

Trials clinici per malattie metaboliche ereditarie in fase di arruolamento o prossimi all'arruolamento, aperti in Italia

Aggiornamento 31/03/2025

Malattia	Clinical trial number	Titolo	Centro	Principal Investigator	Contatto	Email/Tel	Fase	Recruiting /not yet recruiting	Pediatrico /adulto	Link
M Pompe LOPD	NCT06150820	<i>A Study About Antibody Levels and Biomarkers in the Blood in People With Late-onset Pompe Disease</i>	Gussago, Messina, Milano, Pavia, Roma				NA	Recruiting	Adulto	https://clinicaltrials.gov/study/NCT06150820?locStr=Italy&country=Italy&cond=Pompe&aggFilters=status:rec,studyType:int&rank=1#locations
M Pompe LOPD	NCT03911505	<i>ZIP Study-OL Study of Safety, PK, Efficacy, PD, Immunogenicity of ATB200/AT2221 in Pediatrics Aged 0 to < 18 y.o. w/LOPD</i>	Monza San Gerardo	Serena Gasperini	Serena Gasperini	serena.gasperini@irccs-sangerardo.it	Fase 3	Recruiting	Pediatrico	https://clinicaltrials.gov/study/NCT03911505?locStr=Italy&country=Italy&cond=Pompe&aggFilters=status:rec,studyType:int&rank=2
M Pompe LOPD	NCT05951790	<i>Inspiratory Muscle Training (IMT) in Adult People With Pompe Disease</i>	Milano IRCCS S.Maria Nascente – Fondazione Don Carlo Gnocchi		Paolo Banfi	Tel 0240308812 pabanfi@dongnocchi.it	NA	Recruiting	Adulto	https://clinicaltrials.gov/study/NCT05951790?cond=NCT05951790&rank=1
M Pompe IOPD	NCT04808505	<i>A Study to Evaluate the Safety, Efficacy, PK, PD and Immunogenicity of Cipaglucosidase Alfa/Miglustat in IOPD Subjects Aged 0 to <18 (ROSSELLA)</i>	Napoli Federico II, Torino	Giancarlo Parenti	Giancarlo Parenti	parenti@unina.it ;	Fase 3	Not yet recruiting	Pediatrico	https://clinicaltrials.gov/study/NCT04808505?locStr=Italy&country=Italy&cond=Pompe%20Disease&aggFilters=status:not%20rec,studyType:int&rank=5
Mitocondriopatie	NCT06044194	<i>Evaluation of the Effect of Liposomal L-Arg and Vit C Integration on Mitochondrial Function in Patients With Heart Failure</i>	Milano IRCCS Ca Granda		Steano Carugo	0255033579 , stefano.carugo@policlinico.mi.it	Fase 3	Recruiting	Adulto	https://clinicaltrials.gov/study/NCT06044194?cond=NCT06044194&rank=1
MPS II	NCT04573023	<i>A phase III study of JR-141 in mucopolysaccharidosis type II (Hunter syndrome) patients</i>	Monza	Serena Gasperini	Serena Gasperini	serena.gasperini@irccs-sangerardo.it	fase 3	recruiting	Pediatrico	https://clinicaltrials.gov/study/NCT04573023?cond=NCT04573023&rank=1
MPS II	NCT04573023	<i>A phase III study of JR-141 in mucopolysaccharidosis type II (Hunter syndrome) patients</i>	Roma, Ospedale pediatrico Bambino Gesù	Federica Deodato	Federica Deodato	federica.deodato@opbg.net	fase 3	recruiting	Pediatrico	https://clinicaltrials.gov/study/NCT04573023?cond=NCT04573023&rank=1
MPSII	NCT05371613	<i>A Study to Determine the Efficacy and Safety of Tividenofusp Alfa (DNL310) vs Idursulfase in Pediatric Participants With Neuronopathic (nMPS II) or Non-Neuronopathic Mucopolysaccharidosis Type II (nnMPS II) A 24-Month, Multi-Centre, Open Label Phase IV Post Authorisation Efficacy Study to Evaluate the Efficacy, Safety and Immunogenicity of Daily Subcutaneous Metreleptin Treatment in Patients with Partial Lipodystrophy</i>	Udine Santa Maria della Misericordia	Maurizio Scarpa	Maurizio Scarpa; A	maurizio.scarpa@asufc.sanita.fvg.it	Fase 2/3	Recruiting	Pediatrico	https://clinicaltrials.gov/study/NCT05371613?cond=NCT05371613&rank=1
Partial Lipodystrophy	NCT05164341	<i>A 24-Month, Multi-Centre, Open Label Phase IV Post Authorisation Efficacy Study to Evaluate the Efficacy, Safety and Immunogenicity of Daily Subcutaneous Metreleptin Treatment in Patients with Partial Lipodystrophy</i>	Udine Santa Maria della Misericordia	Annalisa Sechi	Annalisa Sechi, Enrica Epifani (Udine)	annalisa.sechi@asufc.sanita.fvg.it , enrica.epifani@asufc.sanita.fvg.it	Fase4	Not yet recruiting	Adulto	https://clinicaltrials.gov/study/NCT05164341?cond=NCT05164341&rank=1
PKU	NCT05166161	<i>A Long-Term Safety Study of PTC923 in Participants With Phenylketonuria</i>	Padova Azienda Ospedaliera	Alberto Burlina	Alberto Burlina	alberto.burlina@unipd.it ;	Fase 3	Recruiting	Pediatrico/adulto	https://clinicaltrials.gov/study/NCT05166161?cond=NCT05166161&rank=1
PKU	NCT05166161	<i>A Long-Term Safety Study of PTC923 in Participants With Phenylketonuria</i>	Roma Umberto I	Filippo Manti	Filippo Manti	filippo.manti@uniroma1.it	Fase 3	Recruiting	Pediatrico/adulto	https://clinicaltrials.gov/study/NCT05166161?cond=NCT05166161&rank=1
PKU	NCT05827536	<i>Open-label, Randomized, 2-way Crossover, Monocentric, Controlled Study to Evaluate the Effect on Daily PHE Fluctuation of PKU GOLIKE Versus SoC in Patients With PKU.</i>	Milano ASST Santi Paolo e Carlo, Ospedale San Paolo	Valentina Rovelli	Valentina Rovelli	02811844554 , valentina.rovelli@asst-santipaolocarlo.it		Recruiting	Pediatrico/adulto	https://clinicaltrials.gov/study/NCT05827536?cond=NCT05827536&rank=1