



Somatic Mutations from Whole Exome Sequencing Analysis of the Patients with Biliary Tract Cancer

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Biliary tract cancer (BTC) is a rare cancer and is associated with a poor prognosis. To understand the genetic characteristics of BTC, we analyzed whole-exome sequencing data and identified somatic mutations in patients with BTC. Tumors and matched blood or normal samples were obtained from seven patients with cholangiocarcinoma who underwent surgical resection. We discovered inactivating mutations of tumor suppressor genes, including *APC*, *TP53*, and *ARID1A*, in three patients. Activating mutations of *KRAS* and *NRAS* were also identified. Our analyses identified somatic mutations in Korean patients with BTC.

Keywords: biliary tract neoplasms, cholangiocarcinoma, somatic mutations, whole-exome sequencing

Introduction

Biliary tract cancer (BTC) is a heterogeneous group of cancers, including gallbladder cancer and extrahepatic and intrahepatic cholangiocarcinoma. BTC has been known as a disease with a poor prognosis, with few treatment options, regardless of its low incidence. In Korea, in 2014, the crude incidence rate of BTC was 11.2 per 100,000 men, and the crude mortality rate was 7.7 per 100,000 in both men and women [1]. Most patients have been diagnosed at an advanced stage. Although surgical resection is a curative treatment, recurrence of disease has been a clinical issue for patients after surgery [2]. Combination treatment with cisplatin plus gemcitabine has been reported as an appropriate treatment for patients with advanced BTC [3].

The genomic characteristics of BTC have been revealed through several studies using high-throughput next-generation sequencing technologies [4-6]. Jiao *et al.* [5] reported exome sequencing results of 32 patients with intrahepatic cholangiocarcinoma. Inactivating mutations in chromatin remodeling genes, including *BAP1*, *ARID1A*, and *PBRM1*, were identified with frequent mutations in *IDH1* and *IDH2*.

The oncogenes *KRAS*, *PIK3CA*, *NRAS*, *GNAS*, and *ERBB2* were also significantly mutated in BTC [6]. Genomic analyses need to be performed in more BTCs to understand their biology and develop therapeutic strategies.

Here, we reported the whole-exome sequencing (WES) data of seven patients with BTC and analyzed their somatic mutations to compare the genetic features between patients.

Methods

Patients and collection of clinical data

All patients had been enrolled in a phase II clinical trial at the National Cancer Center of Korea, as approved by the institutional review board (IRB No. NCCCTC-09-411). Enrolled patients had undergone surgical resection for BTC and were then treated with gemcitabine (1,000 mg/m²) as an adjuvant chemotherapy. The participants voluntarily agreed to donate the materials by signing an informed consent form. Tumor samples were obtained from surgical specimens and blood from seven patients with BTC.

Whole-exome sequencing and data analysis

We extracted genomic DNA from tissue specimens and

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blood samples using the QIAamp DNA mini kit according to the manufacturer's protocol (Qiagen, Valencia, CA, USA). The concentration and quality of DNA were assessed using a NanoDrop spectrophotometer (Thermo Fisher Scientific, Waltham, MA, USA). WES was performed using the SureSelect Human All Exon V5 Target Enrichment kit (Agilent Technologies, Santa Clara, CA, USA) and processed on the HiSeq 2500 platform (Illumina, San Diego, CA, USA). The size and quantity of the libraries were checked using a 2100 Bioanalyzer (Agilent Technologies).

Sequence reads were aligned to the human reference genome hg19 using the Burrows-Wheeler Aligner-MEM algorithm [7]. The alignments were refined using the Genome Analysis Tool Kit (Broad Institute), and duplicate or low-quality reads were excluded [8]. Somatic mutations were called using Mutect and Strelka with default settings by comparing the sequences of tumor samples with those of matched normal samples [9, 10]. Only mutations showing a total read depth of greater than 30 in normal samples were considered for further analysis.

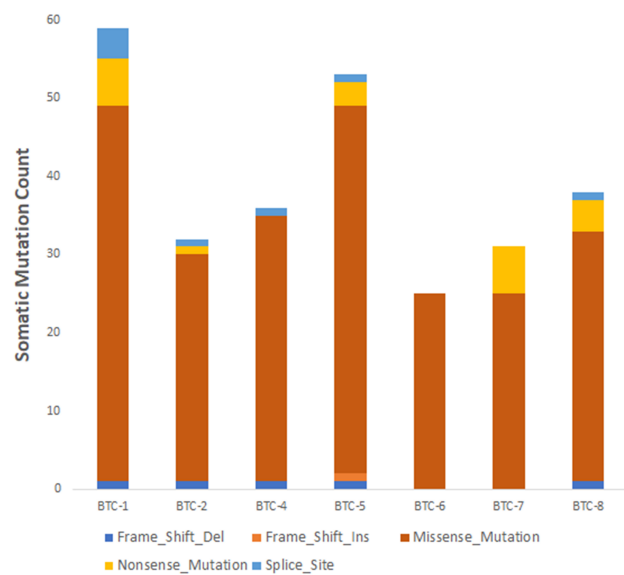


Fig. 1. Somatic mutation count for patients with biliary tract cancer.

Table 1. Demographic information for patients with BTC

Patient ID	Sex	Age (y)	Histologic type	RFS duration (mo)	OS duration (mo)	Total No. of somatic mutations
BTC-1	Male	57	Cholangiocarcinoma	58.2	58.6	59
BTC-2	Female	70	Cholangiocarcinoma	4.1	50.1	32
BTC-4	Male	51	Cholangiocarcinoma	1.9	6.1	36
BTC-5	Female	68	Cholangiocarcinoma	39.8	39.8	53
BTC-6	Female	69	Cholangiocarcinoma	18	38.5	25
BTC-7	Male	55	Cholangiocarcinoma	8.4	23.3	31
BTC-8	Male	55	Cholangiocarcinoma	8.9	15.8	38

BTC, biliary tract cancer; RFS, recurrence-free survival; OS, overall survival.

Table 2. Somatic mutations that were annotated with cancer gene census

Hugo_Symbol	Sample ID	Variant classification	cDNA change	Protein change
<i>APC</i>	BTC-1	Nonsense	c.4660G>T	p.E1554*
<i>APC</i>	BTC-8	Frameshift	c.4661delA	p.E1554fs
<i>ARID1A</i>	BTC-4	Frameshift	c.937delG	p.G314fs
<i>DNMT3A</i>	BTC-7	Missense	c.946A>G	p.I316V
<i>ELN</i>	BTC-4	Missense	c.593C>T	p.P198L
<i>HIP1</i>	BTC-4	Missense	c.1300C>T	p.R434W
<i>KDM5A</i>	BTC-6	Missense	c.2791G>A	p.G931R
<i>KIAA1549</i>	BTC-2	Missense	c.508C>T	p.R170W
<i>KRAS</i>	BTC-7	Missense	c.35G>T	p.G12V
<i>KRAS</i>	BTC-8	Missense	c.38G>A	p.G13D
<i>LPP</i>	BTC-7	Missense	c.457C>T	p.P153S
<i>NRAS</i>	BTC-5	Missense	c.182A>G	p.Q61R
<i>NTRK1</i>	BTC-7	Missense	c.2297G>A	p.R766Q
<i>RB1</i>	BTC-6	Missense	c.2402G>C	p.G801A
<i>RBM15</i>	BTC-1	Missense	c.1519T>G	p.W507G
<i>TP53</i>	BTC-8	Nonsense	c.1024C>T	p.R342*

Results and Discussion

The demographic and clinical characteristics of the patients with BTC, including age, sex, cancer stage, recurrence, and survival, are summarized in Table 1.

The patients included three females and four males with an average age of 60 years. Among the seven patients, five patients were defined as experiencing recurrence of disease after surgery.

We analyzed whole-exome sequencing data of matched tumor and normal DNA from the patients. The mean coverage depth for all samples is demonstrated in Supplementary Table 1. In total, 274 somatic mutations in protein-coding regions were called by both callers, Strelka and Mutect. The average number of somatic mutations per case was 34, showing a range of 25 to 59 (Fig. 1). We found two patients harboring activating mutations in *KRAS*, such as G12V and G13D (Table 2). Missense mutations in codon 61 of the *NRAS* gene and codon 766 of the *NTRK1* gene were also detected in two patients. A nonsense mutation in *TP53* was found in a patient, and *APC* was inactivated by frameshift mutations in two patients. A patient harbored a frameshift mutation that was caused by a deletion of G in the *ARID1A* gene. Detailed information on the somatic mutations is provided in Supplementary Table 2.

This study analyzed somatic mutations using WES in a limited number of patients with BTC. To understand the genetic features of BTC, more data need to be analyzed from more patients.

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Authors' contribution

Conceptualization: KAY, SMW, WJL
 Data curation: KAY, SMW
 Formal analysis: KAY, SMW, YHK, SYK, WJL
 Funding acquisition: SMW, WJL
 Investigation: KAY, SMW, SSH, SJP, WJL
 Methodology: KAY, SMW, YHK, SYK
 Project administration: KAY, SMW, WJL
 Software: KAY
 Supervision: KAY, SMW, SSH, SJP, WJL
 Validation: KAY, SMW, YHK, SYK, WJL
 Visualization: K.-A.Y., S.M.W.
 Writing – original draft: K.-A.Y., S.M.W

Writing – review and editing: KAY, SMW, YHK, SYK, SSH, SJP, WJL

Conflicts of Interest

No potential conflicts of interest relevant to this article was reported.

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Supplementary material

Supplementary data including two tables can be found with this article online at <https://doi.org/10.5808/GI.2018.16.4.e35>.

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SUPPLEMENTARY INFORMATION

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Supplementary Table 1. Coverage Information

Sample	X1	X5	X10	X15	X20	X25	X30	X50	Total bases aligned(bases)	Mean coverage depth
BTC_1N	99.96%	99.77%	99.22%	98.14%	96.33%	93.69%	90.19%	69.94%	6,034,974,927	80.99
BTC_1T	99.97%	99.90%	99.71%	99.38%	98.84%	98.03%	96.92%	89.02%	9,863,779,665	132.38
BTC_2N	99.86%	99.66%	99.15%	98.17%	96.48%	93.95%	90.45%	69.62%	5,978,637,815	80.24
BTC_2T	99.88%	99.79%	99.63%	99.38%	99.00%	98.46%	97.71%	92.10%	11,086,728,430	148.79
BTC_4N	99.97%	99.80%	99.30%	98.32%	96.67%	94.26%	91.07%	72.14%	6,345,622,329	85.16
BTC_4T	99.98%	99.90%	99.72%	99.43%	98.98%	98.33%	97.44%	91.14%	10,760,737,151	144.41
BTC_5N	99.86%	99.62%	98.95%	97.66%	95.53%	92.44%	88.40%	66.57%	5,823,253,443	78.15
BTC_5T	99.87%	99.78%	99.57%	99.21%	98.65%	97.88%	96.83%	89.50%	10,599,020,827	142.24
BTC_6N	99.87%	99.67%	99.16%	98.14%	96.43%	93.80%	90.22%	69.15%	5,968,127,744	80.09
BTC_6T	99.88%	99.79%	99.63%	99.37%	98.99%	98.43%	97.66%	91.94%	10,923,830,027	146.60
BTC_7N	99.97%	99.78%	99.25%	98.21%	96.48%	93.96%	90.63%	71.11%	6,186,543,991	83.03
BTC_7T	99.98%	99.88%	99.66%	99.27%	98.66%	97.75%	96.48%	87.79%	9,956,458,889	133.62
BTC_8N	99.97%	99.76%	99.17%	97.97%	95.96%	93.09%	89.35%	68.81%	5,998,051,912	80.50
BTC_8T	99.98%	99.89%	99.68%	99.34%	98.82%	98.06%	97.01%	89.49%	10,131,605,777	135.97

Supplementary Table 2. Detailed information on total somatic mutations

Patient ID	Hugo_Sym	Entrez_Gen	Chromoso	Start_position	End_position	Variant_Classification	Reference_Tumor_Seq	cDNA_Change	Codon_Ch.	Protein_Ch	Description
BTC-1	APC	324	5	112174759	112174760	Frame_Shift_Del	AG	-	c.3468_3469delAC	c.(3466-34 p.EE1156fs	adenomatous polyposis coli
BTC-1	KRTAP19-ε	337973	21	31914024	31914024	Nonsense_Mutation	G	T	c.129C>A	c.(127-129 p.C43*	keratin associated protein 19-6
BTC-1	PABPC4L	132430	4	135121319	135121319	Nonsense_Mutation	G	A	c.856C>T	c.(856-858 p.R286*	poly(A) binding protein, cytoplasmic 4-like
BTC-1	APC	324	5	112175951	112175951	Nonsense_Mutation	G	T	c.4660G>T	c.(4660-46 p.E1554*	adenomatous polyposis coli
BTC-1	C6orf222	389384	6	36290159	36290159	Nonsense_Mutation	G	C	c.1532C>G	c.(1531-15 p.S511*	chromosome 6 open reading frame 222
BTC-1	ABCA1	19	9	107553269	107553269	Nonsense_Mutation	G	C	c.5861C>G	c.(5860-58 p.S1954*	ATP-binding cassette, sub-family A (ABC1), member 1
BTC-1	AMER1	139285 X		63412530	63412530	Nonsense_Mutation	G	A	c.637C>T	c.(637-639 p.Q213*	APC membrane recruitment protein 1
BTC-1	MFAP2	4237	1	17302995	17302995	Splice_Site	C	A		c.e5+1	microfibrillar-associated protein 2
BTC-1	HIF3A	64344	19	46825222	46825222	Splice_Site	C	T	c.1334C>T	c.(1333-13 p.S445L	hypoxia inducible factor 3, alpha subunit
BTC-1	MAP3K13	9175	3	185155234	185155234	Splice_Site	G	C		c.e3-1	mitogen-activated protein kinase kinase kinase 13
BTC-1	DAB2IP	153090	9	124329507	124329507	Splice_Site	G	A	c.40G>A	c.(40-42)G; p.E14K	DAB2 interacting protein
BTC-1	ABCC2	1244	10	101591752	101591752	Missense_Mutation	C	T	c.3122C>T	c.(3121-31 p.A1041V	ATP-binding cassette, sub-family C (CFTR/MRP), member 2
BTC-1	ACE	1636	17	61570945	61570945	Missense_Mutation	G	A	c.1339G>A	c.(1339-13 p.V447M	angiotensin I converting enzyme
BTC-1	ACTN2	88	1	236899007	236899007	Missense_Mutation	C	T	c.770C>T	c.(769-771 p.A257V	actinin, alpha 2
BTC-1	ACTN4	81	19	39207832	39207832	Missense_Mutation	G	A	c.1019G>A	c.(1018-10 p.R340H	actinin, alpha 4
BTC-1	ACVR1B	91	12	52379016	52379016	Missense_Mutation	C	A	c.864C>A	c.(862-864 p.N288K	activin A receptor, type IB
BTC-1	ADCY5	111	3	123166420	123166420	Missense_Mutation	C	T	c.973G>A	c.(973-975 p.A325T	adenylate cyclase 5
BTC-1	ADCY8	114	8	131949376	131949376	Missense_Mutation	C	T	c.1424G>A	c.(1423-14 p.R475H	adenylate cyclase 8 (brain)
BTC-1	ANO3	63982	11	26463533	26463533	Missense_Mutation	T	C	c.115T>C	c.(115-117 p.C39R	anoctamin 3
BTC-1	APC2	10297	19	1457029	1457029	Missense_Mutation	G	A	c.994G>A	c.(994-996 p.A332T	adenomatosis polyposis coli 2
BTC-1	AQPEP	0	5	115298621	115298621	Missense_Mutation	G	A	c.307G>A	c.(307-309 p.V103M	
BTC-1	AUTS2	26053	7	69364311	69364311	Missense_Mutation	C	T	c.349C>T	c.(349-351 p.R117C	autism susceptibility candidate 2
BTC-1	C11orf63	79864	11	122775018	122775018	Missense_Mutation	C	T	c.730C>T	c.(730-732 p.R244C	chromosome 11 open reading frame 63
BTC-1	C5orf42	65250	5	37153858	37153858	Missense_Mutation	T	A	c.8195A>T	c.(8194-81 p.D2732V	chromosome 5 open reading frame 42
BTC-1	CDH2	1000	18	25572711	25572711	Missense_Mutation	C	T	c.1159G>A	c.(1159-11 p.A387T	cadherin 2, type 1, N-cadherin (neuronal)
BTC-1	CHST9	83539	18	24628462	24628462	Missense_Mutation	C	T	c.127G>A	c.(127-129 p.V43M	carbohydrate (N-acetylgalactosamine 4-0) sulfotransferase 9
BTC-1	CYP11B1	1584	8	143956558	143956558	Missense_Mutation	C	T	c.1426G>A	c.(1426-14 p.V476M	cytochrome P450, family 11, subfamily B, polypeptide 1
BTC-1	DNAH10	196385	12	124341718	124341718	Missense_Mutation	G	A	c.6200G>A	c.(6199-62 p.R2067H	dynein, axonemal, heavy chain 10
BTC-1	DSCAM	1826	21	41559851	41559851	Missense_Mutation	C	T	c.2617G>A	c.(2617-26 p.E873K	Down syndrome cell adhesion molecule
BTC-1	ELF3	1999	1	201983140	201983140	Missense_Mutation	G	A	c.989G>A	c.(988-990 p.S330N	E74-like factor 3 (ets domain transcription factor, epithelial-specific)
BTC-1	FGD3	89846	9	95738639	95738639	Missense_Mutation	C	T	c.101C>T	c.(100-102 p.A34V	FYVE, RhoGEF and PH domain containing 3
BTC-1	FUBP3	8939	9	133493210	133493210	Missense_Mutation	G	A	c.594G>A	c.(592-594 p.M198I	far upstream element (FUSE) binding protein 3
BTC-1	HAND1	9421	5	153855452	153855452	Missense_Mutation	G	T	c.562C>A	c.(562-564 p.P188T	heart and neural crest derivatives expressed 1
BTC-1	KCNS2	3788	8	99440299	99440299	Missense_Mutation	G	A	c.92G>A	c.(91-93)c(p.R31H	potassium voltage-gated channel, delayed-rectifier, subfamily S, member 2
BTC-1	KIAA1429	25962	8	95503865	95503865	Missense_Mutation	C	G	c.5081G>C	c.(5080-50 p.R1694T	KIAA1429
BTC-1	KRTAP19-1	337882	21	31852374	31852374	Missense_Mutation	C	T	c.263G>A	c.(262-264 p.G88D	keratin associated protein 19-1
BTC-1	LRP1B	53353	2	141004693	141004693	Missense_Mutation	G	T	c.13286C>A	c.(13285-1 p.P4429Q	low density lipoprotein receptor-related protein 1B
BTC-1	LRRK2	120892	12	40689336	40689336	Missense_Mutation	G	T	c.2986G>T	c.(2986-29 p.D996Y	leucine-rich repeat kinase 2
BTC-1	MAP2K1	5604	15	66727455	66727455	Missense_Mutation	G	C	c.171G>C	c.(169-171 p.K57N	mitogen-activated protein kinase kinase 1

BTC-1	MBTD1	54799	17	49296318	49296318	Missense_Mutation	A	C	c.376T>G	c.(376-378 p.L126V	mbt domain containing 1
BTC-1	MUC19	283463	12	40820354	40820354	Missense_Mutation	G	A	c.332G>A	c.(331-333 p.R111H	mucin 19, oligomeric
BTC-1	MYLK2	85366	20	30419645	30419645	Missense_Mutation	G	A	c.1564G>A	c.(1564-15 p.V522I	myosin light chain kinase 2
BTC-1	PCDH17	27253	13	58207860	58207860	Missense_Mutation	G	A	c.1180G>A	c.(1180-11 p.G394R	protocadherin 17
BTC-1	PLIN4	729359	19	4510530	4510530	Missense_Mutation	G	A	c.3400C>T	c.(3400-34 p.R1134C	perilipin 4
BTC-1	PRRC2B	84726	9	134322013	134322013	Missense_Mutation	A	G	c.839A>G	c.(838-840 p.D280G	proline-rich coiled-coil 2B
BTC-1	RBM15	64783	1	110883546	110883546	Missense_Mutation	T	G	c.1519T>G	c.(1519-15 p.W507G	RNA binding motif protein 15
BTC-1	RP1-139D8	0	6	42123292	42123292	Missense_Mutation	G	T	c.14G>T	c.(13-15)a(c.p.S5I	
BTC-1	SDK1	221935	7	4273010	4273010	Missense_Mutation	C	T	c.5891C>T	c.(5890-58 p.A1964V	sidekick cell adhesion molecule 1
BTC-1	SPRY2	10253	13	80911584	80911584	Missense_Mutation	G	C	c.257C>G	c.(256-258 p.P86R	sprouty homolog 2 (Drosophila)
BTC-1	ST6GALNA	81849	1	77509964	77509964	Missense_Mutation	C	A	c.337C>A	c.(337-339 p.Q113K	ST6 (alpha-N-acetyl-neuraminy-2,3-beta-galactosyl-1,3)-N-acetylgalactosaminide alpha-2,6-sialyltransferase 5
BTC-1	SYT3	84258	19	51135883	51135883	Missense_Mutation	C	T	c.334G>A	c.(334-336 p.G112R	synaptotagmin III
BTC-1	TADA2A	6871	17	35837062	35837062	Missense_Mutation	G	C	c.1307G>C	c.(1306-13 p.R436T	transcriptional adaptor 2A
BTC-1	TGM2	7052	20	36793594	36793594	Missense_Mutation	C	T	c.7G>A	c.(7-9)Gag p.E3K	transglutaminase 2
BTC-1	TMEM184I	25829	22	38643871	38643871	Missense_Mutation	T	C	c.97A>G	c.(97-99)A(c.p.T33A	transmembrane protein 184B
BTC-1	TRPC1	7220	3	142499703	142499703	Missense_Mutation	A	T	c.792A>T	c.(790-792 p.Q264H	transient receptor potential cation channel, subfamily C, member 1
BTC-1	TSHZ3	57616	19	31770589	31770589	Missense_Mutation	G	C	c.110C>G	c.(109-111 p.A37G	teashirt zinc finger homeobox 3
BTC-1	TSPAN14	81619	10	82271948	82271948	Missense_Mutation	T	G	c.499T>G	c.(499-501 p.Y167D	tetraspanin 14
BTC-1	ZFR	51663	5	32395347	32395347	Missense_Mutation	C	T	c.1897G>A	c.(1897-18 p.E633K	zinc finger RNA binding protein
BTC-1	ZFYVE26	23503	14	68264958	68264958	Missense_Mutation	C	A	c.2021G>T	c.(2020-20 p.G674V	zinc finger, FYVE domain containing 26
BTC-2	EPHA2	1969	1	16464447	16464447	Frame_Shift_Del	G	-	c.1213delC	c.(1213-12 p.H405fs	EPH receptor A2
BTC-2	LGI4	163175	19	35617610	35617610	Nonsense_Mutation	C	T	c.863G>A	c.(862-864 p.W288*	leucine-rich repeat LGI family, member 4
BTC-2	EPHA2	1969	1	16456720	16456720	Splice_Site	C	T		c.e15+1	EPH receptor A2
BTC-2	C17orf97	400566	17	260315	260315	Missense_Mutation	G	T	c.182G>T	c.(181-183 p.G61V	chromosome 17 open reading frame 97
BTC-2	CAMK2N1	55450	1	20810156	20810156	Missense_Mutation	G	A	c.223C>T	c.(223-225 p.P75S	calcium/calmodulin-dependent protein kinase II inhibitor 1
BTC-2	CNBD1	168975	8	88249206	88249206	Missense_Mutation	G	A	c.637G>A	c.(637-639 p.V213M	cyclic nucleotide binding domain containing 1
BTC-2	COL14A1	7373	8	121292323	121292323	Missense_Mutation	G	A	c.3631G>A	c.(3631-36 p.E1211K	collagen, type XIV, alpha 1
BTC-2	FBN3	84467	19	8201283	8201283	Missense_Mutation	C	T	c.1334G>A	c.(1333-13 p.G445D	fibrillin 3
BTC-2	FIGN	55137	2	164467285	164467285	Missense_Mutation	C	A	c.1057G>T	c.(1057-10 p.D353Y	fidgetin
BTC-2	FLNB	2317	3	58118580	58118580	Missense_Mutation	A	G	c.4529A>G	c.(4528-45 p.H1510R	filamin B, beta
BTC-2	INO80D	54891	2	206884534	206884534	Missense_Mutation	G	A	c.1334C>T	c.(1333-13 p.A445V	INO80 complex subunit D
BTC-2	INTS1	26173	7	1544069	1544069	Missense_Mutation	C	G	c.233G>C	c.(232-234 p.C78S	integrator complex subunit 1
BTC-2	KCNIP1	30820	5	170145809	170145809	Missense_Mutation	G	A	c.109G>A	c.(109-111 p.E37K	Kv channel interacting protein 1
BTC-2	KIAA1549	57670	7	138603864	138603864	Missense_Mutation	G	A	c.508C>T	c.(508-510 p.R170W	KIAA1549
BTC-2	LGALS9	3965	17	25975970	25975970	Missense_Mutation	G	A	c.934G>A	c.(934-936 p.V312M	lectin, galactoside-binding, soluble, 9
BTC-2	PLEKHG1	57480	6	151152904	151152904	Missense_Mutation	G	A	c.2657G>A	c.(2656-26 p.R886H	pleckstrin homology domain containing, family G (with RhoGef domain) member 1
BTC-2	PNCK	139728 X		152937421	152937421	Missense_Mutation	C	G	c.328G>C	c.(328-330 p.E110Q	pregnancy up-regulated nonubiquitous CaM kinase
BTC-2	PPDPF	79144	20	62152924	62152924	Missense_Mutation	G	A	c.115G>A	c.(115-117 p.A39T	pancreatic progenitor cell differentiation and proliferation factor
BTC-2	PROKR2	128674	20	5282783	5282783	Missense_Mutation	C	T	c.1058G>A	c.(1057-10 p.R353H	prokineticin receptor 2

BTC-2	RP11-429E	0	20	60294088	60294088	Missense_Mutation	G	A	c.139C>T	c.(139-141 p.L47F	
BTC-2	SCN5A	6331	3	38645514	38645514	Missense_Mutation	C	T	c.1579G>A	c.(1579-15 p.G527R	sodium channel, voltage-gated, type V, alpha subunit
BTC-2	SLC35F1	222553	6	118556702	118556702	Missense_Mutation	G	A	c.380G>A	c.(379-381 p.R127Q	solute carrier family 35, member F1
BTC-2	SLC9A6	10479 X		135067950	135067950	Missense_Mutation	C	T	c.289C>T	c.(289-291 p.R97C	solute carrier family 9, subfamily A (NHE6, cation proton antiporter 6), member 6
BTC-2	SPARC	6678	5	151047056	151047056	Missense_Mutation	T	C	c.557A>G	c.(556-558 p.N186S	secreted protein, acidic, cysteine-rich (osteonectin)
BTC-2	TLL2	7093	10	98144434	98144434	Missense_Mutation	C	G	c.2104G>C	c.(2104-21 p.V702L	tolloid-like 2
BTC-2	TRIM58	25893	1	248028097	248028097	Missense_Mutation	C	T	c.607C>T	c.(607-609 p.R203W	tripartite motif containing 58
BTC-2	TSPAN16	26526	19	11408947	11408947	Missense_Mutation	A	G	c.199A>G	c.(199-201 p.I67V	tetraspanin 16
BTC-2	TSSK2	23617	22	19119034	19119034	Missense_Mutation	A	C	c.122A>C	c.(121-123 p.K41T	testis-specific serine kinase 2
BTC-2	VAV2	7410	9	136635595	136635595	Missense_Mutation	G	A	c.2222C>T	c.(2221-22 p.S741L	vav 2 guanine nucleotide exchange factor
BTC-2	ZC2HC1B	153918	6	144224199	144224199	Missense_Mutation	C	T	c.508C>T	c.(508-510 p.P170S	zinc finger, C2HC-type containing 1B
BTC-2	ZFP69B	65243	1	40928270	40928270	Missense_Mutation	G	A	c.614G>A	c.(613-615 p.G205D	ZFP69 zinc finger protein B
BTC-2	ZNF423	23090	16	49669726	49669726	Missense_Mutation	C	T	c.3337G>A	c.(3337-33 p.A1113T	zinc finger protein 423
BTC-4	ARID1A	8289	1	27023831	27023831	Frame_Shift_Del	G	-	c.937delG	c.(937-939 p.G314fs	AT rich interactive domain 1A (SWI-like)
BTC-4	TULP1	7287	6	35480415	35480415	Splice_Site	C	G		c.e2+1	tubby like protein 1
BTC-4	ACOXL	55289	2	111850501	111850501	Missense_Mutation	T	A	c.1590T>A	c.(1588-15 p.H530Q	acyl-CoA oxidase-like
BTC-4	CACNA1H	8912	16	1270626	1270626	Missense_Mutation	C	A	c.6676C>A	c.(6676-66 p.L2226M	calcium channel, voltage-dependent, T type, alpha 1H subunit
BTC-4	CATSPERB	79820	14	92174496	92174496	Missense_Mutation	T	A	c.455A>T	c.(454-456 p.D152V	catsper channel auxiliary subunit beta
BTC-4	CDKN2A	1029	9	21971035	21971035	Missense_Mutation	T	C	c.170A>G	c.(169-171 p.D57G	cyclin-dependent kinase inhibitor 2A
BTC-4	CES3	23491	16	67006620	67006620	Missense_Mutation	G	A	c.408G>A	c.(406-408 p.M136I	carboxylesterase 3
BTC-4	CLCNKA	1187	1	16349137	16349137	Missense_Mutation	G	A	c.23G>A	c.(22-24)c(p.R8H	chloride channel, voltage-sensitive Ka
BTC-4	COL22A1	169044	8	139890232	139890232	Missense_Mutation	C	T	c.419G>A	c.(418-420 p.R140H	collagen, type XXII, alpha 1
BTC-4	CRISPLD1	83690	8	75941665	75941665	Missense_Mutation	G	A	c.1364G>A	c.(1363-13 p.R455Q	cysteine-rich secretory protein LCCL domain containing 1
BTC-4	DAW1	164781	2	228767727	228767727	Missense_Mutation	T	C	c.550T>C	c.(550-552 p.S184P	dynein assembly factor with WDR repeat domains 1
BTC-4	DYSF	8291	2	71797023	71797023	Missense_Mutation	G	A	c.2938G>A	c.(2938-29 p.G980R	dysferlin
BTC-4	ELN	2006	7	73461047	73461047	Missense_Mutation	C	T	c.593C>T	c.(592-594 p.P198L	elastin
BTC-4	FAM135B	51059	8	139165339	139165339	Missense_Mutation	A	G	c.1379T>C	c.(1378-13 p.L460P	family with sequence similarity 135, member B
BTC-4	FAM208B	54906	10	5803432	5803432	Missense_Mutation	C	A	c.7172C>A	c.(7171-71 p.T2391K	family with sequence similarity 208, member B
BTC-4	GPR65	8477	14	88478153	88478153	Missense_Mutation	G	A	c.962G>A	c.(961-963 p.R321H	G protein-coupled receptor 65
BTC-4	GUCY1A3	2982	4	156643206	156643206	Missense_Mutation	A	T	c.1733A>T	c.(1732-17 p.H578L	guanylate cyclase 1, soluble, alpha 3
BTC-4	HID1	283987	17	72959068	72959068	Missense_Mutation	T	A	c.496A>T	c.(496-498 p.S166C	HID1 domain containing
BTC-4	HIP1	3092	7	75189111	75189111	Missense_Mutation	G	A	c.1300C>T	c.(1300-13 p.R434W	huntingtin interacting protein 1
BTC-4	IRF3	3661	19	50167939	50167939	Missense_Mutation	T	C	c.157A>G	c.(157-159 p.I53V	interferon regulatory factor 3
BTC-4	KCNB1	3745	20	48098906	48098906	Missense_Mutation	C	T	c.112G>A	c.(112-114 p.G38R	potassium voltage-gated channel, Shab-related subfamily, member 1
BTC-4	KCNT1	57582	9	138664603	138664603	Missense_Mutation	C	T	c.2051C>T	c.(2050-20 p.T684M	potassium channel, subfamily T, member 1
BTC-4	LLGL1	3996	17	18143990	18143990	Missense_Mutation	C	T	c.2305C>T	c.(2305-23 p.R769W	lethal giant larvae homolog 1 (Drosophila)
BTC-4	LPHN1	22859	19	14294398	14294398	Missense_Mutation	G	A	c.17C>T	c.(16-18)g(p.A6V	latrophilin 1
BTC-4	MARS	4141	12	57906626	57906626	Missense_Mutation	A	T	c.1846A>T	c.(1846-18 p.I616F	methionyl-tRNA synthetase
BTC-4	MBTPS1	8720	16	84093019	84093019	Missense_Mutation	G	A	c.2719C>T	c.(2719-27 p.R907W	membrane-bound transcription factor peptidase, site 1
BTC-4	PDC	5132	1	186413572	186413572	Missense_Mutation	G	A	c.280C>T	c.(280-282 p.R94C	phosducin
BTC-4	PTTG1IP	754	21	46285352	46285352	Missense_Mutation	C	G	c.126G>C	c.(124-126 p.Q42H	pituitary tumor-transforming 1 interacting protein

BTC-4	RBBP6	5930	16	24581304	24581304	Missense_Mutation	A	T	c.3293A>T	c.(3292-32 p.E1098V	retinoblastoma binding protein 6
BTC-4	RBBP7	5931 X		16871823	16871823	Missense_Mutation	T	A	c.872A>T	c.(871-873 p.D291V	retinoblastoma binding protein 7
BTC-4	RDH10	157506	8	74234976	74234976	Missense_Mutation	A	G	c.833A>G	c.(832-834 p.K278R	retinol dehydrogenase 10 (all-trans)
BTC-4	RFX8	731220	2	102018971	102018971	Missense_Mutation	C	T	c.1172G>A	c.(1171-11 p.R391Q	RFX family member 8, lacking RFX DNA binding domain
BTC-4	SEMA6D	80031	15	48058143	48058143	Missense_Mutation	C	T	c.1505C>T	c.(1504-15 p.A502V	sema domain, transmembrane domain (TM), and cytoplasmic domain, (semaphorin) 6D
BTC-4	SLC6A17	388662	1	110734682	110734682	Missense_Mutation	C	A	c.953C>A	c.(952-954 p.A318D	solute carrier family 6 (neutral amino acid transporter), member 17
BTC-4	TMEM205	374882	19	11455971	11455971	Missense_Mutation	G	A	c.221C>T	c.(220-222 p.S74L	transmembrane protein 205
BTC-4	TRIM4	89122	7	99506284	99506284	Missense_Mutation	T	C	c.719A>G	c.(718-720 p.N240S	tripartite motif containing 4
BTC-5	HMCN1	83872	1	186057081	186057084	Frame_Shift_Del	GTAT	-	c.9381_9384delGT	c.(9379-93 p.QY3127f	hemicentin 1
BTC-5	UPP2	151531	2	158977957	158977958	Frame_Shift_Ins	-	T	c.491_492insT	c.(490-495 p.V165fs	uridine phosphorylase 2
BTC-5	FNTB	2342	14	65511053	65511053	Nonsense_Mutation	C	T	c.847C>T	c.(847-849 p.R283*	farnesyltransferase, CAAX box, beta
BTC-5	ACSM2A	123876	16	20482871	20482871	Nonsense_Mutation	C	T	c.754C>T	c.(754-756 p.Q252*	acyl-CoA synthetase medium-chain family member 2A
BTC-5	PSD3	23362	8	18430175	18430175	Nonsense_Mutation	C	A	c.2647G>T	c.(2647-26 p.E883*	pleckstrin and Sec7 domain containing 3
BTC-5	COL11A1	1301	1	103467527	103467527	Splice_Site	T	C		c.e24-2	collagen, type XI, alpha 1
BTC-5	AC007405.	0	2	171570595	171570595	Missense_Mutation	G	C	c.359C>G	c.(358-360 p.A120G	
BTC-5	AC009365.	0	7	132412557	132412557	Missense_Mutation	G	A	c.409G>A	c.(409-411 p.V137I	
BTC-5	ADAM21	8747	14	70924614	70924614	Missense_Mutation	G	T	c.398G>T	c.(397-399 p.R133L	ADAM metallopeptidase domain 21
BTC-5	APOOL	139322 X		84329077	84329077	Missense_Mutation	G	C	c.560G>C	c.(559-561 p.S187T	apolipoprotein O-like
BTC-5	ASPM	259266	1	197074062	197074062	Missense_Mutation	T	C	c.4319A>G	c.(4318-43 p.Q1440R	asp (abnormal spindle) homolog, microcephaly associated (Drosophila)
BTC-5	ATP7A	538 X		77301051	77301051	Missense_Mutation	C	A	c.4208C>A	c.(4207-42 p.S1403Y	ATPase, Cu++ transporting, alpha polypeptide
BTC-5	ATP8B4	79895	15	50288883	50288883	Missense_Mutation	C	G	c.580G>C	c.(580-582 p.G194R	ATPase, class I, type 8B, member 4
BTC-5	CACNA1A	773	19	13616812	13616812	Missense_Mutation	C	A	c.227G>T	c.(226-228 p.R76L	calcium channel, voltage-dependent, P/Q type, alpha 1A subunit
BTC-5	CCDC175	729665	14	60031860	60031860	Missense_Mutation	T	C	c.625A>G	c.(625-627 p.I209V	coiled-coil domain containing 175
BTC-5	CCL18	6362	17	34398340	34398340	Missense_Mutation	G	A	c.209G>A	c.(208-210 p.C70Y	chemokine (C-C motif) ligand 18 (pulmonary and activation-regulated)
BTC-5	CD163	9332	12	7651580	7651580	Missense_Mutation	A	G	c.662T>C	c.(661-663 p.I221T	CD163 molecule
BTC-5	COL21A1	81578	6	55922544	55922544	Missense_Mutation	G	T	c.2785C>A	c.(2785-27 p.Q929K	collagen, type XXI, alpha 1
BTC-5	CSMD3	114788	8	113697935	113697935	Missense_Mutation	G	C	c.2182C>G	c.(2182-21 p.P728A	CUB and Sushi multiple domains 3
BTC-5	CXorf28	1E+08 X		3189922	3189922	Missense_Mutation	C	T	c.49C>T	c.(49-51)C(p.P17S	chromosome X open reading frame 28
BTC-5	DMXL2	23312	15	51791693	51791693	Missense_Mutation	T	A	c.3728A>T	c.(3727-37 p.D1243V	Dmx-like 2
BTC-5	GAS2	2620	11	22747946	22747946	Missense_Mutation	G	A	c.376G>A	c.(376-378 p.D126N	growth arrest-specific 2
BTC-5	GPR98	84059	5	89939702	89939702	Missense_Mutation	C	T	c.2636C>T	c.(2635-26 p.T879M	G protein-coupled receptor 98
BTC-5	HOXA4	3201	7	27170310	27170310	Missense_Mutation	G	T	c.43C>A	c.(43-45)C(p.P15T	homeobox A4
BTC-5	HP	3240	16	72090131	72090131	Missense_Mutation	C	T	c.77C>T	c.(76-78)a(p.T26M	haptoglobin
BTC-5	ICAM5	7087	19	10405259	10405259	Missense_Mutation	T	C	c.2173T>C	c.(2173-21 p.C725R	intercellular adhesion molecule 5, telencephalin
BTC-5	KIAA1244	57221	6	138576619	138576619	Missense_Mutation	C	T	c.817C>T	c.(817-819 p.H273Y	KIAA1244
BTC-5	KIF21B	23046	1	200948693	200948693	Missense_Mutation	C	T	c.4130G>A	c.(4129-41 p.R1377Q	kinesin family member 21B
BTC-5	KRTAP21-1	337977	21	32127554	32127554	Missense_Mutation	C	A	c.143G>T	c.(142-144 p.C48F	keratin associated protein 21-1
BTC-5	LCA10	0 X		153152431	153152431	Missense_Mutation	G	T	c.311G>T	c.(310-312 p.R104L	

BTC-5	MAP1B	4131	5	71490888	71490888	Missense_Mutation	C	G	c.1706C>G	c.(1705-17 p.T569R	microtubule-associated protein 1B
BTC-5	MLK4	0	1	233518335	233518335	Missense_Mutation	C	T	c.2989C>T	c.(2989-29 p.P997S	
BTC-5	NBEAL2	23218	3	47041430	47041430	Missense_Mutation	C	A	c.3841C>A	c.(3841-38 p.Q1281K	neurobeachin-like 2
BTC-5	NFATC2	4773	20	50140482	50140482	Missense_Mutation	C	T	c.298G>A	c.(298-300 p.A100T	nuclear factor of activated T-cells, cytoplasmic, calcineurin-dependent 2
BTC-5	NRAS	4893	1	115256529	115256529	Missense_Mutation	T	C	c.182A>G	c.(181-183 p.Q61R	neuroblastoma RAS viral (v-ras) oncogene homolog
BTC-5	OLFML2B	25903	1	161967939	161967939	Missense_Mutation	T	A	c.1153A>T	c.(1153-11 p.S385C	olfactomedin-like 2B
BTC-5	OSBPL5	114879	11	3143583	3143583	Missense_Mutation	G	A	c.295C>T	c.(295-297 p.L99F	oxysterol binding protein-like 5
BTC-5	PCDHB1	29930	5	140432385	140432385	Missense_Mutation	G	A	c.1330G>A	c.(1330-13 p.D444N	protocadherin beta 1
BTC-5	PCSK5	5125	9	78936474	78936474	Missense_Mutation	G	A	c.3940G>A	c.(3940-39 p.E1314K	proprotein convertase subtilisin/kexin type 5
BTC-5	PGR	5241	11	100998706	100998706	Missense_Mutation	C	T	c.1096G>A	c.(1096-10 p.D366N	progesterone receptor
BTC-5	PHIP	55023	6	79695085	79695085	Missense_Mutation	T	G	c.2521A>C	c.(2521-25 p.S841R	pleckstrin homology domain interacting protein
BTC-5	PIK3C2G	5288	12	18435656	18435656	Missense_Mutation	C	A	c.641C>A	c.(640-642 p.P214H	phosphatidylinositol-4-phosphate 3-kinase, catalytic subunit type 2 gamma
BTC-5	PRKCD	5580	3	53219710	53219710	Missense_Mutation	A	T	c.979A>T	c.(979-981 p.M327L	protein kinase C, delta
BTC-5	SEC14L2	23541	22	30802345	30802345	Missense_Mutation	C	G	c.145C>G	c.(145-147 p.L49V	SEC14-like 2 (S. cerevisiae)
BTC-5	SEMA6A	57556	5	115782872	115782872	Missense_Mutation	C	T	c.2530G>A	c.(2530-25 p.E844K	sema domain, transmembrane domain (TM), and cytoplasmic domain, (semaphorin) 6A
BTC-5	SKIV2L2	23517	5	54635888	54635888	Missense_Mutation	G	T	c.566G>T	c.(565-567 p.S189I	superkiller viralicidic activity 2-like 2 (S. cerevisiae)
BTC-5	TAF1L	138474	9	32631848	32631848	Missense_Mutation	G	A	c.3730C>T	c.(3730-37 p.R1244W	TAF1 RNA polymerase II, TATA box binding protein (TBP)-associated factor, 210kDa-like
BTC-5	TEX15	56154	8	30702546	30702546	Missense_Mutation	T	G	c.3988A>C	c.(3988-39 p.K1330Q	testis expressed 15
BTC-5	TLR8	51311 X		12939230	12939230	Missense_Mutation	G	C	c.2125G>C	c.(2125-21 p.E709Q	toll-like receptor 8
BTC-5	TMEM200A	114801	6	130762039	130762039	Missense_Mutation	A	G	c.472A>G	c.(472-474 p.I158V	transmembrane protein 200A
BTC-5	TMEM80	283232	11	703112	703112	Missense_Mutation	G	T	c.469G>T	c.(469-471 p.V157F	transmembrane protein 80
BTC-5	TRPM6	140803	9	77377993	77377993	Missense_Mutation	C	A	c.3594G>T	c.(3592-35 p.K1198N	transient receptor potential cation channel, subfamily M, member 6
BTC-5	TSPYL5	85453	8	98290039	98290039	Missense_Mutation	G	A	c.34C>T	c.(34-36)C ₁ p.R12C	TSPYL-like 5
BTC-6	AMIGO2	347902	12	47471598	47471598	Missense_Mutation	A	C	c.1188T>G	c.(1186-11 p.F396L	adhesion molecule with Ig-like domain 2
BTC-6	BIRC6	57448	2	32712788	32712788	Missense_Mutation	A	T	c.7888A>T	c.(7888-78 p.I2630F	baculoviral IAP repeat containing 6
BTC-6	BZW1	9689	2	201680210	201680210	Missense_Mutation	T	G	c.211T>G	c.(211-213 p.F71V	basic leucine zipper and W2 domains 1
BTC-6	CD200R1	131450	3	112648303	112648303	Missense_Mutation	G	A	c.185C>T	c.(184-186 p.P62L	CD200 receptor 1
BTC-6	CRYBG3	131544	3	97593426	97593426	Missense_Mutation	G	T	c.3388G>T	c.(3388-33 p.G1130W	beta-gamma crystallin domain containing 3
BTC-6	DEAF1	10522	11	694992	694992	Missense_Mutation	A	G	c.56T>C	c.(55-57)g ⁺ p.V19A	DEAF1 transcription factor
BTC-6	EFCAB5	374786	17	28295933	28295933	Missense_Mutation	A	T	c.315A>T	c.(313-315 p.E105D	EF-hand calcium binding domain 5
BTC-6	FSIP2	401024	2	186671383	186671383	Missense_Mutation	G	A	c.17350G>A	c.(17350-1 p.A5784T	fibrous sheath interacting protein 2
BTC-6	GAPVD1	26130	9	128064438	128064438	Missense_Mutation	A	T	c.362A>T	c.(361-363 p.N121I	GTPase activating protein and VPS9 domains 1
BTC-6	IQSEC2	23096 X		53349985	53349985	Missense_Mutation	C	T	c.337G>A	c.(337-339 p.D113N	IQ motif and Sec7 domain 2
BTC-6	KDM5A	5927	12	427378	427378	Missense_Mutation	C	T	c.2791G>A	c.(2791-27 p.G931R	lysine (K)-specific demethylase 5A
BTC-6	LPAR3	23566	1	85279695	85279695	Missense_Mutation	T	C	c.896A>G	c.(895-897 p.D299G	lysophosphatidic acid receptor 3
BTC-6	NLRP1	22861	17	5462767	5462767	Missense_Mutation	A	G	c.1249T>C	c.(1249-12 p.W417R	NLR family, pyrin domain containing 1
BTC-6	OR6B3	150681	2	240984973	240984973	Missense_Mutation	C	A	c.517G>T	c.(517-519 p.V173F	olfactory receptor, family 6, subfamily B, member 3
BTC-6	OTUD6A	139562 X		69282627	69282627	Missense_Mutation	G	C	c.253G>C	c.(253-255 p.E85Q	OTU deubiquitinase 6A

BTC-6	PTPRH	5794	19	55715264	55715264	Missense_Mutation	T	C	c.772A>G	c.(772-774 p.T258A	protein tyrosine phosphatase, receptor type, H
BTC-6	RAD50	10111	5	131927660	131927660	Missense_Mutation	T	G	c.1727T>G	c.(1726-17 p.L576R	RAD50 homolog (S. cerevisiae)
BTC-6	RB1	5925	13	49039417	49039417	Missense_Mutation	G	C	c.2402G>C	c.(2401-24 p.G801A	retinoblastoma 1
BTC-6	SAMD9L	219285	7	92763376	92763376	Missense_Mutation	C	T	c.1909G>A	c.(1909-19 p.A637T	sterile alpha motif domain containing 9-like
BTC-6	SERINC5	256987	5	79454696	79454696	Missense_Mutation	C	G	c.949G>C	c.(949-951 p.G317R	serine incorporator 5
BTC-6	ST6GALNA	81849	1	77515950	77515950	Missense_Mutation	T	A	c.679T>A	c.(679-681 p.S227T	ST6 (alpha-N-acetyl-neuraminyl-2,3-beta-galactosyl-1,3)-N-acetylgalactosaminide alpha-2,6-sialyltransferase 5
BTC-6	TMEM47	83604 X		34657486	34657486	Missense_Mutation	A	G	c.245T>C	c.(244-246 p.L82P	transmembrane protein 47
BTC-6	TRIM41	90933	5	180651595	180651595	Missense_Mutation	T	C	c.596T>C	c.(595-597 p.L199P	tripartite motif containing 41
BTC-6	WDSUB1	151525	2	160139525	160139525	Missense_Mutation	G	A	c.56C>T	c.(55-57)g(p.A19V	WD repeat, sterile alpha motif and U-box domain containing 1
BTC-6	ZC3H13	23091	13	46616352	46616352	Missense_Mutation	C	T	c.286G>A	c.(286-288 p.V96M	zinc finger CCCH-type containing 13
BTC-7	CCDC18	343099	1	93705341	93705341	Nonsense_Mutation	G	T	c.2866G>T	c.(2866-28 p.E956*	coiled-coil domain containing 18
BTC-7	KANSL1	284058	17	44115975	44115975	Nonsense_Mutation	G	A	c.2281C>T	c.(2281-22 p.R761*	KAT8 regulatory NSL complex subunit 1
BTC-7	PLEKHG2	64857	19	39912773	39912773	Nonsense_Mutation	G	T	c.1522G>T	c.(1522-15 p.E508*	pleckstrin homology domain containing, family G (with RhoGef domain) member 2
BTC-7	ZNF546	339327	19	40520465	40520465	Nonsense_Mutation	C	T	c.1288C>T	c.(1288-12 p.R430*	zinc finger protein 546
BTC-7	SI	6476	3	164760855	164760855	Nonsense_Mutation	C	A	c.1996G>T	c.(1996-19 p.G666*	sucrase-isomaltase (alpha-glucosidase)
BTC-7	SLITRK2	84631 X		144904042	144904042	Nonsense_Mutation	T	A	c.99T>A	c.(97-99)tg p.C33*	SLIT and NTRK-like family, member 2
BTC-7	CNTNAP5	129684	2	125367471	125367471	Missense_Mutation	C	T	c.1847C>T	c.(1846-18 p.P616L	contactin associated protein-like 5
BTC-7	DNMT3A	1788	2	25467461	25467461	Missense_Mutation	T	C	c.946A>G	c.(946-948 p.I316V	DNA (cytosine-5-)-methyltransferase 3 alpha
BTC-7	DSG4	147409	18	28971077	28971077	Missense_Mutation	G	A	c.721G>A	c.(721-723 p.D241N	desmoglein 4
BTC-7	EPC1	80314	10	32560813	32560813	Missense_Mutation	T	C	c.2038A>G	c.(2038-20 p.T680A	enhancer of polycomb homolog 1 (Drosophila)
BTC-7	ESM1	11082	5	54277867	54277867	Missense_Mutation	A	T	c.409T>A	c.(409-411 p.Y137N	endothelial cell-specific molecule 1
BTC-7	F13A1	2162	6	6266956	6266956	Missense_Mutation	C	A	c.406G>T	c.(406-408 p.V136F	coagulation factor XIII, A1 polypeptide
BTC-7	FCRL3	115352	1	157665154	157665154	Missense_Mutation	G	A	c.1376C>T	c.(1375-13 p.A459V	Fc receptor-like 3
BTC-7	FUT1	2523	19	49254093	49254093	Missense_Mutation	C	A	c.446G>T	c.(445-447 p.W149L	fucosyltransferase 1 (galactoside 2-alpha-L-fucosyltransferase, H blood group)
BTC-7	GLI3	2737	7	42005951	42005951	Missense_Mutation	C	A	c.2720G>T	c.(2719-27 p.S907I	GLI family zinc finger 3
BTC-7	HEATR1	55127	1	236721708	236721708	Missense_Mutation	C	T	c.5033G>A	c.(5032-50 p.C1678Y	HEAT repeat containing 1
BTC-7	KLHL24	54800	3	183368493	183368493	Missense_Mutation	A	G	c.349A>G	c.(349-351 p.M117V	kelch-like family member 24
BTC-7	KLHL9	55958	9	21333179	21333179	Missense_Mutation	C	G	c.1680G>C	c.(1678-16 p.M560I	kelch-like family member 9
BTC-7	KMT2A	4297	11	118392628	118392628	Missense_Mutation	T	A	c.11660T>A	c.(11659-1 p.M3887K	lysine (K)-specific methyltransferase 2A
BTC-7	KRAS	3845	12	25398284	25398284	Missense_Mutation	C	A	c.35G>T	c.(34-36)g(p.G12V	Kirsten rat sarcoma viral oncogene homolog
BTC-7	LPP	4026	3	188326976	188326976	Missense_Mutation	C	T	c.457C>T	c.(457-459 p.P153S	LIM domain containing preferred translocation partner in lipoma
BTC-7	LRP1B	53353	2	141707920	141707920	Missense_Mutation	A	G	c.3020T>C	c.(3019-30 p.F1007S	low density lipoprotein receptor-related protein 1B
BTC-7	MSH3	4437	5	79950739	79950739	Missense_Mutation	G	A	c.193G>A	c.(193-195 p.A65T	mutS homolog 3
BTC-7	NCOR1	9611	17	16049747	16049747	Missense_Mutation	T	C	c.1025A>G	c.(1024-10 p.Y342C	nuclear receptor corepressor 1
BTC-7	NIPBL	25836	5	37064917	37064917	Missense_Mutation	A	G	c.8338A>G	c.(8338-83 p.S2780G	Nipped-B homolog (Drosophila)
BTC-7	NTRK1	4914	1	156851340	156851340	Missense_Mutation	G	A	c.2297G>A	c.(2296-22 p.R766Q	neurotrophic tyrosine kinase, receptor, type 1
BTC-7	PELP1	27043	17	4575146	4575146	Missense_Mutation	C	T	c.3140G>A	c.(3139-31 p.G1047E	proline, glutamate and leucine rich protein 1
BTC-7	RELN	5649	7	103183183	103183183	Missense_Mutation	A	T	c.6666T>A	c.(6664-66 p.H2222Q	reelin

BTC-7	SPTBN1	6711	2	54871578	54871578	Missense_Mutation	G	A	c.4124G>A	c.(4123-41 p.R1375Q	spectrin, beta, non-erythrocytic 1
BTC-7	SUSD2	56241	22	24583685	24583685	Missense_Mutation	G	T	c.2038G>T	c.(2038-20 p.A680S	sushi domain containing 2
BTC-7	ZNF14	7561	19	19822422	19822422	Missense_Mutation	C	A	c.1668G>T	c.(1666-16 p.E556D	zinc finger protein 14
BTC-8	APC	324	5	112175952	112175952	Frame_Shift_Del	A	-	c.4661delA	c.(4660-46 p.E1554fs	adenomatous polyposis coli
BTC-8	LMO7	4008	13	76408475	76408475	Nonsense_Mutation	C	T	c.2479C>T	c.(2479-24 p.Q827*	LIM domain 7
BTC-8	TP53	7157	17	7574003	7574003	Nonsense_Mutation	G	A	c.1024C>T	c.(1024-10 p.R342*	tumor protein p53
BTC-8	DUXA	503835	19	57670637	57670637	Nonsense_Mutation	G	A	c.190C>T	c.(190-192 p.Q64*	double homeobox A
BTC-8	MMS22L	253714	6	97676947	97676947	Nonsense_Mutation	C	T	c.1862G>A	c.(1861-18 p.W621*	MMS22-like, DNA repair protein
BTC-8	KIF15	56992	3	44843467	44843467	Splice_Site	G	C		c.e13+1	kinesin family member 15
BTC-8	ABCA9	10350	17	67025361	67025361	Missense_Mutation	T	G	c.1453A>C	c.(1453-14 p.N485H	ATP-binding cassette, sub-family A (ABC1), member 9
BTC-8	ACSS1	84532	20	25004233	25004233	Missense_Mutation	C	T	c.676G>A	c.(676-678 p.G226R	acyl-CoA synthetase short-chain family member 1
BTC-8	ADAMTS6	11174	5	64595872	64595872	Missense_Mutation	G	A	c.1310C>T	c.(1309-13 p.A437V	ADAM metalloproteinase with thrombospondin type 1 motif, 6
BTC-8	ADORA1	134	1	203098180	203098180	Missense_Mutation	A	G	c.211A>G	c.(211-213 p.I71V	adenosine A1 receptor
BTC-8	AOC3	8639	17	41006705	41006705	Missense_Mutation	C	T	c.1841C>T	c.(1840-18 p.P614L	amine oxidase, copper containing 3
BTC-8	CPZ	8532	4	8603105	8603105	Missense_Mutation	G	A	c.344G>A	c.(343-345 p.R115Q	carboxypeptidase Z
BTC-8	DACT3	147906	19	47151783	47151783	Missense_Mutation	G	C	c.1846C>G	c.(1846-18 p.L616V	dishevelled-binding antagonist of beta-catenin 3
BTC-8	ENPP2	5168	8	120577135	120577135	Missense_Mutation	A	C	c.1097T>G	c.(1096-10 p.V366G	ectonucleotide pyrophosphatase/phosphodiesterase 2
BTC-8	INO80D	54891	2	206870153	206870153	Missense_Mutation	C	A	c.2023G>T	c.(2023-20 p.A675S	INO80 complex subunit D
BTC-8	IRX3	79191	16	54319378	54319378	Missense_Mutation	T	C	c.415A>G	c.(415-417 p.T139A	iroquois homeobox 3
BTC-8	KRAS	3845	12	25398281	25398281	Missense_Mutation	C	T	c.38G>A	c.(37-39)g(p.G13D	Kirsten rat sarcoma viral oncogene homolog
BTC-8	KRT73	319101	12	53004589	53004589	Missense_Mutation	C	T	c.1141G>A	c.(1141-11 p.A381T	keratin 73
BTC-8	LAX1	54900	1	203743063	203743063	Missense_Mutation	G	A	c.451G>A	c.(451-453 p.A151T	lymphocyte transmembrane adaptor 1
BTC-8	MATN4	8785	20	43927089	43927089	Missense_Mutation	G	A	c.1147C>T	c.(1147-11 p.R383W	matrilin 4
BTC-8	NAB1	4664	2	191524551	191524551	Missense_Mutation	G	A	c.649G>A	c.(649-651 p.E217K	NGFI-A binding protein 1 (EGR1 binding protein 1)
BTC-8	NPR3	4883	5	32712527	32712527	Missense_Mutation	C	A	c.645C>A	c.(643-645 p.F215L	natriuretic peptide receptor 3
BTC-8	OTOG	340990	11	17655819	17655819	Missense_Mutation	A	G	c.7288A>G	c.(7288-72 p.T2430A	otogelin
BTC-8	PCDHGB6	56100	5	140788665	140788665	Missense_Mutation	G	A	c.896G>A	c.(895-897 p.G299D	protocadherin gamma subfamily B, 6
BTC-8	POLA1	5422 X		24906154	24906154	Missense_Mutation	G	A	c.4061G>A	c.(4060-40 p.R1354H	polymerase (DNA directed), alpha 1, catalytic subunit
BTC-8	RALGAPA1	253959	14	36096402	36096402	Missense_Mutation	T	A	c.5272A>T	c.(5272-52 p.M1758L	Ral GTPase activating protein, alpha subunit 1 (catalytic)
BTC-8	RIPK2	8767	8	90792331	90792331	Missense_Mutation	G	C	c.882G>C	c.(880-882 p.L294F	receptor-interacting serine-threonine kinase 2
BTC-8	ROBO1	6091	3	78710287	78710287	Missense_Mutation	A	G	c.2213T>C	c.(2212-22 p.I738T	roundabout, axon guidance receptor, homolog 1 (Drosophila)
BTC-8	ROBO2	6092	3	77657029	77657029	Missense_Mutation	C	A	c.3217C>A	c.(3217-32 p.P1073T	roundabout, axon guidance receptor, homolog 2 (Drosophila)
BTC-8	SHISA8	440829	22	42310285	42310285	Missense_Mutation	C	A	c.286G>T	c.(286-288 p.A96S	shisa family member 8
BTC-8	SMARCA2	6595	9	2104152	2104152	Missense_Mutation	T	C	c.3275T>C	c.(3274-32 p.L1092P	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 2
BTC-8	SSTR1	6751	14	38678956	38678956	Missense_Mutation	G	A	c.362G>A	c.(361-363 p.R121H	somatostatin receptor 1
BTC-8	SYN3	8224	22	33265027	33265027	Missense_Mutation	C	T	c.547G>A	c.(547-549 p.G183S	synapsin III
BTC-8	TROVE2	6738	1	193046115	193046115	Missense_Mutation	C	G	c.1021C>G	c.(1021-10 p.L341V	TROVE domain family, member 2
BTC-8	USH2A	7399	1	215844423	215844423	Missense_Mutation	C	T	c.14024G>A	c.(14023-1 p.R4675K	Usher syndrome 2A (autosomal recessive, mild)
BTC-8	USH2A	7399	1	216062042	216062042	Missense_Mutation	G	C	c.7949C>G	c.(7948-79 p.P2650R	Usher syndrome 2A (autosomal recessive, mild)
BTC-8	XCR1	2829	3	46062623	46062623	Missense_Mutation	G	A	c.817C>T	c.(817-819 p.R273C	chemokine (C motif) receptor 1
BTC-8	ZSCAN20	7579	1	33960845	33960845	Missense_Mutation	C	A	c.2901C>A	c.(2899-29 p.F967L	zinc finger and SCAN domain containing 20

Supplementary Table 1. Coverage Information

Sample	X1	X5	X10	X15	X20	X25	X30	X50	Total bases aligned(bases)	Mean coverage depth
BTC_1N	99.96%	99.77%	99.22%	98.14%	96.33%	93.69%	90.19%	69.94%	6,034,974,927	80.99
BTC_1T	99.97%	99.90%	99.71%	99.38%	98.84%	98.03%	96.92%	89.02%	9,863,779,665	132.38
BTC_2N	99.86%	99.66%	99.15%	98.17%	96.48%	93.95%	90.45%	69.62%	5,978,637,815	80.24
BTC_2T	99.88%	99.79%	99.63%	99.38%	99.00%	98.46%	97.71%	92.10%	11,086,728,430	148.79
BTC_4N	99.97%	99.80%	99.30%	98.32%	96.67%	94.26%	91.07%	72.14%	6,345,622,329	85.16
BTC_4T	99.98%	99.90%	99.72%	99.43%	98.98%	98.33%	97.44%	91.14%	10,760,737,151	144.41
BTC_5N	99.86%	99.62%	98.95%	97.66%	95.53%	92.44%	88.40%	66.57%	5,823,253,443	78.15
BTC_5T	99.87%	99.78%	99.57%	99.21%	98.65%	97.88%	96.83%	89.50%	10,599,020,827	142.24
BTC_6N	99.87%	99.67%	99.16%	98.14%	96.43%	93.80%	90.22%	69.15%	5,968,127,744	80.09
BTC_6T	99.88%	99.79%	99.63%	99.37%	98.99%	98.43%	97.66%	91.94%	10,923,830,027	146.60
BTC_7N	99.97%	99.78%	99.25%	98.21%	96.48%	93.96%	90.63%	71.11%	6,186,543,991	83.03
BTC_7T	99.98%	99.88%	99.66%	99.27%	98.66%	97.75%	96.48%	87.79%	9,956,458,889	133.62
BTC_8N	99.97%	99.76%	99.17%	97.97%	95.96%	93.09%	89.35%	68.81%	5,998,051,912	80.50
BTC_8T	99.98%	99.89%	99.68%	99.34%	98.82%	98.06%	97.01%	89.49%	10,131,605,777	135.97

Supplementary Table 2. Detailed information on total somatic mutations

Patient ID	Hugo_Sym	Entrez_Gen	Chromoso	Start_position	End_position	Variant_Classification	Reference_Tumor_Seq	cDNA_Change	Codon_Ch.	Protein_Ch	Description
BTC-1	APC	324	5	112174759	112174760	Frame_Shift_Del	AG	-	c.3468_3469delAC	c.(3466-34 p.EE1156fs	adenomatous polyposis coli
BTC-1	KRTAP19-ε	337973	21	31914024	31914024	Nonsense_Mutation	G	T	c.129C>A	c.(127-129 p.C43*	keratin associated protein 19-6
BTC-1	PABPC4L	132430	4	135121319	135121319	Nonsense_Mutation	G	A	c.856C>T	c.(856-858 p.R286*	poly(A) binding protein, cytoplasmic 4-like
BTC-1	APC	324	5	112175951	112175951	Nonsense_Mutation	G	T	c.4660G>T	c.(4660-46 p.E1554*	adenomatous polyposis coli
BTC-1	C6orf222	389384	6	36290159	36290159	Nonsense_Mutation	G	C	c.1532C>G	c.(1531-15 p.S511*	chromosome 6 open reading frame 222
BTC-1	ABCA1	19	9	107553269	107553269	Nonsense_Mutation	G	C	c.5861C>G	c.(5860-58 p.S1954*	ATP-binding cassette, sub-family A (ABC1), member 1
BTC-1	AMER1	139285 X		63412530	63412530	Nonsense_Mutation	G	A	c.637C>T	c.(637-639 p.Q213*	APC membrane recruitment protein 1
BTC-1	MFAP2	4237	1	17302995	17302995	Splice_Site	C	A		c.e5+1	microfibrillar-associated protein 2
BTC-1	HIF3A	64344	19	46825222	46825222	Splice_Site	C	T	c.1334C>T	c.(1333-13 p.S445L	hypoxia inducible factor 3, alpha subunit
BTC-1	MAP3K13	9175	3	185155234	185155234	Splice_Site	G	C		c.e3-1	mitogen-activated protein kinase kinase kinase 13
BTC-1	DAB2IP	153090	9	124329507	124329507	Splice_Site	G	A	c.40G>A	c.(40-42)G; p.E14K	DAB2 interacting protein
BTC-1	ABCC2	1244	10	101591752	101591752	Missense_Mutation	C	T	c.3122C>T	c.(3121-31 p.A1041V	ATP-binding cassette, sub-family C (CFTR/MRP), member 2
BTC-1	ACE	1636	17	61570945	61570945	Missense_Mutation	G	A	c.1339G>A	c.(1339-13 p.V447M	angiotensin I converting enzyme
BTC-1	ACTN2	88	1	236899007	236899007	Missense_Mutation	C	T	c.770C>T	c.(769-771 p.A257V	actinin, alpha 2
BTC-1	ACTN4	81	19	39207832	39207832	Missense_Mutation	G	A	c.1019G>A	c.(1018-10 p.R340H	actinin, alpha 4
BTC-1	ACVR1B	91	12	52379016	52379016	Missense_Mutation	C	A	c.864C>A	c.(862-864 p.N288K	activin A receptor, type IB
BTC-1	ADCY5	111	3	123166420	123166420	Missense_Mutation	C	T	c.973G>A	c.(973-975 p.A325T	adenylate cyclase 5
BTC-1	ADCY8	114	8	131949376	131949376	Missense_Mutation	C	T	c.1424G>A	c.(1423-14 p.R475H	adenylate cyclase 8 (brain)
BTC-1	ANO3	63982	11	26463533	26463533	Missense_Mutation	T	C	c.115T>C	c.(115-117 p.C39R	anoctamin 3
BTC-1	APC2	10297	19	1457029	1457029	Missense_Mutation	G	A	c.994G>A	c.(994-996 p.A332T	adenomatosis polyposis coli 2
BTC-1	AQPEP	0	5	115298621	115298621	Missense_Mutation	G	A	c.307G>A	c.(307-309 p.V103M	
BTC-1	AUTS2	26053	7	69364311	69364311	Missense_Mutation	C	T	c.349C>T	c.(349-351 p.R117C	autism susceptibility candidate 2
BTC-1	C11orf63	79864	11	122775018	122775018	Missense_Mutation	C	T	c.730C>T	c.(730-732 p.R244C	chromosome 11 open reading frame 63
BTC-1	C5orf42	65250	5	37153858	37153858	Missense_Mutation	T	A	c.8195A>T	c.(8194-81 p.D2732V	chromosome 5 open reading frame 42
BTC-1	CDH2	1000	18	25572711	25572711	Missense_Mutation	C	T	c.1159G>A	c.(1159-11 p.A387T	cadherin 2, type 1, N-cadherin (neuronal)
BTC-1	CHST9	83539	18	24628462	24628462	Missense_Mutation	C	T	c.127G>A	c.(127-129 p.V43M	carbohydrate (N-acetylgalactosamine 4-0) sulfotransferase 9
BTC-1	CYP11B1	1584	8	143956558	143956558	Missense_Mutation	C	T	c.1426G>A	c.(1426-14 p.V476M	cytochrome P450, family 11, subfamily B, polypeptide 1
BTC-1	DNAH10	196385	12	124341718	124341718	Missense_Mutation	G	A	c.6200G>A	c.(6199-62 p.R2067H	dynein, axonemal, heavy chain 10
BTC-1	DSCAM	1826	21	41559851	41559851	Missense_Mutation	C	T	c.2617G>A	c.(2617-26 p.E873K	Down syndrome cell adhesion molecule
BTC-1	ELF3	1999	1	201983140	201983140	Missense_Mutation	G	A	c.989G>A	c.(988-990 p.S330N	E74-like factor 3 (ets domain transcription factor, epithelial-specific)
BTC-1	FGD3	89846	9	95738639	95738639	Missense_Mutation	C	T	c.101C>T	c.(100-102 p.A34V	FYVE, RhoGEF and PH domain containing 3
BTC-1	FUBP3	8939	9	133493210	133493210	Missense_Mutation	G	A	c.594G>A	c.(592-594 p.M198I	far upstream element (FUSE) binding protein 3
BTC-1	HAND1	9421	5	153855452	153855452	Missense_Mutation	G	T	c.562C>A	c.(562-564 p.P188T	heart and neural crest derivatives expressed 1
BTC-1	KCNS2	3788	8	99440299	99440299	Missense_Mutation	G	A	c.92G>A	c.(91-93)c(p.R31H	potassium voltage-gated channel, delayed-rectifier, subfamily S, member 2
BTC-1	KIAA1429	25962	8	95503865	95503865	Missense_Mutation	C	G	c.5081G>C	c.(5080-50 p.R1694T	KIAA1429
BTC-1	KRTAP19-1	337882	21	31852374	31852374	Missense_Mutation	C	T	c.263G>A	c.(262-264 p.G88D	keratin associated protein 19-1
BTC-1	LRP1B	53353	2	141004693	141004693	Missense_Mutation	G	T	c.13286C>A	c.(13285-1 p.P4429Q	low density lipoprotein receptor-related protein 1B
BTC-1	LRRK2	120892	12	40689336	40689336	Missense_Mutation	G	T	c.2986G>T	c.(2986-29 p.D996Y	leucine-rich repeat kinase 2
BTC-1	MAP2K1	5604	15	66727455	66727455	Missense_Mutation	G	C	c.171G>C	c.(169-171 p.K57N	mitogen-activated protein kinase kinase 1

BTC-1	MBTD1	54799	17	49296318	49296318	Missense_Mutation	A	C	c.376T>G	c.(376-378 p.L126V	mbt domain containing 1
BTC-1	MUC19	283463	12	40820354	40820354	Missense_Mutation	G	A	c.332G>A	c.(331-333 p.R111H	mucin 19, oligomeric
BTC-1	MYLK2	85366	20	30419645	30419645	Missense_Mutation	G	A	c.1564G>A	c.(1564-15 p.V522I	myosin light chain kinase 2
BTC-1	PCDH17	27253	13	58207860	58207860	Missense_Mutation	G	A	c.1180G>A	c.(1180-11 p.G394R	protocadherin 17
BTC-1	PLIN4	729359	19	4510530	4510530	Missense_Mutation	G	A	c.3400C>T	c.(3400-34 p.R1134C	perilipin 4
BTC-1	PRRC2B	84726	9	134322013	134322013	Missense_Mutation	A	G	c.839A>G	c.(838-840 p.D280G	proline-rich coiled-coil 2B
BTC-1	RBM15	64783	1	110883546	110883546	Missense_Mutation	T	G	c.1519T>G	c.(1519-15 p.W507G	RNA binding motif protein 15
BTC-1	RP1-139D8	0	6	42123292	42123292	Missense_Mutation	G	T	c.14G>T	c.(13-15)a(c.p.S5I	
BTC-1	SDK1	221935	7	4273010	4273010	Missense_Mutation	C	T	c.5891C>T	c.(5890-58 p.A1964V	sidekick cell adhesion molecule 1
BTC-1	SPRY2	10253	13	80911584	80911584	Missense_Mutation	G	C	c.257C>G	c.(256-258 p.P86R	sprouty homolog 2 (Drosophila)
BTC-1	ST6GALNA	81849	1	77509964	77509964	Missense_Mutation	C	A	c.337C>A	c.(337-339 p.Q113K	ST6 (alpha-N-acetyl-neuraminy-2,3-beta-galactosyl-1,3)-N-acetylgalactosaminide alpha-2,6-sialyltransferase 5
BTC-1	SYT3	84258	19	51135883	51135883	Missense_Mutation	C	T	c.334G>A	c.(334-336 p.G112R	synaptotagmin III
BTC-1	TADA2A	6871	17	35837062	35837062	Missense_Mutation	G	C	c.1307G>C	c.(1306-13 p.R436T	transcriptional adaptor 2A
BTC-1	TGM2	7052	20	36793594	36793594	Missense_Mutation	C	T	c.7G>A	c.(7-9)Gag p.E3K	transglutaminase 2
BTC-1	TMEM184I	25829	22	38643871	38643871	Missense_Mutation	T	C	c.97A>G	c.(97-99)A(c.p.T33A	transmembrane protein 184B
BTC-1	TRPC1	7220	3	142499703	142499703	Missense_Mutation	A	T	c.792A>T	c.(790-792 p.Q264H	transient receptor potential cation channel, subfamily C, member 1
BTC-1	TSHZ3	57616	19	31770589	31770589	Missense_Mutation	G	C	c.110C>G	c.(109-111 p.A37G	teashirt zinc finger homeobox 3
BTC-1	TSPAN14	81619	10	82271948	82271948	Missense_Mutation	T	G	c.499T>G	c.(499-501 p.Y167D	tetraspanin 14
BTC-1	ZFR	51663	5	32395347	32395347	Missense_Mutation	C	T	c.1897G>A	c.(1897-18 p.E633K	zinc finger RNA binding protein
BTC-1	ZFYVE26	23503	14	68264958	68264958	Missense_Mutation	C	A	c.2021G>T	c.(2020-20 p.G674V	zinc finger, FYVE domain containing 26
BTC-2	EPHA2	1969	1	16464447	16464447	Frame_Shift_Del	G	-	c.1213delC	c.(1213-12 p.H405fs	EPH receptor A2
BTC-2	LGI4	163175	19	35617610	35617610	Nonsense_Mutation	C	T	c.863G>A	c.(862-864 p.W288*	leucine-rich repeat LGI family, member 4
BTC-2	EPHA2	1969	1	16456720	16456720	Splice_Site	C	T		c.e15+1	EPH receptor A2
BTC-2	C17orf97	400566	17	260315	260315	Missense_Mutation	G	T	c.182G>T	c.(181-183 p.G61V	chromosome 17 open reading frame 97
BTC-2	CAMK2N1	55450	1	20810156	20810156	Missense_Mutation	G	A	c.223C>T	c.(223-225 p.P75S	calcium/calmodulin-dependent protein kinase II inhibitor 1
BTC-2	CNBD1	168975	8	88249206	88249206	Missense_Mutation	G	A	c.637G>A	c.(637-639 p.V213M	cyclic nucleotide binding domain containing 1
BTC-2	COL14A1	7373	8	121292323	121292323	Missense_Mutation	G	A	c.3631G>A	c.(3631-36 p.E1211K	collagen, type XIV, alpha 1
BTC-2	FBN3	84467	19	8201283	8201283	Missense_Mutation	C	T	c.1334G>A	c.(1333-13 p.G445D	fibrillin 3
BTC-2	FIGN	55137	2	164467285	164467285	Missense_Mutation	C	A	c.1057G>T	c.(1057-10 p.D353Y	fidgetin
BTC-2	FLNB	2317	3	58118580	58118580	Missense_Mutation	A	G	c.4529A>G	c.(4528-45 p.H1510R	filamin B, beta
BTC-2	INO80D	54891	2	206884534	206884534	Missense_Mutation	G	A	c.1334C>T	c.(1333-13 p.A445V	INO80 complex subunit D
BTC-2	INTS1	26173	7	1544069	1544069	Missense_Mutation	C	G	c.233G>C	c.(232-234 p.C78S	integrator complex subunit 1
BTC-2	KCNIP1	30820	5	170145809	170145809	Missense_Mutation	G	A	c.109G>A	c.(109-111 p.E37K	Kv channel interacting protein 1
BTC-2	KIAA1549	57670	7	138603864	138603864	Missense_Mutation	G	A	c.508C>T	c.(508-510 p.R170W	KIAA1549
BTC-2	LGALS9	3965	17	25975970	25975970	Missense_Mutation	G	A	c.934G>A	c.(934-936 p.V312M	lectin, galactoside-binding, soluble, 9
BTC-2	PLEKHG1	57480	6	151152904	151152904	Missense_Mutation	G	A	c.2657G>A	c.(2656-26 p.R886H	pleckstrin homology domain containing, family G (with RhoGef domain) member 1
BTC-2	PNCK	139728 X		152937421	152937421	Missense_Mutation	C	G	c.328G>C	c.(328-330 p.E110Q	pregnancy up-regulated nonubiquitous CaM kinase
BTC-2	PPDPF	79144	20	62152924	62152924	Missense_Mutation	G	A	c.115G>A	c.(115-117 p.A39T	pancreatic progenitor cell differentiation and proliferation factor
BTC-2	PROKR2	128674	20	5282783	5282783	Missense_Mutation	C	T	c.1058G>A	c.(1057-10 p.R353H	prokineticin receptor 2

BTC-2	RP11-429E	0	20	60294088	60294088	Missense_Mutation	G	A	c.139C>T	c.(139-141 p.L47F	
BTC-2	SCN5A	6331	3	38645514	38645514	Missense_Mutation	C	T	c.1579G>A	c.(1579-15 p.G527R	sodium channel, voltage-gated, type V, alpha subunit
BTC-2	SLC35F1	222553	6	118556702	118556702	Missense_Mutation	G	A	c.380G>A	c.(379-381 p.R127Q	solute carrier family 35, member F1
BTC-2	SLC9A6	10479 X		135067950	135067950	Missense_Mutation	C	T	c.289C>T	c.(289-291 p.R97C	solute carrier family 9, subfamily A (NHE6, cation proton antiporter 6), member 6
BTC-2	SPARC	6678	5	151047056	151047056	Missense_Mutation	T	C	c.557A>G	c.(556-558 p.N186S	secreted protein, acidic, cysteine-rich (osteonectin)
BTC-2	TLL2	7093	10	98144434	98144434	Missense_Mutation	C	G	c.2104G>C	c.(2104-21 p.V702L	tolloid-like 2
BTC-2	TRIM58	25893	1	248028097	248028097	Missense_Mutation	C	T	c.607C>T	c.(607-609 p.R203W	tripartite motif containing 58
BTC-2	TSPAN16	26526	19	11408947	11408947	Missense_Mutation	A	G	c.199A>G	c.(199-201 p.I67V	tetraspanin 16
BTC-2	TSSK2	23617	22	19119034	19119034	Missense_Mutation	A	C	c.122A>C	c.(121-123 p.K41T	testis-specific serine kinase 2
BTC-2	VAV2	7410	9	136635595	136635595	Missense_Mutation	G	A	c.2222C>T	c.(2221-22 p.S741L	vav 2 guanine nucleotide exchange factor
BTC-2	ZC2HC1B	153918	6	144224199	144224199	Missense_Mutation	C	T	c.508C>T	c.(508-510 p.P170S	zinc finger, C2HC-type containing 1B
BTC-2	ZFP69B	65243	1	40928270	40928270	Missense_Mutation	G	A	c.614G>A	c.(613-615 p.G205D	ZFP69 zinc finger protein B
BTC-2	ZNF423	23090	16	49669726	49669726	Missense_Mutation	C	T	c.3337G>A	c.(3337-33 p.A1113T	zinc finger protein 423
BTC-4	ARID1A	8289	1	27023831	27023831	Frame_Shift_Del	G	-	c.937delG	c.(937-939 p.G314fs	AT rich interactive domain 1A (SWI-like)
BTC-4	TULP1	7287	6	35480415	35480415	Splice_Site	C	G		c.e2+1	tubby like protein 1
BTC-4	ACOXL	55289	2	111850501	111850501	Missense_Mutation	T	A	c.1590T>A	c.(1588-15 p.H530Q	acyl-CoA oxidase-like
BTC-4	CACNA1H	8912	16	1270626	1270626	Missense_Mutation	C	A	c.6676C>A	c.(6676-66 p.L2226M	calcium channel, voltage-dependent, T type, alpha 1H subunit
BTC-4	CATSPERB	79820	14	92174496	92174496	Missense_Mutation	T	A	c.455A>T	c.(454-456 p.D152V	catsper channel auxiliary subunit beta
BTC-4	CDKN2A	1029	9	21971035	21971035	Missense_Mutation	T	C	c.170A>G	c.(169-171 p.D57G	cyclin-dependent kinase inhibitor 2A
BTC-4	CES3	23491	16	67006620	67006620	Missense_Mutation	G	A	c.408G>A	c.(406-408 p.M136I	carboxylesterase 3
BTC-4	CLCNKA	1187	1	16349137	16349137	Missense_Mutation	G	A	c.23G>A	c.(22-24)c(p.R8H	chloride channel, voltage-sensitive Ka
BTC-4	COL22A1	169044	8	139890232	139890232	Missense_Mutation	C	T	c.419G>A	c.(418-420 p.R140H	collagen, type XXII, alpha 1
BTC-4	CRISPLD1	83690	8	75941665	75941665	Missense_Mutation	G	A	c.1364G>A	c.(1363-13 p.R455Q	cysteine-rich secretory protein LCCL domain containing 1
BTC-4	DAW1	164781	2	228767727	228767727	Missense_Mutation	T	C	c.550T>C	c.(550-552 p.S184P	dynein assembly factor with WDR repeat domains 1
BTC-4	DYSF	8291	2	71797023	71797023	Missense_Mutation	G	A	c.2938G>A	c.(2938-29 p.G980R	dysferlin
BTC-4	ELN	2006	7	73461047	73461047	Missense_Mutation	C	T	c.593C>T	c.(592-594 p.P198L	elastin
BTC-4	FAM135B	51059	8	139165339	139165339	Missense_Mutation	A	G	c.1379T>C	c.(1378-13 p.L460P	family with sequence similarity 135, member B
BTC-4	FAM208B	54906	10	5803432	5803432	Missense_Mutation	C	A	c.7172C>A	c.(7171-71 p.T2391K	family with sequence similarity 208, member B
BTC-4	GPR65	8477	14	88478153	88478153	Missense_Mutation	G	A	c.962G>A	c.(961-963 p.R321H	G protein-coupled receptor 65
BTC-4	GUCY1A3	2982	4	156643206	156643206	Missense_Mutation	A	T	c.1733A>T	c.(1732-17 p.H578L	guanylate cyclase 1, soluble, alpha 3
BTC-4	HID1	283987	17	72959068	72959068	Missense_Mutation	T	A	c.496A>T	c.(496-498 p.S166C	HID1 domain containing
BTC-4	HIP1	3092	7	75189111	75189111	Missense_Mutation	G	A	c.1300C>T	c.(1300-13 p.R434W	huntingtin interacting protein 1
BTC-4	IRF3	3661	19	50167939	50167939	Missense_Mutation	T	C	c.157A>G	c.(157-159 p.I53V	interferon regulatory factor 3
BTC-4	KCNB1	3745	20	48098906	48098906	Missense_Mutation	C	T	c.112G>A	c.(112-114 p.G38R	potassium voltage-gated channel, Shab-related subfamily, member 1
BTC-4	KCNT1	57582	9	138664603	138664603	Missense_Mutation	C	T	c.2051C>T	c.(2050-20 p.T684M	potassium channel, subfamily T, member 1
BTC-4	LLGL1	3996	17	18143990	18143990	Missense_Mutation	C	T	c.2305C>T	c.(2305-23 p.R769W	lethal giant larvae homolog 1 (Drosophila)
BTC-4	LPHN1	22859	19	14294398	14294398	Missense_Mutation	G	A	c.17C>T	c.(16-18)g(p.A6V	latrophilin 1
BTC-4	MARS	4141	12	57906626	57906626	Missense_Mutation	A	T	c.1846A>T	c.(1846-18 p.I616F	methionyl-tRNA synthetase
BTC-4	MBTPS1	8720	16	84093019	84093019	Missense_Mutation	G	A	c.2719C>T	c.(2719-27 p.R907W	membrane-bound transcription factor peptidase, site 1
BTC-4	PDC	5132	1	186413572	186413572	Missense_Mutation	G	A	c.280C>T	c.(280-282 p.R94C	phosducin
BTC-4	PTTG1IP	754	21	46285352	46285352	Missense_Mutation	C	G	c.126G>C	c.(124-126 p.Q42H	pituitary tumor-transforming 1 interacting protein

BTC-4	RBBP6	5930	16	24581304	24581304	Missense_Mutation	A	T	c.3293A>T	c.(3292-32 p.E1098V	retinoblastoma binding protein 6
BTC-4	RBBP7	5931 X		16871823	16871823	Missense_Mutation	T	A	c.872A>T	c.(871-873 p.D291V	retinoblastoma binding protein 7
BTC-4	RDH10	157506	8	74234976	74234976	Missense_Mutation	A	G	c.833A>G	c.(832-834 p.K278R	retinol dehydrogenase 10 (all-trans)
BTC-4	RFX8	731220	2	102018971	102018971	Missense_Mutation	C	T	c.1172G>A	c.(1171-11 p.R391Q	RFX family member 8, lacking RFX DNA binding domain
BTC-4	SEMA6D	80031	15	48058143	48058143	Missense_Mutation	C	T	c.1505C>T	c.(1504-15 p.A502V	sema domain, transmembrane domain (TM), and cytoplasmic domain, (semaphorin) 6D
BTC-4	SLC6A17	388662	1	110734682	110734682	Missense_Mutation	C	A	c.953C>A	c.(952-954 p.A318D	solute carrier family 6 (neutral amino acid transporter), member 17
BTC-4	TMEM205	374882	19	11455971	11455971	Missense_Mutation	G	A	c.221C>T	c.(220-222 p.S74L	transmembrane protein 205
BTC-4	TRIM4	89122	7	99506284	99506284	Missense_Mutation	T	C	c.719A>G	c.(718-720 p.N240S	tripartite motif containing 4
BTC-5	HMCN1	83872	1	186057081	186057084	Frame_Shift_Del	GTAT	-	c.9381_9384delGT	c.(9379-93 p.QY3127f	hemimentin 1
BTC-5	UPP2	151531	2	158977957	158977958	Frame_Shift_Ins	-	T	c.491_492insT	c.(490-495 p.V165fs	uridine phosphorylase 2
BTC-5	FNTB	2342	14	65511053	65511053	Nonsense_Mutation	C	T	c.847C>T	c.(847-849 p.R283*	farnesyltransferase, CAAX box, beta
BTC-5	ACSM2A	123876	16	20482871	20482871	Nonsense_Mutation	C	T	c.754C>T	c.(754-756 p.Q252*	acyl-CoA synthetase medium-chain family member 2A
BTC-5	PSD3	23362	8	18430175	18430175	Nonsense_Mutation	C	A	c.2647G>T	c.(2647-26 p.E883*	pleckstrin and Sec7 domain containing 3
BTC-5	COL11A1	1301	1	103467527	103467527	Splice_Site	T	C		c.e24-2	collagen, type XI, alpha 1
BTC-5	AC007405.	0	2	171570595	171570595	Missense_Mutation	G	C	c.359C>G	c.(358-360 p.A120G	
BTC-5	AC009365.	0	7	132412557	132412557	Missense_Mutation	G	A	c.409G>A	c.(409-411 p.V137I	
BTC-5	ADAM21	8747	14	70924614	70924614	Missense_Mutation	G	T	c.398G>T	c.(397-399 p.R133L	ADAM metalloproteinase domain 21
BTC-5	APOOL	139322 X		84329077	84329077	Missense_Mutation	G	C	c.560G>C	c.(559-561 p.S187T	apolipoprotein O-like
BTC-5	ASPM	259266	1	197074062	197074062	Missense_Mutation	T	C	c.4319A>G	c.(4318-43 p.Q1440R	asp (abnormal spindle) homolog, microcephaly associated (Drosophila)
BTC-5	ATP7A	538 X		77301051	77301051	Missense_Mutation	C	A	c.4208C>A	c.(4207-42 p.S1403Y	ATPase, Cu++ transporting, alpha polypeptide
BTC-5	ATP8B4	79895	15	50288883	50288883	Missense_Mutation	C	G	c.580G>C	c.(580-582 p.G194R	ATPase, class I, type 8B, member 4
BTC-5	CACNA1A	773	19	13616812	13616812	Missense_Mutation	C	A	c.227G>T	c.(226-228 p.R76L	calcium channel, voltage-dependent, P/Q type, alpha 1A subunit
BTC-5	CCDC175	729665	14	60031860	60031860	Missense_Mutation	T	C	c.625A>G	c.(625-627 p.I209V	coiled-coil domain containing 175
BTC-5	CCL18	6362	17	34398340	34398340	Missense_Mutation	G	A	c.209G>A	c.(208-210 p.C70Y	chemokine (C-C motif) ligand 18 (pulmonary and activation-regulated)
BTC-5	CD163	9332	12	7651580	7651580	Missense_Mutation	A	G	c.662T>C	c.(661-663 p.I221T	CD163 molecule
BTC-5	COL21A1	81578	6	55922544	55922544	Missense_Mutation	G	T	c.2785C>A	c.(2785-27 p.Q929K	collagen, type XXI, alpha 1
BTC-5	CSMD3	114788	8	113697935	113697935	Missense_Mutation	G	C	c.2182C>G	c.(2182-21 p.P728A	CUB and Sushi multiple domains 3
BTC-5	CXorf28	1E+08 X		3189922	3189922	Missense_Mutation	C	T	c.49C>T	c.(49-51)C(p.P17S	chromosome X open reading frame 28
BTC-5	DMXL2	23312	15	51791693	51791693	Missense_Mutation	T	A	c.3728A>T	c.(3727-37 p.D1243V	Dmx-like 2
BTC-5	GAS2	2620	11	22747946	22747946	Missense_Mutation	G	A	c.376G>A	c.(376-378 p.D126N	growth arrest-specific 2
BTC-5	GPR98	84059	5	89939702	89939702	Missense_Mutation	C	T	c.2636C>T	c.(2635-26 p.T879M	G protein-coupled receptor 98
BTC-5	HOXA4	3201	7	27170310	27170310	Missense_Mutation	G	T	c.43C>A	c.(43-45)C(p.P15T	homeobox A4
BTC-5	HP	3240	16	72090131	72090131	Missense_Mutation	C	T	c.77C>T	c.(76-78)a(p.T26M	haptoglobin
BTC-5	ICAM5	7087	19	10405259	10405259	Missense_Mutation	T	C	c.2173T>C	c.(2173-21 p.C725R	intercellular adhesion molecule 5, telencephalin
BTC-5	KIAA1244	57221	6	138576619	138576619	Missense_Mutation	C	T	c.817C>T	c.(817-819 p.H273Y	KIAA1244
BTC-5	KIF21B	23046	1	200948693	200948693	Missense_Mutation	C	T	c.4130G>A	c.(4129-41 p.R1377Q	kinesin family member 21B
BTC-5	KRTAP21-1	337977	21	32127554	32127554	Missense_Mutation	C	A	c.143G>T	c.(142-144 p.C48F	keratin associated protein 21-1
BTC-5	LCA10	0 X		153152431	153152431	Missense_Mutation	G	T	c.311G>T	c.(310-312 p.R104L	

BTC-5	MAP1B	4131	5	71490888	71490888	Missense_Mutation	C	G	c.1706C>G	c.(1705-17 p.T569R	microtubule-associated protein 1B
BTC-5	MLK4	0	1	233518335	233518335	Missense_Mutation	C	T	c.2989C>T	c.(2989-29 p.P997S	
BTC-5	NBEAL2	23218	3	47041430	47041430	Missense_Mutation	C	A	c.3841C>A	c.(3841-38 p.Q1281K	neurobeachin-like 2
BTC-5	NFATC2	4773	20	50140482	50140482	Missense_Mutation	C	T	c.298G>A	c.(298-300 p.A100T	nuclear factor of activated T-cells, cytoplasmic, calcineurin-dependent 2
BTC-5	NRAS	4893	1	115256529	115256529	Missense_Mutation	T	C	c.182A>G	c.(181-183 p.Q61R	neuroblastoma RAS viral (v-ras) oncogene homolog
BTC-5	OLFML2B	25903	1	161967939	161967939	Missense_Mutation	T	A	c.1153A>T	c.(1153-11 p.S385C	olfactomedin-like 2B
BTC-5	OSBPL5	114879	11	3143583	3143583	Missense_Mutation	G	A	c.295C>T	c.(295-297 p.L99F	oxysterol binding protein-like 5
BTC-5	PCDHB1	29930	5	140432385	140432385	Missense_Mutation	G	A	c.1330G>A	c.(1330-13 p.D444N	protocadherin beta 1
BTC-5	PCSK5	5125	9	78936474	78936474	Missense_Mutation	G	A	c.3940G>A	c.(3940-39 p.E1314K	proprotein convertase subtilisin/kexin type 5
BTC-5	PGR	5241	11	100998706	100998706	Missense_Mutation	C	T	c.1096G>A	c.(1096-10 p.D366N	progesterone receptor
BTC-5	PHIP	55023	6	79695085	79695085	Missense_Mutation	T	G	c.2521A>C	c.(2521-25 p.S841R	pleckstrin homology domain interacting protein
BTC-5	PIK3C2G	5288	12	18435656	18435656	Missense_Mutation	C	A	c.641C>A	c.(640-642 p.P214H	phosphatidylinositol-4-phosphate 3-kinase, catalytic subunit type 2 gamma
BTC-5	PRKCD	5580	3	53219710	53219710	Missense_Mutation	A	T	c.979A>T	c.(979-981 p.M327L	protein kinase C, delta
BTC-5	SEC14L2	23541	22	30802345	30802345	Missense_Mutation	C	G	c.145C>G	c.(145-147 p.L49V	SEC14-like 2 (S. cerevisiae)
BTC-5	SEMA6A	57556	5	115782872	115782872	Missense_Mutation	C	T	c.2530G>A	c.(2530-25 p.E844K	sema domain, transmembrane domain (TM), and cytoplasmic domain, (semaphorin) 6A
BTC-5	SKIV2L2	23517	5	54635888	54635888	Missense_Mutation	G	T	c.566G>T	c.(565-567 p.S189I	superkiller viralicidic activity 2-like 2 (S. cerevisiae)
BTC-5	TAF1L	138474	9	32631848	32631848	Missense_Mutation	G	A	c.3730C>T	c.(3730-37 p.R1244W	TAF1 RNA polymerase II, TATA box binding protein (TBP)-associated factor, 210kDa-like
BTC-5	TEX15	56154	8	30702546	30702546	Missense_Mutation	T	G	c.3988A>C	c.(3988-39 p.K1330Q	testis expressed 15
BTC-5	TLR8	51311 X		12939230	12939230	Missense_Mutation	G	C	c.2125G>C	c.(2125-21 p.E709Q	toll-like receptor 8
BTC-5	TMEM200A	114801	6	130762039	130762039	Missense_Mutation	A	G	c.472A>G	c.(472-474 p.I158V	transmembrane protein 200A
BTC-5	TMEM80	283232	11	703112	703112	Missense_Mutation	G	T	c.469G>T	c.(469-471 p.V157F	transmembrane protein 80
BTC-5	TRPM6	140803	9	77377993	77377993	Missense_Mutation	C	A	c.3594G>T	c.(3592-35 p.K1198N	transient receptor potential cation channel, subfamily M, member 6
BTC-5	TSPYL5	85453	8	98290039	98290039	Missense_Mutation	G	A	c.34C>T	c.(34-36)C ₁ p.R12C	TSPYL-like 5
BTC-6	AMIGO2	347902	12	47471598	47471598	Missense_Mutation	A	C	c.1188T>G	c.(1186-11 p.F396L	adhesion molecule with Ig-like domain 2
BTC-6	BIRC6	57448	2	32712788	32712788	Missense_Mutation	A	T	c.7888A>T	c.(7888-78 p.I2630F	baculoviral IAP repeat containing 6
BTC-6	BZW1	9689	2	201680210	201680210	Missense_Mutation	T	G	c.211T>G	c.(211-213 p.F71V	basic leucine zipper and W2 domains 1
BTC-6	CD200R1	131450	3	112648303	112648303	Missense_Mutation	G	A	c.185C>T	c.(184-186 p.P62L	CD200 receptor 1
BTC-6	CRYBG3	131544	3	97593426	97593426	Missense_Mutation	G	T	c.3388G>T	c.(3388-33 p.G1130W	beta-gamma crystallin domain containing 3
BTC-6	DEAF1	10522	11	694992	694992	Missense_Mutation	A	G	c.56T>C	c.(55-57)g ⁺ p.V19A	DEAF1 transcription factor
BTC-6	EFCAB5	374786	17	28295933	28295933	Missense_Mutation	A	T	c.315A>T	c.(313-315 p.E105D	EF-hand calcium binding domain 5
BTC-6	FSIP2	401024	2	186671383	186671383	Missense_Mutation	G	A	c.17350G>A	c.(17350-1 p.A5784T	fibrous sheath interacting protein 2
BTC-6	GAPVD1	26130	9	128064438	128064438	Missense_Mutation	A	T	c.362A>T	c.(361-363 p.N121I	GTPase activating protein and VPS9 domains 1
BTC-6	IQSEC2	23096 X		53349985	53349985	Missense_Mutation	C	T	c.337G>A	c.(337-339 p.D113N	IQ motif and Sec7 domain 2
BTC-6	KDM5A	5927	12	427378	427378	Missense_Mutation	C	T	c.2791G>A	c.(2791-27 p.G931R	lysine (K)-specific demethylase 5A
BTC-6	LPAR3	23566	1	85279695	85279695	Missense_Mutation	T	C	c.896A>G	c.(895-897 p.D299G	lysophosphatidic acid receptor 3
BTC-6	NLRP1	22861	17	5462767	5462767	Missense_Mutation	A	G	c.1249T>C	c.(1249-12 p.W417R	NLR family, pyrin domain containing 1
BTC-6	OR6B3	150681	2	240984973	240984973	Missense_Mutation	C	A	c.517G>T	c.(517-519 p.V173F	olfactory receptor, family 6, subfamily B, member 3
BTC-6	OTUD6A	139562 X		69282627	69282627	Missense_Mutation	G	C	c.253G>C	c.(253-255 p.E85Q	OTU deubiquitinase 6A

BTC-6	PTPRH	5794	19	55715264	55715264	Missense_Mutation	T	C	c.772A>G	c.(772-774 p.T258A	protein tyrosine phosphatase, receptor type, H
BTC-6	RAD50	10111	5	131927660	131927660	Missense_Mutation	T	G	c.1727T>G	c.(1726-17 p.L576R	RAD50 homolog (S. cerevisiae)
BTC-6	RB1	5925	13	49039417	49039417	Missense_Mutation	G	C	c.2402G>C	c.(2401-24 p.G801A	retinoblastoma 1
BTC-6	SAMD9L	219285	7	92763376	92763376	Missense_Mutation	C	T	c.1909G>A	c.(1909-19 p.A637T	sterile alpha motif domain containing 9-like
BTC-6	SERINC5	256987	5	79454696	79454696	Missense_Mutation	C	G	c.949G>C	c.(949-951 p.G317R	serine incorporator 5
BTC-6	ST6GALNA	81849	1	77515950	77515950	Missense_Mutation	T	A	c.679T>A	c.(679-681 p.S227T	ST6 (alpha-N-acetyl-neuraminyl-2,3-beta-galactosyl-1,3)-N-acetylgalactosaminide alpha-2,6-sialyltransferase 5
BTC-6	TMEM47	83604 X		34657486	34657486	Missense_Mutation	A	G	c.245T>C	c.(244-246 p.L82P	transmembrane protein 47
BTC-6	TRIM41	90933	5	180651595	180651595	Missense_Mutation	T	C	c.596T>C	c.(595-597 p.L199P	tripartite motif containing 41
BTC-6	WDSUB1	151525	2	160139525	160139525	Missense_Mutation	G	A	c.56C>T	c.(55-57)g(p.A19V	WD repeat, sterile alpha motif and U-box domain containing 1
BTC-6	ZC3H13	23091	13	46616352	46616352	Missense_Mutation	C	T	c.286G>A	c.(286-288 p.V96M	zinc finger CCCH-type containing 13
BTC-7	CCDC18	343099	1	93705341	93705341	Nonsense_Mutation	G	T	c.2866G>T	c.(2866-28 p.E956*	coiled-coil domain containing 18
BTC-7	KANSL1	284058	17	44115975	44115975	Nonsense_Mutation	G	A	c.2281C>T	c.(2281-22 p.R761*	KAT8 regulatory NSL complex subunit 1
BTC-7	PLEKHG2	64857	19	39912773	39912773	Nonsense_Mutation	G	T	c.1522G>T	c.(1522-15 p.E508*	pleckstrin homology domain containing, family G (with RhoGef domain) member 2
BTC-7	ZNF546	339327	19	40520465	40520465	Nonsense_Mutation	C	T	c.1288C>T	c.(1288-12 p.R430*	zinc finger protein 546
BTC-7	SI	6476	3	164760855	164760855	Nonsense_Mutation	C	A	c.1996G>T	c.(1996-19 p.G666*	sucrase-isomaltase (alpha-glucosidase)
BTC-7	SLITRK2	84631 X		144904042	144904042	Nonsense_Mutation	T	A	c.99T>A	c.(97-99)tg p.C33*	SLIT and NTRK-like family, member 2
BTC-7	CNTNAP5	129684	2	125367471	125367471	Missense_Mutation	C	T	c.1847C>T	c.(1846-18 p.P616L	contactin associated protein-like 5
BTC-7	DNMT3A	1788	2	25467461	25467461	Missense_Mutation	T	C	c.946A>G	c.(946-948 p.I316V	DNA (cytosine-5-)-methyltransferase 3 alpha
BTC-7	DSG4	147409	18	28971077	28971077	Missense_Mutation	G	A	c.721G>A	c.(721-723 p.D241N	desmoglein 4
BTC-7	EPC1	80314	10	32560813	32560813	Missense_Mutation	T	C	c.2038A>G	c.(2038-20 p.T680A	enhancer of polycomb homolog 1 (Drosophila)
BTC-7	ESM1	11082	5	54277867	54277867	Missense_Mutation	A	T	c.409T>A	c.(409-411 p.Y137N	endothelial cell-specific molecule 1
BTC-7	F13A1	2162	6	6266956	6266956	Missense_Mutation	C	A	c.406G>T	c.(406-408 p.V136F	coagulation factor XIII, A1 polypeptide
BTC-7	FCRL3	115352	1	157665154	157665154	Missense_Mutation	G	A	c.1376C>T	c.(1375-13 p.A459V	Fc receptor-like 3
BTC-7	FUT1	2523	19	49254093	49254093	Missense_Mutation	C	A	c.446G>T	c.(445-447 p.W149L	fucosyltransferase 1 (galactoside 2-alpha-L-fucosyltransferase, H blood group)
BTC-7	GLI3	2737	7	42005951	42005951	Missense_Mutation	C	A	c.2720G>T	c.(2719-27 p.S907I	GLI family zinc finger 3
BTC-7	HEATR1	55127	1	236721708	236721708	Missense_Mutation	C	T	c.5033G>A	c.(5032-50 p.C1678Y	HEAT repeat containing 1
BTC-7	KLHL24	54800	3	183368493	183368493	Missense_Mutation	A	G	c.349A>G	c.(349-351 p.M117V	kelch-like family member 24
BTC-7	KLHL9	55958	9	21333179	21333179	Missense_Mutation	C	G	c.1680G>C	c.(1678-16 p.M560I	kelch-like family member 9
BTC-7	KMT2A	4297	11	118392628	118392628	Missense_Mutation	T	A	c.11660T>A	c.(11659-1 p.M3887K	lysine (K)-specific methyltransferase 2A
BTC-7	KRAS	3845	12	25398284	25398284	Missense_Mutation	C	A	c.35G>T	c.(34-36)g(p.G12V	Kirsten rat sarcoma viral oncogene homolog
BTC-7	LPP	4026	3	188326976	188326976	Missense_Mutation	C	T	c.457C>T	c.(457-459 p.P153S	LIM domain containing preferred translocation partner in lipoma
BTC-7	LRP1B	53353	2	141707920	141707920	Missense_Mutation	A	G	c.3020T>C	c.(3019-30 p.F1007S	low density lipoprotein receptor-related protein 1B
BTC-7	MSH3	4437	5	79950739	79950739	Missense_Mutation	G	A	c.193G>A	c.(193-195 p.A65T	mutS homolog 3
BTC-7	NCOR1	9611	17	16049747	16049747	Missense_Mutation	T	C	c.1025A>G	c.(1024-10 p.Y342C	nuclear receptor corepressor 1
BTC-7	NIPBL	25836	5	37064917	37064917	Missense_Mutation	A	G	c.8338A>G	c.(8338-83 p.S2780G	Nipped-B homolog (Drosophila)
BTC-7	NTRK1	4914	1	156851340	156851340	Missense_Mutation	G	A	c.2297G>A	c.(2296-22 p.R766Q	neurotrophic tyrosine kinase, receptor, type 1
BTC-7	PELP1	27043	17	4575146	4575146	Missense_Mutation	C	T	c.3140G>A	c.(3139-31 p.G1047E	proline, glutamate and leucine rich protein 1
BTC-7	RELN	5649	7	103183183	103183183	Missense_Mutation	A	T	c.6666T>A	c.(6664-66 p.H2222Q	reelin

BTC-7	SPTBN1	6711	2	54871578	54871578	Missense_Mutation	G	A	c.4124G>A	c.(4123-41 p.R1375Q	spectrin, beta, non-erythrocytic 1
BTC-7	SUSD2	56241	22	24583685	24583685	Missense_Mutation	G	T	c.2038G>T	c.(2038-20 p.A680S	sushi domain containing 2
BTC-7	ZNF14	7561	19	19822422	19822422	Missense_Mutation	C	A	c.1668G>T	c.(1666-16 p.E556D	zinc finger protein 14
BTC-8	APC	324	5	112175952	112175952	Frame_Shift_Del	A	-	c.4661delA	c.(4660-46 p.E1554fs	adenomatous polyposis coli
BTC-8	LMO7	4008	13	76408475	76408475	Nonsense_Mutation	C	T	c.2479C>T	c.(2479-24 p.Q827*	LIM domain 7
BTC-8	TP53	7157	17	7574003	7574003	Nonsense_Mutation	G	A	c.1024C>T	c.(1024-10 p.R342*	tumor protein p53
BTC-8	DUXA	503835	19	57670637	57670637	Nonsense_Mutation	G	A	c.190C>T	c.(190-192 p.Q64*	double homeobox A
BTC-8	MMS22L	253714	6	97676947	97676947	Nonsense_Mutation	C	T	c.1862G>A	c.(1861-18 p.W621*	MMS22-like, DNA repair protein
BTC-8	KIF15	56992	3	44843467	44843467	Splice_Site	G	C		c.e13+1	kinesin family member 15
BTC-8	ABCA9	10350	17	67025361	67025361	Missense_Mutation	T	G	c.1453A>C	c.(1453-14 p.N485H	ATP-binding cassette, sub-family A (ABC1), member 9
BTC-8	ACSS1	84532	20	25004233	25004233	Missense_Mutation	C	T	c.676G>A	c.(676-678 p.G226R	acyl-CoA synthetase short-chain family member 1
BTC-8	ADAMTS6	11174	5	64595872	64595872	Missense_Mutation	G	A	c.1310C>T	c.(1309-13 p.A437V	ADAM metalloproteinase with thrombospondin type 1 motif, 6
BTC-8	ADORA1	134	1	203098180	203098180	Missense_Mutation	A	G	c.211A>G	c.(211-213 p.I71V	adenosine A1 receptor
BTC-8	AOC3	8639	17	41006705	41006705	Missense_Mutation	C	T	c.1841C>T	c.(1840-18 p.P614L	amine oxidase, copper containing 3
BTC-8	CPZ	8532	4	8603105	8603105	Missense_Mutation	G	A	c.344G>A	c.(343-345 p.R115Q	carboxypeptidase Z
BTC-8	DACT3	147906	19	47151783	47151783	Missense_Mutation	G	C	c.1846C>G	c.(1846-18 p.L616V	dishevelled-binding antagonist of beta-catenin 3
BTC-8	ENPP2	5168	8	120577135	120577135	Missense_Mutation	A	C	c.1097T>G	c.(1096-10 p.V366G	ectonucleotide pyrophosphatase/phosphodiesterase 2
BTC-8	INO80D	54891	2	206870153	206870153	Missense_Mutation	C	A	c.2023G>T	c.(2023-20 p.A675S	INO80 complex subunit D
BTC-8	IRX3	79191	16	54319378	54319378	Missense_Mutation	T	C	c.415A>G	c.(415-417 p.T139A	iroquois homeobox 3
BTC-8	KRAS	3845	12	25398281	25398281	Missense_Mutation	C	T	c.38G>A	c.(37-39)g(p.G13D	Kirsten rat sarcoma viral oncogene homolog
BTC-8	KRT73	319101	12	53004589	53004589	Missense_Mutation	C	T	c.1141G>A	c.(1141-11 p.A381T	keratin 73
BTC-8	LAX1	54900	1	203743063	203743063	Missense_Mutation	G	A	c.451G>A	c.(451-453 p.A151T	lymphocyte transmembrane adaptor 1
BTC-8	MATN4	8785	20	43927089	43927089	Missense_Mutation	G	A	c.1147C>T	c.(1147-11 p.R383W	matrilin 4
BTC-8	NAB1	4664	2	191524551	191524551	Missense_Mutation	G	A	c.649G>A	c.(649-651 p.E217K	NGFI-A binding protein 1 (EGR1 binding protein 1)
BTC-8	NPR3	4883	5	32712527	32712527	Missense_Mutation	C	A	c.645C>A	c.(643-645 p.F215L	natriuretic peptide receptor 3
BTC-8	OTOG	340990	11	17655819	17655819	Missense_Mutation	A	G	c.7288A>G	c.(7288-72 p.T2430A	otogelin
BTC-8	PCDHGB6	56100	5	140788665	140788665	Missense_Mutation	G	A	c.896G>A	c.(895-897 p.G299D	protocadherin gamma subfamily B, 6
BTC-8	POLA1	5422 X		24906154	24906154	Missense_Mutation	G	A	c.4061G>A	c.(4060-40 p.R1354H	polymerase (DNA directed), alpha 1, catalytic subunit
BTC-8	RALGAPA1	253959	14	36096402	36096402	Missense_Mutation	T	A	c.5272A>T	c.(5272-52 p.M1758L	Ral GTPase activating protein, alpha subunit 1 (catalytic)
BTC-8	RIPK2	8767	8	90792331	90792331	Missense_Mutation	G	C	c.882G>C	c.(880-882 p.L294F	receptor-interacting serine-threonine kinase 2
BTC-8	ROBO1	6091	3	78710287	78710287	Missense_Mutation	A	G	c.2213T>C	c.(2212-22 p.I738T	roundabout, axon guidance receptor, homolog 1 (Drosophila)
BTC-8	ROBO2	6092	3	77657029	77657029	Missense_Mutation	C	A	c.3217C>A	c.(3217-32 p.P1073T	roundabout, axon guidance receptor, homolog 2 (Drosophila)
BTC-8	SHISA8	440829	22	42310285	42310285	Missense_Mutation	C	A	c.286G>T	c.(286-288 p.A96S	shisa family member 8
BTC-8	SMARCA2	6595	9	2104152	2104152	Missense_Mutation	T	C	c.3275T>C	c.(3274-32 p.L1092P	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 2
BTC-8	SSTR1	6751	14	38678956	38678956	Missense_Mutation	G	A	c.362G>A	c.(361-363 p.R121H	somatostatin receptor 1
BTC-8	SYN3	8224	22	33265027	33265027	Missense_Mutation	C	T	c.547G>A	c.(547-549 p.G183S	synapsin III
BTC-8	TROVE2	6738	1	193046115	193046115	Missense_Mutation	C	G	c.1021C>G	c.(1021-10 p.L341V	TROVE domain family, member 2
BTC-8	USH2A	7399	1	215844423	215844423	Missense_Mutation	C	T	c.14024G>A	c.(14023-1 p.R4675K	Usher syndrome 2A (autosomal recessive, mild)
BTC-8	USH2A	7399	1	216062042	216062042	Missense_Mutation	G	C	c.7949C>G	c.(7948-79 p.P2650R	Usher syndrome 2A (autosomal recessive, mild)
BTC-8	XCR1	2829	3	46062623	46062623	Missense_Mutation	G	A	c.817C>T	c.(817-819 p.R273C	chemokine (C motif) receptor 1
BTC-8	ZSCAN20	7579	1	33960845	33960845	Missense_Mutation	C	A	c.2901C>A	c.(2899-29 p.F967L	zinc finger and SCAN domain containing 20