Supplementary Table 2. Mutation type, genotype group and parathyroid phenotype of all subjects included in the present study.

CDC73 DNA mutations	Total No. of pts	No. of pts w/ parathyroid carcinoma	No. of pts w/ benign parathyroid lesion	No. of pts w/o parathyroid disease	Mutation type [1]	Major/ Minor Group [2]	Protein domain affected [3]
c.-16_8del24	1		1		deletion, gross	High	N+C
c.3G>A	4		4		deletion, gross	High	N+C
deletion of exon 3	1		1		deletion, gross	High	N+C
deletion of exons 1-10	8	2	4	2	deletion, gross	High	N+C
deletion of exons 2-3	1		1		deletion, gross	High	N+C
deletion of exons 4-6	5	1	4		deletion, gross	High	N+C
deletion of exons 7-13	1		1		deletion, gross	High	N+C
deletion of the gene	15	6	4	5	deletion, gross	High	N+C
c.1239delA	4		3	1	deletion, small	High	C
c.1242delA	1	1			deletion, small	High	C
c.1346delG	5	1	1	3	deletion, small	High	C
c.1382delT	4	1	1	2	deletion, small	High	C
c.140_144delAGGAA	12	2	5	5	deletion, small	High	N+C
c.1432_1433delCT	1		1		deletion, small	High	C
c.22delC	1		1		deletion, small	High	N+C
c.242_253del12	2		1	1	deletion, small	Low	N
c.253_258delGTGGTT	5		5		deletion, small	Low	N
c.260_261delGA	2	2			deletion, small	High	N+C
c.30delG	4		4		deletion, small	High	N+C
c.35_41delACATCCA	4		4		deletion, small	High	N+C
c.356delA	14	3	7	4	deletion, small	High	N+C
c.40delC	4	1	3		deletion, small	High	N+C
c.481_484 delAAAG	1			1	deletion, small	High	N+C

c.520_523delTCTG	6	2	4		deletion, small	High	N+C
c.571delG	1	1			deletion, small	High	N+C
c.62_66delAGGGA	1		1		deletion, small	High	N+C
c.626_629delAACA	2	2			deletion, small	High	N+C
c.639delT	5		4	1	deletion, small	High	N+C
c.685_688delAGAG	15	2	5	8	deletion, small	High	N+C
c.687_688delAG	10	3	6	1	deletion, small	High	N+C
c.766_767delGT	30		8	22	deletion, small	High	N+C
c.76delA	5	3	2		deletion, small	High	N+C
c.85delG	2		2		deletion, small	High	N+C
c.20_24delTCCTGinsCCCT	3		3		indels, small	High	N+C
c.433_442delinsAGA	9	1	5	3	indels, small	High	N+C
c.668_669delATinsG	5	2	3		indels, small	High	N+C
41-bp insertion exon 1	7	2	5		insertion, gross	High	N+C
c.1124_1125dupTT	1			1	insertion, small	High	C
c.127_128insC	1	1			insertion, small	High	N+C
c.14_17dupTTAG	2	1	1		insertion, small	High	N+C
c.194dupA	4		3	1	insertion, small	High	N+C
c.205dupC	3		3		insertion, small	High	N+C
c.375dupA	1	1			insertion, small	High	N+C
c.687_688dupAG	45	8	26	11	insertion, small	High	N+C
c.692_693insT	1	1			insertion, small	High	N+C
c.745dup1	1		1		insertion, small	High	N+C
g.95151T>C	2		2		intronic	Low	-
c.1135G>A	1		1		missense	Low	C
c.163T>A	1		1		missense	Low	N
c.164A>C	12		9	3	missense	Low	N
c.176C>T	1	1			missense	Low	N

c.188T>C	9		6	3	missense	Low	N
c.191T>C	5		5		missense	Low	N
c.272G>C	1		1		missense	Low	N
c.284T>C	7		5	2	missense	Low	N
c.293T>C	1		1		missense	Low	N
c.815A>G	1		1		missense	Low	N
c.1475G>A	1		1		nonsense	High	C
c.157G>T	2		2		nonsense	High	N+C
c.162C>G	2		2		nonsense	High	N+C
c.165C>A	8	1	6	1	nonsense	High	N+C
c.165C>G	7	1	5	1	nonsense	High	N+C
c.226C>T	4	3	1		nonsense	High	N+C
c.25C>T	9		8	1	nonsense	High	N+C
c.271C>T	3		3		nonsense	High	N+C
c.343G>T	1	1			nonsense	High	N+C
c.376C>T	2	1	1		nonsense	High	N+C
c.406A>T	6		6		nonsense	High	N+C
c.40C>T	2	1	1		nonsense	High	N+C
c.415C>T	4	3	1		nonsense	High	N+C
c.505C>T	3		3		nonsense	High	N+C
c.664C>T	15	5	8	2	nonsense	High	N+C
c.700C>T	7	3	3	1	nonsense	High	N+C
c.85G>T	2		2		nonsense	High	N+C
c.96G>A	13	2	9	2	nonsense	High	N+C
*12C>A	1		1		regulatory	Low	-
c.-4dupG	1		1		regulatory	Low	-
c.131+1G>A	3		3		splicing	High	N+C
c.237+1G>C	7		5	2	splicing	High	N+C
c.238-1G>A	2	2			splicing	High	N+C

c.306_307+13del	7	1	6		splicing	High	N+C
c.307+1G>A	2	1	1		splicing	High	N+C
c.307+5G>T	1		1		splicing	High	N+C
c.308-9T>A	1		1		splicing	High	N+C
c.423+1G>A	1	1			splicing	High	N+C
c.424-5T>C	1		1		splicing	High	N+C
Sum of distinct CDC73 DNA mutations: 86	Sum of total No. of pts: 419	Sum of No. of pts w/ parathyroid carcinoma: 77	Sum of No. of pts w/ benign parathyroid lesion: 252	Sum of No. of pts w/o parathyroid disease: 90			

1: Mutation type as assigned by HGMD (Human Genetic Mutation Database) from http://www.hgmd.cf.ac.uk/ac/index.php.

2: Group assigned based on the definition described in the Results section.

3: The effect of the mutations was predicted by Mutalyzer (https://mutalyzer.nl/) and Human Splicing Finder 3.0 (http://www.umd.be/HSF3/index.html). N, affecting the NTD only; C, affecting the CTD only, N+C, affecting both the NTD and the CTD.