

Supplementary Table 1. Clinical features of individuals with deletions of 15q24

	Sharp et al. (2007) IMR349	Sharp et al. (2007) C45/06	Sharp et al. (2007) ID204	Sharp et al. (2007) IMR371	Klopocki et al. (2008)	Van Esch et al. (2009)	El-Hattab et al. (2009) Case 1
Del 15q24	72.15–76.01 Mb	72.15–76.01 Mb	72.15–73.85 Mb	70.40–74.21 Mb	72.2–75.9 Mb	70.6–73.7 Mb	72.252–75.937 Mb
Length	3.86 Mb	3.86 Mb	1.7 Mb	3.72 Mb	3.7 Mb	3.1 Mb	3.686 Mb
Inheritance	<i>De novo</i>	<i>De novo</i>	Unknown	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>
Parental origin	Maternal	Maternal	Unknown	Maternal	NA	NA	NA
Age	14 y	14 y	33 y	15 y	10 y	33 y	33 m
Gender	M	M	M	M	M	M	M
ASD	Autistic features	—	—	—	—	—	—
Language	Speaks well	Few words	Speaks well	Simple speech	Speech delay	Limited speech Echolalia Articulation difficulties	NA
Development	Mild MR	Mild MR	Mild MR	Mild MR	Mild MR	Severe MR	Mild delay
Happy facial expression	—	+	+	+	NA	NA	NA
Other behavior	ADHD	—	—	Sleep disturbances	NA	Hyperactivity Aggressiveness	NA
Growth	Height <P3 Low birth weight (<P3) HC <P3	Height <P3 Low birth weight (<P5) HC <P3	Height <P1 Low birth weight (<P2) HC <P3	Height P25 Weight P75 HC P50	Height P50 Truncal obesity HC P75	Tall (185 cm) Obese (94 kg) HC >P97 P (60.5 cm)	Short stature
Facial features	High anterior hair line Long narrow face	High anterior hair line Facial asymmetry	High anterior hair line Facial asymmetry	High anterior hair line Facial asymmetry Long narrow face	High anterior hair line Coarse face Full cheeks	High anterior hair line Long face Small maxilla Large mandible Full cheeks	High anterior hair line Broad forehead
Eye	Broad medial eyebrows Hypertelorism Down-slanting palpebral fissures	Broad medial eyebrows Hypertelorism Down-slanting palpebral fissures Strabismus Microphthalmia	Broad medial eyebrows Hypertelorism Down-slanting palpebral fissures Deep set eyes Epicanthus Strabismus	Broad medial eyebrows Hypertelorism Long palpebral fissures Nystagmus	Broad medial eyebrows Down-slanting palpebral fissures Periorbital fullness Deep set eyes Strabismus Astigmatism	Broad medial eyebrows Hypertelorism Down-slanting small palpebral fissures Strabismus	Sparse eye brows Epicanthus
Nose	Broad nasal base Flaring alae nasi	Broad nasal base Flaring alae nasi	Small nose Hypoplastic alae nasi	High nasal bridge Broad nasal base Flaring alae nasi	Hypoplastic nostrils	Broad nasal base	Depressed nasal bridge
Mouth	Long philtrum Full lower lip High arched palate Crowded teeth	Smooth philtrum Full lower lip	Long smooth philtrum Full lower lip	Long philtrum Full lower lip	Long smooth philtrum	Long smooth philtrum High palate Bifid uvula	Long smooth philtrum Full lower lip
Ear	Ear abnormalities Hearing loss Tinnitus	Ear abnormalities	Small everted ears	Normal ears Hearing loss	Normal appearance	Large ears	Ear lobe pit
Hands/feet	Long slender fingers Proximally implanted thumbs Sandal gap Deep plantar creases	Proximally implanted thumbs Single palmar crease	Small hands Single palmar crease Brachydactyly Syndactyly 2-3, 3-4 fingers and 2-3 toes — (no MRI)	Hypoplastic right thumb Contractures of fingers Pes cavus Camptodactyly of toes — (no MRI)	Broad thumbs Brachydactyly Clinodactyly Delayed carpal ossification	Long slender fingers with distal tapering Clubfeet	Small hands
Neurologic	Wide basal cisterna on brain MRI	Neonatal hypotonia Seizures Brain MRI: dysplastic corpus callosum, pituitary stalk transection, ectopic neurohypophysis, hypoplastic adenohypophysis	— (no MRI)	— (no MRI)	Hypotonia Normal brain CT	Hypotonia Delayed motor development Normal brain imaging	Hypotonia
Genital	Normal	Hypospadias	Hypospadias	Hypospadias	Micropenis Cryptorchidism	Micropenis Cryptorchidism	Normal
Musculoskeletal	Scoliosis Joint laxity	Scoliosis	Joint laxity	Joint laxity Narrow chest Valgus deformity at ankles	Joint laxity Lumbar lordosis Genua valga	Scoliosis	—
Respiratory	Recurrent ear infections	Low tone voice	Recurrent upper airway infections	Recurrent chest infections Asthma	Nasal speech Hoarse voice	High pitched voice	—
Other	Growth hormone deficiency Delayed puberty Bowel atresia Coarse hair of two different colors Hairy elbows	Edema of extremities Growth hormone deficiency Hypogonadotropic hypogonadism Bowel atresia	Feeding difficulties as a child	Diaphragmatic hernia Inguinal hernia	Inguinal and umbilical hernias Skin laxity Splenomegaly Widely spaced inverted nipples Elevated triglycerides and LDL/HDL ratio	Diaphragmatic hernia Inguinal hernias	Acute lymphoblastic leukemia Feeding difficulties Hepatomegaly

Abbreviations: ADHD, attention deficit-hyperactivity disorder; ASD, autism spectrum disorder; CT, computed tomography; F, female; HC, head circumference; ID, intellectual disability; IUGR, intrauterine growth retardation; M, male; MR, mental retardation; MRI, magnetic resonance imaging; NA, not available (not reported); P, percentile; + present; — assessed and absent; ? not assessed

Supplementary Table 1 continued

	El-Hattab et al. (2009) Case 2	El-Hattab et al. (2009) Case 3	El-Hattab et al. (2009) Case 4	Masurel-Paulet et al. (2009)	Smith et al. (2000)*	Marshall et al. (2008) SK0243-003	Present study Patient AU008
Del 15q24	72.130–76.080 Mb	70.750–73.856 Mb	70.708–73.856 Mb	70.755–73.856	70.740–73.860 Mb	69.60–73.89 Mb	69.838–72.897 Mb
Length	3.950 Mb	3.106 Mb	3.148 Mb	3.101 Mb	3.120 Mb	4.289 Mb	3.06 Mb
Inheritance	Unknown	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>
Parental origin	NA	NA	NA	NA	Paternal	Paternal	Paternal
Age	5 m	14 y	9 y	4 y	7 y	?	5 y
Gender	F	M	M	M	F	M	M
ASD	—	—	—	—	Autism	ASD	Autism
Language	NA	NA	NA	2 words only	Language regression, nonverbal	Severe delay	Language regression, nonverbal (2 words)
Development	MR	Delayed	Moderate MR	Moderate to severe MR	Severe global delay	Severe MR	Moderate MR
Happy facial expression	NA	NA	NA	—	—	—	Constant smiling
Other behavior	NA	NA	Aggressiveness	?	Chewing and mouthing behaviors	—	Hyperactive/attention deficits Sleep disturbances Aggressiveness Mouthing behavior
Growth	Short stature	Normal growth	Obesity	IUGR Height P50 Weight P10 HC P10	Normal birth weight Height P10 Weight P5 HC P50	—	Low birth weight (P5) Normal growth afterwards
Facial features	Frontal bossing Brachycephaly	Normal appearance	Facial asymmetry Round face	High anterior hair line Micrognathia	Long narrow face Pointed chin	Severe dysmorphism (no details)	High anterior hair line Long narrow face
Eye	Hypertelorism Epicanthus	Down-slanting palpebral fissures Epicanthus	Normal appearance	Sparse eyebrows Down-slanting palpebral fissures Epicanthus Hypertelorism Strabismus Anisocoria	Epicanthus Strabismus	—	Broad medial eyebrows Epicanthus Hypertelorism Congenital nystagmus
Nose	Broad upturned nasal tip	Normal appearance	Normal appearance	—	Depressed nasal bridge	—	Normal appearance
Mouth	Small mouth	Small mouth	Smooth philtrum	Long philtrum Small mouth Glossoptosis	Thin upper lip Wide dental spacing in the upper jaw Narrow anterior palate	—	Full lower lip Widely spaced teeth
Ear	Normal appearance	Thick, small ears Ear lobe pit	Cup-shaped protruding ears	Anteverted ear lobes	—	—	Protuberant ears
Hands/feet	Overriding second toes Clinodactyly Clubfeet	Long first toes and short second toes	Brachymesophalangy II and V Clinodactyly Pes planus	—	Flat nails	—	Single palmar crease Syndactyly 2-3 toes
Neurologic	Myelomeningocele Hydrocephalus	Hypotonia	Hypotonia	Hypotonia Delayed motor development Brain MRI: multiple cysts of the corpus callosum	Hypotonia Delayed motor development Regression 29 m Brisk tendon reflexes Normal brain CT	—	Brain CT: minimal cortical atrophy Delayed motor development
Genital	Normal	Hypospadias	Micropenis	Micropenis	—	—	Normal
Musculoskeletal	—	Joint laxity Pectus carinatum	Joint laxity	Kyphosis	Pectus excavatum Lax fingers	Severe scoliosis	Mild scoliosis Joint laxity
Respiratory	—	Recurrent ear infections Velo-pharyngeal insufficiency Soft nasal speech	Recurrent ear infections	—	Frequent infections	—	Recurrent respiratory infections Asthma Chronic allergic rhinitis
Other	Tetralogy of Fallot	Café au lait spots Feeding difficulties	Café au lait spots Acanthosis nigricans	—	Light skin, hair and eyes	Diaphragmatic hernia	Polyhydramnios

* The deletion reported by Smith et al. (2002) was originally mapped by FISH to 15q22-q23, but subsequent analysis using Affymetrix SNP 6.0 microarrays revealed a 15q24 deletion (Moyra Smith, personal communication)