

Peroxisomal Disorders: Three Reports

A recent review of peroxisomal disorders has focused attention on them and has allowed their features to be summarized. The list of peroxisomal disorders is increasing with the increased index of suspicion. Thus far, all of the peroxisomal disorders show accumulation of bile acid precursors and very long chain fatty acids, with impaired biosynthesis of plasminogens. In some cases, abnormal peroxisomes can be seen on electron microscopy.

Interestingly, all cases of peroxisomal disorders also have minor congenital anomalies. These include structural abnormalities of the brain due to malmigration of neurons, dysplastic cystic kidneys, retinitis pigmentosa due to dysplasia of the retina, hepatomegaly, deafness, stippled epiphyses, and abnormal facies with high forehead and myopathy. Mental retardation and hypotonia are usually present as well. None of these clinical features are pathognomonic.

The biochemistry of peroxiso-

mal disorders is poorly understood at this time. However, good screening tests of urine, serum, and fibroblasts are beginning to be developed. Presently, Zellweger's syndrome, adrenoleukodystrophy, infantile Refsum's syndrome, infantile Conradi's syndrome, and Leber's disease have been defined as peroxisomal disorders. The index of suspicion for these disorders should be raised when evaluating any child with minor anomalies of the type described above.

The report of pseudo-Zellweger's syndrome by Goldfischer et al¹ indicates that one can have abnormal peroxisomal function in the presence of abundant peroxisomes. The report of Leber's disease by Ek et al² described specific changes of macular hyperpigmentation and absence of electroretinographic responses without the other features of Zellweger's syndrome. This finding suggests that any disorder with dysplasia of the retina needs to be considered as a potential peroxisomal disorder. The description of "infantile" Refsum's syndrome by Sargini et al³ reminds us that the presence of neurosensory deafness and retini-

tis pigmentosa indicate the possibility of the diagnosis of a peroxisomal disorder. This is particularly true in children with hypotonia and hepatomegaly. Sargini et al found four patients within six months when they began to look for the disorder. This suggests that this may be a relatively common condition.

1. Goldfischer S, Collins J, Rapin I, et al. *J Pediatr* 1986;108:25-32.
2. Ek J, Kase BF, Reith A, et al. *J Pediatr* 1986;108:19-24.
3. Sargini S, Budden MD, Kennaway NG, et al. *J Pediatr* 1986;108:34-39.

Editor's comment—*Peroxisomal disorders are a whole new class of inborn errors of metabolism in which the combination of long chain fatty acid metabolism and congenital developmental anomalies is seen. Although the definition of peroxisomal metabolism is in its infancy, peroxisomal disorders appear to be inborn errors of metabolism in which morphogenesis is affected. This is an exciting new area that will surely allow the description of a whole new set of specific single gene disorders.*