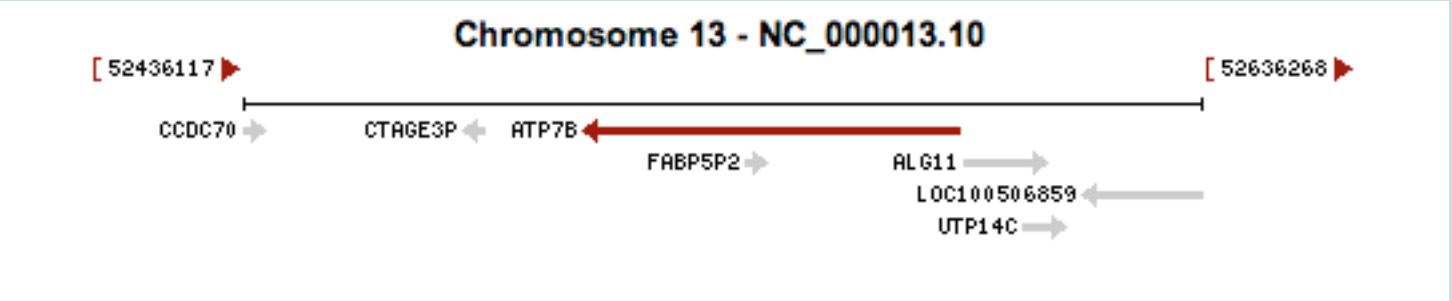
Wilson's Disease (Hepatolenticular Degeneration)

MEREDITH LEHMANN



Effects

- Liver and nervous system
- Copper is absorbed and retained at too high levels
- Symptoms typically begin to show under age 40, and can show as early as 4 years old.

Symptoms

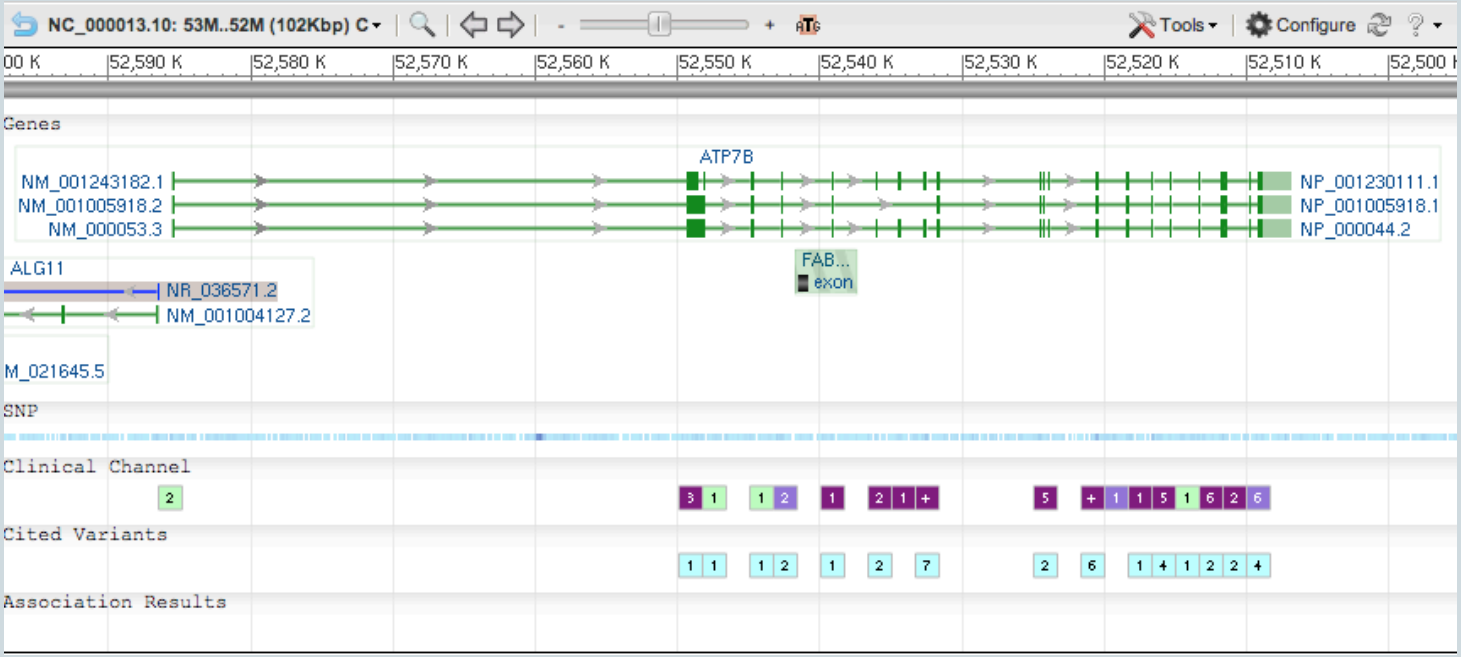
- Abnormal posture of arms and legs
- Confusion or delirium
- Dementia
- Difficulty moving arms and legs, stiffness
- Difficulty walking (ataxia)
- Emotional or behavioral changes
- Enlargement of the abdomen (abdominal distention)
- Personality changes
- Phobias, distress (neuroses)
- Slow movements
- Slow or decreased movement and expressions of the face
- Speech impairment
- Tremors of the arms or hands
- Uncontrollable movement
- Unpredictable and jerky movement
- Vomiting blood
- Weakness
- Yellow skin (jaundice) or yellow color of the white of the eye (icterus)

Classical Diagnosis

- Symptoms can arise as early as childhood
- Blood tests
- Urine test
- Eye test
- Liver biopsy
- MRI/CT scans

Genomics and Diagnosis

- The gene which causes Wilson's disease, ATP7B, has over 200 discovered mutations that can cause Wilson's disease.



Classical Treatment

- Diet of foods with low copper levels
- Medications that bind to copper and push it out with urine
  - Penicillamine
  - Trientine

Modern treatment

- Some patients have received liver transplants
- Tetrathiomolybdate, a currently experimental treatment, has been helpful to some patients

Sources

- OMIM, http://omim.org/entry/606882
- Wilson's Disease Association, http://www.wilsonsdisease.org/wilson-disease/wilsondisease-treatment.php
- NCBI Gene, http://www.ncbi.nlm.nih.gov/gene/540
- PubMed Health, http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001789/