Pt number (DDD number)	**1** (271894)	**2** (262889)	**3** (259077)	**4** (261411)	**5** (264961)
Age	8.3	7	16.75	14	4.39
Sex	F	M	F	F	F
Gestational age at birth	38/40	38/40	38/40	34/40	36/40
Birth weight (kg)	2.636 (19th centile)	2 (1st centile)	2.58 (15th centile)	1.8 (14th centile)	2.5 (42nd centile)
Birth height				*	
Birth OFC (cm)	30.8 (2nd centile)			*	34 (92nd centile)
Current weight (kg)	27 (48th centile)	17.5 (0.4th centile)	73.4 (91st-98th centile)	54.1 (67th)	15.5 (27th centile)
Current height (cm)	123 (9th-25th centile)	108.5 (0.4th centile)	150.5 (0.4th-2nd centile)	148.8 (5th centile)	94 (1st centile)
Current OFC	50.5 (1st centile)	49 (< 0.4th centile)	54 (9th-25th centile)	51 (at chronological age 8.87 years; 11th centile)	47.5 (1st centile)
Developmental delay	Yes	Yes	speech and language delay	Yes	Yes
ID	Yes	Yes	Yes (moderate)	Yes	Yes (moderate/ severe)
Autism	Yes (mild)		Yes	Yes	No
Seizures	No	No	No	No	No
Feeding difficulties	Yes (gastrostomy fed from 4 months to 4 years)	Sensory-based feeding difficulties. He is feed orally as well as through his percutaneous gastrostomy	No	Yes (from infancy, severe gastro-oesophageal reflux).	Poor suck for few days at birth. GO reflux. NGT inserted at 4 months for 3-4 months.
Eye problems	Poor 3-dimensional vision	Divergent squint in his right eye (initially, bilateral squint)	Reading glasses	Lacrimal duct atresia	Astigmatism
Hearing loss	No	No. Grommet insertion February 2010	mild unilateral hearing loss	High frequency sensorineural hearing impairment	Bilateral grommets inserted at 3 years

Pt number (DDD number)	**1** (271894)	**2** (262889)	**3** (259077)	**4** (261411)	**5** (264961)
Facial dysmorphism	Hypertelorism, USPFs, wide nasal bridge, short philtrum, glabellar haemangioma noted at 14 months, now faded.	Arched eyebrows, USPFs, bilateral ptosis, low-set ears, anteverted ears, triangular nose, flat philtrum, small mouth, very thin upper lip	Short palpebral fissures, hypertelorism, USPFs, low-set ears, posteriorly-rotated ears, short philtrum, small mouth	Flat facial profile, epicanthic folds, long eyelashes, depressed nasal bridge, low set and posteriorly rotated ears, a very short nasal columella, long philtrum, thin upper lip, small mouth and tongue	Long eyelashes, telecanthus, epicanthic folds, low-set ears, short philtrum, wide nasal bridge, broad nasal tip, low posterior hairline, capillary haemangioma (faded) over nasal root when seen at 9/12
Digital anomalies	5th finger clinodactyly, extra crease 5th finger	No	Tapering fingers with 5th finger clinodactyly, radial deviation at PIP joint of both 4th fingers, fetal finger pads, broad big toes with hallux valgus	Prominent foetal finger pads, relatively short fingers, deep-set fingernails	Broad tips to fingers
Limbs	Extra skin crease on forearms and back of right thigh	Circumferential skin folds	Circumferential skin folds	Normal	No
Palate	Normal		Short uvula	Normal	No
CVS	ASD and mild pulmonary branch stenosis	Perimembranous VSD with pulmonary valve stenosis	Normal echo	Normal echo and ECG	No but never had an echo
Genital anomalies		Paraphimosis	None. Menarche aged 13.5 years	Not specified	Wide labial opening with partially deficient hymen
Bowel problems		Gastro-oesophageal reflux in infancy	Constipation	Anal stenosis, severe gastro-oesophageal reflux, eosinophilic colitis, recurrent GI infections	No
Urinary problems		No	No	Nil	No
Skin	Normal	Hypertrichosis over elbows, legs and back	possible sebaceous cyst over scalp	Nil	No

Pt number (DDD number)	**1** (271894)	**2** (262889)	**3** (259077)	**4** (261411)	**5** (264961)
Spine	Normal	No	Straight	Nil	No
Neurological findings		Hypotonia	None	Global developmental delay and intellectual disability	No
MRI brain	Severe hypoplasia of the corpus callosum with only the anterior genu present. Arnold-Chiari 1 variant	Underdeveloped corpus callosum	Normal	Normal (including pituitary)	MRI not done
Array	Normal	Normal	Normal	Normal	Not done
CDK13 mutation	p.Gly714Arg	p.Gly717Arg	pGly717Arg (mosaic; higher mutation load in saliva compared to blood)	p.Val719Gly	p.Lys734Arg
Other features	Microdontia	Nasal speech. Sacral dimple. Recurrent mouth ulcers, occasional nosebleeds	Obesity (BMI 33.2; >99.6th centile)	Sacral dimple, delayed ossification of hand bones of 9 months, borderline growth hormone deficiency, oligodontia, thick curly hair, obesity with onset of puberty	Poor sleep
Previous diagnosis	Ohdo-like	Ohdo syndrome	Ohdo syndrome	Kabuki, Angelman and Smith-Magenis considered	

Pt number (DDD number)	**6** (258830)	**7** (265645)	**8** (265813)	**9** (259460)	**10** (270818)
Age	12.68	11	4	4.84	8.16
Sex	F	F	F	F	M
Gestational age at birth	35/40	40/40	38/40	41/40	39/40
Birth weight (kg)	1.7 (3rd centile)	3.64 (70th centile)	2.5kg (9th centile)	3.31 (42nd centile)	2.552 (2nd-9th centile)
Birth height					
Birth OFC (cm)				34 cm (33rd centile)	2nd centile
Current weight (kg)	41 (59th centile)	22.5 (34th centile) at age 7	15.1 (9th centile)	10.06 (9th centile)	19.7 (0.4th-2nd centile)
Current height (cm)	136 (4th centile)	106.8 (1st centile) at age 7	95.8 (0.4th centiel)	81 cm (`5th centile)	121.5 (9th-25th centile)
Current OFC	53 (16th centile)	51.2 (10th centile); 53 cm on 17.11.15 (9th-25th centile) aged 11 years	46.7 (<0.4th)	47 (8th centile)	44 (<0.4th centile at 1.25 y)
Developmental delay	Yes	Yes	Yes, especially speech	Yes	Yes
ID	Yes	Yes	Yes (moderate)	Yes (moderate)	Yes
Autism	No	Yes	No	No	Yes
Seizures	No	History of absence seizures with normal EEG. Single generalised seizure. Diagnosis of autonomic seizures and cyclical vomiting.	yes absences, myoclonic, grand mal	Febrile seizures age 1, none since	Yes (absences, on Valproate)
Feeding difficulties	Neonatal period	NG fed for the first eight to nine months of life, then required gastrostomy.	Yes	Yes, velopharyngeal dysfunction, abnormal swallow	Very slow feeder at birth, irritable with feeds
Eye problems	Squint, myopia	Squint	No	No	No
Hearing loss		No	No	No	No

Pt number (DDD number)	**6** (258830)	**7** (265645)	**8** (265813)	**9** (259460)	**10** (270818)
Facial dysmorphism	Plagiocephaly, short palpebral fissures, ptosis, USPFs, small mouth with downturned corners	Highly-arched, thick eyebrows, USPFs, epicanthic folds, preauricular pit, hypertelorism, low-set ears, posteriorly-rotated ears, prominent nasal bridge, thin upper lip, small mouth, low posterior hairline	Brachycephaly, thick hair, low forehead, thick eyebrows, hypertelorism, telecanthus, epicanthic folds, low-set ears, overfolded helix, small mouth with downturned corners	Hypertelorism, USPFs, bulbous nose, small mouth, thin upper lip. Very curly hair, not in keeping with other family members.	Lots of head hair, DSPFs, abnormality of pinna, short nose, long philtrum, thin upper lip
Digital anomalies	Camptodactyly of 2nd-5th fingers	Hyperextensible finger joints, 5th finger clinodactyly	Tapering fingers, proxinal thumbs	5th finger clinodactyly, 2nd and 3rd toe clinodactyly	5th finger camptodactyly
Limbs				Joint hypermobility	Normal
Palate		No		Normal	Normal
CVS	ASD, VSD	VSD	mild pulmonary artery branch stenosis, ASD	ASD	Secundum ASD - tiny and haemodynamically insignificant
Genital anomalies				No	Normal
Bowel problems		Episodic vomiting, gastro-oesophageal reflux, constipation	Constipation	No	Constipation
Urinary problems				No	No
Skin	Dry skin		hypertrichosis	Normal	Normal

Pt number (DDD number)	**6** (258830)	**7** (265645)	**8** (265813)	**9** (259460)	**10** (270818)
Spine	Hyperlordosis	Scoliosis		Normal	Normal
Neurological findings		Spastic diplegia		Hypotonia	Motor tics
MRI brain	Normal	Periventricular leukomalacia	Normal	MRI not done	Absent corpus callosum
Array	Normal	Normal	Normal	Normal	Normal
CDK13 mutation	p.Arg751Gln	p.Asn842Ser	p.Asn842Ser	p.Asn842Ser	p.Asn842Ser
Other features	Truncal obesity	EEG normal, given diagnosis of Panayiotopoulos sydrome aged 5y		Pica. Central sleep apnoea requiring overnigh oxygen.	Only 2 hours sleep in 24 hours (on melatonin)
Previous diagnosis			CHARGE-like		Rubinstein-Taybi syndrome

Pt number (DDD number)	**11** (not in DDD)	**12** (331720)	**13** (264613)	**14** (270857)	**15** (301509)
Age	4.67	10	8.33	8.42	8.16
Sex	F	F	F	F	F
Gestational age at birth	37/40	37/40 weeks (cesarean section)	38/40	40/40	term
Birth weight (kg)	3.054 (75th centile)	2.484 (2nd-9th centile)	3.061 (55th centile)	3.4 (50th centile)	3.71 (75th to 91st centile)
Birth height	47				
Birth OFC (cm)	34 (75th-91st centile)	34 cm (75th-91st centile)			
Current weight (kg)	13.8 (0.4th-2nd centile)	17 kg (<0.4th centile)	20.5 (2nd-9th centile)	26.3 (50th centile)	33.6 (86th centile)
Current height (cm)	98.7 (3rd centile)	120 cm (<0.4th centile)	116.5 (<2nd centile)	127.5 (25th centile)	126.1 (34th centile)
Current OFC	49.2 (30th centile)	46 cm (<0.4th centile)	52 (25th centile)	53 (25th-50th centile)	51.6 (49th centile)
Developmental delay	Yes (mild) - walked at 19 months, mild speech delay	Yes	Yes	Yes	moderate global
ID	No (low normal range; non verbal Wechsler IQ: 86)	Yes (severe)	Yes	Yes	Currently 3 years behind at school. Gap increasing
Autism	No	No	No (autistic traits)	No	Not diagnosed, but has hand flapping ++ and motor steryotypies
Seizures	No	No	No	Generalised tonic-clonic and focal	
Feeding difficulties	Neonatal period	Yes (neonatal)	Infancy	Yes (tube fed from 2 months, gastrostomy fed from 11 months to 5 years)	Slow to handle mixed textures, but okay now
Eye problems	No	No (narrow lacrimal duct)	Squint, mild ptosis	No	Bilateral esotropia, especially when tired
Hearing loss	No	No	No	No	No hearing loss

Pt number (DDD number)	**11** (not in DDD)	**12** (331720)	**13** (264613)	**14** (270857)	**15** (301509)
Facial dysmorphism	Glabellar congenital oedema and haemangioma, prominent metopic suture, hypertelorism, USPFs, posteriorly rotated ears, wide nasal bridge, broad nasal tip, small mouth	Yes	Medial epicanthic folds, broad nasal bridge, small mouth	Glabellar haemangioma, short USPFs, telecanthus, medial epicanthic folds, broad nasal bridge, posteriorly rotated ears, small mouth.	Blepharophimosis, epicanthic folds, broad nasal tip, tented upper lip, posteriorly rotated ears
Digital anomalies	5th finger clinodactyly, deep palmar creases	Fetal pads and 5th finger clinodactyly		Fetal finger pads	Tapering digits, broad sandle gap in feet
Limbs	No problems	Narrow heel bilat		Joint hypermobility	Pes planus
Palate	No cleft but nasal speech (?VPD)	No	Normal	High-arched	Normal
CVS	Secundum ASD (operated)	ASD		None	No abnormality - has seen cardiology
Genital anomalies	No	No		No	No
Bowel problems	No	Loose stools before 2 yrs of age		Gastro-oesophageal reflux, constipation	No
Urinary problems	No	Urinary tract infection twice		Normal	No
Skin	No problem	No	Unusual wrinkled skin over palms, severe eczema	Normal	normal

Pt number (DDD number)	**11** (not in DDD)	**12** (331720)	**13** (264613)	**14** (270857)	**15** (301509)
Spine	sacral dimple (normal spinal USS)	No		Normal	Straight
Neurological findings	Normal	hypotonia	Hypotonia	Painful dystonic spasms	Hand flapping, steryotypies involving hand and feet
MRI brain	MRI not done	Thin corpus callosum	Normal	Normal	MRI not done
Array	Normal	2008: dup chr3: 2715059-3212741 (pat inherited - suspect benign) and dup chr3: 3562164-3833018 (pat inherited - suspect benign)	Normal	Normal	normal
CDK13 mutation	p.Asn842Ser	p.Asn842Asp	p.Arg860Gln	p.Val874Leu	p.Asp896Asn
Other features	Left congenital torticollis, recurrent ENT and URTS (<4 y)	metopic synostosis, congenital diaphragmatic hernia, choledochal cyst (type 4), recurrent respiratory infections	Low IgA and IgM levels, parental consanguinity, brother has homozygous MICU1 mutation with myopathy, movement disorder, developmental delay, autism and hypothyroidism.	Heterozygous for paternally-inherited p.Thr83Met variant in *TECTA* gene	had some small peg shaped teeth in primary dentition, plus oligodontia, secondary dentition normal. Has skin picking habit with scars on limbs++
Previous diagnosis		No diagnosis			Ohdo considered

Pt number (DDD number)	**16** (271710)
Age	3.54
Sex	F
Gestational age at birth	36/40 (twin pregnancy)
Birth weight (kg)	2.693 (62nd centile)
Birth height	
Birth OFC (cm)	
Current weight (kg)	14.32 (96th centile)
Current height (cm)	92.1 (99th centile)
Current OFC	
Developmental delay	Yes (mild)
ID	Yes
Autism	Yes (mild)
Seizures	No
Feeding difficulties	Yes
Eye problems	No
Hearing loss	

Pt number (DDD number)	**16** (271710)
Facial dysmorphism	Arched eyebrows, prominent forehead, deep-set eyes, hypertelorism, epicanthic folds, low-set ears, posteriorly-rotated ears, prominent ear crus
Digital anomalies	Mild 5th finger clinodactyly
Limbs	
Palate	
CVS	Normal
Genital anomalies	No
Bowel problems	Mild constipation
Urinary problems	No
Skin	Normal

Pt number (DDD number)	**16** (271710)
Spine	Normal
Neurological findings	Hypotonia
MRI brain	MRI not done
Array	Not done
CDK13 mutation	c.2898-1G>A
Other features	No other features
Previous diagnosis	Ohdo-like