CLINICAL SUMMARY

Right superior fundus mass lesion

HISTOPATHOLOGY

- **Specimen type:** Enucleation, Right eye
- **Tumour Size:** 11mm (basal diameter) x 8mm (thickness)
- **Tumour type:** Choroidal Melanoma, Grade 2
- **Tumour stage:** pT2a (AJCC, 8th edition)
- **Cell type:** Spindled, heavily pigmented (region/clone A)
  Predominantly epithelioid, amelanotic (region/clone B)
- **Molecular Studies:** BAP1 retained, chromosome 3 disomy, chromosome 6p gain, chromosome 6q disomy, chromosome 8q disomy, segmental loss of chromosome 17p, loss of chromosome Y
- **Immunohistochemistry:** CD68+ high (A); moderate (B)
  CD163+ moderate (A), moderate to high (B)

NEXT GENERATION GENOME/TRANSCRIPTOME DATA

- **Provider:** Australian Genome Research Facility - 150bp paired end
- **Data yield:**
  - Germline DNA: 519,446,425 paired end, 156.87 Gbp
  - Tumour DNA: Clone A: 792,884,149 paired end, 239.44 Gbp
  - Clone B: 1,202,016,740 paired end, 360.01 Gbp

<table>
<thead>
<tr>
<th>Gene</th>
<th>Alteration</th>
<th>Allele Frequency</th>
<th>Function</th>
</tr>
</thead>
<tbody>
<tr>
<td>GNAQ</td>
<td>c.626A&gt;C, p.Q209P</td>
<td>Clone A: 55%; Clone B: 48%</td>
<td>Gain</td>
</tr>
</tbody>
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No Tumour Dissociates were banked from this sample