**Wissenschaftliches Gutachten**

|  |
| --- |
| **Patientendaten** |
| **Patient** Edgar, Hipp  **Geburtsdatum** test  **Befund** stuff |

|  |
| --- |
| Mutationslast 1 nicht-synonyme SNV 2 |
| Anzahl betroffener Onkogenes 3 |
| Anzahl betroffener Tumorsupressorgene 4 |
| HLA Typ 5 |
| Additional information 6 |

|  |  |  |  |
| --- | --- | --- | --- |
| **Somatic Mutations in known driver Genes** | | | |
| **Gene** | **Mutation** | **Confidence** | **Reference** |
| BRAF | p.Val600Glu | 4 | 1,2,3,4 |
| SF3B1 | p.Pro718Leu | 3 | 1,2,4 |
| DLEC1 | p.Asp215Asn | 2 | 3,5 |

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Somatic Mutations in pharmaceutical target Protein** | | | | |
| **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 |

| **Indirect Association (Other Mutations with known effect on drug)** | | | | | |
| --- | --- | --- | --- | --- | --- |
| **Gene** | **Mutation** | **Therapy** | **Disease** | **Evidence** | **References** |
| BRAF | AGK-BRAF | Sorafenib | Melanoma | C | 29 |
| BRAF | AGK-BRAF | Vemurafenib | Melanoma | D | 29 |
| BRAF | L597R | Vemurafenib | Melanoma | C | 30 |

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Somatic Mutations with known pharmacogenetic effect** | | | | | |
| **Gene** | **Mutation** | **Therapy** | **Disease** | **Evidence** | **References** |
| BRAF | V600E | Cetuximab | Colorectal Cancer | B | 25 |
| BRAF | V600E | Dabrafenib,Trametinib | Melanoma | B | 24 |
| BRAF | V600E | Dabrafenib | Non-small Cell Lung Carcinoma | C | 28 |

References

|  |  |
| --- | --- |
| 1 | Futreal et al., A census of human cancer genes., Nature reviews. Cancer, 4, 3, 2004 |
| 2 | Vogelstein et al., Cancer genome landscapes., Science (New York, N.Y.), 339, 6127, 2013 |
| 3 | Apweiler et al., UniProt: the Universal Protein knowledgebase., Nucleic acids research, 32, Database issue, 2004 |
| 4 | Rubio et al., In silico prescription of anticancer drugs to cohorts of 28 tumor types reveals targeting opportunities., Cancer cell, 27, 3, 2015 |

Appendix

|  |  |  |  |
| --- | --- | --- | --- |
| **Gene** | **Mutation** | **dbSNP** | **COSMIC** |
| TNFRSF8 | p.Pro215Ser | rs267597959 | COSM14024 |
| KIAA0319L | p.Pro278Ser | rs267598576 | COSM36587 |
| FAM46C | p.Thr209Asn | character(0) |  |