

**Appendix 2B** Case-Control Frequency of the MSX1 Mutations and Polymorphisms Diagnosed in this Study

Nucleotide	Variant Frequency <sup>a</sup> /Population	"100"	CL/P	CPO	Control
5'utr					
223	South American	-	0/132	0/14	-
- 247C>T	Caucasian	1/139	1/129	0/10	0/172
	Asian	0/146	0/146	-	-
303	South American	-	0/132	0/14	-
-167C>T	Caucasian	0/140	0/130	0/10	1/171
	Asian	0/146	0/146	-	-
339	South American	-	1/127	0/8	-
- 130C>G	Caucasian	0/140	0/130	0/10	13/157
	Asian	0/146	0/146	-	-
434	South American	-	2/200	0/18	0/108
- 36G>A	Caucasian	9/139	20/284	7/91	15/271
	Asian	0/146	0/298	0/46	-
Exon 1					
516	South American	-	0/202	0/18	2/106
47G>A	Caucasian	0/48	0/214	0/88	0/306
	Asian	0/142	0/432	0/48	-
537	South American	-	1/201	0/18	4/104
68C>T	Caucasian	0/48	0/224	0/88	0/306
	Asian	0/142	0/438	0/48	-
559	South American	-	0/202	0/18	0/108
90C>A	Caucasian	0/48	0/224	0/88	0/308
A30A	Asian	0/142	1/437	0/48	-
570	South American	-	1/201	1/17	5/103
101C>G	Caucasian	12/36	35/189	17/71	57/251
	Asian	5/137	15/423	3/45	4/102
702	South American	-	0/352	0/32	0/108
233A>T	Caucasian	0/48	0/224	0/88	0/310
E78V	Asian	0/142	3/435	0/48	1/671

709	South American	-	1/351	0/32	0/108
240G>A	Caucasian	0/48	0/224	0/88	0/310
V80V	Asian	0/142	0/438	0/48	0/352
741	South American	-	0/352	0/32	0/108
272G>A	Caucasian	0/48	0/224	0/88	0/310
G91D	Asian	0/142	1/437	0/48	-
799	South American	-	3/349	0/32	7/101
330C>T	Caucasian	7/81	15/209	7/81	25/285
G110G	Asian	19/123	63/373	11/37	8/98
810	South American	-	0/352	0/32	0/108
341T>G	Caucasian	0/48	1/223	0/88	0/310
V114G	Asian	0/142	0/436	0/48	-
816	South American	-	1/351	0/32	0/108
347G>A	Caucasian	0/48	0/224	0/88	0/310
G116E	Asian	0/142	0/436	0/48	-
823	South American	-	1/351	0/32	0/108
354C>T	Caucasian	0/48	0/224	0/88	0/310
L118L	Asian	0/142	0/436	0/48	-
Intron 1					
961-971	South American	-	22/80	0/6	-
451+(41-	Caucasian	-	81/227	25/31	19/41
1139	Caucasian	36/112	33/43	3/9	-
451+219G>A	Asian	6/160	6/160	-	-
1152	Caucasian	14/90	14/80	0/10	-
451+232A>T	Asian	105/47	105/47	-	-
1188	Caucasian	0/104	0/94	0/10	-
451+232C>T	Asian	1/175	1/175	-	-
1259-1262	Caucasian	0/164	0/146	0/18	-
451+(339-	Asian	6/172	6/172	-	-
1325	Caucasian	16/152	12/136	4/14	-
451+405G>A	Asian	12/156	12/156	-	-

1348	Caucasian	9/159	9/141	0/18	-
451+428C>T	Asian	16/152	16/152	-	-
1440	Caucasian	7/161	7/143	0/18	-
451+520C>T	Asian	18/160	18/160	-	-
1510	Caucasian	53/121	51/107	2/14	-
451+590G>C	Asian	61/17	61/17	-	-
1807	South American	-	0/202	0/18	-
451+887G>T	Caucasian	0/178	0/412	1/77	0/194
	Asian	0/94	0/294	0/48	0/194
1966	South American	-	0/202	0/18	-
451+1046C>T	Caucasian	0/178	0/412	1/77	0/194
	Asian	0/94	0/294	0/48	0/210
2121	Caucasian	48/120	42/108	6/12	37/147
451+1201T>G	Asian	6/176	7/175	-	-
2450	South American	-		0/34/1/8/3	-
452-780	Caucasian	-			0/310/37/139/64
CA 8-12	Asian	-			2/1162/3/265/60
2564	Caucasian	43/121	35/97	4/14	120/127
452-683C>A	Asian	115/65	115/65	-	-
2573	Caucasian	0/164	0/132	0/18	-
452-674G>A	Asian	2/178	2/178	-	-
2580	Caucasian	111/53	97/49	14/4	116/57
452-667T>G	Asian	45/133	45/133	-	-
2845	Caucasian	45/123	41/109	4/14	-
452-402G>T	Asian	112/66	112/66	-	-
3049	South American	-	0/216	0/28	-
452-198A>G	Caucasian	12/156	11/185	3/17	-
	Asian	0/178	0/178	-	-

3095	Caucasian	8/160	13/181	0/20	-
452-152A>G	Asian	2/176	2/176	-	-
3118	South American	-	0/216	0/28	-
452-129G>A,C	Caucasian	1/10/185	1/10/185	3/17	-
	Asian	2/176	2/176	-	-
3120	South American	-	0/216	0/28	-
452-127 C>A	Caucasian	36/134	37/187	3/25	23/155
	Asian	6/176	6/202	0/4	-
3200	Caucasian	34/96	41/165	3/19	-
452-47 A>G	Asian	8/172	2/338	0/44	-
3232	Caucasian	3/11	63/175	9/13	8/14
452-15delT	Asian	3/89	8/234	3/45	-
3233	Caucasian	0/24	1/238	0/22	-
452-14insC	Asian	0/92	0/242	0/48	-
Exon 2					
3248	South American	-	0/216	0/28	-
453G>T	Caucasian	0/44	0/314	0/32	0/78
R151S	Asian	0/50	0/264	0/50	0/36
3338	South American	-	0/216	0/28	-
543G>A	Caucasian	1/73	4/400	0/32	3/91
L181L	Asian	0/96	0/264	0/50	0/58
3620	South American	-	0/216	0/28	-
825C>T	Caucasian	0/76	0/412	0/32	0/142
A275A	Asian	0/96	1/263	0/50	0/54
3695	South American	-	29/187	3/25	-
*6C>T	Caucasian	17/59	98/308	15/25	24/80
	Asian	3/91	8/254	3/47	3/39
3'utr					
3758	South American	-	9/207	0/28	-
*69C>T	Caucasian	0/20	2/94	-	-
	Asian	11/25	11/25	-	-

3760	South American	-	1/215	0/28	-
*71C>T	Caucasian	0/20	0/96	-	-
	Asian	0/36	0/36	-	-
3969	Caucasian	10/16	10/16	-	-
*280A>G	Asian	22/12	22/12	-	-
4401	South American	-	0/216	0/28	-
*712T>A	Caucasian	0/126	0/108	0/18	0/96
	Asian	1/167	1/167	-	-
4500	South American	-	117/207	9/19	-
*(811-816)del	Caucasian	40/86	34/74	6/12	31/121
	Asian	8/164	8/164	-	-

Notes: <sup>a</sup>Number of variants is number of chromosomes for each allele type, not individuals; <sup>b</sup>CL/P, from patients with nonsyndromic cleft lip with or without cleft palate; <sup>c</sup>CPO, from patients with nonsyndromic cleft of the palate only.