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28 y/o female presenting with foot pain and diagnosed as fabry on kidney biopsy: A case report

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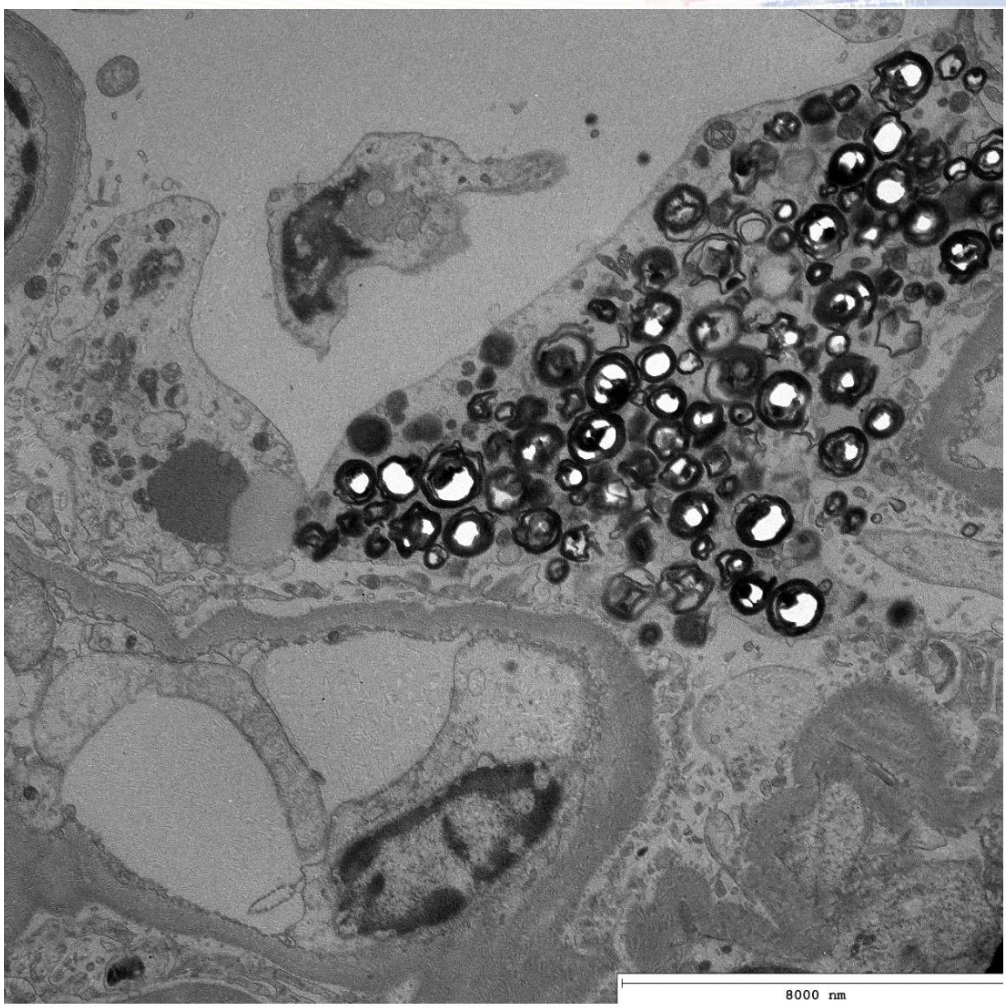
Case Study : Background This is a case of a 28 year old female who presented with fatigue, arthralgias, hypertension, renal insufficiency and skin lesions who underwent kidney biopsy with findings of lamellated inclusion bodies in electron microscopy. Case presentation Patient is a 28 year old female who was admitted due to 6 months history of right foot pain with loss in balance which developed to right foot discoloration described as purplish to bluish in color with associated numbness and paresthesia. This was accompanied with elevated creatinine levels and acute cerebellar infarct on cranial MRI. Other work up showed proteinuria, presence of stenosis on arterial duplex scan, negative C3, ASO, and ANA. She was referred to a nephrologist and a kidney biopsy was done for definitive diagnosis. Discussion Fabry disease is an X-linked lysosomal disorder caused by α-galactosidase A deficiency. It primarily affects males, presenting with neuropathic pain, hypohidrosis, gastrointestinal issues, angiokeratomas, ocular opacities, and chronic fatigue. Females; however, rare may show serious disease manifestations including transit ischemic attacks, stroke and renal failure. Renal manifestations may present as microalbuminuria and over proteinuria with progressive decline in GFR. On light microscopy may present as progressive glomerulosclerosis with capillary wall thickening, tubular atrophy, interstitial fibrosis, arterial and arteriolar sclerosis. Negative immunofluorescence and electron microscopy shows enlarged podocytes with osmiophilic, granular-to-lamellated membrane structures (zebra bodies) Conclusion Fabry disease is diagnosed through genetic testing; however, in a place like the Philippines where it is limited, kidney biopsy could be done not just to confirm the diagnosis of Fabry disease but also to show renal damage.

Picture3. jpg.jpg



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