CASE REPORTS

MENKES' DISEASE (KINKY HAIR DISEASE)

P S Parihar, D C Sharma, S Ramakrishnan and D Kanther

A six-month-old child presented with feeding difficulties, generalised seizures, delayed milestones, pudgy cheeks hypopigmented, sparse and steely wool hair. Serum copper level was 6.0 mg/dl.

Key words : Menkes' disease.

First described by Menkes et al,[1] the characteristic hair changes led O' Brien and Sampson[2] to coin the term kinky hair disease for this X-linked recessive syndrome which is characterized by slow growth, typical hair changes (Pili torti, monilethrix and trichorrhexis nodosa), hypothermia, progressive cerebral degeneration manifesting as irritability, generalised or focal seizures and intracranial hemorrhage, with death before 3 years in most of the cases. These children usually have pudgy cheeks with lack of expressive movements, occasionally with thick and relatively dry skin. The syndrome involves defective copper metabolism with low serum copper and ceruloplasmin levels.[3-5] A few cases have been reported from India also.[6-8] We herein report such a case observed by us.

Case Report

This 6-month-old male child was born to a 20-year-old mother, out of a non-consanguineous marriage. The child was apparently well till three months of age and had acquired social smile and head control. Then onwards, he gradually became increasingly lethargic, uninterested in his surroundings and developed feeding difficulties. He also had four generalised tonic-clonic seizures in the last two months. There was no history of fever, rash or drug intake antenatally by the mother, birth asphyxia or birth trauma. Postnatal period was uneventful. His elder brother died three years ago and had similar complaints and the same hair changes as told by the parents. At 6 months, the child was 5.4 kg with normal temperature, normal anthropometry, pudgy cheeks and hypopigmented, sparse, steely wool hair (Fig. 1). Skin was normal. There was generalised hypertonia. Microscopically, the hair revealed monilethrix and pili torti. Serum copper level was 6.0 mg/dl (normal 22-30 mg/dl). Serum ceruloplasmin could not be done because of lack of facility.

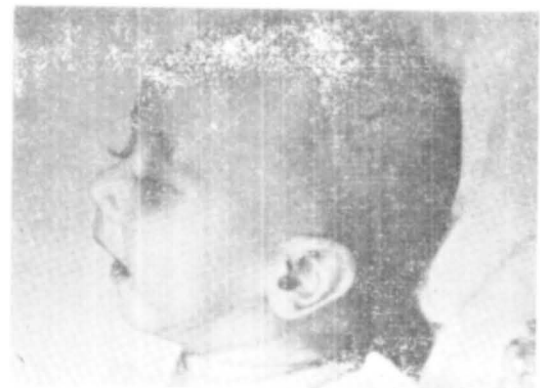

Fig. 1. Sparse, kinky hair and pudgy cheek.

From the Primary Health Centre, Palodara, District Udaipur-313 001 (Rajasthan), India, and the Department of Dermatology, Venereology and Leprology, RNT Medical College, Udaipur, India.

Address correspondence to : Dr P S Parihar, C/o Dr D C Sharma, 123, Kharol Colony, Fatehpura, Udaipur-313 001 (Rajasthan), India.

Skiagrams of long bones revealed scurvy-like changes.

Comments

This child had history of delayed social and motor milestones with neurological abnormalities in the form of lethargy, irritability and seizures. Presence of typical hair changes, pudgy cheeks, generalised hypertonia with low serum copper level and scurvy like bone changes on skiagrams, supported the diagnosis of Menkes' disease.

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