Title: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders *GeneReview* Tables 10 – 12

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

Table 10. Published Pathogenic *CENPJ* Allelic Variants (Based on NM_018451.3 and NP_060921.3 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
c.17delC [Bond et al 2005]	p.Ser7LeufsTer4 (Thr6fsTer3)
c.18delC [Sajid Hussain et al 2013]	p.Ser7ProfsTer2
c.2462C>T [Darvish et al 2010]	p.Thr821Met
c.3243_3246delTCAG [Gul et al 2006a]	p.Ser1081ArgfsTer8
c.3302-1G>C [Al-Dosari et al 2010]	1073_1122del + 1073_1159del (double splicing defect)
c.3704 A>T [Bond et al 2005]	p.Glu1235Val

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

Table 11. Published Pathogenic *STIL* Allelic Variants (Based on NM_003035.2 and NP_003026.2 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
c.2826+1G>A [Kumar et al 2009]	--
c.3655delG [Kumar et al 2009]	p.Val1219Ter (Leu1218fsTer1)
c.3715C>T [Kumar et al 2009]	p.Gln1239Ter
c.2392T>G [Papari et al 2013]	p.Leu798Trp

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

Table 12. Published Pathogenic *CEP135* Allelic Variants (Based on NM_025009.4 and NP_079285 Reference Sequences)

DNA Nucleotide Change	Protein Amino Acid Change
c.970delC [Hussain et al 2012]	p.Gln324SerfsTer2

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

References

Al-Dosari MS, Shaheen R, Colak D, Alkuraya FS. Novel CENPJ mutation causes Seckel syndrome. J Med Genet. 2010 Jun;47:411-4.

Bond J, Roberts E, Springell K, Lizarraga SB, Scott S, Higgins J, Hampshire DJ, Morrison EE, Leal GF, Silva EO, Costa SM, Baralle D, Raponi M, Karbani G, Rashid Y, Jafri H, Bennett C, Corry P, Walsh CA, Woods CG. A centrosomal mechanism involving CDK5RAP2 and CENPJ controls brain size. Nat Genet. 2005 Apr;37:353-5.

Darvish H, Esmaeeli-Nieh S, Monajemi GB, Mohseni M, Ghasemi-Firouzabadi S, Abedini SS, Bahman I, Jamali P, Azimi S, Mojahedi F, Dehghan A, Shafeghati Y, Jankhah A, Falah M, Soltani Banavandi MJ, Ghani-Kakhi M, Garshasbi M, Rakhshani F, Naghavi A, Tzschach A, Neitzel H, Ropers HH, Kuss AW, Behjati F, Kahrizi K, Najmabadi H. A clinical and molecular genetic study of 112 Iranian families with primary microcephaly. J Med Genet. 2010 Dec;47(12):823-8.

Gul A, Hassan MJ, Mahmood S, Chen W, Rahmani S, Naseer MI, Dellefave L, Muhammad N, Rafiq MA, Ansar M, Chishti MS, Ali G, Siddique T, Ahmad W. Genetic studies of autosomal recessive primary microcephaly in 33 Pakistani families: Novel sequence variants in ASPM gene. Neurogenetics. 2006 May;7(2):105-10.

Hussain MS, Baig SM, Neumann S, Nürnberg G, Farooq M, Ahmad I, Alef T, Hennies HC, Technau M, Altmüller J, Frommolt P, Thiele H, Noegel AA, Nürnberg P. A truncating mutation of CEP135 causes primary microcephaly and disturbed centrosomal function. Am J Hum Genet. 2012 May 4;90:871-8.

Kumar A, Girimaji SC, Duvvari MR, Blanton SH. Mutations in STIL, encoding a pericentriolar and centrosomal protein, cause primary microcephaly. Am J Hum Genet. 2009 Feb;84(2):286-90.

Papari E, Bastami M, Farhadi A, Abedini SS, Hosseini M, Bahman I, Mohseni M, Garshasbi M, Moheb LA, Behjati F, Kahrizi K, Ropers HH, Najmabadi H. Investigation of primary microcephaly in Bushehr province of Iran: novel STIL and ASPM mutations. Clin Genet. 2013;83:488-90.

Sajid Hussain M, Marriam Bakhtiar S, Farooq M, Anjum I, Janzen E, Reza Toliat M, Eiberg H, Kjaer KW, Tommerup N, Noegel AA, Nürnberg P, Baig SM, Hansen L. Genetic heterogeneity in Pakistani microcephaly families. Clin Genet. 2013 May;83:446-51.