

**Supplemental Table 1. Clinical characteristics of CS Males by inheritance and mutation type**

	De Novo (n = 15)	Inherited (n = 22)	$t/X^2$ p	Missense (n = 6)	Nonsense (n = 22)	$t/X^2$ p
<b>General</b>						
Age at Enrollment (sd)	12 (5.9)	13 (7.8)	.603	8.1 (6.6)	14 (5.8)	<b>.041</b>
Age at CS Diagnosis, m (sd)	7.1 (5.7)	8.6 (7.6)	.523	6.1 (4.5)	8.5 (6.7)	.403
Able to Walk Currently (%)	11 (85)	17 (81)	.785	5 (83)	16 (76)	.959
History of Regression (%)	7 (54)	11 (52)	.934	2 (40)	11 (52)	.619
<b>Neurological, n (%)</b>						
Microcephaly	14 (100)	19 (95)	.396	5 (100)	20 (100)	-
Ataxia	14 (100)	17 (94)	.370	6 (100)	17 (94)	.555
Autism Spectrum Disorder	4 (29)	2 (10)	.162	1 (17)	5 (25)	.671
Hyperkinesia	14 (100)	19 (86)	.149	3 (100)	18 (75)	.326
Prior Angelman Dx	4 (29)	8 (38)	.561	2 (33)	7 (35)	.940
High Pain Tolerance	14 (100)	19 (86)	.149	6 (100)	19 (90)	.432
Unprovoked Laughter	6 (43)	16 (76)	<b>.046</b>	4 (67)	14 (70)	.877
Contractures	5 (36)	5 (28)	.631	2 (40)	5 (28)	.599
Visual Acuity Problems	9 (64)	9 (43)	.214	4 (67)	10 (50)	.473
Eye Movement Abnormalities	9 (69)	14 (70)	.963	3 (60)	14 (74)	.549
<b>Medical Status, n (%)</b>						
Swallowing Problems	7 (100)	8 (36)	.418	3 (50)	8 (38)	.601
GERD	8 (57)	11 (52)	.782	3 (50)	11 (55)	.829
Current Constipation	5 (42)	13 (62)	.261	4 (67)	11 (61)	.808
Osteopenia	2 (14)	0 (0)	.089	1 (17)	1 (6)	.394
Current Sleep Problems	6 (43)	12 (55)	.494	3 (50)	11 (52)	.918
Frequent Infections	3 (23)	5 (24)	.961	3 (50)	3 (16)	.087
# of Hospitalizations, m (sd)	3.9 (2.7)	5.0 (5.4)	.521	3.2 (1.5)	4.4 (3.8)	.570
<b>Adaptive Functioning and Behavior</b>						
VABS-2: Adaptive Function	35 (13)	34 (14)	.839	40 (12.5)	30 (9.4)	.082
ABC: Irritability	6.0 (6.3)	4.9 (7.4)	.704	5 (7.8)	4.6 (5.6)	.920
ABC: Hyperactivity	31 (6.0)	21 (16)	.136	35 (11.8)	21.6 (13.6)	.135

VABS-2: Vineland Adaptive Behavior Scales-Second Edition; the Adaptive Function score is a composite of the communication, daily living skills, socialization, and motor skills domain scores

ABC: Aberrant Behavior Checklist, a parent questionnaire to assess behavioral problems in individuals with developmental disorders

GERD: gastroesophageal reflux disease