# SNUPN deficiency causes a recessive muscular dystrophy due to RNA mis-splicing and ECM dysregulation

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**Description of Additional Supplementary Files**

**File Name:** Supplementary data 1

**Description:** Clinical description of all the patients carrying SNUPN mutations. Hom, Homozygote; Cmpd. Het., Compound Heterozygote; m1 to m9: mutations; ND, Not Determined; NA, Not Applicable; M, Male; F, Female; y, year(s); m, month(s); h, hour(s); %, percentage; +, mild; ++, moderate; +++, severe.

**File Name:** Supplementary Data 2

**Description:** In-silico analysis results of candidate SNUPN mutations in the patients with muscular dystrophy. The predicted level of pathogenicity using different tools is indicated as follows: 1) pathogenic (red) 2) medium (yellow) 3) benign (green). The conservation threshold varies according to the tool: GERP RS (Conserved: 4-6.18; Not conserved: -12.36-3.99); PhyloP100way Vertebrate (Conserved: ≥4; Not conserved: 0-3.99); PhastCons100way Vertebrate (Conserved: 1 ; Not conserved: 0). The summary is given based on the American College of Medical Genetics and Genomics (ACMG) system for variant classification (www.acmg.net).