Title: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders *GeneReview* Tables 13 – 15

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

Table 13. Published Pathogenic *CEP152* Allelic Variants

DNA Nucleotide Change	Protein Amino Acid Change
NA [Guernsey et al 2010]	p.Gln265Pro
c. [3149T>C; 3676-3678delAAC] [Hussain et al 2013]	p.[Leu1050Pro; Asn1226del]
c.261+1G>C [Kalay et al 2011]	NA (splice donor site mutation with 4 aberrant transcripts)
c.2000A>G [Kalay et al 2011]	p.Lys667Arg
c.2034T>G [Kalay et al 2011]	p.Tyr678Ter
NA [Guernsey et al 2010]	p.Arg987Ter
c.2694+1G>T [Kalay et al 2011]	r.2694G_ins3581, Ile899LeufsTer29
c.2000 A>G [Kalay et al 2011]	p.Lys667Arg
c.4210-4211delGT [Kalay et al 2011]	p.Val1404fsTer2

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

Table 14. Published Pathogenic *PHC1* Allelic Variants (Based on NM_004426.2 and NP_004417.2 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
c.2974C>T [Awad et al 2013]	p.Leu992Phe

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Table 15. Published Pathogenic *CDK6* Allelic Variants (Based on NM_004426.2 and NP_004417.2 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
c.589G>A [Hussain et al 2013]	p.Ala197Thr

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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