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Resources for Genetics Professionals — Genetic Disorders Associated with Founder Variants Common in the Druze 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 Druze ancestry. Note: (1) Pathogenic variants that are common worldwide due to a DNA sequence hot spot are not considered founder variants and thus are not included. (2) Disorders with a carrier frequency ≤1/200 are not included.

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

Gene	Disorder	MOI	DNA Nucleotide Change (Alias [1])	Predicted Protein Change	% of Pathogenic Variants in Gene [2]	Carrier Frequency	Ethnicity (Specific Region)	Reference Sequences	References
ABCA4	Cone-rod dystrophy 3 (OMIM 604116)	AR	c.634C>T	p.Arg212Cys	<100% [3]	Unknown	Druze (Israel)	NM_000350.3 NP_000341.2	Sharon et al [2020]
ASL	Argininosuccinate lyase deficiency	AR	c.346C>T	p.Gln116Ter	~100% [4]	1/41	Druze (Galilee, Israel)	NM_000048.4 NP_000039.2	Falik-Zaccai et al [2008a]
			c.446+1G>A (IVS5+1G>A)	--		1/40	Druze (Israel)	NM_000048.4	Avnat et al [2023]
ATM	Ataxia-telangiectasia; *ATM*-related cancer predisposition	AR AD	c.6672_6680delGGCTCTACGinsCTC	p.Met2224_Arg2227delinsIleSer	~70%	1/3 to 1/6	Druze (N Israel) 5	NM_000051.4 NP_000042.3	Fares et al [2004], Avnat et al [2023]
			c.1339C>T	p.Arg447Ter	~30%	1/6 to 1/15	Druze (N Israel) 6		
						1/59	Druze (Israel)		
ATP7B	Wilson disease	AR	c.3649_3654delGTTCTG	p.Val1217_Leu1218del	<100% [3]	~1/28	Druze (Israel)	NM_000053.4 NP_000044.2	Kalinsky et al [1998]
BCKDHB	Maple syrup urine disease	AR	c.1016C>T	Ser339Leu	~88%	Unknown	Druze (N Israel)	NM_183050.4 NP_898871.1	Wynn et al [2001]
CDH3	Congenital hypotrichosis with juvenile macular dystrophy (OMIM 601553)	AR	c.981delG	p.Met327IlefsTer23	<100% [3]	Unknown	Druze (N Israel)	NM_001793.6 NP_001784.2	Sprecher et al [2001]
CFAP418 (*C8orf37*)	Retinitis pigmentosa	AR	c.545A>G	p.Gln182Arg	~100% [4]	1/45	Druze (Israel)	NM_177965.4 NP_808880.1	Estrada-Cuzcano et al [2012]
CPS1	Carbamoylphosphate synthetase I deficiency (See Urea Cycle Disorders Overview.)	AR	c.3265C>T	p.Arg1089Cys	~100% [4]	1/95	Druze (Galilee, Israel)	NM_001875.5 NP_001866.2	Falik-Zaccai et al [2008a]
CYP1B1	Primary congenital glaucoma	AR	c.1405C>T	p.Arg469Trp	>60%	Unknown	Druze (Israel)	NM_000104.4 NP_000095.2	Geyer et al [2011]
CYP27A1	Cerebrotendinous xanthomatosis	AR	c.355delC	p.Arg119GlyfsTer24	~100% [4]	1/11 to 1/30	Druze (Yarka, Galilee, Israel)	NM_000784.4 NP_000775.1	Leitersdorf et al [1994], Falik-Zaccai et al [2008a], DeBarber et al [2018]

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Gene	Disorder	MOI	DNA Nucleotide Change (Alias [1])	Predicted Protein Change	% of Pathogenic Variants in Gene [2]	Carrier Frequency	Ethnicity (Specific Region)	Reference Sequences	References
DBT	Maple syrup urine disease	AR	c.581C>G	p.Ser194Ter	~100% [4]	Unknown	Druze (N Israel)	NM_001918.5 NP_001909.4	Chuang et al [2004]
DGUOK	Deoxyguanosine kinase deficiency	AR	c.255delA	p.Ala86ProfsTer13	~100% [4]	1/5	Druze (N Israel)	NM_080916.3 NP_550438.1	Mandel et al [2001]
DOLK	Congenital disorder of glycosylation, type Im (See Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview.)	AR	c.1222C>G	p.His408Asp	~100% [4]	Unknown	Druze (Galilee, Israel)	NM_014908.4 NP_055723.1	Lefeber et al [2011]
EHD1	Tubular proteinuria and deafness	AR	c.1192C>T	p.Arg398Trp	~100% [4]	1/196	Druze (Israel)	NM_001282444.2 NP_001269373.1	Issler et al [2022]
ERCC6	Cockayne syndrome	AR	c.1034_1035insT	p.Lys345AsnfsTer24	~100% [4]	1/15	Druze (N Israel)	NM_000124.4 NP_000115.1	Falik-Zaccai et al [2008b]
F7	Factor VII deficiency (OMIM 227500)	AR	c.1109G>T	p.Cys370Phe	<100% [3]	1/40	Druze (Israel)	NM_000131.4 NP_000122.1	Fromovich-Amit et al [2004], Avnat et al [2023]
FANCA	Fanconi anemia	AR	c.3788_3790delTCT	p.Phe1263del	<100% [3]	Unknown	Druze (Israel)	NM_000135.4 NP_000126.2	Steinberg-Shemer et al [2020]
GALC	Krabbe disease	AR	c.1796T>G	p.Ile599Ser	~100% [4]	~1/6 to 1/16	Druze (N Israel)	NM_000153.4 NP_000144.2	Rafi et al [1996], Tappino et al [2010]
GATC	Combined oxidative phosphorylation deficiency 42 (OMIM 618839)	AR	c.233T>G	p.Met78Arg	~100% [4]	1/59	Druze (Israel)	NM_176818.3 NP_789788.1	Friederich et al [2018], Avnat et al [2023]
GNPTG	Mucolipidosis III gamma	AR	c.499dupC	p.Leu167ProfsTer32	<100% [3]	1/59	Druze (N Israel)	NM_032520.5 NP_115909.1	Raas-Rothschild et al [2000], Avnat et al [2023]
HOGA1	Primary hyperoxaluria, type III (OMIM 613616)	AR	c.944_946delAGG	p.Glu315del	~100% [4]	1/20	Druze (Israel)	NM_138413.4 NP_612422.2	Avnat et al [2023]

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Gene	Disorder	MOI	DNA Nucleotide Change (Alias [1])	Predicted Protein Change	% of Pathogenic Variants in Gene [2]	Carrier Frequency	Ethnicity (Specific Region)	Reference Sequences	References
IDUA	Mucopolysaccharidosis type 1	AR	c.1096A>C	p.Thr366Pro	~60%	1/39	Druze (Galilee, Israel)	NM_000203.5 NP_000194.2	Bach et al [1993], Avnat et al [2023]
INSR	*INSR*-related severe syndromic insulin resistance	AR	c.167T>C	p.Ile56Thr	~100% [4]	~1/50	Druze (Israel)	NM_000208.4 NP_000199.2	Falik-Zaccai et al [2014]
LDLR	Familial hypercholesterolemia	AD AR	c.564C>G	p.Tyr188Ter	<100% [3]	Unknown	Druze (Golan Heights)	NM_000527.5 NP_000518.1	Landsberger et al [1992]
MCOLN1	Mucolipidosis IV	AR	c.964C>T	p.Arg322Ter	<100% [3]	Unknown	Druze (Golan Heights)	NM_020533.3 NP_065394.1	Bargal et al [2001]
MEFV	Familial Mediterranean fever	AR	c.442G>C	p.Glu148Gln	52%	Unknown	Druze (Israel)	NM_000243.3 NP_000234.1	Sharkia et al [2013], Avnat et al [2023]
			c.2177T>C	p.Val726Ala	41%				
			c.2082G>A	p.Met694Ile	7%	1/40			
MUTYH	*MUTYH* polyposis	AR	c.1437_1439delGGA	p.Glu480del	~100% [4]	1/34	Druze (Israel)	NM_001128425.2 NP_001121897.1	Avnat et al [2023], Reznick Levi et al [2023]
NARS2	Combined oxidative phosphorylation deficiency 24 (OMIM 616239)	AR	c.500A>G	p.His167Arg	~100% [4]	Unknown	Druze (Israel)	NM_024678.6 NP_078954.4	Mizuguchi et al [2017]
NGLY1	*NGLY1*-related congenital disorder of deglycosylation	AR	c.1294G>T	p.Glu432Ter	~100% [4]	1/25	Druze (village in Israel)	NM_018297.4 NP_060767.2	Kalfon et al [2022]
PCNT	Microcephalic osteodysplastic primordial dwarfism type II	AR	c.3465-1G>A (IVS17-1G>A)	--	~100% [4]	1/30	Druze (N Israel)	NM_006031.6	Weiss et al [2020]
PEPD	Prolidase deficiency	AR	c.605C>T	p.Ser202Phe	~85%	1/21	Druze (Galilee, Israel)	NM_000285.4 NP_000276.2	Falik-Zaccai et al [2008a], Falik-Zaccai et al [2010]
RDH5	Fundus albipunctatus (OMIM 136880)	AD AR	c.712G>T	p.Gly238Trp	~100% [4]	1/40	Druze (Israel)	NM_002905.5 NP_002896.2	Sharon et al [2020], Avnat et al [2023]

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Gene	Disorder	MOI	DNA Nucleotide Change (Alias [1])	Predicted Protein Change	% of Pathogenic Variants in Gene [2]	Carrier Frequency	Ethnicity (Specific Region)	Reference Sequences	References
ROGDI	Kohlschutter-Tonz syndrome (OMIM 226750)	AR	c.469C>T	p.Arg157Ter	~100% [4]	~1/10	Druze (N Israel)	NM_024589.3 NP_078865.1	Mory et al [2012]

Included if ≤3 pathogenic variants account for ≥50% of variants identified in a specific ethnic group
AD = autosomal dominant; AR = autosomal recessive; MOI = mode of inheritance; N = northern
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. At least one additional variant reported in this population in ≥1 family (See Israeli Medical Genetic Database.)
4. To date, additional pathogenic variants in this gene have not been reported in individuals of Druze descent.
5. Druze population originally from Lebanon and Syria.
6. Druze population originally from central Lebanon and Jordan

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Revision History

- 28 September 2023 (sw) Revision: added *ABCA4*, *EHD1*, *FANCA*, *HOGA1*, *MUTYH*, *NGLY1*, and *RDH5*; updated reference sequences
- 16 December 2021 (ha) Revision: added Falik-Zaccai et al [2008a]
- 7 November 2019 (sw) Initial posting

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