

What is WAGR Syndrome

What is WAGR syndrome?

Below is an overview of WAGR syndrome. Please use the sidebars at right to access additional information about the conditions associated with this disorder.

WAGR syndrome is a rare genetic condition that can affect both boys and girls. Babies born with WAGR syndrome often have eye problems, and are at high risk for developing certain types of cancer, and a range of developmental delays. The term “WAGR” stands for the first letters of the physical and mental problems associated with the condition:

- **(W)**ilms’ Tumor, the most common form of kidney cancer in children.
- **(A)**niridia, some or complete absence of the colored part of the eye, called the iris (singular), or irises/irides (plural).
- **(G)**enitourinary problems, such as testicles that are not descended or hypospadias (abnormal location of the opening for urination) in boys, or genital or urinary problems inside the body in girls.
- **(R)**ange of developmental delays.

Most people who have WAGR syndrome have two or more of these conditions. Also, people can have WAGR syndrome, but not have all of the above conditions.

Other names for WAGR syndrome that are used are:

- WAGR Complex
- Wilms’ Tumor-Aniridia-Genitourinary Anomalies-Mental Retardation Syndrome
- Wilms’ Tumor-Aniridia-Gonadoblastoma-Mental Retardation Syndrome
- Chromosome 11p deletion syndrome
- 11p deletion syndrome

The cause of WAGR syndrome is deletion of a group of genes located on chromosome number 11 (11p13 – the “p13” refers to the specific place on chromosome 11 that is affected). Chromosomes are packages of genetic characteristics. There are 22 pairs of chromosomes that are the same in males and females. The 23rd pair determines a person’s sex with males having an X and Y chromosome and females having two X chromosomes.

What are the symptoms of WAGR syndrome?

What are the Symptoms of WAGR Syndrome?

WAGR is called a genetic syndrome. The symptoms of WAGR syndrome are usually seen after the baby is born. The mother's pregnancy and the baby's birth history are not unusual. Enlargement of the baby's kidneys may be seen on a prenatal ultrasound. The eye problems (aniridia) are usually noticed in the newborn period, and for infant boys, the problems with the genitals and urinary systems are also usually obvious in the newborn period.



Individuals born with WAGR syndrome are at higher risk for developing other problems during infancy, childhood, and adulthood. These problems can affect the kidneys, eyes, testes or ovaries. The specific symptoms that happen in a person who has WAGR syndrome depend on the combination of disorders that are present.

Wilms' tumor: About one half of individuals who have WAGR syndrome develop a type of kidney cancer called Wilms tumor. In the early stages of Wilms' tumor there are usually no symptoms. The first signs of this cancer may be blood in the urine, a low-grade fever, loss of appetite, weight loss, lack of energy or swelling of the abdomen.

Aniridia: In infants who are born with aniridia that is associated with WAGR syndrome, the irises of the eyes fail to develop normally before birth. This causes partial or complete absence of the round colored part of the eye (iris). Aniridia is almost always present in babies born with WAGR syndrome. Other eye problems are often present or can develop as the child grows older. These include: clouding of the lens of the eye (cataract); rapid, involuntary movements of the eye (nystagmus); and all or partial loss of vision due to high pressure of the fluid in the eye (glaucoma).

Genital and urinary (GU) problems: A range of GU problems may be present in a baby born with WAGR syndrome. For boys, these may involve the urinary tract opening somewhere along the shaft of the penis rather than at the tip (hypospadias) or undescended testes (cryptorchidism). In girls, these problems may include underdeveloped (streak) ovaries, and malformations of the uterus, fallopian tubes or vagina. In some people with WAGR syndrome, problems with the development of the genitals may make their sexual assignment at birth (male or female) uncertain. Individuals with WAGR syndrome may have a higher risk for a type of cancer called gonadoblastoma, a cancer of the cells that form the testes in males and the ovaries in females.

Range of Developmental Delays: Developmental delays are common in children with WAGR syndrome. The severity of the condition varies from person to person. Some children who have WAGR syndrome may have extremely mild conditions.

Other symptoms of WAGR syndrome may also include:

Other symptoms of WAGR syndrome may also include:

- Developmental, behavioral, and/or psychiatric disorders including autism, attention deficit disorder, obsessive compulsive disorder, anxiety disorders, and depression.
- Early-onset overweight (obesity) and high blood cholesterol levels.
- Excessive food intake (polyphagia/hyperphagia).
- Chronic kidney failure, most often after age 12 years.
- Breathing problems, asthma and pneumonia and breathing problems during sleep (sleep apnea).
- Frequent infections of the ears, nose, and throat, especially during infancy and early childhood.
- Teeth problems – crowded or uneven teeth.

- Problems with muscle tone and strength, especially during infancy and childhood.
- Seizure disorder (epilepsy).
- Inflammation of the pancreas (pancreatitis).

How is WAGR Syndrome diagnosed?

How is WAGR syndrome diagnosed?

Symptoms that suggest WAGR syndrome, like aniridia, are usually noted shortly after birth, and genetic testing for the 11p13 deletion is done. A genetic test called a chromosome analysis or karyotype is done to look for the deleted area (11p13) on chromosome number 11. A more specific genetic test called FISH (fluorescent in situ hybridization) is sometimes done to look for the deletion of specific genes on chromosome number 11.



How is WAGR Syndrome Treated?

Treatment of WAGR syndrome is aimed at the specific symptoms present in the individual. Monitoring to look for problems is also important to catch problems early so that treatment can be given as soon as possible.



Wilms' tumor: Wilms' tumor happens in about half of children with WAGR syndrome. The tumor usually develops between the ages of 1 and 3 years. Most cases of Wilms' tumor have been detected by age 8 years, but in rare cases may occur later. Babies who are suspected to have WAGR syndrome should have ultrasounds of their abdomen at birth. They then need to have abdominal ultrasounds every 3 months until they reach age 8 years. Feeling the abdomen for signs of swelling and masses can be done by both the baby's doctor and the parents, when they are taught how to do this. After age 8 years, watching for signs of Wilms tumor may be done by ultrasound and/or by watching for symptoms such as a low-grade fever, loss of appetite, weight loss, lack of energy or swelling of the abdomen.

Wilms' tumor can often be treated successfully. The overall survival rate of patients with Wilms tumor is excellent and is related to the features of the tumor, and the stage of the disease. Treatment may include surgery to remove the kidney, radiation therapy and chemotherapy.

Aniridia: The treatment of aniridia is aimed at keeping the person's vision. Drugs or surgery may help when there is glaucoma or cataracts. Contact lenses can harm the cornea and should be avoided.

Genital and urinary problems: Children with WAGR syndrome should have regular evaluations to detect abnormal development of their ovaries or testes. Surgery may be needed to remove abnormal gonads or to prevent cancer of the gonads (gonadoblastoma). When both gonads are removed, the individual is given hormone replacement treatment. Surgery may also be done when a boy with WAGR syndrome has undescended testes. When girls with WAGR syndrome have abnormal ovaries, they have routine pelvic ultrasounds or MRI's (magnetic resonance imaging) to watch for the development of gonadoblastoma.

Range of Developmental delays: Individuals with WAGR syndrome may have intellectual delays ranging from severe to mild. Some individuals with WAGR syndrome have normal intelligence.

Children with WAGR syndrome should be referred for Early Intervention Services soon after they are born, or when the diagnosis is made. Treatments include: vision therapy, physical, occupational and speech therapies. Special Education services are also used to help children with WAGR syndrome develop to their fullest ability.

Kidney (renal) failure: The renal failure that can happen in WAGR syndrome often causes the person to have high

blood pressure, high cholesterol, and leakage of protein from the blood into the urine (called proteinuria). All individuals with WAGR syndrome should be routinely screened for high blood pressure and urinary protein. These problems are treated with medications called “ACE inhibitors” or “ARBs.” Some people with WAGR syndrome and renal failure are treated with dialysis or kidney transplant.

Is WAGR Syndrome Inherited?

Is WAGR syndrome inherited?

WAGR syndrome is called a “contiguous gene deletion syndrome.” This means that it is caused by the loss of a section of genes on chromosome 11 (11p13). Most of the time the changes on chromosome 11p13 happen by chance when the egg or sperm are being formed or during the very early stages of the baby’s development in the womb. More rarely, the gene changes are inherited because one of the parents carries a rearrangement (called a translocation) between two chromosomes that can cause the loss of some genes when he or she has a baby. A baby can also have a mixture of normal cells and cells that have the 11p13 changes in his or her body. This is called mosaic WAGR syndrome.

Genetic counseling is helpful for determining whether there may be an increased risk of having another child with WAGR syndrome.

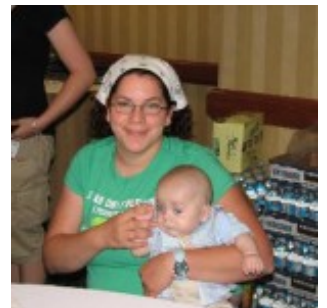
Kelly Trout, BSN, RN

Written in cooperation with the National Human Genome Research Institute

National Institutes of Health

<http://www.genome.gov/26023527>

Last updated: May, 2015



In Time and With Love

If your child is an infant and has recently been diagnosed with WAGR syndrome, you are probably feeling overwhelmed right now. The baby you eagerly anticipated only a short while ago is suddenly a child with a frightening diagnosis. His future may seem dark and full of unknowns. But your child with WAGR syndrome is first and foremost a child. In time, he will smile and laugh, he will learn and grow and enjoy life. And in time, you will too.

Your child with WAGR syndrome is also an individual. It is likely that he will have some, but not all of the symptoms you will read about. Understanding the spectrum of features that may occur with WAGR syndrome is the key to providing your child with the specific forms of help that he or she may need. *Do not expect to learn all of these at once.* In time, your child will show you what he needs, and you will learn how to help him.

If your child is older, you are already aware of many of the challenges and joys of raising a child with this condition. We hope the information on this website will be helpful to you, too.

Good medical care and individualized education are very important for people with WAGR syndrome. Please feel free to share the materials on this website with the professionals who work with your child.

Connecting with other parents of children/adults with WAGR syndrome can make a big difference for you and your child. Other parents can offer helpful experience, practical tips, and priceless understanding and support. On this website you will find a variety of ways to connect, from a private Facebook group, to an email newsletter, to annual in-person get-togethers called "WAGR Weekends."

This website is the online home of the International WAGR Syndrome Association. The IWSA was founded by parents, and is run by an all-volunteer staff of parents who are dedicated to improving the lives of people with WAGR syndrome. We are proud to share our mission, and hope you will consider joining us to fulfill it.

The good news about WAGR syndrome is that individuals who have this disorder can live happy and productive lives. They enrich the lives of everyone around them, and the world is a better place for having them here. The families of International WAGR Syndrome Association are living proof of this.

We look forward to sharing our help and our hope with you!

Coping with the Diagnosis

Coping with the Diagnosis of WAGR syndrome

During pregnancy, most parents have hopes and dreams for their child. When the baby is diagnosed with WAGR syndrome, those hopes and dreams are suddenly changed, and we feel shocked and confused.

Shock

Parents often feel “numb” for a while after receiving the diagnosis. This is your mind’s way of protecting you from more pain than you can handle. As shock fades, you may experience other reactions, such as forgetfulness, feeling as if you are in a bad dream, difficulty concentrating or denial.

Sorrow

When you are told your child has WAGR syndrome, it is natural to feel sad. Most parents need to go through a period of mourning for the child they expected to have. It will help if you express this feeling, and if the people around you allow you to cry and to talk about how you feel.

Anger

Most parents ask, “Why did this happen to us?” and many feel angry. It helps to talk about this feeling too. It is very common to want to assign blame to someone or something. Many parents wonder whether something they did before or during pregnancy caused their child to have WAGR syndrome. It’s important to remember that there is nothing you could have done or not done to cause or prevent this condition.

Guilt

Not all parents feel guilty, but many do. This is also a natural reaction, even though the fact that your child has WAGR syndrome is certainly not your fault.

All of these reactions and feelings are perfectly normal. They may never go away completely, but as time passes they will lessen. Joining a support group can be a good way to cope with these feelings because it’s comforting to know that others have felt the same way. It can also be helpful to learn from other parents and to share the lessons you have learned as well.

Your Marriage

The rate of divorce among parents of children with special needs is thought to be higher than average. There are probably many reasons for this. Men and women often handle crisis differently, and the birth of a child with WAGR syndrome is a crisis. Mothers frequently react with tears and with a great need to talk and express their feelings. Fathers on the other hand may feel the need to contain their emotions by not talking about them. These conflicting needs and coping styles can make each partner feel misunderstood and alone. But simply being aware of and accepting these different needs can be a big help. Couples do need to communicate and a little compromise can go a long way—and can help each partner to have a sense of working together for the benefit of the family. If you find that your marriage is really suffering, consider couples counseling to work through this difficult time. Parents of older children often say that finding constructive ways to weather this crisis brought them closer together.

Siblings

Children usually take their cue from their parents—if you are positive about the new baby, they will be too. You can explain to them that this new baby may need extra care, and that he may learn more slowly than they do. You can also reassure them that in time, this little brother or sister will be a great playmate and someone they will grow to love. Most research shows that children who have a sibling with disabilities get along well with them and are fond of their sibling. Many adults report that growing up with a sibling with disabilities taught them a great deal about

understanding, responsibility, and compassion.

Grandparents

Grandparents can go through many of the same feelings as parents do as they grieve for both their child and their grandchild. Denial of the child's diagnosis is common, and usually reflects an intense need to believe that all will be well. Whenever possible, include grandparents in your child's care, and share information with them. If they know that children with WAGR syndrome do best when their caregivers are both realistic and optimistic, they may be better able to accept their grandchild just as he is. A grandparent's positive and encouraging attitude can be a tremendous asset for a child.

Friends and Others

Friends, co-workers, strangers, and even relatives can unwittingly say thoughtless or hurtful things out of embarrassment and ignorance. Again, most people will take their cue from you. If you are positive about your child and his future, they will learn to be positive also.

This page updated: February, 2015

Kelly Trout, BSN, RN

Health Consultant

International WAGR Syndrome Association

What We Do

Our Mission:

The Mission of the IWSA is to promote international knowledge and awareness of WAGR/11p Deletion syndrome and its complications and treatments, to stimulate research and to reach out those affected by WAGR syndrome in an effort to improve their lives.

The IWSA:

- provides information about WAGR/11p deletion syndrome
- supports and encourages individuals and families affected by this disorder
- promotes research and public awareness
- maintains a website providing information and resources related to WAGR/11p deletion syndrome
- maintains/oversees a “private” parent/caregiver discussion/support group on Facebook, which hosts more than 150 members
- maintains a public Facebook page
- maintains Twitter and Pintrest accounts
- maintains/moderates a yahoo listserv discussion group for parent and caregivers
- produces an electronic newsletter, *WAGR Warrior*, on a regular basis to share current information and resources, fundraising, research, family stories and photos, and activities related to the IWSA and WAGR syndrome
- hosts our annual WAGR Weekend family get-together for families of individuals with WAGR syndrome. WAGR Weekend takes place each summer in a different city in the United States and provides an opportunity for families to meet in person in a casual and friendly atmosphere, to share a few days of camaraderie and friendship, to learn about and share new information, and to be amongst other families who understand the joys and challenges of caring for an individual with WAGR syndrome. Other “mini-WAGR Weekends” take place during the course of the year and have been held in the past in the United Kingdom and Japan as well as the US
- provides information packets for parents, including medical articles, guidelines for physicians, brochures and resource lists. This information is also available on the website at www.wagr.org
- provides access to IWSA Health Consultant, Kelly Trout (Kelly.trout@wagr.org) and IWSA Education Consultant, Rhonda Sena (Rhonda.sena@wagr.org) to address specific questions and/or concerns related WAGR syndrome
- offers participation in the CoRDS/IWSA Patient Registry, which allows patients and caregivers the opportunity to provide information on symptoms, diagnoses, and medical history in a secure online site. Registries can facilitate research and help develop treatments and therapies.

Due to our non-profit status as a public charity we are unable to assist in fundraising for specific individuals/families or assist with medical expenses.