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Significance of early diagnosis of presymptomatic wilson disease: Case study

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Background: Wilson disease is an autosomal recessive inherited disease due to mutation in (ATP>B) gene, 50% of the patients presented with neuropsychiatric manifestations.

Methods & results: We have two Egyptian male siblings with +ve consanguinity, the elder one presented at age of twelve with involuntary movements choreoathetotic then aphasia. He was diagnosed as Wilson disease after 3 years from the onset, he was presented with Kayser-Fleischer ring, high copper in urine and a very low ceruloplasmin enzyme, the patient took penicellamine oral treatment with no response, then he took tetrathimolybdate but no response, he died after 8 years.

The second brother showed very low ceruloplasmin level and he started oral zinc treatment at 8 years, he still takes oral zinc and at sometimes he was non-compliant to zinc, he suffered from tumors in his upper limbs distal more than proximal and slurred speech, he took penicellamine treatment with zinc, the speech is improved, the patient now 28 years old, he goes to his work regularly, married and has one offspring.

Conclusion: Wilson disease is a treatable disease, but it must be discovered very early and managed at once

Biography

Suzette Ibrahim Helal is the assistant prof. of neurology, department of children of special needs, National Research Center, Cairo, Egypt. She has published various papers in reputed journals

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