EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

Curriculum Vitae

PERSONAL INFORMATION

Eva Bermejo-Sanchez

WORK EXPERIENCE

September 2016- Present

Chief of Area

INSTITUTE OF HEALTH CARLOS III (Spain)

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)

March 2018- Present

Principal Investigator for ISCIII

Institute of Health Carlos III (Spain)

European Joint Programme on Rare Diseases (EJP RD). European Union's Horizon 2020 research and innovation programme under grant agreement N°825575.

September 2016- Present

Scientific Director

Institute of Health Carlos III (Spain)

Biobanking at the Spanish "National Biobank of the Institute of Health Carlos III"

December 2016-February 2019

Chief of Group U724

CIBERER (Spain)

Chief of Group U724

January 2009- Present

TENURED SCIENTIST

INSTITUTE OF HEALTH CARLOS III (Spain)

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)

May 1996-December 2008

Researcher

ASEREMAC (Spanish Association for the Registry and Study of Congenital Malformations) (Spain)

Researcher.

Responsible for the Epidemiology Section of ECEMC (Spanish Collaborative Study of Congenital Malformations) and Coordinator.

January 1994-December 1995

Researcher. Specialist of Area

INSALUD (Spain)

Researcher.

Responsible for the Epidemiology Section of ECEMC (Spanish Collaborative Study of Congenital Malformations) and Coordinator.

December 1990-December 1993

Researcher

ASEREMAC (Spain)

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).

February 1987-December 1990

Intern

ASEREMAC (Spain)

Research as intern in the Epidemiology and Terato-epidemiology sections of ECEMC (Spanish Collaborative Study of Congenital Malformations)

EDUCATION AND TRAINING

September 1988-November 1994

PhD

Faculty of Science, Universidad Autonoma de Madrid (Spain)

Science (Genetics). Apto Cum Laude (unanimity)

September 1980-June 1985

Degree in Sciences (Biology) / Master in Science

Faculty of Science, Universidad Autonoma de Madrid (Spain)

Biology (Genetics)

September 1985-June 1986

Certificate of Pedagogic Aptitude

Institute of Education Sciences. Univ. Autonoma de Madrid (Spain)

Education

November 1999-November 1999

Certificate Epidemiology in Action

Emory University (Atlanta, Georgia, USA) (United States)

Epidemiology

March 2005-

Accredited in Genetics

Asociación Española de Genética Humana (AEGH) (Spain)

Genetics

ADDITIONAL INFORMATION

Expertise

- Epidemiology of congenital defects
- Clinical Genetics of congenital defects
- Clinical Teratology
- Dysmorphology
- Prenatal development
- Rare diseases

Publications

[As listed by PubMed on November 2020] Total: 125.

1: Politis MD, Bermejo-Sánchez E, Canfield MA, Contiero P, Cragan JD, Dastgiri S, de Walle HE, 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, ípek 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 Multi-Country Study. Ann Epidemiol. 2020 Nov 27:S1047-2797(20)30415-4. doi:10.1016/j.annepidem.2020.11.007. Online ahead of print.PMID: 33253899

2: 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, ípek 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

3: 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). 2020 Aug 10:S1885-5857(20)30318-2. English, Spanish. doi: 10.1016/j.rec.2020.05.036. Epub ahead of print. PMID: 32792312.

4: 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 Jun 24. doi: 10.1002/humu.24067. Epub ahead of print. PMID: 32579715.

5: Urreizti R, Lopez-Martin E, Martinez-Monseny A, Pujadas M, Castilla-VallmanyaL, 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.

6: 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.

7: 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.

8: Palencia-Campos A, Martínez-Fernández ML, Altunoglu U, Soto-Bielicka P, Torres A, Marín P, Aller E, entürk L, Berköz Ö, Yldran 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.

9: 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.

10: 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.

11: 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.

12: 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 36years. Orphanet J Rare Dis. 2019 Apr 8;14(1):79. doi: 10.1186/s13023-019-1056-y. PMID: 30961612; PMCID: PMC6454694.
13: 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.
14: 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.
15: 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.
16: 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.
17: 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.
18: 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.
19: 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.
20: 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.
21: Taruscio D, Arriola L, Baldi F, Barisic I, Bermejo-Sánchez E, Bianchi F, Calzolari E, Carbone P, Curran R, Garne E, Gatt M, Latos-Bieleska 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.
22: 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.
23: 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.

24: 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.
25: 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.
26: 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. PMID: 22841468.
27: Orioli IM, Amar E, Bakker MK, Bermejo-Sánchez E, Bianchi F, Canfield MA, Clementi M, Correa A, Csáky-Szunyogh M, Feldkamp ML, Landau D, Leoncini E, Li Z, Lowry RB, Mastroiacovo P, Morgan M, Mutchinick OM, Rissmann A, Ritvanen A, Scarano G, Szabova E, Castilla EE. Cyclopia: an epidemiologic study in a large dataset from the International Clearinghouse of Birth Defects Surveillance and Research. Am J Med Genet C Semin Med Genet. 2011 Nov 15;157C(4):344-57. doi: 10.1002/ajmg.c.30323. Epub 2011 Oct 17. PMID: 22006661; PMCID: PMC4484722.
28: Bermejo-Sánchez E, Cuevas L, Amar E, Bakker MK, Bianca S, Bianchi F, Canfield MA, Castilla EE, Clementi M, Cocchi G, Feldkamp ML, Landau D, Leoncini E, Li Z, Lowry RB, Mastroiacovo P, Mutchinick OM, Rissmann A, Ritvanen A, Scarano G, Siffel C, Szabova E, Martínez-Frías ML. Amelia: a multi-center descriptive epidemiologic study in a large dataset from the International Clearinghouse for Birth Defects Surveillance and Research, and overview of the literature. Am J Med Genet C Semin Med Genet. 2011 Nov 15;157C(4):288-304. doi: 10.1002/ajmg.c.30319. Epub 2011 Oct 14. PMID: 22002956; PMCID: PMC4453759.
29: Feldkamp ML, Botto LD, Amar E, Bakker MK, Bermejo-Sánchez E, Bianca S, Canfield MA, Castilla EE, Clementi M, Csaky-Szunyogh M, Leoncini E, Li Z, Lowry RB, Mastroiacovo P, Merlob P, Morgan M, Mutchinick OM, Rissmann A, Ritvanen A, Siffel C, Carey JC. Cloacal exstrophy: an epidemiologic study from the International Clearinghouse for Birth Defects Surveillance and Research. Am J Med Genet C Semin Med Genet. 2011 Nov 15;157C(4):333-43. doi: 10.1002/ajmg.c.30317. Epub 2011 Oct 14. PMID: 22002951.
30: Siffel C, Correa A, Amar E, Bakker MK, Bermejo-Sánchez E, Bianca S, Castilla EE, Clementi M, Cocchi G, Csáky-Szunyogh M, Feldkamp ML, Landau D, Leoncini E, Li Z, Lowry RB, Marengo LK, Mastroiacovo P, Morgan M, Mutchinick OM, Pierini A, Rissmann A, Ritvanen A, Scarano G, Szabova E, Olney RS. Bladder exstrophy: an epidemiologic study from the International Clearinghouse for Birth Defects Surveillance and Research, and an overview of the literature. Am J Med Genet C Semin Med Genet. 2011 Nov 15;157C(4):321-32. doi: 10.1002/ajmg.c.30316. Epub 2011 Oct 14. PMID: 22002949; PMCID: PMC4512232.
31: Bermejo-Sánchez E, Cuevas L, Amar E, Bianca S, Bianchi F, Botto LD, Canfield MA, Castilla EE, Clementi M, Cocchi G, Landau D, Leoncini E, Li Z, Lowry RB, Mastroiacovo P, Mutchinick OM, Rissmann A, Ritvanen A, Scarano G, Siffel C, Szabova E, Martínez-Frías ML. Phocomelia: a worldwide descriptive epidemiologic study in a large series of cases from the International Clearinghouse for Birth Defects Surveillance and Research, and overview of the literature. Am J Med Genet C Semin Med Genet. 2011 Nov 15;157C(4):305-20. doi: 10.1002/ajmg.c.30320. Epub 2011 Oct 14. PMID: 22002800; PMCID: PMC4427055.
32: Palomares M, Delicado A, Mansilla E, de Torres ML, Vallespín E, Fernandez L, Martinez-Glez V, García-Miñaur S, Nevado J, Simarro FS, Ruiz-Perez VL, Lynch SA, Sharkey FH, Thuresson AC, Annerén G, Belligni EF, Martínez-Fernández ML, Bermejo E, Nowakowska B, Kutkowska-Kazmierczak A, Bocian E, Obersztyn E, Martínez-Frías ML, Hennekam RC, Lapunzina P. Characterization of a 8q21.11 microdeletion syndrome associated with intellectual disability and a

recognizable phenotype. Am J Hum Genet. 2011 Aug 12;89(2):295-301. doi: 10.1016/j.ajhg.2011.06.012. Epub 2011 Jul 28. PMID: 21802062; PMCID: PMC3155189.
33: Martínez-Frías ML, Egüés X, Puras A, Hualde J, de Frutos CA, Bermejo E, Nieto MA, Martínez S. Thanatophoric dysplasia type II with encephalocele and semilobar holoprosencephaly: Insights into its pathogenesis. Am J Med Genet A. 2011 Jan;155A(1):197-202. doi: 10.1002/ajmg.a.33765. PMID: 21204232.
34: Bermejo E, Martínez-Frías ML. Prevention, diagnosis and services. Adv Exp Med Biol. 2010;686:55-75. doi: 10.1007/978-90-481-9485-8_4. PMID: 20824439.
35: Martínez-Frías ML, de Frutos CA, Bermejo E, Nieto MA; ECEMC Working Group. Review of the recently defined molecular mechanisms underlying thanatophoric dysplasia and their potential therapeutic implications for achondroplasia. Am J Med Genet A. 2010 Jan;152A(1):245-55. doi: 10.1002/ajmg.a.33188. PMID: 20034074.
36: Martínez-Frías ML, Bermejo E, Mendioroz J, Rodríguez-Pinilla E, Blanco M, Egüés J, Félix V, García A, Huertas H, Nieto C, López JA, López S, Paisán L, Rosa A, Vázquez MS. Epidemiological and clinical analysis of a consecutive series of conjoined twins in Spain. J Pediatr Surg. 2009 Apr;44(4):811-20. doi: 10.1016/j.jpedsurg.2008.07.002. PMID: 19361646.
37: Mendioroz J, Bermejo E, Marshall JD, Naggert JK, Collin GB, Martínez-Frías ML. Presentación de un caso con síndrome de Alström: aspectos clínicos, moleculares y guías diagnósticas y anticipatorias [Alström syndrome: clinical and genetic features, and a diagnostic guide to foresee complications]. Med Clin (Barc). 2008 Nov 29;131(19):741-6. Spanish. doi: 10.1016/s0025-7753(08)75490-3. PMID: 19091203.
38: Rittler M, López-Camelo JS, Castilla EE, Bermejo E, Cocchi G, Correa A, Csaky-Szunyogh M, Danderfer R, De Vigan C, De Walle H, da Graça Dutra M, Hirahara F, Martínez-Frías ML, Merlob P, Mutchinick O, Ritvanen A, Robert-Gnansia E, Scarano G, Siffel C, Stoll C, Mastroiacovo P. Preferential associations between oral clefts and other major congenital anomalies. Cleft Palate Craniofac J. 2008 Sep;45(5):525-32. doi: 10.1597/06-250.1. PMID: 18788868.
39: Martínez-Frías ML, Bermejo E, Pérez B, Desviat LR, Castro M, Leal F, Mansilla E, Martínez-Fernández ML, Rodríguez-Pinilla E, Rodríguez L, Ugarte M; Grupo de Trabajo del Estudio Colaborativo Español de Malformaciones Congénitas (ECEMC). Análisis de las frecuencias de todas las combinaciones genotípicas de 4 polimorfismos de genes implicados en el ciclo del folato en la población española [Analysis of the frequencies of genotype combinations of 4 polymorphisms of genes acting on the folate cycle in the Spanish population]. Med Clin (Barc). 2008 Jun 21;131(3):81-8. Spanish. doi: 10.1157/13124010. PMID: 18590621.
40: Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto D; ECEMC Working Group. Does single umbilical artery (SUA) predict any type of congenital defect? Clinical-epidemiological analysis of a large consecutive series of malformed infants. Am J Med Genet A. 2008 Jan 1;146A(1):15-25. doi: 10.1002/ajmg.a.31911. PMID: 17999408.
41: Mastroiacovo P, Lisi A, Castilla EE, Martínez-Frías ML, Bermejo E, Marengo L, Kucik J, Siffel C, Halliday J, Gatt M, Annerèn G, Bianchi F, Canessa MA, Danderfer R, de Walle H, Harris J, Li Z, Lowry RB, McDonell R, Merlob P, Metneki J, Mutchinick O, Robert-Gnansia E, Scarano G, Sipek A, Pötzsch S, Szabova E, Yevtushok L. Gastroschisis and associated defects: an international study. Am J Med Genet A. 2007 Apr 1;143A(7):660-71. doi: 10.1002/ajmg.a.31607. PMID: 17357116.
42: Rouhani P, Fleming LE, Frías J, Martínez-Frías ML, Bermejo E, Mendioroz J. Pilot study of socioeconomic class, nutrition and birth defects in Spain. Matern Child Health J. 2007 Jul;11(4):403-5. doi: 10.1007/s10995-007-0186-3. Epub 2007 Feb 21. PMID: 17318404.
43: Martínez-Frías ML, Cormier-Daire V, Cohn DH, Mendioroz J, Bermejo E, Mansilla E. Síndrome de Dyggve-Melchior-Clausen: presentación de un caso con una mutación de posible origen español [Dyggve-Melchior-Clausen syndrome: presentation of a case with a mutation of possible Spanish origin]. Med Clin (Barc). 2007 Feb 3;128(4):137-40. Spanish. doi: 10.1157/13098019. PMID: 17288936.
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Projects

[Total: 30 projects and 21 subprojects. Updated on October 2020]

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.)

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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)

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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)

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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).

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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 IIERs group (Partner 1). (Principal Investigator: M. Posada de la Paz)

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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 IIERs biobank, and genetic and molecular study of patients with rare diseases in IIERs 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.