Retinoblastoma: a curable, rare and deadly blinding disease

Every year, thousands of babies and children in low- and middle-income countries lose their sight and their lives to a treatable childhood eye cancer called retinoblastoma; usually because it was not recognised and treated in time.

Although retinoblastoma is relatively uncommon, it can have devastating consequences for the children affected by it. If treated too late, it can lead to the loss of the eye, invasion of the brain and death.

Retinoblastoma does not affect everyone equally. In high-income countries, fewer than 5% of children die as a result of the condition, thanks to early diagnosis and specialist treatment. In Africa, however, it is typical to see 70% of children with retinoblastoma die, mainly because they presented too late. When mothers do present at a tertiary centre with a child who has advanced retinoblastoma, they often report that they have had several interactions with different health professionals over many months or even years, but did not get the referral or care they needed. Every health professional reading this issue of the Community Eye Health Journal has a chance to redress this balance. We need to find and treat children with retinoblastoma early, before it causes disfigurement or death.

Doing so successfully requires adopting a multidisciplinary, multi-level and internationally collaborative approach that looks at the health system as a whole (see page 4). Raising awareness of retinoblastoma in the community, improving the detection and diagnosis of the condition, setting up good referral systems and offering good...
Retinoblastoma is a rare condition with devastating consequences. If left untreated, it can lead to loss of the eye, invasion of the brain and death. In this issue, we offer information and guidance about the detection, diagnosis and treatment of retinoblastoma, including advice about screening for family members when genetic testing is not available, and a step-by-step guide to enucleation. We hope that every health professional reading this journal will raise awareness of this condition so that – globally – we have a better chance to save the life, eyes and sight of children with retinoblastoma.

1 Retinoblastoma: a curable, rare and deadly blindness disease
Richard Bowman

5 A national retinoblastoma network: experiences in Kenya and the UK
Ashwin Reddy and Kahaki Kimani

7 Understanding retinoblastoma: epidemiology and genetics
Ido Didi Fabian and Manoel S Sagoo

8 Germline retinoblastoma: estimating risk and counselling the family
Elisabeth Rosser and Manoel S Sagoo

10 Detecting retinoblastoma
Manoj V Parulekar

11 Classification and staging of retinoblastoma
Ido Didi Fabian, Ashwin Reddy and Manoel S Sagoo

14 Managing and treating intraocular retinoblastoma
Ashwin Reddy, Mukesh Jain and Vikas Khetan

17 Standard reporting of high-risk histopathology features in retinoblastoma
Caroline Thaung and Elin Kotilgo Karra

18 Management of retinoblastoma with extracocular tumour extension
Swathi Kaliki and Vijay Anand Reddy Palkonda

20 How to do an enucleation for retinoblastoma
Swathi Kaliki

23 CLINICAL SKILLS: Testing the red reflex
Richard Bowman and Allen Foster

24 TRACHOMA: Making gains sustainable: partnering with WASH to stop the transmission of trachoma
Leah Wohlgemuth, Helen Hamilton and Tim Jesudason

25 ONCHOCERCIASIS: River blindness: reducing the prevalence of clinical disease
Charles Mackenzie, Martin Kollmann, Sabine Specht and Yao Sodhalon

26 Questions and answers on retinoblastoma

27 Picture quiz

27 Announcements and resources

28 KEY MESSAGES
the parents and/or carers. If they have seen something white or abnormal in their child’s eye, believe what they say, take it seriously and seek specialist advice.

In Tanzania, community nurses have been trained to examine the red reflex (p. 23) using an Arclight ophthalmoscope. The Arclight is an affordable, solar-powered and easy-to-use ophthalmoscope. It has shown preliminary promise; the community nurses found it easy to learn and began picking up cases of cataract and retinoblastoma by using it. Nurses can learn how to examine the red reflex at the same time as examining the child’s other systems.

Tertiary centres

At tertiary centres, histopathologists have a crucial role: once the eye is removed the child may be able to leave hospital completely cured or may need chemotherapy.

Public health awareness campaigns can support the early detection of retinoblastoma

INDIA

Unless otherwise stated, journal content is licensed under a Creative Commons Attribution-NonCommercial (CC BY-NC) license which permits unrestricted use, distribution, and reproduction in any medium for non-commercial purposes, provided that the copyright holders are acknowledged.

ISSN 0953-6833.

Disclaimer

Signed articles are the responsibility of the named authors alone and do not necessarily reflect the views of the London School of Hygiene & Tropical Medicine (the School). Although every effort is made to ensure accuracy, the School does not warrant that the information contained in this publication is complete and correct and shall not be liable for any damages incurred as a result of its use.

The mention of specific companies or of certain manufacturers’ products does not imply that they are endorsed or recommended by the School in preference to others of a similar nature that are not mentioned. The School does not endorse or recommend products or services for which you may view advertisements in this Journal.

The Community Eye Health Journal is supported by:
or radiotherapy; this decision must be based on accurate histopathological staging (page 18).

**International collaboration**

To promote this multi-level, multi-disciplinary and internationally collaborative approach, the Commonwealth Eye Health Consortium has provided start-up funding for an Rb-Network known as Rb-NET, which has already generated specific country plans, a set of core outcome indicators, best practice protocols and a practical resource manual (http://cehc.lshtm.ac.uk/dr-links/rbnet/).

Basic clinical research questions still need to be answered. For instance, researchers in Uganda have shown an improvement in survival by giving chemotherapy before surgery on the basis that so many children have extra-ocular spread at time of presentation. On the other hand, a small study from Tanzania showed that 60% of children for whom there was good histology after enucleation had complete excision of the tumour with low risk and never needed chemotherapy. So which should come first in these settings – chemotherapy or surgery? By combining multi-centre and multi-country clinical research, as Rb-NET has started to do, we can begin to answer these questions and prevent needless tragedies.

This issue of the *Journal* demonstrates that there is real momentum and determination to improve outcomes for children with Rb in all countries across the world. It contains concise, practical information that should help all of us to make a difference.

**Table 1** Roles and responsibilities in the detection, referral and treatment of retinoblastoma

<table>
<thead>
<tr>
<th>Individual responsibilities</th>
<th>Specialist eye centre</th>
</tr>
</thead>
<tbody>
<tr>
<td>Seek help urgently if you see something white inside the centre of the eye (the pupil) OR if you take a photograph and only one eye has a red dot in the centre Do not let anyone turn you away and do not take no for an answer until a doctor at a hospital has examined the child's eyes using a bright light</td>
<td>Same as for ophthalmologists, plus: Learn how to give focal or laser treatment to smaller tumours (usually in the second eye) Create multidisciplinary teams who work closely together to coordinate the treatment of each child Include in this team: ophthalmologists, oncologists, histopathologists, nurses, child life specialists or play therapists and/or counsellors Offer general and genetic counselling to parents/carers Refer parents to other sources of support for their child's learning and development</td>
</tr>
<tr>
<td>Believe the parents if they say they have seen something white inside the pupil and seek specialist advice. Treat it as a medical emergency Learn how to test the red reflex (p. 23). Test all children during routine visits and immunisations</td>
<td>Refer all children with signs of retinoblastoma in two eyes to a national or specialist centre for urgent treatment</td>
</tr>
<tr>
<td>Learn to recognise retinoblastoma and to identify eyes that need enucleation Counsel parents about the good cosmetic outcomes of enucleation with implantation. Show pictures of children with good outcomes Learn how to enucleate, taking more than 15 mm of optic nerve. Always examine the fundus of the fellow eye when you perform an enucleation: there could be a small tumour which is treatable by laser</td>
<td></td>
</tr>
</tbody>
</table>

The Ministry of Health's responsibilities towards the above

Run public awareness campaigns so that parents know that treatment is possible and know when to see a doctor

Ensure that the red reflex test (p. 23) is included in the curriculum for nurses and health workers

Ensure there is at least one ophthalmologist per 100,000 population

Support the development of national retinoblastoma centres and referral networks. Offer subsidised access to specialist treatment for all children with retinoblastoma. Provide screening services for siblings and accommodation or travel subsidies for the parents or carers of these young children.