

Table S5 Polymorphism analysis of rs34201045 (4 bp insertion) in *TP63*

	Polymorphism analysis					Statistical analysis (<i>P</i> -value)			
	Patients				Carriers (n=15)	SHFM vs. SHFLD	Carriers vs. Patients		
	Total (n=27)	SHFM (n=17)	SHFLD (n=8)	GWC (n=2)			Total	SHFM	SHFLD
<Allele frequency>									
Insertion (-)	23	15	7	1	13	0.61	0.62	0.58	0.61
Insertion (+)	31	19	9	3	17				
<Genotype frequency>									
Insertion (-)/(-)	5	3	2	0	2	0.89	0.72	0.73	0.96
Insertion (-)/(+)	13	9	3	1	8				
Insertion (+)/(+)	9	5	3	1	3				
Insertion (-)/(-)	5	3	2	0	2	0.62	0.59	0.67	0.55
Insertion (-)/(+) plus Insertion (+)/(+)	22	14	6	2	11				
Insertion (-)/(-) plus Insertion (-)/(+)	18	12	5	1	10	0.68	0.39	0.43	0.48
Insertion (+)/(+)	9	5	3	1	3				

The genotyping data satisfy the Hardy-Weinberg equilibrium (Fisher's exact test with one degree of freedom).

The (+) and the (-) symbols represent the presence and absence of the 4 bp insertion, respectively.

The 95% confidence intervals for odds ratios include 1.0 in all the comparisons (not shown).

P-values have not been calculated for GWC, because of the small patient number.

The allele frequency in 100 Japanese control subjects is 44% for Insertion (-) and 56% for Insertion (+) (our data).