Figure 2. Chromatogram of the Mutation Identified

Wild type

| T | C | C | T | T | G | C | A | G | A | G | A | G | A | C | C | T | T | G | T | G |

Patient

| T | C | C | T | T | G | C | A | G | A | G | A | G | A | C | C | T | T | G | T | G |

Chromatogram obtained from Sanger sequencing of exon 4 of the elongation of very long-chain fatty acids-like 4 gene and analyzed by Mutation Surveyor version 4.0 (SoftGenetics).

another lipid metabolism gene to the list of genes causing spinocerebellar ataxia.

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OBSERVATION

Successful Antiviral Treatment of Giant Cell Arteritis and Takayasu Arteritis

A patient who satisfies American College of Rheumatology criteria for both giant-cell arteritis (GCA) and Takayasu arteritis had a dramatic favorable response to antiviral treatment. The virological and pathological findings followed by successful antiviral treatment support earlier notions that GCA and Takayasu arteritis may represent a spectrum of the same disease produced by varicella-zoster virus (VZV).

Report of a Case | A woman in her 70s developed severe right-sided temporal pain and jaw claudication. Two months later, she developed bilateral arm pain, which was worse on the left; chest pain on exertion; and shortness of breath. No arm pulses were detected and blood pressure was unobtainable by auscultation or Doppler. Angiography findings revealed bilateral subclavian artery stenosis and left axillary artery occlusion without intracranial vasculopathy. Her erythrocyte sedimentation rate was normal and C-reactive protein level was 1.6 mg/0.1 L (normal <1.0 mg/0.1 L; to convert to nanomoles per liter, multiply by 9.524). Results from a temporal artery (TA) biopsy were initially negative for GCA. Despite treatment with oral prednisone, 30 mg twice daily, she experienced progressive arm pain, intractable fatigue, anorexia, and weight loss.
She underwent additional angiograms, one complicated by deep-seated right hemispheric infarction. Seven months later, she developed gangrene in her left hand and underwent bilateral carotid to brachial artery bypass surgery. She continued prednisone, 20 mg daily, and stopped 2 months later.

Sixteen months after initial presentation, she was cachectic and weighed 30.8 kg. Her fingers were bright red and hypoperesthetic with flexion contractures. Except for weak right popliteal artery pulse, there were no temporal or radial artery pulses, no pulses over the supraclavicular or left popliteal fossa, and both dorsalis pedis pulses were absent. Deep tendon reflexes were increased in the legs with a left extensor plantar response. The erythrocyte sedimentation rate was 30 mm/h (normal <20 mm/h) and C-reactive protein level was 0.3 mg/0.1 L. Computed tomographic angiography revealed extensive large-artery disease involving the right brachiocephalic, left subclavian, and vertebral, bilateral axillary and common carotid arteries; the celiac trunk; and the right renal artery (Figure 1).

Based on detection of VZV in GCA-positive TAs, documented involvement of other large arteries in most patients with GCA, and pathological changes of extensive arteritis with giant cells in both GCA and Takayasu arteritis, we treated our patient with intravenous acyclovir, 15 mg/kg 3 times daily for 2 weeks, followed by oral valacyclovir, 1 g 3 times daily. Immunohistochemical analysis of the TA biopsy obtained 14 months earlier detected VZV antigen, and histopathological examination of 17 sections revealed GCA (Figure 2). The response to antiviral therapy was dramatic. Within a week, she felt energetic and began to eat voraciously. Two weeks later, both TA pulses, left supraclavicular fossa pulse, and left radial and popliteal artery pulses were present. Erythrocyte sedimentation rates and C-reactive protein level during the next

Figure 1. Computed Tomographic Angiograms of Upper Extremities

A. Coronal maximum intensity projection reconstructions at the origin of the great vessels show moderate focal narrowing of the right brachiocephalic artery (black arrowhead) and multiple focal areas of narrowing along the left subclavian and axillary arteries (blue arrowheads) with segmental occlusion distally; the right subclavian artery was partially obscured by contrast in the adjacent vein, although severe stenosis and occlusion were seen distally in the right axillary artery (red arrowhead). Focal narrowing was seen at the origin of the left vertebral artery (yellow arrowhead). B. Volume rendering shows the origin of the great vessels with focal narrowing of the left subclavian artery (blue arrowhead), as well as areas of occlusion distally (red arrowheads). Failure to reconstruct the proximal right subclavian artery is due to reflux of contrast in the adjacent vein, as detailed in panel A. C. An axial section at the level of the common carotid arteries demonstrates bilateral wall thickening, up to 3 mm on the left side (blue arrowheads). Failure to reconstruct the proximal right subclavian artery is due to reflux of contrast in the adjacent vein, as detailed in panel A. D. Aorta and stenotic celiac artery (blue arrowhead) and moderate focal narrowing at the origin of the right renal artery (E, blue arrowhead). Irregularities seen along the aorta (D and E, red arrowheads) reflect calcified atherosclerotic disease.
4 months were normal or mildly elevated. Our patient continues to improve. Four months later, she weighed 39.5 kg, and pulses noted here remain patent. Permanent finger contractures limit mobility and other activities of daily living.

Discussion | Herein, we describe a remarkable case that satisfies American College of Rheumatology criteria for both GCA and Takayasu arteritis. Noteworthy features include development of GCA followed months later by Takayasu arteritis, consistent with findings that large-artery disease frequently complicates GCA. Furthermore, although the original TA biopsy was GCA negative, histopathological examination confirmed the diagnosis of GCA, underscoring the close relationship between VZV antigen and GCA pathology. Most important, however, was the patient’s rapid clinical response to antiviral treatment as manifested by improved energy, appetite, and weight gain, as well as detection of multiple pulses that were absent 2 weeks earlier.

Overall, the virological and pathological findings in this case followed by the favorable response to antiviral therapy sup-
port earlier assumptions that GCA and Takayasu arteritis may represent a spectrum of the same disease produced by VZV.

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Fragile X Tremor Ataxia Syndrome With Rapidly Progressive Myopathy

In this report, we describe a patient with clinically definite fragile X-associated tremor/ataxia syndrome (FTXAS) who experienced rapidly progressive, painless, noninflammatory proximal and distal myopathy after surgery with general anesthesia.

Report of a Case | A right-handed man in his 60s presented with a 10-month history of rapidly progressive motor impairment. His medical history was significant for type 1 diabetes mellitus, peripheral neuropathy, diabetic amyotrophy of the left lower extremity, and complex partial seizures. He also reported a 20-year history of slowly progressive, bilateral hand tremor with action and intention. Family history was significant for fragile X syndrome in his sister’s son.

Ten months earlier, he had undergone total left hip arthroplasty under general anesthesia. After this, he developed rapidly progressive gait and limb ataxia, diffuse muscle weakness and atrophy, impairment of manual dexterity, head tremor, increased hand tremor, dysarthria, mild dysphagia, myoclonic jerking, and frequent falls. He reported orthostatic lightheadedness, nocturia, and erectile dysfunction but no myalgias or muscle cramps and no changes in cognition, motivation, or mood.

Examination showed moderate bilateral dysdiadochokinesis; dysmetria on finger-nose-finger and heel-knee-shin tests; and overshoot on finger follow. There was also mild dysarthria, titubation with standing, and mild-moderate gait ataxia. Extraocular movements were significant for saccadic pursuits and hypometric saccades; square wave jerks and nystagmus were absent. A bilateral jerky hand tremor was present with action and intention; no rest or postural tremor was observed. There was also mild cervical dystonia with left head turn, right head tilt, and a positional yes-yes head tremor. There was no parkinsonism. Diffuse proximal and distal muscle atrophy was noted in all 4 extremities. There was bilateral scapular winging, and right greater than left weakness (4− to 4+ power) in the biceps, finger extensors, and intrinsic hand muscles, particularly the right opponens digiti minimi. There was also weakness (4− to 4+ power) in the left greater than right iliopsoas, of bilateral plantar flexion, and of left ankle and great toe dorsiflexion. Spasticity and fasciculations were absent. There was diffuse hyperreflexia but the ankle jerks were absent. The plantar responses were flexor. There was mild impairment of distal vibratory sensation. The Montreal Cognitive Assessment score was 26 of 30, with impairment of repetition, abstraction, and delayed recall.

Genetic testing showed a fragile X premutation, with 90 CGG repeats in the fragile X mental retardation 1 gene. Brain magnetic resonance imaging showed mild cerebral and cerebellar volume loss and mild T2/fluid-attenuated inversion recovery hyperintensities in the pons and periventricular white matter. The middle cerebellar peduncle and corpus callosum splenium signs were absent. Electrodagnostic testing of the right arm and leg showed evidence of a clear noninflammatory myopathy affecting the proximal and distal muscles and a moderate to severe large-fiber axonal peripheral neuropathy. No active denervation was found. Muscle biopsy from the right quadriceps showed findings consistent with myopathy, including fiber size variation, but no definite signs of mitochondrial dysfunction (Figure). The following test results were all normal: complete blood cell count; electrolytes; renal, hepatic, and thyroid function tests; creatine kinase; erythrocyte sedimentation rate; anti-glutamic acid decarboxylase antibodies; celiac laboratories; other autoimmune and paraneoplastic serologies; vitamin B12 level; heavy metal testing; human immunodeficiency virus test; Lyme antibody; VDRL; serum protein electrophoresis;