

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