TEACHING FILE

Wilson's disease

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Case Report: - A 14 years old girl presented with increased salivation, tremors and spilling of feeds for the past 2 months. She is not able to walk without support and has involuntary movements of hands and protusion of tongue. There is also blackish rash over the skin. She was diagnosed as suffering from Wilson's disease at the age of 6 years in view of involuntary movements, presence of KF ring, low serum ceruloplasmin and elevated urinary copper (1005 µg/day) following which she was on treatment with penicillamine, zinc, pacitane, pyridoxine and phenytoin. There is no history of jaundice. On examination, she had chorea, blackish itchy rash over skin. Other systems are normal. She is diagnosed to have Penicillamine induced rash and penicillamine is stopped. Her present 24 hrs urine copper is 50 µg/dl (Normal = 32 to 64 µg/dl) and slit lamp examination shows presence of KF ring.

Has her Wilson's disease improved?

Expert's opinion: - Though this child has a normal 24 hours urine copper excretion, she still has presence of KF rings. This suggests that copper deposition in tissues is still in excess and chelation seems to be inadequate as extra copper is not excreted in the urine. While monitoring a child with Wilson's disease, it is necessary to achieve maximum excretion of copper in urine. Once tissue copper decreases, the KF ring would disappear and liver copper will also decrease. One way to check would be to do her dry weight of copper in liver. In this child, the worsening of her chorea is due to worsening of her Wilson's disease as liver biopsy showed a high copper content.