

Table 1. Summary of the Xq28 duplication females in the study and other seven symptomatic females, inheriting their duplication from their mothers according to the literature.

Patient		Cytogenetic nomenclature	Size of Xq duplication (kb)	X-inactivation			Clinical features	
				Daughter	Mother	Technique of detection	Daughter	Mother
case1 (this study)		arr[GRCh38] Xq28 (144,988,272-145,271,978)x3 mat	437	0:100 (normal:dup)	100: 0# (normal:dup)	<i>HUMARA</i> / <i>RP2</i> assays	asymptomatic	asymptomatic #
Scott et al.,2014	Case2 Case 3 (twin)	arr[hg18] Xq28 (152,933,008–153,223,072) x 3 mat	290	uninformative	complete skewed XCI#	<i>HUMARA</i> assays	severe	mild#
Shimada et al.,2013	Case 4 (patient4)	arr[hg19] Xq28(152,916,694-153,576,940) x3 mat	660	88:12 (normal:dup)	43:57# (normal:dup)	<i>HUMARA</i> assays	severe	asymptomatic #
Reardon et al.,2010	Case 5 (family 1, III 4)	arr[hg19] Xq28(152,788,477-153,706,446)x3 mat	918	70:30	0:100	<i>HUMARA</i> assays	mild	asymptomatic
Novara et al.,2014	Case 6	arr[hg19] Xq28(153,238,518-153,406,174)×3 mat	168	random	random#	<i>HUMARA</i> assays	moderate	asymptomatic #
	Case 7	arr[hg19] Xq28(152,987,955-153,609,113)×3 mat	621	100% skewed XCI#	100% skewed XCI	<i>HUMARA</i> assays	mild#	asymptomatic
Bijlsma et al.,2012	Case 8 (patient 4)	arr[hg19] Xq28(152,706,073-153,455,489)×3 mat	700	37:67	12:88	<i>HUMARA</i> assays	mild	asymptomatic

females' clinical features are not consistent with the XCI pattern detected by *HUMARA* assays.