Title: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders *GeneReview* Tables 16 – 18

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

Table 16. Published Pathogenic *ATR* Allelic Variants (Based on NM_001184 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
Large deletion (chr3: g.(143,446,412_143,475,187)_(144,013,999_144,022,947)del.) [Mokrani-Benhelli et al 2013]	p.0
c.2022A>G (2101A>G or IVS9-57A>G) [O'Driscoll et al 2003]	Aberrant splicing of exon 9
c.3477G>T [Ogi et al 2012]	p.Met1159Ile
c.5635G>T [Mokrani-Benhelli et al 2013]	p.Asp1879Tyr
c.6897+464C>G [Ogi et al 2012]	p.Val2300Glyfs75Ter

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

Table 17. Published Pathogenic *RBBP8* Allelic Variants (Based on NM_002894.2 and NP_002885.1 Reference Sequences)

DNA Nucleotide Change (Alias [1])	Protein Amino Acid Change
c.1808_1809delTA (1868delTA) [Qvist et al 2011]	p.Ile603LysfTer7
(2347+53T>G) [Qvist et al 2011]	Missplicing and premature termination of protein

See Quick Reference for an explanation of nomenclature. *GeneReviews* follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org).
1. Variant designation that does not conform to current naming conventions

Table 18. Published Pathogenic *CEP63* Allelic Variants

DNA Nucleotide Change	Protein Amino Acid Change
c.129G>A [Sir et al 2011]	p.Trp43Ter

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

References

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