Title: *DEPDC5*-Related Epilepsy *GeneReview* Table 3
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Table 3. Select *DEPDC5* Variants

Variant Classification	DNA Nucleotide Change	Predicted Protein Change	Reference Sequences	References
Pathogenic	c.21C>G	p.Tyr7Ter	NM_001242896.1 NP_001229825.1	Dibbens et al [2013] Scheffer et al [2014]
	c.59-1G>C			Lal et al [2014]
	c.132dupC	p.Asn45GlnfsTer3		D'Gama et al [2015]
	c.193+1G>A			Dibbens et al [2013], Carvill et al [2015]
	c.279+1G>A			Dibbens et al [2013], Scheffer et al [2014]
	c.418C>T	p.Gln140Ter		Scheffer et al [2014]
	c.435G>A	p.Trp145Ter		Ricos et al [2016]
	c.454_455delAT	p.Met152ValfsTer6		Weckhuysen et al [2016]
	c.484-1G>A			Baulac et al [2015]
	c.489_491delGTT	p.Phe164del		Dibbens et al [2013]
	c.492_496delTCGTT	p.Arg165TyrfsTer14		Ricos et al [2016]
	c.526C>T	p.Gln176Ter		Ricos et al [2016]
	c.624+1G>A			D'Gama et al [2015]
	c.730C>T	p.Gln244Ter		Nascimento et al [2015]
	c.715C>T	p.Arg239Ter		Ishida et al [2013], Baulac et al [2015]
	c.727C>T	p.Arg243Ter		Lal et al [2014], Ricos et al [2016]
	c.783_786delTGAG	p.Asn261LysfsTer11		D'Gama et al [2015]
	c.856C>T	p.Arg286Ter		Bagnall et al [2016]
	c.918C>G	p.Tyr306Ter		Striano et al [2015], Pippucci et al [2015]
	c.982C>T	p.Arg328Ter		Ishida et al [2013]
	c.985delA	p.Thr329LeufsTer7		Ricos et al [2016]
	c.1093_1099dupGATTTGG	p.Val367GlyfsTer20		Ricos et al [2016]
	c.1114C>T	p.Gln372Ter		Ishida et al [2013]
	c.1122delA	p.Leu374PhefsTer30		Ishida et al [2013]
	c.1264C>T	p.Arg422Ter		Baulac et al [2015], Ricos et al [2016]
	c.1393C>T	p.Gln465Ter		Weckhuysen et al [2016]
	c.1459C>T	p.Arg487Ter		Picard et al [2014], Dibbens et al [2013]
	c.1555C>T	p.Gln519Ter		Carvill et al [2015]

Variant Classification	DNA Nucleotide Change	Predicted Protein Change	Reference Sequences	References
	c.1663C>T	p.Arg555Ter		Dibbens et al [2013], Scerri et al [2015]
	c.1746_1752delCATGCTG	p.Leu584PhefsTer12		Weckhuysen et al [2016]
	c.1759C>T	p.Arg587Ter		Baulac et al [2015]
	c.1909C>T	p.Arg637Ter		Ricos et al [2016]
	c.2355-2A>G			Picard et al [2014]
	c.2390delA	p.Gln797ArgfsTer18		Carvill et al [2015]
	c.2527C>T	p.Arg843Ter		Dibbens et al [2013], Bagnall et al [2016], Martin et al [2014]
	c.2620C>T	p.Arg874Ter		Lal et al [2014]
	c.3046C>T	p.Gln1016Ter		Bagnall et al [2016]
	c.3259C>T	p.Arg1087Ter		Picard et al [2014]
	c.3265-3C>T		NM_001242896.1 NP_001229825.1	Ricos et al [2016]
	c.3444delA	p.Ile1148MetfsTer24		Lal et al [2014]
	c.3696+5G>A			Ricos et al [2016]
	c.3802C>T	p.Arg1268Ter		Dibbens et al [2013]
	c.3994C>T	p.Arg1332Ter		Ricos et al [2016], Bagnall et al [2016]
	c.4033+5A>G			Ricos et al [2016]
	c.4107G>A	p.Trp1369Ter		Picard et al [2014], Dibbens et al [2013]
	c.4139delT	p.Leu1380ArgfsTer14		Pippucci et al [2015]
	c.4187delC	p.Ala1396GlufsTer21		Mirzaa et al [2016]
	c.4260delG	p.Glu1421ArgfsTer153		Weckhuysen et al [2016]
	c.4397G>A	p.Trp1466Ter		Dibbens et al [2013]
	c.4567C>T	p.Gln1523Ter		Ishida et al [2013]
	c.4606C>T	p.Gln1536Ter		Dibbens et al [2013]
VUS	c.56G>C	p.Ser19Thr		Bagnall et al [2016]
	c.161A>C	p.Gln54Pro		Ricos et al [2016]
	c.268G>A	p.Val90Ile		Lal et al [2014]
	c.640C>G	p.His214Asp		Ricos et al [2016]
	c.814G>T	p.Val272Leu		Lal et al [2014]
	c.842A>T	p.Tyr281Phe		Carvill et al [2015]
	c.1218-18_1218-15delTGTT			D'Gama et al [2015]
	c.1265G>A	p.Arg422Gln		D'Gama et al [2015]
	c.1355C>T	p.Ala452Val		D'Gama et al [2015]
	c.1454G>A	p.Arg485Gln		Ishida et al [2013]
	c.1625A>C	p.Gln542Pro		Ricos et al [2016]
	c.2591C>T	p.Thr864Met		Martin et al [2014]

Variant Classification	DNA Nucleotide Change	Predicted Protein Change	Reference Sequences	References
	c.3092C>A	p.Pro1031His		Carvill et al [2015]
	c.3217A>C	p.Ser1073Arg		Dibbens et al [2013]
	c.3241A>C	p.Thr1081Pro		Ricos et al [2016]
	c.3311C>T	p.Ser1104Leu		Dibbens et al [2013]
	c.3484A>G	p.Ser1162Gly		Lal et al [2014]
	c.3461C>T	p.Ser1154Phe		Ricos et al [2016]
	c.3803G>A	p.Arg1268Gln		Ricos et al [2016]

VUS: variant of uncertain significance

Note on variant classification: Variants listed in the table have been provided by the authors. *GeneReviews* staff have not independently verified the classification of variants.

Note on nomenclature: *GeneReviews* follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org). See Quick Reference for an explanation of nomenclature.

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