Batch name	Developmental disorder	Gene	Mutation (DNA)	Mutation (protein)	Mutation effect	Pathogenic	Sex	Age (years)	DNAmAge
Europe	ASD	NA	NA	NA	NA	NA	Male	23.25	29.94120469
Europe	ASD	NA	NA	NA	NA	NA	Male	25.75	23.66579727
Europe	ASD	NA	NA	NA	NA	NA	Male	23.75	22.89490773
Europe	ASD	NA	NA	NA	NA	NA	Male	26.58	31.33521081
Europe	ASD	NA	NA	NA	NA	NA	Male	11.83	13.55540994
Europe	ASD	NA	NA	NA	NA	NA	Male	12.33	12.62567804
Europe	ASD	NA	NA	NA	NA	NA	Male	11.67	11.91444556
Europe	ASD	NA	NA	NA	NA	NA	Male	12.67	15.1433583
Europe	ASD	NA	NA	NA	NA	NA	Male	15.92	20.69231419
Europe	ASD	NA	NA	NA	NA	NA	Male	16.92	18.37736076
Europe	ASD	NA	NA	NA	NA	NA	Male	15.92	14.74270021
Europe	ASD	NA	NA	NA	NA	NA	Male	19	28.69942806
Europe	ASD	NA	NA	NA	NA	NA	Male	16.75	20.84761017
Europe	ASD	NA	NA	NA	NA	NA	Male	20.16	17.69509361
Europe	ASD	NA	NA	NA	NA	NA	Male	12.92	18.28693655
Europe	ASD	NA	NA	NA	NA	NA	Male	13.25	12.24924728
Europe	ASD	NA	NA	NA	NA	NA	Male	13	15.27709141
Europe	ASD	NA	NA	NA	NA	NA	Male	13.25	15.93247357
Europe	ASD	NA	NA	NA	NA	NA	Male	13.16	17.97126245
Europe	ASD	NA	NA	NA	NA	NA	Male	13.67	18.5985271
Europe	ASD	NA	NA	NA	NA	NA	Male	7.67	9.834525429
Europe	ASD	NA	NA	NA	NA	NA	Male	7.92	8.819610809
Europe	ASD	NA	NA	NA	NA	NA	Male	7.73	10.53639331
Europe	ASD	NA	NA	NA	NA	NA	Male	8	8.782413174
Europe	ASD	NA	NA	NA	NA	NA	Male	7.83	8.331080792
Europe	ASD	NA	NA	NA	NA	NA	Male	8	8.412508081
Europe	ASD	NA	NA	NA	NA	NA	Male	10.83	12.94110542
Europe	ASD	NA	NA	NA	NA	NA	Male	11.5	16.52427744
Europe	ASD	NA	NA	NA	NA	NA	Male	10.83	9.546814402
Europe	ASD	NA	NA	NA	NA	NA	Male	11.5	10.75219435
Europe	ASD	NA	NA	NA	NA	NA	Male	10.83	11.7226536
Europe	ASD	NA	NA	NA	NA	NA	Male	6	8.750320884
Europe	ASD	NA	NA	NA	NA	NA	Male	5.75	8.069349936
Europe	ASD	NA	NA	NA	NA	NA	Male	6	8.205893972
Europe	ASD	NA	NA	NA	NA	NA	Male	5.83	8.765912407
Europe	ASD	NA	NA	NA	NA	NA	Male	6.33	6.903468104
Europe	ASD	NA	NA	NA	NA	NA	Male	5.25	5.648518225
Europe	ASD	NA	NA	NA	NA	NA	Male	5.67	5.896253109
Europe	ASD	NA	NA	NA	NA	NA	Male	5.42	6.160793858
Europe	ASD	NA	NA	NA	NA	NA	Male	5.75	8.719005258
Europe	ASD	NA	NA	NA	NA	NA	Male	5.42	6.49657694
Europe	ASD	NA	NA	NA	NA	NA	Male	3.92	4.884904225
Europe	ASD	NA	NA	NA	NA	NA	Male	4.08	4.766905985
Europe	ASD	NA	NA	NA	NA	NA	Male	4	5.462162993

Europe	ASD	NA	NA	NA	NA	NA	Male	4.08	4.557194499
Europe	ASD	NA	NA	NA	NA	NA	Male	4	4.383741212
Europe	ASD	NA	NA	NA	NA	NA	Male	4.25	5.321367013
Europe	ASD	NA	NA	NA	NA	NA	Male	3.25	2.797437125
Europe	ASD	NA	NA	NA	NA	NA	Male	3.42	3.906912403
Europe	ASD	NA	NA	NA	NA	NA	Male	3.33	4.703272329
Europe	ASD	NA	NA	NA	NA	NA	Male	3.5	3.223456196
Europe	ASD	NA	NA	NA	NA	NA	Male	3.42	4.024449964
Europe	ASD	NA	NA	NA	NA	NA	Male	3.58	4.662665584
Europe	ASD	NA	NA	NA	NA	NA	Male	5.16	7.931806871
Europe	ASD	NA	NA	NA	NA	NA	Male	5.16	6.144088681
Europe	ASD	NA	NA	NA	NA	NA	Male	5.16	5.423886319
Europe	ASD	NA	NA	NA	NA	NA	Male	5.25	6.873520458
Europe	ASD	NA	NA	NA	NA	NA	Male	5.16	6.828746343
Europe	ASD	NA	NA	NA	NA	NA	Male	5.25	6.287392617
Europe	ASD	NA	NA	NA	NA	NA	Male	6.5	7.549817595
Europe	ASD	NA	NA	NA	NA	NA	Male	6.83	5.310188113
Europe	ASD	NA	NA	NA	NA	NA	Male	6.67	8.807848811
Europe	ASD	NA	NA	NA	NA	NA	Male	7.16	7.314048584
Europe	ASD	NA	NA	NA	NA	NA	Male	6.83	7.143809294
Europe	ASD	NA	NA	NA	NA	NA	Male	7.25	4.888587648
Europe	ASD	NA	NA	NA	NA	NA	Male	10.08	11.01168613
Europe	ASD	NA	NA	NA	NA	NA	Male	10.08	9.091817984
Europe	ASD	NA	NA	NA	NA	NA	Male	10.08	12.00962928
Europe	ASD	NA	NA	NA	NA	NA	Male	10.5	11.89814401
Europe	ASD	NA	NA	NA	NA	NA	Male	10.08	10.85200361
Europe	ASD	NA	NA	NA	NA	NA	Male	10.58	15.97655481
Europe	ASD	NA	NA	NA	NA	NA	Male	14.67	19.40830372
Europe	ASD	NA	NA	NA	NA	NA	Male	15.25	17.28948864
Europe	ASD	NA	NA	NA	NA	NA	Male	14.83	18.99313794
Europe	ASD	NA	NA	NA	NA	NA	Male	15.25	17.40182035
Europe	ASD	NA	NA	NA	NA	NA	Male	15.08	20.74719227
Europe	ASD	NA	NA	NA	NA	NA	Male	15.83	17.66494621
Europe	ASD	NA	NA	NA	NA	NA	Male	1.83	2.332369997
Europe	ASD	NA	NA	NA	NA	NA	Male	2.33	2.079645877
Europe	ASD	NA	NA	NA	NA	NA	Male	2.08	3.093728905
Europe	ASD	NA	NA	NA	NA	NA	Male	2.5	3.327332717
Europe	ASD	NA	NA	NA	NA	NA	Male	2.08	3.081702301
Europe	ASD	NA	NA	NA	NA	NA	Male	2.5	3.640188937
Europe	ASD	NA	NA	NA	NA	NA	Male	27.67	5.315328746
Europe	ASD	NA	NA	NA	NA	NA	Male	32.92	35.79080593
Europe	ASD	NA	NA	NA	NA	NA	Male	31.83	35.12415194
Europe	ASD	NA	NA	NA	NA	NA	Male	35.16	34.8152863
Europe	ASD	NA	NA	NA	NA	NA	Male	32.33	33.47894995
Europe	ASD	NA	NA	NA	NA	NA	Male	11.58	14.81256772
Europe	ASD	NA	NA	NA	NA	NA	Male	4.5	3.982793413

Europe	ASD	NA	NA	NA	NA	NA	Male	4.75	6.632731853
Europe	ASD	NA	NA	NA	NA	NA	Male	4.5	5.453577973
Europe	ASD	NA	NA	NA	NA	NA	Male	5	6.0536493
Europe	ASD	NA	NA	NA	NA	NA	Male	4.67	4.665684936
Europe	ASD	NA	NA	NA	NA	NA	Male	5	5.538833496
Europe	ASD	NA	NA	NA	NA	NA	Male	4.33	6.826640979
Europe	ASD	NA	NA	NA	NA	NA	Male	4.42	5.074848057
Europe	ASD	NA	NA	NA	NA	NA	Male	4.33	4.069969605
Europe	ASD	NA	NA	NA	NA	NA	Male	4.5	2.914915908
Europe	ASD	NA	NA	NA	NA	NA	Male	4.33	4.177855824
Europe	ASD	NA	NA	NA	NA	NA	Male	4.5	5.359046992
Europe	ASD	NA	NA	NA	NA	NA	Male	7.33	4.981096393
Europe	ASD	NA	NA	NA	NA	NA	Male	7.5	7.521560211
Europe	ASD	NA	NA	NA	NA	NA	Male	7.33	5.632014057
Europe	ASD	NA	NA	NA	NA	NA	Male	7.58	5.381195679
Europe	ASD	NA	NA	NA	NA	NA	Male	7.42	7.07596058
Europe	ASD	NA	NA	NA	NA	NA	Male	7.58	6.118788705
Europe	ASD	NA	NA	NA	NA	NA	Male	8.83	8.225301829
Europe	ASD	NA	NA	NA	NA	NA	Male	9.08	9.139517533
Europe	ASD	NA	NA	NA	NA	NA	Male	8.83	7.154970232
Europe	ASD	NA	NA	NA	NA	NA	Male	9.67	9.966260719
Europe	ASD	NA	NA	NA	NA	NA	Male	8.92	8.69481855
Europe	ASD	NA	NA	NA	NA	NA	Male	9.67	12.84219838
Europe	ASD	NA	NA	NA	NA	NA	Male	8.08	10.35219735
Europe	ASD	NA	NA	NA	NA	NA	Male	8.25	8.849774575
Europe	ASD	NA	NA	NA	NA	NA	Male	8.16	9.464032218
Europe	ASD	NA	NA	NA	NA	NA	Male	8.33	10.51799454
Europe	ASD	NA	NA	NA	NA	NA	Male	8.16	9.41622481
Europe	ASD	NA	NA	NA	NA	NA	Male	8.75	13.39598874
May_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Female	7	5.473183736
May_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Male	13	15.48878288
May_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Male	55	59.49787491
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Male	1	2.790549766
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Female	4	3.956276247
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Female	15	17.87817565
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Male	1	2.320603044
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Male	4	4.348249902
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Male	1	0.959598999
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Female	1	1.994091886
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Female	10	8.697172131
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Female	14	15.7410421
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Female	6	5.13374965
Nov_2015	Angelman	*UBE3A*	NA	NA	NA	YES	Male	25	32.45470863
May_2015	ATR-X	*ATRX*	c.6254G>A	p.Arg2085His	Missense	YES	Male	6.3	6.19432086
May_2015	ATR-X	*ATRX*	c.736C>T	p.Arg246Cys	Missense	YES	Male	18	13.11825849
May_2015	ATR-X	*ATRX*	c.6593A>G	p.His2198Arg	Missense	YES	Male	1.4	2.604328944

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

Mar_2014	Floating_Harbour	*SRCAP*	c.7316dupC	p.Ala2440Serfs*3	Frameshift	YES	Male	10	4.424381743
Mar_2014	Floating_Harbour	*SRCAP*	c.7165G>T	p.Glu2389*	Nonsense	YES	Female	8	1.524333568
Mar_2014	Floating_Harbour	*SRCAP*	c.7218_7219delTC	p.Gln2407Argfs*35	Frameshift	YES	Male	12	19.26251425
Mar_2014	Floating_Harbour	*SRCAP*	c.7330C>T	p.Arg2444*	Nonsense	YES	Male	5	4.902256866
Mar_2014	Floating_Harbour	*SRCAP*	c.7330C>T	p.Arg2444*	Nonsense	YES	Female	35	38.47378886
Mar_2014	Floating_Harbour	*SRCAP*	c.7330C>T	p.Arg2444*	Nonsense	YES	Female	15	14.81418145
Mar_2014	Floating_Harbour	*SRCAP*	c.7549delC	p.Gln2517Lysfs*5	Frameshift	YES	Male	4	3.645524918
Mar_2014	Floating_Harbour	*SRCAP*	c.7330C>T	p.Arg2444*	Nonsense	YES	Female	6	7.201471688
Mar_2014	Floating_Harbour	*SRCAP*	c.7219C>T	p.Gln2407*	Nonsense	YES	Female	6	6.552720685
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	5	-0.26537653
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	10.41667	4.620596743
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	7.75	9.380603836
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	4.333333	7.378290152
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	0.083333	7.256745087
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	4.166667	6.582911793
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	21	32.38418863
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	34.58333	46.41126929
GSE41273	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	48	58.89975733
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	27	32.354974
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	12	11.03917455
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	42	40.85689027
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	28	31.89965321
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	15	15.3286979
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	17	13.98190146
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	21	21.42017869
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	30	35.16564816
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	28	27.14880628
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	21	24.03936596
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	33	37.84060062
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	29	35.17133434
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	25	25.67600147
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	17	14.45573451
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	33	36.37082822
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	29	34.45261333
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	20	24.86340454
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	41	46.76222649

May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	31	34.61968346
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	27	29.78714348
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	17	19.72629863
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	15	11.78896917
May_2015	FXS	*FMR1*	NA	NA	CGG repeat expansion	YES	Male	14	12.80759084
GSE116300	Kabuki	*KMT2D*	NA	p.Pro443fs	Frameshift	YES	Female	1	0.790826048
GSE116300	Kabuki	*KMT2D*	NA	p.Tyr2199fs	Frameshift	YES	Female	3	4.448848163
GSE116300	Kabuki	*KMT2D*	NA	p.Ser5307fs	Frameshift	YES	Male	5	11.49079359
GSE116300	Kabuki	*KMT2D*	NA	p.Asn4403fs	Frameshift	YES	Male	4.33	6.325934863
GSE116300	Kabuki	*KMT2D*	NA	p.Gln4102*	Nonsense	YES	Male	2	5.566745677
GSE116300	Kabuki	*KMT2D*	NA	p.Gln3934*	Nonsense	YES	Male	3.75	4.443224079
GSE116300	Kabuki	*KMT2D*	c.14515+1G>T	NA	Splice site mutation	YES	Male	2.5	16.55101592
GSE116300	Kabuki	*KMT2D*	NA	p.Gln4090*	Nonsense	YES	Female	1.42	3.379081974
GSE116300	Kabuki	*KMT2D*	NA	p.Thr1708fs	Frameshift	YES	Female	11.5	10.71344707
GSE97362	Kabuki	*KMT2D*	c.15061C>T	p.Arg5021*	Nonsense	YES	Female	14	8.946680052
GSE97362	Kabuki	*KMT2D*	c.16318delG	p.Glu5440Argfs*16	Frameshift	YES	Male	1	0.664960442
GSE97362	Kabuki	*KMT2D*	c.15030dup	p.Glu5011Argfs*13	Frameshift	YES	Male	18	24.00757516
GSE97362	Kabuki	*KMT2D*	c.8172_8173del	p.Pro2724Glnfs*5	Frameshift	YES	Female	16	4.540501556
GSE97362	Kabuki	*KMT2D*	c.6595delT	p.Tyr2199Ilefs*65	Frameshift	YES	Male	15	6.279894046
GSE97362	Kabuki	*KMT2D*	c.14055_14056delCA	p.His4685Glnfs*4	Frameshift	YES	Male	11	9.2260079
GSE97362	Kabuki	*KMT2D*	c.6295C>T	p.Arg2099*	Nonsense	YES	Male	14	6.594838599
GSE97362	Kabuki	*KMT2D*	c.4135delA	p.Met1379Valfs*52	Frameshift	YES	Male	20	10.04269734
GSE97362	Kabuki	*KMT2D*	c.12592C>T	p.Arg4198*	Nonsense	YES	Male	18	9.095825776
GSE97362	Kabuki	*KMT2D*	c.4135delA	p.Met1379Valfs*52	Frameshift	YES	Male	6	8.462691919
GSE97362	Kabuki	*KMT2D*	c.11710C>T	p.Gln3904*	Nonsense	YES	Male	16	12.68670209
GSE97362	Kabuki	*KMT2D*	c.15143G>A	p.Arg5048His	Missense	YES_predicted	Female	7	0.627461504
GSE97362	Kabuki	*KMT2D*	c.16522-5_16522-4delTT	NA	Splice site mutation	YES_predicted	Female	15	12.75508563
Jun_2015	Kabuki	*KMT2D*	c.1801_1822dup22	NA	Frameshift	YES	Male	7	6.044371299
Nov_2015	Kabuki	*KMT2D*	c.13059delG	p.Pro4353fs	Frameshift	YES	Female	6.7	5.526369466
Nov_2015	Kabuki	*KMT2D*	c.839+1delG	NA	Splice site mutation	YES	Male	1.9	2.51325414
Nov_2015	Kabuki	*KMT2D*	c.15844C>T	p.Arg5282*	Nonsense	YES	Female	3.9	3.752004426
Nov_2015	Kabuki	*KMT2D*	c.16294C>T	p.Arg5432Trp	Missense	YES_predicted	Male	21.6	30.3375233
Nov_2015	Kabuki	*KMT2D*	c.8488C>T	p.Arg2830*	Nonsense	YES	Female	0	-0.1055224
Nov_2015	Kabuki	*KMT2D*	c.4168dupG	p.Ala1390fs	Frameshift	YES	Female	3.8	4.177253095
Nov_2015	Kabuki	*KMT2D*	c.15289C>T	p.Arg5097*	Nonsense	YES	Male	4.3	6.455955113
Nov_2015	Kabuki	*KMT2D*	c.4419-2A>G	NA	Splice site mutation	YES	Male	2.6	3.387623395
Nov_2015	Kabuki	*KMT2D*	c.16048A>T	p.Lys5350*	Nonsense	YES	Female	19.1	19.2926115
Nov_2015	Kabuki	*KMT2D*	c.10201C>T	p.Gln3401*	Nonsense	YES	Male	7.1	8.838432826
Nov_2015	Kabuki	*KMT2D*	c.16360C>T	p.Arg5454*	Nonsense	YES	Male	3.4	5.199197126
Nov_2015	Kabuki	*KMT2D*	c.8692C>T	p.Gln2898*	Nonsense	YES	Male	3.1	3.423420462

Nov_2015	Kabuki	*KMT2D*	c.14878C>T	p.Arg4960*	Nonsense	YES	Female	4.1	4.752807097
Nov_2015	Kabuki	*KMT2D*	c.6265A>T	p.Lys2089*	Nonsense	YES	Female	23.1	25.95907184
Nov_2015	Kabuki	*KMT2D*	c.10740+1G>A	NA	Splice site mutation	YES	Female	6.9	6.253113479
Nov_2015	Kabuki	*KMT2D*	c.13652T>A	p.Leu4551*	Nonsense	YES	Male	2.2	3.757460909
Nov_2015	Kabuki	*KMT2D*	c.11596C>T	p.Gln3866*	Nonsense	YES	Female	1	1.193509229
Nov_2015	Kabuki	*KMT2D*	c.548delC	p.Pro183fs	Frameshift	YES	Female	16.6	8.413539447
Nov_2015	Kabuki	*KMT2D*	c.7411C>T	p.Arg2471*	Nonsense	YES	Female	3.3	3.541604601
Nov_2015	Kabuki	*KMT2D*	c.1966dupC	pLeu656fs	Frameshift	YES	Female	24.1	28.78927404
Nov_2015	Kabuki	*KMT2D*	c.6200delA	p.Asn2067fs	Frameshift	YES	Female	9.5	6.485224166
Nov_2015	Kabuki	*KMT2D*	c.7933C>T	p.Arg2645*	Nonsense	YES	Female	9.3	8.701999271
Nov_2015	Kabuki	*KMT2D*	c.13450C>T	p.Arg4484*	Nonsense	YES	Female	5.8	5.430619578
Feb_2016	Noonan	*PTPN11*	c.1403C>T	p.Thr468Met	Missense	YES	Male	9	10.53231848
Feb_2016	Noonan	*PTPN11*	c.1391G>C	p.Gly464Ala	Missense	YES	Female	28	25.06455423
Feb_2016	Noonan	*PTPN11*	c.1493G>T	p.Arg498Leu	Missense	YES	Male	0.4	1.069462128
Feb_2016	Noonan	*PTPN11*	c.836A>G	p.Tyr279Cys	Missense	YES	Male	0.2	0.145725107
Feb_2016	Noonan	*PTPN11*	c.1493G>T	p.Arg498Leu	Missense	YES	Male	7	7.125930003
Feb_2016	Noonan	*PTPN11*	c.1528C>G	p.Gln510Glu	Missense	YES	Female	2	4.906928458
Feb_2016	Noonan	*PTPN11*	c.228G>C	p.Glu76Asp	Missense	YES	Male	17	17.52765019
Feb_2016	Noonan	*PTPN11*	c.215C>G	p.Ala72Gly	Missense	YES	Female	13	9.011977393
Feb_2016	Noonan	*PTPN11*	c.1391G>C	p.Gly464Ala	Missense	YES	Female	0.7	1.172244358
Feb_2016	Noonan	*PTPN11*	c.922A>G	p.Asn308Asp	Missense	YES	Male	15	14.68576639
Feb_2016	Noonan	*PTPN11*	c.836A>G	p.Tyr279Cys	Missense	YES	Male	0.3	0.576697185
Feb_2016	Noonan	*PTPN11*	c.214G>T	p.Ala72Ser	Missense	YES	Male	0.9	1.080594238
Feb_2016	Noonan	*PTPN11*	c.178G>A	p.Gly60Ser	Missense	YES	Male	2	3.079510066
Feb_2016	Noonan	*PTPN11*	c.172A>G	p.Asn58Asp	Missense	YES	Male	37	42.63784241
Feb_2016	Noonan	*PTPN11*	c.174C>A	p.Asn58Lys	Missense	YES	Female	27	32.19911243
Feb_2016	Noonan	*RAF1*	c.781C>T	p.Pro261Ser	Missense	YES	Male	9	11.76954478
Feb_2016	Noonan	*RAF1*	c.770C>T	p.Ser257Leu	Missense	YES	Female	4	6.836828788
Feb_2016	Noonan	*RAF1*	c.788T>G	p.Val263Gly	Missense	YES	Male	8	10.54386119
Feb_2016	Noonan	*RAF1*	c.782C>T	p.Pro261Leu	Missense	YES	Male	3	5.956377653
Feb_2016	Noonan	*RAF1*	c.786T>A	p.Asn262Lys	Missense	YES	Female	3	3.603073783
Feb_2016	Noonan	*RAF1*	c.768G>T	p.Arg256Ser	Missense	YES	Male	20	21.09275241
Feb_2016	Noonan	*RAF1*	c.524A>G	p.His175Arg	Missense	YES	Female	0.7	0.815080545
Feb_2016	Noonan	*RAF1*	c.1837C>G	p.Leu613Val	Missense	YES	Female	10	7.425274033
Feb_2016	Noonan	*RAF1*	c.775T>A	p.Ser259Thr	Missense	YES	Female	8	8.883918263
Feb_2016	Noonan	*RAF1*	c.1472C>T	p.Thr491Ile	Missense	YES	Female	26	29.82312626
Feb_2016	Noonan	*RAF1*	c.781C>A	p.Pro261Thr	Missense	YES	Female	11	12.25565712
Feb_2016	Noonan	*SOS1*	c.2536G>A	p.Glu846Lys	Missense	YES	Female	3	2.62618922
Feb_2016	Noonan	*SOS1*	c.1654A>G	p.Arg552Gly	Missense	YES	Male	16	12.47288243
Feb_2016	Noonan	*SOS1*	c.1310T>C	p.Ile437Thr	Missense	YES	Female	7	7.309199493
Feb_2016	Noonan	*SOS1*	c.806T>C	p.Met269Thr	Missense	YES	Female	35	25.04627009
Feb_2016	Noonan	*SOS1*	c.1642A>C	p.Ser548Arg	Missense	YES	Female	3	4.372134286
Feb_2016	Noonan	*SOS1*	c.925G>T	p.Asp309Tyr	Missense	YES	Female	49	45.20434465
Feb_2016	Noonan	*SOS1*	c.1655G>C	p.Arg552Thr	Missense	YES	Male	1	2.41372048
Feb_2016	Noonan	*SOS1*	c.508A>G	p.Lys170Glu	Missense	YES	Male	0.3	0.944100935
Feb_2016	Noonan	*SOS1*	c.1294T>C	p.Trp432Arg	Missense	YES	Female	14	17.03491762

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

Nov_2015	Saethre_Chotzen	*TWIST1*	c.376G>T	p.Glu126*	Nonsense	YES	Female	0.8	0.92117994
Nov_2015	Saethre_Chotzen	*TWIST1*	c.490C>T	p.Gln164*	Nonsense	YES	Female	28.7	28.56296158
GSE74432	Sotos	*NSD1*	chr5:175,366,008-177,470,488	NA	Long deletion	YES	Female	9	8.442111023
GSE74432	Sotos	*NSD1*	chr5:175,764,262-177,059,256	NA	Long deletion	YES	Female	7	16.4840396
GSE74432	Sotos	*NSD1*	Exons 15-19 deletion	NA	Exonic deletion	YES	Male	10	26.70242296
GSE74432	Sotos	*NSD1*	c.1716delC	p.Cys573Valfs*26	Frameshift	YES	Female	10	14.59121875
GSE74432	Sotos	*NSD1*	c.6454C>T	p.Arg2152*	Nonsense	YES	Female	3.5	9.371834336
GSE74432	Sotos	*NSD1*	c.5445C>G	p.Tyr1815*	Nonsense	YES	Female	13.2	22.67264348
GSE74432	Sotos	*NSD1*	c.4843delT	p.Tyr1615Thrfs*27	Frameshift	YES	Male	3	7.039068162
GSE74432	Sotos	*NSD1*	NA	NA	Microdeletion	YES	Male	2.2	15.1797238
GSE74432	Sotos	*NSD1*	c.6349C>T	p.Arg2117*	Nonsense	YES	Female	12	26.9093016
GSE74432	Sotos	*NSD1*	c.1492C>T	p.Arg498*	Nonsense	YES	Male	2.2	8.399587071
GSE74432	Sotos	*NSD1*	c.6454C>T	p.Arg2152*	Nonsense	YES	Male	18	32.23853498
GSE74432	Sotos	*NSD1*	c.1583delA	p.Lys528Argfs*8	Frameshift	YES	Male	19.7	27.25531484
GSE74432	Sotos	*NSD1*	c.2014_2018delACAGA	p.Thr672Glufs*9	Frameshift	YES	Male	8	26.46585423
GSE74432	Sotos	*NSD1*	c.2014_2018delACAGA	p.Thr672Glufs*9	Frameshift	YES	Male	41	67.36442178
GSE74432	Sotos	*NSD1*	c.2014_2018delACAGA	p.Thr672Glufs*9	Frameshift	YES	Female	2	11.34495985
GSE74432	Sotos	*NSD1*	c.1810C>T	p.Arg604*	Nonsense	YES	Female	1.6	6.2471485
GSE74432	Sotos	*NSD1*	c.1801A>T	p.Lys601*	Nonsense	YES	Male	10.6	30.82670587
GSE74432	Sotos	*NSD1*	c.4977_4978insG	p.Arg1660Alafs*13	Frameshift	YES	Male	20	41.38296452
GSE74432	Sotos	*NSD1*	c.6437G>C	p.Cys2146Ser	Missense	YES_predicted	Male	2	9.83036953
GSE74432	Sotos	*NSD1*	c.6412T>C	p.Cys2138Arg	Missense	YES_predicted	Male	7	29.0788673
GSE74432	Weaver	*EZH2*	c.457_459delTAT	p.Tyr153del	In-frame deletion	YES	Male	30	40.6786865
GSE74432	Weaver	*EZH2*	c.2080C>T	p.His694Tyr	Missense	YES	Female	10.9167	17.28626931
GSE74432	Weaver	*EZH2*	c.2050C>T	p.Arg684Cys	Missense	YES	Male	2.5833	2.611103643
GSE74432	Weaver	*EZH2*	c.398A>G	p.Tyr133Cys	Missense	YES	Female	17	7.870608634
GSE74432	Weaver	*EZH2*	c.553G>C	p.Asp185His	Missense	YES	Male	15.4167	18.04003584
GSE74432	Weaver	*EZH2*	c.394C>T	p.Pro132Ser	Missense	YES	Female	19.75	21.09459251
GSE74432	Weaver	*EZH2*	c.1876G>A	p.Val626Met	Missense	YES	Male	43	42.37721085