BWS AND YOU

An Educational Coloring Book
Dear BWS family,

This book was developed through our conversations with families of children with Beckwith-Wiedemann Syndrome (BWS). We understand that receiving a diagnosis of BWS can be overwhelming. The genetic and epigenetic causes of BWS are some of the most complex to understand and explain. Our descriptions are based on how we explain BWS to patients and families in clinic. We hope that this book helps you on your BWS journey and can be used as a resource to share information with family, friends, and other caregivers.

Through continued conversations and working together, we can further guide and enlighten our knowledge of BWS and continue to answer the many unanswered questions.

We welcome you into our BWS family and hope that through the partnership between our team and BWS families, we can further improve understanding and care for children with BWS.

Warm regards,

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DNA, or deoxyribonucleic acid, is a substance that carries genes from parents to their children.
Intro to Genetics

Genes are pieces of DNA that are passed down from parents to their children and represent traits or characteristics, such as hair color or growth.

Chromosomes are the parts of cells that contain genes. Cells are the smallest machines in your body.
Intro to Genetics

A child will inherit one chromosome from their mother and one chromosome from their father. Usually, a child receives 23 chromosomes from each parent, making a total of 46 chromosomes.
Genetics is the study of the process when a mother and father pass down their genes to their children. Epigenetics is the study of how the function of a person’s genes change. Some genes may or may not express certain traits. Epigenetic changes occur after conception as the embryo turns into a baby.
Beckwith-Wiedemann Syndrome is a disorder that is caused by a change in one or more of the genes at a region of chromosome 11, called 11p15. This specific chromosome region regulates growth, which is why Beckwith-Wiedemann Syndrome is referred to as an overgrowth disorder. Most times, this change happens in some cells, but not every cell.

This is what chromosome 11 looks like under the microscope. The p-arm is the shorter arm and the q-arm is the longer arm.
Most times, the genetic change on chromosome 11 occurs in some, but not every cell in the body. This situation is a result of mosaicism, which is when some cells of the body have normal chromosome 11 and other cells of the body have a change on chromosome 11. The features of Beckwith-Wiedemann Syndrome are a result of the chromosomes in some cells being different from other cells.
The growth of a baby with Beckwith-Wiedemann Syndrome is different from the normal growth of a baby. The diagram of the scale on the left shows how a typical child has a balance between genes that cause growth and genes that limit growth. The diagram of the scale on the right shows how a child who has Beckwith-Wiedemann Syndrome has an imbalance between genes that cause growth and genes that limit growth. This imbalance results in the child having too many genes that cause growth and not enough that limit growth.
People have two copies of chromosome 11, one from their mother and one from their father. Each copy of the chromosome that the child receives is different because one may express certain genes that control growth that the other does not.

This process is called imprinting and is caused by something called methylation. Methylation is a mark on a chromosome that can be thought of as a light switch. This marks the DNA to turn certain genes on or off. When the “light switch” for methylation is on, that means that the signal is “on.” When the “light switch” for methylation is off, that means that the signal is “off.” Having a balance between the “switches” that are on and off is what creates a baby that grows at a normal rate.

Children with Beckwith-Wiedemann Syndrome have changes on chromosome 11 where the “light switches” of methylation are marked or unmarked in a different way than a child without BWS. There are several different changes on chromosome 11 that are known to cause Beckwith-Wiedemann Syndrome.
Normally, a child has a balance of genes that cause growth and genes that limit growth. The DNA is marked so that the mother’s genes make the “don’t grow” signal and the father’s genes make the “grow” signal.

Some types of changes on chromosome 11 that occur with Beckwith-Wiedemann Syndrome may include:

- **IC2, LIT1, KvDMR, loss of methylation**

- **IC1, H19DMR, gain of methylation**
There are also rarer causes such as duplications, deficiencies, or chromosomal rearrangements that lead to an increase in “grow” signal and/or a decrease in “don’t grow” signal.
Some physical differences that may be present in children with Beckwith-Wiedemann Syndrome include:

- Macrosomia – large birth weight and length
- Hypoglycemia – low levels of sugar in the bloodstream
- Macroglossia – an enlarged tongue
- Pits or creases in the earlobe or behind the ear

Draw and color your face here.
Some physical differences that may be present in children with Beckwith-Wiedemann Syndrome include:

- **Omphalocele** – the intestines and other abdominal organs stick outside the body due to a weak spot in the abdominal wall.
- **Umbilical hernia** – when part of the intestine pushes through and creates a bulge, or sac, in a weak spot of the umbilical opening, otherwise known as the belly button.
- **Enlarged abdominal organs** – when parts of the body, such as the kidneys, liver, and pancreas, are larger than normal.
- **Hemihypertrophy/Hemihyperplasia** – when one side or one part of the body is larger than the other side, for example: when one leg is bigger than the other.
Managing BWS

Given that children with Beckwith-Wiedemann Syndrome have an increased risk of developing tumors during childhood, it is recommended that they have regular screenings.

Two types of tumor screenings that are recommended for children with Beckwith-Wiedemann Syndrome are the abdominal ultrasound and having a blood test to measure their alpha-fetoprotein (AFP) concentration.

The abdominal ultrasound is an imaging test that uses sound waves to display views of internal organs.
Alpha-fetoprotein (AFP) is a protein that is released by the liver in the fetus and baby. AFP is released at higher levels by hepatoblastoma (a liver tumor) cells, and AFP levels are normally high when the child is first born and trend downwards towards normal. It is important to follow the trend over time. This test should be ordered and reviewed by pediatricians, geneticists or pediatric oncologists who are familiar with Beckwith-Wiedemann Syndrome.
Some children with Beckwith-Wiedemann Syndrome may need to see other medical specialists.

These may include:

**Endocrinologists** —
Doctors who treat children that have hypoglycemia (low blood sugar)

**Geneticists** —
Doctors who make the clinical diagnosis, order testing and coordinate care

**Oncologists** —
Doctors who manage the prevention, diagnosis and treatment of tumors, and review the results of tests for tumors

**Orthopedists** —
Doctors who manage bone differences in children who have a difference in the size of their legs

**Pediatricians** —
Doctors who treat babies and children

**Plastic Surgeons** —
Doctors who treat children who have an enlarged tongue (macroglossia)

**Pulmonologists** —
Doctors who manage breathing differences
Abdominal ultrasound  
/an imaging test that uses sound waves to display
 views of internal organs

Alpha-fetoprotein (AFP)  
/a protein that is released by the liver in the fetus and
baby

Cells  
/the smallest machines in your body
and the basic unit of all living things

Chromosomes  
/the parts of
cells that contain genes

11p15 on chromosome 11  
/this specific chromosome region regulates growth

DNA  
/is deoxyribonucleic acid, is a substance that carries
genes from parents to their children

Endocrinologists  
/Doctors who treat children that have hypoglycemia
(low blood sugar)

Enlarged abdominal organs  
/when parts of the body, such as the
kidneys, liver, and pancreas, are larger than normal

Epigenetics  
/the study of how
the function of a person’s genes change, where some
genes may or may not express certain traits;
changes occur after conception as the embryo
turns into a baby

Genes  
/pieces of DNA that are passed
down from parents to their children and represent
traits or characteristics, such as hair color or
growth

Geneticists  
/Doctors who make
the clinical diagnosis, order testing, and coordinate
care

Genetics  
/the study of the process
when a mother and father pass down genes to their
children

Hemihypertrophy/Hemihyperplasia  
/when one side
or one part of the body is larger than the other
side, for example: when one leg is bigger than the
other

Hypoglycemia  
/low
levels of sugar in the bloodstream

Macroglossia  
—an enlarged
tongue

 Macrosomia  
— large birth weight
and length

Methylation  
/when DNA is
marked to turn genes “on” or “off”

Mosaicism  
/when some cells of the body have normal chromosome 11 and other
cells of the body have a change on
chromosome 11

Omphalocele  
/the intestines and
other abdominal organs stick outside the body due
to a weak spot in the abdominal wall

Oncologists  
/Doctors who
manage the prevention, diagnosis and treatment of
tumors, and review the results of tests for tumors

Orthopedists  
/Doctors who
manage bone differences in children who have a
difference in the size of their legs

Pediatricians  
/Doctors who treat
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Pulmonologists  
/Doctors who
manage breathing differences

Umbilical hernia  
/when
part of the intestine pushes through and creates a
bulge, or sac, in a weak spot of the umbilical
opening, otherwise known as the belly button
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Let's work together

FAMILIES

CHILDREN

DOCTORS

to spread BWS Awareness