Supplementary Table S2. VEP annotation of 3 missense variants in the shelterin genes in 24 NMTC families (Part 1)

dbSNP	Location	Allele	Consequence	IMPACT	SYMBOL
rs1464083474	14:24709044-24709044	G	missense_variant	MODERATE	TINF2
rs1464083474	14:24709044-24709044	G	missense_variant	MODERATE	TINF2
rs1464083474	14:24709044-24709044	G	missense_variant	MODERATE	TINF2
rs1464083474	14:24709044-24709044	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs202093758	14:24711465-24711465	G	missense_variant	MODERATE	TINF2
rs142662151	16:67693939-67693939	T	missense_variant	MODERATE	ACD
rs142662151	16:67693939-67693939	T	missense_variant	MODERATE	ACD
rs142662151	16:67693939-67693939	T	missense_variant	MODERATE	ACD
rs142662151	16:67693939-67693939	T	missense_variant,splice_region_variant	MODERATE	ACD
rs142662151	16:67693939-67693939	T	missense_variant,splice_region_variant	MODERATE	ACD
rs142662151	16:67693939-67693939	T	missense_variant,splice_region_variant	MODERATE	ACD
rs142662151	16:67693939-67693939	T	missense_variant,splice_region_variant	MODERATE	ACD

Supplementary Table S2, Part 2

dbSNP	SYMBOL	Gene	EXON	cDNA_position	CDS_position	Protein_position	Amino_acids	Codons
rs1464083474	TINF2	ENSG00000092330	9/9	1657	1315	439	V/L	Gtt/Ctt
rs1464083474	TINF2	ENSG00000092330	8/8	1483	1210	404	V/L	Gtt/Ctt
rs1464083474	TINF2	26277	9/9	1657	1315	439	V/L	Gtt/Ctt
rs1464083474	TINF2	26277	8/8	1905	1210	404	V/L	Gtt/Ctt
rs202093758	TINF2	ENSG00000092330	1/9	416	74	25	G/A	gGa/gCa
rs202093758	TINF2	ENSG00000092330	1/6	405	74	25	G/A	gGa/gCa
rs202093758	TINF2	ENSG00000092330	1/8	347	74	25	G/A	gGa/gCa
rs202093758	TINF2	ENSG00000092330	1/2	300	74	25	G/A	gGa/gCa
rs202093758	TINF2	ENSG00000092330	1/4	355	74	25	G/A	gGa/gCa
rs202093758	TINF2	ENSG00000092330	1/2	374	74	25	G/A	gGa/gCa
rs202093758	TINF2	ENSG00000092330	1/7	30	32	11	G/A	gGa/gCa
rs202093758	TINF2	26277	1/9	416	74	25	G/A	gGa/gCa
rs202093758	TINF2	26277	1/6	416	74	25	G/A	gGa/gCa
rs202093758	TINF2	26277	1/9	416	74	25	G/A	gGa/gCa
rs202093758	TINF2	26277	1/8	769	74	25	G/A	gGa/gCa
rs142662151	ACD	ENSG00000102977	2/12	632	367	123	D/N	Gac/Aac
rs142662151	ACD	65057	2/12	704	367	123	D/N	Gac/Aac
rs142662151	ACD	65057	2/11	649	367	123	D/N	Gac/Aac
rs142662151	ACD	ENSG00000102977	2/12	690	358	120	D/N	Gac/Aac
rs142662151	ACD	ENSG00000102977	2/12	119	121	41	D/N	Gac/Aac
rs142662151	ACD	65057	2/12	695	358	120	D/N	Gac/Aac
rs142662151	ACD	65057	2/12	695	358	120	D/N	Gac/Aac

Supplementary Table S2, Part 3

dbSNP	SYMBOL	FLAGS	SYMBOL_SOURCE	HGNC_ID	SOURCE	GIVEN_REF	USED_REF	BAM_EDIT
rs1464083474	TINF2	-	HGNC	11824	Ensembl	C	C	-
rs1464083474	TINF2	-	HGNC	11824	Ensembl	C	C	-
rs1464083474	TINF2	-	EntrezGene	11824	RefSeq	C	C	OK
rs1464083474	TINF2	-	EntrezGene	11824	RefSeq	C	C	-
rs202093758	TINF2	-	HGNC	11824	Ensembl	C	C	-
rs202093758	TINF2	-	HGNC	11824	Ensembl	C	C	-
rs202093758	TINF2	-	HGNC	11824	Ensembl	C	C	-
rs202093758	TINF2	cds_end_NF	HGNC	11824	Ensembl	C	C	-
rs202093758	TINF2	-	HGNC	11824	Ensembl	C	C	-
rs202093758	TINF2	-	HGNC	11824	Ensembl	C	C	-
rs202093758	TINF2	cds_start_NF	HGNC	11824	Ensembl	C	C	-
rs202093758	TINF2	-	EntrezGene	11824	RefSeq	C	C	OK
rs202093758	TINF2	-	EntrezGene	11824	RefSeq	C	C	OK
rs202093758	TINF2	-	EntrezGene	11824	RefSeq	C	C	-
rs202093758	TINF2	-	EntrezGene	11824	RefSeq	C	C	-
rs142662151	ACD	-	HGNC	25070	Ensembl	C	C	-
rs142662151	ACD	-	EntrezGene	25070	RefSeq	C	C	OK
rs142662151	ACD	-	EntrezGene	25070	RefSeq	C	C	-
rs142662151	ACD	-	HGNC	25070	Ensembl	C	C	-
rs142662151	ACD	cds_start_NF	HGNC	25070	Ensembl	C	C	-
rs142662151	ACD	-	EntrezGene	25070	RefSeq	C	C	OK
rs142662151	ACD	-	EntrezGene	25070	RefSeq	C	C	OK

Supplementary Table S2, Part 4

dbSNP	SYMBOL	SIFT	PolyPhen	CLIN_SIG	SOMATIC	PHENO
rs1464083474	TINF2	tolerated_low_confidence(0.1)	benign(0.015)	-	-	-
rs1464083474	TINF2	tolerated_low_confidence(0.15)	benign(0.015)	-	-	-
rs1464083474	TINF2	tolerated_low_confidence(0.1)	benign(0.015)	-	-	-
rs1464083474	TINF2	tolerated_low_confidence(0.15)	benign(0.015)	-	-	-
rs202093758	TINF2	tolerated(0.66)	benign(0.085)	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.68)	benign(0.085)	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.65)	benign(0.085)	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.71)	benign(0.085)	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.78)	benign(0.209)	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.73)	benign(0.358)	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.7)	benign(0.205)	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.66)	benign(0.085)	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.68)	benign(0.085)	benign,likely_benign	-	1
rs202093758	TINF2	-	-	benign,likely_benign	-	1
rs202093758	TINF2	tolerated(0.65)	benign(0.085)	benign,likely_benign	-	1
rs142662151	ACD	deleterious(0)	benign(0.408)	uncertain_significance	-	1
rs142662151	ACD	deleterious(0)	benign(0.408)	uncertain_significance	-	1
rs142662151	ACD	-	-	uncertain_significance	-	1
rs142662151	ACD	tolerated(0.09)	benign(0.31)	uncertain_significance	-	1
rs142662151	ACD	tolerated(0.07)	possibly_damaging(0.471)	uncertain_significance	-	1
rs142662151	ACD	tolerated(0.09)	possibly_damaging(0.76)	uncertain_significance	-	1
rs142662151	ACD	tolerated(0.09)	benign(0.31)	uncertain_significance	-	1

Supplementary Table S2, Part 5

dbSNP	SYMBOL	PUBMED	MOTIF_NAME	CADD_PHRED	CADD_RAW
rs1464083474	TINF2	-	-	0.244	-0.481501
rs1464083474	TINF2	-	-	0.244	-0.481501
rs1464083474	TINF2	-	-	0.244	-0.481501
rs1464083474	TINF2	-	-	0.244	-0.481501
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs202093758	TINF2	-	-	12.31	1.307027
rs142662151	ACD	-	-	25.6	5.237038
rs142662151	ACD	-	-	25.6	5.237038
rs142662151	ACD	-	-	25.6	5.237038
rs142662151	ACD	-	-	25.6	5.237038
rs142662151	ACD	-	-	25.6	5.237038
rs142662151	ACD	-	-	25.6	5.237038
rs142662151	ACD	-	-	25.6	5.237038

VEP, Variant Effect Predictor (https://useast.ensembl.org/info/docs/tools/vep/index.html)