World Eye Cancer Hope
life and sight for every child

Notes for Editors - Retinoblastoma

About Retinoblastoma

- Every year, 300,000 children (0-15 years) develop cancer around the world. 85% live in developing countries and 240,000 (80%) will not survive five years.
- Retinoblastoma (Rb) is a rare cancer that forms in the eyes of babies and young children, usually before the age of three years.
- Rb can affect one eye (unilateral) or both eyes (bilateral) and a child can have multiple tumours in one or both eyes. The cancer does not spread from one eye to the other. Each tumour is caused by a unique genetic change in a single retinal cell.
- Rarely, affected children develop a brain tumour called trilateral retinoblastoma. This has traditionally been fatal, but a growing number of children in developed countries are surviving with aggressive treatment, and they give us great hope for the future.
- Retinoblastoma affects 1:15-20,000 live births, around 9,000 children worldwide each year. It affects boys and girls equally, and is not more common in any race or region.
- About 45 children develop this cancer in the UK each year, 23 in Canada, 280 in the USA and 86 in Kenya. Please contact WE C Hope for other country data.
- Retinoblastoma accounts for about 3% of childhood cancer in developed countries. In large, growing populations, it can account for as much as 15% of childhood cancers due to a higher proportion of the population under five years.

Symptoms and Diagnosis

- The most common early sign of Rb Is a pearly-white glow in the affected eye/s, frequently seen in flash photos. When the cancer is advanced, the white reflection can be seen with the naked eye, most often at dawn and dusk when ambient lighting is low.
- Other symptoms include a squint (misaligned eyes) and a red, sore or swollen eye. In developing countries, many children present with a bulging eye or ruptured eyeball because of poor awareness and access to health care.
- There are other reasons for a white pupil, but in young children, Rb should always be ruled out quickly with an eye exam by an ophthalmologist.
- In developed countries, average age at diagnosis is 30 months (unilateral) and 15 months (bilateral). In Kenya (typical of the developing world), the average age at diagnosis is 36 months (unilateral) and 26 months (bilateral).
- In developing countries, where families often do not have access to a camera, awareness of white pupil is essential. By the time parents see it with their naked eye, the cancer is likely to already threaten life, and the child needs urgent medical care.
- Diagnosis can be confirmed with a thorough eye examination under general anaesthetic, but no biopsy is done as this increases risk of tumour spread beyond the eye. The child should be treated by an ophthalmologist (eye doctor) and oncologist (cancer doctor) working together who have experience of caring for children with retinoblastoma.

Treatment

- Surgical removal of the affected eye/s is the most effective treatment. About 80% of children have at least one eye removed to protect their life, and 5% lose both eyes in developed countries. This is usually because their cancer has not been detected early.
- Greater awareness of white pupil – the early sign of this cancer – will save many children's lives, eyes and sight.
- Other treatments include chemotherapy, laser, cryotherapy (freezing) and radiotherapy. The eye will be removed if these treatments fail to kill the cancer.
- When cancer has spread outside the eye or the child has trilateral Rb, intensive chemotherapy, stem cell or bone marrow transplant, and sometimes radiotherapy are required to give the best chance of cure.

Prognosis

- Children have an excellent chance of cure when retinoblastoma is fully contained in the eye at diagnosis and they receive appropriate treatment and follow up for both eyes by doctors experienced in managing retinoblastoma.
- When Rb has spread outside the eye, the chance of cure is very small, even with aggressive state of the art treatment.
- Early diagnosis is essential for cure.
- 96-99% of children treated in developed countries are cured today.
- In the developing world, early diagnosis and access to specialist care is limited. Most children are not diagnosed until the cancer has already spread outside their eye, and the aggressive treatments needed to give them a chance of cure are either unavailable or too expensive. As a result, global survival is less than 20%.

Genetics

- Most retinoblastoma is caused by errors (mutations) on the *RB1* gene. These may either be inherited or occur during the normal process of cell division.
- In about 50% of children, the *RB1* mutation can be passed to the next generation. These children also have a higher risk of developing other cancers throughout life.
- About 1.5% of children with cancer in one eye and no family history have two normal copies of the *RB1* gene in their tumour. Their cancer is caused by too many copies of *MYCN*, the gene most commonly associated with high risk neuroblastoma. This form of the cancer develops in young babies, is very aggressive and cannot be inherited. The average age at diagnosis is 4.5 months.
- Genetic testing is important to identify which children have the heritable form or Rb, to identify at risk relatives and to eliminate invasive surveillance procedures for children who are not at risk for retinoblastoma.
- *RB1* was the first "tumour suppressor" (cancer preventing) gene discovered. Research of *RB1* has increased fundamental knowledge of how all cancers form, leading to development of new therapies and genetic tests for common cancers affecting millions of adults. Ongoing *RB1* research is essential to further our knowledge and ability to cure – and even prevent – cancer.