84

A New Variation on an Old Theme: Subtleties of the Smith-Lemli-Opitz Syndrome (RSH/SLOS). G. Yoon, M.M. Nezarati, E. Fung, F.F. Snyder, G.E. Graham, Department of Medical Genetics, University of Calgary, Calgary Alberta, Canada.

SLO is a well-characterized autosomal recessive disorder of cholesterol biosynthesis with a classical phenotype that is distinctive and easily recognizable. However, it was not until the recent recognition of the biochemical abnormality that existence of the mild phenotype could be confirmed. Five individuals with biochemically proven mild or "variant" SLO have been described with 7-DHC levels that are intermediate between those measured in classical SLO patients and controls. The variant phenotype also seems to be characterized by cholesterol levels within or just below the normal range, while classical patients are clearly cholesterol-deficient. We now report three additional siblings, products of a first cousin mating, who have clinical and biochemical findings consistent with variant SLO. The oldest, a 12-year-old girl, has microcephaly, mild facial dysmorphism and moderate 2-3 syndactyly of the feet. Her past history is significant for failure to thrive, mild global developmental delay and behavioral problems, including ADD. 7-DHC and cholesterol levels were 64 μ M (controls <10) and 2.7 mM (controls 3.0 - 4.6), respectively. Her 7-year-old brother has microcephaly, a cleft lip, a unilateral single transverse palmar crease and mild 2-3 syndactyly of his feet. He has not been ascertained as a child with developmental delay or behavioral problems. Interestingly, his plasma 7-DHC level was just above the control range at 10 μ M and his plasma cholesterol level was just within the control range at 3.0 mM. We are currently repeating these measurements on a second sample. The youngest child, 5 year old boy, has microcephaly, characteristic facial dysmorphism, a left single transverse palmar crease as well as mild 2-3 syndactyly of his feet. He has a clear history of developmental delay and behavioural problems. His plasma 7-DHC was 52 and 24 μ M on two separate occasions and his plasma cholesterol was 2.9 both times. These children help to further delineate and improve recognition of the mild/variant SLO phenotype, which may account for a substantial percentage of children with minimal dysmorphism, microcephaly and developmental delay.

85

Assessment of Service Load in Clinical Genetics Practice: Report of a Survey Conducted in the GENES Region. R.E. Zinberg¹, K. Greendale², E.S. Salsberg³, R. Rosenthal², K.A. Pass² and the GENES Clinical Services Committee. ¹Mount Sinai School of Medicine, New York, NY, ²New York State Department of Health, Albany, NY, and ³State University of New York at Albany School of Public Health, Albany, NY.

The GENES Clinical Services Committee was convened to monitor genetic services delivery in New York State, Puerto Rico and the Virgin Islands, and to examine the impact of changes in the healthcare delivery system on quality of care in medical genetics. In response to concerns regarding changing practice patterns due to financial constraints in the new medical marketplace, a survey was designed to collect baseline data on current and projected caseload. Such data have not previously been collected in our specialty. A complete list of clinical geneticists in the GENES Region was generated by accessing all relevant databases and directories. 159 met specific criteria and were mailed a survey designed to assess:

(1) demographic profile; (2) % time allocated to various activities such as research or administration; (3) patient volume; (4) appointment waiting times; and (5) impact of changes in practice patterns on quality of care. Those not responding received a second survey and then a reminder telephone call. Individuals not practicing as clinical geneticists were removed from the data set. Preliminary data suggest that most clinicians have experienced an increase in patient volume over the last two years. Financial and quality-of-care implications will be discussed.