

WAGR Syndrome

A Guide for Physicians

International WAGR Syndrome Association

<http://www.wagr.org>

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The International WAGR Syndrome Association gratefully acknowledges the assistance of the following physicians:

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Introduction

This booklet has been designed to assist both primary and specialty care physicians caring for individuals with WAGR syndrome. Information about diagnosis, associated conditions, treatment, and follow-up care throughout life are given, along with resources for additional information and assistance.

WAGR syndrome is a rare genetic disorder. "WAGR" is an acronym for the most common features of the syndrome:

Wilms tumor
Aniridia
Genital abnormalities and/or
Mental Retardation.

Individuals with WAGR syndrome may or may not be affected by the features above, and may also have other physical conditions, as well as behavioral and psychiatric disorders (see Conditions Associated with WAGR Syndrome).

These conditions are the result of a deletion of genetic material from chromosome 11. The primary criteria for diagnosis of WAGR syndrome is evidence of deletion of 11p13.

Synonyms for WAGR syndrome include:

WAGR Complex

WAGRO syndrome

Chromosome 11p deletion syndrome

11p deletion syndrome

Aniridia-Wilms Tumor Association/ATWA

Aniridia-Ambiguous Genitalia-Mental Retardation

AGR Triad

Diagnosis

Most cases of WAGR syndrome are identified in infants with sporadic aniridia, 30% of whom will be positive for the characteristic deletion (11p13). Additional features of WAGR syndrome may not be present or obvious, and should not preclude genetic testing.

In rare cases, aniridia may not be present. Children with Wilms tumor and other conditions associated with WAGR syndrome, such as genital anomalies, mental retardation/learning disabilities, or behavioral disorders may also warrant genetic testing.

Genetic testing for WAGR syndrome should begin with lymphocyte high-resolution chromosome study (at least 550 bands). If the chromosomes are normal, then additional fluorescent *in situ* hybridization (FISH) studies should be done to identify deletion of the PAX6 gene (aniridia) and WT1 gene (Wilms tumor).

Laboratories offering diagnostic molecular cytogenetic testing for WAGR syndrome are listed in **Resources**.

In some patients, there may be clinical overlap of symptoms between WAGR and other WT1-related syndromes. These include Beckwith-Wiedemann syndrome, Denys Drash syndrome, Frasier syndrome, Potocki-Shafer syndrome and/or Multiple Hereditary Exostosis (MHE).

Timely and accurate diagnosis of WAGR syndrome is critical, as many of the conditions associated with the disorder are amenable to treatment if detected early.

Conditions Associated with WAGR Syndrome

Wilms Tumor occurs in up to 50% of children with WAGR syndrome, usually by age 3. In rare cases, Wilms tumor may occur after age 8 in individuals with WAGR syndrome, and has been reported as late as age 25.

Aniridia (iris hypoplasia) is associated with multiple ocular complications. Visual acuity is generally in the 20/100 to 20/200 range, but may be significantly decreased by any or all of the following: foveal hypoplasia, optic nerve hypoplasia, cataract, subluxation of the lens, glaucoma, nystagmus, amblyopia and strabismus. Corneal abnormalities such as markedly increased thickness of the cornea, impaired corneal healing due to limbal cell deficiency, and corneal pannus are common. Other ocular abnormalities include microphthalmia, anterior segment anomalies, and retinal dysplasia.

Genitourinary Anomalies In males, genital abnormalities may include cryptorchidism, hypospadias, small penis, and hypoplastic scrotum. While external genital abnormalities are uncommon in females, internal genital abnormalities may include streak ovaries, and malformations of the uterus, fallopian tubes, or vagina.

Some infants with WAGR syndrome may present with ambiguous genitalia. Duplicate ureters or horseshoe kidney may occur in either males or females. Both sexes have an increased risk of gonadoblastoma.

Retardation/Developmental Delay Mild to moderate mental retardation (IQ 50 to 70) is common in WAGR Syndrome. Some individuals with WAGR Syndrome may have normal intelligence.

Other Conditions Associated with WAGR syndrome:

Head, eyes, ears, nose, throat Frequent, recurrent upper respiratory infections, otitis media, and/or sinusitis. Obstructive sleep apnea, severe dental malocclusion, delayed loss of primary teeth, micrognathia, hearing impairment, cleft palate.

Neurologic Complete or partial agenesis of the corpus callosum, microcephaly, periventricular nodular heterotopias. epilepsy, cerebral palsy, enlarged ventricles, cerebellar hypoplasia, Central Auditory Processing Disorder (CAPD), Sensory Integration Disorder, hypoalgesia (decreased pain perception).

Behavior disorders Autism spectrum disorders, learning disabilities, attention deficit disorders (with or without hyperactivity).

Psychiatric disorders Anxiety, depression, obsessive-compulsive disorder.

Renal Late-onset nephropathy, renal cysts, unilateral renal agenesis, hypoplastic kidney.

Musculoskeletal Hypotonia/hypertonia, hypertense Achilles, talipes, hemihypertrophy, syndactyly/clinodactyly, scoliosis, kyphosis, Multiple Hereditary Exostosis Type II

Metabolic Early Onset Overweight/Obesity, polyphagia/hyperphagia, hyperlipidemia, hypertriglyceridemia.

Gastroenterology Chronic pancreatitis, diaphragmatic hernia, gastroesophageal reflux disease (GERD).biliary atresia, fatty liver.

Cardiopulmonary Asthma, recurrent pneumonia, patent foramen ovale, pulmonary hypertension, valvular hypoplasia, ventricular septal defect, Tetralogy of Fallot, atrial septal defect, tracheomalacia.

Treatment Considerations

Genetic Counseling: WAGR syndrome is most often the result of a sporadic genetic mutation, and thus is not inherited. In rare cases, it may be the result of a balanced translocation in an unaffected parent. Mosaic deletions have been reported, as well as WAGR syndrome arising in the offspring of parents with familial (autosomal dominant) aniridia. Genetic counseling is recommended for parents of children with WAGR syndrome.

Ophthalmologic Care: For infants with aniridia, an initial eye exam under anesthesia will allow the most complete assessment of the patient's ocular conditions. Subsequent exams under anesthesia may be warranted if glaucoma or other potentially progressive conditions are present.

Glaucoma occurs in up to 70% of patients with aniridia. For this reason, ophthalmologic exams are recommended at six to twelve month intervals throughout life.

The cornea of the aniridic eye presents special challenges. Markedly increased central corneal thickness is a recently recognized aspect of congenital aniridia. This may lead to incorrect estimates of intraocular pressure by applanation technique. Patients with aniridia should be monitored for the development of glaucoma through both regular gonioscopy and optic nerve examination.

Corneal limbal stem cell deficiency is thought to be associated with aniridic keratopathy (corneal pannus) as well as poor long-term success rates of penetrating keratoplasty in aniridia. Keratolimbal allograft shows promise in treating both of these conditions.

In the past, painted-iris contact lenses were used to decrease glare and to impart a more natural appearance to the aniridic eye. In view of the fragility of the aniridic cornea, the use of contact lenses should be avoided if possible.

All patients with aniridia should be encouraged to use good quality sunglasses to provide optimal protection from ultraviolet light. Commercial providers of sunglasses for infants and young children are listed in **Resources**.

Additional care of the cornea of individuals with aniridia includes prompt diagnosis of ocular infections and treatment of these with mild antibiotics. When necessary, the use of preservative-free artificial tears and lubricants is also recommended.

Clinical trials using intraocular artificial iris implants began in the US in 2002. While these implants hold promise for the future, surgical challenges and complications including post-operative glaucoma currently limit their use.

Wilms Tumor

The treatment of children with WAGR syndrome/Wilms tumor is usually similar to that of typical children who develop Wilms tumor. However, children with WAGR syndrome may present some additional challenges, such as:

Nephrogenic Rests/nephroblastomatosis These lesions are common in children with WAGR syndrome, and should be monitored closely for malignancy. In some cases, these lesions may require treatment with surgery or chemotherapy.

Increased sensitivity to Vincristine Ptosis and peripheral neuropathy in response to treatment with Vincristine have been reported in children with WAGR syndrome/Wilms tumor. Chemotherapy protocols with this drug may require adjustment in these cases.

Partial nephrectomy The possibility of late-onset renal failure has led some physicians to advocate partial nephrectomy for children with WAGR syndrome and Wilms tumor in order to maximize the amount of renal tissue left after treatment. Physicians and surgeons should be aware that renal failure occurs in individuals with WAGR syndrome who have never had Wilms tumor, and that there are as yet

no studies indicating whether partial nephrectomy is a safe and effective alternative to current Wilms tumor treatment protocols.

Neurologic Care: Valproic acid (Depakote, Depakene, Epilim) (Convulex) should be used with caution in patients with WAGR syndrome, who may have an increased risk for pancreatitis.

Alternatives to risperidone (Risperdal) for the treatment of aggressive behavior should be used if possible, as this drug may exacerbate obesity, hyperglycemia, and/or hyperlipidemia in patients with WAGR syndrome.

Gynecologic Care Females with WAGR syndrome may have streak ovaries, which can increase their risk for gonadoblastoma. Malformations of the vagina and/or uterus may also be present. Diagnosis of these anomalies may be obtained with pelvic ultrasound or MRI. If identified, streak ovaries may warrant removal. If not removed, lifelong periodic surveillance with laboratory tests for tumor markers, such as CA125, HCG (human chorionic gonadotrophin) and AFP (alpha-fetoprotein), as well as pelvic ultrasound or MRI may be appropriate.

Nephrologic Care Late onset nephropathy is common in WAGR syndrome (40% of patients over the age of 12 years). The most frequent lesion reported is focal segmental glomerulosclerosis. Late-onset nephropathy has been reported in individuals with no history of Wilms Tumor. Symptoms include proteinuria, hypertension, and/or hyperlipidemia. Treatment includes ACE (angiotensin converting enzyme) inhibitors and/or ARB (angiotensin-renin blocker) medications to decrease proteinuria, normalize blood pressure, and to prolong adequate renal function. Some patients with WAGR syndrome and end stage renal disease have had renal transplantation.

Behavioral Intervention The presence of vision impairment and/or mental retardation can make evaluation for behavioral or psychiatric disorders challenging in children with WAGR syndrome. A team approach to diagnosis and treatment which includes multiple disciplines may be helpful.

Dietary Consultation Some individuals with WAGR syndrome may have a genetic predisposition to Early Onset Overweight and/or obesity. Consultation with a dietician and assistance with appropriate levels of physical activity may be necessary.

Respiratory/Ear, Nose/Throat The cause of increased susceptibility to upper respiratory illness, ear infections, sinusitis, and pneumonia in children with WAGR syndrome is unknown. Frequent or prolonged antibiotic treatment may be necessary in some cases. Placement of tympanostomy tubes may decrease the frequency of otitis media and protect hearing. Tonsillectomy/adenoidectomy may decrease the frequency of viral/bacterial infections of the throat, and may also contribute to improvement in obstructive sleep apnea. Sleep apnea may require use of continuous positive airway pressure (CPAP).

Orthopedic Care Toe-walking due to hypertense Achilles tendons is common in children with WAGR syndrome. Physical therapy is often helpful. Serial casting and/or ankle-foot orthotics may be necessary for some children.

Gastroenterology Chronic pancreatitis should be considered in the differential diagnosis of patients with WAGR syndrome of any age with symptoms of nausea/vomiting, abdominal pain, weight loss and/or fatty stools.

Early Intervention Infants with WAGR syndrome are at risk for developmental delay, and should be referred to Early Intervention services (physical therapy, speech therapy) wherever these are available. For assistance locating these services, see: **Resources**.

Research

The following institutions are currently conducting studies of WAGR syndrome:

The National Institutes of Health (NIH) is conducting a comprehensive study of WAGR syndrome. This study is sponsored by the National Institute of Child Health and Human Development, and is open to all children and adults with WAGR Syndrome. There is no cost to participants.

For more information, contact:

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10 Center Drive , MSC 1103
Bethesda , MD 20892-1103
Voicemail: 301-435-7820
Fax: 301-402-0574
Page: 301-496-1211 or 102-11838
Email: hanjo@mail.nih.gov

The University of Miami/Miller School of Medicine is conducting a study of ADD/ADHD and Autism in WAGR syndrome. This study is closed to new participants, but the project is ongoing and enrollment may reopen in the future. Please contact Ms. Morales (below) to inquire about current enrollment policies.

For more information, contact:

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Web Site: <http://medgen.med.miami.edu>

Health Supervision of Individuals with WAGR Syndrome

Guidelines for examination, referral, and anticipatory guidance

- I. Birth to 1 Month: Newborns**

- II. 1 Month to 1 Year: Infancy**

- III. 1 to 5 years: Early Childhood**

- IV. 5 to 13 years: Late Childhood**

- V. 13 to 21 years and older: Adolescence and Adulthood**

- VI. Recommended Follow-up for Wilms Tumor Survivors**

BIRTH TO 1 MONTH: NEWBORNS

Examination

Confirm the diagnosis of WAGR syndrome and review the karyotype with the parents. Review the phenotype. Discuss specific findings with both parents whenever possible, and talk about the potential clinical manifestations associated with the syndrome. These should be reviewed again at subsequent meetings, as necessary.

Referrals

- Genetics (confirmation of diagnosis)
- Pediatric Ophthalmology (Aniridia)
- Early Intervention Services (optimization of function)

Evaluate for:

- Nystagmus, cataracts, strabismus
- Hypotonia, hypertonia
- Respiratory tract infections
- Abdominal masses, hematuria

Anticipatory Guidance

- Discuss concept of “Medical Home” (coordination of care by primary care provider)
- Discuss possibility of Wilms tumor. Teach parents importance of regular ultrasound screening and how to perform abdominal palpation.
- Schedule first renal ultrasound.
- Discuss increased susceptibility to respiratory tract infection.
- Discuss the availability and positive impact of early intervention services.
- Inform the family of the availability of support and advice from other parents of children with WAGR syndrome. Provide contact information for support groups (see **Resources**)
- Discuss the strengths of the child and positive family experiences.
- Check on individual resources for support, such as family, clergy, friends.
- Talk about how and what to tell other family members and friends. Review methods of coping with long-term disabilities.

HEALTH SUPERVISION FROM 1 MONTH TO 1 YEAR: INFANCY

Referrals

- Hematology/Oncology, if necessary (Wilms tumor)
- Early Intervention Services (Optimization of function)
- Pediatric Ophthalmology (Aniridia/Ocular complications)
- Pediatric Urologist (Wilms tumor screening, genitourinary anomalies)

Examination

- Physical Examination and Laboratory studies
- Review results of molecular genetic studies.
- Review results of consultations with other specialists.
- Review history of otitis media, respiratory tract infections.
- Check infant's vision at each visit, using developmentally appropriate subjective and objective criteria.
- Schedule renal ultrasound every 3 months.
- Evaluate for abdominal masses, hematuria

Anticipatory Guidance

- Review growth and development.
- Assess the emotional status of parents and interfamily relationships. Educate and support siblings and discuss sibling adjustments.
- Review availability of support groups.
- Review early intervention services relative to the strengths and needs of the infant and family.

HEALTH SUPERVISION FROM 1 TO 5 YEARS:EARLY CHILDHOOD

Referrals

- Hematology/Oncology, if necessary (Wilms tumor)
- Pediatric Neuropsychiatry (diagnosis and management of behavior disorders)
- Pediatric Ophthalmology (Aniridia/Ocular complications)
- Nephrology, if necessary (renal failure)
- Otolaryngology (management of frequent otitis media, sinusitis)
- Orthopedics (movement disorders)
- Dietary Consultation (high risk for obesity)
- Pediatric Dentistry (dental abnormalities)
- Pediatric Gastroenterology (hyperlipidemia/pancreatitis)

Examination

- Physical Examination and Laboratory studies
- Assess for abdominal masses/hematuria. Continue renal ultrasound every 3 months. Peak incidence of Wilm's tumor in children with WAGR syndrome: 0-3 years
- Assess for symptoms of behavior disorders (Autism, PDD, Anxiety, ADD/ADHD)
- Assess frequency, type, and duration of infections.
- Laboratory tests: Lipid Profile

Anticipatory Guidance

- If appropriate, assist with coordination of cancer treatment. Review local, national resources for information and support (see **Resources**)
- Review Early Intervention services, including physical therapy, occupational therapy, and speech therapy.
- Discuss transition from Early Intervention services to preschool Special Education services.
- Discuss the child's behavior, and talk about behavior management, sibling adjustments, socialization, and recreational skills.
- Discuss symptoms related to obstructive sleep apnea, including snoring, restless sleep, and sleep position. Refer for evaluation/treatment as needed.
- Encourage family to establish optimal dietary and physical exercise patterns to help prevent/manage obesity.

HEALTH SUPERVISION FROM 5 TO 13 YEARS: LATE CHILDHOOD

Referrals

- Hematology/Oncology, if necessary (Wilms tumor)
- Pediatric Neuropsychiatry (diagnosis and management of behavior disorders)
- Pediatric Ophthalmology (Aniridia/Ocular complications)
- Nephrology, if necessary (renal failure)
- Orthopedics (movement disorders, scoliosis)
- Dietary Consultation (high risk for obesity)
- Pediatric Dentistry (dental abnormalities)
- Pediatric Gastroenterology (hyperlipidemia/pancreatitis)

Examination

- Physical Examination and Laboratory studies
- Assess for abdominal masses. Continue renal ultrasound for Wilms tumor at 3 month intervals until age 6-8yrs. Controversy exists about minimum age to discontinue ultrasound screening. Wilms tumor has been reported in patients with WAGR syndrome up to age 25. For this reason, some physicians continue ultrasound screening beyond 6-8 years of age, but increase interval to 6-12 months. Maintain high index of suspicion for Wilms tumor.
- Assess for hematuria, proteinuria, hypertension. Refer to Nephrology as needed.
- Assess girls with WAGR syndrome for streak gonads/gonadoblastoma with pelvic ultrasound and/or MRI.
- Assess for symptoms of behavioral and psychiatric disorders.
- Assess frequency, type, and duration of infections. Refer to Otolaryngology as needed.
- Discuss symptoms related to obstructive sleep apnea. Refer for evaluation/treatment as indicated.

Discuss symptoms related to pancreatitis. Refer for evaluation/treatment as indicated.

- Laboratory tests: Lipid Profile.

Anticipatory Guidance

- Review the child's development, and appropriateness of school placement.
- Discuss socialization, family status and relationships.
- Discuss the development of age-appropriate social skills and self-help skills.
- Discuss psychosexual development, physical and sexual development, menstrual hygiene and management.

- Discuss the need for gynecologic care in the pubescent female.

HEALTH SUPERVISION FROM 13 TO 21 YEARS OR OLDER: ADOLESCENCE TO EARLY ADULTHOOD

Referrals

- Neuropsychiatry (diagnosis and management of behavior disorders)
- Ophthalmology (Aniridia and ocular complications)
- Nephrology (renal failure)
- Orthopedics (movement disorders, scoliosis)
- Dietary Consultation (high risk for obesity)
- Dentistry (dental abnormalities)
- Gastroenterology (hyperlipidemia/pancreatitis)

Examination

- Obtain a history and physical examination with attention to developmental status.
- Assess for abdominal masses. Wilms tumor has been reported in patients with WAGR syndrome up to age 25. Maintain high index of suspicion for Wilms tumor.
- Assess for hematuria, proteinuria, hypertension. Refer to Nephrology as needed.
- Assess girls with WAGR syndrome for streak gonads/gonadoblastoma with pelvic ultrasound and/or MRI.
- Assess for symptoms of behavioral and psychiatric disorders.
- Assess nutritional status. Obtain dietary consult as needed.
- Laboratory tests: Lipid Profile.
- Discuss symptoms related to obstructive sleep apnea. Refer for evaluation/treatment as indicated.

Discuss symptoms related to pancreatitis. Refer for evaluation/treatment as indicated.

Anticipatory Guidance

- Discuss issues related to transition to adulthood.
- Discuss appropriateness of school placement with emphasis on adequate vocational training with the school curriculum.
- Discuss sexuality and socialization.
- Discuss group homes and independent living opportunities, workshop settings, and community-supported employment.
- Discuss interfamily relationships, financial planning, and guardianship.

- Facilitate transfer to adult medical care.

Recommended Studies for Standard and Low Risk (Stages I-III) Wilms Tumor Follow-up

Time off Treatment	History and Physical Exam	Urinalysis	Creatinine SGPT, bilirubin, total protein/albumin	Dental Exam	Nuclear GFR	Chest CT scan	Chest radiograph	CT or MRI scan of abdomen and pelvis	Abdominal ultrasound	ECHO/EKG (*Reg. DD-4A Only)
End of Treatment	✓	✓	✓	✓		✓		✓		
3 months	✓	✓	✓		✓		✓		✓	
6 months	✓	✓	✓			✓		✓		
9 months	✓		✓				✓		✓	
12 months	✓	✓	✓	✓	✓	✓		✓		✓
15 months	✓						✓		✓	
18 months	✓	✓	✓			✓		✓		
21 months	✓						✓		✓	
24 months	✓	✓	✓	✓	✓	✓		✓		
27 months	✓						✓		✓	
30 months	✓	✓	✓			✓		✓		
33 months	✓						✓		✓	
36 months	✓	✓	✓	✓	✓	✓		✓		✓
42 months	✓						✓		✓	
48 months	✓	✓	✓	✓	✓		✓		✓	
54 months	✓						✓		✓	
60 months	✓	✓	✓	✓ ¹	✓		✓		✓	✓
Then Every 2 Years	✓	✓								✓

¹Then recommended yearly throughout entire life

If your child received abdominal radiation, the following tests are recommended:

- **For Females:** Gynecological exam: A baseline examination at age 18 and then as indicated by your physician
- **For Males and Females:** Colonoscopy: Every five years beginning at age 35

Recommended Studies for Favorable Wilms Tumor Stage III (LOH) and Stage IV Follow-up

Time off Treatment	History and Physical Exam	CBC, platelets, Urinalysis, creatinine SGPT, bilirubin, total protein/albumin, electrolytes including (Ca ⁺⁺ , PO ₄ , Mg ⁺⁺)	Dental Exam	Chest CT scan	Chest radiograph	CT or MRI scan of abdomen and pelvis	Abdominal ultrasound	ECHO/EKG: Reg. DD-4A	ECHO/EKG: Reg. M
End of Treatment	✓	✓	✓	✓		✓		✓	✓
3 months	✓			✓		✓			
6 months	✓	✓		✓		✓			
9 months	✓			✓		✓			
1 year	✓	✓	✓	✓		✓			
15 months	✓			✓		✓			
18 months	✓	✓		✓		✓			
21 months	✓			✓		✓			
2 years	✓	✓	✓	✓	✓	✓	✓	✓	✓
2 ½ years	✓				✓		✓		
3 years	✓	✓	✓		✓		✓		
3 ½ years	✓				✓		✓		
4 years	✓	✓	✓		✓		✓	✓ ¹	
5 years	✓	✓	✓ ³		✓ ¹		✓ ¹		✓ ⁴

- ¹Then as indicated
- ²Only if carboplatin was taken during chemotherapy
- ³Then recommended yearly throughout entire life
- ⁴Then as per late effect guidelines

Additional lab tests recommended:

- Girls at age 13: LH, FSH, estradiol

Additional lab tests recommended (cont):

- **Boys at age 14:** LH, FSH, testosterone

If your child received abdominal radiation, the following tests are recommended:

- **For Females:**
Gynecological exam: A baseline examination at age 18 and then as indicated by your physician
- **For Males and Females:**
Colonoscopy: Every five years beginning at age 35

If your child received lung radiation, the following tests are recommended:

For Females:

- **Clinical breast exam:** yearly starting at puberty, and then every six months after age 25
- **Mammogram:** Yearly, starting at age 25

For Males and Females:

Time off treatment	Thyroid Blood Tests, T4 and TSH	Pulmonary Function Test (PFT)
End of treatment	✓	
3 months		
6 months		
9 months		
1 year	✓	
15 months		
18 months		
21 months		
2 years	✓	
2 ½ years		
3 years	✓	
3 ½ years		
4 years	✓	
5 years	✓	✓ ¹
6 years	✓	
7 years	✓	
8 years	✓	
9 years	✓	
10 years	✓ ³	

¹Then as indicated

²Then recommended every 3 to 5 years

³Then recommended yearly throughout entire life

Recommendations written and reviewed by: Susanne Ragg, MD, PhD; Deannie Lee, MD, PhD; Mary M Davis MD; James Hill, MD; Mercy Odueyungbo, B.S.; Ann Haddix, RN, MSN, CPNP; Jada Paine, MS; Julie Haydon, BS; Michelle Lucke, MPH; Elaine Skopelja, MALS, AHIP; Kellie Kaneshiro, AMLS, AHIP. Amber McMahon, B.S. Date last reviewed: August 2006 Wilms and Other Renal Tumors: <http://cancer.iu.edu/renaltumors/wilms/> website of the Indiana University Simon Cancer Center.

Resources

Many additional resources for physicians and patients are available at:
<http://www.wagr.org>

Genetic Labs/Diagnostic Testing:

Wessex Regional Genetics Laboratory

Dr. John Crolla, PhD, FRCPath

Note: Clinical diagnostic testing/Strong research interest in aniridia and WAGR syndrome

Salisbury District Hospital

Salisbury Wilts

SP2 8BJ United Kingdom

Tel: +44 (0)1722 429069

Fax: +44 (0)1722 338095

Email: jcrolla@hgmp.mrc.ac.uk

Kleberg Cytogenetics Laboratory

Note: Clinical diagnostic testing only

Baylor College of Medicine

Houston, Texas USA Tel: 713-798-3278

<http://www.bcmgeneticlabs.org/tests/alltests.html>

Aniridia:

Cornea/Cataracts:

Dr. Edward Holland

Dr. Robert Osher

Cincinnati Eye Institute

1945 CEI Drive

Cincinnati, OH 45242

513.984.5133

800-544-5133

http://www.cincinnatieye.com/maps/cei-maps_main.html

Dr. Kenneth Rosenthal

310 East Shore Rd.

Great Neck, NY 11023

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NEW YORK, NEW YORK 10017

516-466-8989 (appointments)

Glaucoma:

<http://www.eyesurgery.org>

Glaucoma:

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930 Madison Avenue, Suite 200
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Wilms Tumor

Children's Oncology Group, Renal Tumors Committee

Assistance with diagnosis and treatment decisions:

Chair: Jeffrey S. Dome, MD
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Email: PGrundy@cha.ab.ca
Web site: <http://www.ualberta.ca/~oncology/faculty/grundy/intro.htm>

Pathology: Elizabeth J. Perlman, MD
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Pediatric Urology Assoc.
1920 E Cambridge Ave. Suite 302
Phoenix, AZ 85006
Phone: (602) 279-1697
Fax: (606) 264-0461
Email: michael.ritchey@gmail.com

Children's Oncology Group

Institutions by State & by Country:
<http://www.curesearch.org/resources/coq.aspx>

International Society of Pediatric Oncology:
<http://www.siop.nl/>

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Downs Road, Sutton
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Genitourinary Anomalies

International Society on Hypospadias and Intersex Disorders
<http://www.ishid.org/index2.htm>

Early Intervention/Special Education:

In the US:

NICHCY - The National Information Center for Children and Youth with Disabilities

State by state contact information:

P.O. Box 1492

Washington, DC 20013-1492

PH: 800-695-0285

Web: <http://www.nichcy.org>

International:

The European Agency for Development in Special Needs Education

26 member countries:

<http://www.european-agency.org/>

Clinical Research:

WAGR Syndrome Research Study

Clinical Study sponsored by the National Institute of Child Health and Human Development

Open to children, adults with WAGR syndrome and their parents

Contact: Dr. Joan Han

(301) 435-7820

Email: hanjo@mail.nih.gov

ADD/ADHD and Autism in WAGR Syndrome

Clinical Research Project

University of Miami School of Medicine

Open to *all* children, adults with WAGR syndrome

Contact: Dr. Yao-Shan Fan

The Dr. John T. Macdonald Foundation Center for Medical Genetics

Leonard Miller School of Medicine

University of Miami

1601 NW 12th Ave, Rm 5037

Miami, Florida , 33136

Phone: 305-243-3823

Fax: 1-866-390-2482

Email: amorales4@med.miami.edu

Web Site: <http://medgen.med.miami.edu>

Phenotype - Genotype Correlation in Aniridia

National Eye Institute

Open to adults and children 4 years of age and older with Aniridia

Contact: Patient Recruitment and Public Liaison Office

Building 61

10 Cloister Court

Bethesda, Maryland 20892-4754

Phone: 800-411-1222
Email: prpl@mail.cc.nih.gov

Resources for Parents

International WAGR Syndrome Association

PO Box 1346
Manassas, Virginia 20108
<http://www.wagr.org>

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