	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
1	000001D01	4	DNA from blood	Tuberous sclerosis	TSC2	ORPHA805	Q85.1	Tuberous sclerosis	191092	Tuberous sclerosis
2	000001Pe02	4	Blood Plasma-EDTA	Tuberous sclerosis	TSC2	ORPHA805	Q85.1	Tuberous sclerosis	191092	Tuberous sclerosis
3	000002F01	20	Fibroblasts	CMT1A	-	ORPHA101081	G60.0	Charcot-Marie-Tooth disease	118220	Charcot-Marie-Tooth Disease, Demyelinating, Type 1A; CMT1A
4	000003F01	35	Fibroblasts	X-linked congenital hydrocephalus	-	ORPHA275543	Q03.8	-	236600	Hydrocephalus
5	000004F01	18	Fibroblasts	Charcot-Marie-Tooth disease 4C	SH3TC2	ORPHA99949	G60.0	Charcot-Marie-Tooth disease	608206	Charcot-Marie-Tooth Disease, Type 4C; CMT4C
6	000005F01	42	Fibroblasts	Charcot-Marie-Tooth disease 4F	PRX	ORPHA99949	G60.0	Charcot-Marie-Tooth disease	605725	Charcot-Marie-Tooth Disease, Demyelinating, Type 4F; CMT4F
7	000006DT01	60	DNA from tissue	Mitochondrial myopathy / external ophthalmoplegia / multiple lipomatosis	-	-	-	-	-	-
8	000006X01	60	PBMCs (in PBS)	Mitochondrial myopathy / external ophthalmoplegia / multiple lipomatosis	-	-	-	-	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
9	000006X02	60	Blood Plasma-EDTA	Mitochondrial myopathy / external ophthalmoplegia / multiple lipomatosis	-	-	-	-	-	-
10	000006X03	60	Urine	Mitochondrial myopathy / external ophthalmoplegia / multiple lipomatosis	-	-	-	-	-	-
11	000007D01	53	DNA from blood	MELAS syndrome	ARNt Leu	ORPHA550	G71.3	Mitochondrial myopathy	540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes; MELAS
12	000007X01	53	PBMCs (in PBS)	MELAS syndrome	ARNt Leu	ORPHA550	G71.3	Mitochondrial myopathy	540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes; MELAS
13	000008X01	33	PBMCs (in PBS)	MELAS syndrome	ARNt Leu	ORPHA550	G71.3	Mitochondrial myopathy	540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes; MELAS

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
14	000008D01	33	DNA from blood	MELAS syndrome	ARNt Leu	ORPHA550	G71.3	Mitochondrial myopathy	540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes; MELAS
15	000010DT01	31	DNA from tissue	Limb-girdle muscular dystrophy		ORPHA263	G71.0	Muscular dystrophy	-	-
16	000011DT01	69	DNA from tissue	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
17	000012Pe01	13	Blood Plasma-EDTA	Acrofacial dysostosis, Nager type	-	ORPHA245	Q75.4	Mandibulofacial dysostosis	154400	Acrofacial Dysostosis 1, Nager Type; AFD1
18	000012D01	13	DNA from blood	Acrofacial dysostosis, Nager type	-	ORPHA245	Q75.4	Mandibulofacial dysostosis	154400	Acrofacial Dysostosis 1, Nager Type; AFD1
19	000013DT01	66	DNA from tissue	Chronic degenerative myelopathy. Complex spastic paraplegia	-	-	-	-	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
20	000013F01	66	Fibroblasts	Chronic degenerative myelopathy. Complex spastic paraplegia	-	-	-	-	-	-
21	000014Pa01	55	Blood Plasma-ACD	Spastic paraparesis	-	-	G11.4	Hereditary spastic paraplegia	-	-
22	000014Pe01	55	Blood Plasma-EDTA	Spastic paraparesis	-	-	G11.4	Hereditary spastic paraplegia	-	-
23	000014D01	55	DNA from blood	Spastic paraparesis	-	-	G11.4	Hereditary spastic paraplegia	-	-
24	000015D01	57	DNA from blood	Spastic paraparesis	-	-	G11.4	Hereditary spastic paraplegia	-	-
25	000015Pe01	57	Blood Plasma-EDTA	Spastic paraparesis	-	-	G11.4	Hereditary spastic paraplegia	-	-
26	000015Pa01	57	Blood Plasma-ACD	Spastic paraparesis	-	-	G11.4	Hereditary spastic paraplegia	-	-
27	000016Pe01	2	Blood Plasma-EDTA	Anti-NMDA-receptor autoimmune encephalitis	-	ORPHA217253	G13.1	Other systemic atrophy primarily affecting central nervous system in neoplastic disease	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
28	000016D01	2	DNA from blood	Anti-NMDA-receptor autoimmune encephalitis	-	ORPHA217253	G13.1	Other systemic atrophy primarily affecting central nervous system in neoplastic disease	-	-
29	000018Pe01	6	Blood Plasma-EDTA	Sotos syndrome	NSD1	ORPHA821	Q87.3	Soto's syndrome (cerebral gigantism)	117550	Sotos syndrome
30	000018R01	6	RNA	Sotos syndrome	NSD1	ORPHA821	Q87.3	Soto's syndrome (cerebral gigantism)	117550	Sotos syndrome
31	000019DT01	35	DNA from tissue	Facioscapulohumeral distrophy	FSHD	ORPHA269	G71.0	Muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
32	000019F01	35	Fibroblasts	Facioscapulohumeral distrophy	FSHD	ORPHA269	G71.0	Muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
33	000020D02	8	DNA from blood	Beckwith Wiedemann syndrome	IC2	ORPHA116	Q87.3	Beckwith Wiedemann syndrome	130650	Beckwith Wiedemann syndrome
34	000020Pe01	8	Blood Plasma-EDTA	Beckwith Wiedemann syndrome	IC2	ORPHA116	Q87.3	Beckwith Wiedemann syndrome	130650	Beckwith Wiedemann syndrome

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35	000020Pa01	8	Blood Plasma-ACD	Beckwith Wiedemann syndrome	IC2	ORPHA116	Q87.3	Beckwith Wiedemann syndrome	130650	Beckwith Wiedemann syndrome
36	000020R01	8	RNA	Beckwith Wiedemann syndrome	IC2	ORPHA116	Q87.3	Beckwith Wiedemann syndrome	130650	Beckwith Wiedemann syndrome
37	000020L01	8	Lymphocytes	Beckwith Wiedemann syndrome	IC2	ORPHA116	Q87.3	Beckwith Wiedemann syndrome	130650	Beckwith Wiedemann syndrome
38	000021D02	4	DNA from blood	Nemaline myopathy	-	ORPHA607	G71.2	Nemaline myopathy	-	-
39	000021Pe01	4	Blood Plasma-EDTA	Nemaline myopathy	-	ORPHA607	G71.2	Nemaline myopathy	-	-
40	000021Pa01	4	Blood Plasma-ACD	Nemaline myopathy	-	ORPHA607	G71.2	Nemaline myopathy	-	-
41	000021L01	4	Lymphocytes	Nemaline myopathy	-	ORPHA607	G71.2	Nemaline myopathy	-	-
42	000021R01	4	RNA	Nemaline myopathy	-	ORPHA607	G71.2	Nemaline myopathy	-	-
43	000022X01	76	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
44	000022X02	76	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
45	000023X01	65	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
46	000024X01	54	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
47	000025X01	54	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
48	000026X01	79	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
49	000027X01	79	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
50	000028X01	79	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
51	000029X01	47	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
52	000030X01	68	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
53	000031X01	64	Immortalized lymphocytes	Amyotrophic lateral sclerosis	-	ORPHA803	G12.2	Progressive muscular atrophy	-	-
54	000032Pe01	56	Blood Plasma-ACD	Steinert myotonic dystrophy	-	ORPHA273	G71.1	Steinert disease or syndrome	160900	Myotonic Dystrophy 1; DM1
55	000032Pa01	56	Blood Plasma-ACD	Steinert myotonic dystrophy	-	ORPHA273	G71.1	Steinert disease or syndrome	160900	Myotonic Dystrophy 1; DM1

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
56	000032L01	56	Lymphocytes	Steinert myotonic dystrophy	-	ORPHA273	G71.1	Steinert disease or syndrome	160900	Myotonic Dystrophy 1
57	000032D02	56	DNA from blood	Steinert myotonic dystrophy	-	ORPHA273	G71.1	Steinert disease or syndrome	160900	Myotonic Dystrophy 1
58	000033Pe01	9	Blood Plasma-EDTA	8q21.11 microdeletion syndrome	ZFHX4	ORPHA284160	Q93.5	-	#614230	Chromosome 8q21.11 deletion syndrome
59	000033Pa01	9	Blood Plasma-ACD	8q21.11 microdeletion syndrome	ZFHX4	ORPHA284160	Q93.5	-	#614230	Chromosome 8q21.11 deletion syndrome
60	000033L01	9	Lymphocytes	8q21.11 microdeletion syndrome	ZFHX4	ORPHA284160	Q93.5	-	#614230	Chromosome 8q21.11 deletion syndrome
61	000033D02	9	DNA from blood	8q21.11 microdeletion syndrome	ZFHX4	ORPHA284160	Q93.5	-	#614230	Chromosome 8q21.11 deletion syndrome
62	000034L01	7	Lymphocytes	8q21.11 microdeletion syndrome	ZFHX4	ORPHA284160	Q93.5	-	#614230	Chromosome 8q21.11 deletion syndrome
63	000034D02	7	DNA from blood	8q21.11 microdeletion syndrome	ZFHX4	ORPHA284160	Q93.5	-	#614230	Chromosome 8q21.11 deletion syndrome
64	000034Pe01	7	Blood Plasma-EDTA	8q21.11 microdeletion syndrome	ZFHX4	ORPHA284160	Q93.5	-	#614230	Chromosome 8q21.11 deletion syndrome
65	000034Pa01	7	Blood Plasma-ACD	8q21.11 microdeletion syndrome	ZFHX4	ORPHA284160	Q93.5	-	#614230	Chromosome 8q21.11 deletion syndrome

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
66	000035X01	0	PBMCs (in PBS)	MELAS syndrome	-	ORPHA550	G71.3	Mitochondrial myopathy	540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes; MELAS
67	000036X01	66	PBMCs (in PBS)	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
68	000036X02	66	Urine	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
69	000036X03	66	Blood Plasma-EDTA	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
70	000036D01	66	DNA from blood	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
71	000037X01	70	PBMCs (in PBS)	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
72	000037X02	70	Urine	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
73	000037X03	70	Blood Plasma-EDTA	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
74	000037D01	70	DNA from blood	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
75	000038D01	37	DNA from blood	Hereditary angioedema, type II	SERPING1	ORPHA100051	D84.1	-	106100	Angioedema, Hereditary, Type II
76	000039D01	59	DNA from blood	Hereditary angioedema, type II	SERPING1	ORPHA100051	D84.1	-	106100	Angioedema, Hereditary, Type II; HAE2
77	000040D01	62	DNA from blood	Hereditary angioedema, type II	SERPING1	ORPHA100051	D84.1	-	106100	Angioedema, Hereditary, Type II; HAE2
78	000040D02	62	DNA from blood	Hereditary angioedema, type II	SERPING1	ORPHA100051	D84.1	-	106100	Angioedema, Hereditary, Type II; HAE2
79	000041D01	34	DNA from blood	Hereditary angioedema, type II	SERPING1	ORPHA100051	D84.1	-	106100	Angioedema, Hereditary, Type II; HAE2
80	000058Pe01	52	Blood Plasma-EDTA	Progressive spinal muscular atrophy	-	-	-	-	-	-
81	000058D01	52	DNA from blood	Progressive spinal muscular atrophy	-	-	-	-	-	-
82	000058F01	52	Fibroblasts	Progressive spinal muscular atrophy	-	-	-	-	-	-
83	000059Pe01	33	Blood Plasma-EDTA	Rett syndrome	MECP2	ORPHA778	F84.2	Rett's disease or syndrome	312750	Rett syndrome; RTT
84	000059L01	33	Lymphocytes	Rett syndrome	MECP2	ORPHA778	F84.2	Rett's disease or syndrome	312750	Rett syndrome; RTT
85	000060Pe01	11	Blood Plasma-EDTA	Periventricular nodular heterotopia	FLNA +c.853>T; p.Arg285Cys	ORPHA98892	Q04.8	-	300049	Heterotopia, periventricular, X-linked dominant

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
86	000060L01	11	Lymphocytes	Periventricular nodular heterotopia	FLNA +c.853>T; p.Arg285Cys	ORPHA98892	Q04.8	-	300049	Heterotopia, periventricular, X-linked dominant
87	000060LI01	11	Lymphocytes	Periventricular nodular heterotopia	FLNA +c.853>T; p.Arg285Cys	ORPHA98892	Q04.8	-	300049	Heterotopia, periventricular, X-linked dominant
88	000060D02	11	DNA from blood	Periventricular nodular heterotopia	FLNA +c.853>T; p.Arg285Cys	ORPHA98892	Q04.8	-	300049	Heterotopia, periventricular, X-linked dominant
89	000061D02	40	DNA from blood	Periventricular nodular heterotopia	FLNA +c.853>T; p.Arg285Cys	ORPHA98892	Q04.8	-	300049	Heterotopia, periventricular, X-linked dominant
90	000061L01	40	Lymphocytes	Periventricular nodular heterotopia	FLNA +c.853>T; p.Arg285Cys	ORPHA98892	Q04.8	-	300049	Heterotopia, periventricular, X-linked dominant
91	000061LI01	40	Immortalized lymphocytes	Periventricular nodular heterotopia	FLNA +c.853>T; p.Arg285Cys	ORPHA98892	Q04.8	-	300049	Heterotopia, periventricular, X-linked dominant
92	000061Pe01	40	Blood Plasma-EDTA	Periventricular nodular heterotopia	FLNA +c.853>T; p.Arg285Cys	ORPHA98892	Q04.8	-	300049	Heterotopia, periventricular, X-linked dominant
93	000062L01	39	Lymphocytes	X-linked nephropathy	-	-	-	-	-	-
94	000062Pa01	39	Blood Plasma-ACD	X-linked nephropathy	-	-	-	-	-	-

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95	000062Pe01	39	Blood Plasma-EDTA	X-linked nephropathy	-	-	-	-	-	-
96	000063Pe01	4	Blood Plasma-EDTA	X-linked nephropathy	-	-	-	-	-	-
97	000063Pa01	4	Blood Plasma-ACD	X-linked nephropathy	-	-	-	-	-	-
98	000063L01	4	Lymphocytes	X-linked nephropathy	-	-	-	-	-	-
99	000063D02	4	DNA from blood	X-linked nephropathy	-	-	-	-	-	-
100	000064Pe01	6	Blood Plasma-EDTA	X-linked nephropathy	-	-	-	-	-	-
101	000064Pa01	6	Blood Plasma-ACD	X-linked nephropathy	-	-	-	-	-	-
102	000064L01	6	Lymphocytes	X-linked nephropathy	-	-	-	-	-	-
103	000064D01	6	DNA from blood	X-linked nephropathy	-	-	-	-	-	-
104	000065Pe01	8	Blood Plasma-EDTA	X-linked nephropathy	-	-	-	-	-	-
105	000065L01	8	Lymphocytes	X-linked nephropathy	-	-	-	-	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
106	000065Pa01	8	Blood Plasma-ACD	X-linked nephropathy	-	-	-	-	-	-
107	000065D01	8	DNA from blood	X-linked nephropathy	-	-	-	-	-	-
108	000066X01	27	Primary fibroblasts	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
109	000066X02	27	Lymphoblastoid cell line	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
110	000067X01	32	Primary fibroblasts	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
111	000067X02	32	Lymphoblastoid cell line	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
112	000067X03	32	Immortalized fibroblasts (telomerase-immortalized)	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
113	000068X01	17	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
114	000069X01	27	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA

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115	000070X01	21	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
116	000071X01	24	Lymphoblastoid cell line	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
117	000071X02	24	Primary fibroblasts	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
118	000071X03	24	Corrected primary fibroblast (lentiviral vector corrected)	Fanconi Anemia corrected (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA corrected
119	000072X01	15	Lymphoblastoid cell line	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
120	000073X01	16	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
121	000074X01	24	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
122	000075X01	15	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
123	000076X01	21	Primary fibroblasts	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
124	000076X02	21	Lymphoblastoid cell line	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
125	000077X01	11	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
126	000078X01	26	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
127	000079X01	11	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
128	000080X01	4	Lymphoblastoid cell line	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
129	000081Pe01	32	Blood Plasma-EDTA	Facioscapulohumeral distrophy, type 2	-	ORPHA269	G71.0	Muscular dystrophy	158901	Facioscapulohumeral Muscular Dystrophy, type 2; FSHD2
130	000081Pa01	32	Blood Plasma-ACD	Facioscapulohumeral distrophy, type 2	-	ORPHA269	G71.0	Muscular dystrophy	158901	Facioscapulohumeral Muscular Dystrophy, type 2; FSHD2

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
131	000081L01	32	Lymphocytes	Facioscapulohumeral distrophy, type 2	-	ORPHA269	G71.0	Muscular dystrophy	158901	Facioscapulohumeral Muscular Dystrophy, type 2; FSHD2
132	000081LI01	32	Lymphocytes	Facioscapulohumeral distrophy, type 2	-	ORPHA269	G71.0	Muscular dystrophy	158901	Facioscapulohumeral Muscular Dystrophy, type 2; FSHD2
133	000081D01	32	DNA from blood	Facioscapulohumeral distrophy, type 2	-	ORPHA269	G71.0	Muscular dystrophy	158901	Facioscapulohumeral Muscular Dystrophy, type 2; FSHD2
134	000082D01	42	DNA from blood	Retinitis pigmentosa	RPGR	ORPHA791	H35.5	Hereditary retinal dystrophy	312610	Retinitis pigmentosa
135	000083D01	50	DNA from blood	Retinitis pigmentosa	RPGR	ORPHA791	H35.5	Hereditary retinal dystrophy	312610	Retinitis pigmentosa
136	000084D01	12	DNA from blood	Retinitis pigmentosa	RPGR	ORPHA791	H35.5	Hereditary retinal dystrophy	312610	Retinitis pigmentosa
137	000085Pe01	48	Blood Plasma-EDTA	Steinert myotonic dystrophy	DMPK	ORPHA273	G71.1	Steinert disease or syndrome	160900	Myotonic Dystrophy 1; DM1

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
138	000085Pa01	48	Blood Plasma-ACD	Steinert myotonic dystrophy	DMPK	ORPHA273	G71.1	Steinert disease or syndrome	160900	Myotonic Dystrophy 1; DM1
139	000085L01	48	Lymphocytes	Steinert myotonic dystrophy	DMPK	ORPHA273	G71.1	Steinert disease or syndrome	160900	Myotonic Dystrophy 1; DM1
140	000086F01	0	Fibroblasts	Short rib-polydactyly syndrome, Verma-Naumoff type	-	ORPHA93271	Q77.2	Short rib syndrome	#263510	Short rib-polydactyly syndrome, type III
141	000087X01	46	Urine	Pearson syndrome	-	ORPHA699	D64.0	-	557000	Pearson marrow-pancreas syndrome
142	000087X02	46	Blood Plasma	Pearson syndrome	-	ORPHA699	D64.0	-	557000	Pearson marrow-pancreas syndrome
143	000087X03	46	PBMCs (in PBS)	Pearson syndrome	-	ORPHA699	D64.0	-	557000	Pearson marrow-pancreas syndrome
144	000088X01	42	Urine	Pearson syndrome	-	ORPHA699	D64.0	-	557000	Pearson marrow-pancreas syndrome
145	000088X02	42	Blood Plasma	Pearson syndrome	-	ORPHA699	D64.0	-	557000	Pearson marrow-pancreas syndrome

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
146	000088X03	42	PBMCs (in PBS)	Pearson syndrome	-	ORPHA699	D64.0	-	557000	Pearson marrow-pancreas syndrome
147	000089X01	89	Urine	Oculopharyngeal muscular dystrophy	-	ORPHA270	G71.0	-	164300	Oculopharyngeal muscular dystrophy
148	000089X02	89	Blood Plasma	Oculopharyngeal muscular dystrophy	-	ORPHA270	G71.0	-	164300	Oculopharyngeal muscular dystrophy
149	000089X03	89	PBMCs (in PBS)	Oculopharyngeal muscular dystrophy	-	ORPHA270	G71.0	-	164300	Oculopharyngeal muscular dystrophy
150	000090X01	76	Urine	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
151	000090X02	76	Blood Plasma	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
152	000090X03	76	PBMCs (in PBS)	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
153	000091X01	56	Urine	Lipid storage myopathy with carnitine deficiency	-	-	-	-	-	-
154	000091X02	56	Blood Plasma	Lipid storage myopathy with carnitine deficiency	-	-	-	-	-	-
155	000091X03	56	PBMCs (in PBS)	Lipid storage myopathy with carnitine deficiency	-	-	-	-	-	-
156	000092X01	44	Urine	Becker muscular dystrophy	-	ORPHA98895	G71.0	-	300376	Muscular Dystrophy, Becker type
157	000092X02	44	Blood Plasma	Becker muscular dystrophy	-	ORPHA98895	G71.0	-	300376	Muscular Dystrophy, Becker type
158	000092X03	44	PBMCs (in PBS)	Becker muscular dystrophy	-	ORPHA98895	G71.0	-	300376	Muscular Dystrophy, Becker type
159	000093X01	83	Urine	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
160	000093X02	83	Blood Plasma	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V
161	000093X03	83	PBMCs (in PBS)	Mc Ardle disease	-	ORPHA368	E74.0	-	232600	Glycogen Storage Disease V

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
162	000094X01	38	Urine	Becker muscular dystrophy	-	ORPHA98895	G71.0	-	300376	Muscular Dystrophy, Becker type
163	000094X02	38	Blood Plasma	Becker muscular dystrophy	-	ORPHA98895	G71.0	-	300376	Muscular Dystrophy, Becker type
164	000094X03	38	PBMCs (in PBS)	Becker muscular dystrophy	-	ORPHA98895	G71.0	-	300376	Muscular Dystrophy, Becker type
165	000095X01	72	Urine	Muscle phosphofructokinase (PFK) deficiency (Tarui's disease)	-	ORPHA371	E74.0	-	232800	Glycogen storage disease VII
166	000095X02	72	Blood Plasma	Muscle phosphofructokinase (PFK) deficiency (Tarui's disease)	-	ORPHA371	E74.0	-	232800	Glycogen storage disease VII
167	000095X03	72	PBMCs (in PBS)	Muscle phosphofructokinase (PFK) deficiency (Tarui's disease)	-	ORPHA371	E74.0	-	232800	Glycogen storage disease VII
168	000096X01	41	Urine	MELAS syndrome	-	ORPHA550	G71.3	Mitochondrial myopathy	540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes; MELAS

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
169	000096X02	41	Blood Plasma	MELAS syndrome	-	ORPHA550	G71.3	Mitochondrial myopathy	540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes; MELAS
170	000096X03	41	PBMCs (in PBS)	MELAS syndrome	-	ORPHA550	G71.3	Mitochondrial myopathy	540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes; MELAS
171	000097X01	63	Urine	Limb-girdle muscular dystrophy	-	ORPHA263	G71.0	Limb-girdle muscular dystrophy	-	-
172	000097X02	63	Blood Plasma	Limb-girdle muscular dystrophy	-	ORPHA263	G71.0	Limb-girdle muscular dystrophy	-	-
173	000097X03	63	PBMCs (in PBS)	Limb-girdle muscular dystrophy	-	ORPHA263	G71.0	Limb-girdle muscular dystrophy	-	-
174	000098X01	72	Urine	Limb-girdle muscular dystrophy	-	ORPHA263	G71.0	Limb-girdle muscular dystrophy	-	-
175	000098X02	72	Blood Plasma	Limb-girdle muscular dystrophy	-	ORPHA263	G71.0	Limb-girdle muscular dystrophy	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
176	000098X03	72	PBMCs (in PBS)	Limb-girdle muscular dystrophy	-	ORPHA263	G71.0	Limb-girdle muscular dystrophy	-	-
178	000099X01	66	Urine	Kearns-Sayre syndrome	-	ORPHA480	H49.8	-	530000	Kearns-Sayre syndrome
178	000099X05	66	Blood Plasma	Kearns-Sayre syndrome	-	ORPHA480	H49.8	-	530000	Kearns-Sayre syndrome
179	000099X06	66	PBMCs (in PBS)	Kearns-Sayre syndrome	-	ORPHA480	H49.8	-	530000	Kearns-Sayre syndrome
180	000100X01	62	Urine	Mitochondrial myopathy	-	-	-	-	251900	Mitochondrial myopathy
181	000100X02	62	Blood Plasma	Mitochondrial myopathy	-	-	-	-	251900	Mitochondrial myopathy
182	000100X03	62	PBMCs (in PBS)	Mitochondrial myopathy	-	-	-	-	251900	Mitochondrial myopathy
183	000101X01	62	Urine	Kearns-Sayre syndrome	-	ORPHA480	H49.8	-	530000	Kearns-Sayre syndrome
184	000101X02	62	Blood Plasma	Kearns-Sayre syndrome	-	ORPHA480	H49.8	-	530000	Kearns-Sayre syndrome
185	000101X03	62	PBMCs (in PBS)	Kearns-Sayre syndrome	-	ORPHA480	H49.8	-	530000	Kearns-Sayre syndrome

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
186	000102X01	57	Blood Plasma	Limb-girdle muscular dystrophy	-	ORPHA263	G71.0	Limb-girdle muscular dystrophy	-	-
187	000102X02	57	PBMCs (in PBS)	Limb-girdle muscular dystrophy	-	ORPHA263	G71.0	Limb-girdle muscular dystrophy	-	-
188	000103X01	56	Urine	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Facioscapulohumeral muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
189	000103X02	56	Blood Plasma	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
190	000103X03	56	PBMCs (in PBS)	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Facioscapulohumeral muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
191	000104X01	37	Urine	Muscular dystrophy due to dysferlin deficiency	-	ORPHA268	G71.0	-	253601	Muscular dystrophy, limb-girdle, type 2B
192	000104X02	37	Blood Plasma	Muscular dystrophy due to dysferlin deficiency	-	ORPHA268	G71.0	-	253601	Muscular dystrophy, limb-girdle, type 2B

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
193	000104X03	37	PBMCs (in PBS)	Muscular dystrophy due to dysferlin deficiency	-	ORPHA268	G71.0	-	253601	Muscular dystrophy, limb-girdle, type 2B
194	000105X01	30	Urine	Adenosine monophosphate deaminase deficiency	-	ORPHA45	-	-	102770	Adenosine monophosphate deaminase deficiency
196	000105X02	30	Blood Plasma	Adenosine monophosphate deaminase deficiency	-	ORPHA45	-	-	102770	Adenosine monophosphate deaminase deficiency
196	000105X03	30	PBMCs (in PBS)	Adenosine monophosphate deaminase deficiency	-	ORPHA45	-	-	102770	Adenosine monophosphate deaminase deficiency
197	000106X01	68	Urine	Muscular dystrophy due to dysferlin deficiency	-	ORPHA268	G71.0	-	253601	Muscular dystrophy, limb-girdle, type 2B
198	000106X02	68	Blood Plasma	Muscular dystrophy due to dysferlin deficiency	-	ORPHA268	G71.0	-	253601	Muscular dystrophy, limb-girdle, type 2B
199	000106X03	68	PBMCs (in PBS)	Muscular dystrophy due to dysferlin deficiency	-	ORPHA268	G71.0	-	253601	Muscular dystrophy, limb-girdle, type 2B
200	000107D02	20	DNA from blood	Deletion 18q	-	ORPHA1600	Q93.8	Monosomy 18q	601808	Chromosome 18q deletion syndrome

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201	000107Pe01	20	Blood Plasma-EDTA	Deletion 18q	-	ORPHA1600	Q93.8	Monosomy 18q	601808	Chromosome 18q deletion syndrome
202	000107Pa01	20	Blood Plasma-ACD	Deletion 18q	-	ORPHA1600	Q93.8	Monosomy 18q	601808	Chromosome 18q deletion syndrome
203	000107LI01	20	Immortalized lymphocytes	Deletion 18q	-	ORPHA1600	Q93.8	Monosomy 18q	601808	Chromosome 18q deletion syndrome
204	000108Pe01	35	Blood Plasma-EDTA	Cowden syndrome	PTEN	ORPHA201	-	-	158350	Cowden síndrome, Multiple hamartoma syndrome, MHAM.
205	000108Pa01	35	Blood Plasma-ACD	Cowden syndrome	PTEN	ORPHA201	Q85.8	-	158350	Cowden syndrome, CS Multiple hamartoma syndrome, MHAM
206	000108L01	35	PBMCs	Cowden syndrome	PTEN	ORPHA201	Q85.8	-	158350	Cowden syndrome, Multiple hamartoma syndrome, MHAM
207	000108D02	35	DNA from blood	Cowden syndrome	PTEN	ORPHA201	Q85.8	-	158350	Cowden syndrome, CS Multiple hamartoma syndrome, MHAM
208	000109Pe01	72	Blood Plasma-EDTA	Retinitis pigmentosa	ABHD12 +c.319delA; p.Arg107fs	ORPHA791	H35.5	Hereditary retinal dystrophy	613599	Retinitis pigmentosa
209	000109L01	72	PBMCs	Retinitis pigmentosa	ABHD12 +c.319delA; p.Arg107fs	ORPHA791	H35.5	Hereditary retinal dystrophy	613599	Retinitis pigmentosa

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210	000109LI01	72	Immortalized lymphocytes	Retinitis pigmentosa	ABHD12 +c.319delA; p.Arg107fs	ORPHA791	H35.5	Hereditary retinal dystrophy	613599	Retinitis pigmentosa
211	000110Pe01	78	Blood Plasma-EDTA	Retinitis pigmentosa	ABHD12 +c.319delA; p.Arg107fs	ORPHA791	H35.5	Hereditary retinal dystrophy	613599	Retinitis pigmentosa
212	000110L01	78	PBMCs	Retinitis pigmentosa	ABHD12 +c.319delA; p.Arg107fs	ORPHA791	H35.5	Hereditary retinal dystrophy	613599	Retinitis pigmentosa
213	000110LI01	78	Immortalized lymphocytes	Retinitis pigmentosa	ABHD12 +c.319delA; p.Arg107fs	ORPHA791	H35.5	Hereditary retinal dystrophy	613599	Retinitis pigmentosa
214	000111F01	26	Fibroblasts	Emery-Dreifuss muscular dystrophy	-	ORPHA261	G71.0	Emery-Dreifuss muscular dystrophy	-	-
215	000112X01	19	Fibroblasts	Neuronal Ceroid Lipofuscinosis	CLN3	ORPHA216	E75.4	Neuronal Ceroid Lipofuscinosis	607042	Ceroid lipofuscinosis, neuronal, 3
216	000112X02	19	Fibroblasts	Neuronal Ceroid Lipofuscinosis	CLN3	ORPHA216	E75.4	Neuronal Ceroid Lipofuscinosis	607042	Ceroid lipofuscinosis, neuronal, 3
217	000113X01	4	Fibroblasts	Neuronal Ceroid Lipofuscinosis	CLN2	-	E75.4	Neuronal Ceroid Lipofuscinosis	#204500	Ceroid lipofuscinosis, neuronal, 2
218	000113X02	4	Fibroblasts	Neuronal Ceroid Lipofuscinosis	CLN2	-	E75.4	Neuronal Ceroid Lipofuscinosis	#204500	Ceroid lipofuscinosis, neuronal, 2

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219	000114Pe01	11	Blood Plasma-EDTA	Epileptic encephalopathy - Cutis laxa	-	-	-	-	-	-
220	000114Pa01	11	Blood Serum	Epileptic encephalopathy - Cutis laxa	-	-	-	-	-	-
221	000114D01	11	DNA from blood	Epileptic encephalopathy - Cutis laxa	-	-	-	-	-	-
222	000119X01	29	PBMCs (in PBS)	Kearns-Sayre syndrome	-	ORPHA480	H49.8	-	530000	Kearns-Sayre syndrome
223	000119X02	29	Blood Plasma-EDTA	Kearns-Sayre syndrome	-	ORPHA480	H49.8	-	530000	Kearns-Sayre syndrome
224	000120X01	80	Urine	Effort rhabdomyolysis	-	-	-	-	-	-
225	000120X02	80	Blood Plasma	Effort rhabdomyolysis	-	-	-	-	-	-
226	000120X03	80	PBMCs (in PBS)	Effort rhabdomyolysis	-	-	-	-	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
227	000121X01	53	Urine	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Facioscapulohumeral muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
228	000121X02	53	Blood Plasma	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Facioscapulohumeral muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
229	000121X03	53	PBMCs (in PBS)	Facioscapulohumeral distrophy	-	ORPHA269	G71.0	Facioscapulohumeral muscular dystrophy	158900; 158901; 160570; 600416	Facioscapulohumeral Muscular Dystrophy
230	000122X01	49	Urine	Tubular aggregate myopathy	-	ORPHA2593	G71.2	-	%160565	Tubular aggregate myopathy
231	000122X02	49	Blood Plasma	Tubular aggregate myopathy	-	ORPHA2593	G71.2	-	%160565	Tubular aggregate myopathy
232	000122X03	49	PBMCs (in PBS)	Tubular aggregate myopathy	-	ORPHA2593	G71.2	-	%160565	Tubular aggregate myopathy
233	000123X01	25	Blood Plasma	4-hydroxybutyric aciduria	-	ORPHA22	E72.8	-	#271980	Succinic semialdehyde dehydrogenase deficiency
234	000123X02	25	PBMCs (in PBS)	4-hydroxybutyric aciduria	-	ORPHA22	E72.8	-	#271980	Succinic semialdehyde dehydrogenase deficiency

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235	000124X01	41	Blood Plasma	Camurati-Engelmann disease	-	ORPHA1328	Q78.3	-	#131300	Camurati-Engelmann disease, CAEND
236	000124X02	41	PBMCs (in PBS)	Camurati-Engelmann disease	-	ORPHA1328	Q78.3	-	#131300	Camurati-Engelmann disease, CAEND
237	000126X01	26	Primary fibroblasts	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
238	000127X01	7	Primary fibroblasts	Fanconi Anemia	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
239	000130D01	26	DNA from blood	Immunodeficiency due to a C2 component complement deficiency	C2	ORPHA169147	D84.1	Defects uin the Complement sytem	613927	Complement Component 2 Deficiency; C2D
240	000130X01	26	Blood Plasma	Immunodeficiency due to a C2 component complement deficiency	C2	ORPHA169147	D84.1	Defects uin the Complement sytem	613927	Complement Component 2 Deficiency; C2D
241	000131D01	22	DNA from blood	Immunodeficiency due to a C2 component complement deficiency	C2	ORPHA169147	D84.1	Defects uin the Complement sytem	613927	Complement Component 2 Deficiency; C2D

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
242	000131X01	22	Blood Plasma	Immunodeficiency due to a C2 component complement deficiency	C2	ORPHA169147	D84.1	Defects uin the Complement sytem	613927	Complement Component 2 Deficiency; C2D
243	000132D01	26	DNA from blood	Immunodeficiency due to Complement Factor 1 deficiency	CF1	ORPHA169147	D84.1	Defects uin the Complement sytem	217030	Complement Factor I Deficiency; CFID
244	000132X01	26	Blood Plasma	Immunodeficiency due to Complement Factor 1 deficiency	CF1	ORPHA169147	D84.1	Defects uin the Complement sytem	217030	Complement Factor I Deficiency; CFID
245	000133X01	11	Lymphoblastoid cell line	Fanconi Anemia (blood mosaic)	FANCA	ORPHA84	D61.0	Fanconi's (congenital pancytopenia)	227650	Fanconi Anemia, Complementation Group A; FANCA
246	000135X01	21	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	261600	Phenylketonuria; PKU
247	000135X02	21	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	261600	Phenylketonuria; PKU
248	000136X01	43	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	261600	Phenylketonuria; PKU
249	000136X012	43	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	261600	Phenylketonuria; PKU

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250	000137X01	21	PBMC (in PBS)	Glutaric aciduria type I; Glutaryl-CoA dehydrogenase deficiency	-	ORPHA25	E72.3	Glutaric aciduria type I	#231670	Glutaric Acidemia
251	000137X02	21	Blood Plasma-EDTA	Glutaric aciduria type I; Glutaryl-CoA dehydrogenase deficiency	-	ORPHA25	E72.3	Glutaric aciduria type I	#231670	Glutaric Acidemia
252	000138X01	20	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	261600	Phenylketonuria; PKU
253	000138X02	20	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
254	000139X01	27	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
255	000139X02	27	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
256	000140X01	22	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
257	000140X02	22	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
258	000141X01	27	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
259	000141X02	27	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
260	000142X01	70	PBMC (in PBS)	Von Recklinghausen Disease or neurofibromatosis type I	-	ORPHA636	Q85.0	Neurofibromatosis (nonmalignant)	#162200	Neurofibromatosis type I
261	000142X02	70	Blood Plasma-EDTA	Von Recklinghausen Disease or neurofibromatosis type I	-	ORPHA636	Q85.0	Neurofibromatosis (nonmalignant)	#162200	Neurofibromatosis type I
262	000143X01	31	PBMC (in PBS)	Methylmalonic acidemia with homocystinuria, type cblC	-	ORPHA79282	E72.1	-	#277400	Methylmalonic acidemia and homocystinuria, cblC type
263	000143X02	31	Blood Plasma-EDTA	Methylmalonic acidemia with homocystinuria, type cblC	-	ORPHA79282	E72.1	-	#277400	Methylmalonic acidemia and homocystinuria, cblC type
264	000144X01	24	PBMC (in PBS)	Citrullinemia	-	ORPHA187	E72.2	Citrullinemia	-	-

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265	000144X02	24	Blood Plasma-EDTA	Citrullinemia	-	ORPHA187	E72.2	Citrullinemia	-	-
266	000145X01	31	PBMC (in PBS)	Galactosemia	-	ORPHA352	E74.2	-	-	-
267	000145X02	31	Blood Plasma-EDTA	Galactosemia	-	ORPHA352	E74.2	-	-	-
268	000146X01	68	PBMC (in PBS)	Control (Galactosemia)	-	-	-	-	-	-
269	000146X02	68	Blood Plasma-EDTA	Control (Galactosemia)	-	-		-	-	-
270	000147X01	56	PBMC (in PBS)	Coenzyme Q 10 deficiency	-	ORPHA35656	-	-	-	-
271	000147X02	56	Blood Plasma-EDTA	Coenzyme Q 10 deficiency	-	ORPHA35656	-	-	-	-
272	000148X01	47	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
273	000148X02	47	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
274	000149X01	21	PBMC (in PBS)	Coenzyme Q 10 deficiency	-	ORPHA35656	-	-	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
275	000149X02	21	Blood Plasma-EDTA	Coenzyme Q 10 deficiency	-	ORPHA35656	-	-	-	-
276	000150X01	38	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
277	000150X02	38	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
278	000151X01	38	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
279	000151X02	38	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
280	000152X01	46	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
281	000152X02	46	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
282	000153X01	65	PBMC (in PBS)	Control (Galactosemia)	-	-	-	-	-	-
283	000153X02	65	Blood Plasma-EDTA	Control (Galactosemia)	-	-	-	-	-	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
284	000154X01	30	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
285	000154X02	30	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
286	000155X01	21	PBMC (in PBS)	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
287	000155X02	21	Blood Plasma-EDTA	Phenylketonuria	-	ORPHA716	E70.0 E70.1	Phenylketonuria	#261600	Phenylketonuria; PKU
288	000156X01	29	PBMC (in PBS)	Galactosemia	-	ORPHA352	E74.2	-	-	-
289	000156X02	29	Blood Plasma-EDTA	Galactosemia	-	ORPHA352	E74.2	-	-	-
290	000157X01	23	PBMC (in PBS)	Biotinidase deficiency	-	ORPHA79241	E53.8	-	#253260	Biotinidase deficiency
291	000157X02	23	Blood Plasma-EDTA	Biotinidase deficiency	-	ORPHA79241	E53.8	-	#253260	Biotinidase deficiency
292	000158X01	18	PBMC (in PBS)	Cerebrotendinous xanthomatosis	-	ORPHA909	E75.5	-	#213700	Cerebrotendinous xanthomatosis

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
293	000158X02	18	Blood Plasma-EDTA	Cerebrotendinous xanthomatosis	-	ORPHA909	E75.5	-	#213700	Cerebrotendinous xanthomatosis
294	000159X01	23	PBMC (in PBS)	Coenzyme Q 10 deficiency	-	ORPHA35656	-	-	-	-
295	000159X02	23	Blood Plasma-EDTA	Coenzyme Q 10 deficiency	-	ORPHA35656	-	-	-	-
296	000160X01	24	PBMC (in PBS)	Propionic acidemia	-	ORPHA35	E71.1	Propionic acidemia	#606054	Propionic acidemia
297	000160X02	24	Blood Plasma-EDTA	Propionic acidemia	-	ORPHA35	E71.1	Propionic acidemia	#606054	Propionic acidemia
298	000220Pe01	28	Blood Plasma-EDTA	Wolfram syndrome	WFS1	ORPHA3463	-	-	606201	Wolfram síndrome I
299	000220D01	28	DNA from blood	Wolfram syndrome	WFS1	ORPHA3463	-	-	606201	Wolfram síndrome I
300	000220LI01	28	Immortalized lymphocytes	Wolfram syndrome	WFS1	ORPHA3463	-	-	606201	Wolfram síndrome I
301	000230F01	2	Primary fibroblasts	Atypical Rhabdoid Tumor (grade IV WHO)		ORPHA99966	-	-	609322	-

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
302	000161X02	34	Blood Plasma-Citrate	Friedreich Ataxia	FXN Mut 1 > 600 Mut 2 > 600 Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
303	000162X02	39	Blood Plasma-Citrate	Friedreich Ataxia	FXN Mut 1 > 300 Mut 2 > 300 Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
304	000163X02	52	Blood Plasma-Citrate	Friedreich Ataxia	FXN Mut 1: 211 Mut 2: 375 Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
305	000164X02	46	Blood Plasma-Citrate	Friedreich Ataxia	-	ORPHA95	G11.1	Friedreich Ataxia	-	Friedreich Ataxia
306	000165X02	39	Blood Plasma-Citrate	Friedreich Ataxia	-	ORPHA95	G11.1	Friedreich Ataxia	-	Friedreich Ataxia
307	000166X02	37	Blood Plasma-Citrate	Friedreich Ataxia	-	ORPHA95	G11.1	Friedreich Ataxia	-	Friedreich Ataxia
308	000167X02	41	Blood Plasma-Citrate	Friedreich Ataxia	-	ORPHA95	G11.1	Friedreich Ataxia	-	Friedreich Ataxia
309	000168X02	37	Blood Plasma-Citrate	Friedreich Ataxia	FXN Mut 1: 930 Mut 2: 630 Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
310	000169X02	35	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA (Genotype: homozygous)	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
311	000170X02	35	Blood Plasma-Citrate	Friedreich Ataxia	FXN Mut 1 > 500 Mut 2 > 700 Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
312	000171X02	46	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA (Genotype: homozygous)	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
313	000172X02	32	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
314	000173X02	28	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
315	000174X02	56	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
316	000175X02	29	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
317	000176X02	38	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
318	000177X02	26	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
319	000178X02	45	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
320	000179X02	19	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
321	000221LI	20	Immortalized lymphocytes	Non-syndromic overgrowth	RNF125	-	-	-	-	-
322	000222LI	13	Immortalized lymphocytes	Non-syndromic overgrowth	RNF135 Mut 1: M112I	-	-	-	-	-
323	000223X02	22	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA (Genotype: homozygous	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
324	000224X02	69	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA (Genotype: homozygous) Mut 1: 75	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
325	000225X02	49	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA (Genotype: homozygous) Mut 1: 250	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
326	000226X02	38	Blood Plasma-Citrate	Friedreich Ataxia	FXN Mut 1: 480 Mut 2: 580 Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
327	000227X02	33	Blood Plasma-Citrate	Friedreich Ataxia	FXN Mut 1: 480 Mut 2: 580 Exp. GAA	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
328	000228X02	50	Blood Plasma-Citrate	Friedreich Ataxia	FXN Exp. GAA (Genotype: homozygous) Mut 1: 314	ORPHA95	G11.1	Friedreich Ataxia	#229300	Friedreich Ataxia 1; FRDA
329	000231LI	36	Immortalized lymphocytes	CMT + Retinitis pigmentosa + hearing loss	PRPS1 c.46T>C; p.Ser16Pro					
330	000232LI	38	Immortalized lymphocytes	Neuropathy + Retinitis pigmentosa	PRPS1 c.46T>C; p.Ser16Pro					
331	000233LI	70	Immortalized lymphocytes	Ataxia + Retinitis pigmentosa	PRPS1 c.46T>C; p.Ser16Pro					

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Orpha Number	ICD-10 Identifier	ICD-10 Name	MIM Number	MIM Name
332	000117LI	36	Immortalized lymphocytes	Retinitis pigmentosa + cognitive impairment + thrombocytopenia + posssible galactosemia	GALE +c.231_232insTGTTC; +c.449C>T p.T150M	-	-	-	-	-
333	000118LI	28	Immortalized lymphocytes	Retinitis pigmentosa + cognitive impairment + thrombocytopenia + posssible galactosemia	GALE +c.231_232insTGTTC;p.K78fs; +p.T150M; c.449C>T	-	-	-	-	-
334	000125LI	62	Immortalized lymphocytes	Retinitis pigmentosa	-	-	-	-		
335	000229LI	6	Immortalized lymphocytes	Leber congenital amaurosis	NMNAT1 +c.769G>A (p.Glu257Lys)	ORPHA65	H35.5	Leber's congenital amaurosis	-	-
336	000234LI	80	Immortalized lymphocytes	Atypical fracture						
337	000235LI	68	Immortalized lymphocytes	Atypical fracture						
338	000236LI	78	Immortalized lymphocytes	Atypical fracture						

	CBK Code	Age	Sample type	Clinical Diagnosis	Mutated Gene	Observervations
	000180-219	-	Blood Plasma-Citrate	Adolescent Idiopathic Scoliosis	-	Plasma citrate collection including samples from 29 patients and 11 controls
	000180-219	-	Blood Plasma-EDTA	Adolescent Idiopathic Scoliosis	-	Plasma EDTA collection including samples from 29 patients and 10 controls
	-	-	DNA from blood	Charcot-Marie-Tooth Disease	-	DNA collection including samples from 231 patients and 158 controls.
	-	-	Blood Plasma-Citrate	Charcot-Marie-Tooth Disease	-	Plasma citrate collection including samples from 45 patients and 15 controls
	-		Blood Plasma-EDTA	Charcot-Marie-Tooth Disease	-	Plasma EDTA collection including samples from 45 patients and 15 controls
	-	-	Primary fibroblasts	Charcot-Marie-Tooth Disease	-	Primary fibroblast collection including samples from 5 patients

CIBERER'S DECENTRALIZED COLLECTIONS:

Node	RD	Number of samples/patients	Sample type
1	Lowe syndrome (LS); Congenital disorder of glycosilation (GDG).	LS: 7 CDG: 20	Primary fibroblasts, DNA, serum.
2	Rare ocular diseases (Norrie, retinal dystrophies, retinosis pigmentaria, Angelman S., alpha-1-antitrypsin, among others); rare cancers; inherited metabolic diseases; neuromuscular and peripheral neuropathies (CMT, Steinert, Duchenne/Becker, Kennedy, among others); neurodegenerative (ataxia, Huntington, Lafora); bone dysplasias (achondroplasia, Holt-Oram); dermatological (Epidermolysis bullosa, incontinentia pigmenti) and others.	Over 40,000 samples	DNA
3	Hereditary hemorrhagic telangiectasia; von Hippel Lindau syndrome (VHL)	HHT: 638 VHL: 140	**HHT:** DNA, plasma; BOECs (blood outgrowth endothelial cells) and HUVECs (Human umbilical vein endothelial cells) primary cultures. **VHL:** DNA, plasma samples, primary cultures.

Node	RD	Number of samples/patients	Sample type
4	Rare diseases and familiar cancer	25,000 samples of familial cancer and genetic diseases.	DNA
5	a): Leber Hereditary Optic Neuropathy, MERRF syndrome, mitochondrial sensorineural deafness, mitochondrial-diabetes; b): Leigh syndrome, severe mitochondrial encephalopathy, autosomal dominant optic atrophy plus syndrome, defects of intergenomic communication with multiple deletions of mtDNA.		Transmitochondrial cybrids (RDs within group **a)**; iPS Cells (RDs within group **b)**
6	Homocystinuria, Krabbe disease, hereditary multiple exostoses, Costello syndrome, Gaucher disease, GM1 gangliosidosis, Sanfilippo disease type C, Niemann-Pick disease types A/B , Niemann-Pick disease type C.	50-200 samples of each pathology except for Opitz C Syndrome (10 samples)	DNA, fibroblasts.
7	Muscular fiber type 2 atrophy, dermatomyositis, muscle distrophy, oculopharyngeal muscular dystrophy, myoadenylate deaminase deficiency, Mc Ardle disease, congenital myopathy, critical illness myopathy, distal myopathy, neutral lipid storage disease, myofibrillar myopathy, mitochondrial myopathy, autoimmune necrotizing myopathy, neuropathic pattern, polymyositis, sporadic inclusion body myositis, muscle toxicity (Sunitinib), vasculitis.	1-50 patients/RD	PBMC, plasma, serum, DNA, tissue.
8	Mitochondrial diseases	2,000 samples aprox.	DNA

Node	RD	Number of samples/patients	Sample type
9	Fragile X and ID		DNA, RNA, tissue
10	Prostatic cancer, Wolfram syndrome and renal diseases	>200 patients/total	DNA, RNA from lymphocytes and tissue
11	Gaucher disease; Fabry disease; Nieman-Pick disease A/B; Nieman-Pick disease C; Lysosomal acid lipase deficiency.	1-300 patients/RD	DNA, plasma, leukocytes, fibroblasts, monocyte-derived macrophages.
12	Rare genetic diseases, including an important collection of overgrowth disorders	35,800 samples of patients with rare genetic diseases. Includes a collection of 1,800 patients with overgrowth disorders (From the Spanish Overgrowth Syndrome Registry).	DNA
13	Hereditary angioedema due to C1-inhibitor deficiency; Acquired angioedema; Angioedema with normal C1 inhibitor activity; Atypical hemolytic-uremic syndrome; C3 glomerulopathy; Barraquer-Simons syndrome; Lawence syndrome; several immunodeficiencies: C1q, C2, C3, C5, C6, C7, C8 , factor H, factor I.	1-600 patients/RD	ADN, ARN, serum and plasma

Node	RD	Number of samples/patients	Sample type
14	Hypoacusis, retinal dystrophies, Usher syndrome, Huntington disease, Charcot-Marie-Tooth disease, Duchenne's muscle distrophy, Steinert myotonic dystrophy, adult onset cerebellar ataxia, Friedreich ataxia, achondroplasia, albinism, spinal muscular atrophy	>500 samples	Tissue, cells and cell lines.
15	Bone disorders (Osteogenesis imperfecta, Ellis-van Creveld syndrom and bone dysplasias)	500 samples aprox.	DNA and fibroblast cell lines
16	Neuromuscular diseases	>1,500 samples	Tissue (muscle, nerve and skin), DNA from blood.
17	Hematological and Immunological diseases	20	DNA
18	Hemolytic anemia due to glucophosphate isomerase deficiency; Hemolytic anemia due to red cell pyruvate kinase deficiency; Triose phosphate-isomerase deficiency; Hereditary elliptocytosis; Hereditary spherocytosis; and other membrane pathologies without definitive diagnosis.	1-50 patients/RD	Leukocyte pellets