

Supplementary Table 1. List of the published case reports/case series included in the present study.

<i>CDC73</i> germline mutation	Published case reports/case series
*12C>A	Frank-Raue (2011) Eur J Endocrinol 165,477
41-bp insertion exon 1	Carpten (2002) Nat Genet 32, 676
c.1124_1125dupTT	Pimenta (2006) Oral Oncol 42, 735
c.1135G>A	Bradley (2006) Clin Endocrinol (Oxf) 64, 299
c.1239delA	Carpten (2002) Nat Genet 32, 676 Bradley (2005) J Intern Med 257, 18
c.127_128insC	Juhlin (2007) Endocr Relat Cancer 14, 501
c.131+1G>A	Cetani (2004) J Clin Endocrinol Metab 89, 5583
c.1346delG	Frank-Raue (2011) Eur J Endocrinol 165, 477
c.1382delT	Chiofalo (2014) Cell Oncol (Dordr) 37, 281
c.14_17dupTTAG	Khadilkar (2015) Endocr Pract 21, 1010
c.140_144delAGGAA	Masi (2008) Endocr Relat Cancer 15, 1115 Kelly (2006) J Bone Miner Res 21, 1666
c.1432_1433delCT	Frank-Raue (2011) Eur J Endocrinol 165, 477
c.1475G>A	Siu (2011) Fam Cancer 10, 695
c.157G>T	Kong (2014) Clin Endocrinol (Oxf) 81, 222
c.-16_8del24	Bellido (2016) Endocr Pathol, 27:142-146
c.163T>A	Bricaire (2013) J Clin Endocrinol Metab 98, E403
c.165C>A	Veiguela (2010) Endocrinol Nutr 57, 391
c.165C>G	Carpten (2002) Nat Genet 32, 676 Bradley (2005) J Intern Med 257, 18
c.176C>T	Haven (2007) Clin Endocrinol (Oxf) 67, 370
c.188T>C	Masi (2008) Endocr Relat Cancer 15, 1115
c.191T>C	Howell (2003) J Med Genet 40, 657

	Villablanca (2004) J Med Genet 41, e32
c.194dupA	Takeuchi (2015) Clin Pediatr Endocrinol 24, 185
c.20_24delTCCTGinsCCCT	Aldred (2006) Oral Surg Oral Med Oral Pathol Oral Radiol Endod 101, 212
c.205dupC	Pichardo-Lowden (2011) Endocr Pract 17, 602
c.226C>T	Siu (2011) Fam Cancer 10, 695
c.22delC	Carlson (2008) Endocr Pract 14, 743
c.237+1G>C	Bricaire (2013) J Clin Endocrinol Metab 98, E403 Villablanca (2004) J Med Genet 41, e32
c.238-1G>A	Moon (2005) J Clin Endocrinol Metab 90, 878
c.242_253del12	Pazienza (2013) PLoS One 8, e82292
c.253_258delGTGGTT	Pazienza (2013) PLoS One 8, e82292
c.25C>T	Carpten (2002) Nat Genet 32, 676 Bradley (2005) J Intern Med 257, 18 Schmitdt (2009) J Oral Maxillofac Surg 67:423
c.260_261delGA	Wang (2012) PLoS One 7, e45567 Kong (2013) Zhonghua Yi Xue Za Zhi 93, 3364
c.271C>T	Bricaire (2013) J Clin Endocrinol Metab 98, E403 Shibata (2015) endocrine journal 62(7) Starker (2012) Horm Cancer 3, 44
c.272G>C	Cetani (2007) Endocr Relat Cancer 14, 493
c.293T>C	Bricaire (2013) J Clin Endocrinol Metab 98, E403
c. 306_307+13del	Carpten (2002) Nat Genet 32, 676
c.307+1G>A	Kong (2014) Clin Endocrinol (Oxf) 81, 222
c.307+5G>T	Frank-Raue (2011) Eur J Endocrinol 165, 477
c.308-9T>A	Bricaire (2013) J Clin Endocrinol Metab 98, E403
c.30delIG	Carpten (2002) Nat Genet 32, 676

c.343G>T	Cetani (2008) J Endocrinol Invest 31, 900
c.35_41delACATCCA	Carpten (2002) Nat Genet 32, 676
c.356delA	Bradley (2005) J Intern Med 257, 18 Carpten (2002) Nat Genet 32, 676
c.375dupA	Shattuck (2003) N Engl J Med 349, 1722
c.376C>T	Bricaire (2013) J Clin Endocrinol Metab 98, E403
c.3G>A	Carpten (2002) Nat Genet 32, 676
c.406A>T	Carpten (2002) Nat Genet 32, 676
c.40C>T	Khadilkar (2015) Endocr Pract 21, 1010 Kong (2013) Zhonghua Yi Xue Za Zhi 93, 3364
c.40delC	Carpten (2002) Nat Genet 32, 676 Bradley (2005) J Intern Med 257, 18 Mizusawa (2006) Clin Endocrinol (Oxf) 65, 9
c.415C>T	Bricaire (2013) J Clin Endocrinol Metab 98, E403 Cetani (2007) Eur J Endocrinol 156, 547 Guarnieri (2012) Cell Oncol (Dordr) 35: 411 Khadilkar (2015) Endocr Pract 21, 1010
c.424-5T>C	Frank-Raue (2011) Eur J Endocrinol 165, 477
c.433_442delinsAGA	Masi (2008) Endocr Relat Cancer 15, 1115
c.481_484 delAAAG	Bricaire (2013) J Clin Endocrinol Metab 98, E403
c.505C>T	Ghemigian (2013) Hormones (Athens) 12, 454
c.520_523delTCTG	Guarnieri (2012) Cell Oncol (Dordr) 35: 411 Mizusawa (2006) Clin Endocrinol (Oxf) 65, 9
c.571delG	Kong (2013) Zhonghua Yi Xue Za Zhi 93, 3364
c.62_66delAGGGA	Mizusawa (2006) Clin Endocrinol (Oxf) 65, 9
c.626_629delAACA	Kong (2013) Zhonghua Yi Xue Za Zhi 93, 3364 Wang (2012) PLoS One 7, e45567

c.639delT	Carpten (2002) Nat Genet 32, 676
c.664C>T	Bricaire (2013) J Clin Endocrinol Metab 98, E403 Juhlin (2007) Endocr Relat Cancer 14, 501 Khadilkar (2015) Endocr Pract 21, 1010 Kong (2013) Zhonghua Yi Xue Za Zhi 93, 3364 Kong (2014) Clin Endocrinol (Oxf) 81, 222 Shattuck (2003) N Engl J Med 349, 1722 Wang (2012) PLoS One 7, e45567
c.668_669delATinsG	Bradley (2005) J Intern Med 257, 18
c.685_688delAGAG	Bricaire (2013) J Clin Endocrinol Metab 98, E403 Guarnieri (2006) J Clin Endocrinol Metab 91, 2827 Guarnieri (2012) Cell Oncol (Dordr) 35: 411 Kong (2013) Zhonghua Yi Xue Za Zhi 93, 3364 Sarquis (2008) Surgery 143, 630
c.687_688delAG	Bricaire (2013) J Clin Endocrinol Metab 98, E403 Guarnieri (2012) Cell Oncol (Dordr) 35: 411 Howell (2003) J Med Genet 40, 657 Kong (2013) Zhonghua Yi Xue Za Zhi 93, 3364 Sarquis (2008) Surgery 143, 630 Wang (2012) PLoS One 7, e45567
c.687_688dupAG	Carpten (2002) Nat Genet 32, 676 Bradley (2005) J Intern Med 257, 18 Shattuck (2003) N Engl J Med 349, 1722
c.692_693insT	Haven (2007) Clin Endocrinol (Oxf) 67, 370
c.700C>T	Bradley (2006) clin Endocrinol (oxf) 64, 299 Bricaire (2013) J Clin Endocrinol Metab 98, E403 Cetani (2007) Eur J Endocrinol 156, 547

	Frank-Raue (2011) Eur J Endocrinol 165, 477
c.745dup1	Bradley (2006) Clin Endocrinol (Oxf) 64, 299
c.766-767delGT	Cavaco (2004) J Clin Endocrinol Metab 89, 1747
c.76delA	Frank-Raue (2011) Eur J Endocrinol 165, 477 Howell (2003) J Med Genet 40, 657
c.815A>G	Juhlin (2006) Endocr Relat Cancer 13, 509
c.85delG	Rekik (2010) Ann Endocrinol (Paris) 71, 121
c.85G>T	Abdulla (2013) Endocr Pract 19: e134 Bricaire (2013) J Clin Endocrinol Metab 98, E403
c.96G>A	Kong (2013) Zhonghua Yi Xue Za Zhi 93, 3364 Kutcher (2013) Head Neck 35:E175 Sarquis (2008) Surgery 143, 630
deletion of exon 3	Bricaire (2013) J Clin Endocrinol Metab 98, E403
deletion of exons 1-10	Korpi-Hyövälti (2014) J Clin Endocrinol Metab 99, 3044
deletion of exons 2-3	Bricaire (2013) J Clin Endocrinol Metab 98, E403
deletion of exons 4-6	Bricaire (2013) J Clin Endocrinol Metab 98, E403 Kong (2014) Clin Endocrinol (Oxf) 81, 222
deletion of exons 7-13	Bricaire (2013) J Clin Endocrinol Metab 98, E403
deletion of the gene	Bricaire (2013) J Clin Endocrinol Metab 98, E403 Cascón (2011) Genes Chromosomes Cancer 50, 922 Domingues (2012) Clin Endocrinol (Oxf) 76, 33
g.95151T>C	Bradley (2006) Clin Endocrinol (Oxf) 64, 299