Case Report:  
Retinal Dysplasia Mimicking Retinoblastoma.

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Abstract: Retinal dysplasia represents a congenital disorder characterized by abnormal proliferation of retinal tissue causing leukocoria. We present a case of an infant with bilateral leukocoria, clinically diagnosed as retinoblastoma, followed by enucleation of the left eye. Microscopy however, demonstrated retinal dysplasia consisting of a disorderly proliferation of retinal tissue with formation of rosettes, mimicking retinoblastoma. Microscopic features that aid in differentiating this lesion from retinoblastoma are discussed.  
Keywords: Retinal Dysplasia; Retinoblastoma; Leukocoria

Introduction  
Leukocoria, an abnormal white reflection from the retina, is a condition caused by several lesions. The causes of leukocoria include Retinoblastoma and also non-tumorous conditions like Persistent Hyperplastic Primary Vitreous, Coat’s disease, congenital cataract and retinal dysplasia. It is of vital importance that Retinoblastoma be differentiated from the rest of the non-tumorous conditions as treatment options and prognosis are different in cases of retinoblastoma. Retinal dysplasia represents a rare cause of leukocoria. It is the congenital anomaly in which the retinal layer forms a disordered proliferative lesion and leads to congenital blindness in children.

Case Report  
We received an enucleated specimen of the left eye from a case of a one year old child with leukocoria in both eyes since birth, the eye being enucleated following a clinical diagnosis of retinoblastoma. The gross specimen consisted of an eyeball measuring 2 x 1.8 x 1.5 cm. The cornea measured 1.2 x 0.9 cm. At the posterior end, soft tissue bit 2.0 cm long was present. Cut section showed reddish brown material with central tiny grey-white area of 0.8 cm diameter in the retrolental region.

On microscopy, portions of normal eyeball tissue were identified in the anterior segment. In addition, the section from the retrolental tissue showed swirls and whorls of uniform cells in multiple layers reminiscent of normal retinal epithelium. These cells had uniform round nuclei with speckled chromatin, scant cytoplasm and indistinct cell membranes. (Fig. 1) At several places these cells were arranged in rosette-like formations with central lumina typical of Flexner-Wintersteiner rosettes. (Fig. 2) No nuclear atypia was evident. No necrosis or calcification was noted. The skeletal muscle fibers and neural fibers from the posterior end of the specimen were within normal limits. Histological features were that of ‘Retinal Dysplasia’. A request for chromosomal study was made, but was declined by the patient’s parents. The other eye was left in situ.

Figure 1: Cells arranged in multilayered swirls and whorls (H&E, X100)
Retinal dysplasia presenting as leukocoria may be mistaken for retinoblastoma clinically. The routine microscopic findings may also simulate a retinoblastoma. Nevertheless, there are microscopic features that are distinctive enough to warrant a confident diagnosis of retinal dysplasia. The need to not misdiagnose this as a retinoblastoma is of extreme importance given the drastic contrast in prognosis between the two conditions.

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References

<table>
<thead>
<tr>
<th>Character</th>
<th>Retinoblastoma</th>
<th>Retinal dysplasia</th>
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<tbody>
<tr>
<td>Cells</td>
<td>Undifferentiated</td>
<td>Uniform</td>
</tr>
<tr>
<td>Mitoses</td>
<td>Frequent</td>
<td>Rare</td>
</tr>
<tr>
<td>Necrosis, calcification</td>
<td>Present</td>
<td>Absent</td>
</tr>
<tr>
<td>Rosettes (IHC)</td>
<td>Positive for cone opsin</td>
<td>Positive for rod opsin and Muller cells</td>
</tr>
<tr>
<td>Layers in rosettes (IHC)</td>
<td>No pattern</td>
<td>Reversal of normal pattern</td>
</tr>
</tbody>
</table>

Other common causes of leukocoria can be excluded on histology: Coat’s disease shows cholesterol clefts and foamy macrophages. Persistent Hyperplastic Primary Vitreous contains fibrovascular tissue and Retinocytoma is composed of numerous rosettes. [7]