Beckwith-Wiedemann Syndrome

What is BWS?

Beckwith-Wiedemann syndrome (BWS) is a genetic disorder, characterized by overgrowth in certain areas of the body. BWS has a wide range of symptoms and severity. Many features of BWS become less obvious as children get older. Many adults have normal growth and appearance.

Children with BWS may have a range of the following features:

- Large body size. (*macrosomia*)
- Overgrowth of one side of the body (*HemiHypertrophy*).
- Large tongue (*macroglossia*). This can cause speech and feeding problems.
- Pits or creases in the earlobes or behind the ear.
- Low blood sugar during the first month of life (*hypoglycemia*).
- Large abdominal organs, including the kidneys, liver, adrenal glands, and pancreas.
- Abdominal wall defects. (*omphalocele or umbilical hernia*).
- During childhood, there is an increased risk of developing certain cancers. This can include Wilm's tumor, tumors of the liver, and other more rare cancers.

What causes BWS?

- **The change is not caused by anything that a mother does during her pregnancy. There is nothing a parent can do to prevent the genetic change.**
- Most children diagnosed with BWS are the first in their family with the diagnosis. Most of the time BWS happens because of a brand new genetic change.
- Rarely, the genetic change is passed from parent to child. In these cases, there is up to a 50% chance, for each pregnancy, that the affected parent will pass on the abnormal gene to their child.
- Because the genetics of BWS is often complex, our team provides parents with genetic counseling to better understand their individual case. Genetic testing may explain the cause of BWS and, if inherited, provide information about future pregnancies.

How is BWS diagnosed?

Physical features: Some features are found more often in children with BWS than in the general population. When a child has a group of features frequently found in children with BWS, he or she may be referred for genetic testing.

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Genetic testing: The genetic test requires a blood sample. This sample will be sent to a lab in order to look for genetic changes on Chromosome 11. Genetic testing will find the cause in a majority of BWS patients. However, it is important to know that a normal test result does not rule out BWS because some genetic changes are still unknown. Children who do not have the genetic change for BWS will have the same medical management and cancer screening recommendations as children who do.

How is BWS treated?

Every child is different! Treatment and outcomes vary and depend on how severe the symptoms are.

- The treatment of BWS is directed toward the specific symptoms that a child may have.
- Medical providers from different specialties may be involved in your child's care. This depends on his or her specific signs and symptoms. For example; your child may meet with surgeons, kidney doctors, dental specialists, speech specialists, genetic specialists, pediatric oncologists, and orthopedic specialists.

Important information to know

Children with BWS usually have normal intelligence and a normal lifespan. They are capable of leading successful, happy, and fulfilling lives. Most children with BWS grow up to be healthy adults. By adolescence, growth normalizes and the risk for childhood cancer decreases.
Rarely, some adults have intellectual or physical delays. Most physical features decrease with time. Some of the features of BWS, such as leg length differences or an enlarged tongue may need surgery to correct.

Is my child at risk for cancer?

Since BWS is an overgrowth syndrome, children are at risk for developing certain childhood cancers. The two most common forms of cancer are Wilm's tumor (kidney tumor) and hepatoblastoma (liver tumor).

- Wilm's tumor is the most common cancer in children with BWS. The risk of developing Wilm's tumor decreases by age eight, but rarely, it can develop at a later age.
- Hepatoblastoma is the second most common form of cancer that can develop. In most cases, this cancer develops by the age of two.
- Other forms of cancer have been found in children with BWS, but are very rare.

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Will my child be tested for cancer?

Children with BWS have about a 5 to 10% chance of developing a cancer in early childhood. For this reason, routine screening is very important! Screening can help us find cancer early. Wilm's tumor and Hepatoblastoma are very treatable forms of cancer if caught early. **Cancers treated at an earlier stage will usually need less treatment and have a higher rate of survival.**

1. **Abdominal Ultrasound:**

Abdominal ultrasounds are safe and painless. They do not expose your child to radiation. We recommend children with BWS have an abdominal ultrasound every 3 months until age eight. This test is done to look for abdominal tumors, including Wilm's tumor.

2. **Measurement of alpha-fetoprotein (AFP)**

We recommend a blood test, AFP (alpha feta protein), every 6 weeks until age four. AFP is a hormone secreted by certain tumors. The AFP test is an easy and effective way to screen for hepatoblastoma.

3. **Routine Follow Up**

We recommend that all children with BWS be seen by a pediatric specialist every 6 months until age eight, and then yearly after that. A pediatric specialist can talk to you about your child's health and examine your child for other signs of cancer.

Where can I read more about BWS?

- Beckwith-Wiedemann Syndrome Family Forum: www.beckwith-wiedemann.info
- Beckwith Wiedemann Support Group: www.bws-support.org.uk
- Beckwith-Wiedemann Children's Foundation: www.beckwith-wiedemannsyndrome.org

For General Health Information:

Call or visit the Family Resource Center (a free health information library) located on the 3rd floor of St. Louis Children's Hospital. Call 314.454.2350. Email at frc@bjc.org

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