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DEGENERATIVE AND METABOLIC DISEASES

HEREDITARY MOTOR AND SENSORY NEUROPATHY (HMSN)

Thirteen affected males and 25 obligate or probable heterozygous females with X-linked HMSN are reported from the Depts Neurology and Pathology, Duke Univ Med Centr, Durham, NC and Dept Neurology, Univ of Pennsylvania, Philadelphia, PA. The German family ancestry was traced back to a female born in 1819 with 2 affected brothers. In 9 generations there were 34 affected males and 54 heterozygous females. No son of an affected male had symptoms or signs of neuropathy. Intelligence and cranial nerves were normal and peripheral nerves were not hypertrophied. The affected or heterozygous patients had at least one of the following: 1) symmetrical distal>proximal weakness and/or atrophy; 2) symmetrical distal sensory loss, 3) hyporeflexia, 4) pes cavus, 5) abnormal NCV'S, 6) obligate heterozygote (i.e. mother of a patient or daughter of an affected male). Affected males suffered from a progressive crippling neuropathy with onset in childhood or adolescence and female carriers were minimally affected or normal. DNA probes placed the gene in the DXYSI-p58-1 region of the X-chromosome. (Rozear MP, Pericak-Vance MA, Fischbeck K et al. Neurology 1987; 37:1460-1465).

> COMMENT. In a child presenting at the age of 5-10 years with a progressively clumsy gait, pes cavus, depressed ankle jerks, and weakness and loss of sensation in the hands and feet, the diagnosis of hereditary motor sensory neuropathy (Charcot-Marie-Tooth disease) must be suspected and nerve conduction and electromyography studies ordered. Ask the parents to remove their shoes and examine them also for pes cavus and absent ankle jerks, if the hereditary nature of the gait disorder has not already been established. Some may have autosomal dominant and others an X-linked inheritance, as in this family. With no documented male-to-male transmission in a family, both autosomal dominant and X-linked forms are possible and need to be considered in counselling.
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> Of 205 patients referred to the Mayo Clinic with undiagnosed peripheral neuropathy, 42% had inherited disorders, most commonly HMSN. (Dyck PJ, Oviatt KF, Lambert EH. Ann Neurol 1981:10:222). Sural nerve biopsy showing demyelination was occasionally useful. Leg cramps were more common and paresthesiae less troublesome in inherited neuropathies than in inflammatory or other acquired neuropathies.

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