Patient Data

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| --- | --- |
| Name |  |
| Birthdate |  |
| Diagnosis |  |

Somatic Mutations in Known Driver Genes

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| --- | --- | --- | --- |
| **Gene** | **Mutation** | **Confidence** | **References** |
| BRAF | p.Val600Glu | 4 | 1,2,3,4 |
| ETS2 | p.Ala64Thr | 2 | 3,5 |
| ETS2 | p.Gly216Ser | 2 | 3,5 |
| ETS2 | p.Val249Ile | 2 | 3,5 |
| PDGFB | p.Gln145Arg | 2 | 1,3 |
| PDGFB | p.Ile88Val | 2 | 1,3 |
| HIRA | p.Ser425Thr | 1 | 5 |
| SUSD2 | p.Arg165Gly | 1 | 3 |
| SUSD2 | p.Arg499Cys | 1 | 3 |
| SUSD2 | p.Arg499His | 1 | 3 |
| SUSD2 | p.Asn436Ser | 1 | 3 |
| SUSD2 | p.Asn466Ser | 1 | 3 |
| SUSD2 | p.Ser110Thr | 1 | 3 |
| SUSD2 | p.Val633Met | 1 | 3 |

Direct Association (Mutation in drug target)

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Gene** | **Mutation** | **Therapy** | **Confidence** | **References** |
| BRAF | p.Val600Glu | sorafenib | 9 | 6,7,8,9,10,11,12,13,14 |
| BRAF | p.Val600Glu | dabrafenib | 4 | 12,14,15 |
| BRAF | p.Val600Glu | vemurafenib | 4 | 12,14,16,17 |
| BRAF | p.Val600Glu | regorafenib | 3 | 12,18 |
| BRAF | p.Val600Glu | gsk2118436 | 1 | 14 |
| BRAF | p.Val600Glu | r7204 | 1 | 14 |

Indirect Association (other Mutations with known effect on drug)

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Gene** | **Mutation** | **Therapy** | **Disease** | **Evidence** | **References** |
| BRAF | AGK-BRAF | Sorafenib | Melanoma | C | 27 |
| BRAF | AGK-BRAF | Vemurafenib | Melanoma | D | 27 |
| BRAF | L597R | Vemurafenib | Melanoma | C | 28 |
| BRAF | MUTATION | Trametinib | Cancer | D | 37 |
| BRAF | PAPSS1-BRAF | Trametinib | Melanoma | D | 36 |
| BRAF | PAPSS1-BRAF | Vemurafenib | Melanoma | D | 36 |
| BRAF | TRIM24-BRAF | Trametinib | Melanoma | D | 36 |
| BRAF | V600 | BAY 86-9766 | Melanoma | C | 30 |
| BRAF | V600 | Dabrafenib,Trametinib | Melanoma | B | 20 |
| BRAF | V600 | Panitumumab | Colorectal Cancer | B | 19 |
| BRAF | V600 | RO4987655 | Melanoma | B | 24 |
| BRAF | V600D | Dabrafenib | Melanoma | B | 21 |
| BRAF | V600E | BEZ235 (NVP-BEZ235, Dactolisib),Selumetinib (AZD6244) | Melanoma | D | 38 |
| BRAF | V600E | Capecitabine,Vemurafenib,Bevacizumab | Colorectal Cancer | D | 35 |
| BRAF | V600E | Cetuximab | Colorectal Cancer | B | 23 |
| BRAF | V600E | Cobimetinib | Cancer | D | 39 |
| BRAF | V600E | Dabrafenib | Non-small Cell Lung Carcinoma | C | 26 |
| BRAF | V600E | Dabrafenib,Trametinib | Melanoma | B | 22 |
| BRAF | V600E | Pictilisib | Melanoma | C | 29 |
| BRAF | V600E | PLX4720,GDC0941 | Colorectal Cancer | D | 33 |
| BRAF | V600E | PLX4720,Nutlin-3 | Colorectal Cancer | D | 34 |
| BRAF | V600E | Sorafenib,Panitumumab | Colorectal Cancer | D | 31 |
| BRAF | V600E | Vemurafenib | Colorectal Cancer | D | 35 |
| BRAF | V600E | Vemurafenib | Melanoma | D | 32 |
| BRAF | V600E AMPLIFICATION | Selumetinib (AZD6244) | Colorectal Cancer | E | 40 |
| BRAF | V600E+V600M | Dabrafenib | Melanoma | C | 25 |

Somatic Mutations with known pharmacogenetic effect

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Gene** | **Mutation** | **Therapy** | **Disease** | **Evidence** | **References** |
| BRAF | V600E | Cetuximab | Colorectal Cancer | B | 23 |
| BRAF | V600E | Dabrafenib,Trametinib | Melanoma | B | 22 |
| BRAF | V600E | Dabrafenib | Non-small Cell Lung Carcinoma | C | 26 |
| BRAF | V600E | Pictilisib | Melanoma | C | 29 |
| BRAF | V600E | BEZ235 (NVP-BEZ235, Dactolisib),Selumetinib (AZD6244) | Melanoma | D | 38 |
| BRAF | V600E | Capecitabine,Vemurafenib,Bevacizumab | Colorectal Cancer | D | 35 |
| BRAF | V600E | Cobimetinib | Cancer | D | 39 |
| BRAF | V600E | PLX4720,GDC0941 | Colorectal Cancer | D | 33 |
| BRAF | V600E | PLX4720,Nutlin-3 | Colorectal Cancer | D | 34 |
| BRAF | V600E | Sorafenib,Panitumumab | Colorectal Cancer | D | 31 |
| BRAF | V600E | Vemurafenib | Colorectal Cancer | D | 35 |
| BRAF | V600E | Vemurafenib | Melanoma | D | 32 |

Appendix

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| --- | --- | --- | --- |
| **Gene** | **Mutation** | **COSMIC** | **dbSNP** |
| BRAF | p.Val600Glu | COSM18443,COSM476,COSM6137 | rs113488022 |
| MRPL39 | p.Thr334Met |  | rs116645811 |
| MRPL39 | p.Asn84Ser |  | rs75377686 |
| MRPL39 | p.Ser31Pro |  | rs3989369 |
| SYNJ1 | p.Met1020Ile | COSM3799865,COSM3799866 | rs115683257 |
| SYNJ1 | p.Ile905Thr |  | rs114053718 |
| SYNJ1 | p.Ala383Thr |  | rs114942253 |
| SYNJ1 | p.Lys334Arg |  | rs2254562 |
| IFNGR2 | p.Thr58Arg | COSM3693766 | rs4986958 |
| IFNGR2 | p.Gln64Arg |  | rs9808753 |
| IFNGR2 | p.Lys182Glu |  | rs17878711 |
| PIGP | p.Tyr118Cys |  | rs16994704 |
| PIGP | p.Ile90Val |  | rs73901833 |
| PIGP | p.Ala9Thr |  | rs2507733 |
| PIGP | p.Pro3Ser |  | rs73200245 |
| ETS2 | p.Ala64Thr |  | rs34373350 |
| ETS2 | p.Gly216Ser | COSM94254 | rs115908228 |
| ETS2 | p.Val249Ile |  | rs116698978 |
| CECR1 | p.His335Arg |  | rs2231495 |
| HIRA | p.Ser425Thr |  | rs115157927 |
| ARVCF | p.Ala805Val |  | rs115736959 |
| ARVCF | p.Val480Ile |  | rs76496156 |
| ARVCF | p.Arg233Trp |  | rs116398106 |
| ARVCF | p.Pro220Leu |  | rs2073748 |
| ARVCF | p.Arg196Gln |  | rs80068543 |
| ARVCF | p.Val175Ala |  | rs2240717 |
| ARVCF | p.Arg34Trp | COSM3693942 | rs116782322 |
| SDF2L1 | p.Asp141Asn |  | rs61739341 |
| SDF2L1 | p.Arg161His |  | rs73166641 |
| TOP3B | p.Leu439Pro |  | rs114712273 |
| TOP3B | p.Asp365Asn |  | rs9610728 |
| RAB36 | p.Ala85Val |  | rs61747143 |
| RAB36 | p.Asn308Asp |  | rs5759612 |
| SUSD2 | p.Ser110Thr | COSM1180456 | rs9680526 |
| SUSD2 | p.Arg165Gly |  | rs115180746 |
| SUSD2 | p.Asn436Ser |  | rs116768581 |
| SUSD2 | p.Asn466Ser |  | rs8141797 |
| SUSD2 | p.Arg499Cys |  | rs115684319 |
| SUSD2 | p.Arg499His |  | rs114116736 |
| SUSD2 | p.Val633Met |  | rs116712499 |
| HSCB | p.Ile163Met |  | rs17884212 |
| SEC14L3 | p.Arg364Cys |  | rs35764129 |
| SEC14L3 | p.Asp335Glu |  | rs2240345 |
| SEC14L3 | p.Val270Leu |  | rs114917409 |
| SEC14L3 | p.Ile232Ser |  | rs115111929 |
| SEC14L3 | p.Arg214His |  | rs2269961 |
| SEC14L3 | p.Arg130His |  | rs115812777 |
| SEC14L3 | p.Ile103Thr | COSM4002142,COSM4002143 | rs4820853 |
| SEC14L3 | p.Arg45Trp |  | rs115083767 |
| SEC14L3 | p.Arg40Cys |  | rs116155456 |
| GAL3ST1 | p.Arg329His |  | rs112976399 |
| GAL3ST1 | p.Arg244Gly |  | rs114565111 |
| GAL3ST1 | p.Val29Met |  | rs2267161 |
| SMTN | p.Pro237Leu |  | rs113930388 |
| SMTN | p.Ala494Val |  | rs11913728 |
| SMTN | p.Arg511His |  | rs116593437 |
| SMTN | p.Ser531Asn |  | rs114015937 |
| SMTN | p.Arg538Gln |  | rs115143483 |
| SMTN | p.Ala547Pro |  | rs3205187 |
| SMTN | p.Ala559Val | COSM4417233,COSM4417234,COSM4417235 | rs5997872 |
| SMTN | p.Arg642Cys |  | rs34292278 |
| LIMK2 | NA |  | rs79923828 |
| LARGE | p.Arg650Gln |  | rs73399520 |
| LARGE | p.Gln48Lys |  | rs115801300 |
| JOSD1 | p.Arg53Trp |  | rs114289284 |
| JOSD1 | p.Ser48Arg |  | rs6001200 |
| PDGFB | p.Gln145Arg |  | rs116555717 |
| PDGFB | p.Ile88Val |  | rs17565 |
| ENTHD1 | p.Ile548Thr |  | rs114948690 |
| ENTHD1 | p.Leu427Ser |  | rs115673469 |
| ENTHD1 | p.Met422Thr |  | rs115302874 |
| ENTHD1 | p.Arg292Thr | COSM3964335 | rs111738363 |
| ENTHD1 | p.Leu239Pro |  | rs115492335 |
| ENTHD1 | p.Leu205Phe |  | rs114100360 |
| FAM83F | p.Thr214Ile |  | rs115365592 |
| FAM83F | p.Arg245Ser |  | rs12330063 |
| FAM83F | p.Asp252Gly |  | rs116438980 |
| FAM83F | p.Ala427Val |  | rs115818503 |
| FAM83F | p.Arg436Gly |  | rs5995794 |
| PHF21B | p.Tyr328Cys |  | rs115264708 |
| PHF21B | p.Gly127Ser |  | rs8135982 |
| PHF21B | p.Ala41%3D |  | rs114043304 |

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