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| Molecular Tumor Report |

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| Case |

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| **Patient** | Doe | Jane |
| **DOB** |  |  |
| **Diagnosis** |  | HCC |

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| --- | --- | --- | --- |
| Mutational Burden | |  |  | | --- | --- | |  |  | |
| Anzahl betroffener Onkogene | 2 |
| Anzahl betroffener Tumorsupressorgene | 4 |
| HLA Typ |  |
| Additional information |  |

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| Somatic Mutations in Driver Genes |

| **Gene** | **Mutation** | **Roles** |
| --- | --- | --- |
| FAM46C | SNV c.626C>A p.Thr209Asn missense | No class |
| SF3B1 | SNV c.2153C>T p.Pro718Leu missense | Activating |
| TNPO1 | SNV c.114G>T p.Gln38His missense | No class |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Activating |
| PCSK5 | SNV c.2240G>A p.Cys747Tyr missense | No class |
| PABPC3 | SNV c.700G>A p.Gly234Arg missense | No class |

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| Somatic Mutations in Pharmaceutical Target Proteins |

**Direct Association (Mutation in drug target)**

| **Gene** | **Mutation** | **Drugs** | **Pubmed** |
| --- | --- | --- | --- |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Sorafenib | Vemurafenib | Regorafenib | Dabrafenib | 16609060 | 16901402 | 17575107 | 16880785 | 20812347 | 11752352 | 23094782 | NA | 23621583 | 15466206 | 22808911 | 22222036 |

**Indirect Associations (other Mutations with known effect on drug )**

| **Gene** | **Mutation** | **Drugs** | **Pubmed** |
| --- | --- | --- | --- |
| EPHB4 | SNV c.1037C>T p.Pro346Leu missense | Bevacizumab | 23579861 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Dabrafenib | Temozolomide | Dacarbazine | PD0325901 | Trametinib | Panitumumab | Sorafenib | Vemurafenib | Selumetinib (AZD6244) | PLX4720 | Nutlin-3 | Bevacizumab | Capecitabine | Cetuximab | Pictilisib | RO4987655 | BAY 86-9766 | BEZ235 (NVP-BEZ235 | Dactolisib) | Cobimetinib | 23031422 | 24586605 | 24576830 | 23325582 | 19001320 | 23524406 | 21098728 | 23020132 | 23463675 | 24583796 | 23845441 | 23812671 | 22180495 | 20619739 | 23890088 | 24345920 | 23715574 | 25370471 | 25989278 | 22169769 | 24947927 | 23434733 | 26678033 | 23934108 |

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| Somatic Mutations with known pharmacogenetic effect |

| **Gene** | **Mutation** | **Drugs** | **Disease** | **Biomarker** | **Effect** | **Evidence** | **Pubmed** |
| --- | --- | --- | --- | --- | --- | --- | --- |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Temozolomide,Dacarbazine | Melanoma | Predictive | N/A | B | 24586605 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Trametinib,Dabrafenib | Melanoma | Predictive | Sensitivity | B | 24583796 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Cetuximab | Colorectal Cancer | Predictive | Resistance or Non-Response | B | 20619739 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Panitumumab,Cetuximab | Colorectal Cancer | Predictive | Resistance or Non-Response | B | 25989278 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Dabrafenib | Non-small Cell Lung Carcinoma | Predictive | Resistance or Non-Response | C | 23524406 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Pictilisib | Melanoma | Predictive | Sensitivity | C | 25370471 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | PD0325901,Trametinib | Melanoma | Predictive | Resistance or Non-Response | D | 24576830 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Sorafenib | Colorectal Cancer | Predictive | Resistance or Non-Response | D | 19001320 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Vemurafenib | Melanoma | Predictive | Resistance or Non-Response | D | 24576830 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | PLX4720,PD0325901 | Colorectal Cancer | Predictive | Sensitivity | D | 23845441 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Nutlin-3,PLX4720 | Colorectal Cancer | Predictive | Sensitivity | D | 23812671 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Bevacizumab,Vemurafenib,Capecitabine | Colorectal Cancer | Predictive | Sensitivity | D | 22180495 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Vemurafenib | Colorectal Cancer | Predictive | Sensitivity | D | 22180495 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | BEZ235 (NVP-BEZ235, Dactolisib),Selumetinib (AZD6244) | Melanoma | Predictive | Sensitivity | D | 26678033 |
| BRAF | SNV c.1799T>A p.Val600Glu missense | Cobimetinib | Cancer | Predictive | Sensitivity | D | 23934108 |

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| Disclaimer |

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