of upper limbs is a common feature of all affected members or an expression of intrafamilial variability.

By linkage analysis we identified the same haplotype in all the patients and the unaffected sibling (II-1); however, by sequence analysis we found a M408V substitution shared only by affected members. These data indicate that this is a de novo mutation arising in the affected member of the second generation (II-3). The mutation identified in this kindred is located close to the carboxyl terminal of atlastin.

Although the function of atlastin is unknown,⁵ the families described thus far indicate that severe lower limb spasticity and early presentation are associated with missense mutations, whereas premature termination such as that caused by the exon 12 insertion may cause milder or delayed disease.⁵

In this report, we present a novel missense mutation in the *SPG3A* gene that, in our family, is associated with upper limbs amyotrophy. We call attention to the possible de novo presentation of *SPG3A* HSP in infancy, and highlight the opportunity to consider HSP in the differential diagnosis of infants presenting pure lower limb motor impairment.

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Aortic dissection presenting as acute ischemic stroke

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Aortic dissection is an uncommon, life-threatening, and treatable emergency that typically presents with tearing chest and back pain.¹ With use of the Stanford classification, aortic dissections can be divided into type A (involving the ascending aorta) and type B (involving the aorta distal to the origin of the left subclavian artery).² Type A dissections may cause ischemic stroke in up to 5 to 10% of patients³ through extension of the dissection into the common carotid arteries or through thromboembolism or cerebral hypoperfusion.

Case report. A 64-year-old man was referred to the neurologist for evaluation of acute stroke. He had a history of hypertension. He was reversing his car when he suddenly grasped his hands to his chest and "blacked out." He was briefly unresponsive but began to speak with garbled speech and had a Wernicke aphasia.

He arrived at the hospital within 30 minutes of onset. His blood pressure was 180/120 mm Hg and was being monitored closely as the patient was being considered for recombinant tissue plasminogen activator (rt-PA) administration (blood pressure must be <180/110 mm Hg).

Urgent MRI revealed a left inferior division middle cerebral artery territory infarct on diffusion-weighted imaging (figure, A) and an occluded left internal carotid artery on intracranial MR angiography (see figure, B). In this case, acute ischemic stroke was initially considered to be the diagnosis. However, there were some unusual features, including the grasping of his hands to his chest at onset and on return to the emergency room, the fall of the blood pressure in the left arm to 100/70 mm Hg and the inability to record blood pressure in the right arm. Right carotid and radial pulses were unpalpable. No visible bolus was seen on MR perfusion imaging, probably owing to poor cardiac output. An aortic dissection had to be excluded, despite the fact that the patient had no chest pain and the 3-hour time window for rt-PA therapy was passing by. On urgent CT of the chest, a type A aortic dissection was found, extending from the aortic valve through the entire length of the aorta (see figure, C).

The patient's neurologic deficit had improved markedly by 3 hours from onset. An emergent repair of the thoracic aorta was performed. The patient, however, developed a left internal carotid artery territory infarct postoperatively and died 7 days later.

Discussion. This case shows the difficulty of both acute ischemic stroke and aortic dissection diagnoses when the presenting symptoms and signs are atypical. RT-PA must be administered within 3 hours from onset,⁴ and it is important that patients with stroke mimics or major contraindications are identified early.

Type A aortic dissection can often mimic acute myocardial

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infarction (through chest pain and EKG changes).⁵ There have been a number of cases of suspected myocardial infarction inadvertently treated with thrombolysis,⁵ complicated by extension of the dissection into the pericardium, leading to cardiac tamponade and death. Because of this known risk, it has been recommended that cardiologists routinely check all peripheral pulses before giving thrombolysis.⁵ The treatment of choice for type A aortic dissection is emergency operative repair because of the immediate risk of aortic rupture.^{1,2} Uncommonly, as in our patient, stroke or TIA is the presenting symptom of type A aortic dissection.⁶ Surgery in this situation has greater hazards but is recommended for all cases except those with profound neurologic deficits.^{1,3,5} RT-PA should not be given for stroke treatment in the setting of aortic dissection because of the risk of aortic rupture⁶ and cardiac tamponade.

Conversely, internal carotid artery dissection is a much commoner cause of stroke, accounting for around 10% of cases in patients younger than 40 years, presenting with neck or head pain often after trauma. The risks of thrombolytic treatment seem to be much lower in this situation than when the dissection occurs in the aorta (presumably a lower risk of cardiac tamponade and aortic rupture). RT-PA therapy has been reported to be feasible in patients with this diagnosis treated within 7 hours of onset, with no deaths.⁷ However, the efficacy of rt-PA in this setting is undetermined.

To avoid giving rt-PA to ischemic stroke patients with aortic dissection, we suggest that, as in cardiac patients, prior to rt-PA infusion, all peripheral pulses be checked.

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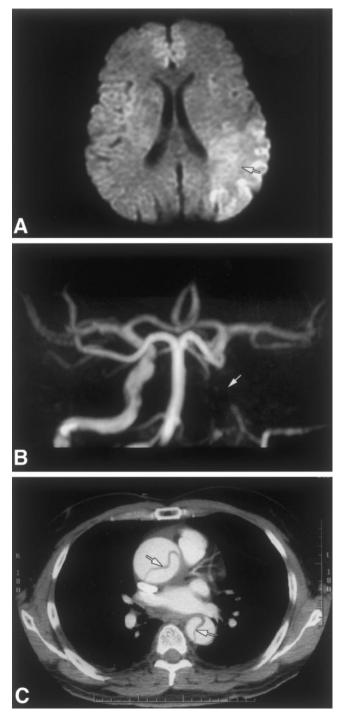


Figure. Urgent MRI revealed a left inferior division middle cerebral artery territory infarct on diffusion-weighted imaging (A, arrow) and an occluded left internal carotid artery on intracranial MR angiography (B, arrow). CT of the chest revealed a type A aortic dissection, extending from the aortic value through the entire length of the aorta (C, arrows).