Door VWS erkende expertisecentra voor stofwisselingsziekten v.a. december 2022

Ziekenhuis	Naam EC (EN)	Aandoening	Orphacode
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Cholesteryl ester storage disease	ORPHA:75234
Amsterdam UMC	Amsterdam UMC Expert Center for immune-mediated and genetic cholestasis syndromes	Crigler-Najjar syndrome	ORPHA:205
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Disorder of carnitine cysle and carnitine transport	ORPHA:309130
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Disorder of fatty acid oxidation and ketone body metabolism	ORPHA:79174
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Disorder of pyridoxine metabolism	ORPHA:79192
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Galactosemia	ORPHA:352
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Gyrate atrophy of choroid and retina	ORPHA:414
Amsterdam UMC	Amsterdam UMC Leukodystrophy Center	Leukodystrophies	ORPHA:68356
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Lysosomal disease	ORPHA:68366
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Peroxisomal disease	ORPHA:68373
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Phenylketonuria	ORPHA:716
Amsterdam UMC	Amsterdam UMC expertise center for rare pediatric kidney and urinary tract diseases	Primary Hyperoxaluria	ORPHA:416
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Rare dyslipidemia	ORPHA:101953
Amsterdam UMC	Amsterdam UMC Expert Center for Inborn Errors of Metabolism	Rare inborn errors of metabolism	ORPHA:68367
Erasmus MC	The ENCORE expertise center for neurodevelopmental disorders	Cockayne Syndrome	ORPHA:191
Erasmus MC	Center for Lysosomal and Metabolic Diseases	Crigler-Najjar syndrome	ORPHA:205

Erasmus MC	Center for Lysosomal and Metabolic Diseases	disorder of amino acid and other organic acid metabolism	ORPHA:79062
Erasmus MC	Center for Lysosomal and Metabolic Diseases	disorder of branched-chain amino acid metabolism	ORPHA:79197
Erasmus MC	Center for Lysosomal and Metabolic Diseases	disorder of lysosomal amino acid transport	ORPHA:79207
Erasmus MC	Center for Lysosomal and Metabolic Diseases	disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
Erasmus MC	Center for Lysosomal and Metabolic Diseases	glycogen storage disease due LAMP2 deficiency	ORPHA:34587
Erasmus MC	Center for Lysosomal and Metabolic Diseases	glycogen storage disease due to acid maltase deficiency	ORPHA:365
Erasmus MC	Center for Lysosomal and Metabolic Diseases	glycoproteinosis	ORPHA:309279
Erasmus MC	Center for Lysosomal and Metabolic Diseases	Lysosomal disease	ORPHA:68366
Erasmus MC	Center for Lysosomal and Metabolic Diseases	mucopolysaccharidosis	ORPHA:79213
Erasmus MC	Center for Lysosomal and Metabolic Diseases	neuronal ceroid lipofucsinosis	ORPHA:216
Erasmus MC	Center for Lysosomal and Metabolic Diseases	organic aciduria	ORPHA:289899
Erasmus MC	Center for Lysosomal and Metabolic Diseases	Porphyria	ORPHA:738
Erasmus MC	Center for Lysosomal and Metabolic Diseases	rare inborn errors of metabolism	ORPHA:68367
Maastricht UMC+	Maastricht UMC+ Center of Expertise for Inborn Errors of Galactose Metabolism	Classic galactosemia	ORPHA:79239
Maastricht UMC+	Maastricht UMC+ Center of Expertise for Inborn Errors of Fructose Metabolism	Disorder of fructose metabolism	ORPHA:308463
Maastricht UMC+	Maastricht UMC+ Center of Expertise for Inborn Errors of Galactose Metabolism	Disorder of galactose metabolism	ORPHA:308467
Maastricht UMC+	Maastricht UMC+ Center of Expertise for Inborn Errors of Fructose Metabolism	Fructose-1,6-biphosphatasedeficiency	ORPHA:348
Maastricht UMC+	Maastricht UMC+ Center of Expertise for Inborn Errors of Galactose Metabolism	Galactokinase deficiency	ORPHA:79237
Maastricht UMC+	Maastricht UMC+ Center of Expertise for Inborn Errors of Galactose Metabolism	Galactose epimerase deficiency	ORPHA:79238

Maastricht UMC+	Maastricht UMC+ Center of Expertise for Inborn Errors of Fructose Metabolism	Hereditary fructose intolerance	ORPHA:469
Radboudumc	Radboudumc Center of Expertise for Inherited metabolic disorders	Congenital disorder of glycosylation	ORPHA:137
Radboudumc	Radboudumc Center of Expertise for Iron disorders	Disorder of iron metabolism and transport	ORPHA:309842
Radboudumc	Radboudumc Expertisecentrum voor Zeldzame nierziekten	Nephropathy sec. to a storage/ other metabolic disease	ORPHA:93593
Radboudumc	Radboudumc Center of Expertise for Inherited metabolic disorders	Disorders of pyridoxine metabolism	ORPHA:79192
Radboudumc	Radboudumc Center of Expertise for Immunodeficiency and autoinflammation	Mevalonate kinase deficiency	ORPHA:309025
Radboudumc	Radboudumc Center of Expertise for Inherited metabolic disorders	Mitochondrial disease	ORPHA:68380
UMC Groningen	UMCG Center of Expertise for Defects in Amino Acid and Organic Acid Metabolism	Disorder of phenylalanine metabolism	ORPHA:284814
UMC Groningen	UMCG Center of Expertise for Carbohydrate, Fatty Acid Oxidation and Ketone Bodies Disorders	Glycogen storage disease	ORPHA:79201
UMC Groningen	UMCG Center of Expertise for Carbohydrate, Fatty Acid Oxidation and Ketone Bodies Disorders	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia	ORPHA:79258
UMC Groningen	UMCG Center of Expertise for Carbohydrate, Fatty Acid Oxidation and Ketone Bodies Disorders	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	ORPHA:79259
UMC Groningen	UMCG Center of Expertise for Carbohydrate, Fatty Acid Oxidation and Ketone Bodies Disorders	Glycogen storage disease due to glycogen debranching enzyme deficiency	ORPHA:366
UMC Groningen	UMCG Center of Expertise for Carbohydrate, Fatty Acid Oxidation and Ketone Bodies Disorders	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
UMC Groningen	UMCG Center of Expertise for Defects in Amino Acid and Organic Acid Metabolism	Molybdenum cofactor deficientie type A	ORPHA:308386
UMC Groningen	UMCG Expertise Center for Movement Disorders Groningen	Neurodegeneration with brain iron accumulation	ORPHA:385
UMC Groningen	UMCG Expertise Center for Movement Disorders Groningen	Neurometabolic disease	ORPHA:68385
UMC Groningen	UMCG Expertise Center for Movement Disorders Groningen	Pantothenate kinase-associated neurodegeneration	ORPHA:157850
UMC Groningen	UMCG Center of Expertise for Defects in Amino Acid and Organic Acid Metabolism	Tyrosinemia type 1	ORPHA:882
UMC Groningen	UMCG Center of Expertise for Hypophosphatemic Rickets	X-linked hypophosphatemia	ORPHA:89936

UMC Utrecht	UMC Utrecht Expertise Centre for inherited metabolic diseases (CIMD-UMCU)	Disorder of carnitine cycle and transport	ORPHA:309130
UMC Utrecht	UMC Utrecht Expertise Centre for inherited metabolic diseases (CIMD-UMCU)	Disorder of fatty acid oxidation and ketone body metabolism	ORPHA:79174
UMC Utrecht	UMC Utrecht Expertise Centre for inherited metabolic diseases (CIMD-UMCU)	Disorder of folate metabolism and transport	ORPHA:285657
UMC Utrecht	UMC Utrecht Expertise Centre for inherited metabolic diseases (CIMD-UMCU)	Disorder of purine or pyrimidine metabolism	ORPHA:79224
UMC Utrecht	Sylvia Toth Center for Multi-disciplinary follow up of Lysosomal Storage Disorders, University Medical Center Utrecht	Hurler disease	ORPHA:93473
UMC Utrecht	UMC Utrecht Expertise Center for rare GI and hepatic diseases	Wilson disease	ORPHA:905