

## Supplementary material genetics

Figure S1. Sanger sequencing confirming the presence of *ATP7B* c.3207C>A variant (primer ATP7BEx13F).

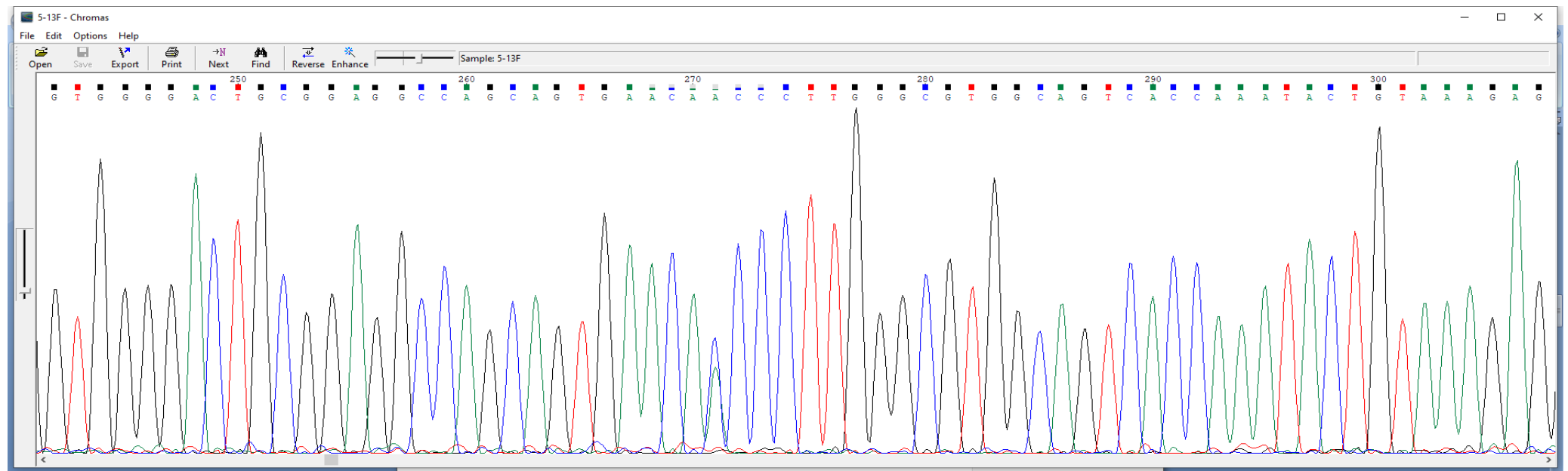


Table S4. Primers used in the study

Gene/application	Primer name	sequence	reference
ATP7B (Sanger sequencing)	ATP7BEx13F	TTTCCAGACGTCCATCACGG	Designed for this study (NCBI primer blast)
	ATP7BEx15R	GGAAGTCCGTGCAGTATCCC	
GAPDH	GAPDHF	GTTCGACAGTCAGCCGCATC	[1]
	GAPDHR	GGAATTTGCCATGGGTGGA	
PPIB	PPIBF	GGTGATCTTTGGTCTCTTCGG	[2]
	PPIBR	TAGATGCTCTTTCCTCCTGTG	
DSCR1	DSCR1F	GCACCAGCTCAAGAAGGAACC	[3]
	DSCR1R	GGGACTCAAATTTGGCCCTGG	
SETX (Real time PCR analysis)	SETXRT10F	GGTCCAGAAAGGGGATGTGA	Designed for this study (NCBI primer blast)
	SETXRT10R	CGTTTTGAGGTTTAGCAAGAGC primers are color highlighted in Scheme 1	
SETX (isoform analysis: PCR and Sanger sequencing)	SETX10F	GGTCCAGAAAGGGGATGTGA	Designed for this study (NCBI primer blast)
	SETX10R	GTCAGGGGCCTGTTCTCTTG primers are color highlighted in Scheme 1	

## References:

1. Chung, S.W.; Choi, B.M.; Kim, J.Y.; Lee, Y.S.; Yoon, J.P.; Oh, K.S.; Park, K.S. Altered Gene and Protein Expressions in Torn Rotator Cuff Tendon Tissues in Diabetic Patients. *Arthroscopy* **2017**, *33*, 518-526 e511,
2. Lefever, S.; Vandesompele, J.; Speleman, F.; Pattyn, F. RTPrimerDB: the portal for real-time PCR primers and probes. *Nucleic Acids Res* **2009**, *37*, D942-945,
3. Gong, C.; Kim, Y.K.; Woeller, C.F.; Tang, Y.; Maquat, L.E. SMD and NMD are competitive pathways that contribute to myogenesis: effects on PAX3 and myogenin mRNAs. *Genes Dev* **2009**, *23*, 54-66,

Table S9. Human Splicing Finder analysis of SETX variant (rs755971927, c.2385\_2387delAAA, p.Ile795\_Lys796delinsMe) detected in the patient.

HSF analysis verdict: new donor splice site, activation of a cryptic donor site. Potential alteration of splicing.

	name	position	sequences	variation
1 (highlighted in magenta color) in Scheme S1	HSF Donor site (matrix GT)	chr9:132329218	GTAATAAAG>CATGTAATA	43.74>77.11 (76.29%)
2 (highlighted in sea green) in Scheme S1	HSF Donor site (matrix GT)	chr9:132329228	GTTATCACA>AAAGTTATC	36.57>70.95 (94.01%)

Scheme S1. The scheme of aberrant splicing in the patient's fibroblasts carrying variant *SETX* c.2385\_2387delAAA (rs755971927) in exon 10 - deletion of AAA (highlighted in red)

SETX Exon 10 sequence (Transcript: ENST00000224140.6; ensembl)

GATTCTGGATGGAGAACAGCCATTTGCCAGATTATTGTCCTAACATGTATGAAGAAATG  
GAAACATTAGCCAGTGTACTTCAGTCAGATATTGGTCAAGACATGCGTGTTTCATAACAGC  
ACATTTCTATGGTTCATCCCTTTTGTCCAGTCCCTCATGGATCTTAAGGATTTGGGTGTG  
GCTTACATAGCACAGGTTGTTAATCATCTGTACTCTGAAGTCAAAGAAGTCCTCAACCAA  
ACAGATGCTGTGTGTGACAAAGTCACTGAATTTTTCTTCTAATTTTGGTATCAGTGATT  
GAACTGCATAGAAATAAAAAATGTTTGCATTTGCTGTGGGTAAAGTCCCAGCAATGGGTG  
GAAGCCGTCGTCAAATGTGCCAAGCTTCCTACCACTGCGTTTACACGGAGTTCTGAGAAA  
TCATCTGGAAATTGCTCCAAAGGAACAGCAATGATATCTTCACTGTCATTGCATTCCATG  
CCATCTAACTCTGTACAATTGCTTATGTGCAGCTGATTAGAAGTCTCCTTAAAGAAGGT  
TATCAGCTTGGGCAGCAGTCTCTTTGCAAGCGATTCTGGGATAAGCTCAACTTATTCCTT  
AGAGGAAATTTATCTCTAGGTTGGCAGTTGACTAGTCAGGAAACCCATGAGCTACAAAGT  
TGCTTAAAGCAAATTATTAGAAACATAAAATTCAAAGCACCTCCATGTAACACTTTTGTG  
GATCTGACTTCTGCATGTAAAATCTCTCCTGCATCTTATAATAAAGAAGAAAGTGAACAA  
ATGGGGAAGACGTCTAGAAAAGATATGCATTGTTTGAAGCTTCCAGCCCAACATTTTCT  
AAAGAACCAATGAAAGTGCAAGACAGTGTATTGATCAAAGCAGATAACACTATAGAAGGT

GACAATAATGAGCAAAATTATATAAAGGATGTGAACTAGAGGACCATCTCTTAGCTGGG  
TCATGCTTAAAGCAGAGTAGTAAAAACATTTTTACTGAAAGAGCTGAAGATCAAATTA  
ATAAGTACAAGGAAGCAGAAGTCTGTAAAAGAGATCTCTTCATATACACCAAAGGACTGT  
ACTTCAAGAAATGGTCCAGAAAGGGGATGTGACAGAGGAATAATAGTATCAACACGTTTG  
TTGACTGATTCTAGCACTGATGCTTTGGAAAAAGTGCCACATCGAATGAAGATTTCTCT  
TTAAAGGATGATGCTCTTGCTAAAACCTCAAACGAAAACTAAGGTACAGAAAGATGAA  
ATCTGTGCAAAATTATCACATGTAATAAAGAAGCAACACAGGAAGAGTACTTTGGTCGAT  
AATACTATCAATTTAGATGAAAATTTGACTGTATCTAACATTGAGAGTTTCTATTCAAGG  
AAAGATACAGGAGTTCAGAAAGGAGATGGTTTCATACACAATCTTTCTTTAGACCCTAGT  
GGTGTCTGGATGATAAGAATGGAGAACAAAAATCTCAAAACAATGTATTGCCAAAAGAG  
AAACAATTAAGAATGAAGAATTAGTTATTTCTCTTTCCATGAAAACAATTGTAAATA  
CAGGAATTTTCATGTTGATGGTAAAGAATTGATCCCTTTTACAGAAATGACCAATGCTTCA  
GAGAAGAAATCATCTCCCTTTAAAGATCTTATGACTGTACCTGAATCAAGAGATGAGGAG  
ATGAGTAATAGTACCAGTGTGATTTATTCTAACTTGAACAGAGAACAGGCCCTGACATC  
AGTCCTAAATCTGACACCTTAACGGATTCTCAGATAGACAGAGACCTTCACAAATTATCT  
TTACTAGCTCAAGCCAGTGTTATTACGTTCCCATCCGATTCACCTCAGAACTCATCGCAG  
CTGCAAAGGAAAAGTAAAAGAAGATAAAAGATGTTTCACAGCTAACCAAAATAATGTTGGA  
GATACCTCCCGTGGACAGGTTATTATTATTTAGATTCTGATGATGATGATGATGAAAGA  
ATCCTGAGTCTTGAGAACTCACTAACAGGACAAAATATGCCTTGAGAGGGAACATCCA  
GAGCAGCACGTTTCAACAGTTAATAGTAAGGAGGAAAAGAATCCAGTAAAGGAAGAAAAG

ACAGAGACTCTTTTTCAGTTTGAGGAATCTGATTCTCAGTGTTTTGAGTTTGAAAGTTCA  
TCTGAAGTGTTTTTCAGTTTGGCAAGATCATCCAGACGATAATAATTCAGTTCAAGATGGT  
GAGAAAAAATGTTTGGCTCCTATAGCCAATACTACAAATGGTCAGGGTTGTACAGATTAT  
GTATCTGAAGTTGTTAAAAAAGGAGCAGAGGGCATTGAAGAACACACAAGACCACGGAGT  
ATTTCTGTTGAAGAATTTTGTGAAATTGAAGTAAAAAGCCTAAGAGAAAACGATCTGAA  
AAACCAATGGCTGAAGATCCTGTGAGGCCTTCATCTTCTGTCAGAAATGAGGGCCAGTCT  
GATACTAATAAGAGAGATCTTGTGGGAAATGATTTTAAAAGTATTGATAGAAGGACTTCA  
ACTCCCAATTCACGTATTCAGAGAGCCACTACGGTTTCACAAAAGAAGTCTTCAAAGCTT  
TGTA CTGTACAGAACCCATCAGGAAAGTTCCAGTTTCTAAGACCCCTAAGAAAACTCAT  
TCAGATGCCAAAAAAGGACAGAATAGAAGTTCAAATTACCTAAGTTGTAGAACAACCTCT  
GCTATAGTGCCGCCAAAGAAATTCGTCAGTGTCCTGAGCCAACTTCAACAGCTGAGAAA  
CTTGGCCTGAAAAAGGGTCTCGTAAGGCATATGAGTTGTCCCAGCGGTCTTTGGATTAT  
GTAGCTCAATTACGTGATCATGGCAAAACTGTTGGAGTAGTTGATACCCGAAAAAAGACT  
AAATTAATTTCTCCTCAGAACCTGTCTGTCAGAAATAATAAGAACTTCTGACTAGTCAA  
GAACTTCAGATGCAAAGGCAGATCAGACCCAAATCACAAAAAATAGACGAAGACTTTCT  
GATTGTGAAAGTACAGATGTTAAAAGAGCAGGGTCACATACAGCACAGAATTCTGACATA  
TTTGTACCAGAATCTGATAGGTCAGATTATAATTGTACAGGAGGAACTGAGGTACTTGCC  
AACAGTAACAGAAAACAGTTAATAAAATGCATGCCTTCTGAACCAGAAACCATAAAAGCA  
AAACATGGGTCTCCAGCAACTGATGATGCTTGCCCTTTGAACCAGTGTGATTCTGTAGTG  
TTAAATGGAACAGTACCAACAAATGAAGTAATTGTCTCCACTTCAGAAGACCCTCTGGGT

GGAGGTGATCCAACAGCACGTCATATAGAGATGGCAGCTTTGAAAGAAGGAGAGCCTGAC  
TCCAGCAGTGATGCAGAGGAAGATAACTTATTTTAACCCAAAATGATCCTGAAGATATG  
GATTTATGTTCAAAATGGAGAATGACAATTATAAACTCATTGAACTAATTCATGGAAAA  
GATACAGTTGAGGTTGAAGAAGATTCTGTAAGTCGGCCTCAGTTGGAATCTTTGAGTGGC  
ACAAAGTGTAAGTACAAAGATTGTCTTGAAACCACAAAAACCAGGGTGAATACTGCCCA  
AAACTCTGAAGTGAAAGCAGCAGATGAAGATGTATTCGTAAACCTGGCTTGCCTCCT  
CCTGCATCTAAACCTTTGAGACCTACCACTAAGATTTTGTAGCTCAAAGAGTACTTCACGA  
ATTGCTGGTCTTTCTAAATCTTTGGAACTTCTTCAGCACTTTCACCGTCTCTAAAAAAT  
AAGTCAAAGGGGATACAGTCGATTTTGAAAGTACCACAGCCAGTTCCTCATAGCTCAG  
AAGCCAGTTGGTGAAATGAAGAATTCGTGCAATGTTCTTCATCCTCAGTCTCCGAATAAT  
TCCAACAGGCAAGGTTGCAAAGTCCATTTGGTGAAAGCAAATATTTTCCATCTTCCTCT  
CCAGTAAACATTCTTTGTCATCACAGTCTGTCTCTGACACCTTCGTTAAAGAGGTCTTA  
AAATGGAAATATGAAATGTTTTGAACTTTGGTCAGTGTGGGCCCCCTGCAAGTCTTTGT  
CAGTCCATCTCAAGACCTGTGCCTGTCAGATTTACAATTATGGAGATTATTTAATGTT  
TTTTCCCTTTGATGGTATTGAATACTTTTGAAACA



The Underlined sequence in SETX Exon 10 sequence above is deleted (358 bp) in isoform A (compare with Figure 2 A and C in the manuscript).

The potential new splice sites determined by Human Splicing Finder analysis are highlighted in magenta and sea green colors in SETX Exon 10 sequence above.

Sequences highlighted in yellow and grey in SETX Exon 10 sequence above correspond to the fragments that remain after 358 bp deletion in isoform A.

>2F.ab1. Sequencing result of band \*A excised from agarose gel (compare with Figure 1 line 6, in the manuscript)

GTATCAACACGTTTGTGACTGATTCTAGCACTGATGCTTTGGAAAAAGTGTCCACATCGAATGAAGATTTCTCTTTAAAGGATGATGCTCTTGCTAAAACCTCAAAACGAAAA  
CTAAGAAATGACCAATGCTTCAGAGAAGAAATCATCTCCCTTTAAAGATCTTATGACTGTACCTGAATCAAGAGATGAGGAGATGAGTAATAGTACCAGTGTGATTTATTCTAA  
CTTGACAAGAGAACAGGCCCTGACA

Figure S4. Sequencing result of band \*A excised from agarose gel (compare with Figure 1 line 6)

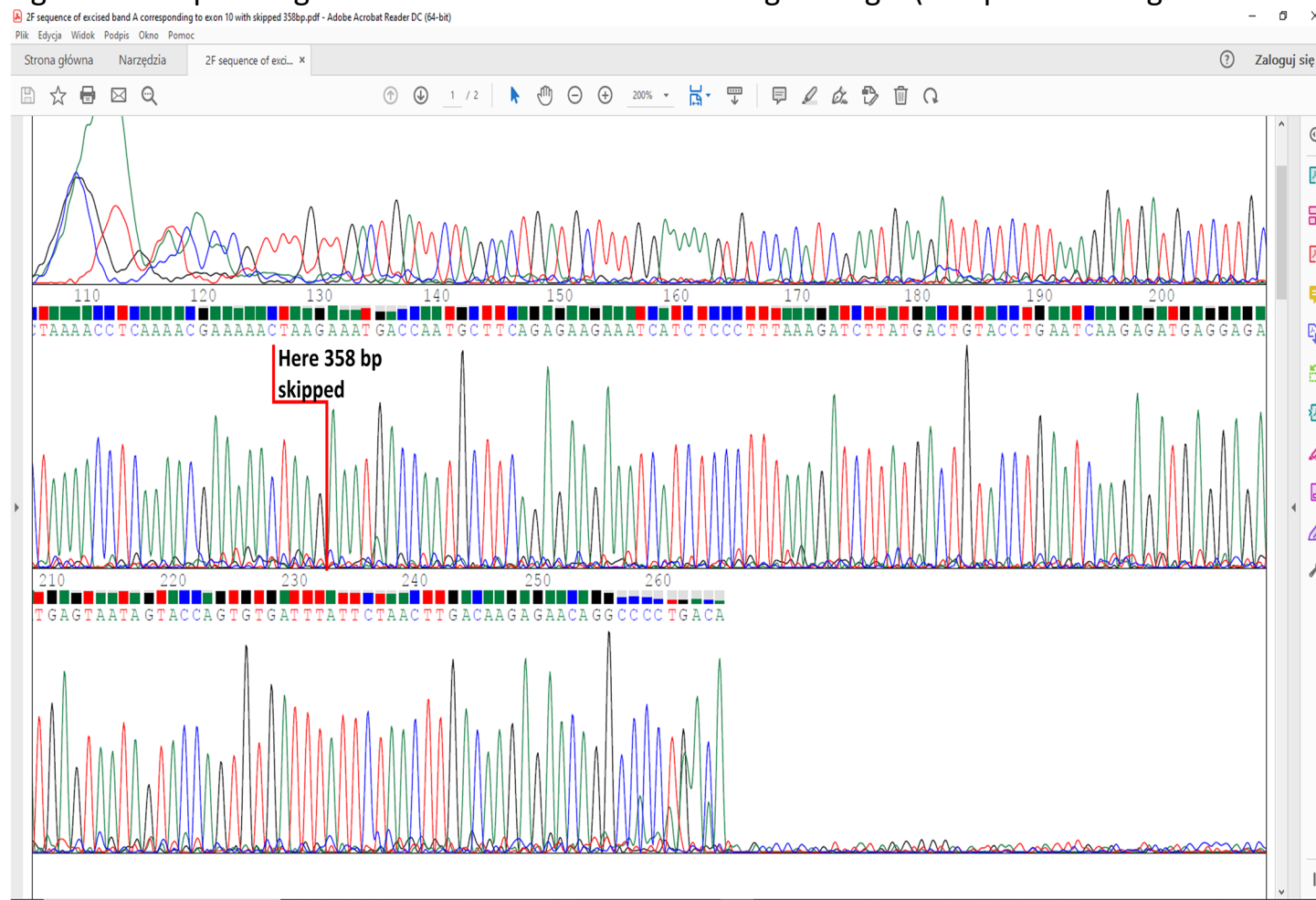


Figure S5. Sequencing result of the main band (see Figure 1 line 6, the strongest band) excised from agarose gel confirmed the presence of *SETX* c.2385\_2387delAAA deletion.

