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Resources for Genetics Professionals — Genetic Disorders Associated with Founder Variants Common in the Mennonite Population

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A founder variant is a pathogenic variant observed at high frequency in a specific population due to the presence of the variant in a single ancestor or small number of ancestors. The presence of a founder variant can affect the approach to molecular genetic testing. When one or more founder variants account for a large percentage of all pathogenic variants found in a population, testing for the founder variant(s) may be performed first.

The table below includes common founder variants – here defined as **three or fewer variants that account for >50% of the pathogenic variants identified in a single gene in individuals of a specific ancestry** – in individuals of Mennonite ancestry. Note: Pathogenic variants that are common worldwide due to a DNA sequence hot spot are not considered founder variants and thus are not included.

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Table. Genetic Disorders Associated with Founder Variants Common in the Mennonite Population

Gene	Disorder	MOI	DNA Nucleotide Change (Alias [1])	Predicted Protein Change (Alias [1])	Proportion of Pathogenic Variants in Gene [2]	Carrier Frequency	Ethnicity (Specific Region or Conference)	Reference Sequences	Reference(s) [3]
ACADM	Medium-chain acyl-coenzyme A dehydrogenase deficiency	AR	c.287-30A>G (IVS4-30A>G)	--	<100% [4]	Unknown	Mennonite (Weaverland & Groffdale)	NM_000016.6	Strauss & Puffenberger [2009]
ALMS1	Alström syndrome	AR	c.10480C>T	p.Gln3494Ter	~100% [5]	Unknown	Mennonite (Durango State, Mexico)	NM_001378454.1 NP_001365383.1	Cruz-Aguilar et al [2017]
ALPL	Hypophosphatasia	AR	c.1001G>A	p.Gly334Asp	<100% [4]	1/25	Mennonite (Manitoba, Canada)	NM_000478.6 NP_000469.3	Greenberg et al [1993]
AMN	Megaloblastic anemia (OMIM 261100)	AR	c.689_733del45	p.Leu230_Arg244del	~100% [5]	Unknown	Mennonite (Weaverland & Groffdale)	NM_030943.4 NP_112205.2	Strauss & Puffenberger [2009]
APC	*APC*-associated polyposis conditions	AD	del promoter 1A & 5'UTR [1]	--	<100%	NA	Mennonite (Manitoba, Canada)	NG_008481.4	Charames et al [2008]
ATM	Ataxia-telangiectasia	AR	c.6200C>A	p.Ala2067Asp	~80%	Unknown	Mennonite (Canada)	NM_000051.4 NP_000042.3	Saunders-Pullman et al [2012], Nakamura et al [2014]
BCKDHA	Maple syrup urine disease	AR	c.1312T>A	p.Tyr438Asn	~100% [5]	1/6 to 1/10	Mennonite (Weaverland, Groffdale, & Stauffer)	NM_000709.4 NP_000700.1	Strauss & Puffenberger [2009]
BTD	Biotinidase deficiency	AR	c.1399T>C (1459T>C)	p.Trp487Arg	<100% [4]	Unknown	Mennonite (Weaverland & Groffdale)	NM_001370658.1 NP_001357587.1	
CACNA1F	Congenital stationary night blindness	XL	c.3166dupC	p.Leu1056ProfsTer11	~80%	NA	Mennonite	NM_005183.4 NP_005174.2	Bech-Hansen et al [1998]
CD3D	SCID type 19 (OMIM 615617)	AR	c.202C>T	p.Arg68Ter	~100% [5]	Unknown	Mennonite	NM_000732.6 NP_000723.1	Dadi et al [2003]
CFTR	Cystic fibrosis	AR	c.1521_1523delCTT	p.Phe508del	<100% [4]	Unknown	Mennonite	NM_000492.4 NP_000483.3	Henderson & Anbar [2009]
CRADD	Intellectual disability with variant lissencephaly (OMIM 614499)	AR	c.382G>C	p.Gly128Arg	~100% [5]	1/51	Mennonite (Pennsylvania)	NM_003805.5 NP_003796.1	Puffenberger et al [2012a]

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CTNS	Cystinosis	AR	c.1015G>A	p.Gly339Arg	~100% [5]	Unknown	Amish Mennonite (Ontario, Canada)	NM_004937.3 NP_004928.2	Rupar et al [2001]
CYP17A1	17-alpha-hydroxylate / 17,20-lyase deficiency (OMIM 202110)	AR	c.1435_1438dupATCC	p.Pro480HisfsTer27	~100% [5]	Unknown	Mennonite (Canada)	NM_000102.4 NP_000093.1	Imai et al [1992]
DGUOK	Deoxyguanosine kinase deficiency	AR	c.763G>T	p.Asp255Tyr	~100% [5]	Unknown	Mennonite (Ontario, Canada)	NM_080916.3 NP_550438.1	Tadiboyina et al [2005]
EDNRB	Hirschsprung disease (OMIM 600155)	AD AR	c.828G>T	p.Trp276Cys	~100% [5]	NA	Mennonite (Groffdale, Stauffer, & Weaverland)	NM_000115.5 NP_000106.1	Puffenberger et al [1994], Strauss & Puffenberger [2009]
EXT2	Seizures, scoliosis, & macrocephaly/ microcephaly syndrome (OMIM 616682)	AR	c.[260T>G;283C>T] [6]	p.[Met87Arg;Arg95Cys] [6]	~100% [5]	1/26	Mennonite (Canada)	NM_207122.2 NP_997005.1	Farhan et al [2015]
F11	Factor XI deficiency (OMIM 612416)	AD AR	c.1327C>T	p.Arg443Cys	~100% [5]	NA	Mennonite (Weaverland & Groffdale)	NM_000128.4 NP_000119.1	Strauss & Puffenberger [2009]
FANCC	Fanconi anemia	AR	c.67delG	p.Asp23IlefsTer23	~100% [5]	Unknown	Mennonite (Manitoba, Canada, & Tamaulipas, Mexico)	NM_000136.3 NP_000127.2	de Vries et al [2012], García-de Teresa et al [2019]
FLVCR1	Posterior column ataxia with retinitis pigmentosa (OMIM 609033)	AR	c.361A>G	p.Asn121Asp	~100% [5]	1/51	Mennonite (Pennsylvania)	NM_014053.4 NP_054772.1	Puffenberger et al [2012a]
GORAB	Geroderma osteodysplasticum (OMIM 231070)	AR	c.136G>T	p.Glu46Ter (Glu123Ter)	~100% [5]	<1/50	Mennonite (Canada, Germany, & Mexico)	NM_152281.3 NP_689494.3	Hennies et al [2008]
GPSM2	Chudley-McCullough syndrome (OMIM 604213)	AR	c.1473delG	p.Phe492SerfsTer5	~71%	Unknown	Mennonite	NM_013296.5 NP_037428.3	Doherty et al [2012]
			c.742delC	p.Gly249GlufsTer32	~29%				

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GUSB	Mucopolysaccharidosis type VII	AR	c.526C>T	p.Leu176Phe	~100% [5]	Unknown	Mennonite	NM_000181.4 NP_000172.2	Wu et al [1994]
HERC2	Intellectual developmental disorder type 38 (OMIM 615516)	AR	c.1781C>T	p.Pro594Leu	~100% [5]	Unknown	Amish Mennonite (US)	NM_004667.6 NP_004658.3	Puffenberger et al [2012b]
HPD	Tyrosinemia type III (OMIM 276710)	AR	c.479A>G	p.Tyr160Cys	<100% [4]	Unknown	Mennonite (W Maryland)	NM_002150.3 NP_002141.2	Strauss & Puffenberger [2009]
			c.1005C>G	p.Ile335Met					
IL7R	SCID type 104 (OMIM 608971)	AR	c.2T>G	p.M1?	~100% [5]	Unknown	Mennonite (Weaverland & Groffdale)	NM_002185.5 NP_002176.2	
LAMB2	Nephrotic syndrome type 5 (OMIM 614199)	AR	c.440A>G	p.His147Arg	~100% [5]	1/48	Mennonite (US)	NM_002292.4 NP_002283.3	Mohney et al [2011]
LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein (OMIM 610798)	AR	c.*23C>A [7] (IVS4+23C>A)	--	~100% [5]	Unknown	Mennonite	NM_014017.4	Bohn et al [2007]
LRP5	Osteoporosis-pseudoglioma syndrome (OMIM 259770)	AR	c.1275G>A	p.Trp425Ter	80%	Unknown	Mennonite (US)	NM_002335.4 NP_002326.2	Streeten et al [2008]
MCCC2	3-methylcrotonyl-coenzyme A carboxylase 2 deficiency (OMIM 210210)	AR	c.517dupT	p.Ser173PhefsTer25	<100% [4]	Unknown	Mennonite	NM_022132.5 NP_071415.1	Gallardo et al [2001], Strauss & Puffenberger [2009]
MLH1	Lynch syndrome	AD	c.2141G>A	p.Trp714Ter	<100% [4]	NA	Mennonite (Manitoba, Canada)	NM_000249.4 NP_000240.1	Orton et al [2008]
MVK	Mevalonate aciduria (OMIM 610377)	AR	c.803T>C	p.Ile268Thr	<100% [4]	Unknown	Mennonite (Weaverland & Groffdale)	NM_000431.4 NP_000422.1	Strauss & Puffenberger [2009]
NOL3	Familial cortical myoclonus (OMIM 614937)	AD	c.61G>C	p.Glu21Gln	~100% [5]	NA	Mennonite (Canada)	NM_001276312.2 NP_001263241.1	Russell et al [2012]
NPHS1	Nephrotic syndrome type 1 (OMIM 256300)	AR	c.1481delC	p.Ser494CysfsTer55	<100% 4	1/12	Old Order Mennonite (Lancaster Co, Pennsylvania)	NM_004646.4 NP_004637.1	Bolk et al [1999]

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NPRL3	Familial focal epilepsy with variable foci 3 (OMIM 617118)	AD	c.349delG	p.Glu117LysfsTer5	<100%	NA	Mennonite (Pennsylvania)	NM_001077350.3 NP_001070818.1	Iffland et al [2022]
PAH	Phenylalanine hydroxylase deficiency	AR	c.1315+1G>A (IVS12+1G>A)	--	<100% [4]	Unknown	Mennonite (Weaverland & Groffdale)	NM_000277.3	Strauss & Puffenberger [2009]
			c.782G>A	p.Arg261Gln				NM_000277.3 NP_000268.1	
PCCB	Propionic acidemia	AR	c.1606A>G	p.Asn536Asp	~100% [5]	Unknown	Mennonite (Weaverland, Groffdale, & Stauffer)	NM_000532.5 NP_000523.2	
PQBP1	Renpenning syndrome (OMIM 309500)	XL	c.640dupC	p.Arg214ProfsTer13	~100% [5]	NA	Mennonite (Canada)	NM_005710.2 NP_005701.1	Lenski et al [2004]
PYGL	Glycogen storage disease type VI	AR	c.1620+1G>A (IVS13+1G>A)	--	~100% [5]	1/17	Mennonite (Weaverland & Groffdale)	NM_002863.5	Chang et al [1998], Strauss & Puffenberger [2009]
SLC3A1	Cystinuria (OMIM 220100)	AD AR	c.1136+2T>C (IVS6+2T>C)	--	~100% [5]	NA	Mennonite (Weaverland & Groffdale)	NM_000341.4	Strauss & Puffenberger [2009]
			c.1354C>T	p.Arg452Trp				NM_000341.4 NP_000332.2	
SLC7A9		AD AR	c.1166C>T	p.Thr389Met	<100% [4]			NM_014270.5 NP_055085.1	
SLC6A3	*SLC6A3*-related dopamine transporter deficiency syndrome	AR	c.1269+1G>T (IVS9+1G>T)	--	~100% [5]	Unknown	Mennonite (Pennsylvania)	NM_001044.5	Puffenberger et al [2012a]
SLC17A5	Salla disease (See Free Sialic Acid Storage Disorders.)	AR	c.115C>T	p.Arg39Cys	~100% [5]	Unknown	Mennonite (W Maryland)	NM_012434.5 NP_036566.1	Strauss et al [2005], Strauss & Puffenberger [2009]
SLC25A4	Hypertrophic cardiomyopathy (OMIM 615418)	AR	c.523delC	p.Gln175ArgfsTer38	~100% [5]	Unknown	Mennonite (W Maryland)	NM_001151.4 NP_001142.2	Strauss et al [2013]

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STRADA	Polyhydramnios, megalencephaly, & symptomatic epilepsy (OMIM 611087)	AR	c.581+1100_*1511del7304	--	~100% [5]	1/25	Mennonite (Weaverland & Groffdale)	NM_001003787.4	Puffenberger et al [2007], Strauss & Puffenberger [2009]
TH	Tyrosine hydroxylase deficiency	AR	c.698G>A	p.Arg233His	~100% [5]	Unknown	Mennonite (Weaverland & Groffdale)	NM_199292.3 NP_954986.2	Strauss & Puffenberger [2009]
THAP1	DYT-THAP1 (OMIM 602629)	AD	c.135_139delTAAACinsGGGTTTA	p.Phe45LeufsTer29	~90%	NA	Amish Mennonite	NM_018105.3 NP_060575.1	Fuchs et al [2009], Saunders-Pullman et al [2014]
TNFRSF1A	Periodic fever (TRAPS) (OMIM 142680)	AD	c.362G>A	p.Arg121Gln	<100%	NA	Mennonite (W Maryland)	NM_001065.4 NP_001056.1	Strauss & Puffenberger [2009]
TUBGCP6	Microcephaly & chorioretinopathy (OMIM 251270)	AR	c.5458T>G	p.Ter1820Gly	~100% [5]	1/51	Mennonite (Pennsylvania)	NM_020461.4 NP_065194.3	Puffenberger et al [2012a]
UGT1A1	Crigler-Najjar syndrome type 1 (OMIM 218800)	AR	c.222C>A	p.Tyr74Ter	~100% [5]	Unknown	Mennonite (Weaverland & Groffdale)	NM_000463.3 NP_000454.1	Strauss & Puffenberger [2009]
ZAP70	*ZAP70*-related combined immunodeficiency	AR	c.1624-11G>A (IVS12-11G>A)	--	~100% [5]	1/10	Mennonite (Ontario, Canada)	NM_001079.4	Schroeder et al [2016]

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ZMPSTE24	Lethal restrictive dermopathy (OMIM 275210)	AR	c.1085dupT	p.Leu362PhefsTer19	~100% [5]	Unknown	Mennonite (Canada & South Dakota)	NM_005857.5 NP_005848.2	Loucks et al [2012]
			c.54dupT	p.Ile19TyrfsTer28			Mennonite (Canada & Pennsylvania)		Moulson et al [2005], Strauss & Puffenberger [2009]

Included if ≤3 pathogenic variants account for ≥50% of variants identified in a specific ethnic group
AD = autosomal dominant; AR = autosomal recessive; Co = county; MOI = mode of inheritance; NA = not applicable; SCID = severe combined immunodeficiency; US = United States; W = Western; XL = X-linked
1. Does not conform to standard HGVS nomenclature
2. This percentage does not account for the possibility of rare *de novo* pathogenic variants occurring in this population.
3. See also Clinic for Special Children and Amish, Mennonite, and Hutterite Genetic Disorders Database.
4. At least one additional variant reported in this population in ≥1 family
5. To date, additional pathogenic variants in this gene have not been reported in individuals of Mennonite decent.
6. Affected individuals have been found to be homozygous for both pathogenic variants listed.
7. Nucleotide substitution is located 23 bp after termination codon and introduces a splice site.

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- 1 June 2023 (sw) Initial posting

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