MassGeneral Hospital for Children℠

Common endocrine issues in Williams syndrome: What you need to know

Various endocrine conditions are more common in people with Williams syndrome, a genetic condition that causes developmental differences. In this handout, you will learn about the most common endocrine conditions found in people with Williams syndrome.

At MGHfC, your child's care team in Pediatric Endocrinology works closely with Barbara Pober, MD, of Medical Genetics. This helps ensure your child receives the best care possible for their genetics and endocrine conditions.

WHAT ARE COMMON ENDOCRINE ISSUES IN PEOPLE WITH WILLIAMS SYNDROME?

Hypercalcinemia and hypercalciuria

People with WS may have **hypercalcemia** (high calcium levels in the blood) or **hypercalciuria** (high levels of calcium in the urine). High calcium levels in the blood are more common in infancy, but could occur any time in life.

Symptoms

Symptoms of hypercalcinemia

- In babies, irritability, vomiting, or constipation. These symptoms can also be caused by other medical conditions.
- In older children, symptoms can include abdominal (belly) pains, fatigue or frequent urination

Symptoms of hypercalcinuria

- There are usually no symptoms of hypercalciuria. Doctors can usually diagnose hypercalciuria through a urine test.
- Over time, the build-up of calcium can lead to a condition called **nephrocalcinosis**. This is <u>not</u> the same as having kidney stones. It usually will be followed by a nephrologist (doctor who specializes in the kidneys).

Treatment (for both conditions)

- Regular blood and urine tests
- A diet that is low in calcium. This means no more than 3 servings of dairy or calcium-fortified foods (foods with calcium added) per day. If you would like to give your child a multivitamin with calcium, please ask the care team for recommendations. Your child will need regular blood tests throughout their life to check their calcium levels and bone density (the amount of material in the bones).
- In rare cases, your child might need more medical care for issues caused by hypercalcinemia. This usually happens if your baby is not feeding well or gets sick. After treatment, calcium levels usually return to normal.
- In some cases, your child might need a renal ultrasound (kidney ultrasound). This helps make sure calcium is not collecting in the kidneys.

Where can I learn more about Williams syndrome?

- **Your child's care team at MGHfC**
- **Williams Syndrome Association**
  https://williams-syndrome.org
- **National Library of Medicine**
  https://ghr.nlm.nih.gov/condition/williams-syndrome

More information on the next page! >>>

Hypothyroid

Hypothyroid is a condition that causes low thyroid function. The thyroid is a gland in the neck that makes thyroid hormone. Thyroid hormone helps regulate the body's energy, metabolism and other functions.

Doctors can diagnose hypothyroid through a blood test. The blood test checks the following:

- Thyroid stimulating hormone (TSH) is a hormone from the brain that tells the thyroid to turn on. If TSH levels are high, it may mean the thyroid is not working properly.
- Thyroxine or free thyroxine (T4 or free T4) level is the actual level of thyroid hormone is flowing throughout the body. This tells how much hormone the body is making.
- Subclinical hypothyroidism. This condition is common in children with WS. It is a condition in which the TSH levels are a little high but the T4 level is normal.

Symptoms

- Fatigue
- Feeling cold more easily
- Constipation
- Dry, coarse (rough) hair and skin
- Hair loss

Treatment

In most cases, hypothyroid issues are mild in people with WS. They might not need medication to replace the body's missing thyroid hormone. If your child does need thyroid medication, it comes in a pill. Children with WS also need blood tests every 1-2 years to check their thyroid

Early Puberty

Many children with WS go through puberty a little earlier than normal. This is usually not a problem unless your child is not emotionally ready for puberty.

Doctors can diagnose early puberty through a physical exam and blood tests. In some cases, a bone age test (an X-ray of your child's left wrist) is also helpful. The bone age test helps doctor see how mature your child's bones have grown. People with early puberty often will have an "advanced bone age." This means the bones look more mature than the age of the person.

Symptoms

The first sign of puberty in girls is breast development. The first sign of puberty in boys is the testes getting larger.

Treatment

If your child starts puberty at a very young age, the care team might do tests to make sure puberty is not starting for other medical reasons.

In children with Williams syndrome, early puberty does not usually need treatment unless your child is not emotionally ready for puberty. If this happens, your child might be able to take medications called GnRH agonists. These medications are given as either a shot or an implant (small plastic rod about the size of a matchstick) in the arm. They stop puberty temporarily.

Are there any recommendations specifically for people with WS?

Like all children, children with WS should have annual exams with their primary care doctor. If the doctor notices the early development of puberty, a pediatric endocrinologist can help recommend further evaluation and discuss options.

More information on the next page! >>>

Hyperglycemia

Hyperglycemia is when there are higher levels of sugar in the blood than usual. Hyperglycemia is more common in teens and adults who have WS. Some teens and adults also have diabetes (a condition that affects how the body uses sugar for energy). Everyone has sugar in the blood to provide energy to the body

Starting at puberty, it is important to check blood sugar regularly in people with WS. Blood sugar can be checked in 3 ways, all of which involve a blood test:

- Fasting blood sugar (also called fasting glucose). This test checks the amount of sugar in the blood before someone has eaten for the day.
- Oral glucose tolerance test. This test is also done before people have eaten for the day. The care team checks the amount of sugar in the blood. Then, your child drinks a beverage with a certain amount of sugar in it. Their sugar is tested again 2 hours later.
- Hemoglobin A1c (HbA1c). This test can be done any time. It gives an average of what the sugar in your child's blood has been over the past 3 months.

Symptoms

Most people with high blood sugar will not feel any differently. If blood sugar is very high, your child might have frequent urination, poor energy levels and increased tiredness.

Treatment

People who have slightly high blood sugar should do their best to eat healthily and to be as active as possible. Walking and other exercises are great ways to lower blood sugar. People with Williams syndrome and diabetes also usually need to take a medication for diabetes. This can be a pill or insulin injections.

Muscle and Body Fat in WS

Body composition refers to how much fat and muscle a person has, and where it is distributed in the body. Children with WS tend to be thin. Sometimes adults with WS are also thin, but some adults may gain weight, especially in their belly, hips or legs. It is healthy and normal for everyone to have some body fat, including those with and without WS.

Doctors are not sure what makes some people store different levels of body fat. It is important for everyone with WS to keep as active as possible. For children, this can include playing and taking part in active sports or physical activities. Early participation can help your child develop lifelong active habits.

Bone Density in WS

Doctors are just starting to learn that bone density might be somewhat lower than expected in people with WS. However, doctors are not sure if fractures are more common in WS than in the general population. Doctors usually recommend monitoring bone density in adults with WS. This is usually done through a bone density scan, also called a DXA scan. This is a painless scan that is like an X-ray.

Strategies that can help improve bone density are:

- Physical activity, especially any weight-bearing activities help strengthen bones.
- Having a normal amount of calcium and vitamin D in the diet. If someone with WS has high calcium levels, it could be necessary to lower calcium and maybe vitamin D intake temporarily. These should go back to normal levels as the problem resolves. Doctors do not recommend that people with WS restrict calcium or vitamin D in their diet.
- Vitamin D supplementation done slowly with periodic blood work to check that calcium levels stay normal. Many people with WS have low vitamin D levels.

Pediatric Endocrine and Diabetes Center

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