



Shaping Tomorrow's Nephrology: Insight-Driven Kidney Care

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Pediatric Wilsons Disease: diagnostic pitfalls

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Case Study : Background: Wilson's disease (WD) is a rare copper (Cu) metabolism disorder with toxic organ accumulation. Diagnosis challenges persist in children with atypical multisystem presentations, including uncommon kidney involvement, that may mimic autoimmune and systemic diseases. We have reported a previously healthy girl with unrecognized WD and non-specific renal manifestations, who had urosepsis that triggered fulminant WD, mimicking autoimmune disease & malignancy. Biochemical study confirmed WD and Early Cu chelation reversed organ damage. Clinical Presentation: A 10-year-old girl presented with recurrent jaundice, abdominal pain, weight loss, poor school performance, and polyuria. She had cachexia, hepatosplenomegaly, kinetic hand tremors, non-immune hemolytic anemia, and mild glomerular and tubular dysfunction. Urosepsis triggered decompensation with worsening hepatic function, acute kidney injury (AKI) with Fanconi-like tubulopathy, coagulopathy, low complements, +ve autoantibodies, and elevated IgG. Despite autoimmune and malignancy mimics, Cu studies confirmed WD; D-penicillamine and Zinc achieved rapid hepatorenal and hemolysis recovery. Fibrosis regressed at 1 year despite late ocular affection Conclusion: This case underscores diagnostic challenges of pediatric WD with multisystem involvement including uncommon renal affections, mimicking autoimmune and malignant disorders, emphasizing early Cu studies in multisystem jaundice. Timely chelation therapy achieved rapid improvement of her liver and renal affection.