

ID	13618.p1	215-13023-0303	13614.p1/T131.03	13818.p1/T142.03	13818.p2/T142.04
Source	SSC	O'Roak et al. 2014	TIGER	TIGER	TIGER
Ascertainment	ASD	ASD	ASD	ASD	ASD
Gene	CHD2	CHD2	CHD2	CHD2	CHD2
Inheritance	De Novo	De Novo	De Novo	De Novo	De Novo
Mutation	Frameshift	Stop-gained	Stop-gained	Frameshift	Frameshift
age	159	72	175	179	179
gender	F	M	M	M	M
male	0	1	1	1	1
female	1	0	0	0	0
White	1	Unk	1	1	1
ID	0	1	1	1	1
ASD	1	1	1	1	1
ADHD	0		0	0	0
Anxiety	0		1	1	1
hearing problem	0		0	0	0
vision problem	0		0	1	1
Giprob	1		1	1	1
neurological problem	1	1	1	1	1
respiratory problem	0		0	0	0
heart problem	0		0	0	0
kidney/urinary problem	0		0	1	1
genital problem	0		0	1	0
birth defect	0		0	1	0
Head Circumference (cm)	51.5	59.4		54.45	54.1
HC Z-score	-1.651006711	4.951724138		-0.3375	-0.55625
VIQ	35	40	90	76	106
NVIQ	49	42	67	62	75
ADOS CSS	9	8	10	8	8
SRS Total T	90		82	74	65
Seizures	1	1	1	1	1
Aggression Caregivers	1		1	1	1
Aggression Outside Home	0		1	0	0
Self-Injury	1		0	0	0
BMI	14.6		26.2	15.7	17.4
Height (cm)	155		169	174.4	173.3
Height z-score	-0.494		0.536	0.604	0.46
Weight (kg)	35.1		74.87	47.85	52.16
Weight z-score	-1.713		1.738	-0.916	-0.406
VABS Total SS	57	36	65	66	80
Sleep Problems	1		1	1	1
Tourettes/Tics	0		0	0	0
CBCL Ext T	69		69	56	51
CBCL Int T	75		59	65	68
BMI z score	-2.2		1.6	-2.2	-1.1
BMI percentile	1.5		95	1.5	13.8
PPVT SS	40		100	90	114
Walking Age (months)	14		13	14	13
1st words age (months)	12		30	18	17

ID	T161.03	ih	12074.p1	25265.p1	MS134
Source	TIGER	Epi4K and EPGP Investigators; Epi4K Consortium et al. 2014	SSC	TIC Gen, Willsey et al. 2017	Rauch et al. 2012
Ascertainment	ASD	Seizure	ASD	Tourettes	ID
Gene	CHD2	CHD2	CHD2	CHD2	CHD2
Inheritance	De Novo	De Novo	De Novo	De Novo	De Novo
Mutation	Missense	Splice-donor and synonymous	Nonsense	Missense	Frameshift-near-splice
age	64	180	75	Unk	72
gender	F	M	F	Unk	F
male	0	1	0	Unk	0
female	1	0	1	Unk	1
White	1	Unk	1	Unk	1
ID	1	0	0	0	1
ASD	1	0	1	0	0
ADHD	0	0	0	1	0
Anxiety	1	0	0	1	0
hearing problem	0		0		
vision problem	0	0	0		
Giprob	1		1		
neurological problem	1	1	1		
respiratory problem	0		0		
heart problem	0		0		
kidney/urinary problem	0		0		
genital problem	0		0		
birth defect	1		0		
Head Circumference (cm)	50.6		49.4		
HC Z-score	-0.060150376		1.31		
VIQ	81		79		
NVIQ	85		82		
ADOS CSS	6		10		
SRS Total T	77		90		
Seizures	0	1	0		
Aggression Caregivers	0		1		
Aggression Outside Home	0		0		
Self-Injury	0		0		
BMI	13.4		13.9		
Height (cm)	123.5		113.5	110	
Height z-score	2.518		-0.626		
Weight (kg)	20.41		17.9		
Weight z-score	0.549		-1.149		
VABS Total SS	82		86		
Sleep Problems	1		1		
Tourettes/Tics	0		1		
CBCL Ext T	58		60		
CBCL Int T	54		61		
BMI z score	-1.8		-1.1		
BMI percentile	3.8		12.9		
PPVT SS	78		89		
Walking Age (months)	17		12		18
1st words age (months)	24		36		24