Family Education Sheet

Beckwith-Wiedemann Syndrome (BWS)

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This family education sheet explains what BWS is and what it means for your baby after birth.

Key points:

- The condition can be inherited, but most often it occurs randomly.
- You should be followed by a maternal fetal medicine doctor if your fetus is affected by BWS.
- Your child should see an oncology specialist for frequent screenings since they're at higher risk of cancer. Tumors are very rare after age 10.
- Children with BWS can live full, healthy lives.

What is BWS?

- Beckwith Wiedemann syndrome is a condition that is caused by genetic changes in a specific region on chromosome 11.
- It occurs in about 1 in 11,000 newborns.
- The condition can be inherited, but most often it occurs randomly.

What features are associated with BWS?

Many of the common features are due to overgrowth of certain body parts. Babies usually have a combination of these features:

- Large birth weight and length
- Large tongue
- Large organs (kidneys, liver, pancreas)
- Overgrowth of 1 side or part of the body
- Earlobe creases or pits behind the ear
- Low blood sugar levels (hypoglycemia)
- Abdominal wall defects, causing some organs to be outside of the body at birth
- Increased risk of some childhood cancers

How is it caused?

Beckwith-Wiedemann can be caused by many types of genetic changes in a specific region of chromosome 11 called 11p15.

Babies usually have 2 copies of chromosome 11, 1 from each parent. The paternal or sperm-derived genes have different functions than the maternal or egg-derived genes at this location. Some of the changes that cause Beckwith-Wiedemann are:

- **Mosaicism** – Some cells in the body have changes in the 11p15 region, while others do not.
- **Methylation** (about 55% of cases) – Certain genes in the 11p15 region are turned "off" instead of "on," or vice versa.
- **Uniparental disomy (UPD)** (about 20% of cases) – Both copies of 11p15 are inherited from 1 parent. Because the sperm and egg-derived copies of 11p15 have unique roles, losing either copy can cause BWS.
- **Chromosomal differences** (about 1% of cases) – There is extra, missing or rearranged genetic material at the 11p15 region.
- ***CDKN1C* gene variant** (about 5% of cases) – A change in this gene located in the 11p15 region may cause features of BWS.

About 20% of the time, the cause of BWS is unknown.

How can you tell if my child has BWS?

Doctors might suspect that your baby has BWS before birth if ultrasound findings show that the baby is large for its gestational age, has an abdominal wall defect (omphalocele) and/or overgrowth of 1 side of the body (hemihyperplasia).

The diagnosis can be confirmed by chorionic villus sampling (CVS) or amniocentesis during pregnancy.

After birth, testing may be done by testing blood or skin cells. Genetic tests that may be ordered to confirm a diagnosis may include:

- **Chromosomal microarray (CMA)** – can detect large pieces of extra or missing genetic material
- **Methylation analysis** – can detect whether certain genes in the 11p15 region are turned "off" or "on"
- ***CDKN1C* gene sequencing** – looks for any small changes in the CDKN1C gene

A negative genetic test result doesn't always mean that a baby is unaffected. Your baby may need to be seen by a geneticist after birth. Other genetic testing may be ordered after your baby is born.

What is the likelihood that another child will be affected?

About 85% of the time, BWS happens by chance and isn't inherited from a parent. In this case, future pregnancies are at a very low risk of being affected.

About 10-15% of the time, BWS runs in the family and can be passed down from parent to child. In this case, there's up to a 50% of BWS affecting a child of first-

degree relatives, such as siblings and future children of the affected person.

A geneticist or genetic counselor may be able to talk to you about whether you have an increased risk of BWS affecting other members of your family.

What should I expect during pregnancy?

If a fetus has BWS, the pregnancy has a higher risk of complications, such as premature birth, too much amniotic fluid and an unusually large placenta or umbilical cord.

You should be followed by a maternal fetal medicine doctor (MFM) if your fetus is affected by BWS. You should have a level II ultrasound and fetal echocardiogram should also be performed. Your child may need surgery after birth if there is an abdominal wall defect present.

How will my child be affected?

Babies with BWS are at an increased risk (about 5-10%) of developing certain cancers during early childhood. These cancers include Wilms' tumor, which is in the kidney, and hepatoblastoma, which is in the liver. Children with BWS should have frequent cancer screenings. Screening includes the following:

- **Abdominal ultrasound** – every 3 months until age 8
- **Serum AFP measurement** – every 3 months until age 4
- **Regular physical exams**

Children with BWS may be referred to other specialists to monitor their growth and development, including orthopedics, plastic surgery, nephrology and endocrinology. Some children need surgery to correct physical differences in growth, such as an abdominal wall defect (AWD). If your child needs surgery for an AWD, it may happen:

- **Shortly after birth** with just 1 operation if the AWD is small
- **In stages,** over several days or weeks if the AWD is large. The staged method may also be used if your baby is premature and doesn't have enough space in the abdominal cavity to fit all of the exposed organs right away.

Your baby may have breathing difficulties during their abdominal repair. Their organs may be swollen and the space inside the abdomen may be small and underdeveloped.

Because of this, your baby may need to use a breathing machine called a ventilator during the repair.

Children with BWS can live full, healthy lives. Many of the physical differences associated with BWS normalize in adolescence. Tumors are very rare after age 10. BWS doesn't affect your child's intelligence or reproductive ability.

Contact us

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