Title: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders *GeneReview* Tables 7 – 8

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

Table 7. Published Pathogenic *CDK5RAP2* Allelic Variants (Based on NM_018249.4 NP_060719.4 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
c.246T>A (reported as c243T>A in Bond et al [2005]) [Hassan et al 2007, Park et al 2011]	p.Tyr82Ter
c.524_528del [Tan et al 2013]	p.Gln175ArgfsTer42
c.700G>T [Pagnamenta et al 2012]	p.Glu234Ter
c.4005-1G>A [Tan et al 2013]	Splice mutation (intron 26)
c.4186-15A>G (IVS26-15A>G) [Bond et al 2005])	p.Arg1334SerfsTer5 [Park et al 2011] (reported as Glu385fsTer4 in Bond et al [2005])
c.4441C>T [Issa et al 2013]	p.Arg1481Ter

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

Table 8. Published Pathogenic *CASC5* Allelic Variants (Based on NM_170589 and NP_733468.2 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
c.6125 G>A [Genin et al 2012]	p.Met2041Ile + abnormal splicing skipping exon 18

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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