Supplementary Online Content


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This supplementary material has been provided by the authors to give readers additional information about their work.
**eTABLE 1: Study Group Definitions in Analyses**

**ANALYSIS 1A:**
- **“BAP1 mutation” group:** patients with an insertion, deletion, insertion/deletion, nonsense mutation, nonsynonymous mutation likely to be damaging to protein function (>75%), or synonymous mutation likely to result in splice site changes (>75%).
- **“Control” group:** patients with no BAP1 polymorphisms.
*Patients with BAP1 polymorphisms which were unlikely to be damaging to protein function based on computational tools were excluded from analysis.

**ANALYSIS 1B:**
- **“BAP1 mutation” group:** patients with an insertion, deletion, insertion/deletion, nonsense mutation, nonsynonymous mutation likely to be damaging to protein function (>75%), or synonymous mutation likely to result in splice site changes (>75%).
- **“Control” group:** patients with no BAP1 polymorphisms and patients with BAP1 polymorphisms predicted not to be deleterious to protein function.

**ANALYSIS 2A:**
- **“BAP1 mutation” group:** patients with an insertion, deletion, insertion/deletion, nonsense mutation, nonsynonymous mutation likely to be damaging to protein function (>75%), or synonymous mutation with any possibility of splice site change.
- **“Control” group:** patients with no BAP1 polymorphisms.
*Patients with BAP1 polymorphisms which were unlikely to be damaging to protein function based on computational tools were excluded from analysis.

**ANALYSIS 2B:**
- **“BAP1 mutation” group:** patients with an insertion, deletion, insertion/deletion, nonsense mutation, nonsynonymous mutation likely to be damaging to protein function (>75%), or synonymous mutation with any possibility of splice site change.
- **“Control” group:** patients with no BAP1 polymorphisms and patients with BAP1 polymorphisms predicted not to be deleterious to protein function.

**ANALYSIS 3A:**
- **“BAP1 mutation” group:** patients with an insertion, deletion, insertion/deletion, nonsense mutation, any nonsynonymous mutation, or synonymous mutation with any possibility of splice site change.
- **“Control” group:** patients with no BAP1 polymorphisms.
*Patients with BAP1 polymorphisms which were unlikely to be damaging to protein function based on computational tools were excluded from analysis.

**ANALYSIS 3B:**
- **“BAP1 mutation” group:** patients with an insertion, deletion, insertion/deletion, nonsense mutation, any nonsynonymous mutation, or synonymous mutation with any possibility of splice site change.
- **“Control” group:** patients with no BAP1 polymorphisms and patients with BAP1 polymorphisms predicted not to be deleterious to protein function.
**eTABLE 2: Analysis 1B**

<table>
<thead>
<tr>
<th></th>
<th>BAP1 mutation</th>
<th>Control</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age at diagnosis (mean, years)</td>
<td>56.8</td>
<td>58.4</td>
<td>0.44</td>
</tr>
<tr>
<td>Gender (% male)</td>
<td>50.0</td>
<td>48.1</td>
<td>1.0</td>
</tr>
<tr>
<td>History of other malignancy (%)</td>
<td>25.0</td>
<td>14.8</td>
<td>0.34</td>
</tr>
<tr>
<td>Family history of malignancy † (%)</td>
<td>100.0</td>
<td>65.7</td>
<td>0.06</td>
</tr>
<tr>
<td>Largest tumor diameter (mean, mm)</td>
<td>15.9</td>
<td>12.3</td>
<td>0.004</td>
</tr>
<tr>
<td>Distance from the fovea (mean, disc diameter) *</td>
<td>1.94</td>
<td>2.59</td>
<td>0.53</td>
</tr>
<tr>
<td>Distance from the optic nerve (mean, disc diameter) *</td>
<td>1.69</td>
<td>2.67</td>
<td>0.28</td>
</tr>
<tr>
<td>Visual acuity (mean, logMAR) [Snellen] &amp;</td>
<td>0.56</td>
<td>0.41</td>
<td>0.13</td>
</tr>
<tr>
<td></td>
<td>[~ 20/72]</td>
<td>[~ 20/50]</td>
<td></td>
</tr>
<tr>
<td>Ciliary body involvement (~) (%)</td>
<td>75</td>
<td>21.9</td>
<td>0.002</td>
</tr>
<tr>
<td>Iris involvement # (%)</td>
<td>12.5</td>
<td>2.2</td>
<td>0.18</td>
</tr>
<tr>
<td>Extrascleral extension # (%)</td>
<td>0</td>
<td>3.6</td>
<td>1.0</td>
</tr>
<tr>
<td>Mean pigmentation score (% with “heavy” pigmentation)</td>
<td>50</td>
<td>47.1</td>
<td>0.54</td>
</tr>
<tr>
<td>Metastatic disease (~) (%)</td>
<td>71.4</td>
<td>17.8</td>
<td>0.003</td>
</tr>
</tbody>
</table>

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### eTABLE 3: Analysis 2A

<table>
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<tr>
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<th>BAPI mutation</th>
<th>Control</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age at diagnosis (mean, years)</td>
<td>57.8</td>
<td>58.5</td>
<td>0.36</td>
</tr>
<tr>
<td>Gender (% male)</td>
<td>63.6</td>
<td>48.1</td>
<td>0.37</td>
</tr>
<tr>
<td>History of other malignancy (%)</td>
<td>18.2</td>
<td>15.2</td>
<td>0.68</td>
</tr>
<tr>
<td>Family history of malignancy (%)</td>
<td>90.0</td>
<td>65.0</td>
<td>0.18</td>
</tr>
<tr>
<td>Largest tumor diameter (mean, mm)</td>
<td>15.1</td>
<td>12.6</td>
<td>0.006</td>
</tr>
<tr>
<td>Distance from the fovea (mean, disc diameter)</td>
<td>2.0</td>
<td>2.6</td>
<td>0.83</td>
</tr>
<tr>
<td>Distance from the optic nerve (mean, disc diameter)</td>
<td>1.5</td>
<td>2.7</td>
<td>0.09</td>
</tr>
<tr>
<td>Visual acuity (mean, logMAR) [Snellen]</td>
<td>0.46</td>
<td>0.40</td>
<td>0.38</td>
</tr>
<tr>
<td></td>
<td>[~ 20/56]</td>
<td>[~ 20/50]</td>
<td></td>
</tr>
<tr>
<td>Ciliary body involvement (%)</td>
<td>54.5</td>
<td>21.6</td>
<td>0.02</td>
</tr>
<tr>
<td>Iris involvement (%)</td>
<td>9.1</td>
<td>2.1</td>
<td>0.23</td>
</tr>
<tr>
<td>Extrascleral extension (%)</td>
<td>0</td>
<td>3.3</td>
<td>1.0</td>
</tr>
<tr>
<td>Mean pigmentation score (% with “heavy” pigmentation)</td>
<td>60</td>
<td>46.2</td>
<td>0.22</td>
</tr>
<tr>
<td>Metastatic disease (%)</td>
<td>55.6</td>
<td>18.0</td>
<td>0.01</td>
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</table>
**eTABLE 4: Analysis 2B**

<table>
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<th>p-value</th>
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<tbody>
<tr>
<td>Age at diagnosis (mean, years)</td>
<td>57.6</td>
<td>58.4</td>
<td>0.37</td>
</tr>
<tr>
<td>Gender (% male)</td>
<td>63.6</td>
<td>47.8</td>
<td>0.37</td>
</tr>
<tr>
<td>History of other malignancy (%)</td>
<td>27.3</td>
<td>14.7</td>
<td>0.22</td>
</tr>
<tr>
<td>Family history of malignancy (%)</td>
<td>81.8</td>
<td>66.0</td>
<td>0.34</td>
</tr>
<tr>
<td>Largest tumor diameter (mean, mm)</td>
<td>15.1</td>
<td>12.3</td>
<td>0.006</td>
</tr>
<tr>
<td>Distance from the fovea (mean, disc diameter)</td>
<td>1.98</td>
<td>2.6</td>
<td>0.84</td>
</tr>
<tr>
<td>Distance from the optic nerve (mean, disc diameter)</td>
<td>1.57</td>
<td>2.69</td>
<td>0.10</td>
</tr>
<tr>
<td>Visual acuity (mean, logMAR) [Snellen]</td>
<td>0.45 [~ 20/56]</td>
<td>0.41 [~ 20/50]</td>
<td>0.41</td>
</tr>
<tr>
<td>Ciliary body involvement (%)</td>
<td>54.5</td>
<td>22.0</td>
<td>0.02</td>
</tr>
<tr>
<td>Iris involvement (%)</td>
<td>9.1</td>
<td>2.2</td>
<td>0.23</td>
</tr>
<tr>
<td>Extrascleral extension (%)</td>
<td>0</td>
<td>3.6</td>
<td>1.0</td>
</tr>
<tr>
<td>Mean pigmentation score (% with “heavy” pigmentation)</td>
<td>60</td>
<td>46.9</td>
<td>0.24</td>
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<tr>
<td>Metastatic disease (%)</td>
<td>55.6</td>
<td>17.9</td>
<td>0.01</td>
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**eTABLE 5: Analysis 3A**

<table>
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<th>p-value</th>
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<tbody>
<tr>
<td>Age at diagnosis (mean, years)</td>
<td>55.0</td>
<td>58.5</td>
<td>0.20</td>
</tr>
<tr>
<td>Gender (% male)</td>
<td>60.0</td>
<td>48.1</td>
<td>0.44</td>
</tr>
<tr>
<td>History of other malignancy (%)</td>
<td>20.0</td>
<td>15.2</td>
<td>0.71</td>
</tr>
<tr>
<td>Family history of malignancy (%)</td>
<td>85.7</td>
<td>65.6</td>
<td>0.15</td>
</tr>
<tr>
<td>Largest tumor diameter (mean, mm)</td>
<td>14.3</td>
<td>12.3</td>
<td>0.06</td>
</tr>
<tr>
<td>Distance from the fovea (mean, disc diameter)</td>
<td>2.0</td>
<td>2.6</td>
<td>0.53</td>
</tr>
<tr>
<td>Distance from the optic nerve (mean, disc diameter)</td>
<td>1.7</td>
<td>2.7</td>
<td>0.08</td>
</tr>
<tr>
<td>Visual acuity (mean, logMAR) [Snellen]</td>
<td>0.37</td>
<td>0.40</td>
<td>0.85</td>
</tr>
<tr>
<td></td>
<td>~ 20/46</td>
<td>~ 20/50</td>
<td></td>
</tr>
<tr>
<td>Ciliary body involvement (%)</td>
<td>46.7</td>
<td>21.7</td>
<td>0.05</td>
</tr>
<tr>
<td>Iris involvement (%)</td>
<td>6.7</td>
<td>2.1</td>
<td>0.29</td>
</tr>
<tr>
<td>Extrascleral extension (%)</td>
<td>0</td>
<td>3.3</td>
<td>1.0</td>
</tr>
<tr>
<td>Mean pigmentation score (% with “heavy” pigmentation)</td>
<td>57.1</td>
<td>46.2</td>
<td>0.13</td>
</tr>
<tr>
<td>Metastastic disease (%)</td>
<td>46.2</td>
<td>18.0</td>
<td>0.02</td>
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</table>
### eTABLE 6: Analysis 3B

<table>
<thead>
<tr>
<th></th>
<th>BAPI mutation</th>
<th>Control</th>
<th>p-value</th>
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</thead>
<tbody>
<tr>
<td>Age at diagnosis (mean, years)</td>
<td>55.0</td>
<td>58.4</td>
<td>0.20</td>
</tr>
<tr>
<td>Gender (% male)</td>
<td>60.0</td>
<td>47.8</td>
<td>0.44</td>
</tr>
<tr>
<td>History of other malignancy (%)</td>
<td>20.0</td>
<td>14.8</td>
<td>0.48</td>
</tr>
<tr>
<td>Family history of malignancy (%)</td>
<td>80.0</td>
<td>65.9</td>
<td>0.40</td>
</tr>
<tr>
<td>Largest tumor diameter (mean, mm)</td>
<td>14.3</td>
<td>12.3</td>
<td>0.06</td>
</tr>
<tr>
<td>Distance from the fovea (mean, disc diameter)</td>
<td>2.04</td>
<td>2.6</td>
<td>0.54</td>
</tr>
<tr>
<td>Distance from the optic nerve (mean, disc diameter)</td>
<td>1.73</td>
<td>2.69</td>
<td>0.08</td>
</tr>
<tr>
<td>Visual acuity (mean, logMAR) [Snellen]</td>
<td>0.37</td>
<td>0.41 [~ 20/46]</td>
<td>0.88 [~ 20/50]</td>
</tr>
<tr>
<td>Ciliary body involvement (%)</td>
<td>46.7</td>
<td>22</td>
<td>0.05</td>
</tr>
<tr>
<td>Iris involvement (%)</td>
<td>6.7</td>
<td>2.2</td>
<td>0.31</td>
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<tr>
<td>Extrascleral extension (%)</td>
<td>0</td>
<td>3.7</td>
<td>1.0</td>
</tr>
<tr>
<td>Mean pigmentation score (% with “heavy” pigmentation)</td>
<td>57.1</td>
<td>46.9</td>
<td>0.31</td>
</tr>
<tr>
<td>Metastatic disease (%)</td>
<td>46.2</td>
<td>17.8</td>
<td>0.02</td>
</tr>
</tbody>
</table>