Title: MEGDEL Syndrome *GeneReview* Table 4
Authors: Wortmann SB, De Brouwer APM, Wevers RA, Morava E
Date: April 2014

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

Table 4. Selected *SERAC1* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference	Reference Sequences
c.442C>T	p.Arg148Ter	Wortmann et al [2012b]	NM_032861.3 NP_116250.3
c.466_467insGCGGGAAATGT	p.Ser156fsCysTer16		
c.576delT	p.Leu193SerfsTer9		
c.1167_1170delTCAG	Altered splicing		
c.1202G>A	p.Gly401Asp		
c.1211G>A	p.Gly404Glu		
c.1309_1313dupACATG	p.Trp438Ter		
c.1403+1G>C	Altered splicing		
c.1435_1437delCTT	p.Leu479del		
c.1493G>C	p.Ser498Thr		
c.1598_1599dupATAGTGTCCCTCATCAT	p.Gly536IlefsTer56		
c.1627_1628dupTC	p.Val544LeufsTer43		
c.1822_1828+10delTCAGCAGGTATTCACTCinsACCAACAGG	Altered splicing		
c.1924C>T	p.Gln642Ter		
c.698-9TG>AGTGATA	p.Leu233Ter	Sarig et al [2013]	
c.128+4A>G	Altered splicing		
c.202C>T	p.Arg68Ter	Tort et al [2013]	

Note on variant classification: Variants listed in the table have been provided by the authors. *GeneReviews* staff have not independently verified the classification of variants.

References

Sarig O, Goldsher D, Nousbeck J, Fuchs-Telem D, Cohen-Katsenelson K, Iancu TC, Manov I, Saada A, Sprecher E, Mandel H. Infantile mitochondrial hepatopathy is a cardinal feature of MEGDEL syndrome (3-methylglutaconic aciduria type IV with sensorineural deafness, encephalopathy and Leigh-like syndrome) caused by novel mutations in SERAC1. Am J Med Genet A. 2013;161:2204-15.

Tort F, García-Silva MT, Ferrer-Cortès X, Navarro-Sastre A, Garcia-Villoria J, Coll MJ, Vidal E, Jiménez-Almazán J, Dopazo J, Briones P, Elpeleg O, Ribes A. Exome sequencing identifies a new mutation in SERAC1 in a patient with 3-methylglutaconic aciduria. Mol Genet Metab. 2013;110:73-7.

Wortmann SB, Vaz FM, Gardeitchik T, Vissers LE, Renkema GH, Schuurs-Hoeijmakers JH, Kulik W, Lammens M, Christin C, Kluijtmans LA, Rodenburg RJ, Nijtmans LG, Grünewald A, Klein C, Gerhold JM, Kozicz T, van Hasselt PM, Harakalova M, Kloosterman W, Barić I, Pronicka E, Ucar SK, Naess K, Singhal KK, Krumina Z, Gilissen C, van Bokhoven H, Veltman JA, Smeitink JA, Lefeber DJ, Spelbrink JN, Wevers RA, Morava E, de Brouwer AP. Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nat Genet. 2012b;44:797-802.