Menkes Disease

Menkes disease is caused by a gene that is flawed. This gene regulates the metabolism of copper in a persons body. Copper is lacking in most tissues in people with this disease. The lack of copper causes structural changes in the hair, brain, bones, liver, and arteries. Brain abnormalities may happen. Weakened bones (*osteoporosis*) may result in fractures. Menkes disease mostly affects males. Affected infants may be born to early (*prematurely*). In some cases, normal or slightly slowed development may proceed for 2 to 3 months.

SYMPTOMS

- Flabby muscle tone.
- Seizures.
- Growth failure (*failure to thrive*).
- Below normal body temperature.
- Abnormal hair that is often white, ivory, or gray in color and is kinky, tangled, sparse, steely and easily broken.

TREATMENT

Early treatment with injections or oral copper supplements may be of some benefit. Other treatment is being supportive of the person and also depends on the symptoms.

WHAT WILL HAPPEN

The prognosis for people with Menkes disease is poor. Most affected persons die within the first decade of life.

Document Released: 12/08/2003 Document Re-Released: 03/26/2009
ExitCare® Patient Information ©2011 ExitCare, LLC.