Differential Diagnoses

FAHR'S DISEASE OR FAHR'S SYNDROME?

by Malathi Latha Perugula, MD, and Steven Lippmann, MD

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This column series compares neurological conditions that pose differential challenges in diagnoses.

Fahr's disease and Fahr's syndrome are two conditions characterized by calcification in certain areas of the brain that results in neurological and/or psychiatric sequelae in patients. While the symptoms and signs of both conditions may resemble one another, there are distinct, critical differences that exist regarding the etiology, location of lesions, prognosis, and treatment. Thus, it is important for clinicians to be familiar with the similarities and differences between Fahr's disease and Fahr's syndrome, so that an accurate diagnosis can be made and appropriate therapy initiated.

DIAGNOSTIC CHECKLIST. Fahr's disease or Fahr's syndrome?[1–6]

A diagnosis of either Fahr's disease or Fahr's syndrome should be considered if some or all of the following symptoms are present:

__ Basal ganglia movement disorder
__ Pyramidal signs
__ Cognitive impairment
__ Gait disorder
__ Cerebellar abnormalities
__ Speech dysfunction
__ Psychiatric presentations
__ Sensory changes.

Consider a diagnosis of Fahr's disease if

__ Age of onset 40 to 60 years
__ Evidence of coarse, progressive, bilateral, symmetrical basal ganglia calcification (Figures 1 and 2)
__ Presence of genetic autosomal dominant or recessive trait

Consider a diagnosis of Fahr's syndrome if

__ Age of onset 30–40 years
__ Evidence of symmetrical, bilateral intracranial calcification (illustrative example can be found here http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3251182/)

and

Presence of any of the following endocrinopathies:

__ idiopathic hypoparathyroidism
__ secondary hypoparathyroidism
__ pseudohypoparathyroidism
__ pseudopseudohypoparathyroidism
__ hyperparathyroidism

or

Presence of any of the following:

__ brucellosis infection, intrauterine or perinatal
__ neuroferritinopathy
__ polycystic lipomembranous osteodysplasia with sclerosing leucoencephaloathy
__ Cockayne syndrome
__ Aicardi-Gouteres syndrome
__ tuberous sclerosis
__ mitochondrial myopathy
__ lipoid proteinosis

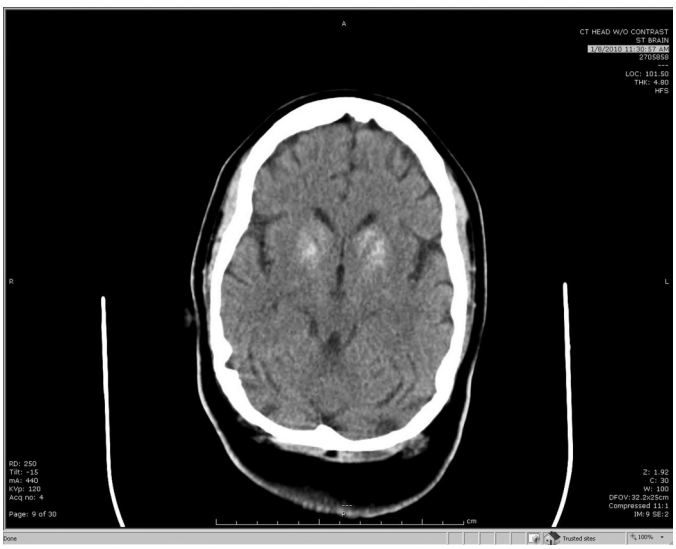
Treatment

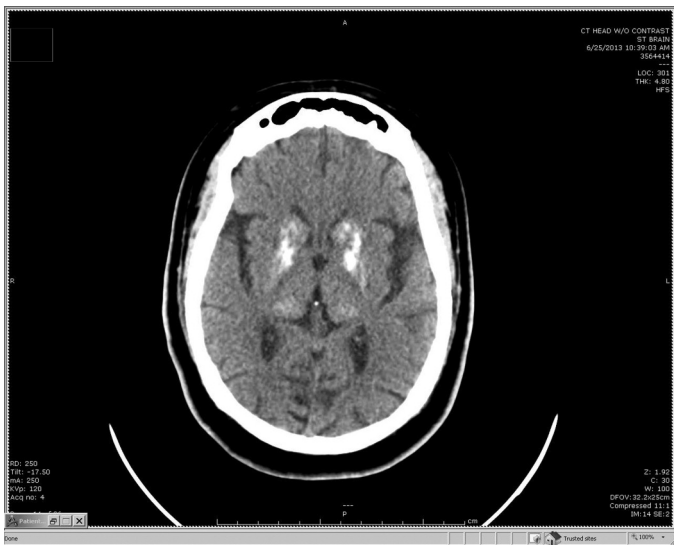
If Fahr's disease—

No specific remediation; only symptomatic therapies.

If Fahr's syndrome—

Treatment should be directed at the specific pathology, with symptomatic therapy adjunctively.





FIGURES 1 and 2. Computerized tomographic scans of the head in 2010 (Figure 1) and 2013 (Figure 2) reveal coarse bilateral, symmetrical calcifications in the basal ganglia, with progression in 2013. Images reprinted with permission. Goyal et al. Would you recognize Fahr's disease if you saw it? *Innov Clin Neurosci.* 2014;11(1–2):26–28

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AUTHOR AFFILIATIONS: The authors are from the Department of Psychiatry and Behavioral Science, University of Louisville School of Medicine, Louisville, Kentucky.

ADDRESS CORRESPONDENCE TO: Steven Lippmann, MD, University of Louisville School of Medicine, 401 E Chestnut Street, Suite 610, Louisville, KY 40202; Email: sblipp01@louisville.edu.