

Appendix 1. Overview of all patients reported with MLS. Familial cases are highlighted in bold. Patients reported in abstracts or patients for which no aberration on the X-chromosome was detected were left out of this table. NA= not applicable, ND= not determined

Patients	Sex	Karyotype	HCCS mutation	Inactivation pattern	Microphthalmia or other ocular anomalies	Skin defects	Short stature	Developmental delay	CNS anomalies	Reference
1	F	46,X,der(X)t(X;3)(p22.2;q12)		skewed	+	-	+	ND	ND	Ropers (1982)
2	F	46,X,der(X)t(X;Y)(p22.3;q11.2)		skewed	+	+	+	-	-	Al-Gazali (1990)
3	F	46,X,der(X)t(X;Y)(p22.3;q11.2)		ND	+	+	+	-	+	Al-Gazali (1990)
4	F	46,X,der(X)t(X;3)(p22.2;p22.2)		ND	+	-	+	-	+	Donnenfeld (1990)
5	F	46,X,del(X)(p22.2)		ND	+	+	ND	-	-	Temple (1990)
6	F	46,X,del(X)(p22.2)		ND	+	+	ND	ND	+	Allanson (1991)
7	F	46,X,del(X)(p22.2)		ND	-	-	+	-	-	Allanson (1991)
8	F	46,X,del(X)(p22.3)		skewed	+	-	-	-	-	Thies (1991)
9	F	46,X,del(X)(p22.1p22.31)		skewed	+	+	+	-	+	Naritomi (1992)
10	F	46,X,del(X)(p22.1p22.31)		random	+	+	ND	ND	+	Naritomi (1992)
11	F	46,X,del(X)(p22.3)		skewed	+	+	ND	ND	-	Lindor (1992)
12	F	46,X,del(X)(p22.3)		skewed	+	+	+	+	-	Lindsay (1994)
13	F	46,X,del(X)(p22.3)		ND	+	+	+	-	-	Lindsay (1994)
14	M	46,X,der(X)t(X;Y)		ND	-	+	NA	NA	+	Lindsay (1994)
15	F	46,X,del(X)(p22.3)		ND	+	+	ND	+	+	Eng (1994)
16	F	46,X,der(X)t(X;Y)(p22.2;q11.2)		ND	+	+	+	-	-	Mucke (1995)
17	F	46,X,der(X)t(X;Y)(p22.2;q11.2)		ND	+	+	+	+	+	Mucke (1995)
18	M	46,X,der(X)t(X;Y)		ND	+	+	+	+	+	Paulger (1997)
19	F	45,X[18]/46,X,der(X)(p22q21)[24]/46,X,del(X)(p22)[58]		random	+	+	+	-	+	Ogata (1998)
20	M	46,X,der(X)t(X;Y)		ND	+	-	ND	ND	ND	Kobayashi (1998)
21	M	46,X,der(X)t(X;Y)		ND	+	+	ND	ND	ND	Stratton (1998)
22	M	46,X,der(X)t(X;Y)(p22.13;p11.2)		ND	+	-	ND	ND	-	Kono (1999)
23	F	46,X,del(X)(p22.3)		ND	+	+	-	+	+	Kayserili (2001)
24	M	46,Y,inv(X)(p22.13~22.2p22.32~22.33)[49]/46,XY[271]		ND	-	+	ND	ND	+	Kutsche (2002)
25	M	46,X,t(X;Y)(p22.3;p11.3)		skewed	+	+	+	-	-	Anguiano (2003)
26	M	46,X,t(X;Y)(p22.3;p11.3)		skewed	+	+	+	-	-	Anguiano (2003)
27	F	46,X,del(X)(p22.3)		ND	+	+	+	-	-	Enright (2003)
28	M	46,X,der(X)t(X;Y)		ND	+	+	ND	ND	+	Cape (2004)
29	M	46,X,der(X)t(X;Y)		ND	+	-	+	ND	-	Cape (2004)
30	F	46,X,der(X)t(X;Y)		ND	+	+	ND	ND	ND	Cape (2004)
31	F	46,X,der(X)t(X;Y)		ND	+	-	+	ND	ND	Cape (2004)
32	F	46,XX,del(X)(p22.22)		ND	+	+	+	+	-	Morleo (2005)
33	F	46,X,der(X)t(X;Y)(p22.22;q11.2)		ND	+	+	+	ND	-	Morleo (2005)
34	F	46,X,der(X)t(X;3)(p22.2;q25.3)		ND	+	+	-	+	-	Morleo (2005)
35	F	46,X,del(X)(p22.22p22.22)		ND	+	+	ND	-	-	Morleo (2005)
36	F	46,XX	8,6 kb microdeletion covering part of HCCS	skewed	+	+	-	+	+	Wimplinger (2006)/Morleo (2005)
37	F	46,XX	8,6 kb microdeletion covering part of HCCS	skewed	+	-	ND	-	-	Wimplinger (2006)
38	F	46,XX	c.589C>T p.R197X	skewed	+	+	-	+	-	Wimplinger (2006)
39	F	46,XX	c.649C>T p.R217C	skewed	+	-	-	ND	+	Wimplinger (2006)
40	F	46,X,del(X)(p22.2) (mosaic in mother without phenotype)		skewed	+	+	ND	ND	ND	Wimplinger (2007)
41	F	46,XX	c.475G>A p.E159K	skewed	+	-	ND	ND	ND	Wimplinger (2007b)
42	F	46,X,der(X)t(X;Y)(p22.3;q11.2)		skewed	+	+	ND	ND	+	Cain (2007)
43	F	46,X,del(X)(p21.3)		skewed	+	+	ND	ND	ND	Schluth (2007)
44	F	46,X,del(X)(p22.3)		ND	+	+	ND	ND	-	Kapur (2008)
45	M	46,X,der(X)t(X;Y)		ND	+	+	ND	ND	-	Kapur (2008)
46	F	46,X,del(X)(p22.2)		ND	+	+	+	-	-	Sharma (2008)
47	F	46,X,del(X)(p22.2)		skewed	+	+	+	+	-	Hobson (2009)
48	F	46,X,del(X)(p22.2)		skewed	+	+	+	+	-	Steichen-Gersdorf (2010)
49	F	46,X,del(X)(p22.3)		skewed	+	+	ND	ND	ND	Alberry (2010)
50	M	46,X,der(X)t(X;Y)(p22.2;p11.2)		ND	+	+	-	NA	+	Qidwai (2010)
51	F	46,X,del(X)(p22.3)		ND	+	+	ND	+	+	Zumwalt (2012)