Retinoblastoma, our experience

Inês Coutinho1, Cristina Santos1, Mário Ramalho1, João Cabral1, Susana Teixeira1
1Hospital Prof. Doutor Fernando Fonseca, EPE

Introduction

Retinoblastoma is the most frequent intraocular tumor in childhood (incidence 1:15 000/20 000 births). In Portugal 5-6 cases/year

Caused by a mutation in the RB1 tumor suppressor gene and both copies of the gene must be mutated in order for a tumor to form. This mutation may be nonheritable (60%) or heritable (40%), the latter by autosomal-dominant transmission with high penetrance.

Retinoblastoma is typically diagnosed during the 1st year in familial and bilateral cases and between ages 1-3 in sporadic unilateral cases. There is no gender or race predilection.

Late diagnosis usually means lost of eye function or even death, that is why early diagnosis and treatment is so critical to preserve life, eye and vision, the key of success.

Goals

Retrospective analysis of patients diagnosed with retinoblastoma, observed and submitted to treatment at Hospital Prof. Doutor Fernando Fonseca (HFF), with collaboration of Instituto Português de Oncologia de Lisboa (IPO) and Hôpital Ophthalmique Jules-Gonin (HJG), in the last 8 years (2004-2012). Presentation of a case.

Clinical case

4 Months old, family history of Retinoblastoma

Retinoblastoma (stage A: < 3mm) on both eyes