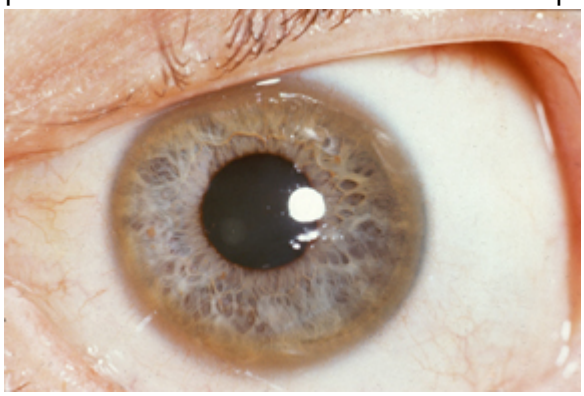
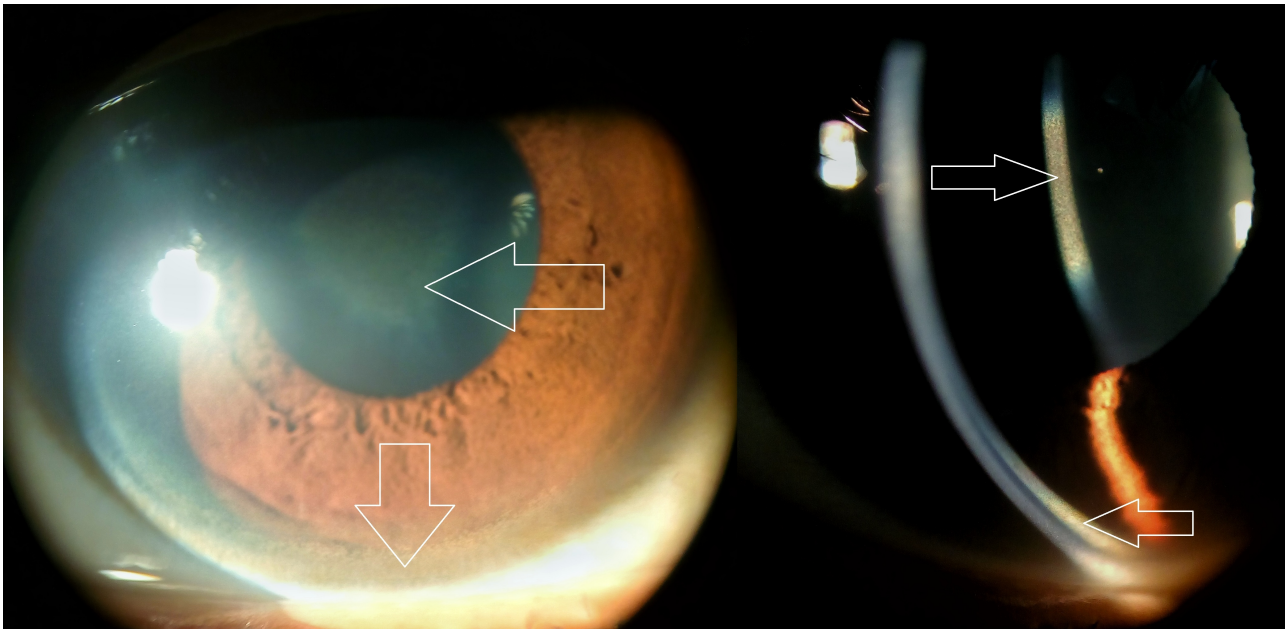
Wilson Disease

Main Features

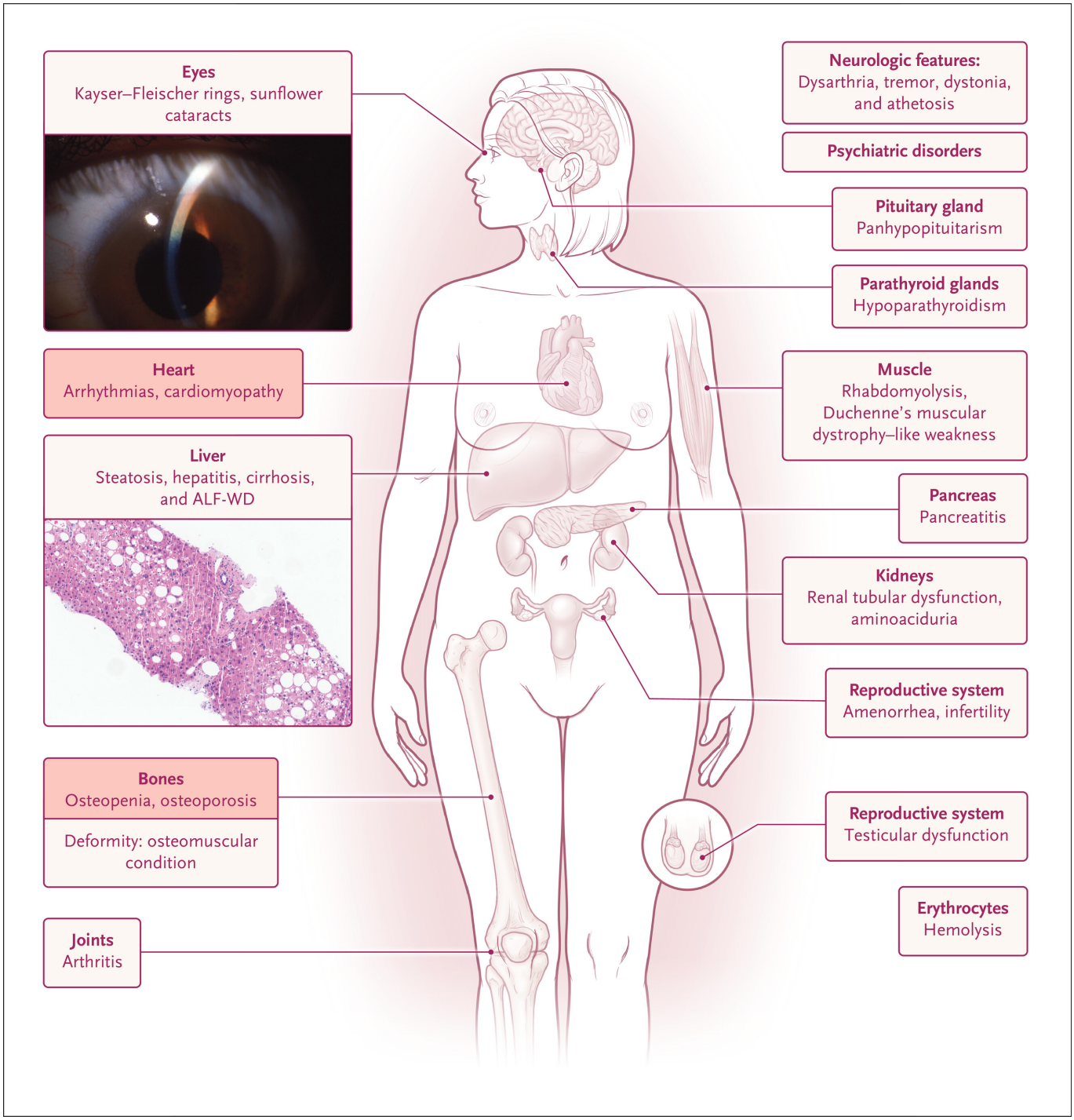
- Hepatic disease due to excess accumulation of copper
- Neurologic impairment from copper overload
- Laboratory findings
  - Low serum ceruloplasmin levels <14 mg/dl
  - 24 hour urine copper excretion >40 µg
- Treatment involves reducing copper concentrations with penicallamine or Tridentine and liver transplant

Eye Findings

- Kayser-Fleischer rings
  - copper deposition in Descemet's membrane
  - present in about 50% in those with hepatic disease
  -  from https://en.wikipedia.org/wiki/Kayser–Fleischer_ring#/media/File:Kayser-Fleischer_ring.jpg
- Sunflower Cataracts- radiating multicolored central opacities, less common

- 
  from https://en.wikipedia.org/wiki/Kayser–Fleischer_ring#/media/File:KF_ring_and_Sunflower_cataract.jpg
- Possible mild retinal neurodegeneration
  - thinning of the retinal nerve fiber layer
  - delayed VEPs
- Gaze palsy, especially up gaze

Other Findings



From Current and Emerging Issues in Wilson's Disease, Roberts EA, Schilsky ML, 2023 NEJM

Etiology

- Autosomal recessive disorder of copper metabolism from mutations in the ATP7B gene leading to impaired copper secretion
- ATP7B encodes for a copper-transporting ATPase
- 1:40,000 to 1:50,000

Reference

- Teschke R. et al. Wilson Disease: Copper-Mediated Cuproptosis, Iron-Related Ferroptosis, and Clinical Highlights, with Comprehensive and Critical Analysis Update. Int.J.Mol.Sci.2024;25:4753
- Wikipedia:Kayser-Fleisher ring
- Albrecht et al. Retinal neurodegeneration in Wilson's disease revealed by spectral domain optical coherence tomography. PLoS One 2012;7(11):e49825

syndrome