Title: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders *GeneReview* Table 6

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

Table 6. Published Pathogenic *WDR62* Allelic Variants (Based on NM_001083961.1 and NP_001077430.1 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
c.193G>A [Nicholas et al 2010, Yu et al 2010]	p.Val65Met
c.332G>C [Sajid Hussain et al 2013]	p.Arg111Thr
c.363delT [Yu et al 2010]	p.Asp112MetfsTer5
c.535_536insA [Bhat et al 2011]	p.Met179fsTer21
c.671G>C [Bilguvar et al 2010]	p.Trp224Ser
c.900C>A [Bhat et al 2011]	p.Cys300Ter
c.1043+1G>A [Yu et al 2010]	p.Ser348ArgfsTer63
c.1194G>A [Sajid Hussain et al 2013]	p.Trp398Ter
c.1198G >A [Bacino et al 2012]	p.Gln400Lys
c.1313G>A [Nicholas et al 2010, Kousar et al 2011, Sajid Hussain et al 2013]	p.Arg438His
c.1408C>T [Bilguvar et al 2010]	p.Gln470Ter
c.1143delA [Memon et al 2013]	p.His381ProfsTer48
c.1531G>A [Nicholas et al 2010, Kousar et al 2011]	p.Asp511Asn
c.1576G>T [Bilguvar et al 2010]	p.Glu526Ter
c.1576G>A [Bilguvar et al 2010]	p.Glu526Lys
c.1942 C>T [Kousar et al 2011]	p.Gln648Ter
c.2083delA [Murdock et al 2011]	p.Ser696AlafsTer4
c.2867 + 4_c2867 + 7delGGTG [Yu et al 2010]	p.Ser956CysfsTer38
c.3232G>A [Nicholas et al 2010]	p.Ala1078Thr
c.3361delG [Sajid Hussain et al 2013]	p.Ala1121GlnfsTer6

DNA Nucleotide Change	Protein Amino Acid Change
c.3503G>A [Sajid Hussain et al 2013]	p.Trp1168
c.3839_3855delGCCAAGAGCCTGCCCTG YU [Bilguvar et al 2010]	p.Gly1280AfsTer21
c.3936dupC/c.3936_3937insC [Yu et al 2010, Kousar et al 2011]	p.Val1314ArgfsTer18/p.Val1313GlyfsTer17
c.4205delTGCC [Bilguvar et al 2010, Nicholas et al 2010]	p.Val1402GlyfsTer12
c.4241dupT [Bilguvar et al 2010]	p.Leu1414LeufsTer41

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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