CV Date	03/12/2021

Part A. PERSONAL INFORMATION

First Name *	Carmen		
Family Name *	Espinós Armero		
Sex *	Female	Date of Birth *	
ID number Social Security, Passport *		Phone Number *	963289680
URL Web	http://espinos.cipf.es		
Email Address	cespinos@cipf.es		
Researcher's identification number	Open Researcher and Contributor ID (ORCID) *	0000-0003-4435-1809	
	Researcher ID	A-9369-2014	
	Scopus Author ID		

* Mandatory

A.1. Current position

Job Title	Senior Independent Researcher		
Starting date	2015		
Institution	Centro de Investigación Príncipe Felipe (CIPF)		
Department / Centre	Unit of Rare Neurodegenerative Diseases / Centro de Investigación Príncipe Felipe (CIPF)		
Country		Phone Number	
Keywords	Clinical medicine and epidemiology; Molecular mechanism of disease; Cell biology; Molecular biology; Genetics		

A.3. Education

Degree/Master/PhD	University / Country	Year
PhD Human Genetics	Universitat de València	1998
Graduate Biology	Universitat de València	1993

A.4. General quality indicators of scientific production

By Publons: Publications in Web of Science: 96; Sum if times cited: 1494; Average citations per item: 17.4; Average citations per year: 62.3.

ANECA accreditation of tenured professor. Associate Professor at the Catholic University of Valencia. Professor in: Master of Research in Cellular, Molecular and Genetics Biology (UVEG), Master of Biomedical Research (UPV). Four research track records recognized by AVAP (periods: 1997-2002, 2003-2008, 2009-2014, 2015-2020).

Doctoral Thesis: 4 (4 ongoing). Master Thesis/Final Degree Project: 25 (3 ongoing). PhD fellowships funded with competitive public calls: 1 (FPU), 1 (PFIS), 2 (GVA).

AEGH accreditation of Human Genetics. Qualification A in the assessment of the Miguel Servet Program (88/100).

44 articles in the last five years (2016-2021). Complete List of Published Work in ORCID: https://orcid.org/0000-0003-4435-1809.

+info: http://www.espinos.cipf ; https://www.cipf.es/

Part C. RELEVANT ACCOMPLISHMENTS

C.1. Most important publications in national or international peer-reviewed journals, books and conferences

AC: corresponding author. (nº x / nº y): position / total authors. If applicable, indicate the number of citations

1 **Scientific paper**. M Peña-Chilet; G Roldán; J Perez-Florido; et al;. 2021. CSVS, a crowdsourcing database of the Spanish population genetic variability Nucleic Acids Research. 49, pp.D1130-D1137. ISSN 0305-1048.

2 **Scientific paper**. P Sancho; A Andrés-Bordería; N Gorría-Redondo; et al; (AC);. (14/15). 2021. Expanding the β-III spectrin-associated phenotypes toward non-progressive congenital ataxias with neurodegeneration International Journal of Molecular Sciences. 22-2505. ISSN 1422-0067.

3 **Scientific paper**. MI Vanegas; A Marce; L Martí; et al; ;. (20/23). 2020. Delineating the motor phenotype of SGCE-myoclonus dystonia syndrome Parkinsonism Relat Disord. 80, pp.165-174. ISSN 1353-8020.

4 **Scientific paper**. A Sánchez-Monteagudo; M Álvarez-Sauco; I Sastre; I Martínez-Torres; V Lupo; M Berenguer; (AC). (7/7). 2020. Genetics of Wilson disease and Wilson-like phenotype in a clinical series from eastern Spain Clinical Genetics. 97-5, pp.758-763. ISSN 0009-9163.

5 **Scientific paper**. M Correa-Vela; V Lupo; M Montpeyó; et al; (AC). (17/17). 2020. Impaired proteasome activity and neurodegeneration with brain iron accumulation in FBXO7 defect Annals of Clinical and Translational Neurology. 7, pp.1436-1442. ISSN 2328-9503.

6 **Scientific paper**. L Matalonga; S Laurie; A Papakonstantinou; et al;. 2020. Improved diagnosis of rare disease patients through systematic detection of runs of homozygosity Journal of Molecular Diagnostics. 22-9, pp.1205-1215. ISSN 1525-1578.

7 **Scientific paper**. L García-Villarreal; A Hernández-Ortega; A Sánchez-Monteagudo; et al; ;. (14/16). 2020. Wilson disease: Revision of diagnostic criteria in a clinical series with great genetic homogeneity Journal of Gastroenterology. 56, pp.78-89. ISSN 0944-1174.

8 **Scientific paper**. P Sancho; L Bartesaghi; O Miossec; et al; (AC). (15/15). 2019. Characterization of molecular mechanisms underlying the axonal Charcot-Marie-Tooth neuropathy caused by MORC2 mutations.Hum Mol Genet. 28, pp.179-186. ISSN 0964-6906.

9 **Scientific paper**. A Darling; S Aguilera; C Tello; et al; ;. (25/26). 2019. PLA2G6-associated neurodegeneration: New insights into brain abnormalities and disease progression.Parkinsonism Relat Disord. 61, pp.179-186. ISSN 1353-8020.

10 **Scientific paper**. V Lupo; M Frasquet; A Sánchez-Monteagudo; et al; ;. (15/16). 2018. Characterizing the phenotype and mode of inheritance of patients with inherited peripheral neuropathies carrying MME mutations.J Med Genet. 55, pp.814-823. ISSN 0946-2716.

11 **Scientific paper**. M Machuca-Arellano; A Vilches; E Clemente; et al; ;. (7/10). 2018. Generation of a human iPSC line from a patient with autosomal recessive spastic ataxia de Charlevoix-Saguenay (ARSACS) caused by a mutation in SACSIN gene Stem Cell Res. 31, pp.249-252. ISSN 1873-5061.

12 **Scientific paper**. E Calpena; V López del Amo; M Chakraborty; B Llamusí; R Artero; C Espinós; MI Galindo. (6/7). 2018. The Drosophila junctophilin gene is functionally equivalent to its four mammalian counterparts and is a modifier of a Huntingtin poly-Q expansion and the Notch pathway Dis Model Mech. 11-1, pp.pii:dmm029082. ISSN 1754-8403.

13 **Scientific paper**. P Sancho; A Sánchez-Monteagudo; A Collado; et al; (AC);. (8/9). 2017. A newly distal hereditary motor neuropathy caused by a rare AIFM1 mutation Neurogenetics. 18, pp.245-250. ISSN 1364-6745.

14 **Scientific paper**. A Darling; C Tello; MJ Martí; et al; ;. (39/41). 2017. Clinical Rating Scale for Pantothenate Kinase-Associated Neurodegeneration: A Pilot Study Mov Disord. 32, pp.1620-1630. ISSN 0885-3185.

15 **Scientific paper**. V Lupo; F García-García; P Sancho; et al; (AC). (14/14). 2016. Assessment of targeted next generation sequencing as a tool for the diagnosis of Charcot-Marie-Tooth and hereditary motor neuropathy J Mol Diagn. 18, pp.225-234. ISSN 1525-1578.

16 **Scientific paper**. T Sevilla; V Lupo; D Martínez-Rubio; et al; (AC). (13/13). 2016. Mutations in the MORC2 gene cause axonal Charcot-Marie-Tooth disease Brain. 139, pp.62-72. ISSN 0006-8950.

17 **Scientific paper**. D Yubero; R Montero; MA Martín; et al; ;. (23/54). 2016. Secondary coenzyme Q10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders Mitochondrion. 30, pp.51-58. ISSN 1567-7249.

18 **Scientific paper**. David Pla-Martín; Eduardo Calpena; Vincenzo Lupo; et al; (AC). (9/9). 2015. Junctophilin-1 is a modifier gene of GDAP1-related Charcot-Marie-Tooth disease Hum Mol Genet. 24, pp.213-229. ISSN 0964-6906.

19 **Scientific paper**. T Sevilla; R Sivera; Dolores Martínez-Rubio; et al; (AC). (10/10). 2015. The EGR2 gene is involved in axonal Charcot-Marie-Tooth disease Eur J Neurol. 22, pp.1548-1555. ISSN 1351-5101.

20 **Review**. A Sánchez-Monteagudo; E Ripollés; M Berenguer; (AC). (4/4). 2021. Wilson's disease: Facing the challenge of diagnosing a rare disease Biomedicines. 9, pp.1097. ISSN 2227-9059.

21 **Bibliographic review**. C Espinós; P Ferenci. (1/2). 2020. Are the new genetic tools for diagnosis of Wilson disease helpful in clinical practice? Journal of Hepatology Reports. 2, pp.100114.

22 **Bibliographic review**. I Hinarejos; C Machuca-Arellano; P Sancho; (AC). (4/4). 2020. Mitochondrial dysfunction, oxidative stress and neuroinflammation in neurodegeneration with brain iron accumulation (NBIA) Antioxidants. 9, pp.1020. ISSN 2076-3921.

23 **Bibliographic review**. (AC); MI Galindo; MA García-Gimeno; et al;. (1/12). 2020. Oxidative stress, a crossroad between rare diseases and neurodegeneration Antioxidants. 9-4, pp.313. ISSN 2076-3921.

24 **Bibliographic review**. C Tello; A Darling; V Lupo; B Pérez-Dueñas; (AC). (5/5). 2018. On the complexity of clinical and molecular bases of neurodegeneration with brain iron accumulation Clin Genet. 93, pp.731-740. ISSN 0009-9163.

C.3. Research projects and contracts

1 **Project**. PI21/00103, Clinical studies, genetic bases and prognostic biomarkers in rare neurodegenerative diseases. Instituto de Salud Carlos III. Carmen Espinós. (Centro de Investigación Príncipe Felipe). 2022-2024. 208.120 €. Principal investigator.

2 **Project**. Bases genéticas y biomarcadores pronóstico de la enfermedad de Wilson y Wilson-like. Fundació Per Amor a l'Art (FPAA). Carmen Espinós. (Centro de Investigación Príncipe Felipe). 2020-2022. 155.790,27 €. Principal investigator.

3 **Project**. Molecular basis of NBIA and NBIA-mimics. EASIGenomics. Carmen Espinós. (Centro de Investigación Príncipe Felipe). 2020-2021. Principal investigator.

4 **Project**. PROMETEO/2018/135, De genes a terapia en enfermedades neurodegenerativas y neuromusculares. Conselleria de Cultura Educació i Esport. FV Pallardó. (Universitat de València). 2018-2021. 310.406 €. Co-PI: Carmen Espinós. Consortium with six PIs. Coordinator: Federico V Pallardó. IPs: Carmen Espinós, José M. Millán, Máximo I Galindo, Teresa Sevilla, Pascual Sanz

5 **Project**. 21500, An integrative approach to develop cellular models and characterize disease mechanisms implicated in CMT2Z, a newly described axonal form of neuropathy. AFMTéléthon. Carmen Espinós. (Centro de Investigación Príncipe Felipe). 2018-2020. 185.000 €. Co-ordinator. Co-PI: Roman Chrast (Karolinska Institutet, Stockholm, Sweden)

6 **Project**. INNEST00/18/007, Biomarcadores para oncología de precisión en cáncer de pulmón, colorrectal y melanoma. Generalitat Valenciana. Carmen Espinós. (Imegen - CIPF - H. Gral Valencia). 2019-2019. 500.000 €. Team member. Consortium led by Imegen with two parthers: Research Center Principe Felipe (CIPF) and Hospital General de Valencia

7 **Project**. PI15/00187, Avanzar en el diagnóstico, la prognosis y la terapia de enfermedades neurodegenerativas raras. Instituto de Salud Carlos III. Carmen Espinós. (Centro de Investigación Príncipe Felipe). 2016-2018. 74.415 €. Principal investigator.

8 **Project**. Whole exome sequencing for clarification of rare causes of axonal Charcot-Marie-Tooth disease. BBMRI-LPC Whole Exome Sequencing Call. Carmen Espinós. (Centro de Investigación Príncipe Felipe). 2016-2018. Co-ordinator.

9 **Project**. Characterization of MORC2, a linker in hereditary sensory and motor neuroapthies. Fundación Ramón Areces. Carmen Espinós. (Centro de Investigación Príncipe Felipe). 2015-2017. 104.690 €. Principal investigator.

10 **Project**. 20143131, Neurodegeneration with Brain Iron Accumulation: Clinical Assessment and Genetic Characterization by means of a Spanish Multi-Centre Research Network.. Fundación La Marató TV3. Carmen Espinós. (Centro de Investigación Príncipe Felipe). 2015-2017. 141.036,25 €. Principal investigator. Coordinator of the total project: Belén Pérez-Dueñas (Ref. 20143130)

11 **Project**. PI12/00453, Investigación traslacional y mecanismos de enfermedad en neuropatías periféricas hereditarias. Instituto de Salud Carlos III. Carmen Espinós. (CIBER ENFERMEDADES RARAS (CIBERER)). 2013-2015. 108.900 €. Principal investigator.

12 **Project**. Translational Research, Experimental Medicine and Therapeutics on Charcot-Marie-Tooth disease (TREAT-CMT). Instituto de Salud Carlos III, International Rare Diseases Research Consortium (IRDiRC). Francesc Palau. (CIBER ENFERMEDADES RARAS (CIBERER)). 2012-2015. 214.650 €. Carmen Espinós: Leader WP2 Partner 6. Consortium with 12 research groups. Coordinator of the total project: Francesc Palau Martínez. Partner no. 6 and Leader of Workpackage 2: Carmen Espinós

13 **Project**. New forms of distal hereditary motor neuropathies (dHMNs): clinical, genetic and cellular characterization (new-dHMNs). Centro Nacional de Análisis Genómico (CNAG). Carmen Espinós. (CIBER ENFERMEDADES RARAS (CIBERER)). 2014-2014. Principal investigator.

14 **Project**. PS09/00095, Bases genéticas y fisiopatología celular de las neuropatías periféricas hereditarias. Instituto de Salud Carlos III. Carmen Espinós. (CIBER ENFERMEDADES RARAS (CIBERER)). 2010-2012. 189.365 €. Principal investigator. Subproject led by Dr. David Blesa.

15 **Project**. CP08/00053, Genética y mecanismos de enfermedad en neuropatías hereditarias. Instituto de Salud Carlos III. Carmen Espinós. (CIBER ENFERMEDADES RARAS (CIBERER)). 2009-2011. 44.952 €. Principal investigator.

16 **Project**. GVPRE/2008/117, Bases moleculares y fisiopatología de la enfermedad de Charcot-Marie-Tooth. Conselleria d'Educació. Carmen Espinós. (CIBER ENFERMEDADES RARAS (CIBERER)). 2008-2008. 22.770 €. Principal investigator.

17 **Contract**. Fellowship PhD student (FI19/00072) Instituto de Salud Carlos III. Isabel Hinarejos. 2020-01/01/2024. 82.400 €.

18 **Contract**. Fellowship Training Dual Bankia Centro de Investigación Príncipe Felipe; Bankia. Victoria Brunchú. 2020-01/01/2022.

19 **Contract**. Fellowship PhD student (ACIF/2019/171): VALi+d Program Generalitat Valenciana. Isabel Hinarejos. 2019-01/01/2020. 22.193 €.

20 **Contract**. Fellowship Training Dual Bankia Centro de Investigación Príncipe Felipe; Bankia. Andrea Català. 2018-01/01/2020.

21 **Contract**. Felloship for a Master Student Fundació Per Amor a l'Art (FPAA); Instituto Médico Valenciano. Sandra Fernández. 2017-01/01/2018. 22.788 €.

22 **Contract**. Fellowship PhD student: Internal Call Centro de Investigación Príncipe Felipe (P.I.06/2017) Centro de Investigación Príncipe Felipe. Candela Machuca. 2017-01/01/2021. 114.992 €.

23 **Contract**. Fellowship PhD student FPU (FPU15/00964) MINISTERIO DE EDUCACION Y CIENCIA. Paula Sancho. 2016-01/12/2018. 58.171 €.

24 **Contract**. Fellowship PhD student (ACIF/2015/123): VALi+d Program Generalitat Valenciana. Paula Sancho. 2015-01/01/2016. 22.193 €.

25 **Contract**. Miguel Servet type II Researcher (CPII14-00002) Instituto de Salud Carlos III. Carmen Espinós. 2014-01/01/2017. 135.000 €.

C.4. Activities of technology / knowledge transfer and results exploitation

Carmen Espinós; Ana Sánchez-Monteagudo. 202131024. Métodos y usos diagnósticos/pronósticos de la enfermedad de Wilson basados en la determinación de los niveles miRNAs Spain. 2021. Centro de Investigación Príncipe Felipe (50%) / Fundació Per Amor a l'Art (50%).