PAEDIATRIC DERMATOLOGY

CO-EXISTING NEUROFIBROMATOSIS TYPE I AND WILSON DISEASE A RARE CASE

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Background: Type 1 neurofibromatosis (NF-1) is one of the most common genetic disorders, with dominant autosomal inheritance. Wilson disease is an autosomal recessive inherited disorder of hepatic copper metabolism resulting in the accumulation of copper in many organs and tissues.

Observation: A case of a girl who had features of both Type I neurofibromatosis and Wilson disease. A 10-year-old girl with hyperpigmented macules and skin colored nodules. On examination, we found more than 6 cafı-au-lait spots (most > 1.5 cm) with axillary and palmar freckling and more than 2 neurofibroma . On ophthalmological examination showed KF ring and lisch nodule. Routine blood tests were normal and brain MRI showed altered signal intensity in bilateral lentiform nucleus and bilateral thalamus and mid brain and 24 hours urinary copper level are raised and ceruloplamin levels are low. These finding were consistent with diagnosis of Wilson disease and this patient also presented fulfilled the criteria of NF1.

Key message: It is important for the physician to be alert to the characteristic signs and symptoms of NF-1 since several complications affecting life expectancy may occur in these patients, such as association with neoplasia and coexistence with other genetic diseases.