

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.

Etiology was unidentified in 30 patients (71%) and termed primary PTC-HH. Secondary causes were detected in 12 (28.6%), defined as secondary PTC, and these included familial Mediterranean fever in 2, preceding trauma (2), and one of each of the following: mycophenolate mofetil-induced PTC, hypervitaminosis A, corticosteroid withdrawal with nephrotic syndrome, oral contraceptives, Guillain-Barre syndrome, urinary tract infection, varicella-zoster virus infection and dural venous sinus thrombosis with otitis media. Treatment included LP, acetazolamide (effective in 14 (37.8%)), and topiramate (effective in 13 of 17 patients (82.4%)). Mean duration of medical treatment was 9 months (range 1-48 months). Ventricular peritoneal shunt was beneficial in 3 patients with impaired visual fields, and visual acuity was normal in all patients at follow-up. (Per H, Canpolat M, Gumus H, et al. Clinical spectrum of the pseudotumor cerebri in children: Etiological, clinical features, treatment and prognosis. **Brain Dev** 2013 Jun;35(6):561-8). (Respond: Huseyin Per, Erciyes University, Division of Pediatric Neurology, Talas, Kayseri 38039, Turkey. E-mail: [hper@erciyes.edu.tr](mailto:hper@erciyes.edu.tr)).

COMMENT. Criteria for the diagnosis of idiopathic intracranial hypertension (HH) or pseudotumor cerebri (PTC) are as follows: 1) symptoms and signs of increased intracranial pressure or papilledema, 2) elevated CSF pressure at LP, 3) normal CSF composition, and 4) normal brain imaging (Per H et al. **Brain Dev** 2013 Jun;35:561-8). Treatable associated disorders should be excluded or treated. Topiramate appeared more effective than acetazolamide in this study and may be used as the drug of choice. Prompt diagnosis and management are important to prevent loss of visual field and acuity.

## **SLEEP DISORDERS**

### **THALAMIC GLUTAMATE/GLUTAMINE IN RESTLESS LEGS SYNDROME**

Investigators at Johns Hopkins University, Baltimore, MD, studied glutaminergic activity and arousal in 28 adults with restless legs syndrome (RLS) and 20 matched controls, using proton magnetic resonance spectroscopy. The thalamic glutamate/glutamine/creatine ratio was higher in patients with RLS than controls ( $p=0.016$ ) and correlated significantly with the wake time during the sleep period ( $p=0.007$ ) and all other RLS-related polysomnographic sleep variables ( $p<0.05$ ) except for periodic leg movements during sleep (PLMS/hour). Glutamate metabolism is strongly related to arousal sleep disturbance but not to PLMS motor features of RLS. This finding contrasts with the reverse for dopamine that shows a limited relation to arousal sleep disturbance but strong relation to PLMS. (Allen RP, Barker PB, Horska A, Earley C J. Thalamic glutamate/glutamine in restless legs syndrome. **Neurology** 2013 May 28;80(22):2028-34). (Response: Dr RP Allen, E-mail: [richardjhu@mac.com](mailto:richardjhu@mac.com)).

COMMENT. An increased glutaminergic activity in RLS demonstrated in this study represents a new RLS abnormality involving thalamocortical activation in a major nondopaminergic neurologic system. The authors (Allen RP, et al) conclude that the combination of glutaminergic (sleep disturbance) and dopaminergic (sensory symptoms, PLMS) abnormalities are involved in the full RLS symptomatology. The elevated