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*Case Report*

# New cytogenetic aberrations found in a case of aggressive retinoblastoma

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**ABSTRACT.** We describe a case of retinoblastoma with an atypical presentation and previously unreported cytogenetic aberrations. A 19-month-old girl with left intraocular retinoblastoma was treated with enucleation and chemotherapy. The disease showed aggressive evolution within a short period between diagnosis and relapse. Eight months after diagnosis, a new large tumor was present in the orbit of the right eye, with diffuse bone pain, pancytopenia and diffuse infiltration into the bone marrow and the central nervous system. The child did not respond to treatment and died. Cytogenetic studies made with G-banding, FISH and SKY analysis showed chromosomal aberrations commonly associated with retinoblastoma, including del(13q), i(6p),

 

+1, and monosomy 16, along with others that had not been reported previously, including dup(5q), dic(15;22) and add(14q). The new chromosomal aberrations may be related to the aggressiveness of the disease in this case.

**Key words:** Retinoblastoma; Cytogenetics; del(13q); i(6p)

 