Genetics in Retinoblastoma: Challenges in Kenya

Dr. Njambi Ombaba
Outline

• Background

• The challenges

• Future

Leucokoria  Squint  Proptosis
Retinoblastoma genetics

- Retinoblastoma is a genetic disease
- Mutation of RB1 gene: Tumor Suppressor gene

- The *RB1* gene is involved in
  - control of cell growth
  - preservation of genome integrity
ALL Retinoblastoma tumours have mutations in both *RB1* copies.

![Diagram showing the progression of RB1 mutations from *RB1*+/+ to *RB1*+/- to *RB1*--.](image-url)
Germline Vs Somatic

- Germline: RB1^+/-
- Somatic: normal
# Clinical Value of Retinoblastoma Genetics

<table>
<thead>
<tr>
<th></th>
<th>Germline</th>
<th>Somatic</th>
</tr>
</thead>
<tbody>
<tr>
<td>Heritability</td>
<td>Heritable</td>
<td>Non heritable</td>
</tr>
<tr>
<td>Laterality</td>
<td>Bilateral, some</td>
<td>Unilateral</td>
</tr>
<tr>
<td></td>
<td>unilateral</td>
<td></td>
</tr>
<tr>
<td>Multiple tumours</td>
<td>Multifocal</td>
<td>Unifocal</td>
</tr>
<tr>
<td>Risk of other</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>cancers-sarcomas</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Risk to siblings/offsprings</td>
<td>Yes</td>
<td>No</td>
</tr>
</tbody>
</table>
Genetic services in retinoblastoma

- Counseling
- Molecular genetic testing
- Surveillance for other cancers
Developing Clinical Cancer Genetics Services in Resource-Limited Countries: The Case of Retinoblastoma in Kenya

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Challenges in Genetic counseling

Doctor’s Perspective
• No genetic counseling unit / training / counselor
• Limited knowledge / skills
• Overworked, limited time
• Lack of genetic testing services
• Frequent examinations under anaesthesia

Patient’s Perspective
• Self blame / denial
• Late presentation
• Poor follow up / costs
• Educational / religious / cultural diversity
• Translation of complex genetic information into local dialects
• Mothers role in the community
• Fathers / clan / community dominance
Future

Medical Education & Testing services

Patient & Family Counseling

Community Engagement
National guidelines on counseling & testing
Thank you