Ziektebeelden - Stofwisselingsziekten

	Indeling RIZIV	OMIM
	A. disorders of amino acid metabolism	
1	classical phenylketonuria and hyperphenylalaninemia	261600
2	phenylketonuria due to PTPS deficiency	261640
3	phenylketonuria due to DHPR deficiency	261630
4	phenylketonuria due to PCD deficiency	264070
5	DOPA-responsive dystonia (TH, SPR, GCH1)	128230
6	leucinose, maple syrup urine disease (MSUD)	248600
7	tyrosinemia type 1	276700
8	tyrosinemia type 2	276600
9	tyrosinemia type 3	276710
10	alkaptonuria	203500
11	homocystinuria, B6 responsive and non responsive	236200
12	homocystinuria due to MTHFR deficiency	236250
13	homocystinuria-megaloblastic anemia Cbl E & G type	236270-250940
14	methionine S-adenosyltransferase deficiency	250850
15	glycine N-methyltransferase deficiency	606664
16	S-adenosylhomocystine hydrolase deficiency	180960
17	hyperammonemia due to CPS deficiency	237300
18	hyperammonemia due to OTC deficiency	311250
19	citrullinemia type I	215700
20	citrullinemia type II	605814-603471
21	argininosuccinic aciduria (ASL deficiency)	207900
22	argininemia (arginase deficiency)	207800
23	hyperammonemia due to NAGS deficiency	237310
24	hyperornithinemia, hyperammonemia, homocitrullinuria (HHH)	238970
25	lysinuric protein intolerance	222700
26	gyrate atrophy, B6 responsive or non responsive	258870
27	hyperlysinemia (alpha-aminoadipic semialdehyde synthase deficiency)	238700
28	non ketotic hyperglycinaemia	238300-330-310
29	hartnup disorder	234500
30	Disorders of serine metabolism	601815-172480
31	Disorders of proline/hydroxyproline/pyrroline 5-carboxylate metabolism	239500-239510-237000-612652-179035
32	hypotonia-cystinuria	606407
33	Lowe oculocerebral syndrome	309000
	B. organic acidemias	
34	methylmalonic aciduria (CblA,B,C,D,F)	251100-251110-277400-277410-277380
35	methylmalonic aciduria mutase deficiency type	251000
36	transcobalamin 2 deficiency	275350
37	propionic acidemia	232000
38	isovaleric aciduria	243500
39	3-methylcrotonylglycinuria	210200
40	3 methylglutaconic aciduria type 1, 2, 3, 4, 5	250950-302060-258501-610198
41	HMG CoA lyase deficiency	246450
42	glutaric aciduria type 1	231670
43	glutaric aciduria type 2 (MADD)	305950
44	L-2 and D-2-OH glutaric aciduria	600721-236792
45	4-hydroxybutyric aciduria (SSADH deficiency)	271980
46	isobutyryl CoA dehydrogenase deficiency (ACAD8)	611283
47	short/branched chain acylCoA dehydrogenase deficiency (SBCADD)	600301
48	malonic aciduria	248360
	C. biotin-responsive disorders	
49	holocarboxylase synthase deficiency	253270
50	biotinidase deficiency	253260
51	biotin responsive basal ganglia disease	607483
	D. disorders of carbohydrate metabolism and glycogen storage diseases	
52	sucrase isomaltase deficiency	222900
53	congenital glucose/galactose malabsorption	606824
54	hereditary fructose intolerance	229600
55	galactosemia (uridylyltransferase deficiency)	230400
56	galactosemia (epimerase deficiency)	230350
57	galactosemia (galactokinase deficiency)	230200
58	transaldolase deficiency	606003
59	pyruvate carboxylase deficiency	266150

60	phosphoenolpyruvate carboxykinase (PEPCK) deficiency	261680
61	fructose 1,6-bisphosphatase deficiency	229700
62	glycogenose 0 (glycogen synthase deficiency)	240600
63	glycogenose Ia and Ib (G6Pase)	232200
64	glycogenose 3 Cori	232400
65	glycogenose 4 Andersen	232500
66	glycogenose 5 Mc Ardle	232600
67	glycogenose 6 Hers	232700-750-740
68	glycogenose 7 Tarui	232800
69	glycogenose IX phosphorylase kinase	306000-261750-613027-300559
70	glycogenosis X (phosphoglycerate mutase deficiency)	261670
71	phosphoglycerate kinase deficiency	300653
72	lactate dehydrogenase deficiency (type XI)	612933
73	aldolase A deficiency (type XII)	611881
74	β-enolase deficiency (type XIII)	612932
75	glucose transporter defect De Vivo syndrome (GLUT-1)	606777
76	fanconi-bickel syndrome (GLUT-2)	227810
	E. hyperinsulinism	
77	hyperinsulinism-hyperammonemia syndrome (HiHa)	606762
78	familial hyperinsulinemic hypoglycemia, types 1-7	256450-602485-609968 and other
	F. disorders of mitochondrial energy metabolism	
79	pyruvate dehydrogenase deficiency	312170
80	mitochondrial complex I deficiency	252010
81	mitochondrial complex II deficiency	252011
82	mitochondrial complex III deficiency	124000
83	mitochondrial complex IV deficiency	220110
84	mitochondrial complex V deficiency	604273
85	mitochondrial DNA mutation/deletion/elongation	540000-551500-545000 and other
86	nuclear DNA mutation affecting mitochondrial structure and/or function	603041-174763 and other
87	mitochondrial DNA depletion syndrome	251880 and other
88	clinical syndrome associated with mitochondrial disorder (report+board advise)	530000-557000-256000-203700-545000 and other
89	krebs cycle enzyme deficiencies	606812 and other
90	coQ10 synthesis defects	607426
	G. mitochondrial beta oxidation defects	
91	camitine transporter deficiency	212140
92	camitine palmitoyltransferase 1 deficiency	255120
93	camitine translocase	212138
94	camitine palmitoyltransferase 2 deficiency	255110
95	short chain acyl coa dehydrogenase deficiency (SCADD)	201470
96	short chain 3 oh acyl coa dehydrogenase deficiency (SCHADD)	601609
97	medium chain acyl coa dehydrogenase deficiency (MCADD)	201450
98	long chain acyl coa dehydrogenase deficiency (LCADD)	201460
99	long chain 3 oh acyl coa dehydrogenase deficiency (LCHADD)	143450
100	very long chain acyl coa dehydrogenase deficiency (VLCADD)	201475
101	mitochondrial trifunctional protein	600890
	H. disorders of ketone body metabolism	
102	3-hydroxy-3-methylglutaryl-coA synthase deficiency	605911
103	3-hydroxy-3methylglutaryl-coA lyase deficiency	246450
104	succinyl-coA 3-oxoacid coA transferase deficiency (scot)	245050
105	β-ketothiolase deficiency	203750
	I. disorders of sterol, bile acid, lipid and lipoprotein metabolism	
106	smith-lemli-opitz syndrome	270400
107	mevalonic aciduria	251170
108	3β-hydroxy δ5 c27 hydroxysteroid dehydrogenase deficiency	607765
109	α methylacyl-CoA racemase deficiency	214950
110	δ4-3-oxosteroid 5β reductase deficiency	604741
111	oxysterol 7α hydroxylase deficiency	603711
112	cerebrotendinous xanthomatosis	213700
113	familial lipoprotein lipase deficiency & apo c2 deficiency	238600
114	Abetalipoproteinemia	200100
115	Tangier disease	205400
116	Inborn hypertriglyceridemia and lipodystrophy syndromes	151660-604367-608600 and other
117	LCAT-deficiency (Norum disease and Fish-eye disease)	245900-136120
118	Hypo-alphalipoproteinemia	107680
119	LPIN1 lipid myopathy	268200
120	Wolman & cholesteryl ester storage disease	278000

	J. peroxisomal disorders	
121	zellweger spectrum (peroxisome biogenesis defects)	214110
122	acyl-CoA oxidase deficiency	264470
123	D-bifunctional protein deficiency	261515
124	2-methyl-CoA racemase deficiency	214950
125	X-linked adrenoleukodystrophy	300100
126	Rhizomelic chondrodysplasia punctata	215100
127	Refsum disease	266500
128	Refsum disease infantile form	266510
	K. lysosomal disorders	
129	glycogen storage disease type 2 (Pompe)	232300
130	glycogen storage disease type 2b (Danon and PRK-AG2)	300257-602743
131	Gaucher disease, type I, II and III	230800-230900-231000
132	Fabry disease	301500
133	Hurler-Scheie disease (MPS I)	607014-607016
134	Hunter disease (MPS II)	309900
135	Sanfilippo A; B; C; D (MPS III)	252900-252920-252930-252940
136	other MPS types (IV, V, VI, VII, VIII)	253000-253010-253200-253220-253230
137	Niemann-Pick type A and B	257200-607616
138	Niemann-Pick type C	257220-607625
139	GM1 and GM2 gangliosidosis	230500-272800 and other
140	Metachromatic Leukodystrophy	250100
141	Krabbe leukodystrophy	254200
142	Mannosidosis	248510
143	Sialidosis and galactosialidosis	256550-256540
144	I-cell disease and mucolipidosis II/III	252500-252600-252605
145	Free sialic storage diseases	269920-604369
146	Mucolipidosis IV	252650
147	Fucosidosis	230000
148	multiple sulfatase deficiency	272200
149	Farber lipogranulomatosis	228000
150	ceroid lipofuscinosis type 1-10	256730-204500-601780-204200 and other
	L. disorders of purine and pyrimidine metabolism	
151	phosphoribosyl pyrophosphate synthetase superactivity	311850-860
152	adenylosuccinate lyase deficiency	103050
153	xanthinuria	278300
154	Lesch-Nyhan syndrome	308000
	M. disorders of creatine metabolism	
155	guanidinoacetate methyltransferase deficiency (GAMT)	601240
156	arginine:glycine amidinotransferase deficiency (AGAT)	602360
157	creatine transporter deficiency	300352
	N. disorders of neurotransmitter and small peptide metabolism	
158	γ-glutamyl transpeptidase deficiency and other disorders of glutathion metabolism	230450-231900-260005 and other
159	folate transporter defects	613068
160	trimethylaminuria & dimethylglycine dehydrogenase dificiency	602079
161	aromatic amino-acid decarboxylase (AADC)	608643
162	gaba transaminase deficiency	613163
163	monoamine oxidase deficiency	300615
164	dopamine beta-hydroxylase deficiency	233360
165	pyridoxine dependent seizures	266100
166	pyridoxamine 5-phosphate oxidase deficiency (pyridoxal-P responsive seizures)	610090
167	molybdenum cofactor deficiency, sulfite oxidase deficiency	2521560-272300
168	acetylaspartic aciduria (Canavan's disease)	271900
	O. Congenital defects of glycosylation (CDG)	
169	Congenital disorders of N- and/or O-glycosylation	212065-602579-611209-611182 and other
	P. porphyrias	
170	5-aminolevulinic acid synthase deficiency	301300
171	5-aminolevulinic acid dehydratase porphyria	125270
172	acute intermittent porphyria	176000
173	hereditary coproporphyria	121300
174	variegate prophyria	176200
175	congenital erythropoietic porphyria	263700
176	erythropoietic protoporphyria	177000
	Q. disorders of copper metabolism	
177	menkes disease	309400
178	Wilson disease	227900

	R. other progressive neurodegenerative diseases	
179	Alexander disease (GFAP)	203450
180	Infantile neuroaxonal dystrophy (PLA2G6)	256600
181	Vanishing white matter disease	603896
182	Pelizaeus-Merzbacher disease	312080
	S. connective tissue disorders	
183	osteogenesis imperfecta	166200-166210-259420 and other
z0	**Niet opgenomen in RIZIV lijst**	
z1	**Andere Aandoening**	
z2	**Ongediagnosticeerd**	