May_2015	ATR-X	ATRX	c.758T>C	p.Leu253Ser	Missense	YES	Male	18.5	6.108170831
May_2015	ATR-X	ATRX	c.4817G>A	p.Ser1606Asn	Missense	YES	Male	21	24.74309568
May_2015	ATR-X	ATRX	c.5786A>G	p.Lys1929Arg	Missense	YES	Male	0.7	-0.14552632
May_2015	ATR-X	ATRX	c.730A>C	p.Ile244Leu	Missense	YES	Male	14	11.30064691
May_2015	ATR-X	ATRX	c.7156C>T	p.Arg2386*	Nonsense	YES	Male	4.6	6.236506951
May_2015	ATR-X	ATRX	c.536A>G	p.Asn179Ser	Missense	YES	Male	4.6	33.54375298
May_2015	ATR-X	ATRX	Exon 207 deletion	NA	Exonic deletion	YES	Male	4.4	4.821921423
May_2015	ATR-X	ATRX	c.7366_7367insA	p.Met2456Asnfs*42	Frameshift	YES	Male	27	39.19917395
May_2015	ATR-X	ATRX	c.109C>T	p.Arg37*	Nonsense	YES	Male	14.5	5.274937882
May_2015	ATR-X	ATRX	c.736C>T	p.Arg246Cys	Missense	YES	Male	2.5	1.113449871
May_2015	ATR-X	ATRX	c.109C>T	p.Arg37*	Nonsense	YES	Male	17.5	22.71435784
May_2015	ATR-X	ATRX	c.109C>T	p.Arg37*	Nonsense	YES	Male	14	11.21597332
Nov_2015	Claes_Jensen	KDM5C	c.1510G>A	p.Val504Met	Missense	YES	Male	30	42.69659356
Nov_2015	Claes_Jensen	KDM5C	c.1439C>T	p.Pro480Leu	Missense	YES_predicted	Male	6	8.103173952
Nov_2015	Claes_Jensen	KDM5C	c.4439_4440delAG	p.Arg1481Glyfs*	Frameshift	YES	Male	26	28.25654272
Nov_2015	Claes_Jensen	KDM5C	Intron 11:+5G>A	NA	Splice site mutation	YES	Male	42	54.3236723
Nov_2015	Claes_Jensen	KDM5C	c.1510G>A	p.Val504Met	Missense	YES	Male	8	10.07007313
Nov_2015	Claes_Jensen	KDM5C	c.1439C>T	p.Pro480Leu	Missense	YES	Male	2	3.619189097
Nov_2015	Claes_Jensen	KDM5C	c.229G>A	p.Ala77Thr	Missense	YES	Male	37	48.42002598
Nov_2015	Claes_Jensen	KDM5C	c.4439_4440delAG	p.Arg1481Glyfs*	Frameshift	YES	Male	28	31.61445991
Nov_2015	Claes_Jensen	KDM5C	c.229G>A	p.Ala77Thr	Missense	YES	Male	13	16.50827759
Nov_2015	Claes_Jensen	KDM5C	c.1510G>A	p.Val504Met	Missense	YES	Male	26	38.69008936
May_2015	Coffin_Lowry	RPS6KA3	c.1520insA	p.Arg507fs	Frameshift	YES	Female	6	4.093225848
May_2015	Coffin_Lowry	RPS6KA3	c.2065C>T	p.Gln689*	Nonsense	YES	Male	11.5	10.63296406
May_2015	Coffin_Lowry	RPS6KA3	c.2186G>A	p.Arg729Gln	Missense	YES_predicted	Male	4	4.62981308
May_2015	Coffin_Lowry	RPS6KA3	c.631_772del142 and c.774+5G>A	NA	Frameshift and intronic mutation	YES_predicted	Male	7	5.068637974
May_2015	Coffin_Lowry	RPS6KA3	c.340C>T	p.Arg114Trp	Missense	YES_predicted	Male	1.3	8.170755226
May_2015	Coffin_Lowry	RPS6KA3	c.727C>T	p.Arg243*	Nonsense	YES	Male	13	14.17141748
May_2015	Coffin_Lowry	RPS6KA3	Intron 14:+1G>A	NA	Splice site mutation	YES	Male	22.8	25.56720654
May_2015	Coffin_Lowry	RPS6KA3	NA	NA	Exonic and intronic deletion	YES	Male	12	10.17620766
May_2015	Coffin_Lowry	RPS6KA3	c.386_387insCTTT	p.Phe130Phefs*141	Frameshift	YES	Male	2	1.808104516
May_2015	Coffin_Lowry	RPS6KA3	c.1155delT	p.Phe385fs*40	Frameshift	YES	Male	8	7.406603271
Mar_2014	Floating_Harbour	SRCAP	c.7303C>T	p.Arg2435*	Nonsense	YES	Female	8	11.29885487
Mar_2014	Floating_Harbour	SRCAP	c.7330C>T	p.Arg2444*	Nonsense	YES	Female	15	16.23135534
Mar_2014	Floating_Harbour	SRCAP	c.7282dupC	p.Arg2428Profs*15	Frameshift	YES	Female	6	5.620915174
Mar_2014	Floating_Harbour	SRCAP	c.7330C>T	p.Arg2444*	Nonsense	YES	Female	10	42.55562244
Mar_2014	Floating_Harbour	SRCAP	c.8117C>G	p.Ser2706*	Nonsense	YES	Male	4	2.815335426
Mar_2014	Floating_Harbour	SRCAP	c.7330C>T	p.Arg2444*	Nonsense	YES	Female	5	4.112348915
Mar_2014	Floating_Harbour	SRCAP	c.7330C>T	p.Arg2444*	Nonsense	YES	Female	42	43.43022309
Mar_2014	Floating_Harbour	SRCAP	c.7330C>T	p.Arg2444*	Nonsense	YES	Male	12	12.37257473