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METABOLIC DISORDERS

ANDERSEN'S SYNDROME: A PERIODIC PARALYSIS

A study of 11 patients from 5 kindreds with Andersen's syndrome (AS), a triad of potassium-sensitive periodic paralysis, ventricular dysrhythmias, and dysmorphic facial features, is reported from the Department of Neurology, University of Milan, Italy; University of Rochester, NY; University of Utah, Salt Lake City; Southwestern Medical Center, Scottish Rite Hospital, Dallas, TX; and Oregon Health Science University, Portland, OR. Episodic attacks of paralysis were associated with hypo-, normo-, or hyperkalemia. A prolonged QT interval (0.47 msec) on ECG was present in all patients and 4 had ventricular arrhythmias, one with cardiac arrest. Dysmorphic features included hypertelorism, broad nose, small mandible, clino- and syndactyly, and two patients had scoliosis. Genetic linkage analysis excluded linkage to the HyperKPP locus on chromosome 17 and the LQT1 locus on chromosome 11. Also, the common HypoKPP dihydropyridine mutations responsible for hypokalemic periodic paralysis were absent. AS is a unique autosomal dominant disorder, genetically distinct from the common forms of periodic paralysis and LQT syndromes. Partial manifestations of the syndrome occur in families, and a prolonged QT interval may be the only sign. (Sansone V, Griggs RC, Meola G et al. Andersen's syndrome: a distinct periodic paralysis. Ann Neurol Sept 1997;42:305-312). (Respond: Dr V Sansone, Department of Neurology, University of Milan, San Donato Hospital, V Morandi 30, 20097-San Donato Milanese, Milan, Italy).

COMMENT. Andersen's syndrome is an autosomal dominant familial disorder characterized by hypo- or hyperkalemic periodic paralysis, prolonged QT interval with or without ventricular dysrhythmias, and facial dysmorphic features. Patients presenting with periodic weakness should have a cardiac workup for prolonged QT interval. Potassium challenges may precipitate life-threatening ventricular arrhythmia, and should be avoided in favor of graded exercise to provoke muscle weakness or serum potassium estimation at times of periodic paralysis.

Other metabolic syndromes which can present with dysmorphic

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features include Smith-Lemli-Opitz, Zellweger, and glutaric acidemia type II (see Menkes JH. In: Progress in Pediatric Neurology III, Millichap JG, ed. Chicago, PNB Publishers, 1997; pp533-535).

PENICILLAMINE-INDUCED SIGNS OF WILSON DISEASE

A 9-year-old boy with asymptomatic Wilson disease who developed neurologic symptoms after treatment with penicillamine is reported from the Department of Pediatrics, Universita Federico II, Naples, Italy. Initially referred for isolated hypertransaminasemia, and hepatomegaly was the only sign of Wilson disease. The diagnosis was confirmed with abnormal urinary copper excretion and high liver copper. After 10 weeks of treatment with penicillamine (20 mg/kg/day) and pyridoxine (25 mg/day), the child developed a tremor of the upper limbs which became progressively severe and incapacitating. Zinc sulfate (100 mg 3x daily) therapy and omission of penicillamine were followed by marked improvement within 2 weeks and remission of tremor at 4 month and 30-month follow-up. Serum transaminase and urinary copper levels were decreased following zinc treatment. (Porzio S, Iorio R, Vajro P, Pensati P, Vegnente A. Penicillamine-related neurologic syndrome in a child affected by Wilson disease with hepatic presentation. Arch Neurol Sept 1997;54:1166-1168). (Reprints: Salvatore Porzio MD, Dipartimento di Pediatria, Universita "Federico II," via S Pansini 5, 80131 Napoli, Italia).

COMMENT. Penicillamine therapy for Wilson disease may rarely precipitate the onset of neurologic symptoms. The authors cite only 5 previous cases, all adults. Zinc appears to be a satisfactory alternative treatment in a limited series. Other reported side-effects of penicillamine include fever, rash, adenopathy, pyridoxine-responsive optic neuritis, nephrotic syndrome, thrombocytopenia, and leukopenia. (Menkes JH. Textbook of Child Neurology, 3rd ed, Philadelphia, Lea & Febiger, 1985; p93).

HEREDO-DEGENERATIVE DISEASES

PELIZAEUS-MERZBACHER-LIKE DISEASE

A family with X-linked inheritance and Pelizaeus-Merzbacher-like disease (PMLD), PMD without the proteolipid protein (PLP) mutation, studied at the UMDNJ-Robert Wood Johnson Medicial School, New Brunswick, NJ, had a new locus on Xq chromosome, more than 10 cM away from PLP. A clinical diagnosis of PMD in the proband was made on the X-linked inheritance, congenital onset, nystagmus, slow progression with pyramidal signs, seizures, and psychomotor deterioration. The patient died at 25 years, and neuropathological study showed calcific vasculopathy in the basal ganglia and dentate nucleus and intact myelin. (Lazzarini A, Schwarz KO, Jiang S, Stenroos ES, Lehner T, Johnson WG. Pelizaeus-Merzbacher-like disease: Exclusion of the proteolipid protein locus and documentation of a new locus on Xq. Neurology Sept 1997;49:824-832). (Reprints: Dr Alice Lazzarini, Department of Neurology, UMDNJ-Robert Wood Johnson Medical School, 97 Paterson St, New Brunswick, NJ 08903).

COMMENT. PMD patients having mutations in the PLP gene have an onset of symptoms in early infancy, nystagmus, spastic diplegia, ataxia, titubation, head bobbing, hypotonia, and mental retardation. Classical type I PMD has 'tigroid' or perivascular preserved myelin and a life span up to 25 years, whereas type II PMD has diffuse demyelination and death before 10