

RESEARCH GROUP BACKGROUND AND ACCOMPLISHMENTS

2014-2019

RARE DISEASES

CURRENT MEMBERS OF THE RESEARCH AREA

Research coordinator

Dra. Clara Cavero Carbonell: cavero_cla@gva.es [ORCID](#) (2015-)

Dr. Óscar Zurriaga Llorens: zurriaga_osc@gva.es [ORCID](#) (2008-2011, 2014-2015)

Dra. Carmen Martos Jiménez: martos_car@gva.es (2011-2014)

Researchers

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Past members of the research group

Elena Gras Colomer: gras_ele@gva.es

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Iris Canet: canet_iri@gva.es

RECORD OF THE RESEARCH AREA (Period 2014-2019)

GROUP ACTIVITIES

The epidemiology programme for rare diseases (EREpi) aims to increase knowledge about the distribution and real situations of diseases and their consequences on the population and uses epidemiological methodology in its analytical design facet as well as the implementation of studies and techniques of spatial epidemiology and the study of quality of life. Compiling records and databases as key elements in the development of research in the field of RD, since they allow sufficient sample sizes to be achieved for clinical or epidemiological research.

LINES OF RESEARCH

- 1. Determination of the prevalence of RD**
- 2. Study of inequalities in RD**
- 3. Analysis of the determinants of RD**

CURRENT COLLABORATIONS WITH OTHER RESEARCH GROUPS

Valencian Community:

National:

CIBERESP

Participación en la acción estratégica proyecto MEDEA: Zurriaga O, Martos Jiménez MC

ORPHANET España:

Miembros del Comité Científico: Zurriaga O, Martos Jiménez MC.

HEMACARE

Editora del Manual for Coding and Reporting Haematological Malignancies” elaborado en el marco del proyecto financiado por la Unión Europea “Cancer Registry based Project on Haematologic Malignancies (HAEMACARE)”, 2010: Martos Jiménez MC.

International:

SCIENTIFIC/TECHNICAL RECORD

■ 1. Research projects funded

2012-2015. *Investigar para prevenir las enfermedades raras pediátricas: Aplicación a las anomalías congénitas cardíacas raras.* Entidad financiadora: Fundació Gent per Gent. Número de proyecto/contrato: 34 ER. Investigador principal: Martos C. Importe: 20.500,00 Euros.

2012-2014. *Cross-Border Patient Registries Initiative PARENT.* Entidad financiadora: Comisión Europea. Número de proyecto/contrato: PR11/096. Investigador principal: Zurriaga O. Importe: 112.945,00 Euros.

2012-2014. *Red Española de registros de Enfermedades Raras para la investigación.* Entidad financiadora: ISCIII. Internacional Rare Diseases Research Consortium (IRDIRC). Número de proyecto/contrato: PR11/122. Investigador principal: Zurriaga O. Importe: 186.600,00 Euros.

2011-2012. *Evaluación de conjunto mínimo básico de datos como fuente de información de las anomalías congénitas en la Comunitat Valenciana.* Entidad financiadora: Conselleria de Sanitat. Número de proyecto/contrato: AP-032/11. Investigador Principal: Martos C. Importe de la subvención: 7.500,00 Euros.

2. Participation in research contracts with companies or administrators.

3. Publications

YEAR 2018

1. Barisic I, Boban L, Akhmedzhanova D, Bergman JEH, Cavero-Carbonell C, Grinfelde I, Materna-Kirylyuk A, Latos-Bielenska A, Randrianaivo H, Zymak-Zakutnya N, Sansovic I, Lanzoni M, Morris JK. Beckwith Wiedemann syndrome:

2. A population-based study on prevalence, prenatal diagnosis, associated anomalies and survival in Europe. *Eur J Med Genet.* 2018;61(9):499-507.· DOI: [10.1016/j.ejmg.2018.05.014](<http://dx.doi.org/10.1016/j.ejmg.2018.05.014>)
3. Bergman JEH, Lutke LR, Gans ROB, Addor MC, Barisic I, Cavero-Carbonell C, Garne E, Gatt M, Klungsoyr K, Lelong N, Lynch C, Mokoroa O, Nelen V, Neville AJ, Pierini A, Randrianaivo H, Rissmann A, Tucker D, Wiesel A, Dolk H, Loane M, Bakker MK. Beta-Blocker Use in Pregnancy and Risk of Specific Congenital Anomalies: A European Case-Malformed Control Study. *Drug Saf.* 2018;41(4):415-27.· DOI: [10.1007/s40264-017-0627-x](<http://dx.doi.org/10.1007/s40264-017-0627-x>)
4. Garne E, Rissmann A, Addor MC, Barisic I, Bergman J, Braz P, Cavero-Carbonell C, Draper ES, Gatt M, Haeusler M, Klungsoyr K, Kurinczuk JJ, Lelong N, Luyt K, Lynch C, O'Mahony MT, Mokoroa O, Nelen V, Neville AJ, Pierini A, Randrianaivo H, Rankin J, Rouget F, Schaub B, Tucker D, Verellen-Dumoulin C, Wellesley D, Wiesel A, Zymak-Zakutnia N, Lanzoni M, Morris JK. Epidemiology of septo-optic dysplasia with focus on prevalence and maternal age - A EUROCAT study. *Eur J Med Genet.* 2018;61(9):483-8.· DOI: [10.1016/j.ejmg.2018.05.010](<http://dx.doi.org/10.1016/j.ejmg.2018.05.010>)
5. Marcos-Gragera R, Solans M, Galceran J, Fernandez-Delgado R, Fernandez-Teijeiro A, Mateos A, Quiros-Garcia JR, Fuster-Camarena N, De Castro V, Sanchez MJ, Franch P, Chirlaque MD, Ardanaz E, Martos C, Salmeron D, Peris-Bonet R, Spanish Childhood Cancer Epidemiology Working G. Childhood and adolescent lymphoma in Spain: incidence and survival trends over 20 years. *Clin Transl Oncol.* 2018;20(10):1289-301.· DOI: [10.1007/s12094-018-1860-1](<http://dx.doi.org/10.1007/s12094-018-1860-1>)

YEAR 2017

1. Boned-Ombuena A, Perez-Panades J, Lopez-Maside A, Miralles-Espi M, Guardiola Vilarroig S, Adam Ruiz D, Zurriaga O. [Prevalence of oral anticoagulation and quality of its management in primary healthcare: A study by the Health Sentinel Network of the Region of Valencia (Spain)]. *Aten Primaria.* 2017.· DOI: [10.1016/j.aprim.2016.11.015](<http://dx.doi.org/10.1016/j.aprim.2016.11.015>)
2. Boyle B, Garne E, Loane M, Addor MC, Arriola L, Cavero-Carbonell C, Gatt M, Lelong N, Lynch C, Nelen V, Neville AJ, O'Mahony M, Pierini A, Rissmann A, Tucker D, Zymak-Zakutnia N, Dolk H. The changing epidemiology of Ebstein's anomaly and its relationship with maternal mental health conditions: a European registry-based study. *Cardiol Young.* 2017;27(4):677-85.· DOI: [10.1017/S1047951116001025](<http://dx.doi.org/10.1017/S1047951116001025>)
3. Cavero-Carbonell C, Gimeno-Martos S, Paramo-Rodriguez L, Rabanaque-Hernandez MJ, Martos-Jimenez C, Zurriaga O. [Drugs use in pregnancy in the Valencia Region and the risk of congenital anomalies]. *An Pediatr (Barc).* 2017;87(3):135-42.· DOI: [10.1016/j.anpedi.2016.08.003](<http://dx.doi.org/10.1016/j.anpedi.2016.08.003>)

4. Cavero-Carbonell C, Gimeno-Martos S, Zurriaga O, Rabanaque-Hernandez MJ, Martos-Jimenez C. [The validity of the Spanish Minimum Basic Data Set to identify congenital anomalies in the Valencian Community (Spain)]. *Gac Sanit.* 2017;31(3):220-26. DOI: [10.1016/j.gaceta.2016.09.014](<http://dx.doi.org/10.1016/j.gaceta.2016.09.014>)
5. Marcos-Gragera R, Galceran J, Martos C, de Munain AL, Vicente-Raneda M, Navarro C, Quiros-Garcia JR, Sanchez MJ, Ardanaz E, Ramos M, Mateos A, Salmeron D, Felipe S, Peris-Bonet R. Incidence and survival time trends for Spanish children and adolescents with leukaemia from 1983 to 2007. *Clin Transl Oncol.* 2017;19(3):301-16. DOI: [10.1007/s12094-016-1531-z](<http://dx.doi.org/10.1007/s12094-016-1531-z>)

YEAR 2016

1. Ata-Ali J, Zurriaga O, Alberich C. Incidence and survival rates for malignant salivary gland tumors. *J Oral Sci.* 2016;58(1):67-73. DOI: [10.2334/josnusd.58.67](<http://dx.doi.org/10.2334/josnusd.58.67>)
2. Cavero-Carbonell C, Gras-Colomer E, Guaita-Calatrava R, Lopez-Briones C, Amoros R, Abaitua I, Posada M, Zurriaga O. Consensus on the criteria needed for creating a rare-disease patient registry. A Delphi study. *J Public Health (Oxf).* 2016;38(2):e178-86. DOI: [10.1093/pubmed/fdv099](<http://dx.doi.org/10.1093/pubmed/fdv099>)
3. Gimeno-Martos S, Cavero-Carbonell C, Lopez-Maside A, Bosch-Sanchez S, Martos-Jimenez C, Zurriaga O. [Chromosomal anomalies: The experience of the Congenital Anomalies Registry of the Valencia Region]. *An Pediatr (Barc).* 2016;84(4):203-10. DOI: [10.1016/j.anpedi.2015.09.010](<http://dx.doi.org/10.1016/j.anpedi.2015.09.010>)
4. Larrañaga N, Sanchez M, Ardanaz E, Felipe S, Marcos-Gragera R, Ramos M, Carulla M, Chirlaque M, Argüelles M, Martos C, Mateo A, Peris-Bonet R. Incidence Patterns and Trends of non-Central Nervous System Solid Tumours in Children and Adolescents. A Collaborative Study of the Spanish Population Based Cancer Registries. *J Cancer.* 2016;7(3):335-43. DOI: [10.7150/jca.12943](<http://dx.doi.org/10.7150/jca.12943>)
5. Mari-Dell'Olmo M, Gotsens M, Palencia L, Rodriguez-Sanz M, Martinez-Beneito MA, Ballesta M, Calvo M, Cirera L, Daponte A, Dominguez-Berjon F, Gandarillas A, Goni NI, Martos C, Moreno-Iribas C, Nolasco A, Salmeron D, Taracido M, Borrell C. Trends in socioeconomic inequalities in mortality in small areas of 33 Spanish cities. *BMC Public Health.* 2016;16(1):663. DOI: [10.1186/s12889-016-3190-y](<http://dx.doi.org/10.1186/s12889-016-3190-y>)
6. Martin Escobar E, Registro Espanol de Enfermos R. The Spanish Renal Registry: 2013 report and evolution from 2007-2013. *NEFROLOGIA.* 2016;36(2):97-120. DOI: [10.1016/j.nefro.2015.10.020](<http://dx.doi.org/10.1016/j.nefro.2015.10.020>)

7. Saez C, Zurriaga O, Perez-Panades J, Melchor I, Robles M, Garcia-Gomez JM. Applying probabilistic temporal and multisite data quality control methods to a public health mortality registry in Spain: a systematic approach to quality control of repositories. *J Am Med Inform Assoc.* 2016;23(6):1085-95. DOI: [10.1093/jamia/ocw010](<http://dx.doi.org/10.1093/jamia/ocw010>)

YEAR 2015

1. Bergman JE, Loane M, Vrijheid M, Pierini A, Nijman RJ, Addor MC, Barisic I, Beres J, Braz P, Budd J, Delaney V, Gatt M, Khoshnood B, Klungsoyr K, Martos C, Mullaney C, Nelen V, Neville AJ, O'Mahony M, Queisser-Luft A, Randrianaivo H, Rissmann A, Rounding C, Tucker D, Wellesley D, Zymak-Zakutnia N, Bakker MK, de Walle HE. Epidemiology of hypospadias in Europe: a registry-based study. *World J Urol.* 2015;33(12):2159-67. DOI: [10.1007/s00345-015-1507-6](<http://dx.doi.org/10.1007/s00345-015-1507-6>)
2. Hruskova Z, Stel VS, Jayne D, Aasarod K, De Meester J, Ekstrand A, Eller K, Heaf JG, Hoitsma A, Martos Jimenez C, Ravani P, Wanner C, Tesar V, Jager KJ. Characteristics and Outcomes of Granulomatosis With Polyangiitis (Wegener) and Microscopic Polyangiitis Requiring Renal Replacement Therapy: Results From the European Renal Association-European Dialysis and Transplant Association Registry. *Am J Kidney Dis.* 2015;66(4):613-20. DOI: [10.1053/j.ajkd.2015.03.025](<http://dx.doi.org/10.1053/j.ajkd.2015.03.025>)
3. McGivern MR, Best KE, Rankin J, Wellesley D, Greenlees R, Addor MC, Arriola L, de Walle H, Barisic I, Beres J, Bianchi F, Calzolari E, Doray B, Draper ES, Garne E, Gatt M, Haeusler M, Khoshnood B, Klungsoyr K, Latos-Bielenska A, O'Mahony M, Braz P, McDonnell B, Mullaney C, Nelen V, Queisser-Luft A, Randrianaivo H, Rissmann A, Rounding C, Sipek A, Thompson R, Tucker D, Wrotecki W, Martos C. Epidemiology of congenital diaphragmatic hernia in Europe: a register-based study. *Arch Dis Child Fetal Neonatal Ed.* 2015;100(2):F137-44. DOI: [10.1136/archdischild-2014-306174](<http://dx.doi.org/10.1136/archdischild-2014-306174>)
4. Nolasco A, Moncho J, Quesada JA, Melchor I, Pereyra-Zamora P, Tamayo-Fonseca N, Martínez-Beneito MA, Zurriaga O, Ballesta M, Daponte A, Gandarillas A, Domínguez-Berjón MF, Marí-Dell'Olmo M, Gotsens M, Izco N, Moreno C, Sáez M, Martos C, Sánchez-Villegas P, Borrell C. Trends in socioeconomic inequalities in preventable mortality in urban areas of 33 Spanish cities, 1996–2007 (MEDEA project). *Int J Equity Health.* 2015;14(1):33. DOI: [10.1186/s12939-015-0164-0](<http://dx.doi.org/10.1186/s12939-015-0164-0>)
5. Paramo-Rodriguez L, Mas Pons R, Cavero-Carbonell C, Martos-Jimenez C, Zurriaga O, Barona Vilar C. [An open heart: experiences of the parents of children with congenital heart disease]. *Gac Sanit.* 2015;29(6):445-50. DOI: [10.1016/j.gaceta.2015.07.009](<http://dx.doi.org/10.1016/j.gaceta.2015.07.009>)
6. Springett A, Wellesley D, Greenlees R, Loane M, Addor MC, Arriola L, Bergman J, Cavero-Carbonell C, Csaky-Szunyogh M, Draper ES, Garne E, Gatt M, Haeusler M, Khoshnood B, Klungsoyr K, Lynch C, Dias CM, McDonnell R,

- Nelen V, O'Mahony M, Pierini A, Queisser-Luft A, Rankin J, Rissmann A, Rounding C, Stoianova S, Tuckerz D, Zymak-Zakutnia N, Morris JK. Congenital anomalies associated with trisomy 18 or trisomy 13: A registry-based study in 16 European countries, 2000-2011. *Am J Med Genet A*. 2015;167A(12):3062-9. DOI: [10.1002/ajmg.a.37355](<http://dx.doi.org/10.1002/ajmg.a.37355>)
7. Zoni AC, Dominguez Berjon MF, Barcelo E, Esteban Vasallo MD, Abaitua I, Jimenez Villa J, Margolles Martins M, Navarro C, Posada M, Ramos Aceitero JM, Vazquez Santos C, Zurriaga Llorens O, Astray Mochales J, Spain RDRG. Identifying data sources for a national population-based registry: the experience of the Spanish Rare Diseases Registry. *Public Health*. 2015;129(3):271-5. DOI: [10.1016/j.puhe.2014.12.013](<http://dx.doi.org/10.1016/j.puhe.2014.12.013>)

YEAR 2014

1. Best KE, Addor M-C, Arriola L, Balku E, Barisic I, Bianchi F, Calzolari E, Curran R, Doray B, Draper E, Garne E, Gatt M, Haeusler M, van Kammen-Bergman J, Khoshnood B, Klungsoyr K, Martos C, Materna-Kiryluk A, Matias Dias C, McDonnell B, Mullaney C, Nelen V, O'Mahony M, Queisser-Luft A, Randrianaivo H, Rissmann A, Rounding C, Sipek A, Thompson R, Tucker D, Wellesley D, Zymak-Zakutnia N, Rankin J. Hirschsprung's disease prevalence in Europe: A register based study. *Birth Defects Research Part A: Clinical and Molecular Teratology*. 2014:n/a-n/a. DOI: [10.1002/bdra.23269](<http://dx.doi.org/10.1002/bdra.23269>)
2. Mari-Dell'olmo M, Gotsens M, Borrell C, Martinez-Beneito MA, Palencia L, Perez G, Cirera L, Daponte A, Dominguez-Berjon F, Esnaola S, Gandarillas A, Lorenzo P, Martos C, Nolasco A, Rodriguez-Sanz M. Trends in socioeconomic inequalities in ischemic heart disease mortality in small areas of nine Spanish cities from 1996 to 2007 using smoothed ANOVA. *J Urban Health*. 2014;91(1):46-61. DOI: [10.1007/s11524-013-9799-6](<http://dx.doi.org/10.1007/s11524-013-9799-6>)

4. Participation in education and training programs

Dr. Óscar Zurriaga, profesor Asociado en el Departamento de Medicina Preventiva y Salud Pública, Ciencias de la Alimentación, Toxicología y Medicina Legal. Universitat de València

Dra. Carmen Martos Jiménez, profesora Asociada en el Departamento de Enfermería de la Universitat de València.

Dra. Carmen Martos Jiménez y Dr. Óscar Zurriaga, profesores en el Máster en Salud Pública y Gestión Sanitaria (Universitat de València-ADEIT-Conselleria de Sanitat)

Dra. Carmen Martos Jiménez, profesora en el Master Oficial Inter-Universitario de Ciencias de la Enfermería –MICCE.

Dra. Carmen Martos Jiménez, coordinadora y profesora del curso “La encuesta como técnica de investigación en ciencias de la salud. Elaboración de cuestionarios y tratamiento estadístico de los datos”. Escuela Universitaria de Ciencias de la Salud de Zaragoza, 27/09/2010 – 05/10/2010.

5. Doctoral theses

- Fecha: 30/06/2017
- Título: Anomalías congénitas: distribución de la prevalencia y factores asociados
- Doctorando: Clara Cavero Carbonell
- Director/es: Dra. Carmen Martos Jiménez, Dra. María José Rabanaque Hernández
- Universidad: Universidad de Zaragoza
- Calificación: Sobresaliente Cum Laude