Supplementary Material

Supplementary Table 1: Characteristics of Subjects with WAGR Syndrome. Deletion boundaries were determined using oligonucleotide array comparative genomic hybridization (NCBI Build 36, hg 18). Subjects with deletion of all or any portion of the *BDNF* gene were designated as *BDNF*+/- (W1-W15, gray shading) while subjects with deletions that did not involve *BDNF* were designated as *BDNF*+/+ (W16-W28, no shading). History of Wilms tumor, aniridia, and genitourinary (GU) anomalies were obtained from medical chart review.

ID	Age (y)	Sex	Deletion Size (Mb)	Telomeric Boundary	Centromeric Boundary	Wilms Tumor	Visual Acuity	GU Anomaly	Included in Han et al. 2008	Included in Xu et al. 2008
W1	22.6	F	19.24	17,181,487	36,424,081	yes	3	yes	no	no
W2	21.8	M	21.62	18,782,898	40,400,884	yes	7	yes	no	no
W3	9.5	F	14.37	20,759,560	35,124,632	no	4	no	yes	yes
W4	8.3	F	11.21	22,437,763	33,647,959	no	2	no	yes	no
W5	11.9	F	18.82	23,887,358	42,707,993	yes	4	no	yes	no
W6	6.5	F	8.00	24,744,722	32,740,440	no	3	no	no	no
W7	12.6	M	11.45	24,748,424	36,203,063	yes	3	yes	yes	yes
W8	16.7	F	13.07	24,770,593	37,835,652	yes	4	yes	yes	no
W9	10.3	M	15.51	25,059,584	40,571,441	yes	4	yes	no	no
W10	22.4	F	10.71	25,330,891	36,039,207	no	4	yes	yes	no
W11	8.0	M	17.81	25,968,860	43,778,471	yes	4	yes	yes	no
W12	20.3	M	14.17	26,005,134	40,174,102	no	7	yes	yes	yes
W13	6.2	M	10.65	26,690,778	37,341,623	yes	4	yes	yes	yes
W14	24.3	F	9.90	27,057,396	36,959,349	yes	5	yes	yes	yes
W15	28.3	M	10.22	27,692,635	37,916,281	yes	6	yes	yes	yes
W16	12.2	M	6.23	27,773,897	33,998,994	no	4	yes	yes	yes
W17	12.0	F	14.80	27,824,678	42,622,913	yes	6	no	no	no
W18	6.3	M	4.62	28,200,894	32,820,697	no	3	yes	yes	no
W19	6.1	F	5.46	28,221,617	33,679,832	no	4	yes	yes	yes
W20	6.1	F	6.64	28,320,098	34,963,800	yes	4	no	yes	yes
W21	13.6	M	10.85	28,901,513	39,755,713	no	4	yes	yes	no
W22	14.0	F	4.90	29,555,113	34,459,864	no	7	no	yes	yes
W23	16.1	F	8.97	29,703,168	38,673,618	yes	4	yes	yes	no
W24	27.1	F	3.57	30,035,387	33,602,848	yes	6	no	yes	no
W25	25.4	F	6.84	30,232,199	37,070,116	no	4	yes	yes	yes
W26	28.3	F	13.47	30,861,844	44,329,536	yes	3	yes	yes	yes
W27	7.1	F	2.22	31,459,056	33,682,710	yes	4	no	yes	no
W28	11.0	M	6.81	32,380,218	39,188,328	yes	2	yes	no	yes

Supplementary Table 2: DSM-IV TR Criteria for Autism – Modified for Individuals with Significant Visual Impairment

1. QUALITATIVE IMPAIRMENTS IN RECIPROCAL SOCIAL INTERACTION
1a. Marked impairment in use of multiple nonverbal behaviors to regulate social interaction I. Consider level and duration of visual impairment in regard to the development and meaningful use of eye contact, gestures, and facial expressions.
1b. Failure to develop peer relationships appropriate to developmental level
1c. Lack of spontaneous seeking to share enjoyment, interests, achievements with others I. Consider level and duration of visual impairment, as skills such as pointing may not be meaningful for some individuals.
1d. Lack of social or emotional reciprocity I. Consider level and duration of visual impairment in regard to comfort with unfamiliar individuals in new environments.
2. QUALITATIVE IMPAIRMENTS IN COMMUNICATION
2a. Delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gestures or mime) I. Consider the level and duration of visual impairment in regard to expectation of "alternative modes of communication." For example, development of sign language/gesture is likely much less meaningful to someone with visual impairment.
2b. In individuals with adequate speech, marked impairment in the ability to initiate or sustain conversation
2c. Stereotyped & repetitive use of language or idiosyncratic
2d. Lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level I. Consider level and duration of visual impairment in regard to expectations for play associated with three dimensional objects. For example, using a doll as an agent of action has limited meaning when the objects are not fully visible to the individual.
3. RESTRICTED, REPETITIVE & STEREOTYPED PATTERNS OF BEHAVIOR, INTERESTS & ACTIVITIES
3a. Encompassing preoccupation with one or more stereotyped patterns of interest that is abnormal either in intensity or focus
3b. Apparently inflexible adherence to specific, nonfunctional routines or rituals I. Consider the how adaptive the "nonfunctional routine or ritual" is based on the individual's visual impairment (i.e., traveling in the same path every time the person walks through their house.).
3c. Stereotyped and repetitive motor mannerisms
3d. Persistent preoccupation with parts of objects

Supplementary Table 3: Characteristics of Subjects with Isolated Aniridia

ID	Age (y)	Sex	*PAX6* Genotype	Visual Acuity Score
A1	7.3	F	heterozygous single base pair deletion in exon 7 (G161VfsX46, amino acid substitution, frame-shift, premature stop-codon)	4
A2	8.3	M	heterozygous W266X (premature stop-codon in exon 10)	3
A3	17.5	M	heterozygous T>A substitution in intron 2 (predicted to alter splicing)	2
A4	23.4	F	heterozygous Q2X (premature stop-codon)	4
A5	28.3	F	heterozygous deletion of exons 1-4	5
A6	29.6	F	heterozygous deletion of exons 6-7	6
A7	32.3	F	heterozygous deletion of exons 8-13	3
A8	33.0	M	heterozygous R240X (premature stop-codon in exon 9, homeobox domain).	3
A9	33.4	M	heterozygous deletion of exons 6-7	7
A10	39.3	M	heterozygous R38W (missense mutation in paired box domain)	3
A11	40.5	F	heterozygous R240X (premature stop-codon in exon 9, homeobox domain)	4
A12	54.0	M	heterozygous E107X (premature stop-codon)	4

Supplementary Table 4: Structural Brain Abnormalities Detected by MRI in Subjects with WAGR/11p Deletion Syndrome

ID	Age (y)	Sex	Deletion Size (Mb)	Telomeric Boundary	Centromeric Boundary	Hypoplastic Corpus Callosum	Grey Matter Heterotopia	Enlarged Ventricles
W1	22.6	F	19.24	17,181,487	36,424,081	no	no	no
W2	21.8	M	21.62	18,782,898	40,400,884	yes	no	no
W3	9.5	F	14.37	20,759,560	35,124,632	no	no	no
W4	8.3	F	11.21	22,437,763	33,647,959	no	no	no
W5	11.9	F	18.82	23,887,358	42,707,993	yes	no	no
W6	6.5	F	8.00	24,744,722	32,740,440	yes	no	no
W7	12.6	M	11.45	24,748,424	36,203,063	no	no	no
W8	16.7	F	13.07	24,770,593	37,835,652	no	no	no
W9	10.3	M	15.51	25,059,584	40,571,441	no	no	no
W10	22.4	F	10.71	25,330,891	36,039,207	no MRI	no MRI	no MRI
W11	8.0	M	17.81	25,968,860	43,778,471	no	no	no
W12	20.3	M	14.17	26,005,134	40,174,102	no	no	no
W13	6.2	M	10.65	26,690,778	37,341,623	yes	no	yes
W14	24.3	F	9.90	27,057,396	36,959,349	no	yes	yes
W15	28.3	M	10.22	27,692,635	37,916,281	no MRI	no MRI	no MRI
W16	12.2	M	6.23	27,773,897	33,998,994	yes	no	no
W17	12.0	F	14.80	27,824,678	42,622,913	no	no	no
W18	6.3	M	4.62	28,200,894	32,820,697	no	no	no
W19	6.1	F	5.46	28,221,617	33,679,832	no	no	no
W20	6.1	F	6.64	28,320,098	34,963,800	yes	no	no
W21	13.6	M	10.85	28,901,513	39,755,713	yes	no	no
W22	14.0	F	4.90	29,555,113	34,459,864	no	no	no
W23	16.1	F	8.97	29,703,168	38,673,618	no	no	no
W24	27.1	F	3.57	30,035,387	33,602,848	no	no	no
W25	25.4	F	6.84	30,232,199	37,070,116	no	no	no
W26	28.3	F	13.47	30,861,844	44,329,536	no	no	no
W27	7.1	F	2.22	31,459,056	33,682,710	yes	yes	no
W28	11.0	M	6.81	32,380,218	39,188,328	no	no	yes