Appendix 1

	Inherited metabolic disorders groups	Specific inherited metabolic disorder diagnoses	**cDNA-ALLELE-1**	**cDNA-ALLELE-2**	**Aa-ALLELE-1**	**Aa- ALLELE-2**	
1	Pompe Disease	Infantile Pompe	c.896T>G	c.896T>G	p.L299R	p.L299R	homozygous
2	Pompe Disease	Infantile Pompe	c.2662G>T	c.2662G>T	p.E888*	p.E888*	homozygous
3	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
4	Pompe Disease	Infantile Pompe	c.1556T>C	c.1556T>C	p.M519T	p.M519T	homozygous
5	Pompe Disease	Infantile Pompe	c.947A>T	c.947A>T	p.N316I	p.N316I	homozygous
6	Pompe Disease	Infantile Pompe	c.1822C>T	c.1822C>T	p.R608*	p.R608*	homozygous
7	Pompe Disease	Infantile Pompe	c.2662G>T	c.2662G>T	p.E888*	p.E888*	homozygous
8	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
9	Pompe Disease	Infantile Pompe	c.896T>G	c.2662G>T	p.L299R	p.E888*	Compound heterozygous
10	Pompe Disease	Infantile Pompe	c.896T>G	c.896T>G	p.L299R	p.L299R	homozygous
11	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
12	Pompe Disease	Infantile Pompe	c.258dupC	c.258dupC	p.N87Qfs*	p.N87Qfs*	homozygous
13	Pompe Disease	Infantile Pompe	c.2237G>A	c.2237G>A	p.W746*	p.W746*	homozygous
14	Pompe Disease	Infantile Pompe	c.896T>G	c.896T>G	p.L299R	p.L299R	homozygous
15	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
16	Pompe Disease	Infantile Pompe	c.406T>G	c.2608C>T	p.Y136D	p.R870*	Compound heterozygous
17	Pompe Disease	Infantile Pompe	c.1430_1430delT	c.1430_1430delT	p.I477Mfs*43	p.I477Mfs*43	homozygous
18	Pompe Disease	Infantile Pompe	c.2741delinsCAG	c.2741delinsCAG	p.Q914Pfs*30	p.Q914Pfs*30	homozygous

19	Pompe Disease	Infantile Pompe	c.1195-17_1199delCCGTGTTGTGGCTGCAGGACGT	c.1195-17_1199delCCGTGTTGTGGCTGCAGGACGT	p.D399Pfs*105	p.D399Pfs*105	homozygous
20	Pompe Disease	Infantile Pompe	c.1978C>T	c.1978C>T	p.R660C	p.R660C	homozygous
21	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
22	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
23	Pompe Disease	Infantile Pompe	c.2662G>T	c.2662G>T	p.E888*	p.E888*	homozygous
24	Pompe Disease	Infantile Pompe	C.1848C>A	C.1848C>A	p.D616E	p.D616E	homozygous
25	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
26	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
27	Pompe Disease	Infantile Pompe	c.1822C>T	c.1822C>T	p.R608*	p.R608*	homozygous
28	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
29	Pompe Disease	Infantile Pompe	c.896T>G	c.896T>G	p.L299R	p.L299R	homozygous
30	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
31	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
32	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
33	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
34	Pompe Disease	Infantile Pompe	c.2014C>T	c.2014C>T	p.R672W	p.R672W	homozygous
35	Pompe Disease	Infantile Pompe	c.1430_1430delT	c.1430_1430delT	p.I477Mfs*43	p.I477Mfs*43	homozygous
36	Pompe Disease	Infantile Pompe	c.258dupC	c.258dupC	p.N87Qfs*	p.N87Qfs*	homozygous
37	Pompe Disease	Infantile Pompe	c.1978C>T	c.1978C>T	p.R660C	p.R660C	homozygous
38	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous

39	Pompe Disease	Infantile Pompe	c.1822C>T	c.2662G>T	p.R608*	p.E888*	Compound heterozygous
40	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
41	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
42	Pompe Disease	Infantile Pompe	c.896T>C	c.118C>T	p.L299P	p.R40*	Compound heterozygous
43	Pompe Disease	Infantile Pompe	c.406T>G	c.406T>G	p.Y136D	p.Y136D	homozygous
44	Pompe Disease	Infantile Pompe	c.896T>C	c.896T>C	p.L299P	p.L299P	homozygous
45	Pompe Disease	Infantile Pompe	c.258_259dupC	c.258_259dupC	p.N87Qfs*9	p.N87Qfs*9	homozygous
46	Pompe Disease	Infantile Pompe	c.2662G>T	c.2662G>T	p.E888*	p.E888*	homozygous
47	Pompe Disease	Infantile Pompe	c.1551+1G>T	c.1551+1G>T	IVS10+1G>T	IVS10+1G>T	homozygous
48	LSD	Mucolipidosis II	c.1547A>T	c.1547A>T	p.D516V	p.D516V	homozygous
49	LSD	MPS tip 1 (mukopolisakkaridoz tip 1)	c.208C>T	c.208C>T	p.Q70*	p.Q70*	homozygous
50	LSD	MPS-1	c.46_57delTCGCTCCTGGCC	c.46_57delTCGCTCCTGGCC	p.16_19delSLLA	p.16_19delSLLA	homozygous
51	LSD	MPS-1	c.208C>T	c.208C>T	p.Q70*	p.Q70*	homozygous
52	LSD	MPS-1	Homozygous deletion on exon 2				homozygous
53	LSD	MPS-1	c.203G>A	c.203G>A	p.W68*	p.W68*	homozygous
54	LSD	MPS-1	c.46_57delTCGCTCCTGGCC	c.46_57delTCGCTCCTGGCC	p.16_19delSLLA	p.16_19delSLLA	homozygous
55	LSD	MPS-1	c.1205G>A	c.1205G>A	p.W402*	p.W402*	homozygous
56	LSD	MPS-2	Hemizygous deletion on exon 9				hemizygous
57	LSD	MPS-2	c.261C>G	N/A	p.S87R	N/A	hemizygous
58	LSD	MPS-2	c.514C>T	N/A	p.R172*	N/A	hemizygous

59	LSD	MPS-2	diagnosed via enzyme analysis				
60	LSD	MPS-2	diagnosed via enzyme analysis				
61	LSD	MPS-6	c.962T>C	c.962T>C	p.L321P	p.L321P	homozygous
62	LSD	MPS-6	c.962T>C	c.962T>C	p.L321P	p.L321P	homozygous
63	LSD	MPS-6	c.465C>A	c.465C>A	p.C155*	p.C155*	homozygous
64	LSD	Cystinosis	c.834_842del	c.834_842del	p.V279_Y281del	p.V279_Y281del	homozygous
65	GSD-3	GSD-III	c.1020delA	c.1020delA	p.E340Dfs*9	p.E340Dfs*9	homozygous
66	GSD-3	GSD-III	Homozygous deletion on exon 3-4				homozygous
67	GSD-3	GSD-III	c.2270_2273delCATT	c.2270_2273delCATT	p.S757fs*18	p.S757fs*18	homozygous
68	GSD-3	GSD-III	c.3772-3776delACATG	c.3772-3776delACATG	p.T1258Dfs*3	p.T1258Dfs*3	homozygous
69	GSD-3	GSD-III	c.4126C>T	c.4126C>T	p.Q1376*	p.Q1376*	homozygous
70	GSD-3	GSD-III	Homozygous deletion on exon 3-4				homozygous
71	GSD-3	GSD-III	c.2590C>T	c.2590C>T	p.P864S	p.P864S	homozygous
72	GSD-3	GSD-III	c.1999C>T	c.1999C>T	p.Q667*	p.Q667*	homozygous
73	GSD-3	GSD-III	c.1999C>T	c.1999C>T	p.Q667*	p.Q667*	homozygous
74	GSD-3	GSD-III	c.4126C>T	c.4126C>T	p.Q1376*	p.Q1376*	homozygous
75	GSD-3	GSD-III	c.440T>C	c.440T>C	p.L147P	p.L147P	homozygous
76	GSD-3	GSD-III	c.2809C>T	c.2809C>T	p.Q937*	p.Q937*	homozygous
77	GSD-3	GSD-IB	diagnosed via enzyme analysis				

78	Fatty acid oxidation disorder	Primary carnitine deficiency	c.1354G>A	c.1354G>A	p.E452K	p.E452K	homozygous
79	Fatty acid oxidation disorders	Primary carnitine deficiency	c.597delG	c.597delG	p.F200Lfs*4	p.F200Lfs*4	homozygous
80	Fatty acid oxidation disorder	Primary carnitine deficiency	c.62_64delTCT	c.62_64delTCT	p.22delS	p.22delS	homozygous
81	Fatty acid oxidation disorder	Primary carnitine deficiency	c.597delG	c.503G>A	p.F200Lfs*4	p.G168D	Compound heterozygous
82	Fatty acid oxidation disorder	Primary carnitine deficiency	c.62_64delTCT	c.62_64delTCT	p.22delS	p.22delS	homozygous
83	Fatty acid oxidation disorder	Primary carnitine deficiency	c.246_250delCCGCTinsTCGCTACCGGCTCGCC	c.246_250delCCGCTinsTCGCTACCGGCTCGCC	p.I89Gfs*45	p.I89Gfs*45	homozygous
84	Fatty acid oxidation disorder	Primary carnitine deficiency	c.246_250delCCGCTinsTCGCTACCGGCTCGCC	c.246_250delCCGCTinsTCGCTACCGGCTCGCC	p.I89Gfs*45	p.I89Gfs*45	homozygous
85	Fatty acid oxidation disorder	CPT II deficiency	c.371G>C	c.371G>C	p.R124P	p.R124P	homozygous
86	Fatty acid oxidation disorder	VLCAD	c.194delC	c.194delC	p.Y398*	p.Y398*	homozygous
87	Fatty acid oxidation disorder	MCAD	c.877C>T	c.877C>T	p.Q293E	p.Q293E	homozygous
88	Fatty acid oxidation disorder	MCAD	c.799G>A	c.799G>A	p.G267R	p.G267R	homozygous
89	Mitochondrial disorder	Sengers syndrome	c.979A>T	c.979A>T	p.K327*	p.K327*	homozygous
90	Mitochondrial disorder	Sengers syndrome	c.297G>T	c.297G>T	p.K99N	p.K99N	homozygous
91	Mitochondrial disorder	Barth syndrome	c.699G>T	N/A	p.Q233H	N/A	hemizygous
92	Mitochondrial disorder	Leigh syndrome	mt.14459G>A (%3)				Mitochondrial DNA mutation

93	Mitochondrial disorder	Complex 1 deficiency	c.406T>G	c.406T>G	p.R136C	p.R136C	homozygous
94	Mitochondrial disorder	Complex 1 deficiency	c.1022T>C	c.1022T>C	p.A34Y	p.A34Y	homozygous
95	Mitochondrial disorder	Complex 1 deficiency	c.796C>T	c.796C>T	p.R266W	p.R266W	homozygous
96	Mitochondrial disorder	COXPD38	c.322C>T	c.322C>T	p.R108C	p.R108C	homozygous
97	Mitochondrial disorder	Mitochondrial complex V (ATP synthase) deficiency	c.238C>T	c.238C>T	p.R80*	p.R80*	homozygous
98	Mitochondrial disorder	Mitochondrial complex V (ATP synthase) deficiency	c.359delC	c.359delC	p.T120Nfs*34	p.T120Nfs*34	homozygous
99	Mitochondrial disorder	Complex 1 deficiency	c.796C>T	c.796C>T	p.R266W	p.R266W	homozygous
100	Amino acid and organic acids metabolism disorder	Methylmalonic acidemia	c.2179C>T	c.2179C>T	p.R727*	p.R727*	homozygous
101	Amino acid and organic acids metabolism disorder	Partial biotinidase deficiency	c.470G>A	c.1330G>C	p.R157H	p.D444H	Compound heterozygous
102	Amino acid and organic acids metabolism disorder	3-MCCD	c.803G>C	c.803G>C	p.R268T	p.R268T	homozygous