

Table S3. The percentages of the low-impact and the high-impact mutations in the subjects who developed jaw disease, kidney disease or uterine disease. The ratio of the above mutations in the subjects who developed the specified disease was 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.

	Percentage of the CDC73 germline mutations		Statistical Significance
	High-impact	Low-impact	
Jaw disease (N=84)	81 (96.4%)	3 (3.6%)	P=0.019
Kidney disease (N=86)	79 (91.9%)	7 (8.1%)	P=0.409
Uterine disease (N=54)	43 (79.6%)	11 (20.4%)	P=0.127