

Early Detection of Metachromatic Leukodystrophy: A Life-Saving Newborn Test in Campania

A new neonatal test for the early diagnosis of metachromatic leukodystrophy (MLD) has been launched in the Campania Region, a rare and severe progressive neurodegenerative genetic disease characterized by rapid and serious deterioration of motor and cognitive functions. Thanks to a pilot project active at CEINGE, the Center for Genetic Engineering and the only Regional Hub for neonatal screening, the regional health system now offers a new life-saving prevention opportunity with the introduction of this new test. Rare diseases and tumors, experts say: "Here, a model of innovation." The effectiveness of the latest generation gene therapies currently available depends solely on early diagnosis: the greatest benefits are achieved when the child is identified and treated before the onset of symptoms and irreversible neurological damage. Recognizing the disease before symptoms appear can therefore allow access to gene therapy at stages when it offers the best results. Screening for MLD, which is not mandatory and not included in the Essential Levels of Care (LEA), is being offered to newborns in Campania as part of a three-year project. Participation is voluntary and requires parents to sign informed consent in the hospital. No additional blood draws are needed: the test uses the same drops of blood already taken from the newborn's heel, between 48 and 72 hours of life, for routine screenings and collected on the Guthrie card. "With the new test, which puts Campania at the forefront of early diagnosis for MLD, we can change the future of affected children. The timeliness of diagnosis can save lives and reduce the effects of the disease on development because it allows for prompt specialist evaluation for therapy access, before irreversible neurological damage occurs," commented President Roberto Fico. This project strengthens the screening pathway in the Campania Region, adding an additional opportunity for early detection of the disease to the existing tests. "The initiative is part of a progressive enrichment path that already sees us engaged in other diseases not included in the LEA, such as lysosomal diseases," explains Mariano Giustino, CEO of CEINGE. "All identified children have been treated promptly." Metachromatic leukodystrophy is a rare, progressive, neurodegenerative genetic disease, with an estimated incidence ranging from one case per 40,000 to one per 160,000 live births. It is caused by a deficiency of the enzyme arylsulfatase A, which leads to the accumulation of substances called sulfatides and consequent damage to the nervous system. In the earliest forms, it can cause rapid loss of motor and cognitive abilities, with very serious consequences for the child's life. Today, a gene therapy authorized in the European Union is available for specific forms and stages of the disease, capable of modifying its course. The greatest benefits are observed in children treated before the onset of symptoms. The new test is part of the regional neonatal screening network, with CEINGE as its sole reference center. The Center already screens all newborns in Campania for the 48 diseases required by law 167/2016 and included in the 2017 LEA. The pilot project is funded by Orchard Therapeutics and has been approved by the Ethics Committee of the University of Naples, protocol 309/2024. The goal is to offer all newborns in the region an additional opportunity for early diagnosis and help reduce inequalities in access to care.

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Il Ceinge, il centro che ha attivato il progetto

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