Curriculum Vitae

Personal information

Eva Bermejo-Sanchez

Work experience

1. Employer: INSTITUTE OF HEALTH CARLOS III
   - Start date: 012022
   - End date:
   - Position: Director of the Institute of Rare Diseases Research (IIER)
   - Activities: Director and Tenured Scientist of the Institute of Rare Diseases Research (IIER) Scientific Coordinator of ECEMC (Spanish Collaborative Study of Congenital Malformations) and it's Clinical Network, and Responsible for the Epidemiology and Clinical Genetics Section of ECEMC. Responsible for the Teratology Information Services (SITTE and SITE) Research Unit on Congenital Anomalies (UIAC)
   - Country: Spain
2. Employer: IRDiRC (International Rare Diseases Research Consortium)
   - Start date: 092022
   - End date: 092025
   - Position: Member of the Interdisciplinary Scientific Committee of IRDiRC
   - Activities: Research on Rare Diseases
   - Country: France
3. Employer: WHO (World Health Organization)
   - Start date: 072022
   - End date:
   - Position: Member of the WHO Technical Working Group on Burden of Birth Defects (TWGBBD)
   - Activities: Study of the burden of birth defects.
   - Country: Switzerland
4. Employer: ICORD (International Collaboration on Rare Diseases & Orphan Drugs)
   - Start date: 042021
   - End date:
   - Position: Auditor of ICORD
   - Activities: Auditor in all the meetings of the Board of ICORD.
   - Country: Spain
5. Employer: INSTITUTE OF HEALTH CARLOS III
   - Start date: 062019
   - End date:
   - Position: Professor of the Doctorate Programme of UNED_ISCIII in Biomedical Sciences and Public Health
   - Activities: Teaching for Doctorate Students
   - Country: Spain
6. Employer: Solve_RD (Solving the Unsolved Rare Diseases) Project
   - Start date: 042019
   - End date:
   - Position: Member of the Clinical Advisory Committee (CAC) of the European Rare Disease Models & Mechanisms Network (RDMM_Europe)
   - Activities: Evaluation of applications for grants for functional validation of genomic variants possibly linked to rare diseases.
   - Country: Germany
7. Employer: FEDER (Spanish Federation of Rare Diseases)
   - Start date: 052017
   - End date:
   - Position: Member of the Advisory Committee of FEDER
   - Activities: Provide advice to FEDER in any issue for which the organization asks for it to the Committee.
   - Country: Spain
8. Employer: INSTITUTE OF HEALTH CARLOS III
   - Start date: 092016
   - End date: 122021
   - Position: Chief of Area at the Institute of Rare Diseases Research (IIER) of ISCIII
   - Activities: Scientist and Chief of Area of the Institute of Rare Diseases Research (IIER) Scientific Coordinator of ECEMC (Spanish Collaborative Study of Congenital Malformations) and it's Clinical Network, and Responsible for the Epidemiology and Clinical Genetics Section of ECEMC. Responsible for the Teratology Information Services (SITTE and SITE) Research Unit on Congenital Anomalies (UIAC)
   - Country: Spain
9. Employer: Institute of Health Carlos III
   - Start date: 012019
   - End date:
   - Position: Principal Investigator for ISCIII atn the European Joint Programme on Rare Diseases (EJP RD). European Union's Horizon 2020 research and innovation programme under grant agreement N°825575.
   - Activities: European Joint Programme on Rare Diseases (EJP RD). European Union's Horizon 2020 research and innovation programme under grant agreement N°825575.
   - Country: Spain
10. Employer: Institute of Health Carlos III
    - Start date: 092016
    - End date:

- Position: Scientific Director of the National Biobank of ISCIII
- Activities: Biobanking at the Spanish "National Biobank of the Institute of Health Carlos III"
- Country: Spain

11. Employer: CIBERER
    - Start date: 122016
    - End date: 022019
    - Position: Chief of Group U724 of CIBERER
    - Activities: Chief of Group U724
    - Country: Spain
12. Employer: INSTITUTE OF HEALTH CARLOS III
    - Start date: 012009
    - End date:
    - Position: TENURED SCIENTIST of the Institute of Rare Diseases Research (IIER) at ISCIII
    - Activities: Scientist of the Institute of Rare Diseases Research (IIER) Responsible for the Epidemiology Section of ECEMC (Spanish Collaborative Study of Congenital Malformations) and Coordinator. Research Center on Congenital Anomalies (CIAC)
    - Country: Spain
13. Employer: ASEREMAC (Spanish Association for the Registry and Study of Congenital Malformations)
    - Start date: 051996
    - End date: 122008
    - Position: Researcher
    - Activities: Researcher. Responsible for the Epidemiology Section of ECEMC (Spanish Collaborative Study of Congenital Malformations) and Coordinator.
    - Country: Spain
14. Employer: INSALUD
    - Start date: 011994
    - End date: 121995
    - Position: Researcher. Specialist of Area at Hospital Clínico San Carlos
    - Activities: Researcher. Responsible for the Epidemiology Section of ECEMC (Spanish Collaborative Study of Congenital Malformations) and Coordinator.
    - Country: Spain
15. Employer: ASEREMAC
    - Start date: 121990
    - End date: 121993
    - Position: Researcher
    - Activities: Researcher in the Epidemiology Section of ECEMC (Spanish Collaborative Study of Congenital Malformations) and Responsible for the control of ECEMC's database. Member of the team of the Teratology Information Service in Spain (SITTE).
    - Country: Spain
16. Employer: ASEREMAC
    - Start date: 021987
    - End date: 121990
    - Position: Intern
    - Activities: Research as intern in the Epidemiology and Terato_epidemiology sections of ECEMC (Spanish Collaborative Study of Congenital Malformations)
    - Country: Spain

Education and training

1. Subject: Faculty of Science, Universidad Autonoma de Madrid
    - Start date: 091988
    - End date: 111994
    - Qualification: PhD
    - Organisation: Science (Genetics). Apto Cum Laude (unanimity)
    - Country: Spain
2. Subject: Faculty of Science, Universidad Autonoma de Madrid
    - Start date: 091980
    - End date: 061985
    - Qualification: Degree in Sciences (Biology) / Master in Science
    - Organisation: Biology (Genetics)
    - Country: Spain
3. Subject: Institute of Education Sciences. Univ. Autonoma de Madrid
    - Start date: 091985
    - End date: 061986
    - Qualification: Certificate of Pedagogic Aptitude
    - Organisation: Education
    - Country: Spain
4. Subject: Emory University (Atlanta, Georgia, USA)
    - Start date: 111999
    - End date: 111999
    - Qualification: Certificate Epidemiology in Action
    - Organisation: Epidemiology
    - Country: United States
5. Subject: Asociación Española de Genética Humana (AEGH)
    - Start date: 032005
    - End date:
    - Qualification: Accredited in Genetics
    - Organisation: Genetics
    - Country: Spain

Additional information

Publications

[As listed by PubMed on October 2022] Total: 131. 1: Baladron B, Mielu LM, López_Martín E, Barrero MJ, Lopez L, Alvarado JI, Monzón S, Varona S, Cuesta I, Cazorla R, Lara J, Iglesias G, Román E, Ros P, Gomez_Mariano G, Cubillo I, Miguel EH, Rivera D, Alonso J, Bermejo_Sánchez E, Posada M, Martínez_Delgado B. Differences in Expression of *IQSEC2* Transcript Isoforms in Male and Female Cases with Loss of Function Variants and Neurodevelopmental Disorder. Int J Mol Sci. 2022 Aug 22;23(16):9480. doi: 10.3390/ijms23169480. PMID: 36012761; PMCID: PMC9409358. 2: Kancherla V, Tandaki L, Sundar M, Lux A, Bakker MK, Bergman JE, Bermejo_ Sánchez E, Canfield MA, Feldkamp ML, Groisman B, Hurtado_Villa P, Källén K, Landau D, Lelong N, Lopez_Camelo J, Mastroiacovo P, Morgan M, Mutchinick OM, Nance AE, Nembhard WN, Pierini A, Šípek A, Stallings EB, Szabova E, Wertelecki W, Zarante I, Rissmann A. A Multicountry Analysis of Prevalence and Mortality among Neonates and Children with Bladder Exstrophy. Am J Perinatol. 2022 May 29. doi: 10.1055/s_0042_1748318. Epub ahead of print. PMID: 35644130. 3: Morris JK, Addor MC, Ballardini E, Barisic I, Barrachina_Bonet L, Braz P, Cavero_Carbonell C, Den Hond E, Garne E, Gatt M, Haeusler M, Khoshnood B, Lelong N, Kinsner_Ovaskainen A, Kiuru_Kuhlefelt S, Klungsoyr K, Latos_Bielenska A, Limb E, O'Mahony MT, Perthus I, Pierini A, Rankin J, Rissmann A, Rouget F, Sayers G, Sipek A Jr, Stevens S, Tucker D, Verellen_Dumoulin C, de Walle HEK, Wellesley D, Wertelecki W, Bermejo_Sanchez E.

Prevention of Neural Tube Defects in Europe: A Public Health Failure. Front Pediatr. 2021 Jun 24;9:647038. doi: 10.3389/fped.2021.647038. PMID: 34249803; PMCID: PMC8264257. 4: Bell JC, Baynam G, Bergman JEH, Bermejo_Sánchez E, Botto LD, Canfield MA, Dastgiri S, Gatt M, Groisman B, Hurtado_Villa P, Kallen K, Khoshnood B, Konrad V, Landau D, Lopez_Camelo JS, Martinez L, Morgan M, Mutchinick OM, Nance AE, Nembhard W, Pierini A, Rissmann A, Shan X, Sipek A, Szabova E, Tagliabue G, Yevtushok LS, Zarante I, Nassar N. Survival of infants born with esophageal atresia among 24 international birth defects surveillance programs. Birth Defects Res. 2021 Jul 15;113(12):945_957. doi: 10.1002/bdr2.1891. Epub 2021 Mar 18. PMID: 33734618; PMCID: PMC8273078. 5: Salinas_Vilca A, Cuevas L; ECEMC Peripheral Group, Bermejo_Sánchez E, Galán I. Smoking during pregnancy: changes and associated risk factors in Spain, 1980_2016. J Public Health (Oxf). 2022 Jun 27;44(2):438_446. doi: 10.1093/pubmed/fdaa277. PMID: 33522592. 6: Martinez_Delgado B, Lopez_Martin E, Lara_Herguedas J, Monzon S, Cuesta I, Juliá M, Aquino V, Rodriguez_Martin C, Damian A, Gonzalo I, Gomez_Mariano G, Baladron B, Cazorla R, Iglesias G, Roman E, Ros P, Tutor P, Mellor S, Jimenez C, Cabrejas MJ, Gonzalez_Vioque E, Alonso J, Bermejo_Sánchez E, Posada M. De novo small deletion affecting transcription start site of short isoform of AUTS2 gene in a patient with syndromic neurodevelopmental defects. Am J Med Genet A. 2021 Mar;185(3):877_883. doi: 10.1002/ajmg.a.62017. Epub 2020 Dec 21. PMID: 33346930. 7: Politis MD, Bermejo_Sánchez E, Canfield MA, Contiero P, Cragan JD, Dastgiri S, de Walle HEK, Feldkamp ML, Nance A, Groisman B, Gatt M, Benavides_Lara A, Hurtado_Villa P, Kallén K, Landau D, Lelong N, Lopez_Camelo J, Martinez L, Morgan M, Mutchinick OM, Pierini A, Rissmann A, Šípek A, Szabova E, Wertelecki W, Zarante I, Bakker MK, Kancherla V, Mastroiacovo P, Nembhard WN; International Clearinghouse for Birth Defects Surveillance and Research. Prevalence and mortality in children with congenital diaphragmatic hernia: a multicountry study. Ann Epidemiol. 2021 Apr;56:61_69.e3. doi: 10.1016/j.annepidem.2020.11.007. Epub 2020 Nov 27. PMID: 33253899; PMCID: PMC8009766. 8: Nembhard WN, Bergman JEH, Politis MD, Arteaga_Vázquez J, Bermejo_Sánchez E, Canfield MA, Cragan JD, Dastgiri S, de Walle HEK, Feldkamp ML, Nance A, Gatt M, Groisman B, Hurtado_Villa P, Kallén K, Landau D, Lelong N, Lopez_Camelo J, Martinez L, Morgan M, Pierini A, Rissmann A, Šípek A, Szabova E, Tagliabue G, Wertelecki W, Zarante I, Bakker MK, Kancherla V, Mastroiacovo P. A multi_country study of prevalence and early childhood mortality among children with omphalocele. Birth Defects Res. 2020 Dec;112(20):1787_1801. doi: 10.1002/bdr2.1822. Epub 2020 Oct 17. PMID: 33067932; PMCID: PMC7722785. 9: Del Castillo Velilla I, Ludeña Del Río M, López_Menchero Oliva JC, Ramos Navarro C, Bermejo_Sánchez E, Bejarano Ramírez N. Neonatal myocardial ischemia and calcifications. Report of a case of generalized arterial calcification of infancy. Rev Esp Cardiol (Engl Ed). 2021 Feb;74(2):187_189. English, Spanish. doi: 10.1016/j.rec.2020.05.036. Epub 2020 Aug 10. PMID: 32792312. 10: Abdelfattah F, Kariminejad A, Kahlert AK, Morrison PJ, Gumus E, Mathews KD, Darbro BW, Amor DJ, Walsh M, Sznajer Y, Weiß L, Weidensee S, Chitayat D, Shannon P, Bermejo_Sánchez E, Riaño_Galán I, Hayes I, Poke G, Rooryck C, Pennamen P, Khung_Savatovsky S, Toutain A, Vuillaume ML, Ghaderi_Sohi S, Kariminejad MH, Weinert S, Sticht H, Zenker M, Schanze D. Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. Hum Mutat. 2020 Sep;41(9):1615_1628. doi: 10.1002/humu.24067. Epub 2020 Jul 15. PMID: 32579715. 11: Urreizti R, Lopez_Martin E, Martinez_Monseny A, Pujadas M, Castilla_ Vallmanya L, Pérez_Jurado LA, Serrano M, Natera_de Benito D, Martínez_Delgado B, Posada_de_la_Paz M, Alonso J, Marin_Reina P, O'Callaghan M, Grinberg D, Bermejo_Sánchez E, Balcells S. Five new cases of syndromic intellectual disability due to KAT6A mutations: widening the molecular and clinical spectrum. Orphanet J Rare Dis. 2020 Feb 10;15(1):44. doi: 10.1186/s13023_020_1317_9. PMID: 32041641; PMCID: PMC7011274. 12: Bakker MK, Kancherla V, Canfield MA, Bermejo_Sanchez E, Cragan JD, Dastgiri S, De Walle HEK, Feldkamp ML, Groisman B, Gatt M, Hurtado_Villa P, Kallen K, Landau D, Lelong N, Lopez Camelo JS, Martínez L, Morgan M, Mutchinick OM, Nembhard WN, Pierini A, Rissmann A, Sipek A, Szabova E, Tagliabue G, Wertelecki W, Zarante I, Mastroiacovo P. Analysis of Mortality among Neonates and Children with Spina Bifida: An International Registry_Based Study, 2001_2012. Paediatr Perinat Epidemiol. 2019 Nov;33(6):436_448. doi: 10.1111/ppe.12589. Epub 2019 Oct 21. PMID: 31637749; PMCID: PMC6899817. 13: Taruscio D, Bermejo_Sánchez E, Salerno P, Mantovani A. Primary prevention as an essential factor ensuring sustainability of health systems: the example of congenital anomalies. Ann Ist Super Sanita. 2019 Jul_Sep;55(3):258_264. doi: 10.4415/ANN_19_03_11. PMID: 31553320. 14: Palencia_Campos A, Martínez_Fernández ML, Altunoglu U, Soto_Bielicka P, Torres A, Marín P, Aller E, Şentürk L, Berköz Ö, Yıldıran M, Kayserili H, Gil_ Camarero E, Colli_Lista G, Sanchís_Calvo A, Carretero A; ECEMC Working Group on Polydactyly, Guillén_Navarro E, López_González V, Ballesta_Martínez M, Rosell J, Aglan MS, Temtamy S, Otaify GA, Cuevas_Catalina L, Torres_Saavedra MN, Nevado J, Tenorio J, Lapunzina P, Bermejo_Sánchez E, Ruiz_Pérez VL. Heterozygous pathogenic variants in GLI1 are a common finding in isolated postaxial polydactyly A/B. Hum Mutat. 2020 Jan;41(1):265_276. doi: 10.1002/humu.23921. Epub 2019 Nov 6. PMID: 31549748. 15: Romero_Rodríguez E, Cuevas L, Simón L; ECEMC Peripheral Group, Bermejo_ Sánchez E, Galán I. Changes in Alcohol Intake During Pregnancy in Spain, 1980 to 2014. Alcohol Clin Exp Res. 2019 Nov;43(11):2367_2373. doi: 10.1111/acer.14193. Epub 2019 Oct 16. PMID: 31509616. 16: Yu X, Nassar N, Mastroiacovo P, Canfield M, Groisman B, Bermejo_Sánchez E, Ritvanen A, Kiuru_Kuhlefelt S, Benavides A, Sipek A, Pierini A, Bianchi F, Källén K, Gatt M, Morgan M, Tucker D, Canessa MA, Gajardo R, Mutchinick OM, Szabova E, Csáky_Szunyogh M, Tagliabue G, Cragan JD, Nembhard WN, Rissmann A, Goetz D, Bower C, Baynam G, Lowry RB, Leon JA, Luo W, Rouleau J, Zarante I, Fernandez N, Amar E, Dastgiri S, Contiero P, Martínez_de_Villarreal LE, Borman B, Bergman JEH, de Walle HEK, Hobbs CA, Nance AE, Agopian AJ. Hypospadias Prevalence and Trends in International Birth Defect Surveillance Systems, 1980_2010. Eur Urol. 2019 Oct;76(4):482_490. doi: 10.1016/j.eururo.2019.06.027. Epub 2019 Jul 9. PMID: 31300237; PMCID: PMC7265200. 17: Groisman B, Bermejo_Sánchez E, Romitti PA, Botto LD, Feldkamp ML, Walani SR, Mastroiacovo P. Join World Birth Defects Day. Pediatr Res. 2019 Jul;86(1):3_4. doi: 10.1038/s41390_019_0392_x. Epub 2019 Apr 9. PMID: 30965352. 18: Llamosas_Falcón L, Bermejo_Sánchez E, Sánchez_Díaz G, Villaverde_Hueso A, Posada de la Paz M, Alonso_Ferreira V. Tetralogy of Fallot in Spain: a nationwide registry_based mortality study across 36 years. Orphanet J Rare Dis. 2019 Apr 8;14(1):79. doi: 10.1186/s13023_019_1056_y. PMID: 30961612; PMCID: PMC6454694. 19: Bermejo_Sánchez E, Botto LD, Feldkamp ML, Groisman B, Mastroiacovo P. Value of sharing and networking among birth defects surveillance programs: an ICBDSR perspective. J Community Genet. 2018 Oct;9(4):411_415. doi: 10.1007/s12687_018_0387_z. Epub 2018 Sep 18. PMID: 30229536; PMCID: PMC6167257. 20: López_Martín E, Martínez_Delgado B, Bermejo_Sánchez E, Alonso J; SpainUDP Network, Posada M. SpainUDP: The Spanish Undiagnosed Rare Diseases Program. Int J Environ Res Public Health. 2018 Aug 14;15(8):1746. doi: 10.3390/ijerph15081746. PMID: 30110963; PMCID: PMC6121381. 21: Alonso_Ferreira V, Sánchez_Díaz G, Villaverde_Hueso A, Posada de la Paz M, Bermejo_Sánchez E. A Nationwide Registry_Based Study on Mortality Due to Rare Congenital Anomalies. Int J Environ Res Public Health. 2018 Aug 10;15(8):1715. doi: 10.3390/ijerph15081715. PMID: 30103420; PMCID: PMC6121521. 22: Bermejo_Sánchez E, Posada de la Paz M. Congenital Anomalies: Cluster Detection and Investigation. Adv Exp Med Biol. 2017;1031:535_557. doi: 10.1007/978_3_319_67144_4_29. PMID: 29214591. 23: Sánchez_Díaz G, Arias_Merino G, Villaverde_Hueso A, Morales_Piga A, Abaitua_ Borda I, Hens M, Bermejo_Sánchez E, Posada de la Paz M, Alonso_Ferreira V. Monitoring Huntington's Disease Mortality across a 30_Year Period: Geographic and Temporal Patterns. Neuroepidemiology. 2016;47(3_4):155_163. doi: 10.1159/000452860. Epub 2016 Nov 25. PMID: 27883994. 24: Marchegiani S, Davis T, Tessadori F, van Haaften G, Brancati F, Hoischen A, Huang H, Valkanas E, Pusey B, Schanze D, Venselaar H, Vulto_van Silfhout AT, Wolfe LA, Tifft CJ, Zerfas PM, Zambruno G, Kariminejad A, Sabbagh_Kermani F, Lee J, Tsokos MG, Lee CC, Ferraz V, da Silva EM, Stevens CA, Roche N, Bartsch O, Farndon P, Bermejo_Sanchez E, Brooks BP, Maduro V, Dallapiccola B, Ramos FJ, Chung HY, Le Caignec C, Martins F, Jacyk WK, Mazzanti L, Brunner HG, Bakkers J, Lin S, Malicdan MC, Boerkoel CF, Gahl WA, de Vries BB, van Haelst MM, Zenker M, Markello TC. Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber_Say Syndromes. Am J Hum Genet. 2015 Jul 2;97(1):99_110. doi: 10.1016/j.ajhg.2015.05.017. Epub 2015 Jun 25. PMID: 26119818; PMCID: PMC4572501. 25: Arroyo_Carrera I, de Zaldívar Tristancho MS, Bermejo_Sánchez E, Martínez_ Fernández ML, López_Lafuente A, MacDonald A, Zúñiga Á, Luis Gómez_Skarmeta J, Luisa Martínez_Frías M. Deletion 1q43_44 in a patient with clinical diagnosis of Warburg_Micro syndrome. Am J Med Genet A. 2015 Jun;167(6):1243_51. doi: 10.1002/ajmg.a.36878. Epub 2015 Apr 21. PMID: 25899426. 26: Martínez_Fernández ML, Fernández_Toral J, Llano_Rivas I, Bermejo_Sánchez E, MacDonald A, Martínez_Frías ML. Delineation of the clinically recognizable 17q22 contiguous gene deletion syndrome in a patient carrying the smallest microdeletion known to date. Am J Med Genet A. 2015 Sep;167A(9):2034_41. doi: 10.1002/ajmg.a.37117. Epub 2015 Apr 21. PMID: 25899082. 27: Taruscio D, Arriola L, Baldi F, Barisic I, Bermejo_Sánchez E, Bianchi F, Calzolari E, Carbone P, Curran R, Garne E, Gatt M, Latos_Bieleńska A, Khoshnood B, Irgens L, Mantovani A, Martínez_Frías ML, Neville A, Rißmann A, Ruggeri S, Wellesley D, Dolk H. European recommendations for primary prevention of congenital anomalies: a joined effort of EUROCAT and EUROPLAN projects to facilitate inclusion of this topic in the National Rare Disease Plans. Public Health

Genomics. 2014;17(2):115_23. doi: 10.1159/000360602. Epub 2014 Apr 3. PMID: 24714026. 28: Martínez_Frías ML, Ocejo_Vinyals JG, Arteaga R, Martínez_Fernández ML, Macdonald A, Pérez_Belmonte E, Bermejo_Sánchez E, Martínez S. Interstitial deletion 14q22.3_q23.2: genotype_phenotype correlation. Am J Med Genet A. 2014 Mar;164A(3):639_47. doi: 10.1002/ajmg.a.36330. Epub 2013 Dec 19. PMID: 24357464. 29: Martínez_Fernández ML, Bermejo_Sánchez E, Fernández B, MacDonald A, Fernández_Toral J, Martínez_Frías ML. Haploinsufficiency of BMP4 gene may be the underlying cause of Frías syndrome. Am J Med Genet A. 2014 Feb;164A(2):338_45. doi: 10.1002/ajmg.a.36224. Epub 2013 Dec 5. PMID: 24311462. 30: Vallejo OG, Benítez Sánchez Mdel C, Cánovas CS, Ontiveros JD, Ruiz Jiménez JI, Bermejo_Sánchez E, Martínez_Frías ML. Patient with disorganization syndrome: surgical procedures, pathology, and potential causes. Birth Defects Res A Clin Mol Teratol. 2013 Dec;97(12):781_5. doi: 10.1002/bdra.23203. Epub 2013 Dec 4. PMID: 24307594. 31: Carrascosa_Romero MC, Suela J, Pardal_Fernández JM, Bermejo_Sánchez E, Vidal_Company A, MacDonald A, Tébar_Gil R, Martínez_Fernández ML, Martínez_Frías ML. A 2.84 Mb deletion at 21q22.11 in a patient clinically diagnosed with Marden_Walker syndrome. Am J Med Genet A. 2013 Sep;161A(9):2281_90. doi: 10.1002/ajmg.a.35862. Epub 2013 Jul 25. PMID: 23894067. 32: Sanchis Calvo A, Roselló_Sastre E, Marcos Puig B, Balanzá Chancosa R, Pérez Ebri ML, Alcover Barrachina I, Camarasa Lillo N, Bermejo_Sánchez E, Escandón Alvarez J. Defectos congénitos en recién nacidos y fetos procedentes de interrupción del embarazo tras diagnóstico prenatal en el período 1982_2009 [Evolution of the frequency of congenital defects in newborn infants and fetuses from terminations of pregnancy after prenatal diagnosis in the period 1982_2009]. Med Clin (Barc). 2013 Aug 17;141(4):152_8. Spanish. doi: 10.1016/j.medcli.2012.05.021. Epub 2012 Jul 28. 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Projects

[Total: 30 projects and 21 subprojects. Updated on November 2021] NAME OF THE PROJECT: "Joint International Study on the Epidemiology of Hypospadias" YEARS: 1987 FUNDING AGENCY: Laboratorio Schering. MAIN OBJECTIVE OF THE PROJECT: To study the main epidemiological characteristics of hypospadias in an international setting (with participation of countries from all over the world, members of ICBDSR), also analyzing, specifically, its possible relationship with the prenatal exposure to anovulatories. PARTICIPATION IN THE PROJECT: Member of the Team, as collaborator of Prof. M.L. Martínez_Frías, dedicating 20 hours/week (Principal Investigator: Prof. Bengt Källén, from Sweden) NAME OF THE PROJECT: "Estudio caso_control del potencial efecto teratogénico de los antibióticos y corticoides en nuestro medio" / "Case_control study of the potential teratogenic effect of antibiotics and corticosteroids in our population" YEARS: 1997_1999 FUNDING AGENCY: Fondo de Investigación Sanitaria. Instituto de Salud Carlos III. Project FIS: 97/0111. MAIN OBJECTIVE OF THE PROJECT: To study consumption of these two groups of drugs during pregnancy and their potential teratogenic effect (through a case_control analysis and controlling possible confounders) in pregnant women exposed during the first trimester. PARTICIPATION IN THE PROJECT: Member of the Research Team, dedicating 30 hours per week (Principal Investigator: Prof. M.L. Martínez_Frías) NAME OF THE PROJECT: "Estudio caso_control de la exposición a benzodiacepinas y relajantes musculares durante el embarazo en nuestra población" / "Case_control study of the exposure to benzodiacepines and muscle relaxant drugs during pregnancy in our population" YEARS: 2000_2001 FUNDING AGENCY: Fondo de Investigación Sanitaria. Instituto de Salud Carlos III. Project FIS: 00/0144. MAIN OBJECTIVE OF THE PROJECT: To study consumption of these two groups of drugs during pregnancy and their potential teratogenic effect (through a case_control analysis and controlling possible confounders) in pregnant women exposed during the first trimester. PARTICIPATION IN THE PROJECT: Member of the Research Team, dedicating 30 hours per week (Principal Investigator: Prof. M.L. Martínez_Frías) NAME OF THE PROJECT: "Investigación Epidemiológica de las Anomalías Congénitas" / "Epidemiological Research of Congenital Anomalies" within the REpIER Centres Network (Network on Epidemiology, of the Research Program on Rare Diseases). YEARS: 2002_2005 FUNDING AGENCY: Network funded in the call for Thematic Cooperative Research Networks of the Instituto de Salud Carlos III. ORDEN SCO/709/2002, of March 22. Project G03/123. MAIN OBJECTIVE: To develop a research program for epidemiological research of Rare Diseases in Spain, that can provide a better knowledge of their situation in clinical, epidemiological an therapeutic terms, while it provides a more appropriate orientation for the development of social_health action guidelines. PARTICIPATION IN THE PROJECT: Researcher of Group 22 (REpIER_CIAC) (Principal Investigator of the Group: M.L. Martínez_Frías; Principal Investigator of the Project: M. Posada de la Paz). NAME OF THE PROJECT: "Investigación Epidemiológica de las Anomalías Congénitas (CIAC)" / "Epidemiological Research of Congenital Anomalies (CIAC)", within the INERGEN Network (Institute of Research on Genetically determined Rare Diseases). YEARS: 2002_2005 FUNDING AGENCY: Network funded in the call for Thematic Cooperative Research Networks of the Instituto de Salud Carlos III. ORDEN SCO/709/2002, of March 22. Project C03/05. MAIN OBJECTIVE: Epidemiological research on genetically determined congenital anomalies. PARTICIPATION IN THE PROJECT: Researcher_Responsible of Group III of the node of Instituto de Salud Carlos III (Madrid). NAME OF THE PROJECT: Convenio ISCIII_ASEREMAC 2002_2011 para "Investigación epidemiológica y causal de los defectos congénitos 2002_2011" / Agreement ISCIII_ASEREMAC 2002_2011 for "Epidemiological and causal research on congenital anomalies 2002_2011" YEARS: 2002_2011 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2002_2011. MAIN OBJECTIVE: To perform an integral approach of the research on congenital anomalies, related to their epidemiological characteristics and their causes, to try to prevent them. PARTICIPATION IN THE PROJECT: Researcher_Responsible for Clinical Genetics and Epidemiology (Principal Investigator: M.L. Martínez_Frías) NAME OF THE PROJECT: "Estudio epidemiológico descriptivo y de factores de riesgo para hernia diafragmática congénita" / "Descriptive epidemiological study and analysis on the risk factors for congenital diaphragmatic hernia" YEARS: 2005_2006 FUNDING AGENCY: Fondo de Investigación Sanitaria. Instituto de Salud Carlos III (Project PI 042661) MAIN OBJECTIVE OF THE PROJECT: To study the epidemiological characteristics of congenital diaphragmatic hernia and to analyze a series of possible risk factors to try to determine the causes of this congenital defect and promote its prevention. PARTICIPATION IN THE PROJECT: Principal Investigator NAME OF THE PROJECT: "INERGEN (Instituto de Investigación de Enfermedades Raras de base Genética): Desarrollo de una red nacional cooperativa de bancos de ADN y muestras biológicas" / "INERGEN (Institute of Research on genetically determined Rare Diseases" YEAR: 2006 FUNDING AGENCY: Fondo de Investigación Sanitaria. Instituto de Salud Carlos III (Project PI 052079). MAIN OBJECTIVE: Development and validation of procedures to collect, store and to make human biological samples available for research. PARTICIPATION IN THE PROJECT: Researcher_Collaborator (Principal Investigator: E. Rodríguez_Pinilla) NAME OF THE PROJECT: "Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER)" / "Centre for Biomedical Research on Rare Diseases as a Network (CIBERER)" YEARS: 2006_2015. FUNDING AGENCY: Instituto de Salud Carlos III. MAIN OBJECTIVE OF THE PROJECT: Incorporation to a stable structure with its own legal entity, on the field of rare diseases, to constitute the Centre for Biomedical Research as a Network (CIBER) on Rare Diseases (CIBERER). PARTICIPATION IN THE PROJECT: Researcher appointed to CIBERER Group U724 (Head of group: M.L. Martínez_Frías). NAME OF THE PROJECT: "Identificación de Defectos Congénitos de Glicosilación asociados a Malformaciones Congénitas (GLICOMAL)" / "Identification of Congenital Glycosilation Defects associated to Congenital Malformations (GLICOMAL)" YEARS: 2007_2009 FUNDING AGENCY: CIBERER (Centro de Investigación Biomédica en Red de Enfermedades Raras) / (Centre for Biomedical Research on Rare Diseases as a Network) MAIN OBJECTIVE OF THE PROJECT: Identification of patients with congenital defects of glycosilation (CDG) in neonates with congenital malformations to link malformation patterns with CDG in humans. PARTICIPATION IN THE PROJECT: Researcher_Collaborator (Principal Investigator: M. Ugarte Pérez) NAME OF THE PROJECT: "Identificación de cambios genómicos constitucionales responsables de síndromes de sobrecrecimiento y cáncer, y múltiples cánceres" / "Identification of constitutional genomic changes that are responsable for overgrowth síndromes and cáncer, and multiple cancers" YEARS:

2007_2009 FUNDING AGENCY: CIBERER (Centro de Investigación Biomédica en Red de Enfermedades Raras) / (Centre for Biomedical Research on Rare Diseases as a Network) MAIN OBJECTIVE OF THE PROJECT: To study infants wit overgrowth or adults with multiple cancers (3 or more unrelated cancers) to confirm or disregard the possible relationship between these disorders and genomic microalterations or epigenetic phenomena". PARTICIPATION IN THE PROJECT: Researcher_Collaborator (Principal Investigator: J. Benítez Ortiz). NAME OF THE PROJECT: "Aplication of DNA chips (arrays) to the identification of new genes and to the diagnosis of some genetic disorders: congenital malformations, eye diseases and epilepsy" YEARS: 2007_2009 FUNDING AGENCY: CIBERER (Centro de Investigación Biomédica en Red de Enfermedades Raras) / (Centre for Biomedical Research on Rare Diseases as a Network) MAIN OBJECTIVE OF THE PROJECT: Identification of new loci/genes linked to diverse rare diseases (congenital malformations, retinal dystrophies, genetic epilepsy) and validation of the new methodology for the diagnostic study of genetic diseases with heterogeneous etiology. PARTICIPATION IN THE PROJECT: Researcher_Collaborator (Principal Investigator: C. Ayuso García). NAME OF THE PROJECT: "Análisis clínico_epidemiológico del Registro de IVEs por defectos congénitos del ECEMC" / "Clinical_epidemiological analysis of the ECEMC 's Registry of TOPFA" YEARS: 2009_2011 FUNDING AGENCY: Instituto de Salud Carlos III (Ministerio de Ciencia e Innovación). Project TPY 028/09 (Program of Grants to Emerging Groups). MAIN OBJECTIVE OF THE PROJECT: To study the clinical characteristics and to epidemiologically analyze the cases of TOPFA registered in the ECEMC Program, given the lack of epidemiological data in Spain to this respect. PARTICIPATION IN THE PROJECT: Principal Investigator _ NAME OF THE PROJECT: "Very Rare Defects Project" (ICBDSR_VRD) YEARS: 2010_2011 FUNDING AGENCY: International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR). MAIN OBJECTIVE OF THE PROJECT: To study the clinical characteristics and to epidemiologically analyze the registered cases in different programs worldwide integrating ICBDSR, with the following very rare congenital defects: Acardia, Amelia, Cyclopia, Cloacal Exstrophy, Bladder Exstrophy, Phocomelia, Conjoined Twins, and Sirenomelia. PARTICIPATION IN THE PROJECT: ROLE IN THE SUBPROJECTS: _ Subproject on Amelia (ICBDSR_VRD_AMELIA): Principal Investigator _ Subproject on Phocomelia (ICBDSR_VRD_PHOCOMELIA): Principal Investigator _ Subproject on Cyclopia (ICBDSR_VRD_CYCLOPIA): Researcher_Collaborator (Principal Investigator: I.M. Orioli, Brasil) _ Subproject on Cloacal Exstrophy (ICBDSR_VRD_CLOACAL EXSTROPHY): Researcher_Collaborator (Principal Investigator: M. Feldkamp, EE.UU.) _ Subproject on Bladder Exstrophy (ICBDSR_VRD_BLADDER EXSTROPHY): Researcher_Collaborator (Principal Investigator: C. Siffel, EE.UU.) NAME OF THE PROJECT: "Red de Biobancos" / "Biobanks Network" YEARS: 2010_2011 FUNDING AGENCY: Instituto de Salud Carlos III. Subprogram for Thematic Cooperative Research in Health Networks, from the Strategic Action in Health. (Project: RD09/0076/00108). MAIN OBJECTIVE: Development of a network of biobanks. PARTICIPATION IN THE PROJECT: Researcher_Collaborator (Principal Investigator: M. Posada de la Paz, for IIER) NAME OF THE PROJECT: "EUROCAT Joint Action" (Ref: 2010 22 04) YEARS: 2011_2013 FUNDING AGENCY: Executive Agency for Health and Consumers. EU Health Programme 2008_2013. MAIN OBJECTIVE: To facilitate the reduction of the problem that congenital anomalies represent for public health, through the epidemiological surveillance developed in the setting of the network of birth defect registries of EUROCAT. PARTICIPATION IN THE PROJECT: Researcher of the group "Spain_Hospital Network (ECEMC)". (Principal Investigator: H. Dolk, United Kingdom) NAME OF THE PROJECT: Convenio ISCIII_ASEREMAC 2012 para "Investigación epidemiológica y causal de los defectos congénitos 2012". / Agreement ISCIII_ASEREMAC 2012 for "Epidemiological and causal research on congenital anomalies 2012" YEARS: 2012 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2012. MAIN OBJECTIVE: To perform an integral approach of the research on congenital anomalies, related to their epidemiological characteristics and their causes, to try to prevent them. PARTICIPATION IN THE PROJECT: Researcher_Collaborator responsible for Epidemiology and Clinical Genetics (Principal Investigator: M.L. Martínez_Frías). NAME OF THE PROJECT: "Spanish Rare Disease Registries Research Network" (Ref: Spain_RDR) YEARS: 2012_2014 FUNDING AGENCY: Instituto de Salud Carlos III (ISCIII) in the frame of the International Rare Disease Research Consortium (IRDiRC). MAIN OBJECTIVE: To develop the National Registry of Rare Diseases in Spain, based on one hand in the regional populational registries for the epidemiological research and health and social planning, and on the other hand in the patient registries for research in specific rare diseases. The National Registry aims to contribute to improve the prevention, diagnosis, prognosis, treatments and quality of life of the patients with rare diseases and their families. PARTICIPATION IN THE PROJECT: Researcher of IIER's group (Partner 1). (Principal Investigator: M. Posada de la Paz) NAME OF THE PROJECT: Convenio ISCIII_ASEREMAC 2013 para "Investigación epidemiológica y causal de los defectos congénitos 2013". / Agreement ISCIII_ASEREMAC 2013 for "Epidemiological and causal research on congenital anomalies 2013" YEARS: 2013 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2012. MAIN OBJECTIVE: To perform an integral approach of the research on congenital anomalies, related to their epidemiological characteristics and their causes, to try to prevent them. PARTICIPATION IN THE PROJECT: Researcher_Collaborator responsible for Epidemiology and Clinical Genetics (Principal Investigator: M.L. Martínez_Frías). NAME OF THE PROJECT: "Gestión y procesado de muestras del biobanco del IIER, y estudio genético y molecular de pacientes con enfermedades raras en el marco del IIER, y con anomalías congénitas en el marco del CIAC, del ISCIII" / "Processing of samples from IIER's biobank, and genetic and molecular study of patients with rare diseases in IIER's setting and with congenital anomalies in the Research Centre on Congenital Anomalies (CIAC), of ISCIII. YEARS: 2013_2015 FUNDING AGENCY: Instituto de Salud Carlos III (Call for hiring Technicians as support to Research in the National Health System. Nr: CA12/00295). MAIN OBJECTIVE: To give support to the research groups at IIER and CIAC, related to work in the biobank and to the genetic and molecular research on patients with congenital anomalies and other rare diseases. Hired researcher: Beatriz Baladrón Jiménez. PARTICIPATION IN THE PROJECT: Researcher_Responsible (Principal Investigator: M. Posada de la Paz). NAME OF THE PROJECT: "Investigación sobre los aspectos clínicos y etiológicos de las fisuras craneo_faciales atípicas congénitas (FCFAC)" / "Research on the clinical and etiological aspects of atypical congenital craniofacial clefts (ACCFC)". YEARS: 2013_2015 FUNDING AGENCY: Instituto de Salud Carlos III (Call for Grants to Research Projects in Health, of the Strategic Action on Health. Nr: PI12/00759). MAIN OBJECTIVE: To unveil the frequency of atypical congenital craniofacial clefts (ACCFC), analyze their clinical aspects, study their epidemiological characteristics, and investigate possible causal factors to obtain clues for their prevention, and to perform the molecular study of patients with ACCFC trying to identify genetic causes of these rare congenital anomalies. PARTICIPATION IN THE PROJECT: Principal Investigator. NAME OF THE PROJECT: Convenio ISCIII_ASEREMAC 2014 para "Investigación epidemiológica y causal de los defectos congénitos 2014". / Agreement ISCIII_ASEREMAC 2014 for "Epidemiological and causal research on congenital anomalies 2014" YEARS: 2014 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2012. MAIN OBJECTIVE: To perform an integral approach of the research on congenital anomalies, related to their epidemiological characteristics and their causes, to try to prevent them. PARTICIPATION IN THE PROJECT: Researcher_Collaborator responsible for Epidemiology and Clinical Genetics (Principal Investigator: M.L. Martínez_Frías). NAME OF THE PROJECT: "Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER)" / "Centre for Biomedical Research on Rare Diseases as a Network" YEARS: 2015_2019. FUNDING AGENCY: Instituto de Salud Carlos III. MAIN OBJECTIVE OF THE PROJECT: Incorporation to a stable structure with its own legal entity, on the field of rare diseases, to constitute the Centre for Biomedical Research as a Network (CIBER) on Rare Diseases (CIBERER). PARTICIPATION IN THE PROJECT: Head of CIBERER Group U724. NAME OF THE PROJECT: Convenio ISCIII_ASEREMAC 2015 para "Investigación epidemiológica y causal de los defectos congénitos 2015". / Agreement ISCIII_ASEREMAC 2015 for "Epidemiological and causal research on congenital anomalies 2015" YEARS: 2015 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2015. MAIN OBJECTIVE: To perform an integral approach of the research on congenital anomalies, related to their epidemiological characteristics and their causes, to try to prevent them. PARTICIPATION IN THE PROJECT: Researcher_Collaborator responsible for Epidemiology and Clinical Genetics (Principal Investigator: M.L. Martínez_Frías). NAME OF THE PROJECT: "ICBDSR Mortality Study" YEARS: 2015_2020 FUNDING AGENCY: International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR). MAIN OBJECTIVE OF THE PROJECT: The final aim of this study is to evaluate the mortality as part of the burden of congenital anomalies. It is planned to examine and describe the mortality of selected non_cardiac malformations in different programs worldwide integrating ICBDSR, including: Hydrocephalus, spina bifida, cleft palate, Robin sequence, cleft lip without cleft palate, cleft lip with cleft palate, esophageal atresia, small intestinal atresia/stenosis, anorectal atresia, bladder exstrophy, diaphragmatic hernia, omphalocele, gastroschisis, Down sundrome, trisomy 18 and trisomy 13. PARTICIPATION IN THE PROJECT: Refinement of the proposal and coordination of the subprojects as a first step. ROLE IN THE SUBPROJECTS: Researcher_Collaborator NAME OF THE PROJECT: Convenio ISCIII_ASEREMAC 2016 para "Investigación epidemiológica y causal de los defectos congénitos 2016". / Agreement ISCIII_ASEREMAC 2016 for "Epidemiological and causal research on congenital anomalies 2016" YEARS: 2016 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2016. MAIN OBJECTIVE: To perform

an integral approach of the research on congenital anomalies, related to their epidemiological characteristics and their causes, to try to prevent them. PARTICIPATION IN THE PROJECT: Researcher_Collaborator responsible for Epidemiology, Clinical Genetics and Clinical Teratology (Principal Investigator: M.L. Martínez_Frías). NAME OF THE PROJECT: Convenio ISCIII_ASEREMAC 2017 para "Investigación epidemiológica y causal de los defectos congénitos 2017". / Agreement ISCIII_ASEREMAC 2017 for "Epidemiological and causal research on congenital anomalies 2017" YEARS: 2017 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2017. MAIN OBJECTIVE: To perform an integral approach of the research on congenital anomalies, related to their epidemiological characteristics and their causes, to try to prevent them. PARTICIPATION IN THE PROJECT: Technical Coordinator & Principal Investigator. NAME OF THE PROJECT: Convenio ISCIII_ASEREMAC 2018 para "Investigación epidemiológica y causal de los defectos congénitos 2018". / Agreement ISCIII_ASEREMAC 2018 for "Epidemiological and causal research on congenital anomalies 2018" YEARS: 2018 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2018. MAIN OBJECTIVE: To perform an integral approach of the research on congenital anomalies, related to their epidemiological characteristics and their causes, to try to prevent them. PARTICIPATION IN THE PROJECT: Technical Coordinator & Principal Investigator. NAME OF THE PROJECT: "Agreement ISCIII_ASEREMAC (Spanish Association for the Registry and Study of Congenital Malformations) for the sustainment of a research unit". YEARS: 2019_2022 FUNDING AGENCY: Instituto de Salud Carlos III. Agreement ISCIII_ASEREMAC 2019_2022. MAIN OBJECTIVE: To collaborate with the aim of ensuring the decrease of congenital anomalies through their research and prevention. PARTICIPATION IN THE PROJECT: Technical Coordinator & Principal Investigator. _ NAME OF THE PROJECT: "European Joint Programme on Rare Diseases – EJP RD" YEARS: 2019_2023 FUNDING AGENCY: European Union (GA 825575). MAIN OBJECTIVE OF THE PROJECT: The European Joint Programme on RD (EJP RD) has two major objectives: (i) To improve the integration, the efficacy, the production and the social impact of research on RD through the development, demonstration and promotion of Europe/ world_wide sharing of research and clinical data, materials, processes, knowledge and know_how; (ii) To implement and further develop an efficient model of financial support for all types of research on RD (fundamental, clinical, epidemiological, social, economic, health service) coupled with accelerated exploitation of research results for benefit of patients. To this end, the EJP RD actions will be organized within four major Pillars assisted by the central coordination: (P1): Funding of research; (P2): Coordinated access to data and services; (P3) Capacity building; (P4): Accelerated translation of research projects and improvement outcomes of clinical studies. PARTICIPATION IN THE PROJECT: Principal Investigator for ISCIII at the EJP RD.

Memberships

_ Member of the Executive Committee of the European Joint Programme on Rare Diseases, for the years 2019_2023. _ Elected Chair of the Executive Committee of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR), since September 2015 to November 2017. Elected Vice_Chair of the Executive Committee of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR), since December 2013 to September 2015. _ Elected Secretary_Treasurer of the Executive Committee of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR), since September 2011 to December 2013. _ Evaluator of the Spanish National Agency of Evaluation and Prospective (ANEP). _ Member of the Spanish Association for the Registry and Study of Congenital Malformations (ASEREMAC) _ Member of the Spanish Association of Human Genetics (AEGH) _ Participation as professor in more tan 100 activities of CME (Continued Medical Education). _ More than 100 Presentations at Scientific Conferences. _ Organization of 36 Scientific Conferences

Other Relevant Information

RESEARCH AWARDS: 1. "Premio Reina Sofía 1988 de Investigación sobre Prevención de las Deficiencias", awarded to Dr. M.L. Martínez_Frías and the Research Group of the Spanish Collaborative Study of Congenital Malformations (ECEMC). 2. "Premio CERMI.ES de investigación científica y social 2004", awarded to María Luisa Martínez_Frías and her research group at the Research Center on Congenital Anomalies (CIAC). 3. "2018 Distinguished Service Award for longstanding excellence, service and leadership. International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR)", awarded to Eva Bermejo_Sánchez.