



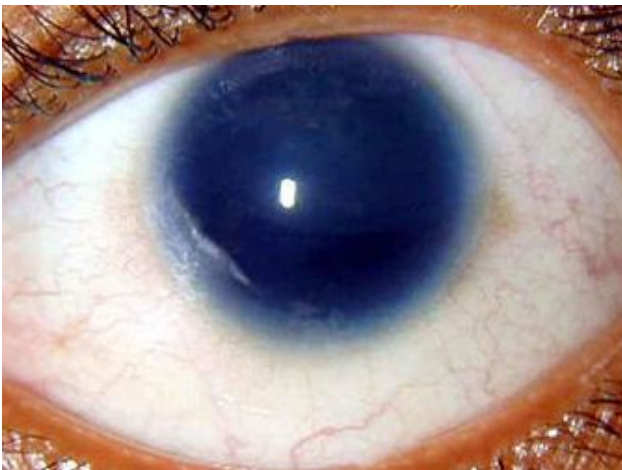
HOLY C.O.W.!

IT'S...

Clinical Question of the Week #5
August 4th, 2008 through August 11th,
2008

Please e-mail your answers to Kuo, Tim, Wendy, and Kevin (klian@mednet.ucla.edu; tprovias@mednet.ucla.edu; wsimon@mednet.ucla.edu; kbreger@mednet.ucla.edu) by 0800 on Monday, August 11th, 2008. The resident or intern with the most correct answers at the end of each month will receive a prize!

Case: A 24-year-old woman presents to you for evaluation after failing the vision test required for obtaining a California driver's license, despite scoring enough to pass the written DMV exam using her glasses. She's had regular ophthalmologic follow up since she was a child, although she says she's had to wear glasses for several years and has had problems with her eyes in the past. Her development was relatively normal and she played high school basketball without incident (they won the state super-sectionals) despite her old pediatrician telling her to "take it easy out there." Examination reveals a well developed, tall slender woman in no distress. Her eye exam revealed a visual acuity of 20/40 on the right, and 20/80 on the left (shown below). Extraocular motions were intact, and the remainder of her exam was notable only for flow murmur on cardiac auscultation.



Questions:

1. What is the diagnosis?
2. Name three other features of this condition.
3. What is the ophthalmologic finding depicted above?
4. What is the genetic basis of this condition, and who discovered it?