

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% (Shaalán MA et al. **Int J Infect Dis** 2002 Sep;6(3):182-6).

NEURO CUTANEOUS 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 embryonic 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.