Genespire Commences Enrollment in International Observational Study, InforMMA, for Patients with Methylmalonic Acidemia

- **The Italian biotech will work with study sites to collect a large dataset detailing the lives of patients with MMA.**
- **Data from InforMMA will help inform Genespire during the development of GENE202, the company's in-development ISLV-based asset for MMA.**
- **The study will take place at world-leading institutions including London's Great Ormond Street Hospital and Milan's Ospedale San Raffaele.**

MILAN, Italy, 25 August 2026 -- Genespire, an Italian biotechnology company focused on developing gene therapies for severe pediatric genetic diseases, has today announced the commencement of patient enrolment in a prospective, multicenter observational study, InforMMA, in children with methylmalonic acidemia (MMA) caused by mutations in the MMUT gene.

The study is designed to follow patients aged up to 16 years who live with severe MMA, with the aim of better characterizing disease progression over time. It will enrol patients both with and without prior liver or kidney transplantation; however, both groups must be assessed as having the "severe" MMA phenotype.

By conducting longitudinal measurements of specific biochemical markers and clinical outcomes linked to metabolic stability and disease trajectory over a 36-month period from baseline, Genespire hopes to deepen the understanding of MMUT-associated MMA. This research will also support the ongoing development of GENE202, the company's immune-shielded lentiviral vector gene therapy designed specifically for MMA.

The study (ClinicalTrials.gov Identifier: NCT07432880/InforMMA) will enrol patients at a total of 7 sites in the US and Europe, all of which are renowned metabolic or pediatric centers of excellence: Children's Hospital of Philadelphia and UPMC Children's Hospital of Pittsburgh in the US; Ospedale San Raffaele and Ospedale Pediatrico

Bambino Gesù in Italy; Hospital Sant Joan de Déu in Barcelona and Hospital Universitario 12 de Octubre in Madrid, Spain; and Great Ormond Street Hospital in the UK.

“Observational studies like this require time, resources and close collaboration with the wider community – but they are fundamental to innovation in rare disease,” said Lucia Faccio, CEO of Genespire. “By investing early in our understanding of MMA, we aim to optimize the later stages of development for GENE202.”

“For far too long, patients with MMA have been underserved by healthcare systems and underrepresented in research, leaving substantial gaps in our understanding of disease progression over time,” added Dr Maddalena Migliavacca, principal investigator at InforMMA’s Ospedale San Raffaele site. “This study represents an important step towards developing a more comprehensive understanding of the real-world patient experience and, ultimately, improving outcomes for patients and their families.”

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About Genespire

Genespire is a biotechnology company, developing off-the-shelf gene therapies based on immune shielded lentiviral vectors (ISLVs) for pediatric patients affected by genetic diseases. ISLVs are designed to be used intravenously and allow the life-long production of the therapy directly from the patient’s liver. Genespire is initially advancing therapeutic programs in inherited metabolic diseases with high unmet medical need. Based in Milan, Italy, Genespire was founded in March 2020 by the gene therapy pioneer Prof. Luigi Naldini and Dr. Alessio Cantore, the Fondazione Telethon, and Ospedale San Raffaele. Genespire is a spin-out of SR-Tiget, a world leading cell and gene therapy research institute. Find out more about us at www.genespire.com.

About Methylmalonic Acidemia (MMA)

MMA is a rare genetic metabolic disorder most frequently caused by a faulty gene coding for the mitochondrial enzyme methylmalonyl-CoA mutase (MUT). People with this condition are unable to break down and use certain proteins and fats found in food and, as a result, circulating methylmalonic acid accumulates in the body, causing damage to the brain, liver, kidneys, and other organs. At present there are no disease-targeted drugs approved for MMA, and affected patients suffer high levels of morbidity and have a heavily reduced life expectancy.