

Teaching NeuroImages: An uncommon cause of carotid artery dissection

Fabry disease

Aikaterini Theodorou, MD, Lina Palaiodimou, MD, Panagiotis Kokotis, MD, PhD, Marianna Papadopoulou, MD, PhD, Spyridon Fradelos, MD, MSc, FEBO, Adamantia Voudouri, MD, PhD, Christina Zompola, MD, Georgios Magoufis, MD, Chrysa Arvaniti, MD, PhD, Anastasios Bonakis, MD, PhD, and Georgios Tsivgoulis, MD, PhD, MSc, FESO

Correspondence
Dr. Theodorou
kattheo24@gmail.com

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Figure 1 Digital subtraction angiography (DSA) findings



Diagnostic DSA reveals left cervical internal carotid artery occlusion due to underlying dissection with characteristic flame-shaped appearance (A). DSA images during (B, C) and at the end of mechanical thrombectomy (D) show extracranial and intracranial internal carotid artery recanalization and mural dissection (D; arrows).

Cervical internal carotid artery dissection (CAD), a leading cause of ischemic stroke in young adults, has been rarely associated with Fabry disease (FD).^{1,2} A 49-year-old man with left acute middle cerebral artery occlusion due to underlying spontaneous CAD received IV thrombolysis and mechanical thrombectomy, achieving complete reperfusion (figure 1).

Further diagnostic workup excluding other stroke etiologies revealed bilateral cornea verticillata and large-fiber sensory polyneuropathy with impaired lower limb temperature perception (figure 2, D–F). Low α -galactosidase levels and molecular genetic testing disclosing pathogenic GLA variant (c427G>A p.[Ala143Thr]) established FD diagnosis.

This case highlights that FD may represent a rare cause of CAD due to sphingolipid accumulation in vessel walls.

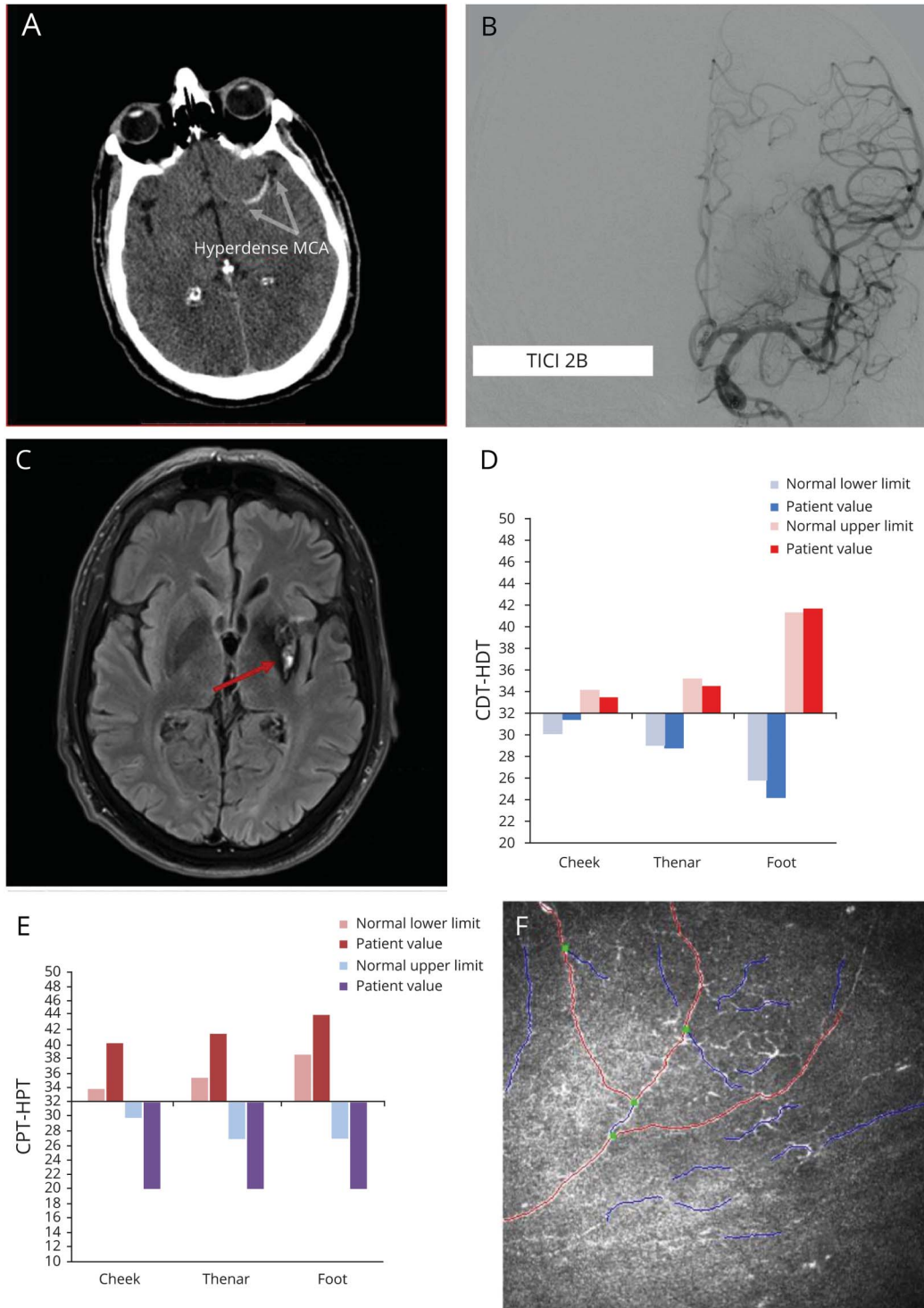
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From the Second Department of Neurology (A.T., L.P., M.P., C.Z., C.A., A.B., G.T.), National and Kapodistrian University of Athens, School of Medicine, "Attikon" University Hospital; Laboratory of Clinical Neurophysiology (P.K.), First Department of Neurology, School of Medicine, National and Kapodistrian University of Athens, "Aiginition" University Hospital; Department of Physiotherapy (M.P.), University of West Attica; Ophthalmological Center "Voudouri Adamantia" (S.F., A.V.), Athens; and Department of Interventional Neuroradiology (G.M.), Metropolitan Hospital, Piraeus, Greece.

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Figure 2 Diagnostic workup in Fabry disease



(A; arrows) Hyperdense middle cerebral artery (MCA) sign in the left M1 segment on baseline brain CT. (B) Post-thrombectomy digital subtraction angiography reveals complete reperfusion (Thrombolysis in Cerebral Infarction [TICI] 2B). (C; arrow) Residual basal ganglia infarction is depicted on follow-up brain MRI. (D, E) Quantitative sensory tests show impairment of cold and warm perception in the lower limbs and (F) corneal confocal microscopy reveals bilateral cornea verticillata. CDT-HDT = cold and heat detection thresholds.

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Appendix Authors

Name	Location	Contribution
Aikaterini Theodorou, MD	“Attikon” University Hospital Athens, Greece	Drafting and revising the manuscript
Lina Palaodimou, MD	“Attikon” University Hospital Athens, Greece	Data collection, critical comments during manuscript revision
Panagiotis Kokotis, MD, PhD	“Aiginiteion” University Hospital, Athens, Greece	Data collection, critical comments during manuscript revision
Marianna Papadopoulou, MD, PhD	University of West Attica, Athens, Greece	Critical comments during manuscript revision
Spyridon Fradelos, MD, MSc, FEBO	Ophthalmologic Center “Voudouri Adamantia,” Athens, Greece	Data collection, critical comments during manuscript revision

Appendix (continued)

Name	Location	Contribution
Adamantia Voudouri, MD, PhD	Ophthalmologic Center “Voudouri Adamantia,” Athens, Greece	Data collection, critical comments during manuscript revision
Christina Zompola, MD	“Attikon” University Hospital Athens, Greece	Data collection, critical comments during manuscript revision
Georgios Magoufis, MD	Metropolitan Hospital, Piraeus, Greece	Data collection, critical comments during manuscript revision
Chrysa Arvaniti, MD, PhD	“Attikon” University Hospital Athens, Greece	Critical comments during manuscript revision
Anastasios Bonakis, MD, PhD	“Attikon” University Hospital Athens, Greece	Critical comments during manuscript revision
Georgios Tsigoulis, MD, PhD, MSc, FESO	“Attikon” University Hospital Athens, Greece	Data collection, drafting the manuscript

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