

**Table S1.** Publications contributing heteroplasmy values m.3243A>G and m.8993T>G for meta-analysis.

m.3243A>G mutation			m.8993T>G mutation		
Pedi-grees	Transmis-sions	Reference	Pedigrees	Transmis-sions	Reference
3	8	(Ciafaloni et al., 1992)	1	3	(Holt et al., 1990)
2	8	(Martinuzzi et al., 1992)	1	1	(Sakuta et al., 1992)
1	5	(Huang et al., 1994)	1	2	(Puddu et al., 1993)
1	4	(Liou et al., 1994)	1	4	(Shoffner et al., 1992)
6	15	(Hammans et al., 1995)	1	5	(Tatuch et al., 1992)
3	8	(Iwanishi et al., 1995)	1	2	(Ciafaloni et al., 1993)
1	2	(Rusanen et al., 1995)	4	6	(Santorelli et al., 1993)
1	1	(Fabrizi et al., 1996)	1	5	(Fryer et al., 1994)
1	3	(Huang et al., 1996)	1	1	(Lodi et al., 1994)
2	8	(Jansen et al., 1997)	1	1	(Pastores et al., 1994)
1	2	(Vilarinho et al., 1997)	1	6	(Degoul et al., 1995)
1	1	(Wilichowski et al., 1998)	1	3	(Houstek et al., 1995)
3	12	(Olsson et al., 1998)	1	8	(Makela-Bengs et al., 1995)
2	2	(Onishi et al., 1998)	1	2	(Tulinius et al., 1995)
1	2	(Chinnery et al., 1999)	1	3	(Bartley et al., 1996)
1	8	(Dubeau et al., 2000)	1	3	(Mak et al., 1996)
1	1	(Hotta et al., 2001)	1	4	(Ferlin et al., 1997)
1	2	(Ko et al., 2001)	4	9	(Uziel et al., 1997)
1	5	(Lien et al., 2001)	4	16	(White et al., 1999)
1	2	(Morovvati et al., 2002)	1	2	(Porto et al., 2001)
1	3	(Cervin et al., 2004)	1	2	(Wong et al., 2002)
1	1	(Chou et al., 2004)	1	3	(Enns et al., 2006)
1	1	(Lu et al., 2002)	1	3	(Steffann et al., 2007)
			2	5	(Mkaouar-Rebai et al., 2009)