

Artículos científicos (publicados el 2º semestre 2022 y 2023).

1. [The gain of function SCN1A disorder spectrum: novel epilepsy phenotypes and therapeutic implications.](#)
Brunklaus A, Brünger T, Feng T, Fons C, Lehikoinen A, Panagiotakaki E, Vintan MA, Symonds J, Andrew J, Arzimanoglou A, Delima S, Gallois J, Hanrahan D, Lesca G, MacLeod S, Marjanovic D, McTague A, Nuñez-Enamorado N, Perez-Palma E, Scott Perry M, Pysden K, Russ-Hall SJ, Scheffer IE, Sully K, Syrbe S, Vaher U, Velayutham M, Vogt J, Weiss S, Wirrell E, Zuberi SM, Lal D, Møller RS, Mantegazza M, Cestèle S. *Brain*. 2022 Nov 21;145(11):3816-3831. doi:10.1093/brain/awac210.
2. [Genotype-phenotype correlations in valosin-containing protein disease: a retrospective multicentre study.](#)
Schiava M, Ikenaga C, Villar-Quiles RN, Caballero-Ávila M, Topf A, Nishino I, Kimonis V, Udd B, Schoser B, Zanoteli E, Souza PVS, Tasca G, Lloyd T, Lopez-de Munain A, Paradas C, Pegoraro E, Nadaj-Pakleza A, De Bleecker J, Badrising U, Alonso-Jiménez A, Kostera-Pruszczyk A, Miralles F, Shin JH, Bevilacqua JA, Olivé M, Vorgerd M, Kley R, Brady S, Williams T, Domínguez-González C, Papadimas GK, Warman-Chardon J, Claeys KG, de Visser M, Muelas N, LaForet P, Malfatti E, Alfano LN, Nair SS, Manousakis G, Kushlaf HA, Harms MB, Nance C, Ramos-Fransi A, Rodolico C, Hewamadduma C, Cetin H, García-García J, Pál E, Farrugia ME, Lamont PJ, Quinn C, Nedkova-Hristova V, Peric S, Luo S, Oldfors A, Taylor K, Ralston S, Stojkovic T, Wehl C, Diaz-Manera J; VCP International Study Group; VCP International Study Group. *J Neurol Neurosurg Psychiatry*. 2022 Jul 27;jnnp-2022-328921. doi: 10.1136/jnnp-2022-328921.
3. [Mutation update for the ACTN2 gene.](#)
Ranta-Aho J, Olive M, Vandroux M, Roticiiani G, Dominguez C, Johari M, Torella A, Böhm J, Turon J, Nigro V, Hackman P, Laporte J, Udd B, Savarese M. *Hum Mutat*. 2022 Dec;43(12):1745-1756. doi: 10.1002/humu.24470
4. [Mucormycosis in COVID-19: The Indian scenario.](#)
Ghosh R, Roy D, Benito-León J.

J Mycol Med. 2022 Aug;32(3):101275. doi:
10.1016/j.mycmed.2022.101275. Epub 2022 Mar 23.

5. [Safety and Outcome of Revascularization Treatment in Patients With Acute Ischemic Stroke and COVID-19: The Global COVID-19 Stroke Registry.](#)

Marto JP, Strambo D, Ntaios G, Nguyen TN, Herzig R, Czlonkowska A, Demeestere J, Mansour OY, Salerno A, Wegener S, Baumgartner P, Cereda CW, Bianco G, Beyeler M, Arnold M, Carrera E, Machi P, Altersberger V, Bonati L, Gensicke H, Bolognese M, Peters N, Wetzel S, Magriço M, Ramos JN, Sargento-Freitas J, Machado R, Maia C, Machado E, Nunes AP, Ferreira P, Pinho E Melo T, Dias MC, Paula A, Correia MA, Castro P, Azevedo E, Albuquerque L, Alves JN, Ferreira-Pinto J, Meira T, Pereira L, Rodrigues M, Araujo AP, Rodrigues M, Rocha M, Pereira-Fonseca Â, Ribeiro L, Varela R, Malheiro S, Cappellari M, Zivelonghi C, Sajeva G, Zini A, Gentile M, Forlivesi S, Migliaccio L, Sessa M, La Gioia S, Pezzini A, Sangalli D, Zedde M, Pascarella R, Ferrarese C, Beretta S, Diamanti S, Schwarz G, Frisullo G, Marcheselli S, Seners P, Sabben C, Escalard S, Piotin M, Maïer B, Charbonnier G, Vuillier F, Legris L, Cuisenier P, Vodret FR, Marnat G, Liegey JS, Sibon I, Flottmann F, Broocks G, Gloyer NO, Bohmann FO, Schaefer JH, Nolte C, Audebert HJ, Siebert E, Sykora M, Lang W, Ferrari J, Mayer-Suess L, Knoflach M, Gizewski ER, Stolp J, Stolze LJ, Coutinho JM, Nederkoorn P, van den Wijngaard I, De Meris J, Lemmens R, De Raedt S, Vandervorst F, Rutgers MP, Guilmot A, Dusart A, Bellante F, Calleja-Castaño P, Ostos F, González-Ortega G, Martín-Jiménez P, García-Madrona S, Cruz-Culebras A, Vera R, Matute MC, Fuentes B, Alonso-de-Leciñana M, Rigual R, Díez-Tejedor E, Perez-Sanchez S, Montaner J, Díaz-Otero F, Pérez-de-la-Ossa N, Flores-Pina B, Muñoz-Narbona L, Chamorro A, Rodríguez-Vázquez A, Renú A, Ayo-Martin O, Hernández-Fernández F, Segura T, Tejada-Meza H, Sagarra-Mur D, Serrano-Ponz M, Hlaing T, See I, Simister R, Werring D, Kristoffersen ES, Nordanstig A, Jood K, Rentzos A, Šimůnek L, Krajíčková D, Krajina A, Mikulik R, Cviková M, Vinklársek J, Školoudík D, Roubec M, Hurtikova E, Hrubý R, Ostry S, Skoda O, Pernicka M, Jurak L, Eichlová Z, Jíra M, Kovar M, Panský M, Menci P, Palouskova H, Tomek A, Janský P, Olšerová A, Sramek M, Havlicek R, Malý P, Trakal L, Fiksa J, Slovák M, Karlinski MA, Nowak M, Sienkiewicz-Jarosz H, Bochynska A, Wrona P, Homa T, Sawczynska K, Slowik A, Wlodarczyk E, Wiacek M, Tomaszewska-Lampart I, Sieczkowski B, Bartosik-Psujek H, Bilik M, Bandzarewicz A, Dorobek M, Zielinska-Turek J, Nowakowska-

Kotas M, Obara K, Urbanowski P, Budrewicz S, Guziński M, Świtońska M, Rutkowska I, Sobieszak-Skura P, Labuz-Roszak BM, Debiec A, Staszewski J, Stępień A, Zwiernik J, Wasilewski G, Tiu C, Terecoasă EO, Radu RA, Negrila A, Dorobat B, Panea C, Tiu V, Petrescu S, Ozdemir A, Mahmoud M, El-Samahy H, Abdelkhalek H, Al-Hashel J, Ismail II, Salmeen A, Ghoreishi A, Sabetay SI, Gross H, Klein P, Abdalkader M, Jabbour P, El Naamani K, Tjoumakaris S, Abbas R, Mohamed GA, Chebl A, Min J, Hovingh M, Tsai JP, Khan M, Nalleballe K, Onteddu S, Masoud H, Michael M, Kaur N, Maali L, Abraham MG, Khandelwal P, Bach I, Ong M, Babici D, Khawaja AM, Hakemi M, Rajamani K, Cano-Nigenda V, Arauz A, Amaya P, Llanos N, Arango A, Vences MÁ, Barrientos Guerra JD, Caetano R, Martins RT, Scollo SD, Yalung PM, Nagendra S, Gaikwad A, Seo KD, Georgiopoulos G, Nogueira RG, Michel P; Global COVID-19 Stroke Registry.

Neurology. 2023 Feb 14;100(7):e739-e750. doi: 10.1212/WNL.0000000000201537.

6. [Isolated Nonreactive Mydriasis in Herpes Zoster Ophthalmicus: An Uncommon Complication.](#)

Blanco-Palmero VA, Ortueta-Olartecoechea A, Uriarte-Pérez de Urabayen D, Sánchez-Tornero M, Méndez-Guerrero A, Matarazzo M.

J Neuroophthalmol. 2022 Jun 1;42(2):e508-e510. doi: 10.1097/WNO.0000000000001289.

7. [Advances in Thymidine Kinase 2 Deficiency: Clinical Aspects, Translational Progress, and Emerging Therapies.](#)

Berardo A, Domínguez-González C, Engelstad K, Hirano M. J Neuromuscul Dis. 2022;9(2):225-235. doi: 10.3233/JND-210786.

8. [Mechanical Thrombectomy for Acute Stroke in a 2-Month-Old Patient and Review of the Literature in Infancy.](#)

Rodríguez-López A, Gimeno I, Vila S, Seoane A, Ballenilla F, Ramos A, Hilaro A, Núñez N, Camacho A, Simón R.

Neuropediatrics. 2022 Oct;53(5):376-380. doi: 10.1055/a-1844-8780

9. [Impact of COVID-19 in Immunosuppressed Children With Neuroimmunologic Disorders.](#)

Olivé-Cirera G, Fonseca E, Cantarín-Extremera V, Vázquez-López M, Jiménez-Legido M, González-Álvarez V, Ribeiro-Constante J, Camacho-Salas A, Martí I, Cancho-Candela R, Martínez-González MJ, Saiz A, Armangué T; Spanish Study Group NeuroimmunoPed-Covid.

Neurol Neuroimmunol Neuroinflamm. 2021 Nov 10;9(1):e1101.
doi: 10.1212/NXI.0000000000001101.

10. [Clinical trials in pediatric ALS: a TRICALS feasibility study.](#)
Kliest T, Van Eijk RPA, Al-Chalabi A, Albanese A, Andersen PM, Amador MDM, BrÅthen G, Brunaud-Danel V, Brylev L, Camu W, De Carvalho M, Cereda C, Cetin H, Chaverri D, Chiò A, Corcia P, Couratier P, De Marchi F, Desnuelle C, Van Es MA, Esteban J, Filosto M, García Redondo A, Grosskreutz J, Hanemann CO, Holmøy T, Høyer H, Ingre C, Koritnik B, Kuzma-Kozakiewicz M, Lambert T, Leigh PN, Lunetta C, Mandrioli J, Mcdermott CJ, Meyer T, Mora JS, Petri S, Povedano M, Reviere E, Riva N, Roes KCB, Rubio MÁ, Salachas F, Sarafov S, Sorarù G, Stevic Z, Svenstrup K, Møller AT, Turner MR, Van Damme P, Van Leeuwen LAG, Varona L, Vázquez Costa JF, Weber M, Hardiman O, Van Den Berg LH.
Amyotroph Lateral Scler Frontotemporal Degener. 2022 Nov;23(7-8):481-488. doi: 10.1080/21678421.2021.2024856.
11. [Muscle MRI characteristic pattern for late-onset TK2 deficiency diagnosis.](#)
Domínguez-González C, Fernández-Torrón R, Moore U, de Fuenmayor-Fernández de la Hoz CP, Vélez-Gómez B, Cabezas JA, Alonso-Pérez J, González-Mera L, Olivé M, García-García J, Moris G, León Hernández JC, Muelas N, Servian-Morilla E, Martín MA, Díaz-Manera J, Paradas C.
J Neurol. 2022 Jul;269(7):3550-3562. doi: 10.1007/s00415-021-10957-0.
12. ['Wine Glass' sign following COVID-19 vaccination in a previously healthy adult.](#)
Ghosh R, Dutta S, Ghosh M, Benito-León J.
Neurologia. 2022 Nov-Dec;37(9):820-823. doi: 10.1016/j.nrl.2022.01.003.
13. [Multiple Myeloma-Induced Hyperammonemic Encephalopathy: Another Important Potential Cause of Triphasic Waves.](#)
León-Ruiz M, Oliva-Navarro J, Alonso-Singer P, Castañeda-Cabrero C, Benito-León J.
J Clin Neurophysiol. 2022 Nov 1;39(7):647. doi: 10.1097/WNP.0000000000000959.
14. [Alien Limb Phenomenon After Diffuse Corpus Callosum Ischemic Stroke.](#)

Sarkar P, Ray BK, Mukherjee D, Pandit A, Ghosh R, Benito-León J, Dubey S.
Neurohospitalist. 2022 Apr;12(2):295-300. doi:
10.1177/19418744211067033.

15. [Onset pattern of nigrostriatal denervation in early Parkinson's disease.](#)
Pineda-Pardo JA, Sánchez-Ferro Á, Monje MHG, Pavese N, Obeso JA.
Brain. 2022 Apr 29;145(3):1018-1028. doi:
10.1093/brain/awab378.
16. [Quality of Life and the Experience of Living with Early-Stage Alzheimer's Disease.](#)
Villarejo-Galende A, García-Arcelay E, Piñol-Ripoll G, Del Olmo-Rodríguez A, Viñuela F, Boada M, Franco-Macías E, Ibañez de la Peña A, Riverol M, Puig-Pijoan A, Abizanda-Soler P, Arroyo R, Baquero-Toledo M, Fera-Vilar I, Balasa M, Berbel Á, Rodríguez-Rodríguez E, Vieira-Campos A, García-Ribas G, Rodrigo-Herrero S, Terrance Á, Prefasi D, Lleó A, Maurino J.
J Alzheimers Dis. 2022;90(2):719-726. doi: 10.3233/JAD-220696.
17. [Adult-onset nemaline myopathy due to a novel homozygous variant in the TNNT1 gene.](#)
Martín-Jiménez P, Fuenmayor-Fernández de la Hoz CP, Hernández-Laín A, Arteche-López A, Quesada-Espinosa JF, Voth AH, Vesperinas A, Olivé M, Domínguez-González C.
Muscle Nerve. 2022 Oct;66(4):E13-E15. doi: 10.1002/mus.27678.
18. [Rapid Molecular Diagnosis of Genetically Inherited Neuromuscular Disorders Using Next-Generation Sequencing Technologies.](#)
Barbosa-Gouveia S, Vázquez-Mosquera ME, González-Vioque E, Hermida-Ameijeiras Á, Sánchez-Pintos P, de Castro MJ, León SR, Gil-Fournier B, Domínguez-González C, Camacho Salas A, Negrão L, Fineza I, Laranjeira F, Couce ML.
J Clin Med. 2022 May 12;11(10):2750. doi:
10.3390/jcm11102750.
19. [BNIP3 Is Involved in Muscle Fiber Atrophy in Late-Onset Pompe Disease Patients.](#)
Carrasco-Rozas A, Fernández-Simón E, Suárez-Calvet X, Piñol-Jurado P, Alonso-Pérez J, de Luna N, Schoser B, Meinke P, Domínguez-González C, Hernández-Laín A, Paradas C, Rivas E, Illa I, Olivé M, Gallardo E, Díaz-Manera J.
Am J Pathol. 2022 Aug;192(8):1151-1166. doi:
10.1016/j.ajpath.2022.05.003.

20. [Muscle MRI in McArdle Disease: A European Multicenter Observational Study.](#)
Løkken N, Revsbech KL, Jacobsen LN, Martinuzzi A, Martin MÁ, Díaz-Manera J, Dominguez-Gonzalez C, Brondani G, Musumeci O, Granata F, Stefan C, Merino-Sanchez C, Peralta CN, Khawajazada T, Alonso-Pérez J, Toscano A, Vissing J.
Neurology. 2022 Jul 19;10.1212/WNL.0000000000200914. doi: 10.1212/WNL.0000000000200914.
21. [Survey on the management of Pompe disease in routine clinical practice in Spain.](#)
Domínguez-González C, Díaz-Marín C, Juntas-Morales R, Nacimiento-Osorio A, Rivera-Gallego A, Díaz-Manera J.
Orphanet J Rare Dis. 2022 Dec 5;17(1):426. doi: 10.1186/s13023-022-02574-5.
22. [Genetic diagnosis of Duchenne and Becker muscular dystrophy through mRNA analysis: new splicing events.](#)
Segarra-Casas A, Domínguez-González C, Hernández-Laín A, Sanchez-Calvin MT, Camacho A, Rivas E, Campo-Barasoain A, Madruga M, Orteiz C, Natera-de Benito D, Nascimento A, Codina A, Rodriguez MJ, Gallano P, Gonzalez-Quereda L.
J Med Genet. 2022 Dec 19:jmg-2022-108828. doi: 10.1136/jmg-2022-108828.
23. [COVID19-associated new-onset movement disorders: a follow-up study.](#)
Schneider SA, Desai S, Phokaewvarangkul O, Rosca EC, Sringean J, Anand P, Bravo GÁ, Cardoso F, Cervantes-Arslanian AM, Chovatiya H, Crosiers D, Dijkstra F, Fearon C, Grandas F, Guedj E, Méndez-Guerrero A, Hassan M, Jankovic J, Lang AE, Makhoul K, Muccioli L, O'Shea SA, Ostovan VR, Perez-Sanchez JR, Ramdhani R, Ros-Castelló V, Schulte C, Shah P, Wojtecki L, Pal PK.
J Neurol. 2023 Mar 21:1-7. doi: 10.1007/s00415-023-11661-x.
24. [Differentially Aquaporin 5 Expression in Submandibular Glands and Cerebral Cortex in Alzheimer's Disease.](#)
Antequera D, Carrero L, Cunha Alves V, Ferrer I, Hernández-Gallego J, Municio C, Carro E.
Biomedicines. 2022 Jul 8;10(7):1645. doi: 10.3390/biomedicines10071645.
25. [Differences in Striatal Metabolism in \[¹⁸F\]FDG PET in Parkinson's Disease and Atypical Parkinsonism.](#)

- Seiffert AP, Gómez-Grande A, Alonso-Gómez L, Méndez-Guerrero A, Villarejo-Galende A, Gómez EJ, Sánchez-González P.
Diagnostics (Basel). 2022 Dec 20;13(1):6. doi: 10.3390/diagnostics13010006.
26. [Does Your Loved One With Cognitive Symptoms Need to See a Doctor? Check It Online.](#)
Agüera-Ortiz L, Martín-Carrasco M, Arriola-Manchola E, Martínez-Lage P, Pérez-Martínez DA, Ojea T, Soler-López B, García-Ribas G.
Front Comput Neurosci. 2022 Jul 14;16:840200. doi: 10.3389/fncom.2022.840200. eCollection 2022.
27. [Rare neurological and neuropsychiatric manifestations of scrub typhus: a case series of 10 cases.](#)
Ghosh R, Mandal A, León-Ruiz M, Roy D, Das S, Dubey S, Benito-León J.
Neurologia (Engl Ed). 2022 Jul 28:S2173-5808(22)00081-5. doi: 10.1016/j.nrleng.2022.07.001.
28. [Awareness of Diagnosis in Persons with Early-Stage Alzheimer's Disease: An Observational Study in Spain.](#)
Villarejo-Galende A, García-Arcelay E, Piñol-Ripoll G, Del Olmo-Rodríguez A, Viñuela F, Boada M, Franco-Macías E, de la Peña AI, Riverol M, Puig-Pijoan A, Abizanda-Soler P, Arroyo R, Baquero-Toledo M, Fera-Vilar I, Balasa M, Berbel Á, Rodríguez-Rodríguez E, Vieira-Campos A, García-Ribas G, Rodrigo-Herrero S, Lleó A, Maurino J.
Neurol Ther. 2022 Sep;11(3):1183-1192. doi: 10.1007/s40120-022-00367-3.
29. [A novel MLIP truncating variant in an 80-year-old patient with late-onset progressive weakness.](#)
Bermejo-Guerrero L, Arteche-López A, de Fuenmayor Fernández de la Hoz C, Hernández-Laín A, Martín MA, Domínguez-González C.
Brain. 2022 Oct 21;145(10):e99-e102. doi: 10.1093/brain/awac286.
30. [Hereditary cerebral small vessel disease: Assessment of a HTRA1 variant using protein stability predictors and 3D modelling.](#)
Hidalgo Mayoral I, Martínez-Salio A, Llamas-Velasco S, Gómez-Majón I, Arteche-López A, Quesada-Espinosa JF, Palma Milla C, Lezana Rosales JM, Pérez de la Fuente R, Juárez Rufián A, Sierra Tomillo O, Sánchez Calvín MT, Gómez Rodríguez MJ,

Ramos Gómez P, Villarejo-Galende A, Díaz-Guzmán J, Ortega-Casarrubios MÁ, Calleja-Castaño P, Moreno-García M.
Eur J Med Genet. 2022 Aug;65(8):104539. doi:
10.1016/j.ejmg.2022.104539.

31. [Toward an Automatic Assessment of Cognitive Dysfunction in Relapsing-Remitting Multiple Sclerosis Patients Using Eye Movement Analysis.](#)
García Cena CE, Gómez-Andrés D, Pulido-Valdeolivas I, Sánchez-Seco VG, Domingo-Santos A, Moreno-García S, Benito-León J.
Sensors (Basel). 2022 Oct 27;22(21):8220. doi:
10.3390/s22218220.
32. [The effect of timing of high-efficacy therapy on processing speed performance in multiple sclerosis.](#)
Labiano-Fontcuberta A, Costa-Frossard L, Sainz de la Maza S, Rodríguez-Jorge F, Chico-García JL, Monreal E.
Mult Scler Relat Disord. 2022 Aug;64:103959. doi:
10.1016/j.msard.2022.103959.
33. [Acute onset movement disorders in diabetes mellitus: A clinical series of 59 patients.](#)
Dubey S, Chatterjee S, Ghosh R, Louis ED, Hazra A, Sengupta S, Das S, Banerjee A, Pandit A, Ray BK, Benito-León J.
Eur J Neurol. 2022 Aug;29(8):2241-2248. doi:
10.1111/ene.15353.
34. [Metrics of progression and prognosis in untreated adults with thymidine kinase 2 deficiency: An observational study.](#)
Domínguez-González C, Hernández-Voth A, de Fuenmayor-Fernández de la Hoz CP, Guerrero LB, Morís G, García-García J, Muelas N, León Hernández JC, Rabasa M, Lora D, Blázquez A, Arenas J, Martín MÁ.
Neuromuscul Disord. 2022 Sep;32(9):728-735. doi:
10.1016/j.nmd.2022.07.399.
35. [Characterizing SOD1 mutations in Spain. The impact of genotype, age, and sex in the natural history of the disease.](#)
Vázquez-Costa JF, Borrego-Hernández D, Paradas C, Gómez-Caravaca MT, Rojas-García R, Varona L, Povedano M, García-Sobrino T, Jericó Pascual I, Gutiérrez A, Riancho J, Turon-Sans J, Assialioui A, Pérez-Tur J, Sevilla T, Esteban Pérez J, García-Redondo A; ALSGESCO.
Eur J Neurol. 2022 Dec 9. doi: 10.1111/ene.15661.

36. [Elevated glutamate and decreased glutamine levels in the cerebrospinal fluid of patients with MELAS syndrome.](#)
Guerrero-Molina MP, Morales-Conejo M, Delmiro A, Morán M, Domínguez-González C, Arranz-Canales E, Ramos-González A, Arenas J, Martín MA, González de la Aleja J.
J Neurol. 2022 Jun;269(6):3238-3248. doi: 10.1007/s00415-021-10942-7.
37. [Comparison of the Results of a Parkinson's Holter Monitor With Patient Diaries, in Real Conditions of Use: A Sub-analysis of the MoMoPa-EC Clinical Trial.](#)
Pérez-López C, Hernández-Vara J, Caballol N, Bayes À, Buongiorno M, Lopez-Ariztegui N, Gironell A, López-Sánchez J, Martínez-Castrillo JC, Saucó M A, López-Manzanares L, Escalante-Arroyo S, Pérez-Martínez DA, Rodríguez-Molinero A; MoMoPa-EC Research Group.
Front Neurol. 2022 May 16;13:835249. doi: 10.3389/fneur.2022.835249. eCollection 2022.
38. [Heterozygous and Homozygous Variants in SORL1 Gene in Alzheimer's Disease Patients: Clinical, Neuroimaging and Neuropathological Findings.](#)
Alvarez-Mora MI, Blanco-Palmero VA, Quesada-Espinosa JF, Arteche-Lopez AR, Llamas-Velasco S, Palma Milla C, Lezana Rosales JM, Gomez-Manjon I, Hernandez-Lain A, Jimenez Almonacid J, Gil-Fournier B, Ramiro-León S, González-Sánchez M, Herrero-San Martín AO, Pérez-Martínez DA, Gómez-Tortosa E, Carro E, Bartolomé F, Gomez-Rodriguez MJ, Sanchez-Calvin MT, Villarejo-Galende A, Moreno-Garcia M.
Int J Mol Sci. 2022 Apr 11;23(8):4230. doi: 10.3390/ijms23084230.
39. [Cardiovascular risk in subjects over 55 years of age and cognitive performance after five years. NEDICES2-RISK study. Study protocol.](#)
Tapias-Merino E, De Hoyos-Alonso MDC, Contador-Castillo I, Rodríguez-Sánchez E, Sanz-Cuesta T, Becerro-Muñoz CM, Hernández-Gallego J, Vega-Quiroga S, Bermejo-Pareja F; NEDICES2-RISK Group.
PLoS One. 2022 Nov 28;17(11):e0274589. doi: 10.1371/journal.pone.0274589. eCollection 2022.
40. [Postural misperception: a biomarker for persistent postural perceptual dizziness.](#)
San Pedro Murillo E, Bancroft MJ, Koohi N, Castro P, Kaski D.

J Neurol Neurosurg Psychiatry. 2023 Feb;94(2):165-166. doi: 10.1136/jnnp-2022-329321.

41. [Therapeutic Decision-Making Under Uncertainty in the Management of Spinal Muscular Atrophy: Results From DECISIONS-SMA Study.](#)
Saposnik G, Camacho A, Díaz-Abós P, Brañas-Pampillón M, Sánchez-Menéndez V, Cabello-Moruno R, Terzaghi M, Maurino J, Málaga I.
Neurol Ther. 2022 Sep;11(3):1209-1219. doi: 10.1007/s40120-022-00366-4.
42. [Home calligraphic exercises as manual dexterity training in patients with Parkinson's disease: a pilot feasibility study.](#)
Llamas-Velasco S, Romero Ferreiro C, González Fuertes Á, García Tell P, Blanco-Palmero VA, Martín-Jimenez P, Pérez Martínez DA, Méndez-Guerrero A.
Disabil Rehabil. 2023 Feb 23:1-5. doi: 10.1080/09638288.2023.2181408.
43. [Loss of function variants in DNAJB4 cause a myopathy with early respiratory failure.](#)
Weihl CC, Töpf A, Bengoechea R, Duff J, Charlton R, Garcia SK, Domínguez-González C, Alsaman A, Hernández-Laín A, Franco LV, Sanchez MEP, Beecroft SJ, Goulee H, Daw J, Bhadra A, True H, Inoue M, Findlay AR, Laing N, Olivé M, Ravenscroft G, Straub V.
Acta Neuropathol. 2023 Jan;145(1):127-143. doi: 10.1007/s00401-022-02510-8.
44. [The Effects of SARS-CoV-2 Infection on the Cognitive Functioning of Patients with Pre-Existing Dementia.](#)
Dubey S, Das S, Ghosh R, Dubey MJ, Chakraborty AP, Roy D, Das G, Dutta A, Santra A, Sengupta S, Benito-León J.
J Alzheimers Dis Rep. 2023 Feb 14;7(1):119-128. doi: 10.3233/ADR-220090.
45. [New Cerebellar Ataxia, Neuropathy, Vestibular Areflexia Syndrome cases are caused by the presence of a nonsense variant in compound heterozygosity with the pathogenic repeat expansion in the RFC1 gene.](#)
Arteche-López A, Avila-Fernandez A, Damian A, Soengas-Gonda E, de la Fuente RP, Gómez PR, Merlo JG, Burgos LH, Fernández CC, Rosales JML, Martínez JFG, Quesada-Espinosa JF, Corton M, Guerrero-Molina MP.

- Clin Genet. 2023 Feb;103(2):236-241. doi: 10.1111/cge.14249.
46. [Myosin post-translational modifications and function in the presence of myopathy-linked truncating MYH2 mutations.](#)
Sonne A, Peverelli L, Hernandez-Lain A, Domínguez-González C, Andersen JL, Milone M, Beggs AH, Ochala J.
Am J Physiol Cell Physiol. 2023 Mar 1;324(3):C769-C776. doi: 10.1152/ajpcell.00002.2023.
47. [Anoctamin-5 related muscle disease: Clinical and genetic findings in a large European cohort.](#)
de Bruyn A, Montagnese F, Holm-Yildiz S, Poulsen NS, Stojkovic T, Behin A, Palmio J, Jokela M, De Bleecker JL, de Visser M, van der Kooi AJ, Ten Dam L, Domínguez González C, Maggi L, Gallone A, Kostera-Pruszczyk A, Macias A, Łusakowska A, Nedkova V, Olive M, Álvarez-Velasco R, Wanschitz J, Paradas C, Mavillard F, Querin G, Fernández-Eulate G, Quinlivan R, Walter MC, Depuydt CE, Udd B, Vissing J, Schoser B, Claeys KG.
Brain. 2023 Mar 13:awad088. doi: 10.1093/brain/awad088.
48. [Impact of a specific consultation for patients with progressive forms of multiple sclerosis on the response to their unmet care needs: a cross-sectional study.](#)
Gómez-López A, Benito-León J, Labiano-Fontcuberta A, Moreno-García S, Salgado-Cámara P.
Mult Scler Relat Disord. 2023 Mar 7;72:104609. doi: 10.1016/j.msard.2023.104609.
49. [Static first-minute-frame \(FMF\) PET imaging after ¹⁸F-labeled amyloid tracer injection is correlated to \[¹⁸F\]FDG PET in patients with primary progressive aphasia.](#)
Gómez-Grande A, Seiffert AP, Villarejo-Galende A, González-Sánchez M, Llamas-Velasco S, Bueno H, Gómez EJ, Tabuenca MJ, Sánchez-González P.
Rev Esp Med Nucl Imagen Mol (Engl Ed). 2023 Feb 7:S2253-8089(23)00014-9. doi: 10.1016/j.remnie.2023.02.001.
50. [High-dose oral glutamine supplementation reduces elevated glutamate levels in cerebrospinal fluid in patients with mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes syndrome.](#)
Guerrero-Molina MP, Morales-Conejo M, Delmiro A, Morán M, Domínguez-González C, Arranz-Canales E, Ramos-González A, Arenas J, Martín MA, de la Aleja JG.
Eur J Neurol. 2023 Feb;30(2):538-547. doi: 10.1111/ene.15626.

