Independent somatic evolution underlies clustered neuroendocrine tumors in the human small intestine

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Abstract

Background

Small intestine neuroendocrine tumor (SI-NET), the most common cancer of the small bowel, often displays a curious multifocal phenotype with several intestinal tumors centered around a regional lymph node metastasis. Although SI-NET patients often present with metastatic disease at the time of diagnosis, there is an unusual absence of somatic driver mutations explaining tumor initiation and metastatic spread. The evolutionary trajectories that underlie multifocal SI-NET lesions could provide insight into the underlying tumor biology, but this question remains unresolved. Here, we determined the complete genome sequences of 65 tumor and tissue samples from 11 patients with multifocal SI-NET, allowing for elucidation of phylogenetic relationships between tumors within individual patients. Intra-individual comparisons of whole genome sequences revealed a lack of shared somatic single-nucleotide variants and copy number alterations among the sampled intestinal lesions, supporting that they were of independent clonal origin. Furthermore, each metastasis originated from a single intestinal tumor, and in three of the patients, two independent tumors had metastasized. We conclude that primary multifocal SI-NETs generally arise from clonally independent cells, suggesting a contribution from of a cancer-priming local factor.

Introduction

Cancer is considered to be an evolutionary process involving cycles of random oncogenic mutations, selection and clonal expansion (Yates and Campbell, 2012). In most malignant epithelial tumors (carcinomas), a single primary tumor will typically form, followed by metastases arising as cells disperse from the tumor. More rarely, carcinomas instead display *multifocality*, i.e. multiple separate tumors at the primary site, which may be due to predisposing germline variants, local mutagenic exposure, or localized spread or expansion of cancer-primed mutated clones (Curtius et al., 2018; Graham et al., 2011).

Of particular interest are small intestine neuroendocrine tumors (SI-NETs), the most common cancer of the small intestine, where ~50% of cases display a striking multifocal phenotype that often involves 10 or more morphologically identical tumors clustered within a limited intestinal segment, commonly centered around a regional lymph node metastasis (Dasari et al., 2017; Gangi et al., 2018). SI-NET has a reported incidence of approx. 1.2 per 100,000 (Dasari et al., 2017) and often presents with distant metastases at diagnosis, thus precluding curative treatment (Choi et al., 2017; Gangi et al., 2018). However, the somatic mutational burden of SI-NET is low and there is an unusual absence of somatic driver mutations (Crona et al., 2015; Francis et al., 2013; Priestley et al., 2019). Consequently, underlying tumorigenic mechanisms are poorly understood and actionable genetic drug targets are lacking.

The evolutionary relationships that govern multiple intestinal tumors and metastases in SI-NET can give insight into the underlying biology, but earlier efforts to determine this have yielded conflicting results (Guo et al., 2000; Katona et al., 2006). A study of copy number alterations (CNA) in multifocal SI-NET primary tumors found that loss of chromosome 18 (chr18), the most frequent CNA, can affect different chromosome homologs in different samples within a patient (Zhang et al., 2020), compatible with an independent clonal origin or late loss of chr18. Alternatively, multifocal SI-NETs have been proposed to represent “drop metastases” originating from regional lymph nodes, consistent

with the limited spatial distribution of the intestinal tumors (Wang et al., 2014). Adding to the challenge is the low mutational burden, which reduces the number of exonic genetic markers.

Here, we performed whole genome sequencing (WGS) on 61 separate intestinal tumors and adjacent metastases from 11 SI-NET patients, which made it possible to conclusively determine the evolutionary trajectory of multifocal SI-NET within single individuals.

Materials and Methods

Patients

11 patients who underwent surgery for SI-NET at Sahlgrenska University Hospital, Gothenburg, Sweden, were included in the study. The clinical characteristics of the patients are summarized in **Supplementary Table S1**. Patients were postoperatively diagnosed with well-differentiated neuroendocrine tumor grade 1-2 of the ileum (WHO 2019). Tumor grade and stage was based on one primary intestinal tumor and extent of lymph node metastases as standard clinical routine. Tumor samples were collected at surgery and were immediately snap-frozen in liquid nitrogen. A piece of each tumor was formalin-fixed and paraffin-embedded for studies by immunohistochemistry. Blood was collected and used as normal tissue in sequencing. We obtained consent from the patients and approval from the Regional Ethical Review Board in Gothenburg (Dnr:558-07, 833-18).

Immunohistochemistry

Sections of all sampled tumors from 11 patients with SI-NET were subjected to antigen retrieval as detailed in **Supplementary Material and Methods**. Histopathological evaluation and assessment of tumor cell content was performed on hematoxylin and eosin-stained sections. Immunohistochemical staining was performed for chromogranin A, synaptophysin, serotonin and somatostatin receptor 2. These antibodies were used: anti-chromogranin A (MAB319; Chemicon; diluted 1:1000), anti-synaptophysin (SY38, M0776; Dako; diluted 1:100), anti-serotonin (H209; Dako; diluted 1:10) and anti-SSTR2 (UMB-1; Abcam; diluted 1:50). All collected tumor samples were reviewed by a board certified surgical pathologist (ON).

DNA extraction

All tumor samples exhibited typical SI-NET morphology and expression of neuroendocrine markers. For DNA extraction, tumor samples of sufficient size and with a purity above 30% were included. DNA from fresh-frozen biopsies as well as blood from each patient was isolated using the allprep DNA/RNA Mini Kit (Qiagen) according to the manufacturer's protocol.

Whole genome sequencing

WGS libraries were constructed for 61 primary and metastatic tumors from the 11 patients. 11 blood normals and 4 adjacent tissue normals were additionally sequenced, for a total of 76 samples. The prepared libraries were sequenced on an Illumina Novaseq 6000 using 150 bp paired-end reads to an average depth of $36.6\times$ (29.8-45.0).

Mapping and somatic variant calling

Sequencing reads were mapped to the human reference genome (hg19) using BWA and somatic variant calling of matched tumor-normal specimens was performed using combined outputs from Mutect2 (GATK v4.1.4.0) and VarScan (v2.3.9). Additional details are provided in **Supplementary Material and Methods**.

Phylogenetic analyses

Phylogenetic analyses were based on SNVs only, as indel calls contained a higher fraction problematic calls. Maximum parsimony phylogenies were reconstructed and visualized using MEGAX with default settings (Stecher et al., 2020). Bootstrapping was performed using 500 replications.

Mutational signatures

The trinucleotide profile for each tumor was matched against known mutational signatures from COSMIC (Alexandrov et al., 2020) (v3 release) using the R package deconstructSigs (Rosenthal et al., 2016) (version 1.9.0). We used default parameters and a maximum of 4 signatures for each sample.

Copy number analysis and homolog phasing

Copy-number alterations (CNAs) were called using XCAVATOR (Magi et al., 2017) using the RC mode and a window size of 2000 bp. The segmented copy number results were filtered to remove small segments (<25 kb) prior to visualization using IGV. Whole-chromosome alterations were phased to determine what chromosome homolog was deleted based on SNP calls reported by VarScan somatic (“germline” and “LOH” sets with a required coverage of 20). Only heterozygous SNPs were considered (variant allele frequency ranging from 0.25 to 0.75 in the blood normal). Variant allele frequencies for individual SNPs were compared between a reference sample, typically the metastatic primary tumor in each patient, and other samples of interest by means of scatter plots.

Structural variant analyses

Structural variants (SVs) were called based on discordant read pairs and split reads using Manta (v.1.5.0) (Chen et al., 2016). Genomic coordinates of high confidence SV calls were annotated using AnnotSV (v.2.1) (Geoffroy et al., 2018). For each patient, all identified translocations were visualised in a circos plot (Krzywinski et al., 2009). A custom script was used to evaluate whether gene-gene fusions were in-frame or out-of-frame. To calculate the frames, exon and coding sequence coordinates were taken from the annotation file provided by AnnotSV.

Results

Evolutionary trajectories in multifocal SI-NET

We initially performed whole genome sequencing (WGS) on 6 intestinal tumors, 3 adjacent lymph node metastases, 2 peritoneal metastases and a blood normal from a patient with suspected SI-NET (Patient 1) that underwent surgery with curative intent (**Fig. 1a**). All lesions showed typical SI-NET morphology and stained positively for established diagnostic SI-NET markers (**Supplementary Fig. 1**). Samples were sequenced at an average coverage of 35.7-43.1× (**Supplementary Table S2**). This was followed by somatic mutation calling using strict filters including rigorous population variant filtering to avoid false positives in phylogenetic analyses. Resulting genome-wide single nucleotide variant (SNV) burdens varied from 353 to 1,749 (0.13-0.62 per Mb; **Supplementary Table S2**).

Figure 1. Whole genome sequencing of 11 primary tumors and metastases from a single SI-NET patient supports independent clonal evolution. (a) Section of the small intestine from a 60 year-old female patient harboring 6 primary tumors (A-F), 3 lymph node metastases (G-I) and 2 peritoneal metastases (J-K). (b) Pairwise analysis of shared somatic SNVs based on whole genome sequencing at 34.5-43.1 $\times$ coverage. Primary tumor C and the 5 metastases shared a common set of 667 mutations. A maximum parsimony phylogenetic tree is shown, with the number of SNVs in each branch given by the scale marker. An indel in *CDKN1B*, a known driver events, is indicated. Bootstrap support was $\geq 98\%$ for all major branches. (c) Proposed model, where all metastases originate from a single primary tumor, and where all primaries are unrelated in terms of somatic evolution.

We next determined pairwise shared somatic SNVs between samples. Surprisingly, a common set of 667 SNVs was shared between a single primary tumor (denoted C) and all 5 metastases (G-K), while overlapping mutations were essentially lacking between the other samples (**Fig. 1b**). The results were thus not compatible with a common clonal origin for the intestinal tumors, expected to result in hundreds to thousands of shared mutations genome-wide in the case of a late-onset cancer. Instead, phylogenetic analysis supported that the intestinal tumors had developed independently, with a single, centrally located, tumor metastasizing first to the lymph nodes and then further to the peritoneum (**Fig. 1b-c**).

To validate the initial findings, we analyzed 50 additional tumors from 10 SI-NET patients, plus matching blood normals, using WGS at 29.8-45.0 $\times$ coverage (Patients 2-11; **Fig. 2; Supplementary Table S2**). The bulk of the material (Patients 3-11) consisted previously sampled tumors stored at a local biobank. Between 3 and 11 intestinal tumors and at least one lymph node metastasis was sequenced for each patient, and three cases included a liver metastasis. Macroscopically normal small intestinal mucosa samples were additionally included in four cases. Samples were selected to have

sufficient tumor material and purity, and all tumor samples stained positively for SI-NET markers (Supplementary Figs. 2-5). One tumor had lower-than-expected mutational burden (143 SNVs), explained by low sample purity, while others varied between 411 and 3390 SNVs (0.15-1.21 per Mb; Supplementary Table S2). In comparison, between 11 and 40 SNVs were called in the normal mucosa samples, where widespread clonal somatic mutations are not expected, supporting that somatic mutation calls had high specificity.

Figure 2. Whole genome sequencing of 50 primary tumors and metastases from 10 additional SI-NET patients confirms independent clonal origin. (a) Section of the small intestine from a 78 year-old male patient harboring 11 primary tumors (A-K) and one lymph node metastasis (L). White labels indicate all identified tumors while letters indicate sequenced samples with sufficient purity and tumor material. (b) Pairwise analysis of shared somatic SNVs. Primary tumor G and the metastasis shared 1219 mutations. The known *CDKN1B* driver event is indicated in the phylogenetic tree (the number of SNVs in each branch is given by the scale marker). (c) Proposed model. (d-l) Similar to panel b, based on archival material from 9 additional patients, ages 48-81, that also included liver metastases and normal mucosa samples. Bootstrap support was 100% for all major branches in the phylogenetic trees. Whole genome sequencing was performed at 29.8-45.0x coverage.

Mirroring the result from Patient 1, shared somatic mutations were essentially absent in between intestinal tumors in all 10 additional patients (**Fig. 2**). In contrast, strong SNV overlaps were seen between individual metastases and specific intestinal tumors, ranging from 82 to 2053 and above 290 SNVs in all pairs but one. For example, in Patient 2, a single intestinal tumor (G) out of 11 that were sampled showed a striking relatedness to an adjacent lymph node metastasis (L; 1,219 shared SNVs; **Fig. 2b**). Similar to Patient 1, the metastatic primary was centrally located in the resected section (**Fig. 2a,c**). Detailed positional data was lacking for the remaining patient samples, as these were derived from archival material.

While individual metastases showed credible overlaps only with a single primary tumor, in three cases (Patients 6, 7 and 10) we found that two independent primary tumors had metastasized to different lymph node or liver metastases (**Fig. 2g,h,k**). Given the lack of a common evolutionary trajectory for the primary tumors, this supports that acquisition of metastatic properties is not a rare event in multifocal SI-NET. In Patient 8, the sample phylogeny suggested two independent metastatic events from a single primary (A), with an earlier event giving rise to a liver metastasis (D) and a later event resulting in a lymph node metastasis (C; **Fig. 2i**). Shared SNVs typically had higher variant allele frequencies (VAFs) than other variants, in both primaries and metastases, consistent with continued somatic evolution and subclonal expansions following the metastatic events (**Supplementary Fig. 6**). In a single case (Patient 9), a lymph node metastasis could not be associated to any of two available intestinal tumors, likely explained by incomplete sampling of intestinal lesions (**Fig. 2j**).

False shared somatic SNVs can arise due to the fact that all samples from a given patient use the same blood normal sequencing data. However, only a small number of additional common SNVs were observed beyond the main genetic relationships (**Fig. 1b** and **Fig. 2b,d-l**), some of which could be dismissed as false positives by manual inspection (**Supplementary Table S3**). In Patient 5, one additional tumor (D) shared 8 credible SNVs with the metastasis (F) (**Fig. 2f**). This overlap grew to 66 when high-confidence variants were whitelisted for relaxed calling in other samples, allowing more sensitive detection of subclonal shared SNVs (**Supplementary Fig. 7**). These variants were present as low-VAF traces in the metastasis while having normal VAFs in the tumor, compatible with

contamination during sample handling or, possibly, metastatic spread to the same lymph node from two tumors with a minor contribution from D (**Supplementary Fig. 8**). Low-VAF variants (13 in total) shared between an intestinal tumor (A) and a lymph node metastasis (C) were also uncovered in Patient 9 (**Supplementary Fig. 9**). In Patient 2, 8 SNVs were shared between the main metastatic primary (G), an adjacent primary (F, 5 mm apart, **Supplementary Fig. 10**) and the metastasis (L) (**Fig. 2b**). All had normal VAF distributions in all 3 samples arguing against contamination (**Supplementary Fig. 11**) and all but one passed manual inspection (**Supplementary Table S3**). Overlaps of this size are too small to represent common clonal origin in a late-onset cancer, and are likely explained by lineage-specific somatic mutations established early during organismal development (Lodato et al., 2015).

Driver mutations and mutational signatures

The median age of the patients was 76 years (one subject, Patient 8, was 48 while others ranged from 68 to 81). The observed burdens and number of pairwise overlapping variants are thus roughly on par with a mutation rate similar to healthy human neurons, estimated to accumulate ~23-40 SNVs/year (Lodato et al., 2018) (**Fig. 3**). Analysis of mutational signatures indeed supported major contributions from COSMIC Signature SBS5, an ubiquitous aging-associated mutational process, or SBS40 which is closely related to SBS5 (Alexandrov et al., 2020) (**Fig. 3**). The observed variability in signature loadings may in part be methodological, since the trinucleotide substitution profiles of the samples were in practice highly similar across the cohort (**Supplementary Fig. 12**). Within-patient burden variability was to a large degree explained by variable sample purity (**Supplementary Fig. 13**). These results confirm the mutationally quiet nature of SI-NET.

Figure 3. Overview of potential driver mutations and mutational signatures for all 11 patients. All CGC genes with non-synonymous mutations in more than one sample are shown. Mutational signature loadings, i.e. relative contributions, refer to COSMIC v3 definitions (Alexandrov et al., 2020) and were dominated by the closely related signatures SBS5 and SBS40 (see **Supplementary Fig. 16** for a full signature legend). Phylogenetic relationships previously inferred from somatic SNVs (**Fig. 1-2**) are indicated at the bottom. Somatic SNV burdens, potential driver mutations (Cancer Gene Census genes mutated in >1 sample) and mutational signature loadings in this figure were determined using less rigorous population variant filtering compared to the phylogenetic analyses (see **Materials and Methods**). CGC, Cancer Gene Census.

Analysis of potential somatic driver mutations in coding genes mirrored earlier published results, with infrequent variants in *CDKN1B* emerging as the main recurrent event (8 samples, 6 independent events; **Fig. 3; Supplementary Table S4**) (Francis et al., 2013). In Patient 1, the same *CDKN1B* frameshift indel occurred in two of the metastases (G, H) in a pattern consistent with the inferred phylogeny (**Fig. 1b-c**). In Patient 2, the same *CDKN1B* frameshift indel was found in the metastatic primary (G) and the metastasis (L), thus again consistent with the phylogeny, while one non-metastatic primary carried a *CDKN1B* stop gain variant (**Fig. 2b-c**). Patients 3 and 7 carried *CDKN1B* variants (stop gain and splice donor) in metastatic primary tumors that were missing in the corresponding metastases (**Fig. 2d,h**). These variants were among a large number of private variants present at relatively low VAF in the

primaries, and may have arisen after metastasis (**Supplementary Figs. 14 and 15**). Other mutated Cancer Gene Census (Tate et al., 2019) genes included *MUC16* (5 samples, 4 independent events), a common false positive gene encoding the second largest human protein (Lawrence et al., 2013), *KMT2C* (5 samples, 3 independent events), *FBLN2* (2 independent events) and *ARID1B* (2 independent events; **Fig. 3**). *TERT* promoter mutations, frequent non-coding driver events in several cancer types (Horn et al., 2013; Huang et al., 2013), were absent, as were notable upstream mutations in other cancer genes (**Supplementary Fig. 16**). Consistent with other reports, there was thus a lack of obvious driver mutations beyond *CDKN1B*, which was not essential for metastasis.

Copy number alterations

Analysis of somatic CNAs gave further support for the phylogenetic relationships inferred from somatic SNVs (**Fig. 4a**). The metastatic tumor in Patient 1 (C) carried a distinct segmental loss on chr11 found in all metastases (G-K) but not the other intestinal tumors. Similarly, the metastatic tumor in Patient 2 (G) exhibited chromothripsis on chr13 (abundant clustered CNAs that oscillated between two states (Korbel and Campbell, 2013), **Supplementary Fig. 18**), and this exact complex pattern was mirrored in the corresponding lymph node metastasis (L) (**Fig. 4b**). Patient 2 and 7 had alterations on chr11 and chr20, respectively, that were present in primary tumors but absent in related metastases and thus presumably occurred after metastasis. Approximately 70-75% of SI-NETs have been shown to harbor hemizygous loss of chr18 (Andersson et al., 2009), and we accordingly observed chr18 loss in 9 of the 11 patients and in 32 of the 61 tumor samples. However, phasing of chr18 loss based on germline single nucleotide polymorphisms (SNPs) revealed that tumors deemed unrelated based on SNVs often had undergone loss of different chromosome homologs, while related tumors always showed loss of the same homolog ($P = 1.2 \times 10^{-4}$, binomial test; **Fig. 4a,c** and **Supplementary Fig. 17**). Other whole chromosome events (4, 5, 14 and 20) showed similar patterns of concordant/discordant chromosome homolog loss or gain in agreement with the established phylogenies (**Fig. 4a**). Among other recurrent events were segmental deletions on chr11 and chr13, as shown previously (Banck et al., 2013; Francis et al., 2013; Walter et al., 2018). No distinctive CNAs were shared in ways that contradicted the SNV-based evolutionary trees (**Fig. 4a**).

Figure 4. Somatic copy number alterations agree with phylogenies inferred from somatic SNVs. (a) Somatic CNA profiles for all samples (blue = deletion, red = amplification; segments <25 kb were excluded for visualization purposes). Phylogenetic relationships previously inferred from somatic SNVs (**Fig. 1-2**) are indicated to the right. Whole-chromosome losses and gains were phased based on germline SNPs (see **Supplementary Fig. 16**), with the two chromosome homologs indicated in green and orange and likely biallelic events shown in yellow. (b) Detailed view of chromothripsis on chr13 in Patient 2, present in the metastatic primary (G) and the metastasis (L). (c) Example of phasing of chr18 deletions in Patient 1.

Chromothripsis, not previously reported in SI-NET to our knowledge, was further supported by analysis of somatic structural alterations. Of 27 in-frame structural events, none of which were obvious drivers,

9 were intrachromosomal alterations on chr13 in Patient 2, all found in both the primary tumor and the metastasis shown by CNA to harbor chromothripsis on this chromosome (G and L; **Supplementary Fig. 19**).

Discussion

We find that in multifocal SI-NET, tumor development is initiated in multiple clonally independent cells, leading to a group of tumors that are unrelated in terms of somatic genetic evolution. Furthermore, we encountered several cases where two independent intestinal tumors had metastasized, each giving rise to a distinct liver or lymph node metastasis. This suggests that acquisition of metastatic properties is not a rare event in multifocal SI-NET and underscores the importance complete surgical removal of all intestinal lesions. Previous exome-based analyses of paired single intestinal and metastatic samples have yielded puzzling results with a highly varying degree of genetic overlap and in some cases no overlap at all (Francis et al., 2013; Walter et al., 2018), seen in one patient also in the present study, and our results thus suggest that the relevant primary tumors have not been sampled in such cases.

Multifocal entero-pancreatic carcinomas are typically seen in hereditary cancer syndromes such as MEN-1 and familial adenomatous polyposis (FAP). Cohort studies support that some SI-NETs may have a heritable component (Dumanski et al., 2017; Walsh et al., 2011), and multifocality has been suggested to occur preferentially in familial cases (Sei et al., 2016; Sei et al., 2015). However, multifocality is common in all SI-NET (Choi et al., 2017; Gangi et al., 2018), and key clinical parameters such as survival or age of diagnosis, which is typically lower for hereditary cancer, are similar in patients with or without multifocality (Choi et al., 2017; Gangi et al., 2018). The observation that the most recurrent somatic genetic event in SI-NET, chr18 loss, can affect both the maternal and the paternal chromosome homologs in the same patient argues against contribution from a predisposing chr18 germline variant by loss of heterozygosity (Zhang et al., 2020). Furthermore, in stark contrast to germline-induced gastrointestinal tumors, SI-NET only affects a limited intestinal segment. Given the general lack of established genetic drivers, and in the light of our results showing clonal independence, we suggest that future studies could focus on cancer-priming local factors that may contribute to

emergence of multifocal SI-NET. Such factors could theoretically include early embryonic genetic events present in blood as well as select EC cell lineages, thus precluding their identification as somatic events in the present study, or local environmental factors. While such factors remain elusive, it can be noted that SI-NET originates from epithelial enterochromaffin cells (EC-cells) that intrinsically interact with their surroundings by paracrine and endocrine serotonin secretion, synaptic-like connections with the enteric nervous system, and receptor-mediated nutrient sensing of luminal content (Bellono et al., 2017).

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Competing interests

None.

References

Alexandrov, L.B., Kim, J., Haradhvala, N.J., Huang, M.N., Tian Ng, A.W., Wu, Y., Boot, A., Covington, K.R., Gordenin, D.A., Bergstrom, E.N., *et al.* (2020). The repertoire of mutational signatures in human cancer. *Nature* 578, 94-101.

- Andersson, E., Sward, C., Stenman, G., Ahlman, H., and Nilsson, O. (2009). High-resolution genomic profiling reveals gain of chromosome 14 as a predictor of poor outcome in ileal carcinoids. *Endocr Relat Cancer* *16*, 953-966.
- Banck, M.S., Kanwar, R., Kulkarni, A.A., Boora, G.K., Metge, F., Kipp, B.R., Zhang, L., Thorland, E.C., Minn, K.T., Tentu, R., *et al.* (2013). The genomic landscape of small intestine neuroendocrine tumors. *J Clin Invest* *123*, 2502-2508.
- Bellono, N.W., Bayrer, J.R., Leitch, D.B., Castro, J., Zhang, C., O'Donnell, T.A., Brierley, S.M., Ingraham, H.A., and Julius, D. (2017). Enterochromaffin Cells Are Gut Chemosensors that Couple to Sensory Neural Pathways. *Cell* *170*, 185-198 e116.
- Chen, X., Schulz-Trieglaff, O., Shaw, R., Barnes, B., Schlesinger, F., Kallberg, M., Cox, A.J., Kruglyak, S., and Saunders, C.T. (2016). Manta: rapid detection of structural variants and indels for germline and cancer sequencing applications. *Bioinformatics* *32*, 1220-1222.
- Choi, A.B., Maxwell, J.E., Keck, K.J., Bellizzi, A.J., Dillon, J.S., O'Dorisio, T.M., and Howe, J.R. (2017). Is Multifocality an Indicator of Aggressive Behavior in Small Bowel Neuroendocrine Tumors? *Pancreas* *46*, 1115-1120.
- Crona, J., Gustavsson, T., Norlen, O., Edfeldt, K., Akerstrom, T., Westin, G., Hellman, P., Bjorklund, P., and Stalberg, P. (2015). Somatic Mutations and Genetic Heterogeneity at the CDKN1B Locus in Small Intestinal Neuroendocrine Tumors. *Ann Surg Oncol* *22 Suppl 3*, S1428-1435.
- Curtius, K., Wright, N.A., and Graham, T.A. (2018). An evolutionary perspective on field cancerization. *Nat Rev Cancer* *18*, 19-32.
- Dasari, A., Shen, C., Halperin, D., Zhao, B., Zhou, S., Xu, Y., Shih, T., and Yao, J.C. (2017). Trends in the Incidence, Prevalence, and Survival Outcomes in Patients With Neuroendocrine Tumors in the United States. *JAMA Oncol* *3*, 1335-1342.
- Dumanski, J.P., Rasi, C., Bjorklund, P., Davies, H., Ali, A.S., Gronberg, M., Welin, S., Sorbye, H., Gronbaek, H., Cunningham, J.L., *et al.* (2017). A MUTYH germline mutation is associated with small intestinal neuroendocrine tumors. *Endocr Relat Cancer* *24*, 427-443.

- Francis, J.M., Kiezun, A., Ramos, A.H., Serra, S., Pedamallu, C.S., Qian, Z.R., Banck, M.S., Kanwar, R., Kulkarni, A.A., Karpathakis, A., *et al.* (2013). Somatic mutation of CDKN1B in small intestine neuroendocrine tumors. *Nat Genet* 45, 1483-1486.
- Gangi, A., Siegel, E., Barmparas, G., Lo, S., Jamil, L.H., Hendifar, A., Nissen, N.N., Wolin, E.M., and Amersi, F. (2018). Multifocality in Small Bowel Neuroendocrine Tumors. *J Gastrointest Surg* 22, 303-309.
- Geoffroy, V., Herenger, Y., Kress, A., Stoetzel, C., Piton, A., Dollfus, H., and Muller, J. (2018). AnnotSV: an integrated tool for structural variations annotation. *Bioinformatics* 34, 3572-3574.
- Graham, T.A., McDonald, S.A., and Wright, N.A. (2011). Field cancerization in the GI tract. *Future Oncol* 7, 981-993.
- Guo, Z., Li, Q., Wilander, E., and Ponten, J. (2000). Clonality analysis of multifocal carcinoid tumours of the small intestine by X-chromosome inactivation analysis. *J Pathol* 190, 76-79.
- Horn, S., Figl, A., Rachakonda, P.S., Fischer, C., Sucker, A., Gast, A., Kadel, S., Moll, I., Nagore, E., Hemminki, K., *et al.* (2013). TERT promoter mutations in familial and sporadic melanoma. *Science* 339, 959-961.
- Huang, F.W., Hodis, E., Xu, M.J., Kryukov, G.V., Chin, L., and Garraway, L.A. (2013). Highly recurrent TERT promoter mutations in human melanoma. *Science* 339, 957-959.
- Katona, T.M., Jones, T.D., Wang, M., Abdul-Karim, F.W., Cummings, O.W., and Cheng, L. (2006). Molecular evidence for independent origin of multifocal neuroendocrine tumors of the enteropancreatic axis. *Cancer Res* 66, 4936-4942.
- Korbel, J.O., and Campbell, P.J. (2013). Criteria for inference of chromothripsis in cancer genomes. *Cell* 152, 1226-1236.
- Krzywinski, M., Schein, J., Birol, I., Connors, J., Gascoyne, R., Horsman, D., Jones, S.J., and Marra, M.A. (2009). Circos: an information aesthetic for comparative genomics. *Genome Res* 19, 1639-1645.
- Lawrence, M.S., Stojanov, P., Polak, P., Kryukov, G.V., Cibulskis, K., Sivachenko, A., Carter, S.L., Stewart, C., Mermel, C.H., Roberts, S.A., *et al.* (2013). Mutational heterogeneity in cancer and the search for new cancer-associated genes. *Nature* 499, 214-218.

- Lodato, M.A., Rodin, R.E., Bohrsen, C.L., Coulter, M.E., Barton, A.R., Kwon, M., Sherman, M.A., Vitzthum, C.M., Luquette, L.J., Yandava, C.N., *et al.* (2018). Aging and neurodegeneration are associated with increased mutations in single human neurons. *Science* 359, 555-559.
- Lodato, M.A., Woodworth, M.B., Lee, S., Evrony, G.D., Mehta, B.K., Karger, A., Lee, S., Chittenden, T.W., D'Gama, A.M., Cai, X., *et al.* (2015). Somatic mutation in single human neurons tracks developmental and transcriptional history. *Science* 350, 94-98.
- Magi, A., Pippucci, T., and Sidore, C. (2017). XCAVATOR: accurate detection and genotyping of copy number variants from second and third generation whole-genome sequencing experiments. *BMC Genomics* 18, 747.
- Priestley, P., Baber, J., Lolkema, M.P., Steeghs, N., de Bruijn, E., Shale, C., Duyvesteyn, K., Haidari, S., van Hoeck, A., Onstenk, W., *et al.* (2019). Pan-cancer whole-genome analyses of metastatic solid tumours. *Nature* 575, 210-216.
- Rosenthal, R., McGranahan, N., Herrero, J., Taylor, B.S., and Swanton, C. (2016). DeconstructSigs: delineating mutational processes in single tumors distinguishes DNA repair deficiencies and patterns of carcinoma evolution. *Genome Biol* 17, 31.
- Sei, Y., Feng, J., Zhao, X., Forbes, J., Tang, D., Nagashima, K., Hanson, J., Quezado, M.M., Hughes, M.S., and Wank, S.A. (2016). Polyclonal Crypt Genesis and Development of Familial Small Intestinal Neuroendocrine Tumors. *Gastroenterology* 151, 140-151.
- Sei, Y., Zhao, X., Forbes, J., Szymczak, S., Li, Q., Trivedi, A., Voellinger, M., Joy, G., Feng, J., Whatley, M., *et al.* (2015). A Hereditary Form of Small Intestinal Carcinoid Associated With a Germline Mutation in Inositol Polyphosphate Multikinase. *Gastroenterology* 149, 67-78.
- Stecher, G., Tamura, K., and Kumar, S. (2020). Molecular Evolutionary Genetics Analysis (MEGA) for macOS. *Mol Biol Evol* 37, 1237-1239.
- Tate, J.G., Bamford, S., Jubb, H.C., Sondka, Z., Beare, D.M., Bindal, N., Boutselakis, H., Cole, C.G., Creatore, C., Dawson, E., *et al.* (2019). COSMIC: the Catalogue Of Somatic Mutations In Cancer. *Nucleic Acids Res* 47, D941-D947.
- Walsh, K.M., Choi, M., Oberg, K., Kulke, M.H., Yao, J.C., Wu, C., Jurkiewicz, M., Hsu, L.I., Hooshmand, S.M., Hassan, M., *et al.* (2011). A pilot genome-wide association study shows genomic

variants enriched in the non-tumor cells of patients with well-differentiated neuroendocrine tumors of the ileum. *Endocr Relat Cancer* 18, 171-180.

Walter, D., Harter, P.N., Battke, F., Winkelmann, R., Schneider, M., Holzer, K., Koch, C., Bojunga, J., Zeuzem, S., Hansmann, M.L., *et al.* (2018). Genetic heterogeneity of primary lesion and metastasis in small intestine neuroendocrine tumors. *Sci Rep* 8, 3811.

Wang, Y.Z., Carrasquillo, J.P., McCord, E., Vidrine, R., Lobo, M.L., Zamin, S.A., Boudreaux, P., and Woltering, E. (2014). Reappraisal of lymphatic mapping for midgut neuroendocrine patients undergoing cytoreductive surgery. *Surgery* 156, 1498-1502; discussion 1502-1493.

Yates, L.R., and Campbell, P.J. (2012). Evolution of the cancer genome. *Nat Rev Genet* 13, 795-806.

Zhang, Z., Makinen, N., Kasai, Y., Kim, G.E., Diosdado, B., Nakakura, E., and Meyerson, M. (2020). Patterns of chromosome 18 loss of heterozygosity in multifocal ileal neuroendocrine tumors. *Genes Chromosomes Cancer*.

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Supplementary appendix

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Supplementary Materials and Methods

Immunohistochemistry

Sections of all sampled tumors from 11 patients with SI-NET were subjected to antigen retrieval using EnVision FLEX Target Retrieval Solution (high pH) in a Dako PT-Link. Immunohistochemical staining was performed in a Dako Autostainer Link using EnVision FLEX according to the manufacturer's instructions (DakoCytomation).

Mapping and somatic variant calling

The reads were mapped to the human reference genome (hg19 assembly) using BWA as part of Sentieon Genomics Tools (bwa-mem v0.7.15.r1140 for patients 1-5 and 0.7.17.r1188 for patients 6-11), which further also performs deduplication, realignment and sorting. Mutect2 was run using default parameters, and the output was fed into the filterMutectCalls tool with a mean median mapping quality score requirement of 10. With VarScan, the somatic tool was used to call variants by default cutoffs and a minimum variant allele frequency of 0.01 from SAMtools (v1.9) pileups of reads with Phred-scaled mapping quality ≥ 15 and variant base quality ≥ 20 . The VarScan processSomatic tool was then used to get only the somatic SNVs and indels, where a minimum support of two reads was required for a variant to be called. The somaticFilter tool was also used to discard SNVs called within SNP clusters or within 3 bp of somatic and germline indels. Variants were further filtered to have coverage ≥ 20 reads in the normal, to be present on both strands, and to be absent in the normal. The final set of variants was yielded by intersecting outputs from the two callers, followed by annotation using ANNOVAR (v2019Oct24) (Wang et al., 2010) and Variant Effect Predictor (VEP) (McLaren et al., 2010). To minimize false positive calls in phylogenetic analyses, population variants in dbSNP150 (provided by ANNOVAR) and the ENSEMBL variant database (provided by VEP) were removed. Analysis of potential driver mutations and mutational signatures was based on less stringent population variant filtering (dbSNP138), to avoid false negatives and since extensive SNP filtering can skew results from mutational signatures analyses. For high-sensitivity analysis of pairwise shared variants, mutations detected by the standard pipeline as described above in any of the two samples in a pair were whitelisted for detection using the unfiltered VarScan somaticFilter output, allowing more sensitive detection of overlapping subclonal mutations.

Sample purity and telomer lengths

Samples were assed for tumor cell content using PurBayes (Larson and Fridley, 2013) and telomere lengths were determined using Computel (Nersisyan and Arakelyan, 2015) using default settings, both presented in **Supplementary Table S2**.

Viral reads and transposition events

Analysis of WGS reads for viral content, which produced only low counts of expected *Herpesviridae* family reads or reads consistent with plasmid vector contamination thus giving no support for a role for DNA viruses in SI-NET, was performed using a previously established pipeline (Tang et al., 2013). TraFiC-mem was used for detection of somatic mobile element insertions (Tubio et al., 2014), which revealed only a single LINE1 transposition event (in *BCAS3* in sample 5F).

Supplementary Figure 1

Pat. 1

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						70%
B						40%
C						50%
D						70%
E						70%
F						70%
G						50%
H						60%
I						70%
J						50%
K						60%

Immunohistochemical analysis of all sequenced samples from Patient 1. All tumors display similar morphology, and all express the neuroendocrine markers, SYP (synaptophysin), CHGA (chromogranin A) and 5-HT (serotonin) as well as the clinically relevant SSTR2 (somatostatin receptor 2). The assessment of tumor cell content was performed visually on hematoxylin and eosin-stained (H&E) sections of the tumors. The letter in each row indicates the sample ID.

Supplementary Figure 2

Pat. 2

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						50%
B						50%
C						60%
D						50%
E						30%
F						30%
G						50%
H						50%
I						60%
J						60%
K						60%
L						50%

Immunohistochemical analysis of all sequenced samples from Patient 2. All tumors display similar morphology, and all express the neuroendocrine markers, SYP (synaptophysin), CHGA (chromogranin A) and 5-HT (serotonin) as well as the clinically relevant SSTR2 (somatostatin receptor 2). The assessment of tumor cell content was performed visually on hematoxylin and eosin-stained (H&E) sections of the tumors. The letter in each row indicates the sample ID.

Supplementary Figure 3

Pat. 3

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						50%
B						50%
C						70%
D						90%

Pat. 4

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						70%
B						70%
C						70%
D						80%

Pat. 5

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						40%
B						50%
C						70%
D						70%
E						70%
F						40%
G						0%

Immunohistochemical analysis of all sequenced samples from Patient 3-5. All tumors display similar morphology, and all express the neuroendocrine markers, SYP (synaptophysin), CHGA (chromogranin A) and 5-HT (serotonin) as well as the clinically relevant SSTR2 (somatostatin receptor 2). The assessment of tumor cell content was performed visually on hematoxylin and eosin-stained (H&E) sections of the tumors. The letter in each row indicates the sample ID. Sample 5G is from normal intestinal mucosa.

Supplementary Figure 4

Pat. 6

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						40%
B						70%
C						90%
D						80%

Pat. 7

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						30%
B						30%
C						40%
D						90%

Pat. 8

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						30%
B						40%
C						70%
D						70%

Immunohistochemical analysis of all sequenced samples from Patient 6-8. All tumors display similar morphology, and all express the neuroendocrine markers, SYP (synaptophysin), CHGA (chromogranin A) and 5-HT (serotonin) as well as the clinically relevant SSTR2 (somatostatin receptor 2). The assessment of tumor cell content was performed visually on hematoxylin and eosin-stained (H&E) sections of the tumors. The letter in each row indicates the sample ID.

Supplementary Figure 5

Pat. 9

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						70%
B						70%
C						70%

Pat. 10

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						80%
B						40%
C						50%
D						90%
E						80%

Pat. 11

Tumor	H&E	SYP	CHGA	5-HT	SSTR2	Tumor content
A						50%
B						30%
C						80%
D						80%

Immunohistochemical analysis of all sequenced samples from Patient 9-11. All tumors display similar morphology, and all express the neuroendocrine markers, SYP (synaptophysin), CHGA (chromogranin A) and 5-HT (serotonin) as well as the clinically relevant SSTR2 (somatostatin receptor 2). The assessment of tumor cell content was performed visually on hematoxylin and eosin-stained (H&E) sections of the tumors. The letter in each row indicates the sample ID.

Supplementary Figure 6

Analysis of variant allele frequency (VAF) distributions of primary-metastasis shared SNVs. VAF distribution plots of SNVs shared between primary tumors and corresponding metastases in all relevant samples. The overall VAF distribution for all high-confidence mutations in each sample is shown as reference (red; normalized to fit y-axis scale). More detailed views and discussions of Patient 3 and Patient 7 are provided in **Supplementary Figs. 14 and 15**.

Supplementary Figure 7

High-sensitivity search for shared variants by whitelisting of SNVs called at high confidence in at least one sample. A two-step procedure was used, where any somatic SNVs called at high confidence (detected by both Mutect and VarScan using various filters including a strand filter as described in Materials and Methods) in at least one sample were “whitelisted”. This was followed by a more sensitive search for presence of the whitelisted variants using VarScan without stringent filters (e.g. exclusion of strand filter, see Materials and Methods). The number and letter in each row/column label indicates the patient number and sample ID, respectively.

Supplementary Figure 8

Variant allele frequency (VAF) distributions indicate low-level contamination between samples D and F from Patient 5. Samples D (primary tumor) and F (lymph node metastasis) in Patient 5 shared 8 SNVs in the main phylogenetic analysis (main Fig. 2). A high-sensitivity search for shared variants (**Supplementary Fig. 7**) revealed 58 additional shared variants called at high confidence in at least one of the samples while being detectable using relaxed filters (e.g. exclusion of strand filter, see Materials and Methods) in the other sample. These 66 shared SNVs were present at high VAFs in sample D (top histogram) while being highly subclonal in sample F (bottom histogram). This supports that the metastasis sample (F) was contaminated with DNA from D, for example during sample handling or, alternatively, that trace amounts of material from D reached F through metastatic spread. Overall VAF distributions for all high-quality mutations in each sample is shown for comparison (red; normalized to fit y-axis scale).

Supplementary Figure 9

Variant allele frequency (VAF) distributions indicate low-level contamination between samples A and C from Patient 9. A high-sensitivity search for subclonal shared SNVs (**Supplementary Fig. 6**) revealed 14 variants shared between samples A (primary tumor) and C (lymph node metastasis) in Patient 9, all present at high confidence in at least one of the samples while being detectable using relaxed filters (e.g. exclusion of strand filter, see Materials and Methods) in the other. The graphs show that variants present at high confidence in A was generally found at low VAF in C, and vice versa. A likely explanation is sample contamination, although transfer of trace amounts of tumor material through metastatic spread could in principle also explain these patterns. Overall VAF distributions for all high-quality mutations in each sample is shown for comparison (red; normalized to fit y-axis scale).

Supplementary Figure 10

Physical positioning of tumors F, G and L in Patient 2.

Supplementary Figure 11

Variant allele frequency (VAF) distributions for 8 variants found in samples F, G and L from Patient 2. A set of 8 SNVs was shared between samples F, G and L (lymph node metastasis) in Patient 2 (main **Fig. 2**). This number stayed constant when using a high-sensitivity approach to uncover subclonal shared SNVs (**Supplementary Fig. 7**). Manual inspection in IGV supported that the shared calls were mostly of high quality, with only one having characteristics of being a false positive call (**Supplementary Table S3**). The shared SNVs showed normal VAF distributions (comparable to other mutations) in all samples. The overall VAF distribution for all high-quality mutations in each sample is shown for comparison (red; normalized to fit y-axis scale).

Supplementary Figure 12

Highly coherent trinucleotide substitution profile across the cohort. (a) Average trinucleotide substitution profile in the cohort. (b) Cosine similarity of per-sample trinucleotide profiles and the cohort average shown in (a). Most samples showed high similarity, and deviation from the average profile was associated with reduced mutation burden, thus reducing the ability to accurately determine the signature. This is most clearly seen for the normal mucosa tissue samples, which all have low mutation counts.

Supplementary Figure 13

Positive correlations between per-sample estimated purity and average variant allele frequency (VAF). Tumor purity, as determined by PurBayes, correlated positively with the SNV burden in each patient. (a) Results using SNV calls used for phylogenetic analyses, which were stringently filtered for known population variants (dbSNP150 and ENSEMBL variants). (b) Results using relaxed population variant filtering (dbSNP138), as used for driver mutations and mutational signature analyses, were nearly identical.

Supplementary Figure 14

Variant allele frequency (VAF)-based analysis of a *CDKN1B* mutation in Patient 3. A *CDKN1B* SNV present in the metastatic primary tumor B is one of a large number of private variants (not shared with the metastasis D) present at relatively low VAF compared to shared variants, which showed higher mean VAF and bimodality in the primary tumor B. This is indicative of subclonality, early whole genome duplication, or a combination of both.

Supplementary Figure 15

Variant allele frequency (VAF)-based analysis of a *CDKN1B* mutation in Patient 7. A *CDKN1B* SNV present in the metastatic primary tumor A is one of a many private variants (not shared with the metastasis D) present at relatively low VAF compared to shared variants.

Supplementary Figure 16

Overview of potential driver mutations including regulatory (upstream and UTR) mutations for all 11 patients. All CGC genes with non-synonymous mutations in more than one sample are shown. Upstream and UTR mutations are shown as reported by AnnoVar. Phylogenetic relationships previously inferred from somatic SNVs (main Fig. 1-2) are indicated at the bottom. Somatic SNV burdens, potential driver mutations (Cancer Gene Census genes mutated in >1 sample) and mutational signature loadings in this figure were determined using less rigorous population variant filtering compared to the phylogenetic analyses (see Materials and Methods). CGC, Cancer Gene Census.

Supplementary Figure 17

Chromosomal phasing of whole-chromosome copy number events based on germline heterozygous SNPs. Heterozygous germline SNPs on chr18 (read coverage 20 and VAF ranging from 0.25 to 0.75 in the blood normal) were used to determine what chromosome homolog was altered. The scatter plots compare VAFs for individual SNPs in a reference sample compared to other relevant samples. The data was down-sampled to 20% to reduce dot density in the plots. The related samples 7A and 7D lacked VAF bimodality on chr4/5 indicative of biallelic gain, further supported by high copy number amplitude in these samples. Lack of bimodality on chr18 in the related samples 3B and 3D may be explained by a biallelic deletion on a whole-genome duplication background (notably the unrelated sample, 3C, deviates and shows bimodality). Alternative explanations include a homozygous deletion (unlikely due to low-level amplitude change in 3D) or a false copy number call (unlikely due to strong amplitude change in 3B).

Supplementary Figure 18

Clustered copy number alterations on chromosome 13 in Patient 2 compatible with chromothripsis. CNAs on chromosome 13 in Patient 2 (samples G and L) oscillated between two distinct amplitude states, which is a hallmark of chromothripsis.

Supplementary Figure 19

Analysis of somatic genomic structural alterations. In-frame fusion gene pairs colored by alteration type, with translocations (BND) shown in green, inversions in red, duplications in purple, and deletions in orange.

Table S1

Patient characteristics

		Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6	Patient 7	Patient 8	Patient 9	Patient 10	Patient 11
Year of birth		1959	1941	1937	1948	1937	1930	1940	1967	1942	1942	1943
Gender		F	M	M	M	F	M	F	F	F	M	F
Medication		0	Amlodipin, Acetylsalicylic acid, Finasteride	Ramipril, Metoprolol, Acetylsalicylic acid, Simvastatin	Ramipril, Metoprolol, Acetylsalicylic acid, Simvastatin	Omeprazol, Ticagrelor, Insulin, Levothyroxine, Prednisolone, Atorvastatin, Ramipril	Insulin, Corticosteroids	Omeprazol, Bendroflumetiazide, Papaverine, Budesonide	0	Omeprazol, Levothyroxine	Finasterid	Tiotropium
Family history of SI-NET or other endocrine tumors		0	0	0	0	0	0	0	0		0	0
Preoperative data	Symptoms	Yes, intestinal obstruction	Unclear/diffuse abdominal pain	No	No/ previous liver resection	Not initially, developed bleeding and abdominal pain.	Carcinoid flush, fatigue	Abdominal pain, carcinoid flush	Carcinoid flush	Abdominal pain	Abdominal pain	Abdominal pain
	S-CGA (mg/L, reference value <102)	104	61	58	na	na	na	12.7	630	965	250	312
	tU-5-HIAA (µmol/24h, reference value 0-50)	41	26	120	87	35	150	92	450	303	160	28
	CT-SCAN	2018 nov. Intestinal tumour and lymph node metastases	2019 jan. Intestinal tumour and lymph node metastases	2014 february, lymph node metastases	2016 january, lymph node metastases	2012 december, lymph node metastases	Liver metastases	Lymphnode metastases, possible liver metastases	Liver metastases	Liver and lymphnode metastases	Liver metastases	Lymphnode metastases
	SSTR2 Scintigraphy	No	No	2014 february, lymph node metastases	2016 january, lymph node metastases	2012 december, lymph node metastases	Liver, lung and lymphnode metastases	Lymphnode metastases	Lymphnode metastases, liver metastases	Lymphnode metastases, bone metastases and liver metastases	No	No
	SSTR2 PET-CT	2018 november, intestinal tumour and lymph node metastases	2019 january, multiple Intestinal tumours and lymphnode metastases	No	No	No	No	No	No	No	Intestinal tumor, lymphnode metastases and liver metastases	Intestinal tumor and lymphnode metastases
Surgical procedure	Year of surgery	2019 january	2019 february	2014 april	2016 february	2014 october	2011 april	2012 april	2015 december	2016 april	2018 september	2019 april
	Radical surgery	No	Yes	Yes	Yes/previous liver resection	Yes	No	Yes	Yes	No	No	Yes
	Type of surgery	Resection of Intestinal tumours, local lymph nodes and peritoneal carcinosis	Resection of Intestinal tumours and local lymph node metastases	Resection of Intestinal tumours and local lymph node metastases	Resection of Intestinal tumours and local lymph node metastases	Resection of Intestinal tumours and local lymph node metastases	Resection of Intestinal tumours and local lymph node metastases	Resection of Intestinal tumours, local lymph node metastases and Liver metastases	Resection of Intestinal tumours, local lymph node metastases and Liver metastases	Resection of Intestinal tumours and local lymph node metastases	Resection of Intestinal tumours, local lymph node metastases and Liver metastases	Resection of Intestinal tumours and local lymph node metastases
Pathology	Grade (WHO 2010)	2	1	1	1	1	1	2	1	1	1	1
	Stage (TNM 7th edition)	IIIB (pT4(m)N2)	IIIB (pT4(m)N2Mx)	IIIB (pT3(m)N1Mx)	IV (pT3(m)N1M1)	IIIB (pT3(m)N1)	IV (pT3(m)N1M1)	IV (pT3(m)N1M1)	IV (pT2(m)N1M1)	IV (pT3(m)N1M1)	IV (pT3(m)N1M1)	IIIB (pT2(m)N1Mx)
Current status 2019-11	Alive	Yes	Yes	Yes	No, 2018-02-21	No, 2015-02-23	No, 2017-01-02	Yes	Yes	Yes	Yes	Yes
	Evidence of disease	Yes, non radical resekction	No	Unclear, minor radiological liver lesions, stable for 2 years	na	na	na	Yes, Liver metastases	Yes, Liver metastases	Yes, non radical resekction	Yes, non radical resekction	No
	S-CGA (mg/L, reference value <102)	66	86	42	na	na	na	1290	36	1410	170	144
	tU-5-HIAA (µmol/24h, reference value 0-50)	13	30	20	na	na	na	68	36	94	65	34
	Progression	No	No	No	na	na	na	Yes	No	Yes	No	No

Table S2

Sample overview and WGS statistics

Library ID	Patient	Sex	Age	Sample ID	Type	Purity (PurBayes)	Telomer length	Reads	Depth	Source	SNVs (strict dbSNP filtering)	Indels (strict dbSNP filtering)	SNVs (relaxed dbSNP filtering)	Indels (relaxed dbSNP filtering)
3854	1	Female	60	A	primary tumor	53	1895.2	398407624	38.5	tumor tissue DNA	591	33	781	54
3856	1	Female	60	B	primary tumor	29	2186.2	446120409	43.1	tumor tissue DNA	631	45	846	80
3857	1	Female	60	C	primary tumor	43	1948.3	403972873	39.1	tumor tissue DNA	1166	51	1458	75
3858	1	Female	60	D	primary tumor	65	2097.1	391424250	37.9	tumor tissue DNA	1154	54	1452	80
3859	1	Female	60	E	primary tumor	29	2262	389950585	37.7	tumor tissue DNA	353	14	498	46
3860	1	Female	60	F	primary tumor	32	2120.2	379365461	36.7	tumor tissue DNA	831	37	1045	65
3861	1	Female	60	G	lymph node metastasis	94	993.5	369624746	35.7	tumor tissue DNA	1615	97	2026	124
3862	1	Female	60	H	lymph node metastasis	82	1192.4	356340972	34.5	tumor tissue DNA	1589	97	1990	132
3863	1	Female	60	I	lymph node metastasis	72	1254.6	379469599	36.7	tumor tissue DNA	1725	89	2116	119
3864	1	Female	60	J	carcinosis	69	1275.9	399174372	38.6	tumor tissue DNA	1731	77	2119	112
3865	1	Female	60	K	carcinosis	92	1054.6	385665478	37.3	tumor tissue DNA	1749	79	2127	112
B3854	1	Female	60	-	blood	NA	2801.3	396919605	38.4	normal blood DNA	NA	NA	NA	NA
3873	2	Male	78	A	primary tumor	51	2059.6	421453654	40.8	tumor tissue DNA	769	54	1014	91
3878	2	Male	78	B	primary tumor	40	2249.8	444369658	43.0	tumor tissue DNA	1108	51	1391	90
3879	2	Male	78	C	primary tumor	26	3275.3	390352470	37.8	tumor tissue DNA	143	8	220	27
3880	2	Male	78	D	primary tumor	68	2077	413563843	40.0	tumor tissue DNA	844	48	1088	77
3882	2	Male	78	E	primary tumor	43	1703.1	406878784	39.4	tumor tissue DNA	894	47	1117	71
3887	2	Male	78	F	primary tumor	38	2138.5	424714790	41.1	tumor tissue DNA	1061	69	1336	114
3888	2	Male	78	G	primary tumor, largest	73	1259.6	400745503	38.8	tumor tissue DNA	2161	85	2635	112
3889	2	Male	78	H	primary tumor	49	1800.5	413256977	40.0	tumor tissue DNA	912	36	1119	73
3898	2	Male	78	I	primary tumor	49	2615.8	381324850	36.9	tumor tissue DNA	1148	57	1431	85
3900	2	Male	78	J	primary tumor	29	2300.6	442407873	42.8	tumor tissue DNA	1271	75	1601	110
3901	2	Male	78	K	primary tumor	44	2524	388846048	37.6	tumor tissue DNA	823	33	1026	62
3902	2	Male	78	L	lymph node metastasis	47	1793.8	370882991	35.9	tumor tissue DNA	1527	67	1882	83
B3873	2	Male	78	-	blood	NA	2646.6	387304060	37.5	normal blood DNA	NA	NA	NA	NA
3482	3	Male	77	A	primary tumor	27	1896.8	379910672	36.7	tumor tissue DNA	762	30	965	44
3483	3	Male	77	B	primary tumor	44	1816	386516909	37.4	tumor tissue DNA	2947	119	3597	154
3484	3	Male	77	C	primary tumor	64	1482.5	432709081	41.9	tumor tissue DNA	3300	120	3843	158
3480	3	Male	77	D	lymph node metastasis	28	2390.4	405481985	39.2	tumor tissue DNA	1566	61	1922	87
B3480	3	Male	77	-	blood	NA	2746.6	465595324	45.0	normal blood DNA	NA	NA	NA	NA
3665	4	Male	68	A	primary tumor	31	2361.2	383800522	37.1	tumor tissue DNA	593	34	784	65
3666	4	Male	68	B	primary tumor, largest	58	2723.2	391904924	37.9	tumor tissue DNA	850	51	1107	81
3667	4	Male	68	C	primary tumor	45	2447.5	388586595	37.6	tumor tissue DNA	1751	70	2151	93
3664	4	Male	68	D	lymph node metastasis	45	1903.8	391908871	37.9	tumor tissue DNA	2471	119	3022	155
B3664	4	Male	68	-	blood	NA	3339.8	385186116	37.3	normal blood DNA	NA	NA	NA	NA
3509	5	Female	77	A	primary tumor	57	2213.9	395371288	38.2	tumor tissue DNA	1521	84	1832	106
3510	5	Female	77	B	primary tumor	62	2529.3	410912098	39.7	tumor tissue DNA	1409	63	1717	88
3511	5	Female	77	C	primary tumor, largest	45	2132.3	440905042	42.6	tumor tissue DNA	1074	54	1416	90
3512	5	Female	77	D	primary tumor	40	2070.7	399759106	38.7	tumor tissue DNA	996	39	1355	79
3513	5	Female	77	E	primary tumor	28	2459.8	406014379	39.3	tumor tissue DNA	810	33	1044	74
3508	5	Female	77	F	lymph node metastasis	28	2344	407683036	39.4	tumor tissue DNA	1662	60	2063	99
N3514	5	Female	77	G	normal mucosa	17	2217.6	403874867	39.1	normal tissue DNA	16	4	82	30
B3508	5	Female	77	-	blood	NA	2593	414206350	40.1	normal blood DNA	NA	NA	NA	NA
3224	6	Male	81	A	primary tumor	57	2109.9	324100879	31.3	tumor tissue DNA	925	50	1194	85
3225	6	Male	81	B	primary tumor, largest	60	2358.5	384806769	37.2	tumor tissue DNA	745	35	1019	90
3223	6	Male	81	C	lymph node metastasis	59	1588.5	350836604	33.9	tumor tissue DNA	1225	67	1557	112
3226	6	Male	81	D	lymph node metastasis	95	1952	352147102	34.1	tumor tissue DNA	765	29	1050	72
3228	6	Male	81	E	normal mucosa	13	1949.8	348620358	33.7	normal tissue DNA	11	8	68	48
B3224	6	Male	81	-	blood	NA	2772	348218764	33.7	normal blood DNA	NA	NA	NA	NA
3299	7	Female	72	A	primary tumor	41	1319.5	393189281	38.0	tumor tissue DNA	1767	83	2222	145
3300	7	Female	72	B	primary tumor, largest	50	1865.7	342654685	33.1	tumor tissue DNA	2365	98	2933	172
3301	7	Female	72	C	lymph node metastasis	64	1640.8	320452865	31.0	tumor tissue DNA	1301	63	1661	97
3303	7	Female	72	D	liver metastasis	61	1067.2	345745784	33.4	tumor tissue DNA	2431	119	2979	162
3302	7	Female	72	E	normal mucosa	16	2994.5	387012589	37.4	normal tissue DNA	40	16	116	55
B3299	7	Female	72	-	blood	NA	3184	383188102	37.1	normal blood DNA	NA	NA	NA	NA
3564	8	Female	48	A	primary tumor, largest	46	3215.2	352065513	34.1	tumor tissue DNA	411	29	584	66
3565	8	Female	48	B	primary tumor	62	2476.9	353743231	34.2	tumor tissue DNA	456	21	643	63
3562	8	Female	48	C	lymph node metastasis	60	1874.4	336038434	32.5	tumor tissue DNA	1220	51	1528	80
3567	8	Female	48	D	liver metastasis	56	2019.5	370689613	35.9	tumor tissue DNA	1677	65	2069	98
3566	8	Female	48	E	normal mucosa	19	2857.7	359606525	34.8	normal tissue DNA	22	6	88	52
B3562	8	Female	48	-	blood	NA	3328.3	354739843	34.3	normal blood DNA	NA	NA	NA	NA
3687	9	Female	74	A	primary tumor, largest	32	2505.2	393090042	38.0	tumor tissue DNA	1459	84	1918	153
3688	9	Female	74	B	primary tumor	46	2544.2	336900791	32.6	tumor tissue DNA	1323	63	1676	109
3686	9	Female	74	C	lymph node metastasis	57	1517.1	337077190	32.6	tumor tissue DNA	1307	55	1689	101
B3686	9	Female	74	-	blood	NA	3335	354812231	34.3	normal blood DNA	NA	NA	NA	NA
3832	10	Male	76	A	primary tumor	71	2910.7	310108169	30.0	tumor tissue DNA	1535	56	1839	93
3833	10	Male	76	B	primary tumor	45	2392.3	348570682	33.7	tumor tissue DNA	1451	65	1812	124
3834	10	Male	76	C	primary tumor	34	3063.7	346026858	33.5	tumor tissue DNA	639	22	832	63
3831	10	Male	76	D	lymph node metastasis	54	2969	335751842	32.5	tumor tissue DNA	1856	63	2220	103
3835	10	Male	76	E	liver metastasis	57	1686.6	349408672	33.8	tumor tissue DNA	1850	115	2275	158
B3831	10	Male	76	-	blood	NA	3689.1	340194676	32.9	normal blood DNA	NA	NA	NA	NA
3921	11	Female	76	A	primary tumor	52	2207	307648042	29.8	tumor tissue DNA	3223	114	3770	152
3922	11	Female	76	B	primary tumor	38	2970.9	350684474	33.9	tumor tissue DNA	2614	84	3194	144
3923	11	Female	76	C	primary tumor	39	2908.1	318831418	30.8	tumor tissue DNA	595	25	771	60
3920	11	Female	76	D	lymph node metastasis	68	1570.8	318611052	30.8	tumor tissue DNA	3390	128	4081	164
B3920	11	Female	76	-	blood	NA	3179	349993880	33.9	normal blood DNA	NA	NA	NA	NA

Average:
36.6

Table S3

Manual inspection of 29 remaining spurious overlapping SNVs (appearing in pairs sharing <10 mutations) in high-confidence mutation calls

Patient	Pair	SNV	IGV comment
1	B-H	2:87799422:A>G	Low mapping quality in region
1	B-I	2:87799422:A>G	
1	B-F	3:13024277:G>A	No visible problems
2	F-G	1:104673351:G>A	No visible problems
2	F-L	1:104673351:G>A	
2	D-F	1:206571894:A>T	Visible in blood normal
2	D-H	1:206571894:A>T	
2	F-H	1:206571894:A>T	
2	B-K	1:245931643:C>A	No visible problems
2	F-G	10:57167423:C>T	No visible problems
2	F-L	10:57167423:C>T	
2	F-G	11:23751995:G>T	No visible problems
2	F-L	11:23751995:G>T	
2	F-G	12:113600342:G>A	No visible problems
2	F-L	12:113600342:G>A	
2	F-G	2:112605597:C>A	No obvious problems (repeat region)
2	F-I	2:112605597:C>A	
2	F-L	2:112605597:C>A	
2	G-I	2:112605597:C>A	
2	I-L	2:112605597:C>A	
2	B-C	2:87625895:A>G	
			Highly problematic region with elevated coverage and mismatches indicative of mapping issues
			Locally elevated coverage indicative of mapping issues, repeat region, flanks a population SNP visible in the blood normal
2	A-B	21:11083762:G>A	
2	A-D	21:11083762:G>A	
2	A-E	21:11083762:G>A	
2	A-F	21:11083762:G>A	
2	A-G	21:11083762:G>A	
2	A-H	21:11083762:G>A	
2	A-I	21:11083762:G>A	
2	A-L	21:11083762:G>A	
2	B-D	21:11083762:G>A	
2	B-E	21:11083762:G>A	
2	B-F	21:11083762:G>A	
2	B-G	21:11083762:G>A	
2	B-H	21:11083762:G>A	
2	B-I	21:11083762:G>A	
2	B-L	21:11083762:G>A	
2	D-E	21:11083762:G>A	
2	D-F	21:11083762:G>A	
2	D-G	21:11083762:G>A	
2	D-H	21:11083762:G>A	
2	D-I	21:11083762:G>A	
2	D-L	21:11083762:G>A	
2	E-F	21:11083762:G>A	
2	E-G	21:11083762:G>A	
2	E-H	21:11083762:G>A	
2	E-I	21:11083762:G>A	
2	E-L	21:11083762:G>A	
2	F-G	21:11083762:G>A	
2	F-H	21:11083762:G>A	
2	F-I	21:11083762:G>A	
2	F-L	21:11083762:G>A	
2	G-H	21:11083762:G>A	
2	G-I	21:11083762:G>A	
2	H-I	21:11083762:G>A	
2	H-L	21:11083762:G>A	
2	I-L	21:11083762:G>A	

2	F-G	4:53985148:G>A	No visible problems
2	F-L	4:53985148:G>A	
2	F-G	7:45320406:T>C	No obvious problems (repeat region)
2	F-L	7:45320406:T>C	
4	A-C	4:159885559:T>G	No obvious problems (repeat region)
5	D-F	1:72880173:A>C	No visible problems
5	D-F	16:19985736:A>G	No visible problems
5	D-F	4:16881745:C>T	No visible problems
5	A-B	4:85800162:C>T	No obvious problems (repeat region)
			No visible problems (overlaps population SNP with different minor allele)
5	D-F	5:178581108:C>A	
5	A-B	5:2954580:A>C	No obvious problems (repeat region)
5	A-G	5:2954580:A>C	
5	B-G	5:2954580:A>C	
5	A-B	6:166155341:G>T	No visible problems
5	A-G	6:166155341:G>T	
5	B-G	6:166155341:G>T	
5	D-F	7:30598385:G>T	No obvious problems (repeat region)
			No visible problems (flanks population SNV)
5	D-F	7:62472926:C>A	
5	D-F	X:10086043:T>G	No visible problems
5	D-F	X:143098302:G>A	No obvious problems (repeat region)
5	A-B	X:49126275:A>C	Visible in blood normal
			Locally elevated coverage indicative of mapping issues, repeat region, variant is visible in the blood normal, flanks a population SNP
7	A-B	1:143283399:G>A	
7	A-E	3:147071941:G>A	No visible problems
7	D-E	3:147071941:G>A	
9	A-C	15:50066578:G>T	No visible problems (repeat region)

Table S4

Somatic non-synonymous, splicing, upstream (promoter) or UTR mutations by gene based on calls filtered for dbSNP138 population variants

Gene	Cancer Gene Census	Sample	Position	Substitution	Function	dbSNP138	dbSNP150	ENSEMBL variant
PPFIA1	no	1A	11:7011661 3	C>T	upstream nonsynonymous	no	no	no
FUT4	no	1A	11:9427730 7	G>A	SNV nonsynonymous	no	no	no
TMEM132C	no	1A	12:1291785 13	T>A	SNV	no	no	no
HOXC9	no	1A	12:5439705 8	C>T	UTR3	no	no	yes
HOXC9	no	6B	12:5439413 7	CGCGCCCAA GCCGGCAGT GTTTCAGCAC GTCGTGG>C	nonframeshift deletion	no	no	no
HOXC9	no	6D	12:5439413 7	GTTTCAGCAC GTCGTGG>C	nonframeshift deletion	no	no	no
DDIT3;MARS	no	1A	12:5791039 2	TAATA>T	UTR3	no	yes	yes
DPY19L2	no	1A	12:6406261 1	C>T	upstream	no	no	no
UNC13C	no	1A	15:5427012 6	T>C	upstream	no	no	no
ALDH3A2	no	1A	17:1957914 9	A>G	UTR3	no	no	no
CD79B	yes	1A	17:6200991 1	G>A	upstream	no	no	no
PHF13	no	1A	1:6673602	A>G	upstream	no	yes	yes
PHF13	no	2A	1:6673749	G>A	upstream nonsynonymous	no	yes	yes
EN1	no	1A	2:11960457 4	G>T	SNV	no	no	yes
TTC31	no	1A	2:74721435 4:18754043	C>A	UTR3 nonsynonymous	no	no	no
FAT1	yes	1A	4	T>C	SNV	no	no	no
FER	no	1A	5:10852986 2	T>G	UTR3	no	no	no
ESR1	yes	1A	6:15213040 8	T>C	splicing	no	yes	yes
ESR1	yes	3D	6:15201124 0	T>G	upstream	no	no	no
VARS2	no	1A	6:30882815	G>A	splicing nonsynonymous	no	no	no
GSTA3	no	1A	6:52770581	G>T	SNV	no	no	no
CCDC129	no	1A	7:31552909	A>G	upstream	no	no	no
HIP1	yes	1A	7:75165054	C>T	UTR3 nonsynonymous	no	yes	yes
DOCK5	no	1A	8:25156605 9:13088101	C>A	SNV	no	no	no
SLC25A25-AS1	no	1A	4	G>A	upstream	no	no	yes
GPM6B	no	1A	X:13789172 10:1499701	T>C	UTR3	no	no	no
DCLRE1C	no	1B	5	C>T	upstream	no	no	no
UBN1	no	1B	16:4930465	A>G	UTR3	no	no	no
OR1E3	no	1B	17:3018985	G>A	upstream	no	no	no

CACNA1G	no	1I	17:4866942 5	C>T	nonsynonymous SNV	no	no	no
NAT9	no	1B	17:7276744 5	G>A	UTR3	no	yes	yes
PNPLA6	no	1B	19:7619892	C>G	nonsynonymous SNV	no	no	no
PNPLA6	no	8D	19:7619887 1:19657836	C>A	nonsynonymous SNV	no	no	no
KCNT2	no	1B	6	C>G	upstream	no	no	no
KCNT2	no	2H	1:19619692 1	A>G	UTR3	no	no	yes
ODF2L	no	1B	1:86861909 20:2342080	A>C	UTR5	no	no	no
CSTL1	no	1B	7	G>A	UTR5	no	no	no
SALL4	yes	1B	20:5040808 2	T>C	nonsynonymous SNV	no	no	no
ARSA	no	1B	22:5106666 9	T>G	upstream	no	no	yes
AGXT	no	1B	2:24181830 4	A>G	UTR3	no	no	no
TPRG1-AS1	no	1B	3:18866589 0	T>A	upstream	no	no	no
SEC22C	no	1B	3:42590686 4:18436577	G>A	UTR3	no	yes	yes
CDKN2AIP	no	1B	4	C>CCGG	UTR5	no	no	no
CDKN2AIP	no	9C	4:18436828 5	T>G	nonsynonymous SNV	no	no	no
LINC02265	no	1B	4:40309052 5:12593070	A>G	upstream	no	no	no
ALDH7A1	no	1B	6	C>A	nonsynonymous SNV	no	no	no
NEDD9	no	1B	6:11184624 6:13018254	G>A	UTR3	no	no	no
TMEM244	no	1B	1	C>T	upstream	no	no	no
FBXL13	no	1B	7:10245364 7	T>C	UTR3	no	no	no
SEMA3A	no	1B	7:83588854	G>A	UTR3	no	no	no
RP1L1	no	1B	8:10468163	C>T	nonsynonymous SNV	no	no	no
MMP16	no	1B	8:89053623	CTCTTGCTT GAATCTCAA GT>C	UTR3	no	no	no
ERCC6L2	no	1B	9:98729325	T>C	UTR3	no	no	no
ERCC6L2	no	6B	9:98735047	G>A	nonsynonymous SNV	no	no	no
ERCC6L2	no	6D	9:98735047 X:13415633	G>A	nonsynonymous SNV	no	no	no
RTL8B	no	1B	7	C>T	nonsynonymous SNV	no	no	no
AFF2	no	1B	X:14807528 9	C>A	UTR3	no	no	no
RHOBTB1	no	1C	10:6269598 6	T>A	UTR5	no	no	no
RHOBTB1	no	1G	10:6269598 6	T>A	UTR5	no	no	no
RHOBTB1	no	1H	10:6269598 6	T>A	UTR5	no	no	no
RHOBTB1	no	1I	10:6269598 6	T>A	UTR5	no	no	no
RHOBTB1	no	1J	10:6269598 6	T>A	UTR5	no	no	no
RHOBTB1	no	1K	10:6269598 6	T>A	UTR5	no	no	no
CYP26A1	no	1C	10:9483494 7	G>C	nonsynonymous SNV	no	no	no
FXVD6	no	1C	11:1177088 42	C>T	UTR3	no	no	no

			11:1177088					
FXVD6	no	1G	42	C>T	UTR3	no	no	no
			11:1177088					
FXVD6	no	1H	42	C>T	UTR3	no	no	no
			11:1177088					
FXVD6	no	1I	42	C>T	UTR3	no	no	no
			11:1177088					
FXVD6	no	1J	42	C>T	UTR3	no	no	no
			11:1177088					
FXVD6	no	1K	42	C>T	UTR3	no	no	no
			11:6229436		nonframeshift			
AHNAK	no	1C	0	ATCT>A	deletion	no	no	no
			11:6229436		nonframeshift			
AHNAK	no	1G	0	ATCT>A	deletion	no	no	no
			11:6229436		nonframeshift			
AHNAK	no	1H	0	ATCT>A	deletion	no	no	no
			11:6229436		nonframeshift			
AHNAK	no	1I	0	ATCT>A	deletion	no	no	no
			11:6229436		nonframeshift			
AHNAK	no	1J	0	ATCT>A	deletion	no	no	no
			11:6229436		nonframeshift			
AHNAK	no	1K	0	ATCT>A	deletion	no	no	no
			11:6726297		nonsynonymous			
PITPNM1	no	1C	1	T>C	SNV	no	no	no
			11:9458714		nonsynonymous			
AMOTL1	no	5C	6	G>T	SNV	no	no	yes
			12:1106559		nonsynonymous			
IFT81	no	1C	76	A>G	SNV	no	no	no
			12:1106559		nonsynonymous			
IFT81	no	1G	76	A>G	SNV	no	no	no
			12:1106559		nonsynonymous			
IFT81	no	1H	76	A>G	SNV	no	no	no
			12:1106559		nonsynonymous			
IFT81	no	1I	76	A>G	SNV	no	no	no
			12:1106559		nonsynonymous			
IFT81	no	1J	76	A>G	SNV	no	no	no
			12:1106559		nonsynonymous			
IFT81	no	1K	76	A>G	SNV	no	no	no
			12:2167020					
GOLT1B	no	1C	1	A>T	UTR3	no	no	yes
			12:2167020					
GOLT1B	no	1G	1	A>T	UTR3	no	no	yes
			12:2167020					
GOLT1B	no	1H	1	A>T	UTR3	no	no	yes
			12:2167020					
GOLT1B	no	1I	1	A>T	UTR3	no	no	yes
			12:2167020					
GOLT1B	no	1J	1	A>T	UTR3	no	no	yes
			12:2167020					
GOLT1B	no	1K	1	A>T	UTR3	no	no	yes
			12:4892182		nonsynonymous			
OR8S1	no	1C	2	C>T	SNV	no	no	no
			12:4892182		nonsynonymous			
OR8S1	no	1G	2	C>T	SNV	no	no	no
			12:4892182		nonsynonymous			
OR8S1	no	1H	2	C>T	SNV	no	no	no
			12:4892182		nonsynonymous			
OR8S1	no	1I	2	C>T	SNV	no	no	no
			12:4892182		nonsynonymous			
OR8S1	no	1J	2	C>T	SNV	no	no	no
			12:4892182		nonsynonymous			
OR8S1	no	1K	2	C>T	SNV	no	no	no
			12:5742405		nonsynonymous			
MYO1A	no	1C	8	T>A	SNV	no	no	no
			14:1013511					
RTL1	no	1C	29	G>A	UTR5	no	yes	yes
			14:5548026		nonsynonymous			
WDHD1	no	1C	8	C>T	SNV	no	no	no
			14:5548026		nonsynonymous			
WDHD1	no	1G	8	C>T	SNV	no	no	no

WDHD1	no	1H	14:5548026 8	C>T	nonsynonymous SNV	no	no	no
WDHD1	no	1I	14:5548026 8	C>T	nonsynonymous SNV	no	no	no
WDHD1	no	1J	14:5548026 8	C>T	nonsynonymous SNV	no	no	no
WDHD1	no	1K	14:5548026 8	C>T	nonsynonymous SNV	no	no	no
RSL1D1	no	1C	16:1194534 7	G>T	stopgain	no	yes	yes
RSL1D1	no	1G	16:1194534 7	G>T	stopgain	no	yes	yes
RSL1D1	no	1H	16:1194534 7	G>T	stopgain	no	yes	yes
RSL1D1	no	1I	16:1194534 7	G>T	stopgain	no	yes	yes
RSL1D1	no	1J	16:1194534 7	G>T	stopgain	no	yes	yes
RSL1D1	no	1K	16:1194534 7	G>T	stopgain	no	yes	yes
TIGD7	no	1C	16:3355280 17:7838956	C>G	UTR5 nonsynonymous SNV	no	no	no
ENDOV	no	1C	17:7838956 6	G>A	nonsynonymous SNV	no	no	no
ENDOV	no	1G	17:7838956 6	G>A	nonsynonymous SNV	no	no	no
ENDOV	no	1H	17:7838956 6	G>A	nonsynonymous SNV	no	no	no
ENDOV	no	1I	17:7838956 6	G>A	nonsynonymous SNV	no	no	no
ENDOV	no	1J	17:7838956 6	G>A	nonsynonymous SNV	no	no	no
ENDOV	no	1K	17:7838956 6	G>A	nonsynonymous SNV	no	no	no
CDH7	no	1C	18:6352992 2	A>G	nonsynonymous SNV	no	no	no
ZNF580	no	1C	19:5615142 8	T>A	upstream	no	no	no
ZNF580	no	1G	19:5615142 8	T>A	upstream	no	no	no
ZNF580	no	1H	19:5615142 8	T>A	upstream	no	no	no
ZNF580	no	1I	19:5615142 8	T>A	upstream	no	no	no
ZNF580	no	1J	19:5615142 8	T>A	upstream	no	no	no
ZNF580	no	1K	19:5615142 8	T>A	upstream	no	no	no
ZNF837	no	1C	19:5889272 3	T>C	upstream	no	no	no
ZNF837	no	1G	19:5889272 3	T>C	upstream	no	no	no
ZNF837	no	1H	19:5889272 3	T>C	upstream	no	no	no
ZNF837	no	1I	19:5889272 3	T>C	upstream	no	no	no
ZNF837	no	1J	19:5889272 3	T>C	upstream	no	no	no
ZNF837	no	1K	19:5889272 3	T>C	upstream	no	no	no
RASSF5	no	1C	1:20676126 6	A>G	UTR3	no	no	no
LRRTM4	no	1C	2:77744941	C>T	UTR3	no	no	no
LRRTM4	no	1G	2:77744941	C>T	UTR3	no	no	no
LRRTM4	no	1H	2:77744941	C>T	UTR3	no	no	no
LRRTM4	no	1I	2:77744941	C>T	UTR3	no	no	no
LRRTM4	no	1J	2:77744941	C>T	UTR3	no	no	no
LRRTM4	no	1K	2:77744941	C>T	UTR3	no	no	no

ZBTB20	no	1C	3:11486665 8	C>T	upstream	no	no	yes
H2AFZ	no	1C	4:10086955 1	G>C	UTR3	no	no	no
HHIP	no	1C	4:14565915 3	T>G	UTR3	no	no	no
HHIP	no	1G	4:14565915 3	T>G	UTR3	no	no	no
HHIP	no	1H	4:14565915 3	T>G	UTR3	no	no	no
HHIP	no	1I	4:14565915 3	T>G	UTR3	no	no	no
HHIP	no	1J	4:14565915 3	T>G	UTR3	no	no	no
HHIP	no	1K	4:14565915 3	T>G	UTR3	no	no	no
NPM1	yes	1C	5:17081484 5	C>T	UTR5	no	yes	yes
NPM1	yes	1G	5:17081484 5	C>T	UTR5	no	yes	yes
NPM1	yes	1H	5:17081484 5	C>T	UTR5	no	yes	yes
NPM1	yes	1I	5:17081484 5	C>T	UTR5	no	yes	yes
NPM1	yes	1J	5:17081484 5	C>T	UTR5	no	yes	yes
NPM1	yes	1K	5:17081484 5	C>T	UTR5	no	yes	yes
RWDD1	no	1C	6:11689231 0	G>A	upstream	no	no	yes
RWDD1	no	1G	6:11689231 0	G>A	upstream	no	no	yes
RWDD1	no	1H	6:11689231 0	G>A	upstream	no	no	yes
RWDD1	no	1I	6:11689231 0	G>A	upstream	no	no	yes
RWDD1	no	1J	6:11689231 0	G>A	upstream	no	no	yes
RWDD1	no	1K	6:11689231 0	G>A	upstream	no	no	yes
BCLAF1	yes	1C	6:13658111 5	G>C	UTR3	no	no	no
BCLAF1	yes	1G	6:13658111 5	G>C	UTR3	no	no	no
BCLAF1	yes	1H	6:13658111 5	G>C	UTR3	no	no	no
BCLAF1	yes	1I	6:13658111 5	G>C	UTR3	no	no	no
BCLAF1	yes	1J	6:13658111 5	G>C	UTR3	no	no	no
BCLAF1	yes	1K	6:13658111 5	G>C	UTR3	no	no	no
BCLAF1	yes	7D	6:13657904 9	G>T	UTR3	no	no	no
FIS1	no	1C	7:10088840 2	C>A	upstream	no	no	no
FIS1	no	1G	7:10088840 2	C>A	upstream	no	no	no
FIS1	no	1H	7:10088840 2	C>A	upstream	no	no	no
FIS1	no	1I	7:10088840 2	C>A	upstream	no	no	no
FIS1	no	1J	7:10088840 2	C>A	upstream	no	no	no
FIS1	no	1K	7:10088840 2	C>A	upstream	no	no	no
NOD1	no	1C	7:30498808	T>C	UTR5	no	no	no
NOD1	no	1G	7:30498808	T>C	UTR5	no	no	no
NOD1	no	1H	7:30498808	T>C	UTR5	no	no	no

NOD1	no	1I	7:30498808	T>C	UTR5	no	no	no
NOD1	no	1J	7:30498808	T>C	UTR5	no	no	no
NOD1	no	1K	7:30498808	T>C	UTR5	no	no	no
IKZF1	yes	1C	7:50468997	C>T	UTR3	no	yes	yes
IKZF1	yes	1G	7:50468997	C>T	UTR3	no	yes	yes
IKZF1	yes	1H	7:50468997	C>T	UTR3	no	yes	yes
IKZF1	yes	1I	7:50468997	C>T	UTR3	no	yes	yes
IKZF1	yes	1J	7:50468997	C>T	UTR3	no	yes	yes
IKZF1	yes	1K	7:50468997	C>T	UTR3	no	yes	yes
IKZF1	yes	10E	7:50470975	G>C	UTR3	no	no	no
LOC101927446	no	1C	7:89963770	G>A	upstream	no	no	no
C8orf34	no	1C	8:69449742	G>A	UTR3	no	no	no
C8orf34	no	1G	8:69449742	G>A	UTR3	no	no	no
C8orf34	no	1H	8:69449742	G>A	UTR3	no	no	no
C8orf34	no	1I	8:69449742	G>A	UTR3	no	no	no
C8orf34	no	1J	8:69449742	G>A	UTR3	no	no	no
C8orf34	no	1K	8:69449742	G>A	UTR3	no	no	no
TMEM38B	no	1C	9:108483966	G>A	nonsynonymous SNV	no	no	yes
TMEM38B	no	1G	9:108483966	G>A	nonsynonymous SNV	no	no	yes
TMEM38B	no	1H	9:108483966	G>A	nonsynonymous SNV	no	no	yes
TMEM38B	no	1I	9:108483966	G>A	nonsynonymous SNV	no	no	yes
TMEM38B	no	1J	9:108483966	G>A	nonsynonymous SNV	no	no	yes
TMEM38B	no	1K	9:108483966	G>A	nonsynonymous SNV	no	no	yes
MPDZ	no	8D	9:131196199:13975510	A>G	nonsynonymous SNV	no	no	no
MAMDC4	no	1C	9:139755107	G>T	UTR3	no	no	no
MAMDC4	no	1G	9:139755107	G>T	UTR3	no	no	no
MAMDC4	no	1H	9:139755107	G>T	UTR3	no	no	no
MAMDC4	no	1I	9:139755107	G>T	UTR3	no	no	no
MAMDC4	no	1J	9:139755107	G>T	UTR3	no	no	no
MAMDC4	no	1K	9:139755107	G>T	UTR3	no	no	no
SLC25A53	no	1C	X:103402062	GTTC>G	upstream	no	no	no
SLC25A53	no	1G	X:103402062	GTTC>G	upstream	no	no	no
SLC25A53	no	1H	X:103402062	GTTC>G	upstream	no	no	no
SLC25A53	no	1I	X:103402062	GTTC>G	upstream	no	no	no
SLC25A53	no	1J	X:103402062	GTTC>G	upstream	no	no	no
SLC25A53	no	1K	X:103402062	GTTC>G	upstream	no	no	no
KIAA1210	no	1C	X:118239092	G>A	nonsynonymous SNV	no	yes	yes
KIAA1210	no	1G	X:118239092	G>A	nonsynonymous SNV	no	yes	yes

KIAA1210	no	1H	X:11823909 2	G>A	nonsynonymous SNV	no	yes	yes
KIAA1210	no	1I	X:11823909 2	G>A	nonsynonymous SNV	no	yes	yes
KIAA1210	no	1J	X:11823909 2	G>A	nonsynonymous SNV	no	yes	yes
KIAA1210	no	1K	X:11823909 2	G>A	nonsynonymous SNV	no	yes	yes
KIAA1210	no	5B	X:11822113 3	C>A	nonsynonymous SNV	no	no	no
CUL4B	no	1C	X:11970843 3	C>A	nonsynonymous SNV	no	no	no
CUL4B	no	1G	X:11970843 3	C>A	nonsynonymous SNV	no	no	no
CUL4B	no	1H	X:11970843 3	C>A	nonsynonymous SNV	no	no	no
CUL4B	no	1I	X:11970843 3	C>A	nonsynonymous SNV	no	no	no
CUL4B	no	1J	X:11970843 3	C>A	nonsynonymous SNV	no	no	no
CUL4B	no	1K	X:11970843 3	C>A	nonsynonymous SNV	no	no	no
OR13H1	no	1C	X:13067748 5	C>T	upstream	no	no	yes
OR13H1	no	1G	X:13067748 5	C>T	upstream	no	no	yes
OR13H1	no	1H	X:13067748 5	C>T	upstream	no	no	yes
OR13H1	no	1I	X:13067748 5	C>T	upstream	no	no	yes
OR13H1	no	1J	X:13067748 5	C>T	upstream	no	no	yes
OR13H1	no	1K	X:13067748 5	C>T	upstream	no	no	yes
F9	no	1C	X:13864535 2	C>G	UTR3	no	no	no
RPL10	yes	1C	X:15362674 7	C>T	UTR5	no	yes	yes
RPL10	yes	1G	X:15362674 7	C>T	UTR5	no	yes	yes
RPL10	yes	1H	X:15362674 7	C>T	UTR5	no	yes	yes
RPL10	yes	1I	X:15362674 7	C>T	UTR5	no	yes	yes
RPL10	yes	1J	X:15362674 7	C>T	UTR5	no	yes	yes
RPL10	yes	1K	X:15362674 7	C>T	UTR5	no	yes	yes
PBDC1	no	1C	X:75392060	C>T	upstream	no	no	no
PBDC1	no	11D	X:75392040	C>T	upstream	no	no	no
FAM9A	no	1C	X:8758960	A>T	UTR3	no	no	no
FAM9A	no	7B	X:8766470	T>C	nonsynonymous SNV	no	yes	yes
FAM9A	no	7C	X:8766470 10:1160550	T>C	nonsynonymous SNV	no	yes	yes
AFAP1L2	no	1D	10:3741392 27	T>A	UTR3	no	no	no
ANKRD30A	no	1D	11:6923968 4	C>A	upstream	no	no	no
LOC102724 265	no	1D	13:5290755 3	G>A	upstream	no	no	no
LINC02333	no	1D	14:2058655 7	C>T	upstream frameshift	no	no	yes
OR4K17	no	1D	14:9341835 2	G>GA	insertion	no	no	no
ITPK1	no	1D	3	C>A	stopgain	no	no	no
WSCD1	no	1D	17:5973114	TGTGTGTGC >T	upstream	no	no	yes

WSCD1	no	6C	17:5973110	TGTGTGTGT GTGC>T	upstream nonsynonymous	no	no	yes
TNK1	no	1D	17:7290416 17:7439221	G>T	SNV nonsynonymous	no	yes	yes
UBE2O	no	1D	2 19:4953915	G>C	SNV nonsynonymous	no	no	no
CGB1	no	1D	4	G>A	SNV nonsynonymous	no	no	no
ERICH3	no	1D	1:75036856	A>G	SNV	no	no	yes
C20orf202	no	1D	20:1183118 20:6051293	C>G	upstream	no	no	no
CDH4	no	1D	5 20:6051467	G>A	UTR3	no	yes	yes
CDH4	no	8B	1 20:6148331	C>T	UTR3	no	no	no
TCFL5	no	1D	3 21:4279272	C>T	UTR3	no	no	no
MX1	no	1D	4 22:1912736	A>G	UTR5	no	no	no
DGCR14	no	1D	6 2:20068408	A>T	splicing	no	no	no
FTCDNL1	no	1D	4	C>G	UTR3 nonsynonymous	no	no	no
APOB	no	1D	2:21227975	T>C CAGCTGGAG CTCTCAAAC AAAAGACAT TTGTTATTTT GGAAAGAA GA>C	SNV	no	no	no
MME	no	1D	3:15490135 6 3:16754082	GA>C	UTR3 nonsynonymous	no	no	no
SERPINI1	no	1D	2	A>G	SNV nonsynonymous	no	no	no
CCR3	no	1D	3:46307260	C>T	SNV	no	no	yes
RYBP	no	1D	3:72427232 4:13512193	T>C	UTR3 nonsynonymous	no	no	yes
PABPC4L	no	1D	0 4:13511934	C>T	SNV	no	no	yes
PABPC4L	no	2F	5	C>A	UTR3 nonsynonymous	no	no	no
C4orf19	no	1D	4:37592434	A>G	SNV	no	no	no
PRR27	no	1D	4:71029544	T>C	UTR3	no	no	no
HERC3	no	1D	4:89577390 5:10014756	T>G	UTR3 nonsynonymous	no	no	no
ST8SIA4	no	1D	7 5:11225880	T>A	SNV	no	no	no
REEP5	no	1D	5 5:13076137	T>C	upstream	no	no	no
RAPGEF6	no	1D	1 6:13199900	T>C	UTR3	no	no	no
ENPP3	no	1D	6 6:15752791	A>T	splicing nonsynonymous	no	no	no
ARID1B	yes	1D	9 6:15715047	T>G C>CAGGCC	SNV frameshift	no	no	no
ARID1B	yes	5C	6 6:15752943	A	insertion	no	no	no
ARID1B	yes	10A	8 6:15752943	G>A	UTR3	no	no	no
ARID1B	yes	10D	8 6:16021147	G>A	UTR3	no	no	no
MRPL18	no	1D	9 7:14811282	G>A	UTR5	no	no	no
CNTNAP2	yes	1D	8 7:14811328	G>A	UTR3	no	no	no
CNTNAP2	yes	2E	4	C>T	UTR3	no	no	no
GTF2I	no	1D	7:74071695	C>A	upstream	no	no	no

LMTK2	no	1D	7:97822878	C>G	nonsynonymous SNV	no	no	no
DDHD2	no	1D	8:38118354 X:13529948	T>G	UTR3	no	no	no
MAP7D3	no	1D	1	T>A	UTR3	no	no	no
CFAP47	no	1D	X:36011367	T>C	nonsynonymous SNV	no	yes	yes
EDA2R	no	1D	X:65816933	C>T	UTR3	no	no	no
ZNF711	no	1D	X:84526794 10:1180559	A>T	nonsynonymous SNV	no	no	no
ECHDC3	no	1E	7	T>C	UTR3	no	no	no
CXCL12	no	1E	10:4486839 12:7218482	C>T	nonsynonymous SNV	no	no	no
RAB21	no	1E	4	T>A	UTR3	no	no	no
TRAPPC6B	no	1E	14:3961941 2	T>C	UTR3	no	yes	yes
MYO1C	no	1E	17:1396758	G>A	upstream	no	yes	yes
TPGS1	no	1E	19:507092 20:5609952	G>C	upstream	no	no	yes
CTCFL	no	1E	2	C>T	UTR5	no	no	yes
CEP68	no	1E	2:65282841	C>T	upstream	no	no	no
ROBO1	no	1E	3:79067925	C>T	UTR5	no	no	no
ROBO1	no	5F	3:79817793 7:13477930	C>A	upstream	no	no	no
C7orf49	no	1E	7	T>C	UTR3	no	yes	yes
EIF4H	no	1E	7:73588649 X:10080895	G>T	upstream nonsynonymous SNV	no	no	yes
ARMCX1	no	1E	0	A>G	SNV	no	no	no
MTMR8	no	1E	X:63616084	A>G	upstream nonsynonymous SNV	no	no	yes
ATRX	yes	1E	X:76931790 10:9909465	T>C	SNV	no	no	no
FRAT2	no	1F	1	C>T	upstream	no	no	no
FRAT2	no	7B	10:9909532 13:2487703	C>G	upstream	no	no	no
SPATA13	no	1F	0	A>T	UTR3	no	no	no
RBM23	no	1F	14:2337052 7	A>C	UTR3	no	no	no
VCPKMT	no	1F	14:5057608 0	A>C	UTR3	no	no	no
VPS13C	no	1F	15:6235258 0	G>A	UTR5	no	no	no
VPS13C	no	9B	15:6215940 6	T>A	UTR3	no	no	no
CCDC113	no	1F	16:5828367 1	T>C	upstream	no	no	no
FAM157B;F	no	1F	16:9016822 8	C>A	upstream	no	no	yes
AM157C	no	1F	17:4590890 3	C>T	UTR5	no	no	yes
MRPL10	no	1F	17:7947915 3	T>C	nonsynonymous SNV	no	no	no
ACTG1	no	1F	19:1065566 5	T>C	nonsynonymous SNV	no	no	no
ATG4D	no	1F	19:2257578 4	C>A	splicing nonsynonymous SNV	no	yes	yes
ZNF98	no	2E	19:9048227	A>G	SNV	no	yes	yes
MUC16	yes	2C	19:9050021	G>A	nonsynonymous SNV	no	yes	yes
MUC16	yes	2K	19:9064422	C>T	stopgain	no	no	no

MUC16	yes	10A	19:9069837	G>A	nonsynonymous SNV	no	yes	yes
MUC16	yes	10D	19:9069837 1:20980142	G>A	nonsynonymous SNV	no	yes	yes
LAMB3	no	1F	4	G>A	nonsynonymous SNV	no	yes	yes
GID8	no	1F	20:6157778 6	T>C	UTR3	no	no	no
TPO	no	1F	2:1491688 3:12114019	T>G	nonsynonymous SNV	no	no	no
STXBP5L	no	1F	1	T>C	UTR3	no	no	no
STXBP5L	no	6C	3:12113962 9	A>G	UTR3	no	no	no
STXBP5L	no	7B	3:12062762 7	C>T	UTR5	no	no	no
SERPINI2	no	1F	3:16715985 0	A>G	UTR3	no	no	no
CAMP	no	1F	3:48266127	A>G	nonsynonymous SNV	no	yes	yes
MST1	no	1F	3:49723397 5:13490709	T>A	splicing	no	no	no
CXCL14	no	1F	5	C>T	UTR3	no	yes	yes
LOC389332	no	1F	5:13552916 3	G>T	upstream	no	no	no
GABRB2	no	1F	5:16072057 0	T>C	UTR3	no	no	no
JARID2	no	1F	6:15501492	G>T	nonsynonymous SNV	no	no	no
JARID2	no	8C	6:15501437	C>T	nonsynonymous SNV	no	no	no
JARID2	no	11B	6:15487724	C>G	nonsynonymous SNV	no	no	no
JARID2	no	11D	6:15487724 6:16010304	C>G	nonsynonymous SNV	no	no	no
SOD2	no	1F	7	G>C	UTR3	no	no	no
ZNF462	no	1F	9:10962505 3	T>C	upstream	no	no	no
AIF1L	no	1F	9:13397221 9	G>A	nonsynonymous SNV	no	yes	yes
CAMSAP1	no	1F	9:13871298 7	C>T	nonsynonymous SNV	no	no	no
IL13RA2	no	1F	X:11423854 7	C>T	UTR3	no	no	no
TEX13C	no	1F	X:12445669 3	G>T	nonsynonymous SNV	no	no	no
EIF1AX	yes	1F	X:20156717	T>A	nonsynonymous SNV	no	no	no
FAM120C	no	1F	X:54095720 11:3182279	A>T	UTR3	no	no	no
PAX6	no	1G	2	T>C	UTR5	no	no	no
PAX6	no	1H	11:3182279 2	T>C	UTR5	no	no	no
CDKN1B	yes	1G	12:1287104 2	GA>G	frameshift deletion	no	no	no
CDKN1B	yes	1H	12:1287104 2	GA>G	frameshift deletion	no	no	no
CDKN1B	yes	2E	12:1287108 6	G>T	stopgain	no	no	no
CDKN1B	yes	2G	12:1287115 6	C>CGTGT	frameshift insertion	no	no	no
CDKN1B	yes	2L	12:1287115 6	C>CGTGT	frameshift insertion	no	no	no
CDKN1B	yes	3B	12:1287095 3	G>A	stopgain	no	no	no
CDKN1B	yes	3C	12:1287188 0	AACAGCTCG >A	UTR3	no	no	no
CDKN1B	yes	4C	12:1287490 2	G>A	UTR3	no	no	no

CDKN1B	yes	7A	12:12871250	T>G	splicing frameshift	no	no	no
CDKN1B	yes	11A	12:12871052	G>GC	insertion nonsynonymous	no	no	yes
LDHB	no	1G	12:21788633	C>A	SNV nonsynonymous	no	no	no
LDHB	no	1H	12:21788633	C>A	SNV nonsynonymous	no	no	no
CHD4	yes	1G	12:66918770	A>C	SNV nonsynonymous	no	no	no
CHD4	yes	1H	12:66918770	A>C	SNV nonsynonymous	no	no	no
GPR12	no	1G	13:27331218	C>G	UTR3	no	no	no
GPR12	no	1H	13:27331218	C>G	UTR3	no	no	no
OR4N2	no	1G	14:20271954	G>T	UTR5	no	no	no
GOSR2	no	1G	17:45017830	CT>C	UTR3	no	no	no
GOSR2	no	1H	17:45017830	CT>C	UTR3	no	no	no
ST6GAL2	no	1G	2:107420274	T>A	UTR3	no	no	no
ST6GAL2	no	1H	2:107420274	T>A	UTR3	no	no	no
KCNG3	no	1G	2:42721086	G>A	UTR5	no	yes	yes
KCNG3	no	1H	2:42721086	G>A	UTR5	no	yes	yes
VAX2	no	1G	2:71127387	C>A	upstream	no	no	no
VAX2	no	1H	2:71127387	C>A	upstream	no	no	no
ARL6	no	1G	3:97517558	G>A	UTR3	no	yes	yes
ARL6	no	1H	3:97517558	G>A	UTR3	no	yes	yes
GABRA2	no	1G	4:46248507	T>A	UTR3	no	no	no
GABRA2	no	1H	4:46248507	T>A	UTR3	no	no	no
ABLM3	no	1G	5:148639801	T>C	UTR3	no	no	yes
ABLM3	no	1H	5:148639801	T>C	UTR3	no	no	yes
TSPO2	no	1G	6:41009747	G>C	upstream	no	no	no
TSPO2	no	1H	6:41009747	G>C	upstream	no	no	no
TSPO2	no	1I	6:41009747	G>C	upstream	no	no	no
TSPO2	no	1J	6:41009747	G>C	upstream	no	no	no
TSPO2	no	1K	6:41009747	G>C	upstream	no	no	no
ANKRD6	no	1G	6:90341754	T>C	UTR3	no	yes	yes
ANKRD6	no	1H	6:90341754	T>C	UTR3	no	yes	yes
ADCY1	no	1G	7:45756223	G>A	UTR3	no	yes	yes
ADCY1	no	1H	7:45756223	G>A	UTR3	no	yes	yes
OBP2A	no	1G	9:138441734	C>T	nonsynonymous SNV	no	no	no
OBP2A	no	1H	9:138441734	C>T	nonsynonymous SNV	no	no	no
EFNB1	no	1G	X:68060657	T>G	UTR3	no	no	no
TBL1X	no	1G	X:9685550	G>T	UTR3	no	no	no
TBL1X	no	1H	X:9685550	G>T	UTR3	no	no	no
PARP16	no	1H	15:65579481	G>A	upstream nonsynonymous	no	no	no
SPOCD1	no	1H	1:32258901	C>T	SNV	no	no	no

C2orf54	no	1H	2:24183589 7	C>T	upstream	no	no	no
SMIM19	no	1H	8:42396871 13:2528393	T>G	UTR5	no	yes	yes
ATP12A	no	1I	4	A>T	nonsynonymous SNV	no	no	no
TOGARAM1	no	3B	14:4551386 6	G>A	nonsynonymous SNV	no	yes	yes
CPSF2;NDU FB1	no	1I	14:9258827 6	G>A	upstream	no	no	no
CHEK2P2	no	1I	15:2048787 5	C>A	upstream	no	no	no
ANKFY1	no	1I	17:4167339 19:1321002	CA>C	upstream	no	no	no
LYL1	yes	1I	7	G>A	UTR3	no	no	no
RINL	no	1I	19:3936160 0	A>G	nonsynonymous SNV	no	no	no
RINL	no	1J	19:3936160 0	A>G	nonsynonymous SNV	no	no	no
RINL	no	1K	19:3936160 0	A>G	nonsynonymous SNV	no	no	no
LILRB4	no	1I	19:5517363 3	A>T	upstream	no	no	no
GPR39	no	1I	2:13317370 2	C>T	upstream	no	no	no
GPR39	no	1J	2:13317370 2	C>T	upstream	no	no	no
GPR39	no	1K	2:13317370 2	C>T	upstream	no	no	no
LTBP1	no	1I	2:33622280	G>C	nonsynonymous SNV	no	no	no
LTBP1	no	2G	2:33335783	G>A	nonsynonymous SNV	no	no	no
GALNT15	no	1I	3:16216479	T>C	UTR5	no	no	no
STT3B	no	1I	3:31661265 6:11169751	A>G	nonsynonymous SNV	no	no	no
REV3L	no	1I	3	C>G	nonsynonymous SNV	no	no	no
HIST1H1C	no	1I	6:26057056	G>C	upstream	no	no	no
UHRF1BP1	no	1I	6:34843958	T>C	UTR3	no	no	no
TMEM217	no	1I	6:37225443	A>G	UTR5	no	yes	yes
TMEM217	no	1J	6:37225443	A>G	UTR5	no	yes	yes
TMEM217	no	1K	6:37225443	A>G	UTR5	no	yes	yes
RBM48	no	1I	7:92169033 9:11816468	A>G	UTR3	no	no	no
DEC1	no	1I	6	A>G	UTR3	no	no	no
DOCK11	no	1I	X:11762955 3	G>A	upstream	no	no	no
PNMA5	no	1I	X:15216325 7	G>C	upstream	no	yes	yes
VWA5A	no	1J	11:1240159 78	C>T	nonsynonymous SNV	no	no	no
VWA5A	no	1K	11:1240159 78	C>T	nonsynonymous SNV	no	no	no
LINC00445	no	1J	13:3627136 1	G>A	upstream	no	no	no
LINC00445	no	1K	13:3627136 1	G>A	upstream	no	no	no
LINC00433	no	1J	13:8919209 0	A>C	upstream	no	no	no
LINC00433	no	1K	13:8919209 0	A>C	upstream	no	no	no
AP4E1	no	1J	15:5129680 0	G>T	UTR3	no	no	no
AP4E1	no	1K	15:5129680 0	G>T	UTR3	no	no	no

POLR3E	no	1J	16:2233728 0	A>G	nonsynonymous SNV	no	no	no
POLR3E	no	1K	16:2233728 0	A>G	nonsynonymous SNV	no	no	no
ARRB2	no	1J	17:4613700	T>G	upstream	no	no	no
LOC105372 028	no	1J	18:2230563 9	T>G	upstream	no	no	no
LOC105372 028	no	1K	18:2230563 9	T>G	upstream	no	no	no
ACP5	no	1J	19:1168887 3	G>A	UTR5	no	no	no
ACP5	no	1K	19:1168887 3	G>A	UTR5	no	no	no
ETHE1	no	1J	19:4403075 7	G>A	nonsynonymous SNV	no	yes	yes
NOSIP	no	1J	19:5008378 5	C>A	UTR5	no	no	no
NOSIP	no	1K	19:5008378 5	C>A	UTR5	no	no	no
SHANK1	no	1J	19:5116548 8	G>A	nonsynonymous SNV	no	yes	yes
FNDC10	no	1J	1:1534015	A>C	UTR3	no	no	no
FNDC10	no	1K	1:1534015	A>C	UTR3	no	no	no
TGFBR3	no	1J	1:92195494	T>C	nonsynonymous SNV	no	no	no
TGFBR3	no	1K	1:92195494 20:6180919	T>C	nonsynonymous SNV	no	no	no
MIR124-3	no	1J	20:6180895 2	A>T	upstream	no	no	no
MIR124-3	no	2B	20:6180895 2	AACG>A	upstream	no	yes	yes
MIR124-3	no	11A	20:6180895 2	AACG>A	upstream	no	yes	yes
MIR124-3	no	11D	20:6180895 2	AACG>A	upstream	no	yes	yes
CLIC6	no	1J	21:3604162 3	C>T	UTR5	no	no	no
CLIC6	no	1K	21:3604162 3	C>T	UTR5	no	no	no
COL6A5	no	1J	3:13014523 8	G>A	nonsynonymous SNV	no	no	no
AMBN	no	1J	4:71472609	CCTT>C	UTR3	no	no	no
AMBN	no	1K	4:71472609 5:12703867	CCTT>C	UTR3	no	no	no
CCDC192	no	5C	6:16399570 3	C>T	upstream	no	no	no
QKI	yes	1J	6:16399570 3	G>T	UTR3	no	no	yes
QKI	yes	1K	6:16399570 3	G>T	UTR3	no	no	yes
AGO2	no	1J	8:14153413 5	TGTTAG>T	UTR3	no	no	no
AGO2	no	1K	8:14153413 5	TGTTAG>T	UTR3	no	no	no
LOC101929 294	no	1J	8:24406616	C>T	upstream	no	no	yes
LOC101929 294	no	1K	8:24406616	C>T	upstream	no	no	yes
XKR5	no	1J	8:6673385	C>A	nonsynonymous SNV	no	yes	yes
XKR5	no	1K	8:6673385	C>A	nonsynonymous SNV	no	yes	yes
ZFH4	no	1J	8:77763722	C>A	nonsynonymous SNV	no	yes	yes
ZFH4	no	1K	8:77763722	C>A	nonsynonymous SNV	no	yes	yes
CTPS2	no	1J	X:16707690	C>T	nonsynonymous SNV	no	no	no

CTPS2	no	1K	X:16707690	C>T	nonsynonymous SNV	no	no	no
ZXDB	no	1J	X:57619430	A>T	nonsynonymous SNV	no	no	no
ZXDB	no	1K	X:57619430	A>T	nonsynonymous SNV	no	no	no
XRCC6P5	no	1J	X:99195180	G>T	upstream	no	no	no
XRCC6P5	no	1K	X:99195180 13:2129657	G>T	upstream	no	no	no
IL17D	no	1K	9 18:1410410	A>C	UTR3	no	no	no
ZNF519	no	1K	8 10:1213402	C>T	UTR3	no	no	yes
TIAL1	no	2A	08 11:2658716	G>C	UTR5	no	no	no
MUC15	no	2A	0	G>T	nonsynonymous SNV	no	no	no
UNKL	no	2A	16:1465250	T>A	upstream	no	no	no
UNKL	no	9B	16:1413700 17:7548688	A>G	UTR3	no	no	yes
SEPT9	yes	2A	0 1:10978039	T>G	nonsynonymous SNV	no	no	no
SARS	no	2A	2 1:10978075	T>C	nonsynonymous SNV	no	no	no
SARS	no	10C	0 1:14528235	C>G	UTR3	no	no	no
NOTCH2NL	no	2A	4 1:14528235	C>T	UTR3	no	yes	yes
NOTCH2NL	no	2C	4	C>T	UTR3	no	yes	yes
SDC3	no	2A	1:31342392	A>C	UTR3	no	no	yes
ERRFI1	no	2A	1:8072296 20:1997747	G>T	UTR3	no	no	no
RIN2	no	2A	0	C>G	nonsynonymous SNV	no	no	no
SMIM11A;S MIM11B	no	2A	21:3574769 22:2106428	C>T	upstream	no	yes	yes
PI4KA	no	2A	1 22:4072548	A>G	nonsynonymous SNV	no	no	no
TNRC6B	no	2A	7 22:4068170	G>T	UTR3	no	no	no
TNRC6B	no	2D	5	AC>A	frameshift deletion	no	no	no
DES11;XRCC 6	no	2A	22:4201712 5	A>G	upstream	no	yes	yes
PDCD6IP	no	2A	3:33839955 4:18369607	A>T	upstream	no	no	no
TENM3	no	2A	7 4:18372225	T>A	stopgain	no	no	no
TENM3	no	2I	8 5:12836929	TA>T	UTR3	no	yes	yes
SLC27A6	no	2A	1 5:16127509	A>G	UTR3	no	no	no
GABRA1	no	2A	1	T>G	UTR5	no	no	no
SAMD3;TM EM200A	no	2A	6:13068739 0	C>T	upstream	no	no	no
FNDC1	no	2A	6:15965507 8	GGAC>G	nonframeshift deletion	no	yes	yes
COL12A1	no	2A	6:75794762	G>T	UTR3	no	yes	yes
POM121C	no	2A	7:75116387 10:1128361	A>T	upstream	no	no	no
ADRA2A	no	2B	67	G>T	upstream	no	no	no
PTDSS2	no	2B	11:447591 11:7001340	G>A	upstream	no	no	no
ANO1	no	2B	3 11:7789989	C>A	nonsynonymous SNV	no	no	yes
USP35	no	2B	2	A>G	UTR5	no	no	yes

KCNH5	no	2B	14:6324653 7	C>G	nonsynonymous SNV	no	no	no
FUT8	no	2B	14:6592239 6	A>C	UTR5	no	no	no
SLC12A1	no	2B	15:4859610 9	A>T	UTR3	no	no	no
SRR	no	2B	17:2218934 17:3376369	C>T	nonsynonymous SNV	no	no	no
SLFN13	no	2B	7	T>A	UTR3	no	no	no
ZNF521	yes	2B	18:2280638 0	C>G	nonsynonymous SNV	no	no	no
EMILIN2	no	2B	18:2885005	A>G	nonsynonymous SNV	no	no	no
TGIF1	no	2B	18:3412861	A>T	UTR5	no	no	no
C19orf25	no	2B	19:1473704 19:4332565	C>G	UTR3	no	no	no
PSG8-AS1	no	2B	0	A>C	upstream	no	no	no
TFAP2C	no	2B	20:5520363 9	C>A	upstream	no	no	no
CSRP3	no	2B	2:16653962 6	G>T	UTR3	no	no	no
CSRP3	no	8B	2:16654500 2	C>G	UTR3	no	no	no
VPS54	no	2B	2:64246608 3:12225271	G>A	upstream	no	yes	yes
PARP9	no	2B	6	A>C	UTR3	no	no	no
TMPPE	no	2B	3:33133528 4:11722047	A>C	UTR3	no	no	no
MIR1973	no	2B	3	C>T	upstream	no	no	no
CHIC2	yes	2B	4:54930707 5:11533683	G>A	UTR5	no	no	no
LVRN	no	2B	1	A>G	nonsynonymous SNV	no	no	no
ARHGEF28	no	2B	5:73237389	T>G	UTR3	no	no	no
EDIL3	no	2B	5:83237288	A>C	UTR3	no	no	no
CDSN	no	2B	6:31085255	G>A	nonsynonymous SNV	no	no	yes
BTBD9	no	2B	6:38224215	AT>A	frameshift deletion	no	no	no
TBCC	no	2B	6:42712434	A>C	UTR3	no	no	no
MIR4464	no	2B	6:91021639 7:10002731	G>T	upstream	no	no	no
MEPCE	no	2B	3	G>A	UTR5	no	no	no
GIMAP2	no	2B	7:15038240 6	T>C	upstream	no	no	no
RAB11FIP1	no	2B	8:37729370	C>G	nonsynonymous SNV	no	no	yes
SNTG1	no	2B	8:51707119 X:15369259	A>G	UTR3	no	no	yes
PLXNA3	no	2B	7	G>C	nonsynonymous SNV	no	no	no
MAGEB2	no	2B	X:30233137	G>T	upstream	no	no	no
DLG3	no	2B	X:69664979	C>T	UTR5	no	yes	yes
TSPY1	no	2B	Y:9304601	G>C	UTR5	no	no	yes
TSPY1	no	2E	Y:9304601	G>C	UTR5	no	no	yes
TSPY1	no	2G	Y:9304601	G>C	UTR5	no	no	yes
TSPY1	no	2K	Y:9304601	G>C	UTR5	no	no	yes
TSPY1	no	2L	Y:9304601 11:4524981	G>C	UTR5	no	no	yes
PRDM11	no	2C	3	ATAT>A	UTR3	no	no	yes

PRDM11	no	5B	11:4524981 1	TTATATTA>T	UTR3	no	no	yes
PRDM11	no	11A	11:4525662 6	T>C	UTR3	no	no	yes
AIM2	no	2C	1:15904722 9	A>C	upstream nonsynonymous SNV	no	no	no
CLCNKA	no	2C	1:16356226	T>G	SNV	no	no	no
TMEM120A	no	2C	7:75624201	G>C	upstream	no	no	no
IKBKB	yes	2C	8:42188861 10:1209253	A>T	UTR3	no	no	no
SFXN4	no	2D	43	G>T	upstream	no	no	no
FRMPD2B	no	2D	10:4886342 4	T>C	upstream	no	yes	yes
FRMPD2B	no	2G	10:4886342 4	T>C	upstream	no	yes	yes
FRMPD2B	no	2H	10:4886342 4	T>C	upstream	no	yes	yes
CRY2	no	2D	11:4586875 0	C>A	UTR5	no	no	no
JRKL- AS1;LOC105 369443	no	2D	11:9624098 9	G>A	upstream	no	no	no
SVOP	no	2D	12:1093058 97	C>A	UTR3	no	no	no
SLCO1C1	no	2D	12:2084837 3	G>A	UTR5	no	no	no
PDE1B	no	2D	12:5497248 1	G>A	UTR3	no	no	no
KNL1	no	2D	15:4088608 3	T>G	upstream	no	no	no
THSD4	no	2D	15:7143339 6	TA>T	upstream	no	no	no
SRL	no	2D	16:4257776 16:7566434	C>T	UTR5 nonsynonymous SNV	no	no	yes
KARS	no	2D	3	T>C	SNV	no	no	no
USP7	no	2D	16:8987061	G>T	UTR3	no	no	no
USP7	no	3C	16:9009378	A>C	nonsynonymous SNV	no	no	no
USP7	no	11B	16:8987889	G>C	nonsynonymous SNV	no	no	no
USP7	no	11D	16:8987889 17:4245374	G>C	nonsynonymous SNV	no	no	no
ITGA2B	no	2D	19:1790552 6	G>C	SNV	no	no	no
B3GNT3	no	2D	19:1790552 0	C>T	upstream	no	no	no
KLF16	no	2D	19:1852770 19:2354415	G>C	UTR3 nonsynonymous SNV	no	no	no
ZNF91	no	2D	19:5896280 7	T>C	SNV	no	yes	yes
ZNF324B	no	2D	7	C>A A>AGGGGAT	upstream	no	no	no
ACTL8	no	2D	1:18153414	G	UTR3	no	no	no
BSDC1	no	2D	1:32832824	T>C	UTR3 nonsynonymous SNV	no	no	no
OSBPL9	no	2D	1:52226381	A>G	SNV	no	no	no
PTBP2	no	2D	1:97278925	T>G	nonsynonymous SNV	no	no	no
PPM1F	no	2D	22:2230792 0	GATAGTCCA GCATC>G	upstream	no	no	no
PPM1F	no	10A	22:2227422 0	C>T	UTR3	no	yes	yes
PPM1F	no	10D	22:2227422 0	C>T	UTR3	no	yes	yes
NEFH	no	2D	22:2988707 2	CATTATGCTT GA>C	UTR3	no	no	no

NEFH	no	2H	22:2987609 3	G>T	upstream	no	no	no
MKRN2	no	2D	3:12624719 4:10073727	T>C	UTR3	no	no	no
DAPP1	no	2D	8 4:11065105	G>A	upstream	no	no	no
PLA2G12A	no	2D	3 4:12977694	G>C	UTR5 nonsynonymous	no	no	no
JADE1	no	2D	4 4:12979301	A>T	SNV nonsynonymous	no	no	no
JADE1	no	4A	1 5:13391513	A>T	SNV	no	no	no
JADE2	no	2D	3 5:14820672	T>G	UTR3 nonsynonymous	no	no	no
ADRB2	no	2D	9	T>C	SNV	no	no	no
HMGCS1	no	2D	5:43287942 6:15852017	G>C	UTR3	no	no	no
SYNJ2	no	2D	4	A>G	UTR3 nonsynonymous	no	no	no
EYS	no	2D	6:64488052 7:11465936	A>G	SNV	no	no	no
MDFIC	no	2D	8	T>C	UTR3	no	no	no
PURB	no	2D	7:44923430	T>C	UTR3	no	yes	yes
PURB	no	3B	7:44922005	C>A	UTR3	no	no	no
DEFA11P	no	2D	8:6887569 10:2650104	G>T	upstream	no	no	no
MYO3A	no	2E	7 10:2752946	G>A	UTR3	no	yes	yes
ACBD5	no	2E	8 10:4970163	G>A	UTR5	no	no	yes
ARHGAP22	no	2E	2 10:7557258	A>T	UTR5	no	no	no
CAMK2G	no	2E	8 10:8600410	T>C	UTR3	no	no	no
RGR	no	2E	1 10:8600454	C>A	upstream	no	no	no
RGR	no	2J	3 12:1075872	C>A	upstream	no	no	no
MAGOHB	no	2E	9 12:4544559	A>C	UTR3	no	no	no
DBX2	no	2E	0 15:7844129	T>C	upstream	no	yes	yes
IDH3A	no	2E	6 17:6035355	A>C	upstream	no	no	no
TBC1D3P2	no	2E	1 19:1793576	G>A	upstream	no	no	yes
JAK3	yes	2E	3 19:3555077	T>C	UTR3	no	no	no
HPN	no	2E	6 19:5484475	A>G	splicing	no	no	no
LILRA4	no	2E	0 19:5484871	C>A	UTR3 nonsynonymous	no	no	no
LILRA4	no	2F	2 1:11430175	T>C	SNV	no	no	no
PHTF1	no	2E	3	C>T	UTR5 nonsynonymous	no	no	no
CYP4A11	no	2E	1:47400741 21:3193458	T>G	SNV	no	no	no
KRTAP19-7	no	2E	0 2:14514637	G>T	upstream	no	yes	yes
ZEB2	no	2E	1 2:14514664	T>A	UTR3	no	no	no
ZEB2	no	3A	6 2:17809524	C>A	UTR3	no	no	no
NFE2L2	yes	2E	4 2:22888413	A>G	UTR3 nonsynonymous	no	no	no
SPHKAP	no	2E	5	T>C	SNV	no	no	no

YIPF4	no	2E	2:32531543	A>T	UTR3	no	no	no
MTA3	no	2E	2:42983761 4:15665602	C>G	UTR3	no	no	no
GUCY1A3	no	2E	9 5:17860810	T>G	UTR3 nonsynonymous	no	no	no
ADAMTS2	no	2E	7 5:17856689	A>T	SNV nonsynonymous	no	no	no
ADAMTS2	no	2J	4 5:17855499	G>A	SNV nonsynonymous	no	no	no
ADAMTS2	no	3C	1 5:17858110	C>G	SNV nonsynonymous	no	no	no
ADAMTS2	no	5D	8 5:17858110	C>A	SNV nonsynonymous	no	no	no
ADAMTS2	no	5F	8	C>A	SNV	no	no	no
IL7R	yes	2E	5:35878869	C>T	UTR3 frameshift	no	yes	yes
IL7R	yes	8D	5:35874574	AC>A	deletion	no	no	no
TMEM270	no	2E	7:73275223	T>G	upstream	no	no	no
SNAI2	no	2E	8:49834530 10:1025888	C>G	upstream	no	no	no
PAX2	no	2F	80 10:1025059	T>G	UTR3	no	no	no
PAX2	no	8C	29 10:7679182	T>G	UTR5	no	no	no
KAT6B	yes	2F	3 11:1101022	A>G	UTR3	no	no	no
RDX	no	2F	20 11:7585371	A>C	UTR3	no	no	no
UVRAG	no	2F	5 12:1009669	A>T	UTR3	no	no	no
GAS2L3	no	2F	53 12:1015246	C>T	upstream	no	no	no
CLEC1B	no	2F	0 12:4933092	A>G	upstream	no	no	no
ARF3	no	2F	3 12:5749901	C>A	UTR3 nonsynonymous	no	yes	yes
STAT6	yes	2F	4 13:2923268	C>A	SNV	no	no	no
POMP	no	2F	0 14:8608972	A>C	upstream nonsynonymous	no	no	no
FLRT2	no	2F	4 14:8608855	C>G	SNV nonsynonymous	no	no	no
FLRT2	no	11B	2 14:8608855	C>T	SNV nonsynonymous	no	no	no
FLRT2	no	11D	2 14:9133486	C>T	SNV	no	no	no
RPS6KA5	no	2F	7 17:3835022	A>T	UTR3	no	no	no
RAPGEFL1	no	2F	1 17:3834891	C>T	UTR3 nonsynonymous	no	no	no
RAPGEFL1	no	9A	2 17:3893332	C>A	SNV nonsynonymous	no	yes	yes
KRT27	no	2F	1 1:16070896	G>A	SNV	no	no	no
SLAMF7	no	2F	3 1:16866570	CA>C	UTR5	no	no	no
DPT	no	2F	8 1:21480387	T>A	UTR3 nonsynonymous	no	no	no
CENPF	no	2F	7	G>C	SNV nonsynonymous	no	no	no
CNR2	no	2F	1:24201234	A>C	SNV nonsynonymous	no	no	no
PLCH2	no	2F	1:2436504	G>A	SNV	no	yes	yes
LPAR3	no	2F	1:85359119	C>T	upstream frameshift	no	no	no
ZNF326	no	2F	1:90492993	TG>T	deletion	no	no	no
MTF2	no	2F	1:93603433	T>G	UTR3	no	no	no

ADAMTS5	no	11A	21:2829171 0	C>T	UTR3	no	no	no
MARCH4	no	2F	2:21712276 1	A>T	UTR3	no	no	no
MARCH4	no	7A	2:21712351 9	G>A	UTR3	no	no	no
MARCH4	no	7D	2:21712351 9	G>A	UTR3	no	no	no
MARCH4	no	8C	2:21723503 2	A>C	UTR5	no	no	no
GAL3ST2	no	2F	2:24271602 4	G>T	upstream	no	yes	yes
BRK1	no	2F	3:10156742	A>G	upstream	no	no	no
ATP2B2	no	2F	3:10370017 3:15717747	C>A	UTR3	no	no	no
VEPH1	no	2F	4	C>T	UTR3	no	no	no
MAP3K13	yes	2F	3:18516560 0	C>A	nonsynonymous SNV	no	no	no
ZNF827	no	2F	4:14668175 8	A>T	UTR3	no	no	no
ASIC5	no	2F	4:15678735 4	C>G	nonsynonymous SNV	no	no	no
LINC01094	no	2F	4:79566835 5:11706538	C>A	upstream	no	no	no
LINC02147	no	2F	3	T>A	upstream	no	no	no
ARHGEF37	no	2F	5:14896038 7	A>G	upstream	no	no	no
MYCT1	no	2F	6:15301895 8	C>A	upstream	no	no	no
MBOAT1	no	2F	6:20102190	C>CA	UTR3	no	no	no
VWA7	no	2F	6:31740760 7:15090886	T>C	nonsynonymous SNV	no	no	no
ABCF2	no	2F	8	G>C	UTR3	no	no	no
STK17A	no	2F	7:43664676	G>A	UTR3	no	no	no
AP5Z1	no	2F	7:4830451 9:11591239	C>T	stopgain	no	no	no
SLC31A2	no	2F	3	T>G	upstream	no	no	no
FRMPD4	no	2F	X:12156319 X:15525808	G>C	upstream	no	yes	yes
DDX11L16	no	2F	2	C>G	upstream	no	yes	yes
DDX11L16	no	3D	X:15525810 3	C>T	upstream	no	no	yes
ADAM8	no	2G	10:1350814 46	C>T	nonsynonymous SNV	no	no	no
ADAM8	no	2L	10:1350814 46	C>T	nonsynonymous SNV	no	no	no
USP54	no	2G	10:7527640 9	G>A	nonsynonymous SNV	no	yes	yes
USP54	no	2L	10:7527640 9	G>A	nonsynonymous SNV	no	yes	yes
CCSER2	no	2G	10:8613207 0	T>C	nonsynonymous SNV	no	yes	yes
PI4K2A	no	2G	10:9940039 1	C>T	upstream	no	no	yes
PI4K2A	no	2L	10:9940039 1	C>T	upstream	no	no	yes
TSG101	no	2G	11:1854850 5	AC>A	upstream	no	no	yes
TSG101	no	2L	11:1854850 5	AC>A	upstream	no	no	yes
PRPF19	no	2G	11:6067501 8	CACACACAC ACAT>C	upstream	no	no	yes
LOC338694	no	2G	11:6893990 8	T>C	upstream	no	no	no

MRE11	no	2G	11:9422694 9	G>T	UTR5	no	no	no
MRE11	no	11B	11:9420370 6	A>T	nonsynonymous SNV	no	no	no
MRE11	no	11D	11:9420370 6	A>T	nonsynonymous SNV	no	no	no
SENP1	no	2G	12:4843771 0	T>C	UTR3	no	no	no
SENP1	no	2L	12:4843771 0	T>C	UTR3	no	no	no
GAPDH	no	2G	12:6643709	C>T	UTR5	no	yes	yes
GAPDH	no	3A	12:6643709 13:1013602	C>T	UTR5	no	yes	yes
NALCN-AS1	no	2G	63	C>T	upstream	no	no	no
NALCN-AS1	no	2L	13:1013599 01	A>G	upstream nonsynonymous SNV	no	no	no
CARS2	no	2G	13:1113403 26	C>T	nonsynonymous SNV	no	no	no
CARS2	no	2L	13:1113403 26	C>T	nonsynonymous SNV	no	no	no
NBEA	yes	2G	13:3573411 1	A>G	nonsynonymous SNV	no	no	no
TNFSF11	no	2G	13:4314088 9	G>A	SNV	no	no	no
TNFSF11	no	2L	13:4314088 9	G>A	UTR5	no	no	no
UCHL3	no	2G	13:7612377 5	G>C	UTR5	no	no	no
UCHL3	no	2L	13:7612377 5	G>C	UTR5	no	no	no
GMFB	no	2G	14:5494362 9	G>GT	UTR3	no	no	no
IGDCC4	no	2G	15:6567550 5	C>A	UTR3	no	no	no
CX3CL1	no	2G	16:5741595 0	C>T	nonsynonymous SNV	no	yes	yes
CX3CL1	no	2L	16:5741595 0	C>T	nonsynonymous SNV	no	yes	yes
CSNK2A2	no	2G	16:5820162 7	T>C	nonsynonymous SNV	no	no	yes
PRMT7	no	2G	16:6835869 5	T>C	nonsynonymous SNV	no	no	no
PRMT7	no	2L	16:6835869 5	T>C	nonsynonymous SNV	no	no	no
HOXB9	no	2G	17:4669900 5	C>A	UTR3	no	no	no
FBXW9	no	2G	19:1280747 9	T>G	upstream	no	no	no
HOOK2	no	2G	19:1288712 3	C>T	upstream	no	yes	yes
HOOK2	no	2L	19:1288712 3	C>T	upstream	no	yes	yes
NWD1	no	2G	19:1692668 8	C>T	UTR3	no	no	no
NWD1	no	2L	19:1692668 8	C>T	UTR3	no	no	no
AP3D1	no	2G	19:2151456	C>A	UTR5	no	no	no
MAP2K2	yes	2G	19:4124199 19:4342043	A>AC	upstream nonsynonymous SNV	no	yes	yes
PSG6	no	2G	0	G>A	nonsynonymous SNV	no	yes	yes
PSG6	no	2L	19:4342043 0	G>A	nonsynonymous SNV	no	yes	yes
TNFSF14	no	2G	19:6665184	G>A	nonsynonymous SNV	no	no	no
TNFSF14	no	2L	19:6665184	G>A	nonsynonymous SNV	no	no	no
TNFSF14	no	10B	19:6671368	C>T	upstream	no	no	yes

TNFSF14	no	10C	19:6663258	G>T	UTR3	no	no	no
TNFSF14	no	10E	19:6671368	C>T	upstream	no	no	yes
			1:15901935		nonsynonymous			
IFI16	no	2G	5	C>T	SNV	no	no	no
			1:15901935		nonsynonymous			
IFI16	no	2L	5	C>T	SNV	no	no	no
MUL1	no	2G	1:20826700	A>C	UTR3	no	no	no
					nonsynonymous			
CAMTA1	yes	2G	1:7798097	A>G	SNV	no	no	no
					nonsynonymous			
CAMTA1	yes	2L	1:7798097	A>G	SNV	no	no	no
			20:3189475		nonsynonymous			
BPIFB1	no	2G	4	G>T	SNV	no	no	no
			20:4004424		nonsynonymous			
CHD6	no	2G	5	C>G	SNV	no	no	no
			20:4004424		nonsynonymous			
CHD6	no	2L	5	C>G	SNV	no	no	no
			20:4469138		nonsynonymous			
NCOA5	no	2G	1	G>A	SNV	no	no	no
			20:4469138		nonsynonymous			
NCOA5	no	2L	1	G>A	SNV	no	no	no
					nonsynonymous			
PLCB1	no	2G	20:8678276	G>A	SNV	no	no	no
					frameshift			
FAM49A	no	2G	2:16745334	A>AT	insertion	no	no	no
			3:10781073					
CD47	no	2G	6	C>G	upstream	no	no	no
			3:10781073					
CD47	no	2L	6	C>G	upstream	no	no	no
			3:10828823		nonsynonymous			
KIAA1524	no	2G	8	C>T	SNV	no	no	no
			3:11936099					
POPDC2	no	2G	5	C>A	UTR3	no	no	no
			3:13832976					
FAIM	no	2G	5	A>T	UTR5	no	no	no
					nonsynonymous			
WDR82	no	2G	3:52294493	G>C	SNV	no	no	no
			4:10887059		nonsynonymous			
CYP2U1	no	2G	7	G>T	SNV	no	no	no
STIM2	no	2G	4:27024624	C>G	UTR3	no	no	no
STIM2	no	2L	4:27024624	C>G	UTR3	no	no	no
PDCL2	no	2G	4:56458344	C>T	UTR5	no	no	no
UNC5C	no	2G	4:96085305	G>C	UTR3	no	no	no
			5:11514076					
CDO1	no	2G	6	C>T	UTR3	no	no	no
			5:17219603		nonsynonymous			
DUSP1	no	2G	4	C>T	SNV	no	yes	yes
			5:17219603		nonsynonymous			
DUSP1	no	2L	4	C>T	SNV	no	yes	yes
SNX18	no	2G	5:53816165	T>G	UTR3	no	no	no
SNX18	no	2L	5:53816165	T>G	UTR3	no	no	no
LRRC70	no	2G	5:61876832	A>T	stopgain	no	no	no
LOC101927			6:11397132					
686	no	2G	8	G>C	upstream	no	no	no
LOC101927			6:11397132					
686	no	2L	8	G>C	upstream	no	no	no
			6:13949934					
HECA	no	2G	9	A>G	UTR3	no	no	no
			6:13949934					
HECA	no	2L	9	A>G	UTR3	no	no	no
E2F3	no	2G	6:20401732	C>G	upstream	no	no	no
E2F3	no	2L	6:20401732	C>G	upstream	no	no	no
RNF8	no	2G	6:37358779	T>A	UTR3	no	no	no

RNF8	no	10E	6:37360075 7:12333478	T>C	UTR3 nonsynonymous	no	yes	yes
WASL	no	2G	0	A>G	SNV	no	no	no
TMEM248	no	2G	7:66421826 8:10313765	A>G	UTR3	no	no	no
MIR5680;N	no	2G	8	T>C	upstream	no	no	no
CALD	no	2L	8:10313765 8	T>C	upstream	no	no	no
MIR5680;N	no	2G	8:11660321	G>T	UTR5	no	no	no
CALD	no	2L	8:11660321	G>T	UTR5	no	no	no
FDFT1	no	2G	9:22452038	T>C	UTR3 nonsynonymous	no	no	no
DMRTA1	no	2G	9:35811210	C>T	SNV	no	no	no
SPAG8	no	2G	9:37917100 11:5617915	T>G	UTR3	no	no	no
SHB	no	2G	8	A>T	upstream	no	no	no
OR5AL1	no	2H	12:5561429 4	TTGTG>T	upstream	no	no	yes
OR10A7	no	2H	16:7041372 4	G>A	UTR3 nonsynonymous	no	no	no
ST3GAL2	no	2H	16:7177951 7	C>A	SNV	no	no	no
AP1G1	no	2H	16:8661255 8	C>A	nonsynonymous SNV	no	no	no
FOXL1	no	2H	19:5288857 9	T>G	UTR3	no	no	no
ZNF880	no	2H	1:15806676 0	C>T	UTR3	no	yes	yes
KIRREL	no	2H	1:18284852 7	G>T	nonsynonymous SNV	no	no	no
DHX9	no	2H	1:22539378 4	G>C	nonsynonymous SNV	no	no	yes
DNAH14	no	2H	1:41092154	C>T	UTR3 nonsynonymous	no	no	no
RIMS3	no	2H	1:92728421 20:3402538	T>G	SNV	no	no	no
GLMN	no	2H	6	G>A	nonsynonymous SNV	no	no	no
GDF5	no	2H	20:3540663 7	A>C	UTR3 nonsynonymous	no	no	no
SOGA1	no	2H	20:3542255 3	T>C	SNV	no	no	no
SOGA1	no	11A	20:6079140 9	C>T	nonsynonymous SNV	no	no	yes
HRH3	no	2H	20:6138613 1	C>T	nonsynonymous SNV	no	yes	yes
NTSR1	no	2H	2:16811589 8	A>C	UTR3 nonsynonymous	no	no	no
XIRP2	no	2H	2:74783685 4:15799712	C>T	SNV	no	no	no
DOK1	no	2H	0	G>C	upstream nonsynonymous	no	no	no
GLRB	no	2H	5:54416272	G>T	SNV	no	no	no
CDC20B	no	2H	5:54408814	G>A	UTR3	no	no	no
CDC20B	no	3B	6:96662040	A>T	UTR3 nonsynonymous	no	no	no
FUT9	no	2H	8:2088809	G>T	SNV	no	no	no
MYOM2	no	2H	8:30585421 9:10007693	G>A	UTR5 nonsynonymous	no	no	no
GSR	no	2H	3	C>T	SNV	no	yes	yes
CCDC180	no	2H	9:12945904 4	G>A	UTR3 frameshift	no	no	no
LMX1B	no	2H	10:1943516 8	TG>T	deletion	no	no	no
MALRD1	no	2I						

KIAA1551	no	2I	12:3211188 3	T>G	upstream	no	no	no
LINC02400	no	2I	12:4215768 8	T>C GGAAACCC>	upstream	no	yes	yes
GNB3	no	2I	12:6950166 13:1006161	G	UTR5	no	no	no
ZIC5	no	2I	06 14:2484567	A>T	UTR3 frameshift	no	no	no
NFATC4	no	2I	8 17:2703946	G>GC	insertion	no	no	no
PROCA1	no	2I	0	C>T	upstream	no	no	no
ZZEF1	no	2I	17:3907982 18:3482432	A>C	UTR3	no	no	yes
CELF4	no	2I	1	A>C	UTR3 nonsynonymous	no	no	no
ANKRD12	no	2I	18:9255549 1:11623841	T>G	SNV	no	no	no
VANGL1	no	2I	7 1:20438126	A>T	UTR3	no	no	no
PPP1R15B	no	2I	5 1:21152679	C>A	upstream nonsynonymous	no	no	no
TRAF5	no	2I	9 1:24175633	G>C	SNV	no	no	no
KMO	no	2I	3	T>A	UTR3	no	no	no
AGO4	no	2I	1:36323462	A>G	UTR3	no	no	no
MPL	yes	2I	1:43802687 20:3358416	GC>G	upstream nonsynonymous	no	no	no
MYH7B	no	2I	2 20:4812443	C>T	SNV	no	yes	yes
PTGIS	no	2I	7 22:3837951	G>T	UTR3 nonsynonymous	no	no	no
SOX10	no	2I	5 2:13180481	G>A	SNV	no	yes	yes
ARHGEF4	no	2I	3 2:18934816	T>C	UTR3 nonsynonymous	no	no	no
GULP1	no	2I	3 2:23703407	A>G	SNV	no	no	yes
AGAP1	no	2I	4 3:15994371	T>G	UTR3 nonsynonymous	no	no	no
C3orf80	no	2I	8 4:17092327	A>G	SNV	no	no	no
MFAP3L	no	2I	7 4:17556360	C>T	UTR5	no	no	no
GLRA3	no	2I	4 5:11451699	TAAAAA>T	UTR3	no	no	no
TRIM36	no	2I	1 5:15657008	G>A	upstream	no	yes	yes
MED7	no	2I	4 5:15689032	G>A	upstream nonsynonymous	no	no	no
NIPAL4	no	2I	0 6:12276615	G>A	SNV	no	no	no
SERINC1	no	2I	7	T>C	UTR3	no	no	no
MCM3	no	2I	6:52128851	G>A	UTR3 nonsynonymous	no	no	no
NME8	no	2I	7:37903988	G>C	SNV nonsynonymous	no	no	no
NME8	no	2J	7:37901694	A>G	SNV	no	no	no
LINC01606	no	2I	8:58130253	C>T	upstream	no	yes	yes
UBE2R2	no	2I	9:33816422	G>A	upstream	no	no	no
UBE2R2	no	7A	9:33817025	G>A	upstream	no	no	no
UBE2R2	no	7D	9:33817025 10:2796380	G>A	upstream	no	no	no
MKX	no	2J	9 10:7349192	T>A	UTR3 nonsynonymous	no	no	no
CDH23	no	2J	3	G>T	SNV	no	no	no

ITIH2	no	2J	10:7759677 11:1428884	G>A	nonsynonymous SNV	no	yes	yes
SPON1	no	2J	8	G>A	UTR3	no	yes	yes
OR9G1;OR9	no	2J	11:5646757					
G9	no	2J	0	GA>G	upstream	no	no	no
CREBZF	no	2J	11:8536867					
			2	T>C	UTR3	no	no	no
OAS3	no	2J	12:1133886		nonsynonymous			
			94	T>G	SNV	no	no	no
VPS33A	no	2J	12:1227174		nonsynonymous			
			72	A>C	SNV	no	no	no
KMT5A	no	2J	12:1238681					
			63	A>G	upstream	no	no	yes
LOC645177	no	2J	12:2517011		nonsynonymous			
			4	A>G	SNV	no	no	no
CD163	no	2J	12:7633784		nonsynonymous			
			12:8008503	C>T	SNV	no	no	no
PAWR	no	2J	5	AGCGCGCCC				
			14:9475633	G>A	upstream	no	no	no
SERPINA10	no	2J	2	T>A	nonsynonymous			
			15:4285981		SNV	no	no	no
HAUS2	no	2J	7	G>T	UTR3	no	no	no
					nonsynonymous			
TELO2	no	2J	16:1552335	T>C	SNV	no	no	no
CLDN9	no	2J	16:3061897	T>C	upstream	no	no	no
			16:3147612					
ARMC5	no	2J	8	G>A	stopgain	no	no	no
					nonframeshift			
FAM173A	no	2J	16:772105	AACGTGT>A	deletion	no	no	no
			17:1056152					
MYH3	no	2J	6	A>C	upstream	no	no	no
			17:6701464		nonsynonymous			
ABCA9	no	8C	1	A>G	SNV	no	no	no
			17:7650364		nonsynonymous			
DNAH17	no	2J	0	G>A	SNV	no	no	no
			19:5574022					
TMEM86B	no	2J	5	A>AT	UTR5	no	no	no
			19:5589577					
TMEM238	no	2J	9	T>A	upstream	no	no	no
			1:17725024		nonsynonymous			
BRINP2	no	2J	8	C>A	SNV	no	no	no
			1:20143367					
PHLDA3	no	2J	8	G>A	UTR3	no	no	yes
					nonsynonymous			
MAN1C1	no	2J	1:26104687	A>T	SNV	no	no	no
					nonsynonymous			
MAN1C1	no	3D	1:26104812	T>G	SNV	no	no	no
GJB4	no	2J	1:35228594	C>T	UTR3	no	no	no
AGO3	no	2J	1:36395739	C>G	upstream	no	yes	yes
AGO3	no	11A	1:36474567	T>G	stopgain	no	no	no
SNRPB	no	2J	20:2451692	T>C	upstream	no	no	no
			2:13141466		nonsynonymous			
POTEJ	no	2J	9	G>A	SNV	no	yes	yes
			2:19708067		nonsynonymous			
HECW2	no	2J	6	G>A	SNV	no	no	no
			2:19745886					
HECW2	no	5C	0	T>G	upstream	no	no	no
LRPPRC	no	2J	2:44114967	G>C	UTR3	no	no	no
RFC1	no	2J	4:39290159	A>G	UTR3	no	no	no
CEP135	no	2J	4:56898215	T>A	UTR3	no	no	no
			5:10230984		nonsynonymous			
PAM	no	2J	0	T>C	SNV	no	yes	yes
			5:10230996		nonsynonymous			
PAM	no	3A	6	C>G	SNV	no	no	no

FBN2	no	2J	5:12759475 7	A>G	UTR3	no	no	no
MFAP3	no	2J	5:15343367 7	A>G	UTR3	no	no	no
ZNF622	no	2J	5:16465653	G>A	nonsynonymous SNV	no	no	no
ADAMTS16	no	2J	5:5306734 6:11267166	A>G	nonsynonymous SNV	no	no	no
RFPL4B	no	2J	8 7:13381201	G>A	SNV	no	yes	yes
LRGUK	no	2J	3	T>A	upstream nonframeshift	no	no	no
AGR2	no	2J	7:16839376	TGAG>T	deletion	no	no	no
LANCL2	no	2J	7:55501097	C>T	UTR3	no	no	yes
LOC101927 141	no	2J	8:83823935	AAAG>A	upstream	no	no	yes
RUNX1T1	yes	2J	8:93115222	G>C	UTR5 nonframeshift	no	no	no
PIGO	no	2J	9:35092680	TGGA>T	deletion	no	no	yes
AUH	no	2J	9:94124396 10:1083334	G>C	upstream	no	no	no
SORCS1	no	2K	99 10:1083386	A>C	UTR3	no	no	no
SORCS1	no	4B	64 12:1039224	G>A	UTR3	no	no	no
KLRD1	no	2K	9 17:3913428	G>A	UTR5	no	yes	yes
KRT40	no	2K	0 18:5289051	G>T	UTR3	no	no	no
TCF4	no	2K	0 19:1979251	A>T	UTR3	no	no	no
ZNF101	no	2K	5 19:2271153	C>T	UTR3	no	yes	yes
LOC101929 124	no	2K	0 19:2271113	G>T	upstream	no	no	no
LOC101929 124	no	7A	2 19:2410374	T>A	upstream	no	no	no
ZNF726	no	2K	6 19:5423772	C>T	UTR3	no	no	no
MIR518D	no	2K	9 1:16846504	T>C	upstream	no	no	no
LOC101928 565	no	2K	9	T>A	upstream	no	no	no
FNDC5	no	2K	1:33330125 20:4370693	T>A	UTR3	no	no	no
STK4	no	2K	5 21:2217550	C>T	UTR3	no	no	no
LINC00320	no	2K	5 3:16079215	A>T	upstream	no	no	no
PPM1L	no	2K	3 3:16078869	G>C	UTR3	no	no	no
PPM1L	no	11D	3	C>T	UTR3	no	no	no
IP6K2	no	2K	3:48731115	G>T	UTR3	no	no	no
MTHFD2P1	no	2K	3:95402303 4:16596130	A>C	upstream nonsynonymous	no	no	no
TRIM60	no	2K	1 5:13236320	C>A	SNV	no	no	no
ZCCHC10	no	2K	3	GC>G	upstream	no	no	no
SETD9	no	2K	5:56223179	G>C	UTR3	no	no	no
SNRNP48	no	2K	6:7590499	CGAGCCTCC ACCTGTG>C	nonframeshift deletion	no	no	no
VCP	no	2K	9:35063043 13:4687259	C>T	nonsynonymous SNV	no	no	no
LINC00563	no	2L	5	T>A	upstream	no	no	no

FPR2	no	2L	19:52272353	G>A	nonsynonymous SNV	no	yes	yes
ZNF160	no	2L	19:53572841	G>A	nonsynonymous SNV	no	yes	yes
WFDC3	no	2L	20:44404133	C>T	nonsynonymous SNV	no	yes	yes
APP	no	2L	21:27354775	G>A	nonsynonymous SNV	no	no	no
B3GALNT1	no	2L	3:160804228	C>G	nonsynonymous SNV	no	no	no
GADL1	no	2L	3:30769878	C>T	nonsynonymous SNV	no	no	no
EVC	no	2L	4:5798864	A>G	nonsynonymous SNV	no	no	no
CDH6	no	2L	5:31193978	A>T	UTR5	no	no	no
CDH6	no	8C	5:31324852	T>C	UTR3	no	no	no
OSGIN2	no	2L	8:90914174	G>T	UTR5	no	no	no
NCR3LG1	no	3A	11:17398465	G>A	UTR3	no	yes	yes
ARL14EP	no	3A	11:30359358	A>C	UTR3	no	no	no
DISP2	no	3A	15:40650458	G>GCCA	UTR5	no	no	yes
CNP	no	3A	17:40126890	C>T	UTR3	no	no	no
XAF1	no	3A	17:6677217	G>C	UTR3	no	no	no
ARHGAP35	no	3A	19:47505680	CT>C	UTR3	no	no	no
OR2T3	no	3A	1:248636180	A>T	upstream	no	no	no
HPCAL4	no	3A	1:40147573	C>A	UTR3	no	no	no
SZT2	no	3A	1:43919835	C>T	UTR3	no	yes	yes
ALG6	no	3A	1:63832771	G>A	upstream	no	no	yes
RALGAPA2	no	3A	20:20372030	A>G	UTR3	no	no	no
GGT7	no	3A	20:33440242	AG>A	frameshift deletion	no	no	no
ARHGAP40	no	3A	20:37278760	G>T	UTR3	no	no	no
LINC01749	no	3A	20:61639829	G>A	upstream	no	no	no
FBLN2	yes	3A	3:13659729	A>T	nonsynonymous SNV	no	no	no
FBLN2	yes	7D	3:13612652	T>A	nonsynonymous SNV	no	no	no
SYNPO2	no	3A	4:119951779	C>T	nonsynonymous SNV	no	no	no
SYNPO2	no	3D	4:119771864	C>T	UTR5	no	no	no
HSPA4L	no	3A	4:128729200	A>G	nonsynonymous SNV	no	yes	yes
BMP3	no	3A	4:81975262	A>G	UTR3	no	no	no
PCDHB7	no	3A	5:14055581	T>A	UTR3	no	no	no
LOC102724511	no	3A	6:170475708	C>A	upstream	no	no	no
GRIN3A	no	3A	9:104331647	T>G	UTR3	no	no	no
IPPK	no	3A	9:95433261	G>A	upstream	no	no	no
KAZALD1	no	3B	10:10282768	C>T	UTR3	no	no	no
GDF10	no	3B	10:48428738	A>T	nonsynonymous SNV	no	yes	yes
TRPC6	no	3B	11:101323350	G>T	UTR3	no	no	no

NTM	no	3B	11:1322057 24	G>A	UTR3 nonsynonymous	no	no	no
KRTAP5-4	no	3B	11:1642822	A>C	SNV nonsynonymous	no	no	no
KRTAP5-4	no	3D	11:1642822 11:6539194	A>C	SNV	no	no	no
PCNX3	no	3B	11:6719573 1	G>T	splicing	no	no	no
RPS6KB2	no	3B	11:6719573 4	C>A	upstream	no	yes	yes
RPS6KB2	no	3D	11:6719573 4	C>A	upstream	no	yes	yes
IGHMBP2	no	3B	11:6870753 7	G>A	UTR3 nonsynonymous	no	no	no
UCP3	no	3B	11:7371679 0	T>C	SNV nonsynonymous	no	no	no
UCP3	no	3D	11:7371679 0	T>C	SNV nonsynonymous	no	no	no
UBTFL1	no	3B	11:8981974 2	G>T	SNV	no	no	no
FAT3	yes	3B	11:9262847 7	C>G	UTR3	no	no	no
SESN3	no	3B	11:9490551 9	A>C	UTR3	no	no	no
UBE3B	no	3B	12:1099154 88	C>T	UTR5	no	no	no
GXYLT1	no	3B	12:4253864 0	G>A	UTR5 nonsynonymous	no	no	yes
TBK1	no	3B	12:6485819 6	C>T	SNV	no	no	no
MFAP5	no	3B	12:8816338	A>T	upstream	no	no	no
MFAP5	no	3D	12:8816338 14:1053617	A>T	upstream	no	no	no
CEP170B	no	3B	14:6097512 87	T>G	UTR3	no	no	no
SIX6	no	3B	16:2884767 3	T>A	upstream nonsynonymous	no	no	no
ATXN2L	no	3B	16:3149442 6	C>T	SNV	no	yes	yes
SLC5A2	no	3B	16:8584039 3	G>A	upstream nonsynonymous	no	no	no
COX4I1	no	3B	17:2795340 2	G>T	SNV	no	no	no
SSH2	no	3B	17:2885069 8	T>C	UTR3	no	no	no
GOSR1	no	3B	17:2935862 2	G>T	UTR3	no	no	no
LOC107984 974	no	3B	17:3485113 4	ATCTTGGC> A TCAGGGAGA AGACAAAG> T	frameshift deletion frameshift deletion nonsynonymous	no no no no no	no no no no no	no no no no no
ZNHIT3	no	3B	17:7480981 18:2148457	T>A	SNV nonsynonymous	no	no	no
EIF4A1	no	3B	18:4440606 9	A>T	SNV	no	no	no
LAMA3	no	3B	18:4440606 7	C>T	UTR3	no	no	no
PIAS2	no	3B	18:596486 19:1668846	C>G	upstream nonsynonymous	no	no	no
CLUL1	no	3B	19:4171240 3	C>T	SNV	no	no	yes
MED26	no	3B	19:4171240 6	T>G	UTR3	no	no	no
CYP2S1	no	11B	19:4171320 3	A>G	UTR3	no	no	no
CYP2S1	no	11D	19:4171320 3	A>G	UTR3	no	no	no
CYP2S1	no	3B	19:4463761 2	A>C	UTR3	no	no	no
ZNF225	no	3B						

EML2	no	3B	19:4612091 9	A>C	nonsynonymous SNV	no	no	no
LAIR1	no	3B	19:5488220 1	G>A	upstream	no	no	no
LAIR1	no	3D	19:5488220 1	G>A	upstream	no	no	no
GP6	no	3B	19:5553005 0	C>T	nonsynonymous SNV	no	yes	yes
MTHFR	no	3B	1:11850906	T>C	nonsynonymous SNV	no	no	no
PRAMEF18	no	3B	1:13038731 1:14459859	T>C	UTR3	no	no	no
NBPF8	no	3B	6	C>T	UTR5	no	no	yes
TMEM240	no	3B	1:1470638 1:15048608	G>T	UTR3	no	no	no
ECM1	no	3B	6	T>G	UTR3	no	no	no
ECM1	no	8C	1:15048062 3	C>G	UTR5	no	no	no
FLG	no	3B	1:15228001 6	C>T	nonsynonymous SNV	no	yes	yes
RASAL2	no	3B	1:17844731 9	A>T	UTR3	no	no	no
RABIF	no	3B	1:20285829 3	A>T	UTR5	no	no	no
KCTD3	no	3B	1:21579414 3	G>T	UTR3	no	no	no
KCTD3	no	3D	1:21579414 3	G>T	UTR3	no	no	no
MARK1	no	3B	1:22083709 2	T>G	UTR3	no	no	no
ITGB3BP	no	3B	1:63974227 20:1839506	A>G	nonsynonymous SNV	no	no	no
DZANK1	no	3B	8	C>T	nonsynonymous SNV	no	no	no
DZANK1	no	11A	20:1842406 4	C>T	nonsynonymous SNV	no	no	yes
CSE1L	no	3B	20:4770730 4	A>G	nonsynonymous SNV	no	no	no
KRTAP10-7	no	3B	21:4602118 3	C>T	nonsynonymous SNV	no	yes	yes
IL1R2	no	3B	2:10260787 2	G>A	upstream	no	no	no
LOC105373 782	no	3B	2:18603218 8	C>A	upstream	no	no	no
PCYOX1	no	3B	2:70505500	G>A	UTR3	no	no	no
SNRNP200	no	3B	2:96967292 3:11862026	A>T	nonsynonymous SNV	no	no	no
IGSF11	no	3B	9	C>T	UTR3	no	yes	yes
C3orf58	no	3B	3:14369045 5	A>C	upstream	no	no	no
LRRC3B	no	3B	3:26664715	G>T	UTR5	no	no	no
EXOG	no	3B	3:38566444	T>A	UTR3	no	no	no
ANO10	no	3B	3:43621929	C>A	nonsynonymous SNV	no	no	no
ANO10	no	3D	3:43621929	C>A	nonsynonymous SNV	no	no	no
ZKSCAN7	no	3B	3:44596849	T>G	UTR5	no	no	no
ZNF660	no	3B	3:44636598	A>C	nonsynonymous SNV	no	no	no
ZNF660	no	3D	3:44636598	A>C	nonsynonymous SNV	no	no	no
CISH	no	3B	3:50644521	C>A	UTR3	no	no	no
LINC01266	no	3B	3:633768	G>A	upstream	no	yes	yes
LINC01266	no	3D	3:633768	G>A	upstream	no	yes	yes

FRMD4B	no	3B	3:69219703	A>T	UTR3	no	no	no
FRMD4B	no	6B	3:69435723 4:12337818	A>T	upstream	no	no	no
IL2	yes	3B	5 4:17525374	A>T	upstream	no	no	no
CEP44	no	3B	9	A>G	UTR3	no	no	no
LOC101928 551	no	3B	4:17575233 7	T>A	upstream nonsynonymous	no	no	no
FGFR3	yes	3B	4:1803622 4:18606408	T>A	SNV	no	no	no
SLC25A4	no	3B	3 4:18606408	C>T	upstream	no	no	no
SLC25A4	no	3D	3 5:12952011	C>T	upstream nonsynonymous	no	no	no
CHSY3	no	3B	3	G>T	SNV	no	no	no
LIFR	yes	3B	5:38480868	A>T	UTR3	no	no	no
LINC02197	no	3B	5:70742618	TAACTCTC>T	upstream nonsynonymous	no	no	no
FCHO2	no	3B	5:72348195 6:15444682	C>G	SNV	no	no	no
OPRM1	no	3B	0	A>T	UTR3	no	no	no
LOC102724 357	no	3B	6:16946690 9	T>A	upstream nonsynonymous	no	no	no
SERPINB1	no	3B	6:2840777	A>T	SNV	no	no	no
DPPA5	no	3B	6:74064561	G>A	upstream	no	no	no
DPPA5	no	3D	6:74064561 7:10017268	G>A	upstream	no	no	no
LRCH4	no	3B	9 7:11426991	G>C	UTR3 nonsynonymous	no	yes	yes
FOXP2	no	3B	5	T>C	SNV nonsynonymous	no	no	no
MPLKIP	no	3B	7:40173920	G>A	SNV nonsynonymous	no	no	yes
MPLKIP	no	3D	7:40173920	G>A	SNV nonsynonymous	no	no	yes
POLD2	no	3B	7:44157268	A>T	SNV	no	no	no
MIR4283- 1;MIR4283- 2	no	3B	7:57024509	T>A	upstream	no	no	no
ATP5J2- PTCD1;PTC D1	no	3B	7:99015916	C>G	UTR3	no	no	no
NAT1	no	3B	8:18080641	TAAA>T	UTR3	no	no	yes
NAT1	no	6B	8:18080641	TAAA>T	UTR3	no	no	yes
LY96	no	3B	8:74903464 9:13145158	A>G	upstream	no	no	no
SET	yes	3B	8	C>T	UTR5 nonsynonymous	no	no	no
ZCCHC7	no	3B	9:37305639	C>G	SNV nonsynonymous	no	no	yes
ZCCHC7	no	3D	9:37354712	T>G	SNV nonsynonymous	no	no	no
GCNT1	no	3B	9:79118112	T>G	SNV nonsynonymous	no	no	no
SPATA31C2	no	3B	9:90745532 X:10635662	G>T	SNV	no	no	no
RBM41	no	3B	6 X:13120996	C>A	splicing	no	no	no
STK26	no	3B	7 X:13512745	C>T	UTR3	no	no	no
SLC9A6	no	3B	9 X:13512745	T>G	UTR3	no	no	no
SLC9A6	no	3D	9	T>G	UTR3	no	no	no

TSR2	no	3B	X:54471120	T>C	UTR3	no	no	no
USP51	no	3B	X:55515905	A>G	upstream	no	no	no
SFR1	no	3C	10:1058811	C>G	upstream	no	no	no
CREM	no	3C	15	G>C	UTR3	no	no	no
FZD8	no	3C	10:3550091	A>C	UTR3	no	no	no
KIF11	no	3C	8	T>C	UTR3	no	yes	yes
GRIA4	no	3C	10:3592727	C>A	UTR3	no	no	no
SCN2B	no	3C	5	C>T	UTR3	no	no	no
OR5T1	no	3C	10:9441498	T>C	nonsynonymous SNV	no	no	yes
OR8H1	no	3C	7	A>G	nonsynonymous SNV	no	no	no
STX3	no	3C	11:1057827	T>C	UTR3	no	no	no
DEUP1	no	3C	52	A>C	upstream	no	no	no
CLEC1A	no	3C	11:1180369	T>C	upstream	no	no	no
STYK1	no	3C	82	G>A	upstream	no	yes	yes
HIP1R	no	3C	11:5604392	T>A	UTR3	no	no	no
RASSF9	no	3C	3	G>C	upstream	no	no	no
RCBTB2	no	3C	11:5605788	TGATCA>T	UTR3	no	no	no
LTB4R	no	3C	6	T>C	UTR3	no	no	no
BLOC1S6	no	3C	12:1082748	GA>G	UTR3	no	no	no
MPI	no	3C	6	G>C	UTR3	no	no	no
PDP2	no	3C	12:1233472	T>G	UTR3	no	no	yes
FBXO31	no	3C	50	G>A	UTR3	no	yes	yes
ELAC2	no	3C	12:8623060	T>C	nonsynonymous SNV	no	yes	yes
TOM1L2	no	3C	9	A>T	UTR3	no	no	no
MIR1253	no	3C	13:4906404	A>C	upstream	no	no	no
KRT222	no	3C	8	G>A	upstream	no	no	no
DHX8	no	3C	14:2478701	T>A	nonsynonymous SNV	no	no	no
ENO3	no	3C	2	A>C	upstream	no	no	no
TOM1L1	no	3C	15:4589924	T>A	stopgain	no	no	no
GYS1	no	3C	6	A>T	nonsynonymous SNV	no	no	no
FCRL1	no	3C	15:7519102	T>A	UTR3	no	no	no
POP3	no	3C	1	T>A	upstream	no	no	no
PRRC2C	no	3C	16:6692264	C>A	nonsynonymous SNV	no	no	no
TOR1AIP2	no	3C	3	T>A	nonsynonymous SNV	no	no	no
ARF1	no	3C	17:1292121	T>C	upstream	no	no	no
RYR2	no	3C	9	G>T	stopgain	no	no	no

TRIM58	no	3C	1:24804206 1	A>G	UTR3	no	no	yes
LINC01767	no	3C	1:56879741	C>T	upstream	no	no	no
ZNF644	no	3C	1:91488572 20:3164325	G>C	upstream nonsynonymous	no	no	no
BPIFB3	no	3C	5	G>T	SNV	no	yes	yes
SPEF1	no	3C	20:3758690 22:3770991	C>T	UTR3	no	no	no
CYTH4	no	3C	8 22:4122239	G>A	UTR3	no	no	no
ST13	no	3C	4 22:4253610	T>C	UTR3	no	no	no
CYP2D7	no	3C	7 22:5094680	G>A	UTR3 nonsynonymous	no	no	yes
NCAPH2	no	3C	0 2:17959922	T>A	SNV nonsynonymous	no	no	no
TTN	no	3C	8 2:20765771	C>A	SNV	no	no	no
FASTKD2	no	3C	4	G>A	UTR3	no	no	yes
RMND5A	no	3C	2:86947420	C>G	UTR5	no	no	yes
CNOT10	no	3C	3:32746301	A>G	splicing	no	no	no
SLC22A13	no	3C	3:38319163	C>A	UTR3	no	no	no
SNRK	no	3C	3:43327887	G>C	upstream	no	no	no
FAM208A	no	3C	3:56717764	C>G	upstream nonsynonymous	no	no	no
ATXN7	no	3C	3:63967994	A>G	SNV	no	no	no
CPNE9	no	3C	3:9745662 4:10983934	T>G	UTR5 nonsynonymous	no	no	no
COL25A1	no	3C	8 4:16765591	G>A	SNV	no	no	no
SPOCK3	no	3C	6 4:18902728	A>T	UTR3	no	no	no
TRIML2	no	3C	1 5:12288881	G>C	upstream nonsynonymous	no	no	no
CSNK1G3	no	3C	3 5:14589065	A>G	SNV	no	no	no
TCERG1	no	3C	0 5:17021072	A>G	UTR3	no	no	no
GABRP	no	3C	7 5:17649497	C>T	UTR5	no	no	no
ZNF346	no	3C	5	G>A	UTR3	no	no	no
SKP2	no	3C	5:36182583	T>G	UTR3	no	no	no
AHRR	no	3C	5:436429 6:12766575	G>C	UTR3	no	no	no
ECHDC1	no	3C	1	T>G	upstream	no	no	no
MCCD1	no	3C	6:31496581	A>C	upstream	no	no	no
BAK1	no	3C	6:33541203	G>T	UTR3 nonsynonymous	no	no	no
FOXP4	no	3C	6:41559055	T>C	SNV	no	no	no
BMP5	yes	3C	6:55618645	A>T	UTR3	no	no	no
KHDRBS2	no	3C	6:62390455	A>T	UTR3	no	no	no
EPHA7	yes	3C	6:94130145	G>T	upstream nonsynonymous	no	no	yes
NACAD	no	3C	7:45125072 8:14399862	A>C	SNV nonsynonymous	no	no	no
CYP11B2	no	3C	0	C>T	SNV	no	no	no
ADAM5	no	3C	8:39171375	G>A	upstream	no	no	no
STK3	no	3C	8:99902005	C>A	UTR5	no	no	no
PRKACG	no	3C	9:71629649	A>T	upstream	no	no	no

OCRL	no	3C	X:12867471 8	C>T	nonsynonymous SNV	no	no	no
MPP1	no	3C	X:15400708 8	A>C	UTR3	no	no	no
YIPF6	no	3C	X:67754417 10:2745888	A>C	UTR3	no	no	no
MASTL	no	3D	2	G>A	nonsynonymous SNV	no	no	no
CD3D	no	3D	11:1182102 01	T>A	nonsynonymous SNV	no	no	no
NAV2	no	3D	11:2005761 4	G>T	nonsynonymous SNV	no	no	no
TRIM48	no	3D	11:5503274 0	C>G	nonsynonymous SNV	no	no	no
SSH1	no	3D	12:1091789 32	T>C	UTR3	no	yes	yes
C12orf65;M PHOSPH9	no	3D	12:1237178 00	C>T	upstream nonsynonymous SNV	no	no	no
LIN7A	no	3D	12:8133145 1	C>G	nonsynonymous SNV	no	no	no
COL4A2	no	3D	13:1110822 83	G>A	nonsynonymous SNV	no	no	no
ATP4B	no	5C	13:1143133 64	G>T	upstream nonsynonymous SNV	no	no	no
PLEKHH1	no	3D	14:6805229 8	C>G	upstream nonsynonymous SNV	no	no	no
TANGO6	no	3D	16:6887707 3	T>G	upstream	no	no	no
FAM222B	no	3D	17:2708449 2	G>T	UTR3	no	no	no
PSMD11	no	3D	17:3080851 0	G>C	UTR3	no	no	no
GPATCH8	no	3D	17:4251419 9	A>C	UTR5	no	no	no
EIF5A	no	3D	17:7211343 17:7667214	C>T	UTR5	no	no	yes
CYTH1	no	3D	6	C>T	UTR3	no	yes	yes
PSMA8	no	3D	18:2377278 3	G>A	UTR3	no	no	no
ZNF564	no	3D	19:1263660 5	A>G	UTR3	no	yes	yes
WTIP	no	3D	19:3499125 6	G>A	UTR3	no	no	no
NLRP8	no	3D	19:5649089 0	G>A	nonsynonymous SNV	no	yes	yes
NBPF15	no	3D	1:14858900 5	G>A	nonsynonymous SNV	no	yes	yes
OR6N1	no	3D	1:15873607 0	G>T	nonsynonymous SNV	no	no	no
OR6N1	no	5F	1:15873602 7	C>A	nonsynonymous SNV	no	yes	yes
ENAH	no	3D	1:22568135 3	C>T	UTR3	no	no	no
LUZP1	no	3D	1:23413001	A>T	UTR3	no	no	no
LUZP1	no	4C	1:23411488	T>G	UTR3	no	no	yes
LUZP1	no	8A	1:23413922	A>G	UTR3	no	no	no
AKT3	yes	3D	1:24385891 3	A>T	nonsynonymous SNV	no	no	no
BPIFB4	no	3D	20:3166940 3	C>T	nonsynonymous SNV	no	no	yes
TTLL12	no	3D	22:4358361 5	G>A	upstream nonsynonymous SNV	no	no	no
MERTK	no	3D	2:11276154 1	T>A	upstream nonsynonymous SNV	no	no	no
INO80D	no	3D	2:20686791 0	T>C	UTR3	no	no	no
INO80D	no	7D	2:20686878 9	G>GT	UTR3	no	no	no

CUL3	yes	3D	2:22537944 0	C>A	nonsynonymous SNV	no	no	no
CRIM1	no	3D	2:36623925	T>C	nonsynonymous SNV	no	no	no
CRIM1	no	11A	2:36583706	G>C	nonsynonymous SNV	no	no	no
KCMF1	no	3D	2:85283054 3:14187653	T>C	UTR3	no	no	no
GK5	no	3D	2 4:12641313	A>G	UTR3	no	no	no
FAT4	yes	3D	5 4:12636746	T>G	UTR3	no	no	no
FAT4	yes	4D	8	A>G	nonsynonymous SNV	no	no	no
DCAF16	no	3D	4:17803792	G>A	UTR3	no	no	no
STX18	no	3D	4:4425265	T>A	nonsynonymous SNV	no	no	no
MMRN1	no	3D	4:90815281	C>A	upstream nonsynonymous SNV	no	no	no
GZMK	no	3D	5:54329734	G>A	SNV	no	no	no
TMEM170B	no	3D	6:11537719	A>T	upstream	no	no	no
LOC101928 253	no	3D	6:12001604	T>C	upstream	no	yes	yes
LOC101928 253	no	8D	6:12001811	CACAT>C	upstream	no	yes	yes
HDGFL1	no	3D	6:22571445 7:11567176	A>T	UTR3	no	no	no
TFEC	no	3D	9	A>T	upstream	no	no	no
ETV1	yes	3D	7:14030294	G>T	UTR5	no	no	yes
ETV1	yes	10C	7:13932287 8:10826357	G>C	UTR3	no	no	no
ANGPT1	no	3D	7	C>T	UTR3	no	no	no
MSR1	no	3D	8:16012653	C>A	nonsynonymous SNV	no	no	no
PTCH1	yes	3D	9:98280163	G>A	upstream	no	no	no
PTCH1	yes	9C	9:98209237 11:1438035	T>C	nonsynonymous SNV	no	no	no
RRAS2	no	4A	0 11:6069000	C>T	nonsynonymous SNV	no	no	no
TMEM109	no	4A	9 11:6564711	C>T	UTR3	no	no	no
CTSW	no	4A	4 11:7125963	A>G	upstream	no	no	no
KRTAP5-9	no	4A	1 12:1134477	C>T	UTR5	no	no	no
OAS2	no	4A	16 12:5385488	G>A	UTR3	no	no	no
PCBP2	no	4A	6 13:3724753	A>G	nonsynonymous SNV	no	no	no
SERTM1	no	4A	7 13:8756390	C>T	upstream	no	no	no
LINC00430	no	4A	1 16:7445583	T>C	upstream	no	no	no
CLEC18B	no	4A	4 16:8806125	G>A	upstream	no	no	yes
BANP	no	4A	7 17:2182447	G>A	splicing	no	no	no
FAM27E5	no	4A	9 17:6176963	T>A	upstream	no	no	no
MAP3K3	no	4A	2 1:15167367	A>G	nonsynonymous SNV	no	no	no
CELF3	no	4A	5 1:17238995	G>A	UTR3	no	no	no
C1orf105	no	4A	3 1:17317670	T>A	UTR5	no	no	no
TNFSF4	no	4A	2	G>A	upstream	no	no	no

RGSL1	no	4A	1:18249686 3	T>C	nonsynonymous SNV	no	no	no
SSX2IP	no	4A	1:85111830 2:12097624	A>T	UTR3	no	no	no
TMEM185B	no	4A	4	C>T	UTR3	no	no	no
TUBA3D	no	4A	2:13223322 4	C>T	upstream nonsynonymous SNV	no	no	no
DNAH6	no	4A	2:84848625	C>G	UTR3	no	no	no
ASAP2	no	4A	2:9545646 3:18479805	T>G	UTR3	no	no	no
C3orf70	no	4A	4	G>T	UTR3	no	no	no
CAMK4	no	4A	5:11082574 5	A>T	UTR3	no	no	no
MNX1	yes	4A	7:15679922 8	A>C	nonsynonymous SNV	no	no	no
ST18	no	4A	8:53076613	C>A	nonsynonymous SNV	no	no	no
PCDH19	no	4A	X:99546872 10:1068495	T>A	UTR3	no	no	no
SORCS3	no	4B	56	A>T	nonsynonymous SNV	no	no	no
SORCS3	no	4D	10:1068495 56	A>T	nonsynonymous SNV	no	no	no
LOC283194	no	4B	11:5882680 9	A>T	upstream	no	no	no
LOC283194	no	4D	11:5882680 9	A>T	upstream	no	no	no
KCTD12	no	4B	13:7745630 4	A>C	UTR3	no	no	no
KCTD12	no	4D	13:7745630 4	A>C	UTR3	no	no	no
ENTPD5	no	4B	14:7443970 4	A>T	nonsynonymous SNV	no	no	no
ENTPD5	no	4D	14:7443970 4	A>T	nonsynonymous SNV	no	no	no
LINC02087	no	4B	17:1572137 5	C>T	upstream	no	no	no
AXL	no	4B	19:4176739 5	T>A	UTR3	no	no	no
AXL	no	4D	19:4176739 5	T>A	UTR3	no	no	no
SULT2A1	no	4B	19:4839012 9	T>C	upstream	no	no	no
SULT2A1	no	4D	19:4839012 9	T>C	upstream	no	no	no
RFWD2	no	4B	1:17617669 3	G>T	upstream	no	no	no
FOXA2	no	4B	20:2256612 8	T>A	upstream	no	no	no
FOXA2	no	4D	20:2256612 8	T>A	upstream	no	no	no
ANAPC1	no	4B	2:11264261 8	G>A	upstream	no	yes	yes
SP3	no	4B	2:17477788 2	G>A	nonsynonymous SNV	no	no	no
SP3	no	4D	2:17477788 2	G>A	nonsynonymous SNV	no	no	no
HNRNPA3; MIR4444- 1;MIR4444- 2	no	4B	2:17807721 5	G>A	upstream	no	yes	yes
HNRNPA3; MIR4444- 1;MIR4444- 2	no	4D	2:17807721 5	G>A	upstream	no	yes	yes
CD28	yes	4B	2:20459437 1	G>A	stopgain	no	no	no
CD28	yes	4D	2:20459437 1	G>A	stopgain	no	no	no

CD28	yes	5E	2:20460148 4	T>A	UTR3	no	no	no
CD28	yes	5F	2:20460148 4	T>A	UTR3	no	no	no
OR5K2	no	4B	3:98215839	C>A	upstream	no	no	no
SPON2	no	4B	4:1202363 4:17273577	C>A	splicing nonsynonymous	no	no	no
GALNTL6	no	4B	8	C>T	SNV	no	no	no
ABCC10	no	4B	6:43395798	T>C	nonsynonymous SNV	no	no	no
ABCC10	no	4D	6:43395798	T>C	nonsynonymous SNV	no	no	no
ABCC10	no	7A	6:43403980	A>C	nonsynonymous SNV	no	no	no
ABCC10	no	7D	6:43403980	A>C	nonsynonymous SNV	no	no	no
PTDSS1	no	4B	8:97311937	C>G	nonsynonymous SNV	no	no	no
PTDSS1	no	4D	8:97311937 9:13630762	C>G	nonsynonymous SNV	no	no	no
ADAMTS13	no	4B	9	G>A	nonsynonymous SNV	no	no	no
ADAMTS13	no	4D	9:13630762 9	G>A	nonsynonymous SNV	no	no	no
TCERG1L	no	4C	10:1329650 97	C>A	nonsynonymous SNV	no	no	no
LOC102724 593	no	4C	10:4695089 3	G>T	upstream	no	no	no
VSTM4	no	4C	10:5031145 3	A>C	UTR3	no	no	no
VSTM4	no	8C	10:5031040 5	T>C	UTR3	no	no	no
TLL2	no	4C	10:9812767 7	A>G	UTR3	no	no	no
TLL2	no	11B	10:9812546 5	C>T	UTR3	no	no	no
TLL2	no	11D	10:9812546 5	C>T	UTR3	no	no	no
PTPN5	no	4C	11:1881327 0	C>T	UTR5	no	yes	yes
LTBP3	no	4C	11:6530868 3	T>C	nonsynonymous SNV	no	no	no
GPR162	no	4C	12:6936501 12:7131518	C>G	UTR3	no	no	no
PTPRR	no	4C	6	G>C	upstream	no	no	no
PTPRR	no	11A	12:7109207 4	G>T	nonsynonymous SNV	no	yes	yes
ADAM20	no	4C	14:7099096 1	T>C	nonsynonymous SNV	no	no	no
MTMR10	no	4C	15:3123163 2	T>A	UTR3	no	no	no
BAHD1	no	4C	15:4075985 0	C>G	UTR3	no	no	no
ICE2	no	4C	15:6074354 4	T>C	UTR3	no	yes	yes
CRAMP1	no	4C	16:1726714 17:6603092	A>C	UTR3	no	no	no
KPNA2	no	4C	5	G>C	upstream	no	no	yes
ANKRD62	no	4C	18:1212804 3	T>C	UTR3	no	yes	yes
SERPINB13	no	4C	18:6126572 2	A>G	UTR3	no	no	no
OR2W5	no	4C	1:24765404 4	C>A	upstream	no	no	no
OR2L5	no	4C	1:24818561 1	G>T	nonsynonymous SNV	no	yes	yes
RRAGC	no	4C	1:39325737	C>G	upstream	no	no	no

SYNDIG1	no	4C	20:2445008 5	C>A	UTR5	no	no	no
CBLN4	no	4C	20:5457249 0	T>C	UTR3	no	no	no
PCK1	no	4C	20:5613553 5	A>G	upstream	no	no	no
DYRK1A	no	4C	21:3873724 7	G>A	upstream	no	no	no
GLI2	no	4C	2:12174938 5	A>T	UTR3	no	no	no
RESP18	no	4C	2:22019808 1	T>C	upstream	no	no	no
TOGARAM2	no	4C	2:29204100	G>A	upstream	no	no	no
SCAP	no	4C	3:47467041	A>C	nonsynonymous SNV	no	no	no
ROBO2	yes	4C	3:77089504	T>A	UTR5	no	no	no
NUP54	no	7C	4:77052429 6:13882037	G>C	nonsynonymous SNV	no	no	no
NHSL1	no	4C	8 7:10209732	T>G	SNV	no	no	no
ALKBH4	no	4C	5 7:15077674	G>C	UTR3	no	no	no
FASTK	no	4C	9	A>T	nonsynonymous SNV	no	no	no
CHST12	no	4C	7:2474076	T>C	UTR3	no	no	no
MIR4651;P OR	no	4C	7:75544331 8:10221139	G>A	upstream	no	no	no
ZNF706	no	4C	6 8:10221848	T>A	UTR3	no	no	no
ZNF706	no	10E	9 9:10088208	C>G	upstream	no	no	no
TRIM14	no	4C	0	G>GTT CGCACCCCT GTGCCCTGC	upstream	no	yes	yes
QRFP	no	4C	9:13377270 9	AT>C	upstream	no	no	yes
HPS1	no	4D	10:1001821 58	T>C	nonsynonymous SNV	no	no	no
NEBL	no	4D	10:2107048 1	G>T	UTR3	no	no	no
GUCY1A2	no	4D	11:1065476 29	A>G	UTR3	no	no	no
ZNF215	no	4D	11:6978782 11:7388240	T>C	UTR3	no	no	no
PPME1	no	4D	9	C>T	UTR5	no	no	no
EP400	no	4D	12:1324983 26	CACAGCCAT TTATT>C	frameshift deletion	no	no	no
STK38L	no	4D	12:2739615 0	C>T	upstream	no	no	no
IQSEC3	no	4D	12:286749	T>A	UTR3	no	no	no
PRMT8	no	4D	12:3662854 15:1021926	T>A	nonsynonymous SNV	no	no	no
TM2D3	no	4D	24 15:1021925	T>C	upstream	no	yes	yes
TM2D3	no	11B	09 16:3140460	A>C	nonsynonymous SNV	no	no	no
ITGAD	no	4D	6 16:3142241	A>T	upstream	no	no	no
ITGAD	no	7D	5	G>T	nonsynonymous SNV	no	no	no
COR07;COR 07-PAM16	no	4D	16:4409361 16:5685763	T>C	nonsynonymous SNV	no	no	no
NUP93	no	4D	1 16:7550807	A>G	SNV	no	no	no
CHST6	no	4D	8	A>G	UTR3	no	no	no

MAF	yes	4D	16:7962782 1	A>C	UTR3	no	no	no
HSD17B2	no	4D	16:8206923 2	T>A	nonsynonymous SNV	no	no	no
GID4	no	4D	17:1796974 2	A>G	UTR3	no	no	no
DUSP3	no	4D	17:4184672 9	C>A	UTR3	no	no	no
GLTPD2	no	4D	17:4691672 17:6650855	T>G	upstream	no	no	no
PRKAR1A	yes	4D	7 17:7320099	A>G	UTR5	no	no	no
NUP85	no	4D	0 19:5843741	C>A	upstream	no	no	yes
ZNF418	no	4D	7 1:15295589	A>C	UTR3	no	no	no
SPRR1A	no	4D	3 1:18237596	A>T	upstream	no	no	no
LINC00272	no	4D	4	T>A	upstream	no	no	no
ELOVL1	no	4D	1:43833395 21:3466054	G>C	UTR5 frameshift	no	no	no
IL10RB	no	4D	3 21:4797593	T>TA	insertion frameshift	no	no	no
DIP2A	no	4D	2 2:20794578	GC>G	deletion	no	no	no
KLF7	no	4D	1 2:21581564	GTA>G	UTR3 frameshift	no	yes	yes
ABCA12	no	4D	8 3:18860367	CT>C	deletion	no	no	no
LPP	yes	4D	2 3:18859986	T>C	UTR3	no	no	no
LPP	yes	6B	1 3:18859986	C>G	UTR3	no	no	no
LPP	yes	6D	1	C>G	UTR3	no	no	no
ABHD5	no	4D	3:43761858 4:18812269	C>T	UTR3	no	no	yes
LINC02374	no	4D	1	G>A	upstream	no	no	yes
GNRHR	no	4D	4:68603267	A>C	UTR3	no	no	no
SMR3B	no	4D	4:71247985 5:12252295	T>C	upstream	no	no	no
PRDM6	no	4D	0 5:13419392	A>G	UTR3	no	no	yes
C5orf24	no	4D	7 5:13419493	A>G	UTR3	no	no	no
C5orf24	no	6A	2 5:16929012	C>A	UTR3	no	no	no
FAM196B	no	4D	9 5:18067264	G>A	UTR3	no	no	no
CTC- 338M12.4	no	4D	1	C>T	upstream	no	no	no
LINC02234	no	4D	5:97176264	C>T	upstream	no	no	no
CARMIL1	no	4D	6:25279455	C>T	upstream	no	no	no
PRIM2	no	4D	6:57513264	A>T	UTR3	no	no	no
TTK	no	4D	6:80713615	A>G	upstream	no	no	no
TSG1	no	4D	6:94416525	A>T	upstream	no	no	no
CDK13	no	4D	7:40027467 8:10536162	C>A	nonsynonymous SNV	no	no	no
DCSTAMP	no	4D	2	C>T	nonsynonymous SNV	no	yes	yes
MIR5692A1; MIR5692A2	no	4D	8:12576386 9:13179847	A>T	upstream	no	no	no
MIGA2	no	4D	1	A>G	upstream	no	no	yes
SPATA31D3	no	4D	9:84563973	C>T	UTR3	no	no	no

SPRN	no	5A	10:1352353 92	A>G	UTR3	no	no	no
MIR1254-2	no	5A	10:2368232 5	G>A	upstream	no	no	yes
CDHR5	no	5A	11:624925 11:6661669	G>A	UTR5	no	no	no
PC	no	5A	9 11:7836949	AC>A	splicing nonsynonymous	no	no	no
TENM4	no	5A	7 11:7836949	C>T	SNV nonsynonymous	no	yes	yes
TENM4	no	5B	7 12:5791992	C>T	SNV nonsynonymous	no	yes	yes
MBD6	no	5A	3	C>T	SNV nonsynonymous	no	no	no
CD163L1	no	5A	12:7556364 13:1144543	G>A	SNV	no	no	no
LINC00552	no	5A	04 13:2172923	C>T	upstream	no	no	no
SKA3	no	5A	9 15:5203022	C>G	UTR3	no	no	no
LYSMD2	no	5A	6 16:7547823	T>C	UTR5	no	no	no
TMEM170A	no	5A	1	G>A	UTR3	no	no	yes
FLII	no	5A	17:1814891 7	TTGGCAAAA GTCGGAGCA >T AGTGTGTGT GTGCGTGCA TGTGTGT>A	frameshift deletion	no	no	no
MLLT6	yes	5A	17:3688260 7		UTR3	no	yes	yes
GJD3	no	5A	17:3852099 9	T>C	upstream	no	yes	yes
MSI2	yes	5A	17:5570993 8	G>T	UTR3	no	no	no
MSI2	yes	5E	17:5576011 5	T>A	UTR3	no	no	no
TMEM94	no	5A	17:7345272 5	G>A	UTR5	no	no	no
LOC400655	no	5A	18:7093261 8	G>T	upstream	no	no	no
CHST8	no	5A	19:3411250 2	C>T	upstream	no	no	no
CHST8	no	6D	19:3411253 9	G>C	upstream	no	no	no
KCNK6	no	5A	19:3880952 1	C>T	upstream nonsynonymous	no	yes	yes
CA11	no	5A	19:4914229 4	A>C	SNV	no	no	no
RORC	no	5A	1:15180524 1	C>G	upstream nonsynonymous	no	no	no
KLHL20	no	5A	1:17370287 7	A>G	SNV	no	yes	yes
RGS21	no	5A	1:19233543 9	C>T	UTR3 nonsynonymous	no	no	no
GLRX2	no	5A	1:19306688 9	T>C	SNV frameshift deletion	no	no	no
KPNA6	no	5A	1:32632855 20:4780482	CCATCA>C		no	no	no
STAU1	no	5A	3 22:3948374	G>C	UTR5	no	no	no
APOBEC3G	no	5A	0 2:10601578	CT>C	UTR3	no	no	no
FHL2	no	5A	7 2:21908088	G>C	UTR5	no	no	no
ARPC2	no	5A	6 2:22651808	A>C	upstream	no	no	no
NYAP2	no	5A	2 3:12480204	G>T	UTR3	no	no	no
SLC12A8	no	5A	1	A>C	UTR3	no	no	no

EPM2AIP1	no	5A	3:37033071	G>A	nonsynonymous SNV	no	no	no
TCAIM	no	5A	3:44380139	G>C	UTR5	no	no	no
SLIT2	no	5A	4:20568893	G>A	nonsynonymous SNV	no	no	no
AFAP1-AS1	no	5A	4:7755658	T>C	upstream	no	no	no
ETF1	no	5A	5:13784224	T>C	UTR3	no	no	yes
CDH9	no	5A	5:26880909	C>T	UTR3	no	no	no
LOC285762	no	5A	6:11981315	A>G	upstream	no	no	no
MOXD1	no	5A	6:13264382	T>A	nonsynonymous SNV	no	no	no
CUL1	no	5A	7:14839504	A>C	upstream	no	no	no
COL28A1	no	5A	7:7575859	G>C	upstream	no	no	no
C9orf50	no	5A	9:13237580	C>T	nonsynonymous SNV	no	no	no
FCN2	no	5A	9:13777234	C>T	upstream	no	no	yes
OLFM1	no	5A	9:13799019	G>A	nonsynonymous SNV	no	no	no
MIR4454	no	5A	X:10911312	G>T	upstream	no	no	no
MIR4454	no	10E	5:7270101	C>T	upstream	no	yes	yes
ADGRG4	no	5A	X:13548009	A>G	nonsynonymous SNV	no	no	no
ATP11C	no	5A	X:13880923	T>A	UTR3	no	no	no
BEND2	no	5A	4	A>T	UTR3	no	no	no
PPP1R3F	no	5A	X:49126275	A>C	upstream	no	no	no
PPP1R3F	no	5B	X:49126275	A>C	upstream	no	no	no
KLF8	no	5A	X:56291846	A>T	nonsynonymous SNV	no	no	no
AMER1	yes	5A	X:63411954	C>G	nonsynonymous SNV	no	no	no
TEAD1	no	5B	11:1295976	A>G	UTR3	no	no	yes
OR52B6	no	5B	9	T>C	nonsynonymous SNV	no	no	no
NUAK1	no	5B	11:5602378	A>G	nonsynonymous SNV	no	no	no
C12orf49	no	5B	12:1064776	G>A	UTR3	no	no	no
C12orf49	no	5D	12:1171529	G>T	UTR3	no	no	no
CCNT1	no	5B	12:1171548	T>G	UTR3	no	no	no
CDK8	no	5B	12:4908447	G>C	UTR5	no	no	no
DCAF4	no	5B	6	T>C	upstream	no	no	no
NUBP1	no	5B	13:2682832	G>A	nonsynonymous SNV	no	no	no
CASKIN1;TR	no	5B	4	T>G	UTR3	no	no	no
AF7	no	5B	14:7339250	C>T	nonsynonymous SNV	no	yes	yes
GLYR1	no	5B	9	G>C	UTR5	no	yes	yes
SUMO2	no	5B	16:1085523	C>T	nonsynonymous SNV	no	no	no
CD68	no	5B	17:7317894	GA>G	upstream	no	no	no
NAPG	no	5B	1	C>A	UTR3	no	no	no
RTTN	no	5B	17:7483329					
			18:1052503					
			7					
			18:6767109					
			4					

ZNF700	no	5B	19:1203535 5	T>C	upstream	no	no	no
ZNF180	no	5B	19:4498009 0	T>C	UTR3	no	no	no
ZNF180	no	10E	19:4500451 7	A>C	UTR5	no	no	no
ZNF180	no	11A	19:4500512 1	C>A	upstream	no	no	no
FBXO42	no	5B	1:16678870	C>T	UTR5	no	no	no
ATPIF1	no	5B	1:28564412 20:5014030	A>G	nonsynonymous SNV	no	no	no
NFATC2	yes	5B	5	C>T	nonsynonymous SNV	no	no	no
LCA5L	no	5B	21:4079269 9	G>C	nonsynonymous SNV	no	no	no
MTFP1	no	5B	22:3082171 9	C>T	UTR5	no	no	no
LRP1B	yes	5B	2:14256794 1	G>A	nonsynonymous SNV	no	no	no
TTLL4	no	5B	2:21960233 5	C>T	UTR5	no	yes	yes
HNRNPLL	no	5B	2:38791029 3:12923119	A>C	UTR3	no	no	no
IFT122	no	5B	1	T>C	nonsynonymous SNV	no	no	no
AZI2	no	5B	3:28373254	T>C	UTR3	no	no	no
LRRFIP2	no	5B	3:37217805	G>C	UTR5	no	no	no
LINC02109	no	5B	5:29143081	G>A	upstream	no	no	no
ERVFRD-1	no	5B	6:11104700	G>A	nonsynonymous SNV	no	no	yes
ERVFRD-1	no	6A	6:11111923	A>G	UTR5	no	no	no
ERVFRD-1	no	6C	6:11111923 6:15892385	A>G	UTR5	no	no	no
TULP4	no	5B	0	C>T	nonsynonymous SNV	no	no	no
CD2AP	no	5B	6:47541892	C>G	nonsynonymous SNV	no	no	no
ZNF716	no	5B	7:57531534	T>C	UTR3	no	no	no
FUT10	no	5B	8:33319197 X:14629567	T>A	UTR5	no	no	no
MIR513A1	no	5B	3	G>A	upstream	no	no	no
AR	yes	5B	X:66948153	T>C	UTR3	no	no	no
AR	yes	9A	X:66788768 12:5089786	G>A	UTR5	no	no	no
DIP2B	no	5C	3	A>C	upstream	no	no	no
MLF2	no	5C	12:6857950 14:4539712	G>A	UTR3	no	no	yes
KLHL28	no	5C	9	A>G	UTR3	no	no	no
ZFYVE26	no	5C	14:6828344 0	T>G	upstream	no	no	no
RASGRP1	no	5C	15:3879552 5	G>C	nonsynonymous SNV	no	no	no
TRIM16	no	5C	17:1558797 8	C>G	upstream	no	no	no
SLC4A1	no	5C	17:4232783 9	G>A	nonsynonymous SNV	no	no	no
VEZF1	no	5C	17:5604927 5	C>T	UTR3	no	no	yes
PIEZO2	no	5C	18:1080713 3	G>A	nonsynonymous SNV	no	yes	yes
LGALS8	no	5C	1:23671151 2	T>G	UTR3	no	no	no
XBP1	no	5C	22:2919710 0	C>T	upstream	no	no	no

XPO1	yes	5C	2:61717857	T>C	nonsynonymous SNV	no	no	no
				AGGGGGCG				
TSGA10	no	5C	2:99771406	G>A	UTR5	no	no	no
			3:12670773		nonsynonymous SNV			
PLXNA1	no	5C	1	C>A	SNV	no	no	no
PPARGC1A	no	5C	4:23815869	T>A	stopgain	no	no	no
MUC21	no	5C	6:30950699	A>T	upstream	no	no	no
			7:14164603		nonsynonymous SNV			
CLECSA	no	5C	7	C>T	SNV	no	no	no
					nonsynonymous SNV			
MACC1	yes	5C	7:20198167	A>G	SNV	no	yes	yes
					nonsynonymous SNV			
SFRP4	yes	5C	7:37955812	G>A	SNV	no	no	no
CLDN4	no	5C	7:73246188	TCTG>T	UTR3	no	no	no
			X:11051468					
CAPN6	no	5C	2	G>T	upstream	no	no	yes
			X:11112523					
TRPC5OS	no	5C	7	G>A	UTR5	no	no	yes
			X:11974901					
MCTS1	no	5C	9	G>A	UTR3	no	no	no
ABCB7	no	5C	X:74376522	C>A	upstream	no	no	no
			10:1265242					
ABRAXAS2	no	5D	74	GGCACA>G	UTR3	no	no	no
			11:5590521					
OR8J3	no	5D	7	G>T	upstream	no	no	no
			11:7182373					
ANAPC15	no	5D	3	G>A	UTR5	no	no	no
			12:2263768		nonsynonymous SNV			
C2CD5	no	5D	9	T>C	SNV	no	no	no
			13:1098593					
MYO16	no	5D	64	G>A	UTR3	no	no	no
			15:4496953					
PATL2	no	5D	8	C>T	upstream	no	no	no
			16:6829318		nonsynonymous SNV			
PLA2G15	no	5D	1	C>A	SNV	no	no	no
			16:6828327		nonsynonymous SNV			
PLA2G15	no	10B	6	A>G	SNV	no	no	no
			16:7083623					
HYDIN	no	5D	9	C>T	UTR3	no	yes	yes
			16:7083632					
HYDIN	no	7C	1	G>A	UTR3	no	no	no
			16:7095474		nonsynonymous SNV			
HYDIN	no	11A	8	G>A	SNV	no	yes	yes
			19:1827383		nonsynonymous SNV			
PIK3R2	no	5D	0	G>A	SNV	no	no	no
NBPF8;NBP			1:14483036					
F9	no	5D	7	G>T	UTR3	no	no	yes
			1:16936516					
BLZF1	no	5D	4	A>C	UTR3	no	no	no
			22:4917514					
MIR4535	no	5D	1	G>A	upstream	no	no	no
			2:12204274					
TFCP2L1	no	5D	9	G>A	UTR5	no	no	yes
			3:14870880					
GYG1	no	5D	6	A>C	upstream	no	no	no
			5:13872893		nonsynonymous SNV			
PROB1	no	5D	7	C>A	SNV	no	no	no
CAST	no	5D	5:95997743	G>A	UTR5	no	no	no
			9:11563189					
SNX30	no	5D	6	G>GT	UTR3	no	yes	yes
			X:10026441					
TRMT2B	no	5D	5	T>C	UTR3	no	no	no
			X:10074371		nonsynonymous SNV			
ARMCX4	no	5D	4	G>T	SNV	no	no	no
ARMCX5-								
GPRASP2;B			X:10200518		nonsynonymous SNV			
HLHB9	no	5D	2	C>T	SNV	no	no	yes

HTR2C	no	5D	X:11381877 7	C>T	UTR5	no	no	no
AVPR2	no	5D	X:15317256 0	G>A	UTR3	no	yes	yes
RPS6KA3	no	5D	X:20285380	G>A	upstream nonsynonymous	no	no	yes
BCOR	yes	5D	X:39930322 10:1027435	G>T	SNV nonsynonymous	no	no	no
MRPL43;SE MA4G	no	5E	42	C>T	SNV	no	yes	yes
MRPL43;SE MA4G	no	5F	10:1027435 42	C>T	nonsynonymous SNV	no	yes	yes
FRMD4A	no	5E	10:1368859 9	C>T	UTR3	no	yes	yes
FRMD4A	no	5F	10:1368859 9	C>T	UTR3	no	yes	yes
RPL27A	no	5E	11:8707514 12:1253992	T>G	UTR3	no	no	no
UBC	no	5E	74	G>A	UTR5	no	no	no
UBC	no	5F	12:1253992 74	G>A	UTR5	no	no	no
NCKAP1L	no	5E	12:5493669 7	G>A	UTR3	no	no	no
C1QTNF8	no	5E	16:1140117	C>T	UTR3	no	no	no
UBE2I	no	5E	16:1375711 17:4294524	C>T	UTR3 nonsynonymous	no	no	yes
EFTUD2	no	5E	6	C>T	SNV	no	no	no
GFAP	no	5E	17:4298734 8	C>T	UTR3	no	no	no
SPRR2A	no	5E	1:15303030 5	T>A	upstream nonsynonymous	no	no	no
PRRX1	yes	5E	1:17068890 9	G>A	SNV	no	no	yes
PRRX1	yes	5F	1:17068890 9	G>A	nonsynonymous SNV	no	no	yes
BLACAT1	no	5E	1:20542525 8	A>C	upstream nonsynonymous	no	no	no
MAP10	no	5E	1:23294142 8	C>T	SNV	no	no	no
MAP10	no	5F	1:23294142 8	C>T	nonsynonymous SNV	no	no	no
ACTRT2	no	5E	1:2938558 20:6267928	G>C	nonsynonymous SNV	no	no	no
SOX18	no	5E	6	G>C	UTR3	no	no	no
PITX2	no	5E	4:11156371 4	G>A	upstream	no	no	no
SH3RF2	no	5E	5:14544275 0	T>A	UTR3	no	no	no
SH3RF2	no	11B	5:14531525 5	C>T	upstream	no	no	no
LOC101926 960	no	5E	5:42175987	T>C	upstream nonsynonymous	no	no	no
AGGF1	no	5E	5:76358994 6:13115720	C>T	SNV	no	yes	yes
SMLR1	no	5E	2	A>G	UTR3	no	no	no
CDKL5	no	5E	X:18651401	T>G	UTR3	no	no	no
CDKL5	no	5F	X:18651401	T>G	UTR3	no	no	no
CDKL5	no	7B	X:18650182	T>C	UTR3	no	no	no
LANCL3	no	5E	X:37430085	G>A	upstream	no	no	no
LANCL3	no	5F	X:37430085 10:7263799	G>A	upstream	no	no	no
SGPL1	no	5F	4	C>T	UTR3	no	no	no
LRRC4C	no	5F	11:4013682 7	C>G	nonsynonymous SNV	no	no	no

CCDC60	no	5F	12:1199785 74	G>A	UTR3	no	yes	yes
TMED2	no	5F	12:1240825 21	C>T	UTR3	no	no	no
KIF21A	no	5F	12:3975116 0	T>C	nonsynonymous SNV	no	no	no
ASB8	no	5F	12:4855151 0	C>G	upstream	no	no	yes
COL4A1	no	5F	13:1108146 15	T>A	nonsynonymous SNV	no	no	no
CAB39L	no	5F	13:4995111 9	T>C	nonsynonymous SNV	no	yes	yes
EXOC3L4	no	5F	14:1035687 24	G>T	stopgain	no	no	no
MIR8071- 1;MIR8071- 2	no	5F	14:1061057 17	G>A	upstream	no	no	yes
MIR8071- 1;MIR8071- 2	no	5G	14:1061057 17	G>A	upstream	no	no	yes
KATNB1	no	5F	16:5778560 8	C>T	nonsynonymous SNV	no	yes	yes
KATNB1	no	8D	16:5778907 3	G>A	nonsynonymous SNV	no	no	no
LINC02089	no	5F	17:5093943 5	T>C	upstream	no	no	no
NDUFAF8	no	5F	17:7921314 2	A>AG	UTR5	no	yes	yes
LOC101927 168	no	5F	18:6730492	A>G	upstream	no	no	no
VAPA	no	5F	18:9913578 19:1472168	A>G	upstream	no	no	no
CLEC17A	no	5F	9	G>C	UTR3	no	no	no
TRIM11	no	5F	1:22858243 7	C>T	nonsynonymous SNV	no	yes	yes
NRDC	no	5F	1:52290977 20:5510003	T>C	nonsynonymous SNV	no	no	yes
FAM209A	no	5F	8	C>G	nonsynonymous SNV	no	no	no
GNAS	yes	5F	20:5742812 1	T>C	UTR5	no	yes	yes
PES1	no	5F	22:3097606 5	C>T	nonsynonymous SNV	no	no	no
EPB41L5	no	5F	2:12085084 3	A>G	nonsynonymous SNV	no	no	no
CDK15	no	5F	2:20276025 8	C>T	UTR3	no	no	no
ASXL2	yes	5F	TGGGATTCA TAGCTTGGC TAGCA>T		frameshift deletion	no	no	no
KCNK3	no	5F	2:25965080	G>T	UTR3	no	no	no
KCNK3	no	10C	2:26954028	A>T	UTR3	no	no	no
LCLAT1	no	5F	2:26952725	CT>C	UTR3	no	no	no
SPTBN1	no	5F	2:30866643	C>CT	UTR3	no	no	no
STAMPB	no	5F	2:54887375	A>G	UTR3	no	no	no
MAT2A	no	5F	2:74090399	T>A	nonsynonymous SNV	no	no	no
CCDC191	no	5F	2:85766424 3:11372967	C>G	nonsynonymous SNV	no	no	no
CNTN6	no	5F	1	G>A	nonsynonymous SNV	no	yes	yes
SLC12A7	no	5F	3:1444012	A>T	nonsynonymous SNV	no	no	no
PCDHGA8	no	5F	5:1053590 5:14077222	G>A	UTR5	no	no	no
MRPL22	no	5F	3 5:15431968 1	G>C	upstream	no	no	no

SERINC5	no	5F	5:79407328	G>A	UTR3 nonsynonymous	no	no	no
BAG6	no	5F	6:31615399 7:10740156	G>C	SNV	no	no	no
CBLL1	no	5F	4 7:13465520	T>A	UTR3	no	no	no
CALD1	no	5F	9 8:14462069	T>C	UTR3 nonsynonymous	no	no	no
ZC3H3	no	5F	5	C>A	SNV	no	no	no
DOK2	no	5F	8:21771743	CAGAG>C	upstream nonsynonymous	no	no	yes
ZNF367	no	5F	9:99150616 X:10596995	C>T	SNV	no	no	no
RNF128	no	5F	4 X:13022063	T>A	UTR5 nonsynonymous	no	no	no
ARHGAP36	no	5F	0	T>C	SNV	no	no	no
SLC25A6	no	5F	X:1511237	A>G	upstream	no	no	yes
SCML1	no	5F	X:17771889	A>G	UTR3 nonsynonymous	no	no	no
FAM47C	yes	5F	X:37028209	G>A	SNV	no	yes	yes
TSPAN7	no	5F	X:38419932	C>T	upstream	no	no	no
KIF6	no	5G	6:39301539	G>A	UTR3	no	yes	yes
KIF6	no	7B	6:39658696	T>A	UTR3	no	no	no
KIF6	no	7C	6:39658696	T>A	UTR3	no	no	no
GPR82	no	5G	X:41583462 10:1327616	A>G	UTR5	no	no	no
MIR378C	no	6A	20 10:1327616	G>A	upstream	no	yes	yes
MIR378C ACBD7;ACB D7-	no	6C	20 10:1513164	G>A	upstream	no	yes	yes
DCLRE1CP1 ACBD7;ACB D7-	no	6A	5 10:1513164	T>C	upstream	no	yes	yes
DCLRE1CP1	no	6C	5 10:4773941	T>C	upstream	no	yes	yes
FAM25BP	no	6A	5 10:4773941	G>T	upstream	no	no	no
FAM25BP	no	6C	5 11:7416778	G>T	upstream	no	no	no
KCNE3	no	6A	7	T>C	UTR3	no	no	no
OR5P3	no	6A	11:7847557	T>C	upstream	no	no	no
OR5P3	no	6C	11:7847557 13:3739326	T>C	upstream	no	no	no
RFXAP LOC105370 306	no	6A	8 13:8879489	T>G	upstream	no	no	no
USP31	no	6A	2 16:2311676	G>A	upstream nonsynonymous	no	no	no
USP31	no	6A	6 16:2311676	T>C	SNV nonsynonymous	no	no	no
USP31	no	6C	6 16:6937345	T>C	SNV nonsynonymous	no	no	no
COG8	no	6A	5	T>C	SNV nonsynonymous	no	yes	yes
ITGAE PNPO;SP2- AS1	no	6A	17:3658545 17:4601885	A>G	SNV	no	no	no
PNPO;SP2- AS1	no	6A	0 17:4601885	C>G	upstream	no	no	no
PNPO;SP2- AS1	no	6C	0 18:2940968	C>G	upstream	no	no	no
TRAPPC8	no	6A	5 18:2940968	A>C	UTR3	no	no	no
TRAPPC8	no	6C	5	A>C	UTR3	no	no	no

LINC01801	no	6A	19:3532472 2	G>C	upstream	no	no	no
LINC01801	no	6C	19:3532472 2	G>C	upstream	no	no	no
ZNF382	no	6A	19:3711889 2	T>G	UTR3	no	no	no
ZNF382	no	6C	19:3711889 2	T>G	UTR3	no	no	no
SBK2	no	6A	19:5604872 5	A>C	upstream nonsynonymous SNV	no	no	no
ANKRD65	no	6A	1:1356352	C>T	SNV	no	no	yes
NASP	no	6A	1:46049299	T>G	upstream	no	no	no
NASP	no	6C	1:46049299	T>G	upstream nonsynonymous SNV	no	no	no
NASP	no	11B	1:46080730	T>A	SNV	no	no	no
NASP	no	11D	1:46080730	T>A	SNV	no	no	no
OMA1	no	6A	1:59012536	C>CG	upstream	no	no	no
OMA1	no	6C	1:59012536	C>CG	upstream	no	no	no
AAK1;ANXA 4	no	6A	2:69870937	G>A	UTR5	no	no	yes
AAK1;ANXA 4	no	6C	2:69870937	G>A	UTR5	no	no	yes
RPIA	no	6A	2:88990633	C>G	upstream	no	no	no
RPIA	no	6C	2:88990633	C>G	upstream	no	no	no
RPL24	no	6A	3:10140581 8	C>T	upstream	no	no	yes
RPL24	no	6C	3:10140581 8	C>T	upstream frameshift deletion	no	no	yes
NECTIN3	no	11D	3:11086374 5	GA>G	deletion	no	no	no
BDH1	no	6A	3:19723738 0	A>AGACCCT GCACTTG	UTR3	no	no	no
BDH1	no	6C	3:19723738 0	A>AGACCCT GCACTTG	UTR3	no	no	no
UGT8	no	6A	4:11559852 4	A>G	UTR3	no	no	no
UGT8	no	6C	4:11559852 4	A>G	UTR3	no	no	no
MAEA	no	6A	4:1282768	T>G	upstream	no	no	no
MAEA	no	6C	4:1282768	T>G	upstream	no	no	no
ABLM2	no	6A	4:8009622	A>C	UTR3	no	no	no
ABLM2	no	6C	4:8009622	A>C	UTR3	no	no	no
HNRNPA0	no	7D	5:13709000 9	G>T	UTR5	no	no	yes
SYNPO	no	6A	5:15003700 4	A>T	UTR3	no	yes	yes
ZNF354B	no	8C	5:17828667 4	A>T	upstream	no	no	no
CANX	no	6A	5:17912579 4	C>G	upstream	no	no	no
CANX	no	6C	5:17912579 4	C>G	upstream	no	no	no
LATS1	yes	6A	6:14998013 0	T>C	UTR3	no	no	no
SENP6	no	6A	6:76310786	C>T	upstream	no	no	no
SENP6	no	6C	6:76310786	C>T	upstream	no	no	no
PTN	no	6A	7:13691258 0	C>T	UTR3	no	no	yes
PTN	no	6C	7:13691258 0	C>T	UTR3	no	no	yes

KMT2C	yes	6A	7:15185148 0	G>A	nonsynonymous SNV	no	no	no
KMT2C	yes	6B	7:15187405 2	G>A	nonsynonymous SNV	no	no	no
KMT2C	yes	6C	7:15185148 0	G>A	nonsynonymous SNV	no	no	no
KMT2C	yes	6D	7:15187405 2	G>A	nonsynonymous SNV	no	no	no
KMT2C	yes	9C	7:15184610 9	G>GTTC	nonframeshift insertion	no	no	no
LSM5	no	6A	7:32525564	A>C	UTR3	no	yes	yes
LSM5	no	6E	7:32525564	A>C	UTR3	no	yes	yes
FBXL18	no	6A	7:5515581	G>C	UTR3	no	no	no
FBXL18	no	6C	7:5515581	G>C	UTR3	no	no	no
MIOS	no	6A	7:7607671	T>G	UTR5	no	no	no
MIOS	no	6C	7:7607671	T>G	UTR5	no	no	no
GABBR2	no	6A	9:10147080 0	C>T	nonsynonymous SNV	no	yes	yes
NIPSNAP3B	no	6A	9:10752538 3	G>A	upstream	no	yes	yes
FAM206A	no	6A	9:11170251 1	T>G	UTR3	no	no	no
FAM206A	no	6C	9:11170251 1	T>G	UTR3	no	no	no
NAA16	no	6B	13:4188476 8	T>C	upstream	no	no	no
NAA16	no	6D	13:4188476 8	T>C	upstream	no	no	no
FAM234A	no	6B	16:284150	C>T	upstream	no	yes	yes
FAM234A	no	6D	16:284150 17:3486902	C>T	upstream	no	yes	yes
MYO19	no	6B	17:3486902 1	C>T	UTR3	no	no	no
MYO19	no	6D	17:3486902 1	C>T	UTR3	no	no	no
RUNDC3A	no	6B	17:4239377 3	A>T	nonsynonymous SNV	no	no	no
RUNDC3A	no	6D	17:4239377 3	A>T	nonsynonymous SNV	no	no	no
KCNJ16	no	6B	17:6812926 4	T>A	nonsynonymous SNV	no	no	no
KCNJ16	no	6D	17:6812926 4	T>A	nonsynonymous SNV	no	no	no
CNDP2	no	6B	18:7218853 0	G>A	UTR3	no	yes	yes
CNDP2	no	6D	18:7218853 0	G>A	UTR3	no	yes	yes
DOHH	no	6B	19:3490966	A>C	UTR3	no	no	no
DHRS3	no	6B	1:12676728	C>T	UTR5	no	no	no
DHRS3	no	6D	1:12676728 1:15590488	C>T	UTR5	no	no	no
KIAA0907	no	6B	1:15590488 6	A>T	upstream	no	no	no
KIAA0907	no	6D	1:15590488 6	A>T	upstream	no	no	no
PIK3CD	no	6B	1:9788571	A>G	UTR3	no	no	no
VPS16	no	6B	20:2845925	AGAGCAGC> A	frameshift deletion	no	no	no
VPS16	no	6D	20:2845925 20:4303483	AGAGCAGC> A	frameshift deletion	no	no	no
HNF4A	no	6B	20:4303483 5	C>T	nonsynonymous SNV	no	yes	yes
HNF4A	no	6D	20:4303483 5	C>T	nonsynonymous SNV	no	yes	yes

MAFF	no	6B	22:3861056 9	A>G	nonsynonymous SNV	no	yes	yes
MAFF	no	6D	22:3861056 9	A>G	nonsynonymous SNV	no	yes	yes
MIR4444- 1;MIR4444- 2	no	6B	3:75262783	G>T	upstream	no	no	no
MIR4444- 1;MIR4444- 2	no	6D	3:75262783	G>T	upstream	no	no	no
FGFBP2	no	6B	4:15962041	C>G	UTR3	no	no	no
FGFBP2	no	6D	4:15962041	C>G	UTR3	no	no	no
FAM13A	no	6B	4:89978410 5:14923780	T>C	upstream	no	no	no
PDE6A	no	6B	4	C>T	UTR3	no	no	no
OSMR	no	6B	5:38935498	C>T	UTR3	no	no	yes
OSMR	no	6D	5:38935498 6:16906810	C>T	UTR3	no	no	yes
SMOC2	no	6B	6	C>T	UTR3	no	yes	yes
NEIL2	no	6B	8:11627600	A>C	UTR5	no	no	no
EXTL3	no	6B	8:28595181	G>GT	splicing	no	no	no
EXTL3	no	6D	8:28595181	G>GT	splicing	no	no	no
ERMP1	no	6B	9:5833356	A>T	upstream	no	no	no
ERMP1	no	6D	9:5833356 12:1222182	A>T	upstream	no	no	no
TMEM120B ARHGAP5;A RHGAP5- AS1	no	6C	31	T>C	UTR3	no	no	no
ARHGAP5;A RHGAP5- AS1	no	6C	14:3254616 2	TTC>T	upstream	no	yes	yes
ARHGAP5;A RHGAP5- AS1	no	10E	14:3254621 2	C>T	upstream	no	no	yes
ZNF598	no	6C	16:2060543 16:2895057	T>G	upstream	no	no	no
CD19	no	6C	4 19:1325090	AGTGT>A	UTR3	no	yes	yes
NACC1 SMIM7;TME M38A	no	6C	9 19:1677126	C>G	UTR3	no	no	no
M38A	no	6C	2 19:4119636	C>T	upstream	no	no	no
NUMBL	no	6C	3	A>C	splicing frameshift deletion	no	no	no
ZNF513	no	6C	2:27600926	TAGTG>T	nonsynonymous SNV	no	no	no
ZDHHC3	no	6C	3:45000672 4:16308541	A>G	SNV	no	no	no
FSTL5	no	6C	9	G>A	upstream	no	no	no
SLC34A2	yes	6C	4:25658157	C>T	UTR5	no	no	no
PRLR	no	6C	5:35060767	A>T	UTR3	no	no	no
PRLR	no	7B	5:35065056	G>A	UTR3	no	no	no
PRLR ADAMTS16; CTD- 2297D10.2	no	7C	5:35065056	G>A	UTR3	no	no	no
MIR548AE2	no	6C	5:5140381	A>C	upstream	no	no	no
	no	6C	5:57826536	G>C	upstream	no	yes	yes
				GGCAGCCGC GGCTGCTGC CGCGGCCGC	nonframeshift deletion	no	yes	yes
SP8	no	6C	7:20825145	C>G	deletion	no	yes	yes
TMEM130	no	6C	7:98445141	G>C	UTR3	no	yes	yes

ZNF789	no	6C	7:99078863	T>G	UTR3 nonsynonymous	no	no	no
SLC39A14	no	11C	8:22267500 10:4369731	G>A	SNV	no	no	no
RASGEF1A	no	6D	2 10:7428359	C>A	stopgain nonsynonymous	no	no	no
MICU1	no	6D	6	T>C	SNV nonsynonymous	no	no	yes
A2M	no	6D	12:9258873	C>T	SNV	no	no	no
PPP1R8	no	6D	1:28157727	T>G	UTR5	no	no	no
LINC01814	no	6D	2:8723961 4:13268570	G>A	upstream	no	no	yes
SNHG27	no	6D	3	G>C	upstream	no	no	yes
C9orf47	no	6D	9:91607353 1:14800425	G>A	UTR3	no	no	no
NBPF26	no	6E	2 10:1177071	C>A	UTR3	no	yes	yes
ATRNL1	no	7A	42 10:1177071	T>G	UTR3	no	no	no
ATRNL1	no	7D	42	T>G	UTR3	no	no	no
KLF6	yes	7A	10:3827777	C>T	upstream	no	no	yes
KLF6	yes	7D	10:3827777 10:5995189	C>T	upstream	no	no	yes
IPMK	no	7A	1 10:5995189	A>G	UTR3	no	no	no
IPMK	no	7D	1	A>G	UTR3	no	no	no
LARP4B	yes	7A	10:855570 11:1183742	A>T	UTR3 nonsynonymous	no	no	no
KMT2A	yes	7A	07 11:1183742	A>G	SNV nonsynonymous	no	no	no
KMT2A	yes	7D	07 12:5010128	A>G	SNV	no	no	no
FMNL3	no	7A	5 12:5291382	A>C	upstream nonsynonymous	no	no	yes
KRT5	no	7A	7 14:1059924	T>C	SNV	no	yes	yes
TMEM121	no	7A	42 14:2386342	C>G	upstream nonsynonymous	no	no	no
MYH6	no	7A	9 14:5172357	C>T	SNV	no	yes	yes
TMX1	no	7A	2 15:6567129	A>G	UTR3	no	no	yes
IGDCC3	no	7A	8 15:6567129	T>C	upstream	no	no	yes
IGDCC3	no	7D	8	T>C	upstream	no	no	yes
LOC105372	no	7A	19:5129012 6	C>T	upstream	no	yes	yes
440	no	7A	19:5625043 7	C>A	upstream	no	no	no
NLRP9	no	7A	19:5625043 7	C>A	upstream	no	no	no
NLRP9	no	7D	1:10144366 3	C>A	UTR3	no	no	no
SLC30A7	no	7A	1:10144366 3	C>A	UTR3	no	no	no
SLC30A7	no	7D	1:11430657 6	C>A	UTR3	no	yes	yes
RSBN1	no	7A	1:15447542 4	T>A	UTR3	no	no	no
TDRD10	no	7A	1:15940929 9	A>G	UTR5	no	no	no
OR10J1	no	7A	1:79110367 20:2128299	G>A	upstream	no	no	no
IFI44L	no	7A	3 2:21992519	C>T	UTR3	no	yes	yes
XRN2	no	7A	3	C>T	upstream	no	yes	yes
IHH	no	7A	3	CGGGGA>C	UTR5	no	no	no

IHH	no	7D	2:21992519 3	CGGGGA>C	UTR5	no	no	no
CLIP4	no	7A	2:29405146	T>A	UTR3	no	no	no
CLIP4	no	7D	2:29405146 3:12016995	T>A	UTR3	no	no	no
FSTL1	no	7A	4	T>C	upstream nonsynonymous SNV	no	no	no
CRYBG3	no	7A	3:97595105 4:11917709	A>T	SNV	no	no	no
NDST3	no	7A	5	T>C	UTR3	no	no	no
NDST3	no	7D	4:11917709 5	T>C	UTR3	no	no	no
KIAA1109	no	7A	4:12316074 1	C>T	nonsynonymous SNV	no	no	no
NSUN7	no	7A	4:40778258	A>G	nonsynonymous SNV	no	no	no
NSUN7	no	7D	4:40778258	A>G	nonsynonymous SNV	no	no	no
ADAMTS3	no	7A	4:73149203 5:17651397	C>A	nonsynonymous SNV	no	no	no
FGFR4	yes	7A	5	G>C	UTR5	no	no	no
FGFR4	yes	7D	5:17651397 5	G>C	UTR5	no	no	no
IL6ST	yes	7A	5:55232181	T>G	UTR3	no	yes	yes
ADGRV1	no	7A	5:90002110 6:14788568	G>A	nonsynonymous SNV	no	no	no
SAMD5	no	7A	4	A>G	UTR3	no	no	no
SAMD5	no	8D	6:14783046 2	T>C	nonsynonymous SNV	no	yes	yes
KDM7A	no	7A	7:13979655 7	A>C	nonsynonymous SNV	no	no	no
KDM7A	no	7D	7:13979655 7	A>C	nonsynonymous SNV	no	no	no
IGFBP3	no	7A	7:45960933 8:14612738	A>C	upstream	no	no	no
ZNF250	no	7A	5	G>C	upstream nonsynonymous SNV	no	no	no
ADAM28	no	7A	8:24199262 9:11782211	G>A	SNV	no	no	yes
TNC	yes	7A	3	G>C	nonsynonymous SNV	no	no	no
TNC	yes	7D	9:11782211 3	G>C	nonsynonymous SNV	no	no	no
PCYT1B	no	7A	X:24597560	G>A	nonsynonymous SNV	no	no	no
PRKX	no	7A	X:3526113	A>T	UTR3	no	no	no
PRKX	no	7D	X:3526113	A>T	UTR3	no	no	no
EDA	no	7A	X:69080743	G>T	UTR3	no	no	no
RLIM	no	7A	X:73803653 10:1327632	C>A	UTR3	no	no	no
UCMA	no	7B	2	G>T	UTR5	no	no	no
ZNF37A	no	7B	10:3838264 3	T>G	upstream	no	no	no
REEP3	no	7B	10:6538377 6	TC>T	UTR3	no	no	no
C11orf87	no	7B	11:1092983 06	T>A	UTR3	no	no	no
FNBP4	no	7B	11:4775415 5	G>C	nonsynonymous SNV	no	no	no
TRIM51	no	7B	11:5565560 7	A>G	nonsynonymous SNV	no	no	no
THAP12	no	7B	11:7606262 7	C>T	nonsynonymous SNV	no	no	yes
KDM2B	no	7B	12:1218669 71	A>G	UTR3	no	no	yes

KDM2B	no	11A	12:1218800 91	C>CG	frameshift insertion	no	no	no
CCDC62	no	7B	12:1232582 17	C>CT	upstream	no	no	no
CCDC62	no	7C	12:1232582 17	C>CT	upstream	no	no	no
PDE3A	no	7B	12:2083694 2	C>G	UTR3	no	no	no
ALX1	no	7B	12:8567426 5	G>A	nonsynonymous SNV	no	no	yes
TMPO	no	7B	12:9892797 2	C>G	nonsynonymous SNV	no	no	no
BORA	no	7B	13:7332795 8	AGGTAC>A	frameshift deletion	no	no	no
FBXL3	no	7B	13:7758075 5	C>T	UTR3	no	no	no
ZNF839	no	7B	14:1027834 25	A>C	upstream	no	no	yes
ZNF839	no	7C	14:1027834 25	A>C	upstream	no	no	yes
NDRG2	no	7B	14:2149320 9	C>T	UTR5	no	no	no
SSTR1	no	7B	14:3868188 6	A>T	UTR3	no	no	no
EEF2K	no	7B	16:2229841 8	G>T	UTR3	no	no	no
EEF2K	no	7C	16:2229841 8	G>T	UTR3	no	no	no
KREMEN2	no	7B	16:3017833 16:3104535	A>C	nonsynonymous SNV	no	no	no
STX4	no	7B	16:3104535 2	G>A	nonsynonymous SNV	no	no	no
STX4	no	7C	16:3104535 2	G>A	nonsynonymous SNV	no	no	no
C16orf78	no	7B	16:4940788 7	C>T	nonsynonymous SNV	no	yes	yes
MT1E	no	7B	16:5665973 0	A>T	UTR5	no	no	no
MT1E	no	7C	16:5665973 0	A>T	UTR5	no	no	no
TMEM186	no	7B	16:8889500 17:2668463	TCA>T	UTR3	no	no	no
TMEM199	no	7B	17:2668463 1	A>G	UTR5	no	no	yes
TMEM199	no	7C	17:2668463 1	A>G	UTR5	no	no	yes
PFAS	no	7B	17:8152533 19:1420337	A>C	upstream	no	no	no
PRKACA	yes	7B	19:3573880 5	ATTG>A	UTR3	no	yes	yes
LSR	no	7B	19:4229378 5	C>T	upstream	no	no	no
EBI3	no	7B	19:6737732 1:15158465	T>G	upstream	no	no	no
GPR108;MI R6791	no	7B	1:15158465 2	T>C	upstream	no	no	yes
SNX27	no	7B	1:15306768 8	G>C	upstream	no	no	no
SPRR2E	no	7B	1:15598964 1:22086373	C>T	upstream nonsynonymous SNV	no	no	no
FHAD1	no	7B	1:22086373 6	A>G	UTR5	no	no	yes
C1orf115	no	7B	1:22783428 0	C>T	nonsynonymous SNV	no	no	no
ZNF678	no	7B	1:22783428 0	C>G	nonsynonymous SNV	no	no	no
ZNF678	no	7C	1:23085070 4	C>G	SNV	no	no	no
AGT	no	7B	1:24792144 9	G>A	upstream nonsynonymous SNV	no	yes	yes
OR1C1	no	7B		G>C		no	no	no

RAB3B	no	7B	1:52376800	A>C	UTR3	no	no	no
RAB3B	no	11A	1:52377792 20:6216879	CTTT>C	UTR3	no	no	yes
PTK6	yes	7B	4 22:4301504	C>G	upstream	no	no	no
CYB5R3	no	7B	9 22:5088305	G>C	UTR3	no	no	no
PPP6R2	no	7B	4 22:5088305	TTTTG>T	UTR3	no	no	yes
PPP6R2	no	7C	4 2:24187263	TTTTG>T	UTR3 frameshift deletion	no	no	yes
CROCC2	no	7B	7	AC>A	deletion	no	no	no
DYNC1LI1	no	7B	3:32613269	C>A	upstream	no	no	no
STAC	no	7B	3:36421065	A>G	upstream	no	no	no
STAC	no	7C	3:36421065	A>G	upstream nonsynonymous	no	no	no
TTLL3	no	7B	3:9876782 4:10689031	C>T	SNV	no	no	yes
NPNT	no	7B	9 4:15828280	T>A	UTR3	no	no	no
GRIA2	no	7B	5 4:15828280	G>T	splicing	no	no	no
GRIA2	no	7C	5 4:19080225	G>T	splicing	no	no	no
LINC01596	no	7B	7	T>G	upstream	no	no	yes
DROSHA	yes	7B	5:31532168 6:13430908	G>C	UTR5	no	no	no
SLC2A12	no	7B	7 6:13430908	T>A	UTR3	no	no	no
SLC2A12	no	7C	7 7:13823961	T>A	UTR3 nonsynonymous	no	no	no
TRIM24	yes	7B	5 7:13823961	G>A	SNV nonsynonymous	no	no	no
TRIM24	yes	7C	5 7:13928173	G>A	SNV nonsynonymous	no	no	no
HIPK2	no	7B	9 7:13928173	A>G	SNV nonsynonymous	no	no	no
HIPK2	no	7C	9	A>G	SNV	no	no	no
PKD1L1	no	7B	7:47814348	C>A	UTR3	no	no	no
FOXK1	no	7B	7:4803863 8:11396002	T>G	UTR3 nonsynonymous	no	no	no
CSMD3	yes	7B	9 8:14221801	A>C	SNV	no	no	no
SLC45A4	no	7B	7 8:14221801	G>A	UTR3	no	no	no
SLC45A4	no	7C	7	G>A	UTR3	no	no	no
RPL30	no	7B	8:99057923 9:12613307	C>T	upstream nonsynonymous	no	no	yes
CRB2	no	7B	5 9:12613307	G>C	SNV nonsynonymous	no	no	no
CRB2	no	7C	5 9:13580429	G>C	SNV	no	no	no
TSC1	yes	7B	2	G>A	UTR5 nonsynonymous	no	yes	yes
UHRF2	no	7B	9:6421114	T>G	SNV nonsynonymous	no	no	no
UHRF2	no	7C	9:6421114 X:12304652	T>G	SNV	no	no	no
XIAP	no	7B	8 X:12304652	C>A	UTR3	no	no	no
XIAP	no	7C	8 X:14507918	C>A	UTR3	no	no	no
MIR892A;M IR892B	no	7B	5 X:15248651	A>T	upstream	no	no	no
MAGEA1	no	7B	7	G>A	upstream	no	no	no

FAM3A	no	7B	X:15373484 8	C>T	UTR3	no	no	yes
FAM3A	no	7C	X:15373484 8	C>T	UTR3 nonsynonymous	no	no	yes
ARSE	no	7B	X:2867516	A>T	SNV	no	no	no
CASK	no	7B	X:41374239	A>G	UTR3	no	no	no
CASK	no	11B	X:41378840	G>T	UTR3	no	no	no
CASK	no	11D	X:41378840	G>T	UTR3	no	no	no
KCND1	no	7B	X:48818893	C>T	UTR3	no	no	no
KCND1	no	7C	X:48818893	C>T	UTR3	no	no	no
TSPYL2	no	7B	X:53111318	G>A	upstream	no	no	no
GNL3L	no	7B	X:54592257 12:1637747	GTT>G	UTR3 nonsynonymous	no	no	yes
SLC15A5	no	7C	1 12:2795187	G>A	SNV	no	no	no
KLHL42	no	7C	7 17:6083725	G>A	UTR3 nonsynonymous	no	no	no
MARCH10	no	7C	7 19:4761573	T>A	SNV nonsynonymous	no	no	yes
ZC3H4	no	7C	8 1:15269189	T>G	SNV	no	no	no
C1orf68	no	7C	0	G>T	upstream	no	no	no
ATP13A2	no	7C	1:17331568 1:21175251	C>A	splicing	no	no	no
SLC30A1	no	7C	0	C>G	upstream nonsynonymous	no	yes	yes
ELAVL4	no	7C	1:50661247	G>A	SNV nonsynonymous	no	no	no
MIER1	no	7C	1:67447527 22:3800525	C>T	SNV	no	no	no
GGA1	no	7C	6 2:23476355	C>A	UTR5	no	no	no
HJURP	no	7C	6	A>T	upstream	no	no	no
CCND3	yes	7C	6:41903734	TGCTGGAGC CCCGGGGG GCTTTGGGC GCTGG>T CTGGAGCCC CGGGGGGCT	nonframeshift deletion	no	no	no
CCND3	yes	11D	6:41903736	T>C	nonframeshift deletion	no	no	no
ARHGEF39	no	7C	9:35661727 10:1051984	T>C	UTR3 nonsynonymous	no	no	no
PDCD11	no	7D	63 10:7464710	G>A	SNV	no	no	no
MCU	no	7D	7 12:2791967	C>T	UTR3 nonsynonymous	no	no	yes
MANSC4	no	7D	9	A>G	SNV nonsynonymous	no	no	no
CLSTN3	no	7D	12:7288060 13:9199996	C>T	SNV	no	no	yes
MIR17HG	no	7D	6 14:1050580	G>T	upstream	no	yes	yes
TMEM179	no	7D	33 14:6951943	C>T	UTR3	no	no	no
DCAF5	no	7D	9 14:7778721	C>A	UTR3	no	no	no
POMT2	no	7D	8 15:4481931	A>C	UTR5	no	no	no
CTDSPL2	no	7D	0	G>A	UTR3 nonsynonymous	no	no	no
C16orf96	no	7D	16:4644726 16:6497961	G>A	SNV	no	yes	yes
CDH11	yes	7D	3	G>A	UTR3	no	no	no

NIP7	no	7D	16:6937644 9	A>G	UTR3	no	no	no
AARS	no	7D	16:7028667 6	A>G	nonsynonymous SNV	no	no	no
RHOT2	no	7D	16:723778 17:2143766	G>C	UTR3	no	yes	yes
C17orf51	no	7D	7 17:4613724	T>C	UTR3	no	no	no
NFE2L1	no	7D	8 17:5593899	G>T	UTR3	no	no	no
CUEDC1	no	7D	8 19:1190939	A>G	UTR3	no	yes	yes
ZNF491	no	7D	0 19:5019307	T>C	upstream	no	no	no
ADM5	no	7D	9	A>G	UTR5	no	yes	yes
PRAMEF6	no	7D	1:13007907 1:16014147	C>A	upstream nonsynonymous	no	no	no
ATP1A4	no	7D	0 1:18261487	G>T	SNV	no	no	no
RGS8	no	7D	9 1:22346590	G>A	UTR3 nonsynonymous	no	no	no
SUSD4	no	7D	1 1:23644526	C>T	SNV	no	no	no
ERO1B	no	7D	5	G>A	UTR5	no	no	no
STMN1	no	7D	1:26227366	G>A	UTR3 nonsynonymous	no	no	no
ARID1A	yes	7D	1:27023106	A>C	SNV	no	no	no
ZBTB8B	no	7D	1:32953263	C>A	UTR3 nonsynonymous	no	no	no
HYI	no	7D	1:43919348	C>G	SNV nonsynonymous	no	no	no
MAST2	no	7D	1:46468539	C>T	SNV	no	no	no
DEFB127	no	7D	20:137730	G>T	upstream	no	no	no
DEFB127	no	10B	20:137622 20:5219227	T>G	upstream nonsynonymous	no	no	no
ZNF217	no	7D	7 20:5493335	G>A	SNV	no	yes	yes
FAM210B	no	7D	3 2:10192513	C>T	upstream	no	no	no
RNF149	no	7D	2 2:21727695	C>T	UTR5	no	yes	yes
SMARCA1	no	7D	0 2:21948745	G>A	upstream nonsynonymous	no	no	no
PLCD4	no	7D	7 3:10219766	G>A	SNV	no	no	no
ZPLD1	no	7D	9 3:18449193	A>T	UTR3	no	no	no
LINC02069	no	7D	6	A>C	upstream nonsynonymous	no	no	no
PLCD1	no	7D	3:38051225	C>A	SNV	no	no	no
ZNF501	no	7D	3:44777008	G>A	UTR3	no	no	no
PRSS42	no	7D	3:46875616 4:11319839	A>G	upstream	no	no	no
TIFA	no	7D	2	A>G	UTR3	no	no	no
MXD4	no	7D	4:2251625	C>A	UTR3	no	no	no
N4BP2	yes	7D	4:40156135	T>C	UTR3	no	no	no
SREK1IP1	no	7D	5:64019260	A>G	UTR3	no	no	no
PACSIN1	no	7D	6:34494065 7:15717765	T>C	UTR5 nonsynonymous	no	no	no
DNAJB6	no	7D	9	A>G	SNV	no	yes	yes
ABHD17A	no	7E	19:1877184 19:4827301	C>T TCCATCATC>	UTR3	no	yes	yes
NOP53-AS1	no	7E	8	T	upstream	no	yes	yes

MFAP2	no	7E	1:17303418 22:2005354	T>G	nonsynonymous SNV	no	no	no
TANGO2	no	7E	7 3:19571718	T>G	UTR3	no	no	no
SDHAP1	no	7E	7 9:12956651	C>A	upstream	no	no	yes
ZBTB43	no	7E	0	A>C	upstream	no	no	no
POLR2L;TSP AN4	no	8A	11:842529	TGCCCCGGCC CGGCCCG>T	upstream	no	no	no
POLR2L;TSP AN4	no	8C	11:842529 12:1260040	TGCCCCGGCC CGGCCCG>T	upstream	no	no	no
TMEM132B	no	8A	71 12:1260040	G>A	stopgain	no	no	no
TMEM132B	no	8C	71 13:1125468	G>A	stopgain	no	no	no
LINC00354	no	8A	23 14:7156871	G>A	upstream nonsynonymous SNV	no	no	no
PCNX1	no	8A	7 14:7156871	C>T	nonsynonymous SNV	no	no	no
PCNX1	no	8C	7 14:7157779	C>T	SNV	no	no	no
PCNX1	no	10A	9 14:7157779	C>A	UTR3	no	no	no
PCNX1	no	10D	9 15:5825619	C>A	UTR3 nonsynonymous SNV	no	no	no
ALDH1A2	no	8A	3 15:5825619	T>C	nonsynonymous SNV	no	no	no
ALDH1A2	no	8C	3 15:9363229	T>C	SNV	no	no	no
RGMA	no	8A	0 15:9363229	C>A	UTR5	no	yes	yes
RGMA	no	8C	0	C>A	UTR5	no	yes	yes
TYW3	no	8A	1:75231489	C>G	UTR3	no	no	no
TYW3	no	8C	1:75231489	C>G	UTR3	no	no	no
TYW3	no	8D	1:75231489	C>G	UTR3	no	no	no
LINC01140	no	8A	1:87594569	C>T	upstream	no	no	no
LINC01140	no	8C	1:87594569	C>T	upstream	no	no	no
LINC01140	no	8D	1:87594569 20:5515205	C>T	upstream	no	no	no
LINC01716	no	8A	7 22:2284149	T>A	upstream	no	no	no
ZNF280B	no	8A	2 22:2284149	A>C	UTR3	no	no	no
ZNF280B	no	8C	2 22:4713399	A>C	UTR3 nonsynonymous SNV	no	no	no
CERK	no	8A	6	T>G	SNV	no	no	no
KLHL5	no	8A	4:39125658 7:13879421	A>G	UTR3	no	no	no
ZC3HAV1	no	8A	4 7:14195861	T>G	UTR5	no	no	no
PRSS58	no	8A	0	G>A	upstream nonsynonymous SNV	no	no	no
CLDN23	no	8A	8:8560695	G>A	nonsynonymous SNV	no	yes	yes
CLDN23	no	8C	8:8560695 11:1336810	G>A	SNV	no	yes	yes
LOC646522 LOC143666; PHRF1	no	8B	32	A>G	upstream	no	no	no
PHRF1 LOC143666; PHRF1	no	8B	11:576235	T>C	upstream	no	no	no
PHRF1	no	10C	11:576170 12:1309266	T>G	upstream nonsynonymous SNV	no	no	no
RIMBP2	no	8B	90	C>T	SNV	no	no	no

BCAT1	no	8B	12:2510237 3	G>C	UTR5	no	no	no
TTC5	no	8B	14:2075759 4	C>T	UTR3	no	no	no
MMP25	no	8B	16:3095951 17:3963752	T>G	upstream	no	no	no
KRT35	no	8B	2	A>T	upstream	no	no	no
CD300LF	no	8B	17:7269086 9	A>T	UTR3	no	no	yes
ABL2	yes	8B	1:17909560 7	G>C	nonsynonymous SNV	no	no	no
ABL2	yes	9A	1:17907652 7	C>T	UTR3	no	no	yes
PLXNA2	no	8B	1:20839109 7	T>G	nonsynonymous SNV	no	no	no
LRRC8C	no	8B	1:90183377 20:3350125	A>G	UTR3	no	no	no
ACSS2	no	8B	3	T>G	nonsynonymous SNV	no	no	no
DSN1	no	8B	20:3538066 0	T>G	UTR3	no	no	no
COL6A6	no	8B	3:13039434 4	C>T	UTR3	no	no	no
VPS52	no	8B	6:33239728	T>C	UTR5	no	no	no
CYP2W1	no	8B	7:1024846	A>C	nonsynonymous SNV	no	no	no
DDX56	no	8B	7:44613736 9:12454500	C>A	UTR5	no	no	no
DAB2IP	no	8B	6	G>A	UTR3	no	no	no
PAOX	no	8C	10:1352049 14	C>G	nonsynonymous SNV	no	no	no
RET	yes	8C	10:4361920 8	T>G	nonsynonymous SNV	no	no	no
ALG10	no	8C	12:3417923 0	G>A	nonsynonymous SNV	no	no	yes
INTS2	no	8C	17:5994325 3	T>C	UTR3	no	no	no
INTS2	no	11A	17:5999914 5	A>T	nonsynonymous SNV	no	no	no
CHMP6	no	8C	17:7897313 6	T>G	UTR3	no	no	no
KATNAL2	no	8C	18:4452634 9	T>A	upstream	no	no	no
INAFM1	no	8C	19:4777755 2	A>C	upstream	no	no	no
ZNF544	no	8C	19:5877322 4	A>T	nonsynonymous SNV	no	no	yes
ARID4B	no	8C	1:23538493 1	A>C	nonsynonymous SNV	no	no	no
PCSK9	no	8C	1:55529927 21:3530270	A>C	UTR3	no	no	no
LINC00649	no	8C	9	C>T	upstream	no	no	no
CCDC117	no	8C	22:2918357 9	T>A	UTR3	no	no	no
CCDC117	no	8D	22:2918357 9	T>A	UTR3	no	no	no
PLA2G3	no	8C	22:3153672 5	A>C	upstream	no	no	no
RHOB	no	8C	2:20646114	A>G	upstream	no	no	no
GPATCH11	no	8C	2:37325510 3:14116256	TATAG>T	UTR3	no	yes	yes
ZBTB38	no	8C	3	C>A	nonsynonymous SNV	no	no	no
TBC1D1	no	8C	4:38140679	T>G	UTR3	no	no	no
KLF3	no	8C	4:38690345	T>G	nonsynonymous SNV	no	no	no
C7	no	8C	5:40982531	C>T	UTR3	no	no	no

LHFPL3	no	8C	7:10396842 9	T>G	upstream	no	no	no
SCIN	no	8C	7:12609997 7:13433148	C>G	upstream	no	no	no
BPGM	no	8C	9 9:13703032	C>T	upstream	no	no	yes
RNU6ATAC	no	8C	7	T>C	upstream	no	no	no
ATP6AP2	no	8C	X:40439341 10:1188981	C>G	upstream	no	no	no
VAX1	no	8D	11	A>G	upstream	no	no	no
ITIH5	no	8D	10:7697635 11:6605051	C>A	stopgain nonsynonymous	no	no	no
CNIH2	no	8D	6	C>T	SNV	no	no	no
CACNA1C- IT2	no	8D	12:2156920 12:3252063	G>T	upstream nonsynonymous	no	no	no
BICD1	no	8D	6 12:5294251	C>A	SNV nonsynonymous	no	no	no
KRT71	no	8D	6	T>G	SNV nonsynonymous	no	no	no
USP5	no	8D	12:6964974 13:2896381	G>T	SNV	no	no	no
FLT1	no	8D	0 13:6643938	A>C	UTR3	no	no	no
LINC01052	no	8D	2 14:2150220	A>G	upstream nonsynonymous	no	no	no
RNASE13	no	8D	7 15:4928406	C>A	SNV	no	no	no
SECISBP2L	no	10B	1 15:5284354	A>G	UTR3	no	no	no
ARPP19	no	8D	7 15:8541340	T>A	UTR3	no	no	no
ALPK3	no	8D	8 17:4147551	A>C	UTR3	no	no	yes
ARL4D	no	8D	1 18:5694117	G>T	upstream	no	no	yes
RAX	no	8D	4 19:3737983	G>A	upstream	no	no	no
ZNF829	no	8D	9 19:5114352	A>C	UTR3	no	no	no
SYT3	no	8D	2	T>C	upstream	no	no	no
DFFA	no	8D	1:10521083 1:11996498	A>T	UTR3 nonsynonymous	no	no	no
HSD3B2	no	8D	7 1:15318944	C>A	SNV	no	no	no
PRR9	no	8D	3 1:24820233	C>A	upstream nonsynonymous	no	no	no
OR2L2	no	8D	2	G>T	SNV	no	no	no
ST6GALNAC 5	no	8D	1:77529083 21:1961757	G>A	UTR3 nonsynonymous	no	no	yes
CHODL	no	8D	4 21:3212046	G>T	SNV	no	no	no
KRTAP21-2	no	8D	5 22:2079608	A>T	upstream	no	no	no
KLHL22	no	8D	3 2:17840543	A>C	UTR3	no	no	no
AGPS	no	8D	9 2:17840478	T>A	UTR3	no	no	no
AGPS	no	11A	8 2:22822245	CAT>C	UTR3	no	yes	yes
MFF	no	8D	8	G>T	UTR3 nonsynonymous	no	no	no
ATRAID	no	8D	2:27439488 3:12416561	G>T	SNV nonsynonymous	no	no	no
KALRN	no	8D	3	C>G	SNV nonsynonymous	no	no	no
SETD2	yes	8D	3:47163154	T>C	SNV	no	no	no

KCTD8	no	8D	4:44451720	A>G	upstream	no	no	no
RUFY3	no	8D	4:71570622	A>T	upstream	no	no	no
LINC01088	no	8D	4:79892392	A>G	upstream	no	no	no
ANTXR2	no	8D	4:80826091	T>C	UTR3	no	no	no
EPB41L4A	no	8D	5:11154562	T>C	nonsynonymous	no	no	no
			0		SNV	no	no	no
KCTD16	no	8D	5:14358672	A>G	nonsynonymous	no	no	no
			5		SNV	no	no	no
GPRIN1	no	8D	5:17602334	T>G	UTR3	no	no	no
			6			no	no	no
CENPQ	no	8D	6:49460384	C>T	UTR3	no	no	no
			7:14958507			no	no	no
ACTR3C	no	8D	5	C>T	UTR3	no	no	no
			7:14958034			no	no	no
ACTR3C	no	10E	5	T>C	UTR3	no	no	no
					nonsynonymous	no	no	no
ABCA13	no	8D	7:48280464	G>C	SNV	no	no	no
ZKSCAN1	no	8D	7:99635864	T>TA	UTR3	no	no	no
			8:10117082			no	no	no
SPAG1	no	8D	7	G>A	UTR5	no	no	no
			12:1079749		nonsynonymous	no	no	no
BTBD11	no	8E	82	A>C	SNV	no	no	no
ARID3A	no	8E	19:972246	TAC>T	UTR3	no	yes	yes
ADARB2	no	9A	10:1227777	G>A	UTR3	no	no	no
			11:1173024			no	no	no
DSCAML1	no	9A	45	C>A	splicing	no	no	no
			11:1226834			no	no	no
UBASH3B	no	9A	77	C>T	UTR3	no	yes	yes
			11:4641851			no	yes	yes
AMBRA1	no	9A	0	C>T	UTR3	no	yes	yes
			11:6070860		nonsynonymous	no	yes	yes
SLC15A3	no	9A	6	C>T	SNV	no	yes	yes
			11:6408826		nonsynonymous	no	no	no
PRDX5	no	9A	3	A>C	SNV	no	no	no
			11:7338738			no	no	no
RAB6A	no	9A	2	G>C	UTR3	no	no	no
			12:1138599			no	no	no
SDSL	no	9A	09	G>A	upstream	no	yes	yes
			12:1216666		nonsynonymous	no	yes	yes
P2RX4	no	9A	07	A>G	SNV	no	yes	yes
			12:1337953			no	no	no
ANHX	no	9A	09	C>T	UTR3	no	no	no
			12:1774241			no	no	no
LINC02378	no	9A	4	A>C	upstream	no	no	no
			12:5812515		nonsynonymous	no	no	no
AGAP2	no	9A	7	T>C	SNV	no	no	no
			15:1014190			no	no	no
ALDH1A3	no	9A	17	C>T	upstream	no	no	no
			15:2492062			no	no	no
NPAP1	no	9A	9	G>T	UTR5	no	no	no
			15:9062305		nonsynonymous	no	no	no
ZNF710	no	9A	7	T>G	SNV	no	no	yes
					nonsynonymous	no	no	no
CLCN7	no	9A	16:1505209	G>C	SNV	no	no	no
					CCTAGGCTG	no	no	no
CHP2	no	9A	16:2376503		GTCTTGAGC	no	no	no
			5		TCCTGGG>C	no	no	no
ADAMTS18	no	9A	16:7746542	G>A	upstream	no	yes	yes
			1		SNV	no	yes	yes
GJC1	no	9A	17:4287692	A>T	UTR3	no	yes	yes
			6			no	yes	yes
FADS6	no	9A	17:7287422	A>C	UTR3	no	no	no
			8			no	no	no
ELOA2	no	9A	18:4456189	A>G	UTR5	no	no	no
			5			no	no	no

PPP4R1	no	9A	18:9547007 19:3016533	C>A	UTR3 nonsynonymous	no	no	no
PLEKHF1	no	9A	0 19:5669668	G>C	SNV	no	no	yes
GALP	no	9A	5	G>C	UTR3	no	no	no
PDPN	no	9A	1:13943516 1:15558186	G>A	UTR3 nonsynonymous	no	no	no
MSTO1	no	9A	6 1:15687644	A>T	SNV nonsynonymous	no	yes	yes
PEAR1	no	9A	9 1:19787188	G>T	SNV nonsynonymous	no	no	no
C1orf53	no	9A	0 20:4727360	G>C	SNV nonsynonymous	no	no	no
PREX1	no	9A	2 22:1707227	A>C	SNV nonsynonymous	no	no	no
CCT8L2	no	9A	0 22:4654609	C>T	SNV	no	yes	yes
PPARA	no	9A	4 22:5060847	G>A	upstream	no	yes	yes
PANX2	no	9A	7 2:11087470	T>G	upstream	no	no	no
MALL	no	9A	5 2:11187856	A>T	upstream	no	no	no
BCL2L11	no	9A	8	C>T	UTR5	no	yes	yes
RAD51AP2	no	9A	2:17712033 2:22474125	A>T	upstream	no	no	no
WDFY1	no	9A	7 2:23066130	T>TA	UTR3	no	no	no
TRIP12	no	9A	5	A>C	splicing	no	no	no
HAAO	no	9A	2:43019902	C>T	upstream nonsynonymous	no	no	no
ARIH2	no	9A	3:49016923 4:17724536	T>A	SNV frameshift	no	no	no
SPCS3	no	9A	9	GTTTC>G	deletion nonsynonymous	no	no	no
KCNIP4	no	9A	4:20733630	T>G	SNV	no	no	no
NIPAL1	no	9A	4:48040457	G>C	UTR3	no	no	no
FRYL	no	9A	4:48592848	C>T	splicing	no	no	no
LOC644145	no	9A	4:56685722	C>G	upstream	no	no	no
FGF5	no	9A	4:81210580	A>G	UTR3 nonsynonymous	no	no	no
ICE1	no	9A	5:5463805	A>T	SNV nonsynonymous	no	no	no
ICE1	no	10B	5:5457844	C>T	SNV nonsynonymous	no	no	yes
ICE1	no	10E	5:5457844 6:12331862	C>T	SNV	no	no	yes
CLVS2	no	9A	5 6:16770425	C>T	UTR5	no	no	no
UNC93A	no	9A	7	G>A	upstream	no	no	no
ELMO1	no	9A	7:36894909	C>G	UTR3	no	no	no
RNF216	no	9A	7:5661388 8:10442700	G>A	UTR3	no	no	no
DCAF13	no	9A	2 8:14437284	G>A	UTR5	no	no	no
ZNF696	no	9A	6 8:14499284	G>C	upstream nonsynonymous	no	no	no
PLEC	no	9A	2 9:11200241	C>G	SNV	no	no	no
EPB41L4B	no	9A	5 9:12964466	A>C	UTR3	no	no	no
ZBTB34	no	9A	3	T>A	UTR3	no	no	no
C9orf135	no	9A	9:72435730	T>TA	UTR5	no	no	yes

KLF9	no	9A	9:73029764	T>C	upstream nonsynonymous SNV	no	no	no
WNK2	yes	9A	9:96061504	T>G		no	no	no
NHS	no	9A	X:17754102	GACTGCAAT TAAAC>G	UTR3	no	no	no
DMD	no	9A	X:31139568	T>C	UTR3	no	no	no
RPGR	no	9A	X:38187197	G>A	upstream nonsynonymous SNV	no	no	no
CXorf36	no	9A	X:45013353	C>T	nonsynonymous SNV	no	no	no
SVIL	no	9B	10:2978815	C>T	nonsynonymous SNV	no	no	no
NRP1	no	9B	10:3354296	T>A	nonsynonymous SNV	no	no	no
PLEKHB1	no	9B	11:7335733	G>A	UTR5	no	yes	yes
TMTC3	no	9B	12:8859150	T>G	UTR3	no	no	no
MAX	yes	9B	14:6556051	AT>A	frameshift deletion	no	no	no
TGFB3	no	9B	14:7644812	G>C	UTR5	no	no	yes
SLC24A4	no	9B	14:9296297	T>A	UTR3	no	no	no
CERS3	no	9B	15:1010848	C>T	UTR5	no	yes	yes
LOC100128 108	no	9B	15:1022846	AATGAGCCC TTCCAGAAC >A	frameshift deletion nonsynonymous SNV	no	no	no
OR4N4	no	9B	15:2238310	C>G	SNV	no	no	no
LINC02204	no	9B	15:7087979	C>T	upstream nonsynonymous SNV	no	yes	yes
UTP4	no	9B	16:6918478	C>G	SNV	no	yes	yes
LINC00304	no	9B	16:8922532	A>G	upstream	no	yes	yes
TRAF4	no	9B	17:2707686	G>A	UTR3	no	no	no
CDH20	no	9B	18:5916661	C>A	nonsynonymous SNV	no	no	no
GNA11	yes	9B	19:3122944	C>A	UTR3	no	yes	yes
ZNF331	yes	9B	19:5408125	C>A	UTR3	no	no	no
PLPPR3	no	9B	19:822842	C>T	upstream nonsynonymous SNV	no	no	yes
OVGP1	no	9B	1:11196423	T>C	SNV	no	yes	yes
DDX59	no	9B	1:20063601	CTT>C	UTR5	no	no	yes
LGALSL	no	9B	2:64687668	G>C	UTR3	no	no	no
PNO1	no	9B	2:68392514	A>C	UTR3	no	no	no
CHCHD6	no	9B	3:12642225	C>T	upstream nonsynonymous SNV	no	no	no
COL8A1	no	9B	3:99509748	G>A	nonsynonymous SNV	no	no	no
TBC1D9	no	9B	4:14158305	T>C	SNV	no	no	no
ZFP42	no	9B	4:18892601	C>T	UTR3	no	no	yes
GDF9	no	9B	5:13220061	T>C	UTR5	no	no	no
NDST1	no	9B	5:14988776	G>A	UTR5	no	yes	yes
IRGM	no	9B	5:15022752	C>T	UTR5	no	no	yes
CPEB4	no	9B	4	A>G	UTR3	no	no	no

NSD1	yes	9B	5:17666288 8	A>C	nonsynonymous SNV	no	no	no
NSD1	yes	11C	5:17655910 5	GC>G	upstream nonsynonymous SNV	no	no	no
DMGDH	no	9B	5:78365362	A>C	nonsynonymous SNV	no	no	no
DXO	no	9B	6:31938510	G>C	nonsynonymous SNV	no	no	no
FBXL4	no	9B	6:99374803 7:11691743	C>T	SNV	no	yes	yes
WNT2	no	9B	6:12862683 7:12862683	G>A	UTR3 nonsynonymous SNV	no	yes	yes
TNPO3	no	9B	4	A>G	SNV	no	no	no
DFNA5	no	9B	7:24797623	C>T	UTR5	no	no	no
CDK6	yes	9B	7:92237932	G>C	UTR3	no	no	no
SLC35D2	no	9B	9:99083522 X:10304595	C>A	UTR3	no	no	no
PLP1	no	9B	3	C>A	UTR3	no	no	no
SPIN4	no	9B	X:62567814 10:1044149	G>A	UTR3 nonsynonymous SNV	no	yes	yes
TRIM8 HSP90B1;M IR3652;TTC 41P	no	9C	39	G>A	SNV	no	no	no
	no	9C	12:1043240 39	A>C	upstream nonsynonymous SNV	no	no	no
GPD1	no	9C	12:5050064 7	C>A	SNV	no	no	no
PYM1	no	9C	12:5629560 2	C>A	UTR3	no	no	no
ATXN7L3B	no	9C	12:7493309 7	C>A	UTR3	no	no	no
SLC25A47	no	9C	14:1007897 46	T>G	UTR5 nonsynonymous SNV	no	no	no
MDGA2	no	9C	14:4738925 6	C>A	SNV	no	yes	yes
MAP3K9 LOC101928 414	no	9C	14:7119462 8	G>C	UTR3	no	no	no
	no	9C	15:4557179 9	G>A	upstream	no	no	no
GLDN	no	9C	15:5169820 5	A>G	UTR3	no	no	no
ANP32A	no	9C	15:6911364 2	C>T	upstream	no	no	no
CLK3	no	9C	15:7489985 9	T>A	upstream	no	no	no
OR1F1	no	9C	16:3253651 16:5855343	C>T	upstream	no	no	no
SETD6	no	9C	6	A>G	UTR3	no	yes	yes
SLFN11	no	9C	17:3369030 5	G>C	stopgain nonsynonymous SNV	no	no	no
SLC38A10	no	9C	17:7921942 0	C>G	SNV	no	no	no
FAM129C	no	9C	19:1763357 4	A>G	upstream	no	no	no
ZNF507	no	9C	19:3287593 4	T>G	UTR3 nonsynonymous SNV	no	no	no
PLEKHG2	no	9C	19:3991543 2	C>T	SNV	no	yes	yes
TTLL10	no	9C	1:1108589	C>T	upstream	no	no	yes
SNHG12	no	9C	1:28910202 22:1995611	C>A	upstream nonsynonymous SNV	no	yes	yes
COMT	no	9C	4	C>A	SNV	no	no	no
ICA1L	no	9C	2:20364275 1	G>T	UTR3	no	no	no
APELA	no	9C	4:16579751 6	A>G	upstream	no	no	no

GUF1	no	9C	4:44680174 5:18004339	C>A	upstream nonsynonymous	no	no	no
FLT4	yes	9C	8 5:18002877	T>C	SNV	no	no	no
FLT4 C5orf22;DR	yes	10E	3	C>T	UTR3	no	yes	yes
OSHA	no	9C	5:31532357 6:11684353	T>C	upstream	no	no	no
FAM26E	no	9C	6 6:14770631	C>T	UTR3	no	yes	yes
STXBP5	no	9C	0 9:12336583	A>G	UTR3	no	no	no
MEGF9	no	9C	4 9:13256504	T>C	UTR3	no	no	yes
TOR1B	no	9C	2 9:13996073	A>G	upstream nonsynonymous	no	no	no
SAPCD2	no	9C	7 X:12903969	C>T	SNV	no	yes	yes
UTP14A	no	9C	7 10:1035303	G>A	upstream frameshift	no	no	no
FGF8	no	10A	74 10:1035303	GCTCT>G	deletion frameshift	no	no	no
FGF8	no	10D	74	GCTCT>G	deletion nonsynonymous	no	no	no
UBQLNL	no	10A	11:5537111 12:1323935	G>C	SNV	no	no	no
ULK1	no	10A	18 12:5412143	T>G	splicing	no	no	no
CALCOCO1	no	10A	8 14:2047034	A>G	upstream nonsynonymous	no	yes	yes
OR4Q2	no	10A	3 14:2047034	G>A	SNV nonsynonymous	no	no	no
OR4Q2	no	10D	3 16:3068091	G>A	SNV nonsynonymous	no	no	no
FBRS	no	10A	0 17:2202320	G>T	SNV	no	no	no
MTRNR2L1	no	10A	9 17:2202320	G>T	UTR5	no	no	no
MTRNR2L1	no	10D	9 17:3736825	G>T	UTR5	no	no	no
STAC2	no	10A	8 17:3736825	T>C	UTR3	no	no	no
STAC2	no	10D	8	T>C	UTR3 nonsynonymous	no	no	no
STXBP2	no	10A	19:7705587	C>G	SNV nonsynonymous	no	no	no
STXBP2	no	10D	19:7705587	C>G	SNV	no	no	no
AZU1	no	10A	19:831994	A>C	UTR3	no	no	no
MARCH2	no	10A	19:8483450	C>G	UTR5 nonsynonymous	no	no	no
HNRNPM	no	10A	19:8530374 1:23706551	T>A	SNV	no	no	no
MTR	no	10A	9	T>A	UTR3	no	no	no
GBP2	no	10A	1:89573310 20:4919953	G>T	UTR3	no	no	no
PTPN1	no	10A	3 20:4919953	G>C	UTR3	no	no	no
PTPN1	no	10D	3 20:5619645	G>C	UTR3	no	no	no
ZBP1	no	10A	9 22:2352429	A>AACCC	upstream nonsynonymous	no	no	no
BCR	yes	10A	5 22:2352429	C>G	SNV nonsynonymous	no	no	no
BCR	yes	10D	5 22:5016937	C>G	SNV nonsynonymous	no	no	no
BRD1	no	10A	1 22:5016937	C>T	SNV nonsynonymous	no	no	yes
BRD1	no	10D	1	C>T	SNV	no	no	yes

BRD1	no	10E	22:5016764 0	A>T	UTR3	no	no	no
GCC2	no	10A	2:10908931 7	A>G	nonsynonymous SNV	no	no	no
GCC2	no	10D	2:10908931 7	A>G	nonsynonymous SNV	no	no	no
ZC3H6	no	10A	2:11303351 3	C>T	UTR5	no	no	no
ZC3H6	no	10D	2:11303351 3	C>T	UTR5	no	no	no
PLCL1	no	10A	2:19901294 3	T>G	UTR3	no	no	no
PLCL1	no	10D	2:19901294 3	T>G	UTR3	no	no	no
SNORA10B	no	10A	2:30410165	T>C	upstream	no	no	no
SNORA10B	no	10D	2:30410165	T>C	upstream	no	no	no
FIGLA	no	10A	2:71018703	TC>T	upstream	no	yes	yes
FIGLA	no	10B	2:71018703	TC>T	upstream	no	yes	yes
FIGLA	no	10C	2:71018703	TC>T	upstream	no	yes	yes
FIGLA	no	10D	2:71018703	TC>T	upstream	no	yes	yes
FIGLA	no	10E	2:71018703	TC>T	upstream	no	yes	yes
REG1A	no	10A	2:79347620	C>G	UTR5	no	no	no
REG1A	no	10D	2:79347620	C>G	UTR5	no	no	no
GRM7-AS3	no	10A	3:6847799 4:13844188	G>T	upstream	no	no	no
PCDH18	no	10A	8 4:16441472	T>C	UTR3	no	yes	yes
TMA16	no	10A	6	T>G	upstream	no	no	no
CXCL3	no	10A	4:74904926	T>C	upstream	no	yes	yes
CXCL3	no	10D	4:74904926 5:10253992	T>C	upstream	no	yes	yes
PPIP5K2	no	10A	5	C>T	UTR3	no	no	no
CCDC152	no	10A	5:42801584	T>A	UTR3	no	no	no
CCDC152	no	10D	5:42801584 6:14414949	T>A	UTR3	no	no	no
PHACTR2	no	10A	0 6:14414949	G>T	UTR3	no	no	no
PHACTR2	no	10D	0	G>T	UTR3	no	no	no
SEMA3D	no	10A	7:84628781 9:12792317	T>C	nonsynonymous SNV	no	no	no
PPP6C	yes	10A	8 9:12792317	T>G	nonsynonymous SNV	no	no	no
PPP6C	yes	10D	8	T>G	nonsynonymous SNV	no	no	no
ASB13	no	10B	10:5681564	T>C	UTR3	no	no	no
ASB13	no	10E	10:5681564 12:1032182	T>C	UTR3	no	no	no
LINC00485	no	10B	66 12:1185023	G>A	upstream	no	no	no
VSIG10	no	10B	92 12:4582556	T>C	UTR3	no	no	no
ANO6	no	10B	7 12:5575835	T>C	UTR3	no	no	no
OR6C75	no	10B	0 12:9309685	A>T	upstream	no	no	no
C12orf74	no	10B	5 12:9309685	G>A	UTR5	no	yes	yes
C12orf74 PARP2;RPP H1	no	10E	5 14:2081160	G>A	UTR5	no	yes	yes
	no	10B	4	T>A	upstream	no	yes	yes

EDDM3A	no	10B	14:2121355 8	G>A	upstream	no	yes	yes
FANCM	no	10B	14:4566952 5	A>G	UTR3	no	no	no
MGA	no	10B	15:4206042 5	A>T	UTR3	no	no	no
ZNF263	no	10B	16:3339971 16:8108000	CCCTA>C	frameshift deletion	no	no	no
ATMIN	no	10B	2	A>C	UTR3	no	no	no
PRPSAP2	no	10B	17:1883428 5	A>G	UTR3	no	no	no
PRPSAP2	no	10E	17:1883428 5	A>G	UTR3	no	no	no
TBX21	no	10B	17:4581075 4	A>G	UTR5	no	no	no
HOXB13	no	10B	17:4680376 3	C>T	UTR3	no	no	no
MED13	no	10B	17:6002308 3	T>C	UTR3	no	no	no
MED13	no	10E	17:6002308 3	T>C	UTR3	no	no	no
BST2	no	10B	19:1751382 2	G>A	UTR3	no	no	no
BST2	no	10E	19:1751382 2	G>A	UTR3	no	no	no
GPR61	no	10B	1:11008679 5	T>C	nonsynonymous SNV	no	no	no
GPR61	no	10E	1:11008679 5	T>C	nonsynonymous SNV	no	no	no
GPR61	no	11B	1:11008668 0	C>G	nonsynonymous SNV	no	no	no
GPR61	no	11D	1:11008668 0	C>G	nonsynonymous SNV	no	no	no
TNFRSF4	no	10B	1:1150490	A>T	upstream	no	no	no
TNFRSF4	no	10E	1:1150490	A>T	upstream	no	no	no
TMEM206	no	10B	1:21258829 8	G>A	upstream	no	no	no
TMEM206	no	10E	1:21258829 8	G>A	upstream	no	no	no
SESN2	no	10B	1:28608497	T>G	UTR3	no	no	no
SESN2	no	10E	1:28608497	T>G	UTR3	no	no	no
DMBX1	no	10B	1:46979799	T>A	UTR3	no	no	no
IFI44	no	10B	1:79128470	G>C	nonsynonymous SNV	no	no	no
SPATA1	no	10B	1:84991728 22:1859334	A>T	nonsynonymous SNV	no	no	no
TUBA8	no	10B	22:3235344 8	A>C	upstream	no	no	no
YWHAH	no	10B	22:3235304 7	T>A	UTR3	no	no	no
YWHAH	no	11C	22:3235304 9	A>G	UTR3	no	no	no
WDR33	no	10B	2:12847782 8	G>A	stopgain	no	no	no
WDR33	no	10E	2:12847782 8	G>A	stopgain	no	no	no
RAB3GAP1	no	10B	2:13591136 2	G>C	nonsynonymous SNV	no	no	no
RAB3GAP1	no	10E	2:13591136 2	G>C	nonsynonymous SNV	no	no	no
MAP2	no	10B	2:21055776 0	T>A	nonsynonymous SNV	no	no	no
MAGEF1	no	10B	3:18442932 5	A>C	nonsynonymous SNV	no	no	no
CCDC50	no	10B	3:19110980 6	T>A	UTR3	no	no	no

IL17RD	no	10B	3:57124856 5:14837673	C>T	UTR3	no	yes	yes
SH3TC2	no	10B	9 5:14837673	T>C	UTR3	no	no	no
SH3TC2	no	10E	9 5:14910982	T>C	UTR3	no	no	no
PPARGC1B	no	10B	6	T>G	UTR5 nonsynonymous	no	no	no
CWC27	no	10B	5:64267637	C>A	SNV	no	no	no
MTRNR2L2	no	10B	5:79946435	T>C	UTR5	no	no	no
MIR183;MI	no	10B	7:12941504					
R96	no	10B	4 7:15006864	A>C	upstream	no	no	no
REPIN1	no	10B	3 7:15006864	C>T	stopgain	no	no	no
REPIN1	no	10E	3	C>T	stopgain	no	no	no
LINC00525	no	10B	7:47800091	A>T	upstream	no	no	yes
HMBOX1;IN	no	10B	8:28747765	T>A	upstream	no	no	no
TS9	no	10B	9:10258398 0					
NR4A3	yes	10B	0	GCGCGCGCG CGCACAC>G	upstream	no	no	no
NR4A3	yes	10E	9:10258398 0 10:1456218	GCGCGCGCG CGCACAC>G	upstream	no	no	no
FAM107B	no	10C	4	A>G	UTR3 nonsynonymous	no	no	no
DCHS1	no	10C	11:6655398 13:4866963	C>T	SNV	no	no	yes
MED4	no	10C	1 19:3998938	G>C	upstream	no	no	no
DLL3	no	10C	7 20:6215209	A>C	upstream	no	no	no
PPDPF	no	10C	5	A>C	upstream	no	no	no
SPARCL1	no	10C	4:88451111	T>C	upstream	no	no	yes
ZBED9	no	10C	6:28555806	C>T	UTR5	no	no	no
SAYS1	no	10C	6:39080565 7:10567355	T>G	UTR5	no	no	no
CDHR3	no	10C	1 7:15113784	T>C	UTR3	no	no	no
CRYGN	no	10C	2	G>T	upstream nonsynonymous	no	no	no
CHRNA3	no	10C	8:42587366 9:13700070	T>A	SNV	no	no	no
WDR5	no	10C	1 X:15305162	C>T	upstream nonsynonymous	no	no	no
IDH3G	no	10C	3 12:4948183	G>C	SNV	no	no	no
DHH	no	10D	1 12:5270826	T>A	UTR3	no	no	no
KRT83	no	10D	3 14:2477819	G>T	UTR3	no	no	no
NOP9	no	10D	0 15:4500780	T>G	UTR3 nonsynonymous	no	no	no
B2M	yes	10D	3 1:15830171	C>G	SNV	no	no	no
CD1B	no	10D	9 1:21223628	T>G	upstream nonsynonymous	no	no	no
DTL	no	10D	3	A>G	SNV nonsynonymous	no	no	no
IPO13	no	10D	1:44424475 22:4379605	T>G	SNV	no	no	no
LINC01639	no	10D	0 2:20258284	G>A	upstream nonsynonymous	no	no	no
ALS2	no	10D	6 3:10123731	A>C	SNV	no	no	no
FAM172BP	no	10D	9	G>A	upstream	no	no	no

MON1A	no	10D	3:49967434	A>G	UTR5 nonsynonymous	no	no	no
PRRC2A	no	11A	6:31595932	C>G	SNV	no	no	no
ERICH5	no	10D	8:99076830	T>G	UTR5	no	no	no
P2RY8	yes	10D	X:1582301 10:9809796	G>A	UTR3 nonsynonymous	no	no	yes
DNTT	no	10E	2 12:1503905	C>T	SNV	no	no	yes
MGP	no	10E	9 12:6641779	CAGAGG>C	upstream	no	no	no
MIR6074	no	10E	9 12:6663873	G>C	upstream	no	no	no
IRAK3	no	10E	7 12:6658233	G>T	stopgain	no	no	no
IRAK3	no	11B	1 12:6658233	G>C	upstream	no	no	no
IRAK3	no	11D	1	G>C	upstream nonsynonymous	no	no	no
LAG3	no	10E	12:6884685 13:8445257	A>G	SNV	no	yes	yes
SLITRK1	no	10E	5 14:7303127	T>A	UTR3	no	no	no
RGS6	no	10E	5 15:4217461	C>T	UTR3 nonsynonymous	no	no	no
SPTBN5	no	10E	8 15:5613038	A>G	SNV	no	no	no
NEDD4	no	10E	0 16:1474366	T>C	splicing nonsynonymous	no	no	no
BFAR	no	10E	4 16:1857393	G>T	SNV	no	no	no
NOMO2	no	10E	4 16:2899377	ACGCACG>A	upstream nonsynonymous	no	no	yes
SPNS1	no	10E	4 16:5328962	G>A	SNV nonsynonymous	no	no	no
CHD9	no	10E	7 18:7050130	T>G	SNV	no	no	no
NETO1	no	10E	6 19:1565131	C>T	UTR3 nonsynonymous	no	no	no
CYP4F22	no	10E	7 19:3700533	G>T	SNV nonsynonymous	no	no	yes
ZNF260	no	10E	2 1:16102628	T>C	SNV frameshift	no	no	no
ARHGAP30	no	10E	0 1:17162169	AT>A	deletion	no	no	no
MYOC	no	10E	7 1:20175270	G>A	stopgain nonsynonymous	no	no	no
NAV1	no	10E	3	C>T	SNV	no	no	no
KIAA0754	no	10E	1:39880980 20:2380782	T>G	UTR3	no	no	no
CST2	no	10E	3	A>G	upstream	no	no	no
RNF24	no	10E	20:3914293 21:4449772	G>C	UTR3	no	no	no
CBS	no	10E	6 21:4760983	G>A	upstream	no	yes	yes
LSS	no	10E	6 22:3757692	C>T	UTR3	no	no	yes
C1QTNF6	no	10E	2	A>C	UTR3	no	no	no
PPP1R21	no	10E	2:48667586 3:12135101	G>A	upstream nonsynonymous	no	no	no
HCLS1	no	10E	1	C>T	SNV	no	no	no
SLC6A6	no	10E	3:14529678	G>A	UTR3	no	no	no
CMC1	no	10E	3:28282374	C>T	upstream nonsynonymous	no	no	no
LOC389199	no	10E	4:7941590	C>G	SNV	no	no	no
SDHA	yes	10E	5:256941	TTC>T	UTR3	no	yes	yes

WDR70	no	10E	5:37752665 6:13848329	G>A	nonsynonymous SNV	no	yes	yes
ARFGEF3	no	10E	3	T>G	nonsynonymous SNV	no	no	no
SYNGAP1	no	10E	6:33412373	G>A	nonsynonymous SNV	no	no	no
MRPS17	no	10E	7:56020983	G>GGT	frameshift insertion	no	no	no
ZNF138	no	10E	7:64254334 8:12421949	G>T	upstream nonsynonymous SNV	no	no	no
FAM83A	no	10E	2	A>C	SNV	no	no	no
CHMP7	no	10E	8:23104150	GGGC>G	UTR5	no	no	no
UNC5D	no	10E	8:35649034 9:13070049	T>A	UTR3	no	no	no
DPM2	no	10E	9	C>G	UTR5	no	no	yes
ANXA1	no	10E	9:75773632 10:1026891	G>T	nonsynonymous SNV	no	yes	yes
SLF2	no	11A	83	T>G	nonsynonymous SNV	no	no	no
FAM53B	no	11A	10:1263104 95	C>G	UTR3	no	no	no
TACR2	no	11A	10:7117701 3	C>G	upstream	no	no	no
TYSND1	no	11A	10:7189832 7	T>C	UTR3	no	no	no
AMPD3	no	11A	11:1048326 8	T>G	nonsynonymous SNV	no	no	no
TMPRSS4	no	11A	11:1179888 44	A>G	UTR3	no	no	no
OR4D5	no	11A	11:1238094 07	A>G	upstream	no	no	no
KCNJ5	yes	11A	11:1287864 76	G>T	nonsynonymous SNV	no	no	no
TP53AIP1	no	11A	11:1288057 82	T>A	UTR3	no	no	no
CELF1	no	11A	11:4749151 4	A>G	UTR3	no	no	no
OR5M9	no	11A	11:5623150 1	T>A	upstream	no	no	no
RBM14- RBM4;RBM 4	no	11A	11:6641139 5	T>C	nonsynonymous SNV	no	yes	yes
NUDT8	no	11A	11:6739748 4	T>G	upstream	no	no	no
WNK1	no	11A	12:1018133 12:1169971	C>T	UTR3	no	no	no
MAP1LC3B2	no	11A	42	T>A	upstream	no	no	no
CDK4	yes	11A	12:5814507 2	A>G	nonsynonymous SNV	no	no	no
LRIG3	yes	11A	12:5926637 0	T>C	nonsynonymous SNV	no	yes	yes
PTPRB	yes	11A	12:7100300 5	T>C	nonsynonymous SNV	no	no	no
AICDA	no	11A	12:8755363 13:1127220	A>G	UTR3	no	no	no
SOX1	no	11A	67	G>T	nonsynonymous SNV	no	no	yes
ZC3H13	no	11A	13:4653641 7	A>T	UTR3	no	no	no
EIF2AK4	no	11A	15:4024147 1	T>G	splicing	no	no	no
TMEM204	no	11A	16:1605369	C>T	UTR3	no	no	no
SRRM2	no	11A	16:2802742 16:3153837	G>C	UTR5	no	no	no
AHSP	no	11A	4	A>T	upstream	no	no	no
RPGRIP1L	no	11A	16:5363438 7	A>C	UTR3	no	no	no

CYB5B	no	11A	16:6949685 5	T>G	UTR3	no	no	no
KCNJ12;KCN			17:2131901		nonsynonymous			
J18	no	11A	3	G>A	SNV	no	yes	yes
			17:3992378		nonsynonymous			
JUP	no	11A	5	T>C	SNV	no	no	no
			17:4117020		nonsynonymous			
VAT1	no	11A	3	C>T	SNV	no	yes	yes
			19:1890238					
COMP	no	11A	4	CTGTG>C	upstream	no	no	yes
			19:2198790					
ZNF43	no	11A	0	A>C	UTR3	no	no	no
					nonsynonymous			
TBXA2R	no	11A	19:3595040	T>A	SNV	no	no	yes
			19:4818526		nonsynonymous			
BICRA	no	11A	0	G>C	SNV	no	no	no
			1:11215095					
LINC01160	no	11A	3	C>T	upstream	no	no	yes
				ACCTGTTCTC				
			1:15066953		CCCCCCCCA			
GOLPH3L	no	11A	1	CCCCCGT>A	UTR5	no	no	no
			1:16253110					
UAP1	no	11A	5	T>C	upstream	no	no	no
			1:16968169					
SELL	no	11A	3	T>C	upstream	no	no	no
			20:4698770					
LINC00494	no	11A	7	T>C	upstream	no	yes	yes
			21:2445063					
MIR6130	no	11A	5	C>T	upstream	no	no	no
			21:3179905					
KRTAP13-3	no	11A	0	A>T	upstream	no	no	no
			22:3243824					
SLC5A1	no	11A	6	C>T	upstream	no	no	no
			22:4679575		nonsynonymous			
CELSR1	no	11A	2	A>T	SNV	no	no	no
					nonsynonymous			
GREB1	no	11A	2:11772155	G>A	SNV	no	no	no
			2:17808545					
HNRNPA3	no	11A	6	C>T	UTR3	no	no	no
					nonsynonymous			
AAK1	no	11A	2:69771586	G>C	SNV	no	no	no
			3:11988658		nonsynonymous			
GPR156	no	11A	5	G>A	SNV	no	no	no
			4:14462191					
FREM3	no	11A	1	C>T	upstream	no	no	no
RBPJ	no	11A	4:26436018	T>G	UTR3	no	no	no
FAM169A	no	11A	5:74073426	A>T	UTR3	no	no	no
					nonsynonymous			
RIOK2	no	11A	5:96512873	A>T	SNV	no	no	no
RGMB	no	11A	5:98129818	G>A	UTR3	no	no	no
TFAP2A	no	11A	6:10397465	A>C	UTR3	no	no	no
			6:10900405					
FOXO3	yes	11A	4	T>C	UTR3	no	no	no
PHACTR1	no	11A	6:12716997	C>G	UTR5	no	no	no
			6:13956380		nonsynonymous			
TXLNB	no	11A	5	T>C	SNV	no	no	no
LOC101929			6:16958356					
504	no	11A	6	C>T	upstream	no	no	yes
PSORS1C3	no	11A	6:31146433	C>A	upstream	no	no	no
PRPF4B	no	11A	6:4020957	A>C	upstream	no	no	no
LOC101929								
555	no	11A	6:40991737	G>A	upstream	no	no	no
DDX43	no	11A	6:74103929	G>C	upstream	no	no	no
			7:11120295					
IMMP2L	no	11A	3	A>C	upstream	no	no	no

TCAF1	no	11A	7:14359921 8	A>T	UTR5	no	no	no
DPP6	no	11A	7:15358479 2	G>T	nonsynonymous SNV	no	no	no
FAM83H	no	11A	8:14480712 8	C>T	UTR3	no	no	no
C9orf152	no	11A	9:11297092 8	T>A	upstream nonsynonymous SNV	no	no	no
SPATA31E1	no	11A	9:90503352 X:13422996	G>C	SNV	no	no	no
SMIM10L2B	no	11A	3	A>C	UTR3	no	no	no
KRBOX4	no	11A	X:46333875	G>T	UTR3	no	no	no
ITM2A	no	11A	X:78616868 10:1197684	T>G	nonsynonymous SNV	no	no	no
RAB11FIP2	no	11B	00	T>C	UTR3	no	no	no
RAB11FIP2	no	11D	10:1197684 00	T>C	UTR3	no	no	no
CACUL1	no	11B	10:1204445 26	A>T	UTR3	no	no	no
CACUL1	no	11D	10:1204445 26	A>T	UTR3	no	no	no
GPR26	no	11B	10:1254491 68	G>C	UTR3	no	no	no
PTPRE	no	11B	10:1298759 63	A>G	nonsynonymous SNV	no	no	no
PTPRE	no	11D	10:1298759 63	A>G	nonsynonymous SNV	no	no	no
SFTPA2	no	11B	10:8131917 9	C>A	stopgain	no	no	no
SFTPA2	no	11D	10:8131917 9	C>A	stopgain	no	no	no
B3GAT1	no	11B	11:1342823 65	C>A	upstream	no	no	no
RCN1	no	11B	11:3212495 2	T>G	nonsynonymous SNV	no	yes	yes
OR4A15	no	11B	11:5513563 3	G>T	nonsynonymous SNV	no	no	no
GPR152	no	11B	11:6721985 9	C>T	nonsynonymous SNV	no	yes	yes
CPT1A	no	11B	11:6857493 9	C>G	nonsynonymous SNV	no	no	no
CPT1A	no	11D	11:6857493 9	C>G	nonsynonymous SNV	no	no	no
OAS1	no	11B	12:1133447 01	G>A	UTR5	no	no	no
LRCOL1	no	11B	12:1331875 38	C>T	upstream	no	no	no
LRCOL1	no	11D	12:1331875 38	C>T	upstream	no	no	no
ATF7	no	11B	12:5391853 3	G>A	stopgain	no	no	no
ATF7	no	11D	12:5391853 3	G>A	stopgain	no	no	no
MIR196A2	no	11B	12:5438542 7	T>C	upstream	no	yes	yes
MIR196A2	no	11D	12:5438542 7	T>C	upstream	no	yes	yes
RNF41	no	11B	12:5660428 0	C>A	nonsynonymous SNV	no	no	no
APAF1	no	11B	12:9904246 2	AT>A	frameshift deletion	no	no	no
APAF1	no	11D	12:9904246 2	AT>A	frameshift deletion	no	no	no
GRK1	no	11B	13:1143221 85	T>A	nonsynonymous SNV	no	no	no
GRK1	no	11D	13:1143221 85	T>A	nonsynonymous SNV	no	no	no

ALG5;EXOS C8	no	11B	13:3757448 0	T>G	upstream	no	no	no
ALG5;EXOS C8	no	11D	13:3757448 0	T>G	upstream	no	no	no
NDFIP2	no	11B	13:8012683 7	A>G	UTR3	no	yes	yes
NDFIP2	no	11D	13:8012683 7	A>G	UTR3	no	yes	yes
EIF5	no	11B	14:1038102 09	T>A	UTR3	no	no	no
HERC2	no	11B	15:2843619 0	A>C	nonsynonymous SNV	no	no	no
HERC2	no	11D	15:2843619 0	A>C	nonsynonymous SNV	no	no	no
STOML1	no	11B	15:7427705 0	A>C	nonsynonymous SNV	no	no	no
LPCAT2	no	11B	16:5561797 5	C>A	UTR3	no	no	no
LPCAT2	no	11D	16:5561797 5	C>A	UTR3	no	no	no
ERN1	no	11B	17:6220785 2	C>G	upstream	no	no	no
ERN1	no	11D	17:6220785 2	C>G	upstream	no	no	no
RGS9	no	11B	17:6313351 7	G>C	UTR5	no	no	no
RGS9	no	11D	17:6313351 7	G>C	UTR5	no	no	no
PHF23	no	11B	17:7138401	C>T	UTR3	no	no	no
PHF23	no	11D	17:7138401 17:7294839	C>T	UTR3 nonsynonymous SNV	no	no	no
HID1	no	11B	19:1530280 4	G>A	nonsynonymous SNV	no	no	yes
NOTCH3	no	11B	19:1530280 4	C>T	nonsynonymous SNV	no	no	no
NOTCH3	no	11D	19:1530280 4	C>T	nonsynonymous SNV	no	no	no
PGPEP1	no	11B	19:1848032 8	T>G	UTR3	no	no	no
ZNF226	no	11B	19:4467095 7	T>G	UTR5	no	no	no
ZNF226	no	11D	19:4467095 7	T>G	UTR5	no	no	no
TGFBR3L	no	11B	19:7983903 1:10120455	A>C	UTR3	no	no	no
VCAM1	no	11B	1:11121748 0	G>A	UTR3	no	no	no
KCNA3	no	11B	1:11121748 1	C>A	UTR5	no	no	no
KCNA3	no	11D	1:11121748 1	C>A	UTR5	no	no	no
SIKE1	no	11B	1:11531218 5	T>C	UTR3	no	no	no
SIKE1	no	11D	1:11531218 5	T>C	UTR3	no	no	no
PTGS2	no	11B	1:18664300 5	A>G	UTR3	no	no	no
PTGS2	no	11D	1:18664300 5	A>G	UTR3	no	no	no
KIF14	no	11B	1:20058790 6	T>G	UTR5	no	no	no
KIF14	no	11D	1:20058790 6	T>G	UTR5	no	no	no
INSM1	no	11B	20:2034973 5	A>C	nonsynonymous SNV	no	no	no
INSM1	no	11D	20:2034973 5	A>C	nonsynonymous SNV	no	no	no
KCNQ2	no	11B	20:6203634 9	C>T	UTR3	no	no	no
KCNQ2	no	11D	20:6203634 9	C>T	UTR3	no	no	no

ACTR3	no	11B	2:11471542 7	C>T	UTR3	no	no	no
ACTR3	no	11D	2:11471542 7	C>T	UTR3	no	no	no
TMEM214	no	11B	2:27255853	G>C	UTR5	no	no	no
TMEM214	no	11D	2:27255853	G>C	UTR5	no	no	no
RNF144A	no	11B	2:7181792 3:13653773	G>C	UTR3	no	no	no
SLC35G2	no	11B	0	A>C	upstream	no	no	no
SLC35G2	no	11D	3:13653773 0	A>C	upstream	no	no	no
ZBTB47	no	11B	3:42707398	A>G	UTR3	no	no	no
ZBTB47	no	11D	3:42707398	A>G	UTR3	no	no	no
CHDH	no	11B	3:53880109	C>G	splicing	no	no	no
CHDH	no	11D	3:53880109	C>G	splicing	no	no	no
GRPEL1	no	11B	4:7061401	T>C	UTR3	no	no	no
GRPEL1	no	11D	4:7061401	T>C	UTR3	no	no	no
PCGF3	no	11B	4:762451	A>G	UTR3	no	no	no
PCGF3	no	11D	4:762451	A>G	UTR3	no	no	no
SKP1	no	11B	5:13349313 0	A>C	UTR3	no	no	no
SKP1	no	11D	5:13349313 0	A>C	UTR3	no	no	no
FAM105A	no	11B	5:14612838	T>C	UTR3	no	no	no
FAM105A	no	11D	5:14612838	T>C	UTR3	no	no	no
BRD9	no	11B	5:864144	C>T	UTR3	no	no	no
ELOVL4	no	11B	6:80657380 7:11272323	T>A	upstream	no	no	no
GPR85	no	11B	2	G>A	UTR3	no	no	no
GPR85	no	11D	7:11272323 2	G>A	UTR3	no	no	no
NDUFA5	no	11B	7:12318134 7	T>C	UTR3	no	no	no
CHRM2	no	11B	7:13655317 8	G>A	upstream	no	no	no
CHRM2	no	11D	7:13655317 8	G>A	upstream	no	no	no
LYNX1	no	11B	8:14385537 8	C>G	UTR3	no	no	no
LYNX1	no	11D	8:14385537 8	C>G	UTR3	no	no	no
NSMAF	no	11B	8:59499151	A>C	stopgain	no	no	no
NSMAF	no	11D	8:59499151	A>C	stopgain	no	no	no
RAB2A	no	11B	8:61535145	T>G	UTR3	no	no	no
RAB2A	no	11D	8:61535145	T>G	UTR3	no	no	no
LINC01508	no	11B	9:93196308	G>T	upstream	no	no	no
LINC01508	no	11D	9:93196308	G>T	upstream	no	no	no
MID2	no	11B	X:10714884 6	A>C	nonsynonymous SNV	no	no	no
MID2	no	11D	X:10714884 6	A>C	nonsynonymous SNV	no	no	no
GABRE	no	11B	X:15112222 4	G>T	UTR3	no	no	no
GABRE	no	11D	X:15112222 4	G>T	UTR3	no	no	no
TM9SF3	no	11C	10:9827798 5	T>C	UTR3	no	yes	yes

ATP8A2	no	11C	13:2594633 4	C>A	UTR5	no	no	yes
SLTM	no	11C	15:5920578 1	A>T	nonsynonymous SNV	no	no	no
UNC45A	no	11C	15:9147954 5	C>G	nonsynonymous SNV	no	no	no
DHX58	no	11C	17:4026130 4	C>T	nonsynonymous SNV	no	no	no
FZD2	no	11C	17:4263774 5	G>A	UTR3	no	no	no
SERPINB10	no	11C	18:6160277 7	C>A	UTR3	no	no	no
KDM5B	no	11C	1:20269805 6	T>C	UTR3	no	yes	yes
MICAL3	no	11C	22:1834877 1	G>GA	frameshift insertion	no	no	no
AP1B1	no	11C	22:2978459 3	C>T	upstream nonsynonymous SNV	no	no	no
GMNC	no	11C	3:19057346 0	C>A	SNV	no	no	no
WDR6	no	11C	3:49044609 5:11044050	T>C	UTR5 nonsynonymous SNV	no	no	no
WDR36	no	11C	8 5:18043366	T>A	SNV	no	no	no
BTNL3	no	11C	0	G>A	UTR3	no	no	no
LSP1P3	no	11C	5:28925989	C>A	upstream	no	no	no
LOC105374 620	no	11C	5:2917725	T>C	upstream nonsynonymous SNV	no	no	no
CKMT2	no	11C	5:80554985	A>C	SNV	no	no	no
LUZP6;MTP N	no	11C	7:13561174 4	A>C	UTR3	no	no	no
MICALL2	no	11C	7:1499555	A>C	upstream	no	no	yes
RPS20;SNO RD54	no	11C	8:56987318 9:13823746	G>C	upstream	no	no	no
C9orf62	no	11C	7	G>T	UTR3	no	no	no
ZDHHC21	no	11C	9:14612730	CTA>C	UTR3	no	no	yes
RASEF	no	11C	9:85677921 10:1316349	C>T	UTR5	no	no	no
EBF3	no	11D	71	T>G	UTR3	no	no	no
QSER1	no	11D	11:3299821 5	A>G	UTR3	no	no	no
MS4A4E	no	11D	11:5996875 5	T>G	UTR3	no	no	no
MS4A10	no	11D	11:6056877 6	G>A	UTR3	no	yes	yes
SYT7	no	11D	11:6129057 2	A>C	nonsynonymous SNV	no	no	no
DIS3	no	11D	13:7333139 2	C>CT	UTR3	no	yes	yes
TTC6	no	11D	14:3815207 7	T>G	nonsynonymous SNV	no	no	no
IGF1R	no	11D	15:9945647 8	A>G	nonsynonymous SNV	no	no	no
MYH1	no	11D	17:1039938 2	A>T	nonsynonymous SNV	no	no	no
ZNF624	no	11D	17:1652483 6	T>C	UTR3	no	no	no
CDC37	no	11D	19:1050191 7	G>C	UTR3	no	no	no
ANO8	no	11D	19:1743972 3	A>G	nonsynonymous SNV	no	no	no
ZNF554	no	11D	19:2835281 1:10974835	T>A	UTR3	no	no	no
KIAA1324	no	11D	1	T>A	UTR3	no	no	no
MAVS	no	11D	20:3856491	T>G	UTR3	no	yes	yes

			22:4132321					
XPNPEP3	no	11D	1	G>A	UTR3	no	no	no
			2:16293184					
DPP4	no	11D	2	A>C	upstream	no	no	no
ARL6IP5	no	11D	3:69134130	A>G	UTR5	no	yes	yes
			5:14099870					
DIAPH1	no	11D	5	G>C	upstream	no	no	no
			5:14956907					
SLC6A7	no	11D	4	ACT>A	upstream	no	no	no
			5:17595456					
RNF44	no	11D	7	A>T	UTR3	no	no	no
			5:17974089		nonsynonymous			
GFPT2	no	11D	8	A>T	SNV	no	no	no
					nonsynonymous			
ADAMTS12	no	11D	5:33641960	C>T	SNV	no	yes	yes
			6:12304630		nonsynonymous			
PKIB	no	11D	7	C>G	SNV	no	no	no
KIF13B	no	11D	8:28927606	C>T	UTR3	no	no	no
					nonsynonymous			
GAS1	no	11D	9:89561201	C>G	SNV	no	no	no
BRWD3	no	11D	X:80065084	C>T	UTR5	no	no	no

References

- Larson, N.B., and Fridley, B.L. (2013). PurBayes: estimating tumor cellularity and subclonality in next-generation sequencing data. *Bioinformatics* 29, 1888-1889.
- McLaren, W., Pritchard, B., Rios, D., Chen, Y., Flicek, P., and Cunningham, F. (2010). Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor. *Bioinformatics* 26, 2069-2070.
- Nersisyan, L., and Arakelyan, A. (2015). Computel: computation of mean telomere length from whole-genome next-generation sequencing data. *PLoS One* 10, e0125201.
- Tang, K.W., Alaei-Mahabadi, B., Samuelsson, T., Lindh, M., and Larsson, E. (2013). The landscape of viral expression and host gene fusion and adaptation in human cancer. *Nat Commun* 4, 2513.
- Tubio, J.M.C., Li, Y., Ju, Y.S., Martincorena, I., Cooke, S.L., Tojo, M., Gundem, G., Pipinikas, C.P., Zamora, J., Raine, K., *et al.* (2014). Mobile DNA in cancer. Extensive transduction of nonrepetitive DNA mediated by L1 retrotransposition in cancer genomes. *Science* 345, 1251343.
- Wang, K., Li, M., and Hakonarson, H. (2010). ANNOVAR: functional annotation of genetic variants from high-throughput sequencing data. *Nucleic Acids Res* 38, e164.