Retinitis Pigmentosa

Presentation by Dominic Fasula
Retinitis Pigmentosa: Background

- Progressive pigmentary retinopathy

- Retinal degenerative disease characterized by pigment deposits predominant in the peripheral retina and by a relative sparing of the central retina.
  - Primary degeneration of the photoreceptor rods, with secondary degeneration of cones.
  - Ring scotoma “tunnel vision”
  - Often develop cataracts
  - Macular edema

- Hereditary
  - Causative gene: Rhodopsin

- Mode of inheritance:
  - Autosomal (dominant/recessive)
  - X-linked
digenic
Retinitis Pigmentosa: Background

- Non-syndromic/syndromic
- The main risk factor is a family history of retinitis pigmentosa.
- Prevalence of non-syndromic RP 1:4,000
- Currently, no effective treatment for this condition
Symptoms

- Symptoms often first appear in childhood, but severe visual problems do not usually develop until early adulthood

- Functional signs
  - Decreased vision in low light or at night
    - “night blindness”
    - Earliest symptom
  - Loss of peripheral vision
    - Ring scotoma
    - “Tunnel vision”
  - Loss of central vision
  - Myopia
  - Inflammatory reactions
  - Often develop cataracts
  - Macular edema
Normal Vision vs RP
Screening for RP

- Color vision
- Examination of retina by ophthalmoscopy
- Fluorescein angiography
- Intraocular pressure
- Electroretinogram
- Pupil reflex response
- Refraction test
- Visual field test
- Slit lamp examination
- Visual acuity
Non-Syndromic

- Typical form

- Usually evolves over several decades
  - However, there are extreme cases:
    - a rapid evolution over two decades
    - Slow progression that never leads to blindness

- Disease course can be divided in three stages:
  - Early, Mid & End
Retinitis Pigmentosa: Early Stage.

- Main symptom: night blindness
- May be peripheral visual field defects in dim light but do not exist in day light
- Bone spicule-shaped pigment deposits not present or rare
- Visual acuity & Optic disc normal
- Attenuation of retinal arterioles modest
- *in most cases, ERG shows decreased amplitude in B wave*
The basic waveform of the ERG

Fig. 13. ERG recordings in a normal patient and one with retinitis pigmentosa.
Retinitis Pigmentosa: Mid Stage

- Night blindness apparent
- Patients become aware of loss of peripheral visual field in day light conditions
- Dyschromatopsia to pale colors
- Difficulties reading
- Presence of bone spicule-shaped pigment deposits in mid periphery, atrophy of retina
- Narrowing of retinal vessels
- Optic disc pale
- Visual field testing reveal mild scotomas
- Early cataract formation
Retinitis Pigmentosa: End Stage

- Patients no longer move autonomously
- Photophobia intense
- Widespread pigment deposits reaching macular area
- Vessels thin
- Optic disc has a waxy pallor
- Chorioretinal atrophy in periphery and in foveomacular area
Syndromic

- Many syndromes associate with various types of pigmentary retinopathies
- Usher syndrome
  - Most frequent syndromic form in which RP is associated with neuorsensory deafness
  - about 14% of all RP cases are Usher Syndrome
  - Bardet Biedl syndrome (BBS)
    - Less prevalent (1:150,000)
    - Digenic inheritance
- Renal abnormalities
- Dysmorphic syndromes
- Metabolic diseases
- Neurological diseases
Treatment

- No effective treatments for this condition
- Wearing sunglasses to protect retina from UV light may help preserve vision
- Clinical trials suggest treatment with Vitamin A and omega-3 fatty acid may slow progression
Future Treatments

- Future treatments may involve:
  - Retinal transplants
  - Stem cells
  - Gene therapy
  - Nutritional supplements
  - Drug therapies
Future Treatments: Current Research

- **2006**: Stem cells: UK Researchers working with mice, transplanted stem cells which were at an advanced stage of development into mice that had been genetically induced to mimic the human conditions of RP and AMD.

- **2008**: Scientists at the Osaka Bioscience Institute have identified a protein, named Pikachurin, which they believe could lead to a treatment for RP.

- **2010**: A possible gene therapy seems to work in mice.
References


http://www.ojrd.com/content/1/1/40


http://www.sciencemag.org/content/early/2010/06/24/science.1190897