

WAGR Spectrum Care Model for Health Management and Wellness Promotion

(A) Comprehensive Work-Up and Diagnostic Approach

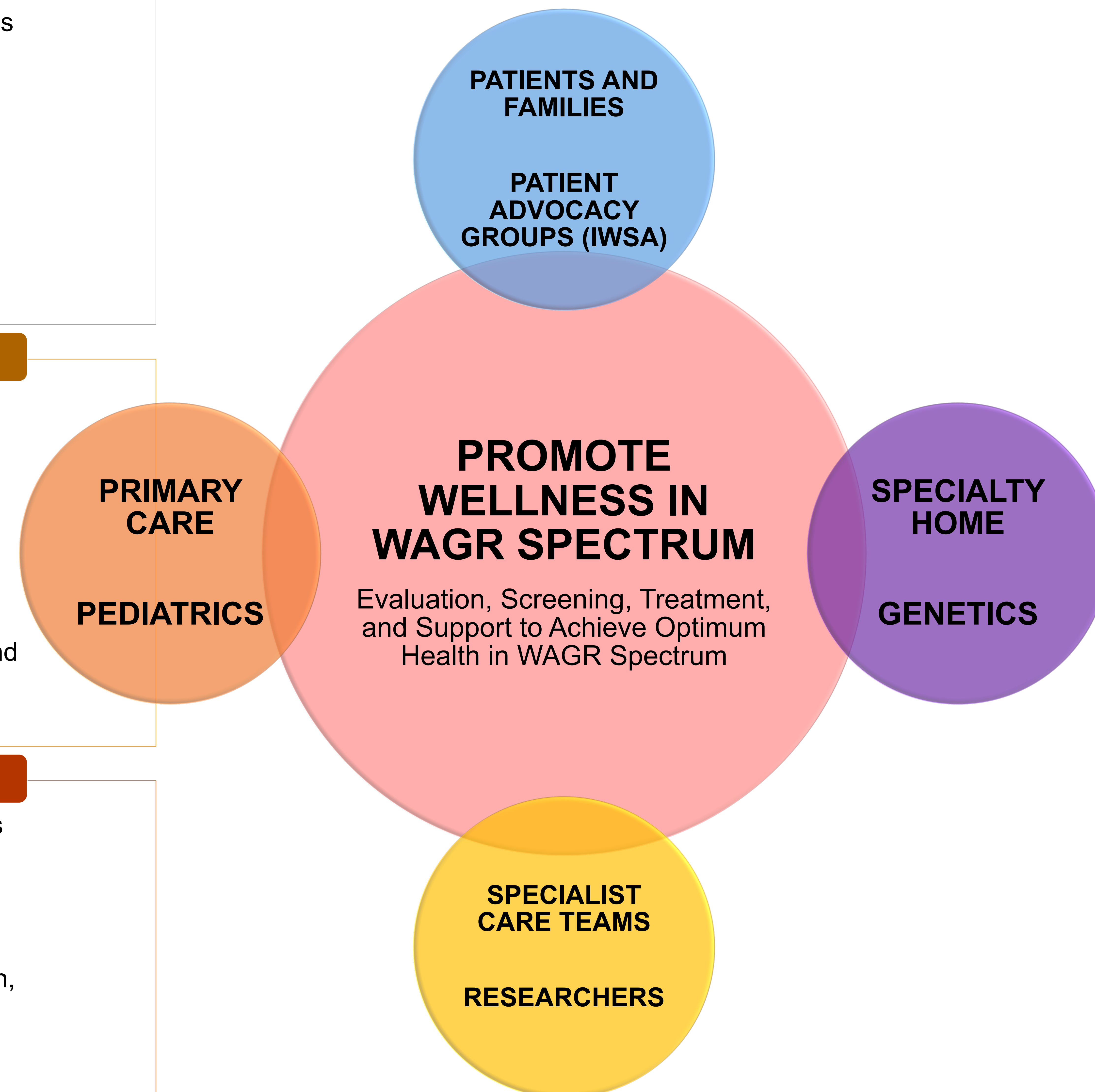
- **ESTABLISH THE CORRECT DIAGNOSIS**
 - Refer to specialists to establish baseline health profile, immediate care needs, and phenotype characteristics
 - Diagnostic work-up should be guided by specialty Genetics team
 - Patients without molecular confirmation and WAGR Spectrum phenotype should receive clinical diagnosis (*continue work-up*)
- **BASELINE HEALTH PROFILE ASSESSMENTS**
 - Perform imaging to assess and diagnose internal GU anomalies and/or abnormalities with organs (CAKUT, tumors, etc)
 - **CORE SPECIALISTS:** Pediatrician, Genetics, Nephrology, Urology, Oncology (or Cancer Predisposition)
 - **SPECIALISTS THAT MAY BE INVOLVED:** Cardiology, Endocrinology, Neurology, Pulmonology, and/or Others as needed for patient

(B) Establish Medical Homes, Individualized Care Plan, and Support Resources

- **MEDICAL HOMES AND SPECIALIST TEAMS**
 - Coordination between patient/family and 'primary home' (pediatrician) and 'specialty home' (care teams) to manage issues
- **CREATE INDIVIDUALIZED CARE PLAN**
 - Initiate **WAGR Spectrum Screening Program**
 - Determine other care needs and follow-up schedule for monitoring and/or treatment (Core Specialists)
- **CORE REFERRALS AND CARE TEAMS:** Specialists should be determined on patients' unique needs
- **REFER TO PSYCHOSOCIAL AND DEVELOPMENTAL SUPPORT SERVICES**
 - Monitor and refer patients to developmental support services to manage Range of behavioral, emotional, and cognitive issues
 - Provide contact information for **International WAGR Syndrome Association (IWSA)**

(C) Care Management and Screening to Improve Long-Term Outcomes

- **GOAL:** Early detection and treatment to mitigate long-term organ damage and adverse health consequences
- **STRATEGIES:**
 - Perform routine 'health profile' assessments to evaluate status and determine WAGR Screening Program follow-up schedule
 - Prompt referral to additional specialists if issue detected or progression of issues
- **KIDNEY HEALTH:** Preserve kidney function and prevent damage; Monitor and screen for UTIs, tumor growth, Kidney Disease (CKD), or other abnormalities
- **CARDIOMETABOLIC HEALTH:** Manage weight to prevent obesity; Monitor cardiac health profile.
- **EYE HEALTH AND VISION:** Use artificial tears frequently (preservative-free eyedrops whenever possible); Avoid cataract removal unless functional vision blocked
- **HEARING:** Consider performing routine hearing evaluations; Preserve hearing status and initiate support to improve quality of life



(A) Summary of General Recommendations for Screening Programs

General Recommendations for Screening Programs in Patient Populations Application to WAGR Population

<p>Children with Predisposition to Wilms Tumor (2016 AACR Childhood Cancer Workshop¹)</p>	<ul style="list-style-type: none"> ‘These recommendations were designed to offer screening in cases <u>with a 1% or greater risk</u> when early detection is minimally invasive and significantly improves outcome.’ ‘We acknowledge that <u>uniform recommendations may result in some patients being screened more frequently and for a longer duration than some clinicians have previously determined to be necessary.</u> Therefore, these recommendations should be discussed with each family...’ ‘<u>Surveillance can be further tailored on the basis of the disorder</u> and knowledge regarding the specific characteristics of the tumors that occur in the syndrome.’ 	<ul style="list-style-type: none"> Lifetime risk for WT cannot be estimated in WAGR Spectrum (>1% risk is conceivable) >50% of patients with WAGR develop WT, nephroblastomatosis, and/or NR during their lifetime <p style="text-align: center;">Risk Stratifications by Age for WAGR Spectrum: High risk (<8 years) Potential risk (8-15 years) Possible risk (>15 years)</p>
<p>Early Identification and Intervention of Chronic Kidney Disease (2019 KDIGO controversies conference²)</p>	<ul style="list-style-type: none"> Conclusion 1: ‘Persons with <u>hypertension, diabetes, or cardiovascular disease</u> should be screened for CKD.’ Conclusion 2: ‘CKD screening and treatment programs should also be implemented in other high-risk individuals and <u>populations based on comorbidities, environmental exposures, or genetic factors.</u>’ Conclusion 3: ‘The initiation, frequency, and cessation of CKD screening should be <u>individualized</u> based on kidney and CVD risk profiles and <u>individual preferences.</u>’ 	<ul style="list-style-type: none"> Adverse cardiometabolic profiles (CVD and Obesity) and CKD are prevalent in patients with and without history of WT There is a high risk for abnormal kidney consequences due to 11p13 deletions and health issues in patients with WAGR Spectrum

(B) Ultrasonography Screening Recommendations for Patients with WAGR Spectrum

Birth or Diagnosis	Full Abd US Pelvic US	Initiate WAGR Spectrum Screening Program: (1) Evaluate baseline kidney and organ status; (2) Diagnose CAKUT and/or internal GU anomalies; (3) Look for signs of NR, nephroblastomatosis, or WT growth and/or potential early UTI to determine follow-up screening required for patient	
Patient Age Groups	Interval and Screening Techniques	Tumor Screening Management Aspects	General Kidney Health Aspects
<8 years of age	3-month RUS*	Monitor high risk for WT, nephroblastomatosis, and/or NR; Manage risks for other adverse kidney issues	<p>GOAL: EARLY DETECTION AND TREATMENT</p> <p>(1)Diagnose and manage CAKUT (2)Screen/Work-Up potential UTIs, CKD, or other signs of kidney damage (3)Monitor CKD status and progression to determine and evaluate treatment (4)Prevent or mitigate cardiometabolic consequences that contribute to severe kidney failure and ESRD</p>
8th birthday	Full Abd US Pelvic US	Evaluate for other abdominal and/or internal GU tumors or abnormalities; Determine follow-up interval for RUS	
8 years - 15 years	3- or 6-month RUS*	Evaluate kidney health status and RUS follow-up interval; Monitor risk for CKD and/or potential risk for WT, Nephroblastomatosis and/or NR	
15 years - 18 years	6-month RUS*	Evaluate kidney health status and RUS follow-up interval; Monitor risk for CKD and/or possible risk for WT, Nephroblastomatosis and/or NR	
18th birthday	Full Abd US Pelvic US	Evaluate for other abdominal and/or internal GU tumors or abnormalities; Determine follow-up interval for RUS	
>18 years of age	Annual RUS*	Monitor overall kidney health status and CKD signs; Manage possible lifetime WT risk	

Frequency Intervals Suggested and Considerations for Implementation Purpose of Screening Interval

1-month and/or 2-month interval	Patients with history of recent UTI or recurrent UTIs to monitor treatment as determined by medical care team Provides shorter duration to evaluate clinical issues or concern for CAKUT, WT, CKD (may provide underlying etiology for clinical signs)	More frequent for patients with clinical signs to enable earlier detection than 3-month interval
3-month interval	Necessary for patients designated as ‘high risk’ for WT based on age (<8 years) and/or clinical characteristics (history of previous WT or NR); Can also assist monitoring CAKUT issues and provide early signs of CKD development and/or progression; Preferences of care team and patient/family should be considered	Routine WT screening and UTI monitoring
6-month interval	Interval period for patients between 8-18 years that do not meet consideration for 3-month frequency Consider implementing more frequent interval for those with clinical issues identified at annual screen and/or if preferences of care team and patient/family direct more frequent screening intervals	Routine WT, UTI, CKD monitoring or screening
Annual interval	Necessary minimum interval to screen for CKD and WT in patients >18 years with stable cardiometabolic and CKD health status; Preferences of care team and patient/family should be considered	Routine CKD monitoring and long-term WT screening