

Lopende onderzoeken in 2025

	Titel onderzoek	Arts/onderzoeker, UMC	Toekenning (in €)	Beoogde Looptijd	Startdatum
1.	Nederlandse App Suite ter ondersteuning van het metabool dieet voor erfelijke stofwisselingsziekten	dr. C. van Karnebeek/ Amsterdam UMC	24.000	5 jaar	1-9-2018
2.	Turning dazzling data into patient solutions – generation of a UMD omics platform for IEM biomarker discovery	dr. D. Lefeber, dr. F. Vaz, dr. J. Jans/ Radboud UMC, Amsterdam UMC, UMC Utrecht	205.204	2 jaar	1-11-2020
3.	Metabolic flux studies in humans by means of stable isotopes	dr. F. Vaz/ Amsterdam UMC	182.332	2 jaar	1-2-2021
4.	New ways, better outcomes: n-of-1 for all	dr. M. Brands, dr. C. van Karnebeek/ Amsterdam UMC	175.684	4 jaar	1-9-2020
5.	Deelprojecten A Bird's Eye View of Gyrate Atrophie	dr. H. Waterham/ Amsterdam UMC	25.000	30 mnd	1-10-2020
6.	CaseDatabase for Inherited Metabolic Disorders (CBD-IMD)	dr. I. Körver Keularts/dr. K.E. Niezen-Koning Maastricht UMC+/ Groningen UMC	50.000	2 jaar, met optie op verlenging in jaar 3	17-12-2021
7.	Hematopoietic stem cell-mediated lentiviral gene therapy for the treatment of patients with Mucopolysaccharidosis type II (MPS II; Hunter syndrome)	dr. P. Pijnappel	338.500	3 jaar	1-3-2022
8.	Cell-permeable glycerol-3-phosphates (G3Ps) as a novel treatment for Leigh Syndrome	dr. W. Koopman, dr. T. Schirris/Radboud UMC	50.000	6 maanden	1-5-2023

9.	Gene editing for correcting mitochondrial mutations and modelling mitochondrial disease	dr. S. Fuchs, drs. M. Koppens/Utrecht UMC	150.000	2 Jaar	1-1-2023
10.	Towards a Prognostic Test for Risk Stratification for Patient-specific Disease Manifestations in Adrenoleukodystrophy (PRISM-ALD)	dr. S. Kemp/Amsterdam UMC	319.820	4 jaar	1 november 2023
11.	New biomarkers for Pompe disease to support newborn screening and to monitor disease progression and effect of (innovative) therapy.	dr. G. Ruijter/Erasmus MC	316.549	4 jaar	1 januari 2024
12.	The use of machine learning to increasing the positive predictive value of newborn screening	dr. M. de Sain – van der Velden/UMC Utrecht	141.231	1,5 jaar	1 september 2024
13.	Improving newbornscreening for primary carnitine deficiency; more health, less anxiety	dr. M. Langeveld /Amsterdam UMC	307.640	3,5 jaar	1 januari 2024
14.	Boosting Residual Enzymatic Activity as a potential therapeutic strategy for Inherited Metabolic Diseases	Dr. A. Garanto/ Radboud UMC	319.975	4 jaar	1 maart 2025

15.	Ex vivo lentiviral gene therapy for late-infantile neuronal ceroid lipofuscinosis (CLN2 disease): from bench to bedside	Dr. H. Huidekoper/ Erasmus MC	320.000	4 jaar	31 maart 2025
16.	Advancing Lentiviral Gene Therapy: Tackling Metabolic Disorders caused by Non-Soluble, Non-Secreted Gene Products Deficiencies	Dr. F. Catalano/ Erasmus MC	320.000	3 jaar	31 januari 2025