Menkes disease is a genetic disorder of copper metabolism that is detectable before birth (prenatally) and which follows a progressively degenerative path involving several organs of the body but especially the brain. It is characterized by seizures, mental retardation, stunted growth, failure to thrive, unstable body temperature, and very unusual color and texture of hair. Menkes disease is characterized by brittle, tangled, sparse, steely or kinky hair that is often white, ivory, or gray in color. Unusual facial features include pudgy cheeks and abnormal eyebrows. Affected infants are often born prematurely. Lower than normal body temperature (hypothermia) and excess bilirubin in the blood (hyperbilirubinemia) may occur causing a yellow appearance (jaundice). Hypothermia may also occur in older infants. In some cases, early symptoms may resolve, and normal or slightly slowed development may proceed for two to three months. Severe developmental delay, loss of early development skills, and convulsions may occur. Brain abnormalities such as a blood clot at the base of the brain (subdural hematoma) and/or rupture or thrombosis of arteries in the brain not infrequently occur. Spastic dementia and seizures may eventually arise. Weakened bones (osteoporosis) are common and can result in fractures. The combination of subdural hematoma and bone fractures may lead to an incorrect diagnosis of child abuse. Emphysema, bladder abnormalities, degeneration of the retina and cysts of the iris have also been described. In rare cases, symptoms are very mild and only a few typical symptoms may appear. Recent studies suggest that the incidence of Menkes disease ranges from about 1 in 100,000 live births to 1 in 250,000 live births. Menkes disease is diagnosed by measurement of a decreased amount of copper and ceruloplasmin in blood plasma but these tests are not always reliable in the newborn period. A new method of diagnosis that can potentially identify affected infants before copper deficiency affects the brain involves measurement of catecholamine levels in blood plasma. Molecular genetic testing for mutations in the APT7A gene is available to confirm the diagnosis. Carrier testing and prenatal diagnosis are available if a specific ATP7A mutation in an affected family member.