

Table S1. *Spectrum of 84 published CTNNB1 cases*

#	Genetic mutation				Gender	Age (yrs)	Birth measurments			Facial dysmorphism	Eye conditions	Micro-encephaly	Axial hypotonia/spasticity	Achieving milestones				Additional clinical features	Source (quality score)
	Exon	Variant	Amino acid change	Mutation type			Lenght (cm)	Weight (g)	OFC (cm)					Sitting (mo)	Crawling (mo)	Walking independetly	Speaking		
1	E3	c.99_100delTG	p.Gly34Asnfs*15	Frameshift	Male	5,5	49	3160	NA	+	Strabismus	+	+/+	14 ; still head-leg	25	6 yrs (cannot stand alone)	Short sentences	Concentration is limited; sensitive to noises, friendly	Kuechler et al. 2015 [2]
2	E3	c.160G>T	p.Glu54*	Nonsense	NA	NA	NA	NA	NA	NA	Strabismus	NA	+/+	NA	NA	Walking with ASO	NA	Dystonia; middle ear effusions	Jin et al. 2020 [5]
3	E3	c.214_215del	p.Gln72fs	Frameshift	Male	5	NA	NA	NA	NA	Normal	+	+/+	NA	NA	NA	Absent speech	GDD, asthma, spastic diplegia	Rossetti et al 2021 [22]
4	E3	c.232C>T	p.Gln78*	Nonsense	Male	11	53	2900	NA	+	Strabismus, hyperopia, astigmatism	-	+/+	NA	Didnt crawl	3 yrs; at 11 yrs coordination problem	Unclear speech; at 11 yrs regression	aggression, anxiety, friendly, sleep problems, stereotypic movements	Winczewska-Wiktor et al. 2016 [23]
5	Intron 3	c.242-1G>C	Splice mutation	Splice	Female	11	NA	NA	NA	NA	Divergent strabismus	-	+/+	NA	NA	NA	NA	NA	Ho et al. 2021 [24]
6	E4	c.274C>T	p.Gln92*	Nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Abnormality of the nervous system	Retterer et al. 2016 [25]
7	E4	c.283C>T	p.Arg95*	Nonsense	Female	4	51	3300	NA	+	Normal	+	+/+	NA	12	4 yrs	Speech apraxia, ~50 words	Aggression when younger, now has stopped	Kuechler et al. 2015 [2]
8	E4	c.283C>T	p.Arg95*	Nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Posey et al. 2017 [26]
9	E4	c. 367 C>T	p.Phe99PhefsTer5	Frameshift	NA	NA	NA	NA	NA	NA	NA	+	+/+	NA	NA	NA	Non-verbal	Bilateral frontal pachygyria; spastic diplegia	Jin et al. 2020 [5]
10	E4	c.400C>T	p.Gln123*	Nonsense	Male	12	NA	NA	NA	NA	Normal	-	+/+	NA	NA	NA	NA	osteogenesis imperfecta, dystonia, blue sclera	Ho et al. 2021 [24]
11	E4	c.423_424insG	p.Tyr142Valfs*4	Frameshift	Female	5,6	51	3450	33	+	Strabismus	+	+/-	NA	24	not yet	Severe, few words (30), sign language	repetitive movements, poor sleeping in infancy	Kuechler et al. 2015 [2]
12	E4	c.493C>T	p.Gln165*	Nonsense	Male	7	NA	NA	NA	NA	Left eye caract, hypermetropia, astigmatism	+	+/+	NA	NA	NA	NA	Dystonia	Ho et al. 2021 [24]
13	E5	c.521; T>C	p.M174T	Missense	Female	34	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Severe simple schizophrenia	Levchenko et al. 2015 [27]
14	E5	c.705dupA	p.Gly236Argfs*35	Frameshift	Female	14	NA	2510	32	+	Strabismus	+	+/-	12	NA	4.5 yrs	Babbling at 3yrs, 14 yrs speaking simple sentences, read simple words	Autism	Tucci et al. 2014 [28]
15	Intron 5	c.734+1G>T	Splice mutation	Splice	Female	32	50	3200	33,5	+	Strabismus	+	+/+	2-5 years (40mo)	NA	No	Absent speech	Autism, aggression, ritualistic behaviours, involuntary gestures	Verhoeven et al. 2020 [1]
16	Intron 5	c.734+1G>A	Splice mutation	Splice	Female	49	NA	NA	NA	+	FEVR	NA	+/+	NA	NA	NA	Absent speech	Involuntary rotation of head	Wang et al. 2019 [29]

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	Exon	Variant	Amino acid change	Mutation type			Lenght (cm)	Weight (g)	OFC (cm)					Sitting (mo)	Crawling (mo)	Walking independently (yrs)	Speaking		
17	Intron 5	c.734+1G>A	Splice mutation	Splice	Female	27 years	NA	NA	NA	+	FEVR	NA	NA	NA	NA	Walking at 49 yrs (ataxic)	Absent speech	deep tendon hyperreflexia, babinski sign, paroxysmal dystonic HN	Wang et al. 2019 [29]
18	E6	c.755delTins AAC	p.Leu252*	Nonsense	Female	15,3	51	2950	33	+	Strabismus, hyperopia mild esotropia, mild hyperopia and astigmatism, strabismus	-	+/+	NA	NA	10 yrs	2 words	Dystonia	Kuechler et al. 2015 [2]
19	E6	c.776_777deITC	p.Leu259Profs*11	Frameshift	Male	7	NA	Poor (NA)	NA	NA		+	NA/+	NA	NA	NA	NA	Aggressive behaviour, GDD, restless leg, bradykinesia, toe walking, spastic diplegia	Rossetti et al 2021 [22]
20	E6	c.796C>T	p.Gln266*	Nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Retterer et al. 2016 [25]
21	E6	c.799_809deIGAAGGAGCTAAinsGAA	p.Gly268TrpfsTer5	Frameshift	Female	7	NA	3100	NA	+	NA	+	+/+	18	NA	3 (broad based gait)	No speech	Intermittent loss of skills	Kharbanda et al. 2017 [30]
22	E6	c.835delC	p.Leu279Cysfs*26	Frameshift	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Grozeva et al. 2015 [31]
23	E7	c.925C>T	p.Gln309*	Nonsense	Male	4,5	NA	NA	NA	+	Hyperopia			18	NA	walking at 4.6 (short distances)	Started speaking at 4 years, but articulation was poor and it was hard to understand	Happy, Feeding difficulties (chewing, swallowing)	Ligt et al. 2012 [4], Tucci et al. 2014 [28]
24	E7	c.998dupA	p.Tyr333*	Nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Prasad et al. 2016 [32]
25	E7	c.998dupA	p.Tyr333Ter	Nonsense	Female	9	NA	2980	NA	NA	NA			14	NA	4,2 (still had difficulties)	Frst words age 4; more fluent speech age 6; said to be 3 years behind with verbal skills	Violent outburst, dystonia	Kharbanda et al. 2017 [30]
26	E7	c.999C> G	p.Tyr333Ter	Nonsense	Female	27 years	NA	2920	NA	+	NA	-	+/+	30	NA	4,5 (ataxic)	First words at 4.5 years; can speak in sentences but speech very unclear	Aggressive behaviour, Asthma; left clubfoot; increased dermatoglyphic whorls	Kharbanda et al. 2017 [30]
27	E7	c.999del	p.Tyr333*	Nonsense	Female	15 mo	NA	NA	NA	NA	NA	NA	+/NA	NA	NA	NA	NA	NA	Sun et al. 2019 [33]
28	E7	c.1016_1025delinsT	p.Thr339_Arg342delinsI	Frameshift	Female	3	NA	NA	NA	NA	TRD, strabismus, hyperopia, astigmatism	+	+/NA	NA	NA	NA	NA	GDD, feeding aversion, and poor weight gain, Type-1 Diabetes	Rossetti et al 2021 [22]
29	E7	c.1005delA	p.Lys335fs	Frameshift	Male	8	NA	NA	NA	NA	hyperopia, astigmatism, esotropia of the left eye, strabismus	+	+/NA	NA	NA	NA	Absent speech	Aggressive, murmur, GDD, chronic diarrhea, dysplastic+ bicuspid pulmonary valve	Rossetti et al 2021 [22]

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30	E7	c.1038_1044delGCTATCTinsGCT	p.Val349AlafsTer9	Frameshift	Female	11	NA	2700	NA	+	Strabismus, hypermetropia	+	+/+	NA	13,5	3,5 (ataxic)	Single words at age 5 years, talks in sentences at age 11 years	Stereotypes, aggressive behaviour, acquired teeth early from 3 months	Kharbanda et al. 2017 [30]
31	E7	c.1043_1044delCT	p.Ser348CysfsX4	Frameshift	Female	8	NA	NA	NA	NA	NA	+	NA	NA	NA	NA	NA	GDD, dystonia	Cordeiro et al. 2018 [34]
32	Intron 7	c.1081+1G>C	IVS6 Intron 7	Splice	Male	3,8	47	2720	>2SD	+	Normal	+	+/+	not yet	not yet	not yet	Unclear speech	dystonia; motor restlessness	Kuechler et al. 2015 [2]
33	E8	c.1104delT	p.His369Thrfs*2	Frameshift	NA	4	NA	NA	NA	NA	FEVR	+	NA	NA	NA	NA	NA	Autism	Sun et al. 2019 [33]
34	E8	c.1105_1109del	p.His369Aspfs*24	Frameshift	Male	22	NA	NA	NA	NA	Divergent strabismus	-	+/+	NA	NA	NA	NA	Dystonia	Ho et al. 2021 [24]
35	E8	c.1127G>A	p.Arg376His	Missense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	ASD	Krupp et al., 2017 [44]
36	E8	c.1163T>C	p.Leu388Pro	Missense	Female	6,8	48	2500	30	+	NA	+	+/+	13	18	2,5	Moderate, first word at 2 ½, 20 words at 4 years but not intelligible	Delayed bone age; swallowing issues	Kuechler et al. 2015 [2]
37	E9	c.1251_1252insACGTG	p.Cys419*	Nonsense	Male	15 mo	50	3060	33	NA	NA	+	+/+	not yet	not yet	not yet	Vocalized only a few syllables	Good eye contact, social smile	Kuechler et al. 2015 [2]
38	E9	c.1272_1275del	p.Ser425Thrfs*11	Frameshift	Female	29	NA	NA	NA	+	NA	+	+/+	24	3 years	8 (due to progressive spasticity now with support)	Started speaking first words between 9-10 years; now able to speak a few words	Aggression, automutilaton	Ligt et al. 2012 [4], Tucci et al. 2014 [28]
39	E9	c.1272_1275del	p.Ser425Thrfs*11	Frameshift	Female	3,25	48	2425	NA	+	Strabismus	-	+/+	NA	NA	not yet	Babbles now (aged 3 3/12 years) constantly and some words are understandable	very happy and friendly, but can show a low frustration tolerance, good eye contact	Kuechler et al. 2015 [2]
40	E9	c. 1344_1345 InsertionA	p.Arg449GlnfsTer24	Frameshift	Male	8	NA	NA	NA	NA	Strabismus	+	NA/+	NA	NA	8	First words at 3 years, at 8 years able to speak short sentences	Spastic displegia; dystonia, hyperflexia, dysphagia, hypospadias,	Jin et al. 2020 [5]
41	E9	c.1420C>T	p.Arg474*	Nonsense	Male	2	51.5	3830	NA	+	Strabismus	-	+/+	18	No	2	limited words	Sleeping and feeding problems, good eye contact, social	Kuechler et al. 2015 [2]
42	E9	c.1420C>T	p.Arg474*	Nonsense	NA	NA	NA	NA	NA	NA	Eye abnormality	NA	NA	NA	NA	NA	NA	NA	Retterer et al. 2016 [25]
43	E9	c.1420C> T	p.Arg474Ter	Nonsense	Female	13	NA	3118	NA	+	Strabismus	+	+/+	13	NA	3,5	First words at 4,5 years	Aggressive, ADHD, mouths objects	Kharbanda et al. 2017 [30]

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Exon	Variant	Amino acid change	Mutation type	Lenght (cm)			Weight (g)	OFC (cm)	Sitting (mo)					Crawling (mo)	Walking independetly (yrs)	Speaking			
44	E9	c.1420C>T	p.Arg474*	Nonsense	Female	5.25	49	2410	31	+	Strabismus	+	+/+	18	23	not yet	No words	Stereotypic outbursts, feeding problems	Kuechler et al. 2015 [2]
45	E9	c.1434_1435insC	p.Glu479Argfs*18	Frameshift	Male	8	NA	NA	NA	+	FEVR and divergent strabismus	+	-/+	NA	NA	NA	NA	NA	Panagiotou et al. 2017 [35]; Ho et al. 2021 [24]
46	E9	c.1512G>A	p.Trp504*	Nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Autism	O’Roak et al. 2017 [44]
47	E9	c.1543C>T	p.Arg515*	Nonsense	Female	51	NA	NA	NA	+	Optic atrophy	+	+/+	NA	NA	No	Not able to speak, but uses sign language	Progressive swallowing difficulties	Ligt et al. 2012 [4], Tucci et al. 2014 [28]
48	E10	c.1603C>T	p.Arg535Ter	Nonsense	Male	3,25	NA	3050	NA	+	Strabismus	+	+/+	8	NA	Unable to walk	Lots of noises but no words	Dystonia	Kharbanda et al. 2017 [30]
49	E10	c.1603C>T	p.Arg535Ter	Nonsense	Female	15 mo	NA	2350	NA	+	FEVR	+	+/-	NA	NA	NA	Can make some sounds	NA	Ke et al. 2020 [39]
50	E10	c.1603C>T	p.Arg535Ter	Nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Retterer et al. 2016 [25]
51	E10	c.1603C>T	p.Arg535Ter	Nonsense	Male	14	NA	3400	NA	+	NA	-	-/+	15	NA	not walking	Moderate; Single words at 14 years	Aggressive, truncal obesity; absent left testis; brachydactyly; achilles tendon contracture	Kharbanda et al. 2017 [30]
52	E10	c.1603C>T	p.Arg535*	Nonsense	Male	Died	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Dieat at 4 months; abnormal lung growth, pulmonary hypertension	Karolak et al. 2019 [42]
53	E10	c.1603C>T	p.Arg535*	Nonsense	Male	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Grozeva et al. 2015 [31]
54	E10	c.1603C>T	p.Arg535*	Nonsense	Female	15 mo	NA	2350	NA	NA	FEVR, strabismus	+	+/-	Not yet	Not yet	Not yet	Only sounds at 15 months	NA	Ke et al. 2020 [39]
55	E10	c.1612C>T	p.Gln538Ter	Nonsense	Female	4,5	NA	2345	NA	+	Strabismus	+	+/+	23	NA	2,5-3 years	First words at 3.4 years	Autism	Kharbanda et al. 2017 [30]
56	E10	c.1652C>T	p.Thr551Met	Missense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Autism	O’Roak et al. 2017 [44], Tucci et al. 2014 [28]
57	E10	c.1665dupG	p.Thr556fs	Frameshift	Male	4	NA	NA	NA	NA	Retinopathy and strabismus	+	+/+	NA	NA	NA	NA	GDD, ASD, spastic diplegia, aggressive, premature birth (24mo), choreiform movements and intention tremor	Rossetti et al. 2021 [22]
58	E10	c.1672C>T	p.Gln558*	Nonsense	Male	15 mo	NA	3600	NA	NA	FEVR	+	+/-	not yet	not yet	not yet	No language at 15 months	Mild thumb adduction	Li et al. 2017 [7]
59	E10	c.1677delA	p.Gln559Hisfs*11	Frameshift	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Multiple congenital anomalies	Retterer et al. 2016 [25]

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	Exon	Variant	Amino acid change	Mutation type			Lenght (cm)	Weight (g)	OFC (cm)					Sitting (mo)	Crawling (mo)	Walking independetly (yrs)	Speaking		
60	Intron 10	c.1683+1G>A	Splice mutation	Splice --> nonsense	Female	2,4	51	3630	34,5	+	Normal	+	+/-	not yet	not yet	not yet	Severe, no words; only syllables	At age 2 months she suffered from skin problems (diaper rash and later eczema), aggressive and emotional problems	Kuechler et al. 2015 [2]
61	E11	c.1723G>A	p.Gly575Arg	Missense	Female	11	NA	NA	NA	NA	FEVR, legal blind on the right eye, totla retinal detachment on left eye	+	+/+	NA	NA	NA	NA	Repetative behaviours of hand-wringling and nail-picking	Rossetti et al. 2021 [22]
62	E11	c.1738_1742delinsACA	p.Leu580Thrfs*28	Frameshift	NA	NA	NA	NA	NA	NA	FEVR	NA	NA	NA	NA	NA	NA	NA	Sun et al. 2019 [33]
63	E11	c.1759C>T	p.Arg587*	Nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Yoo et al. 2017 [40]
64	E11	c.1759C>T	p.Arg587*	Nonsense	Female	4	NA	NA	NA	NA	normal	+	+/+	NA	NA	NA	NA	Bicoronal craniosynostosis	Ho et al. 2021 [24]
65	E11	c.1801C>T	p.Gln601Ter	Nonsense	Male	6,2	NA	2098	NA	+	FEVR	+	+/NA	NA	NA	not yet	Says Mom and Dad with understanding, uses some Makaton signs, points to the body parts	Stereotypic and aggressive behaviour, single supernumerary upper incisor; bilateral orchidopexies	Kharbanda et al. 2017 [30]
66	E12			Frameshift	Male	13	NA	NA	NA	NA	normal	-	+/+	NA	NA	NA	NA	DMD, dystonia	Ho et al. 2021 [24]
67	E12	c.1867C>T	p.Gln623*	Nonsense	NA	4	NA	NA	NA	NA	Retinal detachment	NA	NA	NA	NA	NA	NA	NA	Sun et al. 2019 [33]
68	E12	c.1923dupA	p.Glu642Argfs*6	Frameshift	Male	8,5	50	2880	31	+	Strabismus, hyperopia	+	+/+	NA	14	8 years	Severe; few single words, gestures	Good social contact, aggressive behaviour, frequent infections, syringomyelia	Kuechler et al. 2015 [2]
69	E12	c.1925_1926del	p.Glu642Valfs*5	Frameshift	Female	14,2	48	3030	NA	+	Strabismus	-	-/+	not yet	NA	No	Moderate, first words at 6 years; not speak in sentences	Autism, short eye contact, tantrums, generalized pyramidal symptoms	Kuechler et al. 2015 [2]
70	E12	c.1943A>G	p.N648S	Missense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Severe paranoid schizophrenia	Levchenko et al. 2015 [27]
71	E13	c.1962T>A	p.Tyr654*	Nonsense	Male	2	NA	NA	NA	NA	Intermittent exotropia, mild hyperopia, strabismus	+	+/+	NA	NA	NA	NA	GDD, spastic diplegia	Rossetti et al 2021 [22]
72	E13	c.1981C>T	p.Arg661Ter	Nonsense	Female	9,2	NA	2530	NA	+	NA	+	+/-	11	NA	2,5	First words at 3,4 years	Obsessional behaviour; Dyspraxia; slightly delayed bone age;	Kharbanda et al. 2017 [30]

																		hypermobile joints; glue ear	
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73	E13	c.2038_2041 dup	p.Ser681*	Nonsense	Female	13,2	NA	2523	NA	+	Strabismus, myopia	-	+/+	12	NA	1,5	Mild, full sentences, but delayed	Social, autism, aggressive behaviour, ADHD, scoliosis and a tethered spinal cord	Kuechler et al. 2015 [2]
74	E13	c.2038_2041 dup	p.Ser681*	Nonsense	Female	11	NA	2891	NA	+	Strabismus, myopia	+	+/+	12	17	2	Mild, full sentences, but delayed	Communicative, social, aggression, ADHD	Kuechler et al. 2015 [2]
75	E13	c.2038_2041 dup	p.Ser681*	Nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Multiple congenital anomalies	Retterer et al. 2016 [25]
76	E13	c.2046_2047 del	p.Phe683Glnfs*9	Frameshift	Male	8 mo	NA	3146	NA	NA	Myopia	NA	NA	NA	NA	NA	NA	NA	Coussa et al. 2020 [36]
77	E14	c.2092_2096 dup	p.Ile700Leufs*37	Frameshift	Female	7	NA	NA	NA	NA	Bilateral FEVR, right convergent strabismus	+	-/+	NA	NA	NA	NA	NA	Ho et al. 2021 [24]
78	E14	c.2092_2096 dup	p.Ile700Leufs*37	Frameshift	Female	37	NA	NA	NA	NA	Normal	-	-/-	NA	NA	NA	Full sentences	NA	Ho et al. 2021 [24]
79	E14	c.2112_2116 dup	p.Pro706Glnfs*31	Frameshift	Male	22 mo	NA	1842	NA	NA	FEVR	NA	NA	NA	NA	NA	NA	ASD	Dixon et al. 2016 [37]
80	E14	c.2128C>T	p.Arg710Cys	Missense	inherited	NA	NA	NA	NA	normal	FEVR	NA	Normal	Normal	normal	normal	normal	NA	Panagiotou et al. 2017 [35]
81	E15	c.2142_2157 dup16	p.His720*	Nonsense	inherited	NA	NA	NA	NA	normal	FEVR	NA	Normal	Normal	normal	normal	normal	NA	Panagiotou et al. 2017 [35]
82	E15	c.2273del	p.His758Leufs*30	Frameshift	Male	14	NA	NA	NA	NA	NA	-	NA	NA	NA	NA	NA	Autism, severe ID	Thevenon et al. 2016 [38]
83	Entire gene	333 kb incl. entire gene & ex. 35-37 of ULK4	Gross del	None	Female	5,2	43,5	2100	30,5	+	Hyperopia	+	+/-	14	NA	4,5 years (ataxic)	At 4.5 years could combine several words, count to 10	Friendly, social, short focus, orofacial dyspraxia; sacral dimple	Dubruc et al. 2014 [41]
84	Entire gene	505 kb incl. entire gene	Gross del	None	Male	3	45,5	2107	29,5	+	Esotropia	+	+/-	NA	not yet	not yet	Babbles and say “mama” and “dada” specifically shortly before age 3 years	Happy, good eye contact, dystonic movement	Kuechler et al. 2015 [2]