

Pseudopituitary Dwarfism Due to Resistance to Somatomedin: A New Syndrome

Bierich et al report a patient with elevated circulating growth hormone (GH) and somatomedin-C (Sm-C) concentrations. Although birth length and weight were normal (48 cm and 3 kg), all parameters of growth fell behind quickly. At 12 months of age, the infant's length was 58 cm and the weight 5.6 kg. The bone age was 6 months. Dental eruption occurred at 13 months. Hypoglycemia occurred during the second year. Circulating concentrations of Sm-C were increased for age when measured by bioassay at 10 months (1.99 and 2.03 U/ml). Sm-C by specific radioimmunoassay was elevated for age (1.28 U/ml). Administration of 4 IU of GH daily for four days did not increase the levels.

Fibroblasts from a skin biopsy taken when the patient was 21 months old were incubated with ¹²⁵I Sm-C. Compared with multiple controls, binding to the patient's fibroblasts was diminished by 50%. The

authors attribute the abnormality to defective Sm-C receptors.

This syndrome differs from Laron type dwarfism and the dwarfism described by Hayek et al (*J Peds* 1981;99:868) and Kowarski et al (*JCE&M* 1978;47:461). Sm-C concentrations are low in patients with Laron type dwarfism and do not increase after human growth hormone (hGH) administration. Sm-C levels were low in the patients described by Hayek et al and Kowarski et al, but they did respond to GH injections with increased Sm-C levels. In the patient currently presented, the Sm-C concentration was elevated. The authors term all of these types of dwarfism pseudopituitary dwarfism.

Two different actions of Sm-C are discussed. First are the acute effects upon skeletal muscle, heart muscle, and adipocytes. The second are the long-term metabolic effects that act through fibroblasts and chondrocytes. The authors be-

lieve that Sm-C works through the insulin receptors and affects the classic insulin-dependent tissues in the acute processes. The long-term or later effects influence fibroblasts and chondrocytes, which are induced to proliferate. In the long term, Sm-C is postulated to act primarily through the specific IGF-I receptors.

Bierich JR, Moeller H, Panke MB, et al: *Eur J Pediatr* 1984;142:186.

Editor's comment—This new syndrome is another in the ever increasing list of syndromes in which the patients have GH-deficient-like phenotypes. It is probably one of the least common of such syndromes, but it obviously exists. The authors refer in their bibliography to other patients who may have the same syndrome. We have observed one patient at the University of Virginia who unequivocally has this syndrome. We prefer to use the term "short stature with GH-deficient-like phenotype" for all of these patients who do not have GH deficiency.