Title: Microcephaly-Capillary Malformation Syndrome *GeneReview* Table 3

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

Table 3. Mutations Reported in 11 Families with MIC-CAP Syndrome

Mutation Type	DNA Nucleotide Change	Protein amino acid change	Allele frequency in 10 families with MIC-CAP syndrome
Exonic mutations	c.41G>C	p.Arg14Pro	1/20
	c.112C>T	p.Arg38Cys	3/20
	c.125A>G	p.Glu42Gly	1/20
	c.188A>G	p.Tyr63Cys	1/20
	c.299T>A	p.Phe100Tyr	2/20
	c.411del C	p.Ile138SerfsTer12	1/20
	c.532C>T	p.Arg178Ter	1/20
	c.753_754insT	p.Asp252Ter	1/20
	c.938C>T	p.Thr313Ile	1/20
	c.1134_1138delACTAA	p.Lys378AsnfsTer2	1/20
	c.1270C>T	p.Arg424Ter	3/20 [1]
Intronic mutations	c.203+5G>A	n/a	1/20
	c.279+5G>T	n/a	1/20
	c.1005+358A>G	n/a	2/20 [2]
Deletion/duplication analysis	Exonic or whole-gene deletions or duplications		Unknown, none reported

1. Mutation c.1270C>T was heterozygous in one affected individual and homozygous in another secondary to uniparental isodisomy [McDonell et al 2013].

2. Mutation c.1005+358A>G was homozygous in one affected individual (NM_006463.4) [McDonell et al 2013].