Menkes syndrome, also known as kinky-hair disease, is a rare X-linked recessive, lethal, neurodegenerative disorder of impaired copper transport. The disorder typically is characterized by fine, hypopigmented, wiry hair; doughy skin; bone and connective-tissue disturbances; vascular abnormalities that can result in spontaneous hemorrhaging; and progressive neurologic deterioration. These early findings often are easily confused with child abuse. We report a case of a 6-month-old boy with Menkes syndrome whose symptoms originally were thought to be from child abuse.

Menkes syndrome is a rare multisystemic disorder of impaired copper transport and metabolism. Boys typically present in the first few months of life with symptoms such as failure to thrive, lethargy, hypothermia, hypotonia, seizures, mental and motor retardation, osseous alterations, anemia, intracranial and retinal hemorrhages, and/or other neurologic abnormalities.1-3 We report a case of an infant with Menkes syndrome whose symptoms originally were thought to be from child abuse. Menkes syndrome should be considered when evaluating children with recurrent intracranial hemorrhages and suspected child abuse.

Case Report

A 6-month-old boy presented with acute-onset respiratory distress. He was hypertensive and cyanotic with bulging fontanelles; sluggish pupils; and dysmorphic features such as low-set ears, hypotelorism, polydactyly, and syndactyly. Brain imaging revealed large, bilateral, subacute, subdural hematomas; an acute, small, left-sided occipitoparietal subdural hematoma; and diffuse edema, suggestive of bilateral shearing injuries at the grey-white matter junction. Ophthalmologic examination showed left-sided retinal hemorrhages. The baby’s overall presentation was highly concerning of nonaccidental injuries from shaken baby syndrome; he had already been placed in foster care due to concerns of parental alcohol and drug abuse as well as witnessed reckless parental behavior that endangered his safety. He was born at 36 weeks’ gestation with suspected maternal drug use during pregnancy. His medical history was notable for temperature instability, gastroesophageal reflux disease, laryngotracheomalacia, reactive airway disease, syndactyly of the toes, joint contractures, polydactyly with excision of an extra digit shortly after birth, and a right inguinal hernia. Because of the need for immediate neurosurgical intervention on arrival to the hospital, his scalp hair was shaved for shunt placement, thereby removing any clue of a hair shaft abnormality.

As the hair regrew, it was noted to be somewhat kinky, hypopigmented, and thin (Figures 1 and 2). Dermatology was consulted and a hair sampling was done, which showed characteristic 180° twists of hair consistent with pili torti (Figure 3). The serum copper level was 13 μg/dL (reference range, 38–104 μg/dL) and ceruloplasmin was less than 10 mg/dL (reference range, 15–30 mg/dL). These laboratory findings in combination with his physical examination were presumptively diagnostic of Menkes syndrome. An ATP7A mutation found on genetic analysis further supported the diagnosis. He was treated with subcutaneous injections of copper chloride 200 μg twice daily to prevent further neurologic deterioration.
Menkes syndrome is an X-linked recessive disorder caused by mutations in ATP7A (Xq13.3), a gene encoding the copper-binding enzyme adenosine triphosphatase, that leads to defective copper transport and metabolism with subsequent low levels of serum copper.\textsuperscript{1-3} Menkes syndrome is known to mimic child abuse, specifically shaken baby syndrome. It is exceedingly rare, but 2 cases of a similar nature have been reported in the literature since 1974.\textsuperscript{4,5} The most important dermatologic sign of Menkes syndrome is the structural abnormalities of the hair. The most common hair abnormality is pili torti (180° twists of hair). Other hair abnormalities include segmental shaft narrowing (monilethrix) and brushlike swelling of the hair shaft (trichorrhexis nodosa).\textsuperscript{2}

Clinically there are no hair abnormalities at birth, but the hair appears hypopigmented, lusterless, and sparse in density on regrowth, and looks and feels similar to steel wool. The clinical features, low serum levels of copper and ceruloplasmin, and microscopic hair shaft findings confirm a diagnosis of Menkes syndrome.\textsuperscript{2,6} Central nervous system findings include lethargy, seizures, mental and motor retardation, hypotonia, and hypothermia.\textsuperscript{3} Brain imaging reveals early-onset extensive lesions in white matter extending to the cortical fibers. In more advanced stages, generalized atrophy, subdural hematomas and

![Figure 1. Hair regrowing on the scalp (A and B).](image1)

![Figure 2. Thin, kinky, hypopigmented hair regrowing on the scalp (A). The eyebrows also demonstrated kinky hypopigmented hairs (B).](image2)

![Figure 3. Characteristic 180° twists of hair consistent with pili torti.](image3)
hygromas, and tortuosity of vessels are noted. The majority of children with Menkes syndrome die by 3 years of age with progressive deterioration before death; however, early detection allows for genetic counseling and the institution of copper histidine therapy, an investigational treatment that has shown promising results in some infants if initiated early. The only trials for copper histidine therapy that are available at this time are for asymptomatic infants with no neurologic involvement (http://clinicaltrials.gov; NCT00001262), which would exclude our patient.

Although Menkes syndrome is rare, it should be considered when evaluating a child with recurrent intracranial hemorrhages and suspected child abuse. Particular diligence is needed if the hair is shaved for a neurosurgical procedure, thereby removing the most telltale clue of Menkes syndrome.

REFERENCES