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

Nashabat, Marwan ; Nabavizadeh, Nasrinsadat ; Saraçoğlu, Hilal Pırıl ; Sarıbaş, Burak ; Avcı, Şahin ; Börklü, Esra ; Beillard, Emmanuel ; Yılmaz, Elanur ; Uygur, Seyide Ecesu ; Kayhan, Cavit Kerem ; Bosco, Luca ; Eren, Zeynep Bengi ; Steindl, Katharina ; Richter, Manuela Friederike ; Bademci, Guney ; Rauch, Anita ; Fattahi, Zohreh ; Valentino, Maria Lucia ; Connolly, Anne M ; Bahr, Angela ; Viola, Laura ; Bergmann, Anke Katharina ; Rocha, Maria Eugenia ; Peart, LeShon ; Castro-Rojas, Derly Liseth ; Bültmann, Eva ; Khan, Suliman ; Giarrana, Miriam Liliana ; Teleanu, Raluca Ioana ; Stettner, Georg M ; et al

DOI: <https://doi.org/10.1038/s41467-024-45933-5>

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ZORA URL: <https://doi.org/10.5167/uzh-258319>

Journal Article

Supplemental Material



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Originally published at:

Nashabat, Marwan; Nabavizadeh, Nasrinsadat; Saraçoğlu, Hilal Pırıl; Sarıbaş, Burak; Avcı, Şahin; Börklü, Esra; Beillard, Emmanuel; Yılmaz, Elanur; Uygur, Seyide Ecesu; Kayhan, Cavit Kerem; Bosco, Luca; Eren, Zeynep Bengi; Steindl, Katharina; Richter, Manuela Friederike; Bademci, Guney; Rauch, Anita; Fattahi, Zohreh; Valentino, Maria Lucia; Connolly, Anne M; Bahr, Angela; Viola, Laura; Bergmann, Anke Katharina; Rocha, Maria Eugenia; Peart, LeShon; Castro-Rojas, Derly Liseth; Bültmann, Eva; Khan, Suliman; Giarrana, Miriam Liliana; Teleanu, Raluca Ioana; Stettner, Georg M; et al (2024). SNUPN deficiency causes a recessive muscular dystrophy due to RNA mis-splicing and ECM dysregulation. *Nature Communications*, 15(1758):1758.

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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).