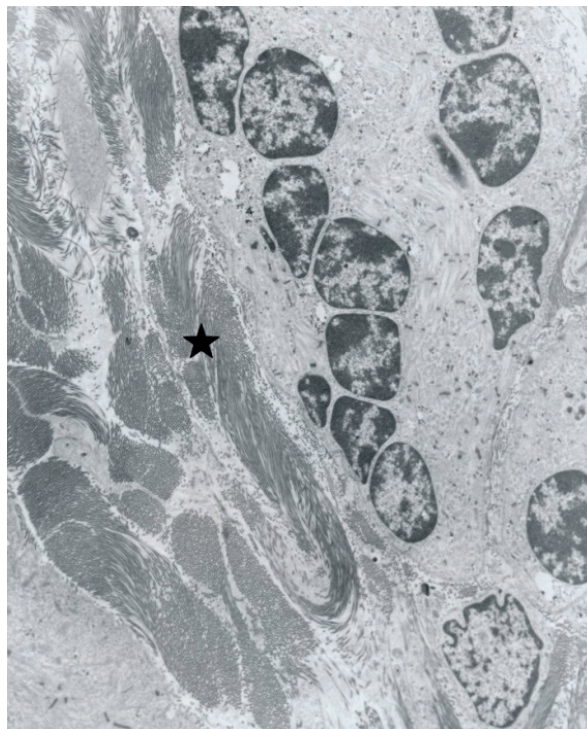
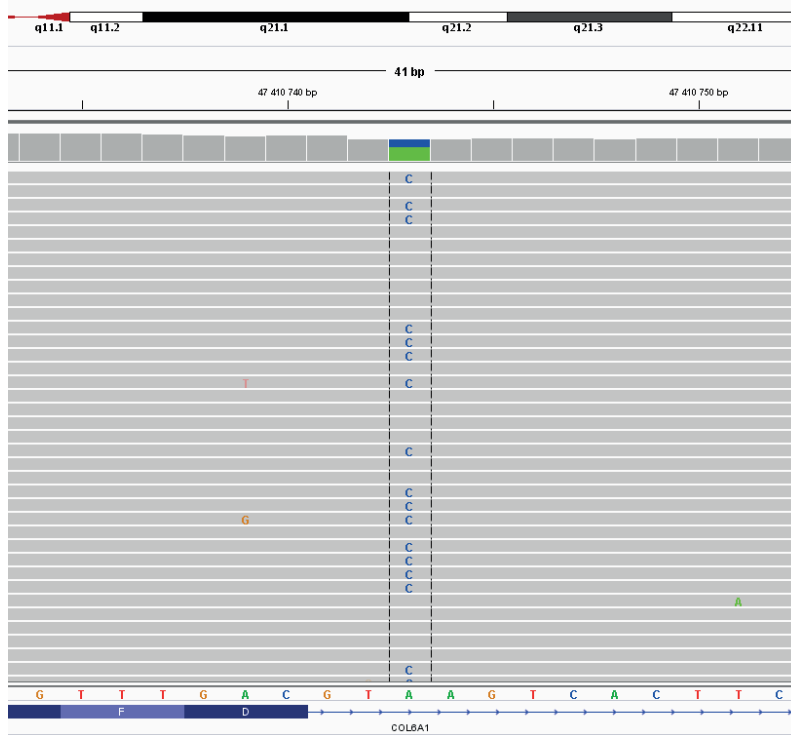


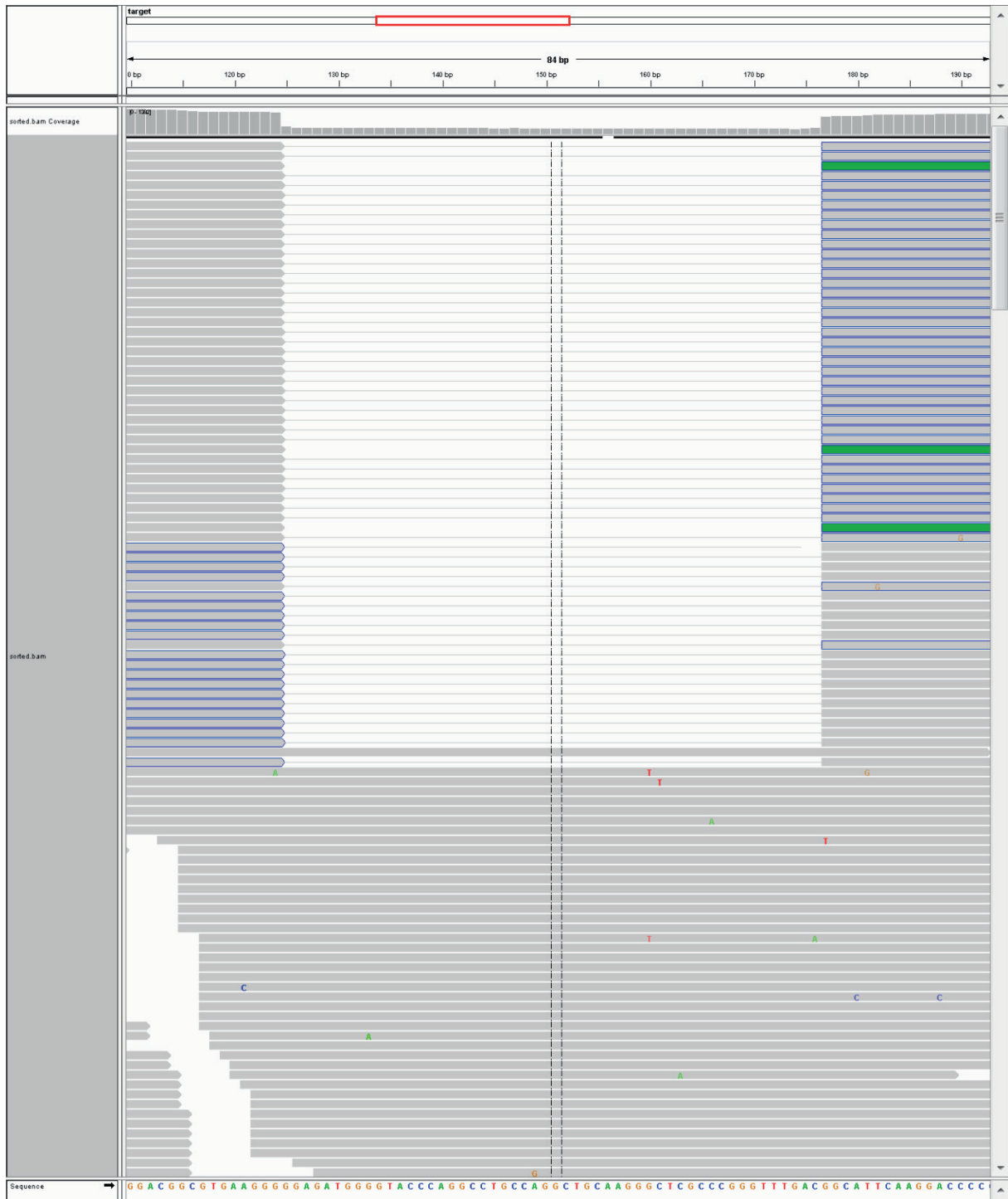
Supplementary Table I. Phenotype–genotype correlations for patients harboring splice-site COL6A1 mutations within 14th intron

COL6A1 mutation	Clinical presentation	Phenotype	Reference
c.1056+1G>A (1 patient)	Slowly progressive limb-girdle myopathy with prominent joint contractures	Mild (BM)	[3]
c.1056+1G>A (2 patients)	Congenital hip dislocation, motor capacity was not affected or was only mildly reduced and distal hyperlaxity and weakness or scoliosis	Mild (BM)	[1]
c.1056+1G>A (7 patients)	Slowly progressive muscle weakness without significant disability until old age, ankle and interphalangeal joint contractures	Mild (BM)	[5]
c.1056+1G>A (patients)	BM	BM	[4]
c.1056+2T>C (3 patients)	Stable muscle weakness, independent walking, Achilles tendon contractures and distal joint hyperlaxity (2) Wheelchair-bound since the age of 35 due to motor disability and multijoint contractures	Mild (BM) Intermediate (moderate progressive)	[6]
c.1056+3A>C (1 patient)	Slowly progressive muscle weakness, walking with orthoses, pronounced distal laxity, Achilles tendon contractures, scoliosis, head drooping	Intermediate (moderate progressive)	This report
c.1056+5G>T (1 patient)	Intermediate ColVI myopathy	Intermediate (moderate progressive)	[2]

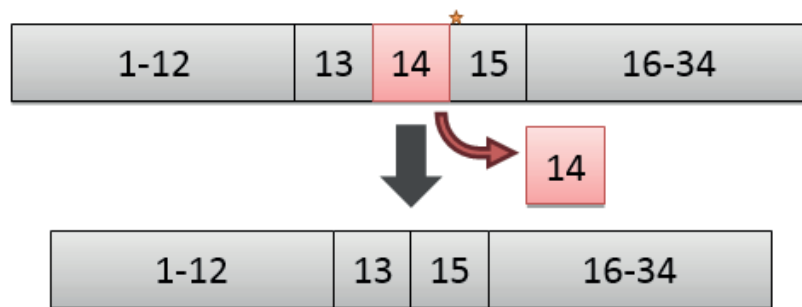

Supplementary Figure 1. Immature muscle fibers surrounded by overgrown collagen (asterix); magnification $\times 2500$



Supplementary Figure 2. Gene panel sequencing results of the proband visualized with IGV. COL6A1 c.1056+3A>C mutation.



Supplementary Figure 3. Visualization of the cDNA sequencing on IGV (Integrative Genomics Viewer).



Supplementary Figure 4. *COL6A1* cDNA – in frame deletion of the entire exon 14. Star indicates the localization of the c.1056+3A>C mutation.

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