Supplementary Information:

Genomic and phenotypic characterization of 404 individuals with neurodevelopmental disorders caused by CTNNB1 variants

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Methods and Materials:

Reverse transcription polymerase chain reaction (RT-PCR)

Relative amounts of endogenous and exogenous *CTNNB1* mRNA were quantified by RT-PCR. HEK293T cells transfected with β-catenin expression constructs and luciferase reporter plasmids were homogenized in TRIzol reagent (Invitrogen, Cat# 15596026) and RNA was isolated using RNeasy kit (QIAGEN, Cat#74104). Complementary DNA (cDNA) was synthesized using random hexamers and the SuperScript IV First-Strand Synthesis System (Invitrogen, Cat# 18091050). Total endogenous and exogenous *CTNNB1* mRNA was detected by primers targeting N-terminal *CTNNB1* cDNA sequence. Exogenous *CTNNB1* mRNA was detected by primers with sequences of a Myc tag or a V5 tag. Equal abundance of total cDNA among samples was confirmed by assessing expression of the housekeeping gene esterase D. Sequences of primers used in RT-PCR can be found in a Supplementary Table 12.

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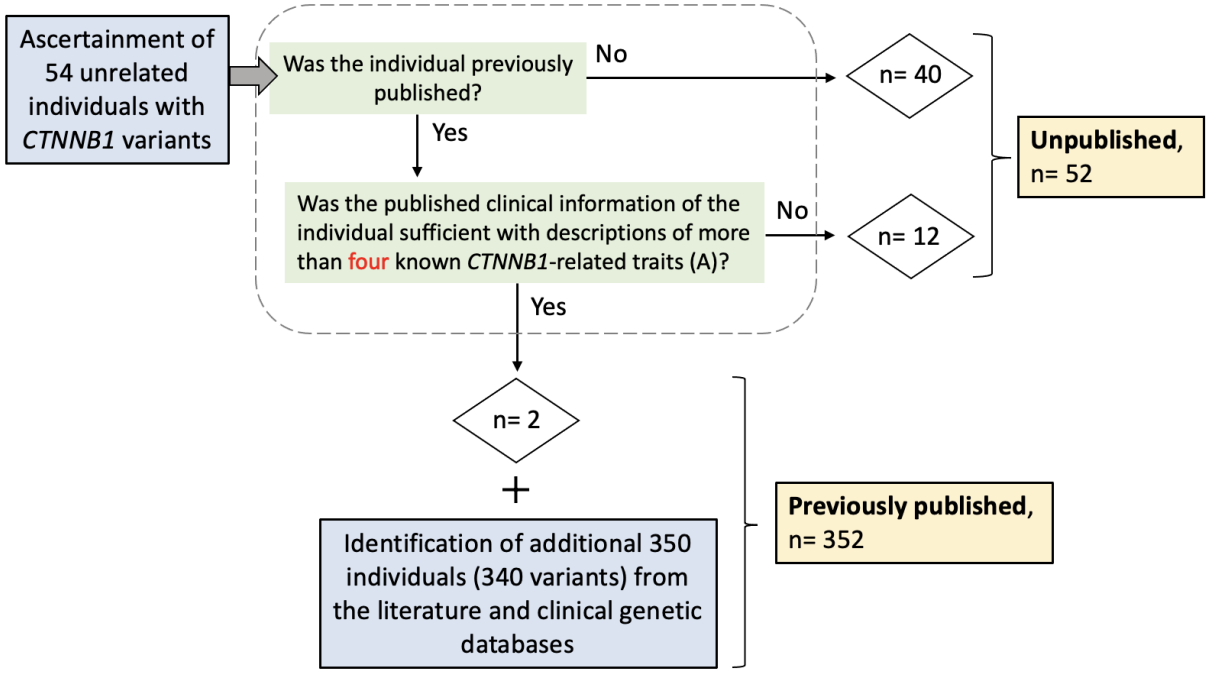
A

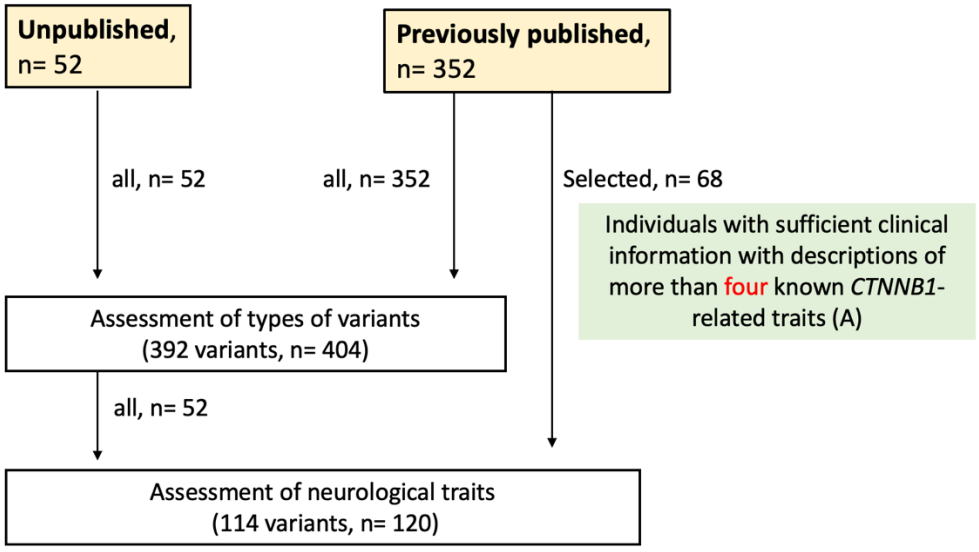
***CTNNB1*-related traits**

i) Cognition
ii) Speech
iii) Movement
iv) Morphology /physiology of the eye
v) Microcephaly
vi) Mild craniofacial dysmorphic features

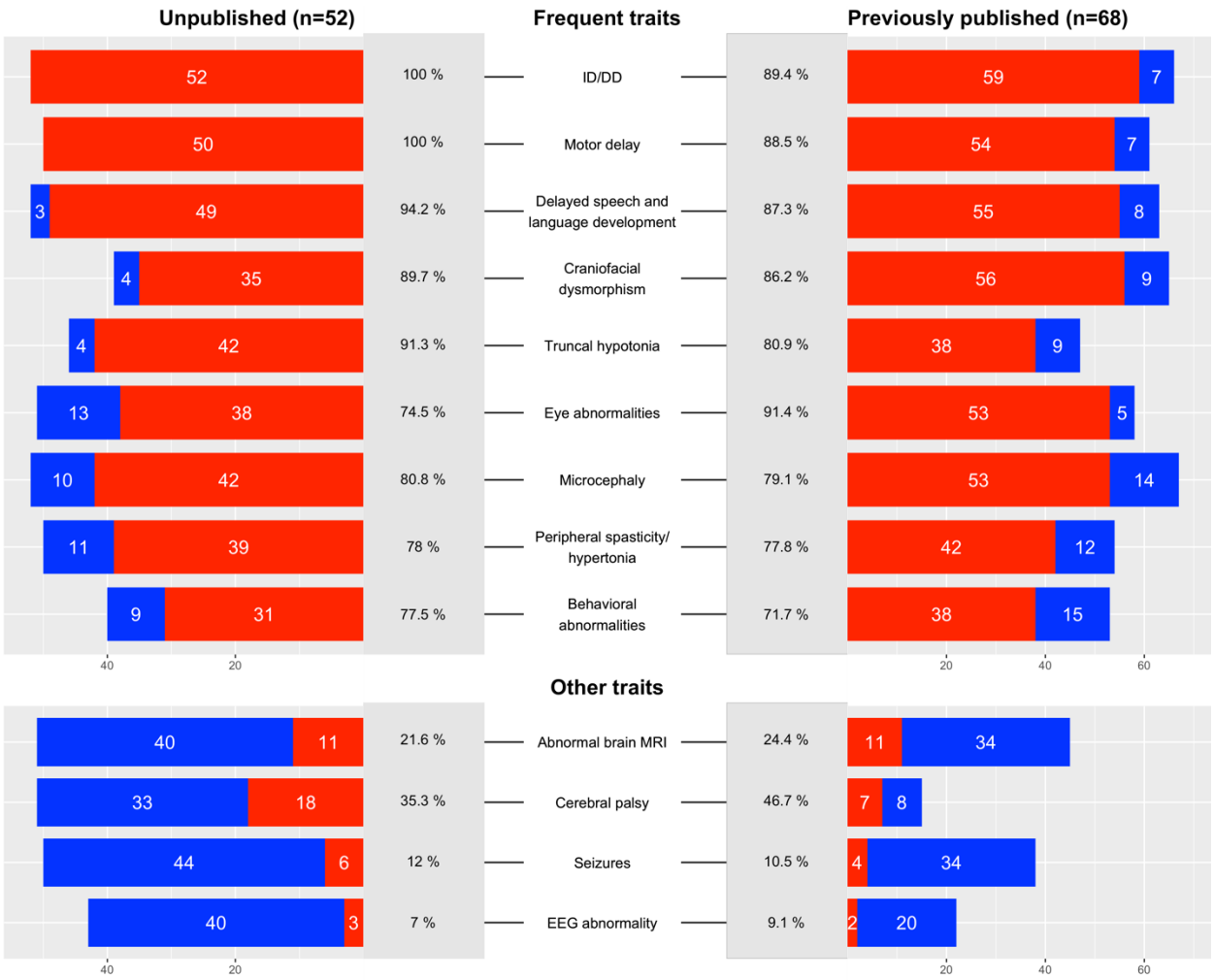
B



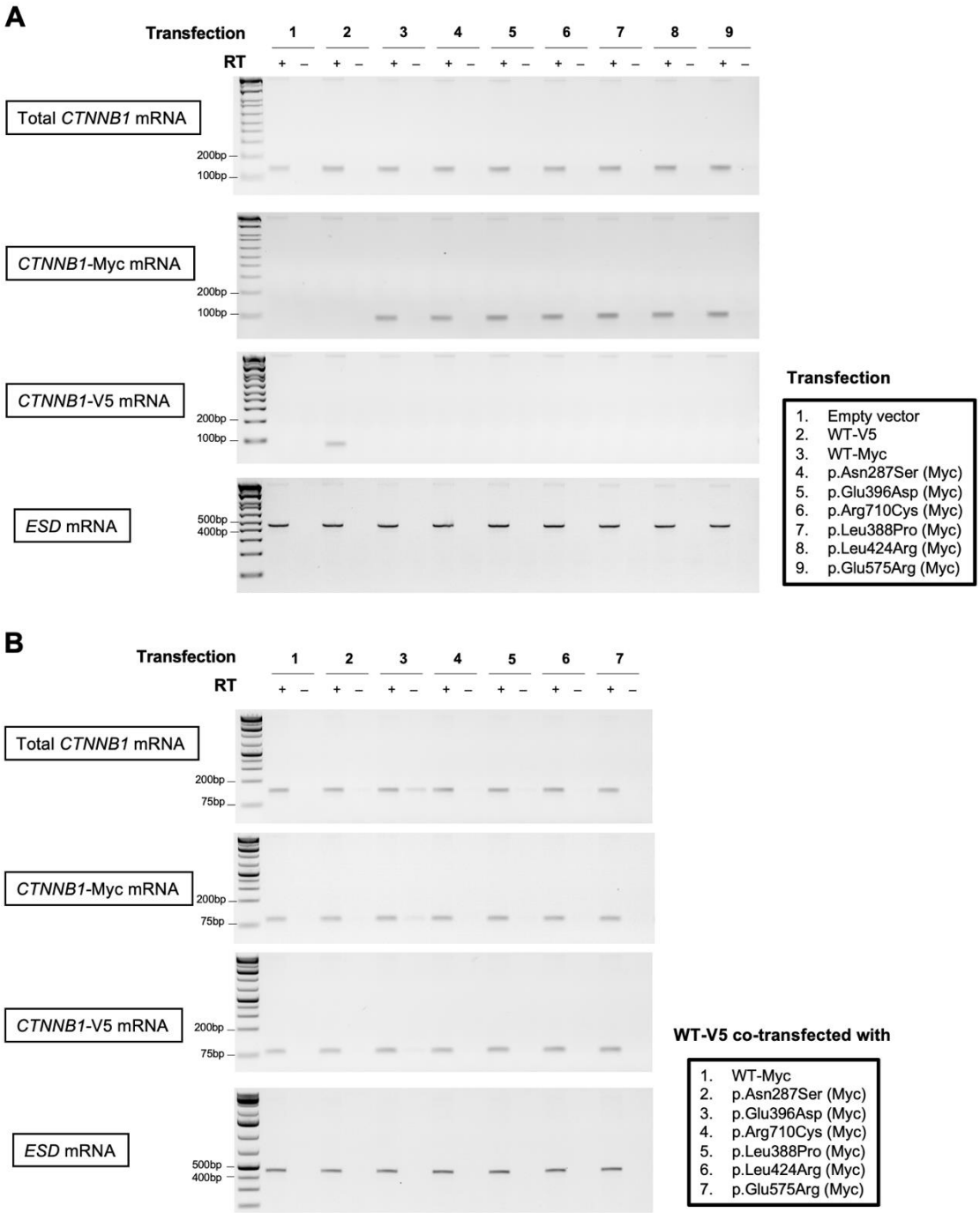
C



Supplementary Figure 1. Identification of 52 unpublished and 352 previously published individuals carrying *CTNNB1* deleterious variants and study design. (A) A summary of six *CTNNB1*-related traits that were defined from previous clinical reviews of individuals carrying pathogenic *CTNNB1* variants. (B) A flow chart shows how we identified 52 unpublished and 352 previously published individuals with *CTNNB1* deleterious variants. (C) A flow chart summarizes design of this study.

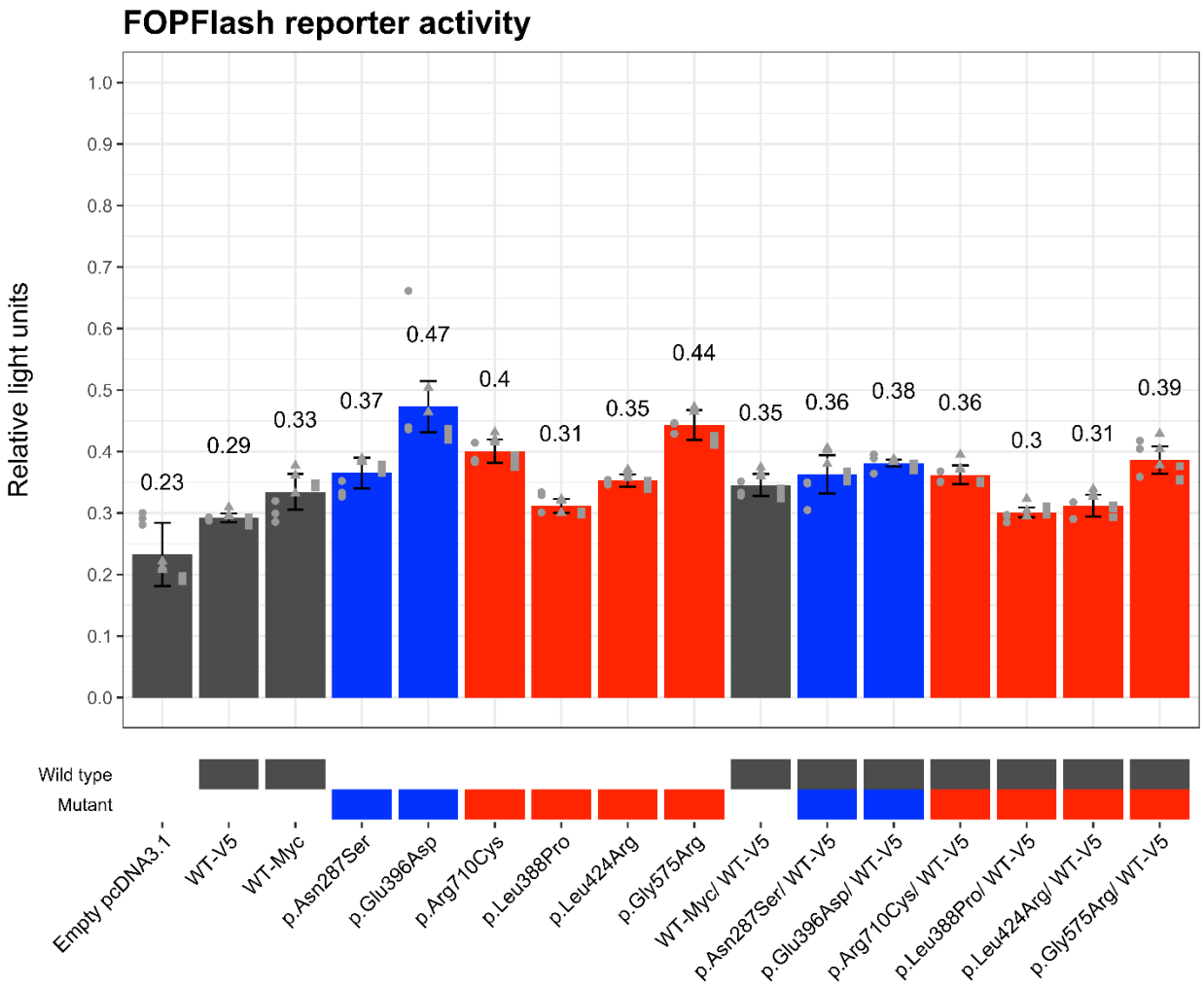


Supplementary Figure 2. Comparison of the frequency of traits between unpublished and previously published individuals. Neurological traits which were frequently identified in unpublished (left) and previously published (right) individuals are summarized at the top. Other relevant traits discussed in the present study are summarized at the bottom. Bar charts show the number of affected (red) and unaffected (blue) individuals per trait. The length of bars reflects the number of individuals with available information per trait. The percentages of affected individuals to total reported cases are shown in the grey boxes.

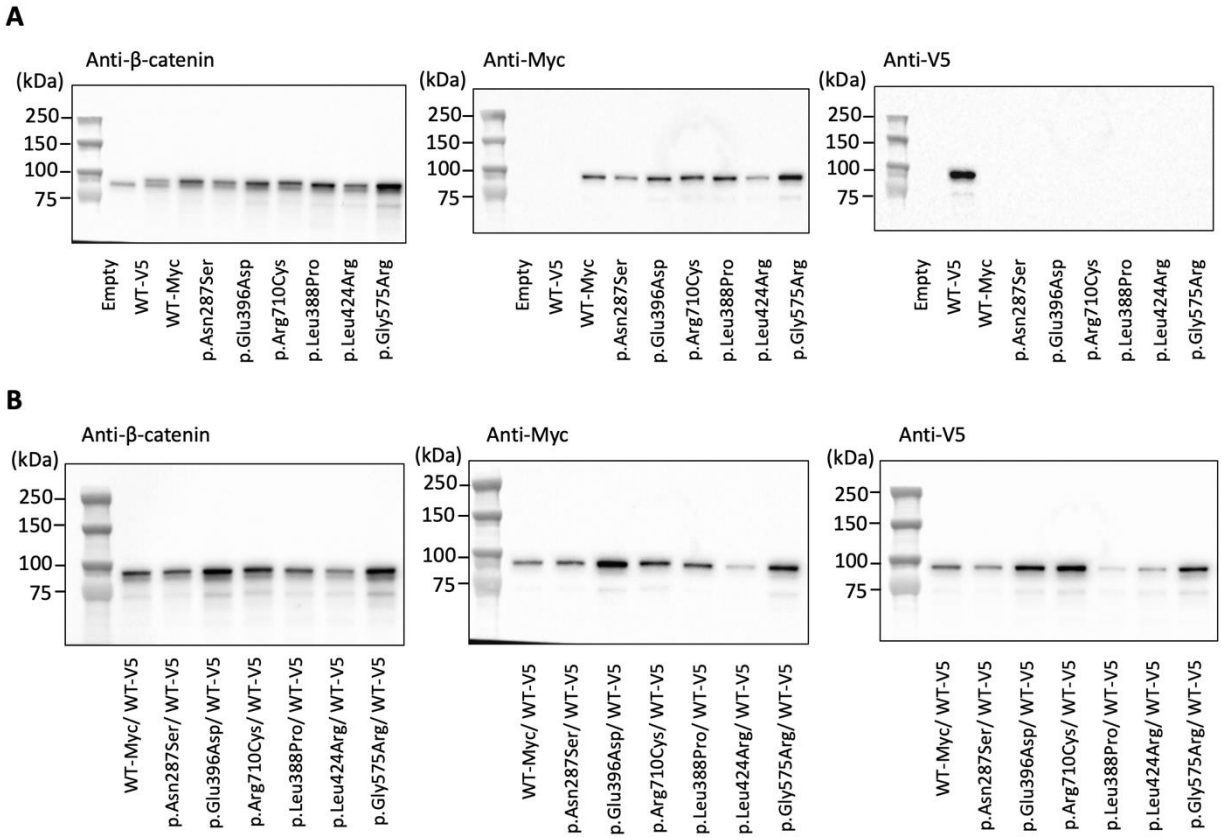


Supplementary Figure 3. Expression of Myc or V5-tagged wildtype β-catenin proteins and Myc-tagged mutant β-catenin proteins transfected into HEK293T cells detected by reverse transcription polymerase chain reaction (RT-PCR). Reverse transcription polymerase chain reaction (RT-PCR) was performed to detect mRNA expression of Myc or V5-tagged wildtype

(WT) β-catenin transcripts and Myc-tagged mutant β-catenin transcripts. Expression constructs were transfected without co-transfection (A) and with co-transfection of a V5-tagged wildtype β-catenin (B). Synthesis of complementary DNA (cDNA) from RNA was performed in the presence (+) or absence (-) of reverse transcriptase (RT). Total *CTNNB1* mRNA was detected with primers which amplify *CTNNB1* coding region (proximal to the N terminus). Exogenous *CTNNB1* mRNA (CTNNB1-Myc or CTNNB1-V5) were selectively amplified using a primer containing a Myc tag or a V5 tag sequence. Amplifications with primers specific to esterase D (ESD) cDNA were performed to confirm success of cDNA synthesis in samples treated with RT.



Supplementary Figure 4. CTNNB1 constructs had minimal effects on luciferase reporter gene expression in the absence of wild type LEF/TCF responsive elements. Relative luciferase activity measured from the FOPFlash reporter construct in HEK293T cells co-transfected with expression vectors for wildtype β-catenin or mutant β-catenin or an equal mix with wildtype β-catenin tagged with V5. Wild type, gnomAD variants, and pathogenic/likely pathogenic variants are highlighted in grey, blue, and red on the X-axis labels, respectively. Assay was performed in triplicate (shown with different shaped data points) with three technical replicate samples for each assay. Error bars indicate standard deviations between the three independent experiments. Results indicate a functional LEF/TCF binding site is required for transactivation.



Supplementary Figure 5. Full blots of western blots shown in Figure 5A and 5B in the main text. (A) Detection of wild-type (WT) and mutant β-catenin proteins transfected as solo into HEK293T cells by western blot, shown in Figure 5A in the main text. (B) Detection of wildtype and mutant β-catenin proteins co-transfected with a V5-tagged wildtype β-catenin into HEK293T cells by western blot, shown in Figure 5B in the main text. Endogenous and exogenous β-catenin were detected with a β-catenin antibody (Anti- β-catenin, left). Myc-tagged wild-type and mutant β-catenin were detected with an anti-Myc antibody (Anti-Myc, middle). A V5-tagged wildtype β-catenin was detected with an anti-V5 antibody (Anti-V5, right).

Supplementary Table 5: Summary of cloning strategies of *CTNNB1* variants tested in TOPFlash dual-luciferase reporter assay.

Variant / purpose	Primer	Sequences (5' -> 3')	Orientation	Primer pair for PCR	Annealing temperature (°C)	Cloning strategy
Overlap PCR	T7	TAATACGACTCACTATAGGG	Forward			
Overlap PCR	CTNNB1_c598 (F1)	CGTACCATGCAGAATACAAATG	Forward			
Overlap PCR	CTNNB1_c1123 (F2)	CAACGTCTTGTTCAGAACTG	Forward			
Overlap PCR	CTNNB1_c1973 (R1)	ACAGCAGCTGCATATGTCG	Reverse			
Overlap PCR	pcDNA_vector (R2)	CCATTGAGTTTAAACCCGCTG	Reverse			
Substitution of a C-terminal tag	CTNNB1_myc_R (sequence of a Myc tag highlighted in red.)	TGGTGATGATGACCGGTACGCAGATCCTCTTCTGAGATGAGTTTTTGTTCTTCGAAGGGCCCTCT	Reverse	F2	62.0	PCR product was purified and digested with AgeI (NEB# R3552S) and EcoRI (NEB# R3101S) prior to ligation into a vector digested with the same set of restriction enzymes.
NM_001904.3: c.860A>G: p.Asn287Ser (rs35288908)	Fragment 1	ATGGTTGCCTTGCTCAGCAAAACAAATGTTA	Forward	R1	72.0	Equal amount of fragment 1 and 2 were mixed in PCR reaction with outer primers (T7 x R1). Amplified product and a vector were digested with Bsu36I (NEB# R0524S) and EcoRI.
	Fragment 2	AACATTTGTTTTGCTGAGCAAGGCAACCATT	Reverse	T7	62.0	
NM_001904.3: c.1188A>C: p.Glu396Asp (rs751375496)	Fragment 1	CTGCAACTAAACAGGACGGGATGGAAGGTC	Forward	R1	72.0	
	Fragment 2	GAGACCTTCCATCCCGTCCTGTTTAGTTGC	Reverse	T7	62.0	
NM_001904.3:	Fragment 1	TGGACTCTCAGGAATCCTTCAGATGCTGCAAC	Forward	R1	72.0	Equal amount of fragment 1 and 2 were mixed in PCR

Variant / purpose	Primer	Sequences (5' -> 3')	Orientation	Primer pair for PCR	Annealing temperature (°C)	Cloning strategy
c.1163T>C: p.Leu388Pro (rs1559474140)	Fragment 2	TAGTTGCAGCATCTGAAGGATTCCTGAGAGTC	Reverse	T7	66.7	reaction with outer primers (T7 x R1). Amplified product was digested with BamHI (R3136S) and EcoRI and cloned into a pcDNA3.1-CTNNB1-V5 vector digested with the same set of enzymes. For this variant only, the V5 tag was replaced with the Myc tag after insertion of CTNNB1 variant of interest.
NM_001904.3: c.1271T>G: p.Leu424Arg (rs863224864)	Fragment 1	CTGTGCAGCTGGAATTCGTTCTAACCTCACTTG	Forward	R2	72.0	Equal amount of fragment 1 and 2 were mixed in PCR reaction with outer primers (F1 x R2). Amplified product and a vector were digested with AgeI and EcoRI.
	Fragment 2	AGTGAGGTTAGAACGAATTCCAGCTGCACAGGT	Reverse	F1	62.0	
NM_001904.3: c.1723G>A: p.Gly575Arg (rs797044875)	Fragment 1	GTTGAAGGTTGTACCAGAGCCCTTCACATC	Forward	R2	62.0	Equal amount of fragment 1 and 2 were mixed in PCR reaction with outer primers (F2 x R2). Amplified product and a vector were digested with AgeI and EcoRI.
	Fragment 2	TGTGAAGGGCTCTGGTACAACCTTCAACTA	Reverse	F2	62.0	
NM_001904.3: c.2128C>T: p.Arg710Cys (rs748653573)	Fragment 1	AACCCCTTGGATATTGCCAGGATGATCC	Forward	R2	72.0	
	Fragment 2	TAGGATCATCCTGGCAATATCCAAGGGGTTC	Reverse	F2	62.0	

"c" in name of primers refers to a position at CTNNB1 cDNA coordinate (NM_001904.3).

Supplementary Table 11: Ratio of different types of *CTNNB1* variants identified in population and neurodevelopmental disorders (NDD).

Variant type	**Population (gnomAD)**		**NDD**	
	Count	Ratio (%)	Count	Ratio (%)
Synonymous	14826	90.96	0	0.00
Missense	612	3.75	28	7.14
Stopgain	0	0.00	176	44.90
Frameshift	0	0.00	141	35.97
In-frame	5	0.03	1	0.26
Splicing	2	0.01	32	8.16
Splice site	854	5.24	6	1.53
Structural variants	0	0.00	8	2.04
Start lost	1	0.01	0	0.00
Total (n)	16300		392	

Supplementary Table 12: Primer sequences designed for RT-PCR.

Target	Primer orientation	Sequence (5’ -> 3’)
ESD (housekeeping)	Forward	GGAGCTTCCCCAACTCATAAATGCC
	Reverse	GCATGATGTCTGATGTGGTCAGTAA
Total CTNNB1 mRNA	Forward	CAGAGTGCTGAAGGTGCTATC
	Reverse	ATTCCTGAGAGTCCAAAGACAG
CTNNB1-Myc mRNA	Forward	CAATCAGCTGGCCTGGTTTG
	Reverse	GATGATGACCGGTACGCAGA
CTNNB1-V5 mRNA	Forward	CAATCAGCTGGCCTGGTTTG
	Reverse	TCGAGACCGAGGAGAGGGTTA

Supplementary Table S1

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Supplementary Table S2

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Supplementary Table S13

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Input Errors and warnings AccNo Genesymbol Variant Reference Sequence Start Descr. Coding DNA Descr. Protein Descr. GeneSymbol Coding DNA Descr. GeneSymbol Protein Descr. Genomic ReferenceCoding Reference Protein ReferenceAffected Transcripts Affected Proteins Restriction Sites Created Restriction Sites Deleted

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NC_000003.11(CTNNB1_v001):c.594del|NC_000003.11(CTNNB1_v002):c.594del|NC_000003.11(CTNNB1_v003):c.594del|NC_000003.11(CTNNB1_v004):c.594del|NC_000003.11(CTNNB1_v005):c.594del|NC_000003.11(CTNNB1_v006):c.594del|NC_000003.11(CTNNB1_v007):c.573del
NC_000003.11(CTNNB1_i001):p.(Ile198Metfs*11)|NC_000003.11(CTNNB1_i002):p.(Ile198Metfs*11)|NC_000003.11(CTNNB1_i003):p.(Ile198Metfs*11)|NC_000003.11(CTNNB1_i004):p.(Ile198Metfs*11)|NC_000003.11(CTNNB1_i005):p.(Ile198Metfs*11)|NC_000003.11(CTNNB1_i006):p.(Ile198Metfs*11)|NC_000003.11(CTNNB1_i007):p.(Ile191Metfs*11)
NC_000003.11:g.41267064G>A (GenRecord): Mutation on splice site in gene CTNNB1 transcript 001.|(GenRecord): Mutation on splice site in gene CTNNB1 transcript 002.|(GenRecord): Mutation on splice site in gene CTNNB1 transcript 003.|(GenRecord): Mutation on splice site in gene CTNNB1 transcript 004.|(GenRecord): Mutation on splice site in gene CTNNB1 transcript 005.|(GenRecord): Mutation on splice site in gene CTNNB1 transcript 006.|(GenRecord): Mutation on splice site in gene CTNNB1 transcript 007.

NC_000003.11(CTNNB1_v001):c.734+1G>A|NC_000003.11(CTNNB1_v002):c.734+1G>A|NC_000003.11(CTNNB1_v003):c.734+1G>A|NC_000003.11(CTNNB1_v004):c.734+1G>A|NC_000003.11(CTNNB1_v005):c.734+1G>A|NC_000003.11(CTNNB1_v006):c.734+1G>A|NC_000003.11(CTNNB1_v007):c.713+1G>A
NC_000003.11(CTNNB1_i001):p.(=)|NC_000003.11(CTNNB1_i002):p.(=)|NC_000003.11(CTNNB1_i003):p.(=)|NC_000003.11(CTNNB1_i004):p.(=)|NC_000003.11(CTNNB1_i005):p.(=)|NC_000003.11(CTNNB1_i006):p.(=)|NC_000003.11(CTNNB1_i007):p.(=)
NC_000003.11:g.41267215G>T

NC_000003.11(CTNNB1_v001):c.799G>T|NC_000003.11(CTNNB1_v002):c.799G>T|NC_000003.11(CTNNB1_v003):c.799G>T|NC_000003.11(CTNNB1_v004):c.799G>T|NC_000003.11(CTNNB1_v005):c.799G>T|NC_000003.11(CTNNB1_v006):c.799G>T|NC_000003.11(CTNNB1_v007):c.778G>T
NC_000003.11(CTNNB1_i001):p.(Glu267*)|NC_000003.11(CTNNB1_i002):p.(Glu267*)|NC_000003.11(CTNNB1_i003):p.(Glu267*)|NC_000003.11(CTNNB1_i004):p.(Glu267*)|NC_000003.11(CTNNB1_i005):p.(Glu267*)|NC_000003.11(CTNNB1_i006):p.(Glu267*)|NC_000003.11(CTNNB1_i007):p.(Glu260*)
Hpy188III,HpyAV
NC_000003.11:g.41268760dupA

NC_000003.11(CTNNB1_v001):c.998dup|NC_000003.11(CTNNB1_v002):c.998dup|NC_000003.11(CTNNB1_v003):c.998dup|NC_000003.11(CTNNB1_v004):c.998dup|

NC_000003.11(CTNNB1_v005):c.998dup|NC_000003.11(CTNNB1_v006):c.998dup|NC_000003.11(CTNNB1_v007):c.977dup

NC_000003.11(CTNNB1_i001):p.(Tyr333*)|NC_000003.11(CTNNB1_i002):p.(Tyr333*)|NC_000003.11(CTNNB1_i003):p.(Tyr333*)|NC_000003.11(CTNNB1_i004):p.(Tyr333*)|NC_000003.11(CTNNB1_i005):p.(Tyr333*)|NC_000003.11(CTNNB1_i006):p.(Tyr333*)|NC_000003.11(CTNNB1_i007):p.(Tyr326*)

MseI

NC_000003.11:g.41268761C>G

NC_000003.11(CTNNB1_v001):c.999C>G|NC_000003.11(CTNNB1_v002):c.999C>G|NC_000003.11(CTNNB1_v003):c.999C>G|NC_000003.11(CTNNB1_v004):c.999C>G|NC_000003.11(CTNNB1_v005):c.999C>G|NC_000003.11(CTNNB1_v006):c.999C>G|NC_000003.11(CTNNB1_v007):c.978C>G

NC_000003.11(CTNNB1_i001):p.(Tyr333*)|NC_000003.11(CTNNB1_i002):p.(Tyr333*)|NC_000003.11(CTNNB1_i003):p.(Tyr333*)|NC_000003.11(CTNNB1_i004):p.(Tyr333*)|NC_000003.11(CTNNB1_i005):p.(Tyr333*)|NC_000003.11(CTNNB1_i006):p.(Tyr333*)|NC_000003.11(CTNNB1_i007):p.(Tyr326*)

DdeI

NC_000003.11:g.41268824_41268833delins18

(grammar): Expected end of text (at char 35), (line:1, col:36)

NC_000003.11:g.41274840C>T

NC_000003.11(CTNNB1_v001):c.1090C>T|NC_000003.11(CTNNB1_v002):c.1090C>T|NC_000003.11(CTNNB1_v003):c.1090C>T|NC_000003.11(CTNNB1_v004):c.1090C>T|NC_000003.11(CTNNB1_v005):c.1090C>T|NC_000003.11(CTNNB1_v006):c.1090C>T|NC_000003.11(CTNNB1_v007):c.1069C>T

NC_000003.11(CTNNB1_i001):p.(Gln364*)|NC_000003.11(CTNNB1_i002):p.(Gln364*)|NC_000003.11(CTNNB1_i003):p.(Gln364*)|NC_000003.11(CTNNB1_i004):p.(Gln364*)|NC_000003.11(CTNNB1_i005):p.(Gln364*)|NC_000003.11(CTNNB1_i006):p.(Gln364*)|NC_000003.11(CTNNB1_i007):p.(Gln357*)

BsmI,Cac8I,HpyCH4V

NC_000003.11:g.41275095del

NC_000003.11(CTNNB1_v001):c.1261del|NC_000003.11(CTNNB1_v002):c.1261del|NC_000003.11(CTNNB1_v003):c.1261del|NC_000003.11(CTNNB1_v004):c.1261del|NC_000003.11(CTNNB1_v005):c.1261del|NC_000003.11(CTNNB1_v006):c.1261del|NC_000003.11(CTNNB1_v007):c.1240del

NC_000003.11(CTNNB1_i001):p.(Ala421Leufs*16)|NC_000003.11(CTNNB1_i002):p.(Ala421Leufs*16)|NC_000003.11(CTNNB1_i003):p.(Ala421Leufs*16)|NC_000003.11(CTNNB1_i004):p.(Ala421Leufs*16)|NC_000003.11(CTNNB1_i005):p.(Ala421Leufs*16)|NC_000003.11(CTNNB1_i006):p.(Ala421Leufs*16)|NC_000003.11(CTNNB1_i007):p.(Ala414Leufs*16)

ApaLI,BaeGI,BsiHKAI,Bsp1286I,BsrI,BtsIMutI,Hpy166II,TspRI

AluI,ApeKI,BbvI,BsgI,CviKI_1,Fnu4HI,MspA1I,PvuII,TseI

NC_000003.11:g.41275137A>T

NC_000003.11(CTNNB1_v001):c.1303A>T|NC_000003.11(CTNNB1_v002):c.1303A>T|NC_000003.11(CTNNB1_v003):c.1303A>T|NC_000003.11(CTNNB1_v004):c.1303A>T|NC_000003.11(CTNNB1_v005):c.1303A>T|NC_000003.11(CTNNB1_v006):c.1303A>T|NC_000003.11(CTNNB1_v007):c.1282A>T

NC_000003.11(CTNNB1_i001):p.(Lys435*)|NC_000003.11(CTNNB1_i002):p.(Lys435*)|NC_000003.11(CTNNB1_i003):p.(Lys435*)|NC_000003.11(CTNNB1_i004):p.(Lys435*)|NC_000003.11(CTNNB1_i005):p.(Lys435*)|NC_000003.11(CTNNB1_i006):p.(Lys435*)|NC_000003.11(CTNNB1_i007):p.(Lys428*)

BfaI

NC_000003.11:g.41275190_41275191insA

NC_000003.11(CTNNB1_v001):c.1356_1357insA|NC_000003.11(CTNNB1_v002):c.1356_1357insA|NC_000003.11(CTNNB1_v003):c.1356_1357insA|NC_000003.11(CTNNB1_v004):c.1356_1357insA|NC_000003.11(CTNNB1_v005):c.1356_1357insA|NC_000003.11(CTNNB1_v006):c.1356_1357insA|NC_000003.11(CTNNB1_v007):c.1335_1336insA

NC_000003.11(CTNNB1_i001):p.(Arg453Thrfs*4)|NC_000003.11(CTNNB1_i002):p.(Arg453Thrfs*4)|NC_000003.11(CTNNB1_i003):p.(Arg453Thrfs*4)|NC_000003.11(CTNNB1_i004):p.(Arg453Thrfs*4)|NC_000003.11(CTNNB1_i005):p.(Arg453Thrfs*4)|NC_000003.11(CTNNB1_i006):p.(Arg453Thrfs*4)|NC_000003.11(CTNNB1_i007):p.(Arg446Thrfs*4) HpyAV

NC_000003.11:g.41275208dup

NC_000003.11(CTNNB1_v001):c.1374dup|NC_000003.11(CTNNB1_v002):c.1374dup|NC_000003.11(CTNNB1_v003):c.1374dup|NC_000003.11(CTNNB1_v004):c.1374dup|NC_000003.11(CTNNB1_v005):c.1374dup|NC_000003.11(CTNNB1_v006):c.1374dup|NC_000003.11(CTNNB1_v007):c.1353dup

NC_000003.11(CTNNB1_i001):p.(Asp459Argfs*4)|NC_000003.11(CTNNB1_i002):p.(Asp459Argfs*4)|NC_000003.11(CTNNB1_i003):p.(Asp459Argfs*4)|NC_000003.11(CTNNB1_i004):p.(Asp459Argfs*4)|NC_000003.11(CTNNB1_i005):p.(Asp459Argfs*4)|NC_000003.11(CTNNB1_i006):p.(Asp459Argfs*4)|NC_000003.11(CTNNB1_i007):p.(Asp452Argfs*4) BbsI,MboII

NC_000003.11:g.41275254C>T

NC_000003.11(CTNNB1_v001):c.1420C>T|NC_000003.11(CTNNB1_v002):c.1420C>T|NC_000003.11(CTNNB1_v003):c.1420C>T|NC_000003.11(CTNNB1_v004):c.1420C>T|NC_000003.11(CTNNB1_v005):c.1420C>T|NC_000003.11(CTNNB1_v006):c.1420C>T|NC_000003.11(CTNNB1_v007):c.1399C>T

NC_000003.11(CTNNB1_i001):p.(Arg474*)|NC_000003.11(CTNNB1_i002):p.(Arg474*)|NC_000003.11(CTNNB1_i003):p.(Arg474*)|NC_000003.11(CTNNB1_i004):p.(Arg474*)|NC_000003.11(CTNNB1_i005):p.(Arg474*)|NC_000003.11(CTNNB1_i006):p.(Arg474*)|NC_000003.11(CTNNB1_i007):p.(Arg467*)

AluI,MspA1I,PvuII

NC_000003.11:g.41275303_41275322del

NC_000003.11(CTNNB1_v001):c.1469_1488del|NC_000003.11(CTNNB1_v002):c.1469_1488del|NC_000003.11(CTNNB1_v003):c.1469_1488del|NC_000003.11(CTNNB1_v004):c.1469_1488del|NC_000003.11(CTNNB1_v005):c.1469_1488del|NC_000003.11(CTNNB1_v006):c.1469_1488del|NC_000003.11(CTNNB1_v007):c.1448_1467del

NC_000003.11(CTNNB1_i001):p.(Gly490Alafs*33)|NC_000003.11(CTNNB1_i002):p.(Gly490Alafs*33)|NC_000003.11(CTNNB1_i003):p.(Gly490Alafs*33)|NC_000003.11(CTNNB1_i004):p.(Gly490Alafs*33)|NC_000003.11(CTNNB1_i005):p.(Gly490Alafs*33)|NC_000003.11(CTNNB1_i006):p.(Gly490Alafs*33)|NC_000003.11(CTNNB1_i007):p.(Gly483Alafs*33) AluI,BsrI,CviKI_1,MseI

NC_000003.11:g.41275660del

NC_000003.11(CTNNB1_v001):c.1555del|NC_000003.11(CTNNB1_v002):c.1555del|NC_000003.11(CTNNB1_v003):c.1555del|NC_000003.11(CTNNB1_v004):c.1555del|NC_000003.11(CTNNB1_v005):c.1555del|NC_000003.11(CTNNB1_v006):c.1555del|NC_000003.11(CTNNB1_v007):c.1534del

NC_000003.11(CTNNB1_i001):p.(Leu519Phefs*18)|NC_000003.11(CTNNB1_i002):p.(Leu519Phefs*18)|NC_000003.11(CTNNB1_i003):p.(Leu519Phefs*18)|NC_000003.11(CTNNB1_i004):p.(Leu519Phefs*18)|NC_000003.11(CTNNB1_i005):p.(Leu519Phefs*18)|NC_000003.11(CTNNB1_i006):p.(Leu519Phefs*18)|NC_000003.11(CTNNB1_i007):p.(Leu512Phefs*18)

NC_000003.11:g.41275685T>A

NC_000003.11(CTNNB1_v001):c.1580T>A|NC_000003.11(CTNNB1_v002):c.1580T>A|NC_000003.11(CTNNB1_v003):c.1580T>A|NC_000003.11(CTNNB1_v004):c.1580T>A|NC_000003.11(CTNNB1_v005):c.1580T>A|NC_000003.11(CTNNB1_v006):c.1580T>A|NC_000003.11(CTNNB1_v007):c.1559T>A

NC_000003.11(CTNNB1_i001):p.(Leu527*)|NC_000003.11(CTNNB1_i002):p.(Leu527*)|NC_000003.11(CTNNB1_i003):p.(Leu527*)|NC_000003.11(CTNNB1_i004):p.(Leu527*)|NC_000003.11(CTNNB1_i005):p.(Leu527*)|NC_000003.11(CTNNB1_i006):p.(Leu527*)|NC_000003.11(CTNNB1_i007):p.(Leu520*) Bpu10I,DdeI

NC_000003.11:g.41275690G>T

NC_000003.11(CTNNB1_v001):c.1585G>T|NC_000003.11(CTNNB1_v002):c.1585G>T|NC_000003.11(CTNNB1_v003):c.1585G>T|NC_000003.11(CTNNB1_v004):c.1585G>T|NC_000003.11(CTNNB1_v005):c.1585G>T|NC_000003.11(CTNNB1_v006):c.1585G>T|NC_000003.11(CTNNB1_v007):c.1564G>T

NC_000003.11(CTNNB1_i001):p.(Glu529*)|NC_000003.11(CTNNB1_i002):p.(Glu529*)|NC_000003.11(CTNNB1_i003):p.(Glu529*)|NC_000003.11(CTNNB1_i004):p.(Glu529*)|NC_000003.11(CTNNB1_i005):p.(Glu529*)|NC_000003.11(CTNNB1_i006):p.(Glu529*)|NC_000003.11(CTNNB1_i007):p.(Glu522*)

NC_000003.11:g.41275708C>T

NC_000003.11(CTNNB1_v001):c.1603C>T|NC_000003.11(CTNNB1_v002):c.1603C>T|NC_000003.11(CTNNB1_v003):c.1603C>T|NC_000003.11(CTNNB1_v004):c.1603C>T|NC_000003.11(CTNNB1_v005):c.1603C>T|NC_000003.11(CTNNB1_v006):c.1603C>T|NC_000003.11(CTNNB1_v007):c.1582C>T

NC_000003.11(CTNNB1_i001):p.(Arg535*)|NC_000003.11(CTNNB1_i002):p.(Arg535*)|NC_000003.11(CTNNB1_i003):p.(Arg535*)|NC_000003.11(CTNNB1_i004):p.(Arg535*)|NC_000003.11(CTNNB1_i005):p.(Arg535*)|NC_000003.11(CTNNB1_i006):p.(Arg535*)|NC_000003.11(CTNNB1_i007):p.(Arg528*)

CviAII,FatI,NlaIII

NC_000003.11:g.41277221delG

NC_000003.11(CTNNB1_v001):c.1690del|NC_000003.11(CTNNB1_v002):c.1690del|NC_000003.11(CTNNB1_v003):c.1690del|NC_000003.11(CTNNB1_v004):c.1690del|NC_000003.11(CTNNB1_v005):c.1690del|NC_000003.11(CTNNB1_v006):c.1690del|NC_000003.11(CTNNB1_v007):c.1669del

NC_000003.11(CTNNB1_i001):p.(Val564Serfs*6)|NC_000003.11(CTNNB1_i002):p.(Val564Serfs*6)|NC_000003.11(CTNNB1_i003):p.(Val564Serfs*6)|NC_000003.11(CTNNB1_i004):p.(Val564Serfs*6)|NC_000003.11(CTNNB1_i005):p.(Val564Serfs*6)|NC_000003.11(CTNNB1_i006):p.(Val564Serfs*6)|NC_000003.11(CTNNB1_i007):p.(Val557Serfs*6)

NC_000003.11:g.41277254G>A

NC_000003.11(CTNNB1_v001):c.1723G>A|NC_000003.11(CTNNB1_v002):c.1723G>A|NC_000003.11(CTNNB1_v003):c.1723G>A|NC_000003.11(CTNNB1_v004):c.1723G>A|NC_000003.11(CTNNB1_v005):c.1723G>A|NC_000003.11(CTNNB1_v006):c.1723G>A|NC_000003.11(CTNNB1_v007):c.1702G>A

NC_000003.11(CTNNB1_i001):p.(Gly575Arg)|NC_000003.11(CTNNB1_i002):p.(Gly575Arg)|NC_000003.11(CTNNB1_i003):p.(Gly575Arg)|NC_000003.11(CTNNB1_i004):p.(Gly575Arg)|NC_000003.11(CTNNB1_i005):p.(Gly575Arg)|NC_000003.11(CTNNB1_i006):p.(Gly575Arg)|NC_000003.11(CTNNB1_i007):p.(Gly568Arg)

BsaWI,HpaII,MspI,NlaIV

NC_000003.11:g.41277290C>T

NC_000003.11(CTNNB1_v001):c.1759C>T|NC_000003.11(CTNNB1_v002):c.1759C>T|NC_000003.11(CTNNB1_v003):c.1759C>T|NC_000003.11(CTNNB1_v004):c.1759C>T|NC_000003.11(CTNNB1_v005):c.1759C>T|NC_000003.11(CTNNB1_v006):c.1759C>T|NC_000003.11(CTNNB1_v007):c.1738C>T
NC_000003.11(CTNNB1_i001):p.(Arg587*)|NC_000003.11(CTNNB1_i002):p.(Arg587*)|NC_000003.11(CTNNB1_i003):p.(Arg587*)|NC_000003.11(CTNNB1_i004):p.(Arg587*)|NC_000003.11(CTNNB1_i005):p.(Arg587*)|NC_000003.11(CTNNB1_i006):p.(Arg587*)|NC_000003.11(CTNNB1_i007):p.(Arg580*)
NC_000003.11:g.41277903C>T

NC_000003.11(CTNNB1_v001):c.1867C>T|NC_000003.11(CTNNB1_v002):c.1867C>T|NC_000003.11(CTNNB1_v003):c.1867C>T|NC_000003.11(CTNNB1_v004):c.1867C>T|NC_000003.11(CTNNB1_v005):c.1867C>T|NC_000003.11(CTNNB1_v006):c.1867C>T|NC_000003.11(CTNNB1_v007):c.1846C>T
NC_000003.11(CTNNB1_i001):p.(Gln623*)|NC_000003.11(CTNNB1_i002):p.(Gln623*)|NC_000003.11(CTNNB1_i003):p.(Gln623*)|NC_000003.11(CTNNB1_i004):p.(Gln623*)|NC_000003.11(CTNNB1_i005):p.(Gln623*)|NC_000003.11(CTNNB1_i006):p.(Gln623*)|NC_000003.11(CTNNB1_i007):p.(Gln616*)
BspCNI,Hpy188III,LpnPI
NC_000003.11:g.41277944dupC

NC_000003.11(CTNNB1_v001):c.1908dup|NC_000003.11(CTNNB1_v002):c.1908dup|NC_000003.11(CTNNB1_v003):c.1908dup|NC_000003.11(CTNNB1_v004):c.1908dup|NC_000003.11(CTNNB1_v005):c.1908dup|NC_000003.11(CTNNB1_v006):c.1908dup|NC_000003.11(CTNNB1_v007):c.1887dup
NC_000003.11(CTNNB1_i001):p.(Thr637Hisfs*11)|NC_000003.11(CTNNB1_i002):p.(Thr637Hisfs*11)|NC_000003.11(CTNNB1_i003):p.(Thr637Hisfs*11)|NC_000003.11(CTNNB1_i004):p.(Thr637Hisfs*11)|NC_000003.11(CTNNB1_i005):p.(Thr637Hisfs*11)|NC_000003.11(CTNNB1_i006):p.(Thr637Hisfs*11)|NC_000003.11(CTNNB1_i007):p.(Thr630Hisfs*11) BanII,Bsp1286I
NC_000003.11:g.41277959dupA

NC_000003.11(CTNNB1_v001):c.1923dup|NC_000003.11(CTNNB1_v002):c.1923dup|NC_000003.11(CTNNB1_v003):c.1923dup|NC_000003.11(CTNNB1_v004):c.1923dup|NC_000003.11(CTNNB1_v005):c.1923dup|NC_000003.11(CTNNB1_v006):c.1923dup|NC_000003.11(CTNNB1_v007):c.1902dup
NC_000003.11(CTNNB1_i001):p.(Glu642Argfs*6)|NC_000003.11(CTNNB1_i002):p.(Glu642Argfs*6)|NC_000003.11(CTNNB1_i003):p.(Glu642Argfs*6)|NC_000003.11(CTNNB1_i004):p.(Glu642Argfs*6)|NC_000003.11(CTNNB1_i005):p.(Glu642Argfs*6)|NC_000003.11(CTNNB1_i006):p.(Glu642Argfs*6)|NC_000003.11(CTNNB1_i007):p.(Glu635Argfs*6)
NC_000003.11:g.41277961_41277962delAG

NC_000003.11(CTNNB1_v001):c.1925_1926del|NC_000003.11(CTNNB1_v002):c.1925_1926del|NC_000003.11(CTNNB1_v003):c.1925_1926del|NC_000003.11(CTNNB1_v004):c.1925_1926del|NC_000003.11(CTNNB1_v005):c.1925_1926del|NC_000003.11(CTNNB1_v006):c.1925_1926del|NC_000003.11(CTNNB1_v007):c.1904_1905del
NC_000003.11(CTNNB1_i001):p.(Glu642Valfs*5)|NC_000003.11(CTNNB1_i002):p.(Glu642Valfs*5)|NC_000003.11(CTNNB1_i003):p.(Glu642Valfs*5)|NC_000003.11(CTNNB1_i004):p.(Glu642Valfs*5)|NC_000003.11(CTNNB1_i005):p.(Glu642Valfs*5)|NC_000003.11(CTNNB1_i006):p.(Glu642Valfs*5)|NC_000003.11(CTNNB1_i007):p.(Glu635Valfs*5) HpyCH4III

NC_000003.11:g.41278087dupG

NC_000003.11(CTNNB1_v001):c.1963dup|NC_000003.11(CTNNB1_v002):c.196
3dup|NC_000003.11(CTNNB1_v003):c.1963dup|NC_000003.11(CTNNB1_v004):c.1963
dup|NC_000003.11(CTNNB1_v005):c.1963dup|NC_000003.11(CTNNB1_v006):c.1963d
up|NC_000003.11(CTNNB1_v007):c.1942dup
NC_000003.11(CTNNB1_i001):p.(Ala655Glyfs*10)|NC_000003.11(CTNNB1_i0
02):p.(Ala655Glyfs*10)|NC_000003.11(CTNNB1_i003):p.(Ala655Glyfs*10)|NC_00
0003.11(CTNNB1_i004):p.(Ala655Glyfs*10)|NC_000003.11(CTNNB1_i005):p.(Ala6
55Glyfs*10)|NC_000003.11(CTNNB1_i006):p.(Ala655Glyfs*10)|NC_000003.11(CTN
NB1_i007):p.(Ala648Glyfs*10) HpyCH4V,MwoI
NC_000003.11:g.41278105C>T

NC_000003.11(CTNNB1_v001):c.1981C>T|NC_000003.11(CTNNB1_v002):c.198
1C>T|NC_000003.11(CTNNB1_v003):c.1981C>T|NC_000003.11(CTNNB1_v004):c.1981
C>T|NC_000003.11(CTNNB1_v005):c.1981C>T|NC_000003.11(CTNNB1_v006):c.1981C
>T|NC_000003.11(CTNNB1_v007):c.1960C>T
NC_000003.11(CTNNB1_i001):p.(Arg661*)|NC_000003.11(CTNNB1_i002):p.(
Arg661*)|NC_000003.11(CTNNB1_i003):p.(Arg661*)|NC_000003.11(CTNNB1_i004):
p.(Arg661*)|NC_000003.11(CTNNB1_i005):p.(Arg661*)|NC_000003.11(CTNNB1_i00
6):p.(Arg661*)|NC_000003.11(CTNNB1_i007):p.(Arg654*)
NC_000003.11:g.41278174_41278175delinsC

NC_000003.11(CTNNB1_v001):c.2050_2051delinsC|NC_000003.11(CTNNB1_v0
02):c.2050_2051delinsC|NC_000003.11(CTNNB1_v003):c.2050_2051delinsC|NC_00
0003.11(CTNNB1_v004):c.2050_2051delinsC|NC_000003.11(CTNNB1_v005):c.2050_
2051delinsC|NC_000003.11(CTNNB1_v006):c.2050_2051delinsC|NC_000003.11(CTN
NB1_v007):c.2029_2030delinsC
NC_000003.11(CTNNB1_i001):p.(Arg684Glnfs*51)|NC_000003.11(CTNNB1_i0
02):p.(Arg684Glnfs*51)|NC_000003.11(CTNNB1_i003):p.(Arg684Glnfs*51)|NC_00
0003.11(CTNNB1_i004):p.(Arg684Glnfs*51)|NC_000003.11(CTNNB1_i005):p.(Arg6
84Glnfs*51)|NC_000003.11(CTNNB1_i006):p.(Arg684Glnfs*51)|NC_000003.11(CTN
NB1_i007):p.(Arg677Glnfs*51) MmeI AcuI,Hpy188I
NC_000003.11:g.41279524_41279525insCC

NC_000003.11(CTNNB1_v001):c.2094_2095insCC|NC_000003.11(CTNNB1_v002
):c.2094_2095insCC|NC_000003.11(CTNNB1_v003):c.2094_2095insCC|NC_000003.1
1(CTNNB1_v004):c.2094_2095insCC|NC_000003.11(CTNNB1_v005):c.2094_2095insC
C|NC_000003.11(CTNNB1_v006):c.2094_2095insCC|NC_000003.11(CTNNB1_v007):c.
2073_2074insCC
NC_000003.11(CTNNB1_i001):p.(Asp699Profs*37)|NC_000003.11(CTNNB1_i0
02):p.(Asp699Profs*37)|NC_000003.11(CTNNB1_i003):p.(Asp699Profs*37)|NC_00
0003.11(CTNNB1_i004):p.(Asp699Profs*37)|NC_000003.11(CTNNB1_i005):p.(Asp6
99Profs*37)|NC_000003.11(CTNNB1_i006):p.(Asp699Profs*37)|NC_000003.11(CTN
NB1_i007):p.(Asp692Profs*37) BcgI (2),Hpy188I
NC_000003.11:g.41279542dupA

NC_000003.11(CTNNB1_v001):c.2112dup|NC_000003.11(CTNNB1_v002):c.211
2dup|NC_000003.11(CTNNB1_v003):c.2112dup|NC_000003.11(CTNNB1_v004):c.2112
dup|NC_000003.11(CTNNB1_v005):c.2112dup|NC_000003.11(CTNNB1_v006):c.2112d
up|NC_000003.11(CTNNB1_v007):c.2091dup
NC_000003.11(CTNNB1_i001):p.(Glu705Argfs*9)|NC_000003.11(CTNNB1_i00
2):p.(Glu705Argfs*9)|NC_000003.11(CTNNB1_i003):p.(Glu705Argfs*9)|NC_00000
3.11(CTNNB1_i004):p.(Glu705Argfs*9)|NC_000003.11(CTNNB1_i005):p.(Glu705Ar

gfs*9)|NC_000003.11(CTNNB1_i006):p.(Glu705Argfs*9)|NC_000003.11(CTNNB1_i0
07):p.(Glu698Argfs*9) MboII
NC_000003.11:g.41280760delA

NC_000003.11(CTNNB1_v001):c.2273del|NC_000003.11(CTNNB1_v002):c.227
3del|NC_000003.11(CTNNB1_v003):c.2273del|NC_000003.11(CTNNB1_v004):c.2273
del|NC_000003.11(CTNNB1_v005):c.2273del|NC_000003.11(CTNNB1_v006):c.2273d
el|NC_000003.11(CTNNB1_v007):c.2252del
NC_000003.11(CTNNB1_i001):p.(His758Leufs*30)|NC_000003.11(CTNNB1_i0
02):p.(His758Leufs*30)|NC_000003.11(CTNNB1_i003):p.(His758Leufs*33)|NC_00
0003.11(CTNNB1_i004):p.(His758Leufs*39)|NC_000003.11(CTNNB1_i005):p.(His7
58Leufs*30)|NC_000003.11(CTNNB1_i006):p.(His758Leufs*30)|NC_000003.11(CTN
NB1_i007):p.(His751Leufs*33) BbvI Cac8I,NspI,SphI

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Ethics Declaration:

"*Genomic and phenotypic characterization of 404 individuals with neurodevelopmental disorders caused by CTNNB1 variants*"
Kayumi et al.

On behalf of all authors, I declare:
This study was approved by the Women's and Children's Health Network Human Research Ethics Committee number 2020/HRE01273. Written informed consent was obtained for all individuals for whom new data are presented in this study. Tabulated, individual-level data (Table S1) in this study are de-identified. Copies of explicit informed written consent was obtained from individuals or their legal guardian for the display of photographs (Figure 1F) and these are archived with the corresponding author.

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