The Impact of Genetics on AMD

Objectives:

With the aging of the population, Age Related Macular Degeneration (ARMD) is becoming more common in optometric offices. The truth of the matter is that most cases are dry AMD and not amendable to current injectable treatments. We need to be aware of all the possible treatment and prevention strategies. In order to be in tune to these goals, we need to be aware of the causative factors of AMD, including genetics. Genetics plays a role in not only who gets AMD, but also likely to contribute to how people respond to treatments.

Goals:
1. Discuss the basic demographics of ARMD.
2. Discuss treatment history of ARMD over time and how it has evolved today.
4. Primer on genetics to help understand pertinence to AMD
5. Genetics of AMD and role that genetics plays in disease.
6. Discuss pharmacogenetics in relation to ARMD nutritional products.
7. Discuss possible pharmacogenetics in relation to intravitreal injections for ARMD.
8. Treatment options and trials supporting their use.
9. Strategies to educate patients and how this education helps influence outcomes.

Outline:
Statistics of AMD
Definition of age related macular degeneration
   Diagnostic criteria-by clinical diagnosis
   Drusen
      Soft vs hard and location
   Atrophic AMD
      AREDS defined categories and treatment implications
      Importance of drusen size
         125 microns confers greater risk
AREDS report 36: using categories/characteristics to predict progression

Risk assessment according to phenotype category

By instrumentation

Drusen or RPE changes not evident by clinical exam

Dark adaptation

Science behind

Reality: device availability

Wet AMD

Historic treatment

MARINA/ANCHOR and other studies

Seven Year outcomes of Lucentis

Overall decline from baseline

Better than no Tx, but continual decrease in efficacy

Avastin

Eylea

VIEW1 and VIEW2

Differences in dosing and efficacy from Lucentis

CATT

How does this affect optometric practices

How does this affect optometric patients

Financial ramifications

Risk Factors for AMD

Non-modifiable

Age

Family history/genetics
Race
Modifiable
Macular pigment optical density
Diet
Nutrition via diet
Nutrition via supplementation
Lutein
Zeaxanthin
Mesozeaxanthin
Other important nutrients to be considered
Smoking
Macular Pigment optical density
What is its significance
How is it tested
Once tested to be low, what can be done about it
Genetics in AMD
Human Genome project and implications
Realilty of genetics in medicine today
Basics of DNA and Gene testing
Genetic testing in other areas of medicine
Pharmacogenetics in medicine
What is a "SNiP"
What SNP’s affect AMD and how can we check
Can some SNiP’s be protective
Testing options
Macula Risk
What genes are tested and what is the risk in each
Who should be tested and when
Sequenome
Other tests available or available soon
Implications of testing
Helping to determine likelihood of progression to visual significance
Affect on treatments and testing regimen based on risk
Helping to determine potential for response to nutrition
Awh paper from Ophthalmology
Description of paper
Potential positive take-aways
Potential controversy in statistics and analysis
Is it repeatable?
Genetic response in relation to Lutein
Wang et al: BMES and RS in Ophthalmology
Lutein and fish oil and genetic influence
Practical integration into clinical routine: when necessary
Diagnosis codes that allow for testing
Cost to patient and to practice of testing (assuming diagnosis codes)
Potential for genetic response to intravitreal (or other) treatments
Studies looking at this
Implications
Examples from other areas of medicine
Home testing for dry AMD
Amsler grid: potential sensitivity and specificity issues
PHP Home
PHP technology
Logistics of home testing
Remote monitoring with alerts with conversion to exudative AMD: sensitivity for RPE detachments

Benefits to patients: earlier detection of conversion

Other potential tests

Reality of computer / internet based tests

Putting it all together

Comprehensive strategy from “pre-AMD” through conversion

Integration of nutrition/vitamins into patient care

Using all genotype and phenotype information to make decisions

Importance of wording and actions taken

Treat “recommendations” as a “prescription”

Resources

Medical journals

Not just ophthalmic journals

Mainstream media

Stories about genetics make “news”

Patients will be aware, so we need to be

Book worth reading: Creative Destruction of Medicine by Eric Toprol