

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 its 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)
- January 2019- 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 2021] Total: 129.
1. 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. eCollection 2021. PMID: 34249803 .
 2. Bell JC, Baynam G, Bergman JEH, Bermejo-Sanchez 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 Free PMC article.
 3. Salinas-Vilca A, Cuevas L; ECEMC Peripheral Group, Bermejo-Sanchez E, Galan I. Smoking during pregnancy: changes and associated risk factors in Spain, 1980-2016. *J Public Health (Oxf).* 2021 Feb 1;fdaa277. doi: 10.1093/pubmed/fdaa277. Online ahead of print. PMID: 33522592
 4. Martinez-Delgado B, Lopez-Martin E, Lara-Herguedas J, Monzon S, Cuesta I, Julia 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-Sanchez 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
 5. Politis MD, Bermejo-Sanchez 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, Kallen K, Landau D, Lelong N, Lopez-Camelo J, Martinez L, Morgan M, Mutchinick OM, Pierini A, Rissmann A, Sipek A, Szabova E, Wertelecki W, Zarante I, Bakker MK, Kancharla 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

6: Nembhard WN, Bergman JEH, Politis MD, Arteaga-Vazquez J, Bermejo-Sanchez E, Canfield MA, Cragan JD, Dastgiri S, de Walle HEK, Feldkamp ML, Nance A, Gatt M, Groisman B, Hurtado-Villa P, Kallen K, Landau D, Lelong N, Lopez-Camelo J, Martinez L, Morgan M, Pierini A, Rissmann A, Sipek 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

7: Del Castillo Velilla I, Ludena Del Rio M, Lopez-Menchero Oliva JC, Ramos Navarro C, Bermejo-Sanchez E, Bejarano Ramirez 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.

8: Abdelfattah F, Kariminejad A, Kahlert AK, Morrison PJ, Gumus E, Mathews KD, Darbro BW, Amor DJ, Walsh M, Sznajder Y, Weiss L, Weidensee S, Chitayat D, Shannon P, Bermejo-Sanchez E, Riano-Galan 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.

9: Urreizti R, Lopez-Martin E, Martinez-Monseny A, Pujadas M, Castilla-VallmanyaL, Perez-Jurado LA, Serrano M, Natera-de Benito D, Martinez-Delgado B, Posada-de-la-Paz M, Alonso J, Marin-Reina P, O'Callaghan M, Grinberg D, Bermejo-Sanchez 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.

10: 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, Martinez 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.

11: Taruscio D, Bermejo-Sanchez 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.

12: Palencia-Campos A, Martinez-Fernandez ML, Altunoglu U, Soto-Bielicka P, Torres A, Marin P, Aller E, Senturk L, Berköz O, Yildiran M, Kayserili H, Gil-Camarero E, Colli-Lista G, Sanchis-Calvo A, Carretero A; ECEMC Working Group on Polydactyly, Guillen-Navarro E, Lopez-Gonzalez V, Ballesta-Martinez M, Rosell J, Aglan MS, Temtamy S, Otaify GA, Cuevas-Catalina L, Torres-Saavedra MN, Nevado J, Tenorio J, Lapunzina P, Bermejo-Sanchez E, Ruiz-Perez 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.

13: Romero-Rodriguez E, Cuevas L, Simon L; ECEMC Peripheral Group, Bermejo-Sanchez E, Galan 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.

14: Yu X, Nassar N, Mastroiacovo P, Canfield M, Groisman B, Bermejo-Sanchez E, Ritvanen A, Kiuru-Kuhlefelt S, Benavides A, Sipek A, Pierini A, Bianchi F, Kallen K, Gatt M, Morgan M, Tucker D, Canessa MA, Gajardo R, Mutchinick OM, Szabova E, Csaky-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, Martinez-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.

15: Groisman B, Bermejo-Sanchez 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.

16: Llamosas-Falcon L, Bermejo-Sanchez E, Sanchez-Diaz 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.
- 17: Bermejo-Sanchez 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.
- 18: Lopez-Martin E, Martinez-Delgado B, Bermejo-Sanchez 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.
- 19: Alonso-Ferreira V, Sanchez-Diaz G, Villaverde-Hueso A, Posada de la Paz M, Bermejo-Sanchez 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.
- 20: Bermejo-Sanchez 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.
- 21: Sanchez-Diaz G, Arias-Merino G, Villaverde-Hueso A, Morales-Piga A, Abaitua-Borda I, Hens M, Bermejo-Sanchez 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.
- 22: 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, Tiftt 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.
- 23: Arroyo-Carrera I, de Zaldivar Tristancho MS, Bermejo-Sanchez E, Martinez-Fernandez ML, Lopez-Lafuente A, MacDonald A, Zuniga A, Luis Gomez-Skarmeta J, Luisa Martinez-Frias 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.
- 24: Martinez-Fernandez ML, Fernandez-Toral J, Llano-Rivas I, Bermejo-Sanchez E, MacDonald A, Martinez-Frias 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.
- 25: Taruscio D, Arriola L, Baldi F, Barisic I, Bermejo-Sanchez E, Bianchi F, Calzolari E, Carbone P, Curran R, Garne E, Gatt M, Latos-Bielenska A, Khoshnood B, Irgens L, Mantovani A, Martinez-Frias 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.
- 26: Martinez-Frias ML, Ocejo-Vinyals JG, Arteaga R, Martinez-Fernandez ML, Macdonald A, Perez-Belmonte E, Bermejo-Sanchez E, Martinez 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.
- 27: Martinez-Fernandez ML, Bermejo-Sanchez E, Fernandez B, MacDonald A, Fernandez-Toral J, Martinez-Frias ML. Haploinsufficiency of BMP4 gene may be the underlying cause of Frias syndrome. *Am J Med Genet A.* 2014 Feb;164A(2):338-45. doi: 10.1002/ajmg.a.36224. Epub 2013 Dec 5. PMID: 24311462.
- 28: Vallejo OG, Benitez Sanchez Mdel C, Canovas CS, Ontiveros JD, Ruiz Jimenez JI, Bermejo-Sanchez E, Martinez-Frias 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.
- 29: Carrascosa-Romero MC, Suela J, Pardal-Fernandez JM, Bermejo-Sanchez E, Vidal-Company A, MacDonald A, Tebar-Gil R, Martinez-Fernandez ML, Martinez-Frias 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.

- 30: Sanchis Calvo A, Rosello-Sastre E, Marcos Puig B, Balanza Chancosa R, Perez Ebri ML, Alcover Barrachina I, Camarasa Lillo N, Bermejo-Sanchez E, Escandon Alvarez J. Defectos congenitos en recién nacidos y fetos procedentes de interrupción del embarazo tras diagnóstico prenatal en el periodo 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.
- 31: Orioli IM, Amar E, Bakker MK, Bermejo-Sanchez E, Bianchi F, Canfield MA, Clementi M, Correa A, Csaky-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.
- 32: Bermejo-Sanchez 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, Martinez-Frias 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.
- 33: Feldkamp ML, Botto LD, Amar E, Bakker MK, Bermejo-Sanchez 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.
- 34: Siffel C, Correa A, Amar E, Bakker MK, Bermejo-Sanchez E, Bianca S, Castilla EE, Clementi M, Cocchi G, Csaky-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.
- 35: Bermejo-Sanchez 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, Martinez-Frias 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.
- 36: Palomares M, Delicado A, Mansilla E, de Torres ML, Vallespin E, Fernandez L, Martinez-Glez V, Garcia-Minaur S, Nevado J, Simarro FS, Ruiz-Perez VL, Lynch SA, Sharkey FH, Thuresson AC, Anneren G, Belligni EF, Martinez-Fernandez ML, Bermejo E, Nowakowska B, Kutkowska-Kazmierczak A, Bocian E, Obersztyn E, Martinez-Frias 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.
- 37: Martinez-Frias ML, Egues X, Puras A, Hualde J, de Frutos CA, Bermejo E, Nieto MA, Martinez 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.
- 38: Bermejo E, Martinez-Frias ML. Prevention, diagnosis and services. *Adv Exp Med Biol*. 2010;686:55-75. doi: 10.1007/978-90-481-9485-8_4. PMID: 20824439.
- 39: Martinez-Frias 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.
- 40: Martinez-Frias ML, Bermejo E, Mendioroz J, Rodriguez-Pinilla E, Blanco M, Egues J, Felix V, Garcia A, Huertas H, Nieto C, Lopez JA, Lopez S, Paisan L, Rosa A, Vazquez 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.

41: Mendioroz J, Bermejo E, Marshall JD, Naggert JK, Collin GB, Martinez-Frias ML. Presentacion de un caso con sindrome de Alstrom: aspectos clinicos, moleculares y guias diagnosticas y anticipatorias [Alstrom 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.

42: Rittler M, Lopez-Camelo JS, Castilla EE, Bermejo E, Cocchi G, Correa A, Csaky-Szunyogh M, Danderfer R, De Vigan C, De Walle H, da Graca Dutra M, Hirahara F, Martinez-Frias 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.

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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 ICBSR), also analyzing, specifically, its possible relationship with the prenatal exposure to anovulatory agents.

PARTICIPATION IN THE PROJECT: Member of the Team, as collaborator of Prof. M.L. Martinez-Frias, dedicating 20 hours/week (Principal Investigator: Prof. Bengt Kallen, from Sweden)

NAME OF THE PROJECT: "Estudio caso-control del potencial efecto teratogenico de los antibioticos 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 Investigacion 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. Martinez-Frias)

NAME OF THE PROJECT: "Estudio caso-control de la exposicion a benzodiazepinas y relajantes musculares durante el embarazo en nuestra poblacion" / "Case-control study of the exposure to benzodiazepines and muscle relaxant drugs during pregnancy in our population"

YEARS: 2000-2001

FUNDING AGENCY: Fondo de Investigacion 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. Martinez-Frias)

NAME OF THE PROJECT: "Investigacion Epidemiologica de las Anomalias Congenitas" / "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 and 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. Martinez-Frias; Principal Investigator of the Project: M. Posada de la Paz).

NAME OF THE PROJECT: "Investigacion Epidemiologica de las Anomalias Congenitas (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 "Investigacion epidemiologica y causal de los defectos congenitos 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. Martinez-Frias)

NAME OF THE PROJECT: "Estudio epidemiologico descriptivo y de factores de riesgo para hernia diafragmatica congenita" / "Descriptive epidemiological study and analysis on the risk factors for congenital diaphragmatic hernia"

YEARS: 2005-2006

FUNDING AGENCY: Fondo de Investigacion 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 Investigacion de Enfermedades Raras de base Genetica): Desarrollo de una red nacional cooperativa de bancos de ADN y muestras biologicas" / "INERGEN (Institute of Research on genetically determined Rare Diseases"

YEAR: 2006

FUNDING AGENCY: Fondo de Investigacion 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. Rodriguez-Pinilla)

NAME OF THE PROJECT: "Centro de Investigacion Biomedica 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. Martinez-Frias).

NAME OF THE PROJECT: "Identificacion de Defectos Congenitos de Glicosilacion asociados a Malformaciones Congenitas (GLICOMAL)" / "Identification of Congenital Glycosilation Defects associated to Congenital Malformations (GLICOMAL)"

YEARS: 2007-2009

FUNDING AGENCY: CIBERER (Centro de Investigacion Biomedica 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 Perez)

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 responsible for overgrowth syndromes and cancer, and multiple cancers"
YEARS: 2007-2009

FUNDING AGENCY: CIBERER (Centro de Investigación Biomedica en Red de Enfermedades Raras) / (Centre for Biomedical Research on Rare Diseases as a Network)

MAIN OBJECTIVE OF THE PROJECT: To study infants with 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. Benitez Ortiz).

NAME OF THE PROJECT: "Aplicación de 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 Biomedica 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 Garcia).

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 "Investigacion epidemiologica y causal de los defectos congenitos 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. Martinez-Frias).

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 "Investigacion epidemiologica y causal de los defectos congenitos 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. Martinez-Frias).

NAME OF THE PROJECT: "Gestion y procesamiento de muestras del biobanco del IIER, y estudio genetico y molecular de pacientes con enfermedades raras en el marco del IIER, y con anomalias congenitas 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 Baladron Jimenez.

PARTICIPATION IN THE PROJECT: Researcher-Responsible (Principal Investigator: M. Posada de la Paz).

NAME OF THE PROJECT: "Investigacion sobre los aspectos clinicos y etiologicos de las fisuras craneo-faciales atipicas congenitas (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 "Investigacion epidemiologica y causal de los defectos congenitos 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. Martinez-Frias).

NAME OF THE PROJECT: "Centro de Investigacion Biomedica 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 "Investigacion epidemiologica y causal de los defectos congenitos 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. Martinez-Frias).

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 syndrome, 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 "Investigacion epidemiologica y causal de los defectos congenitos 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. Martinez-Frias).

NAME OF THE PROJECT: Convenio ISCIII-ASEREMAC 2017 para "Investigacion epidemiologica y causal de los defectos congenitos 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 "Investigacion epidemiologica y causal de los defectos congenitos 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 than 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 Sofia 1988 de Investigacion sobre Prevencion de las Deficiencias", awarded to Dr. M.L. Martinez-Frias and the Research Group of the Spanish Collaborative Study of Congenital Malformations (ECEMC).
 2. "Premio CERMI.ES de investigacion cientifica y social 2004", awarded to Maria Luisa Martinez-Frias 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-Sanchez.