In-service on Wilson's Disease

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Facts about Wilson's Disease....

- Wilson's Disease is also called hepatolenticular degeneration.
- It is a rare, autosomal recessive inherited disease (occurs equally in men and women).
- It causes an accumulation of copper in tissues all over the body, but mainly in the liver, brain, kidneys and cornea.
- Copper is usually excreted in bile.
- Accumulation starts in the liver – once the liver is injured, copper spills into the bloodstream affecting other organs and tissues.
- Mostly occurs in young patients ages eight to 20 years, but can occur up to age 40. In children, symptoms usually start around age four.
- Liver disease occurs in 50% of the patients – can be seen as acute hepatitis, chronic active hepative hepatitis, cirrhosis or fulminant hepatitis.
- Affects one in a million people, mainly Europeans, Sicilians and southern Italians.
- Wilson's Disease is always fatal if not diagnosed and treated.
- Liver disease is most common in children and neurological disease is most common in young adults.
- The gene associated with Wilson's Disease is called ATP7B.
- Wilson's Disease affects chromosome 13.

Diagnosing Wilson's Disease

- Diagnostic triad includes Kayser-Fleischer rings – low serum ceruloplasmin <20 mg/dl and increased amounts of liver and urine copper levels.
- Blood tests, ceruloplasmin, urine copper, eye test for Kayser-Fleischer rings and liver biopsies are used to confirm diagnosis.
- If there is an acute liver injury – this causes hemolytic anemia due to acute release of copper into the bloodstream. Usually seen in fulminant hepatitis.
- If the disease is in a chronic state, then copper accumulates in the brain.

Signs and symptoms

Neurological – resting and intention tremors usually in arms and hands, spasticity, rigidity, chorea.

Dystonic – slowness of speech, unsteady gait, dystonic faces, posturing, difficulty swallowing.

Kayser-Fleischer ring – this is caused by copper deposits on the cornea – greenish or golden brown rings are seen.

Psychiatric – psychosis, neurosis, homicidal/suicidal behaviour, depression, aggression.

Other – may include jaundice, vomiting blood, abdominal swelling.

In women – may experience menstrual irregularities, absent periods, infertility or multiple miscarriages.

Treatment

Medication – must take oral medication for the rest of their lives. If medication is stopped, the copper will accumulate and symptoms will recur usually within three months. Medication choices include: **Penicillamine (Cuprimine, Depen)** – the response on this medication is slow, usually up to one year for maximum effects. This medication binds the copper and increases urinary excretion. **Trientine Hydrochloride (Syprine)** – this medication removes copper from tissues, binds copper and increases excretion in urine. **Zinc Acetate (Galzin)** stops intestines from absorbing copper and promotes copper excretion.

Diet – patients need to take Vitamin B6 and require a low-copper diet. This includes no mushrooms, nuts, liver, chocolate, shellfish and dried fruits.

For more information about Wilson's Disease, feel free to visit the following websites:

http://www.medstudents.com/metdis/metdis2.htm
http://www.1uphealth.com/health/wilson_disease_info.html
http://digestive.niddk.nih.gov/ddiseases/pubs/wilson/index.htm
http://www.ncbi.nlm.nih.gov/disease/Wilson.html
http://www.medhelp.org/wda/lit_what.htm