

Table S1. Clinical details of individuals with a CTNND1 variant

Participant	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6	Patient 7	Patient 8	Patient 9	Patient 10	Patient 11	Patient 12	Patient 13	TOTAL	Kievit et al 2018	Ghoumid et al 2017	
DDD ID	NA	NA	294023	278766 NA	-	-	278000	264418 NA	NA	NA	265667	281120 -	-	-	-	-	
Variant	V148D>T24	V148D>T24	R461*	R461*	R797*	L494H*	L494H*	G533A>G	S68*	S68*	L2302.SA>G	H813P>T3	H813P>T3	-	-	-	
Sex	Female	Female	Male	Male	Female	Female	Male	Female	Male	Male	Male	Male	Male	67/7M	-	-	
<b>Craniofacial</b>																	
Cleft lip/palate	no	no	submucous cleft soft palate & bifid uvula	no	no	yes	right unilateral cleft lip and palate	no	bilateral cleft lip and palate	right unilateral cleft lip and palate	left unilateral cleft lip and palate	left unilateral cleft lip and palate	left unilateral cleft lip and palate	(8/13)	Yes	Yes	
High arched palate	yes	yes	yes	no	no	no	yes	yes	no	no	no	yes	yes	(7/13)	NR	NR	
Thin upper lip	yes	no	no	no	no	no	yes	yes	no	no	yes	yes	no	(2/13)	NR	NR	
Choanal atresia	bilateral	left	no	no	no	no	no	bilateral	no	no	right	no	no	(4/13)	NR	NR	
Dysplastic ears	-	low set ears	asymmetric low set ears; overfolded helices.	slightly antverted	no	small	small	low set ears; overfolded helices.	low set; overfolded helices.	low set; overfolded helices.	low set; overfolded helices.	preauricular right pit, mild conductive hearing loss bilaterally.	-	-	(9/13)	Yes, 1 subject	NR
Wide nasal bridge	yes	no	yes	no	no	no	yes	yes	yes	yes	yes	yes	yes	(11/13)	NR	NR	
Broad nasal tip	yes	no	yes	no	no	no	yes	yes	yes	yes	yes	no	no	(7/13)	NR	NR	
Mid-face hypoplasia	yes	yes	yes	no	no	yes	yes	yes	no	yes	yes	yes	yes	(9/13)	NR	NR	
Mandibular prognathism	yes	no	yes	no	no	no	no	yes	no	yes	yes	no	yes	(5/13)	NR	NR	
Brachycephaly	no	brachycephaly	no	plagioccephaly & brachycephaly	no	no	no	no	no	no	brachycephaly	no	no	(3/13)	NR	NR	
<b>Eyes and eyelids</b>																	
eyelet synchia bilaterally, distichiasis & mild ectropion.	no	anlykoblepharon bilaterally, distichiasis & hyperopia.	narrow upslanted palpebral fissures, hooded eyelids and telecanthus, no ophthalmic concerns.	wide palpebral fissures with scleral show, hypertelorism, ectropion, distichiasis.	sleeps with eyes open; ectropion; everted outer third of the lower eyelids.	narrow palpebral fissures, anlykoblepharon, sleepes with eyes open, watery eyes, distichiasis.	narrow upslanted palpebral fissures, hooded eyelids; had a squint. No ophthalmic concerns.	narrow palpebral fissures, hooded eyelids; bilateral eyelid tags at birth; Tanykoblepharon, telecanthus.	narrow upslanted palpebral fissures, hooded eyelids, telecanthus.	narrow upslanted palpebral fissures, hooded eyelids, telecanthus.	narrow upslanted palpebral fissures, hooded eyelids, telecanthus.	narrow upslanted palpebral fissures, hooded eyelids, telecanthus. No ophthalmic concerns.	narrow upslanted palpebral fissures, hooded eyelids and telecanthus. No ophthalmic concerns.	Narrow palpebral fissures (9/13); hooded eyelids (9/13); telecanthus (7/13); mild ectropion (4/13); anlykoblepharon (3/13)	Reported BCD eyelid malformations.	Reported BCD eyelid malformations.	
Highly arched eyebrows	yes	yes	no	no	no	yes & bushy eyebrows	yes & interrupted eyebrows	yes	no	no	yes	yes	yes	(8/13)	NR	NR	
Thin lateral eyebrows	yes	no	no	no	yes	yes	yes	yes	no	yes	yes	no	no	(8/13)	NR	NR	
<b>Dental/oral</b>																	
Hypodontia	yes, 9 permanent teeth	yes, 5 primary & permanent; other premolars could not be assessed at this age.	yes, 12 permanent teeth	yes (number not determined)	yes, 3 permanent teeth	-	-	yes, 8 permanent teeth	7 missing back molars, ND	7 missing back molars, ND	yes, 3 permanent teeth	no	yes, 3 permanent teeth	(8/13)	Yes	Yes	
Delayed dentition	yes; supernumeration of E3 with apical resorption from 24. Delayed development 34 & 44 with ankylosis submerged and resorbed 64.	yes; impacted E3 and delayed eruption of the second primary molars	-	yes	yes	-	-	ND	ND	ND	yes, ectopic 23	no	delayed development of 21	(6/13)	Yes	NR	
Abnormal crown form	yes; mesoangular eruption of 24, fusion of teeth, long cone-shaped tooth.	yes; macrodontia of E3 & 75; supernumerary tooth in 64-65 area; tooth 53 is dysmorphic and diminutive and tooth 73 is dysmorphic and rotated.	yes; peg 22	no	yes; multiple cortical/peg teeth.	-	disorganised teeth	yes; peg 21, 22 and history of neonatal tooth.	'jointly teeth'	yes; fused incisors	yes; diminutive 22	no	no	(9/13)	Yes	Yes	
<b>Limbs</b>																	
Hands	NAD	NAD	single transverse palmar crease, right hand; thumbs and fifth finger slightly shorter.	NAD	NAD	small hands; short fifth finger.	small hands; short fifth finger.	single transverse palmar crease, right hand.	NAD	NAD	NAD	NAD	NAD	(7/13)	No	No	
Feet	NAD	slight 2,3-syndactyly of feet bilaterally, long halluces.	slight 2,3-syndactyly on feet, lateral deviated halluces.	pes planus, hallux valgus.	NAD	short toes	short toes	NAD	NAD	NAD	NAD	NAD	NAD	(7/13)	Yes	Yes	
<b>Systemic Phenotypes</b>																	
Cardiac	hypoplastic aortic arch, ASD, VSD, MVS.	VSD, PDA, PFO.	NAD	VSD, secundum ASD.	NAD	NAD	NAD	TDP - repair done at 4 months; pulmonary regurgitation - valve replacement at 9 years.	NAD	NAD	hypoplastic aortic arch with severe CoA; postmembranous VSD; surgeries VSD at 1 month & 2 years.	NAD	NAD	(6/13)	No	No	
Neurologic	NAD	sensory processing disorder. Quere autism & ADHD, GDD; aggressive behaviour.	speech delay early life; autism & ADHD, LD, defiant behaviour.	autistic traits, poor coordination.	NAD	NAD	NAD	NAD	NAD	NAD	increasingly challenging behaviour; anxiety; parental concern re:autistic traits.	increasingly challenging behaviour; speech and language delay; developmental delay.	severe speech and language delay; autism; mild LD, OCD.	mild LD.	(8/13)	No	No
Voice anomaly	no	nasal speech	no	no	no	no	no	no	no	no	no	no	no	(3/13)	No	No	
Other skeletal anomalies	severe scoliosis	-	delayed fontanelle closure	joint laxity	-	scoliosis as an adult; short stature	-	-	-	-	stridor; hoarseness	no	no	(5/13)	No	No	
Cancer	no	no	no	no	no	no	no	ovarian dysgerminoma at 12 years of age; stage III in the left ovary; underwent left oophorectomy followed by chemotherapy.	no	no	no	no	no	(1/13)	No	No	
<b>Others</b>	restrictive lung disease	partial agenesis of the corpus callosum	velopharyngeal insufficiency; early onset puberty; bowel problems infant toddler years	-	-	hypothyroid	abnormal nasal morphology; had rhinoplasty for a collapsed nose	macroglossia (enlarged tongue)	glue ear	glue ear	cryptorchidism on the left side; multiple grommets	coronal hypoplasias (repaired); monozygotic twin of Patient 13	monozygotic twin of Patient 12	-	No	No	

Abbreviation: DDD ID, Deciphering Developmental Disorders patient identification number; NA, not applicable; NAD, no abnormality detected; ND, not determined because of non-availability; 23, upper left permanent canine; 63, upper left primary canine; 24, upper left permanent first premolar; 34, lower left permanent first premolar; 44, lower right permanent first premolar; 84, lower right primary first molar; 22, upper left permanent lateral incisor; 21, upper left permanent central incisor; 75, lower left primary second molar; 73, lower left primary canine; 64, upper left primary first molar; 65, upper left primary second molar; VSD,