

Feb_2016	Noonan	SOS1	c.1322G>A	p.Cys441Tyr	Missense	YES	Female	0.6	0.555111083
Feb_2016	Noonan	SOS1	c.806T>G	p.Met269Arg	Missense	YES	Female	0.4	0.844087032
Feb_2016	Noonan	SOS1	c.797C>A	p.Thr266Lys	Missense	YES	Male	1	2.133506512
Feb_2016	Noonan	SOS1	c.1297G>A	p.Glu433Lys	Missense	YES	Male	1	1.481217449
Feb_2016	Noonan	SOS1	c.1300G>A	p.Gly434Arg	Missense	YES	Male	5	8.558246566
May_2015	Rett	MECP2	NA	p.Arg106Trp	Missense	YES	Female	1	1.835127123
May_2015	Rett	MECP2	NA	p.Arg168*	Nonsense	YES	Female	25	29.34649481
May_2015	Rett	MECP2	NA	p.Pro302Arg	Missense	YES	Female	34	35.17904908
May_2015	Rett	MECP2	NA	NA	Exonic deletion	YES	Female	2	2.581071992
May_2015	Rett	MECP2	NA	p.Thr158Met	Missense	YES	Female	1	2.210005617
May_2015	Rett	MECP2	Deletion in exon 4	NA	Exonic deletion	YES	Female	3	5.225511336
May_2015	Rett	MECP2	NA	p.Thr158Met	Missense	YES	Female	1	2.510753024
May_2015	Rett	MECP2	NA	p.Pro225Arg	Missense	YES	Female	4	6.160921221
May_2015	Rett	MECP2	c.1157_1197del41	p.Glu374fs	Frameshift	YES	Female	6	6.2636907
May_2015	Rett	MECP2	NA	p.Arg255*	Nonsense	YES	Female	1.5	1.084382282
May_2015	Rett	MECP2	Deletion in exons 3 and 4	NA	Exonic deletion	YES	Female	6	6.883663479
May_2015	Rett	MECP2	NA	p.Arg106Trp	Missense	YES	Female	29	38.83647398
May_2015	Rett	MECP2	NA	p.Thr158Met	Missense	YES	Female	3	4.77442952
May_2015	Rett	MECP2	NA	p.Arg255*	Nonsense	YES	Female	11	11.74653291
May_2015	Rett	MECP2	Partial deletion of exon 4	NA	Exonic deletion	YES	Female	4	3.072948979
Jun_2015	Saethre_Chotzen	TWIST1	c.385_405dup21	NA	In-frame insertion	YES	Female	0.003	-0.35722332
Nov_2015	Saethre_Chotzen	TWIST1	c.149delC	p.Ala50fs	Frameshift	YES	Male	0.02	0.16785508
Nov_2015	Saethre_Chotzen	TWIST1	c.149delC	p.Ala50fs	Frameshift	YES	Female	0.1	13.96937513
Nov_2015	Saethre_Chotzen	TWIST1	c.376G>T	p.Glu126*	Nonsense	YES	Male	38	41.56611411
Nov_2015	Saethre_Chotzen	TWIST1	c.406_407ins21	NA	In-frame insertion	YES	Male	30	29.61790422
Nov_2015	Saethre_Chotzen	TWIST1	c.156delC	p.Pro52fs	Frameshift	YES	Female	33.5	27.76671901
Nov_2015	Saethre_Chotzen	TWIST1	c.418_419ins21	NA	In-frame insertion	YES	Male	17.7	15.97052177
Nov_2015	Saethre_Chotzen	TWIST1	c.211C>T	p.Gln71*	Nonsense	YES	Female	20.7	18.347741
Nov_2015	Saethre_Chotzen	TWIST1	c.325C>T	p.Gln109*	Nonsense	YES_predicted	Male	0.7	0.45749609
Nov_2015	Saethre_Chotzen	TWIST1	c.396_416dup21	NA	In-frame insertion	YES	Male	0.1	0.386967314
Nov_2015	Saethre_Chotzen	TWIST1	c.193G>T	p.Glu65*	Nonsense	YES	Female	0.01	0.049927484
Nov_2015	Saethre_Chotzen	TWIST1	c.472T>C	p.Phe158Leu	Missense	YES	Female	23.3	0.174364646
Nov_2015	Saethre_Chotzen	TWIST1	NA	NA	Full gene deletion	YES	Female	0.35	0.404844597
Nov_2015	Saethre_Chotzen	TWIST1	NA	NA	Full gene deletion	YES	Female	0.003	7.069271322
Nov_2015	Saethre_Chotzen	TWIST1	c.160G>T	p.Gly54*	Nonsense	YES	Female	0.7	0.830512167
Nov_2015	Saethre_Chotzen	TWIST1	c.397_417dup21	NA	In-frame insertion	YES_predicted	Female	20.5	25.83177177
Nov_2015	Saethre_Chotzen	TWIST1	c.120_145del26	NA	Frameshift	YES	Male	0.6	0.491449014
Nov_2015	Saethre_Chotzen	TWIST1	c.149delC	p.Ala50fs	Frameshift	YES	Female	23.5	18.94806941
Nov_2015	Saethre_Chotzen	TWIST1	c.394_414del21	NA	In-frame deletion	YES	Female	12.3	10.10722932
Nov_2015	Saethre_Chotzen	TWIST1	c.352C>G	p.Arg118Gly	Missense	YES_predicted	Female	21.5	23.41800184