast to Marthe Gautier, who had not published anything on the subject.**
- Jérôme Lejeune often recalled in his diary that his goal was to discover the cause of the disease in order to hopefully find a cure and, one day, treat his beloved patients to whom he showed so much deference. In his notes of January 16, 1959, just before the publication of the first article, he says: "It is not enough to discover an aberrant chromosome, one should also know what 'it' does! (...) To make a mongoloid capable of being named Professor at the Faculty of Medicine of Paris! That is the dream of my existence."
- Jérôme Lejeune was aware of the importance of observation but remained cautious and scrupulously applied a scientific approach. He wanted to have at least three clear cases of observations of 47 chromosomes before publishing, which he obtained in December 1958, as his journal testifies. He also had to convince Professor Turpin, who remained doubtful even after the first publication.

References:

- Titles and scientific works of Jérôme Lejeune, 1972, l'Expansion Française
- Personal diary of January 16, 1959 and March 7, 1959 (Appendix 6)
<https://www.flickr.com/photos/125139824@N04/15351221892/>

G. Did Lejeune take credit for the discovery? Did he receive all the honors afterwards for his role in this discovery, including the Kennedy Prize without sharing the award?

- Jérôme Lejeune, as his diary and public statements show, never attributed the discovery to himself. It was attributed to him in fact. It was Prof. Turpin who recognized Jérôme Lejeune as the discoverer by asking him to sign the lead article first. But he never missed an opportunity to thank or honor his collaborators. In a letter dated November 5, 1958, he thanked Marthe Gautier for her "excellent preparations" while Turpin congratulated him for his preparations (letter from Turpin to Lejeune dated October 27, 1958). During his inaugural lecture at the first chair of fundamental genetics of the Academy of Medicine of Paris in 1965, he quotes and honors at length each member of the team. He salutes his "dear Marthe Gautier" and recognizes her unique contribution to the discovery, citing her "skill" and "tenacity" during "two years of failures and half-successes".
- In 1962, Jérôme Lejeune was awarded the Kennedy Prize for all of his work and not only for his role in the discovery. This award was the result of a survey that the Kennedy

Foundation team visiting Paris had conducted among his colleagues in his absence since he was abroad. The letter from the Kennedy Foundation announces that Lejeune received \$8,333 (about €6,000) for his personal use and \$25,000 (about €18,300) as an "exclusive grant for a research program.

References:

- Lejeune's letter to Gautier of 5/11/1958 shown in Marthe Gautier's article: http://www.medecinesciences.org/articles/medsci/full_html/2009/04/medsci2009253p311/F2.html
- Excerpt from Jérôme Lejeune's inaugural lecture on March 10, 1965 greeting Marthe Gautier Appendix7) <https://www.flickr.com/photos/125139824@N04/14335819821/>
- Transcript of the letter announcing the award of the Kennedy Prize to Lejeune dated 4/12/1962

H. What are the reasons for this controversy?

- In 2009, on the occasion of the 50th anniversary of the discovery, Marthe Gautier made her first accusations in an article in Médecine/Sciences. She says she was "encouraged" by friends, including Professor Simone Gilgenkrantz.
- It is known that Simone Gilgenkrantz was among the first to oppose Jérôme Lejeune, who refused prenatal diagnosis leading to the elimination of children with Down syndrome. Simone Gilgenkrantz writes in one of her books that French genetics missed out on the Nobel Prize because of Jérôme Lejeune's positions.
- It is now a widespread idea that in the scientific field, women have been bullied and confined to a subordinate role. This is an anachronistic and unfair interpretation in the case of Professor Lejeune and Marthe Gauthier.

One is therefore entitled to wonder, in spite of the obvious, whether non-scientific but political motives have not provoked - and biased - Marthe Gautier's accusations and her rereading of history.

References:

- Interview with Marthe Gautier in which she thanks her friends for having encouraged her: <http://www.newengelpublishing.com/andy-engel-interview-with-marthe-gautier-french-translation/>

III. The Foundation's response to the opinion of the INSERM ethics committee (September 2014)

"The opinion of the INSERM ethics committee on the discovery of trisomy 21 calls for the following observations from the Jerome Lejeune Foundation.

A lack of memory or gratitude

As testified by the writings and public statements of the time, Jérôme Lejeune has always recognized and saluted the role of Marthe Gautier in the discovery of trisomy 21, which is the contribution of an American technique of cell tissue culture. By forgetting this tribute, which has never been denied, Mrs. Gautier shows a lack of memory or gratitude. But the contribution of the cell culture technique is not the essence of the discovery. The discoverer is the one who started from a hypothesis, verified it, believed in it and carried the project.

Some counter-truths

Contrary to what Marthe Gautier also maintains, Jérôme Lejeune never attributed the discovery to himself. It was Professor Raymond Turpin, head of the department in which Gautier and Lejeune worked, who recognized Jérôme Lejeune as the discoverer of Down syndrome. Indeed, as shown in his laboratory analysis notebook, on May 22, 1958, Jérôme Lejeune was the first to count all 47 chromosomes and to scientifically establish, for the first time, a link between a chromosomal anomaly and mental retardation. Gautier, who was not a geneticist, had no interest in mongolism, and had not published anything on the subject, could not make the connection alone. On the contrary, Lejeune, who was in charge of the consultation of patients who were then called "mongoloid", had already published several articles on mongolism, and was already a researcher at the CNRS. He was not the unknown person described by Mrs. Gautier.

We have at the disposal of historians the handwritten letter addressed on October 27, 1958 by Prof. Turpin to Jérôme Lejeune (who was traveling abroad at the time) which informs the latter that Marthe Gautier is still counting 46 chromosomes instead of 47 in mongoloid children.

A bias

The fact that the ethics committee of INSERM does not take into account established and documented facts, but prefers to rely on Mrs. Gautier's rereading and memories, 20 years after Jérôme Lejeune's death, must challenge us.

The referral from INSERM and the text of its ethics committee suggest two explanations:

- The desire to highlight the sexist discrimination to which young women scientists in the research sector were subjected by the "mandarins" in the 1960s, which is a chronological contradiction since Jérôme Lejeune was 33 years old at the time of the discovery, and therefore younger than Mrs. Gautier, and was not the head of the department;
- the desire to damage the reputation of a French scientist, of international renown, who devoted part of his life to the defense of human life threatened by abortion (and who, for this reason, caused the team to lose the possibility of obtaining the Nobel Prize). "

IV. Focus: The technique does not make the discovery

One should reread Marthe Gautier's article which is the basis for the opinion of the INSERM ethics committee: she gives pages of details on how to make a cell culture. She does not write anything about the study of mongolism, the reason for the research on chromosomes, the consequences of this discovery. It is a technical element brought to the reflection led by Raymond Turpin and Jérôme Lejeune. Everyone agrees on this point. Jérôme Lejeune never missed an opportunity to thank or honor all his collaborators, including Marthe Gautier. He has always associated Raymond Turpin and Marthe Gautier with the communication of the discovery. But the contribution of the cell culture technique is not the essence of the discovery.

The notice states that "Marthe Gautier will observe the 47 chromosomes". To "observe" does not mean to discover the cause of mongolism. An analogy can help us to understand what is at stake: with regard to the discovery of America, was it Christopher Columbus who first saw the shores of America? Was it not a sailor? But the discoverer, the one that history remembers, is the one who started from a hypothesis, verified it, believed in it and carried the project. We must distinguish between the contribution of a technique and the main role.

On this second point, there is no proof of Marthe Gautier's preponderant role. Moreover, it remains constant that Marthe Gautier did not make the count first. This is known from two written documents: Lejeune reported in his laboratory analysis notebook on May 22, 1958 that he had counted 47 chromosomes, explicitly underlining his questioning of this figure. With regard to Marthe Gautier, a letter from Professor Turpin to Jérôme Lejeune (who was traveling abroad at the time) dated October 27, 1958, indicates that she is still at 46, five months after Lejeune's count.

V. Myths and facts: The real chronology

The objective of the document on the following page is to illustrate in the form of a frieze the chronology of the discovery as it unfolded, by putting in mirror a selection of historical elements supported by archival documents (mail, laboratory notes, university archives, etc.) and certain accusations made by Marthe Gautier.

Available visuals:

- The **graphic "The Discovery of Down Syndrome: Myths and Facts"** copied as an image below and available in PDF, with clickable links to excerpts from archival documents.
- **The archival visuals** cited in the graphic below, visible online, in series or one by one, on the album (FlickrR): <https://www.flickr.com/photos/125139824@N04/>

ANNEXES

LA DECOUVERTE DE LA TRISOMIE 21 : Mythes et Faits

Où a été le premier à découvrir la trisomie 21? J'ai même Leljeune s'est-il approprié la découverte? Cette infographie montre que la découverte fut l'aboutissement d'un long travail d'équipe, comme l'indiquent les trois noms (Leljeune, Gaudier, Turpin) des auteurs de la première publication scientifique sur le sujet. Bien que les notes dans le carnet d'analyse du laboratoire montrent que Leljeune découvrit 47 chromosomes en premier, et plusieurs fois, il a toujours reconnu dignement le rôle de chaque membre de l'équipe, et notamment celui de Martine Gaudier, qui dirigea l'impression et l'intégrité de Leljeune et ce plus de 20 ans après sa mort.

1946

FAIT
Gaudier écrit à Leljeune « Cher ami, très heureux d'avoir de vos nouvelles et de lire au courant de vos travaux ».



A partir de cette date, Leljeune commence à compiler ses observations dans des préparations de chromosomes et « mélange » et prend note dans un carnet d'analyse de laboratoires.

22 Mai



Le carnet d'analyse de laboratoires de Leljeune indique « 1 (trisomie) numéros » et « 47 chromosomes ».

28 Oct



A cette date, Leljeune avait déjà co-écrit avec Raymond Turpin 7 publications sur le chromosome 21. Martine Gaudier qui, elle, ne ren pointe sur le sujet.

27 Oct



Turpin écrit à Leljeune « Mlle Gaudier et Mme Maud en sont encore à 46 ».

1947



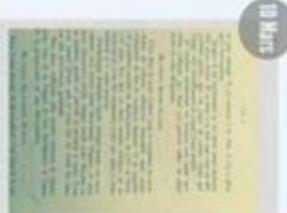
La découverte est publiée pour la première fois dans une revue scientifique pour l'Académie des Sciences. « Leljeune, Gaudier, Turpin, Les chromosomes humains en culture de tissus ».

7 Mars



Le carnet d'analyse de Martine Gaudier porte le commentaire « 47, sûr » sur une photo de caryotype.

10 Mars



Leljeune salue sa « chère Martine Gaudier » et reconnaît la découverte à la « Martine » « habitée » et sa « Martine » pendant « deux années d'échecs et de demi-succès ».

1957

FAIT
A cette date, Gaudier et Leljeune ont déjà en équipe découvert 47 chromosomes en premier. Turpin 5 ans auparavant.

MYTHE

Gaudier affirme dans une interview en mars 2013 à propos de Leljeune : « Je ne le considère pas comme découvreur (avant de publier la culture cellulaire) et dans un article de 2009 qu'il était « un nouveau venu ».

1958

FAIT
Leljeune lui la découverte pour la première fois et la note dans son carnet d'analyse.

MYTHE

Dans son article de 2009, Gaudier déclare avoir reçu « (...) et surtout de mener en évidence une anomalie ».

1958

FAIT
Au moment de la découverte, Leljeune était Chargé de Recherche au CNRS, labor de Turpin, il était déjà à cette époque un scientifique prometteur.

MYTHE

Leljeune n'était que stagiaire CNRS lors du projet.

1959

FAIT
Pendant que Leljeune est en mission à Strasbourg, Leljeune et Turpin ont trouvé 47 chromosomes.

MYTHE

Gaudier a découvert la première trisomie en plus, à partir du mois de mai (interview de mars 2013).

1959

FAIT
C'est seulement après le retour de Leljeune de Strasbourg que Turpin trouve les résultats nécessaires pour confirmer la découverte.

MYTHE

Gaudier affirme que l'article est « l'ensemble » publié, en urgence, uniquement pour prendre de court les médias (interview de 2009).

1965

FAIT
Leljeune continue à trouver des cas de trisomie 21, et confirme la découverte par la trisomie 21.

MYTHE

Gaudier déclare : « un grand succès par 21, il est évident » (interview de 2009).

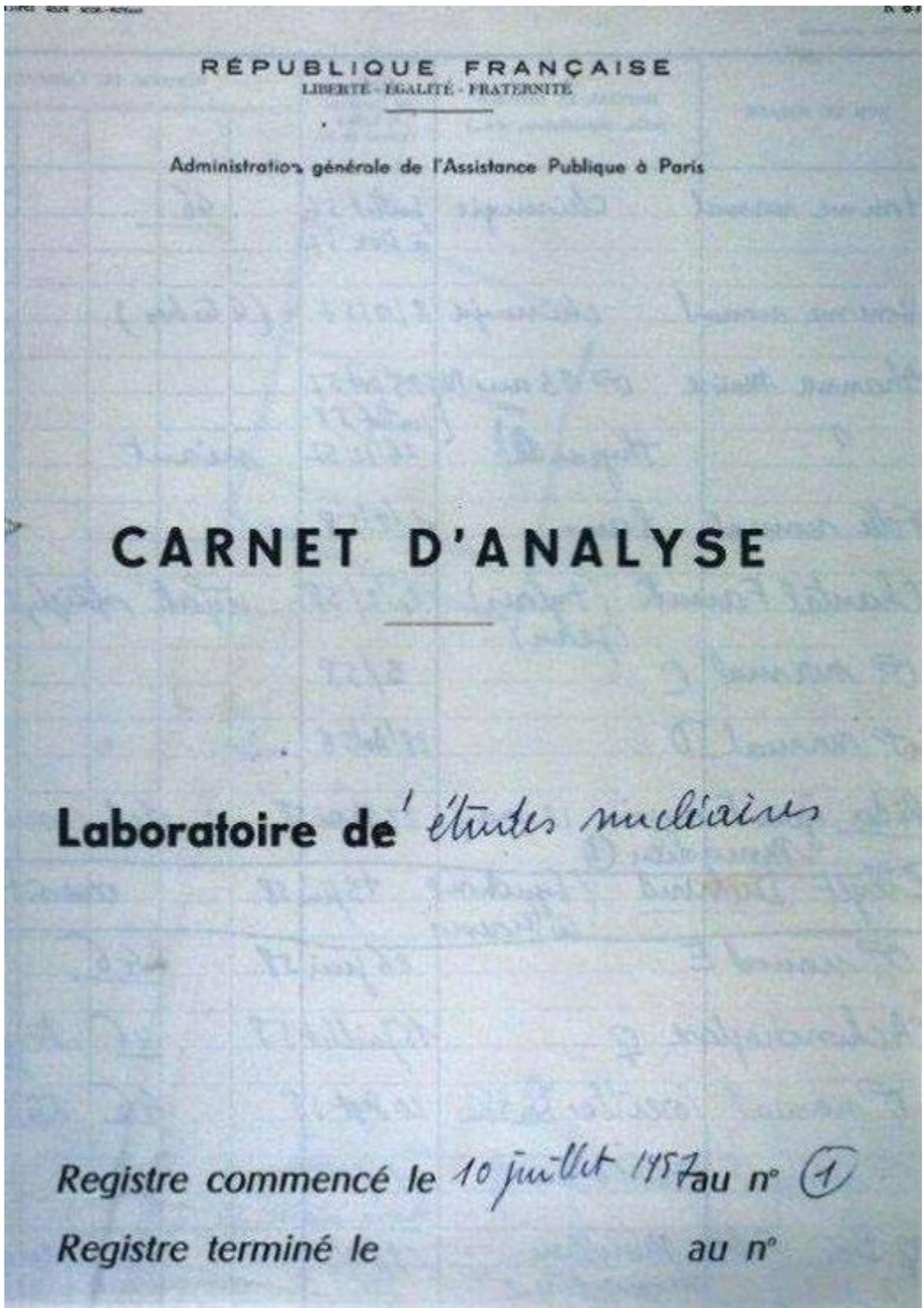
1965

FAIT
Leljeune honore Gaudier dans sa leçon inaugurale.

MYTHE

En 2009, Gaudier déclare que Leljeune s'est attribué la découverte.

Annex 1: Cover of the laboratory notebook kept by Jérôme Lejeune



Annex 2: Page of the laboratory notebook which indicates the first dated observations

N° d'ordre	NOM DU MALADE	HOPITAL ET SERVICE (salle, consultation, etc...)	NATURE de l'analyse et index (valeur en B)	REPONSE DU LABORATOIRE
①	Homme normal	Chirurgie	juillet 57 à Dec 57	46
②	Homme normal	Chirurgie	8/10/57	(4 tubes)
③	(Nom flouté)*	(Symbole mâle) 13 ans N	25 Oct 57	
④	?	Thyroïde	juillet 58 26/12/57	néant
⑤	Fille normale	8 ans	1/2/58	
⑥	(Nom flouté)*	(transcription impossible)** (peau)	4/2/58	(transcription impossible)**
⑦	(Symbole femelle) normal C		3/58	
⑧	(Symbole mâle) normal D		28/Av/58	
⑨	(Nom flouté)* Mongolien 1	(3 mois)	22 Mai 58	1 Chr. surnuméraire
⑩	(Nom flouté)*	(Syndrome intersexué)	13 juin 58	avait 47 chr.
⑪	(Symbole mâle) normal E		26 juin 58	46

N° d'ordre	NOM DU MALADE	HOPITAL ET SERVICE (salle, consultation, etc...)	NATURE de l'analyse et index (valeur en B)	REPONSE DU LABORATOIRE
1	Homme normal	Chirurgie	Juillet 57 à Dec. 57	46
2	Homme normal	Chirurgie	8/10/57	(4 tubes)
3	(Nom flouté)*	(Symbole mâle) 13 ans N	25 Oct. 57 Juillet 58	
4	?	Thyroïde	26/12/1957	Néant
5	Fille normale	8 ans	1/2/58	
6	(Nom flouté)*	(transcription impossible)** (peau)	4/2/58	(transcription impossible)**
7	(Symbole femelle) normal C		3/58	
8	(Symbole mâle) normal D		28/Av/58	
9	(Nom flouté)* Mongolien 1	(3 mois)	22 mai 58	1 Chr. surnuméraire
10	(Nom flouté)*	(Syndrome intersexué)	13 Juin 58	Avait 47 chr.
11	(Symbole mâle) normal E		26 juin 58	46

[page 1/2]
 27-10-58
 L58Turpin1
 S
 S

Mon cher ami,

A l'instant je reçois votre lettre du 22. L'article de Xvitsky et Kimball ne m'a pas échappé, et sur place vous êtes mieux placé pour répondre aux auteurs que nous le serons ici, à votre retour. En outre une riposte prompte est toujours préférable. Si vous êtes en mesure de contre attaquer, n'hésitez pas ; si vous jugez plus sage de reconsidérer vos documents ou d'étudier des faits nouveaux, attendez ; d'ailleurs une argumentation immédiate peut fort bien annoncer des résultats ultérieurs complémentaires. Pour ne pas avoir l'air de vous désolidariser il faut que nous trois nous restions associés. Envoyez-moi alors votre projet ; je le verrai avec Mlle Réthoré.

Deux visiteurs venus me trouver, Cordero Ferreira (pédiatre de Lisbonne) et J. Mohr d'Oslo ont été émerveillés par vos préparations chromosomiques. **Mlle Gautier et Mme Massé en sont toujours à 46.** Deux jumelles de 10 mois, cornitiales, (électro encéphalo identiques) vont nous donner un nouveau matériel.

Quand cette lettre vous parviendra vous n'aurez plus qu'un mois 'à tirer'. Je

27-10-58
 L58Turpin1
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Annex 4: Letter from M. Gautier to J. Lejeune dated 20 Oct. 1958

L. 17950-
20-10-58

page 1/2
Gautier J
page 2/1

Docteur M. GAUTIER
6 rue de Douai
PARIS 10e
Cher ami,

J'ai reçu vos lettres récemment avec un certain retard car elles
étaient fait un stage dans le laboratoire d'une surveillance; Mettez sur
l'adresse = labo Parrot ou mon adresse. Je ne sais si cette lettre
vous parviendra, car peut-être vous avez quitté les lieux
enchantés de la Californie.
J'ai reçu vos lettres récemment avec un certain retard car elles
étaient fait un stage dans le laboratoire d'une surveillance; Mettez sur
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enchantés de la Californie.

Nous avons au récemment de très belles cellules à compléter,
mais toujours au stade de développement chez un
achondroplase. Pour les chr. Flammeux, c'est toujours plus
difficile, vous venez. Toujours d'énormes difficultés pour avoir
des tissus nets. Rien depuis le 15 juin. Je ne suis pas dans le
service le matin et ils sont tellement obscurs que récemment le
patron après un de mes visites à l'ail (légal) deux
hydrocéphales, mais après avoir cherché le tissu
Nous avons au récemment de très belles cellules à compléter,
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hydrocéphales, mais après avoir cherché le tissu

huit jours après puisque nous n'avons pas été prévenues, on les
a retrouvés en bloc chez Bouquet. Votre retour facilitera
l'espérer cette quête de tissus qui devient intolérable. Je m'en
procure des normaux dans d'autres hôpitaux. Rien de pis que la
force à l'essai.
huit jours après puisque nous n'avons pas été prévenues, on les
a retrouvés en bloc chez Bouquet. Votre retour facilitera
l'espérer cette quête de tissus qui devient intolérable. Je m'en
procure des normaux dans d'autres hôpitaux. Rien de pis que la
force à l'essai.

A part cela, les éclairements semblent meilleurs depuis que nous
utilisons le benzo-codécium (genre beven).
A part cela, les éclairements semblent meilleurs depuis que nous
utilisons le benzo-codécium (genre beven).

Donc quelques belles photos en perspective pour l'instant
mais nous n'avancerons pas tant qu'il faudra 3 mois de
patience pour un nouveau tissu.
Donc quelques belles photos en perspective pour l'instant
mais nous n'avancerons pas tant qu'il faudra 3 mois de
patience pour un nouveau tissu.

Fixez tous les luyaux que vous pouvez quant aux cultures de
tissus et tous les fils à part que l'on donne au hasard de vos
visites.
Fixez tous les luyaux que vous pouvez quant aux cultures de
tissus et tous les fils à part que l'on donne au hasard de vos
visites.

Amities.
Amities.

M. Gautier
(signature manuscrite)

20-10-57
LSS Boubal
①

Cher ami,
J'ai reçu vos lettres récemment avec un certain retard car elles
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visites.

Amities.
Amities.

M. Gautier
(signature manuscrite)

Annex 5: Dated summary of titles and research positions of Jérôme Lejeune in an official publication

TITRES UNIVERSITAIRES

- Faculté de Médecine de Paris.
Thèse de Doctorat, juin 1951.
- Faculté des Sciences de Paris.
Certificat de Génétique, 1954.
Certificat de Biochimie, 1955.
Thèse de Doctorat ès Sciences Naturelles, 1960.
- Professeur de Génétique Fondamentale, 1964.
Faculté de Médecine Necker-Enfants Malades.

TITRES HOSPITALIERS

- Biologiste des Hôpitaux. Chef de service à l'Hôpital des Enfants - Malades.
Laboratoire et consultation de Génétique fondamentale, 1964.

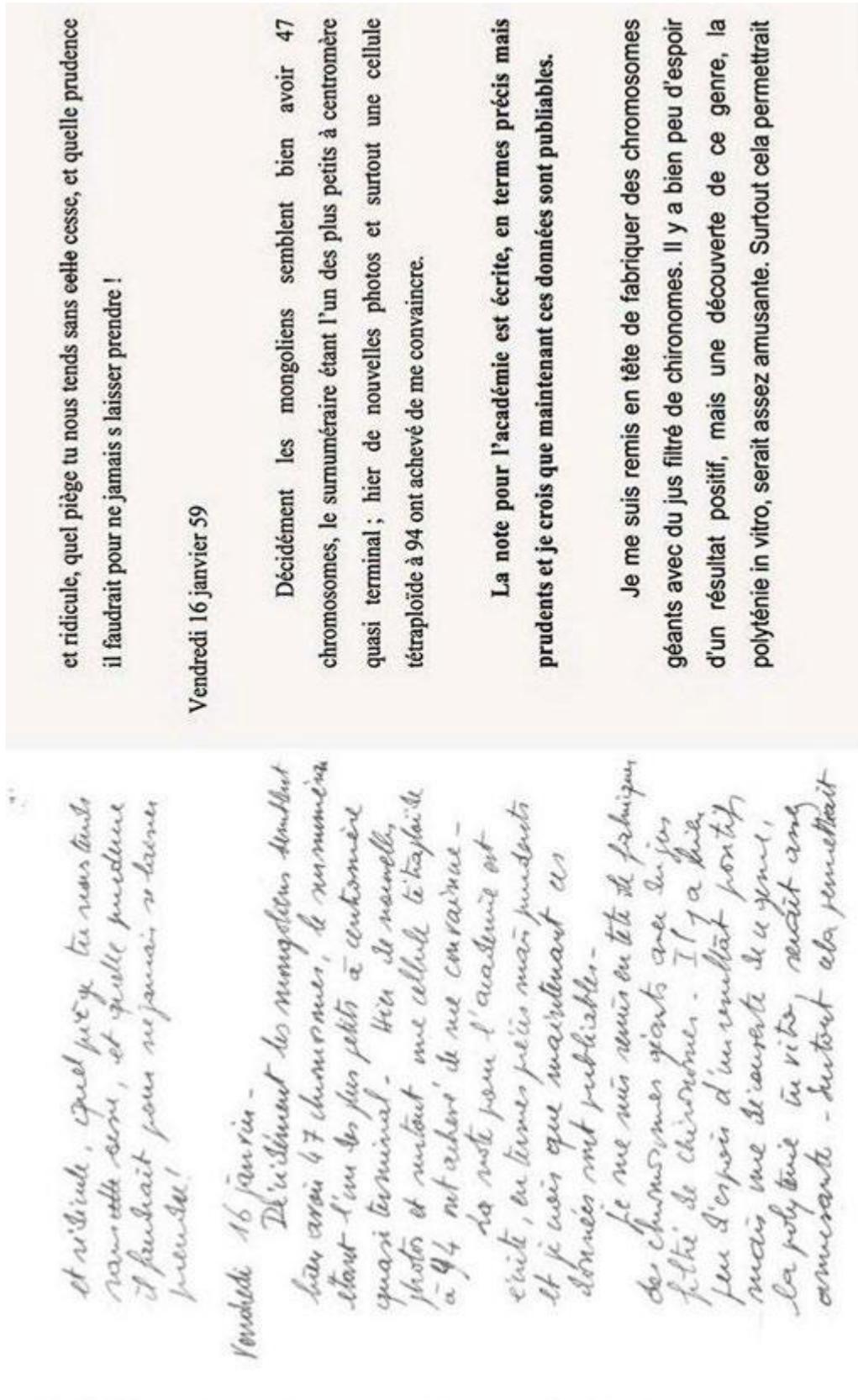
FONCTIONS DE RECHERCHE

Centre National de la Recherche Scientifique

Stagiaire de Recherche,	1952-1954
Attaché	— 1954-1956
Chargé	— 1956-1959
Maitre	— 1959-1962
Directeur	— 1963-1964

- Membre de la Commission de Pathologie expérimentale, Pharmacodynamie et Thérapeutique expérimentale (1960-1968).
- Membre du Comité Consultatif de la Recherche Scientifique et Technique (1965-1968).
- Membre du Conseil Scientifique de l'INSERM depuis 1969.
- Membre du Conseil Scientifique de la Fondation pour la Recherche Médicale Française (1965-1969).
- Membre du Conseil Scientifique de l'Institut Pasteur (1967-1968).
- Directeur exécutif de l'Institut de Progénèse, 1968.

Annex 6: Correspondence from J. Lejeune with his wife and his personal diary, notably of 16 January 1959



se sent soulagé. Si tu consens à le dire, il n'y a plus qu'à l'enregistrer.

Jacques et toi représentez ici les deux aînés recevant leur jeune émule. Et je tiens à te dire que c'est un très profond plaisir que cet appel de noms, voulu par l'usage, soit véritablement le rappel d'une amitié.

Je voudrais maintenant me tourner vers notre troisième collègue, qui, bien que portant robe, ne siège point en cette délégation.

Ma Chère Marthe Gautier,

Vous étiez la seule d'entre nous à connaître les techniques de cultures cellulaires. Ensemble nous avons commencé de lire le caryotype humain. Si je rappelle ces premières années de tâtonnements, en 1957 et 1958, c'est pour dire que ces deux années d'échecs et de demi-succès n'ont été couronnées enfin que grâce à votre habileté et à votre tenacité.

Cardiologue d'enfants réputé, malgré toutes vos occupations vous n'avez point abandonné l'équipe, et nous nous retrouvons encore chaque lundi pour discuter de la semaine passée et prévoir la semaine à venir. C'est avec grand plaisir que je vous dis du fond du cœur ma très affectueuse gratitude.

Nos deux collègues plus jeunes animent le laboratoire, l'un de la fougue de son intelligence et l'autre de la perspicacité de son dévouement.

Ma Chère Marie-Odile Rethoré,

Vous l'avez deviné, le dévouement c'est vous et la per-