

Metabolic disorders

| Reye's syndrome <i>Extremely rare</i> | Wilson disease autosomal recessive ATP7B gene | Hemochromatosis autosomal recessive HFE gene at the short arm of chromosome 6 |
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| Affect <u>brain & liver</u> | <u>Prevents the body from removing extra copper</u> | <u>Absorbing more iron (5 to 20 times than the normal)</u> excess iron is stored in <i>liver, heart, pancreas</i> |
| Most commonly in children (4-14) in recovery <u>from viral infection</u> using aspirin in treating viral infection can cause Reyes syndrome | About 1 in 40,000 people get Wilson disease. <u>equally in men and women</u> , Symptoms usually appear <u>between ages 5 to 35</u> | The HFE protein regulates the production of a protein called hepcidin manufactured in the liver, and it determines how much iron to absorb & release <u>The most known mutation of HFE is C282Y</u> |
| Mitochondrial dysfunction This leads to <u>cerebral edema and increased intracranial pressure (ICP)</u> | Kayser-Fleischer rings is the most unique sign of the disease as a result from buildup of copper in the eyes. They appear as a rusty-brown ring around the edge of the iris and in the rim of the cornea In Liver or spleen: <u>swelling, yellowing of the skin and whites of the eyes</u> . Rarely, acute liver failure In CNS: problems with <u>speech, swallowing, or physical coordination</u> | Phlebotomy , which means <u>removing blood the same way it is drawn from donors at blood banks</u> . |
| No cure , But can be managed by protecting the brain against irreversible damage (by reducing brain swelling, preventing complications in the lungs, and anticipating cardiac arrest) | initially : The removal of excess copper using drugs like (d-penicillamine and trientine) , reduction of copper intake, treating of any liver or central nervous system damage. | Initially : you may have a pint (about 470 milliliters) of blood taken once a week |

