

Coding variants in *SATB2* according to NM_015265.3

Mutation type	Position	DNA variant	Predicted effect on protein	Number of families with variant
Nonsense	Exon 3	c.124G>T	p.Gly42*	1
	Exon 4	c.346G>T	p.Gly116*	1
	Exon 5	c.390T>A	p.Tyr130*	1
	Exon 6	c.505C>T	p.Gln169*	1
	Exon 7	c.688A>T	p.Lys230*	1
	Exon 8	c.715C>T	p.Arg239*	6
	Exon 8	c.748C>T	p.Gln250*	1
	Exon 8	c.847C>T	p.Arg283*	6
	Exon 8	c.868C>T	p.Gln290*	2
	Exon 8	c.988C>T	p.Gln330*	1
	Exon 8	c.997C>T	p.Gln333*	1
	Exon 8	c.1135C>T	p.Gln379*	1
	Exon 8	c.1171C>T	p.Gln391*	1
	Exon 9	c.1255C>T	p.Gln419*	1
	Exon 9	c.1285C>T	p.Arg429*	4
	Exon 9	c.1375C>T	p.Arg459*	5
	Exon 10	c.1495A>T	p.Lys499*	1
Exon 11	c.1756C>T	p.Gln586*	1	
Exon 12	c.2074G>T	p.Glu692*	1	
Missense	Exon 4	c.185T>A	p.Val62Asp	1
	Exon 4	c.257T>G	p.Leu86Arg	1

Exon 4	c.287T>G	p.Leu96Arg	1
Exon 4	c.346G>C\$	p.Gly116Arg	1
Exon 5	c.392T>A	p.Val131Glu	1
Exon 8	c.760C>T	p.His254Tyr	1
Exon 8	c.983C>A	p.Ala328Asp	1
Exon 8	c.1102G>T	p.Val368Phe	1
Exon 8	c.1136A>C	p.Gln379Pro	1
Exon 8	c.1142T>G	p.Val381Gly	1
Exon 8	c.1157C>T	p.Ala386Val	1
Exon 8	c.1165C>T	p.Arg389Cys	10
Exon 8	c.1166G>T	p.Arg389Leu	2
Exon 8	c.1169C>T	p.Thr390Ile	2
Exon 9	c.1174G>C	p.Gly392Arg	1
Exon 9	c.1175G>A	p.Gly392Glu	1
Exon 9	c.1181T>C	p.Leu394Ser	1
Exon 9	c.1186G>C	p.Glu396Gln	1
Exon 9	c.1196G>A	p.Arg399His	4
Exon 9	c.1196G>T	p.Arg399Leu	2
Exon 9	c.1196G>C	p.Arg399Pro	1
Exon 9	c.1204G>A	p.Glu402Lys	1
Exon 9	c.1253T>G	p.Met418Arg	1
Exon 9	c.1286G>A	p.Arg429Gln	4
Exon 10	c.1541A>G	p.Gln514Arg	1
Exon 11	c.1543G>A	p.Gly515Ser	1
Exon 11	c.1554T>G	p.Cys518Trp	1
Exon 11	c.1564C>T	p.Arg522Cys	1
Exon 11	c.1696G>A	p.Glu566Lys	1
Exon 12	c.1903G>T	p.Asp635Tyr	1

	Exon 12	c.1964C>T	p.Pro655Leu	1
Frameshift	Exon 3	c.9_16del	p.Arg4Glu fs*68	1
	Exon 3	c.19dupA	p.Ser7Lys fs*68	1
	Exon 4	c.245_263del	p.Ala82Gly fs*30	1
	Exon 4	c.334delG	p.Ala112Pro fs*6	1
	Exon 5	c.400delG	p.Ala134His fs*17	1
	Exon 6	c.482delA	p.Lys161Ser fs*19	1
	Exon 6	c.583dupT	p.Cys195Leu fs*14	1
	Exon 6	c.594_595delCC	p.Gln199Glu fs*9	1
	Exon 8	c.808_809delCA	p.Gln270Val fs*33	1
	Exon 8	c.816delT	p.His273Thr fs*21	1
	Exon 8	c.832delC	p.His278Thr fs*16	1
	Exon 8	c.1131_1132delGT	p.Ser378Pro fs*18	1
	Exon 8	c.1132_1133insAC	p.Ser378Tyr fs*36	1
	Exon 9	c.1196delG	p.Arg399Leu fs*14	1
	Exon 9	c.1203dupA	p.Glu402Arg fs*35	1
	Exon 9	c.1311_1314dupGGAG	p.Arg439Gly fs*38	1
	Exon 10	c.1478_1479delAG	p.Gln493Arg fs*19	1
	Exon 10	c.1498delG Mosaic	p.Val500Cys fs*46	1
	Exon 10	c.1511_1512insCAAGCCT	p.Phe505Lys fs*10	1
	Exon 10	c.1515delT	p.Phe505Leu fs*41	1
	Exon 11	c.1574delA	p.Glu525Gly fs*21	1
	Exon 11	c.1592dupA	p.Asn531Lys fs*21	1
	Exon 11	c.1627delC	p.Arg543Ala fs*3	1
	Exon 11	c.1639_1642delinsTTT	p.Leu547Phe fs*77	1
	Exon 11	c.1645delC	p.Gln549Ser fs*75	1
	Exon 11	c.1657delG	p.Asp553Met fs*71	1
	Exon 11	c.1728delT	p.Glu577Ser fs*47	1
	Exon 12	c.1945dupT	p.Ser649Phe fs*40	1
	Exon 12	c.1972_1973delAC	p.Thr658His fs*30	1
	Exon 12	c.2005dupC	p.His669Pro fs*20	1

	Exon 12	c.2018dupA	p.His673Glnfs*16	1
	Exon 12	c.2028delG	p.Glu678Serfs*18	1
In frame insertion	Exon 8	c.929_930insTTGTAAGGCAAC	p.Q310delinsHisCysLysAlaThr	1
Splice	Exon 4	c.346+2T>G	p.?	1
	Exon 5	c.473+1delG	p.?	1
	Exon 7	c.598-2A>G	p.?	1
	Exon 8	c.1173+2T>C	p.?	1
	Exon 9	c.1174-2A>G	p.?	1
	Exon 12	c.1741-1G>A	p.?	1