

Case 5 (February 16th 2006): [N Engl J Med 2006;354:741-8.](#)

Case 5-2006 — An 11-Year-Old Girl with Loss of Vision in the Right Eye

Demographic: Female, 11 years, America North, General speciality

Clinical features extracted from the case and entered into the query box:

Decreased vision
Leukokoria
Posterior fibrovascular lesion
Neovascularization of iris
Retinal detachment

Synonyms used by Isabel (optional) for the above query:

Retinal Detachment

STOP !

Before you read further you might want to construct your own:

- Complete differential diagnosis
- Final diagnosis

In the following section you will find:

- The differential diagnosis constructed by the MGH panel
- The final diagnosis of the case
- Which of the MGH panel differential diagnoses did Isabel contain
- Did Isabel suggest the Final diagnosis

Differential Diagnoses considered by the MGH panel:

Retinoblastoma
Cataract
Ocular Toxocariasis
Coats Disease
Ocular Neoplasms

Final Diagnosis of the case according to NEJM:

Retinoblastoma

Differential Diagnoses of the case as given by Isabel:

Retinoblastoma – Isabel 1st page
Ocular Toxocariasis – Isabel 1st page
Coats Disease – Isabel 1st page
Ocular Neoplasms – Isabel 1st page
Cataract – Isabel 2nd page & on clicking on the speciality heading

Was the final diagnosis given by Isabel?

Yes, Retinoblastoma (Isabel 1st page)

Entire presentation cut and pasted into the Isabel query box:

Retinoblastoma – Isabel 1st page
Cataract – Isabel 1st page
Ocular Toxocariasis – Isabel 1st page

Isabel Differential:

<p>NEOPLASTIC DISEASES</p> <p>Retinoblastoma <input type="checkbox"/></p>	<p>OPHTHALMOLOGY</p> <p>Diabetic Eye Disease <input type="checkbox"/></p> <p>Persistent Hyperplastic Primary Vitreous <input type="checkbox"/></p> <p>Familial Exudative Vitreoretinopathy <input type="checkbox"/></p> <p>Uveitis <input type="checkbox"/></p> <p>Retinal Detachment <input type="checkbox"/></p> <p>Neoplasms of the Eye <input type="checkbox"/></p> <p>Coats' Disease <input type="checkbox"/></p> <p>Retinopathy of Blood Disorders <input type="checkbox"/></p> <p>Ectopia Lentis <input type="checkbox"/></p> <p>Coloboma of the Fundus <input type="checkbox"/></p> <p>↳ Refraction Abnormalities <input type="checkbox"/></p> <p>HSV Ocular Infections <input type="checkbox"/></p> <p>Cataract Disorders <input type="checkbox"/></p> <p>Retinitis Pigmentosa <input type="checkbox"/></p> <p>Hypertensive Retinopathy <input type="checkbox"/></p>
<p>INFECTIOUS DISEASES</p> <p>↳ Toxoplasmosis <input type="checkbox"/></p> <p>↳ Toxocariasis <input type="checkbox"/></p> <p>Brucellosis <input type="checkbox"/></p>	<p>CARDIOVASCULAR SYSTEM</p> <p>↳ Left-to-Right Shunt Lesions <input type="checkbox"/></p>
<p>DERMATOLOGY</p> <p>Pseudoxanthoma Elasticum <input type="checkbox"/></p>	<p>HUMAN GENETICS</p> <p>Down Syndrome/Trisomy 21 <input type="checkbox"/></p>
<p>METABOLIC DISEASES</p> <p>Homocystinuria <input type="checkbox"/></p> <p>↳ Urea Cycle Disorders <input type="checkbox"/></p> <p>Albinism <input type="checkbox"/></p>	
<p>NERVOUS SYSTEM DISORDERS</p> <p>Neurofibromatosis <input type="checkbox"/></p> <p>Brain Neoplasms <input type="checkbox"/></p> <p>Tuberous Sclerosis <input type="checkbox"/></p>	
<p>ORTHOPEDIC DISORDERS</p> <p>Marfan Syndrome <input type="checkbox"/></p>	