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A Note on Beckwith-Wiedemann Syndrome

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Perspective

Beckwith-Wiedemann Syndrome (BWS) is the most common overgrowth and cancer predisposition disorder. BWS is caused by changes on chromosome 11p15. 5 and is characterized by a wide spectrum of symptoms and physical findings that vary in range and severity from person to person. Beckwith-Wiedemann syndrome (BWS) is a growth disorder that can affect several parts of the body. Babies and children are larger than normal usually until age 8, when growth slows down, resulting in an average height in adults. Symptoms may include one side or area of the body growing more than the other side, omphalocele or other abdominal wall defect at birth, an abnormally large tongue, low blood sugar in infancy, abnormally large abdominal organs, creases or pits in the skin near the ears, and kidney abnormalities.

Affected children have an increased risk to develop tumors, particularly a rare form of kidney cancer called Wilms tumor, a cancer of muscle tissue called rhabdomyosarcoma. The cause of BWS is complex and is different for different people, but involves genes that control body growth. In most cases BWS is caused by problems with the genomic imprinting of these genes. Genomic imprinting refers to having some genes that are active only when inherited from the father and others that are active only when inherited from the mother. Changes or mutations in the *CDKN1C* gene or larger changes to chromosome 11, such as a deletion, translocation or duplication, may cause BWS. Diagnosis of BWS is based on symptoms with the support of genetic testing. Beckwith-Wiedemann syndrome is a growth disorder variably characterized by neonatal hypoglycemia, macrosomia, macroglossia, hemihyperplasia, omphalocele, visceromegaly, embryonal tumors, adrenocortical cytomegaly, renal abnormalities and ear creases/pits.

BWS is considered a clinical spectrum, in which affected individuals may have many of these features or may have only one or two clinical features. Early death may occur from complications of prematurity, hypoglycemia, cardiomyopathy, macroglossia, or tumors. The previously reported mortality of 20% is likely an overestimate given better recognition of the disorder along with enhanced treatment options. Macroglossia and macrosomia are generally present at birth but may have postnatal onset. Growth rate slows around age seven to eight years. Hemihyperplasia may affect segmental regions of the body or selected organs and tissues. Beckwith-Wiedemann syndrome is associated with abnormal regulation of gene transcription in two imprinted domains on chromosome 11p15.5.

Most individuals with BWS are reported to have normal chromosome studies or karyotypes. Approximately 85% of individuals with BWS have no family history of BWS. Children of subfertile parents conceived by assisted reproductive technology (ART) may be at increased risk for imprinting disorders, including BWS. Identification of the underlying genetic mechanism causing BWS permits better estimation of recurrence risk. Prenatal screening for pregnancies in the general population which identifies findings suggestive of a diagnosis of BWS may lead to the consideration of chromosome analysis, chromosomal microarray, molecular genetic testing. Specific prenatal testing is possible by chromosome analysis for families with an inherited chromosome abnormality by molecular genetic testing for families in which the molecular mechanism of BWS has been defined.

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