A Case of Karyomegalic Tubulointerstitial Nephropathy

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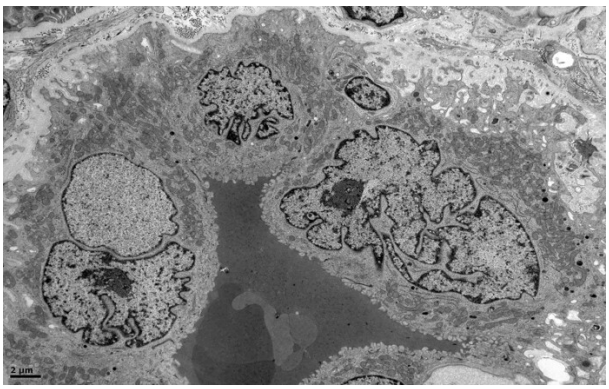
Abstract

The case presented is of a 27-year-old female with persistently raised serum creatinine. Proteinuria was detected during pregnancy and did not resolve following delivery. Renal biopsy revealed 50% glomerular obsolescence by light microscopy and a degree of podocyte injury by electron microscopy however a convincing explanation for the patient's renal impairment remained elusive. Occasional tubular cell nuclei were noted to be large and pleomorphic prompting consideration of regenerative change or viral infection. The case is discussed following the findings of the patient's genetic testing.



Large, pleomorphic tubular cell nuclei in a case of FAN1-related karyomegalic tubulointerstitial nephropathy.

References

1. Clince M, et al. Genotype-phenotype characteristics and disease progression of FAN1-related karyomegalic tubulointerstitial nephropathy. Kidney International (2026) 109, 765-779.