

The Frequency of Genetic Disorders in Children and Young Adults

Utilizing the British Columbia Health Surveillance Registry, a survey was taken of more than one million consecutive births to determine the frequency of genetic diseases in individuals younger than 25 years of age. The survey revealed that 5.3% of individuals develop a disease with an important genetic component. Of the 5.3%, 0.36% are single-gene disorders and 4.6% are clearly defined multifactorial disorders such as cleft lip or diabetes.

The survey was carefully designed to exclude congenital anomalies that were thought to be nongenetic in origin. If data for all congenital anomalies were included in the tabulation, 7.9% of individuals could be identified as having either a congenital anomaly or a known genetic disorder. A further breakdown of the single-gene disorders gives an incidence of 1.4 per 1,000 for autosomal dominant disorders, 1.7 per 1,000 for autosomal recessive disorders, 0.5 per 1,000 for X-linked disorders, and 1.8 per 1,000 for chromosomal abnormalities. Adult-onset diseases were not included in this survey.

It is quite clear from the study that genetic disorders occur very frequently if grouped together, although each individual disorder is relatively rare.

Baird PA, Anderson TW, Newcombe HB, et al. *Am J Hum Genet* 1988;42:677-693.

Editor's comment—*This survey is very useful because it gives a handle on the incidence and frequency of genetic disorders in the general population. It provides the kind of information that policy makers and healthcare planners need.*

Judith G. Hall, M.D.

Knemometry in Childhood: Accuracy and Standardization of a New Technique of Lower Leg Length Measurement

The authors carried out a study of the accuracy of lower leg length measurement by the apparatus introduced some five years ago by Valk and his colleagues. Ninety subjects between the ages of 2.4 and 17.1 years were involved; 46 were referred because of tall or short stature and 44 were normal. Six measurements were taken from each subject at each measurement session; the total number of measurement sessions was 2,200. Subjects and apparatus were displaced and repositioned between each measurement.

The ranges within the six measurements were 0.1 mm (rare) through 0.3 and 0.4 mm (very common) to 1.0 mm, with a few measurements beyond 1.0 mm. Using the median instead of the mean, the average SD of a single estimation was 0.16 mm. When differences between each of the six measurements and the medians of all six were examined, it was clear that the first of the six measurements was both biased and unreliable; when it was dropped, the SD decreased to 0.13 mm. Moreover, when only four measurements were taken and the first discarded, the results were as reliable as when the six measurements were taken and the first discarded. The procedure now recommended for future studies, therefore, is to take four measurements, discard the first, and average the next three.

Although its effect disappears in about two hours, exercise immediately before the lower leg length measurement is taken seems to reduce the measurement in children; strangely, it produces the opposite effect in adults. Therefore, vigorous physical activity should be avoided before measurements are taken. However, the child should be instructed to stand or walk slowly for five to ten minutes before being measured to

provide standardized loading to the lower leg; sitting should not be allowed.

Twelve children were measured on successive days. Over 24 hours the average gain in lower leg length was 0.67 mm, with a range of approximately 1.5 mm to -1.5 mm. Most of the children were also measured weekly and straight lines were fitted to each individual's series of measurements over a period of six months. Deviations among these lines greatly exceeded amounts that could be attributed to measurement error. Thus, the authors state that "estimates of growth velocities of the lower leg on the basis of two measurements taken some weeks apart...are barely reproducible." However, longitudinal series of measurements on individuals, taken weekly over long periods, are a very useful way to investigate growth.

Hermanussen M, Geiger-Benoit K, Burmeister J, et al. *Ann Hum Biol* 1988;15:1-16.

Editor's comment—*This is the definitive study, to date, of the reliability of the Valk technique for lower leg length measurement. The results speak for themselves. The four-measurement technique, in which the first is discarded and the next three averaged, should be adopted as standard. The accuracy of a given measurement is then high; however, due to fluctuations in growth rates, differences between two short-term velocities are unreliable. This conclusion agrees with that of Wales and Milner (abstracted in Growth, Genetics, and Hormones, Vol. 4, No. 1). Anyone using the Valk knemometer or a similar apparatus should read this article before embarking on clinical studies.*

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