Condition: Beckwith-Wiedemann syndrome

Inheritance:

Autosomal dominant/sporadic.

Genetic etiology:

Abnormalities involving an imprinted region of chromosome 11p15. Abnormalities include: paternal isodisomy for 11p15; cytogenetic abnormality (duplication, translocation, or inversion); abnormal methylation of *KCNQ10T1* or *H19;* mutation of *CDKN1C* gene.

Frequency:

Approximately 1/13,000.

Clinical features:

Birth weight is increased and children continue to grow rapidly, slowing around 7-8 years of age. Other associated features include macroglossia, hemihyperplasia, neonatal hypoglycemia, omphalocele, and increased risk of Wilms tumor and hepatoblastoma.

Management:

Treatment of acute problems in infancy, including hypoglycemia and congenital anomalies. Some require surgical reduction of tongue size. A program of monitoring for neoplasia should be instituted.

Genetic counseling:

Approximately 85% of cases are sporadic, but 10-15% are familial, transmitted as autosomal dominant traits, in some cases due to *CDKN1C* mutation.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.