

Editor's comment: Pediatricians, generalists, and internists need to be aware of the deleterious effects of low birth weight on the subsequent morbidity and mortality of adults. These carefully collected and analyzed epidemiologic data of adult men are exceedingly important. Although socioeconomic factors did not influence the incidence of the insulin resistance syndrome, it remains unclear whether personal psychological factors such as the desire to feed and/or overfeed a small baby and/or the need to restrict caloric intake in the large infant might have a bearing on subsequent outcomes.

The findings that smoking and socioeconomic status did not

influence the results are of particular importance. As the insulin resistance syndrome and type 2 diabetes, in particular, have become more prevalent both in children and adults, it is important that significant effort be placed into determining factors that contribute to the onset and persistence of these adversities. A better understanding of the relationships between the variables presented in this article is needed, and perhaps the nutritional principles taught to pediatric residents need to be carefully reviewed if a significant impact is going to be made in the reduction of the near-epidemic disorder of insulin resistance in children.

William L. Clarke, MD

Mosaicism Is a Likely Explanation for the Variability Observed in Androgen Insensitivity Syndrome

Holterhus et al report on 5 patients with somatic mosaicism for abnormalities of the androgen receptor. In all 5 patients, there was a lack of family history; and in all 5 clitoromegaly or micropenis with scrotalization of the labia was present. Each of the 5 patients had a different mutation that had arisen during postzygotic development. It appears that somatic de novo mutations of the androgen receptor occur at a particularly high rate. Thus, somatic mosaicism should be considered when there is more masculinization than expected from a particular mutation.

It is desirable to study both blood leukocytes and tissue fibroblasts to determine whether an individual is mosaic. Individuals with androgen insensitivity mosaicism may need to undergo early gonadectomy in order to avoid further masculinization. Variable expression of wild-type gene products, based on somatic mosaicism, is probably the mechanism for much of the variability that is seen in androgen insensitivity syndrome.

Holterhus P-M, et al. *Pediatr Res* 1999;46:684-690.

Editor's comment: The more we study, the more we learn. Somatic mosaicism appears to be quite common in a number of disorders, but seems to be variable depending on the particular gene. It is important to be aware that the androgen receptor gene seems to be particularly mutable during the course of development and, thus, we can explain the variability seen as related to a specific mutation. Geneticists like to think that there can be genotype-phenotype correlations that are straightforward, but somatic mosaicism leads to confounding situations. Keep an eye out for unexpected variations and consider somatic mosaicism as a possible explanation.

Judith G. Hall, OC, MD

Perceptions of the Outcome of Orthopedic Surgery in Patients With Chondrodysplasias

Hunter has taken the time to carefully interview 197 individuals with disproportionate short stature. Seventy-four of the 197 had undergone a total of 221 surgical procedures. In general, individuals felt they had improved outcomes. However, the attitude very much depended on the particular disorder. The percentage of individuals undergoing surgery ranged from 8.3% for hypochondroplasia to 87.5% for diastrophic dysplasia. The worst outcomes were for foramen magnum-cervical surgery and the best for thoracolumbar procedures to release nerve compression.

Gross points out in his editorial that leg lengthening has been revolutionized by the Ilizarov technique. The complication rate has dropped dramatically as experience has increased, decreasing to only 7% for patients with leg lengthening related to leg length discrepancy or short stature.

Hunter AGW. *Clin Genet* 1999;56:434-440.
Gross R. *Lancet* 1999;354:1574-1575. Editorial.

Editor's comment: Quality-of-life issues have become very important in healthcare outcomes analysis. Clearly, many patients reported by Hunter indicated that they experienced major improvements from orthopedic procedures. However, the perceptions related to specific disorders. The outcomes and natural history also must be related to specific disorders.

Most of the leg lengthening experience in disproportionate short stature is related to hypochondroplasia and achondroplasia (ACH). The procedure can add an extra 4 to 6 inches, which can make an enormous difference in the daily life of individuals whose height is approximately 4 ft. It is terribly important that data continue to be accumulated and combined since each type of disproportionate short stature is relatively rare. Collaborative studies on an international basis are really needed.

Judith G. Hall, OC, MD

2nd Editor's comment: Physicians dealing with short stature need to be aware of these 2 articles and the lead article by Dr. Deborah F. Stanitski, "Limb Lengthening in the Skeletal Dysplasias and Short Stature Conditions: State of the Art in 1997," that appeared in *GGH* (1997;13[2]:17-22). You are invited to read again the excellent presentation by Dr. Stanitski.

The article by Hunter is a general article, reporting levels of patient satisfaction for procedures performed for 2 different types of chondrodysplasias. The information will be of use to you in helping patients who are contemplating surgery, whether of the spine or extremities. However, it will not tell you the information you need to advise the patient.

The commentary by Gross in *Lancet* may be more helpful to