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Atypical case of Wilson's disease without early GI symptoms

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Case presentation: A 19-year-old woman was brought to the emergency department of the Dr Babasaheb Hospital, Kandivli with history of pain and weakness in the lower limbs.

Discussion: Wilson's disease is a rare genetic disorder with an approximate incidence of 1 in 30,000 individuals. [1] The signs and symptoms of this disease is often revealed during the teenage years and up to 40 years. In this condition, there is an accumulation of excessive amounts of copper in the body, especially in the liver, brain, and cornea. Chronic liver disease, hemolytic anemia, or neuropsychiatric disease such as movement disorders, tremors, incoordination, and behavioral changes are the frequent signs and symptoms of the illness (Box 1).23 Sternlieb's criteria are often applied for diagnosing Wilson's disease (Box 2).2 However, these criteria are fulfilled in the advanced stage of the disease. Initially, the patient presented with prominent neuropsychiatric symptoms and the ophthalmic examination revealed K-F rings. The MRI, ophthalmic findings and neuropsychiatric symptoms in this patient was suggestive of Wilson's disease. The choice of treatment and drug dosage will usually depend on the stage or severity of disease (ie., based on the laboratory or histological evidence of aggressive inflammatory injury, neurologic or hepatic impairment). Initial treatment in symptomatic patients is with chelating agents (D-penicillamine or Trientine). In some cases, zinc alone will suffice but if required, it can be administered along with chelating agents as both have diverse mechanism of action.

Conclusion: Wilson's disease is a rare genetic disorder. A confirmed diagnosis of Wilson's disease is easily made in patients with cirrhosis, neurological manifestations, and K-F rings. In general, the neurological and K-F rings appear in the later stage of the disease and in such cases, combined results of clinical findings and biochemical

Biography

Dr. Priya Bharat Mahale, MD, is a distinguished Consultant Physician specializing in Internal Medicine, currently practicing at PRIME Medical Center, Jumeirah, Al Safa 1, Dubai, UAE. She earned her MBBS degree from K.J. Somaiya Medical College & Research Centre, Mumbai, India, in 2005. With over 15 years of clinical experience, Dr. Mahale has served in several reputable healthcare institutions. She began her career as a Consultant Physician at Mangal Murti Hospital, Mumbai, from 2009 to 2012, gaining valuable experience in managing a variety of medical conditions. In 2012, she moved to Dubai and worked as a Senior Registrar in Internal Medicine at Saudi German Hospital. Since 2018, she has been associated with Zulekha Hospital in Dubai and Sharjah, before taking up her current role at PRIME Medical Center. Dr. Mahale is known for her dedication to patient care and her expertise in managing chronic and acute internal medical conditions

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