

Supplementary Table 1: Xq28 duplications in female patients, documented in literature. The first four cases are due to unbalanced subtelomeric translocations to different autosomes, the last one is due to an insertion in chromosome 10.

Reference	Duplicated segment	Rearrangement	Microcephaly	Hypotonia	MR	RI	Seizures
Bialer <i>et al.</i> (2003)	Xq28.qter (size n.d.)	46,XX,der(13)t(X;13)(q28;p11)dn?	+	+	+	+	-
Lachlan <i>et al.</i> (2004)	Xq27.1-qter (15.7-16.6Mb)	46,XX,der(20)t(X;20)(q26.3;q13.33)mat	+	+	+	-	-
Sanlaville <i>et al.</i> (2005)	Xq28.qter (size n.d.)	46,XX add(4)(q34).ish,der(4)t(X;4)(q28;q34)dn?	+	+	+	-	-
Auber <i>et al.</i> (2010)	Xq28-qter (3.81Mb)	46,XX,der(17)t(X;17)(q28;p13.3)dn	+	+	+	+	+
Makrythanasis <i>et al.</i> (2010)	Xq28 (129Kb)	ish ins(10;X)(p15;q28) (RP11-119A22+,wcp10+;RP11-119A22+,wcp10-)	-	+	+	nr	nr

nr, not reported; MR, mental retardation; RI, recurrent infections

Supplementary Figure1: X inactivation results. The allele size of the parents and the case is indicated in brackets.

