Title: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders *GeneReview* Table 5

Authors: Verloes A, Drunat S, Gressens P, Passemard S

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

Table 5. Published Pathogenic *MCPH1* Allelic Variants (Based on Reference Sequences NG_016619 and NP_078872.2)

DNA Nucleotide Change	Protein Amino Acid Change
Deletion exons 1-6 (150-200 kb deletion) [Garshasbi et al 2006, Darvish et al 2010]	p.0?
deletion exons 2 and 3 [Darvish et al 2010]	Not described
Deletion exon 3 [Darvish et al 2010]	Not described
Deletion exon 4 [Darvish et al 2010]	Not described
Large deletions [Garshasbi et al 2006, Darvish et al 2010]	p.0 ?
c.74C>G [Jackson et al 2002]	p.Ser25Ter
c.80 C>G [Trimborn et al 2005]	p.Thr27Arg
c.147C>G [Darvish et al 2010]	p.His49Gln
c.149T>G [Leung et al 2011]	p.Val50Gly
c.215C>T [Trimborn et al 2005, Ghani-Kakhki et al 2012]	p.Ser72Leu
c.233T>C [Ghani-Kakhki et al 2012]	p.Trp75Arg
c.302C>G [Tommerup et al 1993]	p.Ser101Ter
c.427_428insA [Trimborn et al 2004]	p.Thr143AsnfsTer5 [Neitzel et al 2002, Trimborn et al 2004] and p.Thr27Arg [Trimborn et al 2005]
c.436+1G>T [Darvish et al 2010]	Splice mutation not otherwise characterized
c.566_567insA [Darvish et al 2010]	p.Asn189fs
c.1179delG [Sajid Hussain et al 2013]	p.Arg393SerfsTer50

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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