

**Supplementary Table.** Fifty-one pathogenic mutations in PRKAR1A that have been studied in 745 individuals enrolled in NYCP

MUTATIONS in PRKAR1A found in Patients with CNC	
1	c.491_492delTG/p.Val164fsX4
2	IVS7-(5_107)del 103
3	c.438A>T/p.Arg146Ser
4	c.439A>T/p.Ser147Gly
5	c.738T>G/ p.Tyr246X
6	c.82C>T/p.Gln28X
7	IVS7-1G>C or c.839-1G>C
8	c.220C>T/p.Arg74Cys
9	c.786_787GG>CT/p.Glu263X
10	c.1076_77delTTins13/p.Leu359fsPLUS62
11	c.46C>T/p.Arg16X
12	IVS6del(-9>-2)
13	c.1A>G/p.Met1Val
14	c.101_105delCTATT/p.Ser34fsX9
15	IVS9+3A>G or c.891+3A>G
16	IVS5+1G>A
17	c.951delA/p.Arg317fsX13
18	c.124C>T/p.Arg42X
19	c.319G>T/p.Glu107X
20	c.286C>T/p.Arg96X
21	c.69_70duplGA/p.Lys24fsX105
22	c.353_365del13/p.Ile118fsX6
23	IVS2+1 G>A or C177+1G>A
24	IVS7-(2_7)del6 or c.709-7del6(TTTTTA)
25	c.440 + 1G>T
26	c43_58del16/p.Leu15fs104X
27	c.528_531del4/ins11/p.Ile176delinsfsX13
28	c.725dupA / p.Lys242fsX5
29	c.1055_1058del4(GACC)/p.Arg352fsX87
30	IVS4+4delG or c.440+ 4delG
31	c.340delG/p.Val113fsX15
32	c.206C>T/p.Gln69X
33	c.812dupT/p.Leu271fsX6
34	c.566_567AA>CAC/p.Glu189fsX43
35	c.682C>T/p.Arg228X
36	c.712insAA/p.Ser238fsX3
37	c.267delA/p.Pro89fsX39
38	IVS2-2A>G or c.175-2 A>G
39	c.693insT/p.Arg232X
40	c.187A>T/p.Lys63X
41	c.18delC/Thr6fsX122
42	c.531_(539+32)del/p.Ile177fsX12
43	c.679delG/p.Asp227fsX13
44	c1067_1070delAACGinsGCCCA/p.Glu356fsPLUS45
45	c1142_1145delTCTG/p.Val381fsPLUS58
46	c.279_282delTAGG/p.Arg94fsX34
47	c.220_21delCG/p.Arg74fsX6
48	c.547G>T/p.Asp183Tyr
49	IVS4+1insG or c.440+ 1 insG
50	c.672G>A/p.Trp224X
51	IVS4+2T>C, or c.440+2 T>C