



Intragenic mutations of FOXP1-related encephalopathy reported in the literature.

Genetic mutations (amino acid change)	FOXP1 domain	Coding effect	References
c.133_469del337insACCCACCGCCCC ACCCACCGCCCC (p.Pro45Thrfs*39)	N-terminal	Frameshift	[1,2]
c.136C>T (p.Gln46Ter)	N-terminal	Nonsense	[3]
c.201_224del24 (p.Pro66_Gln73del)	N-terminal	Frameshift	[4]
c.214C>T (p.Gln72Ter)	N-terminal	Nonsense	[5]
p.Gln73dup	N-terminal	In-frame	[6]
c.222_223dupGC (p.Pro75Argfs*118)	N-terminal	Frameshift	[1]
c.256dupC (p.Gln86Profs*34)	N-terminal	Frameshift	[7,8]
p.Gln86Profs*35	N-terminal	Frameshift	[6]
c.256delC (p.Gln86Argfs*106)	N-terminal	Frameshift	[8,9]
c.256C>T (p.Gln86Ter)	N-terminal	Nonsense	[1,5,10]
c.263_278del16 (p.Arg88Profs*99)	N-terminal	Frameshift	[10,11]
c.292_295delGGCC (p.Gly98Profs*93)	N-terminal	Frameshift	[4]
c.298del (p.Gln100Serfs*92)	N-terminal	Frameshift	[12]
c.385delG (p. Glu129Serfs*63)	N-terminal	Frameshift	[5]
c.392delG (p.Gly131Alafs*60)	N-terminal	Frameshift	[13]
c.406G>T (p.Glu136Ter)	N-terminal	Nonsense	[5]
p.Glu136Glyfs*39	N-terminal	Frameshift	[6]
c.430G>T (p.Glu144Ter)	N-terminal	Nonsense	[1]
c.460G>T (p.Glu154Ter)	N-terminal	Nonsense	[14]
c.460delG (p.Glu154Argfs*38)	N-terminal	Frameshift	[5]
c.460dupG (p.Glu154Glyfs*301)	N-terminal	Frameshift	[5,10,11,15]
c.461insG (p.Glu155Glyfs*300)	N-terminal	Frameshift	[4,6]
p.Glu155Ter	N-terminal	Nonsense	[6]
p.Lys162Serfs*51	N-terminal	Frameshift	[6]
c.505_506delGGinsT(p.Gly168Serfs*23)	N-terminal	Frameshift	[10]
c.506delG (p.Gly169Alafs*23)	N-terminal	Frameshift	[1]
c.506dup (p.Lys170Glnfs*285)	N-terminal	Frameshift	[16]

c.515_577del63 (p.Gly172_Met192del)	N-terminal	In-frame	[1,15,17]
c.517G>T (p.Glu173Ter)	N-terminal	Nonsense	[5]
c.537C>G (p.Tyr179Ter)	N-terminal	Nonsense	[6]
c.541A>G (p.Lys181Glu)	forkhead cs	Missense	[4]
c.543G>C (p.Lys181Asn)	forkhead cs	Missense	[5]
p.Pro182Leu	forkhead cs	Missense	[6]
c.545C>A (p.Pro182Gln)	forkhead cs	Missense	[5]
c.552dupC (p.Ser185Glnfs*270)	forkhead cs	Frameshift	[6,18]
c.553A>T (p.Ser185Cys)	forkhead cs	Missense	[5]
c.559A>G (p.Asn187Asp)	forkhead cs	Missense	[6,19]
c.561C>A (p.Asn187Lys)	forkhead cs	Missense	[5,6]
c.565C>T (p.Leu189Phe)	forkhead cs	Missense	[5]
c.565C>G (p.Leu189Val)	forkhead cs	Missense	[20]
c.569T>A (p. Ile190Asn)	forkhead cs	Missense	[21]
c.572T>G (p.Met191Arg)	forkhead cs	Missense	[11,15]
c.577G>A (p.Ala193Thr)	forkhead cs	Missense	[1,22]
p.Ile194Serfs*19	forkhead cs	Frameshift	[6]
c.581T>G (p.Ile194Ser)	forkhead cs	Missense	[5]
p.Arg195Pro	forkhead	Missense	[6]
c.586C>T (p.Gln196Ter)	forkhead	Nonsense	[1,6]
c.590G>T (p.Ser197Ile)	forkhead	Missense	[5]
c.592_594delCCC (p.Prp198del)	forkhead	In-frame	[5]
c.609_616del (p.L204Hfs*248)	forkhead	Frameshift	[14]
c.610C>T (p.Leu204Phe)	forkhead	Missense	[6,23]
c.619A>T (p.Ile207Phe)	Forkhead	Missense	[4]
c.624C>G (p.Tyr208Ter)	forkhead	Nonsense	[4,5,18]
p.Tyr208_Ile211del	forkhead	In-frame	[6]
c.643T>C (p.Phe215Leu)	forkhead	Nonsense	[18]
c.651C>G (p.Tyr217Ter)	forkhead	Nonsense	[1]
c.653A>C (p.Tyr218Ser)	forkhead	Missense	[4]
p.Gly224Ser	forkhead	Missense	[6]

c.681C > G (p.Asn227Lys)	forkhead	Missense	[18]
c.688C>G (p.Arg230Gly)	forkhead	Missense	[4]
c.689G>A (p.Arg230His)	forkhead	Missense	[6,24]
c.693C>A (p.Hys231Gln)	forkhead	Missense	[4]
c.694A>T (p.Asn232Tyr)	forkhead	Missense	[25]
c.695A>G (p.Asn232Ser)	forkhead	Missense	[11]
c.700T>C (p.Ser234Pro)	forkhead	Missense	[10]
p.Val242Cysfs*84	forkhead	Frameshift	[6]
c.730C>T (p.Arg244Cys)	forkhead	Missense	[26]
c.732_741del10 (p.His245Thrfs*78)	forkhead	Frameshift	[5]
c.735delC (p.Tyr246Thrfs*80)	forkhead	Frameshift	[1]
c.755G>T (p.Gly252Val)	forkhead	Missense	[1,6]
c.755G>A (p.Gly252Asp)	forkhead	Missense	[5]
c.757A>G (p.Asn253Asp)	forkhead	Missense	[10]
c.762C>G (p.Tyr254Ter)	forkhead	Nonsense	[1]
p.Tyr254Thrfs*72	forkhead	Frameshift	[6]
c.763T>C (p.Trp255Arg)	forkhead	Missense	[6,8]
c.765G>A (p.Trp255Ter)	forkhead	Missense	[27]
p.Leu257Pro	forkhead	Missense	[6]
c.775C>T (p.Pro259Ser)	forkhead	Missense	[25]
c.788_792delACGTG(p.Asp263Valfs*190)	forkhead	Frameshift	[28]
p.Ile266Tyrfs*189	forkhead	Frameshift	[6]
c.858dupC (p.Lys287Glnfs*168)	C-terminal	Frameshift	[29]
c.893C>A (p.Thr298Asn)	C-terminal	Missense	[25]
c.921C>G (p.Try307Ter)	C-terminal	Nonsense	[5]
c.924G>A (p.Trp308Ter)	GBD	Nonsense	[2]
c.946delC (p.Leu316Cysfs*10)	GBD	Frameshift	[11]
c.953_954insC (p.Arg320Profs*135)	C-terminal	Frameshift	[4]
c.969delC (p.Ser323Argfs*3)	C-terminal	Frameshift	[27]
c.974dupT (p.Leu325Phefs*130)	C-terminal	Frameshift	[5]
c.974_975insA (p.Ser326Glufs*129)	C-terminal	Frameshift	[6,30]

c.981C>A (p.Tyr327Ter)	C-terminal	Nonsense	[11]
c.1082dup (p.Leu362Profs*93)	C-terminal	Frameshift	[5]
c.1117_1118insCGTA(p.Ala375Valfs*81)	C-terminal	Frameshift	[4]
c.1141delG (p.Ala381Profs*4)	C-terminal	Frameshift	[5]
c.1186C>A, (p.Cys396Ter)	JBD	Nonsense	[11]
c.1200C>A (p.Tyr400Ter)	JBD	Nonsense	[3]
c.1200C>G (p.Tyr400Ter)	JBD	Nonsense	[2]
p.Asn408Ile	C-terminal	Missense	[6]
c.1248C>G (p.Tyr416Ter)	C-terminal	Missense	[14]
c.1470A>C (p.Ter490Tyext*21)	C-terminal	Stop loss	[4]

¹ Table S1 shows the published *FOXG1* variants.

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