Table 1. Summary of previously reported IDH1 p.R132 mutations in human diseases

<table>
<thead>
<tr>
<th>Diseases</th>
<th>Total number of samples</th>
<th>Samples with mutation</th>
<th>Mutation frequency</th>
<th>Frequency range</th>
<th>Number of IDH1-related reports [References]</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Brain tumors</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Gliomas</td>
<td>18297</td>
<td>6432</td>
<td>35.2%</td>
<td>0.0-100.0%</td>
<td>168 [1-167]</td>
</tr>
<tr>
<td>Non-glioma brain tumors</td>
<td>2232</td>
<td>675</td>
<td>30.2%</td>
<td>0.0-100.0%</td>
<td>30 [1, 3, 7, 12, 45, 59, 80, 149, 168-189]</td>
</tr>
<tr>
<td><strong>Hematological malignancies</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>AML</td>
<td>15509</td>
<td>1053</td>
<td>6.8%</td>
<td>0.0-25.0%</td>
<td>68 [3, 6, 29, 64, 190-253]</td>
</tr>
<tr>
<td>MDS/ MPD / MPN</td>
<td>4376</td>
<td>82</td>
<td>1.9%</td>
<td>0.0-18.8%</td>
<td>28 [193, 196, 199, 200, 206, 209, 213, 218, 226, 250, 254-271]</td>
</tr>
<tr>
<td>NHL</td>
<td>640</td>
<td>1</td>
<td>0.2%</td>
<td>0.0-0.8%</td>
<td>4 [170, 222, 226, 272]</td>
</tr>
<tr>
<td>ALL</td>
<td>567</td>
<td>3</td>
<td>0.5%</td>
<td>0.0-3.2%</td>
<td>6 [3, 6, 196, 215, 222, 229]</td>
</tr>
<tr>
<td>CML</td>
<td>479</td>
<td>0</td>
<td>0.0%</td>
<td>0.0%</td>
<td>7 [6, 196, 209, 218, 226, 273, 274]</td>
</tr>
<tr>
<td>HL</td>
<td>122</td>
<td>0</td>
<td>0.0%</td>
<td>0.0%</td>
<td>3 [170, 222, 226]</td>
</tr>
<tr>
<td>MM</td>
<td>92</td>
<td>0</td>
<td>0.0%</td>
<td>0.0%</td>
<td>2 [3, 226]</td>
</tr>
<tr>
<td>CLL</td>
<td>18</td>
<td>0</td>
<td>0.0%</td>
<td>0.0%</td>
<td>2 [6, 226]</td>
</tr>
<tr>
<td>AMML</td>
<td>3</td>
<td>0</td>
<td>0.0%</td>
<td>0.0%</td>
<td>1 [226]</td>
</tr>
<tr>
<td><strong>Others</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mesenchymal tumour</td>
<td>1200</td>
<td>74</td>
<td>6.2%</td>
<td>6.2%</td>
<td>1 [275]</td>
</tr>
<tr>
<td>Colorectal cancer</td>
<td>926</td>
<td>3</td>
<td>0.3%</td>
<td>0.0-2.9%</td>
<td>8 [2, 3, 6, 23, 149, 170, 276, 277]</td>
</tr>
</tbody>
</table>
Breast cancer 603 1 0.2% 0.0-100.0% 6 [2, 3, 6, 149, 170, 278]
Lung cancer 517 0 0.0% 0.0% 6 [2, 3, 6, 90, 149, 170]
Sarcoma 529 1 0.2% 0.0-100.0% 3 [170, 279, 280]
Pheochromocytoma 314 0 0.0% 0.0% 2 [281, 282]
Prostate cancer 387 7 1.8% 0.0-2.7% 6 [2, 3, 6, 149, 170, 283]
Pancreatic cancer 293 0 0.0% 0.0% 4 [2, 6, 170, 277]
Thyroid Cancer 504 19 3.8% 0.0-15.7% 6 [2, 23, 149, 170, 284, 285]
Cholangiocarcinoma 482 45 9.3% 7.1-14.9% 3 [277, 286, 287]
GIST 180 0 0.0% 0.0% 2 [2, 170]
Enchondroma and related diseases 278 103 37.1% 1.0-90.0% 4 [288-291]
Gastric cancer 190 0 0.0% 0.0% 3 [3, 6, 277]
Ovarian cancer 176 0 0.0% 0.0% 4 [2, 3, 6, 149]
Hepatocellular carcinoma 159 0 0.0% 0.0% 3 [3, 23, 277]
Paraganglioma 155 1 0.6% 0.0-0.8% 2 [281, 282]
Renal cancer 161 0 0.0% 0.0% 5 [2, 3, 149, 170, 292]
Melanoma 173 3 1.7% 0.0-5.1% 4 [2, 149, 279, 293]
Squamous cell carcinoma (oral) 90 0 0.0% 0.0% 1 [229]
Biliary tract cancer 87 9 10.3% 10.3% 1 [277]
Esophageus cancer 73 0 0.0% 0.0% 2 [2, 3]
Bladder cancer 38 0 0.0% 0.0% 1 [2]
Fibrous histiocytoma 36 0 0.0% 0.0% 1 [170]

<table>
<thead>
<tr>
<th>Tumor Type</th>
<th>Cases</th>
<th>IDH1</th>
<th>IDH2</th>
<th>IDH1+IDH2</th>
<th>Refs</th>
</tr>
</thead>
<tbody>
<tr>
<td>Urothelial carcinoma</td>
<td>28</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>1 [3]</td>
</tr>
<tr>
<td>Gallbladder cancer</td>
<td>25</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>3 [149, 277, 286]</td>
</tr>
<tr>
<td>Squamous cell carcinoma (skin)</td>
<td>19</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>1 [3]</td>
</tr>
<tr>
<td>Mesothelioma</td>
<td>18</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>3 [2, 3, 279]</td>
</tr>
<tr>
<td>Endometrial</td>
<td>18</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>1 [149]</td>
</tr>
<tr>
<td>Cervical cancer</td>
<td>11</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>2 [2, 3]</td>
</tr>
<tr>
<td>NPC</td>
<td>7</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>1 [170]</td>
</tr>
<tr>
<td>HNSCC</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>1 [2]</td>
</tr>
</tbody>
</table>


^a The last PubMed search was performed on March 8, 2013.

^b Papers with redundant data were not included in this table.
Lu J et al. IDH1 p.R132 mutations may not be actively involved in the carcinogenesis of hepatocellular carcinoma. Medical Science Monitor, 2013. in press


Lu J et al. IDH1 p.R132 mutations may not be actively involved in the carcinogenesis of hepatocellular carcinoma. Medical Science Monitor, 2013. in press

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77. Utsuki S, Oka H, Miyajima Y, et al: Adult cerebellar glioblastoma cases have different characteristics from supratentorial glioblastoma. Brain Tumor Pathol, 2012; 29(2): 87-95


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16(3): 161-70


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Lu J et al. IDH1 p.R132 mutations may not be actively involved in the carcinogenesis of hepatocellular carcinoma. Medical Science Monitor, 2013. in press

Oncol, 2012; 14(1): 109-16


204. Paschka P, Schlenk RF, Gaidzik VI, et al: IDH1 and IDH2 mutations are frequent genetic alterations in acute myeloid leukemia and confer adverse prognosis in cytogenetically


206. Rocquain J, Carbuccia N, Trouplin V, et al: Combined mutations of ASXL1, CBL, FLT3, IDH1, IDH2, JAK2, KRAS, NPM1, NRAS, RUNX1, TET2 and WT1 genes in myelodysplastic syndromes and acute myeloid leukemias. BMC Cancer, 2010; 10: 401

207. Schnittger S, Haferlach C, Ulke M, et al: IDH1 mutations are detected in 6.6% of 1414 AML patients and are associated with intermediate risk karyotype and unfavorable prognosis in adults younger than 60 years and unmutated NPM1 status. Blood, 2010; 116(25): 5486-96


Lu J et al. IDH1 p.R132 mutations may not be actively involved in the carcinogenesis of hepatocellular carcinoma. Medical Science Monitor, 2013. in press


Lu J et al. IDH1 p.R132 mutations may not be actively involved in the carcinogenesis of hepatocellular carcinoma. Medical Science Monitor, 2013. in press


Lu J et al. IDH1 p.R132 mutations may not be actively involved in the carcinogenesis of hepatocellular carcinoma. Medical Science Monitor, 2013. in press

Leuk Res, 2010; 34(8): 1091-3


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