features of these 4 patients sufficiently similar to suggest a clinical entity not previously described. The focal seizure pattern, hemiparesis, focal epileptiform EEG discharges and focal cortical dysplasia all indicated a localized lesion, yet a generalized abnormality of EEG background activity and widespread cognitive defects pointed to a more diffuse cerebral dysfunction as well. The differential diagnosis included tumor, post-traumatic or ischemic lesions, metabolic disorder, and chronic localized encephalitis of Rasmussen T et al. (Neurology 1958; 8: 435). The report of macrogyria in 3 of the patients and giant astrocytes in both patients with histological studies is suggestive of a forme fruste tuberous sclerosis that might be entertained in the differential diagnosis.

DEGENERATIVE AND METABOLIC DISORDERS

RETT SYNDROME: DIAGNOSTIC CRITERIA

Diagnostic criteria for Rett Syndrome are proposed by the International Rett Syndrome Association and the Centers for Disease Control, Koger Center, F-37, Atlanta, GA. The criteria are separated into three categories: 1) necessary, 2) supportive, and 3) exclusion criteria. Female sex is not included as a necessary criterion because the possibility of undiagnosed male cases cannot be ruled out. Diagnosis is tentative unil 2-5 yrs of age, and the presence of one or more of the exclusion criteria is against the diagnosis, regardless of whether all of the necessary criteria have been met.

Necessary criteria include the following: 1) normal pregnancy, birth and psychomotor development through the first 6 or 18 months; 2) normal head circ at birth and deceleration of head growth between 5 mos and 4 yrs; 3) loss of purposeful hand skills between 6 and 30 months; 4) impaired language and psychomotor development; 5) stereotypic hand movements such as hand wringing; and 6) gait apraxia and ataxia between 1 and 4 years. Supportive criteria include breathing irregularities, EEG abnormalities, seizures, spasticity, scoliosis, growth retardation and small feet. Evidence of intrauterine growth retardation or perinatal acquired brain damage, microcephaly at birth, identifiable metabolic, degenerative or storage diseases are listed as exclusion criteria.

The clinical characteristics of Rett Syndrome and differential diagnoses are listed according to stages and age at onset: 1) Early onset deceleration stage, 6-18 mos; 2) rapid "destructive" stage, 1-3 yrs; 3) pseudostationary stage, 2-10 yrs; 4) late motor deterioration stage, 10+ years. (Trevathan E, Moser HW et al. The Rett Syndrome diagnostic criteria work group. Diagnostic criteria for Rett Syndrome. Ann Neurol April 1988; 23: 425-428).

COMMENT. Heller's dementia, an infantile dementia described in 1908, almost 60 years before the first description of Rett Syndrome, should be added to the differential diagnosis (Millichap JG. Lancet 1987; 1: 440; Rett A and Olsson B. Dev Med & Child Neurol 1987; 29: 835), especially as the female sex is no longer considered a necessary diagnostic criterion for Rett Syndrome. At this stage of our understanding, the diagnostic criteria of Rett Syndrome should not be too strict and too exclusive (Opitz J. Am J Med Genet 1986;

24: 27).

Partington MW (Am J Med Genet March 1988; 29: 633) describes Rett Syndrome in a pair of monozygotic twin girls, pointing out that their development was delayed from birth with no period of normal progress in infancy and subsequent regression, findings at variance with the necessary diagnostic criteria listed above. He states that the cause is not necessarily genetic but could be explained by prenatal toxic or slow viral factors.

Karet D et al. (J Pediat Orthopaedics March/April 1988; 8: 138) reports scoliosis in eight of 10 females with Rett Syndrome treated at the Alfred I. DuPont Institute, Wilmington, Delaware. Curve progression occurred in four and posterior spinal fusion was performed in five. Scoliosis developed at an average age of 11 years and progression was rapid in adolescence. Early surgery is recommended to arrest curve progression and to obtain correction of the deformity.

PEROXISOMAL DISORDERS

Generalized peroxisomal disorders are classified in three main groups in a review article from the Kennedy Institute and the Departments of Neurology and Pediatrics, Johns Hopkins University, 707 N. Broadway, Baltimore, MD. Group 1 includes Zellweger (cerebro-hepato-renal) syndrome, neonatal adrenoleukodystrophy, infantile Refsum disease, and hyperpipecolic acidemia, all characterized by a reduction in the number of peroxisomes and deficiency of multiple peroxisomal enzymes.

Group 2 contains only one rare disorder, rhizomelic chondrodysplasia punctata, characterized by stippled calcification of hyaline cartilage, dwarfing, cataracts, multiple malformations with contractures, koala bear facies, and sever mental retardation. Peroxisomes are normal in number but functionally impaired.

Group 3 includes Refsum disease, X-linked adrenoleukodystrophy, pseudo-Zellweger syndrome, hyperoxaluria type 1, acatalasemia and an undescribed variant. All have a normal number of peroxisomes and the activity of only one peroxisomal enzyme is reduced.

Peroxisomal disorders are a newly recognized and heterogeneous group of diseases with variable manifestations transmitted as autosomal recessive or sex-linked recessive traits and have in common one or more peroxisomal enzyme defects. The term peroxisome is coined from the hydrogen peroxide-forming enzymes found within the subcellular organelle. More than 40 enzymes have now been localized to the peroxisomes. (Naidu S, Moser AE, Moser HW. Phenotypic and Genotypic Variability of generalized peroxisomal disorders. Pediatr Neurol Jan/Feb 1988; 4: 5-12).

COMMENT. This is an excellent review of the various entities now classified as generalized peroxisomal disorders. See Ped Neuro Briefs (March 1988; 2: 22-23, and Oct 1987; 1:32) for case reports of infantile Refsum and Zellweger syndromes.

SPINO-CEREBELLAR DEGENERATION AND CEROID LIPOFUSCINOSIS

Neuronal ceroid lipofuscinosis (NCL) presenting in two different forms within a family is reported from the New York State Office of Mental Retardation and Developmental Disabilities, Institute for Basic Research,