Case Studies

Restrictive Arteriopathy in Late-Onset Pompe Disease: Case Report and Review of the Literature

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Late-onset Pompe disease (LOPD) is an adult type of classical Pompe disease and presents without cardiomyopathy. Neuroimaging in LOPD is typically limited to posterior circulation and involves dilative arteriopathy, especially dolichoectasia and intracranial aneurysms. We report an interesting case of an established diagnosis of asymptomatic LOPD in a young man with a restrictive-variant pattern in posterior vasculature. We discuss the clinical presentation, neuroimaging, existing literature, and prognosis in vascular variants of LOPD. **Key Words:** Pompe—late-onset—stroke—restrictive.

Introduction

Pompe disease is an autosomal recessive, lysosomal storage disease caused by deficiency in acid-α-glucosidase enzyme. Classical Pompe disease manifests in infancy and involves cardiomyopathy. Late-onset Pompe disease (LOPD) is an adult type of Pompe disease (which presents after 1 year of age) and usually manifests with myopathy. Cerebrovascular manifestations have been recently described with neuroimaging, which is typically limited to posterior circulation and involves dilative arteriopathy, especially dolichoectasia and intracranial aneurysms.[1] We report a novel presentation of a restrictive variant of posterior vasculature in a young man with an established diagnosis of LOPD.

Case Report

A man in his 40s with an established diagnosis of asymptomatic LOPD presented with intermittent episodes of dizziness and left faciobrachiocrural numbness. The symptoms began a week before the patient was hospitalized following a sudden neck turning maneuver while shoveling snow. Family history was significant for a younger brother with myopathic features of LOPD receiving enzyme replacement therapy (ERT). Our patient was not maintained on ERT as he remained asymptomatic from LOPD. Intracranial vessel imaging was negative for aneurysms or malformations; however, it demonstrated a beaded appearance of bilateral vertebral arteries with an atherosclerotic plaque concerning for dissection (likely related to recent neck trauma) and a mild stenosis of mid-basilar artery (Fig 1 A,B). Magnetic resonance imaging (MRI) of the brain demonstrated punctate infarction of right middle cerebral peduncle and right cerebellar hemisphere (Fig 1 C,D). The patient remained asymptomatic during hospitalization and was started on aspirin before discharge. Follow-up routine neuroimaging

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Liebeskind: Consultant to Stryker (significant), Medtronic (significant); employed by the University of California (UC), which holds a patent on retriever devices for stroke.

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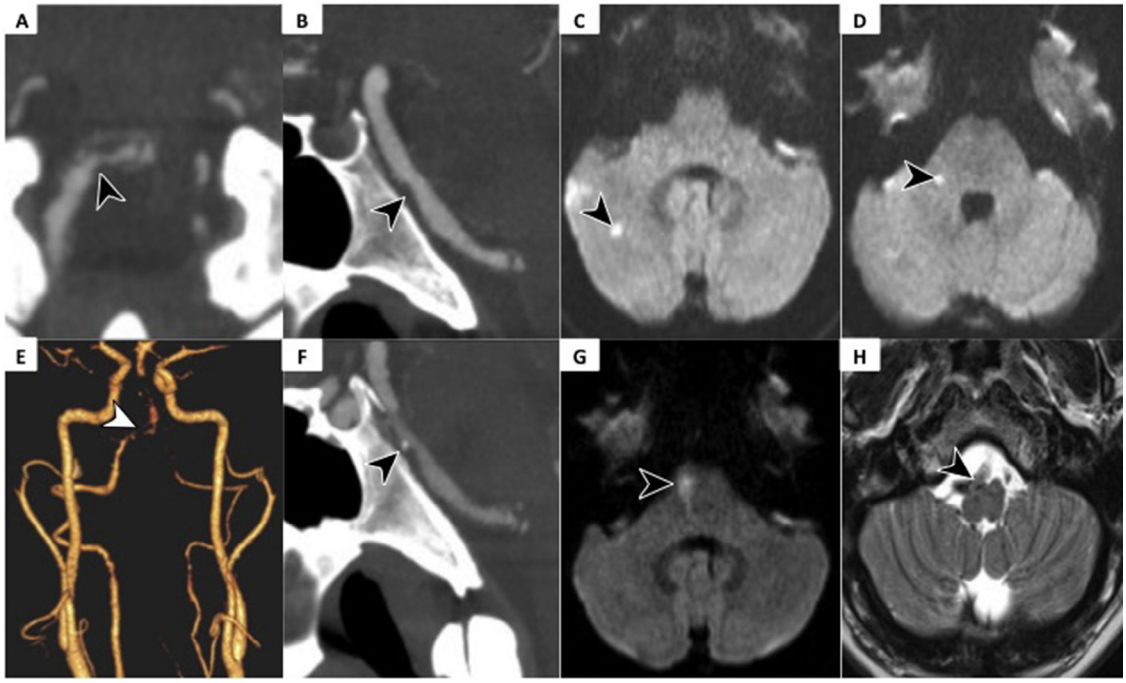

Figure 1. *Neuroimaging of late-onset Pompe disease. CTA depicting (A) right vertebral dissection in axial view with a beaded appearance of vasculature at the vertebrobasilar junction, and (B) mid-basilar stenosis in sagittal view. (C and D) Diffusion-weighted MRI showing punctate infarcts in the right cerebellar and middle cerebral peduncle. Eight months later, the patient presents with left brachiocrural hemiparesis. (E and F) CTA demonstrates progression of stenosis of the vertebrobasilar junction with severe stenosis at mid-basilar segment. (G) MRI brain diffusion-weighted MRI sequence showing right paramedian pontine infarction, and (H) T2-weighted sequence showing right dolichoectatic vertebral artery compressing right pontine hemisphere. Abbreviations: CTA, computed tomographic angiography; MRI, magnetic resonance imaging.*

as outpatient demonstrated progression of stenosis at the vertebrobasilar junction, while the patient remained clinically stable on aspirin monotherapy. Eight months after the index stroke, the patient presented with a fluctuating course of left brachiocrural hemiparesis lasting for 2 weeks' in duration. Computed tomographic angiogram of the head and neck demonstrated progression of steno-occlusive pattern at the vertebrobasilar junction and severe stenosis of mid-basilar segment with robust distal reconstitution (Fig 1 E,F). MRI of the brain revealed acute infarction of the right paramedian pons, with right dolichoectatic vertebral artery compressing the right posterior pontine hemisphere as the likely culprit of acute infarct (Fig 1 G,H). The patient was discharged on dual antiplatelet regimen and was initiated on bi-weekly ERT sessions as outpatient. The clinical and imaging features remained stable on a 3-month follow-up (Fig 2). However, 5 months after the second hospitalization, the patient presented to an outlying facility with vertigo, intermittent weakness, and diplopia. MRI of the brain demonstrated bilateral acute pontine infarcts. His respiratory status deteriorated, which required mechanical ventilation. His symptoms subsequently worsened as he rapidly progressed to a locked-in syndrome. The patient expired

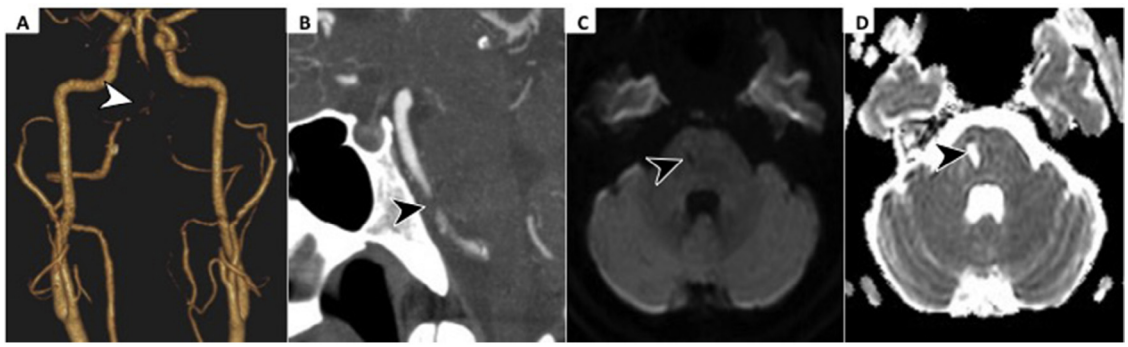

Figure 2. *Neuroimaging 3 months after initiating enzyme replacement therapy. (A and B) CTA of head and neck depicting severe stenosis of the vertebrobasilar junction. (C) Diffusion-weighted MRI and (D) Apparent diffusion coefficient sequences demonstrates old right pontine stroke without new ischemic lesions. Abbreviations: CTA, computed tomographic angiography; MRI, magnetic resonance imaging.*