# Supplementary Results

The *SLC35A2* gene has been previously associated with mild MCD or FCD type 1[1,2] but was more recently confirmed as a genetic marker for the MOGHE phenotype.[3–5] We originally identified four FCD 1a or mMCD samples with *SLC35A2* variants. However, upon in-depth review of histopathological imaging, we determined that the pathological phenotypes of these samples more closely matched MOGHE and reclassified them accordingly. Thus, we detected alterations in *SLC35A2* exclusively in MOGHE samples. Accordingly, we observed a strong association between *SLC35A2* and MOGHE, with 47.1% (16/35) of MOGHE samples carrying a variant in *SLC35A2*.

As expected, alterations in *FGFR1* were exclusively identified in DNET (29%, 7/24), with multiple alterations identified in the majority of cases (71%, 5/7 *FGFR1*-DNET).[6,7] Variants in *BRAF* were strongly associated with gangliogliomas[8], with 77.6% (52/67) of detected *BRAF* alterations occurring in gangliogliomas and 57.5% (46/80) of gangliomas carrying at least one alteration in *BRAF*. Of these, the majority (40/46, 87%) carried the *BRAFV600E* variant. Furthermore, *BRAFV600E* was detected in all three pleomorphic xanthoastrocytomas[7] samples in our cohort. Of note, we observed six gangliogliomas with both a somatic *BRAF* SNV and a somatic duplication covering the *BRAF* gene (7.5%), one sample with two separate somatic duplications covering *BRAF*, and one sample with two different SNV in *BRAF* (V600E and A764V).

Our gene enrichment analysis revealed a strong association between LEAT and *PTPN11* (Figure 3). Our somatic variant gene discovery also revealed novel associations between epileptogenic brain lesions and two different RAS genes, *NRAS* and *KRAS*. Alterations in *NRAS* showed a strong association with polymicrogyria (PMG) and were exclusively detected in 17% of PMG (2/12). We detected a singular somatic *KRAS* SNV in a cancer driving site in a case of meningioangiomatosis (50%, 1/2), a lesion typically associated with the germline neurofibromatosis 2, but not somatic variants in *NF2*.[9,10] To the best of our knowledge, we are the first to report an association between RAS genes and epileptogenic lesions. Lastly, we identified an association between somatic alterations in *NF1* and epileptogenic brain lesions. We detected nine *NF1* alterations across seven LEAT, which included two gangliogliomas, two DNET, and three gliomas (Fig 3A). Among these, we identified DNET sample carrying two distinct somatic *NF1* SNV (a splicing variant and a stopgain) and a pilocytic astrocytoma carrying a somatic *NF1* PTV and a somatic CNN-LOH covering *NF1*. We also identified an additional seven *NF1* alterations across five MCD, which

included four FCD II and a single PMG (Fig 3A). From these, we only detected more than one variant in the PMG, which carried three distinct *NF1* alterations (T30K, G26X, and a somatic CNN-LOH covering *NF1*). Our results expand the spectrum of epileptogenic brain lesions associated to variants in the canonical RAS-RAF-MAPK pathway and serve as a foundation for untangling the role of this pathway in lesional epilepsies.

**Supplementary Table 1: Panel of Normals used in the somatic variant analysis**

| Sample | Sex | Age at surgery | Pathology |
|---|---|---|---|
| PON_CCF_1 | Female | 65 | No visible lesion on histopathology |
| PON_CCF_2 | Female | 21 | No visible lesion on histopathology |
| PON_CCF_3 | Male | 25 | No visible lesion on histopathology |
| PON_CCF_4 | Female | 32 | No visible lesion on histopathology |
| PON_CCF_5 | Male | 47 | No visible lesion on histopathology |
| PON_CCF_6 | Male | 54 | No visible lesion on histopathology |
| PON_CCF_7 | Male | 16 | No visible lesion on histopathology |
| PON_CCF_8 | Female | 61 | No visible lesion on histopathology |
| PON_CCF_9 | Female | 47 | No visible lesion on histopathology |
| PON_EEBB_1 | Male | 33 | Glial Scar |
| PON_EEBB_2 | Male | 38 | Glial Scar |
| PON_EEBB_3 | Male | 25 | No visible lesion on histopathology |
| PON_EEBB_4 | Male | 30 | Encephalitis / Glial Scar |
| PON_EEBB_5 | Male | 29 | No visible lesion on histopathology |
| PON_EEBB_6 | Female | 27 | Rasmussen Encephalitis |
| PON_EEBB_7 | Female | 42 | No visible lesion on histopathology |
| PON_EEBB_8 | Male | 34 | Limbic Encephalitis |
| PON_EEBB_9 | Female | 54 | Glial Scar |
| PON_EEBB_10 | Male | 44 | No visible lesion on histopathology |
| PON_EEBB_11 | Male | 27 | No visible lesion on histopathology |
| PON_EEBB_12 | Female | 44 | No visible lesion on histopathology |
| PON_EEBB_13 | Female | 39 | No visible lesion on histopathology |
| PON_EEBB_14 | Female | 36 | No visible lesion on histopathology |
| PON_EEBB_15 | Male | 37 | No visible lesion on histopathology |

**Supplementary Table 2: Top hits from gene enrichment analysis**

| Gene | Synonymous variants | Missense variants | Nonsense variants | Splice variants | Insertions/ deletions | $P_{global}$ (unadjusted) | $Q_{global}$ |
|---|---|---|---|---|---|---|---|
| **Hippocampal Sclerosis (n=97)** | | | | | | | |
| *NUDT14* | 0 | 0 | 0 | 0 | 1 | 0.001052 | 1 |
| *TEX45* | 0 | 0 | 0 | 0 | 1 | 0.001724 | 1 |
| *FKBP10* | 0 | 0 | 0 | 0 | 1 | 0.001858 | 1 |
| *ARNT* | 0 | 0 | 0 | 0 | 1 | 0.00216 | 1 |
| *UNC5C* | 0 | 0 | 0 | 0 | 1 | 0.002331 | 1 |
| **Malformations of cortical development (n=223)** | | | | | | | |
| *SLC35A2* | 0 | 7 | 2 | 0 | 3 | 0 | **0** |
| *MTOR* | 0 | 11 | 0 | 0 | 1 | 2.56E-10 | **2.57E-06** |
| *AKT3* | 0 | 3 | 0 | 0 | 0 | 3.47E-04 | 1 |
| *OR9G1* | 0 | 4 | 0 | 0 | 0 | 7.59E-04 | 1 |
| *OR9G9* | 0 | 4 | 0 | 0 | 0 | 7.59E-04 | 1 |
| *NRAS* | 0 | 2 | 0 | 0 | 0 | 0.002669 | 1 |
| *CD3G* | 0 | 0 | 0 | 0 | 1 | 0.003661 | 1 |
| **Low-grade epilepsy-associated tumors (n=154)** | | | | | | | |
| *BRAF* | 0 | 39 | 0 | 0 | 3 | 0 | **0** |
| *PTPN11* | 0 | 6 | 0 | 0 | 0 | 4.19E-08 | **4.21E-04** |
| *FGFR1* | 0 | 4 | 0 | 0 | 0 | 2.62E-05 | 0.175515 |
| *OR4C3* | 0 | 4 | 0 | 0 | 0 | 3.16E-04 | 1 |
| *AURKC* | 0 | 1 | 0 | 0 | 1 | 3.59E-04 | 1 |
| *NF1* | 0 | 0 | 0 | 0 | 2 | 7.76E-04 | 1 |
| *RARA* | 0 | 1 | 0 | 0 | 1 | 0.001494 | 1 |
| *HSPA2* | 0 | 1 | 0 | 0 | 1 | 0.002737 | 1 |
| *GNRH2* | 0 | 0 | 0 | 0 | 1 | 0.004502 | 1 |

**Supplementary Table 3: Identified cancer driver variants**

| Gene | Known driver site | Samples with variants | Associated pathology | Specific sample pathologies |
|---|---|---|---|---|
| *BRAF* | p.V600 | 45 | LEAT | Ganglioglioma, PXA, MVNT |
| *MTOR* | p.S2215 | 3 | MCD | FCD type IIb, HME |
| | p.T1977 | 1 | MCD | FCD type IIb |
| | p.L1460 | 1 | MCD | FCD type IIb |
| *NRAS* | p.Q61 | 2 | MCD | PMG |
| *PTPN11* | p.A72 | 2 | LEAT | DNET |
| | p.E76 | 1 | LEAT | Ganglioglioma |
| *KRAS* | p.G12 | 1 | LEAT | Meningioangiomatosis |
| *PIK3CA* | p.V344 | 1 | LEAT | DNET |
| | p.E545 | 1 | MCD | HME |

**Legend:** PXA = Pleomorphic xanthoastrocytoma, MVNT = Multinodular vacuolated neuronal tumor, FCD = Focal cortical dysplasia, HME = Hemimegalencephaly, PMG = Polymicrogyria, DNET = Dysembryoplastic Neuroepithelial tumor

**Supplementary Table 4: Reported SNV in 19 lesional epilepsy genes**

| Sample | Sex | Age at onset | Age at surgery | Lesion group | Pathology | Additional findings | Gene | Variant position(hg19) | Transcript ID | cDNA change | Amino acid change | Variant allelic fraction | Variant class | REVEL score | CADD score | ClinVar class. | ACMG pathogenicity criteria* | ACMG pathogenicity prediction |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| HS_EEBB_20 | F | 3 | 46 | HS | HS 1 | FCD 3a | *MTOR* | chr1:11188137 | NM_004958 | c.C5957G | p.A1986G | 0.47 | Missense | 0.552 | 25.9 | Not in ClinVar | PM2 PP2 PP3 | VUS† |
| HS_EEBB_41 | F | 1 | 35 | HS | HS 1 | | *PTPN11* | chr12:112926885 | NM_002834 | c.C1505T | p.S502L | 0.02 | Missense | | 34 | P/LP | PS1 PM2 PP2 PP3 PP5 | LP† |
| HS_EEBB_53 | M | 1 | 47 | HS | HS 1 | | *TSC1* | chr9:135772010 | NM_000368 | c.G3107A | p.G1036E | 0.5 | Missense | 0.237 | 24.7 | CloP | PM2 | VUS† |
| HS_EEBB_71 | M | 45 | 70 | HS | HS 1 | | *MTOR* | chr1:11301631 | NM_004958 | c.C1520T | p.P507L | 0.47 | Missense | 0.446 | 24.3 | Not in ClinVar | PM2 PP2 | VUS† |
| HS_EEBB_17 | M | 20 | 26 | HS | HS 2 | FCD 3a | *PTEN* | chr10:89720649 | NM_001304717 | c.1321-2A>T | Splicing | 0.03 | PTV | | 35 | Not in ClinVar | PVS1 PM2 PM4 PP3 | P† |
| LEAT_EEBB_1 | F | 14 | 15 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.17 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_10 | F | 6 | 16 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.15 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_10 | F | 6 | 16 | LEAT | GG | | *PTPN11* | chr12:112888210 | NM_002834 | c.G226A | p.E76K | 0.02 | Missense | | 33 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP5 | P |
| LEAT_EEBB_100 | M | 12 | 18 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.17 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_100 | M | 12 | 18 | LEAT | GG | | *BRAF* | chr7:140426307 | NM_001354609 | c.C2291T | p.A764V | 0.03 | Missense | | 22.9 | Not in ClinVar | PS4 PM2 PP2 PP4 | LP |
| LEAT_EEBB_103 | F | 18 | 23 | LEAT | GG | HS Type 1 | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.36 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_106 | F | 0.7 | 31 | LEAT | GG | HS Type 1 | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.04 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_107 | M | 0.3 | 9 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.04 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_108 | F | 7 | 7 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.05 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_12 | M | 1.1 | 4 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.1 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_14 | M | | 17 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.04 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_15 | M | 15 | 17 | LEAT | GG | HS | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.13 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_15 | M | 15 | 17 | LEAT | GG | HS | *PTPN11* | chr12:112888165 | NM_002834 | c.G181A | p.D61N | 0.03 | Missense | | 33 | P | PS1 PS4 PM2 PP2 PP3 PP5 | P |
| LEAT_EEBB_15 | M | 15 | 17 | LEAT | GG | HS | *PTPN11* | chr12:112891083 | NM_002834 | c.G417C | p.E139D | 0.03 | Missense | | 27.4 | P | PS1 PS4 PM2 PP2 PP3 PP5 | P |

| Sample | Sex | Age at onset | Age at surgery | Lesion group | Pathology | Additional findings | Gene | Variant position(hg19) | Transcript ID | cDNA change | Amino acid change | Variant allelic fraction | Variant class | REVEL score | CADD score | ClinVar class. | ACMG pathogenicity criteria* | ACMG pathogenicity prediction |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_22 | M | | 3 | LEAT | GG | HS Type I | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.09 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_22 | M | | 3 | LEAT | GG | HS Type I | *NF1* | chr17:29579966 | NM_001042492 | c.A4121G | p.H1374R | 0.44 | Missense | 0.451 | 23.9 | Not in ClinVar | PS4 PM2 PP2 | LP |
| LEAT_EEBB_22 | M | | 3 | LEAT | GG | HS Type I | *TSC2* | chr16:2130303 | NM_000548 | c.G3535A | p.E1179K | 0.44 | Missense | 0.576 | 23.6 | CloP | PM2 | VUS |
| LEAT_EEBB_27 | M | 16 | 19 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.15 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_32 | F | 20 | 22 | LEAT | GG | | *BRAF* | chr7:140453137 | NM_004333 | c.1798delinsACAG | p.T599_V600insT | 0.26 | Missense | | 22.2 | Not in ClinVar | PS1 PS4 PM2 PM5 PP2 PP4 | P |
| LEAT_EEBB_34 | M | | 4 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.19 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_37 | M | 1 | 44 | LEAT | GG | HS Type I | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.04 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_39 | F | 4 | 28 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.12 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_40 | F | 14 | 25 | LEAT | GG | HS Type I | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.2 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.08 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_43 | F | 0.8 | 2.9 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.25 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_44 | M | 1.1 | 4 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.04 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_47 | F | 0.9 | 4 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.08 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_52 | M | 13 | 20 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.09 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_53 | F | 10 | 19 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.04 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_54 | M | 1 | 6 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.18 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_57 | M | 0.7 | 5 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.13 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_61 | M | 1 | 2 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.15 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_62 | M | 48 | 49 | LEAT | GG | | *TSC1* | chr9:135772994 | NM_000368 | c.G2629T | p.V877L | 0.03 | Missense | 0.344 | 23 | Not in ClinVar | PM2 | VUS |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_63 | M | 13 | 15 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.12 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_65 | M | 1.1 | 5 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.23 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_67 | M | | 5 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.12 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_73 | M | 4 | 6 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.14 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_74 | M | 8 | 12 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.14 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_76 | F | 6 | 15 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.16 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_78 | M | 2 | 8 | LEAT | GG | HS Type 1 | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.04 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_80 | M | 5.7 | 6 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.19 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_84 | M | 0.3 | 2 | LEAT | GG | HS Type 2 | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.21 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_84 | M | 0.3 | 2 | LEAT | GG | HS Type 2 | *PIK3CA* | chr3:178942554 | NM_006218 | c.C2361A | p.D787E | 0.47 | Missense | 0.574 | 25.9 | Not in ClinVar | PM2 PP3 | VUS |
| LEAT_EEBB_85 | M | | 32 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.13 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_90 | F | | 47 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.06 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_90 | F | | 47 | LEAT | GG | | *PTEN* | chr10:89720649 | NM_001304717 | c.1321-2A>T | Splicing | 0.06 | PTV | | 35 | Not in ClinVar | PVS1 PM2 PM4 PP3 | P |
| LEAT_EEBB_91 | F | 6 | 11 | LEAT | GG | | *BRAF* | chr7:140477841 | NM_004333 | c.1465_1467 delinsA | p.A489Tfs*12 | 0.03 | PTV | | 34 | Not in ClinVar | PS4 PM2 PM4 PP3 PP4 | P |
| LEAT_EEBB_94 | M | 6 | 9 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.41 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_96 | M | 8.4 | 9.5 | LEAT | GG | HS Type 1 | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.11 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_98 | M | 15 | 18 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.12 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_EEBB_99 | F | 3 | 8.9 | LEAT | GG | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.12 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| LEAT_CCF_6 | F | 30 | 33.6 | LEAT | DNET | | *PTPN11* | chr12:112926900 | NM_002834 | c.C1520A | p.T507K | 0.02 | Missense | | 34 | P | PS1 PS4 PM2 PP2 PP3 PP5 | P |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_105 | F | 38 | 39 | LEAT | DNET | | *FGFR1* | chr8:38272308 | NM_001174067 | c.A2059G | p.K687E | 0.18 | Missense | 0.9 | 27.8 | P/LP | PS1 PM2 PP3 PP4 PP5 | LP |
| LEAT_EEBB_105 | F | 38 | 39 | LEAT | DNET | | *FGFR1* | chr8:38274850 | NM_001174067 | c.A1730G | p.N577S | 0.11 | Missense | 0.735 | 26.5 | Not in ClinVar | PM2 PP3 PP4 | VUS |
| LEAT_EEBB_105 | F | 38 | 39 | LEAT | DNET | | *NF1* | chr17:29554604 | NM_001042492 | c.2389delG | p.A797Pfs*24 | 0.04 | PTV | | 34 | LP | PVS1 PS4 PM2 PM4 PP3 | P |
| LEAT_EEBB_33 | M | 14 | 17 | LEAT | DNET | | *PIK3CA* | chr3:178952085 | NM_006218 | c.A3140G | p.H1047R | 0.03 | Missense | | 20.1 | P | PS1 PM2 PP5 | LP |
| LEAT_EEBB_49 | F | 30 | 33 | LEAT | DNET | | *FGFR1* | chr8:38274849 | NM_001174067 | c.C1731A | p.N577K | 0.18 | Missense | 0.646 | 29.7 | P | PS1 PM2 PP3 PP4 PP5 | LP |
| LEAT_EEBB_49 | F | 30 | 33 | LEAT | DNET | | *NF1* | chr17:29665035 | NM_001042492 | c.6705-8_6705-1delinsA | Splicing | 0.05 | PTV | | 24.7 | Not in ClinVar | PVS1 PS4 PM2 PM4 | P |
| LEAT_EEBB_49 | F | 30 | 33 | LEAT | DNET | | *NF1* | chr17:29552151 | NM_001042492 | c.C1884G | p.Y628X | 0.05 | PTV | | 34 | Not in ClinVar | PVS1 PS4 PM2 PM4 PM5 PP3 | P |
| LEAT_EEBB_49 | F | 30 | 33 | LEAT | DNET | | *PTPN11* | chr12:112888199 | NM_002834 | c.C215T | p.A72V | 0.03 | Missense | | 32 | CloP | PS4 PM1 PM2 PM5 PP2 PP3 | P |
| LEAT_EEBB_6 | M | 13 | 14 | LEAT | DNET | | *FGFR1* | chr8:38272308 | NM_001174067 | c.A2059G | p.K687E | 0.42 | Missense | 0.9 | 27.8 | P/LP | PS1 PM2 PP3 PP4 PP5 | LP |
| LEAT_EEBB_6 | M | 13 | 14 | LEAT | DNET | | *FGFR1* | chr8:38272308 | NM_001174067 | c.A2059C | p.K687Q | 0.42 | Missense | 0.733 | 27.6 | Not in ClinVar | PM2 PM5 PP3 PP4 | LP |
| LEAT_EEBB_6 | M | 13 | 14 | LEAT | DNET | | *FGFR1* | chr8:38272319 | NM_001174067 | c.A2048G | p.D683G | 0.41 | Missense | 0.866 | 31 | Not in ClinVar | PM2 PP3 PP4 | VUS |
| LEAT_EEBB_60 | M | 19 | 22 | LEAT | DNET | | *FGFR1* | chr8:38272308 | NM_001174067 | c.A2059G | p.K687E | 0.04 | Missense | 0.9 | 27.8 | P/LP | PS1 PM2 PP3 PP4 PP5 | LP |
| LEAT_EEBB_60 | M | 19 | 22 | LEAT | DNET | | *FGFR1* | chr8:38272322 | NM_001174067 | c.T2045C | p.I682T | 0.03 | Missense | 0.721 | 29.4 | Not in ClinVar | PM2 PP3 PP4 | VUS |
| LEAT_EEBB_81 | M | 45 | 59 | LEAT | DNET | | *FGFR1* | chr8:38274849 | NM_001174067 | c.C1731A | p.N577K | 0.23 | Missense | 0.646 | 29.7 | P | PS1 PM2 PP3 PP4 PP5 | LP |
| LEAT_EEBB_81 | M | 45 | 59 | LEAT | DNET | | *PTPN11* | chr12:112888199 | NM_002834 | c.C215G | p.A72G | 0.02 | Missense | | 28.2 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP5 | P |
| LEAT_EEBB_83 | M | 6 | 12 | LEAT | DNET | | *FGFR1* | chr8:38272302 | NM_001174067 | c.A2065C | p.T689P | 0.31 | Missense | 0.673 | 22.8 | Not in ClinVar | PM2 PP4 | VUS |
| LEAT_EEBB_83 | M | 6 | 12 | LEAT | DNET | | *FGFR1* | chr8:38272307 | NM_001174067 | c.A2060T | p.K687M | 0.3 | Missense | 0.876 | 29 | Not in ClinVar | PM2 PM5 PP3 PP4 | LP |
| LEAT_EEBB_83 | M | 6 | 12 | LEAT | DNET | | *FGFR1* | chr8:38272321 | NM_001174067 | c.C2046G | p.I682M | 0.3 | Missense | 0.493 | 23 | Not in ClinVar | PM2 PP4 | VUS |
| LEAT_EEBB_83 | M | 6 | 12 | LEAT | DNET | | *PIK3CA* | chr3:178921549 | NM_006218 | c.T1031G | p.V344G | 0.03 | Missense | | 27.3 | LP | PM1 PM2 PM5 PP3 | LP |
| LEAT_EEBB_2 | F | | 35 | LEAT | AG | | *DEPDC5* | chr22:32206593 | NM_001242896 | c.G1411A | p.G471S | 0.54 | Missense | 0.25 | 24.1 | VUS | PM2 | VUS† |
| LEAT_EEBB_31 | M | 12 | 13 | LEAT | MA | FCD 3c | *KRAS* | chr12:25398284 | NM_033360 | c.G35A | p.G12D | 0.05 | Missense | | 25.3 | P | PS1 PM1 PM2 PP3 PP5 | P |
| LEAT_EEBB_113 | M | 2 | 47 | LEAT | MVNT | | *BRAF* | chr7:140453138 | NM_004333 | c.1797delinsTACA | p.T599_V600insT | 0.14 | Missense | | 22.2 | Not in ClinVar | PS1 PS4 PM2 PP2 | P |
| LEAT_EEBB_95 | F | 6 | 39 | LEAT | PA | | *NF1* | chr17:29677325 | NM_001042492 | c.7446_7450delinsC | p.S2484Ifs*4 | 0.13 | PTV | | | Not in ClinVar | PVS1 PS4 PM2 PM4 | P |
| LEAT_EEBB_38 | M | 11 | 16 | LEAT | PXA | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.12 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP5 | P |

| Sample | Sex | Age at onset | Age at surgery | Lesion group | Pathology | Additional findings | Gene | Variant position(hg19) | Transcript ID | cDNA change | Amino acid change | Variant allelic fraction | Variant class | REVEL score | CADD score | ClinVar class. | ACMG pathogenicity criteria* | ACMG pathogenicity prediction |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.41 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP5 | P |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | *NF1* | chr17:29559097 | NM_001042492 | c.G3204T | p.L1068F | 0.28 | Missense | | 25.6 | LB | PS4 PM2 PP2 PP3 | LP |
| LEAT_EEBB_59 | M | | 16 | LEAT | PXA | | *BRAF* | chr7:140453136 | NM_004333 | c.T1799A | p.V600E | 0.22 | Missense | 0.931 | 32 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP5 | P |
| LEAT_CCF_37 | M | 0.8 | 7.3 | LEAT | TSC | SEGA | *TSC2* | chr16:2106223 | NM_000548 | c.626_628delinsC | p.A210Vfs*24 | 0.02 | PTV | | 21.3 | Not in ClinVar | PVS1 PM2 PM4 PP4 | P |
| LEAT_EEBB_17 | F | 0.1 | 9 | LEAT | TSC | SEGA | *PTPN11* | chr12:112888165 | NM_002834 | c.G181C | p.D61H | 0.05 | Missense | | 29.4 | P | PS1 PS4 PM2 PP2 PP3 PP5 | P |
| LEAT_EEBB_17 | F | 0.1 | 9 | LEAT | TSC | SEGA | *TSC2* | chr16:2106719 | NM_000548 | c.724dupA | p.T242Nfs*96 | 0.46 | PTV | | 25.9 | NP | PVS1 PM2 PM4 PP3 PP4 | P |
| MCD_CCF_19 | M | 10 | 25 | MCD | MOGHE | | *SLC35A2* | chrX:48762551 | NM_005660 | c.632_635del | p.L211Pfs*137 | 0.05 | PTV | | | Not in ClinVar | PVS1 PS4 PM2 PM4 PP3 PP4 | P |
| MCD_CCF_43 | M | 0 | 8.2 | MCD | MOGHE | | *SLC35A2* | chrX:48762557 | NM_005660 | c.G629A | p.C210Y | 0.05 | Missense | 0.667 | 26.2 | Not in ClinVar | PS4 PM2 PP3 PP4 | LP |
| MCD_CCF_77 | F | 1.5 | 2.5 | MCD | MOGHE | FCD1a | *DEPDC5* | chr22:32161000 | NM_001242896 | c.G233C | p.R78P | 0.4 | Missense | 0.21 | 24.8 | VUS | PM2 | VUS† |
| MCD_CCF_79 | F | 1 | 2 | MCD | MOGHE | | *SLC35A2* | chrX:48763748 | NM_005660 | c.C347A | p.A116E | 0.14 | Missense | 0.704 | 28.4 | Not in ClinVar | PS4 PM2 PP3 PP4 | LP |
| MCD_CCF_108 | M | 7 | 25.1 | MCD | MOGHE | | *SLC35A2* | chrX:48767173 | NM_005660 | c.192delinsACCGC | p.F65Pfs*30 | 0.24 | PTV | | 29.1 | Not in ClinVar | PVS1 PS4 PM2 PM4 PP3 PP4 | P |
| MCD_EEBB_2 | F | 0.5 | 7.1 | MCD | MOGHE | | *SLC35A2* | chrX:48762251 | NM_005660 | c.C935T | p.S312F | 0.08 | Missense | 0.657 | 26 | Not in ClinVar | PS4 PM2 PP3 PP4 | LP |
| MCD_EEBB_17 | F | 24 | 59 | MCD | MOGHE | | *SLC35A2* | chrX:48762671 | NM_005660 | c.T515C | p.L172P | 0.03 | Missense | | 26 | LP | PS4 PM2 PP3 PP4 | LP |
| MCD_EEBB_34 | F | 12 | 43.6 | MCD | MOGHE | | *SLC35A2* | chrX:48763729 | NM_005660 | c.366delinsTCTC | p.Y122_T123insL | 0.09 | Missense | | 20.5 | Not in ClinVar | PS4 PM2 PP4 | LP |
| MCD_EEBB_34 | F | 12 | 43.6 | MCD | MOGHE | | *SLC35A2* | chrX:48763730 | NM_005660 | c.364_365insC | p.Y122Sfs*6 | 0.15 | PTV | | 32 | Not in ClinVar | PVS1 PS4 PM2 PM4 PP3 PP4 | P |
| MCD_EEBB_37 | M | 3 | 27 | MCD | MOGHE | | *SLC35A2* | chrX:48762518 | NM_005660 | c.665_668delinsT | p.K222del | 0.29 | Missense | | 20.7 | Not in ClinVar | PS4 PM2 PP4 | LP |
| MCD_EEBB_58 | M | 0 | 4 | MCD | MOGHE | | *SLC35A2* | chrX:48762239 | NM_005660 | c.921_947delinsT | p.S308Wfs*106 | 0.11 | PTV | | 33 | Not in ClinVar | PVS1 PS4 PM2 PM4 PP3 PP4 | P |
| MCD_EEBB_63 | M | 3 | 7.3 | MCD | MOGHE | | *SLC35A2* | chrX:48762510 | NM_005660 | c.675dupA | p.G226Rfs*29 | 0.4 | PTV | | 33 | Not in ClinVar | PVS1 PS4 PM2 PM4 PP3 PP4 | P |
| MCD_EEBB_69 | F | 15 | 44 | MCD | MOGHE | | *SLC35A2* | chrX:48762546 | NM_005660 | c.G640C | p.G214R | 0.08 | Missense | 0.725 | 27.9 | Not in ClinVar | PS4 PM2 PP3 PP4 | LP |
| MCD_EEBB_87 | F | 0 | 6 | MCD | MOGHE | | *SLC35A2* | chrX:48763773 | NM_005660 | c.C322T | p.Q108X | 0.09 | PTV | | 36 | Not in ClinVar | PVS1 PS4 PM2 PM4 PP3 PP4 | P |
| MCD_EEBB_97 | F | 0.5 | 17.1 | MCD | MOGHE | | *NPRL2* | chr3:50385595 | NM_006545 | c.A892C | p.I298L | 0.04 | Missense | | 23 | Not in ClinVar | PM2 | VUS† |
| MCD_EEBB_97 | F | 0.5 | 17.1 | MCD | MOGHE | | *SLC35A2* | chrX:48762281 | NM_005660 | c.C905T | p.S302F | 0.08 | Missense | 0.814 | 26.6 | Not in ClinVar | PS4 PM2 PP3 PP4 | LP |
| MCD_EEBB_100 | M | | 6 | MCD | MOGHE | | *SLC35A2* | chrX:48763773 | NM_005660 | c.C322T | p.Q108X | 0.25 | PTV | | 36 | Not in ClinVar | PVS1 PS4 PM2 PM4 PP3 PP4 | P |
| MCD_EEBB_102 | F | 0.5 | 4.1 | MCD | MOGHE | | *SLC35A2* | chrX:48762251 | NM_005660 | c.C935T | p.S312F | 0.12 | Missense | 0.657 | 26 | Not in ClinVar | PS4 PM2 PP3 PP4 | LP |
| MCD_EEBB_106 | M | 2 | 6.5 | MCD | MOGHE | | *SLC35A2* | chrX:48762354 | NM_005660 | c.C832T | p.Q278X | 0.48 | PTV | | 38 | Not in ClinVar | PVS1 PS4 PM2 PM4 PP3 PP4 | P |

| Sample | Sex | Age at onset | Age at surgery | Lesion group | Pathology | Additional findings | Gene | Variant position(hg19) | Transcript ID | cDNA change | Amino acid change | Variant allelic fraction | Variant class | REVEL score | CADD score | ClinVar class. | ACMG pathogenicity criteria* | ACMG pathogenicity prediction |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| MCD_EEBB_22 | F | 9 | 26 | MCD | FCD 1a | | *MTOR* | chr1:11307914 | NM_004958 | c.C1078T | p.R360W | 0.04 | Missense | 0.479 | 34 | Not in ClinVar | PM2 PP2 PP3 | VUS† |
| MCD_CCF_8 | M | 5 | 21.4 | MCD | FCD 2a | | *NF1* | chr17:29559092 | NM_001042492 | c.G3199T | p.D1067Y | 0.25 | Missense | 0.719 | 24 | LB | PM2 PP2 | VUS† |
| MCD_CCF_13 | M | 2 | 20.4 | MCD | FCD 2a | | *NF1* | chr17:29654761 | NM_001042492 | c.C5513G | p.S1838C | 0.43 | Missense | 0.777 | 27.4 | VUS | PM2 PP2 PP3 | VUS† |
| MCD_CCF_13 | M | 2 | 20.4 | MCD | FCD 2a | | *NPRL2* | chr3:50386328 | NM_006545 | c.C562T | p.Q188X | 0.53 | PTV | | 37 | Not in ClinVar | PVS1 PM2 PM4 PP3 PP4 | P |
| MCD_CCF_30 | M | 2 | 16.5 | MCD | FCD 2a | | *DEPDC5* | chr22:32193632 | NM_001242896 | c.G814T | p.V272L | 0.46 | Missense | 0.144 | 25.2 | CloP | PM2 PP3 PP4 | VUS |
| MCD_CCF_46 | F | 6 | 28.4 | MCD | FCD 2a | | *NPRL3* | chr16:142605 | ENST00000399953 | c.1149dupC | p.A384Rfs*71 | 0.5 | PTV | | 33 | Not in ClinVar | PVS1 PM2 PM4 PP3 PP4 | P |
| MCD_CCF_58 | F | 0 | 2.5 | MCD | FCD 2a | | *NPRL3* | chr16:143291 | ENST00000399953 | c.G957A | p.W319X | 0.56 | PTV | | 39 | Not in ClinVar | PVS1 PM2 PM4 PP3 PP4 | P |
| MCD_CCF_60 | F | 0.1 | 1.5 | MCD | FCD 2a | | *NPRL2* | chr3:50388010 | NM_006545 | c.73dupT | p.Y25Lfs*5 | 0.57 | PTV | | 32 | Not in ClinVar | PVS1 PM2 PM4 PP3 PP4 | P |
| MCD_CCF_80 | F | 47 | 56.3 | MCD | FCD 2a | | *DEPDC5* | chr22:32233122 | NM_001242896 | c.C2335G | p.P779A | 0.49 | Missense | 0.271 | 24.1 | Not in ClinVar | PM2 PP4 | VUS |
| MCD_CCF_83 | F | 0.5 | 9.6 | MCD | FCD 2a | | *NPRL3* | chr16:136827 | ENST00000399953 | c.1584delT | p.M529Cfs*23 | 0.48 | PTV | | 27.4 | Not in ClinVar | PVS1 PM2 PM4 PP3 PP4 | P |
| MCD_EEBB_18 | M | 5.5 | 11 | MCD | FCD 2a | | *BRAF* | chr7:140549970 | NM_004333 | c.A181G | p.I61V | 0.42 | Missense | 0.288 | 20.8 | Not in ClinVar | PM2 PM5 PP2 | VUS† |
| MCD_EEBB_56 | M | 0 | 0.4 | MCD | FCD 2a | | *NPRL3* | chr16:143268 | ENST00000399953 | c.C980T | p.P327L | 0.42 | Missense | 0.885 | 33 | Not in ClinVar | PM2 PP3 PP4 | VUS |
| MCD_EEBB_75 | M | 2.8 | 12 | MCD | FCD 2a | | *DEPDC5* | chr22:32239185 | NM_001242896 | c.C2620T | p.R874X | 0.43 | PTV | 0.114 | 44 | P | PVS1 PS1 PM2 PM4 PP3 PP4 PP5 | P |
| MCD_EEBB_86 | M | 18 | 40 | MCD | FCD 2a | | *MTOR* | chr1:11217230 | NM_004958 | c.G4448A | p.C1483Y | 0.05 | Missense | | 32 | LP | PS4 PM2 PM5 PP2 PP3 PP4 | P |
| MCD_EEBB_88 | M | 0.7 | 5 | MCD | FCD 2a | | *NPRL3* | chr16:143268 | ENST00000399953 | c.C980T | p.P327L | 0.46 | Missense | 0.885 | 33 | Not in ClinVar | PM2 PP3 PP4 | VUS |
| MCD_EEBB_91 | M | 9 | 36 | MCD | FCD 2a | | *DEPDC5* | chr22:32188751 | NM_001242896 | c.C715T | p.R239X | 0.42 | PTV | | 38 | P | PVS1 PS1 PM2 PM4 PP3 PP4 PP5 | P |
| MCD_CCF_28 | F | 0.5 | 6.3 | MCD | FCD 2b | | *TSC2* | chr16:2103361 | NM_000548 | c.T244C | p.W82R | 0.48 | Missense | 0.926 | 25.6 | VUS | PM2 PM5 PP3 PP4 | LP |
| MCD_CCF_38 | M | 4 | 28.7 | MCD | FCD 2b | | *MTOR* | chr1:11217299 | NM_004958 | c.T4379C | p.L1460P | 0.03 | Missense | | 26.1 | P/LP | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| MCD_CCF_48 | F | 7 | 27 | MCD | FCD 2b | | *DEPDC5* | chr22:32161000 | NM_001242896 | c.G233C | p.R78P | 0.37 | Missense | 0.21 | 24.8 | VUS | PM2 PP4 | VUS |
| MCD_CCF_48 | F | 7 | 27 | MCD | FCD 2b | | *TSC2* | chr16:2122364 | NM_000548 | c.G2220T | p.M740I | 0.48 | Missense | 0.803 | 23.2 | Not in ClinVar | PM2 PM5 PP3 PP4 | LP |
| MCD_CCF_49 | F | 2 | 39.2 | MCD | FCD 2b | | *MTOR* | chr1:11188975 | NM_004958 | c.G5748T | p.W1916C | 0.46 | Missense | 0.478 | 27.7 | VUS | PS4 PM2 PP2 PP3 PP4 | LP |
| MCD_CCF_53 | M | 1 | 16.5 | MCD | FCD 2b | | *MTOR* | chr1:11169376 | NM_004958 | c.T7499A | p.I2500N | 0.04 | Missense | 0.455 | 25 | Not in ClinVar | PS4 PM2 PP2 PP3 PP4 | LP |
| MCD_CCF_78 | M | 0.4 | 1.3 | MCD | FCD 2b | | *DEPDC5* | chr22:32200837 | NM_001242896 | c.C1153T | p.R385W | 0.43 | Missense | 0.237 | 34 | VUS | PM2 PP3 PP4 | VUS |
| MCD_CCF_78 | M | 0.4 | 1.3 | MCD | FCD 2b | | *NPRL3* | chr16:188253 | ENST00000399953 | c.C14T | p.T5I | 0.55 | Missense | 0.094 | 23.3 | VUS | PM2 PP4 | VUS |
| MCD_CCF_92 | M | 6 | 14.8 | MCD | FCD 2b | | *MTOR* | chr1:11187847 | NM_004958 | c.T6050C | p.I2017T | 0.08 | Missense | 0.724 | 29.5 | Not in ClinVar | PS4 PM2 PP2 PP3 PP4 | LP |

| Sample | Sex | Age at onset | Age at surgery | Lesion group | Pathology | Additional findings | Gene | Variant position(hg19) | Transcript ID | cDNA change | Amino acid change | Variant allelic fraction | Variant class | REVEL score | CADD score | ClinVar class. | ACMG pathogenicity criteria* | ACMG pathogenicity prediction |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| MCD_CCF_101 | F | 0 | 0.9 | MCD | FCD 2b | | *MTOR* | chr1:11184573 | NM_004958 | c.C6644A | p.S2215Y | 0.03 | Missense | 0.857 | 32 | P/LP | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| MCD_CCF_102 | M | 7 | 19.5 | MCD | FCD 2b | | *MTOR* | chr1:11169377 | NM_004958 | c.A7498T | p.I2500F | 0.04 | Missense | | 33 | LP | PS4 PM2 PP2 PP3 PP4 | LP |
| MCD_CCF_102 | M | 7 | 19.5 | MCD | FCD 2b | | *TSC1* | chr9:135804196 | NM_000368 | c.C64T | p.R22W | 0.42 | Missense | 0.471 | 25.9 | CloP | PM2 PP3 PP4 | VUS |
| MCD_EEBB_32 | M | 13 | 24 | MCD | FCD 2b | | *NF1* | chr17:29701091 | NM_001042492 | c.C8438T | p.T2813I | 0.21 | Missense | 0.067 | 23.3 | Not in ClinVar | PM2 PP2 | VUS† |
| MCD_EEBB_67 | M | 20 | 32 | MCD | FCD 2b | | *NF1* | chr17:29677224 | NM_001042492 | c.G7345C | p.V2449L | 0.4 | Missense | 0.2 | 25.8 | Not in ClinVar | PM2 PP2 PP3 | VUS† |
| MCD_EEBB_71 | F | 3 | 10 | MCD | FCD 2b | | *MTOR* | chr1:11188164 | NM_004958 | c.C5930A | p.T1977K | 0.05 | Missense | | 34 | NP | PS4 PM1 PM2 PM5 PP2 PP3 PP4 | P |
| MCD_EEBB_80 | M | 9 | 19 | MCD | FCD 2b | | *MTOR* | chr1:11184573 | NM_004958 | c.C6644T | p.S2215F | 0.03 | Missense | | 33 | P/LP | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| MCD_EEBB_85 | F | 4 | 32 | MCD | FCD 2b | | *PTEN* | chr10:89623887 | NM_001304717 | c.180_181ins AA | p.L61Nfs*2 | 0.49 | PTV | | | Not in ClinVar | PVS1 PM2 PM4 PM5 PP4 | P |
| MCD_EEBB_92 | M | 0.2 | 0.9 | MCD | FCD 2b | | *MTOR* | chr1:11174399 | NM_004958 | c.7276delins CCCT | p.P2425_L2426insP | 0.03 | Missense | | 19.06 | Not in ClinVar | PS4 PM2 PP2 PP4 | LP |
| MCD_CCF_93 | F | 0 | 4.1 | MCD | cMCD | HME / PMG / FCD 2a | *AKT3* | chr1:243859016 | NM_005465 | c.G49A | p.E17K | 0.08 | Missense | 0.372 | 29.9 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| MCD_CCF_94 | F | 0 | 0.4 | MCD | cMCD | HME / PMG / FCD 2a / FCD 1a | *PTEN* | chr10:89720720 | NM_000314 | c.872dupA | p.N292Kfs*6 | 0.74 | PTV | | 34 | P | PVS1 PS1 PM2 PM4 PP3 PP4 PP5 | P |
| MCD_CCF_110 | M | 0 | 0.2 | MCD | cMCD | HME / PMG / FCD 2a | *AKT3* | chr1:243859016 | NM_005465 | c.G49A | p.E17K | 0.13 | Missense | 0.372 | 29.9 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| MCD_EEBB_14 | F | | 1 | MCD | cMCD | HME / FCD 2a | *MTOR* | chr1:11184573 | NM_004958 | c.C6644A | p.S2215Y | 0.1 | Missense | 0.857 | 32 | P/LP | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| MCD_EEBB_21 | F | 0.2 | 1 | MCD | cMCD | HME / PMG / FCD 2a | *PIK3CA* | chr3:178936091 | NM_006218 | c.G1633A | p.E545K | 0.16 | Missense | 0.654 | 33 | P/LP | PS1 PM1 PM2 PP3 PP4 PP5 | P |
| MCD_EEBB_26 | F | 0.5 | 2 | MCD | cMCD | HME / PMG / FCD 2a | *PIK3CA* | chr3:178952085 | NM_006218 | c.A3140T | p.H1047L | 0.19 | Missense | 0.359 | 11.75 | P | PS1 PM2 PP4 PP5 | LP |
| MCD_EEBB_28 | M | 0.3 | 0.8 | MCD | cMCD | HME / FCD 2a | *MTOR* | chr1:11169377 | NM_004958 | c.A7498T | p.I2500F | 0.03 | Missense | | 33 | LP | PS4 PM2 PP2 PP3 PP4 | LP |
| MCD_EEBB_52 | F | 0 | 0.7 | MCD | cMCD | HME / PMG / FCD 2a | *PIK3CA* | chr3:178936082 | NM_006218 | c.G1624A | p.E542K | 0.27 | Missense | 0.439 | 27.3 | P/LP | PS1 PM2 PP3 PP4 PP5 | LP |
| MCD_EEBB_57 | F | 0.1 | 0.5 | MCD | cMCD | HME / PMG / FCD 2a | *AKT3* | chr1:243859016 | NM_005465 | c.G49A | p.E17K | 0.08 | Missense | 0.372 | 29.9 | P | PS1 PS4 PM1 PM2 PP2 PP3 PP4 PP5 | P |
| MCD_EEBB_57 | F | 0.1 | 0.5 | MCD | cMCD | HME / PMG / FCD 2a | *MTOR* | chr1:11313993 | NM_004958 | c.C743T | p.T248I | 0.47 | Missense | 0.223 | 21.4 | VUS | PS4 PM2 PP2 PP4 | LP |
| MCD_CCF_32 | M | 0 | 1.1 | MCD | cMCD | PMG / FCD 2a / FCD 1a | *NPRL3* | chr16:169168 | ENST00000399953 | c.G275A | p.R92Q | 0.3 | Missense | 0.311 | 33 | VUS | PM2 PM5 PP3 PP4 | LP |
| MCD_CCF_104 | F | 0 | 0.1 | MCD | cMCD | PMG / FCD 2b | *MTOR* | chr1:11301647 | NM_004958 | c.A1504C | p.K502Q | 0.09 | Missense | 0.215 | 25 | Not in ClinVar | PS4 PM2 PP2 PP3 PP4 | LP |
| MCD_CCF_104 | F | 0 | 0.1 | MCD | cMCD | PMG / FCD 2b | *TSC2* | chr16:2135289 | NM_000548 | c.A4628G | p.H1543R | 0.41 | Missense | 0.947 | 24.4 | Not in ClinVar | PM2 PM5 PP3 PP4 | LP |

| Sample | Sex | Age at onset | Age at surgery | Lesion group | Pathology | Additional findings | Gene | Variant position(hg19) | Transcript ID | cDNA change | Amino acid change | Variant allelic fraction | Variant class | REVEL score | CADD score | ClinVar class. | ACMG pathogenicity criteria* | ACMG pathogenicity prediction |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| MCD_EEBB_10 | M | 14 | 25 | MCD | cMCD | PMG / NH / FCD 2a / GG | *NRAS* | chr1:115256529 | NM_002524 | c.A182G | p.Q61R | 0.13 | Missense | 0.888 | 23.1 | P | PS1 PS4 PM1 PM2 PP3 PP5 | P |
| MCD_EEBB_65 | F | 1 | 20 | MCD | cMCD | PMG / NH / FCD 2a / DNET | *NRAS* | chr1:115256530 | NM_002524 | c.C181A | p.Q61K | 0.24 | Missense | 0.841 | 24.1 | VUS | PS4 PM1 PM2 PM5 PP3 | P |
| MCD_EEBB_76 | M | 0 | 0.7 | MCD | cMCD | PMG / FCD 2b | *MTOR* | chr1:11174399 | NM_004958 | c.7276delinsCCCT | p.P2425_L2426insP | 0.04 | Missense | | 19.06 | Not in ClinVar | PS4 PM2 PP2 PP4 | LP |
| MCD_EEBB_89 | M | 7 | 20 | MCD | cMCD | PMG / FCD 1b | *NF1* | chr17:29483029 | NM_001042492 | c.C89A | p.T30K | 0.03 | Missense | 0.332 | 25.8 | Not in ClinVar | PM2 PP2 PP3 | VUS† |
| MCD_EEBB_89 | M | 7 | 20 | MCD | cMCD | PMG / FCD 1b | *NF1* | chr17:29483016 | NM_001042492 | c.G76T | p.G26X | 0.46 | PTV | | 36 | Not in ClinVar | PVS1 PM2 PM4 PP3 | P† |
| MCD_CCF_27 | F | 0 | 6.5 | MCD | TSC | FCD2b | *TSC2* | chr16:2130169 | NM_000548 | c.3402dupC | p.H1135Pfs*33 | 0.45 | PTV | | 32 | Not in ClinVar | PVS1 PM2 PM4 PP3 PP4 | P |
| MCD_CCF_29 | F | 0.2 | 3.3 | MCD | TSC | FCD 2b | *TSC2* | chr16:2121603 | NM_000548 | c.C1932A | p.C644X | 0.46 | PTV | | 38 | Not in ClinVar | PVS1 PM2 PM4 PP3 PP4 | P |
| MCD_CCF_55 | M | 0.3 | 5.1 | MCD | TSC | FCD2b | *TSC2* | chr16:2136739 | NM_000548 | c.T4856C | p.F1619S | 0.46 | Missense | 0.986 | 28.5 | NP | PM2 PP3 PP4 | VUS |
| MCD_CCF_56 | F | 4 | 8.5 | MCD | TSC | FCD2b | *TSC2* | chr16:2107157 | NM_000548 | c.826_827del | p.M276Vfs*61 | 0.43 | PTV | | 26.7 | P | PVS1 PS1 PM2 PM4 PP3 PP4 PP5 | P |

**Legend to supplementary table 4**: HS = Hippocampal sclerosis, MCD = Malformation of cortical development, LEAT = Low-grade epilepsy-associated tumor, FCD = Focal cortical dysplasia, MOGHE = mild MCD with oligodendroglial hyperplasia in epilepsy, HME = Hemimegalencephaly, PMG = Polymicrogyria, cMCD = complex MCD, TSC = tuberous sclerosis complex, MA = meningioangiomatosis, GG = Ganglioglioma, AG = Angiocentric Glioma, DNET = Dysembryoplastic neuroepithelial tumor, PA = Pilocytic Astrocytoma, PXA = Pleomorphic xanthoastrocytoma, MVNT = multinodular vacuolated neurol tumor, SEGA = Subependymal giant cell astrocytoma, PTV = Protein-truncating variant, P = Pathogenic, LP = Likely pathogenic, B = Benign, LB = Likely benign, CIoP = Conflicting interpretations of pathogenicity, VUS = Variant of uncertain significance, NP = Not provided.

***** Variants were evaluated based on the following ACMG criteria: PVS1, PS1, PS4, PM1, PM2, PM4, PM5, PP2, PP3, and PP4. (PSV1 = predicted null variant in a gene where loss-of-function is a known mechanism of disease, PS1 = Same amino acid change as an established pathogenic variant, PS4 = statistically significant association with pathology, PM1 = Mutational hot sport or well-studied functional domain, PM2 = Absent in population databases, PM4 = protein length changing variant, PM5 = Novel missense change at an amino acid residue where a different pathogenic missense change has been seen before, PP2 = Missense in gene with low rate of benign missense variants, PP3 = Computational evidence supports a deleterious effect on the gene / gene product, PP4 = patient's phenotype is associated with the gene.)

† Gene-disease validity has not been established. ACMG pathogenicity criteria cannot be applied so classification was done only for research purposes,

**Supplementary Table 5: Somatic CNV and CNN-LOH**

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| HS_EEBB_55 | F | 16 | 49 | HS | HS (NOS) | | 9:86179563-141213431 | 9q21.33-34.2 | Loss | 55.03 | 0.03 | 2 | *TSC1* | *ABCA2, DNM1, GABBR2, GRIN1, NOTCH1, PHF19, SCAI, SET, SPTAN1, STXBP1, WDR5* | *SETX, TRAF2, OLFM1, LRRC8A, KIAA0368, RALGPS1, DAB2IP, GABBR2, ERP44, BRINP1, MAPKAP1, FAM120A, ZNF462, NOTCH1, SMC2, ZNF618, ZER1, WNK2, TSC1, ZMYND19, BRD3, ZNF483, SEC16A, RPL7A, ANAPC2, PTCH1, ANP32B, ABCA2, COL27A1, BICD2, PRRC2B, RAD23B, WDR5, PHF2, NUP214, ASTN2, NAA35, CACNA1B, GRIN1, RABL6, COL5A1, RALGDS, RAPGEF1, ABL1, NUP188, SET, SPTAN1, DNM1, ENG, STXBP1, ZBTB34, ZBTB43, SCAI, NR5A1, LHX2, DENND1A, RABGAP1, RC3H2, RAB14. PHF19, MEGF9, PRPF4, UGCG, EPB41L4B, KLF4, SLC44A1, TEX10, NCBP1, CDC14B, SYK, SPIN1, ZCCHC6, NTRK2, HNRNPK, UBQLN1, CAMSAP1, NTNG2, PPP2R4, GAPVD1, LHX6, SVEP1, DAPK1, GOLGA2, STRBP, SEMA4D, EHMT1, RXRA, NR6A1, FBXW2* |
| HS_EEBB_7 | M | 5 | 51 | HS | HS 1 | | 22:18520612-51304566 | 22q | CNN-LOH | 32.78 | 0.03 | 5 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, GNAZ, RAC2, CELSR1, SHANK3, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA11, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, HIC2, PLXNB2, TNRC6B, ZNRF3* |
| HS_EEBB_81 | F | 2 | 25 | HS | HS 1 | FCD 3a | 19:50401320-59128983 | 19q13.42-13.43 | CNN-LOH | 8.73 | 0.03 | 2 | | *BRSK1, PPP2R1A, PRKCG, PRPF31, SHANK1, U2AF2* | *TRIM28, ZNF331, PRKCG, SHANK1, U2AF2, BRSK1, PRPF31, PPP2R1A, LENG8, TTYH1, CNOT3, PPP6R1, RPS5, NUP62, MYH14* |
| HS_EEBB_17 | M | 20 | 26 | HS | HS 2 | FCD 3a | 19:54462369-59128983 | 19q13.42-13.43 | CNN-LOH | 4.67 | 0.02 | 2 | | *BRSK1, PRPF31, U2AF2* | *TRIM28, U2AF2, BRSK1, PRPF31, LENG8, TTYH1, CNOT3, PPP6R1, RPS5* |
| LEAT_EEBB_46 | M | 10 | 20 | LEAT | AG | | 6:135521495-141454143 | 6q23.3-24.1 | Loss | 5.93 | 0.69 | 2 | *MYB* | | *TNFAIP3, KIAA1244, REPS1, MAP3K5* |
| LEAT_EEBB_110 | M | 6 | 15 | LEAT | AG | | 6:135543837-163969815 | 6q23.3-24.1 | Loss | 28.43 | 0.20 | 2 | | | *FBXO5, TNFAIP3, KIAA1244, REPS1, TCP1, STXBP5, ZBTB2, ARID1B, IGF2R, WTAP, MAP3K5, PLAGL1, TULP4, SCAF8, TAB2, HIVEP2, MAP3K4, ESR1, LATS1* |
| LEAT_EEBB_19 | F | 15 | 16 | LEAT | DGG | | 5:71661131-180915260 | 5q13.2-35,2 | Loss | 109.25 | 0.23 | 1 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, RBM22, LOX, LNPEP, NR3C1, ELL2, IRF1, ADAMTS2, PDLIM4, NDFIP1, AP3B1, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, PDE8B, SOX30, VCAN, CPEB4, FBXW11, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, TCERG1, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, CTNNA1, GPRIN1, HSPA4, ZNF608, PCDHGC4, PWWP2A, KLHL3, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, NKX2-5, UNC5A, PURA, EFNA5, HOMER1, LARP1, PPP2R2B, TNPO1, DPYSL3, SEMA6A, DDX46, HNRNPH1, ETF1, NRG2, KIF3A, SNX2, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, FCHO2, NSD1, MGAT1, APC, PPP2CA, TCOF1, KCTD16, FBN2, FNIP1, KCNN2, GNB2L1, CTC-432M15.3, FAM193B, CSNK1A1, TENM2, GRIA1, RNF145, SLIT3, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_19 | F | 15 | 16 | LEAT | DGG | | 9:73909871-86175639 | 9q21.12-21.32 | Loss | 12.27 | 0.22 | 1 | | | *GDA, GNAQ, ALDH1A1, TLE1, TLE4, RORB* |
| LEAT_EEBB_19 | F | 15 | 16 | LEAT | DGG | | 9:88234954-137530346 | 9q21.33-34.2 | Loss | 49.30 | 0.23 | 2 | *TSC1* | *DNM1, GABBR2, PHF19, SCAI, SET, SPTAN1, STXBP1, WDR5* | *SETX, LRRC8A, KIAA0368, RALGPS1, DAB2IP, GABBR2, ERP44, BRINP1, MAPKAP1, FAM120A, ZNF462, SMC2, ZNF618, ZER1, WNK2, TSC1, BRD3, ZNF483, RPL7A, PTCH1, ANP32B, COL27A1, BICD2, PRRC2B, RAD23B, WDR5, PHF2, NUP214, ASTN2, NAA35, RALGDS, RAPGEF1, ABL1, NUP188, SET, SPTAN1, DNM1, ENG, STXBP1, ZBTB34, ZBTB43, SCAI, NR5A1, LHX2, DENND1A, RABGAP1, RC3H2, RAB14, PHF19, MEGF9, PRPF4, UGCG, EPB41L4B, KLF4, SLC44A1, TEX10, NCBP1, CDC14B, SYK, SPIN1, ZCCHC6, NTNG2, PPP2R4, GAPVD1, LHX6, SVEP1, DAPK1, GOLGA2, STRBP, SEMA4D, RXRA, NR6A1, FBXW2* |
| LEAT_EEBB_19 | F | 15 | 16 | LEAT | DGG | | 22:23754578-51304566 | 22q | Loss | 27.55 | 0.22 | 4 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *SF3A1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, RAC2, CELSR1, SHANK3, SMARCB1, EP300, PATZ1, KCNJ4, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA11, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, PLXNB2, TNRC6B, ZNRF3* |
| LEAT_EEBB_33 | M | 14 | 17 | LEAT | DNET | | 6:0-171115067 | 6 | Gain | 171.12 | 0.01 | 8 | *MYB* | *GTPBP2, JARID2, KLHDC3, PPP2R5D, TMEM63B, TRERF1, XPO5* | *TFAP2D, RANBP9, TRAM2, FBXO5, TNFAIP3, DST, ATXN1, KIAA1244, CUL9, BACH2, REPS1, FAM65B, KIF13A, TMEM63B, TTBK1, GCM1, POU5F1, PHF3, NUP153, PSMB1, XPO5, SRF, SCUBE3, PHIP, C6orf136, PPARD, GLTSCR1L, EEF1A1, DEF6, TCP1, STXBP5, COL12A1, ZBTB2, RPS10, KLHDC3, TUBB, EPB41L2, PRPF4B, JARID2, KCNQ5, ARID1B, E2F3, DOPEY1, FYN, IGF2R, BRPF3, WTAP, REV3L, MAP3K5, ANKS1A, SLC35F1, PLAGL1, ZNF318, DLL1, THBS2, TULP4, SCAF8, TAB2, HIVEP2, PTPRK, HSF2, NUS1, AMD1, CDK19, CDC40, NR2E1, ATG5, PRDM1, SIM1, PNISR, EPHA7, MAP3K7, MDN1, ZNF292, SYNCRIP, TBX18, SMAP1, BAI3, LRRC1, RUNX2, CDC5L, HSP90AB1, UBR2, TRERF1, CCND3, USP49, FOXP4, CMTR1, RNF8, SRSF3, SPDEF, PHF1, RXRB, COL11A2, PBX2, ZBTB12, BAG6, PRRC2A, PPP1R10, GNL1, TRIM39, GABBR1, TRIM27, C6orf62, HIVEP1, TFAP2A, DSP, RREB1, GMDS, MLLT4, MAP3K4, WASF1, TFAP2B, PFDN6, BRD2, AGPAT1, DDX39B, CDYL, DAAM2, SYNGAP1, FAM135A, GRIK2, BEND3, MDGA1, SNAP91, RPS18, ESR1, SENP6, PTK7, PPP2R5D, HDAC2, CCNC, RIMS1, L3MBTL3, GRM4, PACSIN1, PDE10A, LATS1* |
| LEAT_EEBB_36 | M | 3 | 8 | LEAT | DNET | | 7:138562547-140335570 | 7q34 | Gain | 1.77 | 0.20 | 2 | | | *MKRN1, HIPK2, KIAA1549, UBN2* |
| LEAT_EEBB_41 | M | 2 | 3 | LEAT | DNET | | 8:0-38684853 | 8p | CNN-LOH | 38.68 | 0.11 | 2 | *FGFR1* | *XPO7* | *XPO7, DLC1, ATP6V1B2, CCAR2, DPYSL2, PPP2R2A, WHSC1L1, EGR3, RBPMS, ASH2L, NRG1, FGF17, MAK16, CNOT7, LZTS1, HMBOX1, PTK2B, INTS10, UNC5D, XKR6, DLGAP2, FGFR1, PURG, EBF2* |
| LEAT_EEBB_49 | F | 30 | 33 | LEAT | DNET | | 19:44544237-59128983 | 19q13.31-13.4 | Loss | 14.58 | 0.05 | 1 | | *AP2A1, BRSK1, CA11, CALM3, CLPTM1, GRIN2D, IRF2BP1, LMTK3, NOVA2, PPFIA3, PPP2R1A, PPP5C, PRKCG,* | *PPP5C, BCL3, RELB, SLC17A7, RASIP1, VASP, SYMPK, TSKS, PVRL2, TRIM28, ZC3H4, ZNF331, MARK4, GRIN2D, PRKCG, SAE1, RPS11, SHANK1, IRF2BP1, ZNF296, U2AF2, BRSK1, PRPF31, PPP2R1A, LENG8, PPFIA3, CLPTM1, FOSB, AP2A1, SCAF1, TTYH1, STRN4, GLTSCR1, ARHGAP35, CNOT3, PPP6R1, PRR12, TOMM40, PRKD2, PRMT1, RPL18, RUVBL2, RPS5, NUP62, SNRNP70, MYH14, NPAS1* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | *PRMT1, PRPF31, PRR12, SHANK1, SLC8A2, TOMM40, U2AF2* | |
| LEAT_EEBB_49 | F | 30 | 33 | LEAT | DNET | | 20:0-22746255 | 20p | Loss | 22.75 | 0.03 | 1 | | *CENPB* | *CSNK2A1, JAG1, SNAP25, PCSK2, ATRN, LZTS3, PLCB1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH* |
| LEAT_EEBB_55 | M | 23 | 25 | LEAT | DNET | | 2:0-25424397 | 2p25.3-23.3 | Loss | 25.42 | 0.26 | 1 | | *VSNL1* | *TRIB2, ATAD2B, KIDINS220, ASAP2, RPS7, ADAM17, ROCK2, RNF144A, PUM2, DDX1, NCOA1, MYT1L* |
| LEAT_EEBB_55 | M | 23 | 25 | LEAT | DNET | | 2:25568821-63939417 | 2p23.3-15 | Gain | 38.37 | 0.21 | 1 | | *CAD, FBXO11, PPP1CB, XPO1* | *MSH2, NRBP1, PAPOLG, SIX3, EHBP1, CAD, RAB10, KIF3C, FOSL2, CRIM1, ZFP36L2, DPYSL5, REL, MEMO1, SIX2, PRKCE, SRSF7, SPAST, BCL11A, CCDC88A, PPM1G, ZNF512, SPTBN1, GTF3C2, CCT4, EFEMP1, PPP1CB, USP34, XPO1, SLC8A1, FBXO11, PSME4, NRXN1, WDR43, HNRNPLL, BIRC6, SOS1, ASXL2, EIF2B4* |
| LEAT_EEBB_55 | M | 23 | 25 | LEAT | DNET | | 8:49729020-94305178 | 8q11.21-22.1 | Loss | 44.58 | 0.11 | 1 | *MYBL1* | | *RB1CC1, RDH10, ARFGEF1, RAB2A, ST18, MMP16, VCPIP1, PLAG1, XKR4, ZNF704, EYA1, RPL7, COPS5, TOX, CHD7, ZBTB10, RUNX1T1, NCOA2, WWP1, STMN2, LYN, ZFHX4, MYBL1, KCNB2, STAU2, YTHDF3* |
| LEAT_EEBB_55 | M | 23 | 25 | LEAT | DNET | | 13:28150993-115169878 | 13 | Loss | 87.02 | 0.26 | 2 | | *MYCBP2, NBEA* | *AKAP11, FLT3, EFNB2, DCLK1, IPO5, KPNA3, RB1, RBM26, ZC3H13, FLT1, DACH1, INTS6, PDS5B, HSPH1, COG3, FBXL3, MYO16, CHAMP1, TFDP1, CUL4A, ARHGEF7, COL4A1, TNFSF13B, FAM155A, TPP2, TM9SF2, DOCK9, HS6ST3, SLITRK1, SPRY2, KLF12, KBTBD6, FOXO1, FRY, SLC7A1, PAN3, LRCH1, NBEA, MTUS2, TSC22D1, FNDC3A, MYCBP2* |
| LEAT_EEBB_55 | M | 23 | 25 | LEAT | DNET | | 15:23833737-60664030 | 15q11.2-22.2 | Loss | 36.83 | 0.05 | 1 | | *RTF1* | *AQR, MGA, DLL4, CTDSPL2, THBS1, ADAM10, HERC2, TTBK2, UBR1, SPRED1, COPS2, LEO1, MAP1A, PDIA3, PYGO1, OTUD7A, RASGRP1, GABRB3, SEMA6D, FBN1, TJP1, CASC5, ZNF770, INO80, SLTM, TP53BP1, RYR3, RTF1, UBE3A, MYO5A, BAHD1, FRMD5, RFX7, USP8, MAGEL2, DMXL2, RNF111, MEIS2* |
| LEAT_EEBB_75 | M | 6 | 12 | LEAT | DNET | | 2:0-243199373 | 2 | Gain | 243.20 | 0.01 | 3 | | *CAD, FBXO11, HDLBP, MAP4K4, PPP1CB, PPP3R1, RQCD1, SCN2A, SF3B1, SNRNP200, UNC80, VSNL1, XPO1* | *CYP26B1, INPP4A, TRIB2, TMEM131, MSH2, NRBP1, PCGF1, IGFBP5, PPP1R7, DLX2, PAPOLG, ATAD2B, AFTPH, ACVR2A, RIF1, ILKAP, PER2, KIDINS220, CCT7, USP37, REV1, EPC2, SIX3, PPIG, HECW2, SLC40A1, ACTR3, POLR1B, TFCP2L1, PTPN4, CLASP1, LRP2, POLR1A, EHBP1, ATF2, MCM6, CCNT2, R3HDM1, AGPS, ADAM23, CUL3, ADD2, CAD, RAB10, KIF3C, FOSL2, VPS54, MEIS1, AMMECR1L, CTDSP1, RQCD1, CRIM1, ASAP2, EPHA4, ZFP36L2, RANBP2, SCN3A, TRIP12, FMNL2, DPYSL5, EIF5B, REL, MEMO1, KCNJ3, EN1, INHBB, IWS1, FZD5, ARPC2, CTLA4, SIX2, SCN1A, ANTXR1, AGAP1, COL3A1, RPS7, SEMA4C, PRKCE, MAT2A, PSMD1, KLF7, CFLAR, SP3, SNED1, ADAM17, SPEG, SRSF7, SPAST, ROCK2, RAPH1, GLS, RNF144A, WDR33, NCL, SNRNP200, DIS3L2, FIGN, SF3B1, NPAS2, BCL11A, CCDC88A, PUM2, NR4A2, ERBB4, PPM1G, HDAC4, MAP4K4, FN1, ZNF512, STK39, AFF3, SPTBN1, GFPT1, SCN2A, ARID5A, SAP130, GTF3C2, INPP5D, MAP2, NCKAP1, STAT1, DLX1, BMPR2, COL5A2, RND3, ACTR2, DDX1, HSPD1, LRP1B, TBR1, HDLBP, ATG16L1, SPHKAP, XRCC5, STAT4, OSBPL6, HNRNPA3, BAZ2B, GALNT13, DPP10, CCT4, EFEMP1, SP9, PPP1CB, MOB1A, RAPGEF4, USP34, XPO1, SLC8A1, INO80D, FBXO11, PSME4, ATG4B, NRXN1, NCOA1, MBD5, WDR43, AAK1, SLC39A10, ARHGAP25, TET3, GIGYF2, ZNF638, KDM3A, ATG9A, HNRNPLL, MGAT5, PSMD14, KCMF1, MAP3K2, AGFG1, CALCRL, SATB2, UBR3, BIRC6, SOS1, TMEM185B, MYT1L, SESTD1, PAX8, CNTNAP5, TLK1, KANSL3, CREB1, KIF5C, ASXL2, UNC80, EIF2B4, GLI2, BZW1, IKZF2, KIF1A, FBXO41, REEP1, ZEB2, HSPE1-MOB4* |
| LEAT_EEBB_75 | M | 6 | 12 | LEAT | DNET | | 7:0-147731239 | 7 | Gain | 147.73 | 0.02 | 18 | *BRAF* | *CAMK2B, OGDH, PSMC2, RAC1* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, PTPN12, LRRC4, MKRN1, IGF2BP3, TTYH3, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, DGKI, BRAF, C7orf60, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, ZKSCAN1, GTF2I, AHCYL2, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, GRB10, TNS3, SEPT7, HIPK2, TBX20, FOXP2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, LHFPL3, CNOT4* |
| LEAT_EEBB_75 | M | 6 | 12 | LEAT | DNET | | 7:138550994-140481402 | 7q34 | Gain | 1.93 | 0.30 | 2 | *BRAF* | | *MKRN1, BRAF, HIPK2, KIAA1549, UBN2* |
| LEAT_EEBB_75 | M | 6 | 12 | LEAT | DNET | | 12:49424065-51343851 | 12q13.12 | Gain | 1.92 | 0.32 | 1 | | *SMARCD1* | *ASIC1, KCNH3, KMT2D, TUBA1A, DIP2B, TUBA1B, SMARCD1, LARP4, PRPF40B, SPATS2* |
| LEAT_EEBB_81 | M | 45 | 59 | LEAT | DNET | | 8:0-42446238 | 8p | CNN-LOH | 42.45 | 0.04 | 2 | *FGFR1* | *XPO7* | *SFRP1, XPO7, ANK1, DLC1, ATP6V1B2, CCAR2, DPYSL2, PPP2R2A, WHSC1L1, EGR3, RBPMS, SLC20A2, ASH2L, NRG1, FGF17, MAK16, CNOT7, LZTS1, KAT6A, HMBOX1, PTK2B, INTS10, UNC5D, XKR6, DLGAP2, FGFR1, PURG, EBF2, IKBKB, VDAC3* |
| LEAT_EEBB_104 | M | 5 | 17 | LEAT | DNET | | 4:0-191154276 | 4 | Gain | 191.15 | 0.12 | 5 | | *ABCE1, ANKRD17, CTBP1, DHX15, POLR2B, PPP3CA, SMARCA5, WDFY3* | *BOD1L1, PHOX2B, NFKB1, PDGFRA, UBE2K, KDR, PPAT, COPS4, RASGEF1B, RAPGEF2, FNIP2, ADD1, PPARGC1A, SEPT11, CNOT6L, CENPE, LEF1, SEC24B, CCNA2, NAF1, RPS3A, GPM6A, FBXW7, GRID2, SMARCA5, UCHL1, KIT, CTBP1, GABRB1, WDFY3, HMGB2, GRIA2, NAA15, HHIP, ABCE1, DCLK2, PDS5A, LDB2, PITX2, KIAA0232, REST, CLOCK, LIMCH1, HNRNPD, EDNRA, FAM193A, CRMP1, DHX15, TRIM2, RAP1GDS1, LIN54, FBXL5, RBPJ, YTHDC1, CLCN3, HTT, ANK2, NR3C2, FRYL, ANKRD17, SMARCAD1, G3BP2, PCGF3, ZNF827, POLR2B, ATP8A1, RBM47, RFC1, WHSC1, ELF2, FAT4, ELOVL6, PPP3CA, AFF1, CPE, JAKMIP1, KIAA0922, SLC4A4, BMPR1B, RPL9, RELL1, OTUD4, PLRG1, WDR1, BEND4, PDGFC, ANKRD50, SLIT2, CDKN2AIP, MAML3, GABRA2, TENM3, LPHN3, KLHL2, SMAD1* |
| LEAT_EEBB_104 | M | 5 | 17 | LEAT | DNET | | 6:0-171115067 | 6 | Gain | 171.12 | 0.11 | 8 | *MYB* | *GTPBP2, JARID2, KLHDC3, PPP2R5D, TMEM63B, TRERF1, XPO5* | *TFAP2D, RANBP9, TRAM2, FBXO5, TNFAIP3, DST, ATXN1, KIAA1244, CUL9, BACH2, REPS1, FAM65B, KIF13A, TMEM63B, TTBK1, GCM1, POU5F1, PHF3, NUP153, PSMB1, XPO5, SRF, SCUBE3, PHIP, C6orf136, PPARD, GLTSCR1L, EEF1A1, DEF6, TCP1, STXBP5, COL12A1, ZBTB2, RPS10, KLHDC3, TUBB, EPB41L2, PRPF4B, JARID2, KCNQ5, ARID1B, E2F3, DOPEY1, FYN, IGF2R, BRPF3, WTAP, REV3L, MAP3K5, ANKS1A, SLC35F1, PLAGL1, ZNF318, DLL1, THBS2, TULP4, SCAF8, TAB2, HIVEP2, PTPRK, HSF2, NUS1, AMD1, CDK19, CDC40, NR2E1, ATG5, PRDM1, SIM1, PNISR, EPHA7, MAP3K7, MDN1, ZNF292, SYNCRIP, TBX18, SMAP1, BAI3, LRRC1, RUNX2, CDC5L, HSP90AB1, UBR2, TRERF1, CCND3, USP49, FOXP4, CMTR1, RNF8, SRSF3, SPDEF, PHF1, RXRB, COL11A2, PBX2,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | *ZBTB12, BAG6, PRRC2A, PPP1R10, GNL1, TRIM39, GABBR1, TRIM27, C6orf62, HIVEP1, TFAP2A, DSP, RREB1, GMDS, MLLT4, MAP3K4, WASF1, TFAP2B, PFDN6, BRD2, AGPAT1, DDX39B, CDYL, DAAM2, SYNGAP1, FAM135A, GRIK2, BEND3, MDGA1, SNAP91, RPS18, ESR1, SENP6, PTK7, PPP2R5D, HDAC2, CCNC, RIMS1, L3MBTL3, GRM4, PACSIN1, PDE10A, LATS1* |
| LEAT_EEBB_104 | M | 5 | 17 | LEAT | DNET | | 7:0-159138663 | 7 | Gain | 159.14 | 0.12 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_104 | M | 5 | 17 | LEAT | DNET | | 9:0-141213431 | 9 | Gain | 141.21 | 0.12 | 2 | *TSC1* | *ABCA2, DNM1, GABBR2, GRIN1, NOTCH1, PHF19, SCAI, SET, SMU1, SPTAN1, STXBP1, TLN1, VCP, WDR5* | *SETX, GDA, CLTA, TAF1L, TRAF2, OLFM1, LRRC8A, KIAA0368, RALGPS1, DAB2IP, GABBR2, RNF38, ERP44, UBE2R2, BRINP1, MAPKAP1, UHRF2, FAM120A, ZNF462, NOTCH1, SMC2, GNAQ, ZNF618, ZER1, ALDH1A1, WNK2, NOL6, TSC1, ZMYND19, BRD3, ZNF483, SEC16A, TLN1, RPL7A, ANAPC2, PTCH1, TESK1, ANP32B, ABCA2, COL27A1, BICD2, PRRC2B, RAD23B, PAX5, WDR5, VCP, PHF2, NUP214, TOPORS, ASTN2, NAA35, CACNA1B, GRIN1, RABL6, COL5A1, RALGDS, RAPGEF1, ABL1, NUP188, SET, SPTAN1, DNM1, ENG, STXBP1, ZBTB34, ZBTB43, SCAI, NR5A1, LHX2, DENND1A, RABGAP1, RC3H2, RAB14, PHF19, MEGF9, PRPF4, UGCG, EPB41L4B, KLF4, SLC44A1, TEX10, NCBP1, CDC14B, SYK, SPIN1, ZCCHC6, NTRK2, HNRNPK, UBQLN1, TLE1, TLE4, RORB, SHB, CNTFR, TEK, MLLT3, RPS6, BNC2, PSIP1, NFIB, PTPRD, JAK2, CDC37L1, RFX3, SMARCA2, CAMSAP1, NTNG2, PPP2R4, GAPVD1, LHX6, SMU1, ELAVL2, SVEP1, DAPK1, GOLGA2, STRBP, SEMA4D, RUSC2, EHMT1, RXRA, NR6A1, FBXW2* |
| LEAT_EEBB_104 | M | 5 | 17 | LEAT | DNET | | 11:0-135006516 | 11 | Gain | 135.01 | 0.13 | 5 | | *ADRBK1, B3GNT1, CKAP5, CNIH2, DDB1, DPF2, GANAB, GNG3, GRAMD1B, KDM2A, KMT2A, LTBP3, MARK2, MEN1, MUC5B, NXF1, PCNXL3, PLCB3, PPP2R5B, PRPF19, PSMC3, RPS6KA4, SART1, SSRP1, SYVN1* | *TTC17, PPP2R5B, SPI1, PRPF19, MAPK8IP1, COPB1, PPFIA1, RRAS2, DAGLA, PRKRIR, DCUN1D5, EED, ARHGEF17, POLD3, FNBP4, OSBP, DDX6, ARCN1, NRXN2, FBXO3, GTF2H1, KCNC1, FAM160A2, LRRC4C, SERPING1, SSRP1, TENM4, ZC3H12C, HMBS, MYRF, FADS2, CWC15, VPS26B, YAP1, ZDHHC5, SIK3, C11orf84, LRP5, FAT3, PCF11, PKNOX2, PSMC3, APBB1, DCHS1, PATL1, RRM1, DDB1, HNRNPUL2, LTBP3, SUV420H1, SIK2, STIP1, CHRM1, RSF1, ADRBK1, RBM14, ARHGAP32, RAB6A, NPAS4, CNIH2, SART1, CLCF1, PPP1CA, AMBRA1, ZFP91, FJX1, PACS1, EHD1, DSCAML1, SF3B2, CSTF3, NUP98, KCNA4, PPME1, EEF1G, RPS6KA4, PDE2A, BCL9L, ZBTB16, SPTY2D1, C2CD2L, MEN1, USP47, CPSF7, CAPRIN1, GANAB, FAR1, ANO1, PCNXL3, UVRAG, FAM168A, MPPED2, CTR9, GAB2, DLG2, SYVN1, ATG2A, SF1, AHNAK, IPO7, BRSK2, ARNTL, CUL5, PICALM, NUMA1, PPP6R3, INCENP, NAV2, SOX6, ARHGEF12, CTNND1, QSER1, MARK2, HYOU1, RDX, RELA, RBM4, RBM14-RBM4, KLC2, PHF21A, GRM5, PSMA1, PAX6, ARRB1, TBCEL, PRDM11, PSMD13, CHRM4, AP2A2, WEE1, MAML2, TRIM3, KIRREL3, ZBTB44, TRAF6, FLI1, DPF2, ATG13, KDM2A, C11orf30, CREB3L1, CKAP5, MUC5B, GRAMD1B, API5, FZD4, RNF214, CELF1, NXF1, EIF3F, IGSF9B, ST5, KMT2A, HSPA8, OTUB1, PLCB3, SYT7* |
| LEAT_EEBB_104 | M | 5 | 17 | LEAT | DNET | | 14:19000000-107349540 | 14q | Gain | 88.35 | 0.12 | 2 | | *ACIN1, CHD8, DYNC1H1, JPH4, MTA1, PSMC6, RBM25, RNF31, SUPT16H, UNC79, ZNF219* | *MAP4K5, PPP1R13B, G2E3, PAPOLA, SUPT16H, SOS2, PSMA3, AHSA1, EIF5, NFKBIA, TGFB3, BMP4, ZC3H14, UNC79, VRTN, EIF2S1, PSMC1, PSMA6, SNW1, CYP46A1, RBM25, RPS6KA5, YY1, RCOR1, ACIN1, RTN1, ITPK1, AKAP6, ELMSAN1, FBXO33, TRIM9, PCNX, RALGAPA1, MMP14, HNRNPC, DDHD1, RNF31, PRMT5, PSEN1, PPM1A, YLPM1, TTC7B, MTA1, JAG2, EML1, SEL1L, PPP2R5E, DCAF5, FBLN5, FERMT2, ARHGAP5, ARID4A, PRPF39, NPAS3, CALM1, BCL11B, ATL1, TECPR2, DYNC1H1, BAZ1A, ZNF219, CDC42BPB, PSMB5, SPTB, CCNK, SAMD4A, INF2, EVL, ACTN1, ZBTB1, DAAM1, JPH4, HECTD1, CHD8, TOX4, EXOC5, CEP170B, PPP2R5C, MDGA2, BEGAIN, PSMC6, PACS2, SCFD1, GPHN, DICER1, HIF1A, NOVA1, GTF2A1, AKT1, SMEK1, NRXN3, SIPA1L1, NAA30, SRP54, TRAF3* |
| LEAT_EEBB_104 | M | 5 | 17 | LEAT | DNET | | 20:0-63025520 | 20 | Gain | 63.03 | 0.12 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_105 | F | 38 | 39 | LEAT | DNET | | 6:0-171115067 | 6 | Gain | 171.12 | 0.02 | 8 | *MYB* | *GTPBP2, JARID2, KLHDC3, PPP2R5D, TMEM63B, TRERF1, XPO5* | *TFAP2D, RANBP9, TRAM2, FBXO5, TNFAIP3, DST, ATXN1, KIAA1244, CUL9, BACH2, REPS1, FAM65B, KIF13A, TMEM63B, TTBK1, GCM1, POU5F1, PHF3, NUP153, PSMB1, XPO5, SRF, SCUBE3, PHIP, C6orf136, PPARD, GLTSCR1L, EEF1A1, DEF6, TCP1, STXBP5, COL12A1, ZBTB2, RPS10, KLHDC3, TUBB, EPB41L2, PRPF4B, JARID2, KCNQ5, ARID1B, E2F3, DOPEY1, FYN, IGF2R, BRPF3, WTAP, REV3L, MAP3K5, ANKS1A, SLC35F1, PLAGL1, ZNF318, DLL1, THBS2, TULP4, SCAF8, TAB2, HIVEP2, PTPRK, HSF2, NUS1, AMD1, CDK19, CDC40, NR2E1, ATG5, PRDM1, SIM1, PNISR, EPHA7, MAP3K7, MDN1, ZNF292, SYNCRIP, TBX18, SMAP1, BAI3, LRRC1, RUNX2, CDC5L, HSP90AB1, UBR2, TRERF1, CCND3, USP49, FOXP4, CMTR1, RNF8, SRSF3, SPDEF, PHF1, RXRB, COL11A2, PBX2, ZBTB12, BAG6, PRRC2A, PPP1R10, GNL1, TRIM39, GABBR1, TRIM27, C6orf62, HIVEP1, TFAP2A, DSP, RREB1, GMDS, MLLT4, MAP3K4, WASF1, TFAP2B, PFDN6, BRD2, AGPAT1, DDX39B, CDYL, DAAM2, SYNGAP1, FAM135A, GRIK2, BEND3, MDGA1, SNAP91, RPS18, ESR1, SENP6, PTK7, PPP2R5D, HDAC2, CCNC, RIMS1, L3MBTL3, GRM4, PACSIN1, PDE10A, LATS1* |
| LEAT_EEBB_105 | F | 38 | 39 | LEAT | DNET | | 7:0-159138663 | 7 | Gain | 159.14 | 0.08 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | *HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_1 | F | 14 | 15 | LEAT | GG | | 5:0-180915260 | 5 | Gain | 180.92 | 0.20 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_1 | F | 14 | 15 | LEAT | GG | | 7:0-159138663 | 7 | Gain | 159.14 | 0.21 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_1 | F | 14 | 15 | LEAT | GG | | 11:0-135006516 | 11 | Gain | 135.01 | 0.20 | 5 | | *ADRBK1, B3GNT1, CKAP5, CNIH2, DDB1, DPF2, GANAB, GNG3, GRAMD1B, KDM2A, KMT2A, LTBP3, MARK2, MEN1, MUC5B, NXF1, PCNXL3, PLCB3, PPP2R5B, PRPF19, PSMC3, RPS6KA4, SART1, SSRP1, SYVN1* | *TTC17, PPP2R5B, SPI1, PRPF19, MAPK8IP1, COPB1, PPFIA1, RRAS2, DAGLA, PRKRIR, DCUN1D5, EED, ARHGEF17, POLD3, FNBP4, OSBP, DDX6, ARCN1, NRXN2, FBXO3, GTF2H1, KCNC1, FAM160A2, LRRC4C, SERPING1, SSRP1, TENM4, ZC3H12C, HMBS, MYRF, FADS2, CWC15, VPS26B, YAP1, ZDHHC5, SIK3, C11orf84, LRP5, FAT3, PCF11, PKNOX2, PSMC3, APBB1, DCHS1, PATL1, RRM1, DDB1, HNRNPUL2, LTBP3, SUV420H1, SIK2, STIP1, CHRM1, RSF1, ADRBK1, RBM14, ARHGAP32, RAB6A, NPAS4, CNIH2, SART1, CLCF1, PPP1CA, AMBRA1, ZFP91, FJX1, PACS1, EHD1, DSCAML1, SF3B2, CSTF3, NUP98, KCNA4, PPME1, EEF1G, RPS6KA4, PDE2A, BCL9L, ZBTB16, SPTY2D1, C2CD2L, MEN1, USP47, CPSF7, CAPRIN1, GANAB, FAR1, ANO1, PCNXL3, UVRAG, FAM168A, MPPED2, CTR9, GAB2, DLG2, SYVN1, ATG2A, SF1, AHNAK, IPO7, BRSK2, ARNTL, CUL5, PICALM, NUMA1, PPP6R3, INCENP, NAV2, SOX6, ARHGEF12, CTNND1, QSER1, MARK2, HYOU1, RDX, RELA, RBM4, RBM14-RBM4, KLC2, PHF21A, GRM5, PSMA1, PAX6, ARRB1, TBCEL, PRDM11, PSMD13, CHRM4, AP2A2, WEE1, MAML2, TRIM3, KIRREL3, ZBTB44, TRAF6, FLI1, DPF2, ATG13, KDM2A, C11orf30, CREB3L1, CKAP5, MUC5B, GRAMD1B, API5, FZD4, RNF214, CELF1, NXF1, EIF3F, IGSF9B, ST5, KMT2A, HSPA8, OTUB1, PLCB3, SYT7* |
| LEAT_EEBB_1 | F | 14 | 15 | LEAT | GG | | 12:0-133851895 | 12 | Gain | 133.85 | 0.20 | 8 | *KRAS, PTPN11* | *ANKRD52, ATP2B1, CHD4, CS, DDX23, GCN1L1, HECTD4, LRP1, NAA25, NAB2, PA2G4, PLXNC1, RARG, SCN8A, SETD1B, SMARCC2, SMARCD1, STAT2, USP5* | *SENP1, HDAC7, FOXJ2, TNFRSF1A, CBX5, SART3, ASIC1, CSRNP2, CNOT2, USP5, HCFC2, RAB35, LRP1, SLC38A2, MSI1, HNF1A, TBX3, GDF11, ESPL1, KCNH3, KCNMB4, PLXNC1, SLC41A2, WBP11, ATF7IP, NAP1L1, ASUN, CDK17, CCND2, LRP6, CORO1C, LPCAT3, CUX2, LHX5, NAA25, ATP5B, PTGES3, EIF4B, COPZ1, CLSTN3, SMARCC2, ANKRD52, SETD1B, DUSP6, USP15, BTBD11, EPS8, MED13L, NCKAP1L, ITGA5, FRS2, CCT2, TMEM132B, HSP90B1, STAT6, GCN1L1, KMT2D, TUBA1A, DIP2B, PA2G4, LEMD3, DHX37, TBX5, STAT2, WNK1, MLXIP, PITPNM2, ULK1, EEA1, E2F7, SP1, FBXO21, RSRC2, XPOT, ARID2, PTPRB, TUBA1B, SARNP, ZCCHC8, FAM60A, NOS1, KSR2, PPP1CC, HNRNPA1, NTN4, DYRK2, RNF41, R3HDM2, CACNA1C, CS, PTPN11, SPPL3, SCN8A, SRGAP1, GIT2, MBD6, ATN1, CHD4, DNAJC14, RFX4, PCBP2, SCAF11, KDM2B, ATXN2, ZFC3H1, NABP2, COL2A1, NOP2, PRMT8, STK38L, DENND5B, EP400, CIT, TAOK3, RIC8B, MON2, SMARCD1, ETV6, ZNF384, CCDC64, LARP4, SLC38A1, AEBP2, KDM5A, SETD8, RASSF8, NCOR2, ATF7, SBNO1, TMEM132D, RPL6, RARG, PAN2, ZDHHC17, ATP2B1, PLEKHA5, DCTN2, CPSF6, NELL2, PPP1R12A, SOX5, SLC4A8, KIF5A, PRICKLE1, PTPN6, MDM2, NAV3, ATP2A2, CLIP1, ACVR1B, SFSWAP, RAN, PHC1, CAND1, MAP3K12, AGAP2, ANKS1B, PRPF40B, PPFIA2, HECTD4, BAZ2A, SPATS2, GRIN2B* |
| LEAT_EEBB_1 | F | 14 | 15 | LEAT | GG | | 19:0-59128983 | 19 | Gain | 59.13 | 0.20 | 5 | | *ACTN4, AP2A1, ATP1A3, BRD4, BRSK1, CA11, CACNA1A, CACTIN, CADM4, CALM3, CARM1, CHERP, CLPTM1, DAZAP1, DCAF15, DNMT1, DYRK1B, EGLN2, FZR1, GRIN2D, GSK3A, IRF2BP1, KHSRP, LMTK3, MAP4K1, MAST1, MAU2, NACC1, NOVA2, NUMBL, PIAS4, PPFIA3, PPP2R1A, PPP5C, PRKACA, PRKCG, PRMT1, PRPF31, PRR12, RFX1,* | *PPP5C, CD22, MBD3, ANO8, KDM4B, BCL3, REXO1, CDC34, HNRNPL, RELB, SLC17A7, SGTA, FBL, RASIP1, RPL18A, RAB3A, KMT2B, USF2, ERF, GSK3A, SIPA1L3, CADM4, DAZAP1, TSHZ3, C3, VASP, SYMPK, UBA2, TSKS, SUGP1, ABHD8, WDR18, MAST1, PVRL2, SAFB2, MLLT1, ACTN4, NUMBL, TRIM28, ZC3H4, EIF3G, ZNF331, RFX1, DCAF15, CSNK1G2, HPN, CCNE1, UPF1, ELL, MAST3, MARK4, GRIK5, MAP2K2, DPP9, PIAS4, GRIN2D, BRD4, WIZ, NOTCH3, PRKCG, ARID3A, SAE1, ADAMTS10, RPS11, RNF126, NACC1, CELF5, SHANK1, KIAA0355, RAB8A, CHAF1A, IRF2BP1, RFX2, ZNF296, LTBP4, PRKACA, U2AF2, EEF2, BRSK1, SAMD4B, PRPF31, PPP2R1A, LMNB2, HNRNPM, LENG8, STK11, CARM1, PPFIA3, PIP5K1C, CLPTM1, ARHGEF1, LRRC25, RANBP3, LPHN1, PDE4A, FOSB, AP3D1, DPF1, ZNF536, TMEM259, PTBP1, ATP13A1, PTPRS, XAB2, AP2A1, BABAM1, DNMT1, DDA1, CACNA1A, GATAD2A, SCAF1, LONP1, TTYH1, SIN3B, DNM2, STRN4, HNRNPUL1, MAU2, FZR1, GLTSCR1, NFIX, MAP2K7, KHSRP, DOT1L, ARHGAP35, CNOT3, ELAVL1, PPP6R1, PRR12, TNPO2, TOMM40, CACTIN, SMARCA4, PRKD2, CAMSAP3, ILF3, PRMT1, EPS15L1, ZNF146, UNC13A, POU2F2, APC2, EVI5L, HOMER3, ATP1A3, CHERP, RPL18, CIC, LRFN3, SAFB, MAP4K1, DKFZP761J1410, DYRK1B, FCHO1, RUVBL2, MYO9B, RPS5, NUP62, SNRNP70, SUPT5H, MYH14, SUGP2, VAV1, NPAS1, FKBP8* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | *SHANK1, SIN3B, SLC8A2, SMARCA4, SPTBN4, SUPT5H, TMEM259, TNPO2, TOMM40, U2AF2, UPF1, XAB2* | |
| LEAT_EEBB_1 | F | 14 | 15 | LEAT | GG | | 20:0-63025520 | 20 | Gain | 63.03 | 0.19 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_4 | M | 1 | 17 | LEAT | GG | | 10:118766183-123216161 | 10q25.3-26.13 | Loss | 4.45 | 0.22 | 1 | | | *PDZD8, MCMBP, TIAL1, EIF3A, CACUL1, EMX2* |
| LEAT_EEBB_11 | F | 8 | 14 | LEAT | GG | | 1:20142866-249250621 | 1 | Gain | 229.11 | 0.03 | 1 | *AKT3* | *AGO1, AGO3, AHCYL1, AKT3, ARF1, ARID1A, ATP1A1, CACNA1E, CELF3, CLK2, COPA, DENND4B, DHX9, ILF2, INTS3, IPO13, KDM1A, KDM4A, LMNA, MACF1, MEF2D, NCDN, NDUFS2, PEA15, PRPF3, RBBP4, RBBP5, SF3B4, TOMM40L, TRIM46, UBE2Q1, USP21, USP24, WDR26, ZMYM4* | *LRRC7, SARS, CHRM3, SORT1, NOTCH2, ZCCHC11, TAF5L, LAMC1, ARHGAP29, SUCO, TAF12, ARID4B, RBBP5, PSMA5, CELSR2, ILDR2, PRCC, SF3B4, SETDB1, POGZ, KCNN3, HNRNPU, WNT3A, NMNAT2, ZFYVE9, RAP1GAP, UBE2Q1, ZYG11B, USP24, GMEB1, ZNF281, ZNF496, NCSTN, SPRTN, CCT3, RGL1, SHE, LRP8, USP48, GLUL, PPP1R8, HP1BP3, ZMYM4, KCND3, RORC, INTS3, LPHN2, FMN2, YBX1, ANKRD34A, SRRM1, ZNF687, ARID1A, PRPF3, AHCTF1, KHDRBS1, LIN9, CSF1, LRRC8B, TRIM46, LCK, LRRC8D, PRRC2C, NFASC, USP1, LMX1A, JAK1, LRRC41, PTP4A2, PTPRU, MEF2D, LEPR, NASP, GRHL3, THRAP3, RNF220, MAN1A2, SFPQ, NEGR1, TRIM33, ARNT, ZBTB18, CAMSAP2, RAB3GAP2, KIRREL, ELF3, PTPRF, EXOC8, CLK2, DENND4B, ARHGEF2, BRINP2, IPO9, WDTC1, HORMAD1, ASTN1, ILF2, AKT3, RYR2, ACTN2, HEATR1, SIPA1L2, EGLN1, TRIM67, CDC42BPA, ENAH, FBXO28, SLC30A10, TGFB2, PROX1, LPGAT1, SLC30A1, IRF6, MAPKAPK2, NUCKS1, MDM4, PIK3C2B, SOX13, SYT2, NAV1, LHX9, CFH, CDC73, PTGS2, TPR, IVNS1ABP, SWT1, RNF2, DHX9, CACNA1E, XPR1, CEP350, RASAL2, RFWD2, TNR, RC3H1, SERPINC1, ZBTB37, ATP1B1, TIPRL, POU2F1, DEDD, ARHGAP30, COPA, DCAF8, ETV3, ARHGEF11, LMNA, UBQLN4, KIAA0907, RUSC1, PYGO2, CRTC2, GATAD2B, SNX27, PI4KB, PSMD4, MCL1, RPRD2, FAM46C, SYCP1, HIPK1, RBM15, AHCYL1, AMIGO1, COL11A1, HIAT1, GCLM, MTF2, RPL5, ZNF644, PKN2, FUBP1, USP33, ZZZ3, ZRANB2, ANKRD13C, SRSF11, SERBP1, SGIP1, DNAJC6, ROR1, NFIA, DAB1, SSBP3, NDC1, IPO13, KDM4A, FOXJ3, HIVEP3, CTPS1, RLF, PABPC4, RRAGC, SF3A3, MTF1, GRIK3, STK40, MAP7D1, AGO3, AGO1, AGO4, NCDN, DLGAP3, S100PBP, RBBP4, EIF3I, TXLNA, KPNA6, BAI2, YTHDF2, EYA3, STX12, AHDC1, TMEM57, LYPLA2, RPL11, HNRNPR, EPHB2, ECE1, LYST, ASH1L, PIAS3, FAF1, RPS8, KDM1A, KIF26B, MOV10, WDR26, TCEB3, RCOR3, PBX1, SYT14, PUM1, C1orf226, SLC2A1, PTBP2, UBAP2L, ITPKB, WASF2, GON4L, CSDE1, PTPRC, TDRD5, OSBPL9, ATP6V0B, SH3GLB1, SMG7, MEX3A, ATP1A1, CEPT1, ZC3H11A, MACF1, EIF4G3* |
| LEAT_EEBB_11 | F | 8 | 14 | LEAT | GG | | 5:0-180915260 | 5 | Gain | 180.92 | 0.03 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_11 | F | 8 | 14 | LEAT | GG | | 20:921971-63025520 | 20 | Gain | 62.10 | 0.07 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_15 | M | 15 | 17 | LEAT | GG | HS | 9:0-141213431 | 9 | Loss | 141.21 | 0.06 | 3 | *TSC1* | *ABCA2, DNM1, GABBR2, GRIN1, NOTCH1, PHF19, SCAI, SET, SMU1, SPTAN1, STXBP1, TLN1, VCP, WDR5* | *SETX, GDA, CLTA, TAF1L, TRAF2, OLFM1, LRRC8A, KIAA0368, RALGPS1, DAB2IP, GABBR2, RNF38, ERP44, UBE2R2, BRINP1, MAPKAP1, UHRF2, FAM120A, ZNF462, NOTCH1, SMC2, GNAQ, ZNF618, ZER1, ALDH1A1, WNK2, NOL6, TSC1, ZMYND19, BRD3, ZNF483, SEC16A, TLN1, RPL7A, ANAPC2, PTCH1, TESK1, ANP32B, ABCA2, COL27A1, BICD2, PRRC2B, RAD23B, PAX5, WDR5, VCP, PHF2, NUP214, TOPORS, ASTN2, NAA35, CACNA1B, GRIN1, RABL6, COL5A1, RALGDS, RAPGEF1, ABL1, NUP188, SET, SPTAN1, DNM1, ENG, STXBP1, ZBTB34, ZBTB43, SCAI, NR5A1, LHX2, DENND1A, RABGAP1, RC3H2, RAB14, PHF19, MEGF9, PRPF4, UGCG, EPB41L4B, KLF4, SLC44A1, TEX10, NCBP1, CDC14B, SYK, SPIN1, ZCCHC6, NTRK2, HNRNPK, UBQLN1, TLE1, TLE4, RORB, SHB, CNTFR, TEK, MLLT3, RPS6, BNC2, PSIP1, NFIB, PTPRD, JAK2, CDC37L1, RFX3, SMARCA2, CAMSAP1, NTNG2, PPP2R4, GAPVD1, LHX6, SMU1, ELAVL2, SVEP1, DAPK1, GOLGA2, STRBP, SEMA4D, RUSC2, EHMT1, RXRA, NR6A1, FBXW2* |
| LEAT_EEBB_16 | M | 7 | 20 | LEAT | GG | | 3:0-56942973 | 3p26.3-14.3 | Loss | 56.94 | 0.04 | 1 | *NPRL2, RAF1* | *ATP2B2, CELSR3, CTNNB1, DHX30, PBRM1, RBM5, TOP2B* | *SEMA3F, CELSR3, ZBTB47, NKTR, DAZL, SMARCC1, BHLHE40, KAT2B, RBM6, DOCK3, DYNC1LI1, IQSEC1, UBP1, FGD5, SLC6A1, CACNA1D, ERC2, SLC4A7, EOMES, STT3B, SNRK, BSN, WDR48, ITPR1, TWF2, RPL15, LRRN1, NR2C2, RBM15B, IP6K2, RARB, NISCH, RBM5, CTNNB1, ARIH2, GRM7, MAP4, ATP2B2, TRIM71, BRPF1, SRGAP3, PRKCD, SFMBT1, PBRM1, QRICH1, THRB,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | *UBE2E2, ANKRD28, SETD5, RAD54L2, SETD2, SCN5A, SATB1, PLCL2, ZNF445, ARPC4, USP19, TOP2B, DHX30, NBEAL2, CNOT10, SLC6A6, BAP1, CLASP2, WNT5A, CACNA2D3, CAMKV, CACNA2D2, FAM208A, DAG1* |
| LEAT_EEBB_16 | M | 7 | 20 | LEAT | GG | | 7:0-41948020 | 7 | Undetermined | 41.95 | 0.02 | 1 | | *RAC1* | *RALA, HOXA11, CDK13, ITGB8, SP4, AHR, INHBA, IGF2BP3, TTYH3, GNA12, KBTBD2, USP42, ELMO1, ACTB, RAPGEF5, C7orf26, HNRNPA2B1, EIF3B, HERPUD2, HOXA3, CARD11, SEPT7, TBX20, THSD7A, ELFN1, ETV1, TNRC18, HDAC9* |
| LEAT_EEBB_16 | M | 7 | 20 | LEAT | GG | | 22:17725150-51304566 | 22q | Loss | 33.58 | 0.04 | 4 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, GNAZ, RAC2, CELSR1, SHANK3, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA1I, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, MICAL3, HIC2, PLXNB2, TNRC6B, ZNRF3* |
| LEAT_EEBB_32 | F | 20 | 22 | LEAT | GG | | 5:0-180915260 | 5 | Gain | 180.92 | 0.29 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_32 | F | 20 | 22 | LEAT | GG | | 7:0-159138663 | 7 | Gain | 159.14 | 0.29 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_32 | F | 20 | 22 | LEAT | GG | | 12:0-133851895 | 12 | Gain | 133.85 | 0.29 | 8 | *KRAS, PTPN11* | *ANKRD52, ATP2B1, CHD4, CS, DDX23, GCN1L1, HECTD4, LRP1, NAA25, NAB2, PA2G4, PLXNC1, RARG, SCN8A, SETD1B, SMARCC2, SMARCD1, STAT2, USP5* | *SENP1, HDAC7, FOXJ2, TNFRSF1A, CBX5, SART3, ASIC1, CSRNP2, CNOT2, USP5, HCFC2, RAB35, LRP1, SLC38A2, MSI1, HNF1A, TBX3, GDF11, ESPL1, KCNH3, KCNMB4, PLXNC1, SLC41A2, WBP11, ATF7IP, NAP1L1, ASUN, CDK17, CCND2, LRP6, CORO1C, LPCAT3, CUX2, LHX5, NAA25, ATP5B, PTGES3, EIF4B, COPZ1, CLSTN3, SMARCC2, ANKRD52, SETD1B, DUSP6, USP15, BTBD11, EPS8, MED13L, NCKAP1L, ITGA5, FRS2, CCT2, TMEM132B, HSP90B1, STAT6, GCN1L1, KMT2D, TUBA1A, DIP2B, PA2G4, LEMD3, DHX37, TBX5, STAT2, WNK1, MLXIP, PITPNM2, ULK1, EEA1, E2F7, SP1, FBXO21, RSRC2, XPOT, ARID2, PTPRB, TUBA1B, SARNP, ZCCHC8, FAM60A, NOS1, KSR2, PPP1CC, HNRNPA1, NTN4, DYRK2, RNF41, R3HDM2, CACNA1C, CS, PTPN11, SPPL3, SCN8A, SRGAP1, GIT2, MBD6, ATN1, CHD4, DNAJC14, RFX4, PCBP2, SCAF11, KDM2B, ATXN2, ZFC3H1, NABP2, COL2A1, NOP2, PRMT8, STK38L, DENND5B, EP400, CIT, TAOK3, RIC8B, MON2, SMARCD1, ETV6, ZNF384, CCDC64, LARP4, SLC38A1, AEBP2, KDM5A, SETD8, RASSF8, NCOR2, ATF7, SBNO1, TMEM132D, RPL6, RARG, PAN2, ZDHHC17, ATP2B1, PLEKHA5, DCTN2, CPSF6, NELL2, PPP1R12A, SOX5, SLC4A8, KIF5A, PRICKLE1, PTPN6, MDM2, NAV3, ATP2A2, CLIP1, ACVR1B, SFSWAP, RAN, PHC1, CAND1, MAP3K12, AGAP2, ANKS1B, PRPF40B, PPFIA2, HECTD4, BAZ2A, SPATS2, GRIN2B* |
| LEAT_EEBB_40 | F | 14 | 25 | LEAT | GG | HS Type 1 | 19:0-59128983 | 19 | Gain | 59.13 | 0.18 | 5 | | *ACTN4, AP2A1, ATP1A3, BRD4, BRSK1, CA11, CACNA1A, CACTIN, CADM4, CALM3, CARM1, CHERP, CLPTM1, DAZAP1, DCAF15, DNMT1, DYRK1B, EGLN2, FZR1, GRIN2D, GSK3A, IRF2BP1, KHSRP, LMTK3, MAP4K1, MAST1, MAU2, NACC1, NOVA2, NUMBL, PIAS4, PPFIA3, PPP2R1A, PPP5C, PRKACA, PRKCG, PRMT1, PRPF31, PRR12, RFX1, SHANK1, SIN3B, SLC8A2, SMARCA4, SPTBN4, SUPT5H, TMEM259, TNPO2, TOMM40, U2AF2, UPF1, XAB2* | *PPP5C, CD22, MBD3, ANO8, KDM4B, BCL3, REXO1, CDC34, HNRNPL, RELB, SLC17A7, SGTA, FBL, RASIP1, RPL18A, RAB3A, KMT2B, USF2, ERF, GSK3A, SIPA1L3, CADM4, DAZAP1, TSHZ3, C3, VASP, SYMPK, UBA2, TSKS, SUGP1, ABHD8, WDR18, MAST1, PVRL2, SAFB2, MLLT1, ACTN4, NUMBL, TRIM28, ZC3H4, EIF3G, ZNF331, RFX1, DCAF15, CSNK1G2, HPN, CCNE1, UPF1, ELL, MAST3, MARK4, GRIK5, MAP2K2, DPP9, PIAS4, GRIN2D, BRD4, WIZ, NOTCH3, PRKCG, ARID3A, SAE1, ADAMTS10, RPS11, RNF126, NACC1, CELF5, SHANK1, KIAA0355, RAB8A, CHAF1A, IRF2BP1, RFX2, ZNF296, LTBP4, PRKACA, U2AF2, EEF2, BRSK1, SAMD4B, PRPF31, PPP2R1A, LMNB2, HNRNPM, LENG8, STK11, CARM1, PPFIA3, PIP5K1C, CLPTM1, ARHGEF1, LRRC25, RANBP3, LPHN1, PDE4A, FOSB, AP3D1, DPF1, ZNF536, TMEM259, PTBP1, ATP13A1, PTPRS, XAB2, AP2A1, BABAM1, DNMT1, DDA1, CACNA1A, GATAD2A, SCAF1, LONP1, TTYH1, SIN3B, DNM2, STRN4, HNRNPUL1, MAU2, FZR1, GLTSCR1, NFIX, MAP2K7, KHSRP, DOT1L, ARHGAP35, CNOT3, ELAVL1, PPP6R1, PRR12, TNPO2, TOMM40, CACTIN, SMARCA4, PRKD2, CAMSAP3, ILF3, PRMT1, EPS15L1, ZNF146, UNC13A, POU2F2, APC2, EVI5L, HOMER3, ATP1A3, CHERP, RPL18, CIC, LRFN3, SAFB, MAP4K1, DKFZP761J1410, DYRK1B, FCHO1, RUVBL2, MYO9B, RPS5, NUP62, SNRNP70, SUPT5H, MYH14, SUGP2, VAV1, NPAS1, FKBP8* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_40 | F | 14 | 25 | LEAT | GG | HS Type 1 | 20:0-63025520 | 20 | Gain | 63.03 | 0.19 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | 2:0-243199373 | 2 | Gain | 243.20 | 0.07 | 3 | | *CAD, FBXO11, HDLBP, MAP4K4, PPP1CB, PPP3R1, RQCD1, SCN2A, SF3B1, SNRNP200, UNC80, VSNL1, XPO1* | *CYP26B1, INPP4A, TRIB2, TMEM131, MSH2, NRBP1, PCGF1, IGFBP5, PPP1R7, DLX2, PAPOLG, ATAD2B, AFTPH, ACVR2A, RIF1, ILKAP, PER2, KIDINS220, CCT7, USP37, REV1, EPC2, SIX3, PPIG, HECW2, SLC40A1, ACTR3, POLR1B, TFCP2L1, PTPN4, CLASP1, LRP2, POLR1A, EHBP1, ATF2, MCM6, CCNT2, R3HDM1, AGPS, ADAM23, CUL3, ADD2, CAD, RAB10, KIF3C, FOSL2, VPS54, MEIS1, AMMECR1L, CTDSP1, RQCD1, CRIM1, ASAP2, EPHA4, ZFP36L2, RANBP2, SCN3A, TRIP12, FMNL2, DPYSL5, EIF5B, REL, MEMO1, KCNJ3, EN1, INHBB, IWS1, FZD5, ARPC2, CTLA4, SIX2, SCN1A, ANTXR1, AGAP1, COL3A1, RPS7, SEMA4C, PRKCE, MAT2A, PSMD1, KLF7, CFLAR, SP3, SNED1, ADAM17, SPEG, SRSF7, SPAST, ROCK2, RAPH1, GLS, RNF144A, WDR33, NCL, SNRNP200, DIS3L2, FIGN, SF3B1, NPAS2, BCL11A, CCDC88A, PUM2, NR4A2, ERBB4, PPM1G, HDAC4, MAP4K4, FN1, ZNF512, STK39, AFF3, SPTBN1, GFPT1, SCN2A, ARID5A, SAP130, GTF3C2, INPP5D, MAP2, NCKAP1, STAT1, DLX1, BMPR2, COL5A2, RND3, ACTR2, DDX1, HSPD1, LRP1B, TBR1, HDLBP, ATG16L1, SPHKAP, XRCC5, STAT4, OSBPL6, HNRNPA3, BAZ2B, GALNT13, DPP10, CCT4, EFEMP1, SP9, PPP1CB, MOB1A, RAPGEF4, USP34, XPO1, SLC8A1, INO80D, FBXO11, PSME4, ATG4B, NRXN1, NCOA1, MBD5, WDR43, AAK1, SLC39A10, ARHGAP25, TET3, GIGYF2, ZNF638, KDM3A, ATG9A, HNRNPLL, MGAT5, PSMD14, KCMF1, MAP3K2, AGFG1, CALCRL, SATB2, UBR3, BIRC6, SOS1, TMEM185B, MYT1L, SESTD1, PAX8, CNTNAP5, TLK1, KANSL3, CREB1, KIF5C, ASXL2, UNC80, EIF2B4, GLI2, BZW1, IKZF2, KIF1A, FBXO41, REEP1, ZEB2, HSPE1-MOB4* |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | 5:0-180915260 | 5 | Gain | 180.92 | 0.05 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | 7:0-159138663 | 7 | Gain | 159.14 | 0.04 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNAI2, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | 12:0-133851895 | 12 | Gain | 133.85 | 0.07 | 8 | *KRAS, PTPN11* | *ANKRD52, ATP2B1, CHD4, CS, DDX23, GCN1L1, HECTD4, LRP1, NAA25, NAB2, PA2G4, PLXNC1, RARG, SCN8A, SETD1B, SMARCC2, SMARCD1, STAT2, USP5* | *SENP1, HDAC7, FOXJ2, TNFRSF1A, CBX5, SART3, ASIC1, CSRNP2, CNOT2, USP5, HCFC2, RAB35, LRP1, SLC38A2, MSI1, HNF1A, TBX3, GDF11, ESPL1, KCNH3, KCNMB4, PLXNC1, SLC41A2, WBP11, ATF7IP, NAP1L1, ASUN, CDK17, CCND2, LRP6, CORO1C, LPCAT3, CUX2, LHX5, NAA25, ATP5B, PTGES3, EIF4B, COPZ1, CLSTN3, SMARCC2, ANKRD52, SETD1B, DUSP6, USP15, BTBD11, EPS8, MED13L, NCKAP1L, ITGA5, FRS2, CCT2, TMEM132B, HSP90B1, STAT6, GCN1L1, KMT2D, TUBA1A, DIP2B, PA2G4, LEMD3, DHX37, TBX5, STAT2, WNK1, MLXIP, PITPNM2, ULK1, EEA1, E2F7, SP1, FBXO21, RSRC2, XPOT, ARID2, PTPRB, TUBA1B, SARNP, ZCCHC8, FAM60A, NOS1, KSR2, PPP1CC, HNRNPA1, NTN4, DYRK2, RNF41, R3HDM2, CACNA1C, CS, PTPN11, SPPL3, SCN8A, SRGAP1, GIT2, MBD6, ATN1, CHD4, DNAJC14, RFX4, PCBP2, SCAF11, KDM2B, ATXN2, ZFC3H1, NABP2, COL2A1, NOP2, PRMT8, STK38L, DENND5B, EP400, CIT, TAOK3, RIC8B, MON2, SMARCD1, ETV6, ZNF384, CCDC64, LARP4, SLC38A1, AEBP2, KDM5A, SETD8, RASSF8, NCOR2, ATF7, SBNO1, TMEM132D, RPL6, RARG, PAN2, ZDHHC17, ATP2B1, PLEKHA5, DCTN2, CPSF6, NELL2, PPP1R12A, SOX5, SLC4A8, KIF5A, PRICKLE1, PTPN6, MDM2, NAV3, ATP2A2, CLIP1, ACVR1B, SFSWAP, RAN, PHC1, CAND1, MAP3K12, AGAP2, ANKS1B, PRPF40B, PPFIA2, HECTD4, BAZ2A, SPATS2, GRIN2B* |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | 14:24860592-107349540 | 14q | Gain | 82.49 | 0.06 | 2 | | *DYNC1H1, MTA1, PSMC6, RBM25, UNC79* | *MAP4K5, PPP1R13B, G2E3, PAPOLA, SOS2, PSMA3, AHSA1, EIF5, NFKBIA, TGFB3, BMP4, ZC3H14, UNC79, VRTN, EIF2S1, PSMC1, PSMA6, SNW1, CYP46A1, RBM25, RPS6KA5, YY1, RCOR1, RTN1, ITPK1, AKAP6, ELMSAN1, FBXO33, TRIM9, PCNX, RALGAPA1, DDHD1, PSEN1, PPM1A, YLPM1, TTC7B, MTA1, JAG2, EML1, SEL1L, PPP2R5E, DCAF5, FBLN5, FERMT2, ARHGAP5, ARID4A, PRPF39, NPAS3, CALM1, BCL11B, ATL1, TECPR2, DYNC1H1, BAZ1A, CDC42BPB, SPTB, CCNK, SAMD4A, INF2, EVL, ACTN1, ZBTB1, DAAM1, HECTD1, EXOC5, CEP170B, PPP2R5C, MDGA2, BEGAIN, PSMC6, PACS2, SCFD1, GPHN, DICER1, HIF1A, NOVA1, GTF2A1, AKT1, SMEK1, NRXN3, SIPA1L1, NAA30, SRP54, TRAF3* |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | 15:23226254-102531392 | 15q | Gain | 79.31 | 0.04 | 3 | | *RTF1, SIN3A* | *AQR, MGA, CSK, PIAS1, DLL4, SNRPA1, ABHD17C, IREB2, CTDSPL2, THBS1, KIF23, ADAM10, HERC2, AP3B2, SNX1, RAB11A, HCN4, TTBK2, IGF1R, ZNF710, FURIN, IQGAP1, UBR1, SPRED1, COPS2, LEO1, HDGFRP3, ZNF592, CLPX, MAP1A, PDIA3, PYGO1, CSNK1G1, ARNT2, OTUD7A, RPL4, FBXO22, RASGRP1, GABRB3, SEMA6D, FBN1, ZNF609, MORF4L1, ASB7, NEO1, DPP8, NPTN, BNC1, TJP1, CASC5, MEF2A, LINGO1, MYO9A, ZNF770, NTRK3, AKAP13, INO80, ARIH1, USP3, SLTM, TP53BP1, RYR3, RTF1,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | *NR2F2, CHD2, SIN3A, UBE3A, MYO5A, BAHD1, FRMD5, RASGRF1, RFX7, USP8, ACAN, DENND4A, HERC1, ANP32A, MAGEL2, DMXL2, TLE3, RNF111, MEIS2, TLN2, CPEB1, PARP6* |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | 21:15515843-48129895 | 21q | CNN-LOH | 32.61 | 0.05 | 2 | | | *HUNK, SIK1, USP25, CCT8, TIAM1, SCAF4, IFNGR2, PKNOX1, U2AF1, TRAPPC10, ZBTB21, PAXBP1, BRWD1, GABPA, SON, ETS2, COL6A1, SLC5A3, ITSN1, AGPAT3, DYRK1A, NRIP1, DSCAM, MORC3, ERG, SYNJ1* |
| LEAT_EEBB_42 | F | 20 | 33 | LEAT | GG | | 22:17221495-51304566 | 22q | CNN-LOH | 34.08 | 0.06 | 5 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, GNAZ, RAC2, CELSR1, SHANK3, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA1I, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, MICAL3, HIC2, PLXNB2, TNRC6B, ZNRF3* |
| LEAT_EEBB_48 | M | 27 | 40 | LEAT | GG | HS Type 1 | 4:58377516-191154276 | 4q | Gain | 132.78 | 0.02 | 1 | | *ABCE1, ANKRD17, PPP3CA, SMARCA5, WDFY3* | *NFKB1, COPS4, RASGEF1B, RAPGEF2, FNIP2, SEPT11, CNOT6L, CENPE, LEF1, SEC24B, CCNA2, NAF1, RPS3A, GPM6A, FBXW7, GRID2, SMARCA5, WDFY3, HMGB2, GRIA2, NAA15, HHIP, ABCE1, DCLK2, PITX2, HNRNPD, EDNRA, TRIM2, RAP1GDS1, LIN54, YTHDC1, CLCN3, ANK2, NR3C2, ANKRD17, SMARCAD1, G3BP2, ZNF827, ELF2, FAT4, ELOVL6, PPP3CA, AFF1, CPE, KIAA0922, SLC4A4, BMPR1B, OTUD4, PLRG1, PDGFC, ANKRD50, CDKN2AIP, MAML3, TENM3, LPHN3, KLHL2, SMAD1* |
| LEAT_EEBB_63 | M | 13 | 15 | LEAT | GG | | 12:0-133851895 | 12 | Gain | 133.85 | 0.05 | 8 | *KRAS, PTPN11* | *ANKRD52, ATP2B1, CHD4, CS, DDX23, GCN1L1, HECTD4, LRP1, NAA25, NAB2, PA2G4, PLXNC1, RARG, SCN8A, SETD1B, SMARCC2, SMARCD1, STAT2, USP5* | *SENP1, HDAC7, FOXJ2, TNFRSF1A, CBX5, SART3, ASIC1, CSRNP2, CNOT2, USP5, HCFC2, RAB35, LRP1, SLC38A2, MSI1, HNF1A, TBX3, GDF11, ESPL1, KCNH3, KCNMB4, PLXNC1, SLC41A2, WBP11, ATF7IP, NAP1L1, ASUN, CDK17, CCND2, LRP6, CORO1C, LPCAT3, CUX2, LHX5, NAA25, ATP5B, PTGES3, EIF4B, COPZ1, CLSTN3, SMARCC2, ANKRD52, SETD1B, DUSP6, USP15, BTBD11, EPS8, MED13L, NCKAP1L, ITGA5, FRS2, CCT2, TMEM132B, HSP90B1, STAT6, GCN1L1, KMT2D, TUBA1A, DIP2B, PA2G4, LEMD3, DHX37, TBX5, STAT2, WNK1, MLXIP, PITPNM2, ULK1, EEA1, E2F7, SP1, FBXO21, RSRC2, XPOT, ARID2, PTPRB, TUBA1B, SARNP, ZCCHC8, FAM60A, NOS1, KSR2, PPP1CC, HNRNPA1, NTN4, DYRK2, RNF41, R3HDM2, CACNA1C, CS, PTPN11, SPPL3, SCN8A, SRGAP1, GIT2, MBD6, ATN1, CHD4, DNAJC14, RFX4, PCBP2, SCAF11, KDM2B, ATXN2, ZFC3H1, NABP2, COL2A1, NOP2, PRMT8, STK38L, DENND5B, EP400, CIT, TAOK3, RIC8B, MON2, SMARCD1, ETV6, ZNF384, CCDC64, LARP4, SLC38A1, AEBP2, KDM5A, SETD8, RASSF8, NCOR2, ATF7, SBNO1, TMEM132D, RPL6, RARG, PAN2, ZDHHC17, ATP2B1, PLEKHA5, DCTN2, CPSF6, NELL2, PPP1R12A, SOX5, SLC4A8, KIF5A, PRICKLE1, PTPN6, MDM2, NAV3, ATP2A2, CLIP1, ACVR1B, SFSWAP, RAN, PHC1, CAND1, MAP3K12, AGAP2, ANKS1B, PRPF40B, PPFIA2, HECTD4, BAZ2A, SPATS2, GRIN2B* |
| LEAT_EEBB_63 | M | 13 | 15 | LEAT | GG | | 13:21050575-115169878 | 13 | Loss | 94.12 | 0.05 | 2 | | *MYCBP2, NBEA* | *AKAP11, FLT3, EFNB2, XPO4, RNF17, DCLK1, IPO5, KPNA3, RB1, RBM26, ZC3H13, USP12, FLT1, DACH1, INTS6, PDS5B, HSPH1, COG3, FBXL3, MYO16, CHAMP1, TFDP1, CUL4A, ARHGEF7, COL4A1, TNFSF13B, FAM155A, TPP2, TM9SF2, DOCK9, HS6ST3, SLITRK1, SPRY2, KLF12, KBTBD6, FOXO1, FRY, SLC7A1, PAN3, NUPL1, FGF9, LATS2, LRCH1, NBEA, MTUS2, TSC22D1, FNDC3A, MYCBP2* |
| LEAT_EEBB_63 | M | 13 | 15 | LEAT | GG | | 18:0-78077248 | 18 | Gain | 78.08 | 0.05 | 1 | | *MAPRE2, SMAD7, TCF4* | *ZNF236, WDR7, DSG1, ZNF24, NOL4, THOC1, USP14, ANKRD12, PHLPP1, CDH2, ASXL3, ESCO1, C18orf8, RNF165, ZCCHC2, SETBP1, ATP5A1, TRAPPC8, ZNF407, PTPN2, DLGAP1, SMCHD1, TSHZ1, GNAL, ZNF532, SMAD4, MALT1, ZNF521, CTIF, SOCS6, KIAA1468, TCF4, ROCK1, NEDD4L, PPP4R1, SMAD2, CXXC1, CELF4, DCC, ZNF516, PTPRM, PIAS2, MBD1* |
| LEAT_EEBB_63 | M | 13 | 15 | LEAT | GG | | 19:6547141-59128983 | 19 | Gain | 52.58 | 0.05 | 5 | | *ACTN4, AP2A1, ATP1A3, BRD4, BRSK1, CA11, CACNA1A, CADM4, CALM3, CARM1, CHERP, CLPTM1, DCAF15, DNMT1, DYRK1B, EGLN2, GRIN2D, GSK3A, IRF2BP1, LMTK3, MAP4K1, MAST1, MAU2, NACC1, NOVA2, NUMBL, PPFIA3, PPP2R1A, PPP5C, PRKACA, PRKCG, PRMT1, PRPF31, PRR12, RFX1, SHANK1, SIN3B, SLC8A2, SMARCA4, SPTBN4, SUPT5H, TNPO2, TOMM40, U2AF2, UPF1, XAB2* | *PPP5C, CD22, ANO8, BCL3, HNRNPL, RELB, SLC17A7, FBL, RASIP1, RPL18A, RAB3A, KMT2B, USF2, ERF, GSK3A, SIPA1L3, CADM4, TSHZ3, C3, VASP, SYMPK, UBA2, TSKS, SUGP1, ABHD8, MAST1, PVRL2, ACTN4, NUMBL, TRIM28, ZC3H4, EIF3G, ZNF331, RFX1, DCAF15, HPN, CCNE1, UPF1, ELL, MAST3, MARK4, GRIK5, GRIN2D, BRD4, WIZ, NOTCH3, PRKCG, SAE1, ADAMTS10, RPS11, NACC1, SHANK1, KIAA0355, RAB8A, IRF2BP1, ZNF296, LTBP4, PRKACA, U2AF2, BRSK1, SAMD4B, PRPF31, PPP2R1A, HNRNPM, LENG8, CARM1, PPFIA3, CLPTM1, ARHGEF1, LRRC25, LPHN1, PDE4A, FOSB, DPF1, ZNF536, ATP13A1, XAB2, AP2A1, BABAM1, DNMT1, DDA1, CACNA1A, GATAD2A, SCAF1, TTYH1, SIN3B, DNM2, STRN4, HNRNPUL1, MAU2, GLTSCR1, NFIX, MAP2K7, ARHGAP35, CNOT3, ELAVL1, PPP6R1, PRR12, TNPO2, TOMM40, SMARCA4, PRKD2, CAMSAP3, ILF3, PRMT1, EPS15L1, ZNF146, UNC13A, POU2F2, EVI5L, HOMER3, ATP1A3, CHERP, RPL18, CIC, LRFN3, MAP4K1, DKFZP761J1410, DYRK1B, FCHO1, RUVBL2, MYO9B, RPS5, NUP62, SNRNP70, SUPT5H, MYH14, SUGP2, VAV1, NPAS1, FKBP8* |
| LEAT_EEBB_63 | M | 13 | 15 | LEAT | GG | | 20:0-63025520 | 20 | Gain | 63.03 | 0.05 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_70 | F | 18 | 22 | LEAT | GG | | 5:0-180915260 | 5 | Gain | 180.92 | 0.05 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK,* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | *KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_70 | F | 18 | 22 | LEAT | GG | | 7:0-159138663 | 7 | Gain | 159.14 | 0.05 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_70 | F | 18 | 22 | LEAT | GG | | 11:0-135006516 | 11 | Gain | 135.01 | 0.11 | 5 | | *ADRBK1, B3GNT1, CKAP5, CNIH2, DDB1, DPF2, GANAB, GNG3, GRAMD1B, KDM2A, KMT2A, LTBP3, MARK2, MEN1, MUC5B, NXF1, PCNXL3, PLCB3, PPP2R5B, PRPF19, PSMC3, RPS6KA4, SART1, SSRP1, SYVN1* | *TTC17, PPP2R5B, SPI1, PRPF19, MAPK8IP1, COPB1, PPFIA1, RRAS2, DAGLA, PRKRIR, DCUN1D5, EED, ARHGEF17, POLD3, FNBP4, OSBP, DDX6, ARCN1, NRXN2, FBXO3, GTF2H1, KCNC1, FAM160A2, LRRC4C, SERPING1, SSRP1, TENM4, ZC3H12C, HMBS, MYRF, FADS2, CWC15, VPS26B, YAP1, ZDHHC5, SIK3, C11orf84, LRP5, FAT3, PCF11, PKNOX2, PSMC3, APBB1, DCHS1, PATL1, RRM1, DDB1, HNRNPUL2, LTBP3, SUV420H1, SIK2, STIP1, CHRM1, RSF1, ADRBK1, RBM14, ARHGAP32, RAB6A, NPAS4, CNIH2, SART1, CLCF1, PPP1CA, AMBRA1, ZFP91, FJX1, PACS1, EHD1, DSCAML1, SF3B2, CSTF3, NUP98, KCNA4, PPME1, EEF1G, RPS6KA4, PDE2A, BCL9L, ZBTB16, SPTY2D1, C2CD2L, MEN1, USP47, CPSF7, CAPRIN1, GANAB, FAR1, ANO1, PCNXL3, UVRAG, FAM168A, MPPED2, CTR9, GAB2, DLG2, SYVN1, ATG2A, SF1, AHNAK, IPO7, BRSK2, ARNTL, CUL5, PICALM, NUMA1, PPP6R3, INCENP, NAV2, SOX6, ARHGEF12, CTNND1, QSER1, MARK2, HYOU1, RDX, RELA, RBM4, RBM14-RBM4, KLC2, PHF21A, GRM5, PSMA1, PAX6, ARRB1, TBCEL, PRDM11, PSMD13, CHRM4, AP2A2, WEE1, MAML2, TRIM3, KIRREL3, ZBTB44, TRAF6, FLI1, DPF2, ATG13, KDM2A, C11orf30, CREB3L1, CKAP5, MUC5B, GRAMD1B, API5, FZD4, RNF214, CELF1, NXF1, EIF3F, IGSF9B, ST5, KMT2A, HSPA8, OTUB1, PLCB3, SYT7* |
| LEAT_EEBB_70 | F | 18 | 22 | LEAT | GG | | 13:19307875-115169878 | 13q | Gain | 95.86 | 0.11 | 2 | | *MYCBP2, NBEA* | *AKAP11, FLT3, EFNB2, XPO4, RNF17, DCLK1, IPO5, KPNA3, RB1, RBM26, ZC3H13, USP12, FLT1, DACH1, INTS6, PDS5B, HSPH1, PSPC1, COG3, FBXL3, MYO16, CHAMP1, TFDP1, CUL4A, ARHGEF7, COL4A1, TNFSF13B, FAM155A, TPP2, TM9SF2, DOCK9, HS6ST3, SLITRK1, SPRY2, KLF12, KBTBD6, FOXO1, FRY, SLC7A1, PAN3, NUPL1, FGF9, LATS2, ZMYM2, LRCH1, NBEA, MTUS2, TSC22D1, FNDC3A, MYCBP2* |
| LEAT_EEBB_74 | M | 8 | 12 | LEAT | GG | | 4:0-191154276 | 4 | Gain | 191.15 | 0.09 | 5 | | *ABCE1, ANKRD17, CTBP1, DHX15, POLR2B, PPP3CA, SMARCA5, WDFY3* | *BOD1L1, PHOX2B, NFKB1, PDGFRA, UBE2K, KDR, PPAT, COPS4, RASGEF1B, RAPGEF2, FNIP2, ADD1, PPARGC1A, SEPT11, CNOT6L, CENPE, LEF1, SEC24B, CCNA2, NAF1, RPS3A, GPM6A, FBXW7, GRID2, SMARCA5, UCHL1, KIT, CTBP1, GABRB1, WDFY3, HMGB2, GRIA2, NAA15, HHIP, ABCE1, DCLK2, PDS5A, LDB2, PITX2, KIAA0232, REST, CLOCK, LIMCH1, HNRNPD, EDNRA, FAM193A, CRMP1, DHX15, TRIM2, RAP1GDS1, LIN54, FBXL5, RBPJ, YTHDC1, CLCN3, HTT, ANK2, NR3C2, FRYL, ANKRD17, SMARCAD1, G3BP2, PCGF3, ZNF827, POLR2B, ATP8A1, RBM47, RFC1, WHSC1, ELF2, FAT4, ELOVL6, PPP3CA, AFF1, CPE, JAKMIP1, KIAA0922, SLC4A4, BMPR1B, RPL9, RELL1, OTUD4, PLRG1, WDR1, BEND4, PDGFC, ANKRD50, SLIT2, CDKN2AIP, MAML3, GABRA2, TENM3, LPHN3, KLHL2, SMAD1* |
| LEAT_EEBB_74 | M | 8 | 12 | LEAT | GG | | 6:0-171115067 | 6 | Gain | 171.12 | 0.06 | 8 | *MYB* | *GTPBP2, JARID2, KLHDC3, PPP2R5D, TMEM63B, TRERF1, XPO5* | *TFAP2D, RANBP9, TRAM2, FBXO5, TNFAIP3, DST, ATXN1, KIAA1244, CUL9, BACH2, REPS1, FAM65B, KIF13A, TMEM63B, TTBK1, GCM1, POU5F1, PHF3, NUP153, PSMB1, XPO5, SRF, SCUBE3, PHIP, C6orf136, PPARD, GLTSCR1L, EEF1A1, DEF6, TCP1, STXBP5, COL12A1, ZBTB2, RPS10, KLHDC3, TUBB, EPB41L2, PRPF4B, JARID2, KCNQ5, ARID1B, E2F3, DOPEY1, FYN, IGF2R, BRPF3, WTAP, REV3L, MAP3K5, ANKS1A, SLC35F1, PLAGL1, ZNF318, DLL1, THBS2, TULP4, SCAF8, TAB2, HIVEP2, PTPRK, HSF2, NUS1, AMD1, CDK19, CDC40, NR2E1, ATG5, PRDM1, SIM1, PNISR, EPHA7, MAP3K7, MDN1, ZNF292, SYNCRIP, TBX18, SMAP1, BAI3, LRRC1, RUNX2, CDC5L, HSP90AB1, UBR2, TRERF1, CCND3, USP49, FOXP4, CMTR1, RNF8, SRSF3, SPDEF, PHF1, RXRB, COL11A2, PBX2, ZBTB12, BAG6, PRRC2A, PPP1R10, GNL1, TRIM39, GABBR1, TRIM27, C6orf62, HIVEP1, TFAP2A, DSP, RREB1, GMDS, MLLT4, MAP3K4, WASF1, TFAP2B, PFDN6, BRD2, AGPAT1, DDX39B, CDYL, DAAM2, SYNGAP1, FAM135A, GRIK2, BEND3, MDGA1, SNAP91, RPS18, ESR1, SENP6, PTK7, PPP2R5D, HDAC2, CCNC, RIMS1, L3MBTL3, GRM4, PACSIN1, PDE10A, LATS1* |
| LEAT_EEBB_74 | M | 8 | 12 | LEAT | GG | | 7:0-159138663 | 7 | Gain | 159.14 | 0.06 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_74 | M | 8 | 12 | LEAT | GG | | 9:0-141213431 | 9 | Gain | 141.21 | 0.08 | 2 | *TSC1* | *ABCA2, DNM1, GABBR2, GRIN1, NOTCH1, PHF19, SCAI, SET, SMU1,* | *SETX, GDA, CLTA, TAF1L, TRAF2, OLFM1, LRRC8A, KIAA0368, RALGPS1, DAB2IP, GABBR2, RNF38, ERP44, UBE2R2, BRINP1, MAPKAP1, UHRF2, FAM120A, ZNF462, NOTCH1, SMC2, GNAQ, ZNF618, ZER1, ALDH1A1, WNK2, NOL6, TSC1, ZMYND19, BRD3, ZNF483, SEC16A, TLN1, RPL7A, ANAPC2, PTCH1, TESK1, ANP32B, ABCA2, COL27A1, BICD2, PRRC2B, RAD23B, PAX5,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | *SPTAN1, STXBP1, TLN1, VCP, WDR5* | *WDR5, VCP, PHF2, NUP214, TOPORS, ASTN2, NAA35, CACNA1B, GRIN1, RABL6, COL5A1, RALGDS, RAPGEF1, ABL1, NUP188, SET, SPTAN1, DNM1, ENG, STXBP1, ZBTB34, ZBTB43, SCAI, NR5A1, LHX2, DENND1A, RABGAP1, RC3H2, RAB14, PHF19, MEGF9, PRPF4, UGCG, EPB41L4B, KLF4, SLC44A1, TEX10, NCBP1, CDC14B, SYK, SPIN1, ZCCHC6, NTRK2, HNRNPK, UBQLN1, TLE1, TLE4, RORB, SHB, CNTFR, TEK, MLLT3, RPS6, BNC2, PSIP1, NFIB, PTPRD, JAK2, CDC37L1, RFX3, SMARCA2, CAMSAP1, NTNG2, PPP2R4, GAPVD1, LHX6, SMU1, ELAVL2, SVEP1, DAPK1, GOLGA2, STRBP, SEMA4D, RUSC2, EHMT1, RXRA, NR6A1, FBXW2* |
| LEAT_EEBB_74 | M | 8 | 12 | LEAT | GG | | 12:0-133851895 | 12 | Gain | 133.85 | 0.06 | 8 | *KRAS, PTPN11* | *ANKRD52, ATP2B1, CHD4, CS, DDX23, GCN1L1, HECTD4, LRP1, NAA25, NAB2, PA2G4, PLXNC1, RARG, SCN8A, SETD1B, SMARCC2, SMARCD1, STAT2, USP5* | *SENP1, HDAC7, FOXJ2, TNFRSF1A, CBX5, SART3, ASIC1, CSRNP2, CNOT2, USP5, HCFC2, RAB35, LRP1, SLC38A2, MSI1, HNF1A, TBX3, GDF11, ESPL1, KCNH3, KCNMB4, PLXNC1, SLC41A2, WBP11, ATF7IP, NAP1L1, ASUN, CDK17, CCND2, LRP6, CORO1C, LPCAT3, CUX2, LHX5, NAA25, ATP5B, PTGES3, EIF4B, COPZ1, CLSTN3, SMARCC2, ANKRD52, SETD1B, DUSP6, USP15, BTBD11, EPS8, MED13L, NCKAP1L, ITGA5, FRS2, CCT2, TMEM132B, HSP90B1, STAT6, GCN1L1, KMT2D, TUBA1A, DIP2B, PA2G4, LEMD3, DHX37, TBX5, STAT2, WNK1, MLXIP, PITPNM2, ULK1, EEA1, E2F7, SP1, FBXO21, RSRC2, XPOT, ARID2, PTPRB, TUBA1B, SARNP, ZCCHC8, FAM60A, NOS1, KSR2, PPP1CC, HNRNPA1, NTN4, DYRK2, RNF41, R3HDM2, CACNA1C, CS, PTPN11, SPPL3, SCN8A, SRGAP1, GIT2, MBD6, ATN1, CHD4, DNAJC14, RFX4, PCBP2, SCAF11, KDM2B, ATXN2, ZFC3H1, NABP2, COL2A1, NOP2, PRMT8, STK38L, DENND5B, EP400, CIT, TAOK3, RIC8B, MON2, SMARCD1, ETV6, ZNF384, CCDC64, LARP4, SLC38A1, AEBP2, KDM5A, SETD8, RASSF8, NCOR2, ATF7, SBNO1, TMEM132D, RPL6, RARG, PAN2, ZDHHC17, ATP2B1, PLEKHA5, DCTN2, CPSF6, NELL2, PPP1R12A, SOX5, SLC4A8, KIF5A, PRICKLE1, PTPN6, MDM2, NAV3, ATP2A2, CLIP1, ACVR1B, SFSWAP, RAN, PHC1, CAND1, MAP3K12, AGAP2, ANKS1B, PRPF40B, PPFIA2, HECTD4, BAZ2A, SPATS2, GRIN2B* |
| LEAT_EEBB_74 | M | 8 | 12 | LEAT | GG | | 16:0-90354753 | 16 | Gain | 90.35 | 0.08 | 3 | *NPRL3, TSC2* | *CNOT1, CTCF, EDC4, FBXL19, MAPK8IP3, MAZ, NUDT21, SF3B3, STX1B, UBE2I, USP7* | *CACNG3, TFAP4, GLG1, STX1B, C16orf70, CORO1A, NUTF2, USP10, TSC2, CAPN15, TOX3, MAPK8IP3, SALL1, FUS, VPS4A, DYNC1LI2, XYLT1, RBL2, PKD1, RAB11FIP3, CREBBP, FOXF1, GNAO1, CSNK2A2, SRCAP, SETD1A, ZNF629, CTCF, NKD1, ZFHX3, CDH11, ZCCHC14, IRF8, JPH3, ATP6V0D1, ADCY9, VPS35, ZNF319, PLK1, NUDT21, ZC3H18, ANKRD11, SF3B3, PRKCB, XPO6, TAOK2, RBFOX1, TUBB3, DNAJA2, CNOT1, RANBP10, MKL2, RBBP6, GLYR1, SH2B1, RNF40, C16orf72, IL21R, MTSS1L, PDPK1, CASKIN1, USP7, NFATC3, UBE2I, FBRS, GTF3C1, PLCG2, FBXL19, RPL13, BANP, AP1G1, BRD7, ATXN2L, TNRC6A, MYH11, KIAA0430, ZC3H7A, GRIN2A, UBN1, FBXL16, TBC1D10B, CBFB, BCAR1, FAM65A, CYLD, NFAT5, GSPT1, PAPD5, HERPUD1, SMG1, CMIP, RSPRY1, ZNF423, RNPS1, CHD9, CDH8, TERF2* |
| LEAT_EEBB_74 | M | 8 | 12 | LEAT | GG | | 20:0-63025520 | 20 | Gain | 63.03 | 0.06 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_80 | M | 5.7 | 6 | LEAT | GG | | 1:0-120110175 | 1p | Loss | 120.11 | 0.08 | 1 | *MTOR* | *AGO1, AGO3, AHCYL1, ARID1A, ATP1A1, CAMTA1, IPO13, KDM1A, KDM4A, MACF1, MTOR, NCDN, RBBP4, USP24, ZMYM4* | *LRRC7, SARS, MFN2, PRDM2, TARDBP, SORT1, ZCCHC11, ARHGAP29, CHD5, KIF1B, TAF12, PRDM16, PGD, PSMA5, CELSR2, ZFYVE9, RAP1GAP, ZYG11B, USP24, GMEB1, CAMTA1, LRP8, USP48, PPP1R8, HP1BP3, ZMYM4, KCND3, LPHN2, YBX1, SRRM1, ARID1A, KHDRBS1, CSF1, MRTO4, LRRC8B, LCK, LRRC8D, RERE, USP1, TMEM201, JAK1, UBE4B, LRRC41, PTP4A2, PTPRU, LEPR, NASP, GRHL3, THRAP3, RNF220, MAN1A2, SFPQ, NEGR1, VPS13D, TRIM33, PTPRF, MTOR, WDTC1, FAM46C, SYCP1, HIPK1, RBM15, AHCYL1, AMIGO1, COL11A1, HIAT1, GCLM, MTF2, RPL5, ZNF644, PKN2, FUBP1, USP33, ZZZ3, ZRANB2, ANKRD13C, SRSF11, SERBP1, SGIP1, DNAJC6, ROR1, NFIA, DAB1, SSBP3, NDC1, IPO13, KDM4A, FOXJ3, HIVEP3, CTPS1, RLF, PABPC4, RRAGC, SF3A3, MTF1, GRIK3, STK40, MAP7D1, AGO3, AGO1, AGO4, NCDN, DLGAP3, S100PBP, RBBP4, EIF3I, TXLNA, KPNA6, BAI2, YTHDF2, EYA3, STX12, AHDC1, TMEM57, LYPLA2, RPLI1, HNRNPR, EPHB2, ECE1, UBR4, RCC2, FBXO42, ZBTB17, SPEN, CASZ1, CLSTN1, PIK3CD, DNAJC11, PHF13, AJAP1, TP73, PANK4, SKI, GABRD, GNB1, FAF1, RPS8, KDM1A, MOV10, TCEB3, PUM1, SLC2A1, PTBP2, WASF2, CSDE1, OSBPL9, ATP6V0B, DDI2, SH3GLB1, ATP1A1, CEPT1, MACF1, EIF4G3* |
| LEAT_EEBB_80 | M | 5.7 | 6 | LEAT | GG | | 5:0-180915260 | 5 | Gain | 180.92 | 0.03 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_80 | M | 5.7 | 6 | LEAT | GG | | 6:0-171115067 | 6 | Gain | 171.12 | 0.03 | 8 | *MYB* | *GTPBP2, JARID2, KLHDC3, PPP2R5D, TMEM63B, TRERF1, XPO5* | *TFAP2D, RANBP9, TRAM2, FBXO5, TNFAIP3, DST, ATXN1, KIAA1244, CUL9, BACH2, REPS1, FAM65B, KIF13A, TMEM63B, TTBK1, GCM1, POU5F1, PHF3, NUP153, PSMB1, XPO5, SRF, SCUBE3, PHIP, C6orf136, PPARD, GLTSCR1L, EEF1A1, DEF6, TCP1, STXBP5, COL12A1, ZBTB2, RPS10, KLHDC3, TUBB, EPB41L2, PRPF4B, JARID2, KCNQ5, ARID1B, E2F3, DOPEY1, FYN, IGF2R, BRPF3, WTAP, REV3L, MAP3K5, ANKS1A, SLC35F1, PLAGL1, ZNF318, DLL1, THBS2, TULP4, SCAF8, TAB2, HIVEP2, PTPRK, HSF2, NUS1, AMD1, CDK19, CDC40, NR2E1, ATG5, PRDM1, SIM1, PNISR, EPHA7, MAP3K7, MDN1, ZNF292, SYNCRIP, TBX18, SMAP1, BAI3, LRRC1, RUNX2, CDC5L, HSP90AB1, UBR2, TRERF1, CCND3, USP49, FOXP4, CMTR1, RNF8, SRSF3, SPDEF, PHF1, RXRB, COL11A2, PBX2, ZBTB12, BAG6, PRRC2A, PPP1R10, GNL1, TRIM39, GABBR1, TRIM27, C6orf62, HIVEP1, TFAP2A, DSP, RREB1, GMDS, MLLT4, MAP3K4, WASF1, TFAP2B, PFDN6, BRD2, AGPAT1, DDX39B, CDYL, DAAM2, SYNGAP1, FAM135A, GRIK2, BEND3, MDGA1, SNAP91, RPS18, ESR1, SENP6, PTK7, PPP2R5D, HDAC2, CCNC, RIMS1, L3MBTL3, GRM4, PACSIN1, PDE10A, LATS1* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_80 | M | 5.7 | 6 | LEAT | GG | | 7:0-159138663 | 7 | Gain | 159.14 | 0.03 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_80 | M | 5.7 | 6 | LEAT | GG | | 19:0-23207354 | 19p | Gain | 23.21 | 0.12 | 1 | | *BRD4, CACNA1A, CACTIN, CARM1, CHERP, DAZAP1, DCAF15, DNMT1, FZR1, KHSRP, MAST1, MAU2, NACC1, PIAS4, PRKACA, RFX1, SIN3B, SMARCA4, TMEM259, TNPO2, UPF1, XAB2* | *MBD3, ANO8, KDM4B, REXO1, CDC34, SGTA, RPL18A, RAB3A, DAZAP1, C3, SUGP1, ABHD8, WDR18, MAST1, SAFB2, MLLT1, EIF3G, RFX1, DCAF15, CSNK1G2, UPF1, ELL, MAST3, MAP2K2, DPP9, PIAS4, BRD4, WIZ, NOTCH3, ARID3A, ADAMTS10, RNF126, NACC1, CELF5, RAB8A, CHAF1A, RFX2, PRKACA, EEF2, LMNB2, HNRNPM, STK11, CARM1, PIP5K1C, LRRC25, RANBP3, LPHN1, PDE4A, AP3D1, TMEM259, PTBP1, ATP13A1, PTPRS, XAB2, BABAM1, DNMT1, DDA1, CACNA1A, GATAD2A, LONP1, SIN3B, DNM2, MAU2, FZR1, NFIX, MAP2K7, KHSRP, DOT1L, ELAVL1, TNPO2, CACTIN, SMARCA4, CAMSAP3, ILF3, EPS15L1, UNC13A, APC2, EVI5L, HOMER3, CHERP, SAFB, DKFZP761J1410, FCHO1, MYO9B, SUGP2, VAV1, FKBP8* |
| LEAT_EEBB_80 | M | 5.7 | 6 | LEAT | GG | | 22:17897544-51304566 | 22q | Loss | 33.41 | 0.06 | 4 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, GNAZ, RAC2, CELSR1, SHANK3, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA1I, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, MICAL3, HIC2, PLXNB2, TNRC6B, ZNRF3* |
| LEAT_EEBB_85 | M | NA | 32 | LEAT | GG | | 4:0-191154276 | 4 | Gain | 191.15 | 0.04 | 5 | | *ABCE1, ANKRD17, CTBP1, DHX15, POLR2B, PPP3CA, SMARCA5, WDFY3* | *BOD1L1, PHOX2B, NFKB1, PDGFRA, UBE2K, KDR, PPAT, COPS4, RASGEF1B, RAPGEF2, FNIP2, ADD1, PPARGC1A, SEPT11, CNOT6L, CENPE, LEF1, SEC24B, CCNA2, NAF1, RPS3A, GPM6A, FBXW7, GRID2, SMARCA5, UCHL1, KIT, CTBP1, GABRB1, WDFY3, HMGB2, GRIA2, NAA15, HHIP, ABCE1, DCLK2, PDS5A, LDB2, PITX2, KIAA0232, REST, CLOCK, LIMCH1, HNRNPD, EDNRA, FAM193A, CRMP1, DHX15, TRIM2, RAP1GDS1, LIN54, FBXL5, RBPJ, YTHDC1, CLCN3, HTT, ANK2, NR3C2, FRYL, ANKRD17, SMARCAD1, G3BP2, PCGF3, ZNF827, POLR2B, ATP8A1, RBM47, RFC1, WHSC1, ELF2, FAT4, ELOVL6, PPP3CA, AFF1, CPE, JAKMIP1, KIAA0922, SLC4A4, BMPR1B, RPL9, RELL1, OTUD4, PLRG1, WDR1, BEND4, PDGFC, ANKRD50, SLIT2, CDKN2AIP, MAML3, GABRA2, TENM3, LPHN3, KLHL2, SMAD1* |
| LEAT_EEBB_85 | M | NA | 32 | LEAT | GG | | 6:0-171115067 | 6 | Gain | 171.12 | 0.04 | 8 | *MYB* | *GTPBP2, JARID2, KLHDC3, PPP2R5D, TMEM63B, TRERF1, XPO5* | *TFAP2D, RANBP9, TRAM2, FBXO5, TNFAIP3, DST, ATXN1, KIAA1244, CUL9, BACH2, REPS1, FAM65B, KIF13A, TMEM63B, TTBK1, GCM1, POU5F1, PHF3, NUP153, PSMB1, XPO5, SRF, SCUBE3, PHIP, C6orf136, PPARD, GLTSCR1L, EEF1A1, DEF6, TCP1, STXBP5, COL12A1, ZBTB2, RPS10, KLHDC3, TUBB, EPB41L2, PRPF4B, JARID2, KCNQ5, ARID1B, E2F3, DOPEY1, FYN, IGF2R, BRPF3, WTAP, REV3L, MAP3K5, ANKS1A, SLC35F1, PLAGL1, ZNF318, DLL1, THBS2, TULP4, SCAF8, TAB2, HIVEP2, PTPRK, HSF2, NUS1, AMD1, CDK19, CDC40, NR2E1, ATG5, PRDM1, SIM1, PNISR, EPHA7, MAP3K7, MDN1, ZNF292, SYNCRIP, TBX18, SMAP1, BAI3, LRRC1, RUNX2, CDC5L, HSP90AB1, UBR2, TRERF1, CCND3, USP49, FOXP4, CMTR1, RNF8, SRSF3, SPDEF, PHF1, RXRB, COL11A2, PBX2, ZBTB12, BAG6, PRRC2A, PPP1R10, GNL1, TRIM39, GABBR1, TRIM27, C6orf62, HIVEP1, TFAP2A, DSP, RREB1, GMDS, MLLT4, MAP3K4, WASF1, TFAP2B, PFDN6, BRD2, AGPAT1, DDX39B, CDYL, DAAM2, SYNGAP1, FAM135A, GRIK2, BEND3, MDGA1, SNAP91, RPS18, ESR1, SENP6, PTK7, PPP2R5D, HDAC2, CCNC, RIMS1, L3MBTL3, GRM4, PACSIN1, PDE10A, LATS1* |
| LEAT_EEBB_87 | F | 14 | 54 | LEAT | GG | | 5:0-180915260 | 5 | Gain | 180.92 | 0.04 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_87 | F | 14 | 54 | LEAT | GG | | 7:0-159138663 | 7 | Gain | 159.14 | 0.04 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_87 | F | 14 | 54 | LEAT | GG | | 12:0-133851895 | 12 | Gain | 133.85 | 0.04 | 8 | *KRAS, PTPN11* | *ANKRD52, ATP2B1, CHD4, CS, DDX23, GCN1L1, HECTD4, LRP1, NAA25, NAB2, PA2G4, PLXNC1, RARG, SCN8A, SETD1B, SMARCC2, SMARCD1, STAT2, USP5* | *SENP1, HDAC7, FOXJ2, TNFRSF1A, CBX5, SART3, ASIC1, CSRNP2, CNOT2, USP5, HCFC2, RAB35, LRP1, SLC38A2, MSI1, HNF1A, TBX3, GDF11, ESPL1, KCNH3, KCNMB4, PLXNC1, SLC41A2, WBP11, ATF7IP, NAP1L1, ASUN, CDK17, CCND2, LRP6, CORO1C, LPCAT3, CUX2, LHX5, NAA25, ATP5B, PTGES3, EIF4B, COPZ1, CLSTN3, SMARCC2, ANKRD52, SETD1B, DUSP6, USP15, BTBD11, EPS8, MED13L, NCKAP1L, ITGA5, FRS2, CCT2, TMEM132B, HSP90B1, STAT6, GCN1L1, KMT2D, TUBA1A, DIP2B, PA2G4, LEMD3, DHX37, TBX5, STAT2, WNK1, MLXIP, PITPNM2, ULK1, EEA1, E2F7, SP1, FBXO21, RSRC2, XPOT, ARID2, PTPRB, TUBA1B, SARNP, ZCCHC8, FAM60A, NOS1, KSR2, PPP1CC, HNRNPA1, NTN4, DYRK2, RNF41, R3HDM2, CACNA1C, CS, PTPN11, SPPL3, SCN8A, SRGAP1, GIT2, MBD6, ATN1, CHD4, DNAJC14, RFX4, PCBP2, SCAF11, KDM2B, ATXN2, ZFC3H1, NABP2, COL2A1, NOP2, PRMT8, STK38L, DENND5B, EP400, CIT, TAOK3, RIC8B, MON2, SMARCD1, ETV6, ZNF384, CCDC64, LARP4, SLC38A1, AEBP2, KDM5A, SETD8, RASSF8, NCOR2, ATF7, SBNO1, TMEM132D, RPL6, RARG, PAN2, ZDHHC17, ATP2B1, PLEKHA5, DCTN2, CPSF6, NELL2, PPP1R12A, SOX5, SLC4A8, KIF5A, PRICKLE1, PTPN6, MDM2, NAV3, ATP2A2, CLIP1, ACVR1B, SFSWAP, RAN, PHC1, CAND1, MAP3K12, AGAP2, ANKS1B, PRPF40B, PPFIA2, HECTD4, BAZ2A, SPATS2, GRIN2B* |
| LEAT_EEBB_87 | F | 14 | 54 | LEAT | GG | | 20:0-63025520 | 20 | Gain | 63.03 | 0.02 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_98 | M | 15 | 18 | LEAT | GG | | 4:0-191154276 | 4 | Gain | 191.15 | 0.02 | 5 | | *ABCE1, ANKRD17, CTBP1, DHX15, POLR2B, PPP3CA, SMARCA5, WDFY3* | *BOD1L1, PHOX2B, NFKB1, PDGFRA, UBE2K, KDR, PPAT, COPS4, RASGEF1B, RAPGEF2, FNIP2, ADD1, PPARGC1A, SEPT11, CNOT6L, CENPE, LEF1, SEC24B, CCNA2, NAF1, RPS3A, GPM6A, FBXW7, GRID2, SMARCA5, UCHL1, KIT, CTBP1, GABRB1, WDFY3, HMGB2, GRIA2, NAA15, HHIP, ABCE1, DCLK2, PDSS A, LDB2, PITX2, KIAA0232, REST, CLOCK, LIMCH1, HNRNPD, EDNRA, FAM193A, CRMP1, DHX15, TRIM2, RAP1GDS1, LIN54, FBXL5, RBPJ, YTHDC1, CLCN3, HTT, ANK2, NR3C2, FRYL, ANKRD17, SMARCAD1, G3BP2, PCGF3, ZNF827, POLR2B, ATP8A1, RBM47, RFC1, WHSC1, ELF2, FAT4, ELOVL6, PPP3CA, AFF1, CPE, JAKMIP1, KIAA0922, SLC4A4, BMPR1B, RPL9, RELL1, OTUD4, PLRG1, WDR1, BEND4, PDGFC, ANKRD50, SLIT2, CDKN2AIP, MAML3, GABRA2, TENM3, LPHN3, KLHL2, SMAD1* |
| LEAT_EEBB_98 | M | 15 | 18 | LEAT | GG | | 7:0-134639161 | 7 | Gain | 134.64 | 0.03 | 18 | | *CAMK2B, OGDH, PSMC2, RAC1* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, PTPN12, LRRC4, IGF2BP3, TTYH3, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, C7orf60, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, ZKSCAN1, GTF2I, AHCYL2, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, RAPGEF5, C7orf26, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, GRB10, TNS3, SEPT7, TBX20, FOXP2, ATXN7L1, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, CADPS2, GTF2IRD1, LHFPL3* |
| LEAT_EEBB_98 | M | 15 | 18 | LEAT | GG | | 11:0-135006516 | 11 | Gain | 135.01 | 0.04 | 5 | | *ADRBK1, B3GNT1, CKAP5, CNIH2, DDB1, DPF2, GANAB, GNG3, GRAMD1B, KDM2A, KMT2A, LTBP3, MARK2, MEN1, MUC5B, NXF1, PCNXL3, PLCB3, PPP2R5B, PRPF19, PSMC3, RPS6KA4, SART1, SSRP1, SYVN1* | *TTC17, PPP2R5B, SPI1, PRPF19, MAPK8IP1, COPB1, PPFIA1, RRAS2, DAGLA, PRKRIR, DCUN1D5, EED, ARHGEF17, POLD3, FNBP4, OSBP, DDX6, ARCN1, NRXN2, FBXO3, GTF2H1, KCNC1, FAM160A2, LRRC4C, SERPING1, SSRP1, TENM4, ZC3H12C, HMBS, MYRF, FADS2, CWC15, VPS26B, YAP1, ZDHHC5, SIK3, C11orf84, LRP5, FAT3, PCF11, PKNOX2, PSMC3, APBB1, DCHS1, PATL1, RRM1, DDB1, HNRNPUL2, LTBP3, SUV420H1, SIK2, STIP1, CHRM1, RSF1, ADRBK1, RBM14, ARHGAP32, RAB6A, NPAS4, CNIH2, SART1, CLCF1, PPP1CA, AMBRA1, ZFP91, FJX1, PACS1, EHD1, DSCAML1, SF3B2, CSTF3, NUP98, KCNA4, PPME1, EEF1G, RPS6KA4, PDE2A, BCL9L, ZBTB16, SPTY2D1, C2CD2L, MEN1, USP47, CPSF7, CAPRIN1, GANAB, FAR1, ANO1, PCNXL3, UVRAG, FAM168A, MPPED2, CTR9, GAB2, DLG2, SYVN1, ATG2A, SF1, AHNAK, IPO7, BRSK2, ARNTL, CUL5, PICALM, NUMA1, PPP6R3, INCENP, NAV2, SOX6, ARHGEF12, CTNND1, QSER1, MARK2, HYOU1, RDX, RELA, RBM4, RBM14-RBM4, KLC2, PHF21A, GRM5, PSMA1, PAX6, ARRB1, TBCEL, PRDM11, PSMD13, CHRM4, AP2A2, WEE1, MAML2, TRIM3, KIRREL3, ZBTB44, TRAF6, FLI1, DPF2, ATG13, KDM2A, C11orf30, CREB3L1, CKAP5, MUC5B, GRAMD1B, API5, FZD4, RNF214, CELF1, NXF1, EIF3F, IGSF9B, ST5, KMT2A, HSPA8, OTUB1, PLCB3, SYT7* |
| LEAT_EEBB_98 | M | 15 | 18 | LEAT | GG | | 18:18570235-78077248 | 18q | CNN-LOH | 59.51 | 0.01 | 1 | | *MAPRE2, SMAD7, TCF4* | *ZNF236, WDR7, DSG1, ZNF24, NOL4, PHLPP1, CDH2, ASXL3, ESCO1, C18orf8, RNF165, ZCCHC2, SETBP1, ATP5A1, TRAPPC8, ZNF407, TSHZ1, ZNF532, SMAD4, MALT1, ZNF521, CTIF, SOCS6, KIAA1468, TCF4, ROCK1, NEDD4L, SMAD2, CXXC1, CELF4, DCC, ZNF516, PIAS2, MBD1* |
| LEAT_EEBB_98 | M | 15 | 18 | LEAT | GG | | 21:20190260-48129895 | 21q | CNN-LOH | 27.94 | 0.03 | 2 | | | *HUNK, SIK1, CCT8, TIAM1, SCAF4, IFNGR2, PKNOX1, U2AF1, TRAPPC10, ZBTB21, PAXBP1, BRWD1, GABPA, SON, ETS2, COL6A1, SLC5A3, ITSN1, AGPAT3, DYRK1A, DSCAM, MORC3, ERG, SYNJ1* |
| LEAT_EEBB_101 | F | 9 | 13 | LEAT | GG | | 5:0-180915260 | 5 | Gain | 180.92 | 0.29 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_101 | F | 9 | 13 | LEAT | GG | | 6:0-171115067 | 6 | Gain | 171.12 | 0.33 | 8 | *MYB* | *GTPBP2, JARID2, KLHDC3, PPP2R5D, TMEM63B, TRERF1, XPO5* | *TFAP2D, RANBP9, TRAM2, FBXO5, TNFAIP3, DST, ATXN1, KIAA1244, CUL9, BACH2, REPS1, FAM65B, KIF13A, TMEM63B, TTBK1, GCM1, POU5F1, PHF3, NUP153, PSMB1, XPO5, SRF, SCUBE3, PHIP, C6orf136, PPARD, GLTSCR1L, EEF1A1, DEF6, TCP1, STXBP5, COL12A1, ZBTB2, RPS10, KLHDC3, TUBB, EPB41L2, PRPF4B, JARID2, KCNQ5, ARID1B, E2F3, DOPEY1, FYN, IGF2R, BRPF3, WTAP, REV3L, MAP3K5, ANKS1A, SLC35F1, PLAGL1, ZNF318, DLL1, THBS2, TULP4, SCAF8, TAB2, HIVEP2, PTPRK, HSF2, NUS1, AMD1, CDK19, CDC40, NR2E1, ATG5, PRDM1, SIM1, PNISR, EPHA7, MAP3K7, MDN1, ZNF292, SYNCRIP, TBX18, SMAP1, BAI3, LRRC1, RUNX2, CDC5L, HSP90AB1, UBR2, TRERF1, CCND3, USP49, FOXP4, CMTR1, RNF8, SRSF3, SPDEF, PHF1, RXRB, COL11A2, PBX2, ZBTB12, BAG6, PRRC2A, PPP1R10, GNL1, TRIM39, GABBR1, TRIM27, C6orf62, HIVEP1, TFAP2A, DSP, RREB1, GMDS, MLLT4, MAP3K4, WASF1, TFAP2B, PFDN6, BRD2, AGPAT1, DDX39B, CDYL, DAAM2, SYNGAP1, FAM135A, GRIK2, BEND3, MDGA1, SNAP91, RPS18, ESR1, SENP6, PTK7, PPP2R5D, HDAC2, CCNC, RIMS1, L3MBTL3, GRM4, PACSIN1, PDE10A, LATS1*|
| LEAT_EEBB_101 | F | 9 | 13 | LEAT | GG | | 7:0-159138663 | 7 | Gain | 159.14 | 0.33 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4*|
| LEAT_EEBB_101 | F | 9 | 13 | LEAT | GG | | 10:0-135269349 | 10 | Gain | 135.27 | 0.32 | 3 | *PTEN* | *LDB1, PPP3CB, TRIM8, ZSWIM8* | *PSD, TDRD1, KIF11, MAP3K8, EPC1, MTPAP, NRP1, HNRNPH3, CCAR1, BTAF1, CPEB3, SLIT1, ABLIM1, PPRC1, ARID5B, DOCK1, ANK3, DIP2C, C10orf12, NODAL, KAT6B, HECTD2, WAPAL, KIF5B, TRIM8, MLLT10, LARP4B, CAMK2G, SEPHS1, GRID1, FAM208B, PDZD8, ZMIZ1, PCGF5, PIK3AP1, SEC24C, TAF3, SORCS1, NKX2-3, HELLS, DDX21, WAC, ATRNL1, RET, SH3PXD2A, ADD3, UPF2, FRMD4A, ZRANB1, MCMBP, GTPBP4, LRRTM3, MGEA5, ZEB1, SGMS1, SMC3, SFMBT2, INPP5A, EBF3, TIAL1, EIF3A, CACUL1, HSPA12A, FAM160B1, SHOC2, SLK, TAF5, CNNM2, SUFU, ACTR1A, NFKB2, SCD, CHUK, LCOR, TM9SF3, LGI1, ZCCHC24, DLG5, ZNF503, H2AFY2, TET1, BICC1, IPMK, PRKG1, MAPK8, PARD3, FAM171A1, CAMK1D, PPP3CB, PSAP, HERC4, CDK1, RASGEF1A, ITGB1, ARHGAP21, ZMYND11, ZSWIM8, JMJD1C, HK1, KCNMA1, P4HA1, LDB1, TSPAN14, HNRNPF, RBM17, CELF2, FGFR2, CCNJ, KLF6, CUL2, TCF7L2, EMX2*|
| LEAT_EEBB_101 | F | 9 | 13 | LEAT | GG | | 15:20052938-102531392 | 15q | Gain | 82.48 | 0.32 | 3 | | *RTF1, SIN3A* | *AQR, MGA, CSK, PIAS1, DLL4, SNRPA1, ABHD17C, IREB2, CTDSPL2, THBS1, KIF23, ADAM10, HERC2, AP3B2, SNX1, RAB11A, HCN4, TTBK2, IGF1R, ZNF710, FURIN, IQGAP1, UBR1, SPRED1, COPS2, LEO1, HDGFRP3, ZNF592, CLPX, MAP1A, PDIA3, PYGO1, CSNK1G1, ARNT2, OTUD7A, RPL4, FBXO22, RASGRP1, GABRB3, CYFIP1, SEMA6D, FBN1, ZNF609, MORF4L1, ASB7, NEO1, DPP8, NPTN, BNC1, TJP1, CASC5, MEF2A, LING01, MYO9A, ZNF770, NTRK3, AKAP13, INO80, ARIH1, USP3, SLTM, TP53BP1, RYR3, RTF1, NR2F2, CHD2, SIN3A, UBE3A, MYO5A, BAHD1, FRMD5, RASGRF1, RFX7, USP8, ACAN, DENND4A, HERC1, ANP32A, MAGEL2, DMXL2, TLE3, RNF111, MEIS2, TLN2, CPEB1, PARP6*|
| LEAT_EEBB_101 | F | 9 | 13 | LEAT | GG | | 20:0-63025520 | 20 | Gain | 63.03 | 0.32 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8*|
| LEAT_EEBB_101 | F | 9 | 13 | LEAT | GG | | 22:16000000-51304566 | 22q | Gain | 35.30 | 0.31 | 1 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, GNAZ, RAC2, CELSR1, SHANK3, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA1I, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, MICAL3, HIC2, PLXNB2, TNRC6B, ZNRF3*|
| LEAT_EEBB_103 | F | 18 | 23 | LEAT | GG | HS Type I | 2:0-243199373 | 2 | Gain | 243.20 | 0.02 | 3 | | *CAD, FBXO11, HDLBP, MAP4K4, PPP1CB, PPP3R1, RQCD1, SCN2A, SF3B1, SNRNP200, UNC80, VSNL1, XPO1* | *CYP26B1, INPP4A, TRIB2, TMEM131, MSH2, NRBP1, PCGF1, IGFBP5, PPP1R7, DLX2, PAPOLG, ATAD2B, AFTPH, ACVR2A, RIF1, ILKAP, PER2, KIDINS220, CCT7, USP37, REV1, EPC2, SIX3, PPIG, HECW2, SLC40A1, ACTR3, POLR1B, TFCP2L1, PTPN4, CLASP1, LRP2, POLR1A, EHBP1, ATF2, MCM6, CCNT2, R3HDM1, AGPS, ADAM23, CUL3, ADD2, CAD, RAB10, KIF3C, FOSL2, VPS54, MEIS1, AMMECR1L, CTDSP1, RQCD1, CRIM1, ASAP2, EPHA4, ZFP36L2, RANBP2, SCN3A, TRIP12, FMNL2, DPYSL5, EIF5B, REL, MEMO1, KCNJ3, EN1, INHBB, IWS1, FZD5, ARPC2, CTLA4, SIX2, SCN1A, ANTXR1, AGAP1, COL3A1, RPS7, SEMA4C, PRKCE, MAT2A, PSMD1, KLF7, CFLAR, SP3, SNED1, ADAM17, SPEG, SRSF7, SPAST, ROCK2, RAPH1, GLS, RNF144A, WDR33, NCL, SNRNP200, DIS3L2, FIGN, SF3B1, NPAS2, BCL11A, CCDC88A, PUM2, NR4A2, ERBB4, PPM1G, HDAC4, MAP4K4, FN1, ZNF512, STK39, AFF3, SPTBN1, GFPT1, SCN2A, ARID5A, SAP130, GTF3C2, INPP5D, MAP2, NCKAP1, STAT1, DLX1, BMPR2, COL5A2, RND3, ACTR2, DDX1, HSPD1, LRP1B, TBR1, HDLBP, ATG16L1, SPHKAP, XRCC5, STAT4, OSBPL6, HNRNPA3, BAZ2B, GALNT13, DPP10, CCT4, EFEMP1, SP9, PPP1CB, MOB1A, RAPGEF4, USP34, XPO1, SLC8A1, INO80D, FBXO11, PSME4, ATG4B, NRXN1, NCOA1, MBD5, WDR43, AAK1, SLC39A10, ARHGAP25, TET3, GIGYF2, ZNF638, KDM3A, ATG9A, HNRNPLL, MGAT5, PSMD14, KCMF1, MAP3K2, AGFG1, CALCRL, SATB2, UBR3, BIRC6, SOS1, TMEM185B, MYT1L, SESTD1, PAX8, CNTNAP5, TLK1, KANSL3, CREB1, KIF5C, ASXL2, UNC80, EIF2B4, GLI2, BZW1, IKZF2, KIF1A, FBXO41, REEP1, ZEB2, HSPE1-MOB4*|
| LEAT_EEBB_103 | F | 18 | 23 | LEAT | GG | HS Type I | 3:0-188200897 | 3 | Gain | 188.20 | 0.02 | 1 | *NPRL2, RAF1, PIK3CA* | *AP2M1, ATP2B2, ATP6V1A, CELSR3, CTNNB1, DHX30, EIF4G1, GSK3B, PBRM1,* | *SEMA3F, CELSR3, ZBTB47, NKTR, KALRN, SEC61A1, DAZL, SMARCC1, BHLHE40, TOPBP1, DNAJC13, PDZRN3, KAT2B, PIK3CA, CBLB, ARHGAP31, MECOM, XRN1, MAP3K13, RAB7A, RBM6, DOCK3, DYNC1LI1, IQSEC1, ISY1, STXBP5L, FEZF2, UBP1, TOMM70A, FGD5, RAB6B, SLC6A1, CACNA1D, ERC2, APPL1, DCUN1D1, AP2M1, SLC4A7, EOMES, STT3B, PSMD6, PRICKLE2, CCNL1, SLMAP, SNRK, SENP2, BSN, WDR48, ITPR1, ARF4, YEATS2, TWF2, ETV5, RPL15, ZNF654, PSMD2, KIAA2018, GSK3B,*|

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | *PIK3CA, PLXND1, RBM5, SEC61A1, STAG1, TOP2B* | *TMEM39A, LRRN1, TMEM108, RUVBL1, NR2C2, RBM15B, EIF4A2, PLXND1, NFKBIZ, IP6K2, CD86, RARB, COPB2, KPNA4, FNDC3B, SEC62, PLCH1, GATA2, PODXL2, KPNA1, NISCH, RBM5, CTNNB1, MITF, ARIH2, PPP4R2, FXR1, GRM7, MAP4, ATP2B2, ZNF148, TSC22D2, STAG1, TRIM71, BRPF1, SRGAP3, PDCD10, ATP2C1, PLXNA1, PRKCD, SFMBT1, PBRM1, QRICH1, THRB, UBE2E2, EPHB1, FRMD4B, ATXN7, ANKRD28, SETD5, MAGI1, BCL6, RAD54L2, SETD2, SCN5A, SATB1, PLCL2, ISY1-RAB43, EIF4G1, ZNF445, ACTL6A, TBL1XR1, ARPC4, USP19, TOP2B, TNIK, CNBP, DHX30, SCHIP1, NBEAL2, TRA2B, CNOT10, SLC6A6, SKIL, BAP1, TIPARP, ADCY5, CLASP2, U2SURP, WNT5A, MED12L, ZBTB20, CACNA2D3, SLITRK3, CAMKV, PIK3CB, CACNA2D2, ARMC8, ROBO2, TFDP2, LSAMP, FOXP1, FAM208A, PHC3, GMPS, DHX36, ZBTB38, DAG1* |
| LEAT_EEBB_103 | F | 18 | 23 | LEAT | GG | HS Type 1 | 7:0-159138663 | 7 | Gain | 159.14 | 0.27 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_103 | F | 18 | 23 | LEAT | GG | HS Type 1 | 9:0-141213431 | 9 | Loss | 141.21 | 0.36 | 3 | *TSC1* | *ABCA2, DNM1, GABBR2, GRIN1, NOTCH1, PHF19, SCAI, SET, SMU1, SPTAN1, STXBP1, TLN1, VCP, WDR5* | *SETX, GDA, CLTA, TAF1L, TRAF2, OLFM1, LRRC8A, KIAA0368, RALGPS1, DAB2IP, GABBR2, RNF38, ERP44, UBE2R2, BRINP1, MAPKAP1, UHRF2, FAM120A, ZNF462, NOTCH1, SMC2, GNAQ, ZNF618, ZER1, ALDH1A1, WNK2, NOL6, TSC1, ZMYND19, BRD3, ZNF483, SEC16A, TLN1, RPL7A, ANAPC2, PTCH1, TESK1, ANP32B, ABCA2, COL27A1, BICD2, PRRC2B, RAD23B, PAX5, WDR5, VCP, PHF2, NUP214, TOPORS, ASTN2, NAA35, CACNA1B, GRIN1, RABL6, COL5A1, RALGDS, RAPGEF1, ABL1, NUP188, SET, SPTAN1, DNM1, ENG, STXBP1, ZBTB34, ZBTB43, SCAI, NR5A1, LHX2, DENND1A, RABGAP1, RC3H2, RAB14, PHF19, MEGF9, PRPF4, UGCG, EPB41L4B, KLF4, SLC44A1, TEX10, NCBP1, CDC14B, SYK, SPIN1, ZCCHC6, NTRK2, HNRNPK, UBQLN1, TLE1, TLE4, RORB, SHB, CNTFR, TEK, MLLT3, RPS6, BNC2, PSIP1, NFIB, PTPRD, JAK2, CDC37L1, RFX3, SMARCA2, CAMSAP1, NTNG2, PPP2R4, GAPVD1, LHX6, SMU1, ELAVL2, SVEP1, DAPK1, GOLGA2, STRBP, SEMA4D, RUSC2, EHMT1, RXRA, NR6A1, FBXW2* |
| LEAT_EEBB_103 | F | 18 | 23 | LEAT | GG | HS Type 1 | 16:0-90354753 | 16 | Gain | 90.35 | 0.02 | 3 | *NPRL3, TSC2* | *CNOT1, CTCF, EDC4, FBXL19, MAPK8IP3, MAZ, NUDT21, SF3B3, STX1B, UBE2I, USP7* | *CACNG3, TFAP4, GLG1, STX1B, C16orf70, CORO1A, NUTF2, USP10, TSC2, CAPN15, TOX3, MAPK8IP3, SALL1, FUS, VPS4A, DYNC1LI2, XYLT1, RBL2, PKD1, RAB11FIP3, CREBBP, FOXF1, GNAO1, CSNK2A2, SRCAP, SETD1A, ZNF629, CTCF, NKD1, ZFHX3, CDH11, ZCCHC14, IRF8, JPH3, ATP6V0D1, ADCY9, VPS35, ZNF319, PLK1, NUDT21, ZC3H18, ANKRD11, SF3B3, PRKCB, XPO6, TAOK2, RBFOX1, TUBB3, DNAJA2, CNOT1, RANBP10, MKL2, RBBP6, GLYR1, SH2B1, RNF40, C16orf72, IL21R, MTSS1L, PDPK1, CASKIN1, USP7, NFATC3, UBE2I, FBRS, GTF3C1, PLCG2, FBXL19, RPL13, BANP, AP1G1, BRD7, ATXN2L, TNRC6A, MYH11, KIAA0430, ZC3H7A, GRIN2A, UBN1, FBXL16, TBC1D10B, CBFB, BCAR1, FAM65A, CYLD, NFAT5, GSPT1, PAPD5, HERPUD1, SMG1, CMIP, RSPRY1, ZNF423, RNPS1, CHD9, CDH8, TERF2* |
| LEAT_EEBB_103 | F | 18 | 23 | LEAT | GG | HS Type 1 | 17:0-75075664 | 17 | Loss | 75.08 | 0.05 | 1 | *NF1* | *ACACA, CACNA1G, CHD3, CLTC, CLUH, COL1A1, DLG4, EFTUD2, EZH1, FXR2, GIT1, HDAC5, KAT2A, KPNB1, MBTD1, MINK1, MTMR4, NEURL4, NF1, NLK, PIP4K2B, POLR2A, PRPF8, SENP3, SLC25A11, SREBF1, STAT3, STAT5A, STAT5B, SUPT6H, THRA, UBTF, UNK* | *YBX2, XYLT2, APPBP2, DERL2, MNT, TBX21, NUFIP2, PSMB3, RPL19, RPS6KB1, DDX5, ASIC2, HNF1B, COL1A1, HDAC5, HLF, FOXN1, TBX2, SOX9, NUP85, OMG, FXR2, RARA, RAP1GAP2, KDM6B, SRSF1, KAT7, PSMD11, TAOK1, SPAG9, VMP1, SLC4A1, KANSL1, SMURF2, RABEP1, PITPNM3, SMG6, YWHAE, PSMD3, ATP6V0A1, STAT3, FBXL20, SRCIN1, KCNH4, NCOR1, COPS3, SSH2, CLTC, PIP4K2B, MSI2, KPNB1, IGF2BP1, KLHL10, STAT5B, PSME3, EIF4A1, CRK, MED1, USP32, NEUROD2, UBTF, NLGN2, MRC2, GGNBP2, BPTF, PPM1E, ZBTB4, YPEL2, AP2B1, PITPNA, SUPT6H, PPP1R9B, LASP1, UBE2O, PHF23, KIF1C, CASKIN2, ATAD5, NPEPPS, POLR2A, SUZ12, FOXJ1, MTMR4, HSF5, MLLT6, RAB11FIP4, RTN4RL1, FMNL1, HEXIM1, PHF12, NDEL1, NXN, SYNRG, STAT5A, TLK2, IKZF3, SMARCE1, ACACA, RAI1, MAP2K4, MINK1, PSMD12, NF1, RHOT1, HELZ, CACNA1G, MYH10, MAP3K3, BECN1, NFE2L1, SP2, ARHGAP44, CHD3, ZZEF1, C17orf85, SPOP, CBX1, CNP, GIT1, MPRIP, PAFAH1B1, MED13, ALKBH5, NEURL4, DLG4, NLK, C17orf104, CAMTA2, DCAF7, TOP2A, TANC2, EFTUD2, GJC1, ERN1, INTS2, CDK12, ATXN7L3, DNAJC7, TOB1, LUC7L3, MYO18A, CDC27, ANKFY1, CLUH, PRPF8, CTDNEP1, PELP1, SOCS7, DDX42, MSL1, VEZF1, PITPNC1, MBTD1, PRKAR1A, UNK, LSM12, GPATCH8, NMT1, AC005358.1* |
| LEAT_EEBB_103 | F | 18 | 23 | LEAT | GG | HS Type 1 | 22:17223938-51304566 | 22q | Loss | 34.08 | 0.08 | 4 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, GNAZ, RAC2, CELSR1, SHANK3, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA1I, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, MICAL3, HIC2, PLXNB2, TNRC6B, ZNRF3* |
| LEAT_EEBB_50 | M | 26 | 27 | LEAT | IDA | HS Type 1 | 5:0-180915260 | 5 | Gain | 180.92 | 0.35 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| LEAT_EEBB_50 | M | 26 | 27 | LEAT | IDA | HS Type 1 | 7:0-159138663 | 7 | Gain | 159.14 | 0.35 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4*` |
| LEAT_EEBB_50 | M | 26 | 27 | LEAT | IDA | HS Type 1 | 11:0-135006516 | 11 | Gain | 135.01 | 0.34 | 5 | | *ADRBK1, B3GNT1, CKAP5, CNIH2, DDB1, DPF2, GANAB, GNG3, GRAMD1B, KDM2A, KMT2A, LTBP3, MARK2, MEN1, MUC5B, NXF1, PCNXL3, PLCB3, PPP2R5B, PRPF19, PSMC3, RPS6KA4, SART1, SSRP1, SYVN1* | *TTC17, PPP2R5B, SPI1, PRPF19, MAPK8IP1, COPB1, PPFIA1, RRAS2, DAGLA, PRKRIR, DCUN1D5, EED, ARHGEF17, POLD3, FNBP4, OSBP, DDX6, ARCN1, NRXN2, FBXO3, GTF2H1, KCNC1, FAM160A2, LRRC4C, SERPING1, SSRP1, TENM4, ZC3H12C, HMBS, MYRF, FADS2, CWC15, VPS26B, YAP1, ZDHHC5, SIK3, C11orf84, LRP5, FAT3, PCF11, PKNOX2, PSMC3, APBB1, DCHS1, PATL1, RRM1, DDB1, HNRNPUL2, LTBP3, SUV420H1, SIK2, STIP1, CHRM1, RSF1, ADRBK1, RBM14, ARHGAP32, RAB6A, NPAS4, CNIH2, SART1, CLCF1, PPP1CA, AMBRA1, ZFP91, FJX1, PACS1, EHD1, DSCAML1, SF3B2, CSTF3, NUP98, KCNA4, PPME1, EEF1G, RPS6KA4, PDE2A, BCL9L, ZBTB16, SPTY2D1, C2CD2L, MEN1, USP47, CPSF7, CAPRIN1, GANAB, FAR1, ANO1, PCNXL3, UVRAG, FAM168A, MPPED2, CTR9, GAB2, DLG2, SYVN1, ATG2A, SF1, AHNAK, IPO7, BRSK2, ARNTL, CUL5, PICALM, NUMA1, PPP6R3, INCENP, NAV2, SOX6, ARHGEF12, CTNND1, QSER1, MARK2, HYOU1, RDX, RELA, RBM4, RBM14-RBM4, KLC2, PHF21A, GRM5, PSMA1, PAX6, ARRB1, TBCEL, PRDM11, PSMD13, CHRM4, AP2A2, WEE1, MAML2, TRIM3, KIRREL3, ZBTB44, TRAF6, FLI1, DPF2, ATG13, KDM2A, C11orf30, CREB3L1, CKAP5, MUC5B, GRAMD1B, API5, FZD4, RNF214, CELF1, NXF1, EIF3F, IGSF9B, ST5, KMT2A, HSPA8, OTUB1, PLCB3, SYT7*` |
| LEAT_EEBB_50 | M | 26 | 27 | LEAT | IDA | HS Type 1 | 12:0-133851895 | 12 | Gain | 133.85 | 0.06 | 8 | *KRAS, PTPN11* | *ANKRD52, ATP2B1, CHD4, CS, DDX23, GCN1L1, HECTD4, LRP1, NAA25, NAB2, PA2G4, PLXNC1, RARG, SCN8A, SETD1B, SMARCC2, SMARCD1, STAT2, USP5* | *SENP1, HDAC7, FOXJ2, TNFRSF1A, CBX5, SART3, ASIC1, CSRNP2, CNOT2, USP5, HCFC2, RAB35, LRP1, SLC38A2, MSI1, HNF1A, TBX3, GDF11, ESPL1, KCNH3, KCNMB4, PLXNC1, SLC41A2, WBP11, ATF7IP, NAP1L1, ASUN, CDK17, CCND2, LRP6, CORO1C, LPCAT3, CUX2, LHX5, NAA25, ATP5B, PTGES3, EIF4B, COPZ1, CLSTN3, SMARCC2, ANKRD52, SETD1B, DUSP6, USP15, BTBD11, EPS8, MED13L, NCKAP1L, ITGA5, FRS2, CCT2, TMEM132B, HSP90B1, STAT6, GCN1L1, KMT2D, TUBA1A, DIP2B, PA2G4, LEMD3, DHX37, TBX5, STAT2, WNK1, MLXIP, PITPNM2, ULK1, EEA1, E2F7, SP1, FBXO21, RSRC2, XPOT, ARID2, PTPRB, TUBA1B, SARNP, ZCCHC8, FAM60A, NOS1, KSR2, PPP1CC, HNRNPA1, NTN4, DYRK2, RNF41, R3HDM2, CACNA1C, CS, PTPN11, SPPL3, SCN8A, SRGAP1, GIT2, MBD6, ATN1, CHD4, DNAJC14, RFX4, PCBP2, SCAF11, KDM2B, ATXN2, ZFC3H1, NABP2, COL2A1, NOP2, PRMT8, STK38L, DENND5B, EP400, CIT, TAOK3, RIC8B, MON2, SMARCD1, ETV6, ZNF384, CCDC64, LARP4, SLC38A1, AEBP2, KDM5A, SETD8, RASSF8, NCOR2, ATF7, SBNO1, TMEM132D, RPL6, RARG, PAN2, ZDHHC17, ATP2B1, PLEKHA5, DCTN2, CPSF6, NELL2, PPP1R12A, SOX5, SLC4A8, KIF5A, PRICKLE1, PTPN6, MDM2, NAV3, ATP2A2, CLIP1, ACVR1B, SFSWAP, RAN, PHC1, CAND1, MAP3K12, AGAP2, ANKS1B, PRPF40B, PPFIA2, HECTD4, BAZ2A, SPATS2, GRIN2B*` |
| LEAT_EEBB_50 | M | 26 | 27 | LEAT | IDA | HS Type 1 | 13:19377679-115169878 | 13q | Gain | 95.79 | 0.06 | 2 | | *MYCBP2, NBEA* | *AKAP11, FLT3, EFNB2, XPO4, RNF17, DCLK1, IPO5, KPNA3, RB1, RBM26, ZC3H13, USP12, FLT1, DACH1, INTS6, PDS5B, HSPH1, PSPC1, COG3, FBXL3, MYO16, CHAMP1, TFDP1, CUL4A, ARHGEF7, COL4A1, TNFSF13B, FAM155A, TPP2, TM9SF2, DOCK9, HS6ST3, SLITRK1, SPRY2, KLF12, KBTBD6, FOXO1, FRY, SLC7A1, PAN3, NUPL1, FGF9, LATS2, ZMYM2, LRCH1, NBEA, MTUS2, TSC22D1, FNDC3A, MYCBP2*` |
| LEAT_EEBB_50 | M | 26 | 27 | LEAT | IDA | HS Type 1 | 17:0-81195210 | 17 | Gain | 81.20 | 0.24 | 1 | *NF1* | *ACACA, CACNA1G, CHD3, CLTC, CLUH, COL1A1, DLG4, EFTUD2, EZH1, FXR2, GIT1, HDAC5, KAT2A, KPNB1, MBTD1, MINK1, MTMR4, NEURL4, NF1, NLK, PIP4K2B, POLR2A, PRPF8, SENP3, SLC25A11, SREBF1, STAT3, STAT5A, STAT5B, SUPT6H, THRA, UBTF, UNK* | *YBX2, XYLT2, APPBP2, DERL2, MNT, TBX21, NUFIP2, PSMB3, RPL19, RPS6KB1, DDX5, ASIC2, HNF1B, COL1A1, HDAC5, HLF, FOXN1, TBX2, SOX9, NUP85, OMG, FXR2, RARA, RAP1GAP2, KDM6B, SRSF1, KAT7, PSMD11, TAOK1, SPAG9, VMP1, SLC4A1, KANSL1, SMURF2, RABEP1, PITPNM3, SMG6, YWHAE, PSMD3, ATP6V0A1, STAT3, FBXL20, SRCIN1, KCNH4, NCOR1, COPS3, SSH2, CLTC, EIF4A3, CBX8, CBX4, PIP4K2B, MSI2, KPNB1, IGF2BP1, KLHL10, STAT5B, PSME3, EIF4A1, CRK, MED1, USP32, NEUROD2, UBTF, NLGN2, MRC2, GGNBP2, BPTF, FASN, RPTOR, RP11-1055B8.7, PPM1E, CBX2, ZBTB4, YPEL2, AP2B1, PITPNA, CSNK1D, SUPT6H, PPP1R9B, LASP1, UBE2O, PHF23, KIF1C, CASKIN2, ATAD5, NPEPPS, POLR2A, SUZ12, FOXJ1, MTMR4, HSF5, CHMP6, MLLT6, RAB11FIP4, NPLOC4, RTN4RL1, P4HB, FMNL1, HEXIM1, PHF12, NDEL1, TNRC6C, NXN, SYNRG, STAT5A, TLK2, IKZF3, SMARCE1, ACACA, RAI1, MAP2K4, MINK1, PSMD12, NF1, RHOT1, HELZ, CACNA1G, MYH10, CYTH1, MAP3K3, BECN1, NFE2L1, SP2, ARHGAP44, CHD3, ZZEF1, C17orf85, GPS1, SPOP, CBX1, CNP, GIT1, MPRIP, PAFAH1B1, MED13, ALKBH5, NEURL4, DLG4, NLK, C17orf104, CAMTA2, DCAF7, TOP2A, TANC2, EFTUD2, GJC1, SEPT9, ERN1, INTS2, CDK12, ATXN7L3, DNAJC7, TOB1, LUC7L3, MYO18A, CDC27, ANKFY1, CLUH, PRPF8, CTDNEP1, PELP1, SOCS7, DDX42, MSL1, VEZF1, PITPNC1, MBTD1, PRKAR1A, UNK, LSM12, GPATCH8, NMT1, AC005358.1*` |
| LEAT_EEBB_50 | M | 26 | 27 | LEAT | IDA | HS Type 1 | 19:0-59128983 | 19 | Gain | 59.13 | 0.33 | 5 | | *ACTN4, AP2A1, ATP1A3, BRD4, BRSK1, CA11, CACNA1A, CACTIN, CADM4, CALM3, CARM1, CHERP, CLPTM1, DAZAP1, DCAF15, DNMT1, DYRK1B, EGLN2, FZR1, GRIN2D, GSK3A, IRF2BP1, KHSRP, LMTK3, MAP4K1, MAST1, MAU2, NACC1, NOVA2, NUMBL, PIAS4,* | *PPP5C, CD22, MBD3, ANO8, KDM4B, BCL3, REXO1, CDC34, HNRNPL, RELB, SLC17A7, SGTA, FBL, RASIP1, RPL18A, RAB3A, KMT2B, USF2, ERF, GSK3A, SIPA1L3, CADM4, DAZAP1, TSHZ3, C3, VASP, SYMPK, UBA2, TSKS, SUGP1, ABHD8, WDR18, MAST1, PVRL2, SAFB2, MLLT1, ACTN4, NUMBL, TRIM28, ZC3H4, EIF3G, ZNF331, RFX1, DCAF15, CSNK1G2, HPN, CCNE1, UPF1, ELL, MAST3, MARK4, GRIK5, MAP2K2, DPP9, PIAS4, GRIN2D, BRD4, WIZ, NOTCH3, PRKCG, ARID3A, SAE1, ADAMTS10, RPS11, RNF126, NACC1, CELF5, SHANK1, KIAA0355, RAB8A, CHAF1A, IRF2BP1, RFX2, ZNF296, LTBP4, PRKACA, U2AF2, EEF2, BRSK1, SAMD4B, PRPF31, PPP2R1A, LMNB2, HNRNPM, LENG8, STK11, CARM1, PPFIA3, PIP5K1C, CLPTM1, ARHGEF1, LRRC25, RANBP3, LPHN1, PDE4A, FOSB, AP3D1, DPF1, ZNF536, TMEM259, PTBP1, ATP13A1, PTPRS, XAB2, AP2A1, BABAM1, DNMT1, DDA1, CACNA1A, GATAD2A, SCAF1, LONP1, TTYH1, SIN3B, DNM2, STRN4, HNRNPUL1, MAU2, FZR1, GLTSCR1, NFIX, MAP2K7, KHSRP, DOT1L, ARHGAP35, CNOT3, ELAVL1, PPP6R1, PRR12, TNPO2, TOMM40, CACTIN, SMARCA4, PRKD2, CAMSAP3, ILF3, PRMT1, EPS15L1, ZNF146, UNC13A, POU2F2, APC2, EVI5L, HOMER3, ATP1A3, CHERP, RPL18, CIC, LRFN3, SAFB, MAP4K1, DKFZP761J1410, DYRK1B, FCHO1, RUVBL2, MYO9B, RPS5, NUP62, SNRNP70, SUPT5H, MYH14, SUGP2, VAV1, NPAS1, FKBP8*` |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | *PPFIA3, PPP2R1A, PPP5C, PRKACA, PRKCG, PRMT1, PRPF31, PRR12, RFX1, SHANK1, SIN3B, SLC8A2, SMARCA4, SPTBN4, SUPT5H, TMEM259, TNPO2, TOMM40, U2AF2, UPF1, XAB2* | |
| LEAT_EEBB_50 | M | 26 | 27 | LEAT | IDA | HS Type I | 20:0-63025520 | 20 | Gain | 63.03 | 0.03 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_112 | M | 8 | 26 | LEAT | IDA | | 9:0-141213431 | 9 | Loss | 141.21 | 0.22 | 3 | *TSC1* | *ABCA2, DNM1, GABBR2, GRIN1, NOTCH1, PHF19, SCAI, SET, SMU1, SPTAN1, STXBP1, TLN1, VCP, WDR5* | *SETX, GDA, CLTA, TAF1L, TRAF2, OLFM1, LRRC8A, KIAA0368, RALGPS1, DAB2IP, GABBR2, RNF38, ERP44, UBE2R2, BRINP1, MAPKAP1, UHRF2, FAM120A, ZNF462, NOTCH1, SMC2, GNAQ, ZNF618, ZER1, ALDH1A1, WNK2, NOL6, TSC1, ZMYND19, BRD3, ZNF483, SEC16A, TLN1, RPL7A, ANAPC2, PTCH1, TESK1, ANP32B, ABCA2, COL27A1, BICD2, PRRC2B, RAD23B, PAX5, WDR5, VCP, PHF2, NUP214, TOPORS, ASTN2, NAA35, CACNA1B, GRIN1, RABL6, COL5A1, RALGDS, RAPGEF1, ABL1, NUP188, SET, SPTAN1, DNM1, ENG, STXBP1, ZBTB34, ZBTB43, SCAI, NR5A1, LHX2, DENND1A, RABGAP1, RC3H2, RAB14, PHF19, MEGF9, PRPF4, UGCG, EPB41L4B, KLF4, SLC44A1, TEX10, NCBP1, CDC14B, SYK, SPIN1, ZCCHC6, NTRK2, HNRNPK, UBQLN1, TLE1, TLE4, RORB, SHB, CNTFR, TEK, MLLT3, RPS6, BNC2, PSIP1, NFIB, PTPRD, JAK2, CDC37L1, RFX3, SMARCA2, CAMSAP1, NTNG2, PPP2R4, GAPVD1, LHX6, SMU1, ELAVL2, SVEP1, DAPK1, GOLGA2, STRBP, SEMA4D, RUSC2, EHMT1, RXRA, NR6A1, FBXW2* |
| LEAT_EEBB_112 | M | 8 | 26 | LEAT | IDA | | 18:0-78077248 | 18 | Loss | 78.08 | 0.24 | 1 | | *MAPRE2, SMAD7, TCF4* | *ZNF236, WDR7, DSG1, ZNF24, NOL4, THOC1, USP14, ANKRD12, PHLPP1, CDH2, ASXL3, ESCO1, C18orf8, RNF165, ZCCHC2, SETBP1, ATP5A1, TRAPPC8, ZNF407, PTPN2, DLGAP1, SMCHD1, TSHZ1, GNAL, ZNF532, SMAD4, MALT1, ZNF521, CTIF, SOCS6, KIAA1468, TCF4, ROCK1, NEDD4L, PPP4R1, SMAD2, CXXC1, CELF4, DCC, ZNF516, PTPRM, PIAS2, MBD1* |
| LEAT_EEBB_113 | M | 2 | 47 | LEAT | MVNT | | 5:0-180915260 | 5 | Gain | 180.92 | 0.02 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A, DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_113 | M | 2 | 47 | LEAT | MVNT | | 7:0-159138663 | 7 | Gain | 159.14 | 0.06 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_95 | F | 6 | 39 | LEAT | PA | | 4:0-191154276 | 4 | Gain | 191.15 | 0.15 | 5 | | *ABCE1, ANKRD17, CTBP1, DHX15, POLR2B, PPP3CA, SMARCA5, WDFY3* | *BOD1L1, PHOX2B, NFKB1, PDGFRA, UBE2K, KDR, PPAT, COPS4, RASGEF1B, RAPGEF2, FNIP2, ADD1, PPARGC1A, SEPT11, CNOT6L, CENPE, LEF1, SEC24B, CCNA2, NAF1, RPS3A, GPM6A, FBXW7, GRID2, SMARCA5, UCHL1, KIT, CTBP1, GABRB1, WDFY3, HMGB2, GRIA2, NAA15, HHIP, ABCE1, DCLK2, PDS5A, LDB2, PITX2, KIAA0232, REST, CLOCK, LIMCH1, HNRNPD, EDNRA, FAM193A, CRMP1, DHX15, TRIM2, RAP1GDS1, LIN54, FBXL5, RBPJ, YTHDC1, CLCN3, HTT, ANK2, NR3C2, FRYL, ANKRD17, SMARCAD1, G3BP2, PCGF3, ZNF827, POLR2B, ATP8A1, RBM47, RFC1, WHSC1, ELF2, FAT4, ELOVL6, PPP3CA, AFF1, CPE, JAKMIP1, KIAA0922, SLC4A4, BMPR1B, RPL9, RELL1, OTUD4, PLRG1, WDR1, BEND4, PDGFC, ANKRD50, SLIT2, CDKN2AIP, MAML3, GABRA2, TENM3, LPHN3, KLHL2, SMAD1* |
| LEAT_EEBB_95 | F | 6 | 39 | LEAT | PA | | 6:0-171115067 | 6 | Gain | 171.12 | 0.06 | 8 | *MYB* | *GTPBP2, JARID2, KLHDC3, PPP2R5D, TMEM63B, TRERF1, XPO5* | *TFAP2D, RANBP9, TRAM2, FBXO5, TNFAIP3, DST, ATXN1, KIAA1244, CUL9, BACH2, REPS1, FAM65B, KIF13A, TMEM63B, TTBK1, GCM1, POU5F1, PHF3, NUP153, PSMB1, XPO5, SRF, SCUBE3, PHIP, C6orf136, PPARD, GLTSCR1L, EEF1A1, DEF6, TCP1, STXBP5, COL12A1, ZBTB2, RPS10, KLHDC3, TUBB, EPB41L2, PRPF4B, JARID2, KCNQ5, ARID1B, E2F3, DOPEY1, FYN, IGF2R, BRPF3, WTAP, REV3L, MAP3K5, ANKS1A, SLC35F1, PLAGL1, ZNF318, DLL1, THBS2, TULP4, SCAF8, TAB2, HIVEP2, PTPRK, HSF2, NUS1, AMD1, CDK19, CDC40, NR2E1, ATG5, PRDM1, SIM1, PNISR, EPHA7, MAP3K7, MDN1, ZNF292, SYNCRIP, TBX18, SMAP1, BAI3, LRRC1, RUNX2, CDC5L, HSP90AB1, UBR2, TRERF1, CCND3, USP49, FOXP4, CMTR1, RNF8, SRSF3, SPDEF, PHF1, RXRB, COL11A2, PBX2, ZBTB12, BAG6, PRRC2A, PPP1R10, GNL1, TRIM39, GABBR1, TRIM27, C6orf62, HIVEP1, TFAP2A, DSP, RREB1, GMDS, MLLT4,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | *MAP3K4, WASF1, TFAP2B, PFDN6, BRD2, AGPAT1, DDX39B, CDYL, DAAM2, SYNGAP1, FAM135A, GRIK2, BEND3, MDGA1, SNAP91, RPS18, ESR1, SENP6, PTK7, PPP2R5D, HDAC2, CCNC, RIMS1, L3MBTL3, GRM4, PACSIN1, PDE10A, LATS1* |
| LEAT_EEBB_95 | F | 6 | 39 | LEAT | PA | | 7:0-159138663 | 7 | Gain | 159.14 | 0.15 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_95 | F | 6 | 39 | LEAT | PA | | 10:0-135534747 | 10 | Gain | 135.53 | 0.14 | 3 | *PTEN* | *LDB1, PPP3CB, TRIM8, ZSWIM8* | *PSD, TDRD1, KIF11, MAP3K8, EPC1, MTPAP, NRP1, HNRNPH3, CCAR1, BTAF1, CPEB3, SLIT1, ABLIM1, PPRC1, ARID5B, DOCK1, ANK3, DIP2C, C10orf12, NODAL, KAT6B, HECTD2, WAPAL, KIF5B, TRIM8, MLLT10, LARP4B, CAMK2G, SEPHS1, GRID1, FAM208B, PDZD8, ZMIZ1, PCGF5, PIK3AP1, SEC24C, TAF3, SORCS1, NKX2-3, HELLS, DDX21, WAC, ATRNL1, RET, SH3PXD2A, ADD3, UPF2, FRMD4A, ZRANB1, MCMBP, GTPBP4, LRRTM3, MGEA5, ZEB1, SGMS1, SMC3, SFMBT2, INPP5A, EBF3, TIAL1, EIF3A, CACUL1, HSPA12A, FAM160B1, SHOC2, SLK, TAF5, CNNM2, SUFU, ACTR1A, NFKB2, SCD, CHUK, LCOR, TM9SF3, LGI1, ZCCHC24, DLG5, ZNF503, H2AFY2, TET1, BICC1, IPMK, PRKG1, MAPK8, PARD3, FAM171A1, CAMK1D, PPP3CB, PSAP, HERC4, CDK1, RASGEF1A, ITGB1, ARHGAP21, ZMYND11, ZSWIM8, JMJD1C, HK1, KCNMA1, P4HA1, LDB1, TSPAN14, HNRNPF, RBM17, CELF2, FGFR2, CCNJ, KLF6, CUL2, TCF7L2, EMX2* |
| LEAT_EEBB_95 | F | 6 | 39 | LEAT | PA | | 17:26851501-81195210 | 17q | CNN-LOH | 54.34 | 0.12 | 2 | *NF1* | *ACACA, CACNA1G, CLTC, COL1A1, EFTUD2, EZH1, GIT1, HDAC5, KAT2A, KPNB1, MBTD1, MTMR4, NF1, PIP4K2B, STAT3, STAT5A, STAT5B, SUPT6H, THRA, UBTF, UNK* | *XYLT2, APPBP2, TBX21, NUFIP2, PSMB3, RPL19, RPS6KB1, DDX5, ASIC2, HNF1B, COL1A1, HDAC5, HLF, FOXN1, TBX2, SOX9, NUP85, OMG, RARA, SRSF1, KAT7, PSMD11, TAOK1, SPAG9, VMP1, SLC4A1, KANSL1, SMURF2, PSMD3, ATP6V0A1, STAT3, FBXL20, SRCIN1, KCNH4, SSH2, CLTC, EIF4A3, CBX8, CBX4, PIP4K2B, MSI2, KPNB1, IGF2BP1, KLHL10, STAT5B, PSME3, MED1, USP32, NEUROD2, UBTF, MRC2, GGNBP2, BPTF, FASN, RPTOR, RP11-1055B8.7, PPM1E, CBX2, YPEL2, AP2B1, CSNK1D, SUPT6H, PPP1R9B, LASP1, UBE2O, CASKIN2, ATAD5, NPEPPS, SUZ12, FOXJ1, MTMR4, HSF5, CHMP6, MLLT6, RAB11FIP4, NPLOC4, P4HB, FMNL1, HEXIM1, PHF12, TNRC6C, SYNRG, STAT5A, TLK2, IKZF3, SMARCE1, ACACA, PSMD12, NF1, RHOT1, HELZ, CACNA1G, CYTH1, MAP3K3, BECN1, NFE2L1, SP2, GPS1, SPOP, CBX1, CNP, GIT1, MED13, C17orf104, DCAF7, TOP2A, TANC2, EFTUD2, GJC1, SEPT9, ERN1, INTS2, CDK12, ATXN7L3, DNAJC7, TOB1, LUC7L3, MYO18A, CDC27, SOCS7, DDX42, MSL1, VEZF1, PITPNC1, MBTD1, PRKAR1A, UNK, LSM12, GPATCH8, NMT1* |
| LEAT_EEBB_95 | F | 6 | 39 | LEAT | PA | | 19:0-59128983 | 19 | Gain | 59.13 | 0.07 | 5 | | *ACTN4, AP2A1, ATP1A3, BRD4, BRSK1, CA11, CACNA1A, CACTIN, CADM4, CALM3, CARM1, CHERP, CLPTM1, DAZAP1, DCAF15, DNMT1, DYRK1B, EGLN2, FZR1, GRIN2D, GSK3A, IRF2BP1, KHSRP, LMTK3, MAP4K1, MAST1, MAU2, NACC1, NOVA2, NUMBL, PIAS4, PPFIA3, PPP2R1A, PPP5C, PRKACA, PRKCG, PRMT1, PRPF31, PRR12, RFX1, SHANK1, SIN3B, SLC8A2, SMARCA4, SPTBN4, SUPT5H, TMEM259, TNPO2, TOMM40, U2AF2, UPF1, XAB2* | *PPP5C, CD22, MBD3, ANO8, KDM4B, BCL3, REXO1, CDC34, HNRNPL, RELB, SLC17A7, SGTA, FBL, RASIP1, RPL18A, RAB3A, KMT2B, USF2, ERF, GSK3A, SIPA1L3, CADM4, DAZAP1, TSHZ3, C3, VASP, SYMPK, UBA2, TSKS, SUGP1, ABHD8, WDR18, MAST1, PVRL2, SAFB2, MLLT1, ACTN4, NUMBL, TRIM28, ZC3H4, EIF3G, ZNF331, RFX1, DCAF15, CSNK1G2, HPN, CCNE1, UPF1, ELL, MAST3, MARK4, GRIK5, MAP2K2, DPP9, PIAS4, GRIN2D, BRD4, WIZ, NOTCH3, PRKCG, ARID3A, SAE1, ADAMTS10, RPS11, RNF126, NACC1, CELF5, SHANK1, KIAA0355, RAB8A, CHAF1A, IRF2BP1, RFX2, ZNF296, LTBP4, PRKACA, U2AF2, EEF2, BRSK1, SAMD4B, PRPF31, PPP2R1A, LMNB2, HNRNPM, LENG8, STK11, CARM1, PPFIA3, PIP5K1C, CLPTM1, ARHGEF1, LRRC25, RANBP3, LPHN1, PDE4A, FOSB, AP3D1, DPF1, ZNF536, TMEM259, PTBP1, ATP13A1, PTPRS, XAB2, AP2A1, BABAM1, DNMT1, DDA1, CACNA1A, GATAD2A, SCAF1, LONP1, TTYH1, SIN3B, DNM2, STRN4, HNRNPUL1, MAU2, FZR1, GLTSCR1, NFIX, MAP2K7, KHSRP, DOT1L, ARHGAP35, CNOT3, ELAVL1, PPP6R1, PRR12, TNPO2, TOMM40, CACTIN, SMARCA4, PRKD2, CAMSAP3, ILF3, PRMT1, EPS15L1, ZNF146, UNC13A, POU2F2, APC2, EVI5L, HOMER3, ATP1A3, CHERP, RPL18, CIC, LRFN3, SAFB, MAP4K1, DKFZP761J1410, DYRK1B, FCHO1, RUVBL2, MYO9B, RPS5, NUP62, SNRNP70, SUPT5H, MYH14, SUGP2, VAV1, NPAS1, FKBP8* |
| LEAT_EEBB_102 | F | 5 | 11 | LEAT | PA | | 17:52121998-81195210 | 17q22-25.3 | Gain | 29.07 | 0.23 | 1 | | *CLTC, MTMR4, UNK* | *APPBP2, RPS6KB1, DDX5, HLF, TBX2, SOX9, NUP85, SRSF1, VMP1, SMURF2, CLTC, EIF4A3, CBX8, CBX4, MSI2, USP32, MRC2, BPTF, FASN, RPTOR, RP11-1055B8.7, PPM1E, CBX2, YPEL2, CSNK1D, UBE2O, CASKIN2, FOXJ1, MTMR4, HSF5, CHMP6, NPLOC4, P4HB, TNRC6C, TLK2, PSMD12, HELZ, CYTH1, MAP3K3, GPS1, MED13, DCAF7, TANC2, SEPT9, ERN1, INTS2, DDX42, VEZF1, PITPNC1, PRKAR1A, UNK* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 5:0-180915260 | 5 | Gain | 180.92 | 0.10 | 11 | | *ANKHD1, CAMK2A, CANX, CSNK1A1, CYFIP2, DDX46, ETF1, HDAC3, HNRNPH1, IK, KDM3B, LARP1, NR2F1, PPP2CA, PURA, RBM22, TCERG1, UNC5A, ZMAT2* | *LCP2, YTHDC2, TRIP13, RBM22, SKIV2L2, PAPD7, LOX, LNPEP, NUP155, NR3C1, ELL2, IRF1, ADAMTS2, ZSWIM6, PDLIM4, NDFIP1, AP3B1, FST, DOCK2, NDST1, PDGFRB, FLT4, FAF2, SLC12A2, NNT, FGF10, PDE8B, SLC9A3, SOX30, ZFR, CDH6, VCAN, CPEB4, FBXW11, SLC1A3, ARRDC3, RBM27, JAKMIP2, VDAC1, AFF4, SLC6A3, MARCH6, SKP2, PLK2, RASA1, MEGF10, YIPF5, ARHGAP26, ZMAT2, CCT5, NIPBL, CHD1, PCDH1, HMGCR, PCDHAC2, DBN1, MAML1, KIAA0947, TCERG1, MAP1B, UTP15, RAPGEF6, NPM1, CREBRF, ANKHD1, HSPA9, PTGER4, CTNNA1, HCN1, GPRIN1, CTNND2, HSPA4, ZNF608, PCDHGC4, PAIP1, PWWP2A, KLHL3, TERT, DMXL1, EBF1, KDM3B, TRIM41, SSBP2, HMGCS1, SNX18, NKX2-5, UNC5A, PURA, EFNA5, RGS7BP, HOMER1, LARP1, PPP2R2B, TNPO1, ADCY2, PDE4D, DPYSL3, SEMA6A, TRIO, DDX46, HNRNPH1, RICTOR, ETF1, NRG2, KIF3A, SNX2, ADAMTS6, MIER3, IL6ST, PRLR, MTMR12, FAM169A, DRD1, SYNPO, FBXO38, MATR3, CDC23, CSNK1G3, JMY, CAMK2A,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | *DIAPH1, UBE2D2, MAP3K1, KIF2A, FCHO2, PDZD2, NSD1, MGAT1, APC, PPP2CA, TCOF1, DDX4, ERBB2IP, KCTD16, FBN2, ZNF131, FNIP1, DROSHA, KCNN2, GNB2L1, EXOC3, CTC-432M15.3, FAM193B, CSNK1A1, RAI14, TENM2, GRIA1, RNF145, SLIT3, PIK3R1, CYFIP2, ANKHD1-EIF4EBP3, SCAMP1, FBXL17, SMAD5* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 7:0-65796278 | 7p22.3-q11.21 | Gain | 65.80 | 0.11 | 1 | | *CAMK2B, OGDH, RAC1* | *RALA, HOXA11, CDK13, ITGB8, SP4, OGDH, PSMA2, AHR, INHBA, IGF2BP3, TTYH3, GNA12, EGFR, CCT6A, ADCY1, KBTBD2, USP42, ZMIZ2, ELMO1, ACTB, VWC2, RAPGEF5, C7orf26, HNRNPA2B1, EIF3B, HECW1, GLI3, HERPUD2, HOXA3, CARD11, GRB10, TNS3, SEPT7, TBX20, THSD7A, ELFN1, ETV1, TNRC18, IKZF1, HDAC9* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 7:65796279-159138663 | 7q | Loss | 93.34 | 0.24 | 1 | *RHEB, BRAF* | *AGAP3, CDK5, CUL1, EZH2, PSMC2, SLC4A2* | *ARF5, HGF, NAMPT, STX1A, CLIP2, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, NYAP1, TSC22D4, ACHE, GNB2, COPS6, CCDC132, YWHAG, MEPCE, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, PCLO, LIMK1, HIP1, BAZ1B, AUTS2, TRIM24, UBE3C, MKLN1, MAGI2, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, AGAP3, RBM33, PAXIP1, HIPK2, FOXP2, NCAPG2, ATXN7L1, KIAA1549, RELN, PSMC2, DOCK4, CBLL1, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 10:0-135534747 | 10 | Gain | 135.53 | 0.12 | 3 | *PTEN* | *LDB1, PPP3CB, TRIM8, ZSWIM8* | *PSD, TDRD1, KIF11, MAP3K8, EPC1, MTPAP, NRP1, HNRNPH3, CCAR1, BTAF1, CPEB3, SLIT1, ABLIM1, PPRC1, ARID5B, DOCK1, ANK3, DIP2C, C10orf12, NODAL, KAT6B, HECTD2, WAPAL, KIF5B, TRIM8, MLLT10, LARP4B, CAMK2G, SEPHS1, GRID1, FAM208B, PDZD8, ZMIZ1, PCGF5, PIK3AP1, SEC24C, TAF3, SORCS1, NKX2-3, HELLS, DDX21, WAC, ATRNL1, RET, SH3PXD2A, ADD3, UPF2, FRMD4A, ZRANB1, MCMBP, GTPBP4, LRRTM3, MGEA5, ZEB1, SGMS1, SMC3, SFMBT2, INPP5A, EBF3, TIAL1, EIF3A, CACUL1, HSPA12A, FAM160B1, SHOC2, SLK, TAF5, CNNM2, SUFU, ACTR1A, NFKB2, SCD, CHUK, LCOR, TM9SF3, LGI1, ZCCHC24, DLG5, ZNF503, H2AFY2, TET1, BICC1, IPMK, PRKG1, MAPK8, PARD3, FAM171A1, CAMK1D, PPP3CB, PSAP, HERC4, CDK1, RASGEF1A, ITGB1, ARHGAP21, ZMYND11, ZSWIM8, JMJD1C, HK1, KCNMA1, P4HA1, LDB1, TSPAN14, HNRNPF, RBM17, CELF2, FGFR2, CCNJ, KLF6, CUL2, TCF7L2, EMX2* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 12:0-133851895 | 12 | Gain | 133.85 | 0.12 | 8 | *KRAS, PTPN11* | *ANKRD52, ATP2B1, CHD4, CS, DDX23, GCN1L1, HECTD4, LRP1, NAA25, NAB2, PA2G4, PLXNC1, RARG, SCN8A, SETD1B, SMARCC2, SMARCD1, STAT2, USP5* | *SENP1, HDAC7, FOXJ2, TNFRSF1A, CBX5, SART3, ASIC1, CSRNP2, CNOT2, USP5, HCFC2, RAB35, LRP1, SLC38A2, MSI1, HNF1A, TBX3, GDF11, ESPL1, KCNH3, KCNMB4, PLXNC1, SLC41A2, WBP11, ATF7IP, NAP1L1, ASUN, CDK17, CCND2, LRP6, CORO1C, LPCAT3, CUX2, LHX5, NAA25, ATP5B, PTGES3, EIF4B, COPZ1, CLSTN3, SMARCC2, ANKRD52, SETD1B, DUSP6, USP15, BTBD11, EPS8, MED13L, NCKAP1L, ITGA5, FRS2, CCT2, TMEM132B, HSP90B1, STAT6, GCN1L1, KMT2D, TUBA1A, DIP2B, PA2G4, LEMD3, DHX37, TBX5, STAT2, WNK1, MLXIP, PITPNM2, ULK1, EEA1, E2F7, SP1, FBXO21, RSRC2, XPOT, ARID2, PTPRB, TUBA1B, SARNP, ZCCHC8, FAM60A, NOS1, KSR2, PPP1CC, HNRNPA1, NTN4, DYRK2, RNF41, R3HDM2, CACNA1C, CS, PTPN11, SPPL3, SCN8A, SRGAP1, GIT2, MBD6, ATN1, CHD4, DNAJC14, RFX4, PCBP2, SCAF11, KDM2B, ATXN2, ZFC3H1, NABP2, COL2A1, NOP2, PRMT8, STK38L, DENND5B, EP400, CIT, TAOK3, RIC8B, MON2, SMARCD1, ETV6, ZNF384, CCDC64, LARP4, SLC38A1, AEBP2, KDM5A, SETD8, RASSF8, NCOR2, ATF7, SBNO1, TMEM132D, RPL6, RARG, PAN2, ZDHHC17, ATP2B1, PLEKHA5, DCTN2, CPSF6, NELL2, PPP1R12A, SOX5, SLC4A8, KIF5A, PRICKLE1, PTPN6, MDM2, NAV3, ATP2A2, CLIP1, ACVR1B, SFSWAP, RAN, PHC1, CAND1, MAP3K12, AGAP2, ANKS1B, PRPF40B, PPFIA2, HECTD4, BAZ2A, SPATS2, GRIN2B* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 15:24148457-102531392 | 15q | Gain | 78.38 | 0.12 | 3 | | *RTF1, SIN3A* | *AQR, MGA, CSK, PIAS1, DLL4, SNRPA1, ABHD17C, IREB2, CTDSPL2, THBS1, KIF23, ADAM10, HERC2, AP3B2, SNX1, RAB11A, HCN4, TTBK2, IGF1R, ZNF710, FURIN, IQGAP1, UBR1, SPRED1, COPS2, LEO1, HDGFRP3, ZNF592, CLPX, MAP1A, PDIA3, PYGO1, CSNK1G1, ARNT2, OTUD7A, RPL4, FBXO22, RASGRP1, GABRB3, SEMA6D, FBN1, ZNF609, MORF4L1, ASB7, NEO1, DPP8, NPTN, BNC1, TJP1, CASC5, MEF2A, LINGO1, MYO9A, ZNF770, NTRK3, AKAP13, INO80, ARIH1, USP3, SLTM, TP53BP1, RYR3, RTF1, NR2F2, CHD2, SIN3A, UBE3A, MYO5A, BAHD1, FRMD5, RASGRF1, RFX7, USP8, ACAN, DENND4A, HERC1, ANP32A, DMXL2, TLE3, RNF111, MEIS2, TLN2, CPEB1, PARP6* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 16:0-90354753 | 16 | Gain | 90.35 | 0.12 | 3 | *NPRL3, TSC2* | *CNOT1, CTCF, EDC4, FBXL19, MAPK8IP3, MAZ, NUDT21, SF3B3, STX1B, UBE2I, USP7* | *CACNG3, TFAP4, GLG1, STX1B, C16orf70, CORO1A, NUTF2, USP10, TSC2, CAPN15, TOX3, MAPK8IP3, SALL1, FUS, VPS4A, DYNC1LI2, XYLT1, RBL2, PKD1, RAB11FIP3, CREBBP, FOXF1, GNAO1, CSNK2A2, SRCAP, SETD1A, ZNF629, CTCF, NKD1, ZFHX3, CDH11, ZCCHC14, IRF8, JPH3, ATP6V0D1, ADCY9, VPS35, ZNF319, PLK1, NUDT21, ZC3H18, ANKRD11, SF3B3, PRKCB, XPO6, TAOK2, RBFOX1, TUBB3, DNAJA2, CNOT1, RANBP10, MKL2, RBBP6, GLYR1, SH2B1, RNF40, C16orf72, IL21R, MTSS1L, PDPK1, CASKIN1, USP7, NFATC3, UBE2I, FBRS, GTF3C1, PLCG2, FBXL19, RPLI3, BANP, AP1G1, BRD7, ATXN2L, TNRC6A, MYH11, KIAA0430, ZC3H7A, GRIN2A, UBN1, FBXL16, TBC1D10B, CBFB, BCAR1, FAM65A, CYLD, NFAT5, GSPT1, PAPD5, HERPUD1, SMG1, CMIP, RSPRY1, ZNF423, RNPS1, CHD9, CDH8, TERF2* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 20:0-63025520 | 20 | Gain | 63.03 | 0.12 | 10 | | *CENPB, TOP1* | *SALL4, CSTF1, EEF1A2, CSNK2A1, CHMP4B, SOGA1, PLAGL2, TAF4, LAMA5, ADRM1, RBM39, JAG1, SNAP25, PCSK2, ITCH, RALGAPB, ATRN, CSE1L, GMEB2, DIDO1, LSM14B, NCOA5, PPP1R16B, TPX2, BCL2L1, MYT1, LZTS3, SS18L1, TCFL5, GGT7, PLCB1, EPB41L1, ATP9A, PMEPA1, E2F1, CBFA2T2, SYCP2, KCNQ2, C20orf112, TOP1, YTHDF1, MRGBP, PSMA7, PHACTR3, ZNF217, TSHZ2, B4GALT5, KCNB1, STAU1, ARFGEF2, PREX1, NCOA3, PCIF1, YWHAB, PTPRT, CHD6, SRC, DLGAP4, PHF20, NCOA6, EIF2S2, MAPRE1, POFUT1, PAK7, BMP2, PCNA, CENPB, PTPRA, SNPH, RGS19, RAE1, NFATC2, ADNP, ZNFX1, TM9SF4, SPATA2, ZNF512B, SLC12A5, ZMYND8* |
| LEAT_EEBB_56 | M | 11 | 15 | LEAT | PXA | | 21:15463956-48129895 | 21q | Gain | 32.67 | 0.14 | 1 | | | *HUNK, SIK1, USP25, CCT8, TIAM1, SCAF4, IFNGR2, PKNOX1, U2AF1, TRAPPC10, ZBTB21, PAXBP1, BRWD1, GABPA, SON, ETS2, COL6A1, SLC5A3, ITSN1, AGPAT3, DYRK1A, NRIP1, DSCAM, MORC3, ERG, SYNJ1* |
| LEAT_CCF_37 | M | 0.833 | 7.29 | LEAT | SEGA | TSC | 7:0-159138663 | 7 | Gain | 159.14 | 0.14 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | *HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1, KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| LEAT_CCF_37 | M | 0.833 | 7.29 | LEAT | SEGA | TSC | 16:0-9806375 | 16p13.3 | CNN-LOH | 9.81 | 0.03 | 4 | *NPRL3, TSC2* | *MAPK8IP3, UBE2I, USP7* | *TFAP4, TSC2, CAPN15, MAPK8IP3, PKD1, RAB11FIP3, CREBBP, ADCY9, RBFOX1, GLYR1, C16orf72, PDPK1, CASKIN1, USP7, UBE2I, UBN1, FBXL16, RNPS1* |
| LEAT_EEBB_17 | F | 0.076923077 | 9 | LEAT | SEGA | TSC | 16:0-16081737 | 16p13.3 | CNN-LOH | 16.08 | 0.09 | 4 | *NPRL3, TSC2* | *MAPK8IP3, UBE2I, USP7* | *TFAP4, TSC2, CAPN15, MAPK8IP3, PKD1, RAB11FIP3, CREBBP, ADCY9, RBFOX1, MKL2, GLYR1, C16orf72, PDPK1, CASKIN1, USP7, UBE2I, MYH11, KIAA0430, ZC3H7A, GRIN2A, UBN1, FBXL16, GSPT1, RNPS1* |
| MCD_CCF_62 | F | 5 | 8.33 | MCD | FCD 2a | Hyaline astrocytic inclusions | 1:146523045-249250621 | 1q | Gain | 102.73 | 0.06 | 2 | *AKT3* | *AKT3, ARF1, CACNA1E, CELF3, CLK2, COPA, DENND4B, DHX9, ILF2, INTS3, LMNA, MEF2D, NDUFS2, PEA15, PRPF3, RBBP5, SF3B4, TOMM40L, TRIM46, UBE2Q1, USP21, WDR26* | *CHRM3, TAF5L, LAMC1, SUCO, ARID4B, RBBP5, ILDR2, PRCC, SF3B4, SETDB1, POGZ, KCNN3, HNRNPU, WNT3A, NMNAT2, UBE2Q1, ZNF281, ZNF496, NCSTN, SPRTN, CCT3, RGL1, SHE, GLUL, RORC, INTS3, FMN2, ZNF687, PRPF3, AHCTF1, LIN9, TRIM46, PRRC2C, NFASC, LMX1A, MEF2D, ARNT, ZBTB18, CAMSAP2, RAB3GAP2, KIRREL, ELF3, EXOC8, CLK2, DENND4B, ARHGEF2, BRINP2, IPO9, HORMAD1, ASTN1, ILF2, AKT3, RYR2, ACTN2, HEATR1, SIPA1L2, EGLN1, TRIM67, CDC42BPA, ENAH, FBXO28, SLC30A10, TGFB2, PROX1, LPGAT1, SLC30A1, IRF6, MAPKAPK2, NUCKS1, MDM4, PIK3C2B, SOX13, SYT2, NAV1, LHX9, CFH, CDC73, PTGS2, TPR, IVNS1ABP, SWT1, RNF2, DHX9, CACNA1E, XPR1, CEP350, RASAL2, RFWD2, TNR, RC3H1, SERPINC1, ZBTB37, ATP1B1, TIPRL, POU2F1, DEDD, ARHGAP30, COPA, DCAF8, ETV3, ARHGEF11, LMNA, UBQLN4, KIAA0907, RUSC1, PYGO2, CRTC2, GATAD2B, SNX27, PI4KB, PSMD4, MCL1, RPRD2, LYST, ASH1L, KIF26B, WDR26, RCOR3, PBX1, SYT14, C1orf226, UBAP2L, ITPKB, GON4L, PTPRC, TDRD5, SMG7, MEX3A, ZC3H11A* |
| MCD_CCF_62 | F | 5 | 8.33 | MCD | FCD 2a | Hyaline astrocytic inclusions | 16:47304567-90354753 | 16q12.1-24.3 | Loss | 43.05 | 0.06 | 1 | | *CNOT1, CTCF, EDC4, NUDT21, SF3B3* | *GLG1, C16orf70, NUTF2, USP10, TOX3, SALL1, VPS4A, DYNC1LI2, RBL2, FOXF1, GNAO1, CSNK2A2, CTCF, NKD1, ZFHX3, CDH11, ZCCHC14, IRF8, JPH3, ATP6V0D1, ZNF319, NUDT21, ZC3H18, ANKRD11, SF3B3, TUBB3, CNOT1, RANBP10, MTSS1L, NFATC3, PLCG2, RPL13, BANP, AP1G1, BRD7, CBFB, BCAR1, FAM65A, CYLD, NFAT5, PAPD5, HERPUD1, CMIP, RSPRY1, ZNF423, CHD9, CDH8, TERF2* |
| MCD_EEBB_68 | M | 6 | 43 | MCD | FCD 2a | | 22:18009909-49085205 | 22q | CNN-LOH | 31.08 | 0.04 | 5 | *DEPDC5* | *CSNK1E, PATZ1, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, GNAZ, RAC2, CELSR1, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, SCUBE1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA1I, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, MICAL3, HIC2, TNRC6B, ZNRF3* |
| MCD_EEBB_75 | M | 2.8 | 12 | MCD | FCD 2a | | 22:19183787-51304566 | 22q | CNN-LOH | 32.12 | 0.04 | 5 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, GNAZ, RAC2, CELSR1, SHANK3, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA1I, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, HIC2, PLXNB2, TNRC6B, ZNRF3* |
| MCD_EEBB_94 | F | 0.25 | 3 | MCD | FCD 2a | | 22:16000000-51304566 | 22q | CNN-LOH | 35.30 | 0.04 | 5 | *DEPDC5* | *CSNK1E, PATZ1, PLXNB2, SF3A1* | *RTN4R, SF3A1, MAPK1, MORC2, RPL3, MYH9, EIF3D, RBX1, BRD1, GNAZ, RAC2, CELSR1, SHANK3, SMARCB1, UFD1L, MED15, HIRA, EP300, PATZ1, KCNJ4, BCR, PHF21B, SPECC1L, PRR14L, FBLN1, MAPK8IP2, SULT4A1, PITPNB, NF2, GGA1, DGCR8, ZC3H7B, LARGE, MKL1, AP1B1, XRCC6, TCF20, PIM3, SCUBE1, SBF1, DEPDC5, CSNK1E, SOX10, EIF4ENIF1, MTMR3, CACNA1I, ELFN2, GRAMD4, CBX6, EWSR1, RBFOX2, MICAL3, HIC2, PLXNB2, TNRC6B, ZNRF3* |
| MCD_EEBB_110 | M | 0.5 | 7.5 | MCD | FCD 2a | Hyaline astrocytic inclusions | 1:145723739-249250621 | 1q | Gain | 103.53 | 0.08 | 2 | *AKT3* | *AKT3, ARF1, CACNA1E, CELF3, CLK2, COPA, DENND4B, DHX9, ILF2, INTS3, LMNA, MEF2D, NDUFS2, PEA15, PRPF3, RBBP5, SF3B4, TOMM40L, TRIM46, UBE2Q1, USP21, WDR26* | *CHRM3, TAF5L, LAMC1, SUCO, ARID4B, RBBP5, ILDR2, PRCC, SF3B4, SETDB1, POGZ, KCNN3, HNRNPU, WNT3A, NMNAT2, UBE2Q1, ZNF281, ZNF496, NCSTN, SPRTN, CCT3, RGL1, SHE, GLUL, RORC, INTS3, FMN2, ZNF687, PRPF3, AHCTF1, LIN9, TRIM46, PRRC2C, NFASC, LMX1A, MEF2D, ARNT, ZBTB18, CAMSAP2, RAB3GAP2, KIRREL, ELF3, EXOC8, CLK2, DENND4B, ARHGEF2, BRINP2, IPO9, HORMAD1, ASTN1, ILF2, AKT3, RYR2, ACTN2, HEATR1, SIPA1L2, EGLN1, TRIM67, CDC42BPA, ENAH, FBXO28, SLC30A10, TGFB2, PROX1, LPGAT1, SLC30A1, IRF6, MAPKAPK2, NUCKS1, MDM4, PIK3C2B, SOX13, SYT2, NAV1, LHX9, CFH, CDC73, PTGS2, TPR, IVNS1ABP, SWT1, RNF2, DHX9, CACNA1E, XPR1, CEP350, RASAL2, RFWD2, TNR, RC3H1, SERPINC1, ZBTB37, ATP1B1, TIPRL, POU2F1, DEDD, ARHGAP30, COPA, DCAF8, ETV3, ARHGEF11, LMNA, UBQLN4, KIAA0907, RUSC1, PYGO2, CRTC2, GATAD2B, SNX27, PI4KB, PSMD4, MCL1, RPRD2, LYST, ASH1L, KIF26B, WDR26, RCOR3, PBX1, SYT14, C1orf226, UBAP2L, ITPKB, GON4L, PTPRC, TDRD5, SMG7, MEX3A, ZC3H11A* |
| MCD_CCF_48 | F | 7 | 26.97 | MCD | FCD 2b | | 9:71034203-141213431 | 9q21.11-34.3 | CNN-LOH | 70.18 | 0.09 | 1 | *TSC1* | *ABCA2, DNM1, GABBR2, GRIN1, NOTCH1, PHF19, SCAI, SET, SPTAN1, STXBP1, WDR5* | *SETX, GDA, TRAF2, OLFM1, LRRC8A, KIAA0368, RALGPS1, DAB2IP, GABBR2, ERP44, BRINP1, MAPKAP1, FAM120A, ZNF462, NOTCH1, SMC2, GNAQ, ZNF618, ZER1, ALDH1A1, WNK2, TSC1, ZMYND19, BRD3, ZNF483, SEC16A, RPL7A, ANAPC2, PTCH1, ANP32B, ABCA2, COL27A1, BICD2, PRRC2B, RAD23B, WDR5, PHF2, NUP214, ASTN2, NAA35, CACNA1B, GRIN1, RABL6, COL5A1, RALGDS, RAPGEF1, ABL1, NUP188, SET, SPTAN1, DNM1, ENG, STXBP1, ZBTB34, ZBTB43, SCAI, NR5A1, LHX2, DENND1A, RABGAP1, RC3H2, RAB14, PHF19, MEGF9, PRPF4, UGCG, EPB41L4B, KLF4, SLC44A1, TEX10, NCBP1, CDC14B, SYK, SPIN1, ZCCHC6, NTRK2, HNRNPK, UBQLN1, TLE1, TLE4, RORB, CAMSAP1, NTNG2, PPP2R4, GAPVD1, LHX6, SVEP1, DAPK1, GOLGA2, STRBP, SEMA4D, EHMT1, RXRA, NR6A1, FBXW2* |
| MCD_CCF_48 | F | 7 | 26.97 | MCD | FCD 2b | | 16:0-3367061 | 16p13.3 | CNN-LOH | 3.37 | 0.05 | 4 | *NPRL3, TSC2* | *MAPK8IP3, UBE2I* | *TSC2, CAPN15, MAPK8IP3, PKD1, RAB11FIP3, PDPK1, CASKIN1, UBE2I, FBXL16, RNPS1* |
| MCD_CCF_90 | F | 0.67 | 2.92 | MCD | FCD 2b | | 7:0-159138663 | 7 | Gain | 159.14 | 0.05 | 18 | *RHEB, BRAF* | *AGAP3, CAMK2B, CDK5, CUL1, EZH2, OGDH, PSMC2, RAC1, SLC4A2* | *ARF5, RALA, HOXA11, CDK13, HGF, NAMPT, ITGB8, SP4, OGDH, STX1A, PSMA2, CLIP2, AHR, INHBA, ZNF777, PTPN12, LRRC4, MKRN1, ACTR3B, IGF2BP3, TTYH3, KCNH2, RHEB, KMT2C, SEMA3A, ST7, PRKAR2B, DBF4, CDK6, ANKIB1, GNA12, EGFR, CCT6A, NUP205, PRKAG2, DGKI, BRAF, C7orf60, SHH, COL1A2, LMTK2, ADCY1, NYAP1, TSC22D4, ACHE, GNB2, COPS6, KBTBD2, CCDC132, USP42, YWHAG, ZMIZ2, MEPCE, ELMO1, KMT2E, ING3, MET, EZH2, ZKSCAN1, GTF2I, AHCYL2, CUL1, FLNC, ACTB, PCLO, LIMK1, HIP1, BAZ1B, VWC2, AUTS2, TRIM24, RAPGEF5, C7orf26, UBE3C, MKLN1, MAGI2, HNRNPA2B1, SND1, CACNA2D1, PLXNA4, TRRAP, CUX1, EIF3B, CAPZA2, CALD1, DNAJC2, NRF1, ZNF800, PTPRZ1, SRPK2, DMTF1, HECW1, GLI3, HERPUD2, HOXA3, CARD11, AGAP3, GRB10, TNS3, SEPT7, RBM33, PAXIP1, HIPK2, TBX20, FOXP2, NCAPG2, ATXN7L1,* |

| Sample | Sex | Age at Onset | Age at Surgery | Lesion Group | Pathology | Additional Findings | Variant Position (hg19) | Chromosomal Region | Variant Type | CNV size (Mbp) | CNV Allelic Fraction | Samples with CNV | Included LFE Genes | Included Triplosensitive Genes | Included Haploinsufficient Genes |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | *KIAA1549, THSD7A, ELFN1, RELN, ETV1, TNRC18, PSMC2, DOCK4, IKZF1, CBLL1, HDAC9, FEZF1, FAM131B, CADPS2, GTF2IRD1, UBN2, SLC4A2, LHFPL3, CNOT4* |
| MCD_EEBB_104 | F | NA | 15 | MCD | FCD 2b | | 4:93918343-191154276 | 4q22.2-35.2 | CNN-LOH | 97.24 | 0.02 | 1 | | *ABCE1, PPP3CA, SMARCA5* | *NFKB1, RAPGEF2, FNIP2, CENPE, LEF1, SEC24B, CCNA2, NAF1, RPS3A, GPM6A, FBXW7, GRID2, SMARCA5, HMGB2, GRIA2, NAA15, HHIP, ABCE1, DCLK2, PITX2, EDNRA, TRIM2, RAP1GDS1, CLCN3, ANK2, NR3C2, SMARCAD1, ZNF827, ELF2, FAT4, ELOVL6, PPP3CA, CPE, KIAA0922, BMPR1B, OTUD4, PLRG1, PDGFC, ANKRD50, CDKN2AIP, MAML3, TENM3, KLHL2, SMAD1* |
| MCD_CCF_71 | F | 0.83 | 1.82 | MCD | cMCD | NH | 1:203644628-234525940 | 1q32.1-42.2 | Gain | 30.88 | 0.06 | 1 | | *ARF1, RBBP5, WDR26* | *TAF5L, RBBP5, WNT3A, SPRTN, LIN9, NFASC, RAB3GAP2, EXOC8, SIPA1L2, EGLN1, TRIM67, CDC42BPA, ENAH, FBXO28, SLC30A10, TGFB2, PROX1, LPGAT1, SLC30A1, IRF6, MAPKAPK2, NUCKS1, MDM4, PIK3C2B, SOX13, WDR26, RCOR3, SYT14, ITPKB, ZC3H11A* |
| MCD_CCF_94 | F | 0.0082 | 0.37 | MCD | cMCD | PMG / FCD 2a / FCD 1a | 10:75324903-135534747 | 10q22.2-26.3 | CNN-LOH | 60.21 | 0.15 | 1 | *PTEN* | *LDB1, TRIM8, ZSWIM8* | *PSD, TDRD1, KIF11, BTAF1, CPEB3, SLIT1, ABLIM1, PPRC1, DOCK1, C10orf12, KAT6B, HECTD2, WAPAL, TRIM8, CAMK2G, GRID1, PDZD8, ZMIZ1, PCGF5, PIK3AP1, SEC24C, SORCS1, NKX2-3, HELLS, ATRNL1, SH3PXD2A, ADD3, ZRANB1, MCMBP, MGEA5, SMC3, INPP5A, EBF3, TIAL1, EIF3A, CACUL1, HSPA12A, FAM160B1, SHOC2, SLK, TAF5, CNNM2, SUFU, ACTR1A, NFKB2, SCD, CHUK, LCOR, TM9SF3, LGI1, ZCCHC24, DLG5, ZNF503, ZSWIM8, KCNMA1, LDB1, TSPAN14, FGFR2, CCNJ, TCF7L2, EMX2* |
| MCD_CCF_104 | F | 0 | 0.12 | MCD | cMCD | FCD 2b | 16:0-12353187 | 16p13.3 | CNN-LOH | 12.35 | 0.04 | 4 | *NPRL3, TSC2* | *MAPK8IP3, UBE2I, USP7* | *TFAP4, TSC2, CAPN15, MAPK8IP3, PKD1, RAB11FIP3, CREBBP, ADCY9, RBFOX1, GLYR1, C16orf72, PDPK1, CASKIN1, USP7, UBE2I, ZC3H7A, GRIN2A, UBN1, FBXL16, GSPT1, RNPS1* |
| MCD_EEBB_89 | M | 7 | 20 | MCD | cMCD | FCD 1b | 17:28305017-81195210 | 17q | CNN-LOH | 52.89 | 0.06 | 2 | *NF1* | *ACACA, CACNA1G, CLTC, COL1A1, EFTUD2, EZH1, HDAC5, KAT2A, KPNB1, MBTD1, MTMR4, NF1, PIP4K2B, STAT3, STAT5A, STAT5B, THRA, UBTF, UNK* | *XYLT2, APPBP2, TBX21, PSMB3, RPL19, RPS6KB1, DDX5, ASIC2, HNF1B, COL1A1, HDAC5, HLF, TBX2, SOX9, NUP85, OMG, RARA, SRSF1, KAT7, PSMD11, SPAG9, VMP1, SLC4A1, KANSL1, SMURF2, PSMD3, ATP6V0A1, STAT3, FBXL20, SRCIN1, KCNH4, CLTC, EIF4A3, CBX8, CBX4, PIP4K2B, MSI2, KPNB1, IGF2BP1, KLHL10, STAT5B, PSME3, MED1, USP32, NEUROD2, UBTF, MRC2, GGNBP2, BPTF, FASN, RPTOR, RP11-1055B8.7, PPM1E, CBX2, YPEL2, AP2B1, CSNK1D, PPP1R9B, LASP1, UBE2O, CASKIN2, ATAD5, NPEPPS, SUZ12, FOXJ1, MTMR4, HSF5, CHMP6, MLLT6, RAB11FIP4, NPLOC4, P4HB, FMNL1, HEXIM1, TNRC6C, SYNRG, STAT5A, TLK2, IKZF3, SMARCE1, ACACA, PSMD12, NF1, RHOT1, HELZ, CACNA1G, CYTH1, MAP3K3, BECN1, NFE2L1, SP2, GPS1, SPOP, CBX1, CNP, MED13, C17orf104, DCAF7, TOP2A, TANC2, EFTUD2, GJC1, SEPT9, ERN1, INTS2, CDK12, ATXN7L3, DNAJC7, TOB1, LUC7L3, CDC27, SOCS7, DDX42, MSL1, VEZF1, PITPNC1, MBTD1, PRKAR1A, UNK, LSM12, GPATCH8, NMT1* |

**Legend to supplementary table 5:** LFE = lesional focal epilepsy, CNV = Copy number variant, LEAT = Low-grade epilepsy-associated tumor, MCD = Malformation of cortical development, HS = Hippocampal sclerosis, AG = Angiocentric Glioma, DNET = Dysembryoplastic neuroepithelial tumor, GG = Ganglioglioma, DGG = Desmoplastic ganglioglioma, IDA = Isomorphic diffuse astrocytoma, PA = Pilocytic astrocytoma, PXA = Pleomorphic xanthoastrocytoma, MVNT = multinodular vacuolated neurol tumor, SEGA = Subependymal giant cell astrocytoma, TSC = Tuberous sclerosis complex, FCD = Focal cortical dysplasia, HME = Hemimegalencephaly, cMCD = complex MCD, PMG = Polymicrogyria, CNN-LOH = Copy number neutral loss of heterozygosity

**Supplementary Table 6: Summary of *PTPN11* variant pathogenicity evaluation according to the ACMG guidelines**

| Sample | Lesion Group | Pathology | *PTPN11* Variant | ACMG Strong Evidence | ACMG Moderate Evidence | ACMG Supporting Evidence | ACMG Variant Classification |
|---|---|---|---|---|---|---|---|
| HS_EEBB_41 | HS | HS Type 1 | p.S502L | PS1 | PM2 | PP2, PP3 | Likely Pathogenic |
| LEAT_EEBB_10 | LEAT | Ganglioglioma | p.E76K | PS1, PS4 | PM1, PM2 | PP2, PP3 | Pathogenic |
| LEAT_EEBB_15 | LEAT | Ganglioglioma | p.D61N | PS1, PS4 | PM2 | PP2, PP3 | Pathogenic |
| | | | p.E139D | PS1, PS4 | PM2 | PP2, PP3 | Pathogenic |
| LEAT_CCF_6 | LEAT | DNET | p.T507K | PS1, PS4 | PM2 | PP2, PP3 | Pathogenic |
| LEAT_EEBB_49 | LEAT | DNET | p.A72V | PS4 | PM1, PM2, PM5 | PP2, PP3 | Pathogenic |
| LEAT_EEBB_81 | LEAT | DNET | p.A72G | PS1, PS4 | PM1, PM2 | PP2, PP3 | Pathogenic |
| LEAT_EEBB_17 | LEAT | TSC (SEGA) | p.D61H | PS1, PS4 | PM2 | PP2, PP3 | Pathogenic |

**Legend to Supplementary Table 6**: ACMG = American College of Medical Genetics and Genomics, HS = Hippocampal sclerosis, LEAT = Low-grade epilepsy-associated neuroepithelial tumor, DNET = Dysembryoplastic neuroepithelial tumor, TSC = Tuberous sclerosis complex, SEGA = Subependymal giant cell astrocytoma, PS1 = Same amino acid change as an established pathogenic variant, PS4 = statistically significant association with pathology, PM1 = Mutational hot sport or well-studied functional domain, PM2 = Absent in population databases, PM5 = Novel missense change at an amino acid residue where a different pathogenic missense change has been seen before, PP2 = Missense in gene with low rate of benign missense variants, PP3 = Computational evidence supports a deleterious effect on the gene / gene product