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Supplementary Data

Haploinsufficiency of a Spliceosomal GTPase

Encoded by *EFTUD2* Causes Mandibulofacial

Dysostosis with Microcephaly

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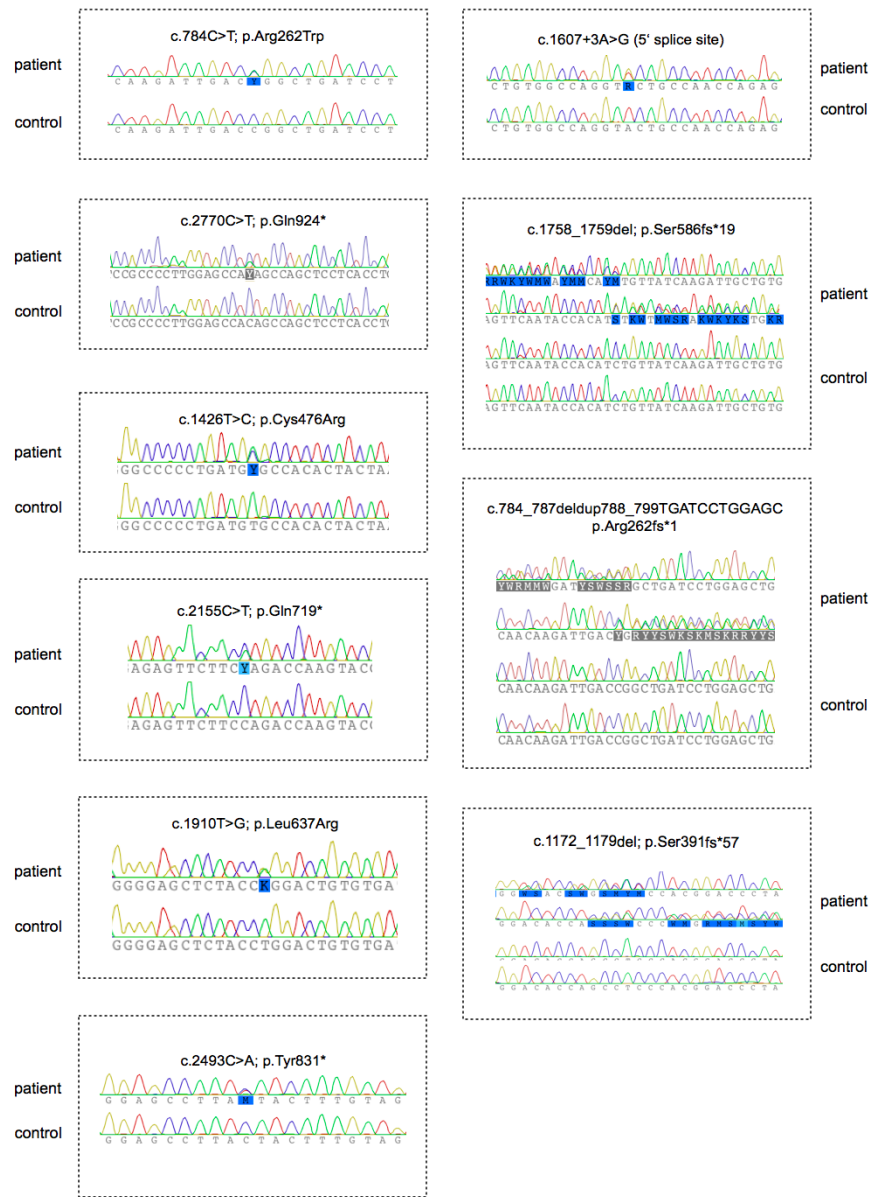


Figure S1. Chromatograms Demonstrating Mutations in *EFTUD2*

Chromatograms demonstrating MFDM-causing mutations (Table 3) are depicted above. All mutations were confirmed by bidirectional Sanger sequencing.



Figure S2. Chromosomal Rearrangements Identified in Individuals with MFDM

Image adapted from UCSC genome browser (hg19) showing microdeletions (red) and microduplications (blue) in probands 1 and 5. Also shown are benign copy number variants from the Database of Genomic Variants and potentially pathogenic rearrangements from DECIPHER (see Web Resources). The small partial deletion in individual 1 involves only *EFTUD2* and a single neighbouring gene, *HIGD1B*. The complex rearrangement in individual 5 includes a 17q21.31 microdeletion (involving eleven genes, including *EFTUD2*), a microduplication of a further ten genes within a distinct but adjacent interval on 17q21.31, and an agenic deletion at 4q12 (59109311-59427079; not depicted). Apart from the presence of VSD, this individual's phenotype otherwise resembles that of patients with *EFTUD2* point mutations.

Table S1. Primers for *EFTUD2* Amplification and Sequencing

Name	Forward	Reverse
EFTUD2_ex1 (noncoding)	GTGATCTGCCCGAAGGC	GCATATAAGGCCCTTCCAG
EFTUD2_ex2	AGGCATGTGAAAGGGGTATG	TCACTTAAGCCCAGGAGGTG
EFTUD2_ex3	GTCAGTGCTGTCACTCACGG	GAACTTTTAAGGTTTGCTGAGAGC
EFTUD2_ex4	TGAAAGGTCAGTCTAATGGAATAGTG	GCCAAAAGCCACCTCTTATC
EFTUD2_ex5	GCCTGAGCACTCTCCAGTTG	CACCCCTAGTCAGGAGGTG
EFTUD2_ex6	ATGTGGTGTATTGGGAGCTG	AGGAAGGGTGAAGGAAGGAG
EFTUD2_ex7	TGAGGGAAAGTGATTGATGG	AAAGGCTCCTCTACATTCCC
EFTUD2_ex8	TTACTAGCAACTGGGGAGCC	AAAGATGCACTGCTCCGTTT
EFTUD2_ex9	CCCTCAGTTCACCCTACCAG	GTGCTCTGGGTATTGTTGCG
EFTUD2_ex10	GCCTTCITTGGTTCTGCTTTG	AATCACAGTTGTTCCCAGAGG
EFTUD2_ex11	GGAGAGTGGCTGTGATACCG	AGTCATTGAGCAGGAAAGGC
EFTUD2_ex12	AAGGAACTGCCTGGACCC	TAGGGGAGGAAAGACGGTAG
EFTUD2_ex13	TGGAAACTTTACAGTCACAC	TTAGTGACCAACAACCACCC
EFTUD2_ex14	TCCCTGTCTCCCTCTAGC	AGATTTCTGACCTCCATCGC
EFTUD2_ex15	TGGCTCAGTAACTCCTGCTC	TGGGAAAGTGGGCATTACTC
EFTUD2_ex16	GAGCTCATGTCCTTGCTGTC	TCACTCCTTCCAGTCCTTG
EFTUD2_ex17	GGGAATGCTCTGCCTAAGTTT	AGGACATGGCAATGACTGTG
EFTUD2_ex18	TGATCCTAAGCAAGTCCTCTTC	GAAGCGGTGTGCAGCTC
EFTUD2_ex19	TGGCCTTGATTTTGTGTTGTG	CCAGGAATTCAAGGATTTGG
EFTUD2_ex20	AGGCAGTTGGGGTTTTCAC	GTGCATAGTGCTCATGGTGG
EFTUD2_ex21_22	AGCTGTCCGCTCTTGTTACCC	CTTTTGGGAAAAGAACAACCTTG
EFTUD2_ex23	AGGCACATCTGTCTTCTCCC	AATCCTAAAGATGGTGAGCCC
EFTUD2_ex24_25	AGCTGCAGTGTTCAGGAG	TGTAGGAGCCGAGGTGACTC
EFTUD2_ex26	GCTGGAATGCATTTGGGG	GATCAGGACAGAAGGGGATG
EFTUD2_ex27	AGAAGAGTCTGCTTCTGGG	GGGCCAAAAGAAAGAGAGAG
EFTUD2_ex28	AGGTCTTGGATAGTCTCCTTGC	AATTAAGGAGGAGGCGGC