Medical Xpress

Cancer researchers discover new type of retinoblastoma in babies

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A team of Canadian and international cancer researchers led by Dr. Brenda Gallie at the Princess Margaret Cancer Centre, University Health Network (UHN), has discovered a new type of retinoblastoma, a rapidly developing eye cancer that affects very young babies– a finding that can immediately change clinical practice and optimize care for these children.

The finding, published online today in Lancet Oncology, is a breakthrough in recognizing that a single cancer gene (an oncogene) drives an aggressive retinoblastoma that starts long before birth in families with no history of the disease, says surgeon Dr. Gallie, an ophthalmologist who is also affiliated with The Hospital for Sick Children (SickKids) and a Professor in the Faculty of Medicine, University of Toronto.

"This research completely challenges conventional thinking and clinical practice," says Dr. Gallie. "The common type of retinoblastoma is initiated by damage to both copies of the RB1 tumor suppressor gene; the predisposition to this type of retinoblastoma can be inherited, so the other eye of the child and those of infant relatives are at risk to develop tumours. When we remove the eye with a large tumour in very young babies and show it is the new oncogene-driven type of retinoblastoma, there is believed to be zero risk for retinoblastoma developing in the other eye or in other infants in the family. This is a major advance in personalized cancer medicine for these children and families."

The oncogene-driven tumours are much larger than those anticipated in children with inherited retinoblastoma at the same age. "The earliest diagnosis comes when parents observe a white (instead of black) pupil of the eye, and the doctors listen to their observations and understand the urgency of referral. Sometimes Mom really does know best and clinicians should pay close attention."

Although less than 2% of unilateral retinoblastoma tumors are driven by the oncogene, the early age of onset predicts that about 1 in 5 babies diagnosed under six months of age actually has oncogene-driven retinoblastoma. "All the babies were completely cured by surgery," says Dr. Gallie.

"We've thought for a long time that all retinoblastoma were caused by loss of the retinoblastoma gene. Our study now reveals that's not the whole story: a new type of retinoblastoma, with normal retinoblastoma genes, is instead driven by extra copies of a powerful cancer gene, causing the cancer to grow very rapidly long before birth. The average age of diagnosis is four months."

This study, on which several clinical laboratories collaborated, demonstrates that molecular diagnostics can identify novel malignant diseases that elude traditional microscopic study of tissue. The researchers analysed more than 1,000 primary unilateral non-familial retinoblastoma tumours to validate oncogene-driven retinoblastoma. The Canadian research team included three UHN Research Institutes; Impact Genetics, Toronto; the B.C. Cancer Research Centre and University of British Columbia, Vancouver; the Cross Centre, Edmonton; The Hospital for Sick Children and the Ontario Institute for Cancer Research. The international collaborators were from the Netherlands, Germany, France and New Zealand.