

anormales⁴¹⁸. Como se ilustra en la [figura 25B](#), la lengua aparece extruida medialmente con los dedos índice y pulgar adyacentes a ésta, lo cual explica las peculiaridades de este síndrome lacunar aquí descrito.

La última observación de la que nos ocuparemos tiene un interés congresual y presencial histórico, que ni el Dr Polo Esteban ni este autor olvidarán jamás. Una mujer de 37 años en su 23 semana de gestación ingresó por un cuadro de visión borrosa y debilidad generalizada. En la exploración había una oftalmoplejía externa y paresia facial bilateral, voz nasal, disartria, y tetraparesia³¹⁴. La evaluación obstétrica fue normal. La madre de la paciente estaba afectada; el brote de botulismo fue desencadenado por una conserva casera de judía verde; tiene interés que todas las gallinas de su gallinero alimentadas con este producto contaminado hubieran muerto. No se identificó el tipo de *Clostridium botulinum* responsable. La estimulación repetitiva a 50 Hz demostró una potenciación del 70%. Se administraron 5 viales de antitoxina botulínica AB. La debilidad progresó hasta la tetraplejía requiriendo soporte ventilatorio; los movimientos fetales eran entonces los únicos visibles. En este periodo las ecografías abdominales sucesivas demostraron un desarrollo fetal normal. Fue dada de alta a los tres meses del inicio sintomático con mínima debilidad residual. El parto fue eutócico, con un bebé normal. Así, pues, el botulismo en esta paciente no tuvo ninguna repercusión negativa en el embarazo, lo cual apoya la decisión de tratar a la mujer embarazada con BoNT/A, cuando sea necesario, al menos en la segunda parte del embarazo. Este trabajo se publicó como una *Letter to the Editor* en *Lancet* el 20 de julio de 1996³¹⁴. Un mes antes, lo presentamos como póster en el *European Neurological Society Meeting* de la Haya⁴¹⁹. Como era nuestra costumbre, el Dr. Polo Esteban y un servidor acudimos a colgar el póster muy pronto (en torno a las 7:30). En

una sala inmensa y casi vacía, a unos escasos metros detrás de nosotros, advertimos la silueta de una mujer pacientemente esperando para hablar con el senior del trabajo, Dr. Polo Esteban. Era, ni más ni menos, que la Dra. Angela Vincent, reputadísima neuroinmunóloga de la Universidad de Oxford (UK), quien entabló un distendido diálogo del que fui testigo mudo. Al agradecer el trabajo afirmó: “esto va en contra de lo que hasta ahora habíamos pensado”.

6.5. Postscriptum: un reconocimiento para el Dr. Onofre Combarros Pascual

Como hemos esbozado anteriormente, en 1994 el Dr. Combarros Pascual puso en marcha el Laboratorio de Neurogenética en un espacio adosado al del Laboratorio de Genética Molecular del HUMV dirigido por el Dr. José Luis Fernández Luna. En sus casi tres décadas de existencia, ha cambiado tres veces de emplazamiento: sótano de la Escuela Universitaria de Enfermería, planta sexta de este edificio, y finalmente Facultad de Medicina. El Dr. Combarros Pascual orientó su investigación al análisis de la asociaciones de diversos *loci* genéticos de riesgo con la enfermedad de Alzheimer esporádica; más adelante, y dentro de un programa internacional, condujo varios estudios de epistasis en esta enfermedad. Su rendimiento investigador fue excepcional con 103 trabajos indexados en *PubMed* (*Search, Combarros O and Alzheimer disease*). Por añadidura, el Dr. Combarros Pascual creó un centro de atracción para investigadores jóvenes, por donde han pasado y siguen pasando Médicos Adjuntos y Especialistas becarios del IDIVAL y del ISCIII, lo cual ha sido un pilar esencial para la investigación del Servicio de Neurología. Además, él fue IP de nuestro de Grupo de Enfermedades Neurodegenerativas, tanto del CIBERNED como del IDIVAL. Tras su

jubilación, el Dr. Jon Infante Ceberio le ha relevado contando con el inestimable apoyo del Dr. Pascual Sánchez-Juan.

7. Conclusiones

Han sido casi cinco décadas de ininterrumpida investigación neurológica, en gran medida derivada de nuestra sólida formación postgraduada, y del inestimable apoyo de los compañeros de trabajo. Nuestra actividad investigadora recibió pronto reconocimiento internacional que, desde luego, ha ejercido una espléndida función estimulante.

Conflicto de interés

El autor no declara ningún conflicto de intereses. No se ha recibido ninguna financiación pública ni privada.

Agradecimientos

Quisiéramos manifestar nuestro profundo reconocimiento a todos quienes han colaborado con el Servicio de Neurología del HUMV, y particularmente a aquellos cuyos nombres no figuran en el texto. Vaya una mención especial para Doña Marta de la Fuente (Secretaria de Neurología) por su inestimable ayuda en el mecanografiado de nuestros manuscritos, y a Don Mario Corral (Responsable de la Biblioteca Marquesa de Pelayo) por su apoyo en la búsqueda de la bibliografía.

Hay dos personas a quienes quisiéramos dedicar este trabajo histórico. A mi esposa, May, siempre a mi lado dándome ánimo para seguir adelante en los 48 años que llevamos juntos. Y al Profesor Juan Martínez López de Letona (1937-2012) a quien debo el haber alcanzado mi madurez como médico en el amplio sentido de la palabra; cuando evoco su memoria siempre brota el último párrafo del obituario que le dedicó Ignacio Sotelo (El País, 17 de enero de 2013): *No hizo nunca el menor intento por recibir honores, todo lo contrario, se lo ponía difícil al que lo intentara, quedando en una débil penumbra en una España que, al brillar los oropeles y oírse tan solo los ecos, en vez de las voces, ha terminado por ignorar a los mejores.*

Bibliografía

1. Muñoz Molina A. Todo lo que era sólido. Madrid: Ediciones Seix, SA; 2013: 197.
2. Berciano Blanco JA. Neurología. En: Izquierdo Rojo JM, ed. 70 Años de Valdecilla. Santander: Gráficas Calima, SA; 1999: 113-133.
3. Jiménez González L. Sistema nervioso central. Anatomía funcional y fundamentos de patología. Zaragoza: Ediciones Heraldo de Aragón; 1962.
4. Segovia de Arana JM. Desarrollo del sistema sanitario público español. 1964-2004 Hospital Universitario Puerta de Hierro. Madrid: Edimsa Editores Médicos, SA; 2005: 15-19.
5. Segovia de Arana JM. La Clínica Puerta de Hierro: 20 años después (Tribuna libre). El País, 16 de julio, 1985.
6. Segovia de Arana JM. 1964 Génesis del proyecto hospitalario Puerta de Hierro. En: 50 Aniversario (1964-2014) Hospital Puerta de Hierro Majadahonda, p. 17-19
[file:///C:/Users/Usuario/Downloads/LIBRO+CUBIERTAS_50_ANIVERSARIO_HUPHM%20\(1\).pdf](file:///C:/Users/Usuario/Downloads/LIBRO+CUBIERTAS_50_ANIVERSARIO_HUPHM%20(1).pdf)
7. Mackay AL. Diccionario de citas científicas. La cosecha de una mirada serena. Madrid: CSIC, Ediciones de la Torre; 1992: 36.
8. Rubín García JM, Maestre Sánchez A. Perspectiva histórica general. En: Izquierdo Rojo JM, ed. 70 Años de Valdecilla. Santander: Gráficas Calima, SA; 1999: 19-31.
9. Thomas A. Le cervelet. Paris, Stenheil; 1897: 164-231.
10. Dejerine J, Thomas A. L'atrophie olivo-ponto-cérébelleuse. *Nouv Iconogr Salpêtr.* 1900; 13: 330-370.
11. Thomas A. Atrophie du cervelet et sclérose en plaque. *Rev Neurol.* 1903; 1: 121-131.
12. Ley R. Forme atypique d'atrophie cérébelleuse ayant évolué en syndrome rigide. *J Belge Neurol Psychiat.* 1925; 47: 92-108.
13. Guillain G, Mathieu P, Bertrand I. Étude anatomo-clinique sur deux cas d'atrophie olivo-ponto-cérébelleuse avec development progressif d'un état hypertonique. *Ann de Méd* 1926; 20: 417-459.
14. Guillain G, Bertand I, Godet-Guillain J. Examen anatomo-pathologique d'un cas de syndrome cérébelleuse progressive non hereditaire avec abolition des réflexes tendineux des membres. *Rev Neurol (Paris).* 1942; 74: 330-333.
15. Veron JP. Contribution á l'étude de l'atrophie olivo-ponto-cérébelleuse et des ses lésions associées. Thèse de médecin. Paris: Foulon ed.; 1968.
16. Bonduelle M, Escourolle R, Bouygues P, Lormeau G, Gray F. [Familial olivo-ponto-cerebellar atrophy with myoclonus. Limits of cerebellar myoclonic dyssynergia (Ramsay-Hunt syndrome)]. *Rev Neurol (Paris).* 1976; 132:113-124.
17. Berciano J. Nuevas contribuciones al conocimiento clínico y patológico de la atrofia olivopontocerebelosa. Tesis doctoral, Universidad de Bilbao, 1978.
18. Berciano J. Olivopontocerebellar atrophy. A review of 117 cases. *J Neurol Sci.* 1982; 53: 253-272.
19. Berciano J, Ricoy JR, Rebollo M, Combarros O, Coria F, Val F. [Familial olivopontocerebellar atrophy (Menzel type). Apropos of a family followed for 46 years]. *Arch Neurobiol (Madr).* 1983; 46: 51-58.
20. Greenfield JG. The spino-cerebellar degenerations. Oxford: Blackwell; 1954.
21. Bassetti CL, Jagella EC. Joseph Jules Dejerine (1849-1917). *J Neurol.* 2006; 253: 823-824.
22. Duckett S. André-Thomas (1867-1963). *J Neurol.* 2000; 247: 235-236.

23. Recondo J. Sur quelques cas anatomocliniques d'atrophie cérébelleuse associée à des dégénérescences systématisées (cordons postérieures, voies d'association, système pallido-nigrique) avec une disgression sur le modalités de la paralysie des mouvement conjugués oculaires. Thèse médecin, Université de Paris; 1964.
24. Scherer HJ. Beiträge zur pathologischen Anatomie des Kleinhirns: 3. Mitteilung (genuine Kleinhirnatrophien). *Z Ges Neurol Psychiatr.* 1933; 145: 335-405.
25. Scherer HJ. Extrapyramidale Störungen bei der olivopontocerebellären Atrophie: ein beitrage zum Problem des lokalen vorseitigen Alterns. *Z Ges Neurol Psychiatr.* 1933; 145: 406-419.
26. Berciano J, Combarros O, Polo JM, Pascual J, Oterino A. An early description of striatonigral degeneration. *J Neurol.* 1999; 246: 462-466.
27. Berciano J, Pascual J, Polo JM. History of ataxia research. En: Klockgether T ed. *Handbook of ataxia disorders.* New York: Marcel Dekker, Inc.; 2000: 77-100.
28. Berciano J. Historical overview of hereditary ataxias with an annotation on the legacy of Hans Joachim Scherer. *Neurosci History.* 2018; 6: 85-100.
29. Berciano J. Hans Joachim Scherer (1906-1945). *J Neurol.* 2021; 268:3052-3053.
30. Van der Eecken H, Adams RD, van Bogaert L. Striopallidal-nigral degeneration. A hitherto undescribed lesion in paralysis agitans. *J Neuropathol Exp Neurol.* 1960; 19: 159-60.
31. Adams R, van Bogaert L, van der Eecken H. Dégénérescences nigro-striées et cerebello-nigro-striées (Unicité clinique et variabilité pathologique des dégénérescences préséniles à forme de rigidité extrapyramidale). *Psychiatr Neurol (Basel).* 1961; 142: 219-59.
32. Adams RD, van Bogaert L, van der Eecken H. Striato-nigral degeneration. *J Neuropathol Exp Neurol.* 1964; 23: 584-608.
33. Pascual J, Pazos A, del Olmo E, Figols J, Leno C, Berciano J. Presynaptic parkinsonism in olivopontocerebellar atrophy: clinical, pathological, and neurochemical evidence. *Ann Neurol.* 1991; 30: 425-428.
34. Berciano J, Valldeoriola F, Ferrer I, Rumià J, Pascual J, Marín C, et al. Presynaptic parkinsonism in multiple system atrophy mimicking Parkinson's disease: a clinicopathological case study. *Mov Disord.* 2002; 17: 812-816.
35. Quinn N. Multiple system atrophy--the nature of the beast. *J Neurol Neurosurg Psychiatry.* 1989; Suppl:78-89.
36. Berciano J. Olivopontocerebellar atrophy. En: Jankovic J, Tolosa E, eds. *Parkinson's disease and movement disorders.* Baltimore: Williams Wilkins; 1998: 263-95.
37. Berciano J. Multiple system atrophy and idiopathic late-onset cerebellar ataxia. En: Manto M and Pandolfo M (eds.), *The cerebellum and its disorders.* Cambridge: Cambridge University Press; 2002: 178-197.
38. Berciano J. Olivopontocerebellar atrophy (OPCA). En: Gilman et al, eds. *Neurobiology of disease.* Elsevier: Amsterdam; 2006: 95-104.
39. Berciano J, Boesch S, Pérez-Ramos JM, Wenning GK. Olivopontocerebellar atrophy: toward a better nosological definition. *Mov Disord.* 2006; 21: 1607-1613.
40. Marie P. Sur l'hérédoataxie cérébelleuse. *Sem Mèd (Paris).* 1893; 13: 444-447.
41. Holmes G. A form of familial degeneration of the cerebellum. *Brain.* 1907; 30: 466-89.

42. Berciano J, Rebollo M, Coria F, Pérez JL, Leno C. [Marie's heredoataxia. New considerations on the use of this term and its nosologic independence]. *Med Clin (Barc)*. 1983; 80: 506-508.
43. Harding AE. Classification of the hereditary ataxias and paraplegias. *Lancet*. 1983; 1(8334): 1151-1155.
44. Harding AE. The hereditary ataxias and related disorders. Edinburgh: Churchill Livingstone, 1984.
45. Berciano J, Amado JA, Freijanes J, Rebollo M, Vaquero A. Familial cerebellar ataxia and hypogonadotropic hypogonadism: evidence for hypothalamic LHRH deficiency. *J Neurol Neurosurg Psychiatry*. 1982; 45: 747-51.
46. Combarros O, Calleja J, Leno C, Berciano J. Association of an ataxia indistinguishable from Friedreich's ataxia and congenital glaucoma in a family: a new syndrome. *J Med Genet*. 1988; 25:44-46.
47. Ramos A. Nuevas contribuciones del estudio con tomografía computadorizada y resonancia magnética al conocimiento de los síndromes espinocerebelosos. Tesis doctoral, Universidad de Cantabria, 1992.
48. Ramos A, Quintana F, Díez C, Leno C, Berciano J. CT findings in spinocerebellar degeneration. *AJNR Am J Neuroradiol*. 1987; 8: 635-640.
49. Polo JM. Prevalencia de los síndromes espinocerebelosos familiares en Cantabria. Estudio nosológico, genético, clínico y neurofisiológico de la forma pura de la paraplejía espástica hereditaria. Tesis doctoral, Universidad de Cantabria, 1988.
50. Polo JM, Calleja J, Combarros O, Berciano J. Hereditary ataxias and paraplegias in Cantabria, Spain. An epidemiological and clinical study. *Brain*. 1991; 114: 855-866.
51. Polo JM, Calleja J, Combarros O, Berciano J. Hereditary "pure" spastic paraplegia: a study of nine families. *J Neurol Neurosurg Psychiatry*. 1993; 56: 175-181.
52. Chamberlain S, Shaw J, Rowland A, Wallis J, South S, Nakamura Y, et al. Mapping of mutation causing Friedreich's ataxia to human chromosome 9. *Nature*. 1988; 334(6179): 248-250.
53. Chamberlain S, Shaw J, Wallis J, Rowland A, Chow L, Farrall M, et al. Genetic homogeneity at the Friedreich ataxia locus on chromosome 9. *Am J Hum Genet*. 1989; 44: 518-521.
54. Campuzano V, Montermini L, Moltò MD, Pianese L, Cossée M, Cavalcanti F, et al. Friedreich's ataxia: autosomal recessive disease caused by an intronic GAA triplet repeat expansion. *Science*. 1996; 271(5254): 1423-1427.
55. Dürr A, Cossée M, Agid Y, Campuzano V, Mignard C, Penet C, et al. Clinical and genetic abnormalities in patients with Friedreich's ataxia. *N Engl J Med*. 1996; 335: 1169-1175.
56. Berciano J, Mateo I, De Pablos C, Polo JM, Combarros O. Friedreich ataxia with minimal GAA expansion presenting as adult-onset spastic ataxia. *J Neurol Sci*. 2002; 194: 75-82.
57. Mateo I, Llorca J, Volpini V, Corral J, Berciano J, Combarros O. GAA expansion size and age at onset of Friedreich's ataxia. *Neurology*. 2003; 61: 274-275.
58. Mateo I, Llorca J, Volpini V, Corral J, Berciano J, Combarros O. Expanded GAA repeats and clinical variation in Friedreich's ataxia. *Acta Neurol Scand*. 2004; 109: 75-78.
59. Berciano J, Combarros O, Calleja J, Polo JM, Pascual J, Leno C. Friedreich's ataxia presenting with pure sensory ataxia: a long-term follow-up study of two patients. *J Neurol*. 1993; 240: 177-180.
60. Berciano J, Combarros O, De Castro M, Palau F. Intronic GAA triplet repeat expansion in Friedreich's ataxia presenting with pure sensory ataxia. *J Neurol*. 1997; 244:390-391.
61. Berciano J, Infante J, García A, Polo JM, Volpini V, Combarros O. Very late-onset Friedreich's ataxia with minimal GAA1 expansion mimicking multiple system atrophy of cerebellar type. *Mov Disord*. 2005; 20: 1643-1645.

62. McKusick VA. Mendelian inheritance in man: catalogs of autosomal dominant, autosomal recessive, and X-linked phenotypes (Eight edition). Baltimore and London: The Johns Hopkins University Press; 1988.
63. Higgins JJ, Morton DH, Patronas N, Nee LE. An autosomal recessive disorder with posterior column ataxia and retinitis pigmentosa. *Neurology*. 1997; 49: 1717-1720.
64. Berciano J, Polo JM. Autosomal recessive posterior column ataxia and retinitis pigmentosa. *Neurology* 1998; 51: 772-1773.
65. Higgins JJ. Autosomal recessive posterior column ataxia and retinitis pigmentosa (reply from the authors). *Neurology* 1998; 51: 1773.
66. Higgins JJ, Morton DH, Loveless JM. Posterior column ataxia with retinitis pigmentosa (AXPC1) maps to chromosome 1q31-q32. *Neurology*. 1999; 52:146-150.
67. Higgins JJ, Klutzman K, Berciano J, Combarros O, Loveless JM. Posterior column ataxia and retinitis pigmentosa: a distinct clinical and genetic disorder. *Mov Disord*. 2000; 15: 575-578.
68. Rajadhyaksha AM, Elemento O, Puffenberger EG, Schierberl KC, Xiang JZ, Putorti ML, et al. Mutations in FLVCR1 cause posterior column ataxia and retinitis pigmentosa. *Am J Hum Genet*. 2010; 87: 643-654.
69. Infante J. Ataxias dominantes: caracterización genética, análisis del fenotipo y correlaciones clínico-genéticas en genealogías españolas. Tesis doctoral, Universidad de Cantabria, 2003.
70. Infante J, Combarros O, Volpini V, Corral J, Llorca J, Berciano J. Autosomal dominant cerebellar ataxias in Spain: molecular and clinical correlations, prevalence estimation and survival analysis. *Acta Neurol Scand*. 2005; 111: 391-399.
71. Álvarez Paradelo S. Estudio neurofisiológico multimodal en las ataxias espinocerebelosas con herencia autosómica dominante de tipo SCA2 y SCA3. Tesis doctoral, Universidad de Cantabria, 2007.
72. Álvarez-Paradelo S, García A, Infante J, Berciano J. Multimodal neurophysiological study of SCA2 and SCA3 autosomal dominant hereditary spinocerebellar ataxias. *Neurologia*. 2011; 26: 157-165.
73. García A, Alvarez S, Infante J, Berciano J. Masseter reflex in the study of spinocerebellar ataxia type 2 and type 3. *Muscle Nerve*. 2009; 40:640-642.
74. Infante J, Berciano J, Volpini V, Corral J, Polo JM, Pascual J, et al. Spinocerebellar ataxia type 2 with Levodopa-responsive parkinsonism culminating in motor neuron disease. *Mov Disord*. 2004; 19:848-852.
75. Berciano J, Infante J, García A, de Pablos C, Amer G, Polo JM, et al. Stiff man-like syndrome and generalized myokymia in spinocerebellar ataxia type 3. *Mov Disord*. 2006; 21:1031-1035.
76. Berciano J, Ferrer I. Glial and neuronal cytoplasmic inclusions in familial olivopontocerebellar atrophy. *Ann Neurol*. 1996; 40: 819-820.
77. Berciano J, Ferrer I. Glial cell cytoplasmic inclusions in SCA2 do not express alpha-synuclein. *J Neurol*. 2005; 252: 742-744.
78. Lewis P. Familial orthostatic hypotension. *Brain*. 1964;87:719-728.
79. Berciano J, Wenning GK. The Lewis family revisited: no evidence for autosomal dominant multiple system atrophy. *Parkinsonism Relat Disord*. 2005; 11: 363-365.
80. Schmitz-Hübsch T, du Montcel ST, Baliko L, Berciano J, Boesch S, Depondt C et al. Scale for the assessment and rating of ataxia: development of a new clinical scale. *Neurology*. 2006; 66: 1717-1720.
81. Jacobi H, Bauer P, Giunti P, Labrum R, Sweeney MG, Charles P, et al. The natural history of spinocerebellar ataxia type 1, 2, 3, and 6: a 2-year follow-up study. *Neurology*. 2011; 77: 1035-1041.
82. Tezenas du Montcel S, Durr A, Rakowicz M, Nanetti L, Charles P, Sulek A, et al. Prediction of the age at onset in spinocerebellar ataxia type 1, 2, 3 and 6. *J Med Genet*. 2014; 51: 479-486.

83. Tezenas du Montcel S, Durr A, Bauer P, Figueroa KP, Ichikawa Y, Brussino A, et al. Clinical Research Consortium for Spinocerebellar Ataxia (CRC-SCA); EUROSCA network. *Brain*. 2014; 137: 2444-2455.
84. Linnemann C, Tezenas du Montcel S, Rakowicz M, Schmitz-Hübsch T, Szymanski S, Berciano J, et al. Peripheral neuropathy in spinocerebellar ataxia type 1, 2, 3, and 6. *Cerebellum*. 2016; 15: 165-173.
85. Jacobi H, du Montcel ST, Bauer P, Giunti P, Cook A, Labrum R, et al. Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. *Lancet Neurol*. 2015; 14: 1101-1108.
86. Diallo A, Jacobi H, Schmitz-Hübsch T, Cook A, Labrum R, Durr A, et al. Body mass index decline is related to spinocerebellar ataxia disease progression. *Mov Disord Clin Pract*. 2017; 11: 689-697.
87. Diallo A, Jacobi H, Cook A, Labrum R, Durr A, Brice A, et al. Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study. *Lancet Neurol*. 2018; 17: 327-334.
88. Jacobi H, du Montcel ST, Bauer P, Giunti P, Cook A, Labrum R, et al. Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. *J Neurol*. 2018; 265: 2040-2051.
89. Diallo A, Jacobi H, Cook A, Giunti P, Parkinson MH, Labrum R, et al. Prediction of survival with long-term disease progression in most common spinocerebellar ataxia. *Mov Disord*. 2019; 34: 1220-1227.
90. Di Gregorio E, Borroni B, Giorgio E, Lacerenza D, Ferrero M, Lo Buono N, et al. ELOVL5 mutations cause spinocerebellar ataxia 38. *Am J Hum Genet*. 2014; 95: 209-217.
91. Gazulla J, Orduna-Hospital E, Benavente I, Rodríguez-Valle A, Osorio-Cacedo P, Alvarez-de Andrés S, et al. Contributions to the study of spinocerebellar ataxia type 38 (SCA38). *J Neurol*. 2020; 267: 2288-2295.
92. Bouchard JP, Richter A, Mathieu J, Brunet D, Hudson TJ, Morgan K, et al. Autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Neuromuscul Disord*. 1998; 8: 474-479.
93. Engert JC, Bérubé P, Mercier J, Doré C, Lepage P, Ge B, et al. ARSACS, a spastic ataxia common in northeastern Québec, is caused by mutations in a new gene encoding an 11.5-kb ORF. *Nat Genet*. 2000; 24: 120-125.
94. Criscuolo C, Banfi S, Orio M, Gasparini P, Monticelli A, Scarano V, et al. A novel mutation in SACS gene in a family from southern Italy. *Neurology*. 2004; 13; 62:100-102.
95. Criscuolo C, Saccà F, De Michele G, Mancini P, Combarros O, Infante J, et al. Novel mutation of SACS gene in a Spanish family with autosomal recessive spastic ataxia. *Mov Disord*. 2005; 20:1358-1361.
96. García A, Criscuolo C, de Michele G, Berciano J. Neurophysiological study in a Spanish family with recessive spastic ataxia of Charlevoix-Saguenay. *Muscle Nerve*. 2008; 37: 107-110.
97. Berciano J, García A, Infante J. Peripheral nerve involvement in hereditary cerebellar and multisystem degenerative disorders. *Handb Clin Neurol*. 2013; 115: 907-932.
98. Gazulla J, Vela AC, Marín MA, Pablo L, Santorelli FM, Benavente I, et al. Is the ataxia of Charlevoix-Saguenay a developmental disease? *Med Hypotheses*. 2011; 77: 347-352.
99. Gazulla J, Benavente I, Vela AC, Marín MA, Pablo LE, Tessa A, et al. New findings in the ataxia of Charlevoix-Saguenay. *J Neurol*. 2012; 259: 869-878.
100. Gazulla J, Mayayo-Sinués E, Benavente I, Modrego PJ, Berciano J. Ataxia of Charlevoix-Saguenay: MR and Clinical Results in Lower-Limb Musculature. *Can J Neurol Sci*. 2014; 41:37-41.
101. Szmulewicz DJ, Waterston JA, Halmagyi GM, Mossman S, Chancellor AM, McLean CA, et al. Sensory neuropathy as part of the cerebellar ataxia neuropathy vestibular areflexia syndrome. *Neurology*. 2011; 76: 1903-1910.

102. Infante J, García A, Serrano-Cárdenas KM, González-Aguado R, Gazulla J, de Lucas EM, et al. Cerebellar ataxia, neuropathy, vestibular areflexia syndrome (CANVAS) with chronic cough and preserved muscle stretch reflexes: evidence for selective sparing of afferent Ia fibres. *J Neurol*. 2018; 265: 1454-1462.
103. Cortese A, Simone R, Sullivan R, Vandrovcova J, Tariq H, Yau WY, et al. Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. *Nat Genet*. 2019; 51: 649-658.
104. Rafehi H, Szmulewicz DJ, Bennett MF, Sobreira NLM, Pope K, Smith KR, et al. Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. *Am J Hum Genet*. 2019; 105: 151-165.
105. Träschütz A, Cortese A, Reich S, Dominik N, Faber J, Jacobi H, et al; RFC1 Study Group. Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. *Neurology*. 2021; 96: e1369-e1382.
106. Gazulla J, Ferrer I, Izquierdo-Alvarez S, Alvarez S, Sánchez-Alcudia R, Bestué-Cardiel M, et al. Hereditary primary lateral sclerosis and progressive nonfluent aphasia. *J Neurol*. 2019; 266:1079-1090.
107. Gazulla J, Ferrer I, Berciano J. The clinical and radiological profile of primary lateral sclerosis: an annotation on its pathological and clinical background. *J Neurol*. 2020; 267:574.
108. Gazulla J, Ferrer I, Berciano J. Reader response: The underacknowledged PPA-ALS: A unique clinicopathologic subtype with strong heritability. *Neurology*. 2020; 94: 282-283.
109. Gazulla J, Izquierdo-Alvarez S, Ruiz-Fernández E, Berciano J. Initial Cerebellar Ataxia in hereditary adult-onset primary lateral sclerosis. *Case Rep Neurol*. 2021; 13: 414-421.
110. González Sánchez M, Izquierdo S, Álvarez S, Bautista Alonso RE, Berciano J, Gazulla J. Clinical manifestations of episodic ataxia type 5. *Neurol Clin Pract*. 2019; 9: 503-504.
111. Gazulla J, Izquierdo-Alvarez S, Ruiz-Fernández E, Lázaro-Romero A, Berciano J. Episodic vestibulocerebellar ataxia associated with a *CACNA1G* missense variant. *Case Rep Neurol*. 2021; 13: 347-354.
112. Combarros Pascual O. Aportaciones al estudio de la atrofia muscular peroneal. Tesis doctoral, Universidad de Cantabria, 1980.
113. Combarros O, Rebollo M, Calleja J, Berciano J. [Prevalence of peroneal muscular atrophy in Cantabria]. *Rev Clin Esp*. 1982; 166:281-28
114. Combarros O, Calleja J, Polo JM, Berciano J. Prevalence of hereditary motor and sensory neuropathy in Cantabria. *Acta Neurol Scand*. 1987; 75: 9-12.
115. Combarros O, Calleja J, Figols J, Cabello A, Berciano J. Dominantly inherited motor and sensory neuropathy type I. Genetic, clinical, electrophysiological and pathological features in four families. *J Neurol Sci*. 1983; 61: 181-191.
116. Bird TD, Kraft GH. Charcot-Marie-Tooth disease: data for genetic counseling relating age to risk. *Clin Genet*. 1978; 14: 43-49.
117. Berciano J, Combarros O, Calleja J, Polo JM, Leno C. The application of nerve conduction and clinical studies to genetic counseling in hereditary motor and sensory neuropathy type I. *Muscle Nerve*. 1989; 12: 302-306.
118. Guiloff RJ, Thomas PK, Contreras M, Armitage S, Schwarz G, Sedgwick EM. Linkage of autosomal dominant type I hereditary motor and sensory neuropathy to the Duffy locus on chromosome 1. *J Neurol Neurosurg Psychiatry*. 1982; 45: 669-674.
119. Middleton-Price HR, Harding AE, Berciano J, Pastor JM, Huson SM, Malcolm S. Absence of linkage of hereditary motor and sensory neuropathy type I to chromosome 1 markers. *Genomics*. 1989; 4: 192-197.

120. Vance JM, Nicholson GA, Yamaoka LH, Stajich J, Stewart CS, Speer MC, et al. Linkage of Charcot-Marie-Tooth neuropathy type 1a to chromosome 17. *Exp Neurol.* 1989; 104: 186-189.
121. Middleton-Price HR, Harding AE, Monteiro C, Berciano J, Malcolm S. Linkage of hereditary motor and sensory neuropathy type I to the pericentromeric region of chromosome 17. *Am J Hum Genet.* 1990; 46:92-94.
122. Raeymaekers P, Timmerman V, Nelis E, De Jonghe P, Hoogendijk JE, Baas F, et al. Duplication in chromosome 17p11.2 in Charcot-Marie-Tooth neuropathy type 1a (CMT 1a). The HMSN Collaborative Research Group. *Neuromuscul Disord.* 1991; 1: 93-97.
123. Lupski JR, de Oca-Luna RM, Slaugenhaupt S, Pentao L, Guzzetta V, Trask BJ, et al. DNA duplication associated with Charcot-Marie-Tooth disease type 1A. *Cell.* 1991; 66: 219-232.
124. Hallam PJ, Harding AE, Berciano J, Barker DF, Malcolm S. Duplication of part of chromosome 17 is commonly associated with hereditary motor and sensory neuropathy type I (Charcot-Marie-Tooth disease type 1). *Ann Neurol.* 1992; 31: 570-572.
125. Berciano J, Sevilla T, Casanovas C, Sivera R, Vílchez JJ, Infante J, et al; Programa 3 (Enfermedades Neuromusculares) del Centro de Investigación Biomédica en Red de Enfermedades Neurodegenerativas (CIBERNED) del Instituto de Salud Carlos III. [Guidelines for molecular diagnosis of Charcot-Marie-Tooth disease]. *Neurologia.* 2012; 27: 169-178.
126. García A. Contribución del estudio neurofisiológico al diagnóstico de la enfermedad de Charcot-Marie-Tooth tipo 1A en la infancia. Tesis doctoral, Universidad de Cantabria, 1996.
127. García A, Combarros O, Calleja J, Berciano J. Charcot-Marie-Tooth disease type 1A with 17p duplication in infancy and early childhood: a longitudinal clinical and electrophysiologic study. *Neurology.* 1998; 50: 1061-1067.
128. García A, Calleja J, Antolín FM, Berciano J. Peripheral motor and sensory nerve conduction studies in normal infants and children. *Clin Neurophysiol.* 2000; 111:513-520.
129. Berciano J, García A, Calleja J, Combarros O. Clinico-electrophysiological correlation of extensor digitorum brevis muscle atrophy in children with charcot-marie-tooth disease 1A duplication. *Neuromuscul Disord.* 2000; 10: 419-424.
130. Berciano J, Berciano MT, Combarros O. Original descriptions of peroneal muscular atrophy. *Muscle Nerve.* 2003; 28:251-252.
131. García A, Pelayo-Negro AL, Álvarez-Paradelo S, Antolín FM, Berciano J. Electromyographic tendon reflex recording: An accurate and comfortable method for diagnosis of Charcot-Marie-Tooth disease type 1a. *Muscle Nerve.* 2015; 52: 39-44.
132. Sabir M, Lyttle D. Pathogenesis of pes cavus in Charcot-Marie-Tooth disease. *Clin Orthop Relat Res.* 1983; 175: 173-178.
133. Gallardo E. Contribuciones de la Resonancia Magnética al estudio de la amiotrofia de las extremidades inferiores en la enfermedad de Charcot-Marie-Tooth. Tesis doctoral, Universidad de Cantabria, 2008.
134. Gallardo E, García A, Combarros O, Berciano J. Charcot-Marie-Tooth disease type 1A duplication: spectrum of clinical and magnetic resonance imaging features in leg and foot muscles. *Brain.* 2006; 129: 426-437.
135. Berciano J, Gallardo E, García A, Infante J, Mateo I, Combarros O. Charcot-Marie-Tooth disease type 1A duplication with severe paresis of the proximal lower limb muscles: a long-term follow-up study. *J Neurol Neurosurg Psychiatry.* 2006; 77: 1169-1176.

136. Berciano J, Gallardo E, García A, Ramón C, Mateo I, Infante J, et al. CMT1A duplication: refining the minimal adult phenotype. *J Peripher Nerv Syst.* 2008; 13: 310-312.
137. Berciano J, García A, Gallardo E, Ramón C, Combarros O. Phenotype and clinical evolution of Charcot-Marie-Tooth disease type 1A duplication. *Adv Exp Med Biol.* 2009; 652:183-200.
138. Berciano J, Gallardo E, García A, Ramón C, Infante J, Combarros O. Clinical progression in Charcot-Marie-Tooth disease type 1A duplication: clinico-electrophysiological and MRI longitudinal study of a family. *J Neurol.* 2010; 257: 1633-1641.
139. Berciano J, Gallardo E, García A, Pelayo-Negro AL, Infante J, Combarros O. New insights into the pathophysiology of pes cavus in Charcot-Marie-Tooth disease type 1A duplication. *J Neurol.* 2011; 258: 1594-1602.
140. Berciano J, Gallardo E. Charcot-Marie-Tooth disease. En: Saba L edit. *Imaging in neurodegenerative disorders.* Oxford: Oxford University Press; 2015: 437-460.
141. Birouk N, Gouider R, Le Guern E, Gugenheim M, Tardieu S, Maisonobe T, et al. Charcot-Marie-Tooth disease type 1A with 17p11.2 duplication. Clinical and electrophysiological phenotype study and factors influencing disease severity in 119 cases. *Brain.* 1997;120: 813-823.
142. Shy ME, Blake J, Krajewski K, Fuerst DR, Laura M, Hahn AF, et al. Reliability and validity of the CMT neuropathy score as a measure of disability. *Neurology.* 2005 12; 64: 1209-1214.
143. Berciano J, García A, Combarros O. Initial semeiology in children with Charcot-Marie-Tooth disease 1A duplication. *Muscle Nerve.* 2003; 27:34-39.
144. Gazulla J, Almárcegui C, Berciano J. Reversible inflammatory neuropathy superimposed on Charcot-Marie-Tooth type 1A disease. *Neurol Sci.* 2018; 39: 793-794.
145. Verhamme C, van Schaik IN, Koelman JH, de Haan RJ, de Visser M. The natural history of Charcot-Marie-Tooth type 1A in adults: a 5-year follow-up study. *Brain.* 2009; 132: 3252-62.
146. Gallardo E, Claeys KG, Nelis E, García A, Canga A, Combarros O, et al. Magnetic resonance imaging findings of leg musculature in Charcot-Marie-Tooth disease type 2 due to dynamin 2 mutation. *J Neurol.* 2008; 255:986-992.
147. Claeys KG, Züchner S, Kennerson M, Berciano J, Garcia A, Verhoeven K, et al. Phenotypic spectrum of dynamin 2 mutations in Charcot-Marie-Tooth neuropathy. *Brain.* 2009; 132: 1741-1752.
148. Gallardo E, García A, Ramón C, Maraví E, Infante J, Gastón I, et al. Charcot-Marie-Tooth disease type 2J with MPZ Thr124Met mutation: clinico-electrophysiological and MRI study of a family. *J Neurol.* 2009; 256:2061-2071.
149. Zimoń M, Baets J, Auer-Grumbach M, Berciano J, Garcia A, Lopez-Laso E, et al. Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 cause an unusual spectrum of neuropathies. *Brain.* 2010; 133: 1798-1809.
150. Berciano J, Baets J, Gallardo E, Zimoń M, García A, López-Laso E, et al. Reduced penetrance in hereditary motor neuropathy caused by TRPV4 Arg269Cys mutation. *J Neurol.* 2011; 258: 1413-1421.
151. Berciano J, García A, Peeters K, Gallardo E, De Vriendt E, Pelayo-Negro AL, et al. NEFL E396K mutation is associated with a novel dominant intermediate Charcot-Marie-Tooth disease phenotype. *J Neurol.* 2015; 262:1289-1300.
152. Berciano J, Peeters K, García A, López-Alburquerque T, Gallardo E, Hernández-Fabián A, et al. NEFL N98S mutation: another cause of dominant intermediate Charcot-Marie-Tooth disease with heterogeneous early-onset phenotype. *J Neurol.* 2016; 263: 361-369.

153. Berciano J, García A, Gallardo E, Peeters K, Pelayo-Negro AL, Álvarez-Paradelo S et al. Intermediate Charcot-Marie-Tooth disease: an electrophysiological reappraisal and systematic review. *J Neurol*. 2017; 264: 1655-1677.
154. Berciano J, Combarros O, Figols J, Calleja J, Cabello A, Silos I et al. Hereditary motor and sensory neuropathy type II. Clinicopathological study of a family. *Brain*. 1986; 109: 897-914.
155. Nelis E, Berciano J, Verpoorten N, Coen K, Dierick I, Van Gerwen V, et al. Autosomal dominant axonal Charcot-Marie-Tooth disease type 2 (CMT2G) maps to chromosome 12q12-q13.3. *J Med Genet*. 2004; 41: 193-197.
156. Peeters K, Palaima P, Pelayo-Negro AL, García A, Gallardo E, García-Barredo R et al. Charcot-Marie-Tooth disease type 2G redefined by a novel mutation in LRSAM1. *Ann Neurol*. 2016; 80: 823-833.
157. Palaima P, Berciano J, Peeters K, Jordanova A. LRSAM1 and the RING domain: Charcot-Marie-Tooth disease and beyond. *Orphanet J Rare Dis*. 2021; 10;16:74.
158. Shy M. LRSAM1 lessons. *Ann Neurol*. 2016; 80: 821-822.
159. Willison HJ, Jacobs BC, van Doorn PA. Guillain-Barré syndrome. *Lancet* 2016; 388: 717-27.
160. Sedano Tous MJ. Estudio del síndrome de Guillain Barré en Cantabria. Tesis doctoral, Universidad de Cantabria, 1991.
161. Sedano MJ, Calleja J, Canga E, Berciano J. Guillain-Barré syndrome in Cantabria, Spain. An epidemiological and clinical study. *Acta Neurol Scand*. 1994; 89: 287-292.
162. Sedano MJ, Orizaola P, Gallardo E, García A, Pelayo-Negro AL, Sánchez-Juan P et al. A unicenter, prospective study of Guillain-Barré syndrome in Spain. *Acta Neurol Scand*. 2019; 139: 546-554.
163. Berciano J, Coria F, Montón F, Calleja J, Figols J, LaFarga M. Axonal form of Guillain-Barresyndrome: evidence for macrophage-associated demyelination. *Muscle Nerve*. 1993; 16:744-751.
164. Berciano J, Figols J, García A, Calle E, Illa I, Lafarga M, et al. Fulminant Guillain-Barresyndrome with universal inexcitability of peripheral nerves: a clinicopathological study. *Muscle Nerve*. 1997; 20: 846-857.
165. Berciano J, García A, Figols J, Muñoz R, Berciano MT, Lafarga M. Perineurium contributes to axonal damage in acute inflammatory demyelinating polyneuropathy. *Neurology*. 2000; 55: 552-559.
166. Berciano J, García A, Villagrà NT, González F, Ramón C, Illa I, et al. Severe Guillain-Barresyndrome: sorting out the pathological hallmark in an electrophysiological axonal case. *J Peripher Nerv Syst*. 2009; 14: 54-63.
167. Gallardo E, Sedano MJ, Orizaola P, Sánchez-Juan P, González-Suárez A, García A, et al. Spinal nerve involvement in early Guillain-Barre syndrome: a clinico-electrophysiological, ultrasonographic and pathological study. *Clin Neurophysiol*. 2015; 126: 810-819.
168. Berciano J. Guillain-Barré síndrome. Saarbrücken (Germany): LAP LAMBERT Academic Publishing, 2015.
169. Berciano MT, Calle E, Andres MA, Berciano J, Lafarga M. Schwann cell nuclear remodelling and formation of nuclear and coiled bodies in Guillain-Barresyndrome. *Acta Neuropathol*. 1996; 92:386-394.
170. Berciano J. MR imaging in Guillain-Barresyndrome. *Radiology*. 1999; 211:290-291.
171. Berciano J. Physiologic-pathologic correlation in Guillain-Barresyndrome in children. *Neurology*. 2000; 55:1762.
172. Berciano J. Cerebrospinal fluid filtration for treating Guillain-Barresyndrome: is there any pathological background? *Anaesthesia*. 2000; 55:408.
173. Berciano J, García A. Nerve ischemia in Guillain-Barresyndrome: an alternative mechanism for early conduction failure. *Rev Neurol (Paris)*. 2002; 158: 364-365.

174. Berciano J. [Axonal pathology in Guillain-Barresyndrome: a complex pathophysiology]. *Neurologia*. 2003; 18:121-131.
175. Villagra NT, Berciano J, Altable M, Navascues J, Casafont I, Lafarga M, et al. PML bodies in reactive sensory ganglion neurons of the Guillain-Barresyndrome. *Neurobiol Dis*. 200; 16: 158-168.
176. Berciano J, Berciano MT, Lafarga M. Cerebrospinal fluid pleocytosis with neutrophil leukocytes in Guillain-Barresyndrome. *Eur J Neurol*. 2004; 11: 645-646
177. Berciano J. Thickening and contrast enhancement of spinal roots on MR imaging in Guillain-Barresyndrome: thoughts on pathologic background. *AJNR Am J Neuroradiol*. 2011; 32: E179.
178. Berciano J, Garca A, Berciano MT, Lafarga M. Criteria for Guillain-Barresyndrome: additional insights from clinico-pathological studies. *Clin Neurophysiol*. 2013; 124:819-821.
179. Berciano J. Reply to "Motor selectivity: important role in the diagnosis of acute motor axonal neuropathy". *Clin Neurophysiol*. 2013; 124: 1702-1703.
180. Berciano J, Garca A. Sural-sparing in Guillain-Barresyndrome: Does it mean lack of histopathological changes? *Clin Neurophysiol*. 2016; 127: 969-970.
181. Berciano J. Early Guillain-Barresyndrome with normal peripheral conduction: which is the pathological hallmark? *Clin Neurol Neurosurg*. 2015; 137: 11.
182. Berciano J, Sedano MJ, Pelayo-Negro AL, Garca A, Orizaola P, Gallardo E, et al. Proximal nerve lesions in early Guillain-Barresyndrome: implications for pathogenesis and disease classification. *J Neurol*. 2017; 264: 221-236.
183. Berciano J. Spinal nerve involvement in early Guillain-Barresyndrome: The Haymaker and Kernohan's legacy. *J Neurol Sci*. 2017; 382: 1-9.
184. Berciano J. Neuropathic pain in early Guillain-BarreSyndrome. *Pain Physician*. 2018; 21:E279-E280.
185. Berciano J. Axonal pathology in early stages of Guillain-Barresyndrome. *Neurologia (Engl Ed)*. 2018 Jul 26:S0213-4853(18)30176-2. doi: 10.1016/j.nrl.2018.06.002.
186. Garca A, Sedano MJ, lvarez-Paradelo S, Berciano J. Reversible conduction failure on the deep tendon reflex response recording in early Guillain-Barresyndrome. *Clin Neurophysiol Pract*. 2018; 3: 159-163.
187. Berciano J, Orizaola P, Gallardo E, Pelayo-Negro AL, Sanchez-Juan P, Infante J, et al. Very early Guillain-Barresyndrome: A clinical-electrophysiological and ultrasonographic study. *Clin Neurophysiol Pract*. 2019; 5:1-9.
188. Berciano J. Inflammatory oedema of nerve trunks may be pathogenic in very early Guillain-Barresyndrome. *Acta Neurol Belg*. 2020;120: 1061-1065.
189. Berciano J. Axonal degeneration in Guillain-Barresyndrome: a reappraisal. *J Neurol*. 2021; 268: 3728-3743.
190. Berciano J, Gallardo E. Spinal nerve pathology in Guillain-Barresyndrome associated with COVID-19 infection. *Muscle Nerve*. 2020; 62: E74-E75
191. Nedkova V, Gutierrez-Gutierrez G, Navacerrada-Barrero FJ, Berciano J, Casasnovas C. Re-evaluating the accuracy of optimized electrodiagnostic criteria in very early Guillain-Barresyndrome: a sequential study. *Acta Neurol Belg*. 2021;121: 1141-1150.
192. Berciano J. Pathogenic events in very early Guillain-Barresyndrome: neither demyelination nor axonal degeneration but endoneurial inflammatory oedema. *J Neurol*. 2021 Sep 3. doi: 10.1007/s00415-021-10773-6
193. Feasby TE, Gilbert JJ, Brown WF, Bolton CF, Hahn AF, Koopman WF, et al. An acute axonal form of Guillain-Barre polyneuropathy. *Brain*. 1986; 109:1115-1126.

194. Cros D, Triggs WJ. There are no neurophysiologic features characteristic of "axonal" Guillain-Barré syndrome. *Muscle Nerve*.1994; 17: 675-677.
195. Feasby TE. Axonal Guillain-Barré syndrome. *Muscle Nerve*.1994; 17:678-679.
196. Yuki N. Pathogenesis of axonal Guillain-Barré syndrome:hypothesis. *Muscle Nerve*. 1994;17:680-681.
197. Izumo S, Linington C, Wekerle H, Meyermann R.Morphologic study on experimental allergic neuritis mediated by T cell line specific for bovine P2 protein in Lewis rats.*Lab Invest* 1985; 53: 209-218.
198. Hahn AF, Feasby TE, Steele A, Lovgren DS, Berry J.Demyelination and axonal degeneration in Lewis rat experimental allergic neuritis depend on the myelin dosage.*Lab Invest*. 1988; 59: 115-125.
199. Hahn AF, Feasby TE, Wilkie L, Lovgren D. P2-peptide induced experimental allergic neuritis: a model to study axonal degeneration.*Acta Neuropathol*. 1991; 82: 60-65
200. Berthold CH, Fraher JP, King RHM, Rydmark M. Microscopical anatomy of the peripheral nervous system. In: Dyck PJ and Thomas PK. eds., *Peripheral neuropathy*. Philadelphia: WB Saunders, vol 1, 2005: 35-91.
201. Haymaker WE &Kernohan JW. The Landry-Guillain-Barré syndrome; a clinicopathologic report of 50 fatal cases and a critique of the literature.*Medicine (Baltimore)*. 1949; 28: 59-141.
202. Powell HC, Myers RR, Mizisin AP, Olee T, Brostoff SW.Response of the axon and barrier endothelium to experimental allergic neuritis induced by autoreactive T cell lines.*Acta Neuropathol* 1991; 82: 364-77.
203. Kamath S, Venkatanarasimha N, Walsh MA, Hughes PMMRI appearance of muscle denervation.*Skeletal Radiol*. 2008; 37: 397-404.
204. Sedano MJ, Canga A, de Pablos C, Polo JM, Berciano J. Muscle MRI in severe Guillain-Barré syndrome with motor nerve inexcitability.*J Neurol*. 2013; 260: 1624-1630.
205. Berciano J, Gallardo E, Fernández-Torre JL, González-Quintanilla V, Infante J Magnetic resonance imaging of lower limb musculature in acute motor axonal neuropathy.*J Neurol*. 2012; 259: 1111-1116.
206. Oguz-Akarsu E, Ozpar R, Mirzayev H, Acet-Ozturk NA, Hakyemez B, Ediger D, et al.; Pandemic Study Team. Guillain-Barré Syndrome in a Patient With Minimal Symptoms of COVID-19 Infection.*Muscle Nerve*. 2020; 62: E54-E5
207. Martín-Aguilar L, Camps-Renom P, Lleixà C, Pascual-Goñi E, Díaz-Manera J, Rojas-García R, et al. Serum neurofilament light chain predicts long-term prognosis in Guillain-Barré syndrome patients.*J Neurol Neurosurg Psychiatry*. 2020 Nov 5;jnnp-2020-323899. doi: 10.1136/jnnp-2020-323899.
208. Berciano J. Rational therapy of Guillain-Barré syndrome.*Lancet*. 1998; 351: 733-735.
209. Berciano J. The rationale for the use of corticosteroids in early severe Guillain-Barré syndrome.*Autoimmun Rev*. 2021; 20:102907.
210. Preston DC, Shapiro BE. Electromyography and neuromuscular disorders. *Clinical-electrophysiologic correlations*, 2nd edition. Philadelphia: Elsevier/Butterworth Heinemann, 2005; 9-22.
211. García A, Infante J, Berciano J. Ia afferent fibers in peripheral nerve disorders: Evidence for divergent vulnerability.*Clin Neurophysiol Pract*. 2021; 6: 133-134.
212. Berciano J. [The nosology of the olivopontocerebellar atrophy. (Critical review) (author's transl)].*Arch Neurobiol (Madr)*. 1981; 44: 163-182.
213. Berciano J, Gutiérrez J, Rebollo M, Dierssen G Thoracic spinal cord ependymoma presenting with ejaculatory failure. Case report. *J Neurosurg*. 1982; 56: 143-144.
214. Rebollo M, Quintana F, Berciano JA.[Bilateral congenital absence of the internal carotid artery].*Med Clin (Barc)*. 1982; 78: 120-121.

215. Coria F, Rebollo M, Quintana F, Polo JM, Berciano J. Occipitoatlantal instability and vertebrobasilar ischemia: case report. *Neurology*. 1982; 32: 303-305.
216. Calleja J, De Pablos C, Fernández R, Berciano J. [Diagnostic and evolutionary signification of the EEG in herpetic encephalitis]. *Arch Neurobiol (Madr)*. 1982; 45: 213-224.
217. Gutiérrez J, Berciano J. [Pontine hemorrhage in a patient addicted to amphetamines]. *Med Clin (Barc)*. 1983; 8: 91.
218. Rebollo M, Quintana F, Combarros O, Berciano J. Giant aneurysm of the intracavernous carotid artery and bilateral carotid fibromuscular dysplasia. *J Neurol Neurosurg Psychiatry*. 1983; 46: 284-285.
219. Coria F, Quintana F, Villalba M, Rebollo M, Berciano J. Craniocervical abnormalities in Down's syndrome. *Dev Med Child Neurol*. 1983; 25: 252-225.
220. Fernández R, Calleja J, de Pablos C, Berciano J. Clinico-neurophysiologic correlation in the Guillain-Barré syndrome. A propos of a serial study of 20 patients]. *Arch Neurobiol (Madr)*. 1983; 46: 287-298.
221. Berciano J, Gutiérrez J, Figols J, Calleja J, Rebollo M, Combarros O. [Sensory polyneuropathy associated with pancreatic adenocarcinoma. Clinico-pathological study of a case observed for 5 years]. *Rev Clin Esp*. 1983; 170: 295-297.
222. Berciano J, Pérez-López JL, Fernández F, Val F, Leno C. Voluminous benign osteoblastoma of the skull. *Surg Neurol*. 1983; 20: 383-386.
223. Rebollo M, Val JF, Garijo F, Quintana F, Berciano J. Livedo reticularis and cerebrovascular lesions (Sneddon's syndrome). Clinical, radiological and pathological features in eight cases. *Brain*. 1983; 106: 965-979.
224. Coria F, Quintana F, Rebollo M, Combarros O, Berciano J. Occipital dysplasia and Chiari type I deformity in a family. Clinical and radiological study of three generations. *J Neurol Sci*. 1983; 62: 147-158.
225. Combarros O, Val F, Figols J, Leno C, Rebollo M, Berciano J. [Subacute ascending polyneuropathy (Guillain-Barré syndrome) as a presenting form of panarteritis nodosa. Apropos of 2 cases with necropsy study]. *Rev Clin Esp*. 1984; 172: 119-121.
226. Leno C, Rebollo M, Combarros O, Figols J, Berciano J. [Encapsulated pontine abscess opening to a ventricle. Apropos of a case with necropsy examination]. *Rev Clin Esp*. 1984; 172: 173-174.
227. Montón F, Rebollo M, Quintana F, Berciano J. Cerebral arterial occlusion and intracranial venous thrombosis in a woman taking oral contraceptives. *Postgrad Med J*. 1984; 60: 426-428.
228. Leno C, Combarros O, Polo JM, Rebollo M, Berciano J. [Bilateral obturator neuralgia secondary to osteitis of the pubis]. *Arch Neurobiol (Madr)*. 1984; 47: 347-352.
229. Rebollo M, Berciano J. [Idiopathic livedo reticularis and cerebrovascular accidents (Sneddon syndrome)]. *Med Clin (Barc)*. 1984; 83: 644-647.
230. Miró J, Rebollo M, Combarros O, Polo JM, Leno C, Berciano J. [Multiple sclerosis in Cantabria. Retrospective study of 30 cases]. *Rev Clin Esp*. 1984; 175: 153-156.
231. Coria F, Berciano J. [Occipito-cervical dysplasia]. *Med Clin (Barc)*. 1985; 84: 199-205.
232. Mayo J, Berciano J. Cephalic tetanus presenting with Bell's palsy. *J Neurol Neurosurg Psychiatry*. 1985; 48: 290.
233. Pérez López JL, Longo J, Quintana F, Diez C, Berciano J. Late onset epileptic seizures. A retrospective study of 250 patients. *Acta Neurol Scand*. 1985; 72: 380-384.
234. Rebollo M, Leno C, Pascual J, Berciano JA. [Organic cerebral syndrome caused by barbiturate deprivation]. *Rev Clin Esp*. 1985; 177: 424.

235. Berciano J, Combarros O. [Myoclonic cerebellar dyssynergia (Ramsay Hunt syndrome)]. *Med Clin (Barc)*. 1986; 86: 78-82.
236. Montón F, Carda JR, Quintana F, Berciano J. Neurogenic claudication and ependymoma. *Neurosurgery*. 1986; 18: 390.
237. Mayo J, Arias M, Leno C, Berciano J. Vascular parkinsonism and periarteritis nodosa. *Neurology*. 1986; 36: 874-875.
238. Berciano J, Ribalta T. [A 28-year-old male with fever, neck rigidity and loss of consciousness]. *Med Clin (Barc)*. 1986; 87: 554-563.
239. Fernández F, Leno C, Combarros O, Berciano J. Cricopharyngeal dysfunction due to syringobulbia. *Neurology*. 1986; 36: 1623-1625.
240. Leno C, Combarros O, Berciano J. Lumbosacral plexopathy due to dermoid cyst of the greater omentum. *Postgrad Med J*. 1987; 63: 45-46.
241. García-Moncó C, Quintana F, Berciano J. Epidural thoracic spinal cord metastasis presenting with cerebellar gait ataxia. *Surg Neurol*. 1987; 28: 316-317.
242. Polo JM, Pascual J, Díez C, Leno C, Berciano J. [Recurrent herpes simplex encephalitis and acyclovir]. *Med Clin (Barc)*. 1987; 89: 423-425.
243. Berciano J. Diffuse cerebral angiomas. *AJNR Am J Neuroradiol*. 1987; 8: 1146.
244. Pascual J, Combarros O, Berciano J. Partial status epilepticus following single low dose of chlorimipramine in a patient on MAO-inhibitor treatment. *Clin Neuropharmacol*. 1987; 10: 565-567.
245. Pascual J, Berciano J. [Idiopathic intracranial hypertension. Diagnosis and treatment]. *Med Clin (Barc)*. 1987; 89: 873-876.
246. de Pablos C, Calleja J, Fernández F, Berciano J. Miller Fisher syndrome: an electrophysiologic case study. *Electromyogr Clin Neurophysiol*. 1988; 28: 21-25.
247. Miró J, García-Moncó C, Leno C, Berciano J. Pelvic pain: an undescribed paroxysmal manifestation of multiple sclerosis. *Pain*. 1988; 32: 73-75.
248. López-Vega JM, Calleja J, Combarros O, Polo JM, Berciano J. Motor neuron disease in Cantabria. *Acta Neurol Scand*. 1988; 77: 1-5.
249. Leno C, Berciano J. [Cerebral ischemia in patients under 45 years of age]. *Med Clin (Barc)*. 1988; 90: 160-161.
250. Combarros O, Calleja J, Pascual J, Berciano J. Numbness of the tip of the tongue as the presenting symptom of chronic inflammatory polyradiculoneuropathy. *Neurology*. 1988; 38: 333.
251. Pascual J, Sedano MJ, Polo JM, Berciano J. Intravenous lidocaine for status epilepticus. *Epilepsia*. 1988; 29: 584-589.
252. Calleja J, Carpizo R, Berciano J. Orgasmic epilepsy. *Epilepsia*. 1988; 29: 635-639.
253. Pascual J, Combarros O, Polo JM, Berciano J. Localized CNS brucellosis: report of 7 cases. *Acta Neurol Scand*. 1988; 78: 282-289.
254. Berciano J. Sneddon syndrome: another mendelian etiology of stroke. *Ann Neurol*. 1988; 24: 586-587.
255. Pascual J, Combarros O, Berciano J. Gaze-evoked amaurosis in pseudotumor cerebri. *Neurology*. 1988; 38: 1654-1655.
256. García-Moncó C, Berciano J. Sarcoid meningitis, high adenosine deaminase levels in CSF and results of cranial irradiation. *J Neurol Neurosurg Psychiatry*. 1988; 51: 1594-1596.

257. de Pablos C, Calleja J, Combarros O, Berciano J. Spanish toxic oil syndrome neuropathy in three patients with hereditary motor and sensory neuropathy type I. *Arch Neurol*. 1989; 46: 202-204.
258. Leno C, Sedano MJ, Combarros O, Berciano J. Congenital giant pigmented nevus and intracranial arteriovenous malformation. *Surg Neurol*. 1989; 31: 407-408.
259. Pascual J, Berciano J. Failure of mexiletine to control trigeminal neuralgia. *Headache*. 1989; 29: 517-518.
260. Polo JM, Berciano J. [Creutzfeldt-Jakob disease: an historical fact]. *Med Clin (Barc)*. 1989; 93: 317.
261. Pascual J, Gutiérrez A, Polo JM, Berciano J. [Occipital condyle syndrome: presentation of a case]. *Neurologia*. 1989; 4: 293-295.
262. Pascual J, Polo JM, Berciano J. The dose of propranolol for migraine prophylaxis. Efficacy of low doses. *Cephalalgia*. 1989; 9: 287-291.
263. Berciano J, Berciano MT, Polo JM, Figols J, Ciudad J, Lafarga M. Creutzfeldt-Jakob disease with severe involvement of cerebral white matter and cerebellum. *Virchows Arch A Pathol Anat Histopathol*. 1990; 417: 533-538.
264. Combarros O, Gutiérrez A, Pascual J, Berciano J. Oral dyskinesias associated with bilateral thalamo-capsular infarction. *J Neurol Neurosurg Psychiatry*. 1990; 53: 168-169.
265. Miró J, Peña-Sagredo JL, Berciano J, Insúa S, Leno C, Velarde R. Prevalence of primary Sjögren's syndrome in patients with multiple sclerosis. *Ann Neurol*. 1990; 27: 582-584.
266. García-Moncó JC, Miró Jornet J, Fernández Villar B, Benach JL, Guerrero Espejo A, Berciano JA [Multiple sclerosis or Lyme disease? a diagnosis problem of exclusion]. *Med Clin (Barc)*. 1990; 94: 685-688.
267. Berciano J, Pascual J. [Pharmacotherapy of the spino-cerebellar syndromes]. *Neurologia*. 1990; 5: 200-204.
268. Miró J, Amado JA, Pesquera C, López-Cordovilla JJ, Berciano J. Assessment of the hypothalamic-pituitary-adrenal axis function after corticosteroid therapy for MS relapses. *Acta Neurol Scand*. 1990; 81: 524-528.
269. Pascual J, Polo JM, Berciano J. Serious migraine: a study of some epidemiological aspects. *Headache*. 1990; 30: 481-484.
270. Berciano J. Dysphagia in Chiari malformations. *Neurology*. 1990; 40: 1637-1638.
271. Vega A, Quintana F, Berciano J. Basichondrocranium anomalies in adult Chiari type I malformation: a morphometric study. *J Neurol Sci*. 1990; 99: 137-145.
272. Berciano J. MR imaging of degenerative disorders of brainstem and cerebellum. *Magn Reson Imaging*. 1991; 9: 467.
273. Lafarga M, Berciano MT, Suarez I, Viadero CF, Andres MA, Berciano J. Cytology and organization of reactive astroglia in human cerebellar cortex with severe loss of granule cells: a study on the ataxic form of Creutzfeldt-Jakob disease. *Neuroscience*. 1991; 40: 337-352.
274. Berciano J, Díez C, Polo JM, Pascual J, Figols J. CT appearance of panencephalopathic and ataxic type of Creutzfeldt-Jakob disease. *J Comput Assist Tomogr*. 1991; 15: 332-334.
275. Arias Bal MA, Vázquez-Barquero JL, Peña C, Miro J, Berciano JA. Psychiatric aspects of multiple sclerosis. *Acta Psychiatr Scand*. 1991; 83: 292-296.
276. Pascual J, Berciano J. An open trial of buspirone in migraine prophylaxis. Preliminary report. *Clin Neuropharmacol*. 1991; 14: 245-250.
277. Combarros O, Calleja J, Hernández L, Polo JM, Berciano J. Guillain-Barré syndrome associated with idiopathic thrombocytopenic purpura. *J Neurol Neurosurg Psychiatry*. 1991; 54: 654-655.
278. De Pablos C, Berciano J, Calleja J. Brain-stem auditory evoked potentials and blink reflex in Friedreich's ataxia. *J Neurol*. 1991; 238: 212-216.

279. Berciano J, Oterino A, Rebollo M, Pascual J. Myasthenia gravis unmasked by cocaine abuse. *N Engl J Med.* 1991; 325: 892.
280. Polo JM, Oterino A, Setién S, Berciano J. [Epilepsy in Spain: comments concerning an unusual factor in decompensation]. *Med Clin (Barc).* 1991; 97: 596-597.
281. Combarros O, Polo JM, Pascual J, Berciano J. Evidence of somatotopic organization of the sensory thalamus based on infarction in the nucleus ventralis posterior. *Stroke.* 1991; 22: 1445-1447.
282. Miró J, Aguayo F, Garrido JC, Alvarez C, Polo JM, Pascual J, et al. Intrathecal immunoglobulin synthesis in multiple sclerosis: effect of corticosteroids and azathioprine. *Eur Neurol.* 1992; 32: 349-353.
283. Pascual J, Ciudad J, Berciano J. Role of lidocaine (lignocaine) in managing status epilepticus. *J Neurol Neurosurg Psychiatry.* 1992; 55: 49-51.
284. Arias Bal MA, Vázquez Barquero JL, Miró J, Peña C, Berciano J. [Depressive states in multiple sclerosis. Critical bibliographic review]. *Actas Luso Esp Neurol Psiquiatr Cienc Afines.* 1992; 20: 97-103.
285. Berciano J, Coria F. Occipitotlantal instability: a hemodynamic cause of vertebrobasilar ischemia after neck motion. *Stroke.* 1992; 23: 921.
286. Leno C, Pascual J, Polo JM, Berciano J, Sedano C. Nephrotic syndrome, accelerated atherosclerosis, and stroke. *Stroke.* 1992; 23: 921-912.
287. Pascual J, Oterino A, Berciano J. Headache in type I Chiari malformation. *Neurology.* 1992; 42: 1519-1521.
288. Combarros O, Díez C, Cano J, Berciano J. Ataxic hemiparesis with cheiro-oral syndrome in capsular infarction. *J Neurol Neurosurg Psychiatry.* 1992; 55: 859-860.
289. Polo JM, Fábrega E, Casafont F, Fariñas MC, Salesa R, Vázquez A, et al. Treatment of cerebral aspergillosis after liver transplantation. *Neurology.* 1992; 42: 1817-1819.
290. Combarros O, Fábrega E, Polo JM, Berciano J. Cyclosporine-induced chorea after liver transplantation for Wilson's disease. *Ann Neurol.* 1993; 33: 108-109.
291. Lafarga M, Berciano MT, Saurez I, Andres MA, Berciano J. Reactive astroglia-neuron relationships in the human cerebellar cortex: a quantitative, morphological and immunocytochemical study in Creutzfeldt-Jakob disease. *Int J Dev Neurosci.* 1993; 11: 199-213.
292. Pascual J, Berciano J. Relief of cluster-tic syndrome by the combination of lithium and carbamazepine. *Cephalalgia.* 1993; 13:205-206.
293. Leno C, Berciano J, Combarros O, Polo JM, Pascual J, Quintana F, et al. A prospective study of stroke in young adults in Cantabria, Spain. *Stroke.* 1993; 24: 792-795.
294. Tolosa E, Berciano J. Choreas, hereditary and other ataxias, tics, myoclonus, and other movement disorders. *Curr Opin Neurol Neurosurg.* 1993; 6: 358-368.
295. Pascual J, Berciano J. [Daily chronic headache in patients with migraine induced by abuse of ergotamine-analgesics: response due to a protocol of outpatient treatment]. *Neurologia.* 1993; 8: 212-215.
296. Peña-Sagredo JL, Miró J, Aguayo F, Alvarez C, Berciano J. [Primary Sjogren syndrome with involvement of the nervous system. Description of 4 cases]. *Neurologia.* 1993; 8: 231-234.
297. Berciano J. [Gangliosides and the Guillain-Barré syndrome]. *Med Clin (Barc).* 1993; 101: 758-759.
298. Combarros O, Miró J, Berciano J. Ageusia associated with thalamic plaque in multiple sclerosis. *Eur Neurol.* 1994; 34: 344-346.
299. Berciano J, Jiménez C, Figols J, Ferreres JC, Combarros O, Arjona R, et al. Primary leptomeningeal lymphoma presenting as cerebellopontine angle lesion. *Neuroradiology.* 1994; 36: 369-371.

300. Pascual J, Berciano J. Experience in the diagnosis of headaches that start in elderly people. *J Neurol Neurosurg Psychiatry*. 1994; 57: 1255-1257.
301. Berciano J, Calleja J, Combarros O. Charcot-Marie-Tooth disease. *Neurology*. 1994; 44: 1985-1986.
302. Berciano J. [The pupil in Alzheimer's disease]. *Neurologia*. 1995; 10: 63-64.
Pascual J, Combarros O, Leno C, Polo JM, Rebollo M, Berciano J. [Distribution of headache by diagnosis as the reason for neurologic consultation]. *Med Clin (Barc)*. 1995; 104: 161-164.
303. Berciano J. [Slowly developing Creutzfeldt-Jakob disease]. *Neurologia*. 1995; 10: 213-214.
304. Combarros O, Sánchez-Pernaute R, Orizaola P, Berciano J. Absence of F-waves as an early electrodiagnostic finding in infarction of the conus medullaris. *Muscle Nerve*. 1995; 18: 552-554.
305. Leno C, Berciano J, Combarros O, Sedano C, Alvarez C, Merino J, et al. Etiologic study of stroke in 95 young adults. *Neurologia*. 1995; 10: 283-287.
306. Pascual J, Berciano J. Clinical experience with headaches in preadolescent children. *Headache*. 1995; 35: 551-553.
307. Serratos JM, Delgado-Escueta AV, Posada I, Shih S, Drury I, Berciano J, et al. The gene for progressive myoclonus epilepsy of the Lafora type maps to chromosome 6q. *Hum Mol Genet*. 1995; 4: 1657-1663.
308. Berciano J, Figols J, Combarros O, Calleja J, Pascual J, Oterino A. Plexiform neurofibroma of the cauda equina presenting as peroneal muscular atrophy. *Muscle Nerve*. 1996; 19: 250-253.
309. Torres-Aleman I, Barrios V, Lledo A, Berciano J. The insulin-like growth factor I system in cerebellar degeneration. *Ann Neurol*. 1996; 39: 335-342.
310. Sánchez Pernaute R, Berciano J, Rebollo M, Pascual J. Intramedullary tuberculoma of the spinal cord with syringomyelia. *Neuroradiology*. 1996; 38 Suppl 1: S105-106.
311. Combarros O, Iglesias F, Guitera MV, Berciano J. MRI in radiation-induced myelopathy and pharyngocutaneous fistula. *Neuroradiology*. 1996; 38 Suppl 1: S130-132.
312. Pascual J, Iglesias F, Oterino A, Vázquez-Barquero A, Berciano J. Cough, exertional, and sexual headaches: an analysis of 72 benign and symptomatic cases. *Neurology*. 1996; 46: 1520-1524.
313. Polo JM, Berciano J. [Mad cows: neurologists in their place]. *Neurologia*. 1996; 11: 201-204.
314. Polo JM, Martin J, Berciano J. Botulism and pregnancy. *Lancet*. 1996; 348(9021): 195.
315. Combarros O, Pascual J, de Pablos C, Ortega F, Berciano J. Taste loss as an initial symptom of Guillain-Barré syndrome. *Neurology*. 1996; 47: 1604-1605.
316. Berciano J, Pascual J. [The new editors' message]. *Neurologia*. 1997; 12: 49-50.
317. Berciano J, Pascual J, Polo JM, Combarros O, Figols J, Díez C. Ataxic type of Creutzfeldt-Jakob disease with disproportionate enlargement of the fourth ventricle: a serial CT study. *J Neurol Neurosurg Psychiatry*. 1997; 62: 295-297.
318. Berciano J, Combarros O. [Nerve enlargement in a girl with Charcot-Marie-Tooth disease type 1A]. *Neurologia*. 1997; 12: 184.
319. Morís G, Figols J, Combarros O, Berciano J. [An asymmetric form of chronic spinal muscular atrophy]. *Rev Clin Esp*. 1997; 197: 726-727.
320. Pascual J, Oterino A, Berciano J. [Headaches due to cough, exertion and sexual intercourse]. *Neurologia*. 1997; 12 Suppl 5: 61-65.
321. Combarros O, Alvarez de Arcaya A, Berciano J. Isolated unilateral hypoglossal nerve palsy: nine cases. *J Neurol*. 1998; 245: 98-100.
322. Pascual J, Berciano J. Buspirone in primary headaches. *Acta Neurol Scand*. 1998; 97: 142.

323. Torres-Aleman I, Barrios V, Berciano J. The peripheral insulin-like growth factor system in amyotrophic lateral sclerosis and in multiple sclerosis. *Neurology*. 1998; 50: 772-776.
324. Casado Naranjo I, Berciano J, Durán Herrera C, Aguirre Sánchez J. [Sixty-seven-year-old male with subacute progressive ataxia and extensive involvement of the lower cranial nerves]. *Neurologia*. 1998; 13: 22-28.
325. Leno C, Mateo I, Cid C, Berciano J, Sedano C. Autoimmunity in Down's syndrome: another possible mechanism of Moyamoya disease. *Stroke*. 1998; 29: 868-869.
326. Combarros O, Alvarez de Arcaya A, Quintana F, Berciano J. [Paralysis of the hypoglossal nerve in the presentation of dural arteriovenous fistula of the posterior fossa]. *Neurologia*. 1998; 13: 260-262.
327. Pascual J, Cid C, Berciano J. High-dose i.v. immunoglobulin for peripheral neuropathy associated with Sjögren's syndrome. *Neurology*. 1998; 51: 650-651.
328. Combarros O, Escribano J, Sánchez-Velasco P, Leyva-Cobián F, Oterino A, Leno C, et al. Association of the HLA-A2 allele with an earlier age of onset of Alzheimer's disease. *Acta Neurol Scand*. 1998; 98: 140-141.
329. Combarros O, Misiego M, Oterino A, Berciano J. [Sensitive deficit of pseudo-polyneuritis distribution as the initial manifestation of spondylotic cervical myelopathy]. *Neurologia*. 1998; 13: 407-409.
330. Pascual J, Berciano J. [When a headache has to be investigated?]. *An Med Interna*. 1999; 16: 165-166.
331. Fernández-Torre JL, Polo JM, Calleja J, Berciano J. Castleman's disease associated with chronic inflammatory demyelinating polyradiculoneuropathy: a clinical and electrophysiological follow-up study. *Clin Neurophysiol*. 1999; 110: 1133-1138
332. Pascual J, Cerezal L, Canga A, Alvarez de Arcaya A, Polo JM, Berciano J. Tolosa-Hunt syndrome: focus on MRI diagnosis. *Cephalalgia*. 1999; 19 Suppl 25: 36-38.
333. Berciano J. [From the genetics to the prevention of stroke]. *Rev Neurol*. 1999; 29: 836-847.
334. Martini R, Berciano J, Van Broeckhoven C. 5th Workshop of the European CMT Consortium, 69th ENMC International Workshop: therapeutic approaches in CMT neuropathies and related disorders 23-25 April 1999, Soestduinen, The Netherlands. *Neuromuscul Disord*. 2000; 10: 69-74.
335. Combarros O, Infante J, Berciano J. Akinetic mutism from frontal lobe damage responding to levodopa. *J Neurol*. 2000; 247: 568-569.
336. Berciano J. [Ataxia and genetics]. *Med Clin (Barc)*. 2000; 115: 135-136.
337. Sánchez-Guerra M, Cerezal L, Leno C, Díez C, Figols J, Berciano J. Primary brain lymphoma presenting as Parkinson's disease. *Neuroradiology*. 2001; 43: 36-40.
338. Berciano J, Pascual J. [Message of the editors]. *Neurologia*. 2001; 16: 55-56.
339. Berciano J, Lafarga M. Pioneers in neurology. Santiago Ramón y Cajal (1852-1934). *J Neurol*. 2001; 248: 152-153.
340. Berciano J, Lafarga M, Berciano M. Santiago Ramón y Cajal. *Neurologia*. 2001; 16: 118-121.
341. Berciano J. [Genetics in Parkinson's disease: toward a new nosological era]. *Med Clin (Barc)*. 2001; 116: 614-616.
342. Infante J, García A, Combarros O, Mateo JI, Berciano J, Sedano MJ, et al. Diagnostic strategy for familial and sporadic cases of neuropathy associated with 17p11.2 deletion. *Muscle Nerve*. 2001; 24: 1149-1155
343. Berciano J. Axonal dysfunction in adoptive transfer of experimental autoimmune neuritis with P2-specific CD4+ cells: the role of nerve ischemia. *J Neuropathol Exp Neurol*. 2001; 60: 1233-1244.
344. Berciano J, Infante J, Mateo I, Combarros O. [Hereditary ataxias and paraplegias: a clinicogenetic review]. *Neurologia*. 2002; 17: 40-51.

345. Mateo I, Sánchez-Guerra M, Combarros O, Llorca J, Infante J, González-García J, et al Lack of association between cathepsin D genetic polymorphism and Alzheimer disease in a Spanish sample. *Am J Med Genet.* 2002; 114: 31-33
346. Berciano J. Intramedullary spinal tuberculoma and syringomyelia. *Can J Neurol Sci.* 2002; 29: 102.
347. Combarros O, Vadillo A, Gutiérrez-Pérez R, Berciano J. Cervical spinal cord infarction simulating myocardial infarction. *Eur Neurol.* 2002; 47: 185-186.
348. Berciano J, Montón FI, Maeso MC, Ferrer I. [Man aged 49 years suffering from progressive clinical picture with palatal tremor, segmental myoclonus, ataxia, parkinsonism, amyotrophy, pyramidal signs, supranuclear ophthalmoplegia and cognitive decline]. *Neurologia.* 2002; 17: 237-243.
349. Combarros O, Infante J, Llorca J, Peña N, Fernández-Viadero C, Berciano J. The myeloperoxidase gene in Alzheimer's disease: a case-control study and meta-analysis. *Neurosci Lett.* 2002; 326: 33-36.
350. Combarros O, Alvarez-Arcaya A, Sánchez-Guerra M, Infante J, Berciano J. Candidate gene association studies in sporadic Alzheimer's disease. *Dement Geriatr Cogn Disord.* 2002; 14: 41-54.
351. Polo JM, Alvarez de Arcaya A, Cid C, Berciano J. Aphasia in a farmer following viper bite. *Lancet.* 2002; 359(9324): 2164.
352. Combarros O, Sánchez-Guerra M, Infante J, Llorca J, Berciano J. Gene dose-dependent association of interleukin-1A [-889] allele 2 polymorphism with Alzheimer's disease. *J Neurol.* 2002; 249:1242-1245.
353. Combarros O, Sánchez-Guerra M, Infante J, Llorca J, Berciano J. Interaction of the H63D mutation in the hemochromatosis gene with the apolipoprotein E epsilon 4 allele modulates age at onset of Alzheimer's disease. *Dement Geriatr Cogn Disord.* 2003; 15: 151-154.
354. Combarros O, Rodero L, Infante J, Palacio E, Llorca J, Fernández-Viadero C, et al. Age-dependent association between the Q7R polymorphism in the Saitohin gene and sporadic Alzheimer's disease. *Dement Geriatr Cogn Disord.* 2003; 16: 132-135.
355. Berciano J, Pascual J. Selective contrast enhancement of anterior spinal nerve roots on magnetic resonance imaging: a suggestive sign of Guillain-Barré syndrome and neurobrucellosis. *J Peripher Nerv Syst.* 2003; 8: 135.
356. Combarros O, Llorca J, Sánchez-Guerra M, Infante J, Berciano J. Age-dependent association between interleukin-1A (-889) genetic polymorphism and sporadic Alzheimer's disease. A meta-analysis. *J Neurol.* 2003; 250: 987-989.
357. Berciano J, Combarros O. Hereditary neuropathies. *Curr Opin Neurol.* 2003; 16: 613-622.
358. Combarros O, Infante J, Llorca J, Berciano J. Interleukin-1A (-889) genetic polymorphism increases the risk of multiple system atrophy. *Mov Disord.* 2003; 18: 1385-1386.
359. Morís G, Berciano J, Miró J. A clinical longitudinal study of multiple sclerosis in Cantabria, Spain. *Neurologia.* 2003; 18: 723-730.
360. Berciano J, García A. Acute motor conduction block neuropathy: Another Guillain-Barré syndrome variant. *Neurology.* 2004; 62: 1026-1027.
361. Berciano J, Leno C, Figols J, García A, Polo JM, Berciano MT, et al. Epilepsia partialis continua in progressive multifocal leukoencephalopathy: a motor cortex isolation syndrome. *Mov Disord.* 2003; 18: 1559-1564.
362. Combarros O, Infante J, Llorca J, Berciano J. No evidence for association of the monocyte chemoattractant protein-1 (-2518) gene polymorphism and Alzheimer's disease. *Neurosci Lett.* 2004; 360: 25-28.

363. Combarros O, Infante J, Llorca J, Berciano J. Polymorphism at codon 66 of the brain-derived neurotrophic factor gene is not associated with sporadic Alzheimer's disease. *Dement Geriatr Cogn Disord*. 2004; 18: 55-58.
364. Combarros O, Infante J, Llorca J, Peña N, Fernández-Viadero C, Berciano J. The chemokine receptor CCR5-Delta32 gene mutation is not protective against Alzheimer's disease. *Neurosci Lett*. 2004; 366: 312-314.
365. Fernández-Torre JL, Calleja J, Pascual J, Galdós P, De Pablos C, Berciano J. Epilepsia partialis continua of the abdominal muscles: a detailed electrophysiological study of a case. *Mov Disord*. 2004; 19: 1375-1378.
366. Berciano J, Pascual J. [Message from the outgoing editors]. *Neurologia*. 2004; 19: 702-703.
367. Llorca J, Dierssen-Sotos T, Combarros O, Berciano J. Consistency in gene-Alzheimer's disease association studies. *J Epidemiol Community Health*. 2005; 59: 83-85.
368. Berciano J. Subcortical pathophysiological mechanisms in epilepsia partialis continua. *Cerebellum*. 2004; 3: 248.
369. Berciano J, Illa I. Does *Campylobacter jejuni* infection elicit "demyelinating" Guillain-Barré syndrome?. *Neurology*. 2005; 64: 766-767;
370. Berciano J. [Contribution of dopamine transporters brain SPECT to multisystemic atrophy diagnosis]. *Med Clin (Barc)*. 2005; 124: 78.
371. Combarros O, Infante J, Llorca J, Peña N, Fernández-Viadero C, Berciano J. Interaction between interleukin-6 and intercellular adhesion molecule-1 genes and Alzheimer's disease risk. *J Neurol*. 2005; 252: 485-487.
372. Combarros O, Infante J, Rodríguez E, Llorca J, Peña N, Fernández-Viadero C, Berciano J. CD14 receptor polymorphism and Alzheimer's disease risk. *Neurosci Lett*. 2005; 380: 193-196.
373. Combarros O, Riancho JA, Infante J, Sañudo C, Llorca J, Zarrabeitia MT, et al. Interaction between CYP19 aromatase and butyrylcholinesterase genes increases Alzheimer's disease risk. *Dement Geriatr Cogn Disord* 2005; 20: 153-157.
374. Infante J, Rodríguez E, Combarros O, Mateo I, Fontalba A, Pascual J, et al. LRRK2 G2019S is a common mutation in Spanish patients with late-onset Parkinson's disease. *Neurosci Lett*. 2006; 395: 224-226.
375. Berciano J, Poca MA, García A, Sahuquillo J. Paroxysmal cervicobrachial cough-induced pain in a patient with syringomyelia extending into spinal cord posterior gray horns. *J Neurol*. 2007; 254: 678-81.
376. Combarros O, Riancho JA, Arozamena J, Mateo I, Llorca J, Infante J, et al. Interaction between estrogen receptor-alpha and butyrylcholinesterase genes modulates Alzheimer's disease risk. *J Neurol*. 2007; 254: 1290-1292
377. Gómez-Garre P, Gutiérrez-Delicado E, Gómez-Abad C, Morales-Corraliza J, Villanueva VE, Rodríguez de Córdoba S, et al. Hepatic disease as the first manifestation of progressive myoclonus epilepsy of Lafora. *Neurology*. 2007; 68: 1369-1373.
378. Combarros O, Llorca J, Sánchez-Juan P, Mateo I, Infante J, Rodríguez E, et al. Interaction between prion protein and interleukin-1A genes increases early-onset Alzheimer's disease risk. *J Neurol*. 2007; 254: 115-117.
379. Berciano J, Gallardo E, Domínguez-Perles R, Gallardo E, García A, García-Barredo R, et al. Autosomal-dominant distal myopathy with a myotilin S55F mutation: sorting out the phenotype. *J Neurol Neurosurg Psychiatry*. 2008; 79: 205-208.

380. Combarros O, Sánchez-Juan P, Riancho JA, Mateo I, Rodríguez-Rodríguez E, Infante J, et al. Aromatase and interleukin-10 genetic variants interactively modulate Alzheimer's disease risk. *J Neural Transm (Vienna)*. 2008; 115: 863-867.
381. Infante J, Berciano J, Sánchez-Juan P, García A, Di Fonzo A, Breedveld G, et al. Pseudo-orthostatic and resting leg tremor in a large Spanish family with homozygous truncating parkin mutation. *Mov Disord*. 2009; 24: 144-147.
382. Berciano J. Peripheral neuropathies: Molecular diagnosis of Charcot-Marie-Tooth disease. *Nat Rev Neurol*. 2011; 7: 305-306.
383. Berciano J, Fernández-Torre JL, Ramón C, Pelayo-Negro AL, Infante J. Subclinical Charcot-Marie-Tooth disease type 1A in an ex-professional cyclist. *Clin Neurol Neurosurg*. 2012; 114: 394-395.
384. Berciano J, de Lucas EM, Combarros O. Thumb, forefinger, and lip numbness: a distinctive thalamic lacunar syndrome. *Neurol Sci*. 2013; 34: 253-254.
385. Delgado-Alvarado M, Palacio-Portilla E, Pelayo-Negro AL, Lerena P, Berciano J. From ileostomy to sudden quadriplegia with electrocardiographic abnormalities: a short and unfortunate path. *Neurol Sci*. 2013; 34: 1471-1473.
386. Berciano J. Recent advances in clinical neurogenetics. *J Neurol*. 2013; 260: 2451-2457.
387. Sierra M, Infante J, Berciano J. Substantia nigra echogenicity in Friedreich's ataxia patients. *Cerebellum*. 2013; 12: 437-440.
388. Riancho J, Infante J, Orizaola P, Mateo I, Viadero R, Delgado-Alvarado M, et al. [Reversible cerebral vasoconstriction and acute encephalopathy as a presentation form of Guillain-Barré syndrome]. *Rev Clin Esp (Barc)*. 2013; 213: e23-6.
389. Delgado-Alvarado M, Sedano MJ, González-Quintanilla V, de Lucas EM, Polo JM, Berciano J. Progressive multifocal leukoencephalopathy and idiopathic CD4 lymphocytopenia. *J Neurol Sci*. 2013; 327: 75-79.
390. Riancho J, Agea L, Infante J, Berciano J. [Progressive binocular diplopia]. *Med Clin (Barc)*. 2013; 141: e11.
391. Delgado-Alvarado M, Gómez-Román J, Sánchez-Salmón E, Rodríguez-Rodríguez E, Polo JM, García-Castaño A, et al. Nonanaplastic pleomorphic xanthoastrocytoma with meningeal dissemination presenting with bilateral visual loss. *J Neuroimaging*. 2014; 24: 533-535.
392. Riancho J, Delgado-Alvarado M, Sedano MJ, Polo JM, Berciano J. Herpes simplex encephalitis: clinical presentation, neurological sequelae and new prognostic factors. Ten years of experience. *Neurol Sci*. 2013; 34: 1879-1881.
393. Berciano J, Martínez-Agüeros JA, Gallardo E, Martínez-Martínez MÁ, Infante J, García A, et al. Hereditary neuropathy with liability to pressure palsy: fulminant radicular dysfunction during anterolateral lumbar interbody fusion. *J Neurol*. 2013; 260: 2411-2413.
394. Berciano J, Gallardo E. Proximal lower-limb weakness in Charcot-Marie-Tooth disease. *JAMA Neurol*. 2013; 70: 1587.
395. Berciano J. Commentary. *Mov Disord*. 2014; 29: 38-39.
396. Gazulla J, Berciano J. Multiple-system atrophy. *N Engl J Med*. 2015; 372: 1374.
397. Riancho J, Jiménez-López Y, Marco-de Lucas E, Berciano J. Sudden onset of facial diplegia and aphagia. *Rev Clin Esp (Barc)*. 2015; 215: 540-541.
398. Riancho J, Ruiz-Soto M, Berciano MT, Berciano J, Lafarga M. Neuroprotective Effect of Bexarotene in the SOD1(G93A) Mouse Model of Amyotrophic Lateral Sclerosis. *Front Cell Neurosci*. 2015; 9: 250.

399. Riancho J, Berciano MT, Ruiz-Soto M, Berciano J, Landreth G, Lafarga M. Retinoids and motor neuron disease: Potential role in amyotrophic lateral sclerosis. *J Neurol Sci.* 2016; 360: 115-120.
400. Riancho J, Berciano MT, Berciano J, Lafarga M. Relaunching an old drug: the potential role of bexarotene in neurodegenerative diseases. *J Neurol.* 2016; 263: 177-178.
401. Riancho J, Gonzalo I, Ruiz-Soto M, Berciano J. Why do motor neurons degenerate? Actualization in the pathogenesis of amyotrophic lateral sclerosis. *Neurologia (Engl Ed).* 2019; 34: 27-37.
402. Riancho J, Lozano-Cuesta P, Santurtún A, Sánchez-Juan P, López-Vega JM, Berciano J, et al. Amyotrophic lateral sclerosis in Northern Spain 40 years Later: What has changed? *Neurodegener Dis.* 2016; 16: 337-341.
403. Berciano J, Gallardo E, Orizaola P, Marco de Lucas E, García A, Pelayo-Negro AL, et al. Comment on paraparetic Guillain-Barré syndrome: Non-demyelinating reversible conduction failure restricted to the lower limbs. *Muscle Nerve.* 2017; 55: 445-446.
404. Berciano J, García A. Letter regarding the article: "A novel missense variant (Gln220Arg) of GNB4 encoding guanine nucleotide-binding protein, subunit beta-4 in a Japanese family with autosomal dominant motor and sensory neuropathy". *Eur J Med Genet.* 2018; 61: 43-44.
405. Berciano J. Progressive multifocal leukoencephalopathy in an immunocompetent patient. *Neurologia (Engl Ed).* 2020; 35: 58.
406. Berciano J. Additional arguments supporting that Franklin Delano Roosevelt's paralytic illness was related to Guillain-Barré syndrome. *J Med Biogr.* 2018; 26: 142-143.
407. Berciano J, Terán-Villagrà N. Sneddon syndrome and non-bacterial thrombotic endocarditis: a clinicopathological study. *J Neurol.* 2018; 265: 2143-2145.
408. Natera-de Benito D, Berciano J, García A, M de Lucas E, Ortez C, Nascimento A. Acute Flaccid Myelitis With Early, Severe Compound Muscle Action Potential Amplitude Reduction: A 3-Year Follow-up of a Child Patient. *J Clin Neuromuscul Dis.* 2018; 20: 100-101.
409. Berciano J, García A. Novel GARS mutation presenting as autosomal dominant intermediate Charcot-Marie-Tooth disease: Intermediate or axonal? *J Peripher Nerv Syst.* 2019; 24: 161.
410. Berciano J. Primary central nervous system lymphoma mimicking cerebellopontine angle lesion. *Neurologia (Engl Ed).* 2020; 35: 506-507.
411. Berciano J. Livedo racemosa generalisata: an anthological vision through Vladímir Lébedev painting. *J Neurol.* 2019; 266: 1801-1802.
412. Berciano J, Natera-de Benito D. Description of restrictively defined acute flaccid myelitis. *JAMA Pediatr.* 2019; 173: 701-702.
413. Sneddon IB. Cerebro-vascular lesions and livedo reticularis. *Br J Dermatol.* 1965; 77:180-185.
414. Rebollo M. Livedo reticularis idiopática: manifestaciones predominantes de una arteriopatía genéticamente determinada. Tesis doctoral, Universidad de Cantabria, 1982.
415. Natowicz M, Kelley RI. Mendelian etiologies of stroke. *Ann Neurol.* 1987; 22:175-192.
416. Zaragoza E. Malformaciones de la charnela occípito-cervical y sus correlaciones clínicas. Madrid: Ministerio de Trabajo (Instituto Nacional de Previsión), Madrid: 1974.
417. Marin-Padilla M. Study of the skull in human cranioschisis. *Acta Anat (Basel).* 1965; 62:1-20.
418. Marin-Padilla M. Mesodermal alterations induced by hypervitaminosis A. *J Embryol Exp Morphol.* 1966; 15: 261-269.

419. Marin-Padilla M, Marin-Padilla TM. Morphogenesis of experimentally induced Arnold-Chiari malformation. *J Neurol Sci.* 1981; 50:29-55.
420. Vega Bolivar A. Estudio radiológico y tomodensitométrico de la fosa craneal posterior en pacientes con la deformidad de Chiari tipo I. Tesis doctoral, Universidad de Cantabria, 1988.
421. Sahuquillo J, Rubio E, Poca MA, Rovira A, Rodriguez-Baeza A, Cervera C. Posterior fossa reconstruction: a surgical technique for the treatment of Chiari I malformation and Chiari I/syringomyelia complex-- preliminary results and magnetic resonance imaging quantitative assessment of hindbrain migration. *Neurosurgery.* 1994; 35: 874-884.
422. Leno C. Estudio prospectivo del ictus juvenil en Cantabria, Tesis dooctoral, Universidad de Cantabria, 1989.
423. Miró J. Clínica, curso y pronóstico de la Esclerosis en placas. Tesina de licenciatura, Universidad de Cantabria 1983.
424. Miró J. Esclerosis múltiple en Cantabria. Tesis doctoral, Universidad de Cantabrai, 1990.
425. Alexander E, McFarland H. Sjögren's syndrome mimicking multiple sclerosis. *Ann Neurol.* 1990; 27: 587-588.
426. López Vega JM. Epidemiología y clínica de la esclerosis lateral amiotrófica y de otras formas de enfermedad de la motoneurona en Cantabria. Estudio retrospectivo del periodo 1974-1985. Tesina de licenciatura, Universidad de Cantabria, 1986.
427. Riancho J. Efecto del bexaroteno en ratones transgénicos con esclerosis lateral amiotrófica. Tesis doctoral, Universidad de Cantabria, 2015.
428. Ohye C. Thalamus. En: Praxinos G edit. *The human nervous system*, San diego, California, Academic University Press, 1990: 439-468.
429. Polo JM, Martín J, Duyos JA, Combarros O, Berciano J. Botulism in the second half of pregnancy: a case report. Sixth Meeting of the European Neurological Society, La Haya, junio 1996. Resumen publicado en *J Neurol* 1996; 243 (suppl 2): S67.

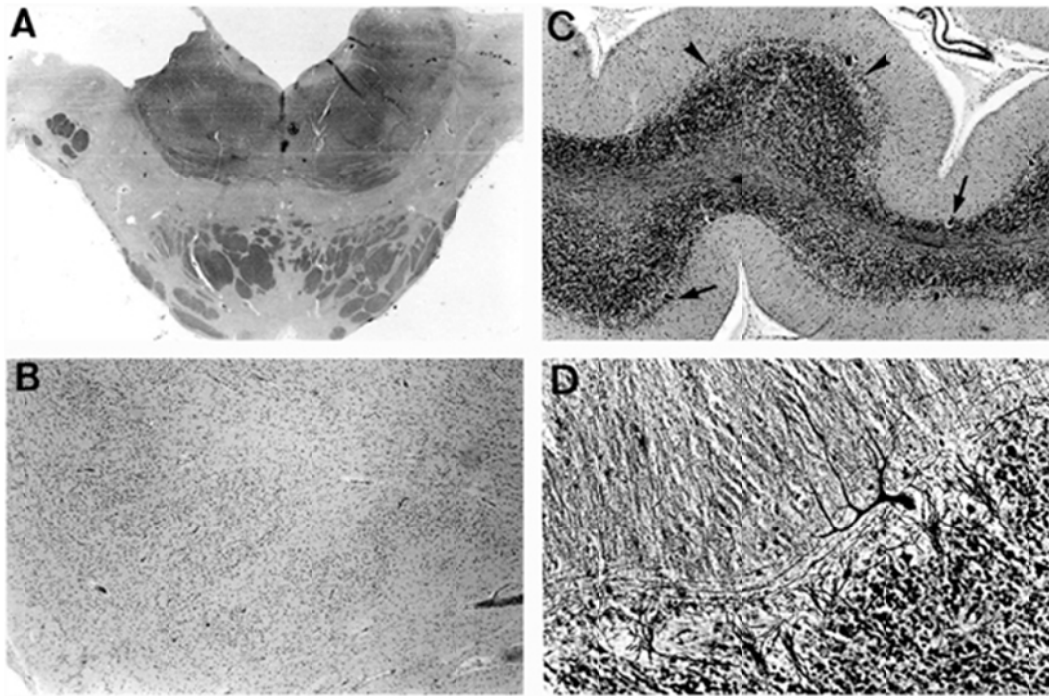


Figura 1. Hallazgos histológicos en la atrofia olivo-ponto-cerebelosa tipo Menzel¹⁹. **(A)** Sección transversa de la protuberancia en la que se observa una desmielinización completa de las fibras ponto-cerebelosas (Spielmeyer). **(B)** Detalle de la oliva bulbar con pérdida completa de neuronas; de hecho, su contorno abollonado se insinúa merced a la intensa gliosis (Nissl). **(C)** Folia cerebelosa en la que hay una pérdida casi completa de células de Purkinje (las flechas señalan dos remanentes) y acusada proliferación de la glía de Bergmann (puntas de flecha). **(D)** La pérdida de células de Purkinje da lugar a la presencia de cestas vacías (Naoumenko-Feigin).