Appendix 1: Survey

Caregiver Demographic Information

What is your age in years?

_____

What sex were you assigned at birth (on your original birth certificate)?

◯ Female
◯ Male
◯ Unsure
◯ Not listed. Please describe in the below text box
◯ Prefer not to answer

Sex assigned at birth:

_____

What is your current gender identity? Please select all that apply.

☐ Woman
☐ Man
☐ Transgender or of trans experience
☐ Non-binary
☐ Genderqueer
☐ Genderfluid
☐ Agender
☐ Questioning
☐ Not listed. Please describe in the below text box
☐ Prefer not to answer

Current gender identity:

_____

How would you describe your racial/ethnic identity? Please select all that apply.

☐ Black, African American, or African
☐ American Indian or Alaska Native
☐ Asian
☐ Middle Eastern or North African
☐ Native Hawaiian or other Pacific Islander
☐ Hispanic, Latino, or Spanish
☐ White
☐ None of these fully describe me. Please describe in the below text box
☐ Prefer not to answer

Racial/ethnic identity:

_____

What is your employment status?

◯ Full-time
◯ Part-time
◯ Not currently working outside the home
◯ Prefer not to answer

What is the highest level of education that you completed?

- ◯ Less than a high school degree
- ◯ High school diploma/GED
- ◯ Vocational/professional certificate
- ◯ Some college
- ◯ Associate's degree
- ◯ Bachelor's degree
- ◯ Master's or graduate degree
- ◯ Doctoral or other professional degree
- ◯ Not listed. Please describe in the below text box

Highest education level: _____

How many children are living in your household (including your loved one with WAGR)? _____

What is your relationship to your loved one with WAGR?

- ◯ Parent
- ◯ Relative
- ◯ Not listed
- ◯ Prefer not to answer

Please specify type of parent.

- ◯ Biological parent
- ◯ Adoptive parent
- ◯ Stepparent
- ◯ Foster parent
- ◯ Not listed
- ◯ Prefer not to answer

Please specify. _____

Please specify relationship. _____

Please specify relationship. _____

_____

Demographic Information for the Individual with WAGR Spectrum Disorder

What is the name of your loved one with WAGR? _____

What is the age of your loved one with WAGR (in years)? _____

What sex was your loved one with WAGR assigned at birth (on the original birth certificate)?

- ◯ Female
- ◯ Male
- ◯ Unsure
- ◯ Not listed. Please describe in the below text box
- ◯ Prefer not to answer

Sex assigned at birth:

_____

What is your loved one with WAGR's current gender identity? Please select all that apply.

- ☐ Woman/girl
- ☐ Man/boy
- ☐ Transgender or of trans experience
- ☐ Non-binary
- ☐ Genderqueer
- ☐ Genderfluid
- ☐ Agender
- ☐ Questioning
- ☐ Not listed. Please describe in the below text box
- ☐ Prefer not to answer

Current gender identity:

_____

How would you describe the racial/ethnic identity of your loved one with WAGR? Please select all that apply.

- ☐ Black, African American, or African
- ☐ American Indian or Alaska Native
- ☐ Asian
- ☐ Middle Eastern or North African
- ☐ Native Hawaiian or other Pacific Islander
- ☐ Hispanic, Latino, or Spanish
- ☐ White
- ☐ None of these fully describe my loved one with WAGR. Please describe in the below text box
- ☐ Prefer not to answer

Racial/ethnic identity:

_____

_____

Birth History

Please fill in the birth weight of your loved one with WAGR. Please specify units (pounds, ounces, or grams).

_____

Please fill in the birth length of your loved one with WAGR. Please specify units (inches or centimeters).

_____

Please fill in the gestational age at birth (in weeks) of your loved one with WAGR.

_____

Were there any complications during delivery or within the first month after birth for your loved one with WAGR?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please describe the delivery and/or birth complications.

_____

_____

Eye/Vision Conditions

Either currently or in the past, has your loved one with WAGR had: structural differences in the eye(s) or vision loss?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Aniridia (partial or complete absence of the iris)
- ☐ Aniridia fibrosis syndrome
- ☐ Nystagmus (rapid, uncontrollable eye movements)
- ☐ Cataract(s)
- ☐ Glaucoma
- ☐ Optic nerve hypoplasia (small or underdeveloped optic nerve)
- ☐ Foveal/macular hypoplasia (underdevelopment of the fovea)
- ☐ Amblyopia (lazy eye)
- ☐ Strabismus (crossing of the eye(s))
- ☐ Corneal keratopathy/pannus (growth of blood vessels on the cornea)
- ☐ Hyperopia (difficulty seeing close up)
- ☐ Myopia (difficulty seeing far)
- ☐ Peter's Anomaly
- ☐ Retinal detachment
- ☐ Aphakia (missing lens in the eye that occurs, for example, as the result of cataract surgery)
- ☐ Coloboma (missing tissue in part of the eye)
- ☐ Anisocoria (different sized pupils)
- ☐ Heterochromia (different colored eyes)
- ☐ Microcornea (small cornea)
- ☐ Microphthalmos (very small eye(s))
- ☐ Anophthalmia (absence of one or both eyes)
- ☐ Other

Please specify "other."

_____

_____

Cardiac Conditions

Either currently or in the past, has your loved one with WAGR experienced problems with: blood pressure, cholesterol, the structure of the heart, or the function of the heart?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.	☐ Hypertension (high blood pressure) ☐ Heart murmur ☐ Atrial septal defect (ASD) ☐ Patent foramen ovale (PFO) ☐ Ventricular septal defect (VSD) ☐ Coarctation of the aorta ☐ Interrupted aortic arch ☐ Patent ductus arteriosus (PDA) ☐ Pulmonary valve stenosis ☐ Pulmonary valve regurgitation ☐ Pulmonary atresia ☐ Mesocardia (heart in the middle of the chest) ☐ Hyperlipidemia (high cholesterol) ☐ Other
Please specify other heart or blood pressure conditions.	_____
Did any of these heart differences require surgery?	◯ Yes ◯ No ◯ Unsure
Please specify which heart difference(s) required surgery:	_____

_____

Gastrointestinal and Feeding Conditions

Either currently or in the past, has your loved one with WAGR experienced problems with: feeding or with the gastrointestinal (GI) system (stomach, liver, pancreas, small intestine, large intestine/colon, esophagus, rectum, or anus)?	◯ Yes ◯ No ◯ Unsure

Please select all that apply.

- ☐ Feeding problems
- ☐ Pyloric stenosis (thickening of the pylorus muscle leading to projectile vomiting)
- ☐ Chronic vomiting
- ☐ Chronic constipation
- ☐ Chronic diarrhea
- ☐ Irritable bowel syndrome (IBS)
- ☐ Inflammatory bowel disease (IBD)
- ☐ Gastroesophageal reflux disease (GERD)
- ☐ Diaphragmatic hernia
- ☐ Inguinal hernia
- ☐ Umbilical hernia
- ☐ Intestinal malrotation
- ☐ Peptic ulcers (open sores in the duodenum (top of the small intestine) or in the stomach lining)
- ☐ Gastroparesis (slow movement of food through the stomach)
- ☐ Pancreatitis (inflammation of the pancreas)
- ☐ Gallstones
- ☐ Gallbladder removal (cholecystectomy)
- ☐ Anal stenosis (narrowing of the anal canal)
- ☐ Anorectal malformation (birth differences of the anus or rectum that interfere with passing stool/bowel movements)
- ☐ Other

Please select all that apply.

- ☐ Nasogastric tube (NG tube)
- ☐ Gastrostomy tube (G tube)
- ☐ Dysphagia (difficulty swallowing)
- ☐ Choking with feeds
- ☐ Difficulty latching
- ☐ Hyperphagia (extreme hunger leading to overeating)
- ☐ Other

Please specify other feeding problems.

_____

Please specify other GI conditions.

_____

_____

Ear, Nose, and Throat Conditions

Either currently or in the past, has your loved one with WAGR had structural differences of, problems with, or surgeries on: the ear(s), nose, or throat?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Ear differences present at birth
- ☐ Hearing loss/Deafness
- ☐ Anosmia (partial or full loss of smell)
- ☐ Obstructive Sleep Apnea (OSA)
- ☐ Tonsillectomy (surgery that removes the tonsils)
- ☐ Adenoidectomy (surgery that removes the adenoids)
- ☐ Tympanostomy tube placement (ear tube insertion)
- ☐ Other

Please specify "ear differences" (e.g., low-set ears; atypical shape or fold of the outside portion of the ear (pinna/auricle)).	_____
Is the hearing loss on both sides or one side?	◯ Both sides (bilateral) ◯ One side (unilateral) ◯ Unsure
Is the hearing loss conductive or sensorineural?	◯ Conductive ◯ Sensorineural ◯ Both conductive and sensorineural ◯ Unsure
Does your loved one with WAGR use any devices to help them hear?	◯ Yes ◯ No
Please select all that apply.	☐ Cochlear implant(s) ☐ Hearing aid(s) ☐ Other
Please specify other hearing device.	_____
Please specify other ear, nose, or throat conditions.	_____
	_____

Dental/Palate/Jaw Conditions

Either currently or in the past, has a dentist or other healthcare provider noted that your loved one with WAGR has: differences in their teeth (like differences in the size, shape, color, amount, or location of the teeth) or in their mouth (like differences in the palate or jaw)?	◯ Yes ◯ No ◯ Unsure
Please select all that apply.	☐ Dental cavity/cavities ☐ Missing teeth ☐ Extra teeth ☐ Crowded teeth ☐ Widely spaced teeth ☐ Large gap between top two front teeth ☐ Large teeth ☐ Tooth in palate/roof of mouth ☐ Narrow palate ☐ High-arched palate ☐ Cleft palate ☐ Cleft lip ☐ Enamel hypoplasia (missing or absent tooth enamel) ☐ Weak/soft tooth enamel ☐ Malocclusion (underbite or overbite) ☐ Other
Please specify "other."	_____

Conditions involving the Extremities

Either currently or in the past, has your loved one with WAGR experienced differences in the structure or size of: the hands, feet, legs, arms, or nails?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Lateralized overgrowth/hemihypertrophy/hemihyperplasia (overgrowth of one side or one part of the body)
- ☐ Small hands/fingers for age
- ☐ Syndactyly (fused/webbed fingers or toes)
- ☐ Polydactyly (extra fingers or toes)
- ☐ Fifth finger clinodactyly (curved, short fifth finger)
- ☐ Small feet/toes for age
- ☐ Flat feet
- ☐ Club foot
- ☐ Toe-walking
- ☐ In-toeing (turning in of feet while walking)
- ☐ Recurrent ingrown toenails
- ☐ Small nails for age
- ☐ Dysplastic (atypical) nails
- ☐ Contractures of fingers/toes (unable to fully open the fingers or toes)
- ☐ Other

Please specify "other."

_____

Musculoskeletal Conditions

Either currently or in the past, has your loved one with WAGR experienced problems with: the bones or muscles?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Scoliosis (curvature of the spine)
- ☐ Multiple hereditary exostoses (MHE) (benign bone tumors)
- ☐ Osteopenia (moderately decreased bone density)
- ☐ Osteoporosis (severely decreased bone density that can lead to fractures)
- ☐ Fracture (broken bone)
- ☐ Hypotonia (low muscle tone)
- ☐ Hypertonia (high muscle tone)
- ☐ Other

Please specify "other."

_____

_____

Conditions involving Tumors, Cancer, and Polyps

Either currently or in the past, has your loved one with WAGR had: tumors, cancer, and/or colon polyps?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Wilms tumor
- ☐ Nephrogenic rests
- ☐ Colon polyps
- ☐ Skin cancer
- ☐ Gonadoblastoma
- ☐ Other

Has your loved one with WAGR received treatment for the tumor(s), cancer, and/or colon polyp(s)?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all treatments that apply.

- ☐ Surgery
- ☐ Chemotherapy
- ☐ Radiation therapy
- ☐ Other

Please specify the other type of treatment.

_____

Has your loved one with WAGR had more than one Wilms tumor?

- ◯ Yes
- ◯ No
- ◯ Unsure

At what age was the (first) Wilms tumor identified? Please specify months or years.

_____

Where was the (first) Wilms tumor?

- ◯ Right kidney
- ◯ Left kidney
- ◯ Unsure

When the (first) Wilms tumor was identified, what was the stage of the tumor?

- ◯ Stage I
- ◯ Stage II
- ◯ Stage III
- ◯ Stage IV
- ◯ Stage V
- ◯ Unsure

Was the (first) Wilms tumor found on ultrasound screening?

- ◯ Yes
- ◯ No
- ◯ Unsure

What led to the identification of the (first) Wilms tumor?

_____

At what age was the second Wilms tumor identified? Please specify months or years.

_____

Where was the second Wilms tumor?	◯ Right kidney ◯ Left kidney ◯ Unsure
When the second Wilms tumor was identified, what was the stage of the tumor?	◯ Stage I ◯ Stage II ◯ Stage III ◯ Stage IV ◯ Stage V ◯ Unsure
Was the second Wilms tumor found on ultrasound screening?	◯ Yes ◯ No ◯ Unsure
What led to the identification of the second Wilms tumor?	_____
Has your loved one with WAGR had more than two Wilms tumors?	◯ Yes ◯ No ◯ Unsure
For each additional Wilms tumor, please specify 1) the age that the Wilms tumor was identified (and specify months or years); 2) whether the Wilms tumor was in the right kidney or the left kidney; 3) the staging of the Wilms tumor; and 4) what led to the identification of the Wilms tumor.	_____
At what age were the colon polyps identified? Please specify months or years.	_____
At what age was the skin cancer identified? Please specify months or years.	_____
At what age was the gonadoblastoma identified? Please specify months or years.	_____
Please specify the other type(s) of tumor(s), cancer, or polyp(s).	_____
Please specify at what age the other type(s) was found (and specify months or years).	_____
	_____

Genitourinary Conditions

Either currently or in the past, has your loved one with WAGR experienced problems with: the kidney(s), urinary tract, bladder, urine, or reproductive organs/genitalia?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Chronic kidney disease
- ☐ Polycystic / Cystic kidney
- ☐ Hypoplastic kidney (underdevelopment of the kidney)
- ☐ Duplicate kidney
- ☐ Horseshoe kidney
- ☐ Renal agenesis (absence of a kidney from the time of birth)
- ☐ Renal tissue disorganization
- ☐ Focal segmental glomerulosclerosis (FSGS) (development of scar tissue in the glomeruli, the kidney's filtering units)
- ☐ Hydronephrosis/dilated renal pelvis (swelling of one or both kidneys due to buildup of urine)
- ☐ Ureteric reflux / vesicoureteral reflux (VUR) (backward flow of urine from the bladder into the ureters and kidneys)
- ☐ Ureteral duplication (an extra ureter on one or both sides)
- ☐ More than three urinary tract infections (UTIs) per year
- ☐ Difficulty emptying the bladder
- ☐ Small bladder
- ☐ Large bladder
- ☐ Proteinuria (protein in the urine)
- ☐ Hematuria (blood in the urine)
- ☐ Ambiguous genitalia
- ☐ Differences in male/male-appearing reproductive organs/genitalia
- ☐ Differences in female/female-appearing reproductive organs/genitalia
- ☐ Other

Please specify the current stage of chronic kidney disease.

- ◯ Stage 1
- ◯ Stage 2
- ◯ Stage 3a
- ◯ Stage 3b
- ◯ Stage 4
- ◯ Stage 5 (kidney failure)
- ◯ Unsure

Please select all that apply.

- ☐ Cryptorchidism (undescended testicle(s))
- ☐ Hypospadias (opening of the penis on the underside rather than the tip)
- ☐ Micropenis
- ☐ Bifid scrotum
- ☐ Other

Please specify the additional male/male-appearing reproductive organ/genitalia difference(s).

_____

Please select all that apply.

- ☐ Streak ovaries (underdeveloped ovaries)
- ☐ Bicornuate uterus (heart-shaped/divided uterus)
- ☐ Small or hypoplastic (underdeveloped) uterus
- ☐ Ovarian cysts / Polycystic ovarian syndrome (PCOS)
- ☐ Other

Please specify the additional female/female-appearing reproductive organ/genitalia difference(s).

_____

Please specify other genitourinary conditions.

_____

_____

Endocrine Conditions

Either currently or in the past, has your loved one with WAGR experienced problems with: growth, weight, sugar levels, hormones, puberty, or the thyroid?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Short stature
- ☐ Tall stature
- ☐ Obesity
- ☐ Diabetes
- ☐ Prediabetes
- ☐ Hyperglycemia (high blood sugar)
- ☐ Hypoglycemia (low blood sugar)
- ☐ Precocious (early) puberty
- ☐ Delayed puberty
- ☐ Low testosterone
- ☐ Early menopause
- ☐ Hyperthyroidism (overactive thyroid)
- ☐ Hypothyroidism (underactive thyroid)
- ☐ Other

Please specify "other."

_____

Has your loved one with WAGR used growth hormone?

- ◯ Yes
- ◯ No
- ◯ Unsure

_____

Respiratory Conditions

Either currently or in the past, has your loved one with WAGR experienced problems with: the lungs, breathing, or with respiratory illnesses?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Lung hypoplasia (underdevelopment of the lungs)
- ☐ Laryngomalacia (noisy breathing due to floppy tissue in the voice box)
- ☐ Tracheomalacia (noisy breathing due to floppy tissue below the voice box)
- ☐ Pharyngomalacia (collapse of the upper airway when breathing)
- ☐ Bronchomalacia (collapse of the lower airway when breathing)
- ☐ Bronchiectasis (widening/thickening of the airways (bronchi) from inflammation and infection)
- ☐ Chronic obstructive pulmonary disease (COPD)
- ☐ Reactive airway syndrome/pre-asthma
- ☐ Asthma
- ☐ Shallow breathing
- ☐ Shortness of breath
- ☐ Apnea (pauses in breathing, especially during sleep)
- ☐ Obstructive sleep apnea (OSA)
- ☐ Pneumonia
- ☐ Recurrent (more than twice per year) pneumonia
- ☐ Recurrent sinusitis
- ☐ More than seven colds per year
- ☐ Respiratory tract infections requiring antibiotics
- ☐ Other

Please specify "other."

_____

_____

Developmental, Learning, Psychiatric, Behavioral, and Emotional Conditions

Does your loved one with WAGR have a history of: developmental delay, intellectual disability, challenges with learning, autism spectrum disorder, ADHD, anxiety, depression, or other developmental/emotional/psychiatric/behavioral diagnoses?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all conditions for which your loved one has a diagnosis.

- ☐ Global developmental delay
- ☐ Intellectual disability
- ☐ Learning disability in reading
- ☐ Learning disability in math
- ☐ Dyslexia
- ☐ Speech (expressive) delay
- ☐ Language (receptive) delay
- ☐ Auditory processing disorder
- ☐ Sensory integration disorder
- ☐ Social communication disorder
- ☐ Visual-perceptive disorder
- ☐ Visual motor deficit
- ☐ Autism spectrum disorder (ASD)
- ☐ Attention deficit/hyperactivity disorder (ADD/ADHD)
- ☐ Obsessive compulsive disorder (OCD)
- ☐ Oppositional defiance disorder (ODD)
- ☐ Anxiety
- ☐ Depression
- ☐ Panic attacks
- ☐ Bipolar disorder / manic depression
- ☐ Schizophrenia
- ☐ Pseudobulbar affect (neurological condition that causes outbursts of uncontrolled or inappropriate laughing or crying; these episodes don't match a person's internal emotional state)
- ☐ Other

Please specify "other."

_____

_____

Neurological Conditions

Either currently or in the past, has your loved one with WAGR had: seizures, migraines, structural differences of the brain, an atypical head size, an unusual response to pain, or differences in muscle tone?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Epilepsy (seizures)
- ☐ Migraines
- ☐ Microcephaly (smaller than typical head size for your child's age)
- ☐ Macrocephaly (larger than the typical head size for your child's age)
- ☐ Agenesis of the corpus callosum (partial or complete absence of the area that connects the two sides of the brain)
- ☐ Intracranial hypertension / pseudotumor cerebri (buildup of pressure around the brain)
- ☐ Hypotonia (low muscle tone)
- ☐ Hypertonia (increased muscle tone)
- ☐ Spasticity (stiff or rigid muscles)
- ☐ Ataxia
- ☐ Insensitivity to pain / high pain tolerance
- ☐ Cerebral palsy
- ☐ Other

Please specify "other."

_____

_____

Allergy Conditions

Either currently or in the past, has your loved one with WAGR had: allergies/allergic reactions?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please select all that apply.

- ☐ Animal dander / dust allergy
- ☐ Hay fever/allergic rhinitis
- ☐ Latex / skin irritant allergy
- ☐ Atopic dermatitis/eczema
- ☐ Medication allergies
- ☐ Lactose intolerance
- ☐ Food allergies
- ☐ Other

Please specify "other."

_____

_____

Other Conditions

Does your loved one with WAGR have a history of situs inversus (where the organs in the chest and abdomen are on the opposite side of the body)?

- ◯ Yes
- ◯ No
- ◯ Unsure

Please describe any other conditions that your loved one with WAGR is experiencing now or has experienced in the past that were not listed above.

_____