Supplemental Table 1. Comprehensive review of characteristics of pts with 1p36 del syn

Authors	No. of Pts with 1p36-	Total No. of Pts	Age	Ethnicity	Gender	Specific genetic alteration	Dysmorphic features	Developmental features	Neurological symptoms	Brain malformations	Hearing	Vision	Behavioral	ENT	Gastrointestinal fx	Cardiac fx	Endocrine fx	Urinary fx	Skin fx	Musculoskeletal fx	Others
Yang CY et al[1]	1	1	15 wks (fetus)	0	unknown (fetus)	1p36 del	0	0	0	0	0	0	0	0	Echogenic bowel on US	b/l ventriculomegaly on US	0	0	0	0	0
Wang Q et al[2]	2	2	#1: 8y, #2: 2y	#1: unknown, #2: Chinese	M	#1: 575 Kb microdel at 1p36.11 encompassing *AHDC1* gene. #2: 491 Kb microduplication at 1p36.11p35.3 covering the entire *AHDC1* gene	# 1: sparse hair, triangular face, microcephaly, craniosynostosis, prominent forehead, protuberant ears, hypertelorism, teeth dysplasia & micrognathia. #2: macrocephaly, prominent forehead, low-set ear, deep-set eye, esotropia, downslanting palpebral fissures, broad nose with depressed nasal bridge, deep philtrum & teeth dysplasia.	#1: expressive language delay, moderate intellectual disability. #2: delayed milestones, unstable gait, ADHD, absent expressive language, moderate cognitive impairment.	#1: epileptic seizures	#1: corpus callosum thinning & delayed myelination. #2: corpus callosum thinning, delayed myelination, brain white matter dysplasia & hydrocephalus	# 1: mild hearing impairment	0	#1: self injurious behavior. #2: tongue stretching, drooling & insensitive to pain, mild anxiety	#2: laryngomalacia, OSA	#1failure to thrive	0	0	#1:nocturnal urine & fecal incontinence. #2:urinary incontinence	0	#2: overgrowth & X-ray showed advanced bone age	0

Battaglia A et al[3]	60	60	0-24 y	0	41 F, 19 M	52 had a “pure” 1p36 del, other 8 had a more complex rearrangement	Microbrachycephaly (n=39), Large, late-closing AF (n=30), Straight eyebrows (n=60), Deep-set eyes (n=60), Epicanthus (n=30), Broad nasal root/bridge (n=60), Midface hypoplasia (n=60), Posteriorly rotated/low-set/abnormal ears (n=20), Long philtrum (n=60), Pointed chin (n=60)	Expressive language absent in(n=45, 75%), few isolated words(n=10, 17%), & 2 word phrase (n=5, 8%)	Congenital hypotonia (n=57), Seizures (n=26), EEG abnormalities (n= 34), Infantile spasms: 25%	enlargement of lateral ventricles (18 pts), cortical atrophy (10 pts), enlargement of subarachnoid spaces (11 pts), diffuse brain atrophy (5 pts), & enlargement of the frontotemporal operculum (2 pts). Focal pachygyria (1 pt), coarse & nodular aspect of the cortex (1 pt). White matter anomalies (8 pts) & included delay in myelination (4 pts), PVL (3 pts), Chiari 1 malformation (2 pts). Anomalies or morphologic variants of commissural structures (8 pts), including hypoplasia, thinning, & total or partial broadness of the corpus callosum (6 pts); cavum septum pellucidum (2	SNHL (n=9, Conductive HL (n=3, Mixed HL (n=3)	strabismus (35%), hypermetropia, myopia, astigmatism (23%), nystagmus (26.5%), u/l cataract (5.9%), retinal albinism (5.9%), & u/l optic nerve colobo ma (2.9%). Visual inattention (n=28)	self-biting of hands & wrists (30%), temper tantrums (22%), reduced social interaction (52%), stereotypies like holding hands in front of face, hand washing or flapping, head shaking or banging & rocking (34%), tendency to smell or beating or rolling objects in a repetitive & purposeless way (10%), & hyperphagia (13%)		GER 7%, hiatal hernia(n=1). Hypertrophic pyloric stenosis (n=2), intestinal malrotation with malposition of the cecum (n=1), Anteriorly placed anus 3% & imperforate anus (2%). Hooked or bilobed gallbladder in 3 (17%), congenital gallstones in 1 (5.5%), & a small spleen in 1 (5.5%)	ASD (28%), VSD (23%), PDA (12.8%), bicommissural AV, PV stenosis/dysplasia/atresia, & MV insufficiency (20.5%), TOF (7.7%), CoA (5.1%), infundibular stenosis of the RV (2%), & EA (2%). Cardiomyopathy in 13 pts (27%); “noncompaction” type in 11 (23%) pts & dilated type in 2 (4%) pts.	congenital hypothyroidism(n=)3	u/l renal pelvis with hydronephrosis of the upper pole, kidney ectopia with R kidney cyst, & u/l pelvic ectasia. Cryptorchidism (40%), hypospadias (20%), scrotal hypoplasia (13%), & micropenis (7%) in males. Small labia minora (12%), clitoris hypertrophy (7%), & labia majora hypertrophy (2%) in females	Sacral/coccygeal dimples in 9	rib anomalies such as 11 ribs, or bifid/fused/enlarged ribs (16%), lower-limb asymmetry (6.5%), scoliosis (16%), congenital hip dysplasia (3%), valgus deformity of the femoral neck (3%), thinning of the long bones (3%), increased height of the vertebral bodies (3%), congenital b/l talipes valgus (3%), b/l calcaneovalgus (3%), & phalangeal hypoplasia of the hands with cone-shaped epiphyses of hands & feet (3%). Delayed bone age 22%.	0

										pts); & cyst of the cavum vergae (1 pt). Colpocephaly (1 pt), mild ventriculomegaly (1 pt), coronal & lambdoidal craniosynostosis (1 pt), d colpocephaly (1 pt), mild ventriculomegaly (1 pt), coronal & lambdoidal craniosynostosis (1 pt)											
Zhang Z et al[4]	1	1	2m	0	F	4.4 Mb interstitial del of the 1p36.33–p36.32 region.	broad face, prominent forehead, straight eyebrows & broad nasal bridge	Severe motor delay	0	ventriculomegaly	0	0	0	0	0	ASD, pulmonary HTN & LV dysfunction (poor systolic function) with DCM	0	0	wrinkled skin on abdomen, hirsutism on forehead, extensive extra skin on her back & thighs (Cutis Laxa)	0	0

Nistico' D[5]	2	2	#1: 6y, #2: 8y	Chinese	F	del 1p36.32 in both	#1: midfacial hypoplasia, b/l epicanthic fold, upslanting palpebral fissures, nasal bone hypoplasia, dental occlusion. #2: b/l epicanthus, low-set ears, severe midfacial hypoplasia, flat occiput, two posterior hair whorls, high palate	#1: normal, #2: normal except mild language delay	#1: none. #2: R frontal subdural hemorrhage with epileptic seizure, moderate hypotonia	0	neg in both	strabismus in #2	0	0	#2: poor growth due to feeding difficulties,	#2: PDA & PFO, flattening of IVS, mild tricuspid deficiency & DCM with LV non-compaction. LHF at 5mo age.	0	0	#1: one hyperpigmented macule on the abdomen & on the L-knee, hockey-stick palm crease on her L hand. #2: Hockey-stick palmar crease on her R hand, one hyperpigmented macule on the back	#1: dental anomalies of shape, number & position. ligamentous laxity of elbows & hands (especially of the thumb), b/l fifth finger clinodactyly. #2: dental anomalies, ligamentous laxity of the elbows, b/l short 5th finger	0
Yokoyama E et al[6]	1	1	13	0	M	interstitial del of 7.2 Mb with breakpoints at bands 1p36.31p36.21	coarse facies; narrow forehead; telecanthus; epicanthus; convergent strabismus; synophrys; hirsutism; a broad nasal bridge; large ears; teletelia; diastasis recti	severe dd	0	0	0	limitations to abduction with palpebral retraction & shots, hypermetropia of the R eye (b/l DRS type 1).	0	0	0	echo normal	0	Retractile testes	absent palmar creases	brachydactyly	0

Heilstedt HA et al[7]	61	61	0-11 y	0	23 M, 38 F	true terminal dels, interstitial dels, derivative chromosomes, & complex rearrangements of 1p36. Majority in most distal 4.0 Mb region of 1p36	Large AF 22/26 (85%) Microcephaly 18/30 (60%) Brachycephaly 18/30 (60%) Deep-set eyes 24/30 (80%) Flat nasal bridge 23/30 (77%) Flat nose 20/30 (67%) Thickened ear helices 16/30 (53%) Asymmetric ears 16/30 (53%) Posteriorly rotated ears 7/30 (23%) Low-set ears 7/30 (23%) Pointed chin 20/30 (67%) Short fifth finger 26/30 (87%) Fifth-finger clinodactyly 12/30 (40%)	dd in all	Hypotonia 26/30 (87%) Oropharyngeal dysphasia 21/29 (72%) Feeding difficulties in infancy 19/30 (63%) Epilepsy 15/31 (48%)	0	HL in 23	Hypermetropia 20/30 (67%) Visual inattentiveness 9/30 (30%) Strabismus 9/30 (30%) Myopia 5/30 (17%) Nystagmus 4/30 (13%)	0	0	0	DCM7/30 (23%) PDA5/30 (17%) VSD 4/30 (13%) Dilated aortic root 3/30 (10%) ASD2/30 (7%) LV dilation without cardiomyopathy 2/30 (7%) Bicommissural AV 2/30 (75) EA 1/30 (3%)	Hypothyroidism in6	0	0	Orofacial cL anomalies in 5.	0
Rudnik-Schöneborn S et al[8]	1	1	2mo	German	F	1p36 del ranging form b&1p36.13to1p36.23,comprising a loss of 8.7Mb	microcephaly, low body weight, a broad nasal bridge, hypertelorism, long eyelashes, infraorbital bulging, fleshy ears & a large mouth	dd	MR, hypotonia	agenesis of the anterior commissure & of rostral corpus callosum & partial agenesis of the septum pellucidum	0	0	0	0	FTT	0	0	0	0	0	0

Philip Campeau PM et al[9]	2	2	#1: 0 m #2: fetus aborted at 21 weeks	0	#1: F, #2: unknown	monosomy 1p36	#1: prominent occiput, high forehead, large anterior fontanel, flat facial profile, deep-set eyes, small nose with a broad base, low-set posteriorly rotated ears which were abnormally convoluted, narrow palpebral fissures, micrognathia, L single palmar crease. #2: large head, wide base of the nose, high-arched palate, low-set ears,	severe global dd	#1: hypotonia. #2: hydrocephalus with a posterior interhemispheric ventricular cyst, focal polymicrogyria of the cerebral hemispheres, & marked cerebellar hypoplasia.	#1: ventriculomegaly, enlarged lateral & 3rd ventricles, colpocephaly. moderate to severe non-obstructive hydrocephalus. Skeletal survey showed decreased ossification of the skull & cervical spine.	#1: SNHL	0	0	#1: submucosal cL palate & velopharyngeal incompetence	#1: severe gastroesophageal reflux, a	#1: asymmetrical ventricles, the RV being larger than the L, a muscular VSD & a tortuous aortic arch, PDA, hypoplastic distal aortic arch.	#1: hypothyroidism	0	#2: b/l single palmar creases.	#1: Short femurs, u/l club foot, IUGR, L pes cavus & calcaneovalgus deformity. #2: flexion contractures of both lower limbs, b/l antecubital pterygia, prominent heels & misaligned toes	0
Burszstejn AC et al[10]	1	1	8y	0	F	monosomy 1p36	deep-set eyes, low-set & posteriorly rotated ears & hypertrichosis	delayed	infantile spasms	Infantile spasms, partial seizure, asymmetrical pattern on EEG with left temporo-rolandic slow waves a/w left rolandic spikes and right temporal slow waves. cerebral malformations with agenesis of corpus callosum and a ventricular dilation.	0	b/l papillary colobo ma,	0	0	0	inter-atrial & a trabeculated inter-ventricular communications	0	0	0	brachydactyly	0

Dod HS et al[11]	1	1	25y	Caucasian	M	1p36 del	deep-set eyes, a prominent nasal bridge, large ears, elongated face with mild prognathism	profound intellectual impairment with minimal expressive language	seizures	0	0	0	0	0	0	LVNC: mildly dilated L atrium & ventricle	0	0	0	0	0
D'Angelo CS et al[12]	9	237	1-31 y	0	0	monosomy 1p36 in 9 (the study focuses on these 9 pts)	microcephaly, brachycephaly, straight eyebrows, deep set eyes, epicanthal folds, flat nasal bridge, posteriorly rotated low set ears, pointed chin,	global delay in all	hypotonia, seizures	hyperintensity in areas of white matter, craniosynostosis, lissencephaly	none	strabismus, myopia, congenital cataract, astigmatism	hyperphagia, self injury, higher pain threshold, stubbornness, aggressive, ADHD, skin picking, OCD		0	AV stenosis in 1 only	Hypothyroidism	micropenis, cryptorchidism, scrotal hypoplasia, labia majora hypoplasia	palmar creases	clinodactyly, advanced bone age	0
El-Hattab AW et al[13]	1	1	0y (newborn)	0	F	1p36 del	microbrachycephaly, a large AF, low set ears, deep set eyes, epicanthal folds, flat nasal bridge, midface hypoplasia, & high arched palate	0	germinal matrix cysts	0	nl	nl	0	0	omphalocele, cloacal exstrophy, imperforate anus	VSD, ASD, PDA, R aortic arch	0	ambiguous genitalia, prominent rugated labioscrotal folds, no apparent genital tubercle. Renal malposition & malrotation, with both kidneys positioned more inferior & the lower poles facing mediallly. 2 hemi-uteri & 2 hemi-vagina widely separated & terminating blindly	0	superior deviation of the L great toe with a large cL between the great toe & remaining toes, a syndactyly between 2nd & 3rd L toes, fusion of R 9th & 10thrib, multiple sacral segmentation defects, & diastasis of the symphysis pubis	0

																		within the pelvis.			

Rosenfeld JA et al[14]	5	5	#1: 2y, #2: 20y, #3: 17.5y, #4: 4.5y	0	#1: M, #2: M, #3: F, #4: M	monosomy 1p36	#1: telangiectasia on cheeks, straight eyebrows with synophrys, long lashes, short palpebral fissures, scooped-out, upturned, bulbous nose, pointed chin, & ears with prominent antihelix & extra helix. #2: straight eyebrows, a long & prominent nose, a high & arched palate, a thick & rolled out lower lip, retrognathia, a long neck, large hands without arachnodactyly, & mild pectus excavatum. #3: downslanting & narrow palpebral fissures, mild epicanthal folds, deficiency of inferior orbital ridges & hypoplasia of the zygomatic arches, long nose with a broad tip, overfolded helices on small ears, highly arched palate, crowding of maxillary dentition, mild maxillary hypoplasi	delay in all	#2: CT head nl, #3: nl, #4: seizures, #5: hypotonia, epilepsy	0	#2: mild u/l conductive HL	0	#1: hand flapping, sensory integration issues, high pain tolerance. #2: anger control issues, impulsivity, ADD. #3: anger, aggressive, skin picking, hyperphagia. #5:hyperphagia, outbursts of biting & screaming, chewing at his clothes, rocking while seated, & skin picking, high pain tolerance	0	#3: hyperphagia with obesity, poor feeding as a kid	0	0	0	0	0	0

							a. #4: straight eyebrows, deep-set eyes, midface hypoplasia with flattened nasal bridge & bulbous nose, & pointed chin. #5: long face with an elongated & prognathic chin, small cupped ears with a prominent helix, deep-set eyes with short palpebral fissures, low-set but not straight eyebrows, & a prominent nose													

Verrotti A et al[15]	22	22	3-31y	0	10 M, 12F	1p36 del	0	0	0	ventricular dilatations (2 pts), thinned corpus callosum (2 pts, 1 with L opercula dysplasia), corpus callosum hypoplasia (3 pts, of which 2 with ventricular dilatations), arachnoid cyst (2 pts), septum pellucid cyst, b/l alterations of the middle-posterior white matter 1 pt) & diffuse brain atrophy (1 pt)	0	0	0	0	0	0	0	0	0	0	This paper only talks about seizures & epilepsy
Carter LB et al[16]	72	72	newborns	0	0	1p36	0	0	seizures	White matter abnormalities: PVL		0	0	0	0	0	0	0	0	0	Talks more about perinatal stress & HIE type picture
Redon R et al[17]	6	6	16 m - 19y	0	5F,1M	1p36	brachycephaly, plagiocephaly,large AF, large forehead,flat midface, deep set eyes,short palpebral fissures, epicanthal folds, straight eyebrows, prominent supra-orbital ridges, small low set ears with dysplastic	MR & delay in all		0	HL in 1	nystagmus, strabismus	hyperphagia, aggressiveness, autistic features	orofacial cLing	feeding difficulties in 3/6	ASD,VSD in 1. EA in 2	0	0	0	small hands, 5th finger clinodactyly, short 5th finger	0

							helices, small mouth with downturned corners, pointed chin													
Lee ML et al[18]	1	1	6m	0	F	46,XX,del(1)(p36.3-pter)	brachycephaly, microcephaly, large AF, small low-set ears, deep-set eyes, short down-slanting palpebral fissures, hypertelorism, flat nose, flat nasal bridge, prognathism, & small hands	dd	none	none	SNHL	0	0	0	0	Bounding pulses, cardiomegaly on CXR, tachycardia & LV hypertrophy on EKG, large PDA	0	0	0	0	0
Isidor B et al[19]	1	1	9y	0	F	46,XX.ish del(1)(p36.3p36.3)	arched eyebrows, fullness of the periorbital region, moderate hypertelorism, down turned lips with a thin upper lip, long & wide spaced central incisors, low set ears, 5th finger clinodactyly & short 4th & 5th toes.	delay	axial hypotonia with hyperlaxity & peripheral hypertonia, epilepsy	corpus callosum hypoplasia.	0	0	0	0	0	0	0	0	0	hyperlaxity with recurvatum of both knees.	0
Saito Y et al[20]	1	1	2y	0	M	46,XY,der(1)t(1;4)(p36.23;q35)	broad forehead, depressed nasal bridge, long philtrum, thin lips, hemangioma on the L lower cheek, round & thick auricles, b/l single palmer crease & clinodactyly of the 5th toes		seizures, mild hypotonia with exaggerated knee tendon reflex & extensor plantar response	b/l perisylvian PMG with R-sided predominance	0	0	0	0	0	mild overriding of Valsalva sinus on the ventricular septum	0	0	0	0	0

D'Angelo CS et al[21]	1	41	13y (only one had 1p36 del)	Brazilian	F	1p36 terminal del	high & prominent forehead, narrow bitemporal diameter, thick eyebrows with mild synophrys, slightly deep-set eyes, downslanting palpebral fissures, posteriorly rotated ears, prominent cheeks & a small mouth	delay	hypotonia as neonate	normal	normal	Congenital b/l cataract, myopia & astigmatism	hyperphagia, obesity, hyperactive, crying episodes, irritable, ADD, skin picking	0	0	normal echo	diabetes	0	hypertrichosis	b/l fifth-finger clinodactyly	0
Fitzgibbon GJ et al[22]	1	1	9y	Caucasian	F	duplication of a region of 1p32.3, del within 1p36.32	small mouth with heaped up palate, a small chin & a small over folded ear, straight eyebrows, 5th finger clinodactyly & short toes	delayed	0	0	0	hyperm etropia	0	0	0	0	0	0	0	0	0
Puvbandits in S et al[23]	1	1	11y	0	M	46,XY,del(1)(p36.33)	enlarged AF, epicanthal folds, high arch palate, deep-set eyes, short palpebral fissures, broad & flat nasal bridge, midface hypoplasia, pointed chin, & abnormal ears	delay	0	choroid plexuses & b/l ventriculomegaly	0	R esotropia	temper outbursts, episodes of violent physical activity, banging or throwing objects, striking/biting people, & self-injuring behavior with hand & wrist biting	0	0	dextrocardia, a large muscular VSD, & absence of the L SVC.	0	undescended testes	single crease, grey hair at 11 yrs of age	R clavicular hyperplasia, hypoplasia of the middle phalange of the 5th finger, clinodactyly	0
Shapira SK et al[24]	13	13	prenatal - 11y	0	7 M, 6F	1p36 del	Microcephaly, Brachycephaly Large AF Low anterior hairline Small ears Large ears Thickened ear helices Ear-pinna dysplasia Ear	delayed	hypotonia, seizures, MR, infantile spasms, myoclonus	lateral ventricle asymmetry, ventricular enlargement, & focal atrophy.	HL in 4	Eye problems in 6	hand biting, banging or throwing objects, striking people, & episodes of violent physical activity.	0	0	infantile cardiomyopathy, minor congenital heart defects	precocious puberty in 2, growth delay in 8	cryptorchidism in 1	0	0	0

							asymmetry Low-set ears Posteriorly rotated ears Short palpebral fissures Palpebral fissures (up) Palpebral fissures (down) Deep-set eyes Hypotelorism Flat nasal bridge Flat nose High nasal bridge Long-appearing philtrum Prognathism Pointed chin Small hands/feet, 5th finger short/clinodactyly Scrotal hypoplasia													
Tan TY et al[25]	1	1	0y (newborn)	0	M	1p36	b/l epicanthal folds, deep-set eyes, flat nasal bridge, small nose, prominent chin, & short neck	delayed	0	0	nl	0	poor social interaction, repetitive stereotypic non-choreiform movements, poor eye contact & poorly intelligible speech	0	Elevated LFTs (GGT, ALP, & ALT) s/o partial biliary tract obstruction. Liver biopsy showed periportal, pericellular, & perivenular fibrosis a/w macrovesicular fatty change, suggestive of non-specific non-alcoholic steatohepatitis. Gastric antral nodularity, with evidence of H.pylori	CXR showed cardiomegaly, but echo nl.	hypercholesterolemia, type 2 diabetes	0	Hypertrichosis, chronic subungual warts	osteoporosis, multiple repeated fractures, vit D def, broad ribs, thick calvarium with mild verticalization of the skull base, & mild thoracic scoliosis, mild anterior wedging of T11 & T12 vertebral bodies, & more marked anterior wedging of L3 with an irregular superior end-plate, b/l coxa valga, broad distal epiphysis & metaphyseal flare of femur	0

															infection. Increased cerulloplasmin & proportionate increase in serum copper.					
Neal J et al[26]	1	1	3y	0	F	1p36.22-> 1pter del.	epicanthal folds b/lly, posteriorly rotated & slightly low set ears, a broad nasal bridge, thin upper lip, & anteverted nares	delayed	0	several nodular gray matter heterotopia along the wall of the L lateral ventricle. Rostrum of corpus callosum was truncated, the ventricles were slightly enlarged, patchy areas of hyperintensity were seen in periventricular & subcortical white matter b/lly consistent with delayed myelination, T12 to T6 syrinx	HL	Duane syn, limited abduction of L eye	laughs spontaneously & makes guttural utterances.	0	0	0	0	0	0	3rd digit overrides the 2nd digit in both feet, short 5th digit in R hand, scoliosis	0
Sab Rudnik-Schöneborn S et al[27]	1	1	0y (newborn)	0	F	1p36	microcephaly, low body weight, a broad nasal bridge, hypertelorism, long eyelashes, infraorbital bulging, fleshy ears & a large mouth	delayed	0	agenesis of the anterior commissure & of the rostral corpus callosum & partial agenesis of the septum pellucidum (	0	0	0	0	0	0	0	0	0	0	0
Chen E et al[28]	1	1	0y (newborn)	0	N	46,XY, dup(1)(p36.31p36.33).	short palpebral fissures, blepharophimosis, congenital cataracts, posteriorly angulated	delayed	severe hypotonia, seizures	asymmetry of gray matter in cerebral hemispheres & diffuse atrophy & delay	SNHL	Dense congenital cataracts, amblyopia, exotropia, aphakia	0	choanal atresia	b/l inguinal hernias, poor feeding, dilated esophagus with abnormal	0	0	micropenis	sacral dimple	broad proximal phalanges, tapered distal digits, fifth finger clinodactyly, b/l 2–3 toe syndactyly, & overlapping toes, &	transient hypogammaglobulinemia of infancy & combined T & B cell deficiency

							& low-set ears			of white matter myelination, respectively.		, ptosis.			peristalsis					severe osteopenia of all bones.	
Saito S et al[29]	1	1	0y (newborn)	0	F	1p36	round face with short palpebral fissures, a depressed nasal bridge, small mouth, high arched palate, micro/retrognathia, & low set ears	delayed	seizures	b/l perisylvian polymicrogyria & periventricular nodular heterotopia	0	0	0	0	0	two VSDs (membranous & muscular), PDA, EA with moderate TV regurgitation, & LVNC, CHF & pulmonary HTN	0	0	0	0	0
Breckpot J et al[30]	1	1	0y (newborn)	Caucasian	F	1p36	short palpebral fissures, hypertelorism, dysplastic low-set ears & large AF	delayed	seizures	lateral ventricles were enlarged, mild periventricular leukomalacia	SNHL	0	0	narrow piriform aperture on CT, slight choanal atresia, , a triangular-shaped palatum & abnormal maxillary dentition (with a single medial incisor bud	anal stenosis	Dextroposition of aorta & a mild Ebstein malformation of TV with mild ventricular hypertrophy	0	0	sacral dimple	b/l clubfeet, limited hip abduction, platyspondyly & small iliac wings	0
Rankin J et al[31]	1	1	0y (newborn)	0	F	1p36	prominent forehead, straight & low set eyebrows, upslanting & short palpebral fissures, small mouth, pointed chin, small low set ears, brachycephaly	delayed	hypotonia	0	0	0	0	0	Duodenal atresia	ASD	0	0	0	0	0
Tsuyusaki Y et al[32]	2	2	#1: 0y (newborn), #2: 10y	#1: Japanese, #2: Russian	#1: M, #2: M	1p36	#1: deep-set eyes a/w almond shaped palpebral fissures, straight eyebrows, prominent forehead, broad & flat nasal root, & pointed chin. #2: deep-set	delayed in both	hypotonia & difficulty in sucking in both. Seizures in #2.		0	strabismus in #2	#1: hyperphagia, #2: hyperphagia, temper outbursts & impulsivity	0	0	0	decreased GH in #2	0	hypopigmented skin in both	#1: small & narrow hands with a straight ulnar border & small feet with short toes.	0

							eyes, straight eyebrows, pointed chin													
Knight-Jones E et al[33]	4	4	0-3 y	Caucasian	#1, 2, 3: M. #4: F	partial monosomy for 1p36.3.	#1: wide AF, midface hypoplasia, wide asymmetric brow, deep-set eyes, small convex nose with flattened tip, small mouth with thin narrow lips, ridged palate, ears low-set, microcephaly, plagiocephaly. #2: plagiocephaly, wide asymmetric brow, unusual scalp hair whorls, epicanthic folds, horizontal palpebral fissures, stellate irides, small mouth with thin lower lip, flat philtrum, & round lower face. #3: microcephalic, widely patent AF & PF, brachycephaly & plagiocephaly. Prominent brow, deep-set eyes, sculptured appearance of the orbits, flattened pinnae. #4: ears were low set & asymmetrical, the R ear being cup	dd in all	#1: L hemiplegia due to R MCA occlusion, Lennox Gastaut syn. #2: hypotonia, seizures. #3: hypotonia, seizures. #4: hypotonia, seizures	#1 & 2: cerebral atrophy. #3: moderate dilatation of the 3rd & lateral ventricles, slight prominence of the cerebral sulci. #4: dilated lateral ventricles	#4: HL	#1: fine rotary nystagmus, intermittent alternating convergent squint. #2: myopia & rotary nystagmus. #4: optic atrophy	#1: biting his fingers. #2: self-injuring behaviour	#4: u/l cL lip	0	#1: cardiomyopathy #3: VSD	0	#3: R inguinal hernia, undescended testes, & coronal hypospadias. #4: large labia	#1: nail dysplasia. #3: preponderance of whorl patterns on the fingertips	#1: unusual hands with flattened spatulate digits, mild soft-tissue syndactyly, partial webbing from thumb to index finger. #2: scoliosis. #3: tapering fingers. #4: clinodactyly of 5th fingers. Only 11 ossified rib pairs. D6 vertebral body showed a sagittal cL (butterfly vertebra), dorsal & lumbar vertebral bodies with short AP diameter. small & osteopenic pelvis. slightly limited abduction of the L hip in early infancy, scoliosis	0

							shaped, plagiocephaly & a 3rd fontanelle													
Bahi-Buisson N et al[34]	91	91	1-25 y	0	64 F, 27 M	pure 1p36 del in 80, complex rearrangements in 11.	straight eyebrows, deep-set eyes, flat nasal bridge, midface hypoplasia, & pointed chin.	delayed	seizures, MR	0	0	0	0	0	0	0	0	0	0	talks more about seizures & their types.
Girardeau F et al[35]	1	1	16y	0	M	del of 1p36.3	large AF & widely separated sagittal sutures, plagiocephaly with a broad flat forehead & hypotelorism, R ear with no lobe that was 5 mm shorter than the L ear, small nose with deviation of nasal septum, small mouth, overcrowded dentition, a flat ridged palate, & midface hypoplasia.	delayed	seizures	0	0	Moderate b/l optic atrophy, b/l rotatory nystagmus,	0	0	0	0	0	small genitalia, & no evidence of puberty	0	scoliosis, mild syndactyly of the 2nd & 3rd fingers & short thumbs, short digits & both halluces showed distal valgus deviation.	0
Keppler-Noreuil KM et al[36]	3	3	0y (newborn)	0	all F	1p36.22-+pter del	#1: large AF, small, upward slanting palpebral fissures, low set ears with R preauricular ear pits, microstomia, & polydactyly of the L hand. #2: large AF, small palpebral fissures, b/l iris colobomas, low set, small ears, & microstomia. #3:	delayed	#1: hydrocephalus. #2: grade 2 IVH, hydrocephalus. #3: seizures	0	0	#1: esotropia,	0	0	#3: GERD, poor feeding	#1: patent foramen ovale, patent ductus arteriosus (PDA), & L ventricular dysfunction	0	0	#3: telangiectatic skin lesion on her mid forehead with several hyperpigmented macules on her extremities	#3: tapered fingers with fifth finger clinodactyly,	0

							hypotonia, large AF, brachycephaly, hypertelorism, inferior epicanthic folds, low set ears with prominent auricular roots, flat nasal bridge														
Minami K et al[37]	1	1	1y	Japanese	M	46, XY, del (1)(p36.2)	microcephalus, large AF, low-set ears, deep-set eyes, flat nasal bridge, nasal deformation with cL lip & palate, pointed chin, & generalised hypotonia	delayed	seizures	0	0	0	0	0	midgut volvulus a/w malrotation & annular pancreas	VSD & CoA.	0	0	0	0	0
Zenker M et al[38]	4	4	0-9.5 y	0	1,2,3: F. 4: M	1p36	#1: brachycephaly, frontal bossing, deep set eyes with straight eyebrows, a hypoplastic midface, & relative prognathism, low-set, posteriorly rotated, & protruding ears, flat nose & nasal bridge, pointed chin, small mouth, highly arched palate, small hands & feet, & 5th finger brachydactyly. #2: prominent forehead & supraorbital ridges, straight	delayed	#1: Poor sucking & hypotonia, seizures, MR. #2: Hypotonia & severe psychomotor retardation. #3: hypotonia, MR. #4: Poor suck & hypotonia, seizures	#2: moderately enlarged cerebral ventricles, hypotonia, MR. #3: slightly enlarged ventricles. #4: slightly enlarged lateral ventricles & narrowing of the corpus callosum.	#1: SNHL	#1: Visual inattentiveness, rotatory nystagmus. #2: b/l optic atrophy. #3: decreased visual acuity.	#1: self injurious behaviour. #3: Hyperactive, aggressive behaviour		0	#3: PDA. #4: small muscular VSD	#1: hypothyroidism	0	0	#2: Diminished muscular mass & tone lead to severe valgus deformity of the ankles	0

							eyebrows, deep set eyes, a hypoplastic midface, & relative prognathism, Posteriorly rotated, mildly dysplastic ears, thick scalp hair & a low-set posterior hairline. #3: bossing forehead with prominent supraorbital ridges, straight eyebrows, deep set eyes, a flat midface, & thick hair. #4: large extending down to the nasal root, prominent forehead & deep set eyes with epicanthus inversus. low set ears, high palate, atypical single palmar crease, & proximally set 2nd toes.													
Wang BT et al[39]	1	1	0y (newborn)	Taiwanese	F	1p36	prominent forehead, loose/redundant skin over the nape (persistent since birth), adducted thumbs/halluces, narrow vermillion border of the lips, & downturned corners of the mouth	delayed	hypotonia	0	HL	0	0	0	0	0	0	0	0	0	0
Ferril GR et al[40]	1	1	0y (newb	0	0	del1p36.33-1p36.32, dup1q44.	0	0	0	0	0	0	0	buccopharyngeal membra	0	0	0	0	0	0	0

			orn)										ne & cLed soft palate. The tongue appeare d to be trapped in the posterio r orophar ynx & hypoph arynx by the membra ne thus required tracheos tomy. retrogna thia, glossopt osis, & an anterior & small larynx								
Zhu X et al[41]	1	1	0y (ne wb orn)	Chinese	M	576 kb del of 1p36.33–1p36.32	upslanting palpebral fissures, broad nasal tip, elongated philtrum, a pointed chin & small mouth	delayed	seizures	enlarged cerebral ventricle s.	0	0	0	0	0	ASD	0	0	0	hypoplastic thumb of L hand, b/l fifth finger clinodactyly	0
Shim ada S et al[42]	5 0	50	1-25y	Japanese	15 M, 35 F	1p36	Microcep haly 83%, Brachyce phaly 65% , Straight 84% , Deep-set eyes 93% , Epicanthu s 86%, Broad nasal root/bridg e 97% , Long philtrum 63% , Low set ears 88%, Pointed chin 89% , Late closure of the AF56% , High palate 29%, CL palate 22%, CL lip 16% , CL jaw + 2%	delayed	Epileps y in 70% , Infantil e spasms in 16% , hypoton ia, difficult y swallow ing	Perivent ricular nodular heteroto pia (PVNH) , Polymic rogyria, Cortical dysplasi a Enlarge ment of lateral ventricle s, Delay in myelinat ion, Hypopla sia of corpus callosum , Chiari type II malform ation Cavum septum pellucid um, Choroid plexus cyst, Enlarge ment of subdural space,	HL	Strabis mus 15%, ametro pia 26%, nystag mus 20%, oculom otor disturb ance 2%, colobo ma 2%, congen ital microp hthalmi a 2%, retinitis pigmen tosa 2%	temper tantrums , self injury	stenosis of nasal cavity, laryngo malacia, adenoid hypertro phy, OSA, atresia of external acoustic opening	NASH, constipat ion	PDA: 37%, VSD: 37%, ASD: 16%, AS 4%, PS: 4%, Aortic valve prolapse: 2%, EA 6%, DORV: 6%, HLHS: 2%, PFO: 2%, PAPVC 2%, PA: 2%, Positional anomaly of valsalva sinus 2%, Pulmonary HTN: 8%, LVNC 10%, dilated cardiomyop athy 4%	hypothyroi dism, obesity, precocious puberty	Renal hypopla sia 2%, hydrone phrosis 5%, pyelecta sis 2%, nephron ophthisi s 5%, ectopic kidney 2%, ambigu ous external genitali a 7%, cryptorc hidism 645, scrotal hypopla sia 14%	atopic dermatiti s	Finger abnormalities 15%, Limb deformity 4%. 11 ribs 4% , Scoliosis 25% Congenital dislocated hip 4% Developmental dysplasia of the hip 2%	0

										Arachnoid cyst										
Haimi M et al[43]	1	1	0y (newborn)	mother is of KurdisheTunisian origin, & the father of GermanePolisheMoroccan origin	F	1p36	flat occiput, mild hypoplasia of the midface, deep-set eyes, downslanting palpebral fissures, short philtrum, small ears, straight eyebrows	delayed	hypotonia, mild to mod MR	mild dilatation of the frontal subarachnoid spaces	0	strabismus	hyperphagia		elevated LFTs, lysosomal disorder	0	precocious puberty	0	hirsutism	short 4th toes	0
Di Donato N et al[44]	2	2	#1: 27y #2: 30 (brothers)	0	M	#1: del in 1p36.32,	#1: d high forehead, deep set eyes & prominent ears. #2: a flat face with a high forehead, horizontal eyebrows, upslanted palpebral fissures & thin vermilion of the upper lip.	#1: mild learning disability. #2: mild speech delay	0	0	0	0	0	0	0	0	0	0	0	oligosyndactyly of the R hand with mesomelic shortening of the R arm, length asymmetry of the lower extremities & the hypoplastic R foot with absent 5th metatarsal & fifth toe. Progressive maxillary retraction	0
Stagi S et al[45]	2	2	#1: 6y, #2: 10y	0	F	#1: del of approximately 1.5 Mb that involved the 1p36.33 region. #2: del of approximately 2.5 Mb that involved the 1p36.33 – 1p36.32 region.	#1: small mouth with heaped-up palate, small chin, small folded ear, straight eyebrows, fifth finger clinodactyly & short toes. #2: truncular obesity, scoliosis, small hands & feet, a small mouth with heaped-up palate, small chin & a small folded ear, straight eyebrows, fifth finger clinodactyly & short	delayed in both	0	0	SNHL in #2	0	hyperphagia in #2	0	0	persistent PDA in #2 at 6 ms of age & elevated BP.	hyperinsulinism & insulin resistance in #1, excessive weight gain, hyperinsulinism, insulin resistance & DM type 2 in #2	0	0	0	0

							toes, & mild learning disability.													
Gamba BF et al[46]	1	1	0y (newborn)	0	M	five dels adjacent to one duplication spread over p35.3-1p36.33	microcephaly; square face; straight/sparse eyebrows; short palpebral fissures; blepharophimosis; posteriorly angulated, low-set, & abnormally modeled ears; midface hypoplasia; b/l cL lip & palate	delayed	lumbar meningocele, Infantile spasms	cerebral ventricular dilation				choanal atresia				micropenis		broad proximal phalanges; tapered distal digits; 5th finger clinodactyly; & overlapping toes	0
Shiba N et al[47]	1	1	0y (newborn)	0	F	46,XX,del(1)(p36.31)	b/l low set small malformed ears, prominent forehead, deep set eyes, flat nasal bridge, pointed chin, short neck, & b/l single transverse creases.	delayed	hypotonia, seizures	cervicomedullary compression at the skull base, b/l ventriculomegaly	SNHL	myopia with astigmatism	Self-injurious behavior (arm chewing) & teeth grinding	0	severe conj hyperbili, feeding difficulties, GERD	PDA,VSD. dysplasia of atrioventricular valves, but no cardiomyopathy on autopsy.	GH deficiency, Precocious 2ndary sexual characteristics & clitoromegaly	0	0	0	0
Kawashima H et al[48]	1	1	12y	0	F	1p36 del	saddle nose & frontal bossing without long philtrum, chin was sharp & her eyes were deep set	MR & delay	0	0	0	0	0	0	anomalous arrangement of pancreatobiliary duct with accessory pancreatic duct, which formed a loop in front of the musculus papillaris & ran to the CBD (complex type)	PDA	0	0	0	0	0
Nicoulaz A et al[49]	1	1	0y (newborn)	Swiss	F	16 Mb terminal del of the 1p36.33-1p36.13 region	deep set eyes, small palpebral fissures, pointed chin & low-set ears with thickened helices	pt died on DOL 2	0	ventriculomegaly & marked pachygyria, absent septum pellucidum & thinned	0	0	0	0	intestinal obstruction & suspected duodenal atresia	TOF	0	0	0	camptodactyly of both digits IV, & joint contractures	died 48 hrs after birth

										corpus callosum										
Gajecka M et al[50]	2	2	#1: 3m, #2: 1m	0	F	1p36	#1: upslanting palpebral fissures, characteristic eyebrows, slightly depressed nasal bridge with minimal midfacial flattening. #2: mid-facial flattening, depressed nasal bridge & mildly upslanting palpebral fissures	dd	hypotonia	0	#1 & 2: mild SNHL	0	0	0	0	#1: LVNC of free wall & apex as well as aortic regurgitation, trivial tricuspid regurgitation, borderline normal LV shortening, & mild LV dysfunction with DCM. #2: mild non-compaction cardiomyopathy (clinically silent)	premature thelarche	0	0	#1: generalized ligamentous laxity including decreased hip rotation & joint movement including pronation of the ankles	0
Vieira GH et al[51]	1	1	15y	0	F	1p36	square-shaped face, relative prognathism, microbrachycephaly, hypoplasia of the midface, deep-set & closely spaced eyes, everted & ‘‘tented’’ upper lip, & dental anomalies. short, broad hands, recurrent ear infections, onychotillomania, & sleep disturbances	dd	sleep disturbance, seizures, hypotonia	0	0	0	aggressive behavior & ‘‘biting the hands’’, onychotillomania	repeated ear infections	0	0	0	0	0	Brachydactyly/ camptodactyly. Short hands & feet.	0
Buck A et al[52]	1	1	16y	Korean	F	de novo single copy number loss of 210 kb (hg18, chr1:2908089-3118435) at 1p36.32.	midface hypoplasia, short nose, anteverted nares, thin upper lip, oligodontia. prominent forehead, bitemporal narrowing, low-set ears with prominent antihelix, hypoplasti	delayed	hypotonia	0	0	strabismus, esotropia	self-injurious skin picking, tic-like phenomena (eye blinking, squinting, nasal twitching, & throat clearing), & unprovoked	0	0	0	0	0	hypoplastic nails	brachydactyly	0

							c tragus, upslanting palpebral fissures, midface hypoplasia, microdontia, oligodontia, & abnormally shaped tooth cusps						temper tantrums							
Shimada S et al[53]	2	2	#1: 5y, #2: 18y	Japanese	F	1p36	#1: deep-set eyes, epicanthal folds, bulbous & small nose, broad nasal root, long philtrum, & pointed chin. #2: straight eyebrows, deep-set eyes, & pointed chin.	delayed	hypotonia	0	0	#2: strabismus, astigmatism, & hyperm etropia	#1: hyperphagia. #2: temper tantrums	#1: OSA, repeated ear infections. #2: cL palate	0	#1:ASD, VSD (both closed spontaneously). #2: PAPVC.	0	0	transverse palmar crease, hyperpigmentation & depigmentation.	0	0
Brazil A et al[54]	40	40	12-15y: 14, 16-19y: 7, 20-23: 10, 24-27: 6, 28-31: 1, 34y: 1, 46y: 1	0	29 F, 11M	1p36	widely spaced nipples, high arched palate, bulbous nose, wide nasal bridge, pointed chin	delayed	hypotonia: 26. seizures: 13, spasticity: 9	0	Conductive: 7, SNHL: 5, mixed: 3	myopia: 15, strabismus: 15, hyperm etropia: 3. Others like cortically visually impaired, exotropia, astigmatism, ptosis, nystagmus, cortical blindness, divergent squint in 9.	0	cL palate in 4, cL lip l	constipation 16, diarrhea 10, reflux: 8, abdominal pain: 8, ulcer: 2, celiac disease in 1.	VSD: 8, PDA: 5, EA 2, PFO1,ASD 1, narrow aortic arch1, bicuspid AV in 1, tricuspid aortic valve in 1, TOF in 1, transient myocardial function in 1. LVNC.	0	kidney infection n 9	0	s scoliosis, kyphosis, sacral dimple, short stature, clinodactyly, syndactyly, tapered fingers, shortened fingers, “toes growing over each other,” broad & angulated thumbs & great toes, calcaneovalgus, rocker bottom feet in 9.	0
Çöllü M et al[55]	1	1	4y	Iraqi	M	1.77 Mb terminal del of chromosome 1p36	microbrachycephaly, a narrow forehead, flat supraorbital ridges, straight eyebrows, midface hypoplasia, broad nasal bridge, b/l epicanthal folds, a	delayed	0	0	0	0	0	0	anus was patent, located anteriorly. b/l paracardiac diaphragmatic hernia	0	0	CBE was visible above symphysis pubis, through which urine was dripping. Epispadias, hypopla	absent umbilicus	pectus excavatum, absent lower half of the sternum, externally rotated legs & a waddling gait. Very wide pelvis with absent pubic symphysis	0

							short nose with broad nasal tip, & smooth long philtrum with thick lips & pointed chin											stic scrotum & absence of palpable testes			
Murakoshi M et al[56]	1	1	24y	Japanese	F	1p36	deep-set eyes, prominent nasal bridge, pointed chin, & clinodactyly	delayed	seizures, hypotonia	partial agenesis of corpus callosum	0	0	0	0	0	DCM, HTN	abdominal paraganglioma	0	0	0	0
Zagalo A et al[57]	1	1	9y	0	0	terminal del with 139 Mb corresponding to the region 1p36.33	straight eyebrows, deep-set eyes, broad nasal root, flat nasal bridge & pointed chin & prepubertal genitalia.	delayed	0	0	0	0	0	OSA, recurrent resp infections	0	HTN	hypercholesterolemia	0	0	0	0
Lee J et al[58]	1	1	25y	0	F	1p36	0	0	0	0	0	0	0	0	0	LVNC, LV hypertrophy with a strain pattern, prominent LV trabeculation, end diastolic compacted myocardium to noncompacted myocardium ratio met the CMR diagnostic criteria of > 2.3	0	0	0	0	0
Pearce FB et al[59]	1	1	4.5 y	0	F	1p36	large AF, brachycephaly, deep-set eyes with epicanthal folds, & midface hypoplasia	0	0	0	0	0	0	0	Urine organic acids revealed elevation of p-OHphenylacetic acid	EKG: sinus tachycardia with L atrial enlargement & nonspecific ST-T changes. Echo: normal segmental anatomy with dilated LV with severe systolic dysfunction (shortening fraction less than 10%) & localized noncompaction, which was present in the LV apex & extended to	0	0	0	0	0

																a prominent area on the lateral wall. Myocardium showed fibrosis, severe myocyte hypertrophy, & subendocardial myofibrillar loss, especially immediately below the endocardial layer. Extensive endocardial fibroelastosis .					
Ting TW et al[60]	2	2	0y (newborn)	#1: Malay, #2: chinese	F both	1p36	#1: minor facial anomalies, including deep set eyes & midface hypoplasia, but not suggestive of any specific syndromic diagnosis.	dd	hypotonia, seizures in both	#1: R subependymal heterotopia	0	0	0	#2: laryngomalacia	0	#1: large PDA, DCM, LVNC. #2: persistent pulmonary HTN, PDA, LVNC	0	0	0	0	0
Zanardo ÉA et al[61]	1	1	0y (newborn)	0	F	1p36	prominent forehead, epicanthus, ocular hypertelorism, low-set ears, anteverted nares, ogival palate, delayed dentition, synophrys, prominent supraorbital ridges, straight eyebrows, long & prominent philtrum & a deep sacral pit	delay	hypotonia, seizures	0	0	0	0	0	0	stenosis of the L pulmonary artery, with significant hemodynamic effects;	hypothyroidism	0	hypertrichosis	5th finger clinodactyly of the hands, & overlapping of the 3rd to the 2nd toe	0
El Waly B et al[62]	1	1	35y	0	F	1p36.23 & 12p13.1 duplications	0	delay	b/l opercular syn, dysphasia, difficulties in chewing & swallowing, continuous drooling, accomp	abnormal temporo-parietal gyration which is consistent with b/l polymicrogyria	0	0	0	0	0	0	0	0	0	kyphoscoliosis	0

									anied by hemiparesis, microcephaly, seizures											
Reish O et al[63]	5	5	#1: 5y, #2: 44y #3: 47y, #4: 6y, #5: 3y.	#1,2,3, 4, 5: Caucasian	1,2,3,4: F. 5: M	#1: 46,XX,- 1, +der(l)t(1;19)(p36.31;q1 3.42)pa. #2: 46,XX,- l,+der(l)t(l;19)(p 36.31;q13.42. #3: del 1p36.3. #4: del of the distal b&s 1p36.32 & 1p36.3. #5: 45,XY-1,- 13,+der(l)t(1;13) (p36.33;q12.1)	#1: large anterior fontanelle, large forehead, deep-set eyes with b/l epicanthal folds. Flattened midface, depressed nasal bridge. Short mouth with downturned corners, palate was high & arched, low set ears with poor helix formation. #2: frontal prominence & prominent supraorbital ridges. Deeply set eyes with down slanting palpebral fissure & notable synophrys. Midface was hypoplastic with a depressed nasal bridge. Low set ears, small with folded pinnas. Her mouth was small with high-arched palate & down-turned corners, & relative prognathism. #3: long forehead, deep-set eyes with b/l cataract &	delayed	#1: seizures, #2: seizures, disturbed sleeping patterns & aggressive, self-abusive behavior. #3: seizures, quadriparesis & disuse atrophy of her legs.	#1: "split" sutures & "atrophy of the brain. #2: quadriparesis due to congenital spinal stenosis of her cervical spine. Disk herniation of (25-6 & C6-7, with significant cord compression, & spinal stenosis of C4-7. #4: ventricular system & cortical sulci were mildly prominent consistent with cerebral atrophy. #5: prominence of the sulci with generalized atrophy.	#1: SNHL. #2: partial HL in L ear.	#1: b/l optic atrophy, #5: R eye exotropia	#1: temper tantrums, self-abusive behavior #3: crying out & screaming with no evident provocation. Teeth grinding, self abusive behavior, hitting her head, scratching her peroneal region, biting & chewing her hands & pinching herself. severe temper tantrums. #4: attention span was very short, & she was easily frustrated with frequent temper tantrums & head-banging. #5: temper tantrums & self-abusive behavior including head-banging & biting himself	0	0	#4: heart failure & DCM with diminished LV activity at 4 m age. #5: PDA	#3: small breasts, sparse pubic & axillary hair	#3: urinary & bowel incontinence. #5: R undescended testicle	#1: deep sacral dimple with a small tuft of hair.	#1" L 2nd incisor & canine were fused. Short hands & fingers, clinodactyly of the 5th finger. Small feet with short toes. Unsteady gait with a wide base. B/l genua valga legs. Proximal thumb proximally inserted & midphalanx hypoplasia of the 5th fingers. #2: thoracolumbar kyphosis, short hands & fingers & hypoplastic proximal 5th phalanges b/lly. B/l camptodactyly of the 2nd, 3rd & 4th fingers. Small feet with 3rd toe overriding the 4th toe on the L. #3: thoracolumbar kyphoscoliosis. Small feet & toes. #4: short hands & feet with tapered fingers & a short thumb on the L hand. #5: postaxial hexadactyly of R foot, . Lambdoid craniosynostosis. atrophy of his R sternocleidomastoid. 2nd toe overlapped the 3rd toe b/lly. Flexible joints & mild symmetric hypotonicity.	Pt 2 is the paternal aunt of pt 1.

							hyperopia, down-slanting palpebral fissures & an amaurotic expression. Hypoplastic midface, mouth was down-turned at the corners, & higharched palate. Prominent chin with square jaws laterally. Low set ears with prominent antihelix. #4: flat midface, a large forehead & a depressed nasal bridge. deep-set eyes & L esotropia. mouth small, high-arched palate & capped teeth, chin was prominent with prognathic appearance low-set ears, small & cup-shaped ears. #5: large forehead & a flat occiput. hair had an abnormal frontal whorl, deep-set eyes, hypertelorism & synophrys. Midface hypoplasia, a small mouth with high-arched																

							palate & a pointed chin. low-set ears were 4.5 cm (<-2 S.D.) long, with an underdeveloped helix in the upper portion & prominent antihelix. B/l supernumerary nipples.														
Eugster EA et al[64]	1	1	4.5 y	0	F	mos46,XX,del(1)(p36.33).ish del(1)(p36.33)(D1Z2−),15q11.2(D15S10 × 2, SNRPN × 2)[47]/46,XX.ish 1p36.3(D1Z2 × 2),15q11.2(D15S10 × 2, SNRPN × 2)	narrow forehead, deep-set eyes, with ''almond'' -shaped palpebral fissures, flat midface, & a small down-turned mouth with a high arched palate, small ears	delayed	seizures, hypotonia	0	HL	strabismus	temper outbursts, impulsivity, & self-injurious behaviors (biting her hands when angry). Hyperphagia	0	0	0	0	0	0	short fingers, 5th finger clinodactyly	0
Riegel M et al[65]	1	1	4y	0	F	del of 1p36.3-1pter	skull was brachycephalic, asymmetric face, prominent forehead with high frontal hairline, synophrys, downward slanting palpebral fissures, inner epicanthal folds, broad & short nose with upturned nares; long & flat philtrum, narrow vermilion of the lips, small mouth with downturned corners, prominent lateral palatine ridges, R ear larger	delayed	seizures, hypotonia	brain atrophy with enlarged ventricles & subarachnoid spaces	0	0	0	0	0	EA & PDA(PDA	0	0	0	small hands with transient transverse palmar crease on the R side, camptodactyly of both fifth fingers with shortness of the middle phalanx & only one flexion crease on the R side; small feet with short metatarsals of 2nd toes with broad terminal phalanges & nails	0

							than the L & posteriorly rotated; asymmetry of the thorax with the L side smaller than the R.													
Finelli P et al[66]	1	1	0y (newborn)	0	M	46,XY,der(1)-inv(1)(p22p36.2)del(1)(p21p22)	large AF, midface hypoplasia, hypertelorism, deep-set eyes with epicanthic folds, ¯at nasal bridge with bulbous nasal tip & anteverted nares, large ears, long philtrum, small mouth, high palate, & a short frenulum linguae	delayed	0	minimal pituitary hypoplasia	0	0	hyperphagia	0	0	0	0	0	0	0	0
Neumann LM et al[67]	1	1	12y	German	M	46XYish del(1)(pter->p36.33~36.32)	obesity, mildly prominent chin, broad neck, truncal obesity	delayed	seizures, episodes of atonic hemiparesis, wide based ataxic gait		0	mild b/l ptosis, b/l strabismus	stereotypic hand movements	0	0	0	0	0	circumscribed occipital alopecia	fusion of R upper canine & 1st premolar tooth	Died from sepsis & multiorgan failure
Digilio MC et al[68]	1	44	0.2-37y	0	27 M, 17 F	del 1p36 & del 8p23.1 are the most frequent chromosomal imbalances a/w EA, common genes include GATA4 (pts with del 8p23.1), NKX2.5 (published pts with isolated EA), & a hypothetical locus in the 1p36 region (pts with del 1p36)	0	0	0	0	0	0	0	0	0	EA was a/w an additional CHD in 16/44 (36%) pts, including 6/12 (50%) syndromic, & 10/ 32 (31%) nonsyndromic	0	0	0	0	EA was syndromic in 12 (27%) pts, nonsyndromic in 32 (73%). (CHARGE syn in 2 pts, VACTERL association, Noonan syn, Kabuki syn, Holt-Oram syn, Cornelia-de Lange syn two each).

Abbreviations:

Week	Wk
Ultrasound	US
Year	Y
Female	F
Male	M
Month	M
Patient	Pt
Unilateral	u/l
Bilateral	b/l
Hearing loss	HL
Sensorineural	SN
And	&
Aortic valve	AV
Pulmonic valve	PV
Mitral valve	MV
Tricuspid valve	TV
Atrial septal defect	ASD
Ventricular septal defect	VSD
Patent ductus arteriosus	PDA
Tetralogy of Fallot	TOF
Coarctation of aorta	CoA
Right ventricle	RV
Left ventricle	LV
Features	Fx
Hypertension	HTN
Dilated cardiomyopathy	DCM
Patent foramen ovale	PFO
Interventricular septum	IVS
Left heart failure	LHF
Hemorrhage	H'ge
Deletion	Del
Developmental delay	Dd
Mental retardation	MR
Associated with	a/w
Congenital heart disease	CHD
Ebstein anomaly	EA
Syndrome	Syn
Anterior fontanelle	AF
Right	R
Left	L
Second	2nd
First	1st
Third	3rd
Periventricular leukomalacia	PVL
Gastroesophageal reflux	GER

Fifth	5th
Anterior fontanelle	AF
Left ventricular noncompaction	LVNC
Attention deficit hyperactivity disorder	ADHD
Obsessive compulsive disorder	OCD
Superior vena cava	SVC
Liver function tests	LFTs
Congestive heart failure	CHF
Middle cerebral artery	MCA
Posterior fontanelle	PF
Common bile duct	CBD
Partial anomalous pulmonary venous connection	PAPVC

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