rett syndrome research trust

RSRT MECP2 iPSC Collection at Coriell

Mutation (protein)	Mutation (DNA)	Isogenic control available	Sex	Source
Point Mutations				
p.R106W	c.C316T	Yes	F	lymphocytes
p.R133C	c.C397T	Yes	F	lymphocytes
p.T158M	c.C473T	Yes	F	lymphocytes
p.R255X	c.C763T	Yes	F	lymphocytes
p.R306C	c.C916T	Yes	F	lymphocytes

RSRT MECP2 Fibroblast Collection at RUCDRs

Mutation (protein)	Mutation (DNA)	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 Deletion Mutations			
p.P72Rfs*53	c.215del	M	skin biopsy
p.G252Afs	c.753delC	F	skin biopsy
p.G269Afs*20	c.806delG	M and F	skin biopsy
Duplicated Region Mutation			
p.R20EfsX29	c.44_57dup14	F	skin biopsy
Inversion 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