Teaching NeuroImages:
A prematurely aging patient presenting with severe leukoaraiosis and stroke

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A 27-year-old woman presented with sudden onset left-sided numbness and double vision. In the last 10 years, she developed diabetes mellitus, cataract, osteoarthritis, osteoporosis, benign neoplasm of the skull, epilepsy, and nonscarring alopecia. Her parents were first-degree relatives and there were no relatives with similar disease. On examination there were right internuclear ophthalmoplegia, short stature, tight skin, hyperkeratosis (figure 1), cataract, and mild cognitive impairment. Brain MRI disclosed acute brainstem ischemic infarct and severe leukoaraiosis with multiple old lacunar infarcts secondary to small-vessel disease (figure 2). Antinuclear antibodies and anticardiolipin and lupus anticoagulant antibodies were normal or negative. Genetic testing was not available.

The patient presents with 5 cardinal and 3 minor features of Werner syndrome, which is an unusual autosomal recessive inherited disorder caused by mutations in the WRN gene on chromosome 8. It encodes a protein with helicase and exonuclease activities, absence of which leads to abnormalities in several DNA repair and processing pathways. Werner syndrome is the most common adult-onset progeria. The disease is characterized by premature aging and propensity for cancer.

Werner syndrome is an uncommon cause of stroke, but it should be considered in young patients with premature aging.

AUTHOR CONTRIBUTIONS
Dr. Seixas, Dr. Baiense: design or conceptualization of the study. Dr. Seixas, Dr. Pedroso, Dr. Fukuda, Dr. De Figueiredo, Dr. Baiense, Dr. Yared, Dr. Ferraz, Dr. Barsottini: analysis or interpretation of the data. Dr. Seixas, Dr. Pedroso, Dr. Fukuda, Dr. De Figueiredo, Dr. Ferraz, Dr. Barsottini: drafting or revising the manuscript for intellectual content.

REFERENCES

MYSTERY CASE RESPONSES
The Mystery Case series was initiated by the Neurology® Resident & Fellow Section to develop the clinical
reasoning skills of trainees. Residency programs, medical student preceptors, and individuals were invited to use this Mystery Case as an educational tool. Responses were solicited through a group e-mail sent to the American Academy of Neurology Consortium of Neurology Residents and Fellows and through social media (Facebook and Twitter).

The answers that we received for this Mystery Case came from 3 different continents through e-mail and social media, in English and Spanish, and from individual residents rather than groups. All of them were well-reasoned and thoughtful.

Many respondents (46%) correctly identified Werner syndrome or adult progeria based on the concomitant occurrence of multiple pathologies characteristic for premature aging in a rather young patient. The second most common diagnosis (20%) was cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy. Other diagnostic considerations included Cockayne syndrome (cachectic dwarfism, photosensitivity, progeroid appearance, pigmentary retinopathy, sensorineural hearing loss, and progressive neurologic degeneration), Rothmund-Thomson syndrome (photosensitivity, poikilodermatous skin changes, cataracts, skeletal dysplasias, and predisposition to osteosarcoma and skin cancer), as well as systemic lupus erythematosus, myotonic dystrophy, and Proteus syndrome.

This Mystery Case illustrates a rare cause of stroke that should be considered in young patients with premature aging.

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Brain MRI diffusion-weighted imaging and apparent diffusion coefficient disclosed small area of restricted diffusion on right pons (A). Fluid-attenuated inversion recovery sequence showed diffuse subcortical hyperintensity (B) and susceptibility-weighted imaging showed numerous hypointense lesions suggesting microhemorrhages (C). These findings are suggestive of small-vessel disease.
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