A CRITICAL EVALUATION OF COPPER METABOLISM IN INDIAN
WILSON'S CHILDREN WITH SPECIAL REFERENCE TO THEIR
PHENOTYPES AND RELATIVES. R. PRASAD, G. KAUR AND S. N. S. WADIA.
DEPARTMENT OF BIOCHEMISTRY AND PAEDIATRICS, PIMER,
CHANDIGARH, INDIA

WILSON'S DISEASE IS AN AUTOSOMAL RECESSIVE DISORDER OF
COPPER ACCUMULATION RESULTING FROM A DELETION TO A
HOMOZYGOTE'S DNA, LEADING TO INCREASED COPPER DAMAGE, SERUM COPPER AND CERULOPLASMIN IN CONTROL
SUBJECTS (141 CASES OF DIFFERENT TYPES OF LIVER CIRRHOSIS) WHO
WAS SIGNIFICANTLY HIGHER THAN COMPARABLE WILSON'S DISEASE
(51) AND THEIR RELATIVES (50) WHO HAD HEPATOCOPEXION OF COPPER AND C-AMP WERE SIGNIFICANTLY
(P<0.01) ELEVATED IN WILSON'S CHILDREN ASSOCIATED WITH RENAL
TUBULAR ACIDOSIS. THEREFORE, 13 CASES OF WILSON'S WERE CONFIRMED BY
MEASURING HEPATIC COPPER (0.5-6.5 ug WET TISSUE; MEAN + SD). DURING THE FAMILY SCREENING BY SERUM COPPER,
CERULOPLASMIN AND URINARY COPPER AND HEPATIC COPPER, 10 SUBJECTS WERE TREATED TO PREVENT PRESYMPTOMATIC WILSON'S
DISEASE. THESE SUBJECTS WERE THEN STARTED THE D-PENCILLAMINE THERAPY, BECAUSE PRESYMPTOMATIC TREATMENT
PREVENTS PROGRESSION OF THE DISEASE AND ITS COMPLICATIONS.

Identification of two novel polymorphisms in the glucocerebrosidase
gene region. E. K. Lau, S. Winfield, G. Ambrose, A. M. Ziman, N. Tayebi, and E. I. Ginz1, Clinical Neurology Branch, IRP,
NIMH, NIH, Bethesda, MD; 2Shaare Zedek Medical Center, Jerusalem, Israel.

Niemann-Pick disease Type C (NPC) a lipidosis, is caused by a unique
behavioural defect in the enzymatic protein manifestations of the
condition often cause diagnostic confusion in the early stages. We present 3
cases highly suggestive of NPC, and that may be diagnostically useful in
Gaucher patients, and that may be diagnostically useful in
Gaucher patients.

In conclusion, these cases illustrate the diverse, but common presentations of a rare disorder. Pulmonary manifestations (as in case 3) are rarely described in classical NPC, but they have been reported in late-life NPC.

In each of the three cases the definitive diagnosis was established by demonstration of impaired cholesteric esterification in skin fibroblasts. In conclusion, these cases illustrate the diverse, but common presentations of a rare disorder. Pulmonary manifestations (as in case 3) are rarely described in classical NPC, but they have been reported in late-life NPC.

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