Erythropoietic Protoporphyria

Maddy Scannell
What is it?

- shortage of ferrochelatase
- buildup of protoporphyrin
- sensitivity to sunlight
- UVA (longer wavelengths)
- origin of vampire legends
Acquisition

- FECH gene underexpresses ferrochelatase
- all races, both male and females
  - common in East Asians
- usually begins in childhood
- 1/50,000 people
Diagnosis

- symptom based
- test for levels of protoporphyrin (blood or stool)
Symptoms

- abnormal sensitivity to sunlight
  - burning, swelling
  - hours to resolve
- variation between individuals
- common on tops of hands, feet, face, ears
Effects

- serious impact on quality of life
  - social exclusion
  - depression, anxiety
- higher unemployment rate
- small scars, skin thickens/reddens
- hepatotoxicity
Treatment

- symptomatic relief
- avoid sunlight, tint windows, wear protective clothing
- sunscreen
- cysteine, beta-carotene, cholestyramine (photosensitivity)
- narrowband UVB phototherapy (skin thickening)
Progression

- sensitivity in infancy/childhood
- visible skin lesions ensue only after long or multiple exposures to sunlight
- chronic conditions may develop, liver toxicity
- not deadly