

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