

Curriculum Vitae

PERSONAL INFORMATION



Giulio Piluso

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Sex M | Date of birth 05/13/1961 | Nationality Italian

WORK EXPERIENCE

- 2017 – today **Associate Professor of Medical Genetics**
Università degli Studi della Campania “Luigi Vanvitelli”
▪ University and Research
- 2014 – 2017 **Researcher in Medical Genetics**
Seconda Università degli studi di Napoli
▪ University and Research
- 2001 – 2014 **Researcher in General Pathology**
Seconda Università degli Studi di Napoli
▪ University and Research
- 1999 - 2001 **Post-doctoral Fellowship**
Seconda Università degli Studi di Napoli
▪ Research
- 1997 - 1999 **TELETHON Research Fellowship**
Seconda Università degli Studi di Napoli
▪ Research
- 1996 - 1997 **Research Fellowship for young researchers by U.I.L.D.M.**
Seconda Università degli Studi di Napoli
▪ Research
- 1994 - 1996 **Post-graduate training**
Seconda Università degli Studi di Napoli
▪ Research

TEACHING ACTIVITIES

- 2018 – today C. di L. Magistrale in Medicina e Chirurgia; Università della Campania “L. Vanvitelli” (sede di Caserta), affidamento dell'insegnamento di Genetica Medica (24 ore; CFU 2.5) nel C.I. di Patologia e Fisiopatologia Generale e Genetica Medica
- 2018 – today C. di L. triennale in Ostetricia, Università degli Studi della Campania “L. Vanvitelli” (sede di Napoli), affidamento dell'insegnamento di Genetica Medica (15 ore).
- 2018 – 2020 C. di L. triennale in Biotecnologia, Università degli Studi della Campania “L. Vanvitelli”, affidamento dell'insegnamento di Genetica Medica (48 ore; CFU 6).
- 2016 – 2019 C. di L. triennale in Ostetricia, Università degli Studi della Campania “L. Vanvitelli” (sede di Caserta), affidamento dell'insegnamento di Genetica Medica (15 ore).
- 2014 – 2018 C. di L. Magistrale in Medicina e Chirurgia dell'Università della Campania “L. Vanvitelli” (sede di Caserta), affidamento dell'insegnamento di Genetica Medica (24 ore; CFU 2.5) nel C.I. di Patologia e Fisiopatologia Generale e Genetica Medica
- 2008 – 2012 C. di L. triennale in Ostetricia, Seconda Università degli Studi di Napoli (sede di Salerno), affidamento dell'insegnamento di Fisiopatologia e Patologia generale (38 ore).

2006 – 2015	C. di L. triennale in Ostetricia e C. di L. triennale in Infermieristica, Seconda Università degli Studi di Napoli (sede di Caserta), affidamento dell'insegnamento di Fisiopatologia e Patologia generale (38 ore).
2005 – 2013	C. di L. triennale in Tecniche di Laboratorio Biomedico, Seconda Università degli Studi di Napoli, affidamento dell'insegnamento di Tecnologie Ricombinanti (12 ore).
2005 – 2006	C. di L. triennale in Tecnico della Prevenzione nell'Ambiente e nei Luoghi di Lavoro, Seconda Università degli Studi di Napoli, affidamento dell'insegnamento di Genetica (12 ore).
2004 – 2012	C. di L. triennale in Fisioterapia, Seconda Università degli Studi di Napoli (sede di Salerno), affidamento dell'insegnamento di Fisiopatologia e Patologia generale (36 ore) - Anni Accademici: dal 2004-05 al 2011-12.
2003 – 2015	C. di L. triennale in Tecnica della riabilitazione psichiatrica, C. di L. triennale in Terapia della neuro e psicomotricità dell'età evolutiva, e C. di L. triennale in Ortottica e assistenza oftalmologica, Seconda Università degli Studi di Napoli, affidamento dell'insegnamento di Fisiopatologia e Patologia generale (38 ore).
2003 – 2015	C. di L. triennale in Logopedia, Seconda Università degli Studi di Napoli, affidamento dell'insegnamento di Patologia generale (15 ore).
2002 – 2012	C. di L. triennale in Ostetricia, Seconda Università degli Studi di Napoli (sede di Salerno), affidamento dell'insegnamento di Genetica Medica (15 ore).

CARE ACTIVITIES

2002 – today	<p>With the decree of inclusion in the welfare activities of the Policlinico University Company of the Second University of Naples, P.D. no. 126 of 12.02.2002, he was assigned to the welfare activities of the 1st Clinical Chemical Analysis Service, afferent to the Department of Laboratory Medicine of the AUP. As part of the assisting activities of the Laboratory Medicine Department, he performed molecular diagnostics in medical genetics in the field of muscular dystrophies, especially Duchenne/Becker muscular dystrophy, cingular muscular dystrophies, and other neuromuscular disorders. She has been working continuously since 2002 as Manager (1st level) and is currently working at the Laboratory of Medical Genetics, U.O.S.D. Medical Genetics and Karyomatology, D.A.I. of Internal Medicine, Geriatrics and Neurology, of the A.O.U. "Luigi Vanvitelli".</p> <p>He coordinates the activities of the Medical Genetics Laboratory for the molecular diagnostics of Neurofibromatosis Type 1, Legius Syndrome and RASopathies, including Next Generation Sequencing. He coordinates the analysis activities using CGH arrays. He is involved in the diagnosis of some X-linked mental retardation disorders, in particular FG syndrome.</p>
2016- oggi	Since April 2016, he also carries out his care activities as a manager within the Emergency Diagnostic Analysis Services, afferent to the O.U. of Clinical and Molecular Pathology.

EDUCATION AND TRAINING

1995 – 1998	Post-graduate course in Biochemistry and Clinical Chemistry Facoltà di Medicina e Chirurgia, Seconda Università degli Studi di Napoli
1994	Graduated in Life Sciences Facoltà di Scienze MM.FF.NN., Università degli Studi "Federico II" di Napoli

PERSONAL SKILLS

Mother tongue(s)	Italian
Other language(s)	English

Organisational / managerial skills	<p>Management of Resources and Working Groups.</p> <ul style="list-style-type: none"> ▪ 2023-2025 - Scientific Unit Lead in the research project "PRECISION DIAGNOSIS AND THERAPY FOR RARE DISEASES BY INTERPRETING NON-CODING GENOMES (PARADIGM)" PNRR Health funding (€1,000,000), unit share €244,495. ▪ 2019-Present - Scientific Co-Chair of the research project "Beyond the exome: dissecting the
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- missing heritability of Mendelian disease with high genetic heterogeneity (DisHetGeDPI)" funded by the University (325,000 €), within the VALERE program.
- 2018-22 - Participation in the project "Solve-RD - solving the unsolved rare diseases" funded by the European Commission, in which the University of Campania "L. Vanvitelli" participates with a research unit (<https://solve-rd.eu/the-group/consortium/university-of-naples/>).
 - 2012 - Scientific responsible for annual research project funded by the Neurofibromatosis Association (ANF onlus): total project cost € 22,000.
 - PRIN 2010-11 - Scientific responsible for three-year research project co-funded by MIUR: co-funding fee 117,600 €.
 - PRIN 2004 - Scientific head of two-year research project co-funded by MIUR: co-funding fee 30,000 €.

ADDITIONAL INFORMATION

Honours and awards

2000: 1st october. "L. Gioia" Prize of "Gaetano Conte Accademy" for the best post presentation at 5th Congress of Mediterranean Society of Myology.

Reviewer for international journals, for national and international fund calls, and participation to scientific committees

Reviewer for: Annals of Neurology, Acta Neuropatologica, Epilepsia; PLoS One, The Journal of Biomedical Science, Journal of Pediatric Genetics, Neuromuscular Disorders, DNA & Cell Biology, Cancer, Frontiers in Molecular Neuroscience, European Journal of Medical Genetics, American Journal of Medical Genetics, Scientific Reports.

External reviewer for: Agence Nationale de la Recherche (ANR, France), call 2019.

Member of the scientific committee of the International Congress "Neurofibromatosis type 1 and 2: from genetics to neurosurgery", Napoli 18th-19th october 2018.

Member of Scientific Societies

The American Society of Human Genetics (ASHG), European Society of Human Genetics (ESHG), World Muscle Society (WMS), Società Italiana di Genetica Umana (SIGU) and Associazione Italiana Miologia (AIM)

Communications to Congresses and Seminars as invited speaker

Verona, 24-27 settembre 2002 – 5° Congresso Nazionale SIGU (Società Italiana di Genetica Umana), Palazzo della Gran Guardia, Verona.

"La sindrome FG in una famiglia italiana: identificazione di un nuovo locus (FGS4) a Xp11.4-p11.3"

Napoli, 14 novembre 2002 – "La Patologia generale a Napoli a cento anni dalla nascita di Luigi Califano", Sala Conferenze della Facoltà di Medicina e Chirurgia, Seconda Università degli Studi di Napoli.

"La sindrome FG in una famiglia italiana: identificazione di un nuovo locus (FGS4) a Xp11.4-p11.3 e probabile ruolo del gene CASK" (*invited speaker*)

Roma, 31 gennaio 2006 – "Aggiornamenti in Genetica Clinica: la sindrome FG", Policlinico Gemelli (aula 617), Facoltà di Medicina e Chirurgia, Università Cattolica del Sacro Cuore, Roma.

Relazione dal titolo: "Analisi molecolare di una famiglia italiana con sindrome FG" (*su invito*)

Roma, 27 febbraio 2007 – "IX Incontro di Genetica Clinica", Sala Italia, Centro Congressi, Università Cattolica del Sacro Cuore, Roma.

"Il gene FGS4" (*invited speaker*)

Napoli, 16 maggio 2009 – "Il Giornata campana sulla Neurofibromatosi di tipo I", Sala Conferenze della Facoltà di Medicina e Chirurgia, Seconda Università degli Studi di Napoli.

"Genetica e diagnostica molecolare della Neurofibromatosi di tipo 1" (*invited speaker*)

Genova, 17-18 aprile 2010 – "V Congresso Nazionale sulle Neurofibromatosi", Santuario Madonna della Guardia, Genova.

"Neurofibromatosi di tipo 1: tecniche diagnostiche" (*invited speaker*)

Napoli, 17-22 luglio 2010 – XII International Congress on Neuromuscular Diseases, Centro Congressi "Federico II", Napoli.

"Motor Chip: a CGH microarray for neuromuscular disorders"

Roma, 30 ottobre 2011 – "Assemblea Associazione Neurofibromatosi", Istituto Salesiani del Sacro Cuore, Roma

"L'importanza del genetista in un Centro per la Neurofibromatosi tipo 1" (*invited speaker*)

Milano, 13-16 novembre 2011 – 14° Congresso Nazionale SIGU (Società Italiana di Genetica Umana), Centro Congressi MIC, Milano.

"Motor Chip: un CGH microarray per l'identificazione di mutazioni del numero di copie in 245 disordini neuromuscolari"

Napoli, 3-4 aprile 2014 – “Sindromi Genetiche e Tumori in Età Pediatrica”, Corso di Aggiornamento SIGU, Napoli

“Diagnosi molecolare differenziale in età pediatrica: Neurofibromatosi di tipo 1 vs Sindrome di Legius” (*invited speaker*)

Roma, 12-13 febbraio 2018 – “XX Incontro di Genetica Clinica”, Sala Brasca, Centro Congressi, Università Cattolica del Sacro cuore, Roma.

“Sindrome FG” (*invited speaker*)

Napoli, 18-19 ottobre 2018 – “Neurofibromatosis type 1 and 2: from genetics to neurosurgery”, Centro Congressi Complesso Monumentale Donnaregina, Napoli.

Relazione dal titolo: “NGS in molecular diagnosis of NF1” (*invited speaker*)

Napoli, 23-24 novembre 2018 – “La ricerca sulle malattie rare in Campania”, Auditorium Regione Campania, Napoli. Relazione dal titolo: “Il network campano per le RASopatie” (*invited speaker*)

Salerno, 19-20 maggio 2023 – “Patologie del Pathway RAS/MAPK: l’importanza delle rete multidisciplinare”, Grand Hotel Salerno, Salerno.

Relazione dal titolo: “Genetics of Neurofibromatosis type 1” (*invited speaker*)

Napoli, 14 ottobre 2023 – “Convegno nazionale ANF”, Clinica Pediatria, AOU Vanvitelli, Napoli.

Relazione dal titolo: “Analisi genetica nella neurofibromatosi 1,2,e 3: dalla diagnosi alle informazioni utili circa il fenotipo” (*invited speaker*)

Publications

Bibliometric indexes of Prof. Giulio Piluso are available on Scopus:

<https://www.scopus.com/authid/detail.uri?authorId=6603527210&origin=AuthorEval>

1. Caiazza M, Budillon A, Monda E, Aruta G, Esposito A, Del Vecchio Blanco F, Piluso G, Nigro V, Scarano G, Limongelli G. An atypical Aymé-Gripp phenotype detected by exome sequencing. *Am J Med Genet A*. 2023 Sep 15. doi:10.1002/ajmg.a.63406. Epub ahead of print. PMID: 37712597.
2. Morleo M, Venditti R, Theodorou E, Briere LC, Rosello M, Tirozzi A, Tammaro R, Al-Badri N, High FA, Shi J; Undiagnosed Diseases Network; Telethon Undiagnosed Diseases Program; Putti E, Ferrante L, Cetengizoglu V, Torella A, Walker MA, Tenconi R, Iascone M, Mei D, Guerrini R, van der Smagt J, Kroes HY, van Gassen KLI, Bilal M, Umair M, Pingault V, Attle-Bitach T, Amiel J, Ejaz R, Rodan L, Zollino M, Agrawal PB, Del Bene F, Nigro V, Sweetser DA, Franco B. De novo missense variants in phosphatidylinositol kinase PIPI5kly underlie a neurodevelopmental syndrome associated with altered phosphoinositide signaling. *Am J Hum Genet*. 2023 Aug 3;110(8):1377-1393. doi: 10.1016/j.ajhg.2023.06.012. Epub 2023 Jul 13. PMID: 37451268; PMCID: PMC10432144.
3. Torella A, Ricca I, Piluso G, Galatolo D, De Michele G, Zanobio M, Trovato R, De Michele G, Zeuli R, Pane C, Cocozza S, Saccà F, Santorelli FM, Nigro V, Filla A. A new genetic cause of spastic ataxia: the p.Glu415Lys variant in TUBA4A. *J Neurol*. 2023 Oct;270(10):5057-5063. doi: 10.1007/s00415-023-11816-w. Epub 2023 Jul 7. PMID: 37418012; PMCID: PMC10511369.
4. Torella A, Budillon A, Zanobio M, Del Vecchio Blanco F, Picollo E, Politano L, Nigro V, Piluso G. Alu-Mediated Insertions in the DMD Gene: A Difficult Puzzle to Interpret Clinically. *Int J Mol Sci*. 2023 May 25;24(11):9241. doi: 10.3390/ijms24119241. PMID: 37298193; PMCID: PMC10252317.
5. Capasso S, Cardiero G, Musollino G, Preziosi R, Testa R, Dembech S, Piluso G, Nigro V, Digilio FA, Lacerda G. Functional analysis of three new alpha-thalassemia deletions involving MCS-R2 reveals the presence of an additional enhancer element in the 5' boundary region. *PLoS Genet*. 2023 May 22;19(5):e1010727. doi: 10.1371/journal.pgen.1010727. PMID: 37216374; PMCID: PMC10202303.
7. Yaldis B, Kucuk E, Hampstead J, Hofste T, Pfundt R, Coroninas Galbany J, Rinne T, Yntema HG, Hoischen A, Nelen M, Gilissen C, Solve-RD consortium. Twist exome capture allows for lower average sequence coverage in clinical exome sequencing. *Hum Genomics*. 2023 May 3;17(1):39. doi: 10.1186/s40246-023-00485-5. PMID: 37138343; PMCID: PMC10155373.
8. Jackson A, Lin SJ, Jones EA, Chandler KE, Orr D, Moss C, Haider Z, Ryan G, Holden S, Harrison M, Burrows N, Jones WD, Loveless M, Petree C, Stewart H, Low K, Donnelly D, Lovell S, Drosou K; Genomics England Research Consortium; Solve-RD consortium; Varshey GK, Banks S. Clinical, genetic, epidemiologic, evolutionary, and functional delineation of TSPEAR-related autosomal recessive ectodermal dysplasia 14. *HGG Adv*. 2023 Mar 3;4(2):100186. doi: 10.1016/j.xhgg.2023.100186. PMID: 37009414; PMCID: PMC10064225.
9. Zucchini M, Capolongo G, Del Vecchio Blanco F, Secondulfo F, Gupta N, Blasio G, Pollastro RM, Cervesato A, Piluso G, Gigliotti G, Torella A, Nigro V, Perna AF, Capasso G, Trepiccione F. Next-Generation Sequencing (NGS) Analysis Illustrates the Phenotypic Variability of Collagen Type IV Nephropathies. *Genes (Basel)*. 2023 Mar 21;14(3):764. doi: 10.3390/genes14030764. PMID: 36981034; PMCID: PMC10048128.
10. Peduto C, Zanobio M, Nigro V, Perrotta S, Piluso G, Santoro C. Neurofibromatosis Type 1: Pediatric Aspects and Review of Genotype-Phenotype Correlations. *Cancers (Basel)*. 2023 Feb 14;15(4):1217. doi: 10.3390/cancers15041217. PMID: 36831560; PMCID: PMC954221.
11. Denommé-Pichon AS, Mataalonga L, de Boer E, Jackson A, Benetti E, Banks S, Bruel AL, Ciolfi A, Clayton-Smith J, Dallapiccola B, Duffourd Y, Ellwanger K, Fallerini C, Gilissen C, Graessner H, Haack TB, Havlovicova M, Hoischen A, Jean-Marcis N, Kleefstra T, López-Martín E, Macek M, Mencarelli MA, Mouton S, Pfundt R, Pizzi S, Posada M, Radio FC, Renieri A, Rooryck C, Ryba L, Safranow H, Schwarzbach M, Tartaglia M, Thauvin-Robinet C, Thevenon J, Tran Mau-Them F, Trimouille A, Votypka P, de Vries BBA, Willemse MH, Zurek B, Verloes A, Philippe C; Solve-RD DITF-ITHACA; Solve-RD SNV-indel Working Group; Solve-RD Consortia; Orphanomix Group; Vitobello A, Visser LELM, Faivre L. A Solve-RD ClinVar-based reanalysis of 1522 index cases from ERN-ITHACA reveals common pitfalls and misinterpretations in exome sequencing. *Genet Med*. 2023 Jan 20;25(4):100018. doi: 10.1016/j.gme.2023.100018. Epub ahead of print. PMID: 36681873.
12. Viggiani E, Picollo E, Passamano L, Onore ME, Piluso G, Scutifero M, Torella A, Nigro V, Politano L. Spectrum of Genetic Variants in the Dystrophin Gene: A Single Centre Retrospective Analysis of 750 Duchenne and Becker Patients from Southern Italy. *Genes (Basel)*. 2023 Jan 14;14(1):214. doi: 10.3390/genes14010214. PMID: 36672955; PMCID: QMC895256.
13. Patera R, Bettinaglio P, Borghesi A, Mangano E, Tritto V, Cesaretti C, Schettino C, Bordoni R, Santoro C, Avignone S, Moscatelli M, Melone MAB, Saletti V, Piluso G, Natacci F, Riva P, Eoli M. A Translational Approach to Spinal Neurofibromatosis: Clinical and Molecular Insights from a Wide Italian Cohort. *Cancers (Basel)*. 2022 Dec 22;15(1):59. doi: 10.3390/cancers15010059. PMID: 36612057; PMCID: PMC981775.
14. Santoro C, Boccià R, Iovino C, Piluso G, Perrotta S, Simonelli F. Patients carrying Arg1809 substitution with no choroidal abnormalities: a further proof of a "Quasi-Incomplete" NF1 phenotype. *Eur J Hum Genet*. 2022 Nov 21. doi: 10.1038/s41431-022-01236-1. Epub ahead of print. PMID: 36404348.
15. Peduto C, Piluso G, Nigro V, Brunetti-Pierri N. Are SHROOM4 loss-of-function variants pathogenic? *Am J Med Genet A*. 2022 Nov;188(11):3374-3375. doi: 10.1002/ajmg.a.62935. Epub 2022 Aug 16. PMID: 36209347.
16. Santoro C, Mironi G, Zanobio M, Ranucci G, D'Arriko A, Ciccalà D, Iascone M, Bernardo P, Piccolo V, Ronchi A, Limongelli G, Carotenuto M, Nigro V, Cinalli G, Piluso G. Mystery(n) Phenotypic Presentation in Europeans: Report of Three Further Novel Missense RNF213 Variants Leading to Severe Syndromic Forms of Moyamoya Angiopathy and Literature Review. *Int J Mol Sci*. 2022 Aug 11;23(16):8952. doi: 10.3390/ijms23168952. PMID: 36012218
17. Napolitano F, Dell'Aquila M, Terracciano C, Franzese G, Gentile MT, Piluso G, Santoro C, Colavito D, Patanè A, De Blasis P, Sampaolesi S, Melone MAB. Genotype-Phenotype Correlations in Neurofibromatosis Type 1: Identification of Novel and Recurrent NF1 Gene Variants and Correlations with Neurocognitive Phenotype. *Genes (Basel)*. 2022 Jun 23;13(7):1130. doi: 10.3390/genes13071130. PMID: 35885913
18. Tomabene P, Ferla R, Llado-Santaelaria M, Centruolo M, Dell'Anno M, Esposito F, Marrocco E, Pone E, Minopoli R, Iodice C, Nusco E, Rossi S, Lyubenova H, Manfredi A, Di Filippo L, Iuliano A, Torella A, Piluso G, Musacchia F, Surace EM, Cacchiaroli D, Nigro V, Auricchio A. Therapeutic homology-independent targeted integration in retina and liver. *Nat Commun*. 2022 Apr 12;13(1):1963. doi: 10.1038/s41467-022-29550-8. PMID: 35414130
19. Gicchino MF, Piluso G, Giugliano T, Cirillo M, Oliveri AN, Santoro C. Expanding the Neuroradiological Phenotype of 18q Deletion Syndrome. *Indian Pediatr*. 2021 Dec 15;58(12):1187-1188. PMID: 34939585.
20. Santoro C, Gaudino G, Torella A, Piluso G, Perrotta S, Miraglia Del Giudice E; Telethon Undiagnosed Diseases Program, Nigro V, Grandone A. Intermittent macrothrombocytopenia in a novel patient with Takenouchi-Kosaki syndrome and review of literature. *Eur J Med Genet*. 2021 Dec;64(12):104358. doi: 10.1016/j.ejmg.2021.104358. Epub 2021 Oct 5. PMID: 34624555.
21. D'Amico A, Rosano C, Pannone L, Pinna V, Assunto A, Motta M, Uggla L, Daniele P, Mandile R, Mariniello L, Siano MA, Santoro C, Piluso G, Martinelli S, Strisciuglio P, De Luca A, Tartaglia M, Melis D. Clinical variability of neurofibromatosis 1: A modifying role of cooccurring PTPN11 variants and atypical brain MRI findings. *Clin Genet*. 2021 Nov;100(5):563-572. doi: 10.1111/cge.14040. Epub 2021 Aug 17. PMID: 34346503.
22. Piccolo V, Russi T, Di Pinto D, Pota E, Di Martino M, Piluso G, Ronchi A, Argenziano G, Di Brizzi EV, Santoro C. Poikiloderma With Neutropenia and Mastocytosis: A Case Report and a Review of Dermatological Signs. *Front Med (Lausanne)*. 2021 Jun 10;8:680363. doi: 10.3389/fmed.2021.680363.

- PMID: 34179048; PMCID: PMC82222900.
23. Mataalonga L, Hernández-Ferrer C, Piscia D; Solve-RD SNV-indel working group, Schüle R, Synofzik M, Töpf A, Vissers LELM, de Voer R; Solve-RD DITF-GENTURIS; Solve-RD DITF-IITHACA; Solve-RD DITF-euroNMD; Solve-RD DITF-RND, Tonda R, Laurie S, Fernandez-Callejo M, Picó D, Garcia-Linares C, Papakonstantinou A, Corvá A, Joshi R, Diez H, Gut I, Hoischen A, Graessner H, Beltran S; Solve-RD Consortium. Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. *Eur J Hum Genet*. 2021 Sep;29(9):1337-1347. doi: 10.1038/s41431-021-00852-7. Epub 2021 Jun 1. Erratum in: *Eur J Hum Genet*. 2021 Aug 16;. PMID: 34075210; PMCID: PMC8440686.
 24. Töpf A, Pyle A, Griffin H, Mataalonga L, Schon K; Solve-RD SNV-indel working group; Solve-RD DITF-euroNMD, Sickmann A, Schara-Schmidt U, Hentschel A, Chinnery PF, Köbel H, Root A, Horvath R. Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). *Eur J Hum Genet*. 2021 Sep;29(9):1348-1353. doi:10.1038/s41431-021-00851-8. Epub 2021 Jun 1. PMID: 34075209; PMCID: PMC8440675.
 25. Zurek B, Ellwanger K, Vissers LELM, Schüle R, Synofzik M, Töpf A, de Voer RM, Laurie S, Mataalonga L, Gilissen C, Ossowski S, 't Hoen PAC, Vitobello A, Schulze-Hentrich JM, Riess O, Brunner HG, Brookes AJ, Rath A, Bonne G, Gummus G, Verloes A, Hoogerbrugge N, Evangelista T, Harmuth T, Swertz M, Spalding D, Hoischen A, Beltran S, Graessner H; Solve-RD consortium. Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. *Eur J Hum Genet*. 2021 Sep;29(9):1325-1331. doi: 10.1038/s41431-021-00859-0. Epub 2021 Jun 1. Erratum in: *Eur J Hum Genet*. 2021 Aug 13;. PMID: 34075208; PMCID: PMC8440542.
 26. Johari M, Sarparanta J, Viñola A, Jonson PH, Savarese M, Jokela M, Torella A, Piluso G, Said E, Vella N, Cauchi M, Magot A, Magri F, Mauri E, Kornblum C, Reimann J, Stojković T, Romero NB, Luquin H, Huovinen S, Lahermo P, Donner K, Comi GP, Nigro V, Hackman P, Udd B. Missense mutations in small muscle protein X-linked (SMPX) cause distal myopathy with protein inclusions. *Acta Neuropathol*. 2021 Aug;142(2):375-393. doi: 10.1007/s00401-021-02319-x. Epub 2021 May 11. PMID: 33974137; PMCID: PMC8270885.
 27. Schüle R, Timmann D, Erasmus CE, Reichbauer J, Wayand M; Solve-RD-DITF-RND, van de Warrenburg B, Schöls L, Wilke C, Bevot A, Zuchner S, Beltran S, Laurie S, Mataalonga L, Graessner H, Synofzik M; Solve-RD Consortium. Solving unsolved rare neurological diseases—a Solve-RD viewpoint. *Eur J Hum Genet*. 2021 Sep;29(9):1332-1336. doi: 10.1038/s41431-021-00901-1. Epub 2021 May 10. Erratum in: *Eur J Hum Genet*. 2021 Aug 25;. PMID: 33972714; PMCID: PMC8440537.
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