PEDIATRIC NOTES

THE WEEKLY PEDIATRIC COMMENTARY

Sydney S. Gellis, M.D., Editor

Volume 18, Number 1 January 6, 1994

Pages 2-3

Treatment of Rett Syndrome With L-Carnitine

Mitochondrial abnormalities have been described in muscle and nerve biopsy specimens in Rett syndrome, and because carnitine transports long-chain fatty acids into mitochondria, the authors (Univ. Illinois, Chicago) report its administration to a 17-year-old female with Rett syndrome. The mother's pregnancy and delivery had been normal. Developmental delay in the infant was questioned at 6 months. She began to walk at 16 months. At 5 years of age she had a cognitive level of a 3-year-old. At 6 years she had an IQ of 39. Between 6 and 7 years her gait started to deteriorate and she began to lose her verbal abilities. At 8 years of age her head circumference was below the 2nd percentile (it had been at the 50th percentile at birth), and she had a developmental age of 32 months. Over the next few years she lost all walking and speaking abilities. At age 17 years her total serum carnitine was 39.7 nmol/mL (normal 30 to 73) and free serum carnitine was 36.8 (normal 19 to 60). She was treated with 500 mg of L-carnitine 3 times a day (50 mg/kg/day). Within 2 months she showed significant improvement. She started to respond to verbal commands and to say words. Her total serum level was 66.6 and free level was 59. After 2 months the carnitine was discontinued and within a week she became lethargic and disinterested in her environment. Her levels had fallen to 35.1 and 29.9 respectively. Resumption of carnitine restored within a week her previous improvements.

(Plioplys AV and Kasnicka I. South Med J 86:1411-1413, Dec 1993)

Comment: With no dearth of patients with Rett syndrome, the authors should have tried carnitine in more than one patient before publication, and I should not have abstracted this paper. On the other hand, the parents of Rett patients, watching helplessly the deterioration of the children, are desperate and will quickly insist on the therapy so that we should soon know if it has any merit. Perhaps, too, this patient doesn't have Rett syndrome.

For earlier papers on Rett syndrome, see NOTES, vol. 16:p.121 (cerebellar and cerebral abnormalities); vol. 15:p.22 (genetic transmission) and p.44 (respiratory abnormalities); and vol. 12:p.69 (criteria for diagnosis) and p.150 (hyperventilation). These will refer you back to more items if you want them.

PEDIATRIC NOTES™ is published weekly but mailed every two weeks by first class mail (airmail to foreign countries). Subscription fee (binder and index included) for U.S., Canada, and Mexico for 52 issues: $75.00 (U.S. Funds). Students and house officers in U.S., Canada, and Mexico: $34.00 (U.S. Funds). To all countries other than U.S., Canada, and Mexico (all subscriptions are sent by airmail only and binder by surface mail): $85.00 (U.S. Funds drawn on a U.S. bank). All Subscriptions to PEDIATRIC NOTES 1994 begin with issue No. 1, January 6, 1994. Subscriptions entered at any time during 1994 are retroactive to issue No. 1. Payment must accompany subscription orders. Correspondence and subscription requests should be sent to:

PEDIATRIC NOTES POST OFFICE BOX 59 NEWTONVILLE, MA 02160

Dr. Sidney Gellis is one of the most prominent academic pediatricians in the United States (and the world). He works at one of the Harvard Hospitals and edits Pediatric Notes. This publication is sent to most pediatricians in this country. The cited article appeared in print at the very end of December. He picked up on it and published a summary on January 6, 1994. Clearly, he was very interested in these results. (Dr. Gellis is incorrect when he states that there is "no dearth" of Rett syndrome patients--I have only seen three cases in four years of working in Chicago). Thus, a small step at Marklund Nursing Home for Children, may lead to a bigger step for mankind (to paraphrase Neil Armstrong). --AP