

	Johansson, 2014	Johansson, 2014	Johansson, 2014	Johansson, 2014	Crowley, 2010	Chen, 2016
Patient ID	Family 1	Family 2	Family 3	Family 4	Patient 1	Patient 1
Sex	3F/1M	1F/1M	M	M	M	M
15q14 deletion (hg19)	NR	NR	NR	NR	g.(?_37190737)_ (37313594_?)del	g.(?_33865665)_ (38723737_?)del
Size (Mb)	58 kb dup	0.6 Mb	1 Mb	1.9 Mb	123 kb (mosaicΔ)	4.9 Mb
CGH platform	*	*	*	*	SNP array	CytoChip 60k
inheritance	maternally#	maternally#	de novo	de novo	de novo	de novo
other genetic defects	NR	NR	NR	NR	NR	NR
age	3 – 32 y	3 – 33 y	3 y	14 y	NR	foetus
cleft palate	4/4 CP (2), submucous CP (2)	2/2 CP (1), CLP (1)	normal	normal	cleft soft palate	NR
cardiac malformation	0/4	VSD (1)	normal	VSD	VSD	ToF
facial features	broad forehead, high anterior hairline, fine arched eyebrows	broad forehead, high anterior hairline, fine arched eyebrows	broad forehead, high anterior hairline, fine arched eyebrows	NR	NR	long face, low-set ears, beaked nose, prominent nasal bridge, narrow forehead, micrognathia, pointed chin
biometry	NR	NR	NR	NR	NR	W: p75 L: p50
Intellectual disability	learning problems	NR	mild ID	learning problems	NR	NR
walked at age	14 – 24 mo	21 – 36 mo	20 mo	27 mo	NR	NR
brain imaging by MRI	NR	NR	NR	NR	NR	NR
behavioral problems	NR	NR	autism	social problems	NR	NR
other features	NR	NR	NR	CAL spots	hearing loss	NR

Supplementary table 2. Molecular and clinical data from previously published patients with 15q14 deletions, harboring MEIS2. Patients with more extended 15q14 deletions proximally reaching beyond 15q13.3, or distally beyond 15q15, were excluded. 15q14 deletions, which were not delineated by means of array CGH, were excluded as well. Phenotypes from patients with microscopically visible 15q14 deletions were summarized by Erdogan *et al* (Erdogan et al. 2007) and by Chen *et al.* (Chen et al. 2016). *Array-based genomic copy number analyses were either done on the Affymetrix 250K SNP array, the Affymetrix SNP Array 6.0, or the Agilent 105K Human Genome CGH Oligo Microarray. #This copy number variant was inherited from an affected parent. ΔThis deletion is found in approximately 40% of cells. Abbreviations: CAL spots: café-au-lait spots, CLP: cleft lip and palate, CP: cleft palate, HC: head circumference, ID: intellectual disability, L: length, NA: not available, NR: not reported, mo: months, PFO: patent foramen ovale, ToF: tetralogy of Fallot, VSD: ventricular septal defect, y: years, W: weight

	Chen, 2008	Brunetti-Pierri, 2008	Erdogan, 2007	Roberti 2011	Shimajima 2017	Total
patient ID	Patient 1	patient 1	patient 1	patient 1	patient 1	15 patients
Sex	M	M	F	M	M	M/F = 10/5
15q14 deletion (hg19)	g.(?_34045708)_ (39689708_?)del	g.(?_34439336)_ (38667981_?)del	g.(?_32378020)_ (37589340_?)del	g.(?_35684649)_ (37285184_?)del	g.(?_34105933)_ (37270012_?)del	1 intragenic duplication, 10 deletions
Size (Mb)	5.6 Mb	4.2 Mb	5.3 Mb	1.6 Mb	3.17 Mb	2.83 Mb (mean)
CGH platform	Nimblegen oligo	Agilent 244k	32k tiling array	Agilent 105k	Agilent 60k	variable
inheritance	de novo	de novo	de novo	de novo	de novo	9 de novo, 1 familial deletion, 1 familial intragenic duplication
other genetic defects	NR	NR	NR	balanced translocation	NR	
age	4 y	6 y	4 y	18 y 6 mo	9 y	NA
cleft palate	cleft palate	bifid uvula	cleft palate	cleft palate	submucous CP	12/14 (85%)
cardiac malformation	VSD	PFO	atrial septal defect	normal	VSD	7/15 (46%)
facial features	long face, low-set ears, beaked nose, prominent nasal bridge, narrow forehead, small pointed chin	deep-set eyes, thin upper lip, smooth philtrum	low-set ears, short philtrum, pointed chin	deep-set eyes, short philtrum, bitemporal narrowing, pointed chin	NR	variable: bitemporal narrowing, pointed chin, beaked nose
biometry	W <p3 L <p3 HC <p3	W p25 L <p5 HC p75	W p3 L p50 HC p3	W p10-25 L <p3 HC p3	W p10 L p3 HC p50	short stature (3/6), microcephaly (3/5)
Intellectual disability	moderate ID	mild ID	moderate ID	learning problems	mild ID	mild (9), moderate (2), NA (4)
walked at age	NR	24 mo	18 mo	30 mo	20 mo	14 – 36 mo
brain imaging by MRI	normal	normal	NR	hypoplastic frontal lobes (CT)	NR	normal
behavioral problems	NR	repetitive behavior, autism	NR	NR	autism	autism (4)
other features	epilepsy	inguinal hernia, pectus excavatum, pes planus	hypotonia	irregular dentition, mild scoliosis	recurrent ear infections	

Supplementary table 2 (continued). Molecular and clinical data from previously published patients with 15q14 deletions, harboring MEIS2. Abbreviations: ASD: autism spectrum disorder, ID: intellectual disability, NA: not available, NR: not reported, mo: months, HC: head circumference, L: length, PFO: patent foramen ovale, y: years, W: weight