Copper Metabolism in the Steely-Hair Syndrome

Key Words: kinky-hair syndrome, steely-hair syndrome, copper metabolism, copper deficiency

Menkes and co-workers\(^1\) described (in 1962) children with developmental regression, seizures, temperature instability, scurvy-like bone changes, and a peculiar steel-like hair. Danks and his co-workers\(^2\) reported a defect in copper absorption.

Lott et al.\(^3\) investigated the copper metabolism in an 11-month-old male with this syndrome. Serum copper was 15 \(\mu\)g per 100 ml and serum ceruloplasmin undetectable. Less than 1 percent of orally administered \(^{64}\)Cu was absorbed. This patient was given supplemental copper as cupric sulfate in two divided doses totalling 0.52 mg per kilogram per day. Within seven days of such supplemental oral copper, the serum copper rose three-fold (from 15 \(\mu\)g per 100 ml) and remained at approximately 75 percent of the normal concentration for the remaining 22 days of therapy. After the end of supplementation, the serum copper returned to baseline in about ten days. Although no oxidase activity could be found indicating ceruloplasmin, all samples did contain an immunoprecipitation band, although in a reduced amount. It is concluded that the copper absorption block is only partial in the Steely-hair syndrome and is overcome by high doses of oral supplements.


Alpha-1-Antitrypsin Deficiency—Liver Disease

Key Words: cirrhosis, sialic acid, cell transport, alpha-1-antitrypsin, antitrypsin sialyltransferase

Eriksson and Larsson\(^1\) purified the PAS-positive inclusion bodies from cases of cirrhosis associated with homozygous alpha-1-antitrypsin deficiency. The main component was a protein of the same molecular size as the serum alpha-1-antitrypsin and has immunological similarity. Chemical analysis of this liver material showed a complete absence of sialic acid, as a major difference with the normal serum protein.

It is now established that deficiency of serum alpha-1-antitrypsin is associated with some cases of chronic obstructive pulmonary disease and some cases of fatal cirrhosis in the perinatal or juvenile period. More recently, hepatocellular carcinoma has been associated with a partial deficiency.\(^2\) Although the pulmonary damage is postulated to represent the uninhibited