

Teaching NeuroImages: Neuroimaging leads to recognition of previously undiagnosed Fabry disease

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A 41-year-old man with chronic hypertension presented in a transient, global confusional state. The patient had neuropathic pain and kidney disease, the significance not recognized by his doctors. Family history was positive for hypertension and kidney disease. Brain CT, MRI, and magnetic resonance angiography showed abnormalities typical of Fabry disease (figure).¹ The diagnosis was confirmed by genotype (p.Arg227Gln) and enzymology. Cutaneous and ocular signs indicative for Fabry disease were absent.

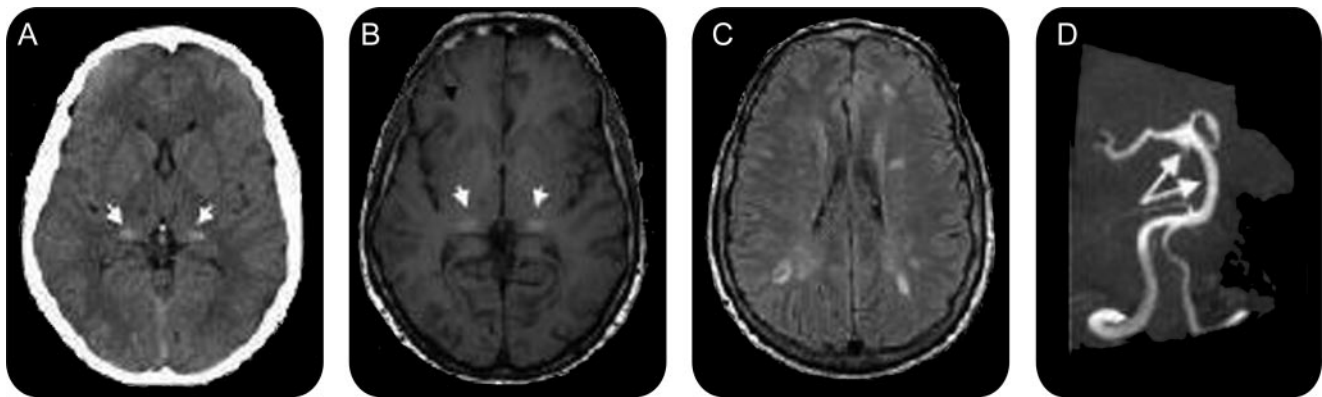
For the past year, the patient has been treated with IV α -galactosidase-A, 0.2 mg/Kg every 2 weeks.² His neuropathic pain improved, although

renal function continued to deteriorate. Fabry disease is probably underestimated and diagnosis is frequently delayed.² Typical radiologic manifestations allow diagnosis.¹ Early enzyme replacement therapy reduces the risk of clinical worsening and often ameliorates the prognosis.²

REFERENCES

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Figure Brain CT, brain MRI, and magnetic resonance angiography



Axial CT scan demonstrated increased attenuation in the pulvinar region (arrows, A), corresponding to the sites of hyperintensity on T1-weighted MRI (the pulvinar sign) (arrows, B). Fluid-attenuated inversion recovery-weighted axial MRI section showed multiple white matter lesions in cerebral hemispheres (C). Magnetic resonance angiography showed basilar dolichoectasia (arrows, D).

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