List of autosomal recessive diseases included in the UMCG gene panel

Disease	Links	Gene
Andermann Syndrome	http://www.omim.org/entry/218000 http://www.orpha.net/consor4.01/www/cgi-bin/OC_Exp.php?lng=EN&Expert=1496	SLC12A6
Aspartylglucosaminuria	http://www.omim.org/entry/208400 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=93	AGA
Ataxia Telangiectasia	http://www.omim.org/entry/208900 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=100	ATM
Bloom Syndrome	http://www.omim.org/entry/210900 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=125	BLM (RECQL3)
Canavan's Disease	http://omim.org/entry/271900 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=141	ASPA
Citrullinemia type 1	http://www.omim.org/entry/215700 http://www.orpha.net/consor4.01/www/cgi-bin/OC_Exp.php?lng=EN&Expert=247525	ASS1
Congenital disorder of glycosylation type 1A	http://www.omim.org/entry/212065 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=79318	PMM2
Congenital Nephrotic Syndrome, Finnish Type	http://www.omim.org/entry/256300 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=839	NPHS1
D-Bifunctional Protein Deficiëncy	http://www.omim.org/entry/261515	HSD17B4
Epidermolysis Bullosa	http://www.omim.org/entry/226700 http://www.omim.org/entry/226600	LAMA3, LAMB3, LAMC2, COL7A1
Familial Dysautonomia	http://www.omim.org/entry/223900 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=1764	IKBKAP
GM2 Gangliosidosis, mainly Tay Sachs Disease	http://www.omim.org/entry/272800 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=845	HEXA
GRACILE Syndrome	http://www.omim.org/entry/603358 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=53693	BCS1L
Hypophosphatasemia	http://omim.org/entry/241500 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=436	ALPL
Infantile Sialic Acid Storage Disease	http://www.omim.org/entry/269920 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=834	SLC17A5
Krabbe's Disease	http://www.omim.org/entry/245200 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=487	GALC
Lipoamide Dehydrogenase Deficiëncy	http://www.omim.org/entry/246900 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=2394	DLD
Leukoencephalopathy with Vanishing White Matter	http://www.omim.org/entry/603896 http://www.orpha.net//consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=135	EIF2B1, EIF2B2, EIF2B3,

Disease	Links	Gene
		EIF2B4, EIF2B5
Metachromatic Leukodystrophy	http://omim.org/entry/250100 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=512	ARSA
Mitochondrial Recessive Ataxia Syndrome	http://www.omim.org/entry/607459	POLG
Mitochondrial DNA depletion syndromes type 4A	http://www.omim.org/entry/203700	POLG
Mitochondrial DNA Depletion Syndrome type 4 B	http://www.omim.org/entry/613662	POLG
Mucolipidosis IV	http://omim.org/entry/252650 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=578	MCOLN1
Mucopolysaccharidosis I (Hurler, Hurler-Scheie)	http://omim.org/entry/607014 http://omim.org/entry/607015 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=579	IDUA
Mucopolysaccharidosis III, Sanfilippo Syndrome	http://omim.org/entry/252900 http://omim.org/entry/252920 http://omim.org/entry/252930 http://omim.org/entry/252940 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=581	SGSH, NAGLU, HGSNAT, GNS
Mucopolysaccharidosis IV, Morquio Syndrome	http://omim.org/entry/253000 http://omim.org/entry/253010 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=582	GALNS, GLB1
Mucopolysaccharidosis VI, Maroteaux-Lamy Syndrome	http://omim.org/entry/253200 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=583 http://www.maroteaux-lamy.com/en-gb/pages/hcp/index.aspx	ARSB
Mucopolysaccharidosis VII, Sly Syndrome	http://omim.org/entry/253220 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=584	GUSB
Muscle Eye Brain Disease/ Muscular Dystrophy-Dystroglycanopathy (congenital with Brain and Eye Anomalies)	http://www.omim.org/entry/253280 http://omim.org/entry/614643 http://omim.org/entry/615041	POMGNT1; FKRP, ISPD, TMEM5,
Neuronal Ceroid Lipofuscinosis, Types 1,2	http://omim.org/entry/256730 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=79263 http://omim.org/entry/204500 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=168491	PPT1, TPP1
Neuronal Ceroid Lipofuscinosis Type 3, Batten-Spielmeyer-Vogt's Disease	http://omim.org/entry/204200 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=79264	CLN3
Neuronal Ceroid Lipofuscinosis Type 5, Finnish Variant	http://www.omim.org/entry/256731 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=168491	CLN5
Neuronal Ceroid Lipofuscinosis Type 8 & Progressive Epilepsy and	http://omim.org/entry/610003 http://www.orpha.net/consor/cgi-	CLN8

Disease	Links	Gene
Mental Retardation	bin/OC_Exp.php?Expert=1947	
Niemann-Pick's Disease	http://omim.org/entry/257220 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=646 http://omim.org/entry/257200 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=77292	NPC1, SMPD1
Nijmegen Breakage Syndrome	http://omim.org/entry/251260 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=647	NBN
Osteopetrosis	http://www.ojrd.com/content/4/1/5 http://omim.org/entry/259700 http://www.orpha.net/consor4.01/www/cgi-bin/OC_Exp.php?lng=EN&Expert=667	TCIRG1
Osteogenesis Imperfecta, Type VII	http://omim.org/entry/610682 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=666	CRTAP
Polycystic Kidney Disease	http://omim.org/entry/263200 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=731	PKHD1
Pontocerebellar Hypoplasia Type 1	http://omim.org/entry/607596 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=2254	VRK1
Pontocerebellar Hypoplasia Type 2 (A,B & C)	http://www.omim.org/entry/277470 http://www.omim.org/entry/612389 http://www.omim.org/entry/612390 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2524	TSEN54, TSEN2, TSEN34
Pontocerebellar Hypoplasia Type 6	http://omim.org/entry/611523 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=166073	RARS2
Rhizomelic Chondrodysplasia Punctata	http://omim.org/entry/215100 http://omim.org/entry/222765 http://omim.org/entry/600121 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=177	PEX7, GNPAT, AGPS
Sjögren-Larsson Syndrome	http://omim.org/entry/270200 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=816	ALDH3A2
Spinal Musculaire Atrophy (SMA), type 0/1)	http://omim.org/entry/253300 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=70	SMN1
Smith-Lemli-Opitz Syndrome	http://omim.org/entry/270400 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=818	DHCR7
Zellweger Syndrome	http://omim.org/entry/214100 http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=912	PEX1