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Resources for Genetics Professionals — Genetic Disorders Associated with Founder Variants Common in the Hutterite 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 more than 50% of the pathogenic variants identified in a single gene in individuals of a specific ancestry** – in individuals of Hutterite 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 Hutterite Population

Gene	Disorder	MOI	DNA Nucleotide Change	Predicted Protein Change	Proportion of Pathogenic Variants in Gene	Carrier Frequency	Leut/Ethnicity (Specific Region)	Reference Sequences	References
ABCC8	Permanent neonatal diabetes mellitus	AR	See footnote 1.	–	~100% [2]	Unknown	Schmeideleut	NM_000352.4	Triggs-Raine et al [2016]
ABCG8	Sitosterolemia	AR	c.320C>G	p.Ser107Ter	~100% [2]	1/12 to 1/16	Schmeideleut	NM_022437.2 NP_071882.1	Chong et al [2012], Triggs-Raine et al [2016]
ALPL	Hypophosphatasia	AR	c.1001G>A	p.Gly334Asp	~100% [2]	Unknown	Dariusleut	NM_000478.5 NP_000469.3	Triggs-Raine et al [2016]
AR	Androgen insensitivity syndrome	XL	c.2033T>C	p.Leu678Pro	~100% [2]	Unknown	Schmiedeleut (Manitoba)	NM_000044.4 NP_000035.2	Belsham et al [1995]
BBS2	Bardet-Biedl syndrome	AR	c.472-2A>G	–	~100% [2]	1/22 to 1/36	Dariusleut & Schmiedeleut	NM_031885.3	Chong et al [2012], Triggs-Raine et al [2016]
BCHE	Succinylcholine sensitivity (OMIM 617936)	AR	c.293A>G	p.Asp98Gly	~100% [2]	1/21	Schmiedeleut	NM_000055.3 NP_000046.1	Zelinski et al [2007], Triggs-Raine et al [2016]
BCKDHB	Maple syrup urine disease	AR	c.595_596delAG	p.Pro200Ter	Unknown	Unknown	Hutterite	NM_183050.3 NP_898871.1	Mroch et al [2014]
CFTR	Cystic fibrosis	AR	c.1521_1523delCTT	p.Phe508del	31%	1/33 to 1/45	Dariusleut, Lehrerleut, & Schmiedeleut	NM_000492.3 NP_000483.3	Zielenski et al [1993], Chong et al [2012], Triggs-Raine et al [2016]
			c.3302T>A	p.Met1101Lys	69%	1/14 to 1/16			
CPT1A	Carnitine palmitoyltransferase 1A deficiency	AR	c.2129G>A	p.Gly710Glu	~100% [2]	1/15 to 1/16	Dariusleut & Schmiedeleut	NM_001876.3 NP_001867.2	Prasad et al [2001], Triggs-Raine et al [2016]
CSPP1	Joubert syndrome	AR	c.363_364delTA	p.His121GlnfsTer22	~100% [2]	Unknown	Schmiedeleut	NM_024790.6 NP_079066.5	Shaheen et al [2014]

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Gene	Disorder	MOI	DNA Nucleotide Change	Predicted Protein Change	Proportion of Pathogenic Variants in Gene	Carrier Frequency	Leut/Ethnicity (Specific Region)	Reference Sequences	References
DNAJC19	Dilated cardiomyopathy with ataxia syndrome (OMIM 610198)	AR	c.130-1G>C	–	~100% [2]	1/21 to 1/34	Dariusleut, Lehrerleut, & Schmiedeleut	NM_145261.3	Davey et al [2006], Chong et al [2012], Triggs-Raine et al [2016]
DPH1	Louks-Innes syndrome (OMIM 616901)	AR	c.17T>A	p.Met6Lys	~100% [2]	1/32	Dariusleut	NM_001383.4 NP_001374.3	Loucks et al [2015], Triggs-Raine et al [2016]
DSC2	Arrhythmogenic right ventricular cardiomyopathy	AR	c.1660C>T	p.Gln554Ter	~100% [2]	1/8 to 1/11	Dariusleut, Lehrerleut, & Schmiedeleut	NM_024422.4 NP_077740.1	Gerull et al [2013], Wong et al [2014], Triggs-Raine et al [2016]
EMG1	Bowen-Conradi syndrome (OMIM 211180)	AR	c.257A>G	p.Asp86Gly	~100% [2]	1/10 to 1/21	Dariusleut, Lehrerleut, & Schmiedeleut	NM_006331.7 NP_006322.4	Armistead et al [2009], Triggs-Raine et al [2016]
FKRP	Limb-girdle muscular dystrophy type 2I (OMIM 607155)	AR	c.826C>A	p.Leu276Ile	~100% [2]	1/7 to 1/9	Dariusleut, Lehrerleut, & Schmiedeleut	NM_024301.4 NP_077277.1	Frosk et al [2005], Chong et al [2012], Triggs-Raine et al [2016]
GJB2	*GJB2*-related autosomal recessive nonsyndromic hearing loss	AR	c.35delG	p.Gly12ValfsTer2	~100% [2]	1/28 to 1/65	Dariusleut & Schmiedeleut	NM_004004.5 NP_003995.2	Chong et al [2012], Triggs-Raine et al [2016]
LEMD2	Juvenile-onset cataract and arrhythmic cardiomyopathy (OMIM 212500)	AR	c.38T>G	p.Leu13Arg	~100% [2]	1/8	Schmiedeleut, Lehrerleut	NM_181336.3 NP_851853.1	Boone et al [2015], Abdelfatah et al [2019]

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Gene	Disorder	MOI	DNA Nucleotide Change	Predicted Protein Change	Proportion of Pathogenic Variants in Gene	Carrier Frequency	Leut/Ethnicity (Specific Region)	Reference Sequences	References
LMNA	Emery-Dreifuss muscular dystrophy	AR	c.1445G>A	p.Arg482Gln	~100% [2]	1/69	Dariusleut & Lehrerleut (Alberta, Canada)	NM_170707.3 NP_733821.1	Wiltshire et al [2013]
MMUT	Methylmalonic acidemia	AR	c.1420C>T	p.Arg474Ter	~100% [2]	1/63	Dariusleut	NM_000255.3 NP_000246.2	Triggs-Raine et al [2016]
MYO7A	Usher syndrome 1	AR	c.52C>T	p.Gln18Ter	~100% [2]	Unknown	Dariusleut	NM_000260.3 NP_000251.3	Chong et al [2012]
NDUFS4	Leigh syndrome	AR	c.393dupA	p.Glu132ArgfsTer15	~100% [2]	1/13	Dariusleut & Schmiedeleut	NM_002495.3 NP_002486.1	Triggs-Raine et al [2016]
PCDH15	Usher syndrome 1	AR	c.1088delT	p.Leu363TrpfsTer58	~100% [2]	1/21 to 1/40	Schmeideleut [3]	NM_033056.3 NP_149045.3	Alagramam et al [2001], Chong et al [2012]
PIBF1	Joubert syndrome	AR	c.1910A>C	p.Asp637Ala	~100% [2]	Unknown	Schmiedeleut	NM_006346.3 NP_006337.2	Wheway et al [2015]
PROP1	*PROP1*-related combined pituitary hormone deficiency	AR	c.301_302delAG	p.Leu102CysfsTer8	~100% [2]	Unknown	Hutterite	NM_006261.4 NP_006252.3	Wu et al [1998]
SLC39A8	Congenital disorder of glycosylation type IIn	AR	c.112G>C	p.Gly38Arg	~100% [2]	1/59	Lehrerleut	NM_022154.5 NP_071437.3	Boycott et al [2015]
						1/26	Dariusleut		
SLC5A5	Thyroid dyshormonogenesis I (OMIM 274400)	AR	c.1183G>A	p.Gly395Arg	~100% [2]	Unknown	Dariusleut, Lehrerleut, & Schmiedeleut	NM_000453.2 NP_000444.1	Kosugi et al [1999]
SMN1	Spinal muscular atrophy type III	AR	Del entire gene [4]	–	<100% [5]	1/8	Lehrerleut, Schmeideleut (South Dakota) [3]	NM_000344.3 NP_000335.1	Chong et al [2011]
SQOR	Leigh syndrome	AR	c.637G>A	p.Glu213Lys	~100% [2]	1/13	Lehrerleut	NM_021199.4 NP_067022.1	Friederich et al [2020]

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Gene	Disorder	MOI	DNA Nucleotide Change	Predicted Protein Change	Proportion of Pathogenic Variants in Gene	Carrier Frequency	Leut/Ethnicity (Specific Region)	Reference Sequences	References
TECR	Nonsyndromic mental retardation (OMIM 614020)	AR	c.545C>T	p.Pro182Leu	~100% [2]	1/14 to 1/63	Schmeideleut	NM_138501.5 NP_612510.1	Chong et al [2012], Triggs-Raine et al [2016]
TH	Tyrosine hydroxylase deficiency	AR	c.1481C>T	p.Thr494Met	~100% [2]	Unknown	Dariusleut	NM_199292.2 NP_954986.2	Boycott et al [2008]
THOC6	Beaulieu-Boycott-Innes syndrome	AR	c.136G>A	p.Gly46Arg	~100% [2]	1/33	Dariusleut	NM_024339.4 NP_0778315.2	Beaulieu et al [2013]
						1/50	Lehrerleut		
TMEM237	Joubert syndrome	AR	c.52C>T	p.Arg18Ter	~100% [2]	1/12.5	Schmiedeleut	NM_001044385.2 NP_001037850.1	Chong et al [2012]
						1/17	Canadian Hutterites		Huang et al [2011]
TRAPPC11	Limb-girdle muscular dystrophy type 18 (OMIM 615356)	AR	c.1287+5G>A	p.Ala372_Ser429del	~100% [1]	1/8 to 1/14	Dariusleut & Schmeideleut	NM_021942.5	Bögershausen et al [2013], Triggs-Raine et al [2016]
TRIM32	Limb-girdle muscular dystrophy type 2H (OMIM 254110)	AR	c.1459G>A	p.Asp487Asn	~100% [2]	1/6.5 to 1/31	Dariusleut, Lehrerleut, & Schmiedeleut	NM_012210.3 NP_036342.2	Chong et al [2012], Triggs-Raine et al [2016]
TYR	Oculocutaneous albinism type 1 (OMIM 203100, 606952)	AR	c.272G>A	p.Cys91Tyr	~100% [2]	1/7 to 1/9	Dariusleut, Lehrerleut, & Schmiedeleut	NM_000372.4 NP_000363.1	Chong et al [2012], Triggs-Raine et al [2016]
VLDLR	VLDLR cerebellar hypoplasia	AR	Del entire gene Chr9:2479657-2678818del	–	~100% [2]	1/15	Lehrerleut & Dariusleut	NC_000009.12 (GRCh18.p13)	Glass et al [2005]

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Gene	Disorder	MOI	DNA Nucleotide Change	Predicted Protein Change	Proportion of Pathogenic Variants in Gene	Carrier Frequency	Leut/Ethnicity (Specific Region)	Reference Sequences	References
ZMPSTE24	Restrictive dermopathy (OMIM 275210)	AR	c.1085dupT	p.Leu362PhefsTer19	~100% [2]	1/15.5 to 1/21	Dariusleut, Lehrerleut, & Schmiedeleut	NM_005857.4 NP_005848.2	Loucks et al [2012], Chong et al [2012]

Included if ≤3 pathogenic variants account for ≥50% of variants identified in a specific ethnic group; see also www.biochemgenetics.ca/plainpeople.

AR = autosomal recessive; MOI = mode of inheritance; XL = X-linked

1. The c.823-7T>A variant is reported in Triggs-Raine et al [2016], attributed to a personal communication. No data regarding the pathogenicity of the variant are provided.

2. To date, additional pathogenic variants in this gene have not been reported in individuals of Hutterite descent.

3. Carrier frequency reported in Schmiedeleut population residing in United States [Chong et al 2011, Chong et al 2012, Gerull et al 2013]; carrier frequency in Lehrerleut and Dariusleut not reported to date

4. Individuals of Hutterite ancestry with *SMN1* founder deletion have 0 *SMN1* and 2 *SMN2* copies per chromosome.

5. De novo pathogenic variants have been reported in Hutterite population [Chong et al 2011].

Revision History

- 1 December 2022 (sw) Revision: removed *NPHP1* variants
- 7 May 2020 (sw) Initial posting

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