

Online Supplemental Table 1

21qter critical region microsatellite markers* and SNPs* for the CFEOM/U family members

Marker Name	Position on Chromosome 21 (bp)	I-3	II-8	II-9	II-10	II-12	II-13	II-14	III-4	III-5	III-10	III-11	III-14	IV-1
D21S1260	41 616 421		208 210	210 218	204 210	204 210	204 218	204 210	204 208	204 208	208 218	208 210	204 206	208 208
rs234781	43 324 461			C C	C C			C C			C T	C T		
rs2839627	43 342 854			C T	C C			C C			T C	C C		
rs170916	43 342 899			C G	C C			C C			G G	C G		
rs234728	43 342 911			A G	A A			A A			G G	A G		
D21S1890	43 704 380	162 164	149 162	162 162	162 162	162 164	162 164	162 162	149 162	149 156	149 162	149 162	149 162	149 149
D21S1259	44 180 066	204 218	216 218	204 204	204 204	204 218	218 218	204 204	218 218	218 218	204 218	204 218	204 216	218 218
D21S1912	44 433 938	169 197	169 191	169 169	169 169	169 197	197 199	169 169	191 194	191 197	169 191	169 191	169 199	191 191
1305K* *	44 748 500	243 253	245 245	243 243	243 243	243 253	247 253	243 243	245 247	245 249	243 245	243 245	243 247	245 245
D21S171	44 848 869	113 123	115 115	113 113	113 113	113 123	117 123	113 113	115 117	115 119	113 115	113 115	113 117	115 115
D21S1903	44 997 254	239 255	245 253	255 255	255 255	239 255	239 243	255 255	248 253	245 253	253 255	253 255	249 255	253 253

Marker Name	Position on Chromosome 21 (bp)																	
		I-3	II-8	II-9	II-10	II-12	II-13	II-14	III-4	III-5	III-10	III-11	III-14	IV-1				
D21S18	45 400	200																
97	825	202	196 200	202 202	202 202	200 202	200 202	202 202	196 196	196 200	196 202	196 202	202	196				
rs19993	45 526	C C		C C							C G	C G						
33	603																	
rs19993	45 526	A A		A A							A G	A G						
34	659																	
2044K*	45 585	248	250 254	248 248	248 248	248 248	244 248	248 248	244 250	250 254	248 250	248 250	244	250				
*	432	248											248	250				
rs28389	45 696	C C		C C							C C	C C						
17	106																	
rs72535	46 004	T T		T T							T T	T T						
8	439																	
2849K*	46 292	228	226 228	228 228	228 228	228 228	228 228	228 228	226 228	226 228	228 228	228 228	228	228				
*	500	228											230	228				
rs28391	46 517	T T		T T							T T	T T						
68	817																	
rs10606	46 518	G G		G G							G G	G G						
09	648																	
3086K*	46 529	251	251 259	251 251	251 251	251 251	251 251	251 251	251 251	251 259	251 251	251 251	251	251				
*	500	251											253	251				
rs28392	46 656	T T		T T							T T	T T						
35	794																	
3258K*	46 701	215	215 218	215 215	215 215	215 215	215 215	215 215	215 224	215 218	215 215	215 215	215	215				
*	500	215											221	215				
rs28392	46 754	C C		C C							C C	C C						

Marker Name	Position on Chromosome 21 (bp)	Chromosomes													
		I-3	II-8	II-9	II-10	II-12	II-13	II-14	III-4	III-5	III-10	III-11	III-14	IV-1	
81	169														
D21S14	46 793	213	213 224	213 213	213 213	213 213	213 213	213 213	213 224	213 224	213 213	213	213	213	
46	784	213										213	224	213	
rs9722	46 875 441	C C		C C								C C	C C		
rs88182	46 876 070	C T	C C	C C	C C	C T	C T	C C	C C	C C	C C	C C	C C	C C	

*Markers sizes in bp or SNP base.

**Newly designed markers. For primer sequence information, see online Supplemental Table 2.

Affected individuals are shown in bold. The critical region homozygous in all affected individuals is framed in bold.

Online Supplemental Table 2

Primer information of the newly designed microsatellite markers

A

Marker	Primer Sequence		Observed Allele Sizes (bp)
	Forward	Reverse	
1305K	5'-ACCGCTGGTACATGTGAATG-3'	5'-TGCTATCTTTGGGGTCCTTG-3'	243–253
2044K	5'-CATCAACACCCACACCAGAG-3'	5'-GTGTGCACAGTGCTCTCCAG-3'	248–254
2849K	5'-GGACCGATGGACAGAAGGTA-3'	5'-TGCAGCCATGAATTAGATGC-3'	226–230
3086K	5'-CACATGTTGGTGGTGGACA-3'	5'-CCGCCACTGACTCTCTCTCT-3'	251–259
3858K	5'-TTACCCTGTGCATTTCTGGA-3'	5'-GCAGTGAGCTGAGATTGTGC-3'	215–221