

Title: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders *GeneReview* Table 9

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 9. Published Pathogenic *ASPM* Allelic Variants (Based on NM_009791.4 and NP_033921 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
Large deletion [Passemard et al 2009]	p.0 ?
c.74delG [Nicholas et al 2009]	p.Arg25fs
c.77delG [Passemard et al 2009, Kousar et al 2010]	p.Gly26AlafsTer42Ter
c.117_118del [Tan et al 2013]	p.Leu41GlnfsTer
c.297+1460_3391-242del21844 [Nicholas et al 2009]	Loss of microtubular binding domain
c.349C>T [Bond et al 2002, Kumar et al 2004]	p.Arg117Ter
c.440delA [Nicholas et al 2009]	p.Lys147fs
c.577C>T [Nicholas et al 2009]	p.Gln193Ter
c.637del [Tan et al 2013]	p.Ile213Tyrfs
c.719delCT [Bond et al 2002, Bond et al 2003]	p.Ser240fs
c.803_804del [Tan et al 2013]	p.Lys268Serfs
c.1002delA [Muhammad et al 2009]	p.Val365fs
c.1138C>T [Tan et al 2013]	p.Gln380Ter
c.1152_1153delAG [Nicholas et al 2009]	p.Ser384fs
c.1179delT [Nicholas et al 2009]	p.Pro393fs
c.1258delTCTCAAT [Bond et al 2002, Bond et al 2003]	p.Ser420fs
c.1260delTCAAGTC [Gul et al 2006, Kousar et al 2010]	p.Ser420fs
c.1366G>T [Nicholas et al 2009]	p.Glu456Ter
c.1406_1413delATCCTAAA [Nicholas et al 2009]	p.Asn469fs
c.1590delA [Nicholas et al 2009]	p.Lys530fsTer
c.1630_1634delATCTT [Passemard et al 2009]	p.Tyr544SerfsTer9

DNA Nucleotide Change	Protein Amino Acid Change
c.1726_1729del [Tan et al 2013]	p.Lys576Alafs
1727delAG [Bond et al 2003]	p.Lys576fs
c.1959_1962del [Tan et al 2013, Bond et al 2003]	p.Asn653Lysfs
c.1990C>T [Bond et al 2003]	p.Gln664Ter
c.2101C>T [Kousar et al 2010]	p.Gln701Ter
c.2389 C>T [Passemaid et al 2009, Saadi et al 2009]	p.Arg797Ter
c.2419+2T>C [Tan et al 2013]	p.Leu3035Ter
c.2571G>A [Tan et al 2013]	p.Trp857Ter
c.2761-25A-G [Nicholas et al 2009]	Intronic mutation modifying splicing: exon 10 skipped, exon 11 frameshift with 30 novel aa then stop
c.2791C>T [Tan et al 2013]	p.Arg931Ter
c.2936dup [Tan et al 2013]	p.Arg980Alafs
c.2936+5G-T [Bond et al 2003]	Removes splice donor site, additional 2 aa then stop
c.2938C>T [Muhammad et al 2009]	p.Arg980Ter
c.2967G>A [Nicholas et al 2009]	p.Trp989Ter
c.2968del [Tan et al 2013]	p.Asp990Thrfs
c.3055C>T [Muhammad et al 2009, Nicholas et al 2009]	p.Arg1019Ter
c.3082G>A [Bond et al 2003]	Removes splice donor site, additional 3 aa then stop
3188T>G [Nicholas et al 2009]	p.Leu1063Ter
c.3327T>G [Tan et al 2013]	p.Tyr1109Ter
c.3390+3_6del [Tan et al 2013]	Not defined by author
c.3477delCGCTA [Muhammad et al 2009]	p.Ala1160fs
c.3527C>G [Bond et al 2002, Bond et al 2003]	p.Ser1176Ter
c.3663delG [Bond et al 2002]	p.Arg1221fs
c.3710C>G [Nicholas et al 2009]	p.Ser1237Ter
c.3741+1G-A [Nicholas et al 2009]	Removes splice donor site, additional 9 novel aa then stop

DNA Nucleotide Change	Protein Amino Acid Change
c.3796G>T [Nicholas et al 2009, Ariani et al 2013]	p.Glu1266Ter
c.3811C>T [Bond et al 2003, Nicholas et al 2009, Passemard et al 2009, Tan et al 2013]	p.Arg1271Ter
c.3853_3854del [Tan et al 2013]	p.Asp1285Serfs
c.3945_3946del [Tan et al 2013]	p.Arg1315Serfs
c.3960_3961insA [Tan et al 2013]	p.Val1321Serfs
c.3978G>A [Kumar et al 2004, Gul et al 2006, Gul et al 2007, Muhammad et al 2009, Kousar et al 2010, Sajid Hussain et al 2013]	p.Trp1326Ter
c.3979C>T [Sajid Hussain et al 2013]	p.Arg1327Ter
c. 4074 G>A [Passemard et al 2009]	p.Trp1358Ter
c.4195_4196insA [Desir et al 2008]	p.Thr1399AsnfsTer20
c.4581delA [Bond et al 2003]	p.Gly1527fs
c.4728_4729del [Tan et al 2013]	p.Arg1576Serfs
c.4795C>T [Bond et al 2003, Tan et al 2013]	p.Arg1599Ter
c.4849C>T [Papari et al 2013]	p.R1617Ter
c.4855delTA [Nicholas et al 2009]	p.Tyr1619fs
c.5136C>A [Bond et al 2003, Gul et al 2007]	p.Tyr1712Ter
c.5149delA [Gul et al 2007]	p.Ile1717fs
c.5196T>A [Tan et al 2013]	p.Cys1732Ter
c.5584A>G [Hussain et al 2013]	p.Lys1862Glu
c.6189T>G [Shen et al 2005, Tan et al 2013]	p.Tyr2063Ter
c. 6232 T_C [Passemard et al 2009]	p.Arg2078Ter
c.6335delAT [Trimborn et al 2004, Nicholas et al 2009]	p.His2112fs
c.6651_6654delAACA [Passemard et al 2009]	p.Thr2218TyrfTer8
c.6686delGAAA [Passemard et al 2009, Kousar et al 2010]	p.Arg2229ThrfsTer10
c.6732delA [Muhammad et al 2009]	p.Tyr2245fs
c.7308dup [Tan et al 2013]	p.Val2437Cysfs
c.7489_7493delTATAT [Nicholas et al 2009]	p.Tyr2497fs

DNA Nucleotide Change	Protein Amino Acid Change
c.7612C>T [Tan et al 2013]	p.Gln2538Ter
c.7665del [Tan et al 2013]	p.Ala2556Leufs
c.7761T>G [Bond et al 2002, Nicholas et al 2009]	p.Tyr2587Ter
c.7781_7782delAG [Saadi et al 2009]	p.Gln2594fsTer6
c.7782_7783del [Tan et al 2013, Nicholas et al 2009, Passemard et al 2009]	p.Lys2595Serfs
c.7815_7816del [Ariani et al 2013]	p.E2605fs
c.7825C>T [Tan et al 2013]	p.Gln2609Ter
c.7857dup [Tan et al 2013]	p.Gln2620ThrfsTer
c.7859_7860delAG [Nicholas et al 2009]	p.Gln2620fs
c.7894C>T [Muhammad et al 2009]	p.Gln2632Ter
c.8017C>T [Tan et al 2013]	p.Gln2673Ter
c.8130_8131delAA [Nicholas et al 2009]	p.Thr2710fs
c.8133_8136del [Tan et al 2013]	p.Lys2712Leufs
c.8190_8191delAG [Passemard et al 2009]	p.Glu2731LysfsTer18
c.8191_8194delGAAA [Passemard et al 2009]	p.Arg2732LysfsTer4
c.8200_8201delAA [Hussain et al 2013]	p.Asn2734LeufsTer16
c.8273 T>A [Passemard et al 2009]	p.Leu2758Ter
c.8378delT [Nicholas et al 2009]	p.Met2793fs
c.8508_8509delGA [Bond et al 2003, Gul et al 2006, Muhammad et al 2009, Nicholas et al 2009, Hussain et al 2013]	p.Lys2837Metfs35Ter
c.8668C>T [Muhammad et al 2009, Hussain et al 2013]	p.Gln2890Ter
c.8711_8712del [Tan et al 2013]	p.Gln2904Argfs
c.8844delC [Nicholas et al 2009]	p.Ala2948fs
c.8903G>A [Tan et al 2013]	p.Trp2968Ter
c.9091C>T [Tan et al 2013]	p.Arg3031Ter
c.9118insCATT [Gul et al 2006]	p.Tyr3040fs
c.9159delA [Bond et al 2002, Kousar et al 2010]	p.Lys3053fs
c.9178C>T [Kumar 2004, Nicholas et al 2009, Tan et al 2013]	p.Gln3060Ter

DNA Nucleotide Change	Protein Amino Acid Change
c.9190C>T [Bond et al 2002, Nicholas et al 2009]	p.Arg3064Ter
c.9238A>T [Gul et al 2006, Nicholas et al 2009]	p.Leu3080Ter
c.9309_9310del [Tan et al 2013]	p.Arg3103Serfs
c.9319C>T [Muhammad et al 2009, Passemard et al 2009]	p.Arg3107Ter
c.9454C>T [Tan et al 2013]	p.Arg3152Ter
c.9492T>G [Hussain et al 2013, Muhammad et al 2009, Kousar et al 2010]	p.Tyr3164Ter
c.9507delG [Passemard et al 2009]	p.Ile3170LeufsTer9
c.9539A>C [Gul et al 2006]	p.Gln3180Pro
c.9557C>G [Bond et al 2002, Bond et al 2003, Gul et al 2006, Hussain et al 2013, Tan et al 2013]	p.Ser3186Ter
c.9595A>T [Muhammad et al 2009]	p.Lys3199Ter
c.9677_9678insG [Muhammad et al 2009]	p.Cys3226fs
c.9681delA [Nicholas et al 2009]	p.Thr3227fs
c.9686_9690delTTAAA [Bond et al 2003]	p.Ile3229SerfsTer10
c.9697C>T [Muhammad et al 2009, Tan et al 2013]	p.Arg3233Ter
c.9730C>T [Gul et al 2007, Hussain et al 2013, Tan et al 2013]	p.Arg3244Ter
c.9745delCT [Nicholas et al 2009]	p.Leu3249fs
9754delA [Bond et al 2003]	p.Arg3252fs
c.9789T>A [Nicholas et al 2009, Hussain et al 2013]	p.Tyr3263Ter
c.9910C>T [Tan et al 2013]	p.Arg3304Ter
c.9984+1G-T [Bond et al 2003]	Removes splice donor site, additional 29 novel aa then stop
c.10059C>A [Gul et al 2007]	p.Tyr3353Ter

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

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