

Table S4. The percentages of the mutations impacting on the NTD only, the CTD only, or both domains in the subjects with jaw disease, kidney disease, or uterine disease. The ratios of the above mutations in the subjects who developed the specified disease were compared to the ratio in all the subjects. The significance of the difference was calculated using chi-square or Fisher's exact test, as appropriate.

	Protein domain affected by the mutations			Statistical Significance
	NTD	NTD+CTD	CTD	
Jaw disease (N=83)	2 (2.4%)	73 (88.0%)	8 (9.6%)	P=0.007
Kidney disease (N=85)	7 (8.2%)	76 (89.4%)	2 (2.4%)	P=0.616
Uterine disease (N=52)	8 (15.4%)	40 (76.9%)	4 (7.7%)	P=0.275