INTERNATIONAL WAGR
IWSA
SYNDROME ASSOCIATION

Aniridia and Pax6-related conditions in individuals with WAGR syndrome

Key points for families with children who have WAGR syndrome

Based on Prof. Mariya Moosajee's presentation at WAGR Weekend UK 2025

Eye Care and Vision Support

Aniridia is present in almost all children with WAGR and affects every part of the eye

- Early eye shaking (nystagmus) often shows by 6 weeks of age
- Vision can be reduced by retinal changes, cataracts, glaucoma, or cornea problems

Regular eye monitoring is essential

- Children under 8: eye exams every 4 months, glasses updated yearly, and checks for glaucoma
- Older children: frequency depends on individual eye health

Protecting vision

- Sunglasses or tinted glasses for glare
- Blue-light filters on screens
- Low-vision aids and digital tools
- Healthy diet (fruits, vegetables, fish twice a week) and regular exercise

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Why Genetic Testing Matters

- Confirms the diagnosis and identifies which health problems to watch for

- Helps doctors bring together the right **multidisciplinary team (MDT):** eye doctors, kidney specialists, pediatricians, developmental specialists, and genetic counselors
- Provides information on inheritance and family planning options
- Gives access to new research, treatments, and clinical trials
- Testing looks for changes in **PAX6** and **WT1** (and sometimes **BDNF** linked to obesity)

Health Monitoring Beyond the Eyes

- **Kidneys:** ultrasound every 3 months until age 8, then every 6 months until age 18, then annually
- **Development and behavior:** regular assessments for learning differences, ADHD, autism, or anxiety
- **Hormones and metabolism:** watch for obesity, insulin resistance, or diabetes
- **Hearing and sleep:** monitor for auditory processing issues or sleep disorders

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What If Your Doctor Isn't Familiar With WAGR or Aniridia?

- You can request a second opinion at a center with expertise
    - **Professor Mariya Moosajee** (Moorfields Eye Hospital, UK) for overall WAGR/aniridia care
    - Specific pediatric specialists for glaucoma or cornea care if needed
- A polite way to ask
    - *"We attended a WAGR weekend where Prof. Moosajee spoke and offered to review patients for aniridia and include families in research."*

Research and New Therapies in Development

- **Boosting PAX6 protein:** medicines, microRNA therapy, and gene therapy are being studied

- **Non-viral gene therapy** shows promise and may avoid immune reactions seen with viral approaches
- Families can help by
    - Contacting **m.moosajee@nhs.net** for research enrollment
    - Registering at **Moorfields ROAM** for research opportunities
    - Staying in touch with IWSA and Aniridia Network for trial updates

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Support and Resources

- www.gene.vision – clear information on rare genetic eye conditions
- **International WAGR Syndrome Association (IWSA)** – family connections and research updates https://wagr.org/
- **Aniridia Network** and **Aniridia Europe** – peer support and resources

  https://aniridia.org.uk/ https://www.aniridia.eu/
- **RNIB and Guide Dogs** – vision support services
- **Esme's Umbrella** – info on Charles Bonnet Syndrome (visual hallucinations from low vision, not mental illness)

Bottom line:

- Keep up with regular eye, kidney, and developmental checks
- Use sunglasses, digital aids, and vision registration to support daily life
- Genetic testing connects you to the best care and cutting-edge research
- Stay linked to expert centers and family networks for ongoing support

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