



GEN59 - Ocular Phenotype in Marfan syndrome

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

ORGANIZATION

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BACKGROUND AND RATIONALE

Ectopia lentis is a major characteristic in the diagnosis of Marfan syndrome. The seminal paper by Maumenee, et al in 1981 reported a prevalence of 60%. Other secondary complications include retinal detachment (10%), with a significant amount occurring in the few months post lens distraction surgery (Maumenee IH, et al. *Trans Am Ophthalmol Soc* 1981:79; 684-733). There has been a paucity of literature about ocular features of ectopia lentis and retinal detachment in that surgery, and clinical questions abound regarding risk of retinal detachment post surgery and/or age-related risk. More recent reports in the literature state widely varying ectopia lentis prevalences of 40-80% (Sultan G, et al. *In Ophthal & Vis Science* 2002:46(6); 1757-1764, Konradsen TR, et al. *Acta Ophthal* 2013).

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.

