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

| Ziekenhuis | Naam EC (EN) | Aandoening | Orphacode |
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| 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 |

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| 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 |

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| 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 |
| CWZ Nijmegen | Center for Cerebrotendinous xanthomatosis | Cerebrotendinous xanthomatosis | ORPHA:909 |
| 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 |

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| 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 |