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Genomic characterization of Chinese ovarian clear cell carcinoma identifies driver genes by whole exome sequencing

Qin Yang [a,b,1]; Cancan Zhang [a,1]; Yuan Ren [a,1]; Huan Yi [a,c,1]; Tianjiao Luo [b,1]; Fangliang Xing [d]; Xuefeng Bai [e]; Lining Cui [f]; Linyan Zhu [f]; Jun Ouyang [g]; Pengcheng Jiang [h]; Weirong Fan [a]; Jianping Qiu [i]; Fengmian Wang [a]; Xin Xing [a]; Zhigang Zhang [b]; Xueli Zhang [b,*]; Rong Zhang [a,*]

[a] Fengxian Hospital, The Third School of Clinical Medicine, Southern Medical University, Shanghai, PR China; [b] State Key Laboratory of Oncogenes and Related Genes, Shanghai Cancer Institute, Ren Ji Hospital, School of Medicine, Shanghai Jiao Tong University, Shanghai, PR China; [c] Fujian Provincial Maternity and Children's Hospital, Affiliated Hospital of Fujian Medical University, Fujian, PR China; [d] Department of Pathology, Tongliao City Hospital West District, Tongliao, Inner Mongolia, PR China; [e] Department of Pathology, Baotou Medical College Affiliated Baotou Cancer Hospital, Baotou, Inner Mongolia, PR China; [f] Department of Obstetrics and Gynecology, Ningbo First Hospital, Ningbo Hospital of Zhejiang University, Ningbo, Zhejiang, PR China; [g] Department of Obstetrics and Gynecology, Nanjing Medical University Affiliated Changzhou Maternal and Child Health Care Hospital, Changzhou, Jiangsu, PR China; [h] Department of Obstetrics and Gynecology, Nanjing Medical University Affiliated Changzhou No.2 People's Hospital, Changzhou, Jiangsu, PR China; [i] Department of Obstetrics and Gynecology, Nanjing Medical University Affiliated Suzhou Munipical Hospital, Suzhou, Jiangsu, PR China

Abstract

Little is known about the genetic alterations characteristic of ovarian clear cell carcinoma (OCCC). Our aim was to identify targetable genomic alterations in this type of cancer. Forty-two OCCC formalin-fixed, paraffin-embedded (FFPE) tissue samples were analyzed by whole-exome sequencing (WES), and 74 FFPE tissue samples underwent targeted sequencing (TS) to confirm the relevant driver mutations. Cell proliferation was assessed by cell counting kit-8 (CCK8) assays. In the 42 samples, ARID1A (64.3%) and PIK3CA (28.5%) were frequently mutated, as were PPP2R1A (11.9%), PTEN (7.1%) and KRAS (4.8%), which have been reported in previous OCCC studies. We also detected mutations in MUC4 (28.6%), MAGEE1 (19%), and ARID3A (16.7%); associations with these genes have not been previously reported. The functional protein-activated pathways were associated with proliferation and survival (including the PI3K/AKT, TP53, and ERBB2 pathways) in 83% of OCCCs and with chromatin remodeling in 71% of OCCCs. Patients with alterations in MAGEE1 (64% in the targeted sequencing cohort) had worse clinical outcomes (log-rank $p < 0.05$). A functional study revealed that two MAGEE1 mutants, one lacking two MAGE domains and the other containing two MAGE domains, significantly decreased the proliferative capacity of OCCC cells. We successfully identified novel genetic alterations in OCCC using whole-exome sequencing and targeted sequencing of OCCC patient samples and potential therapeutic targets for the treatment of this malignancy.

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Keywords: Ovarian clear cell carcinomas, Driver mutation, MAGEE1, Whole exome sequencing, Target sequencing

Introduction

Among gynecologic malignancies, ovarian cancer is the second most common and the most deadly globally in 2020 cancer statistics [1]. Epithelial ovarian cancer accounts for over 80% of the malignant ovarian cancer cases [2] and consists of four major histological tumor subtypes, including serous, clear-cell, endometrioid, and mucinous [3]. Among the histological subtypes, ovarian clear cell carcinoma (OCCC) accounts for approximately 5% of all epithelial ovarian cancers, with an occurrence rate above 20% in certain Asian populations [4]. In addition, women with

* Corresponding authors.
e-mail addresses: xlzhang@shsci.org (X. Zhang), rongzhang@163.com (R. Zhang).
[1] These authors contributed equally to this work.

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advanced OCCC have poorer survival outcomes. OCCC is a distinct subtype with a lower response rate than the serous subtype to platinum-based chemotherapy [5–7]. Although OCCC is the second leading cause of death from ovarian cancer, the etiology and pathogenesis of this devastating disease are poorly understood.

The molecular characterization of solid tumors by whole genome or exome sequencing has provided important insights in cancer biology. Somatic mutations found in cancer may suggest personalized treatments options. OCCC-specific somatic mutations are clustered in AT-rich interactive domain 1A (SWI-like) (*ARID1A*), phosphatidylinositol-4, 5-bisphosphate 3-kinase, catalytic subunit alpha (*PIK3CA*), protein phosphatase 2 scaffold subunit alpha (*PPP2R1A*) and K-ras (*KRAS*) [8–12]. Somatic copy number amplification of the *ZNF217* gene in OCCC was also previously reported [13].

In this study, we obtained whole-exome sequencing (WES) data for Asian OCCC samples via next-generation sequencing (NGS) and integrated the independent single-nucleotide variant (SNV) and copy number variant (CNV) analyses to better elucidate the genomic architecture of our OCCC samples. To detect both high- and low-frequency pathogenic mutations, we performed targeted sequencing of a large cohort of 69 OCCC tumors and matched normal tissues. Finally, we validated recurrent mutants in several genes, including *MAGEE1* in OCCC. Patients with alterations in *MAGEE1* had worse clinical outcomes. The expression of mutant MAGEE1 significantly decreased the proliferative capacity of OCCC cells. These results indicate that MAGEE1-targeted drugs could guide the future development of therapeutic strategies for OCCC.

Materials and methods

Study population

Patients enrolled in this study were pathologically diagnosed with ovarian clear cell carcinoma between January 2008 and December 2016. Tissue samples were collected from formalin-fixed, paraffin-embedded (FFPE) blocks of tumor tissue from 69 ovarian clear cell cancer patients. The use of samples and medical records was approved by the research ethics committees of Shanghai University of Medicine & Health Sciences Affiliated with Sixth People's Hospital South Campus (approval number: 2017-KY-01), Fujian Provincial Maternity and Children's Hospital (approval number: 2017049), Nanjing Medical University Affiliated with Changzhou Maternal and Child Health Care Hospital (approval number: 2017005), Nanjing Medical University Affiliated with Changzhou No. 2 People's Hospital (approval number: 2016-017-01), and Nanjing Medical University Affiliated with Suzhou Municipal Hospital (approval number: L2017003). Genomic DNA was extracted from tumor areas of tissue sections from the FFPE blocks using the QIAGEN GeneRead DNA FFPE Kit (ID: 762174; this kit helps reduce errors due to DNA deamination caused by formalin fixation and aging).

Whole exome sequencing

Sequencing data were generated as detailed previously. In brief, whole-exome capture libraries were constructed from tumor and normal DNA after sample shearing, end repair, phosphorylation, and ligation to barcoded sequencing adaptors. DNA then underwent solution-phase hybrid capture with SureSelect v.2 Exome bait (Agilent Technologies), followed by sample multiplexing and sequencing on an Illumina HiSeq X Ten instrument. Raw sequencing reads were trimmed with Trimmomatic to filter low-quality reads. Clean reads were aligned to the reference human genome (UCSC, hg19) using Burrows-Wheeler Aligner (BWA). Duplicates were identified by Picard, and the remaining outputs were locally realigned using the Genome Analysis Toolkit (GATK). We detected somatic mutations with the MuTect algorithm and somatic indels based on concordant events identified by the Indelocator algorithm. To remove artifacts from the hydrolytic deamination of cytosine to uracil in FFPE samples, we filtered out C > T mutations consistent with a 20:1 single-strand bias based on read pair orientation.

We used the eDriver, OncodriveFML, ActiveDriver, MutSigCV, Genome MuSic and OncodriveCLUST tools to infer significantly mutated genes. Hypermutated tumors are defined as those with a mutation count >1000. All somatic variants were annotated in dbSNP138, the 1000 Genomes Project and EXAC by ANNOVAR. Variants with allele frequencies greater than 0.5% of the allelic fraction in these databases were removed.

Validation with target sequencing

For targeted ultradeep sequencing of 56 genes, the Illumina HiSeq platform was used. After samples were library prepped and run on the HiSeq platform, reads were mapped to hg19 using BWA, and realigned using GATK. Variants were called using GATK. For targeted ultradeep sequencing, we required a depth $\geq$200 and a quality score $\geq$20.

Pathway enrichment analysis

Canonical cancer pathways were selected from MSigDB (including 1329 gene sets). The list of genes with recurrent mutations, including functional SNVs and CNVs, was analyzed.

Pathway enrichment analyses of genes harboring somatic SNVs and CNVs were performed with KEGG or Gene Ontology by using the clusterProfiler package in R. P-values were calculated based on a hypergeometric distribution with FDR correction using the Benjamini method.

Cell culture

The OCCC cell lines OVISE and ES-2 were both preserved in Shanghai Cancer Institute, Ren Ji Hospital, School of Medicine, Shanghai Jiao Tong University. OVISE was cultured in RPMI 1640 medium, and ES-2 cells were cultured in McCoy's 5A medium, containing 10% fetal bovine serum (FBS) and 1% antibiotics (100 μg/ml streptomycin and 100 units/ml penicillin). All cells were incubated at 37 °C in a humidified atmosphere containing 5% CO_2.

Plasmid construction and cell transfection

The MAGEE1 wild-type (MAGEE1-WT) sequence was obtained from Asia-vector Biotechnology Co., Ltd (Shanghai, China). Two mutants were constructed, mut1 lacking two MAGE domains and mut2 containing two MAGE domains. The hemagglutinin (HA) tag was added to the C terminus of the MAGEE1 mutants. All constructs were verified by sequencing. Cell transduction steps were performed according to the manufacturer's protocols using Lipofectamine™ 3000 Transfection Reagent (Thermo Fisher Scientific).

Quantitative Real-Time PCR (qPCR)

Total RNA was extracted from OCCC cell lines by using TRIzol (Invitrogen, Carlsbad, CA, USA). Reverse transcription was performed as previously described [14]. β-Actin was used as internal control for quantification. The data were analyzed using the $2^{-\Delta\Delta Ct}$ method. The primer sequences used in our study were as follows: MAGEE1-F, 5′-CCCAGAAGAGGTGACGG-3′; MAGEE1-R, 5′-GCAGCAGGAAGATG AGGA-3′; β-Actin-F, 5′-CTCTGGCTCCTAGCACCATGAAGA-3′; β-Actin-R, 5′-GTAAAACGCAGCTCAGTAACAGTCCG-3′.

Western blot

Western blot analysis was performed as described previously [14]. The membranes were incubated overnight at 4 °C with primary antibodies against HA (ab18181; Abcam) and β-actin (M1210-1, Huabio, Hangzhou, China), followed by incubation with species-specific secondary antibodies for 1 h. The signals were detected by an Odyssey infrared imaging system (LI-COR, Lincoln, NE) and further quantified by ImageJ software.

Cell viability

To measure cell proliferation, cells were seeded at 2000 cells/well in a 96-well plate and detected by Cell Counting Kit-8 (CCK8, Dojindo, Japan) after 0, 1, 2, 3, and 4 days. The experiments were performed in triplicate and repeated twice.

Statistical analysis

Known prognostic factors for OCCC were recorded for each patient, as shown in Table 1. The characteristics of patients in different groups were compared using Fisher's exact test (for categorical data) or the Mann-Whitney test (for continuous data). Only 57 patients with complete follow-up clinical data were included in the survival analysis. OS was calculated based from the date of OCCC diagnosis to the data of death from disease or the last follow-up. The Kaplan-Meier method was used to estimate the OS distribution, and differences in survival between groups were assessed using the log rank test. P values less than 0.05 indicated statistical significance. R software was used to perform all statistical analyses and generate all graphs.

Results

Clinical cohort

A cohort of 69 patients diagnosed with ovarian clear cell cancer and treated surgically from 2008 to 2016 was recruited. Ovarian tumor tissue samples and matched solid normal tissue samples were obtained. This cohort was included in the WES cohort (n = 42) and the targeted sequencing cohort (n = 69 including the WES cohort). The detailed clinical pathological characteristics of the patients in the cohorts, including age, tumor stage, chemotherapy, the presence of endometriosis and survival, are presented in Table 1. The median age at diagnosis was 52 years, but a notable percentage of the patients (10%) were younger than 40 years at diagnosis. The tumor was stage I in 45 patients, stage II in 12 subjects and stage III/IV in 12 individuals.

Mutation detection analysis

To detect somatic mutations in tumor samples, whole-exome sequencing was performed on 84 samples (42 ovarian tissues, including both cancer and adjacent normal tissues). For the tumor and matched normal DNA samples, each targeted base was sequenced by a mean of 180 independent reads. A total of 5868 somatic exonic mutations were identified in all tumor samples, including nonsynonymous variants, in_frameshift variants, frameshift variants, nonsense variants and splice site variants, with a median of 153 alterations per tumor (range, 44–794).

The number of somatic mutations was higher in patients with stage III/IV disease than in patients with stage I/II disease (P = 0.039; Wilcoxon test) (1). However, no correlation was observed between the number of somatic mutations and patient age (group 1 ≤52 years; group 2 >52 years; P = 0.31) or the presence of endometriosis (P = 0.50) (Fig. S1).

The previously reported hotspot genes with somatic mutations in OCCC included *ARID1A*, *PIK3CA*, *KRAS*, *PPP2R1A*, *PTEN*, *MLL3*, *ARID1B* and *PIK3R1* (Fig. S2). In total, 63 mutations were discovered across these 8 genes in 42 OCCCs (Table S1). Of these, *ARID1A* and PIK3CA were the two most frequently mutated genes in all patients. *ARID1A* accounted for 34 mutations in 27 patients, which was consistent with a previous report, but these mutations were scattered along the entire length of the genes. *PIK3CA* accounted for 14 mutations in 12 patients, and 4 of these patients had *PIK3CA* hotspot mutations (c.1624G > A [p. Glu542Lys] and c.1633G > A [p. Glu545Lys]).

Genes predicted by more than one method may be more likely to be drivers. By applying six independent mutation prediction algorithms, eDriver (Table S2), OncodriveFML (Table S3), ActiveDriver (Table S4), MutSigCV (Table S5), Genome MuSic (Table S6) and OncodriveCLUST (Table S7), we computed the significance levels of the frequently mutated genes, accounting for the gene size and the background mutation rate. Fig. 1a shows the most significantly mutated genes identified by more than two cancer driver detection methods in our panel of 43 cases, with their mutation frequencies derived from whole-exome sequencing; these genes included *ARID1A*, *MAGEE1*, *PIK3CA*, *MUC4*, *ARID3A*, *FLG2*, *TCHH*, *GRM3*, *MUC17*, *ZNF208* and *GAGE12J*. In addition to *ARID1A* and *PIK3CA*, other genes were newly identified as novel SMGs in our study. The frequencies of significant somatic mutations in the oncogenes *MUC4* and *ARID3A* were 28.6% and 16.7%, respectively. *MAGEE1*, *FLG2*, *TCHH*, *GRM3*, *MUC17*, *ZNF208*, and *GAGE12J* were mutated in 19.0%, 26.2%, 23.8%, 4.8%, 23.8%, 9.5% and 9.5% of samples, respectively (Fig. 1b).

The mutation spectrum revealed C: G > T: A transitions (69.41%), as the most abundant alteration, and other transversions, including C: G > G: C (6.91%), T: A > C: G (7.83%), C: G > A: T (1.02%), T: A > G: C (2.25%) and T: A > A: T (3.37%) (Fig. 1c and d). Using known mutational signatures reported in the COSMIC database, the most frequent mutation signature was signature 6 (likely with defective DNA mismatch repair), which is most frequently found in colorectal and uterine cancers. The second most common signature observed was signature 1, a pattern associated with spontaneous deamination of 5-methylcytosine that accounts for a significant percentage of the critical somatic driver mutations observed in most cancers, including OVs (Fig. 1e).

Validation with target sequencing

To further validate the frequently mutated genes found in the WES cohort, we performed targeted sequencing of all 69 tumor samples. The targeted gene panel included over 56 previously identified genes that are prone to mutation and involved in OCCC (Table S8). This panel is composed of genes from four categories: MutSigCV-positive cancer driver genes (6 genes), genes with a high variant frequency (19 genes), previously reported genes (9 genes) and other candidate genes (22 genes). We analyzed tumor DNA from a total of 138 OCCC samples (ovarian tissues including both cancer and adjacent normal tissues). After quality control for library preparation, 124 tumors (62 tumor samples) were suitable for analysis. On average, 96% of the reads mapped uniquely to the targeted sequences, and we obtained an average read depth of 800 reads per base in the targeted region.

After filtering out changes in intronic regions and polymorphisms (present in dbSNP134), we identified a total of 1816 variants in all sequenced exons of the 62 OCCC patients. After discarding alterations in noncoding RNAs or the 5′ or 3′ untranslated regions, and synonymous mutations, only 1096 single nucleotide variants and indels were identified that resulted in missense mutations, frameshift mutations, inframe mutations or nonsense mutations, with an average of 17.62 mutations per case (Table 2). *AHNAK2* was the most frequently mutated gene (60.3%), followed by *OBSCN*

Table 1. The clinical pathological characteristics in OCCC patients.

Sample	Age	Grade	Status	OS	Chemotherapy	Endometriosis	WES	Target
OCCC_01	64	II	Alive	2124	Y	N	Y	Y
OCCC_02	35	I	Alive	1946	Y	N	Y	Y
OCCC_03	50	IC	Alive	933	Y	N	Y	Y
OCCC_04	52	IA	Alive	455	Y	Y	Y	Y
OCCC_05	59	IIIA	Dead	902	Y	Y	Y	Y
OCCC_06	45	IA	Alive	455	Y	N	Y	Y
OCCC_07	43	IIC	Dead	908	Y	N	Y	Y
OCCC_08	56	IA	Alive	455	Y	Y	Y	Y
OCCC_09	59	IC	Alive	1978	Y	N	Y	Y
OCCC_10	60	IC	Alive	360	Y	N	Y	Y
OCCC_11	46	IIC	Dead	1234	Y	N	Y	Y
OCCC_12	50	IC	Alive	392	Y	N	Y	Y
OCCC_13	43	IC	Alive	850	N	Y	Y	Y
OCCC_14	48	IC	Alive	265	Y	Y	Y	Y
OCCC_15	53	I	Alive	758	Y	Y	Y	Y
OCCC_16	49	II	Dead	545	Y	Y	Y	Y
OCCC_17	50	IC	Alive	708	Y	Y	Y	Y
OCCC_18	50	II	Alive	568	N	N	Y	Y
OCCC_19	64	IA	Alive	1551	Y	N	Y	Y
OCCC_20	53	II	Alive	600	Y	Y	Y	Y
OCCC_21	53	I	Alive	517	Y	Y	Y	Y
OCCC_22	40	IA	Alive	600	Y	N	Y	Y
OCCC_23	33	IA	Alive	517	Y	N	Y	Y
OCCC_24	63	IA	Alive	517	Y	N	Y	Y
OCCC_25	37	IIIC	Dead	336	Y	Y	Y	Y
OCCC_26	44	IC	Alive	1267	Y	N	Y	Y
OCCC_27	56	IC	Dead	631	Y	N	Y	Y
OCCC_28	52	IB	Alive	423	N	N	Y	Y
OCCC_29	70	IIB	Dead	301	N	N	Y	Y
OCCC_30	61	IC	Alive	455	Y	N	Y	Y
OCCC_31	38	IA	Alive	1946	Y	N	Y	Y
OCCC_32	51	IA	Alive	933	Y	N	Y	Y
OCCC_33	65	III	Alive	1580	Y	N	Y	Y
OCCC_34	47	IA	Alive	789	Y	N	Y	Y
OCCC_35	40	IC	Alive	1393	Y	Y	Y	Y
OCCC_36	54	IC	Alive	423	Y	Y	Y	Y
OCCC_37	32	IC	Dead	268	Y	Y	Y	Y
OCCC_38	58	IIIC	Alive	1293	Y	N	Y	Y
OCCC_39	54	IIIC	Alive	1632	Y	Y	Y	Y
OCCC_40	41	IIIB	Alive	392	Y	Y	Y	Y
OCCC_41	74	IIIC	Dead	63	N	N	Y	Y
OCCC_42	51	IIIC	Dead	533	Y	Y	Y	Y
OCCC_43	56	IIIB	Dead	122	N	N	N	Y
OCCC_44	66	II	Alive	392	Y	N	N	Y
OCCC_45	81	IB	Dead	2854	N	N	N	Y
OCCC_46	54	IIIC	Alive	2584	Y	N	N	Y
OCCC_47	42	IA	Alive	1821	Y	N	N	Y
OCCC_48	45	I	Alive	3282	Y	Y	N	Y
OCCC_49	68	IIIC	Dead	300	Y	N	N	Y
OCCC_50	54	IA	Alive	1361	Y	N	N	Y
OCCC_51	54	IA	Alive	1361	Y	N	N	Y
OCCC_52	43	IIC	Alive	1519	Y	Y	N	Y
OCCC_53	55	IC	Alive	1393	Y	N	N	Y
OCCC_54	58	IA	Alive	1884	Y	N	N	Y
OCCC_55	34	IA	Alive	2708	Y	N	N	Y
OCCC_56	53	I	Alive	2394	Y	Y	N	Y
OCCC_57	53	II	Alive	758	Y	Y	N	Y
OCCC_58	57	IIB	Alive	2310	Y	N	N	Y
OCCC_59	45	I	Alive	2311	Y	Y	N	Y
OCCC_60	46	IC	Alive	1695	Y	N	N	Y
OCCC_61	60	IA	Alive	600	Y	N	N	Y
OCCC_62	57	IC	Alive	663	Y	N	N	Y
OCCC_63	50	III	Alive	1425	Y	N	N	Y
OCCC_64	45	IC	Dead	724	Y	Y	N	Y
OCCC_65	54	IC	Alive	600	Y	N	N	Y
OCCC_66	30	IB	Alive	663	Y	N	N	Y
OCCC_67	51	IA	Alive	2886	Y	Y	N	Y
OCCC_68	47	II	Alive	2332	Y	N	N	Y
OCCC_69	64	I	Alive	120	Y	Y	N	Y

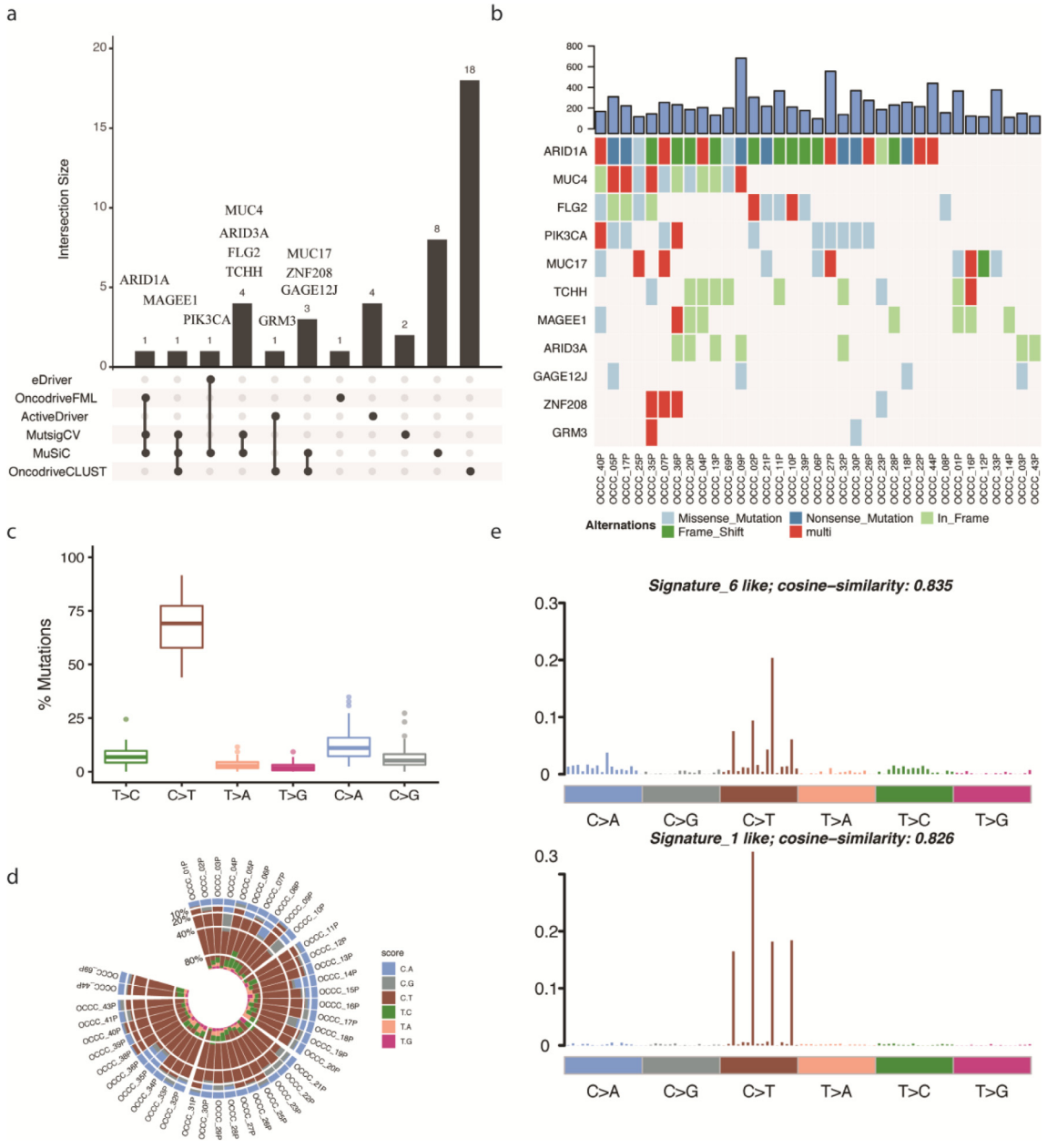

Fig. 1. Mutations identified by whole-exome sequencing in OCCC patients. (a) Eleven SMGs identified in 42 OCCC samples. Venn diagram of the overlap of the significantly mutated genes as assessed by eDriver, OncodriveFML, ActiveDriver, MutSigCV, Genome MuSic and OncodriveCLUST. (b) Mutation oncoprint of the 11 SMGs identified by at least two of the three methods. (c) Distribution of specific nucleotide changes among somatic variations by whole exome sequencing in cancer tissues compared to adjacent tissues. (d) The bars show the percentage of somatic single nucleotide variations identified in each sample. (e) Two mutational signatures were observed in the genomes of OCCC samples: a defective DNA mismatch repair signature (signature 6) and an age-associated signature (signature 1). These data were adapted from COSMIC database (http://cancer.sanger.ac.uk/cosmic/signatures).

(55.5%), *ARID1A* (54.0%), and *AHNAK* (49.2%), *MUC19* (46.0%), *DSPP* (42.9%), *PIK3CA* (41.3%), *FLG* (41.3%) and *MUC17* (41.3%) (Fig. S2). In *ARID1A*, we identified 45 heterozygous variants (21 frameshift, 11 nonsense, 7 missense changes, 3 in frame variants and 3 splice site variants) spread across the coding exons of the gene in 33 patients. Heterozygous mutations were also identified in PIK3CA, and these mutations mostly clustered in exons 10 and 21. The majority of the *PIK3CA* mutations were missense mutations ($n = 26$). In *MAGEE1*, we identified 13 heterozygous variants including 7 inFrameDel mutations, 4 missense mutations, 1 frameshiftIns mutation and 1 inFrameIn mutation (Fig. S3).

Copy number variant analysis

We next applied the software tool Control-FREEC to detect unique CNVs in 42 ovarian clear cell tumors. The merged copy number pattern of the 42 patients showed arm-level and focal SCNAs across all chromosomes (Fig. 2). While many small amplified/deleted regions were detected across the genome, there were five large blocks of amplifications (spanning >1 Mb) involving chr8q, which includes MYC; chr20q, which includes ZNF217; and chr17q, which includes ERBB2, PPP1R1B, and TBC1D3. Other loci, including the PIK3CA, EIF3E and CDH17 loci, were also

Table 2. Gene mutations identified by targeted sequencing in 62 patients with OCCC.

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
ARID1A	1	27087346	27087346	+	Splice_Site	G	T	OCCC_22	
OBSCN	1	228528833	228528833	+	Missense_Mutation	G	A	OCCC_22	p.R5912H
MSH3	5	79950724	79950724	+	Missense_Mutation	G	C	OCCC_22	p.A60P
MUC17	7	100683482	100683482	+	Missense_Mutation	C	T	OCCC_22	p.P2929S
AHNAK	11	62296059	62296059	+	Missense_Mutation	C	A	OCCC_22	p.V1944L
AHNAK2	14	105413318	105413318	+	Missense_Mutation	C	T	OCCC_22	p.G2824R
AHNAK2	14	105413471	105413471	+	Missense_Mutation	T	C	OCCC_22	p.K2773E
TCHH	1	152083777	152083777	+	Missense_Mutation	T	G	OCCC_37	p.Q639P
FLG	1	152277704	152277704	+	Missense_Mutation	C	G	OCCC_37	p.D3220H
FLG	1	152281635	152281635	+	Missense_Mutation	C	A	OCCC_37	p.R1909S
OBSCN	1	228462377	228462377	+	Missense_Mutation	G	A	OCCC_37	p.V1930M
OBSCN	1	228468087	228468087	+	Missense_Mutation	C	T	OCCC_37	p.A2624V
DSPP	4	88537412	88537412	+	Missense_Mutation	G	A	OCCC_37	p.D1200N
PTPRN2	7	157959931	157959931	+	Missense_Mutation	G	A	OCCC_37	p.A201V
PLEC	8	144998243	144998243	+	Missense_Mutation	G	A	OCCC_37	p.R2089C
PLEC	8	145024703	145024703	+	Missense_Mutation	G	A	OCCC_37	p.R58W
AHNAK	11	62293968	62293968	+	Missense_Mutation	C	T	OCCC_37	p.V2641M
MUC19	12	40873781	40873781	+	Missense_Mutation	T	C	OCCC_37	p.S1776P
MUC19	12	40873989	40873989	+	Missense_Mutation	C	T	OCCC_37	p.S1845L
AHNAK2	14	105407327	105407327	+	Missense_Mutation	G	A	OCCC_37	p.P4821S
AHNAK2	14	105407525	105407525	+	Missense_Mutation	G	A	OCCC_37	p.P4755S
AHNAK2	14	105413516	105413516	+	Missense_Mutation	C	G	OCCC_37	p.V2758L
AHNAK2	14	105413517	105413517	+	Missense_Mutation	G	C	OCCC_37	p.N2757K
AHNAK2	14	105413791	105413791	+	Missense_Mutation	G	A	OCCC_37	p.S2666F
AHNAK2	14	105419549	105419549	+	Missense_Mutation	G	T	OCCC_37	p.P747T
AHNAK2	14	105419610	105419610	+	Missense_Mutation	C	G	OCCC_37	p.Q726H
CASKIN1	16	2231127	2231127	+	Missense_Mutation	C	T	OCCC_37	p.G748S
ERBB2	17	37863287	37863287	+	Missense_Mutation	G	A	OCCC_37	p.E40K
SPTBN4	19	41062992	41062992	+	Missense_Mutation	G	A	OCCC_37	p.A1785T
PPP2R1A	19	52715982	52715982	+	Missense_Mutation	C	T	OCCC_37	p.R183W
LAMA5	20	60909579	60909579	+	Splice_Site	C	A	OCCC_37	p.E861*
HRNR	1	152191230	152191230	+	Missense_Mutation	G	T	OCCC_50	p.Q959K
OBSCN	1	228474615	228474615	+	Missense_Mutation	G	A	OCCC_50	p.R3140Q
PIK3CA	3	178952085	178952085	+	Missense_Mutation	A	G	OCCC_50	p.H1047R
DSPP	4	88536737	88536737	+	Missense_Mutation	G	A	OCCC_50	p.D975N
PLEC	8	145009181	145009181	+	Missense_Mutation	T	A	OCCC_50	p.N412Y
EVPL	17	74010545	74010545	+	Missense_Mutation	C	A	OCCC_50	p.G779C
HELZ2	20	62194375	62194375	+	Missense_Mutation	G	A	OCCC_50	p.R1934W
ARID1A	1	27105565	27105565	+	Nonsense_Mutation	G	T	OCCC_04	p.E1726*
ARID1A	1	27106159	27106159	+	Missense_Mutation	G	A	OCCC_04	p.E1924K
DST	6	56357781	56357781	+	Missense_Mutation	C	T	OCCC_04	p.R6514Q
AHNAK2	14	105413284	105413284	+	Missense_Mutation	G	A	OCCC_04	p.S2835L
SPTA1	1	158592867	158592867	+	Missense_Mutation	C	T	OCCC_47	p.R2009H
SPTA1	1	158617396	158617396	+	Missense_Mutation	G	A	OCCC_47	p.R1277C
OBSCN	1	228462332	228462332	+	Missense_Mutation	G	A	OCCC_47	p.E1915K
OBSCN	1	228464316	228464316	+	Missense_Mutation	C	G	OCCC_47	p.P2129R
OBSCN	1	228468069	228468069	+	Missense_Mutation	G	A	OCCC_47	p.S2618N
OBSCN	1	228475581	228475581	+	Missense_Mutation	G	A	OCCC_47	p.R3244Q
OBSCN	1	228503679	228503679	+	Missense_Mutation	A	G	OCCC_47	p.T4382A
OBSCN	1	228506756	228506756	+	Missense_Mutation	C	T	OCCC_47	p.S4768L
OBSCN	1	228509733	228509733	+	Missense_Mutation	C	T	OCCC_47	p.A5064V
OBSCN	1	228555619	228555619	+	Missense_Mutation	A	T	OCCC_47	p.Y6554F
OBSCN	1	228559933	228559933	+	Missense_Mutation	A	T	OCCC_47	p.T7152S
OBSCN	1	228562312	228562312	+	Missense_Mutation	C	T	OCCC_47	p.R7508C
OBSCN	1	228564758	228564758	+	Missense_Mutation	G	A	OCCC_47	p.R7682H
OBSCN	1	228566387	228566387	+	Missense_Mutation	G	A	OCCC_47	p.R7933Q
PIK3CA	3	178916623	178916623	+	Nonsense_Mutation	C	T	OCCC_47	p.R4*
MUC4	3	195511070	195511070	+	Missense_Mutation	C	G	OCCC_47	p.D2461H
MUC4	3	195511208	195511208	+	Missense_Mutation	T	G	OCCC_47	p.T2415P
MUC4	3	195513530	195513530	+	Missense_Mutation	A	C	OCCC_47	p.L1641V
SHROOM3	4	77662408	77662408	+	Missense_Mutation	G	A	OCCC_47	p.E1028K
DST	6	56357740	56357740	+	Missense_Mutation	C	T	OCCC_47	p.E6528K
MUC17	7	100683993	100683993	+	Missense_Mutation	C	A	OCCC_47	p.T3099N
AHNAK	11	62299325	62299325	+	Missense_Mutation	T	G	OCCC_47	p.K855T
SPTBN2	11	66457571	66457571	+	Missense_Mutation	G	A	OCCC_47	p.R1917W
SPTBN2	11	66478182	66478182	+	Missense_Mutation	G	A	OCCC_47	p.S315L
SPTBN2	11	66478411	66478411	+	Missense_Mutation	C	T	OCCC_47	p.V288M

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
KRAS	12	25398284	25398284	+	Missense_Mutation	C	T	OCCC_47	p.G12D
MUC19	12	40838017	40838017	+	Missense_Mutation	G	A	OCCC_47	p.A867T
SPTB	14	65260543	65260543	+	Missense_Mutation	C	A	OCCC_47	p.S613I
AHNAK2	14	105413318	105413318	+	Missense_Mutation	C	T	OCCC_47	p.G2824R
AHNAK2	14	105415607	105415607	+	Missense_Mutation	C	T	OCCC_47	p.V2061M
AHNAK2	14	105417358	105417358	+	Missense_Mutation	T	C	OCCC_47	p.E1477G
CASKIN1	16	2231918	2231918	+	Missense_Mutation	C	T	OCCC_47	p.V548M
XPO6	16	28167402	28167402	+	Missense_Mutation	C	T	OCCC_47	p.D364N
SPTBN4	19	41060134	41060134	+	Missense_Mutation	A	C	OCCC_47	p.S1586R
SPTBN4	19	41062987	41062987	+	Missense_Mutation	G	A	OCCC_47	p.R1783Q
VASP	19	46021266	46021266	+	Missense_Mutation	G	A	OCCC_47	p.R86H
PPP2R1A	19	52716323	52716323	+	Missense_Mutation	C	A	OCCC_47	p.S256Y
LAMA5	20	60889985	60889985	+	Missense_Mutation	G	A	OCCC_47	p.T2689M
LAMA5	20	60899525	60899525	+	Missense_Mutation	C	T	OCCC_47	p.R1872H
HELZ2	20	62190630	62190630	+	Missense_Mutation	C	T	OCCC_47	p.R2640H
MAPK1	22	22123579	22123579	+	Missense_Mutation	T	C	OCCC_47	p.M333V
MAPK1	22	22153396	22153396	+	Missense_Mutation	G	A	OCCC_47	p.R172C
ARID1A	1	27057788	27057788	+	Missense_Mutation	C	T	OCCC_25	p.S499L
ARID1A	1	27106648	27106648	+	Missense_Mutation	G	A	OCCC_25	p.G2087R
RPTN	1	152128065	152128065	+	Missense_Mutation	C	T	OCCC_25	p.G504R
HRNR	1	152191050	152191050	+	Missense_Mutation	C	T	OCCC_25	p.G1019R
FLG	1	152281145	152281145	+	Missense_Mutation	T	G	OCCC_25	p.K2073Q
FLG	1	152282178	152282178	+	Missense_Mutation	C	G	OCCC_25	p.E1728D
FLG	1	152283430	152283430	+	Missense_Mutation	A	G	OCCC_25	p.F1311S
FLG2	1	152324114	152324114	+	Missense_Mutation	C	T	OCCC_25	p.A2050T
OBSCN	1	228464337	228464337	+	Missense_Mutation	C	T	OCCC_25	p.S2136L
LRP1B	2	141625794	141625794	+	Missense_Mutation	C	T	OCCC_25	p.R1403H
MUC4	3	195508586	195508586	+	Missense_Mutation	A	C	OCCC_25	p.S3289A
DSP	6	7584072	7584072	+	Missense_Mutation	G	A	OCCC_25	p.E2193K
DST	6	56480547	56480547	+	Missense_Mutation	C	T	OCCC_25	p.R2573Q
MUC17	7	100679633	100679633	+	Missense_Mutation	C	T	OCCC_25	p.P1646S
MUC17	7	100682351	100682351	+	Missense_Mutation	A	G	OCCC_25	p.I2552V
AHNAK	11	62295870	62295870	+	Missense_Mutation	T	C	OCCC_25	p.M2007V
AHNAK	11	62296335	62296335	+	Missense_Mutation	C	T	OCCC_25	p.A1852T
SPTB	14	65251050	65251050	+	Missense_Mutation	C	T	OCCC_25	p.R1306Q
SPTB	14	65260495	65260495	+	Missense_Mutation	C	T	OCCC_25	p.R629Q
SPTB	14	65263370	65263370	+	Missense_Mutation	G	A	OCCC_25	p.R416W
AHNAK2	14	105405284	105405284	+	Missense_Mutation	G	A	OCCC_25	p.R5502W
AHNAK2	14	105408638	105408638	+	Missense_Mutation	G	C	OCCC_25	p.L4384V
AHNAK2	14	105410804	105410804	+	Missense_Mutation	C	G	OCCC_25	p.D3662H
CASKIN1	16	2231447	2231447	+	Missense_Mutation	G	A	OCCC_25	p.P641L
CASKIN1	16	2239089	2239089	+	Missense_Mutation	C	T	OCCC_25	p.D186N
EVPL	17	74018494	74018494	+	Splice_Site	C	T	OCCC_25	p.P202P
PPP2R1A	19	52715982	52715982	+	Missense_Mutation	C	T	OCCC_25	p.R183W
ARID1A	1	27099947	27099947	+	Nonsense_Mutation	C	T	OCCC_01	p.R1276*
HRNR	1	152191019	152191019	+	Missense_Mutation	G	T	OCCC_01	p.S1029Y
HRNR	1	152191565	152191565	+	Missense_Mutation	G	A	OCCC_01	p.T847M
SPTA1	1	158581062	158581062	+	Missense_Mutation	C	A	OCCC_01	p.G2418C
OBSCN	1	228520965	228520965	+	Missense_Mutation	T	A	OCCC_01	p.L5266Q
MUC4	3	195515435	195515435	+	Missense_Mutation	C	T	OCCC_01	p.A1006T
SHROOM3	4	77631404	77631404	+	Missense_Mutation	C	T	OCCC_01	p.A140V
SHROOM3	4	77676219	77676219	+	Missense_Mutation	C	A	OCCC_01	p.P1528Q
SHROOM3	4	77700047	77700047	+	Missense_Mutation	G	A	OCCC_01	p.R1903Q
ANK3	10	61965552	61965552	+	Missense_Mutation	C	T	OCCC_01	p.E431K
AHNAK	11	62292816	62292816	+	Missense_Mutation	C	T	OCCC_01	p.V3025M
AHNAK2	14	105407965	105407965	+	Missense_Mutation	G	A	OCCC_01	p.A4608V
CASKIN1	16	2237236	2237236	+	Missense_Mutation	T	A	OCCC_01	p.T256S
EVPL	17	74005072	74005072	+	Missense_Mutation	A	C	OCCC_01	p.L1405R
EVPL	17	74005073	74005073	+	Missense_Mutation	G	T	OCCC_01	p.L1405I
EYA2	20	45801465	45801465	+	Missense_Mutation	G	A	OCCC_01	p.R383H
ARID1A	1	27056349	27056349	+	Nonsense_Mutation	C	T	OCCC_32	p.Q449*
PIK3CA	3	178936091	178936091	+	Missense_Mutation	G	A	OCCC_32	p.E545K
AHNAK2	14	105415160	105415160	+	Missense_Mutation	C	G	OCCC_32	p.V2210L
KRT10	17	38978372	38978372	+	Missense_Mutation	G	A	OCCC_32	p.R156C
AKT2	19	40745962	40745962	+	Missense_Mutation	G	A	OCCC_32	p.P210L
ARID1A	1	27106558	27106558	+	Missense_Mutation	C	T	OCCC_34	p.R2057W

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
MUC17	7	100680354	100680354	+	Missense_Mutation	C	G	OCCC_34	p.T1886S
TCHH	1	152081230	152081230	+	Missense_Mutation	A	C	OCCC_35	p.L1488R
FLG	1	152280731	152280731	+	Missense_Mutation	G	C	OCCC_35	p.H2211D
PIK3CA	3	178936082	178936082	+	Missense_Mutation	G	A	OCCC_35	p.E542K
MUC4	3	195489015	195489015	+	Missense_Mutation	G	A	OCCC_35	p.L583F
DST	6	56417211	56417211	+	Missense_Mutation	A	T	OCCC_35	p.V5249D
SPTB	14	65289745	65289745	+	Missense_Mutation	C	A	OCCC_35	p.W23L
DST	6	56505405	56505405	+	Splice_Site	T	-	OCCC_35	
HRNR	1	152188862	152188862	+	Missense_Mutation	G	A	OCCC_61	p.S1748L
FLG	1	152276886	152276886	+	Missense_Mutation	G	C	OCCC_61	p.D3492E
FLG	1	152279527	152279527	+	Missense_Mutation	T	C	OCCC_61	p.D2612G
FLG	1	152280002	152280002	+	Missense_Mutation	T	C	OCCC_61	p.T2454A
FLG2	1	152325817	152325817	+	Missense_Mutation	T	C	OCCC_61	p.H1482R
FLG2	1	152325818	152325818	+	Missense_Mutation	G	C	OCCC_61	p.H1482D
FLG2	1	152325820	152325820	+	Missense_Mutation	T	G	OCCC_61	p.Y1481S
MUC4	3	195510896	195510896	+	Missense_Mutation	G	A	OCCC_61	p.P2519S
MUC17	7	100680017	100680017	+	Missense_Mutation	A	G	OCCC_61	p.I1774V
MUC17	7	100682427	100682427	+	Missense_Mutation	G	C	OCCC_61	p.R2577T
MUC17	7	100682967	100682967	+	Missense_Mutation	T	C	OCCC_61	p.L2757P
PLEC	8	145003304	145003304	+	Missense_Mutation	T	C	OCCC_61	p.Q1213R
AHNAK2	14	105407327	105407327	+	Missense_Mutation	G	A	OCCC_61	p.P4821S
AHNAK2	14	105413791	105413791	+	Missense_Mutation	G	A	OCCC_61	p.S2666F
AHNAK2	14	105414923	105414923	+	Missense_Mutation	T	C	OCCC_61	p.K2289E
PPP2R1A	19	52722956	52722956	+	Missense_Mutation	C	T	OCCC_61	p.R381W
LAMA5	20	60909671	60909671	+	Missense_Mutation	C	T	OCCC_61	p.R830Q
DST	6	56515830	56515831	+	Splice_Site	TC	-	OCCC_61	p.G232fs
FLG	1	152276490	152276490	+	Missense_Mutation	C	G	OCCC_03	p.E3624D
FLG	1	152282852	152282852	+	Missense_Mutation	A	G	OCCC_03	p.Y1504H
FLG	1	152283256	152283256	+	Missense_Mutation	C	G	OCCC_03	p.R1369T
ZNF717	3	75788028	75788028	+	Missense_Mutation	G	A	OCCC_03	p.S199L
MUC17	7	100678724	100678724	+	Missense_Mutation	C	A	OCCC_03	p.P1343T
MUC17	7	100682922	100682922	+	Missense_Mutation	G	C	OCCC_03	p.R2742P
KRAS	12	25398284	25398284	+	Missense_Mutation	C	G	OCCC_03	p.G12A
MUC19	12	40820396	40820396	+	Missense_Mutation	G	A	OCCC_03	p.R125Q
AHNAK2	14	105410804	105410804	+	Missense_Mutation	C	G	OCCC_03	p.D3662H
ARID1A	1	27106915	27106915	+	Nonsense_Mutation	C	T	OCCC_30	p.Q2176*
PIK3CA	3	178952077	178952077	+	Missense_Mutation	T	G	OCCC_30	p.N1044K
FLG	1	152277137	152277137	+	Missense_Mutation	G	C	OCCC_69	p.R3409G
FLG	1	152281635	152281635	+	Missense_Mutation	C	A	OCCC_69	p.R1909S
CELSR3	3	48689423	48689423	+	Missense_Mutation	C	A	OCCC_69	p.R1937L
MUC4	3	195513010	195513010	+	Missense_Mutation	G	A	OCCC_69	p.P1814L
MUC17	7	100679169	100679169	+	Missense_Mutation	C	A	OCCC_69	p.A1491E
MUC17	7	100679388	100679388	+	Missense_Mutation	A	C	OCCC_69	p.Q1564P
MUC17	7	100682261	100682261	+	Missense_Mutation	G	C	OCCC_69	p.V2522L
PLEC	8	144995266	144995266	+	Missense_Mutation	G	T	OCCC_69	p.A3045D
PLEC	8	144998339	144998339	+	Missense_Mutation	G	A	OCCC_69	p.R2057W
AHNAK	11	62295071	62295071	+	Missense_Mutation	T	A	OCCC_69	p.D2273V
SPTBN4	19	40993723	40993723	+	Missense_Mutation	C	T	OCCC_69	p.R97W
UNC13B	9	35397632	35397632	+	Splice_Site	-	CCATCGGGAAGGTGCT GATGATGTC TGCATACTGCATCAGC ACCTT	OCCC_69	p.T1144fs
ARID1A	1	27106559	27106559	+	Missense_Mutation	G	A	OCCC_63	p.R2057Q
OBSCN	1	228456294	228456294	+	Missense_Mutation	C	T	OCCC_63	p.A1642V
SHROOM3	4	77662468	77662468	+	Missense_Mutation	C	T	OCCC_63	p.R1048C
DST	6	56485366	56485366	+	Nonsense_Mutation	G	A	OCCC_63	p.R1156*
AHNAK2	14	105412913	105412913	+	Missense_Mutation	G	A	OCCC_63	p.R2959W
SPTBN4	19	41019388	41019388	+	Missense_Mutation	G	A	OCCC_63	p.G898S
MAPK1	22	22153395	22153395	+	Missense_Mutation	C	T	OCCC_63	p.R172H
KRT5	12	52913524	52913524	+	Splice_Site	A	-	OCCC_63	
HELZ2	20	62197545	62197545	+	Missense_Mutation	G	T	OCCC_67	p.T877N
MAPK8IP2	22	51042980	51042980	+	Missense_Mutation	C	T	OCCC_67	p.P152L
OBSCN	1	228412271	228412271	+	Missense_Mutation	C	T	OCCC_41	p.T922M
SHROOM3	4	77678005	77678005	+	Nonsense_Mutation	C	T	OCCC_41	p.R1705*
DST	6	56417706	56417706	+	Missense_Mutation	T	C	OCCC_41	p.Q5084R
PLEC	8	144992259	144992259	+	Missense_Mutation	C	A	OCCC_41	p.Q4047H

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
PLEC	8	145003701	145003701	+	Missense_Mutation	C	T	OCCC_41	p.E1125K
AHNAK	11	62285364	62285364	+	Missense_Mutation	C	T	OCCC_41	p.V5509M
KRT5	12	52911474	52911474	+	Missense_Mutation	C	T	OCCC_41	p.R331H
AHNAK2	14	105412598	105412598	+	Missense_Mutation	A	C	OCCC_41	p.L3064V
AHNAK2	14	105412633	105412633	+	Missense_Mutation	G	A	OCCC_41	p.P3052L
MAGEE1	X	75650211	75650211	+	Missense_Mutation	G	A	OCCC_41	p.V630M
ARID1A	1	27100961	27100961	+	Nonsense_Mutation	C	T	OCCC_17	p.Q1415*
FLG	1	152280568	152280568	+	Missense_Mutation	C	A	OCCC_17	p.R2265I
CTNNB1	3	41275648	41275648	+	Nonsense_Mutation	C	T	OCCC_17	p.R515*
ZNF717	3	75788088	75788088	+	Missense_Mutation	C	T	OCCC_17	p.R179K
ZNF717	3	75790800	75790800	+	Missense_Mutation	C	T	OCCC_17	p.D49N
PIK3CA	3	178936095	178936095	+	Missense_Mutation	A	T	OCCC_17	p.Q546L
MUC4	3	195508955	195508955	+	Missense_Mutation	C	T	OCCC_17	p.A3166T
SHROOM3	4	77675528	77675528	+	Missense_Mutation	C	T	OCCC_17	p.R1298C
SHROOM3	4	77677730	77677730	+	Missense_Mutation	C	T	OCCC_17	p.T1613I
KRT5	12	52912805	52912805	+	Missense_Mutation	C	T	OCCC_17	p.S232N
AHNAK2	14	105408107	105408107	+	Missense_Mutation	G	T	OCCC_17	p.L4561I
AHNAK2	14	105412871	105412871	+	Missense_Mutation	C	T	OCCC_17	p.A2973T
ARID1A	1	27023818	27023818	+	Nonsense_Mutation	C	G	OCCC_18	p.Y308*
HRNR	1	152186877	152186877	+	Missense_Mutation	C	G	OCCC_18	p.G2410R
FLG	1	152281113	152281113	+	Missense_Mutation	G	C	OCCC_18	p.S2083R
FLG	1	152283589	152283589	+	Nonsense_Mutation	G	T	OCCC_18	p.S1258*
FLG	1	152283590	152283590	+	Missense_Mutation	A	C	OCCC_18	p.S1258A
FLG	1	152284318	152284318	+	Missense_Mutation	C	T	OCCC_18	p.G1015D
FLG	1	152284319	152284319	+	Missense_Mutation	C	G	OCCC_18	p.G1015R
FLG2	1	152328326	152328326	+	Missense_Mutation	C	T	OCCC_18	p.G646R
ZNF717	3	75788260	75788260	+	Missense_Mutation	C	A	OCCC_18	p.G122W
MUC17	7	100677793	100677793	+	Missense_Mutation	G	A	OCCC_18	p.M1032I
MUC17	7	100680228	100680228	+	Missense_Mutation	C	A	OCCC_18	p.A1844E
MUC17	7	100682045	100682045	+	Missense_Mutation	C	A	OCCC_18	p.P2450T
MUC17	7	100682552	100682552	+	Missense_Mutation	A	G	OCCC_18	p.K2619E
MUC17	7	100682556	100682556	+	Missense_Mutation	A	G	OCCC_18	p.D2620G
UNC13B	9	35398227	35398227	+	Missense_Mutation	A	T	OCCC_18	p.N1176I
AHNAK	11	62298074	62298074	+	Missense_Mutation	C	G	OCCC_18	p.R1272P
AHNAK2	14	105413471	105413471	+	Missense_Mutation	T	C	OCCC_18	p.K2773E
SPTB	14	65258436	65258452	+	Splice_Site	CCTGGTGTTCAGATGGT	-	OCCC_18	p.DHLNTR930fs
FLG	1	152276045	152276045	+	Missense_Mutation	A	G	OCCC_48	p.Y3773H
FLG	1	152284263	152284263	+	Missense_Mutation	G	C	OCCC_48	p.H1033Q
FLG	1	152284673	152284673	+	Missense_Mutation	G	A	OCCC_48	p.R897C
FLG	1	152284791	152284791	+	Missense_Mutation	G	T	OCCC_48	p.H857Q
FLG2	1	152329942	152329942	+	Missense_Mutation	C	T	OCCC_48	p.R107Q
OBSCN	1	228476577	228476577	+	Missense_Mutation	C	G	OCCC_48	p.L3443V
ZNF717	3	75786921	75786921	+	Missense_Mutation	G	A	OCCC_48	p.P568L
MUC4	3	195477841	195477841	+	Missense_Mutation	C	T	OCCC_48	p.V1028I
MUC4	3	195513605	195513605	+	Missense_Mutation	G	A	OCCC_48	p.P1616S
MUC17	7	100681211	100681211	+	Missense_Mutation	G	C	OCCC_48	p.V2172L
MUC17	7	100681533	100681533	+	Missense_Mutation	C	A	OCCC_48	p.T2279N
MUC17	7	100681985	100681985	+	Missense_Mutation	A	T	OCCC_48	p.T2430S
PLEC	8	144991243	144991243	+	Missense_Mutation	G	A	OCCC_48	p.S4386L
PLEC	8	144998614	144998614	+	Missense_Mutation	C	T	OCCC_48	p.R1965Q
ANK3	10	61829483	61829483	+	Missense_Mutation	C	T	OCCC_48	p.R3719H
ANK3	10	61831651	61831651	+	Missense_Mutation	C	G	OCCC_48	p.Q2996H
AHNAK	11	62295656	62295656	+	Missense_Mutation	A	T	OCCC_48	p.V2078D
MUC19	12	40878695	40878695	+	Missense_Mutation	A	G	OCCC_48	p.I3414V
MUC19	12	40878697	40878697	+	Missense_Mutation	A	G	OCCC_48	p.I3414M
MUC19	12	40924074	40924074	+	Splice_Site	G	T	OCCC_48	
KRT5	12	52908794	52908794	+	Missense_Mutation	C	T	OCCC_48	p.G569R
AHNAK2	14	105409546	105409546	+	Missense_Mutation	T	G	OCCC_48	p.K4081T
AHNAK2	14	105413266	105413266	+	Missense_Mutation	A	G	OCCC_48	p.L2841P
AHNAK2	14	105416046	105416046	+	Missense_Mutation	G	C	OCCC_48	p.F1914L
AHNAK2	14	105419446	105419446	+	Missense_Mutation	T	G	OCCC_48	p.K781T
CASKIN1	16	2236759	2236759	+	Missense_Mutation	G	A	OCCC_48	p.R333W
CDC27	17	45219364	45219364	+	Missense_Mutation	A	T	OCCC_48	p.M469K
ARID1A	1	27106142	27106142	+	Missense_Mutation	G	A	OCCC_66	p.R1918Q
HRNR	1	152191206	152191206	+	Missense_Mutation	A	G	OCCC_66	p.S967P
OBSCN	1	228432236	228432236	+	Missense_Mutation	G	A	OCCC_66	p.G1149R
PIK3CA	3	178952085	178952085	+	Missense_Mutation	A	G	OCCC_66	p.H1047R

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
MUC4	3	195506411	195506411	+	Missense_Mutation	C	T	OCCC_66	p.A4014T
MUC4	3	195508937	195508937	+	Missense_Mutation	T	A	OCCC_66	p.T3172S
SHROOM3	4	77661446	77661446	+	Missense_Mutation	A	G	OCCC_66	p.K707R
MSH3	5	80088643	80088643	+	Missense_Mutation	C	A	OCCC_66	p.P879T
MSH3	5	80088644	80088644	+	Missense_Mutation	C	A	OCCC_66	p.P879Q
ANK3	10	61829936	61829936	+	Missense_Mutation	C	T	OCCC_66	p.G3568E
KRAS	12	25398284	25398284	+	Missense_Mutation	C	A	OCCC_66	p.G12V
AHNAK2	14	105413284	105413284	+	Missense_Mutation	G	A	OCCC_66	p.S2835L
CASKIN1	16	2230814	2230814	+	Missense_Mutation	G	A	OCCC_66	p.A852V
CDC27	17	45235598	45235598	+	Missense_Mutation	G	T	OCCC_66	p.S150Y
OBSCN	1	228522798	228522798	+	Missense_Mutation	G	T	OCCC_19	p.V5402L
LRP1B	2	141625780	141625780	+	Missense_Mutation	A	T	OCCC_19	p.S1408T
LRP1B	2	141625782	141625782	+	Missense_Mutation	G	T	OCCC_19	p.A1407D
MUC4	3	195477909	195477909	+	Missense_Mutation	C	T	OCCC_19	p.R1005Q
MUC17	7	100683723	100683723	+	Missense_Mutation	G	C	OCCC_19	p.R3009T
AHNAK	11	62295549	62295549	+	Missense_Mutation	C	T	OCCC_19	p.A2114T
EVPL	17	74003794	74003794	+	Missense_Mutation	T	C	OCCC_19	p.Y1831C
VASP	19	46021012	46021012	+	Missense_Mutation	T	A	OCCC_19	p.F33I
VASP	19	46021013	46021013	+	Missense_Mutation	T	G	OCCC_19	p.F33C
LAMA5	20	60905992	60905992	+	Missense_Mutation	C	A	OCCC_19	p.C1220F
FLG	1	152280430	152280430	+	Missense_Mutation	G	C	OCCC_51	p.S2311C
FLG2	1	152324093	152324093	+	Missense_Mutation	G	C	OCCC_51	p.Q2057E
FLG2	1	152324146	152324146	+	Missense_Mutation	T	G	OCCC_51	p.H2039P
FLG2	1	152328192	152328192	+	Missense_Mutation	A	C	OCCC_51	p.H690Q
SPTA1	1	158604407	158604407	+	Missense_Mutation	C	G	OCCC_51	p.E1831Q
OBSCN	1	228463635	228463635	+	Missense_Mutation	G	A	OCCC_51	p.R2043H
OBSCN	1	228475464	228475464	+	Missense_Mutation	G	A	OCCC_51	p.R3205H
CELSR3	3	48689456	48689456	+	Missense_Mutation	G	C	OCCC_51	p.S1926C
MUC4	3	195512117	195512117	+	Missense_Mutation	C	G	OCCC_51	p.A2112P
DSP	6	7585846	7585846	+	Missense_Mutation	C	T	OCCC_51	p.S2784F
PLEC	8	144999110	144999110	+	Missense_Mutation	C	G	OCCC_51	p.E1800Q
AHNAK	11	62284251	62284251	+	Missense_Mutation	C	T	OCCC_51	p.E5880K
MUC19	12	40876710	40876710	+	Missense_Mutation	C	T	OCCC_51	p.P2752L
AHNAK2	14	105412801	105412801	+	Missense_Mutation	G	C	OCCC_51	p.S2996C
CASKIN1	16	2230236	2230236	+	Missense_Mutation	C	G	OCCC_51	p.A1045P
KRT10	17	38975232	38975232	+	Missense_Mutation	T	C	OCCC_51	p.S519G
EVPL	17	74010545	74010545	+	Missense_Mutation	C	A	OCCC_51	p.G779C
ARID3A	19	932572	932572	+	Nonsense_Mutation	C	T	OCCC_51	p.R175*
LAMA5	20	60889911	60889911	+	Missense_Mutation	C	T	OCCC_51	p.A2714T
FLG2	1	152324440	152324440	+	Missense_Mutation	A	G	OCCC_33	p.I1941T
AHNAK2	14	105412100	105412100	+	Missense_Mutation	G	C	OCCC_33	p.Q3230E
LAMA5	20	60909672	60909672	+	Missense_Mutation	G	A	OCCC_33	p.R830W
SPTBN2	11	66460644	66460666	+	Splice_Site	TCCTGGCCCTCAC CTTGGCCTTC	-	OCCC_33	p.EKAKV1615fs
PIK3CA	3	178952085	178952085	+	Missense_Mutation	A	G	OCCC_06	p.H1047R
DMRTB1	1	53925412	53925412	+	Missense_Mutation	C	T	OCCC_59	p.R96C
TCHH	1	152084066	152084066	+	Missense_Mutation	C	T	OCCC_59	p.E543K
HRNR	1	152189067	152189067	+	Missense_Mutation	C	T	OCCC_59	p.G1680R
FLG	1	152282178	152282178	+	Missense_Mutation	C	G	OCCC_59	p.E1728D
CELSR3	3	48685349	48685349	+	Nonsense_Mutation	G	A	OCCC_59	p.R2352*
MUC4	3	195515449	195515449	+	Missense_Mutation	A	G	OCCC_59	p.V1001A
DSP	6	7579931	7579931	+	Missense_Mutation	G	A	OCCC_59	p.E1170K
MUC17	7	100683644	100683644	+	Missense_Mutation	G	A	OCCC_59	p.G2983S
PLEC	8	144995227	144995227	+	Missense_Mutation	G	A	OCCC_59	p.T3058M
SPTBN2	11	66461627	66461627	+	Missense_Mutation	C	T	OCCC_59	p.D1496N
KRT5	12	52912805	52912805	+	Missense_Mutation	C	T	OCCC_59	p.S232N
AHNAK2	14	105408172	105408172	+	Missense_Mutation	A	C	OCCC_59	p.V4539G
AHNAK2	14	105412720	105412720	+	Missense_Mutation	G	T	OCCC_59	p.T3023N
AHNAK2	14	105419313	105419313	+	Missense_Mutation	C	G	OCCC_59	p.E825D
XPO6	16	28128712	28128712	+	Missense_Mutation	C	T	OCCC_59	p.R644Q
EVPL	17	74005742	74005742	+	Missense_Mutation	C	T	OCCC_59	p.V1182M
DST	6	56457157	56457158	+	Splice_Site	CT	-	OCCC_59	
MUC19	12	40858220	40858228	+	Splice_Site	CAGGAAGTA	-	OCCC_59	p.QEV1199del
HRNR	1	152186844	152186844	+	Missense_Mutation	G	A	OCCC_44	p.R2421C
HRNR	1	152187048	152187048	+	Missense_Mutation	C	T	OCCC_44	p.G2353S
HRNR	1	152187194	152187194	+	Missense_Mutation	C	A	OCCC_44	p.R2304L
HRNR	1	152192259	152192259	+	Missense_Mutation	T	C	OCCC_44	p.T616A

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
OBSCN	1	228432198	228432198	+	Missense_Mutation	C	T	OCCC_44	p.A1136V
ZNF717	3	75786036	75786036	+	Missense_Mutation	A	G	OCCC_44	p.F863S
ZNF717	3	75788217	75788217	+	Missense_Mutation	G	A	OCCC_44	p.T136I
MSH3	5	79950724	79950724	+	Missense_Mutation	G	C	OCCC_44	p.A60P
MUC17	7	100679782	100679782	+	Missense_Mutation	A	G	OCCC_44	p.I1695M
MUC17	7	100679783	100679783	+	Missense_Mutation	A	C	OCCC_44	p.T1696P
MUC17	7	100683099	100683099	+	Missense_Mutation	C	G	OCCC_44	p.P2801R
AHNAK2	14	105413588	105413588	+	Missense_Mutation	G	C	OCCC_44	p.L2734V
EVPL	17	74019680	74019680	+	Missense_Mutation	C	T	OCCC_44	p.R85H
ARID1A	1	27099122	27099122	+	Splice_Site	-	G	OCCC_44	p.S1180fs
HRNR	1	152185695	152185695	+	Missense_Mutation	C	A	OCCC_08	p.G2804C
HRNR	1	152187879	152187879	+	Missense_Mutation	T	C	OCCC_08	p.R2076G
OBSCN	1	228400030	228400030	+	Nonsense_Mutation	C	A	OCCC_08	p.Y182*
OBSCN	1	228505767	228505767	+	Missense_Mutation	G	T	OCCC_08	p.G4675V
MUC17	7	100680579	100680579	+	Missense_Mutation	C	A	OCCC_08	p.S1961Y
HELZ2	20	62195277	62195277	+	Missense_Mutation	G	A	OCCC_08	p.P1633L
LRP1B	2	141267574	141267574	+	Missense_Mutation	C	A	OCCC_15	p.C2774F
EYA2	20	45801437	45801437	+	Missense_Mutation	G	A	OCCC_15	p.V374M
OBSCN	1	228463548	228463548	+	Missense_Mutation	C	A	OCCC_49	p.P2014Q
FLG	1	152286344	152286344	+	Missense_Mutation	C	G	OCCC_39	p.D340H
ANK3	10	61832367	61832367	+	Missense_Mutation	C	T	OCCC_39	p.V2758I
SPTBN2	11	66481105	66481105	+	Missense_Mutation	C	T	OCCC_39	p.E257K
MUC19	12	40876494	40876494	+	Missense_Mutation	T	C	OCCC_39	p.I2680T
EYA2	20	45812004	45812004	+	Missense_Mutation	G	A	OCCC_39	p.G501S
MAPK1	22	22160188	22160188	+	Missense_Mutation	C	T	OCCC_39	p.R148H
PIK3CA	3	178952007	178952007	+	Missense_Mutation	A	G	OCCC_62	p.Y1021C
UNC13B	9	35376074	35376074	+	Missense_Mutation	C	T	OCCC_62	p.S473L
KRT1	12	53071200	53071200	+	Missense_Mutation	C	T	OCCC_62	p.R343H
AHNAK2	14	105418508	105418508	+	Missense_Mutation	C	A	OCCC_62	p.V1094L
LAMA5	20	60908258	60908258	+	Missense_Mutation	G	A	OCCC_62	p.S1057L
PIK3CA	3	178936067	178936067	+	Nonsense_Mutation	C	T	OCCC_31	p.R537*
MUC4	3	195515006	195515006	+	Missense_Mutation	G	C	OCCC_31	p.H1149D
MUC17	7	100683472	100683472	+	Missense_Mutation	A	T	OCCC_31	p.E2925D
MUC17	7	100683474	100683474	+	Missense_Mutation	T	G	OCCC_31	p.V2926G
UNC13B	9	35382464	35382464	+	Missense_Mutation	A	C	OCCC_31	p.N840T
AHNAK	11	62289981	62289981	+	Missense_Mutation	C	T	OCCC_31	p.V3970I
OBSCN	1	228566387	228566387	+	Missense_Mutation	G	A	OCCC_11	p.R7933Q
CTNNB1	3	41266097	41266097	+	Missense_Mutation	G	T	OCCC_11	p.D32Y
PLEC	8	144997792	144997792	+	Missense_Mutation	C	T	OCCC_11	p.R2239H
AHNAK2	14	105412466	105412466	+	Missense_Mutation	C	T	OCCC_11	p.G3108S
CDC27	17	45249432	45249432	+	Splice_Site	T	A	OCCC_11	
CDC27	17	45249431	45249431	+	Splice_Site	C	-	OCCC_11	
EVPL	17	74019372	74019372	+	Splice_Site	-	CTGTTTCTGCTC CAGCACGC GTGCCCAGAAACA GGTCAGGAA	OCCC_11	
ARID1A	1	27100070	27100070	+	Splice_Site	G	A	OCCC_07	
FLG2	1	152331330	152331330	+	Missense_Mutation	C	T	OCCC_07	p.V11I
PIK3CA	3	178952085	178952085	+	Missense_Mutation	A	G	OCCC_07	p.H1047R
PPP2R1A	19	52715970	52715970	+	Missense_Mutation	C	A	OCCC_07	p.P179T
PLEC	8	145004373	145004373	+	Missense_Mutation	C	T	OCCC_20	p.V988M
PLEC	8	145005812	145005812	+	Missense_Mutation	G	A	OCCC_20	p.R869W
KRT10	17	38975261	38975261	+	Missense_Mutation	G	A	OCCC_20	p.S509F
LAMA5	20	60910229	60910295	+	Splice_Site	CCAGTGTGCCCCC AAATCCCCCACAC CTTGGTCCTCAGAC TCACCGGCTGGCA CTCAGCAACTCCAC	-	OCCC_20	p.GGVAECQP785fs
PIK3CA	3	178919287	178919287	+	Missense_Mutation	G	T	OCCC_14	p.D258Y
MUC4	3	195489009	195489009	+	Missense_Mutation	C	G	OCCC_14	p.A585P
MUC19	12	40878078	40878078	+	Missense_Mutation	C	T	OCCC_14	p.T3208I
AHNAK2	14	105420865	105420865	+	Missense_Mutation	C	T	OCCC_14	p.R308H
TCHH	1	152083846	152083846	+	Missense_Mutation	C	A	OCCC_43	p.R616L
FLG	1	152284450	152284450	+	Missense_Mutation	C	T	OCCC_43	p.R971H
OBSCN	1	228399923	228399923	+	Missense_Mutation	G	A	OCCC_43	p.G147R
CELSR3	3	48696988	48696988	+	Missense_Mutation	C	T	OCCC_43	p.R1027Q

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
DST	6	56346870	56346870	+	Missense_Mutation	C	T	OCCC_43	p.R6850Q
PLEC	8	144999694	144999694	+	Missense_Mutation	C	T	OCCC_43	p.R1605H
PLEC	8	145001886	145001886	+	Missense_Mutation	G	A	OCCC_43	p.R1287W
UNC13B	9	35403941	35403941	+	Missense_Mutation	C	T	OCCC_43	p.R1563W
ANK3	10	61834631	61834631	+	Missense_Mutation	T	A	OCCC_43	p.Q2003L
SPTB	14	65240080	65240080	+	Missense_Mutation	G	A	OCCC_43	p.A1679V
AHNAK2	14	105418067	105418067	+	Missense_Mutation	C	T	OCCC_43	p.G1241S
EYA2	20	45725802	45725802	+	Missense_Mutation	G	A	OCCC_43	p.G295R
LAMA5	20	60885861	60885861	+	Missense_Mutation	G	A	OCCC_43	p.R3436W
MAPK8IP2	22	51042592	51042592	+	Missense_Mutation	C	T	OCCC_43	p.R23C
MAGEE1	X	75648374	75648374	+	Missense_Mutation	G	T	OCCC_43	p.K17N
RPTN	1	152128196	152128196	+	Missense_Mutation	C	T	OCCC_46	p.S460N
DSPP	4	88536980	88536980	+	Missense_Mutation	A	G	OCCC_46	p.N1056D
MUC17	7	100683179	100683179	+	Missense_Mutation	G	A	OCCC_46	p.G2828S
PLEC	8	144991180	144991180	+	Nonsense_Mutation	C	T	OCCC_46	p.W4407*
AHNAK2	14	105413066	105413066	+	Missense_Mutation	T	C	OCCC_46	p.K2908E
AHNAK2	14	105418918	105418918	+	Missense_Mutation	C	A	OCCC_46	p.G957V
ARID1A	1	27101099	27101099	+	Nonsense_Mutation	C	T	OCCC_21	p.R1461*
FLG	1	152282672	152282672	+	Missense_Mutation	G	C	OCCC_21	p.P1564A
LRP1B	2	141459361	141459361	+	Missense_Mutation	G	A	OCCC_21	p.T2119M
CELSR3	3	48681704	48681704	+	Missense_Mutation	C	T	OCCC_21	p.A2704T
CELSR3	3	48697838	48697838	+	Missense_Mutation	C	T	OCCC_21	p.V744M
PIK3CA	3	178936083	178936083	+	Missense_Mutation	A	G	OCCC_21	p.E542G
PIK3CA	3	178952013	178952013	+	Missense_Mutation	G	A	OCCC_21	p.R1023Q
MUC17	7	100685327	100685327	+	Missense_Mutation	T	A	OCCC_21	p.S3544T
MUC19	12	40873956	40873956	+	Missense_Mutation	G	T	OCCC_21	p.G1834V
MAPK8IP2	22	51044262	51044262	+	Missense_Mutation	G	A	OCCC_21	p.R409Q
HRNR	1	152187606	152187606	+	Missense_Mutation	G	A	OCCC_53	p.R2167C
LOR	1	153233513	153233513	+	Missense_Mutation	G	A	OCCC_53	p.G30S
LRP1B	2	141660736	141660736	+	Splice_Site	T	A	OCCC_53	
PIK3CA	3	178936082	178936082	+	Missense_Mutation	G	C	OCCC_53	p.E542Q
PLEC	8	144990707	144990707	+	Missense_Mutation	C	T	OCCC_53	p.V4565M
ANK3	10	61956385	61956385	+	Splice_Site	T	A	OCCC_53	
AHNAK2	14	105411020	105411020	+	Missense_Mutation	C	T	OCCC_53	p.A3590T
AHNAK2	14	105418481	105418481	+	Missense_Mutation	C	T	OCCC_53	p.V1103I
AKT2	19	40741948	40741948	+	Missense_Mutation	C	T	OCCC_53	p.E342K
MAPK1	22	22153396	22153396	+	Missense_Mutation	G	A	OCCC_53	p.R172C
OBSCN	1	228494303	228494342	+	Splice_Site	GGCTCCCAGGCCACCAGTG CCACCCTCACTGTCACAGGTG	-	OCCC_53	p.GSQATSATLTVTG3964fs
CASKIN1	16	2239567	2239567	+	Splice_Site	-	G	OCCC_53	
PIK3CA	3	178936082	178936082	+	Missense_Mutation	G	A	OCCC_26	p.E542K
UNC13B	9	35403873	35403873	+	Missense_Mutation	G	A	OCCC_26	p.G1540D
CDK12	17	37646866	37646866	+	Missense_Mutation	G	A	OCCC_26	p.R663H
ARID1A	1	27058029	27058029	+	Nonsense_Mutation	T	G	OCCC_27	p.Y579*
ARID1A	1	27106621	27106621	+	Nonsense_Mutation	G	T	OCCC_27	p.E2078*
FLG	1	152285252	152285252	+	Missense_Mutation	C	G	OCCC_27	p.A704P
OBSCN	1	228564757	228564757	+	Missense_Mutation	C	T	OCCC_27	p.R7682C
CTNNB1	3	41266097	41266097	+	Missense_Mutation	G	T	OCCC_27	p.D32Y
CTNNB1	3	41275239	41275239	+	Missense_Mutation	C	T	OCCC_27	p.R469C
PIK3CA	3	178951955	178951955	+	Missense_Mutation	A	G	OCCC_27	p.M1004V
PLEC	8	145007424	145007424	+	Missense_Mutation	G	T	OCCC_27	p.F590L
KRT1	12	53070208	53070208	+	Missense_Mutation	G	C	OCCC_27	p.N442K
AHNAK2	14	105412500	105412500	+	Missense_Mutation	G	C	OCCC_27	p.D3096E
AHNAK2	14	105413588	105413588	+	Missense_Mutation	G	C	OCCC_27	p.L2734V
AHNAK2	14	105416455	105416455	+	Missense_Mutation	A	G	OCCC_27	p.M1778T
PPP2R1A	19	52715983	52715983	+	Missense_Mutation	G	A	OCCC_27	p.R183Q
MAPK8IP2	22	51041925	51041925	+	Nonsense_Mutation	C	T	OCCC_27	p.Q149*
ARID1A	1	27023904	27023904	+	Nonsense_Mutation	G	A	OCCC_05	p.W337*
FLG2	1	152328546	152328546	+	Missense_Mutation	C	A	OCCC_05	p.L572F
FLG2	1	152328547	152328547	+	Missense_Mutation	A	G	OCCC_05	p.L572S
PIK3CA	3	178928226	178928226	+	Missense_Mutation	C	T	OCCC_05	p.P471L
UNC13B	9	35403903	35403903	+	Missense_Mutation	G	A	OCCC_05	p.R1550Q
ANK3	10	61829919	61829919	+	Missense_Mutation	G	A	OCCC_05	p.R3574C
AHNAK	11	62288810	62288810	+	Missense_Mutation	C	T	OCCC_05	p.S4360N
HRNR	1	152190900	152190900	+	Missense_Mutation	T	A	OCCC_29	p.S1069C
HRNR	1	152191862	152191862	+	Missense_Mutation	A	G	OCCC_29	p.L748S
HRNR	1	152191964	152191964	+	Missense_Mutation	C	G	OCCC_29	p.S714T

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
HRNR	1	152193378	152193378	+	Missense_Mutation	C	T	OCCC_29	p.G243S
FLG	1	152277315	152277315	+	Missense_Mutation	C	G	OCCC_29	p.E3349D
OBSCN	1	228494989	228494989	+	Missense_Mutation	C	A	OCCC_29	p.Q4075K
MUC17	7	100679059	100679059	+	Missense_Mutation	G	C	OCCC_29	p.K1454N
MUC17	7	100679919	100679919	+	Missense_Mutation	A	C	OCCC_29	p.N1741T
AHNAK	11	62290470	62290470	+	Missense_Mutation	C	T	OCCC_29	p.V3807M
CASKIN1	16	2228904	2228904	+	Splice_Site	G	A	OCCC_29	p.R1400C
OBSCN	1	228504548	228504548	+	Missense_Mutation	C	T	OCCC_64	p.P4475L
OBSCN	1	228526694	228526694	+	Missense_Mutation	G	A	OCCC_64	p.R5742H
LRP1B	2	141457870	141457870	+	Missense_Mutation	T	C	OCCC_64	p.N2250D
PIK3CA	3	178952085	178952085	+	Missense_Mutation	A	G	OCCC_64	p.H1047R
PLEC	8	145008841	145008841	+	Missense_Mutation	C	T	OCCC_64	p.D472N
CDK12	17	37687306	37687306	+	Missense_Mutation	C	T	OCCC_64	p.R1404C
LAMA5	20	60892464	60892464	+	Missense_Mutation	C	T	OCCC_64	p.R2483H
HELZ2	20	62191879	62191879	+	Missense_Mutation	C	T	OCCC_64	p.E2485K
TCHH	1	152083325	152083325	+	Missense_Mutation	A	T	OCCC_60	p.L790M
OBSCN	1	228559450	228559450	+	Missense_Mutation	G	A	OCCC_60	p.G6991R
PIK3CA	3	178936094	178936094	+	Missense_Mutation	C	G	OCCC_60	p.Q546E
PLEC	8	144999935	144999935	+	Missense_Mutation	G	A	OCCC_60	p.R1525W
AHNAK	11	62291300	62291300	+	Missense_Mutation	T	C	OCCC_60	p.K3530R
KRAS	12	25398284	25398284	+	Missense_Mutation	C	T	OCCC_60	p.G12D
CASKIN1	16	2239305	2239305	+	Missense_Mutation	G	T	OCCC_60	p.N140K
MINK1	17	4794986	4794986	+	Missense_Mutation	G	A	OCCC_60	p.R659H
SPTBN4	19	40978640	40978640	+	Missense_Mutation	G	A	OCCC_60	p.A38T
PPP2R1A	19	52715983	52715983	+	Missense_Mutation	G	T	OCCC_60	p.R183L
HELZ2	20	62191410	62191410	+	Missense_Mutation	T	C	OCCC_60	p.T2566A
HELZ2	20	62194713	62194713	+	Missense_Mutation	A	T	OCCC_60	p.L1821Q
FLG	1	152278643	152278643	+	Missense_Mutation	A	G	OCCC_57	p.W2907R
FLG	1	152282506	152282506	+	Missense_Mutation	T	A	OCCC_57	p.Y1619F
FLG	1	152282507	152282507	+	Missense_Mutation	A	G	OCCC_57	p.Y1619H
FLG	1	152284081	152284081	+	Missense_Mutation	G	T	OCCC_57	p.P1094H
FLG2	1	152326837	152326837	+	Missense_Mutation	G	C	OCCC_57	p.A1142G
MUC4	3	195506887	195506887	+	Missense_Mutation	A	G	OCCC_57	p.M3855T
MSH3	5	80064770	80064770	+	Missense_Mutation	G	A	OCCC_57	p.R734Q
AHNAK	11	62292801	62292801	+	Missense_Mutation	A	T	OCCC_57	p.F3030I
AHNAK	11	62295216	62295216	+	Missense_Mutation	C	T	OCCC_57	p.V2225M
KRAS	12	25398281	25398281	+	Missense_Mutation	C	T	OCCC_57	p.G13D
AHNAK2	14	105411448	105411448	+	Missense_Mutation	C	T	OCCC_57	p.R3447K
AHNAK2	14	105413093	105413093	+	Missense_Mutation	C	A	OCCC_57	p.V2899L
CDC27	17	45214689	45214689	+	Missense_Mutation	C	T	OCCC_57	p.R581Q
HRNR	1	152191577	152191577	+	Missense_Mutation	C	T	OCCC_13	p.R843Q
FLG2	1	152326978	152326978	+	Missense_Mutation	T	C	OCCC_13	p.N1095S
PIK3CA	3	178936091	178936091	+	Missense_Mutation	G	A	OCCC_13	p.E545K
DST	6	56480838	56480838	+	Missense_Mutation	G	A	OCCC_13	p.A2476V
MUC17	7	100681363	100681363	+	Missense_Mutation	C	A	OCCC_13	p.F2222L
AHNAK	11	62296059	62296059	+	Missense_Mutation	C	A	OCCC_13	p.V1944L
KRAS	12	25398281	25398281	+	Missense_Mutation	C	T	OCCC_13	p.G13D
MUC19	12	40835768	40835768	+	Nonsense_Mutation	A	T	OCCC_13	p.K706*
MUC19	12	40878843	40878843	+	Missense_Mutation	A	G	OCCC_13	p.E3463G
AHNAK2	14	105412294	105412294	+	Missense_Mutation	A	G	OCCC_13	p.L3165S
PPP2R1A	19	52716326	52716326	+	Missense_Mutation	G	T	OCCC_13	p.W257L
EVPL	17	74006625	74006625	+	Splice_Site	-	CTGACTTCTGGGCCCT TCCTTCCTTT TTTTTTTTTTTTTTTT	OCCC_13	
ARID1A	1	27100369	27100369	+	Missense_Mutation	A	G	OCCC_56	p.M1361V
FLG	1	152286281	152286281	+	Missense_Mutation	C	T	OCCC_56	p.A361T
PIK3CA	3	178952085	178952085	+	Missense_Mutation	A	G	OCCC_56	p.H1047R
PTPRN2	7	157449185	157449185	+	Missense_Mutation	G	A	OCCC_56	p.T687M
MUC19	12	40878258	40878258	+	Missense_Mutation	C	T	OCCC_56	p.T3268I
MUC19	12	40878260	40878260	+	Missense_Mutation	A	G	OCCC_56	p.T3269A
KRT10	17	38975298	38975298	+	Missense_Mutation	C	A	OCCC_56	p.G497C
TCHH	1	152084549	152084549	+	Missense_Mutation	G	C	OCCC_09	p.Q382E
HRNR	1	152191783	152191783	+	Missense_Mutation	G	C	OCCC_09	p.H774Q
FLG	1	152277106	152277106	+	Missense_Mutation	C	T	OCCC_09	p.R3419Q
FLG	1	152279642	152279642	+	Missense_Mutation	G	C	OCCC_09	p.Q2574E
FLG	1	152279952	152279952	+	Missense_Mutation	A	T	OCCC_09	p.S2470R
OBSCN	1	228432152	228432152	+	Missense_Mutation	G	C	OCCC_09	p.G1121R

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
OBSCN	1	228467542	228467542	+	Missense_Mutation	C	T	OCCC_09	p.R2473W
OBSCN	1	228476583	228476583	+	Missense_Mutation	G	A	OCCC_09	p.V3445M
OBSCN	1	228509289	228509289	+	Missense_Mutation	G	A	OCCC_09	p.R4916Q
CELSR3	3	48679349	48679349	+	Missense_Mutation	C	T	OCCC_09	p.R2920Q
SHROOM3	4	77662516	77662516	+	Missense_Mutation	G	A	OCCC_09	p.D1064N
SHROOM3	4	77662664	77662664	+	Missense_Mutation	G	A	OCCC_09	p.R1113H
MUC17	7	100683102	100683102	+	Missense_Mutation	C	T	OCCC_09	p.T2802I
MUC17	7	100683800	100683800	+	Missense_Mutation	G	A	OCCC_09	p.G3035S
MUC17	7	100684875	100684875	+	Missense_Mutation	G	C	OCCC_09	p.S3393T
PTPRN2	7	157370768	157370768	+	Missense_Mutation	C	T	OCCC_09	p.R854Q
UNC13B	9	35396918	35396918	+	Missense_Mutation	G	A	OCCC_09	p.R1090Q
UNC13B	9	35403978	35403978	+	Missense_Mutation	G	A	OCCC_09	p.R1575Q
AHNAK	11	62303477	62303477	+	Missense_Mutation	C	T	OCCC_09	p.V32I
AHNAK2	14	105410716	105410716	+	Missense_Mutation	G	T	OCCC_09	p.P3691H
AHNAK2	14	105414686	105414686	+	Missense_Mutation	C	T	OCCC_09	p.V2368I
AHNAK2	14	105414938	105414938	+	Missense_Mutation	C	T	OCCC_09	p.V2284M
VASP	19	46021211	46021211	+	Missense_Mutation	C	T	OCCC_09	p.R68W
HELZ2	20	62195085	62195085	+	Missense_Mutation	G	A	OCCC_09	p.A1697V
MAPK8IP2	22	51042637	51042637	+	Missense_Mutation	G	A	OCCC_09	p.A38T
MAPK8IP2	22	51045156	51045156	+	Missense_Mutation	G	A	OCCC_09	p.R466Q
SPTA1	1	158637741	158637741	+	Nonsense_Mutation	C	A	OCCC_36	p.E649*
PIK3CA	3	178916876	178916876	+	Missense_Mutation	G	A	OCCC_36	p.R88Q
PIK3CA	3	178952072	178952072	+	Missense_Mutation	A	G	OCCC_36	p.M1043V
MUC17	7	100683008	100683008	+	Missense_Mutation	G	C	OCCC_36	p.V2771L
KRT5	12	52910517	52910517	+	Missense_Mutation	C	T	OCCC_36	p.R448Q
HRNR	1	152187900	152187900	+	Missense_Mutation	C	T	OCCC_02	p.G2069S
HRNR	1	152191425	152191425	+	Missense_Mutation	C	T	OCCC_02	p.G894S
FLG	1	152282147	152282147	+	Missense_Mutation	G	T	OCCC_02	p.Q1739K
FLG	1	152282753	152282753	+	Missense_Mutation	G	A	OCCC_02	p.P1537S
FLG	1	152283598	152283598	+	Missense_Mutation	T	C	OCCC_02	p.E1255G
FLG	1	152285686	152285686	+	Missense_Mutation	T	C	OCCC_02	p.H559R
PIK3CA	3	178936082	178936082	+	Missense_Mutation	G	A	OCCC_02	p.E542K
MUC4	3	195506438	195506438	+	Missense_Mutation	T	C	OCCC_02	p.S4005G
AHNAK2	14	105416839	105416839	+	Missense_Mutation	G	T	OCCC_02	p.A1650E
CASKIN1	16	2229188	2229188	+	Missense_Mutation	C	T	OCCC_02	p.R1305Q
MINK1	17	4788808	4788808	+	Missense_Mutation	G	A	OCCC_02	p.R180H
HELZ2	20	62203578	62203578	+	Missense_Mutation	G	C	OCCC_02	p.A54G
FLG2	1	152327408	152327408	+	Missense_Mutation	T	C	OCCC_24	p.R952G
FLG2	1	152328778	152328778	+	Missense_Mutation	C	T	OCCC_24	p.C495Y
SPTA1	1	158605725	158605725	+	Missense_Mutation	G	A	OCCC_24	p.L1804F
OBSCN	1	228469825	228469825	+	Missense_Mutation	C	T	OCCC_24	p.R2797W
SHROOM3	4	77677618	77677618	+	Missense_Mutation	A	G	OCCC_24	p.K1576E
MUC17	7	100675451	100675451	+	Missense_Mutation	C	G	OCCC_24	p.Q252E
MUC17	7	100676667	100676667	+	Missense_Mutation	A	G	OCCC_24	p.N657S
MUC17	7	100677995	100677995	+	Missense_Mutation	A	G	OCCC_24	p.S1100G
MUC17	7	100678029	100678029	+	Missense_Mutation	G	C	OCCC_24	p.R1111T
PTPRN2	7	157929370	157929370	+	Missense_Mutation	C	T	OCCC_24	p.D384N
KRT5	12	52912805	52912805	+	Missense_Mutation	C	T	OCCC_24	p.S232N
PPP2R1A	19	52716323	52716323	+	Missense_Mutation	C	T	OCCC_24	p.S256F
CDC27	17	45232154	45232154	+	Splice_Site	-	TGGGGTTAATG	OCCC_24	
DSP	6	7580658	7580658	+	Missense_Mutation	T	A	OCCC_28	p.L1412Q
DSP	6	7585432	7585432	+	Missense_Mutation	C	T	OCCC_28	p.T2646M
LAMA5	20	60909062	60909062	+	Missense_Mutation	G	A	OCCC_28	p.P925S
MUC4	3	195506386	195506386	+	Missense_Mutation	G	A	OCCC_65	p.P4022L
AHNAK	11	62298252	62298252	+	Missense_Mutation	C	T	OCCC_65	p.V1213M
KRAS	12	25398285	25398285	+	Missense_Mutation	C	A	OCCC_65	p.G12C
DST	6	56417688	56417688	+	Missense_Mutation	G	A	OCCC_10	p.S5090L
MUC17	7	100677582	100677582	+	Missense_Mutation	C	G	OCCC_10	p.T962S
FLG2	1	152327579	152327579	+	Missense_Mutation	T	A	OCCC_58	p.S895C
SPTA1	1	158592886	158592886	+	Missense_Mutation	A	T	OCCC_58	p.S2003T
OBSCN	1	228433270	228433270	+	Missense_Mutation	G	A	OCCC_58	p.R1213H
OBSCN	1	228506756	228506756	+	Missense_Mutation	C	T	OCCC_58	p.S4768L
PIK3CA	3	178936082	178936082	+	Missense_Mutation	G	A	OCCC_58	p.E542K
MUC4	3	195506555	195506555	+	Missense_Mutation	C	T	OCCC_58	p.A3966T
SHROOM3	4	77676308	77676308	+	Missense_Mutation	C	G	OCCC_58	p.Q1558E
MUC17	7	100680105	100680105	+	Missense_Mutation	T	C	OCCC_58	p.L1803P

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
AHNAK	11	62295576	62295576	+	Missense_Mutation	G	A	OCCC_58	p.L2105F
AHNAK2	14	105414686	105414686	+	Missense_Mutation	C	T	OCCC_58	p.V2368I
AHNAK2	14	105419180	105419180	+	Missense_Mutation	A	G	OCCC_58	p.S870P
HELZ2	20	62191383	62191383	+	Missense_Mutation	G	A	OCCC_58	p.R2575W
NEURL1B	5	172113300	172113301	+	Splice_Site	GT	-	OCCC_58	
FLG	1	152278594	152278594	+	Missense_Mutation	A	T	OCCC_54	p.L2923Q
FLG2	1	152323277	152323277	+	Nonsense_Mutation	G	A	OCCC_54	p.Q2329*
MUC19	12	40876126	40876126	+	Missense_Mutation	T	G	OCCC_54	p.H2557Q
CDC27	17	45235598	45235598	+	Missense_Mutation	G	T	OCCC_54	p.S150Y
DSP	6	7570774	7570804	+	Splice_Site	TCAGGGCCATGACAATC GCCAAGGTATGTCC	-	OCCC_54	p.IRAMTIAK560fs
FLG2	1	152326057	152326057	+	Missense_Mutation	C	G	OCCC_12	p.G1402A
FLG2	1	152326058	152326058	+	Missense_Mutation	C	A	OCCC_12	p.G1402C
SPTA1	1	158606445	158606445	+	Missense_Mutation	C	T	OCCC_12	p.E1766K
MUC19	12	40878306	40878306	+	Missense_Mutation	G	A	OCCC_12	p.G3284E
ARID1A	1	27099958	27099958	+	Nonsense_Mutation	T	A	OCCC_40	p.Y1279*
FLG	1	152280083	152280083	+	Missense_Mutation	C	T	OCCC_40	p.A2427T
FLG2	1	152325292	152325292	+	Missense_Mutation	C	G	OCCC_40	p.S1657T
FLG2	1	152328192	152328192	+	Missense_Mutation	A	C	OCCC_40	p.H690Q
CELSR3	3	48698864	48698864	+	Missense_Mutation	G	A	OCCC_40	p.R402C
PIK3CA	3	178936082	178936082	+	Missense_Mutation	G	A	OCCC_40	p.E542K
PIK3CA	3	178938935	178938935	+	Missense_Mutation	A	C	OCCC_40	p.E726A
MSH3	5	79950724	79950724	+	Missense_Mutation	G	C	OCCC_40	p.A60P
DSP	6	7558461	7558461	+	Missense_Mutation	G	A	OCCC_40	p.R129Q
DST	6	56485116	56485116	+	Missense_Mutation	C	T	OCCC_40	p.R1239H
MUC17	7	100676360	100676360	+	Missense_Mutation	C	G	OCCC_40	p.P555A
PLEC	8	144997144	144997144	+	Missense_Mutation	G	A	OCCC_40	p.A2455V
AHNAK2	14	105411236	105411236	+	Missense_Mutation	C	T	OCCC_40	p.A3518T
AHNAK2	14	105413372	105413372	+	Missense_Mutation	T	C	OCCC_40	p.M2806V
AHNAK2	14	105414134	105414134	+	Missense_Mutation	G	A	OCCC_40	p.P2552S
AHNAK2	14	105414343	105414343	+	Missense_Mutation	C	T	OCCC_40	p.R2482K
AHNAK2	14	105418818	105418818	+	Missense_Mutation	G	C	OCCC_40	p.D990E
XPO6	16	28167850	28167850	+	Splice_Site	T	A	OCCC_40	
EVPL	17	74005187	74005187	+	Missense_Mutation	C	T	OCCC_40	p.E1367K
EYA2	20	45801390	45801390	+	Missense_Mutation	C	T	OCCC_40	p.S358L
HELZ2	20	62191321	62191321	+	Missense_Mutation	A	T	OCCC_40	p.N2595K
MAGEE1	X	75648993	75648993	+	Missense_Mutation	G	A	OCCC_40	p.V224M
MAGEE1	X	75650068	75650068	+	Missense_Mutation	G	T	OCCC_40	p.G582V
RPTN	1	152129136	152129136	+	Missense_Mutation	C	T	OCCC_23	p.G147S
FLG	1	152282120	152282120	+	Missense_Mutation	G	T	OCCC_23	p.Q1748K
FLG2	1	152327858	152327858	+	Missense_Mutation	T	C	OCCC_23	p.S802G
MUC17	7	100676109	100676109	+	Missense_Mutation	A	C	OCCC_23	p.N471T
AHNAK	11	62295801	62295801	+	Missense_Mutation	T	C	OCCC_23	p.M2030V
AHNAK2	14	105410322	105410322	+	Missense_Mutation	A	C	OCCC_23	p.I3822M
AHNAK2	14	105419653	105419653	+	Missense_Mutation	A	G	OCCC_23	p.L712P
AHNAK2	14	105419654	105419654	+	Missense_Mutation	G	A	OCCC_23	p.L712F
CDC27	17	45235670	45235670	+	Splice_Site	-	AATATACA	OCCC_23	
FLG	1	152284655	152284655	+	Missense_Mutation	T	C	OCCC_38	p.R903G
OBSCN	1	228464942	228464942	+	Missense_Mutation	G	A	OCCC_38	p.E2228K
SHROOM3	4	77661952	77661952	+	Missense_Mutation	C	T	OCCC_38	p.R876C
DST	6	56329528	56329528	+	Missense_Mutation	G	A	OCCC_38	p.R7252C
DST	6	56481864	56481864	+	Missense_Mutation	G	A	OCCC_38	p.T2134M
PLEC	8	144996830	144996830	+	Missense_Mutation	C	T	OCCC_38	p.A2560T
AHNAK	11	62292766	62292766	+	Missense_Mutation	A	T	OCCC_38	p.D3041E
AHNAK	11	62294381	62294381	+	Missense_Mutation	G	C	OCCC_38	p.A2503G
SPTBN2	11	66461717	66461717	+	Missense_Mutation	C	T	OCCC_38	p.V1466M
XPO6	16	28187305	28187305	+	Missense_Mutation	G	A	OCCC_38	p.R107W
PPP2R1A	19	52716301	52716301	+	Missense_Mutation	C	T	OCCC_38	p.R249C

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
ARID1A	1	27099889	27099889	+	Frame_Shift_Del	G	-	OCCC_22	p.M1256fs
DMRTB1	1	53925429	53925441	+	Frame_Shift_Del	GACTCCCTCCGGA	-	OCCC_22	p.GTPSG101fs
DSPP	4	88535833	88535850	+	In_Frame_Del	TAGCAGTGACAGCAGCAG	-	OCCC_22	p.SSDSSS674del
MUC19	12	40876443	40876475	+	In_Frame_Del	CAGCTGGAGTGACAGGTAC AAATGGACCATCAT	-	OCCC_22	p.AGVTGTNGPSS2664del
MAPK8IP2	22	51042073	51042073	+	Frame_Shift_Ins	-	GAGGCTTTGACCTGGTGCGT	OCCC_22	p.G156fs

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
							CCGGCCTAAGCCTCCGTTGTT		
ARID1A	1	27023145	27023162	+	In_Frame_Del	GCGGCGGCGGAGCCGGCA	-	OCCC_37	p.GGGAGS85del
LOR	1	153233487	153233510	+	In_Frame_Del	GCGGCGGTGGCGGTGGCG GCGGCA	-	OCCC_37	p.GGGGGGGS22del
ZNF717	3	75787230	75787313	+	In_Frame_Del	GTTTTCCCACATTCATTGC ATTCGTAGGGTTTTTCCCC TGTGTGAGTCCATTGATGG ATAGTGAGGAATGACTTACG GTGAAAC	-	OCCC_37	p.437_465TFHRKSFL TIHQWTHTGEKPYECNECGKT>T
DST	6	56470589	56470589	+	In_Frame_Ins	-	TAT	OCCC_37	p.2734_2735insN
PLEC	8	145003635	145003679	+	In_Frame_Del	GCAGCCGCAGGCGGTGCACGGT GCGCGTCTCACAGGCCTCCAGCT	-	OCCC_37	p.QLEACETRTVHRLRL1132del
ANK3	10	61828704	61828745	+	In_Frame_Del	TGGTGGTGGTGGTAGTGGTGGT AGTGGTGGTGGTGGTGGCAG	-	OCCC_37	p.TATTTTTTTTTTTT3965del
KRTAP5-2	11	1618958	1619017	+	In_Frame_Del	ACTGGCAGCACACAGGGACACAA CAGTTGGACTGGCAGCAACAGGG CTTGCAGCAGCTGG	-	OCCC_37	p.SSCCKPCCCQSNCCVPVCCQ155del
MUC19	12	40878865	40878924	+	In_Frame_Del	ATCACCTGGGGTGACAGGGACAAC TGGATCATCAACTGGGGTGACAGG GATAACTGGATT	-	OCCC_37	p.SPGVTGTTGSSTGVTGITGL3471del
KRT5	12	52913704	52913781	+	In_Frame_Del	CCACCTCCAAAGCCAGCTCCGCC ACCGAGCCCAAAGCCACCACCAGC TCCACCGCCGAAACCAAATCCACT ACCGGCA	-	OCCC_37	p.100_126GAGSGFGFGGGAGGG FGLGGGAGFGGG>G
SPTB	14	65216035	65216035	+	Frame_Shift_Ins	-	G	OCCC_37	p.K2326fs
CDK12	17	37687471	37687471	+	Frame_Shift_Ins	-	G	OCCC_37	p.W1459fs
MAGEE1	X	75648505	75648540	+	In_Frame_Del	CTGAGGGCCCAAGCACCTCCGT TCTGCCCACCTCCG	-	OCCC_37	p.EGPSTSVLPTSA62del
ARID1A	1	27099418	27099418	+	Frame_Shift_Ins	-	A	OCCC_50	p.D1219fs
ARID1A	1	27106877	27106877	+	Frame_Shift_Ins	-	C	OCCC_50	p.C2163fs
TCHH	1	152080845	152080868	+	In_Frame_Del	CTCCTGGCGCAGCTGTTGTTGGCC	-	OCCC_50	p.GQQQLRQE1609del
FLG	1	152282104	152282104	+	Frame_Shift_Ins	-	CACGTGTGGACTCTTGGTGGC TCTGCTGATGGGGCCCAGCCA CCAAGAGTCCACACGTGGCCAG TCAGGGGAAAGGTCTGGA	OCCC_50	p.G1753fs
FLG2	1	152323402	152323402	+	Frame_Shift_Ins	-	CTGGCTGTCTTTGTTGAGATCC AGCTTGGCCCTGAATGTGTCCTGA ATGTGTGTGTGAG	OCCC_50	p.G2287fs
SPTA1	1	158641210	158641210	+	Frame_Shift_Ins	-	GAAGAGAAGA	OCCC_50	p.L508fs
CELSR3	3	48677161	48677206	+	Frame_Shift_Del	GAGGGCCCAAGCACAGAGGCTGTGG CAGAAGGTGTGGCAGTGGTGT	-	OCCC_50	p.HTTATPSATASVLGPS3271fs
SHROOM3	4	77677654	77677655	+	Frame_Shift_Del	GC	-	OCCC_50	p.A1589fs
DSPP	4	88535832	88535832	+	In_Frame_Ins	-	CAGTAGTGACAGCAGCAG	OCCC_50	p.672_673insAVVTAA
DSPP	4	88536263	88536277	+	In_Frame_Del	GATAGCGACAGCAGC	-	OCCC_50	p.DSDSS817del
PLEC	8	144997994	144998029	+	In_Frame_Del	GCCGCGCCGACTCCTGCTCCGCTCGC TCCCGCAGGC	-	OCCC_50	p.RLRERAEQESAR2160del
MUC19	12	40873358	40873387	+	In_Frame_Del	ACAACTGGACCATCAGCTGACGGGTCAGGA	-	OCCC_50	p.TTGPSADGSG1635del
MUC19	12	40878199	40878228	+	In_Frame_Del	AACTGGACTATCAGCTGAAGCAACAGAGAT	-	OCCC_50	p.TGLSAEATEI3249del
MINK1	17	4789807	4789848	+	In_Frame_Del	CCCACGGAGCAGCTACTGAAGTTTCCCTT CATCCGGGACCAG	-	OCCC_50	p.PTEQLLKFPFIRDQ279del
KRT10	17	38975120	38975137	+	In_Frame_Del	CCGCCGCCGGAGCTGCTG	-	OCCC_50	p.550_556GSSSGGG>G
EVPL	17	74010547	74010551	+	Frame_Shift_Del	TCGCT	-	OCCC_50	p.SD777fs
EVPL	17	74023206	74023232	+	In_Frame_Del	TTGGGGGACCCCTTGGCGGGGGAGCCC	-	OCCC_50	p.16_25KGSPAKGSPK>K
SPTBN4	19	41062156	41062156	+	Frame_Shift_Ins	-	A	OCCC_50	p.G1751fs
MAPK1	22	22221709	22221714	+	In_Frame_Del	CCGCCG	-	OCCC_50	p.AA6del
TCHH	1	152081063	152081063	+	In_Frame_Ins	-	CTGGCGGCGCAGCTGCTGTT CCTCCTGGAGGAATTTTCTCT GCCGTTG	OCCC_04	p.1543_1544insQRQRKFL QEEQQLRRQ
FLG2	1	152327352	152327352	+	In_Frame_Ins	-	TGACCTGAGCCTGATCCATGTTG GCCAAAGCCAGAGGAT	OCCC_04	p.970_970Q>QSSGFGQHGSGSGQ
FLG2	1	152328234	152328234	+	In_Frame_Ins	-	CCTGAGCCAGAAACATGTTGTCC AAAGCCAGAGGACTGA	OCCC_04	p.676_676G>GQSSGFGQHVSGSG
MUC4	3	195512933	195512933	+	In_Frame_Ins	-	GGTGGTGTGACCTGAAGATGCTG AGGAAGGGATGGTGA CAGGAAGAGAGGTGGTGTCACCT GTGGATGCTGAGGAA GCGTCGGTGACAGGAAGAGG	OCCC_04	p.1839_1840insPLPVTDASSASTG DTTSLPVTIPSSASSGHTT

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
DSPP	4	88535719	88535719	+	In_Frame_Ins	-	AGTGACAGTAAGTCAGACAGCAGT GACAGCAACAGC	OCCC_04	p.636_636S>RVTVSQTAVTATA
DSPP	4	88536388	88536388	+	In_Frame_Ins	-	AGTGACAGTAGTAATAGTAGTGAC AGCAGCGATAGCAG TGACAGCAGCAACAGC	OCCC_04	p.859_859S>RVTVVIVVTAAI AVTAATA
MUC19	12	40876933	40876933	+	In_Frame_Ins	-	ACAGGGACAACTGGACCATCACCT GGAGTAACAGGTAC AACTGGAACACCAGCTGGGGTT	OCCC_04	p.2827_2828insQGQLDHHLE* QVQLEHQLGL
MUC19	12	40877751	40877751	+	In_Frame_Ins	-	TGGATCATCAGCTGGGG TGACAGGGAAAAC	OCCC_04	p.3099_3099I>MDHQLG*QGKL
ARID1A	1	27100182	27100182	+	Frame_Shift_Del	G	-	OCCC_47	p.P1326fs
LRP1B	2	141093297	141093297	+	Frame_Shift_Ins	-	CAGTGAGTAGTGTAGTAA CTGAACCAATGCATTCTAG AATGCATTGGTT	OCCC_47	p.W4001fs
MUC17	7	100678287	100678287	+	In_Frame_Ins	-	CAGTTCACCTCCTCCAAC TGCTGAAGTTACCAG	OCCC_47	p.1197_1197A>AVHLLQLLKLPA
MUC19	12	40877705	40877764	+	In_Frame_Del	GGGGTGACAGGGACAACTGGAC TATCAGCTGGGGAGACAGGGAA AATTGGATCATCAGCT	-	OCCC_47	p.GVTGTTGLSAGETGKIGSS A3084del
KRT1	12	53073801	53073830	+	In_Frame_Del	CCACCAAAGCCACCACCACCAA AGCCACCA	-	OCCC_47	p.101_111GGGFGGGGFGG>G
AHNAK2	14	105419178	105419178	+	Frame_Shift_Del	G	-	OCCC_47	p.S871fs
MINK1	17	4793912	4793932	+	In_Frame_Del	AACAGCAGCAGCAGCTTCAGA	-	OCCC_47	p.QQQQLQK484del
HELZ2	20	62196884	62196884	+	In_Frame_Ins	-	ACG	OCCC_47	p.1098_1099insV
FLG	1	152280877	152280878	+	Frame_Shift_Del	CC	-	OCCC_25	p.G2162fs
MUC4	3	195506651	195506652	+	Frame_Shift_Del	CA	-	OCCC_25	p.A3934fs
TCHH	1	152083757	152083795	+	In_Frame_Del	GCTGCTGGCGCCTCTCCTCCT GCTCCTCGCTCTTCAGCA	-	OCCC_01	p.LLKSEEQEERRQQ633del
FLG2	1	152324557	152324557	+	Frame_Shift_Ins	-	TG	OCCC_01	p.S1902fs
FLG2	1	152326888	152326926	+	In_Frame_Del	GACTGACCTGAGCCCGATCCA TATTGGCCAAAGCCAGAG	-	OCCC_01	p.1112_1125SSGFGQYGSGSGQS>S
LOR	1	153233584	153233628	+	In_Frame_Del	CTCCGGCGGCGGTGGCTACTCT GGCGGCGGCTGCGGCGGGGGCTC	-	OCCC_01	p.SGGGGYSGGGCGGGS54del
SPTA1	1	158581061	158581061	+	Frame_Shift_Del	C	-	OCCC_01	p.G2418fs
OBSCN	1	228520964	228520964	+	Frame_Shift_Del	C	-	OCCC_01	p.L5266fs
OBSCN	1	228523964	228523964	+	In_Frame_Ins	-	AGGAAAGGTACAGTCAGGGT GGGTGCATGCTTGACTGTAC CTTTCCTCTGCCCACCA	OCCC_01	p.5511_5512insGKVQSGWVHA* LYLSSAHQ
DSPP	4	88536974	88536982	+	In_Frame_Del	AGCAGCAAT	-	OCCC_01	p.SSN1054del
DST	6	56437786	56437786	+	In_Frame_Ins	-	CTTTTTGACTTTTTTTTTTT	OCCC_01	p.4227_4227T>KKKKSQKA
MUC19	12	40876900	40876900	+	Frame_Shift_Ins	-	GTGACAGGGACAACTAGACTA TCAGCTGGAGTGACAGTCT	OCCC_01	p.-2816fs
MUC19	12	40877413	40877442	+	In_Frame_Del	GACAGGGACAACTGGACC ATCAGCTGGGGC	-	OCCC_01	p.TGTTGPSAGA2987del
WWP2	16	69967951	69967951	+	In_Frame_Ins	-	TGCCTGCAGATCAACCCCGC CTCCTCCATCAACCCGGGT	OCCC_01	p.588_589insACRSTPPPPSTRV
WWP2	16	69972971	69972971	+	Frame_Shift_Ins	-	CAGTTTGTCACCGGTACCTG CCGCCTGCCCGGTGACAAAC TGCAGCAGCCGGATC	OCCC_01	p.Q796fs
CDK12	17	37687209	37687209	+	Frame_Shift_Ins	-	AAGAACAGGACCTTCTCAGGC TCTCTGAGCCACCTTGGAAGG TCCTGTTCTTCACCAGGGTC	OCCC_01	p.-1372fs
HELZ2	20	62197069	62197069	+	Frame_Shift_Ins	-	CACCACGTCTTCCTTCACTGC GTCTCCTGCTGGTGAAGGAAG ACGTGGTGCCCGGGGCACCACGT	OCCC_01	p.P1036fs
DSPP	4	88536515	88536520	+	In_Frame_Del	AGTGAT	-	OCCC_32	p.SD903del
MUC19	12	40877650	40877679	+	In_Frame_Del	GACAGGGACAACTGAACC ATTAGCTGGAGG	-	OCCC_32	p.TGTTEPLAGG3066del
ARID3A	19	971949	971949	+	In_Frame_Ins	-	GCA	OCCC_32	p.555_556insA
RPTN	1	152128233	152128268	+	In_Frame_Del	TCTGGCCTTGTCTGTCTG GCTGACCATAGTGGGAAC	-	OCCC_34	p.SSHYGQPDRQGQ436del
ARID1A	1	27105826	27105826	+	Frame_Shift_Del	C	-	OCCC_35	p.P1813fs
TCHH	1	152083743	152083781	+	In_Frame_Del	CTCGCGCCTTAGTTGCTGC TGGCGCCTCTCCTCCTGCTC	-	OCCC_35	p.EQEERRQQQLRRE638del
RPTN	1	152127670	152127675	+	In_Frame_Del	TCCCTG	-	OCCC_35	p.QG634del
OBSCN	1	228560130	228560171	+	In_Frame_Del	GTCCCAGTCAGAGGAGGAGGA GCAGGAGGAGGCCAGGGCTGA	-	OCCC_35	p.SQSEEEEQEEARAE7218del
DSPP	4	88537521	88537556	+	In_Frame_Del	ACAGCAGTGACAGCAGCGATA	-	OCCC_35	p.SSDSSDSSDSSN1237del

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
						GCAGTGACAGCAGCA			
DSP	6	7542206	7542206	+	In_Frame_Ins	-	CCGAGTCTGGCCCGGACCTGCG CTACGAGGTGACCAGGT	OCCC_35	p.19_20insPSLARTCATR*PG
DST	6	56417210	56417210	+	Frame_Shift_Del	A	-	OCCC_35	p.V5249fs
AHNAK	11	62291447	62291461	+	In_Frame_Del	TTGGGCATTTTCATC	-	OCCC_35	p.3476_3481KMKMPK>K
MUC19	12	40875869	40875898	+	In_Frame_Del	CAATCAGCTGGAGTCACAATG ACATCTATC	-	OCCC_35	p.QSAGVTMTSI2472del
MUC19	12	40877650	40877679	+	In_Frame_Del	GACAGGGACAACTGAACCATT AGCTGGAGG	-	OCCC_35	p.TGTTEPLAGG3066del
MUC19	12	40878242	40878271	+	In_Frame_Del	GCTGGGGTGACAGGGACAACT GGACTATCT	-	OCCC_35	p.AGVTGTTGLS3263del
KRT5	12	52908926	52908949	+	In_Frame_Del	CAAGACCTCCACCGAGGCCGCCGC	-	OCCC_35	p.GGGLGGGL517del
SPTB	14	65236348	65236348	+	Frame_Shift_Ins	-	CAAGCTCCAGGCAGGCACTGAAGT TCTTGCTACGGAGCTTGGCGAGT	OCCC_35	p.G1966fs
AHNAK2	14	105408153	105408182	+	In_Frame_Del	GTGCCCTTTGAGGCCGGCTACC TCGGGCAT	-	OCCC_35	p.MPEVAGLKGH4536del
WWP2	16	69965477	69965478	+	Frame_Shift_Del	CC	-	OCCC_35	p.FQ529fs
MINK1	17	4797305	4797337	+	In_Frame_Del	GTCAGCACCATGGTGGTCCACGA CGTCGAGGAG	-	OCCC_35	p.VSTMVVHDVEE863del
MINK1	17	4797342	4797368	+	In_Frame_Del	CCGGGACCCAGCCCCCATACGG GGGCG	-	OCCC_35	p.875_884TGTQPPYGGG>S
ARID3A	19	968463	968465	+	In_Frame_Del	CAT	-	OCCC_35	p.I519del
SPTBN4	19	41009746	41009747	+	Frame_Shift_Del	GG	-	OCCC_35	p.G458fs
SPTBN4	19	41009747	41009747	+	In_Frame_Ins	-	TTT	OCCC_35	p.458_458G>VW
VASP	19	46025639	46025674	+	In_Frame_Del	CCAGGACCTCCCCCTCCTCCA GGTCCCCCCCCACCC	-	OCCC_35	p.PGPPPPPGPPPP174del
MAGEE1	X	75648778	75648813	+	In_Frame_Del	TGCCGCCCACCGCCTCTGAG GTACCGAGCACCTCCC	-	OCCC_35	p.PPTASEVPSTSL153del
RPTN	1	152129116	152129151	+	In_Frame_Del	GTCTTGTCTCTCAGGCTGAC CATGGTGGGAATCTCC	-	OCCC_03	p.GDSHHGQPERQD142del
SPTA1	1	158641157	158641157	+	Frame_Shift_Ins	-	AAGGCTTCCTCAAAGTCTTCATGC TTTGTCTTCATGCTTCTGAAGCCC TTCTTCAGAAGCATGAAGACTTT	OCCC_03	p.F525fs
HRNR	1	152191631	152191669	+	In_Frame_Del	CCTGAGCCAGACTCGTGTTG CCCAAAACCAGAAGCCTGG	-	OCCC_69	p.812_825GQASGFGQHESGSG>G
HRNR	1	152192822	152192860	+	In_Frame_Del	TGGCCGTGGCCTGGAGACT GGCCAGATCCAGAGCTGTGT	-	OCCC_69	p.415_428QHSSGSGQSPGHGQ>Q
HRNR	1	152193038	152193076	+	In_Frame_Del	CCTGAGCCAGACTCATGTTG CCCAAAGCCAGAAGTCTGG	-	OCCC_69	p.343_356GQTSGFGQHESGSG>G
FLG	1	152281747	152281749	+	In_Frame_Del	TTC	-	OCCC_69	p.K1872del
LRP1B	2	141771184	141771184	+	Frame_Shift_Ins	-	GCAATGTCACCTCACTTGTTATCA AATCTAGTTGAGGTGACATTGCTG AGGCATGAAAGACCACCCCTATTTGG	OCCC_69	p.L774fs
DST	6	56469384	56469384	+	Frame_Shift_Ins	-	CAATTCAGGAATCTGAACCTCCGCC TTGTCTCCAAGATCCCAAGAAAAGG AGGTTCAGATCCCAAGAAAAGGAGGTT	OCCC_69	p.S3137fs
MUC17	7	100677572	100677572	+	In_Frame_Ins	-	CTGAAGCCACTTCATCTCCTACAAC TGCTGAAGGTACCAGCAGTT	OCCC_69	p.958_959insLKPLHLLQLLKVPAV
PLEC	8	144992791	144992791	+	In_Frame_Ins	-	GGAAGCCTGTGGCTGCCTGTGCCA CAGGCTTCCTGCTGGACCCGGTGA AGGGGGAGC	OCCC_69	p.3869_3870insRSPFTGS SRKPVAQAATGF
PLEC	8	144993014	144993014	+	Frame_Shift_Ins	-	GATGATCCGAGCCTCGAACAGGTCC TCAGCCGTGAGGCGGCGGCGCACCT	OCCC_69	p.-3795fs
AHNAK	11	62293497	62293497	+	In_Frame_Ins	-	GACATCAATGTCAGCCTTGGGCAGG TTCACATTGATGTCTCAGGACCGAAA GTGGATGTTGAATGTCCCGAT	OCCC_69	p.2797_2798insIGTFNIHFRS* DINVNLPKADIDV
SPTBN2	11	66459096	66459096	+	In_Frame_Ins	-	CCGGGAGAACTCTCGGAATTTGTCT CGGAGCATCTGCTTCCTCTACCAGAT GCTCCGAGACAAATT	OCCC_69	p.1741_1742insNLSRSIW* RKQMLRDKFREFSR
SPTBN2	11	66481836	66481836	+	In_Frame_Ins	-	CTTCTCCTTGTTGTCTTCTGTCTCC ACACTGATGACAACAAGGAGAAGAAGT CAGCCAAGGATGCCCTG	OCCC_69	p.175_176insQGILG*LLLL VVISVETEDNKEK
MUC19	12	40876495	40876524	+	In_Frame_Del	AACTGGATCATCACCTGGA GTGACAGGGAC	-	OCCC_69	p.TGSSPGVTGT2691del
MUC19	12	40877680	40877709	+	In_Frame_Del	GACAGGGACAACTGGAAT ATTAGCCGGGGT	-	OCCC_69	p.TGTTGILAGV3076del
EVPL	17	74004643	74004643	+	In_Frame_Ins	-	TGCGCTCCCTGTTGAGCATCTCCCA CACGCGGGCCC	OCCC_69	p.1547_1548insRARVWEMLNRER

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
SPTBN4	19	41066181	41066181	+	Frame_Shift_Ins	-	GCCTGTGAGGATGCCCGCCTGCATG TCAGCTCCACAGCCGACAT	OCCC_69	p.-1930fs
TCHH	1	152084176	152084193	+	In_Frame_Del	TGCTGCTCGCGCCTCTCC	-	OCCC_63	p.500_506QERREQQ>Q
RPTN	1	152127299	152127340	+	In_Frame_Del	CTGTCTCGTCTCTGATGGC TCTGCTCATGT TCATGGGTTTGT	-	OCCC_63	p.745_759RQTHEHEQSHQRRDR>R
RPTN	1	152129036	152129107	+	In_Frame_Del	GAATCTCTGTCTTGTCTCTC AGGCTGACTG TGGTGGGAATCTCTGTCTTG TTTCTCAGAC TGACCATGGTGG	-	OCCC_63	p.156_180SHHGQSEKQDRDSHH SQPERQDRDS>S
OBSCN	1	228560156	228560191	+	In_Frame_Del	AGGAGGCCAGGGCTGAGTC CCAGTCGGAG GAGCAGC	-	OCCC_63	p.EARAESQSEEQQ7227del
CELSR3	3	48677161	48677206	+	Frame_Shift_Del	GAGGGCCCAAGCACAGAGGC TGTGGCAGA AGGTGTGGCAGTGGTGT	-	OCCC_63	p.HTTATPSATASVLGPS3271fs
SHROOM3	4	77662005	77662005	+	In_Frame_Ins	-	CTCTCCAGCGAGCCGGGCCTGTC CCGCCACTCGGGCTCC	OCCC_63	p.894_895insSPASRACPATRAP
SPTBN2	11	66463902	66463903	+	Frame_Shift_Del	TT	-	OCCC_63	p.K1375fs
MUC19	12	40878780	40878869	+	In_Frame_Del	CTGGGGTGACAGGGAAAACTGGACTATCT GCTGGAGTGACAGAGACAATTGGACTATC AGCTGAAGCGACAGGGACAATTGGATCA TCAC	-	OCCC_63	p.GVTGKTGLSAGVTETIGLSA EATGTIGSSP3443del
KRT5	12	52908926	52908949	+	In_Frame_Del	CAAGACCTCCACCGAGGCCGCCGC	-	OCCC_63	p.GGGLGGGL517del
KRT1	12	53073827	53073853	+	In_Frame_Del	ACCACCACCAAAGCCACCACCACCATA	-	OCCC_63	p.YGGGGFGGG94del
ERBB2	17	37866709	37866709	+	Frame_Shift_Ins	-	GCCAGCTGTGTGACTGCCTGTC CCTGTGAGT	OCCC_63	p.-293fs
MAPK1	22	22123530	22123530	+	Frame_Shift_Ins	-	CAAAAATTAGTTCTTTGAGCT AATTTTTGAAGAGACTG	OCCC_63	p.E349fs
AHNAK	11	62296259	62296259	+	Frame_Shift_Del	A	-	OCCC_67	p.V1877fs
AHNAK2	14	105405032	105405032	+	Frame_Shift_Ins	-	ACAGC	OCCC_67	p.Q5586fs
AHNAK2	14	105419006	105419020	+	In_Frame_Del	TGGGCATCTTGAAAC	-	OCCC_67	p.SFKMP923del
SPTBN4	19	41074024	41074101	+	In_Frame_Del	GCGGCGGCCGGAGCGGCAGGAGTCAGCGGA GCACGAGGCGGCACACAGCCTTACCCTGGGC CGCTATGAGCAGATGGA	-	OCCC_67	p.RRPERQESAEHEAAHSLTL GRYEQME2265del
KRT10	17	38975164	38975166	+	In_Frame_Del	GCC	-	OCCC_41	p.G541del
HRNR	1	152187804	152187842	+	In_Frame_Del	AGCCAGGCCCATGTTGGCCACTGCTGGAAG ACCGACCGG	-	OCCC_17	p.SGRSSSSGQHGPG2088del
LOR	1	153233825	153233923	+	In_Frame_Del	GGCGGCTCCTCCGGGGGCGGCTCCGGCT GCTTCTCCTCCGGCGGCGGCGGCTTCTC GGGCCAGGCGGTCCAGTGCCAGAGCTACG GAGGCGTCTCTAGC	-	OCCC_17	p.GGSSGGGSGCFSSGGGGFSG QAVQCQSYGGVSS134del
LRP1B	2	141055447	141055447	+	Frame_Shift_Ins	-	CAGGTTCCTCCATTTTGACAAAA ATGGAGGAACCTGCATTGTGA	OCCC_17	p.-4299fs
MUC4	3	195510219	195510266	+	In_Frame_Del	TGAGGAAGTCTCGGTGACAAGAAGAGGGG TGGTGTCACCTGTGGATGA	-	OCCC_17	p.SSTGDTTPLLVTETSS2729del
DSPP	4	88535504	88535518	+	In_Frame_Del	AGTGACAGCAGTGAC	-	OCCC_17	p.SDSSD569del
NEURL1B	5	172113743	172113743	+	Frame_Shift_Ins	-	TCAG	OCCC_17	p.P495fs
DST	6	56481871	56481871	+	Frame_Shift_Ins	-	CTTGTTAAGAGTTTTCTG AACTTCTTCAACT	OCCC_17	p.F2132fs
PLEC	8	144997814	144997843	+	In_Frame_Del	CCTCCGCCTCCTCAGCCGCCCGCCGGGCCG	-	OCCC_17	p.AARRAAEEAE2222del
KRT1	12	53069244	53069303	+	In_Frame_Del	ACCTCCGGAGCCATAGCTGCCACGGCCGCC GCCGCCGCCACCTCCAGAACCATAGCTACC	-	OCCC_17	p.GSYGSGGGGGGGRGSYGSGG537del
MINK1	17	4797309	4797368	+	In_Frame_Del	GCACCATGGTGGTCCACGACGTCGAGGAG ATCACCGGGACCCAGCCCCCATACGGGGGCG	-	OCCC_17	p.TMVVHDVEEITGTQPPYGGG865del
SPTBN4	19	41018722	41018730	+	In_Frame_Del	GCGGGCGCA	-	OCCC_17	p.AGA679del
SPTBN4	19	41018936	41018982	+	Frame_Shift_Del	GCGCGGCGAGCGCCCGGCGCCGCTGGCAG AGGCTGGAAGAGGCGGCG	-	OCCC_17	p.RAASARRRWQRLEEAA747fs
LAMA5	20	60904886	60904886	+	In_Frame_Ins	-	ACCGTGCGT	OCCC_17	p.1355_1356insTHG
RPTN	1	152129044	152129151	+	In_Frame_Del	GTCTTGTCTCTCAGGCTGACTGTGGTGGGA ATCTCTGTCTTGTTTCTCAGACTGACCATGG TGGGAATCTCTGTCTTGTCTCTCAGGCTGAC CATGGTGGGAATCTCC	-	OCCC_18	p.GDSHHGQPERQDRDSHHG QSEKQDRDSHHSQPERQD142del
LRP1B	2	141986949	141986949	+	Frame_Shift_Ins	-	AACAATA	OCCC_18	p.Y218fs
LRP1B	2	141986951	141986951	+	Frame_Shift_Ins	-	TGGTGCA	OCCC_18	p.-217fs
SHROOM3	4	77680731	77680731	+	Frame_Shift_Ins	-	A	OCCC_18	p.N1745fs
DSPP	4	88535567	88535626	+	In_Frame_Del	AGTGACAGCAGTGATAGCAGTGACAGTGATA	-	OCCC_18	p.SDSSDSSDSDSSDSSNSSDS585del

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
						GTAGTGATAGCAGCAATAGCAGTGACAGT			
PLEC	8	144996883	144996883	+	Frame_Shift_Ins	-	AG	OCCC_18	p.R2542fs
AHNAK2	14	105416683	105416683	+	Frame_Shift_Ins	-	TGAGG	OCCC_18	p.K1702fs
HRNR	1	152187854	152187931	+	In_Frame_Del	TGTCGGCCATAGCTGGGAGACTGCCTTGAC CCAGACCCACGCTGGCCGTGGCCTGGAGAC TGGCCAGATCCAGAGCCC	-	OCCC_48	p.QGSGSGQSPGHGQRGSGS RQSPSYGR2058del
MUC4	3	195508174	195508174	+	In_Frame_Ins	-	GAAGAGGGGTGGCGTGACCTG TGGATGCTGAGGAAGGGCTGG TGACAT	OCCC_48	p.3425_3426insHVTSPSSASTGHATPL
UNC13B	9	35236530	35236530	+	Frame_Shift_Ins	-	CCATGGTGGGGACTGTGTGGATT GCGCTGAAGACTATTCTGACGAAT AGTCTTCAGCGCAAT	OCCC_48	p.T73fs
SPTBN2	11	66472923	66472923	+	In_Frame_Ins	-	TCGCAAGGTCTATACTCTGAGAAA GTCCCTCTCCTGTGACTT	OCCC_48	p.607_608insESHRRGTFSEYRPC
MAPK8IP2	22	51041768	51041768	+	In_Frame_Ins	-	GAG	OCCC_48	p.97_98insR
RPTN	1	152127563	152127563	+	Frame_Shift_Ins	-	AGAGACTGGCAATCATGCAGTA	OCCC_66	p.P671fs
CELSR3	3	48686212	48686223	+	In_Frame_Del	GGCCAGCAGGTG	-	OCCC_66	p.HLLA2236del
ANK3	10	61828704	61828706	+	In_Frame_Del	TGG	-	OCCC_66	p.T3978del
MUC19	12	40873080	40873109	+	In_Frame_Del	TATCAGGGACAACTGGACCATTAGCTGAAA	-	OCCC_66	p.SGTTGPLAEI1543del
MUC19	12	40873358	40873387	+	In_Frame_Del	ACAACTGGACCATCAGCTGACGGGTCAGGA	-	OCCC_66	p.TTGPSADGSG1635del
CTNNB1	3	41266973	41266973	+	Frame_Shift_Ins	-	TGGGACCTTGCATAACCT TTCCCATC ATCGTGATGGCCAGTAAG CCCTCACGATGA	OCCC_19	p.A215fs
MUC4	3	195514433	195514433	+	Frame_Shift_Ins	-	GACGCTTCCTCAGTATCCACAGGTCA CACCACCCCTCTTCA	OCCC_19	p.G1340fs
DSPP	4	88536764	88536781	+	In_Frame_Del	GATAGCAGTGACAGCAGT	-	OCCC_19	p.DSSDSS996del
AHNAK	11	62287913	62287927	+	In_Frame_Del	TTGGGCATTTTCACT	-	OCCC_19	p.4654_4659KVKMPK>K
KRT10	17	38978482	38978550	+	In_Frame_Del	CCACCAAAGCTGCCCCCACCAAAGCTGCC ACCTCCGAAACTGCCCCCTCCAAAGATGC CTCCATAACTC	-	OCCC_19	p.96_119GSYGGIFGGGSFGGG SFGGGSFGG>G
SPTBN4	19	41040166	41040166	+	In_Frame_Ins	-	CTCCTTCACATGGAGAGCCAG CTGCAAGACGTGGTCTCCTCC AGGGTCCACGTCTTGCAGCTGGCT	OCCC_19	p.1426_1426L>LSFTWRASCK TWSPPGSTSCSWL
MAGEE1	X	75649425	75649460	+	In_Frame_Del	GTGCCGCCCACCGCCTCTGATGGATCGG ACACCTCC	-	OCCC_19	p.VPPTASDGSDTS368del
OBSCN	1	228560130	228560171	+	In_Frame_Del	GTCCCAGTCAGAGGAGGAGGAGCAGGAG GAGGCCAGGGCTGA	-	OCCC_51	p.SQSEEEEQEEARAE7218del
LRP1B	2	141457914	141457914	+	Frame_Shift_Del	C	-	OCCC_51	p.R2235fs
MUC4	3	195489005	195489067	+	In_Frame_Del	GAGGCGTGGAGGATGTTGGAGAGCGCG ATCACCGAGACGGTGGCCCAGCCGTCGA AGCTGGCC	-	OCCC_51	p.565_586SASFDGWATVS VIALSNILHAS>S
MSH3	5	79950724	79950724	+	In_Frame_Ins	-	CAGCGC	OCCC_51	p.59_60insQR
MUC17	7	100681304	100681304	+	Frame_Shift_Ins	-	CAACTTCTGAAGGTACCAGC ATGCCAACCTT	OCCC_51	p.T2203fs
AHNAK	11	62286529	62286529	+	Frame_Shift_Ins	-	GGTGCCTGGAGTTCACCCTCCAG TTTGGGGCTAGGGAGAG	OCCC_51	p.-5120fs
MUC19	12	40873080	40873109	+	In_Frame_Del	TATCAGGGACAACTGGACCATTAGCTGAAA	-	OCCC_51	p.SGTTGPLAEI1543del
MUC19	12	40878944	40878973	+	In_Frame_Del	ACAACTGGATTGTCAACTGAAGTGACAGGA	-	OCCC_51	p.TTGLSTEVTG3497del
AHNAK2	14	105415500	105415520	+	In_Frame_Del	GGGGCCCTTGATGTCCACCTG	-	OCCC_51	p.QVDIKGP2090del
CDK12	17	37627658	37627711	+	In_Frame_Del	ACCCCTCCACCTCTTCCCACAATTGCTTCT CCCCCACCCCCTCTACCAACTACT	-	OCCC_51	p.TPPPLPTIASPPPPLPTT525del
ERBB2	17	37881624	37881624	+	Frame_Shift_Ins	-	AGAA	OCCC_51	p.F899fs
EVPL	17	74010547	74010551	+	Frame_Shift_Del	TCGCT	-	OCCC_51	p.SD777fs
SPTBN4	19	41018722	41018730	+	In_Frame_Del	GCGGGCGCA	-	OCCC_51	p.AGA679del
MAGEE1	X	75648566	75648601	+	In_Frame_Del	GCCCACCATCTCTGAGGCCTCAAGCGCC TCCGGGCA	-	OCCC_51	p.PTISEASSASGQ82del
TCHH	1	152080326	152080397	+	In_Frame_Del	GCGCAGCTGCTGTTCCTCCCTCTCCTGG CGGAGCTGTTCCTCCTCGCGGAATTTTCT GTCGCGCTCCTGGCG	-	OCCC_33	p.RQERDRKFREEEQLRQE REEQQLR1766del
HRNR	1	152192096	152192096	+	Frame_Shift_Ins	-	TGGAA	OCCC_33	p.G670fs
FLG	1	152276467	152276467	+	In_Frame_Ins	-	GGA	OCCC_33	p.3631_3632insL
OBSCN	1	228521376	228521376	+	Frame_Shift_Ins	-	CCCTGGAGATCATCTCCGTCACCC GGGAGGACTCTGGCCAGAGTCCT	OCCC_33	p.T5317fs
CELSR3	3	48681045	48681060	+	Frame_Shift_Del	GCCAGGAGCCCAAAGA	-	OCCC_33	p.LFGLLA2739fs
CELSR3	3	48685368	48685369	+	Frame_Shift_Del	GC	-	OCCC_33	p.R2345fs
DSPP	4	88535504	88535518	+	In_Frame_Del	AGTGACAGCAGTGAC	-	OCCC_33	p.SDSSD569del
PLEC	8	144997814	144997843	+	In_Frame_Del	CCTCCGCCTCCTCAGCCGCCC	-	OCCC_33	p.AARRAAEEAE2222del

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
						GCCGGGCCG			
KRT10	17	38978482	38978496	+	In_Frame_Del	CCACCAAAGCTGCCC	-	OCCC_33	p.114_119GGSFGG>G
EVPL	17	74005040	74005080	+	Frame_Shift_Del	CGCCGGCCCGCAGCTGCTGCACCTCAAG CTCTAGCTGGCGC	-	OCCC_33	p.RQLELEVQQLRAGV1403fs
HELZ2	20	62195541	62195541	+	In_Frame_Ins	-	ACTCGCTGCCCACCAG GAACTCAGCGAG TGCACGCGGAGTTCCT GGTGGGCAGC GAGTGCACGCGG	OCCC_33	p.1544_1545insSACTRCPPGT PRALAEFLVGSE
ARID1A	1	27087533	27087533	+	Frame_Shift_Del	C	-	OCCC_06	p.P703fs
TCHH	1	152080564	152080564	+	In_Frame_Ins	-	GGAATTTTCTCTCTCGTTCCTGAC GGCGGAGCTGCTGTTCCTCTTCGC	OCCC_06	p.1709_1710insREEEQQLRRQERERKF
DSPP	4	88536046	88536046	+	In_Frame_Ins	-	GATAGCAGTGACAGCAGCAACAGCA GTGACAGTAGC	OCCC_06	p.745_745D>EIAVTAATAVTVA
PLEC	8	144997640	144997696	+	In_Frame_Del	CCGCCTGTGCCCGCCGCGCCGC CTCTTGCTCGGCCTCCTTGCGC AGCTTCTCTGCAG	-	OCCC_06	p.AAEKLRKEAEQEAARRAQA2271del
MUC19	12	40877181	40877181	+	In_Frame_Ins	-	AACTAGACCATCAGCTGGGGTAACAGG GACAACAG GACTATCAGCTGAAGTGACAGAGAT	OCCC_06	p.2909_2909T>KLDHQLG*Q GQQDYQLK*QRS
MUC19	12	40878586	40878586	+	In_Frame_Ins	-	ACTGGACTATCAGCTGAGGTGACAGGGACA	OCCC_06	p.3378_3379insLDYQLR*QGH
KRT1	12	53073844	53073844	+	In_Frame_Ins	-	ACCACCATAACCACCACCAAAGCCACT	OCCC_06	p.96_97insSGFGGGYGG
ARID1A	1	27087544	27087544	+	Frame_Shift_Ins	-	A	OCCC_59	p.A707fs
SPTA1	1	158617450	158617450	+	In_Frame_Ins	-	CTCAGTGGCATCTGGATGGGAC TCACTGAGCCG	OCCC_59	p.1258_1259insRLSESHPDATE
NEURL1B	5	172110551	172110551	+	Frame_Shift_Ins	-	CCAGCTTGGCCACCACCTGGTTGTT	OCCC_59	p.-236fs
DSP	6	7576632	7576633	+	Frame_Shift_Del	AG	-	OCCC_59	p.E913fs
PLEC	8	144999765	144999765	+	Frame_Shift_Ins	-	ATCGAGGAGGAGA	OCCC_59	p.E1581fs
XPO6	16	28167475	28167475	+	Frame_Shift_Ins	-	ACCAAGGATAACAA	OCCC_59	p.Q339fs
ARID1A	1	27100182	27100187	+	In_Frame_Del	GCAGCA	-	OCCC_44	p.QQ1333del
ARID1A	1	27101132	27101132	+	Frame_Shift_Del	G	-	OCCC_44	p.A1472fs
OBSCN	1	228403319	228403319	+	Frame_Shift_Ins	-	GAGGTCACTTTCTCCGTGGACCTC CCGCTGAG GCCACCGTGAGGTCCACCGT	OCCC_44	p.-629fs
OBSCN	1	228404312	228404312	+	Frame_Shift_Ins	-	CTCCTTGTGCGAGATGTGTCCC GGGCCACAT	OCCC_44	p.-763fs
OBSCN	1	228505627	228505627	+	Frame_Shift_Ins	-	GCCCCTGAGAGCCGGCAGGTGGCAGCTGGT GACATCTTCACCAGCT	OCCC_44	p.A4629fs
SHROOM3	4	77675891	77675891	+	Frame_Shift_Ins	-	GGGTCTCGCTGCCTCAGTGGCCACCTC CTTCTCGAGCAAAGTGGGT	OCCC_44	p.R1419fs
MUC17	7	100678734	100678735	+	Frame_Shift_Del	CC	-	OCCC_44	p.T1347fs
SPTBN2	11	66463844	66463844	+	Frame_Shift_Ins	-	GCACAGCAGCTCTGAGCTGTTTGCCCA GAGCTGCTGTGCCCTGGAGA	OCCC_44	p.-1394fs
PPP2R1A	19	52719102	52719102	+	Frame_Shift_Ins	-	CTGTGAGGCCGAGGTGAGGGCCGC AGCCTCCCACAAGGAGG	OCCC_44	p.D293fs
DSPP	4	88535833	88535850	+	In_Frame_Del	TAGCAGTGACAGCAGCAG	-	OCCC_15	p.SSDSSS674del
DMRTB1	1	53925474	53925474	+	Frame_Shift_Ins	-	CTCG	OCCC_49	p.-117fs
DSPP	4	88536774	88536818	+	In_Frame_Del	ACAGCAGTGATAGCAGTGA CAGCAGTGA CAGCAGTGATAGCAGCA	-	OCCC_49	p.SSDSSDSSDSSDSSN1003del
UNC13B	9	35397682	35397682	+	Frame_Shift_Ins	-	CCAGCCTATTGCACAAAGGAGAAAC TGGTAGGTTCAGGCCCTGGGACTA	OCCC_49	p.-1161fs
AHNAK	11	62290020	62290020	+	In_Frame_Ins	-	CAGGTTCACATCCACTTCTGGACCT TCTCCTTAAGTGGATGTGAACCTGCC CAAGGCTGACCTTGACGT	OCCC_49	p.3956_3957insTSRSALGRFTS T*GEGPEVDVNL
LAMA5	20	60922041	60922052	+	In_Frame_Del	CGCAGGTGCCCC	-	OCCC_49	p.GGTC330del
ARID1A	1	27105726	27105726	+	Frame_Shift_Del	G	-	OCCC_39	p.E1780fs
SPTA1	1	158654960	158654960	+	In_Frame_Ins	-	CTTCCCCAGATCATCTGCATCTCGCTTG AAAACTTGT AAGTGATAGCTTGAGGATTCCTATCAC TTACAAGTTTT	OCCC_39	p.67_68insKTCK**ESSSYHL QVFKRDADDLGK
RPTN	1	152127835	152127835	+	In_Frame_Ins	-	CTCTGGCCTTGTCTGTCTGTCTGAC CATAATGATAG	OCCC_62	p.580_580S>SYHYGQTDRQGQS
DSPP	4	88535772	88535772	+	In_Frame_Ins	-	CAGTGATAGCAGCGACAGCAGCAATA GCAGTAACAG CAGTGATAGTAGTGACAG	OCCC_62	p.653_653N>TVIAATAAIAVTAVIVVTD
ANK3	10	61828704	61828706	+	In_Frame_Del	TGG	-	OCCC_62	p.T3978del
ANK3	10	61829748	61829748	+	Frame_Shift_Ins	-	TTGGAGCCTCTCTTCAACAAAATCCCTC	OCCC_62	p.F3631fs

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
MINK1	17	4795793	4795793	+	Frame_Shift_Ins	-	TTG CCCAACGCCTCTAGGTAATAGAGT TGTCC	OCCC_62	p.-729fs
EVPL	17	74005156	74005170	+	In_Frame_Del	ACCACCACCTCCTGC	-	OCCC_62	p.1372_1377VQEVVV>V
MUC4	3	195513398	195513398	+	In_Frame_Ins	-	GGTGACAGGAAGAGGGGTGGCGT GAGCTGTGG ATGCTGAGGAAGTGCT	OCCC_31	p.1684_1685insSTSSASTAHATPLPVT
DSPP	4	88534443	88534444	+	Frame_Shift_Del	GG	-	OCCC_31	p.G369fs
DST	6	56434706	56434706	+	Frame_Shift_Ins	-	CAATGGGAACTTTTTTTGTTGTTT CTTTTAT CCATGACTT	OCCC_31	p.V4398fs
KRT5	12	52908978	52908983	+	In_Frame_Del	GCCACT	-	OCCC_31	p.SG506del
ARID1A	1	27106336	27106337	+	Frame_Shift_Del	TG	-	OCCC_11	p.C1983fs
FLG2	1	152326379	152326379	+	Frame_Shift_Ins	-	TGTGTGAATGTGTTCTGAATGTC	OCCC_11	p.Q1295fs
OBSCN	1	228528930	228528930	+	Frame_Shift_Ins	-	TACGTGTTCCGGAACATGATGA AGGTCTG CAGGCTC TGGAACACGTAGCTGACGG	OCCC_11	p.-5945fs
MUC4	3	195475790	195475791	+	Frame_Shift_Del	GG	-	OCCC_11	p.P1103fs
SHROOM3	4	77476849	77476849	+	In_Frame_Ins	-	GCTCCAGAAAGGAGGCAGTTT CCCTGGGT CTTGTAG GATCCTTTCACCAGGGAAACT	OCCC_11	p.85_86insAPERRQFPWVL*DPFTRET
DST	6	56496075	56496075	+	Frame_Shift_Del	G	-	OCCC_11	p.S1148fs
AHNAK	11	62293469	62293469	+	Frame_Shift_Ins	-	GACATTCAACATCCACTTTCGGTCCTGAG ACATCAA TGTCAGCCTTGAACCTGCCCAAGGCT	OCCC_11	p.P2807fs
MUC19	12	40852569	40852569	+	In_Frame_Ins	-	AGGTCAGTATATTAATTCCTGGTT TTACATGTT AAATAAAAACCAGGAATTAATATACTGACCT GTAGAAAA	OCCC_11	p.1114_1115insKVSILIPGFTC *IKTRN*YTDL*K
SPTB	14	65270403	65270403	+	Frame_Shift_Ins	-	ACATTGTAGA	OCCC_11	p.-132fs
AHNAK2	14	105407516	105407517	+	Frame_Shift_Del	GC	-	OCCC_11	p.MQ4757fs
MINK1	17	4800542	4800542	+	Frame_Shift_Ins	-	CTTCATGACTCTGGGGGCAGCAG CCAAGTTTT	OCCC_11	p.Y1320fs
ERBB2	17	37866644	37866644	+	Frame_Shift_Ins	-	CAGTTGCTACCCCTCTGGATCTA ACATGACTT TTTTTTTTTTT	OCCC_11	p.L271fs
ARID1A	1	27101161	27101161	+	Frame_Shift_Del	G	-	OCCC_07	p.M1481fs
TCHH	1	152084549	152084549	+	In_Frame_Ins	-	CTGCTCGCGCCTCAGCTGCTG CTCGCGCCTC TCCTCCTC	OCCC_07	p.381_382insEEERREQQLRREQ
DSPP	4	88536521	88536526	+	In_Frame_Del	AGTGAC	-	OCCC_07	p.SD903del
ARID1A	1	27106630	27106630	+	Frame_Shift_Ins	-	G	OCCC_20	p.C2081fs
TCHH	1	152084210	152084210	+	In_Frame_Ins	-	CTGCTGCTCGCGCCT	OCCC_20	p.494_495insRREQQ
FLG2	1	152326888	152326926	+	In_Frame_Del	GACTGACCTGAGCCCGATC CATATTG GCCAAAGCCAGAG	-	OCCC_20	p.1112_1125SSGFGQYGSGSGQS>S
FLG2	1	152331310	152331313	+	Frame_Shift_Del	TTTG	-	OCCC_20	p.YK16fs
OBSCN	1	228479814	228479814	+	Frame_Shift_Ins	-	TG	OCCC_20	p.L3519fs
CTNNB1	3	41280738	41280776	+	In_Frame_Del	GATGGGCTGCCAGATCTGG GGCATG CCCAGGACCTCATG	-	OCCC_20	p.DGLPDLGHAQDLM751del
SHROOM3	4	77677692	77677698	+	Frame_Shift_Del	ACTCCAA	-	OCCC_20	p.RLQ1600fs
PLEC	8	144999805	144999805	+	In_Frame_Ins	-	CCTCCGAGCTCTGCCGCAGC TGCTGCAGCT	OCCC_20	p.1567_1568insELQQLRQSSE
MUC19	12	40877380	40877409	+	In_Frame_Del	AGTGACAGGGACAACTGGACT GTCAACTGA	-	OCCC_20	p.VTGTTGLSTE2976del
MUC19	12	40877680	40877709	+	In_Frame_Del	GACAGGGACAACTGGAATATTAGCCGGGGT	-	OCCC_20	p.TGTTGILAGV3076del
MUC19	12	40877853	40877882	+	In_Frame_Del	CTGGAGTGACGGGGACAACTGGACTATCAC	-	OCCC_20	p.GVTGTTGLSP3134del
MUC19	12	40878604	40878633	+	In_Frame_Del	GGTGACAGGGACAACTGGACTATCAGTTGG	-	OCCC_20	p.VTGTTGLSVG3384del
AHNAK2	14	105411949	105411949	+	Frame_Shift_Ins	-	A	OCCC_20	p.A3280fs
MINK1	17	4792947	4793018	+	In_Frame_Del	GCGGCGGGAGCGGGAGCAGCGGAAGCTGC AGGAGAAGGAGCAGCAGCGGCGGCTGGAG GACATGCAGGCTCT	-	OCCC_20	p.RREREQRKLQEKEQQRR LEDMQAL413del
KRT10	17	38975137	38975137	+	In_Frame_Ins	-	CCGCCGCCGTAT	OCCC_20	p.550_550G>GYGGG
RPTN	1	152128355	152128390	+	In_Frame_Del	TCTGTCTGACCATAGTGAGAACTTTGGTCT TGTCTG	-	OCCC_14	p.DRQDQSSHYGQT395del

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
DSPP	4	88535459	88535509	+	In_Frame_Del	AGCAGTGACAGTGATAGTAGTGATAGCAGC AATAGCAGTGATAGTAGTGAC	-	OCCC_14	p.SSDSDSSDSSNSSDSSD549del
DSPP	4	88536515	88536520	+	In_Frame_Del	AGTGAT	-	OCCC_14	p.SD903del
UNC13B	9	35310668	35310668	+	In_Frame_Ins	-	GCAGAGAAGGAGGCAGCATGTGAACCCA AGGAGATGAAAGCT	OCCC_14	p.323_324insQRRRQHVNPRR*KL
TCHH	1	152084406	152084477	+	In_Frame_Del	CTGCTCGCGCCTCAGCTGCTGCTCGCGCC TCAGCTGCTGCTCGCGCCTCAGCTGCTGCT CGCGCCTCAGCTG	-	OCCC_43	p.QLRREQQLRREQQLRREQ QLRREQ406del
SPTA1	1	158639269	158639270	+	Frame_Shift_Del	AC	-	OCCC_43	p.Y588fs
OBSCN	1	228505740	228505740	+	Frame_Shift_Ins	-	TGGGAATGGAGCGCATCCAGCCCGT	OCCC_43	p.G4666fs
CELSR3	3	48694195	48694196	+	Frame_Shift_Del	GC	-	OCCC_43	p.R1445fs
DSP	6	7581743	7581743	+	Frame_Shift_Ins	-	AG	OCCC_43	p.Q1774fs
PLEC	8	144998456	144998488	+	In_Frame_Del	CCAGCCGCCGCCGCTGGAAGGCCTCGT CCTCCG	-	OCCC_43	p.AEDEAFQRRRL2007del
PLEC	8	145003857	145003857	+	In_Frame_Ins	-	GAGCCGTACTCGCGCTCAGCCATCA GCCGGTCCTCGGCT	OCCC_43	p.1097_1097S>SAEDRLMAEREYGS
UNC13B	9	35375191	35375191	+	Frame_Shift_Del	A	-	OCCC_43	p.E455fs
AHNAK	11	62291447	62291461	+	In_Frame_Del	TTGGGCATTTTCATC	-	OCCC_43	p.3476_3481KMKMPK>K
AHNAK2	14	105408570	105408571	+	Frame_Shift_Del	GG	-	OCCC_43	p.P4406fs
EVPL	17	74023206	74023232	+	In_Frame_Del	TTGGGGGACCCCTTGGCGGGGGAGCCC	-	OCCC_43	p.16_25KGSPAKGSPK>K
HELZ2	20	62196712	62196712	+	Frame_Shift_Ins	-	GGGGCCCGAGGAGGCATCGTCCAGCGG GATGGCTGACGCCTCCTCGGGCCCCAT CCAGGTCA	OCCC_43	p.I1155fs
HELZ2	20	62196752	62196752	+	Frame_Shift_Ins	-	TCCT	OCCC_43	p.-1141fs
MAGEE1	X	75649528	75649528	+	In_Frame_Ins	-	CTCCGTGCTGCCTAACCCTGGT GAGGGCCCTCAATGTGCTCGGGCCCT	OCCC_43	p.402_402S>TPCCLTLVRALNVLGPC
TCHH	1	152080254	152080325	+	In_Frame_Del	GCGCAGCTGCTGTTCTTCCCTCTCCT GGCGTAGCTGTTCCTCCTCGCGGAAT TTTCTGTCAGAC TCTTGGCT	-	OCCC_46	p.SQESDRKFREEEQLRQ EREEQQLR1790del
TCHH	1	152082863	152082883	+	In_Frame_Del	GCAGCTGCTCTTCCTCCTGCT	-	OCCC_46	p.QQEEEQL937del
HRNR	1	152191328	152191366	+	In_Frame_Del	GAGCCAGACCCATGTCGGCCAC TGCTGGAAGACCGACCG	-	OCCC_46	p.913_926SGRSSSSGRHGSGS>S
FLG2	1	152326621	152326659	+	In_Frame_Del	TGACCTGAGCCTGAACCATATT GGCCAAATCCAGTGGAC	-	OCCC_46	p.1201_1214QSTGFGQYGSGSGQ>Q
FLG2	1	152327758	152327796	+	In_Frame_Del	TGTCCAAAGCCAGAGGATTGTCC TGAGCCAGACCCATGT	-	OCCC_46	p.822_835QHGSGSGQSSGFGQ>Q
FLG2	1	152327846	152327848	+	In_Frame_Del	CAG	-	OCCC_46	p.805_806TG>S
PIK3CA	3	178922313	178922315	+	In_Frame_Del	ACC	-	OCCC_46	p.H362del
MUC4	3	195538627	195538638	+	In_Frame_Del	AGGCAGAGACAC	-	OCCC_46	p.17_21LCLCL>L
DST	6	56357771	56357781	+	Frame_Shift_Del	GAGACTTGGCC	-	OCCC_46	p.RPSL6514fs
AHNAK	11	62289760	62289760	+	In_Frame_Ins	-	TCCACTTTGGGGCCCTTGATGTC AACTTCAGGGGCCTTTAGATCACCT	OCCC_46	p.4042_4043insEGDLKAPEVDIKGPKV
MUC19	12	40877680	40877709	+	In_Frame_Del	GACAGGGACAACTGGAATAT TAGCCGGGGT	-	OCCC_46	p.TGTTGILAGV3076del
KRT5	12	52908926	52908949	+	In_Frame_Del	CAAGACCTCCACCGAGGCCGCCGC	-	OCCC_46	p.GGGLGGGL517del
SPTB	14	65216761	65216796	+	In_Frame_Del	CAGGGCCAGGGGTTCCTCCCCATGGT AGGGCATCCC	-	OCCC_46	p.GMPYHGEEPLAL2227del
WWP2	16	69874149	69874149	+	In_Frame_Ins	-	CAGTGCCCTGACAGATGGTGAGT GCCGCCCTGCTCCTCAGTCCTGAG GAGCAGGGCGGCACTCACCATCTGT	OCCC_46	p.154_154G>AVP*QMVSAALL LSPEEQGGTHHLC
ERBB2	17	37884039	37884040	+	Frame_Shift_Del	CA	-	OCCC_46	p.K1171fs
LAMA5	20	60898688	60898689	+	Frame_Shift_Del	CC	-	OCCC_46	p.G1963fs
LAMA5	20	60902992	60902995	+	Frame_Shift_Del	GGGC	-	OCCC_46	p.RP1575fs
MAGEE1	X	75649234	75649341	+	In_Frame_Del	TGAGCACCTCCGTGCAGCCCACTGC TGGTGAGGGAT CGAGCACCTCCGTGCCGCCCACCCC TGGTGGGGGA CTGAGCACCTCCGTGCCGCCCACCG CCACTGAGGAGT	-	OCCC_46	p.304_340LSTSVQPTAGEGSS TSVPPTPGGGLSTSVPPTATEEL>L
TCHH	1	152084549	152084549	+	In_Frame_Ins	-	CTGCTCGCGCCTCTCCTCCTC	OCCC_21	p.381_382insEEERREQ
OBSCN	1	228560130	228560171	+	In_Frame_Del	GTCCCAGTCAGAGGAGGAGGAGCAG GAGGAGGCCAGG GCTGA	-	OCCC_21	p.SQSEEEEQEEARAE7218del
EVPL	17	74004871	74004871	+	Frame_Shift_Ins	-	GGTCCTTCTCCAGCTTGACCAC TTCCTCCATGATGATCTTCTCCC ACGGTGCAGGAGAAGATCATCAT GGAGGAAGT	OCCC_21	p.P1472fs

(continued on next page)

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
ARID1A	1	27106589	27106589	+	Frame_Shift_Ins	-	C	OCCC_53	p.I2067fs
TCHH	1	152080606	152080606	+	In_Frame_Ins	-	CCC	OCCC_53	p.1695_1696insG
TCHH	1	152084176	152084193	+	In_Frame_Del	TGCTGCTCGCGCCTCTCC	-	OCCC_53	p.500_506QERREQQ>Q
HRNR	1	152188530	152188530	+	In_Frame_Ins	-	GCG	OCCC_53	p.1858_1859insR
FLG2	1	152328664	152328702	+	In_Frame_Del	TGTCCAAAACCAGAGGATTGT CCTGAGACAGACCC ATGC	-	OCCC_53	p.520_533QHGSVSGQSSGFGQ>Q
OBSCN	1	228444399	228444443	+	In_Frame_Del	CAGGCCCAGGCGGGGGCC AGCACCACACTCAGCT GCGAGGTGGCT	-	OCCC_53	p.QAQAGASTTLSCEVA1453del
OBSCN	1	228562291	228562291	+	Frame_Shift_Ins	-	C	OCCC_53	p.A7501fs
DSP	6	7542183	7542183	+	Frame_Shift_Ins	-	CACTCTGGGCCGCATGATCCGCG CCGAGTCTGGCCCGGAT	OCCC_53	p.N12fs
ANK3	10	62023687	62023687	+	Frame_Shift_Ins	-	GCACTTTTCCTTTGGTGTCATTCA AAGGAAAAGTGCGTCTCCCA	OCCC_53	p.R202fs
AHNAK	11	62293578	62293578	+	Frame_Shift_Del	T	-	OCCC_53	p.M2771fs
MUC19	12	40876788	40876817	+	In_Frame_Del	GGACAACTGGACTATCACC TGGAGTGACAA	-	OCCC_53	p.TTGLSPGVTR2779del
MUC19	12	40877680	40877709	+	In_Frame_Del	GACAGGGACAACTGGAATAT TAGCCGGGGT	-	OCCC_53	p.TGTTGILAGV3076del
MUC19	12	40878635	40878664	+	In_Frame_Del	GTGACAGGGATAGCTGGACT CTCAGCTGGC	-	OCCC_53	p.VTGIAGLSAG3394del
AHNAK2	14	105411019	105411019	+	Frame_Shift_Del	G	-	OCCC_53	p.A3590fs
MINK1	17	4789807	4789848	+	In_Frame_Del	CCCACGGAGCAGCTACTGAA GTTTCCCTTCAT CCGGGACCAG	-	OCCC_53	p.PTEQLLKFPFIRDQ279del
EVPL	17	74023206	74023232	+	In_Frame_Del	TTGGGGGACCCCTTGGCGG GGGAGCCC	-	OCCC_53	p.16_25KGSPAKGSPK>K
LAMA5	20	60897166	60897166	+	Frame_Shift_Ins	-	G	OCCC_53	p.P2135fs
ARID1A	1	27023116	27023116	+	Frame_Shift_Del	G	-	OCCC_26	p.Q74fs
HRNR	1	152192595	152192595	+	Frame_Shift_Ins	-	CCTAGATGACTGAACAAACCTG AGCTAGATCCGTGTTGTTCACT	OCCC_26	p.-503fs
FLG2	1	152324722	152324722	+	Frame_Shift_Ins	-	TAGTTCCATGTCTCTCGTCAAC TATGGATTCTGACTCTCCAGG CTGGATCTCAACATGGAGAGT CAGAATCCA	OCCC_26	p.T1847fs
DSPP	4	88536737	88536772	+	In_Frame_Del	GACAGCAGTGACAGCAGCAACAGCAGCGAT AGCAGT	-	OCCC_26	p.DSSDSSNSSDSS975del
CDK12	17	37627658	37627711	+	In_Frame_Del	ACCCCTCCACCTCTTCCCACAATTGCTTCT CCCCCACCCCCTCTACCAACTACT	-	OCCC_26	p.TPPPLPTIASPPPPLPTT525del
HELZ2	20	62196806	62196806	+	In_Frame_Ins	-	CGCAGCAGC	OCCC_26	p.1123_1123H>QLLR
HRNR	1	152193038	152193076	+	In_Frame_Del	CCTGAGCCAGACTCATGTTGCCCAAAGCC AGAAGTCTGG	-	OCCC_27	p.343_356GQTSGFGQHESGSG>G
CELSR3	3	48693701	48693701	+	Frame_Shift_Ins	-	GCGGCGCCTT	OCCC_27	p.R1499fs
MAPK8IP2	22	51042892	51042892	+	Frame_Shift_Ins	-	ACAC	OCCC_27	p.G123fs
MAGEE1	X	75648768	75648803	+	In_Frame_Del	AGCACCTCCGTGCCGCCCACCGCCTCTGA GGTACCG	-	OCCC_27	p.STSVPPTASEVP149del
MSH3	5	79950724	79950724	+	In_Frame_Ins	-	CAGCGC	OCCC_05	p.59_60insQR
AHNAK	11	62300860	62300860	+	Frame_Shift_Ins	-	ACTAT	OCCC_05	p.-343fs
AHNAK	11	62300862	62300862	+	Frame_Shift_Ins	-	AA	OCCC_05	p.G343fs
MAPK8IP2	22	51042919	51042937	+	Frame_Shift_Del	GAGGCGGCCGCGGGGCCCG	-	OCCC_05	p.EAAAGPG132fs
TCHH	1	152082863	152082883	+	In_Frame_Del	GCAGCTGCTCTTCCTCCTGCT	-	OCCC_29	p.QQEEEQL937del
TCHH	1	152083645	152083665	+	In_Frame_Del	TGCTCGCGCCTCTCTTCCTCA	-	OCCC_29	p.HEEERRE676del
HRNR	1	152190899	152190899	+	Frame_Shift_Del	C	-	OCCC_29	p.S1070fs
SPTBN2	11	66463850	66463850	+	Frame_Shift_Ins	-	CAGCTCTGGGCAAACAGCT	OCCC_29	p.C1392fs
WWP2	16	69965462	69965462	+	In_Frame_Ins	-	ATC	OCCC_29	p.525_525F>LS
MINK1	17	4797305	4797337	+	In_Frame_Del	GTCAGCACCATGGTGGTCCACGACGTCGAGGAG	-	OCCC_29	p.VSTMVVHDVEE863del
MINK1	17	4797342	4797368	+	In_Frame_Del	CCGGGACCCAGCCCCCATACGGGGGCG	-	OCCC_29	p.875_884TGTQPPYGGG>S
EVPL	17	74005040	74005080	+	Frame_Shift_Del	CGCCGGCCCGCAGCTGCTGCACCTCAAG CTCTAGCTGGCGC	-	OCCC_29	p.RQLELEVQQLRAGV1403fs
EYA2	20	45717929	45717929	+	In_Frame_Ins	-	CATCTCAAAAAAAAAAAAAA	OCCC_29	p.238_238T>TSQKKKKT
TCHH	1	152080546	152080593	+	In_Frame_Del	AGCTGCTGTTCCTCCTGGAGGAATTTTCTC TCTCGTTCCTGACGGCGG	-	OCCC_64	p.1700_1716LRRQERERKFL QEEQQL>L
TCHH	1	152082863	152082883	+	In_Frame_Del	GCAGCTGCTCTTCCTCCTGCT	-	OCCC_64	p.QQEEEQL937del
TCHH	1	152083757	152083795	+	In_Frame_Del	GCTGCTGGCGCCTCTCCTCCTGCTCCTCG CTCTTCAGCA	-	OCCC_64	p.LLKSEEQEERRQQ633del
FLG2	1	152328434	152328472	+	In_Frame_Del	CATGTTGTCCAAAGCCAGAGGATTGTCCTG AGCCAGACC	-	OCCC_64	p.GSGSGQSSGFGQH597del

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
AHNAK2	14	105408026	105408027	+	Frame_Shift_Del	CC	-	OCCC_64	p.E4588fs
RPTN	1	152127299	152127340	+	In_Frame_Del	CTGTCTCGTCTCTGATGGCTCTGCTCATG TTCATGGGTTTGT	-	OCCC_60	p.745_759RQTHEHEQSHQRRDR>R
RPTN	1	152129095	152129130	+	In_Frame_Del	CTGACCATGGTGGGAATCTCTGTCTT GTCTCTCAGG	-	OCCC_60	p.PERQDRDSHHGQ149del
MSH3	5	79950700	79950717	+	In_Frame_Del	GCAGCGGCTGCAGCGGCC	-	OCCC_60	p.AAAAAA52del
DST	6	56485271	56485287	+	Frame_Shift_Del	TATCTTCTGCAGAGAGA	-	OCCC_60	p.ISLQKI1182fs
AHNAK	11	62298941	62298941	+	Frame_Shift_Ins	-	CTCCAGGTGTGTGTGTGTG	OCCC_60	p.S983fs
KRT5	12	52908926	52908949	+	In_Frame_Del	CAAGACCTCCACCGAGGCCGCCGC	-	OCCC_60	p.GGGLGGGL517del
CASKIN1	16	2239304	2239304	+	In_Frame_Ins	-	CTAATTTTAATTTTT	OCCC_60	p.140_141insKN*N*
KRT10	17	38975103	38975103	+	In_Frame_Ins	-	GCT	OCCC_60	p.562_562S>SS
PPP2R1A	19	52724342	52724342	+	Frame_Shift_Ins	-	ACCCCAACTACCTGCAC CGCATGACT	OCCC_60	p.D492fs
FLG	1	152281412	152281413	+	Frame_Shift_Del	AC	-	OCCC_57	p.ES1983fs
LOR	1	153233699	153233701	+	In_Frame_Del	GGA	-	OCCC_57	p.G95del
SPTA1	1	158632528	158632528	+	Frame_Shift_Ins	-	GATCCAGGCCTCCTCATCC TCTGTGTCTCTACAAATCAG AGGATGAG GAGGCCTGGATCCAAGAGAC TGAACCCTCA	OCCC_57	p.Q810fs
MUC4	3	195507226	195507226	+	Frame_Shift_Ins	-	CCTGTGGATACTGAGGAAG	OCCC_57	p.V3742fs
AHNAK	11	62288844	62288844	+	Frame_Shift_Ins	-	TG	OCCC_57	p.G4349fs
ERBB2	17	37881624	37881624	+	Frame_Shift_Ins	-	AGAA	OCCC_57	p.F899fs
ARID1A	1	27094341	27094341	+	Frame_Shift_Ins	-	A	OCCC_13	p.E1017fs
TCHH	1	152081378	152081378	+	In_Frame_Ins	-	CTGCTGTTCCTCTTCACGGA ATTTTCTGTCACGCTCTTGG CGGCTCAG	OCCC_13	p.1438_1439insLSRQERDRKFREEEQQ
TCHH	1	152081530	152081530	+	In_Frame_Ins	-	GTTCCTGGCGGCGCAGCCGC TGTTCCTCCTCGAGGAATTTT CTCCCTT	OCCC_13	p.1387_1388insQGRKFLEEEQRLRRQE
MUC4	3	195507241	195507241	+	In_Frame_Ins	-	CTGAGGAAGGGCTGGTGACAT GAAGAGGGGTGACGTGACCTGTAGATA	OCCC_13	p.3736_3737insVSTGHVTPLHVTSPSS
MUC19	12	40877361	40877361	+	In_Frame_Ins	-	AACTGGACTATCAGCTGGAGTG ACAGGGACAACTGGACTGTCAACTGA AGTGACAGGGAC	OCCC_13	p.2969_2969I>KLDYQLE*QGQ LDCQLK*QGL
MUC19	12	40878531	40878531	+	In_Frame_Ins	-	TGGACTATCAGCTGGAGTGACAGGGACAAA	OCCC_13	p.3358_3359insMDYQLE*QGQ
XPO6	16	28118877	28118877	+	Frame_Shift_Ins	-	CTTCGGGTGGGGTGGTGTGTGTGTGTGT GTGTGT	OCCC_13	p.-821fs
ARID1A	1	27107135	27107136	+	Frame_Shift_Del	CA	-	OCCC_56	p.S2249fs
HRNR	1	152185812	152185812	+	Frame_Shift_Ins	-	AG	OCCC_56	p.G2765fs
MAPK8IP2	22	51042308	51042308	+	In_Frame_Ins	-	GCGGGGCGCAGTCGCCAGT GCGCCCGGGTTGCGACT	OCCC_56	p.193_194insAGRSRQCARVAT
HRNR	1	152187559	152187559	+	In_Frame_Ins	-	CTAGGA	OCCC_09	p.2182_2182S>SPS
MUC4	3	195515915	195515915	+	Frame_Shift_Ins	-	AT	OCCC_09	p.E846fs
TCHH	1	152083803	152083844	+	In_Frame_Del	GCGCCTCTCTTCCTCCGGCTCCTCGC GCTTCAGCCGCTGCTC	-	OCCC_36	p.EQRLKREEPEEERR617del
TCHH	1	152083909	152083977	+	In_Frame_Del	TCCTGCTCGCGCTTCAGCCGCTGCTG GCGCCTCTCCTCCTCGCG CTTCAGCAGCTGATCGCGCCTCTCC	-	OCCC_36	p.572_595EERRDQLLKREEER RQQRLKREQE>E
RPTN	1	152127241	152127282	+	In_Frame_Del	GTCTTCATGGGTTTGCCTGTCTCGTCT CTGACGGTTCTGCTT	-	OCCC_36	p.KQNRQRRDRQTHED765del
OBSCN	1	228444409	228444453	+	In_Frame_Del	CGGGGGCCAGCACCACACTCAGCTGCGA GGTGGCTCAGGCCCAGA	-	OCCC_36	p.GASTTLSCEVAQAQT1457del
CELSR3	3	48677161	48677206	+	Frame_Shift_Del	GAGGGCCCAAGCACAGAGGCTGTGGCAG AAGGTGTGGCAGTGGTGT	-	OCCC_36	p.HTTATPSATASVLGPS3271fs
SHROOM3	4	77675655	77675655	+	Frame_Shift_Ins	-	AGGGCTGGTCACAGACACC	OCCC_36	p.-1340fs
DSPP	4	88536230	88536238	+	In_Frame_Del	AGCAGTGAT	-	OCCC_36	p.SSD815del
DST	6	56510688	56510688	+	Frame_Shift_Ins	-	TC	OCCC_36	p.K374fs
DST	6	56765382	56765382	+	In_Frame_Ins	-	CTCGTCTTCTAAGATGCC GAGGGCTTGCTCAGGGAT TCAGAGCAAGCC	OCCC_36	p.84_85insGLALNP*ASPRHLRRR
PTPRN2	7	157449106	157449159	+	In_Frame_Del	GGATGAGGCGCTGCTGCGTGCGGAGGG GCTGGGGATCGGCCCGT CGCTGAACTG	-	OCCC_36	p.QFSDGPIPSPSARSSASS696del
KRTAP5-2	11	1619347	1619388	+	In_Frame_Del	CAGCCCCCACAGCCAGAGCCACAGCCCCC ACGGCCGGAGCCA	-	OCCC_36	p.31_45CGSGRGGCGSGCGGC>C
MAGEE1	X	75648505	75648540	+	In_Frame_Del	CTGAGGGCCCAAGCACCTCCGTTCTGCC CACCTCCG	-	OCCC_36	p.EGPSTSVLPTSA62del

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Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
ARID1A	1	27106335	27106335	+	Frame_Shift_Ins	-	TG	OCCC_02	p.C1983fs
RPTN	1	152129036	152129107	+	In_Frame_Del	GAATCTCTGTCTTGTCTCTCAGGCTGACT GTGGTGGGAATC TCTGTCTTGTTTCTCAGAC TGACCATGGTGG	-	OCCC_02	p.156_180SHHGQSEKQDRDS HHSQPERQDRDS>S
KRTAP5-2	11	1619173	1619232	+	In_Frame_Del	CCCCCACAGGAGCCACAGCCCC CCTTGGAGCCCCCACAGG AGCCACAGCCCCCCTTGGAG	-	OCCC_02	p.83_103GSKGGCGSCGGS KGGCGSCGG>G
KRT10	17	38975103	38975103	+	In_Frame_Ins	-	GCTGCCGCCGCCGTAT CCGCCGCCGCCGCT	OCCC_02	p.562_562S>SGGGGYGGGSS
HRNR	1	152188893	152188893	+	Frame_Shift_Ins	-	CC	OCCC_24	p.S1738fs
FLG2	1	152326334	152326334	+	Frame_Shift_Ins	-	GTCA	OCCC_24	p.R1310fs
LOR	1	153233702	153233776	+	In_Frame_Del	GGCGGCGGCTCCTCCGGCGG GGGCTCTGGCTGTTTCTCCA GCGGTGGGGGCGGCTCCGGC TGCTTCTCCTCCGGT	-	OCCC_24	p.GGGSSGGGSGCFSSGGG GSGCFSSG93del
DSPP	4	88535567	88535626	+	In_Frame_Del	AGTGACAGCAGTGATAGCAGT GACAGTGATAGTAGTGATAGC AGCAATAGCAGTGACAGT	-	OCCC_24	p.SDSSDSSDSDSSDSSNSSDS585del
ARID1A	1	27057730	27057792	+	In_Frame_Del	CAGCAGCAGCCACCCTACTCCC AGCAACCACCGTCCCAGACCCCT CATGCCCAACCTTCGTAT	-	OCCC_28	p.QQQPPYSQQPPSQTPHAQPSY480del
OBSCN	1	228560156	228560191	+	In_Frame_Del	AGGAGGCCAGGGCTGAGTCCCAG TCGGAGGAGCAGC	-	OCCC_28	p.EARAESQSEEQQ7227del
DSPP	4	88535624	88535659	+	In_Frame_Del	AGTAGTGACAGCAGTGATAGCAGT GACAGTAGTGAT	-	OCCC_28	p.SSDSSDSSDSSD604del
DSP	6	7580657	7580657	+	Frame_Shift_Ins	-	A	OCCC_28	p.L1412fs
SPTBN4	19	41078059	41078060	+	Frame_Shift_Del	CT	-	OCCC_28	p.A2485fs
OBSCN	1	228528946	228528946	+	In_Frame_Ins	-	CGTAGC	OCCC_65	p.5949_5950insRS
DSPP	4	88536488	88536520	+	In_Frame_Del	AGTGACAGCAGTGATAGCAGCAACA GCAGTGAT	-	OCCC_65	p.SDSSDSSNSSD903del
ARID1A	1	27094400	27094400	+	Frame_Shift_Del	G	-	OCCC_10	p.M1036fs
TCHH	1	152080326	152080397	+	In_Frame_Del	GCGCAGCTGCTGTTCCTCCCTCTC CTGGCGGAGCTGTTCCTCCTCGCGG AATTTTCTGTCGCGCTCCTGGCG	-	OCCC_10	p.RQERDRKFREEEQLRQER EEQQLR1766del
TCHH	1	152081350	152081350	+	Frame_Shift_Ins	-	GCGTC	OCCC_10	p.F1448fs
SPTBN4	19	40998889	40998889	+	Frame_Shift_Ins	-	GACTGAGGACAACAGAGAG ACACGCTCAGCCAAGGATG CAGAGCATCCTT	OCCC_10	p.E172fs
ARID1A	1	27087894	27087897	+	Frame_Shift_Del	GCCA	-	OCCC_58	p.RP727fs
ARID1A	1	27107135	27107135	+	Frame_Shift_Ins	-	A	OCCC_58	p.S2249fs
TCHH	1	152080594	152080641	+	In_Frame_Del	AGCTGCTGTTCCTCTTCGCGGAAT TTTCTGTCACGCTCTTGGCGGCGC	-	OCCC_58	p.1684_1700LRRQERDRK FREEEQQL>L
FLG2	1	152328527	152328529	+	In_Frame_Del	CAG	-	OCCC_58	p.578_579TG>S
OBSCN	1	228560130	228560171	+	In_Frame_Del	GTCCCAGTCAGAGGAGGAGGAGC AGGAGGAGGCCAGGGCTGA	-	OCCC_58	p.SQSEEEEQEEARAE7218del
SHROOM3	4	77676310	77676310	+	Frame_Shift_Ins	-	GGAAGTCATCCATGCTATA CAACGGGGTTTC	OCCC_58	p.-1559fs
DSPP	4	88535624	88535659	+	In_Frame_Del	AGTAGTGACAGCAGTGATAGCAGT GACAGTAGTGAT	-	OCCC_58	p.SSDSSDSSDSSD604del
DSPP	4	88536635	88536652	+	In_Frame_Del	AGCAGTGACAGCAGTGAT	-	OCCC_58	p.SSDSSD941del
AHNAK	11	62291344	62291364	+	In_Frame_Del	TGGGCCCTCAATGTTCATACT	-	OCCC_58	p.SMNIEGP3509del
AHNAK	11	62291447	62291461	+	In_Frame_Del	TTGGGCATTTTCATC	-	OCCC_58	p.3476_3481KMKMPK>K
AHNAK	11	62297524	62297524	+	In_Frame_Ins	-	TCTGGTGTGGATATCTT CTGAGGCTTTATACTCATT	OCCC_58	p.1454_1455insEMSIKPQKISTP
AHNAK	11	62299078	62299078	+	Frame_Shift_Ins	-	GGCATCTTGAACTTGG	OCCC_58	p.-937fs
MUC19	12	40877967	40878026	+	In_Frame_Del	TATCAGCTGGGGTGACAGGGACAA CTGGATCACTAGCTGGAGGGACAG GGACAATTGGAC	-	OCCC_58	p.SAGVTGTTGSLAGGTG TIGL3172del
KRT1	12	53073970	53073970	+	In_Frame_Ins	-	ACC	OCCC_58	p.54_55insG
MINK1	17	4793912	4793932	+	In_Frame_Del	AACAGCAGCAGCAGCTTCAGA	-	OCCC_58	p.QQQQLQK484del
LAMA5	20	60903371	60903371	+	Frame_Shift_Ins	-	TCACAGCCGACCAGGGGGTG GCAGCCAAAGGTCTGGGACT GCCTTCTGTGCCAGCCCCAG ACCTT	OCCC_58	p.-1526fs
MAGEE1	X	75648522	75648522	+	Frame_Shift_Ins	-	TTGTG	OCCC_58	p.S67fs
FLG	1	152279728	152279728	+	In_Frame_Ins	-	AAT	OCCC_54	p.2545_2545G>D*
FLG2	1	152323276	152323276	+	Frame_Shift_Del	T	-	OCCC_54	p.Q2329fs

Table 2 (*continued*)

Hugo_Symbol	chromosome	Start_Position	End_Position	Strand	Variant_Classification	Tumor_Seq_Allele1	Tumor_Seq_Allele2	Tumor_Samle_Barcode	protein_change
OBSCN	1	228509582	228509583	+	Frame_Shift_Del	CT	-	OCCC_54	p.L5014fs
OBSCN	1	228509583	228509583	+	In_Frame_Ins	-	AAG	OCCC_54	p.5014_5014L>QV
OBSCN	1	228524709	228524709	+	Frame_Shift_Ins	-	CCTCTGGCTGCCAAGGAGGC CTCCGAGGGCCT	OCCC_54	p.-5515fs
DSPP	4	88535714	88535719	+	In_Frame_Del	AGTGAT	-	OCCC_54	p.SD636del
AHNAK	11	62291447	62291461	+	In_Frame_Del	TTGGGCATTTTCATC	-	OCCC_54	p.3476_3481KMKMPK>K
MUC19	12	40876299	40876328	+	In_Frame_Del	CAGTATCAGGGACAACTGTACAAT CTCTTA	-	OCCC_54	p.VSGTTVQSLT2616del
MUC19	12	40879293	40879322	+	In_Frame_Del	ACAGGGACAACTGGACTATCAGC TGGAGTC	-	OCCC_54	p.RDNWTISWSH3614del
AHNAK2	14	105411021	105411041	+	In_Frame_Del	CTTGGGGCCTTTCAGGTCCAG	-	OCCC_54	p.LDLKGPK3583del
MINK1	17	4793920	4793934	+	In_Frame_Del	CAGCAGCTTCAGAAA	-	OCCC_54	p.QQLQK486del
LAMA5	20	60892819	60892819	+	Frame_Shift_Ins	-	CCACC	OCCC_54	p.L2419fs
HELZ2	20	62196229	62196229	+	In_Frame_Ins	-	GGTGGCCTGGTCCGACGGGG GCACCCTCAAGCTGAG	OCCC_54	p.1315_1316insLSLRVPPSDQAT
HRNR	1	152191626	152191626	+	In_Frame_Ins	-	AGAGGA	OCCC_12	p.826_827insSS
ARID1A	1	27057983	27057983	+	Frame_Shift_Ins	-	AC	OCCC_40	p.Q564fs
DST	6	56357778	56357778	+	In_Frame_Ins	-	ACGAACACAAGGTATGTA	OCCC_40	p.6515_6515P>LHTLCSS
AHNAK	11	62297907	62297907	+	Frame_Shift_Ins	-	ATCAGGCATGGAGATCTTGGGG GCCTTGAAGTGCAAGATCTCCAT GCCTGATGTGGACCTGA	OCCC_40	p.V1328fs
AHNAK	11	62298807	62298807	+	In_Frame_Ins	-	CACATTCGCTTTGGACAGGTT	OCCC_40	p.1027_1028insNLSKANV
MUC19	12	40877680	40877709	+	In_Frame_Del	GACAGGGACAACTGG AATATTAGCCGGGGT	-	OCCC_40	p.TGTTGILAGV3076del
SPTB	14	65241890	65241890	+	In_Frame_Ins	-	GGCCTCAGCCTCGTCTGC ATCCAGGTAGTACTACGAG GCACAGCAGTACTACCT	OCCC_40	p.1598_1599insR*YCCAS* YYLDADEAEA
AHNAK2	14	105412190	105412193	+	Frame_Shift_Del	GTTG	-	OCCC_40	p.QL3199fs
MINK1	17	4796340	4796340	+	Frame_Shift_Ins	-	CTCACGGCCAGGCCG GCCCGCAGTGAGTCAC CTGGTGA	OCCC_40	p.R793fs
SPTBN4	19	41025967	41025978	+	In_Frame_Del	GGGAGGCGCGCA	-	OCCC_40	p.EARR1189del
OBSCN	1	228560156	228560191	+	In_Frame_Del	AGGAGGCCAGGGC TGAGTCCCAGTCG GAGGAGCAGC	-	OCCC_23	p.EARAESQSEEQQ7227del
DSPP	4	88536089	88536142	+	In_Frame_Del	AGTGACAGCAGCAACA GCAGTGACAGCAGTGAT AGCAGTGACAGCAGTGATAGT	-	OCCC_23	p.SDSSNSSDSSDSSDSSDS759del
DST	6	56480945	56480946	+	Frame_Shift_Del	GG	-	OCCC_23	p.P2440fs
EVPL	17	74005307	74005333	+	In_Frame_Del	CCAGCACCGGGTCCTTCT CGTGGCGCA	-	OCCC_23	p.VRHEKDPVL1318del
EVPL	17	74017566	74017566	+	Frame_Shift_Del	C	-	OCCC_23	p.V332fs
MAPK1	22	22221709	22221714	+	In_Frame_Del	CCGCCG	-	OCCC_23	p.AA6del
MAPK8IP2	22	51044027	51044027	+	Frame_Shift_Ins	-	AACCTGCCGT	OCCC_23	p.V361fs
FLG	1	152280802	152280802	+	Frame_Shift_Ins	-	TGGATCCTGACTGCC CACGGGAGGCATCAG ACCTTCCCTGGGATGA	OCCC_38	p.R2187fs
LOR	1	153233780	153233824	+	In_Frame_Del	GGCGGCTCCTCCGGGGG CGGCTCCGGCTGCTTCTC CAGCGGTGGG	-	OCCC_38	p.GGSSGGGSGCFSSGG134del
MSH3	5	79950709	79950717	+	In_Frame_Del	GCAGCGGCC	-	OCCC_38	p.AAA58del

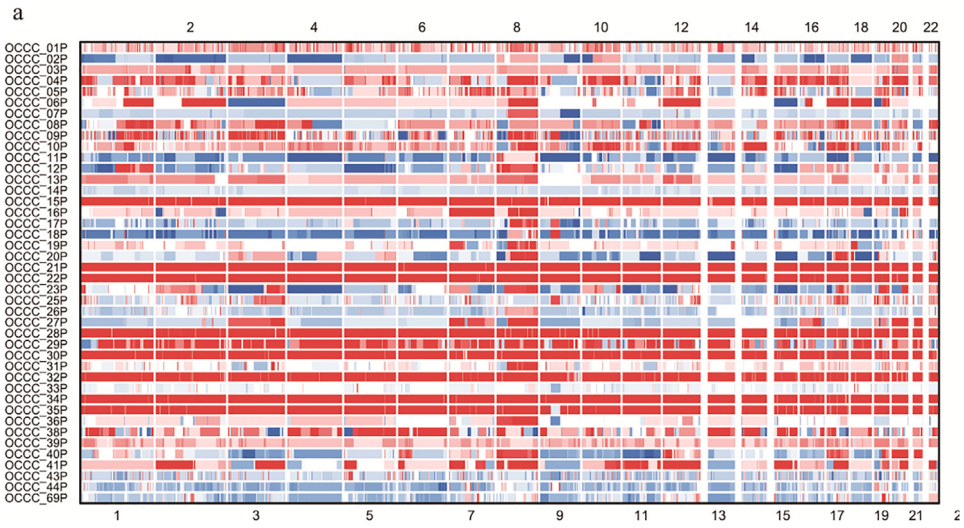

Fig. 2. Somatic copy number aberrations (SCNA) landscape in 42 OCCC samples. Deletions and amplifications are indicated by boxes in different shades of blue and red, respectively. (For interpretation of the references to colour in this figure legend, the reader is referred to the web version of this article.)

amplified (11.9%, 47.6%, and 35.7% of samples, respectively). On the other hand, frequent large blocks of deletions were observed on chr9q in the region including *NOTCH1* and *PAEP* (19% and 16.6%).

At the chromosome arm level, we examined focal somatic copy number alterations by performing GISTIC 2.0 analysis and found that the amplified loci in OCCC samples were at 19q, 2q, 8q, 17q, 1p, 1q, 5q, 12q, and 20q, and the deletions common to the data set were at 19p, 11p, 16p, 9q, 21q, 1p, 7q, 17q, 11q, 16q, 19q, 4p and 2q (cutoff q-value <0.01) (Fig. S4). All focal genes identified by GISTIC 2.0 are shown in Table S9 (amplification cut-off = 0.1, deletion cutoff = -0.1, cutoff *q* value <0.05).

Pathways in ovarian clear cell carcinoma

To discover the important altered pathways in OCCC, we performed an integrative analysis of single nucleotide variant (SNV) data and copy number variant (CNV) data and selected recurrently mutated genes in two or more individuals that mapped to canonical cancer pathways using MSigDB and frequent alterations across multiple pathways, including p53 effectors, NGF signaling, focal adhesion, and PTEN-dependent cell cycle arrest and apoptosis (Table 3 and Fig. 5S). KEGG pathway enrichment analysis further identified significant overlaps (*q* value <0.001) with

Table 3. MSigDB canonical pathway database enrichment for recurrent mutant genes in OCCC samples. This table presents the top 20 modules. Module: name of the module.

Gene Set Name	Genes in Gene Set (K)	Description	Genes in Overlap (k)	k/K	*p*-value	FDR *q*-value
KEGG_PATHWAYS_IN_CANCER	328	Pathways in cancer	25	0.0762	5.43E−18	7.22E−15
PID_P53_DOWNSTREAM_PATHWAY	137	Direct p53 effectors	13	0.0949	3.58E−11	2.18E−08
KEGG_TYPE_II_DIABETES_MELLITUS	47	Type II diabetes mellitus	9	0.1915	6.19E−11	2.18E−08
KEGG_PROSTATE_CANCER	89	Prostate cancer	11	0.1236	6.55E−11	2.18E−08
REACTOME_DEVELOPMENTAL_BIOLOGY	396	Genes involved in Developmental Biology	19	0.048	1.82E−10	4.84E−08
REACTOME_SIGNALLING_BY_NGF	217	Genes involved in Signalling by NGF	14	0.0645	1.08E−09	2.39E−07
KEGG_THYROID_CANCER	29	Thyroid cancer	7	0.2414	1.52E−09	2.84E−07
SIG_PIP3_SIGNALING_IN_CARDIAC_MYOCTES	67	Genes related to PIP3 signaling in cardiac myocytes	9	0.1343	1.71E−09	2.84E−07
KEGG_FOCAL_ADHESION	201	Focal adhesion	13	0.0647	4.15E−09	6.13E−07
REACTOME_IMMUNE_SYSTEM	933	Genes involved in Immune System	26	0.0279	9.84E−09	1.31E−06
KEGG_COLORECTAL_CANCER	62	Colorectal cancer	8	0.129	1.93E−08	2.33E−06
REACTOME_ADAPTIVE_IMMUNE_SYSTEM	539	Genes involved in Adaptive Immune System	19	0.0353	2.77E−08	3.07E−06
KEGG_NOTCH_SIGNALING_PATHWAY	47	Notch signaling pathway	7	0.1489	5.45E−08	5.58E−06
PID_HES_HEY_PATHWAY	48	Notch−mediated HES/HEY network	7	0.1458	6.34E−08	6.02E−06
SIG_INSULIN_RECEPTOR_PATHWAY_IN_CARDIAC_MYOCYTES	51	Genes related to the insulin receptor pathway	7	0.1373	9.79E−08	8.67E−06
KEGG_ENDOMETRIAL_CANCER	52	Endometrial cancer	7	0.1346	1.12E−07	9.34E−06
REACTOME_SIGNALING_BY_FGFR	112	Genes involved in Signaling by FGFR	9	0.0804	1.64E−07	1.12E−05
PID_FGF_PATHWAY	55	FGF signaling pathway	7	0.1273	1.67E−07	1.12E−05
BIOCARTA_ERK5_PATHWAY	18	Role of Erk5 in Neuronal Survival	5	0.2778	1.68E−07	1.12E−05
BIOCARTA_PTEN_PATHWAY	18	PTEN dependent cell cycle arrest and apoptosis	5	0.2778	1.68E−07	1.12E−05

KEGG cancer, platinum drug resistance and the AMPK signaling pathway (Table S10).

Given that the four functional protein activating pathways, including the (PI3K)/AKT/mammalian target of rapamycin (mTOR) pathway, TP53 pathway, ERBB2 pathway, and chromatin remolding pathways were more frequently activated in OCCCs, these pathways represent potential therapeutic pathways for targeted treatment approaches (Fig. 3). Of the 42 samples, 35 (83%) contained at least one mutation in one of the four pathways. The PI3K/AKT, TP53, ERBB2, and chromatin remolding pathways were mutated in 83%, 67%, 40% and 71% of cases, respectively (Fig. S6).

Clinical relevance of aberrant genes mutations

For the WES cohort of 42 OCCC cases, the median follow-up time was 27.6 months, and 78% of patients were alive at the time of last follow-up. Kaplan-Meier analysis with the log-rank test revealed a shorter survival period for the 42 OCCC patients with *PLEC* mutations (HR 0.27, 95% confidence interval 0.76 to 0.04, $P = 0.004$) or *CDC27* mutations (HR 3.6, 95% confidence interval 1.2 to 10.7, $P = 0.035$) than for those without the corresponding mutation (Fig. S7). In addition, based on the targeted sequencing data, the OS was not significantly different between the *PLEC* or *CDC27* mutation-positive patients and the wild-type patients ($P = 0.227$ or $P = 0.954$) (Fig. S7). However, the OS analysis of OCCC patients grouped by *MAGEE1* mutation revealed that OCCC patients with *MAGEE1* mutations had a shorter survival time than those without MAGEE1 mutations (log-rank $p < 0.05$) (Fig. 4). The OBSCN mutation was found to have a significant effect on overall survival in all OCCC patients (Fig. 4). The human OBSCN gene on chromosome 1q42.13 region is comprised of over 80 exons and encodes a ~720 kDa protein. The OBSCN gene is frequently and consistently mutated in various cancers with a strong correlation with breast, colorectal and other

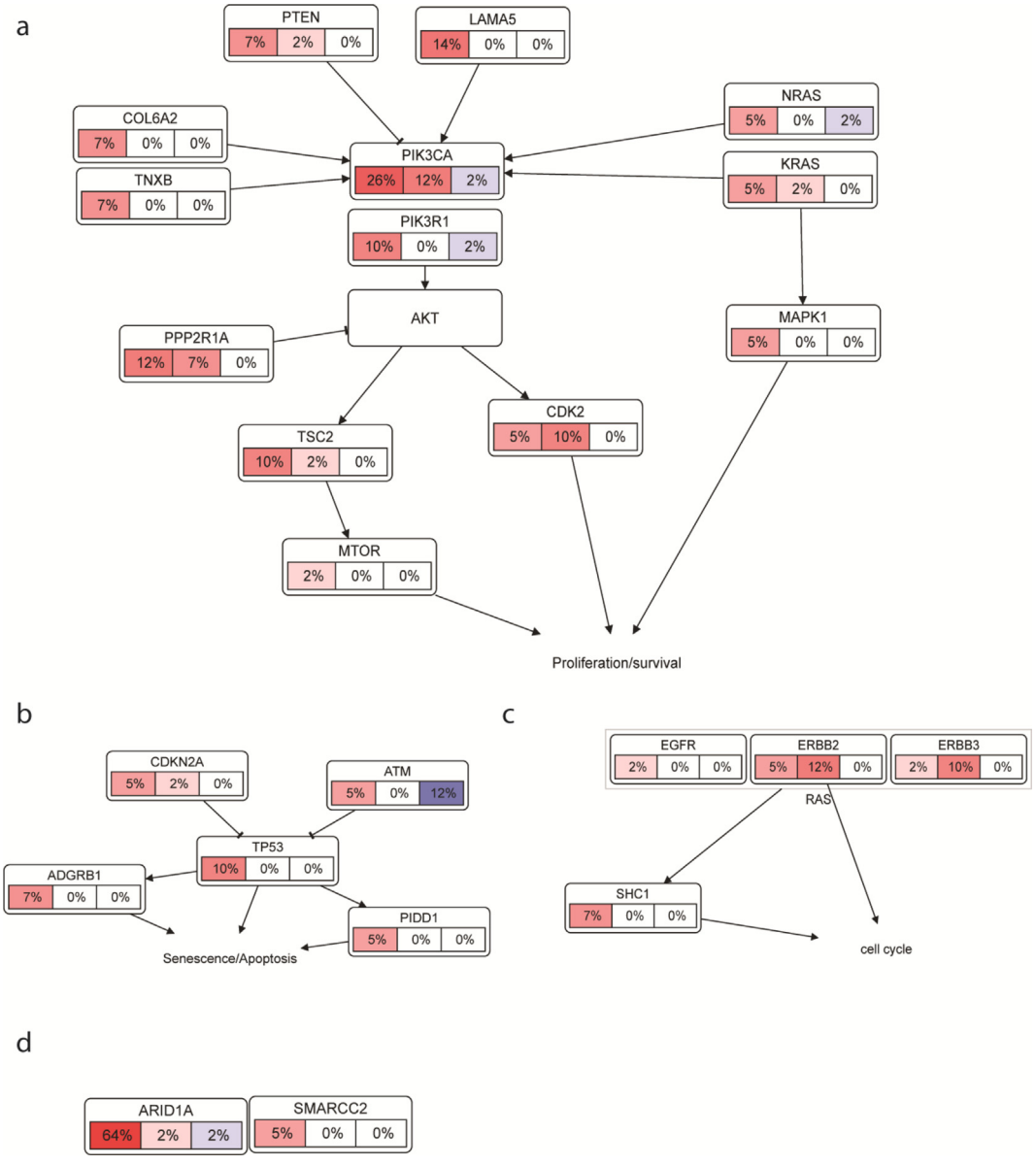

Fig. 3. Somatically altered pathways in OCCC patients. (a) Somatically altered genes in the PI3K/AKT, (b) TP53, (c) ERBB2 and (d) chromatin remolding pathways. Non-synonymous somatic mutations and copy number deletions were considered as inactivating mutations (shades of blue), while copy number amplifications were considered as activating mutations (shades of red). (For interpretation of the references to colour in this figure legend, the reader is referred to the web version of this article.)

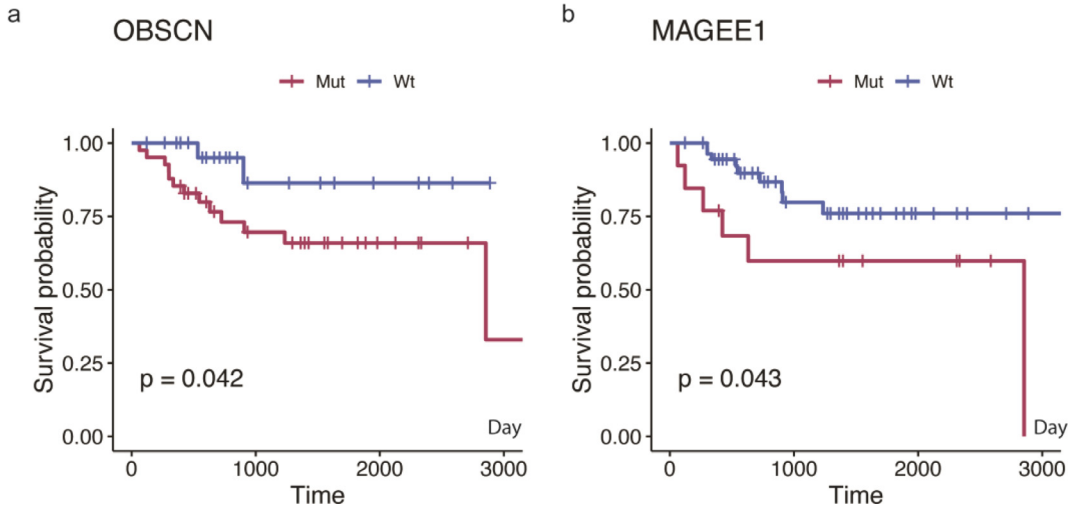

Fig. 4. Kaplan-Meier estimate of overall survival by OBSCN mutational status (a) and MAGEE1 mutational status (b). Patients with tumors harboring mutations had significantly worse overall survival than those with corresponding WT genes in their tumors (log-rank test, OBSCN $P = 0.042$; MAGEE1 $P = 0.043$).

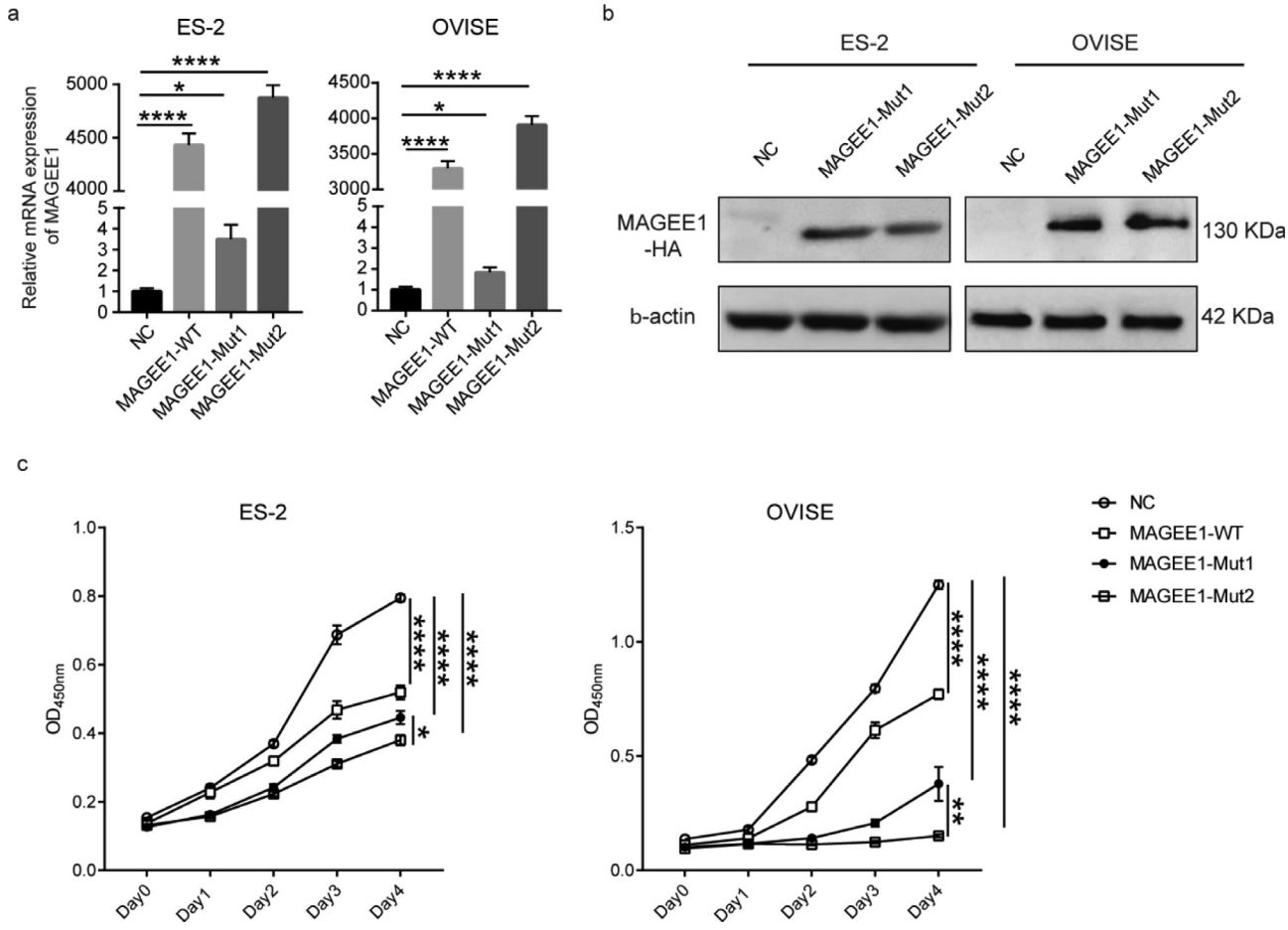

Fig. 5. MAGEE1 overexpression inhibits cell viability in vitro. (a) The expression of MAGEE1 in the ES-2 and OVISE cell lines transfected with empty vector control (NC), MAGEE1-WT, MAGEE1-Mut1 or Magee1-Mut2 was determined by RT-PCR. (b) Western blotting of MAGEE1 expression levels in ES-2 and OVISE cells transfected with empty vector control (NC), MAGEE1-WT, MAGEE1-Mut1 or Magee1-Mut2. (c) CCK8 assay showed a significant reduction in the viability of cells transfected with MAGEE1-WT, MAGEE1-Mut1 or Magee1-Mut2 compared to cells transdfected with empty vector control (NC).

female related cancers [15]. For known OCCC driver genes, such as *ARID1A*, *PIK3CA* and *PPP2R1A*, no survival difference was observed based on the presence of genetic abnormalities. These data suggest that mutations in these genes may represent a poor prognostic factor and are likely involved in the pathogenesis of OCCC.

MAGEE1 mutants affects cell growth in OCCC cells

To further elucidate the relevance of MAGEE1 to OCCC progression, we first analyzed MAGEE1 levels in OCCC cell lines (Fig. 5a). Then, we performed a functional assay and found that MAGEE1 overexpression significantly decreased the viability of OCCC cells (Fig. 5c). MAGEE1 contains a nuclear localization signal in the N-terminal region and two MAGE domains in the C-terminal region. To determine whether the two domains of the MAGEE1 gene are associated with proliferation, OVISIE and ES-2 cells expressing the MAGEE1 mutants (MAGEE1-mut1 and MAGEE1-mut2) were constructed and analyzed (Fig. 5b). CCK8 assays revealed that both OVISIE and ES-2 cells transfected with MAGEE1-mut1 or MAGEE1-mut2 exhibited a significant decrease in cell proliferation (Fig. 5c), suggesting that both domains of the MAGEE1 gene are closely associated with OCCC cell proliferation. However, the viability of OCCC cells transfected with the MAGEE1-mut2 was lower than that of cells transfected with the MAGEE1-mut1, revealing that the second domain of MAGEE1 has a greater contribution to OCCC cell proliferation.

Discussion

OCCC is one of the most malignant subtypes of epithelial ovarian cancer and is more prevalent in Asians (11.1% of total EOC cases) than in Caucasians or Africans in the USA (4.8 and 3.1%, respectively) [16,17]. OCCC is a rare form or subtype of epithelial ovarian cancer that has a unique structure (morphology), unfavorable prognosis, and chemotherapeutic resistance. Most notably, ARID1A and PIK3CA were the most frequently mutated genes in OCCC patients. Yasuda et al. indicated that ARID1A and PIK3CA was the most frequently mutated gene, occurring in approximately 66.7% and 50% of patients with OCCC ($n = 48$) in Japanese women using whole exome sequencing [12]. In addition, Seo et al. revealed that PIK3CA mutations and ARID1A were found in 40% and 40% in the 15 Korean OCCCs using whole exome sequencing [11]. In another study, ARID1A mutations and PIK3CA mutations were detected in 77.8% and 66.7% of OCCC ($n = 16$) in the Taiwanese population using targeted sequencing [18]. In the present study, we successfully characterized the genomic landscape of 69 Chinese patients with OCCC. To our knowledge, this is the first report of an NGS WES study in Chinese patients with OCCC.

The most frequent mutated gene identified in this study was the ARID1A gene (66.7%, 32/48), which encodes a key component of the SWI/SNF chromatin-remodeling complex that is conserved in all eukaryotes, plays an important role in controlling gene expression and is critical in development, differentiation, and tumor suppression. Although the frequency of somatic ARID1A mutations in OCCC was recently published to be 46% and 57% based on whole-exome and transcriptome sequencing analyses, respectively, the frequency reported in this study was 66.7%. The frequency of *ARID1A* mutations in this study was slightly higher than that in a previous report, but the difference was not statistically significant. Mutation of the SWI/SNF-related gene *SMARCA2* was identified in six cases; hence other genes in the ARID1 pathway may be mutated in the remaining cases. Other known OCCC-related genes were also frequently mutated, including *PIK3CA* (50%, 24/48), *PPP2R1A* (18.8%, 9/48) and *KRAS* (16.7%, 8/48) (Fig. 3).

MAGEE1 was also predicted to be a cancer driver gene by three independent mutation prediction algorithms. *MAGEE1* is a member of the melanoma antigen gene (MAGE) family and is encoded on the X chromosome, spanning one exon, containing 957 amino acids. The MAGE family has garnered growing interest as cancer biomarkers and immunotherapy targets because a subset of these human proteins has been classified as cancer-testis antigens (CTAs), which have restricted expression in the testis (and occasionally in the ovary and placenta) and are aberrantly re-expressed in cancer and can be immunogenic. Collectively, MAGE genes have been found to be broadly expressed in many tumor types, including colon, melanoma, brain, lung, prostate, and breast cancer, among others. Furthermore, *MAGEE1* is mutated frequently enough to be classified as a candidate cancer gene (CAN-gene) in breast cancer and thus potentially a driver of tumorigenesis. In this study, mutations in *MAGEE1* were found in eight patients in the WES cohort and fifth patients in the targeted sequencing cohort.

The other genes, including *MUC4, ARID3A, FLG2, TCHH, GRM3, MUC17, ZNF208* and *GAGE12J*, were identified as novel SMGs in our study. *MUC4* and *MUC17* are related to cell apoptosis/anti-adhesive [19] and cell restitution processes [20], respectively, and we identified somatic mutations in these genes in 28.6% and 23.8% of OCCC samples, respectively.

ARID3A, FLG2, TCHH, GRM3, ZNF208, and *GAGE12J* were mutated in 4.8% to 28.6% of samples, respectively. ARID3A is a member of the human AT-rich interaction domain (ARID) family, is located at 19p13.3, and is a nuclear matrix-associated transcription factor that blocks cell differentiation and promotes cell proliferation [21]. The *FLG2* gene encodes a histidine- and glutamine-rich protein of approximately 248 kDa belonging to the fibrinogen-related protein superfamily that has apoptotic effects on effector T-cells and prevents the maturation of dendritic cells [22]. *TCHH* (trichohyalin) is a member of the S100-fused type proteins (SFTP) family and probably contributes to tumorigenic processes such as cell proliferation, metastasis, angiogenesis and immune evasion [23]. *GRM3* is a group II metabotropic glutamate receptors that activates the AKT signaling pathway [24]. Additionally, mutations in *GRM3* that lead to constitutive receptor activation have been shown to provide cell proliferation and survival signals in melanoma [25]. *ZNF208* is a member of the zinc finger family of proteins that bind to DNA through a series of zinc finger motifs and regulate gene transcription [26]. Mutations in *ZNF208* have been observed in gastric cancer [27]. The *GAGE12J* gene is located on the X chromosome and encodes a cancer-testis antigens that promotes gastric cancer growth and metastasis by modulating the expression of gastric cancer metastasis-related genes [28].

This current study has several limitations. First, the study population of this retrospective case-control study was small ($n = 69$). Second, the median follow-up time was 58 months from the SEER database for patients with white and Asian. And early-stage OCCC confined to ovary has favorable prognosis. Regarding the stage of disease ($n = 69$) in our cohort, it was stage I in 45(65.2%), stage II in 12 (17.4%), stage III in 12(17.4%). The median followup of 27.6 months in our cohort is relatively short and 78% of patients were alive at last followup. Therefore, this is a limit regarding the impact of reported mutations on survival outcomes. Third, the expression of the genes with somatic mutations was not investigated. Paired tumor/normal whole transcriptome sequencing or microarray analyses to discover the effects of genetic alterations would improve the accuracy and completeness of the genomic profiling results. Despite these limitations, this study is the first to use whole exome sequencing to genetically characterize OCCC in an Asian population. By analyzing cancer tissue samples and matched normal samples from individual OCCC patients, integrative somatic analyses were completed.

Conclusions

In conclusion, the present study successfully characterized the genomic landscape of 69 patients with OCCC. We identified potential therapeutic targets for the treatment of OCCC. Additional larger studies including whole transcriptome sequencing to determine the effects of genetic alterations are warranted.

Conflict of interest

No potential conflicts of interest were disclosed.

Acknowledgements

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Appendix A. Supplementary data

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