

7 born to mothers with hyperthyroidism developed transient hyperthyroidism, but none showed changes in the MDI. Motor nerve conduction, somatosensory evoked potentials, and Bayley scales showed no differences among groups. (Smit BJ, Kok JH, Vulsma T et al. Neurologic development of the newborn and young child in relation to maternal thyroid function. Acta Paediatr March 2000;89:291-295). (Respond: Dr BJ Smit, Department of Neonatology, H3N-148, Academic Medical Center, University of Amsterdam, Emma Children's Hospital AMC, PO Box 22700, 1100 DE Amsterdam, The Netherlands).

COMMENT. Maternal subclinical hypothyroidism in the first half of pregnancy is associated with a lower mental development score in the infants during the first year of life. Optimal treatment of maternal thyroid disease throughout pregnancy is emphasized.

Low triiodothyronine levels in preterm infants are correlated with a 6.6 point deficit in Full Scale IQ scores on the WISC Scales at 8 year follow-up, and an increased risk of developing cerebral palsy and cognitive deficits is reported in premature infants with low thyroxine levels in the first week of life. Some authorities caution that thyroxine therapy for hypothyroid mothers is unlikely to benefit premature babies, and triiodothyronine replacement in premature infants may be dangerous. Neonatologists generally withhold therapy pending a retest after the infant reaches term. (See Progress in Pediatric Neurology III, 1997;pp 281-2).

HEREDITARY NEUROPATHIES

GENETICS OF MOTOR AND SENSORY NEUROPATHY TYPE 2C

Thirty-three subjects in a large family with hereditary motor and sensory neuropathy (HMSN) type 2C were examined by linkage analysis at the Mayo Clinic, Rochester, MN. The HMSN 2C phenotype, including 12 affected and 11 at risk, were not linked to either the HMSN 2B or 2D loci. HMSN 2C is genetically distinct from HMSN 2A, 2B, and 2D. (Nagamatsu M, Jenkins RB, Schaid DJ, Klein DM, Dyck PJ. Hereditary motor and sensory neuropathy type 2C is genetically distinct from types 2B and 2D. Arch Neurol May 2000;57:669-672). ((Reprints: Peter James Dyck MD, Peripheral Neuropathy Research Center, Mayo Clinic, 200 First St SW, Rochester, MN 55905).

COMMENT. HMSN type 2C is a clinical variety of HMSN 2 manifested by motor and sensory neuropathy of the limbs and progressive weakness of the vocal cords, diaphragm, and intercostal muscles. HMSN 2 shows genetic heterogeneity, and at least 4 genetic varieties have been confirmed: HMSN 2A (1p35-p36), HMSN 2B (3q13-q22), HMSN 2D (7p14), and HMSN 2C (not linked to any of these loci).

The many faces of Charcot-Marie-Tooth disease (HMSN) are reviewed in an editorial (Vance JM. Arch Neurol May 2000;57:638-640). Phenotype classifications in CMT include CMT1 (autosomal dominant, demyelinating), CMT2 (autosomal dominant, axonal presentation), CMT3 (autosomal recessive, axonal presentation), CMT4 (autosomal recessive, demyelinating presentation), and CMTX (X-linked, axonal, or demyelinating presentation). Both clinical and molecular classifications are discussed.

Neurotrophins and their relevance to neurologic disease are reviewed by Kernie SG, Parada LF (*Arch Neurol* May 2000;57:654-657). Neurotrophin therapy has potential in diseases of the peripheral nervous system especially because its function has been studied in peripheral sensory ganglia. Trials of subcutaneous injections of nerve growth factor (NGF), the prototypical neurotrophin, in diabetic polyneuropathy have shown promising results.

RETT SYNDROME

MECP2 MUTATIONS AND RETT SYNDROME PHENOTYPES

Seventy-one sporadic and 7 familial Rett syndrome (RTT) patients were screened for *MECP2* mutations by direct sequencing and the pattern of X chromosome inactivation (XCI) was determined in 39 RTT patients at the Baylor College of Medicine, Houston, TX. Twenty-three different disease-causing *MECP2* mutations were identified in 54 of 71 (76%) sporadic and in 2 of 7 (29%) familial cases. Thirty-one of 34 patients (91%) with classic RTT had random XCI. Nonrandom XCI was associated with milder phenotypes. RTT is caused by a partial loss of *MeCP2* function. (Amir RE, van den Veyver IB, Schultz R et al. Influence of mutation type and X chromosome inactivation on Rett syndrome phenotypes. *Ann Neurol* May 2000;47:670-679). (Respond: Dr Huda Y Zoghbi, Howard Hughes Medical Institute, Baylor College of Medicine, One Baylor Plaza, Houston, TX 77030).

COMMENT. Different *MECP2* mutations have similar Rett syndrome phenotypic consequences, and random X chromosome inactivation plays a role in the full phenotypic spectrum of classic RTT.

Amino acid receptors in frontal cortex in RTT syndrome. A study at Johns Hopkins University, Kennedy Krieger Institute, showed that the densities of N-methyl-D-aspartate, AMPA, and GABA, measured autoradiographically in the superior frontal gyrus, were higher in younger patients and lower in older patients when compared with controls. The age-related changes in amino acid receptor density could be correlated with the stages of RTT syndrome, younger age-stage II/III regression and seizures to a less epileptic plateau stage in older girls. (Blue ME, Naidu S, Johnston MV. Development of amino acid receptors in frontal cortex from girls with Rett syndrome. *Ann Neurol* 1999;45:541-545).

ATTENTION DEFICIT DISORDERS

DAMP DIAGNOSIS

A simplified pediatric school entry screening examination for the syndrome of deficits in attention, motor control and perception (DAMP) is suggested from the Goteborg University, Sweden. A population-based cohort of 113 children, 6-7 years of age (62 with and 51 without DAMP), were compared on measures of attention, motor function, language, and cognition. Attention deficits were identified by both parents and pediatrician. Four of nine motor function tests and visual reaction times discriminated between the DAMP and control groups. Design copying for diagnosing perceptual disorders was better than block design and object assembly WISC subtests. Full-scale WISC IQs were lower in the DAMP group, and children with DAMP had greater phonological processing difficulties. (Landgren M, Kjellman B, Gillberg C. Deficits in attention, motor control and perception (DAMP): a simplified school entry examination. *Acta Paediatr* March 2000;89:302-309). (Respond: Christopher Gillberg MD, Department of Child