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Ultrastructural alterations of renal tissue in a male patient with fabry's disease

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Fabry disease is an X-linked lipid storage disorder due to deficient lysosomal alpha galactosidase A. Kidney biopsy was done on a 19 year old male patient with complaint of acroparesthesia, maculopapular skin lesions and cornea verticillata. Kidney biopsy tissue was processed and examined by electron microscopy. Changes were inclusion bodies in the cytoplasm of the renal cells. These inclusions were osmophilic with concentric lamellation of clear and dark layers, showing onion skin appearance. The podocytes were mostly affected and some of the foot processes were fused. Cross-sections of collagen fibers were also evident, indicating fibrosis. The ultra-structure of the kidney clearly showed the intracytoplasmic glycosphingolipid accumulation in renal cells, responsible for progressive decline in renal function which could lead to kidney failure. The final diagnosis of Fabry disease was confirmed. In the present case-study, electron microscopy proved to be a valuable diagnostic aid.

Biography

Amir Abbas Farshid is a Professor of Veterinary Pathology, Faculty of Veterinary Medicine, as well as Head of Electron Microscope Center, Urmia University, Urmia, Iran, with more than 85 research papers published in reputed journals.

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