Retinoblastoma, our experience

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Introduction

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

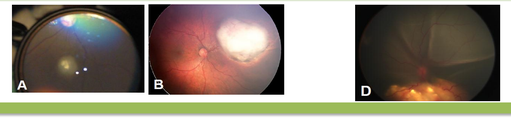
Caused by a mutation in the RB1 tumor suppressor gene and both copies of the gene must be mutated in order for a tumor to form.
This mutation may be nonheritable (60%) or heritable (40%), the latter by autosomal-dominant transmission with high penetrance.

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.

Presentation	Number of cases (%)	
	Goddard et al[2]	Abramson et al[3]
Leukocoria	52	54
Strabismus	29	19
Changes in the appearance of the eye	10	5
Visual acuity decrease	9	4

International Intraocular Retinoblastoma Classification
Group A- Very low risk
Group B- Low risk
Group C- Moderate risk
Group D- High risk
Group E- Very high risk



Therapeutic approaches	
Enucleation	Thermotherapy
Local chemotherapy	Brachytherapy
Cryotherapy	Radiotherapy by protons beam irradiation
Photocoagulation	Systemic chemotherapy

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.

Table 1- Epidemiological data

Variable	Laterality		Global
	unilateral	bilateral	
Nº of patients	7 (64%)	4 (36%)	11
Nº of eyes	7	8	15
Average age at diagnosis (months)	28	8	21
Gender (F/M)	5/2	1/3	6/5 (54%/46%)
Race (caucasian/ black)	6/1	2/2	8/3 (73%/27%)
Family history	-	1	1 (9%)

Table 2 and 3- Presentation and staging

Presentation	Number of patients
Asymptomatic (screening)	2 (18%) (1- family history; 1- prematurity)
Leukocoria	7 (64%)
Strabismus	2 (18%)

Stage	Number of eyes
A	3 (20%)
B	3 (20%)
C	-
D	6 (40%)
E	3 (20%)

Table 4 and 5- Treatment modalities

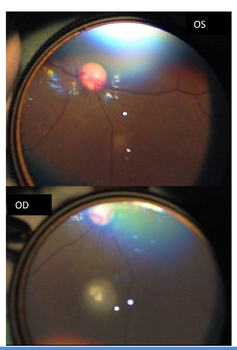
Treatment	Number of eyes	
	HFF/IPO	HJG
Systemic chemotherapy	15 (100%)	-
Photocoagulation	11 (73%)	-
Cryotherapy	10 (66%)	-
Transpupillary termotherapy (Diode LASER)	-	6 (40%)
Radiotherapy	-	3 (20%)
Local chemotherapy (intravitreal or intraarterial)	-	2 (13%)
Primary enucleation	4 (27%)	-
Secondary enucleation	3 (20%)	-

Parameters	Laterality		Total
	unilateral	bilateral	
Nº of eyes	7	8	15
Conservative treatment (nº of eyes)	4 (27%)	4 (27%)	8 (53%)
Enucleation (nº of eyes)	3 (20%)	4 (27%)	7 (47%)
Bilateral enucleation (nº of patients)	-	1	-

Clinical case

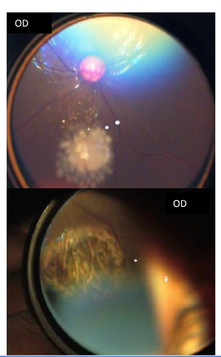
4 Months old, family history of Retinoblastoma
Retinoblastoma (stage A: < 3mm) on both eyes

Before treatment



Systemic chemotherapy + Photocoagulation + Cryotherapy

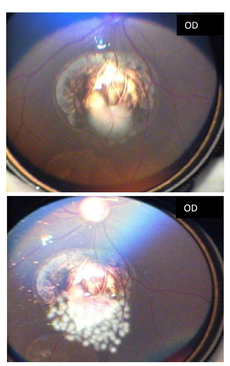
After treatment



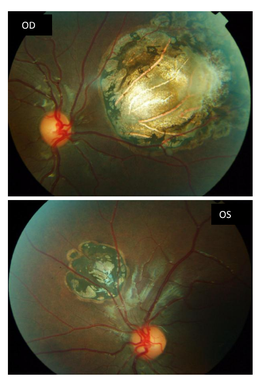
Immediately

15 days: scarring lesions

Relapse 6 months after first treatment



Photocoagulation + Cryotherapy



After 5 years: no sign of relapse
Visual acuity 1.0 (both eyes)

The best treatment is, and will always be, early diagnosis and treatment

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