DIPARTIMENTO DI MEDICINA SPECIALISTICA,
DIAGNOSTICA E SPERIMENTALE

Subject: Research on trisomy 21

Bologna, 2 October 2021

Down Syndrome (DS) is the most frequent human chromosomal disorder, with a frequency of 1 in ~700 births. People with DS have **intellectual disability**, which is present to some degree of severity in all affected individuals and which involves symbolic thought, whereas affectivity and social skills are conserved. It is common for children with DS to arouse a climate of affective intensity greater than the norm.

Although it is a syndrome observed since ancient times, the cause was not known until the '50s of the last century, and instead it was attributed to a "degeneration of the race" as a consequence of supposed "fault" of parents (alcoholism, syphilis). In 1959, the young French doctor Jérôme Lejeune identified the cause of DS as the presence of three copies of human chromosome 21 (Hsa21), instead of the normal two, in the cells of the affected individuals. This condition has been called trisomy 21.

This discovery introduced the notion that a given clinical symptom may be connected to a specific alteration of the human genetic material for the first time, giving origin to the field of **medical genetics**: Prof. Lejeune held the first Chair of Genetics at University of Paris. Moreover, the syndrome is expected to be associated with a spontaneous and unpredictable genetic mutation, with a constant frequency in all populations of the Earth, losing any negative "moral" connotation; the derogatory term "mongolism" was abandoned in favor of "Down Syndrome" or "trisomy 21". Studyng in detail the mechanism of the syndrome becomes possible, in particular how the presence of an extra chromosome 21 can be associated with the symptoms, in view of a possible pharmacological intervention as a therapy for trisomic subjects. Prof. Lejeune firmly believed in the possibility to find a **therapy**, currently investigated by a limited number of research groups.

Our research group aims to systematically study possible **genotype-phenotype relationships in DS** in order to achieve fully understand the genetic mechanisms of the syndrome to therefore identify therapy targets. In the last years, we have identified a new gene not identified in the original report of the complete sequence of chromosome 21, we have performed a large-scale analysis of the structure and expression of chromosome 21, we have designed and developed bioinformatics tools able to process the original information on the structure and expression of genes and genomes. On April 22nd 2016, our paper about the “critical region” for Down syndrome was published in the journal *Human Molecular Genetics*. The study, confirmed by a subsequent article published in 2019, suggests that a “critical region” responsible for the main symptoms of Down syndrome corresponds to only one thousandth of the whole chromosome 21.

The **research project** downloadable from this same website shows the results of international collaboration obtained by our group and details our ideas for further research on trisomy 21. Funding for this research was influenced, on the one hand, by the limited availability of funds for the local experimental research, and on the other, by the intense efforts toward prenatal diagnosis oriented to selective abortion rather than toward basic and applied research aimed at finding effective therapies. For this reason, every contribution is crucial to support our research activities of the Laboratory.

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To make a donation online for our project, you can use this link (up to 1,999 Euros):
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I sincerely thank you very much for your attention, and I am available for any clarification, further information or just to meet, if you wish.

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