

The Cognitive and Behavioral Phenotype of the 16p11.2 Deletion in a Clinically Ascertained Population

Supplemental Information

Subject ID	Previously Published*	Age (years)	Gender female male	Relationship to 16p11.2 father mother sibling gm in un	Inheritance d gm in un	ASD false true	ADHD false true	Anxiety and Mood Disorder false true	Behavior Disorder false true	Articulation Disorder false true	Language Disorder false true	Learning Disorder false true	Intellectual Disability false true	Coordination Disorder false true	Tic Disorder false true	Enuresis false true	ASD severity (ADOS CSS)	SRS parent raw total score	Best FSIQ	Best VIQ	Best NVIQ	Adaptive Behavior Composite
14701.x7	*	1.7																71	56	80	73	
14702.x3	*	7.9															2	77	80	82	82	77
14703.x3	*	11.1															6	84	86	68	99	72
14704.x7	*	3															1	68	64	73	89	
14706.x2	*	6.3															1	105	102	107	99	60
14708.x10		9.4															2	20	89	85	94	102
14709.x13		3.8															10	96	95	97	72	
14711.x7		5.6															1	56	89	87	94	91
14713.x7	*	7.3															6	34	80	86	83	103
14714.x18		20.8																101	98	104	79	
14715.x36*		13.3															1	76	88	98	84	73
14715.x37*		8.4															2	49	77	80	83	73
14715.x38*		6.7															1	34	82	83	85	93
14716.x3	*	2															1	75	67	83	98	
14718.x5	*	10.6															1	60	68	63	74	87
14719.x25*		3.8															3	81	73	89	74	
14721.x5		14.5															6	94	92	86	96	71
14726.x12*		5.6															4	54	74	86	73	89
14729.x5		8.3															4	77	62	55	76	71
14730.x7		13.5															1	122	72	49	87	43
14731.x8	*	3.2															1	74	76	77	70	
14732.x6	*	10.9															4	114	54	55	53	67
14733.x15*		9.3															2	98	86	77	93	75
14736.x7		5.1															1	48	105	111	100	81
14737.x7	*	3.9															3	79	85	79	81	
14739.x3	*	7.9															1	122	63	48	77	63
14741.x22*		48																83	82	86	81	
14741.x23*		14.7															2	18	75	82	75	78
14744.x5	*	15.5															2	96	91	88	94	74
14745.x3	*	2.9															2	71	73	62	77	
14746.x8	*	9.9															5	80	60	54	64	87
14746.x9	*	8.8															3	76	79	67	89	82
14747.x10*		6.8															6	77	84	77	89	78
14748.x1	*	10															1	31	70	68	74	78

Figure S1, continued next page.

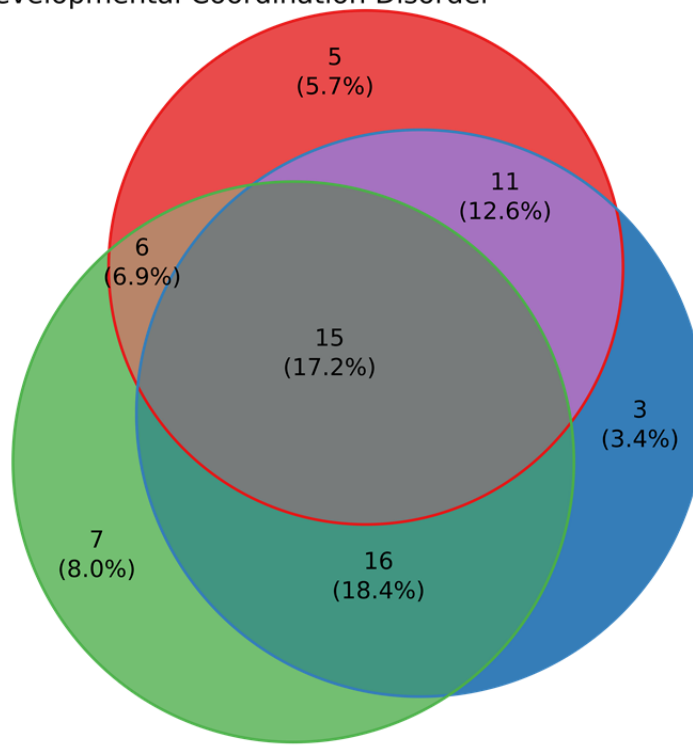
Subject ID	Age (years)	Gender	Relationship to IIP	Inheritance	ASD	ADHD	Anxiety and Mood Disorders	Behavior Disorder	Articulation Disorder	Language Disorder	Learning Disorder	Intellectual Disability	Coordination Disorder	Tic Disorder	Enuresis	ASD severity (ADOS CSS)	SRS parent raw total score	Best FSIQ	Best VIQ	Best NVIQ	Adaptive Behavior Composite
14750.x15	3.3	female	father	gm	false											2	97	96	74	76	
14753.x5	13.8	female	mother	in	true											9	116	79	72	86	68
14754.x7	11.9	female	sibling	un												1	122	88	71	100	69
14755.x15	38	female															106	109	101		
14755.x17	13.1	female														1	131	75	73	79	73
14758.x3 *	6.3	female														6	81	71	61	82	79
14760.x10*	5.6	female														2	134	99	99	100	61
14761.x13*	10.3	female														1	9	98	100	97	103
14761.x14*	12.1	female														2	6	103	106	102	93
14763.x7 *	5.2	female														1	72	96	76	110	81
14764.x2	3.7	female														2	95	80	106	88	
14765.x2 *	12.7	female														3	84	66	64	69	65
14772.x12*	10.5	female														1	66	72	55	84	77
14776.x3 *	5.4	female														6	103	49	23	48	62
14777.x3 *	7.8	female														3	106	60	59	66	68
14778.x12*	3.2	female														2	89	93	86	79	
14779.x7	16.8	female														2	67	81	84	83	77
14780.x3 *	12.4	female														6	60	76	57	89	83
14781.x16*	8.8	female														6	28	97	86	104	95
14782.x3 *	9.9	female														2	162	85	82	89	63
14785.x5 *	6.1	female														3	57	76	91	72	84
14786.x23*	40.3	female															89	80	102	93	
14786.x24*	12.3	female														2	50	63	63	65	74
14789.x7	10.7	female														3	67	103	100	104	83
14790.x1	4.1	female														2	66	46	38	54	71
14791.x26*	7.1	female														3	82	93	89	95	80
14792.x10	7.8	female														6	24	105	115	99	97
14793.x3 *	7.8	female														1	54	100	114	92	83
14795.x17*	8	female														2	39	87	87	89	80
14796.x3 *	12.3	female														6	96	59	63	57	66
14797.x8 *	16.2	female														93	86	100	81	67	
14799.x1 *	14.5	female														7	87	122	106	130	85
14802.x7	0.8	female															103	82	124	93	
14805.x16	1.2	female															95	87	103	79	
14807.x10	4.4	female														4	36	89	85	95	82
14808.x7	1.4	female															77	51	109	68	
14809.x1	4	female														4	42	79	75	84	78
14818.x20	8	female														3	33	81	80	84	85
14823.x14	10.8	female														1	91	90	89	92	80
14824.x12	14.3	female														6	117	103	98	105	65
14824.x13	14.3	female														9	125	93	87	98	63
14826.x9	7.8	female														3	72	94	92	98	80
14830.x7	8.3	female														4	60	87	80	93	76
14831.x17	5.8	female														2	100	92	79	102	69
14833.x8	12.3	female														3	64	111	106	116	83
14838.x3	2.4	female															103	115	91	122	
14840.x22	8.4	female														9	130	67	84	60	74

Figure S1, continued next page.

Subject ID	Previously Published*	Age (years)	Gender female male	Relationship to IIP father mother sibling	Inheritance gm in un	ASD false true	ADHD false true	Anxiety and Mood Disorder false true	Behavior Disorder false true	Articulation Disorder false true	Language Disorder false true	Learning Disorder false true	Intellectual Disability false true	Coordination Disorder false true	Tic Disorder false true	Enuresis false true	ASD severity (ADOS CSS)	SRS parent raw total score	Best FSIQ	Best VIQ	Best NVIQ	Adaptive Behavior Composite
14850.x22		5	female	mother	in	true											6	85		95	65	
14858.x1		34.9	female	mother	in													78	63	99	51	
14858.x2		8.5	female	mother	in												1	115	71	64	78	82
14858.x3		12.6	female	mother	in												1	102	84	87	85	103
14858.x4		11.5	female	mother	in												1	112	80	64	93	110
14858.x6		3.7	female	mother	in																	104
14859.x4		3.8	female	mother	in												1	75	56	91	73	
14863.x7		0.9	female	mother	in													112	95	129	91	
14869.x13		14.9	female	mother	in													53	118	113	118	102
14874.x13		3.7	female	mother	in	true		true					true				7	68	58	63	59	
14877.x7		4.2	female	mother	in				true				true				4	94	55	54	55	62
14888.x3		31.7	female	mother	in													72	65	84		
14888.x5		2.9	female	mother	in												3	58	51	59	88	
14893.x7		7.4	female	mother	in									true			2	25	86	86	89	93
14895.x1		44.4	female	mother	in			true										90	90	93		
14895.x3		11	female	mother	in	true							true	true			6	85	85	80	90	70

Figure S1. Psychiatric diagnoses and demographic information for the individuals with 16p11.2 deletion. The previously published variable indicates that data from this individual was reported in Zufferey *et al.* (1). Age is reported in years. Gender, relationship to initially identified patient, and inheritance status are color coded, with the key by the variable heading. A black square indicates the presence of a diagnosis for the psychiatric diagnoses listed. Autism spectrum disorder (ASD) severity (as measured by the calibrated severity score (CSS) and by Social Responsiveness Scale (SRS) parent total raw score) is represented by colored shading, from dark to light, with lighter green indicating a higher score (more impairment). Full Scale IQ (FSIQ), Verbal IQ (VIQ), and Nonverbal IQ (NVIQ) are represented by colored shading, from dark to light, with lighter green indicating a higher score (higher IQ). Adaptive functioning (as measured by the adaptive behavior composite from the Vineland) is represented by colored shading, from dark to light, with lighter green indicating a higher score (greater adaptive ability). ADHD, attention-deficit/hyperactivity disorder; ADOS, Autism Diagnostic Observation Schedule.

- Expressive and Mixed Receptive-Expressive Language Disorder
- Phonological Disorder
- Developmental Coordination Disorder



- Expressive and Mixed Receptive-Expressive Language Disorder
- Autism Spectrum Disorders
- Developmental Coordination Disorder

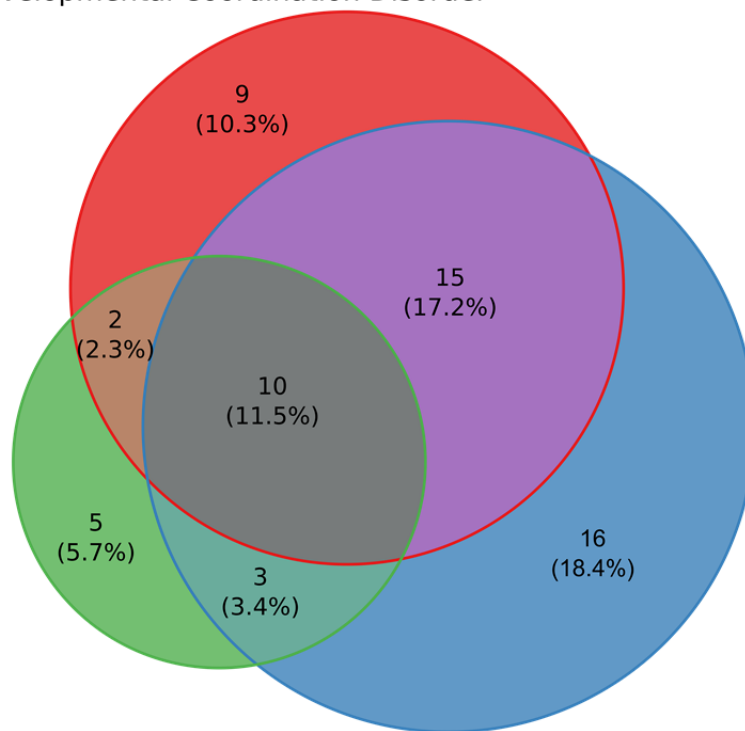


Figure S2. Diagnostic comorbidity for most commonly observed psychiatric diagnoses in individuals with 16p11.2 deletion; $n = 87$. This figure demonstrates the overlap in diagnoses within individuals to highlight the constellation of symptoms and most commonly occurring comorbidities.

Table S1. Analyses of Carriers vs Non-Carriers and De-Novo vs. Inherited Carriers

	Intercept		De Novo Carrier vs. Inherited Carrier		Carrier vs. Non Carrier	
	est	<i>p</i>	est	<i>p</i>	est	<i>p</i>
Vineland Composite Score	104.24	<.0001	-1.68	0.7247	-24.92	<.0001
Vineland Social Score	103.24	<.0001	-4.86	0.261	-21.81	<.0001
Vineland Communication Score	104.15	<.0001	-0.13	0.973	-27.47	<.0001
Vineland Daily Living Score	104.13	<.0001	-4.84	0.2307	-21.85	<.0001
CASL Total Score	105.88	<.0001	8.31	0.07	-31.58	<.0001
CBCL Total	47.03	<.0001	-2.05	0.6199	14.93	<.0001
CBCL Externalizing	48.21	<.0001	-0.02	0.995	6.61	0.0058
CBCL Internalizing	49.42	<.0001	1.88	0.6405	10.37	<.0001
CBCL Affective	54.42	<.0001	0.20	0.9347	6.10	0.0004
CBCL Anxiety	54.16	<.0001	5.05	0.1175	3.91	0.0276
CBCL Somatic	54.51	<.0001	2.62	0.3614	4.81	0.0109
CBCL ADHD	53.51	<.0001	-4.07	0.1058	9.92	<.0001
CBCL Oppositional Defiant Conduct	54.38	<.0001	0.77	0.7532	3.72	0.0255
Conduct	52.25	<.0001	-0.70	0.7415	4.60	0.0016
Articulation Disorder	-3.95	<.0001	0.64	0.4211	0.98	<.0001
Coordination Disorder	-28.37	-	-	-	-	-
Language Disorder	-5.06	<.0001	0.66	0.2381	0.99	<.0001
Enuresis	-4.34	<.0001	0.27	0.34	0.95	0.0002
ADHD	-3.21	<.0001	0.53	0.917	0.84	0.0072
Number of DSM Diagnoses	-1.23	<.0001	1.06	0.7852	9.89	<.0001
CTOPP Standard Score	7.77	<.0001	0.70	0.4172	-2.48	<.0001
BSIQ	2.99	0.0019	4.50	0.0263	6.04	<.0001
Handedness	-1.91	0.0002	0.17	0.0391	0.91	0.0002
Full Scale IQ (FSIQ)	109.18	<.0001	8.69	0.0419	-28.35	<.0001
Non-Verbal IQ (NVIQ)	110.11	<.0001	7.33	0.0999	-24.95	<.0001
Verbal IQ (VIQ)	106.16	<.0001	8.59	0.0644	-29.12	<.0001
SRS Total	22.48	<.0001	-0.60	0.9562	56.01	<.0001

ADHD, attention-deficit/hyperactivity disorder; BSIQ, Behavior and Sensory Interests Questionnaire; CASL, Comprehensive Assessment of Spoken Language; CBCL, Child Behavior Checklist; CTOPP, Comprehensive Test of Phonological Processing; SM, Diagnostic and Statistical Manual; SRS, Social Responsiveness Scale.

Table S2. Analyses of Carriers vs Non-Carriers and De-Novo vs Inherited Carriers Controlling for Age, Sex, and NVIQ

	Intercept		NVIQ		Age (Years)		Sex: Female vs. Male		De Novo Carrier vs. Inherited Carrier		Carrier vs. Non Carrier	
	est	p	est	p	est	p	est	p	est	p	est	p
Vineland Composite Score	77.68	<.0001	0.24	0.0322	0.15	0.5108	-1.21	0.6656	-2.32	0.6423	-19.82	<.0001
Vineland Social Score	88.70	<.0001	0.15	0.1073	-0.15	0.4253	-0.28	0.9056	-4.90	0.2746	-18.60	<.0001
Vineland Communication Score	85.30	<.0001	0.19	0.0312	-0.15	0.4051	0.55	0.8023	-0.23	0.9538	-23.61	<.0001
Vineland Daily Living Score	93.99	<.0001	0.06	0.4714	0.16	0.3886	2.20	0.341	-4.52	0.2863	-20.11	<.0001
CASL Total Score	38.83	0.0002	0.65	<.0001	-0.19	0.4978	0.48	0.8276	1.75	0.6565	-18.65	<.0001
CBCL Total	76.73	<.0001	-0.24	0.013	-0.31	0.4101	-0.48	0.8315	0.34	0.9369	9.11	0.0052
CBCL Externalizing	74.20	<.0001	-0.18	0.0454	-0.66	0.0689	1.93	0.3744	2.18	0.5789	1.82	0.5369
CBCL Internalizing	65.85	<.0001	-0.16	0.0829	0.10	0.7812	-0.60	0.7763	3.59	0.3978	6.49	0.0312
CBCL Affective	55.69	<.0001	-0.03	0.6717	0.12	0.627	0.61	0.6939	0.75	0.7806	5.50	0.0138
CBCL Anxiety	71.78	<.0001	-0.18	0.0115	0.17	0.5357	-0.39	0.8073	7.30	0.0356	-0.23	0.9154
CBCL Somatic	69.71	<.0001	-0.14	0.0636	-0.08	0.7662	0.60	0.7329	4.23	0.1661	1.63	0.4994
CBCL ADHD	59.49	<.0001	-0.07	0.2764	0.10	0.6727	0.21	0.8885	-3.19	0.2373	8.47	0.0002
CBCL Oppositional Defiant Conduct	78.54	<.0001	-0.18	0.0052	-0.54	0.0259	1.55	0.2922	3.02	0.2265	-0.81	0.6865
Articulation Disorder	67.60	<.0001	-0.13	0.0205	-0.22	0.2764	1.22	0.3443	0.98	0.6537	1.57	0.3741
Coordination Disorder	-2.71	0.1617	0.01	0.4069	-0.14	0.0003	-0.04	0.9282	0.52	0.9005	0.95	0.0002
Language Disorder	-26.01	-	0.01	-	-0.18	-	-0.31	-	-	-	-	-
Enuresis	-1.78	0.4287	-0.01	0.5944	-0.09	0.0256	-0.54	0.2506	0.59	0.557	0.97	0.0028
ADHD	-4.25	0.099	0.01	0.4739	-0.09	0.0028	0.71	0.2543	0.22	0.2416	0.92	0.0347
Number of DSM Diagnoses	-1.47	0.4398	0.00	0.8893	-0.06	0.0149	-0.99	0.0472	0.37	0.6626	0.71	0.2237
CTOPP Standard Score	0.73	0.0692	-0.01	0.0017	-0.02	0.0026	-0.26	0.0065	0.93	0.7537	5.07	<.0001
BSIQ	4.10	0.0352	0.04	0.0328	-0.05	0.1658	0.19	0.6432	0.27	0.7632	-1.69	0.007
Handedness	9.24	0.0355	-0.05	0.175	0.05	0.7386	-1.54	0.1471	4.53	0.0333	4.78	0.0021
Full Scale IQ (FSIQ)	2.28	0.2452	-0.03	0.0757	-0.03	0.6071	-0.58	0.1673	0.18	0.0811	0.81	0.0278
Non-Verbal IQ (NVIQ)	104.35	<.0001	-	-	0.20	0.0027	-2.30	0.1709	8.33	0.0468	-24.66	<.0001
Verbal IQ (VIQ)	103.33	<.0001	-	-	0.28	0.0001	-2.77	0.1125	6.79	0.1164	-19.85	<.0001
SRS Total	103.20	<.0001	-	-	0.12	0.109	-0.99	0.5833	8.39	0.0706	-26.96	<.0001
	35.45	0.1149	-0.28	0.1685	1.51	0.0629	1.45	0.7785	6.26	0.5814	50.52	<.0001

See Table S1 for abbreviations.

Table S3. Analyses of Carriers vs Non-Carriers and De-Novo vs Inherited Carriers Controlling for Age, Sex, NVIQ, and ASD

	Intercept		Best NVIQ		Age (Years)		Sex: Female vs. Male		ASD: Yes vs. No		De Novo Carrier vs. Inherited Carrier		Carrier vs. Non Carrier	
	est	p	est	p	est	p	est	p	est	p	est	p	est	p
Vineland Composite Score	77.19	<.0001	0.24	0.03	0.13	0.565	-1.38	0.6346	-0.4	0.9279	-3.50	0.5114	-19.38	<.0001
Vineland Social Score	90.74	<.0001	0.14	0.1304	-0.14	0.4589	-1.14	0.6236	-9.2	0.0096	-4.39	0.3275	-17.27	<.0001
Vineland Communication Score	86.46	<.0001	0.18	0.0361	-0.16	0.3726	-0.29	0.8965	-6.5	0.0508	0.42	0.9189	-22.40	<.0001
Vineland Daily Living Score	95.44	<.0001	0.06	0.5023	0.14	0.4377	1.07	0.6461	-8.0	0.0239	-4.13	0.3467	-18.51	<.0001
CASL Total Score	39.37	0.0002	0.65	<.0001	-0.23	0.4315	-0.06	0.979	-1.6	0.6373	1.20	0.7755	-18.10	<.0001
CBCL Total	77.00	<.0001	-0.24	0.0143	-0.32	0.4002	-0.80	0.7276	-0.1	0.9845	0.42	0.9224	9.24	0.0064
CBCL Externalizing	75.07	<.0001	-0.18	0.0488	-0.78	0.0346	1.99	0.3602	5.0	0.1283	1.26	0.7487	0.90	0.7634
CBCL Internalizing	66.45	<.0001	-0.16	0.089	0.02	0.9623	-0.79	0.7162	2.9	0.3809	3.17	0.4529	6.09	0.049
CBCL Affective	56.53	<.0001	-0.03	0.6749	0.04	0.8672	0.49	0.7549	2.2	0.3368	0.48	0.8585	5.14	0.0235
CBCL Anxiety	72.06	<.0001	-0.17	0.0136	0.03	0.9049	-0.18	0.9082	6.2	0.0162	5.98	0.0727	-1.15	0.6022
CBCL Somatic	70.72	<.0001	-0.13	0.0635	-0.18	0.5188	0.54	0.7585	3.2	0.2222	3.70	0.2215	0.99	0.6855
CBCL ADHD	60.68	<.0001	-0.07	0.279	-0.04	0.8789	0.34	0.8188	5.5	0.0176	-4.31	0.1052	7.34	0.0012
CBCL Oppositional Defiant Conduct	79.10	<.0001	-0.17	0.0058	-0.61	0.0137	1.53	0.3032	2.4	0.2722	2.63	0.2956	-1.23	0.5499
Articulation Disorder	68.20	<.0001	-0.12	0.0216	-0.31	0.1447	1.33	0.3052	3.2	0.1014	0.40	0.8531	0.92	0.6063
Coordination Disorder	-2.41	0.2286	0.01	0.4696	-0.14	0.0002	-0.39	0.4411	-1.8	0.0222	0.60	0.6308	0.96	<.0001
Language Disorder	-27.13	-	0.01	-	-0.18	-	-0.18	-	0.4	-	-	-	-	-
Enuresis	-1.95	0.3961	-0.01	0.6441	-0.09	0.038	-0.34	0.4791	0.7	0.2549	0.56	0.7048	0.96	0.005
ADHD	-4.49	0.0788	0.01	0.4176	-0.10	0.0032	0.90	0.1657	1.0	0.1558	0.20	0.2052	0.90	0.055
Number of DSM Diagnoses	-1.44	0.442	0.00	0.8929	-0.06	0.0146	-1.01	0.0435	-0.1	0.9047	0.38	0.6926	0.71	0.2211
CTOPP Standard Score	0.59	0.1505	-0.01	0.003	-0.02	0.0035	-0.21	0.0363	0.3	0.0648	0.87	0.5729	4.93	<.0001
BSIQ	4.13	0.034	0.04	0.0343	-0.05	0.2	0.13	0.7558	-0.9	0.1939	0.40	0.658	-1.52	0.0165
Handedness	8.93	0.0426	-0.05	0.2367	0.00	0.9801	-1.53	0.1663	1.7	0.2864	3.49	0.1246	4.80	0.0028
SRS Total	2.99	0.18	-0.04	0.056	-0.01	0.8232	-0.70	0.098	-1.5	0.0317	0.23	0.196	0.84	0.024
	33.02	0.1286	-0.23	0.2305	1.21	0.1286	1.75	0.7295	17.9	0.0227	2.40	0.8327	48.45	<.0001

See Table S1 for abbreviations.

Table S4. Analyses of Carriers vs Non-Carriers and De-Novo vs Inherited Carriers Controlling for Age, Sex, NVIQ, and CSS

	Intercept		Best NVIQ		Age (Years)		Sex: Female vs. Male		CSS		De Novo Carrier vs. Inherited Carrier		Carrier vs. Non Carrier	
	est	p	est	p	est	p	est	p	est	p	est	p	est	p
Vineland Composite Score	76.26	<.0001	0.23	0.0618	0.35	0.4352	-1.13	0.7203	0.22	0.7904	-2.20	0.6827	-19.81	<.0001
Vineland Social Score	102.62	<.0001	0.09	0.3528	-0.85	0.025	-0.78	0.7442	-0.80	0.2256	-4.49	0.3452	-19.22	<.0001
Vineland Communication Score	89.12	<.0001	0.17	0.0454	-0.42	0.2062	0.20	0.9264	0.08	0.8969	-0.82	0.8401	-24.51	<.0001
Vineland Daily Living Score	103.05	<.0001	0.01	0.902	-0.10	0.7709	1.08	0.6617	-0.71	0.2799	-3.18	0.4759	-19.99	<.0001
CASL Total Score	39.96	0.0004	0.65	<.0001	-0.41	0.2413	1.40	0.5497	0.63	0.3023	-0.67	0.8677	-20.76	<.0001
CBCL Total	73.87	<.0001	-0.20	0.0592	-0.40	0.3742	-0.89	0.7186	-0.87	0.2004	1.13	0.8014	11.92	0.0017
CBCL Externalizing	71.63	<.0001	-0.15	0.1185	-0.84	0.0612	2.23	0.3573	0.32	0.6183	2.04	0.6227	3.06	0.3582
CBCL Internalizing	59.65	<.0001	-0.09	0.3221	0.04	0.9327	-0.20	0.9284	-0.43	0.493	4.05	0.3619	9.25	0.0068
CBCL Affective	53.64	<.0001	0.00	0.9834	0.00	0.9911	1.16	0.4921	-0.20	0.6473	1.55	0.5749	7.02	0.0058
CBCL Anxiety	67.93	<.0001	-0.16	0.031	0.30	0.34	0.51	0.7674	0.47	0.3313	6.88	0.0609	-1.05	0.6574
CBCL Somatic	67.13	<.0001	-0.11	0.1569	-0.22	0.5165	1.30	0.4988	0.65	0.2063	2.87	0.3701	1.06	0.6896
CBCL ADHD	55.60	<.0001	-0.04	0.5463	0.03	0.9191	0.85	0.606	0.71	0.1145	-3.72	0.1899	8.54	0.0009
CBCL Oppositional Defiant	79.18	<.0001	-0.18	0.0089	-0.73	0.0167	1.96	0.2293	0.66	0.1274	2.54	0.3379	-1.14	0.6072
Conduct	67.39	<.0001	-0.13	0.0342	-0.32	0.215	1.53	0.2901	0.52	0.1767	0.66	0.7772	1.23	0.5367
Articulation Disorder	-2.36	0.2712	0.01	0.4557	-0.08	0.3199	-0.34	0.4911	-0.35	0.0118	0.68	0.4075	0.97	0.0001
Coordination Disorder	-27.01	-	0.01	-	-0.10	-	-0.31	-	-0.12	-	-	-	-	-
Language Disorder	-1.59	0.5029	-0.01	0.7048	-0.13	0.116	-0.68	0.198	0.06	0.6669	0.50	0.9999	0.96	0.0044
Enuresis	-5.30	0.0476	0.01	0.4759	0.03	0.6615	0.88	0.1858	0.07	0.5832	0.30	0.4265	0.89	0.0688
ADHD	-2.54	0.237	0.01	0.803	0.04	0.6788	-1.47	0.0178	-0.07	0.6268	0.47	0.9497	0.79	0.1117
Number of DSM Diagnoses	-0.21	0.6751	-0.01	0.0388	0.00	0.7646	-0.20	0.076	0.02	0.5335	1.05	0.8464	6.80	<.0001
CTOPP Standard Score	5.53	0.0199	0.03	0.1104	-0.12	0.1605	0.07	0.8835	0.03	0.8509	-0.02	0.9818	-1.95	0.0074
SRS Total	16.25	0.5087	-0.16	0.4576	1.74	0.0664	4.07	0.4705	2.43	0.1325	5.08	0.6718	49.67	<.0001

See Table S1 for abbreviations.

Table S5. Analyses of Carriers vs Non-Carriers and De-Novo vs Inherited Carriers Controlling for Age, Sex, NVIQ, and SRS

	Intercept		Best NVIQ		Age (Years)		Sex: Female vs. Male		SRS Total Score		De Novo Carrier vs. Inherited Carrier		Carrier vs. Non Carrier	
	est	p	est	p	est	p	est	p	est	p	est	p	est	p
Vineland Composite Score	78.66	<.0001	0.22	0.0737	0.53	0.2464	-1.35	0.67	-0.13	0.0335	-2.89	0.6014	-13.00	0.0173
Vineland Social Score	105.92	<.0001	0.06	0.4289	-0.18	0.5363	-1.10	0.55	-0.31	<.0001	-3.84	0.3471	-3.83	0.228
Vineland Communication Score	92.40	<.0001	0.14	0.087	0.06	0.8373	0.96	0.64	-0.19	<.0001	-0.60	0.8724	-14.49	0.0001
Vineland Daily Living Score	98.84	<.0001	0.03	0.7162	0.34	0.3559	2.48	0.32	-0.16	0.0015	-4.58	0.3306	-12.55	0.0045
CASL Total Score	44.01	0.0001	0.62	<.0001	-0.21	0.5295	0.10	0.97	-0.05	0.2526	0.24	0.9557	-16.76	0.0002
CBCL Total	70.59	<.0001	-0.18	0.0306	-0.68	0.048	-1.14	0.56	0.20	<.0001	-0.98	0.798	-0.75	0.817
CBCL Externalizing	69.13	<.0001	-0.13	0.1126	-0.98	0.0051	1.20	0.54	0.17	<.0001	1.09	0.7661	-7.10	0.0332
CBCL Internalizing	59.36	<.0001	-0.10	0.2005	-0.23	0.4538	-1.51	0.41	0.20	<.0001	2.11	0.5488	-3.45	0.2634
CBCL Affective	51.53	<.0001	0.02	0.7635	-0.15	0.477	-0.07	0.96	0.14	<.0001	-0.15	0.9458	-1.93	0.3879
CBCL Anxiety	68.18	<.0001	-0.15	0.0171	-0.04	0.8702	-0.78	0.58	0.13	<.0001	6.49	0.044	-6.92	0.0068
CBCL Somatic	67.29	<.0001	-0.11	0.1234	-0.23	0.4041	0.27	0.87	0.08	0.0183	3.73	0.2065	-2.55	0.377
CBCL ADHD	55.77	<.0001	-0.03	0.5841	-0.11	0.6384	-0.30	0.83	0.11	0.0001	-3.95	0.1134	2.58	0.2652
CBCL Oppositional Defiant Conduct	76.23	<.0001	-0.15	0.0124	-0.70	0.0048	1.32	0.35	0.08	0.0027	2.60	0.2947	-5.49	0.0244
Articulation Disorder	64.67	<.0001	-0.10	0.0542	-0.39	0.0533	0.78	0.51	0.09	0.0002	0.32	0.8768	-3.32	0.1
Coordination Disorder	-2.75	0.1987	0.00	0.7974	-0.09	0.2262	0.22	0.65	0.00	0.565	0.64	0.4876	0.94	0.0063
Language Disorder	-26.86	-	0.01	-	-0.18	-	-0.31	-	0.02	-	-	-	-	-
Enuresis	-25.91	-	0.00	-	-0.11	-	-0.92	-	0.01	-	-	-	-	-
ADHD	-3.96	0.1500	0.01	0.6321	-0.04	0.5916	0.80	0.23	-0.01	0.5019	0.30	0.4322	0.94	0.0742
Number of DSM Diagnoses	-2.91	0.1459	0.01	0.5132	0.00	0.9601	-1.36	0.01	0.00	0.9086	0.38	0.7182	0.78	0.2249
CTOPP Standard Score	-0.03	0.9492	-0.01	0.0096	0.00	0.7752	-0.25	0.02	0.00	0.21	1.08	0.7481	6.16	<.0001
	4.36	0.0403	0.04	0.0399	-0.07	0.359	0.17	0.69	0.00	0.5771	0.18	0.8516	-1.36	0.0882

See Table S1 for abbreviations.

Supplemental Reference

1. Zufferey F, Sherr EH, Beckmann ND, Hanson E, Maillard AM, Hippolyte L, *et al.* (2012): A 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. *J Med Genet.* 49:660-8.