GEN59 - Ocular Phenotype in Marfan Syndrome

OBJECTIVE: Describe ocular phenotype (ectopia lentis, retinal detachment, cataracts) in Marfan syndrome.

ORGANIZATION

Lead Investigator: Jennifer Pardo Habashi, MD
Co-Investigators: Gretchen Oswald MS CGC
Funding Source: GenTAC and JHU

BACKGROUND AND RATIONALE


DESIGN

Method: Describe ocular phenotype in Marfan syndrome and compare to other disorders to show that it is a disease-specific process

Inclusion criteria:
• Subjects with confirmed MFS, LDS, vEDS, and Familial TAAD diagnosis.

Samples: None

Data:
• Organ system review
• Genetic
• Demographics
• Quality of life

CONCLUSIONS

Results: This review of ocular data within the GenTAC registry identifies a >50% prevalence of lens dislocation in MFS and a notable absence of lens dislocation in LDS as previously reported; therefore, presence or absence of lens dislocation is a useful diagnostic tool in differentiating patients. Furthermore, this data supports the theoretical risk that LASIK correction may increase the risk of retinal detachment in patients with connective tissue disorders however more detailed studies investigating the temporal relationship need to be done.