

Supplemental material

Biallelic *NDUF44* deletion causes Mitochondrial Complex IV deficiency in a patient with Leigh syndrome

Sanger sequencing of the PCR product obtained with primers flanking the deletion in the patient

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ATCATCCCAC TTCTTAAGAC TTACAGCTCT GGTGTAAAAG CCAGTCACCT 50
TGTTTCATGTG TTGAAATCAA TGATATTTGC AATAAAAAGA TTATTTAATC 100
AGATTGTTAA CAGCTGAATA AGCTTTTACC AAATAGTCCT TTTCTTTCCA 150
AGTTTTATAT AATATGGCTT TTAGAATTTT CCCCGccagg catggtggct 200
cacgcctgta atcccagtac ttcaggaggt caaggcagga agatcacgac 250
gtgaagagac tgagaccatc ctggccaata tggtgaaaac tcgtctctac 300
caaaaataca aaaaattacc ccagtgtggt ggcattgcacc tgtagtccca 350
gctactcggg aggctgaggc aggagaattg cttgaaccgc ggagacagag 400
gttgcagtga gccgagttcg caccactgca ctccagcctg ggcaatatga 450
gcaaaaactct gtctcagaaa aaaaaaaaaa aattgcttga actcaggaga 500
tagaggttgc agtgagccaa gattgcacca ctgcactcca actgggtgac 550
agagtggagac tccatctctg aaacaaaaat aaaataaaaa TAACAAAGAA 600
AACCAGctgg gcatgttggc ataaccgtag tcccacctac ttgggaggct 650
aaagcgggag gattgtttta actcaggagt tgcaggctac aatgaactaa 700
gatcgcgccca ctgcactcca gcctgggtga cagagagaaa ctctgtccca 750
aaaTAAGTGA TGCCTTTAtt tattttat ttttttggag 800
atggagtctc gctctgtggc ccaacctgga gtgtggtggc acgatctcgg 850
ctcactgcaa cctctgcctc ccagggttcaa gcgattctcc tgcctcagcc 900
tccccaatag ctgggactac aggcacatgc caccacaccc ggttaatttt 950
ttgtatTTTT agtagagatg gggtttcacc gtgttagtca ggatggtctc 1000
gatctctgga cctcgtgatc tgcccacctc ggccctccca agtgctggga 1050
ttacagggtg gagccaccgc acctggccTA AGTGATGCCT TTAGAGATGT 1100
TAACAAAGCA AGGTTACAAA TATAAATTTC ATTAAATATT AACCAAATG 1150
TCAAATTGTG AAAGTTTAAA TTTTGCACAT TGAAGTTAGT GGA
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Color legend.

Black DNA mapping at chr7:10969033-10969448 with 100% identity.

Blue DNA mapping at chr19:14427161-14427497, 337 bases, with 100% identity.

Green DNA mapping at chr7:10982429-10982868 with 100% identity.

The 337 bases have also 82.1% identity to chr7:101018817-101019244, spanning 428 bases

(<https://genome.ucsc.edu/>, BLAT Tool).

Small letters indicate repetitive elements.

Alignment between chr19:14427161-14427497 and AluSc8 at chr7:10982444-10982753

Identities	Gaps	Strand
91/116 (78%)	2/116 (1%)	Plus/Minus

Chr19:14427223 aGAATTGCTTGAACCTCAGGAGATAGAGGTTGCAGTGAGCCAAAGATTGCACCACCTGCACCTC Chr19:14427282
| | | | | | | | | | | | | | | | | | | | | | | | | | | |
Chr7:10982563 AGAATCGCTTGAACCTGGGAGGCAGAGGTTGCAGTGAGCCGAGATCGTGCCACCACACTC Chr7:10982503

Chr19:14427283 CA-ACTGGGTGACAGAGTGAGACTCCATCTCTGaaacaaaataaaaaataaa Chr19:14427337
| | | | | | | | | | | | | | | | | | | | | | | | | | | |
Chr7:10982503 CAGGTTGGGGCCACAGAGCGAGACTCCATCTC-AAAAAATAAACAAAACAAAAATAA Chr7:10982449

Alignment between chr19:14427161-14427497 and AluSc at chr7:10969217-10969522

Range 1: 239 to 296

Identities	Gaps	Strand
52/60 (87%)	2/60 (3%)	Plus/Plus

Chr19:14427167 CCACTGCACTCCAGCCTGGGCAATATGAGCAAACACTGTGTCTCAGaaaaaaaaaaaaaGAA Chr19:14427226
| | | | | | | | | | | | | | | | | | | | | | | |
Chr7:10969455 CCACTGCACTCCAGCCT-GGCAACA-GAGCAAGACTCCATCTCAAAAAAAAAAAAAAAAAA Chr7: 10969513

Range 2: 190 to 304

Identities	Gaps	Strand
95/115 (83%)	0/115 (0%)	Plus/Plus

Chr19:14427223	aGAATTGCTTGAACCTCAGGAGATAGAGGTTGCAGTGAGCCAAAGATTGCACCACTGCACTC	Chr19:14427282
Chr7:10969406	AGAATTGCTTGAACCCGGGAGACAGAGGTTGCAGTGAGCCGAGATGGTGCCACTGCACTC	Chr7:10969465
Chr19:14427283	CAACTGGGTGACAGAGTGAGACTCCATCTCTGaaaacaaaataaaaataaa	Chr19:14427337
Chr7:10969455	CAGCGTGGCAACAGAGCAAGACTCCATCTCAAAAAAAAAAAAAAAAAAAAAA	Chr7:10969513

Range 3: 143 to 283

Identities	Gaps	Strand
107/142 (75%)	3/142 (2%)	Plus/Plus

Chr19:14427358	TGTTGGCATA-ACC-GTAGTCCCACCTACTTGGGAGGCTAAAGCGGGAGGATTGTTTAAA	Chr19:14427415
Chr7:10969359	TGGTGGCATGCACCTGTAGTCCCAGCTACTCGGGAGGCTGAGGCAGGAGAATTGCTTGAA	Chr7:10969408
Chr19:14427416	CTCAGGAGTTGCAGGCTACAATGAACTAAGATCGCGCCACTGCACTCCAGCCTGGGTGAC	Chr19:14427475
Chr7: 10969409	CCCCGGAGACAGAGGTTGCAGTGAGCCGAGATGGTGCCACTGCACTCCAGCCT-GGCAAC	Chr7:10969458
Chr19:14427476	AGAGAGAAACTCTGTCCCAAAA	Chr19:14427497
Chr7: 10969459	AGAGCAAGACTCCATCTCAAAA	Chr7:10969499