

# Bioinformatics reveals severe tumor mutations in children

Di Wynne Dinwiddie

Thanks to bioinformatics, some rare genetic variants have been identified that predispose to neuroblastoma, a malignant tumor of the nervous system that affects about 15,000 children and adolescents worldwide every year, more than a hundred in Italy, and is considered the first cause of death and the third tumor in frequency after leukemia. Blood and brain tumors in childhood. The result, which opens new horizons for early diagnosis and personalized treatment of the disease, is published in the journal *eBioMedicine* by researchers led by Mario Capasso and Achille Iolascon del Ceinge, respectively, Associate Professor and Federico II Professor of Medical Genetics at the University of Naples.

The research, which was funded by Open Onlus, the Italian Neuroblastoma Foundation and the Airc Foundation for Cancer Research, is based on the analysis of the largest case series studied to date. All genetic data has been made available in an online database so that other scientists can refer to it freely to develop new research.

“We analyzed the DNA of nearly 700 children with neuroblastoma and more than 800 controls using advanced sequencing, an innovative technology that can reliably and rapidly decode all genes known to date,” Capasso explains. “This is the largest number of cases studied to date as we have found that 12% of children with neuroblastoma have at least one inherited genetic mutation that increases the risk of cancer.”

The study was made possible by advanced computational analyzes conducted by a team of experts working at the next-generation sequencing bioinformatics facility in Ceinge. In particular, these are the investigations carried out by bioinformatics expert Ferdinando Bonfiglio, first author of the work.

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“The results of this research have relevant clinical implications,” adds Iolascon. “They are useful for improving diagnosis, making it more early and certain than ever before, and for improving clinical management of the patient by guiding the clinician toward the use of personalized treatments.”

Another interesting fact that emerged from the research, Capasso asserts, “is that some of the mutations found in these children are also associated with the development of neurodevelopmental diseases, such as autism spectrum disorders. The results achieved are also useful for a better

understanding of the mechanisms and molecules that underlie the development of non-cancerous diseases.”

[Bioinformatics reveals severe tumor mutations in children \(wireservice.ca\)](https://wireservice.ca)

The screenshot shows a news article on the WIRE SERVICE CANADA website. The article is titled "Bioinformatics reveals severe tumor mutations in children" and is categorized under "SCIENCE". It is written by Wynne Dwyll and was published 2 days ago, with a 2-minute read time. The article features a microscopic image of a neuroblastoma tumor. The text explains that bioinformatics has identified rare genetic variants that predispose to neuroblastoma, a malignant tumor of the nervous system that affects about 15,000 children and adolescents worldwide every year, more than a hundred in Italy, and is considered the first cause of death and the third tumor in frequency after leukemia, blood and brain tumors in childhood. The result, which opens new horizons for early diagnosis and personalized treatment of the disease, is published in the journal *Cell* and is the work of researchers led by Mario Capasso and Achille Iolascon del Ceinge, respectively, Associate Professor and Federico II Professor of Medical Genetics at the University of Naples. The research, which was funded by Open Onlus, the Italian Neuroblastoma Foundation and the AirC Foundation for Cancer Research, is based on the analysis of the largest case series studied to date. All genetic data has been made available in an online database so that other scientists can refer to it freely to develop new research. A quote from the article states: "We analyzed the DNA of nearly 700 children with neuroblastoma and more than 800 controls using advanced sequencing, an innovative technology that can reliably and rapidly decode all genes known to date," Capasso explained. "This is the largest number of cases studied to date as we have found that 13% of children with neuroblastoma have at least one inherited genetic mutation that increases the risk of cancer".

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