

Entity	AAE	Type	Alteration (physical location)	dbSNP or rs	COSMIC Mutation ID	Previously reported	Number of FTC patients with the mutation (n)	Number of FTA patients with the mutation (n)	
<i>APC</i>	FTC	p.Thr1493 Thr	synonymous	chr5:112175 770 G>A	rs41115	-	Familial colorectal cancer Familial adenomatous polyposis 1 Hereditary cancer-predisposing syndrome APC-Associated Polyposis Disorders	4	1
<i>CSF1R</i>	FTC FTA	-	3 prime UTR variant	chr5:149433 596 T>G	rs206693 4	-	Hereditary diffuse leukoencephalopathy with spheroids	6	5
<i>ERBB4</i>	FTC FTA	-	intron variant	chr2:212812 097	rs839541	-	-	3	2
<i>FGFR3</i>	FTC FTA	p.Thr653T hr	synonymous	chr4:180789 4 G>A	rs768860 9	-	Craniosynostosis	7	7
<i>FLT3</i>	FTC FTA	c.1310- 3T>C	splice region	chr13:28610 183 A>G	rs249123 1	-	-	18	10
<i>FLT3</i>	FTC	-	intron variant	chr13:28602 292	rs755808 65	-	-	5	0

T>C									
<i>HRAS</i>	FTC FTA	p.His27His	synonymous	chr11:53424 2 A>G//T>C	rs12628	COSM249860	Costello syndrome Rasopathy	6	7
<i>KDR</i>	FTC FTA	-	intron variant	chr4:559802 39	rs769279 1	-	-	3	2
<i>PDGFR A</i>	FTC FTA	p.Val824Val	synonymous	chr4:551520 40 C>T	rs222823 0	COSM22413	Gastrointestinal stroma tumor; Idiopathic hypereosinophilic syndrome	3	2
<i>PDGFR A</i>	FTC FTA	p.Pro567Pro	synonymous	chr4:551410 55 A>G	rs187377 8	-	Gastrointestinal stroma tumor; Idiopathic hypereosinophilic syndrome	4	5
<i>RET</i>	FTC FTA	p.Leu769Leu	synonymous	chr10:43613 843 G>T	rs180086 1	-	Multiple endocrine neoplasia, type 2	11	10
<i>RET</i>	FTC FTA	p.Ser904Ser	synonymous	chr10:43615 633 C>G	rs180086 3	-	Pheochromocytoma; Renal adysplasia; Hereditary cancer- predisposing syndrome; Multiple endocrine neoplasia; Hirschsprung Disease, Dominant	5	3
<i>TP53</i>	FTC	p.Pro72Arg	missense	chr17:75794 72 G>C	rs104252 2	-	Li-Fraumeni syndrome; Hereditary cancer-predisposing syndrome	10	0