

“burning feet syndrome.” The cause may be nutritional, vitamin B deficiencies, diabetes, hypothyroidism, spinal stenosis, or genetic (Dyck PJ et al. Burning feet as the only manifestation of dominantly inherited sensory neuropathy. **Mayo Clin Proc** 1983 Jul;58(7):426-9), but often idiopathic (Ropper AH, Samuels MA eds. Diseases of the peripheral nerves. In: **Adams and Victor’s Principals of Neurology**. New York, McGraw Hill Medical, 2009, Chap 46;1296-7). SFPN in children is rarely reported. It appears to be an autoimmune disorder, but symptoms of dysesthesia and hyperesthesia are also encountered in children with ADHD and PDD, associated with an abnormal microarray, deletion 17p12 (Personal case report) (Grozeva D, et al. **Schizophr Res** 2012 Mar;135(1-3):1-7).

Diagnostic testing for small fiber neuropathy.

SFPN is characterized by loss of sensation (thermal and pinprick hypoesthesia) and positive sensory symptoms (burning pain, allodynia, hyperalgesia). Clinical neurological examination and routine neurophysiologic tests are often insufficiently sensitive, but skin biopsy and recent availability of normative reference values are of proven value in the diagnosis of damaged small nerve fibers (Lauria G, Lombardi R. Small fiber neuropathy: is skin biopsy the holy grail? **Curr Diab Rep** 2012 Aug;12(4):384-92). In an earlier study of 486 patients with SFPN, skin biopsy had a diagnostic efficiency of 88.4%, clinical examination of 54.6% and quantitative sensory testing of 46.9% (Devigili G, et al. The diagnostic criteria for small fibre neuropathy: from symptoms to neuropathology. **Brain** 2008 Jul;131(Pt 7):1912-25).

COATS SYNDROME IN FACIOSCAPULOHUMERAL DYSTROPHY TYPE 1

Investigators at University of Rochester Medical Center, NY; Hopital Archet-CHU de Nice, France; and Albert Einstein College of Medicine, NY, studied the frequency of Coats syndrome and its association with D4Z4 contraction size in 408 patients identified with facioscapulohumeral dystrophy type 1 (FSHD1). Three patients (0.8%) had a history of Coats disease, and 14 patients had FSHD and Coats syndrome confirmed by ophthalmologic examination. Median age at diagnosis of Coats syndrome was 10 years. The median D4Z4 fragment size was 13 kilobases. Close surveillance for retinal complications is recommended in FSHD1 patients with D4Z4 fragments <15kb. (Statland JM, Sacconi S, Farmakidis C, Donlin-Smith CM, Chung M, Tawil R. Coats syndrome in facioscapulohumeral dystrophy type 1. **Neurology** 2013 Mar 26;80(13):1247-50). (Response: Dr Statland: Jeffrey_Statland@URMC.Rochester.edu).

COMMENT. Coats disease, characterized by exudative retinitis and telangiectases of the retina, with slow progression to retinal detachment, may occur alone as an idiopathic form, unilateral, in males with onset at age 1-20 years, or as a syndrome, associated with FSHD1 and high-frequency hearing loss. FSHD1 is caused by a loss of D4Z4 repeat units on chromosome 4q35. Coats disease in the FSHD-associated syndrome is bilateral, it occurs at any age, and most frequently in female patients with FSHD and large contractions (allele size <15 kb). Contraction size (<15 kb) rather than age or FSHD severity should determine the need for annual retinal examinations for retinal vascular

involvement and possible surgery. An early sign of Coats disease is a yellow-eye in flash photography, a reflection off cholesterol deposits in retinal blood vessels.

Coats disease is named after George Coats (Coats G. Forms of retinal disease with massive exudation. **Royal London Ophthalmic Hospital Reports**. 1908;17(3):440-525). A syndrome characterized by retinal, hearing, muscle and mental disorders was described 60 years later by Robert G. Small (Small RG. Coats' disease and muscular dystrophy. **Transactions of the American Academy of Ophthalmology and Oto-Laryngology, Rochester, MN**. 1968 Mar-Apr;72(2):225-31).

SMA TYPE III MIMICS MUSCULAR DYSTROPHY

Researchers at the National Neuroscience Institute, Riyadh, Saudi Arabia, report a series of 8 patients with type III spinal muscular atrophy who were referred with a diagnosis of muscular dystrophy. Developmental milestones were normal until early juvenile or teens years when they showed a slowly progressive proximal weakness involving limb-girdle muscles. A clumsy gait was associated with frequent falls and difficulty in climbing stairs. Seven patients were products of consanguineous marriage. Hypertrophy of calves in 3 patients contrasted with generalized muscle wasting. Tongue fasciculation occurred in 2 patients, deep tendon reflexes were diminished in 7, and spinal scoliosis developed in 5. Muscle biopsy had nonspecific myopathic features in 3 patients, and nerve conduction studies showed normal, mildly neurogenic or myopathic changes. Serum creatine kinase levels varied from normal to significantly elevated. The diagnosis of SMA III was confirmed by gene testing where deletions of exon 7 were detected in all patients. (Alsaman AS, AlShaikh NM. Type III spinal muscular atrophy mimicking muscular dystrophies. **Pediatr Neurol** 2013 May;48(5):363-6). (Response: Dr Alsaman. E-mail: aalsaman@kfmc.med.sa).

COMMENT. In the diagnosis of SMA type III, the presence of dystrophic features such as calf muscle hypertrophy, limb-girdle muscle weakness, elevated serum CPK, and myopathic or dystrophic muscle biopsy findings will sometimes lead to confusion with muscular dystrophy. Diagnosis is confirmed with a molecular genetic polymerase chain reaction-based test for 5q telomeric SMN1 mutation.

VASCULAR DISORDERS

INTRACEREBRAL HEMORRHAGE, ACUTE SYMPTOMATIC SEIZURES, AND EPILEPSY

Investigators at Yale University School of Medicine, New Haven, CT; Children's Hospital of Philadelphia; Vanderbilt University, Nashville, TN; and Johns Hopkins University, studied the incidence and risk factors for seizures and epilepsy in 73 children with spontaneous intracerebral hemorrhage (ICH) including 20 perinatal subjects (>37 weeks gestation to 28 days) and 53 aged >28 days to <18 years at presentation. Acute symptomatic seizures occurred in 35 subjects (48%); they were a presenting symptom of ICH in 12 perinatal (60%) and 19 childhood (36%) subjects, and they occurred after