Genetics in Glaucoma

CE-NY

November 8, 2015

Genes and their environmental triggers will become important as we look to better understand how to manage or even prevent glaucoma.

Exfoliative Syndrome and Exfoliative Glaucoma

The most common, identifiable cause of glaucoma

- Increased prevalence with age
- Higher incidence in some populations
- Aggressive glaucoma
- Complications in cataract surgery
- Association with systemic disease?
LOXL 1 Gene and Prevalence of ES

Iceland
LOXL1 gene variants
Cases = 98%
Controls = 80%
Prevalence of ES ~20% in people older than age 60

Australia
LOXL1 gene variants
Cases = 98%
Controls = 80%
Prevalence of ES ~1% in people older than age 60

Therefore, LOXL1 gene associated with condition BUT presence of gene variants not strongly correlated with prevalence of the disease


Geographic and Climatic Factors

Exfoliation Across the US

Conclusions:
Ambient temperature and sun exposure may be important environmental triggers of ES.

Discovery of environmental factors linked to ES could lead to primary prevention measures for this condition.

Arch Ophthalmol. 2011;129(8):1053-1060

Geographic and Climatic Factors

Results:
Compared with middle-tier residence, northern-tier residence (above 42°N) was associated with an increased hazard of ES of 114%

Compared with middle-tier residence, southern-tier (below 37°N) was associated with a reduced hazard of ES of 17%

For every 1° increase in January low temperature, the hazard of ES decreased by 3%

For every additional sunny day annually, the hazard of ES increased by 1.5% in locations with average levels of other climatic factors.

Arch Ophthalmol. 2011;129(8):1053-1060

Demographic and Geographic Features of Exfoliation Glaucoma

Female Nurses Health Study
78,955 women
Male Health Professionals Follow-up Study
41,191 men

Followed for more than 20 years
NHS: 288 cases
HPFS: 60 cases

Demographic and Geographic Features of Exfoliation Glaucoma

In this mainly white cohort from the United States:

Increasing age and female gender were significant risk factors for EG or EGS

However, Scandinavian heritage was not

Living in the middle or southern regions of the United States relative to living in the northern region was associated with a reduced risk of EG or EGS. Where you lived at age 15 and age 60 were particularly important.

Ambient temperature and sun exposure may serve as environmental triggers

Ophthalmology 2012;119:27–35

Exfoliation Risks (beyond Genetics)

Colder temperature and UV exposure

Increased risk with time spent outdoors

Risk increases based upon sunny days

Risk increases with lower temperatures

Risk increases with increased elevation

Conclusions and Relevance

Lifetime outdoor activities may contribute to XFS. The association with work over snow or water and the lack of association with brimmed hat wear suggests that ocular exposure to light from reflective surfaces may be an important type of exposure in XFS etiology.