Letter to the Editor



Ophthalmic Genetics 1381-6810/02/ US\$ 16.00

Ophthalmic Genetics – 2002, Vol. 23 No. 2, pp. 135–136 © Swets & Zeitlinger 2002

An infant with Down syndrome and retinoblastoma. A possible non-fortuitous association

Annette C. Moll Saskia M. Imhof Lex Bouter Willem den Otter Jan Willem Koten

Dear editor,

Satge et al. reported in *Ophthalmic Genetics* an infant with Down syndrome and retinoblastoma and presented also a review of the literature. They are to be commended with a very interesting study.

As the authors stated in their article, it is unlikely that all cases with both Down syndrome and retinoblastoma are reported in the literature and included in their study. In fact, they missed at least one study² published in your journal regarding the prevalence of mental retardation in patients with hereditary retinoblastoma. We did an epidemiological study and found in the Dutch retinoblastoma Register 8.7% cumulative incidence of mental retardation in 241 patients with hereditary retinoblastoma born in a 50 year period between 1945–1994. Two patients (females) of the 241 hereditary retinoblastoma patients had both Down syndrome (trisomy 21) and sporadic bilateral retinoblastoma (no 13q deletion found).² That is much higher than the 1 Down syndrome in 690 life births reported by the WHO.

In summary, our epidemiological study supports the idea of Statge et al.' that hereditary retinoblastoma patients may have an increased risk for Down syndrome.

Sincerely,
Annette C. Moll
Saskia M. Imhof
Lex Bouter
Amsterdam, the Netherlands
Willem den Otter
Jan Willem Koten
Utrecht, the Netherlands

Correspondence and reprint requests to:

Annette C. Moll, M.D., Ph.D.
Ophthalmologist, Epidemiologist
Department of Ophthalmology
VU University Medical Center
De Boelelaan 1117
1081 HV Amsterdam
The Netherlands
Tel: +31-20-4444795

Fax: +31-20-4444745 E-mail: a.moll@vumc.nl

References

- I Satge D, Gembara P, Sasco AJ, Francannet C, Desjardin L, Vekemans M, Demeocq F. An infant with Down syndrome and retinoblastoma: A possible nonfortuitous association. *Ophthalmic Genetics* 2001;22:117–123.
- 2 Moll AC, Imhof SM, Bleeker-Wagemakers EM, Den Otter W, Bouter LM, Koten JW, Tan KEWP.
- Prevalence of mental retardation in patients with hereditary retinoblastoma. *Ophthalmic Genetics* 1997;18:217–220.
- 3 World Health Organization.
 Down syndrome. In: Congenital
 Malformations worldwide. A report
 from the International Clearinghouse
 for Birth Defects Monitoring
 Systems. Amsterdam: Elsevier,
 1991;157–162.