

eTable 1. Diagnostician and age of diagnosis of non-RTT and *MECP2* duplication participants.**Non-RTT females**

Diagnostician	Number	Percent	Median Age (y)	Interquartile Range	Minimum Age	Maximum Age
Pediatrician	3	7.3%	5.8	n.c.	4.2	5.9
Developmental pediatrician	10	24.4%	6.0	2.9-15.9	1.3	17.4
Neurologist	18	43.9%	5.0	2.8-16.5	1.0	51.6
Geneticist	9	22.0%	3.9	3.3-27.3	1.2	30.0
Other specialist	1	2.4%	6.7	n.c.	n.c.	n.c.
Other primary care provider	0					
Family member or teacher	0					
Overall	41	100.0%	5.4	3.0-14.4	1.0	51.6

Non-RTT males

Diagnostician	Number	Percent	Median Age (y)	Interquartile Range	Minimum Age	Maximum Age
Pediatrician	2	10.0%	n.c.	n.c.	1.3	4.2
Developmental pediatrician	2	10.0%	n.c.	n.c.	11.9	20.4
Neurologist	5	25.0%	4.7	2.6-9.2	1.6	12.7
Geneticist	9	45.0%	2.2	0.9-3.1	0.7	4.3
Other specialist	2	10.0%	n.c.	n.c.	4.4	18.1
Other primary care provider	0					
Family member or teacher	0					
Missing	0					
Overall	20	100.0%	3.5	1.5-5.5	0.7	20.4

eTable 1 continued.

MECP2 duplication females

Diagnostician	Number	Percent	Median Age (y)	Interquartile Range	Minimum Age	Maximum Age
Pediatrician	0					
Developmental pediatrician	0					
Neurologist	3	42.9%	37.1	n.c.	4.5	40.6
Geneticist	4	57.1%	38.5	n.c.	1.2	64.8
Other specialist	0					
Other primary care provider	0					
Family member or teacher	0					
Missing	0					
Overall	7	100.0%	37.8	3.7-46.6	1.2	64.8

MECP2 duplication males

Diagnostician	Number	Percent	Median Age (y)	Interquartile Range	Minimum Age	Maximum Age
Pediatrician	0					
Developmental pediatrician	4	14.3%	9.0	3.8-14.7	3.2	15.5
Neurologist	9	32.1%	7.3	3.2-10.5	0.6	14.8
Geneticist	15	53.6%	7.5	1.6-13.5	0.5	20.0
Other specialist	0					
Other primary care provider	0					
Family member or teacher	0					
Missing	0					
Overall	28	100.0%	7.3	2.9-12.0	0.5	20.0

n.c. = not calculated

eTable 2. Demographic characteristics of population and diagnostic category (Total N=1087).

		Classic		Atypical	
		(n=919)		(n=166)	
Ethnicity	Neither Hispanic, Latino or Spanish origin	783	85%	141	85%
	Hispanic, Latino, or Spanish origin	136	15%	25	15%
Race	Unknown or refused	23	3%	4	2%
	American Indian	6	1%	2	1%
	Asian	35	4%	4	2%
	African American	43	5%	9	5%
	Hawaiian or Pacific Islander	2	<1%	0	0%
	White	757	82%	142	86%
	Mixed race	53	6%	5	3%
Primary Residence	Home	898	98%	163	98%
	Group home	18	2%	0	0%
	Institution	3	<1%	3	2%
Population Density	Rural	404	48%	66	45%
	Suburban	241	29%	45	30%
	Urban	193	23%	37	25%
Estimated Income	Below national median	348	42%	60	41%
	Middle class	340	41%	67	45%
	Upper middle class	138	17%	21	14%

Data do not sum to total number of participants in all cases due to missing data.

Characteristics of diagnosis

Distribution: Participants were diagnosed between 1983 and 2013. Age of diagnosis ranged from 7 months to 53.0 years. Median age of diagnosis was 2.7 years (IQR 2.0 – 4.1) in classic and 3.8 years (IQR 2.3 – 6.9) in atypical RTT (Table 2).