RETT SYNDROME RESEARCH TRUST

MECP2 iPSC Collection at Coriell Institute

Protein variant	DNA variant	Clonally isolated isogenic control	Sex	Source
Point Mutations				
p.R106W	c.C316T	Yes	F	lymphocyte
p.R133C	c.C397T	Yes	F	lymphocyte
p.R133H	c.G398A	Yes	F	lymphocyte
p.T158M	c.C473T	Yes	F	lymphocyte
p.R168X	c.C502T	Yes	F	lymphocyte
p.R255X	c.C763T	Yes	F	lymphocyte
p.R270X	c.C808T	Yes	F	lymphocyte
p.R294X	c.C880T	Yes	F	fibroblast
p.R306C	c.C916T	Yes	F	lymphocyte
Splicing Mutation				
---	c.62+1delGT (intron 1)	Yes	F	Coriell fibro. GM25456

MECP2 Fibroblast Collection at Sampled

Protein variant	DNA variant	Sex	Source
Point Mutations			
p.P72L	c.C215T	F	skin biopsy
p.R106W	c.C316T	F	skin biopsy
p.R133C	c.C397T	F	skin biopsy
p.T158M	c.C473T	F	skin biopsy
p.R168X	c.C502T	F	skin biopsy
p.R255X	c.C763T	F	skin biopsy
p.R270X	c.C808T	F	skin biopsy
p.R294X	c.C880T	F	skin biopsy
p.R306C	c.C916T	F	skin biopsy
Single Base Deletion Mutations			
p.P72Rfs*53	c.215del	M	skin biopsy
p.G252Afs	c.753delC	F	skin biopsy
p.G269Afs*20	c.806delG	M/F	skin biopsy
Duplicated Region Mutation			
p.R20EfsX29	c.44_57dup14	F	skin biopsy
Splicing Mutations			
---	c.62+2T>G: IVSI+2T>G in intron 1	M	skin biopsy
---	IVS3-2A>G	F	skin biopsy
Large Deletion Mutations			
---	exon 4 partial deletion: codon 377 into 3' UTR	F	skin biopsy
---	97kb interstitial deletion of XQ28-Q28	F	skin biopsy