INFECTIOUS DISEASES

DIAGNOSTIC FEATURES OF NEUROBRUCELLOSIS

Investigators at Ankara Numune Training and Research Hospital, a tertiary care community hospital in Turkey, performed a prospective observational study between February 2002 and March 2005 of patients >16 years of age with laboratory confirmed brucellosis. The diagnosis of brucellosis was based on clinical findings and a serum agglutinin titer of >1:160 in serum tube agglutination or a positive blood culture. Lumbar puncture was performed on patients with neurological symptoms or signs, including headache, neck stiffness, confusion, or changes in personality. Neurobrucellosis among laboratory-confirmed brucellosis patients was diagnosed by any one of the following criteria: 1) neurologic symptoms or signs; 2) brucella organism isolated from the CSF and/or anti-Brucella antibodies in CSF; 3) lymphocytosis, increased protein, and decreased glucose in CSF; or 4) MRI or CT abnormalities.

Of 128 patients with LP, 48 (37.5%) were diagnosed with neurobrucellosis, 45 had a CSF agglutination titer of >1.8, and 7 (15%) had Brucella bacteria isolated from CSF. Of 48 patients with neurobrucellosis, 16 (33%) were female, ages ranged from 13-77 years (median age 42 years), and 32 (65%) raised livestock. Consumption of cheese produced from unpasteurized milk was the source of infection in 41 (85%) patients. In addition to fever, myalgia, sweating, and weight loss, neurobrucellosis patients presented with headache, blurred vision, loss of hearing, and confusion. Neurological symptoms also included behavioral changes, agitation, muscle weakness, disorientation, neck rigidity, paresthesias, and rarely, diplopia, facial paralysis, and ataxia. Following treatment with ceftriaxone, rifampicin, and doxycycline for 6 months, one patient died of cardiac failure while the remainder showed no relapse after 3, 6, and 9 months follow-up. Three patients with cranial nerve involvement (facial paralysis in 1, and sensorineural hearing loss in 2) recovered with sequelae. (Guven T, Uguriu K, Ergonui O, et al. Neurobrucellosis: Clinical and diagnostic features. **Clin Infect Dis** 2013 May;56(10):1407-12). (Response: Onder Ergonui MD, MPH, Koc University, School of Medicine, Istanbul, Turkey. E-mail: oergonui@ku.edu.tr).

COMMENT. Brucellosis in children is usually a mild self-limited disease compared with the more chronic disease in adults. Physical findings include lymphadenopathy, hepatosplenomegaly, and arthritis. Serious complications include meningitis, endocarditis, and osteomyelitis. Most cases occur in travelers returning from endemic areas such as the Mediterranean or Middle East (AAP. Brucellosis. In: Pickering LK, ed. **Red Book**: 2012 Report of the Committee on Infectious Diseases. 29th ed. Elk Grove Village, IL: AAP; 2012:256-258).

Manifestations of neurobrucellosis include encephalitis, meningoencephalitis, radiculitis, myelitis, and neuropathies. In the present study, the diagnosis is based on neurological symptoms and signs, and laboratory findings. In endemic regions, the diagnosis should be considered in a patient with severe and persistent headache.

More than 20 references to studies of neurobrucellosis in children are listed in a PubMed search for the last decade 2002-12, the majority from Turkey or Saudi Arabia. Children <12 years of age constituted 21% (115/545) of the total brucellosis admissions

to a major Riyadh hospital in the period 1984-1995. Consumption of unpasteurized camel milk was the main source of infection. Arthritis was the dominant symptom in 70% (Shaalan MA et al. **Int J Infect Dis** 2002 Sep;6(3):182-6).

NEUROCUTANEOUS DISORDERS

STURGE-WEBER SYNDROME LINKED TO GNAQ MUTATION

Investigators from Johns Hopkins School of Medicine, the Hugo W Moser Research Institute at Kennedy Krieger, Baltimore; Duke University; and Medical College of Wisconsin, Milwaukee, performed whole-genome sequencing of DNA from paired samples of tissue from 3 persons with the Sturge-Weber syndrome (SWS). GNAQ somatic mosaic mutations were identified in 88% of participants (23 of 26) with the SWS and from 92% of participants (12 of 13) with nonsyndromic port-wine stains, but not in any of samples from 4 participants with an unrelated cerebrovascular malformation or in any of the samples from 6 controls. The prevalence of the mutant allele in affected tissues ranged from 1.0 to 18.1%. SWS and port-wine stains are caused by a somatic activating mutation in GNAQ. (Shirley MD, Tang H, Gallione CJ, et al. Sturge-Weber syndrome and port-wine stains caused by somatic mutation in GNAQ. **N Engl J Med** 2013 May 23;368(21):1971-9). (Reprints: Dr Pevsner, Department of Neurology, Kennedy Krieger Institute, 707 N Broadway, Baltimore, MD 21205. Email: Pevsner@kennedykrieger.org).

COMMENT. These findings identify a single mechanism for the SWS and nonsyndromic port-wine stains and they document a molecular basis for these malformations, causally related to a mutation in a specific gene, GNAQ. The authors hypothesize that the port-wine stains may represent a late origin of the somatic GNAQ mutation in vascular endothelial cells, whereas the SWS mutation may occur earlier in embryotic development. A child born with a port-wine stain in the distribution of the ophthalmic branch of the trigeminal nerve has a 26% chance of having SWS (Ch'ng S, Tan ST. **J Plast Reconstr Aesthet Surg** 2008 Aug;61(8):889-93; cited by Shirley MD et al. 2013).

INTRACRANIAL HYPERTENSION

CLINICAL SPECTRUM OF PSEUDOTUMOR CEREBRI

Investigators at Erciyes University, Kayseri, Turkey, studied the etiological and clinical features, treatment, and prognosis of pseudotumor cerebri (PTC) in 42 consecutive patients (average age at symptom onset 10 years; range 12 months to 17 years). Girls outnumbered boys, 27 (64%) to 15 (36%). Obesity was associated in 11 (26.2%) patients. Headache in 32 (76%) was the most common presenting symptom. Headache was acute in 13 (31%), chronic daily in 12 (28.8%), acute recurrent in 4 (9.5%), and chronic relapsing in 3 (7.1%). Diplopia occurred in 18 (42.9%), visual loss in 14 (33.3%), vomiting in 15 (35.7%). Papilledema was present in all patients, and VIth cranial nerve paralysis in 8 (19.1%), one bilateral. Mean CSF opening pressure was 350 +/- 96 mm water. One had venous sinus thrombosis on MR venography.