PEDIATRIC NEUROLOGY BRIEFS
A MONTHLY JOURNAL REVIEW

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Vol. 12, No. 12 December 1998

SEIZURE DISORDERS

DIPSTICK DIAGNOSIS OF NEONATAL SEIZURE DISORDER

A dipstick test strip (Sulfistix) showing elevated urinary sulfite concentrations (400 mg/l) was used in diagnosis of *molybdenum cofactor deficiency* (MCD), an inborn errror of metabolism, in a male infant presenting with convulsions and cerebral dysgenesis at birth and followed for 3 years at the Klinik fur Kinder und Jugendmedizin, St Marlenhospital, Vechta, Germany. He was irritable, tremulous, and had brief, repetitive, generalized tonic-clonic seizures on the first day of life. The EEG showed multifocal epileptic discharges, and a brain ultrasound showed microgyria, agenesis of the corpus callosum, and multicystic leucoencephalopathy. A sister had died at age 3 years after a similar illness, diagnosis undetermined. Serum lactate, pyruvate, and ammonia, urine aminoacid and organic acid concentrations, muscle and nerve biopsies, and the mitochondrial genome were normal. Sulfite oxidase deficiency, a diagnosis considered on the first day, was dismissed following a negative test for sulfite made on urine collected and allowed to stand at room temperature. The test was repeated at 2 years, using freshly collected urine, and sulfites were present in increased concentration. Other characteristic laboratory findings for MCD were also positive: serum and urinary uric acid concentrations, not previously tested, were very low, urinary xanthine excretion was increased, and genetic testing found the patient homozygous for the MCD gene mutation R73W on exon 1. At 3 year follow-up, the child was severely retarded and hypotonic, and had bilateral lens dislocations. (Koch H. Dipsticks and convulsions. Lancet Dec 5, 1998;352:1824). (Respond: Dr Hartmut Koch, Klinik fur Kinder und Jugendmedizin, St Marlenhospital, D-49375 Vechta, Germany).

COMMENT. A dipstick urine test (Sulfistix,® Merck), commonly used to measure sulfite in wine, should be included in the routine work-up for persistent neonatal seizures, especially in infants born to consanguineous parents. A fresh urine specimen is essential, since sulfite is rapidly oxidized to sulfate at room temperature, nullifying the test. If sulfites are present, measurements of serum and urine concentrations of uric acid should follow.

PEDIATRIC NEUROLOGY BRIEFS (ISSN 1043-3155) © 1998 covers selected articles from the world literature and is published monthly. Send subscription requests ($58 US; $60 Canada; $68 airmail outside N America) to **Pediatric Neurology Briefs - J. Gordon Millichap, M.D., F.R.C.P. - Editor,** P.O. Box 11391, Chicago, Illinois, 60611, USA.
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Molybdenum cofactor deficiency, with resultant sulfite oxidase and xanthine dehydrogenase deficiencies, is an inherited, autosomal recessive, inborn error of metabolism manifesting with neonatal seizures, opisthotonus, craniofacial dysmorphic features, ectopia lentis, and progressive, usually fatal, neurologic deterioration. Although rare, with a total of approximately 15 cases reported, the diagnosis is probably overlooked or delayed, as in the present case-report. Additional reports, not cited by the author, include an infant showing moderate short-term clinical improvement with a diet low in methionine and supplemented with cysteine. (see Progress in Pediatric Neurology Vol II, PNB Publ, 1994;pp474-476; Ped Neur Briefs August 1998;12:58-59). In the US, sulfite dipsticks (Quantofix, Maehery-Nagel) may be purchased at $20/100 through Gallard-Schlesinger, Tel:516-333-5600.

STARING SPELLS: EPILEPTIC OR NONEPILEPTIC EVENTS

A parental questionnaire containing 25 items to distinguish epileptic (absence seizures [AS]) and nonepileptic staring [NES] characteristics in 40 children presenting with staring spells was tested at the Cleveland Clinic, Ohio. Results from 17 children with AS and 23 with NES, diagnosed by neurologic exam, EEG, and video EEG, were compared. Features with high specificity for NES (responsive to touch, uninterrupted play, event recognized by teacher or health professional) occurred more frequently in NES than in AS. Each of these features had moderate sensitivity. Body rocking occurred only in patients with NES, but sensitivity was low. Features with high specificity for AS included limb twitches, upward gaze, and urinary incontinence during staring episode; but sensitivity for these features was low. Problems with learning or attention were reported in 40% of children in both groups. A more frequent positive family history for staring or AS in the AS group was not significant. (Rosenow F, Wyllie E, Kotagal P et al. Staring spells in children: descriptive features distinguishing epileptic and nonepileptic events. J Pediatr Nov 1998;133:660-663). (Reprints: Elaine Wyllie MD, Department of Neurology, S51, Cleveland Clinic Foundation, 9500 Euclid Ave, Cleveland, OH 44195).

COMMENT. A history of staring spells is often elicited during the pediatric neurology evaluation of children presenting with short attention, distractibility, and other symptoms of ADHD. The need for an EEG in patients with ADHD and in those presenting with staring spells requires clinical judgement. The questionnaire employed in the above report assists in the differentiation of nonepileptic staring (NES) events and absence epilepsy, and offers a group of features that may be considered strongly exclusive of a diagnosis of epilepsy. The diagnosis of NES is most likely in children who respond to touch while staring, continue playing, exhibit body rocking, and are initially identified by a teacher, nurse, or psychologist. In a child with these clinical features and a normal interictal EEG, a video recording may be deferred. The occurrence of associated limb twitching, upward eye movements, interrupted play, or urinary incontinence are indicative of absence epilepsy, and EEG documentation of epileptiform discharges, with or without video-recording, is essential.

Epileptic seizures temporally associated with nonepileptic seizures are reported in 4 patients, one child and 3 adults, evaluated at the New York University Medical Center (Devinsky O, Gordon E. Neurology Nov 1998;51:1293-1296). A 12-year-old girl with medically refractory partial seizures since age 8 years had video-EEG documented epileptic and nonepileptic