

CPAI **c.165G>C** **p.G55=** **p.G55G**

Citations:

Witt H, Beer S, Rosendahl J, Chen JM, Chandak GR, Masamune A, Bence M, Szmola R, Oracz G, Macek M Jr, Bhatia E, Steigenberger S, Lasher D, Bühler F, Delaporte C, Tebbing J, Ludwig M, Pilsak C, Saum K, Bugert P, Masson E, Paliwal S, Bhaskar S, Sobczynska-Tomaszewska A, Bak D, Balascak I, Choudhuri G, Nageshwar Reddy D, Rao GV, Thomas V, Kume K, Nakano E, Kakuta Y, Shimosegawa T, Durko L, Szabó A, Schnúr A, Hegyi P, Rakonczay Z Jr, Pfützer R, Schneider A, Groneberg DA, Braun M, Schmidt H, Witt U, Friess H, Algül H, Landt O, Schuelke M, Krüger R, Wiedenmann B, Schmidt F, Zimmer KP, Kovacs P, Stumvoll M, Blüher M, Müller T, Janecke A, Teich N, Grützmann R, Schulz HU, Mössner J, Keim V, Löhr M, Férec C, Sahin-Tóth M. (2013) **Variants in *CPAI* are strongly associated with early onset chronic pancreatitis.** Nat Genet 45, 2116-2120

2 affected

Dai LN, Chen YW, Yan WH, Lu LN, Tao YJ, Cai W. (2016) **Hereditary pancreatitis of 3 Chinese children: Case report and literature review.** Medicine (Baltimore) 95, e4604

1 affected (homozygous), 3 unaffected (1 homozygous)

Mutation not described; identified from electropherogram; described as homozygous but shown as heterozygous