

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

Las publicaciones han sido ordenadas por año de publicación. La numeración de las publicaciones es correlativa a lo largo de los años, siguiendo el orden alfabético de primeros autores.

Año 1977

1. Castilla E, Martínez-Frías ML, Paz J. Patterns of combined limb anomalies. *Teratology* 1977; 16,2:203-210.
2. Martínez-Frías ML, Castilla E, Paz J. Descriptive systems for congenital limb anomalies. *Teratology* 1977; 15,2:163-169.

Año 1981

3. Lortie-Monette F, Kucera J, Weatherall JAC, Knudsen LN, Guibaud P, Czeizel A, Klingberg MA, Mastroiacovo P, Ashizawa M, Mutchinick O, Foster F, Nevin NC, Bjerkedal T, Castilla E, Martínez-Frías ML, Kallen B, Oakley GP, Stickle G, Hay S. A communication from the International Clearinghouse for Birth Defects Monitoring Systems. *Internat J Epidemiol* 1981; 10,3:245-246.

Año 1982

4. Martínez-Frías ML, Salvador J, Prieto L. Spanish toxic oil and congenital malformations. *The Lancet* 1982; 2:1349.

Año 1984

5. Martínez-Frías ML, Salvador J, Prieto L, Zaplana J. Epidemiological study of gastroschisis and omphalocele in Spain. *Teratology* 1984; 29:377-382.

Año 1985

6. Mori MA, Huertas H, Pinel I, Giralt P, Martínez-Frías ML. Trisomy 13 in the child of two carriers of a 13/15 translocation. *Am J Med Genet* 1985; 20:17-20.

Año 1986

7. Fullana A, García-Frías E, Martínez-Frías ML, Razquin S, Quero J. Caudal deficiency and asplenia anomalies in sibs. *Am J Med Genet Suppl* 1986; 2:23-29. Developmental field concept (233-239).
8. Källén B, Bertollini R, Castilla E, Czeizel A, Knudsen LB, Martínez-Frías ML, Mastroiacovo P, Mutchinick O. A joint international study on the epidemiology of hypospadias. *Acta Paediatr Scand* 1986; Suppl 324:1-52.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

9. Martínez-Frías ML, Parralo JA, Salvador J, Frías J. Sex ratios in neural tube defects. *The Lancet* 1986; 2:871-872.

Año 1987

10. Lancaster PAL, Knudsen LB, Goujard J, Robert E, Mastroiacovo P, Mutchinick O, Irgens L, Erickson D, Martínez-Frías ML. Epidemiology of Bladder Exstrophy and Epispadias: A Communication from the International Clearinghouse for Birth Defects Monitoring Systems. *Teratology* 1987; 36:221-227.
11. Mori MA, Gomar JL, Díaz de Bustamante A, Ananías A, Pinel I, Martínez-Frías ML. Partial duplication 16p resulting from a 3:1 segregation of a maternal reciprocal translocation. *Am J Med Genet* 1987; 26:203-206.
12. Mori MA, Rodríguez L, Pinel I, Casas JM, Díaz de Bustamante A, Martínez-Frías ML. Partial monosomy 15q due to de novo t(15;22)(q15;p11). *Ann Genet* 1987; 30,4:246-248.

Año 1988

13. Martínez-Frías ML, Herranz I, Salvador J, Prieto L, Ramos MA, Rodríguez-Pinilla E, Cordero JF. Prevalence of dominant mutations in Spain: Effect of changes in maternal age distribution. *Am J Med Genet* 1988; 31:845-852.
14. Martínez-Frías ML, Ramos MA, Salvador J. Thanatophoric dysplasia: An autosomal dominant condition? *Am J Med Genet* 1988; 31:815-820.
15. Martínez-Frías ML, Salvador J. Megadose vitamin A and teratogenicity. *The Lancet* 1988; 1:236.
16. Mori MA, Gómez-Sabrido F, Díaz de Bustamante A, Pinel I, Martínez-Frías ML. De novo 10q23 interstitial deletion. *J Med Genet* 1988; 25:209-210.
17. Pinel I, Díaz de Bustamante A, Urioste M, Félix V, Martínez-Frías ML. An unusual variant of chromosome 16. Two new cases. *Hum Genet* 1988; 80:194.
18. Urioste M, Valcárcel E, Gómez MA, Pinel I, García de León R, Díaz de Bustamante A, Tebar R, Martínez-Frías ML. Holoprosencephaly and Trisomy 21 in a child born to a nondiabetic mother. *Am J Med Genet* 1988; 30:925-928.

Año 1989

19. Källén B, Robert E, Mastroiacovo P, Martínez-Frías ML, Castilla EE, Cocchi G. Anticonvulsant drugs and malformations is there a drug specificity?. *Eur J Epidemiol* 1989; 5:31-36.
20. Martínez-Frías ML. Association of Holoprosencephaly and Down Syndrome (Letter to the Editor). *Am J Med Genet* 1989; 32:435.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

21. Martínez-Frías ML, Prieto L, Herranz I, Rodríguez-Pinilla E, Salvador J. Prenatal exposure to valproic acid and spina bifida. Some methodological aspects in case-control studies. *Eur J Epidemiol* 1989; 5:252.
22. Martínez-Frías ML, Rodríguez-Pinilla E, Salvador J. Valproate and spina bifida. *The Lancet* 1989; 1:611-612.
23. Martínez-Frías ML, Salvador J, Rodríguez-Pinilla E. Reply to Dr. Rybicki (Letter to the Editor). *Am J Med Genet* 1989; 34:299.

Año 1990

24. López Pajares I, Delicado A, Díaz de Bustamante A, Pellicer A, Pinel I, Pardo M, Martín M. Tetraploidy in a liveborn infant. *J Med Genet* 1990; 27:782-783.
25. Martínez-Frías ML. Clinical Manifestation of Prenatal Exposure to Valproic Acid Using Case Reports and Epidemiologic Information. *Am J Med Genet* 1990; 37:277-282.
26. Martínez-Frías ML, Frías JL, Salvador J. Clinical/Epidemiological Analysis of Malformations. *Am J Med Genet* 1990; 35:121-125.
27. Martínez-Frías ML, Salvador J. Epidemiological aspects of prenatal exposure to high doses of vitamin A in Spain. *Eur J Epidemiol* 1990; 6:118-123.
28. Pinel I, Urioste M, Martínez-Frías ML, Gomar JL. Endoreduplications in a family with a reciprocal translocation (9q;16p) (Letter to the Editor). *Clin Genet* 1990; 38:399-400.
29. Rodríguez JI, Palacios J, Urioste M. New acrofacial dysostosis syndrome in 3 sibs. *Am J Med Genet* 1990; 35:484-489.
30. Urioste M, Pinel I, Gomar JL, Skinner C, Martínez-Frías ML. Diploid/Tetraploid mosaicism in a stillborn infant with Prune Belly Anomaly. *Ann Génét* 1990; 33,1:49-51.

Año 1991

31. Källén B, Castilla E, Kringelbach M, Lancaster PAL, Martínez-Frías ML, Mastroiacovo P, Mutchinick O, Robert E. Parental Fertility and Infant Hypospadias: An International Case-Control Study. *Teratology* 1991; 44:629-634.
32. Källén B, Mastroiacovo P, Lancaster PAL, Mutchinick O, Kringelbach M, Martínez-Frías ML, Robert E, Castilla EE. Oral contraceptives in the etiology of isolated hypospadias. *Contraception* 1991; 44:173-182.
33. Lancaster PAL, Kucera J, Knudsen LB, Botting BJ, Robert E, Goujard J, Elek C, Mastroiacovo P, Cocchi G, Borman B, Irgens L, Castilla E, Martínez-Frías ML, Ericson A. Conjoined twins – An epidemiological study based on 312 cases. *Acta Genet Gemellol* 1991; 40:325-335.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

34. Martínez-Frías ML como coautora. Congenital malformations worldwide. A Report from the International Clearinghouse for Birth Defects Monitoring Systems. Ed. Elsevier Science Publishers BV. Amsterdam, 1991.
35. Martínez-Frías ML. Valproic acid and spina bifida (Letter to the Editor). *The Lancet* 1991; 338:196-197.
36. Martínez-Frías ML, Bermejo E, Cereijo A, Sánchez M, López M, Gonzalo C. Epidemiological aspects of mendelian syndromes in a Spanish population sample: II. Autosomal recessive malformation syndromes. *Am J Med Genet* 1991; 38:626-629.
37. Martínez-Frías ML, Cereijo A, Bermejo E, López M, Sánchez M, Gonzalo C. Epidemiological aspects of mendelian syndromes in a Spanish population sample: I. Autosomal dominant malformation syndromes. *Am J Med Genet* 1991; 38:622-625.
38. Martínez-Frías ML, Frías JL, Rodríguez-Pinilla E, Urioste M, Bermejo E, Cereijo A, Gayá F. Value of clinical analysis in epidemiological research: The Spanish Registry experience. *Am J Med Genet* 1991; 41:192-195.
39. Martínez-Frías ML, Frías JL, Vázquez I, Fernández J. Bartsocas-Papas syndrome: Three familial cases from Spain. *Am J Med Genet* 1991; 39:34-37.
40. Martínez-Frías ML, Rodríguez-Pinilla E. Tracheoesophageal and anal atresia in prenatal children exposed to a high dose of alcohol (Letter to the Editor). *Am J Med Genet* 1991; 40:128.
41. Robert E, Martínez-Frías ML. What does it mean to be a member of the Clearinghouse? *Int J Risk & Saf Med* 1991; 2:249-254.
42. Urioste M, Arroyo A, Martínez-Frías ML. Campomelia, polycystic dysplasia, and lymphocele in two sibs. *Am J Med Genet* 1991; 41:475-477.
43. Urioste M, Martínez-Frías ML. Anorectal anomalies and Down Syndrome (Letter to the Editor). *Am J Med Genet* 1991; 39:493.

Año 1992

44. Arroyo I, García MJ, Cimadevilla CE, Carretero V, Bermejo E, Martínez-Frías ML. Bilateral anophthalmia, esophageal atresia and right cryptorchidism: A new entity? *Am J Med Genet* 1992; 43:686-687.
45. Cereijo AI, Martínez-Frías ML. Prevalence of other birth defects among relatives of oral cleft probands (Letter to the Editor). *J Med Genet* 1992; 29:516.
46. Cohen Jr MM, Krieborg S, Lammer EJ, Cordero JF, Mastroiacovo P, Erickson JD, Roeper P, Martínez-Frías ML. Birth prevalence study of the Apert Syndrome. *Am J Med Genet* 1992; 42:655-659.

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47. Källén B, Castilla EE, Lancaster PAL, Mutchinick O, Knudsen LB, Martínez-Frías ML, Mastroiacovo P, Robert E. The cyclops and the mermaid: An epidemiological study of two types of rare malformations. *J Med Genet* 1992; 29:30-35.
48. Källén B, Castilla EE, Robert E, Lancaster PAL, Kringelbach M, Mutchinick O, Martínez-Frías ML, Mastroiacovo P. An international case-control study on hypospadias. The problem with variability and the beauty of diversity. *Eur J Epidemiol* 1992; 8:256-263.
49. Källén BAJ, Martínez-Frías ML, Castilla EE, Robert E, Lancaster PAL, Kringelbach M, Mutchinick OM, Mastroiacovo P. Hormone therapy during pregnancy and isolated hypospadias: An international case-control study. *Int J Risk & Saf Med* 1992; 3:183-198.
50. Martínez-Frías ML, Bermejo E. Prevalence of congenital anomaly syndromes in a Spanish gypsy population. *J Med Genet* 1992; 29:483-486.
51. Martínez-Frías ML, Bermejo E, Cereijo A. Preaxial polydactyly of feet in infants of diabetic mothers: Epidemiological test of a clinical hypothesis. *Am J Med Genet* 1992; 42:643-646.
52. Martínez-Frías ML, Cereijo A, Rodríguez-Pinilla E, Urioste M. Methimazole in animal feed and congenital aplasia cutis (Letter to the Editor). *The Lancet* 1992; 339:742-743.
53. Martínez-Frías ML, Cucalón F, Urioste M. New case of limb body-wall complex associated with sirenomelia sequence. *Am J Med Genet* 1992; 44:583-585.
54. Martínez-Frías ML, Frías JL, Galán E, Domingo R, Paisán L, Blanco M. New Syndrome?: Tracheoesophageal fistula, gastrointestinal abnormalities, hypospadias, and prenatal growth deficiency. *Am J Med Genet* 1992; 44:352-355.
55. Martínez-Frías ML, Rodríguez-Pinilla E. Reply to Drs. Karasik and Marion (Letter to the Editor). *Am J Med Genet* 1992; 42:854.
56. Martínez-Frías ML, Rodríguez-Pinilla E. Folic acid supplementation and neural tube defects (Letter to the Editor). *Lancet* 1992; 340:620.
57. Martínez-Frías ML, Urioste M, Cereijo A, Rodríguez Pinilla E. Anorectal and esophageal anomalies in Down Syndrome (Letter to the Editor). *Am J Med Genet* 1992; 44:848-849.
58. Martínez-Frías ML, Urioste M, Martín M, Frías JL. Pseudotrisonomy 13 Syndrome (Letter to the Editor). *Am J Med Genet* 1992; 43:633-635.
59. Ramos Arroyo MA, Rodríguez-Pinilla E, Cordero JF. Maternal diabetes: The risk for specific birth defects. *Eur J Epidemiol* 1992; 8:503-508.
60. Rodríguez JI, Palacios J, Urioste M. Response to Dr. Hecht (Letter to the Editor). *Am J Med Genet* 1992; 42:401.
61. Rodríguez JI, Palacios J, Urioste M. Acrofacial dysostosis syndromes (Letter to the Editor). *Am J Med Genet* 1992; 42:851.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 62.** Rodríguez JI, Palacios J, Urioste M, Rodríguez-Peralto JL. Tetra-phocomelia with multiple malformations: X-linked Amelia, or Roberts syndrome, or DK-phocomelia syndrome? (Letter to the Editor). *Am J Med Genet* 1992; 43:630-631.
- 63.** Rodríguez JI, Rodríguez-Peralto JL, Muro M, Urioste M, Palacios J. Anencephaly and limb deficiencies. *Am J Med Genet* 1992; 44:66-71.

Año 1993

- 64.** Cereijo AI, Martínez-Frías ML. Consanguineous marriages among parents of patients with Down syndrome (Letter to the Editor). *Clin Genet* 1993; 44:221-222.
- 65.** Cohen MM Jr, Kreiborg S, Lammer EJ, Cordero JF, Mastroiacovo P, Erickson JD, Roeper P, Martínez-Frías ML. Reply to Drs. Czeizel et al. (Letter to the Editor). *Am J Med Genet* 1993; 45:393.
- 66.** Martínez-Frías ML. Interviewer bias and maternal bias (Letter to the Editor). *Teratology* 1993; 47:531-532.
- 67.** Martínez-Frías ML (como coautora). Recommendations and protocols for prenatal diagnosis. European Study Group on Prenatal Diagnosis. JM Carrera y GC di Renzo eds. Barcelona. Publicado también en *Prog Diagn Pren* 1993; 5:3-66.
- 68.** Martínez-Frías ML, Bermejo E, Urioste M, Egüés J, López Soler JA. Short rib-polydactyly syndrome (SRPS) with anencephaly and other central nervous system anomalies: A new type of SRPS or a more severe expression of a known SRPS entity? *Am J Med Genet* 1993; 47:782-787.
- 69.** Martínez-Frías ML, Bermejo E, Urioste M, Huertas H, Arroyo I. Lethal short rib-polydactyly syndromes: Further evidence for their overlapping in a continuous spectrum. *J Med Genet* 1993; 30:937-941.
- 70.** Martínez-Frías ML, Cereijo A, Rodríguez-Pinilla E. Smoking in pregnancy (Letter to the Editor). *The Lancet* 1993; 341:1350-1351.
- 71.** Martínez Santana S, Pérez Alvarez F, Frías JL, Martínez-Frías ML. Hypertrichosis, atrophic skin, ectropion and macrostomia (Barber-Say Syndrome): Report of a new case. *Am J Med Genet* 1993; 47:20-23.
- 72.** Urioste M. Chromosome cultures from human cartilage. *Am J Med Genet* 1993; 46:123-125.
- 73.** Urioste M, Rodríguez JI, Barcia JM, Martín M, Escribá R, Pardo M, Camino J, Martínez-Frías ML. New syndrome: Persistence of Müllerian derivatives, lymphangiectasis, hepatic failure, postaxial polydactyly, renal and craniofacial anomalies. *Am J Med Genet* 1993; 47:494-503.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

Año 1994

- 74.** Martínez-Frías ML. Developmental field defects and associations: Epidemiological evidence of their relationship. *Am J Med Genet* 1994; 49:45-51.
- 75.** Martínez-Frías ML. Gastroschisis: Is the prevalence increasing? (Letter to the Editor). *Am J Med Genet* 1994; 49:128.
- 76.** Martínez-Frías ML. Another way to interpret the description of the Monster of Ravenna of the sixteenth century (Letter to the Editor). *Am J Med Genet* 1994; 49:362.
- 77.** Martínez-Frías ML. Epidemiological analysis of outcomes of pregnancy in diabetic mothers: Identification of the most characteristic and most frequent congenital anomalies. *Am J Med Genet* 1994; 51:108-113.
- 78.** Martínez-Frías ML. Spina bifida and hypospadias: A non random association or an X-linked recessive condition? *Am J Med Genet* 1994; 52:5-8.
- 79.** Martínez-Frías ML, Alcaraz M, Espejo P, Gómez MA, García de León R, González Moro L. Laurin-Sandrow syndrome (mirror hands and feet and nasal defects): Description of a new case. *J Med Genet* 1994; 31:410-412.
- 80.** Martínez-Frías ML, Bermejo E, García A, Galán E, Prieto L. Holoprosencephaly associated with caudal dysgenesis: A clinical-epidemiological analysis. *Am J Med Genet* 1994; 53:46-51.
- 81.** Martínez-Frías ML, Bermejo E, Paisán L, Martín M, Egüés J, López JA, Martínez S, Orbea C, Cucalón F, Gairi JM, Urioste M, De La Cruz MA. Severe spondylocostal dysostosis associated with other congenital anomalies: A clinical/epidemiologic analysis and description of ten cases from the Spanish Registry. *Am J Med Genet* 1994; 51:203-212.
- 82.** Martínez-Frías ML, Bermejo E, Sánchez Otero T, Urioste M, Morena V, Cruz E. New Syndrome: Sclerocornea, Hypertelorism, Syndactyly, and Ambiguous Genitalia. *Am J Med Genet* 1994; 49:195-197.
- 83.** Martínez-Frías ML, Gomar JL. New case of axial mesodermal dysplasia sequence: Epidemiologic evidence of a single entity. *Am J Med Genet* 1994; 49:74-76.
- 84.** Martínez-Frías ML, Martín M, Pardo M, Torres M, Cohen MM Jr. Holoprosencephaly and hypognathia with two proboscides: Report of a case and review of unusual proboscides. *J Craniofac Genet Dev Biol* 1994; 14:231-234.
- 85.** Martínez-Frías ML, Urioste M. Segmentation anomalies of the vertebrae and ribs: A developmental field defect: Epidemiologic evidence. *Am J Med Genet* 1994; 49:36-44.
- 86.** Urioste M, Martínez-Frías ML, Aparicio P. Ectrodactyly in Trisomy 13 syndrome (Letter to the Editor). *Am J Med Genet* 1994; 53:390-392.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 87.** Urioste M, Martínez-Frías ML, Bermejo E, Jiménez N, Romero D, Nieto C, Villa A. Short rib-polydactyly syndrome and pericentric inversion of chromosome 4. *Am J Med Genet* 1994; 49:94-97.
- 88.** Urioste M, Martínez-Frías ML, Bermejo E, Villa A, Jiménez N, Romero D, Nieto C. Chromosome 4p16 and osteochondrodysplasias. *Nature Genetics* 1994; 6:334.
- 89.** Urioste M, Paisán L, Martínez-Frías ML. DK-Phocomelia syndrome in a child with a long follow-up. *Am J Med Genet* 1994; 52:269-271.
- 90.** Urioste M, Visedo G, Sanchis A, Sentís C, Villa A, Ludeña P, Hortigüela JL, Martínez-Frías ML, Fernández-Piqueras J. Dynamic mosaicism involving an unstable supernumerary der(22) chromosome in Cat Eye Syndrome. *Am J Med Genet* 1994; 49:77-82.

Año 1995

- 91.** Castilla EE, Martínez-Frías ML. Congenital Healed Cleft Lip. *Am J Med Genet* 1995; 58:106-112.
- 92.** Galán-Gómez E, Cardesa-García JJ, Campo-Sampedro FM, Salamanca-Maesso C, Martínez-Frías ML, Frías JL. Kabuki Make-up (Niikawa-Kuroki) Syndrome in Five Spanish Children. *Am J Med Genet* 1995; 59:276-282.
- 93.** Martínez-Frías ML. Primary midline developmental field. I. Clinical and epidemiological characteristics. *Am J Med Genet* 1995; 56:374-381.
- 94.** Martínez-Frías ML, Martín M, Pardo M, Fernández de las Heras F, Frías JL. Distal aphyalangia, syndactyly, and extra metatarsal, associated with short stature, microcephaly and borderline intelligence: A New Autosomal Dominant Disorder. *Am J Med Genet* 1995; 55:213-216.
- 95.** Martínez-Frías ML, Urioste M, Bermejo E, Rodríguez-Pinilla E, Félix V, Paisán L, Martínez S, Egués J, Gómez F, Aparicio P, Cucalón F, Arroyo A, Meipp C, Vázquez S, Rodríguez JI, Rosa A, García J, Jiménez N, Moro C. Primary midline developmental field. II. Clinical/epidemiological analysis of alteration of laterality (normal body symmetry and asymmetry). *Am J Med Genet* 1995; 56:382-388.
- 96.** Rodríguez-Pinilla E, Martínez-Frías ML. Video display terminals: Risk of trisomy 18? (Letter to the Editor). *Clin Genet* 1995; 47:335-336.
- 97.** Urioste M, Arroyo I, Villa A, Lorda-Sánchez I, Barrio R, López-Cuesta MJ, Rueda J. Distal deletion of chromosome 13 in a child with the "Opitz" GBBB syndrome. *Am J Med Genet* 1995; 59:114-122.
- 98.** Villa A, Urioste M, Bofarull JM, Martínez-Frías ML. De novo interstitial deletion q16.2q21 on chromosome 6. *Am J Med Genet* 1995; 55:379-383.
- 99.** Villa A, Urioste M, Carrascosa MC, Vázquez S, Martínez A, Martínez-Frías ML. Pericentric inversions of chromosome 4: Report of a new family and review of the literature. *Clin Genet* 1995; 48:255-260.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

Año 1996

- 100.** Castilla EE, Lugarinho da Fonseca R, Da Graça Dutra M, Bermejo E, Cuevas L, Martínez-Frías ML. Epidemiological analysis of rare polydactylies. *Am J Med Genet* 1996; 65:295-303.
- 101.** Martínez-Frías ML. Epidemiological analysis of the association of congenital diaphragmatic hernia with upper-limb deficiencies: A primary polytopic developmental field defect. *Am J Med Genet* 1996; 62:68-70.
- 102.** Martínez-Frías ML. Multiple vertebral segmentation defects and rib anomalies (Letter to the Editor). *Am J Med Genet* 1996; 66:91.
- 103.** Martínez-Frías ML, García A, Cuevas J, Rodríguez JI, Urioste M. A new case of fibrochondrogenesis from Spain. *J Med Genet* 1996; 33:429-431.
- 104.** Martínez-Frías ML, Prieto L, Urioste M, Bermejo E. Clinical/Epidemiological analysis of congenital anomalies associated with diaphragmatic hernia. *Am J Med Genet* 1996; 62:71-76.
- 105.** Martínez-Frías ML, Urioste M, Bermejo E, Sanchis A, Rodríguez-Pinilla E. Epidemiological analysis of multi-site closure failure of neural tube in humans. *Am J Med Genet* 1996; 66:64-68.
- 106.** Prieto L, Martínez-Frías ML. Epidemiological analysis of the association between two congenital anomalies in the same child: A method for adjusting nonspecific clustering. *Am J Med Genet* 1996; 62:61-67.
- 107.** Schaefer C, Amoura-Elefant E, Vial T, Ornoy A, Garbis H, Robert E, Rodríguez-Pinilla E, Pexieder T, Prapas N, Merlob P. Pregnancy outcome after prenatal quinolone exposure. Evaluation of a case registry of the European Network of Teratology Information Services (ENTIS). *Europ J Obstet Gynecol and Reprod Biol* 1996; 69:83-89.
- 108.** Urioste M, Lorda-Sánchez I, Blanco M, Burón E, Aparicio P, Martínez-Frías ML. Severe congenital limb deficiencies, vertebral hypersegmentation, absent thymus and mirror polydactyly: A defect expression of a developmental control gene? *Hum Genet* 1996; 97:214-217.

Año 1997

- 109.** Botto LD, Khoury MJ, Mastroiacovo P, Castilla EE, Moore CA, Skjaerven R, Mutchinick OM, Borman B, Cocchi G, Czeizel AE, Goujard J, Irgens LM, Lancaster PAL, Martínez-Frías ML, Merlob P, Ruusinen A, Stoll C, Sumiyoshi Y. The spectrum of congenital anomalies of the VATER Association: An international study. *Am J Med Genet* 1997; 71:8-15.
- 110.** Caro-Patón T, Carvajal A, Martín de Diego I, Martín-Arias LH, Alvarez Requejo A, Rodríguez-Pinilla E. Is metronidazole teratogenic? A meta-analysis. *Br J Clin Pharmacol* 1997; 44:179-182.
- 111.** Feingold M, Hall BD, Lacassie Y, Martínez-Frías ML. Syndrome of microcephaly, facial and hand abnormalities, tracheoesophageal fistula, duodenal atresia, and developmental delay. *Am J Med Genet* 1997; 69:245-249.

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- 112.** Lorda-Sánchez I, Urioste M, Villa A, Carrascosa MC, Vázquez MS, Martínez A, Martínez-Frías ML. Proximal partial 5p trisomy resulting from a maternal (19;5) insertion. *Am J Med Genet* 1997; 68:476-480.
- 113.** Lorda-Sánchez I, Villa A, Urioste M, Bernal E, Jaso E, García A, Martínez-Frías ML. Tetrasomy 5p mosaicism due to an extra i(5p) in a severely affected girl. *Am J Med Genet* 1997; 68:481-484.
- 114.** Martínez-Frías ML. Clinical and epidemiological characteristics of infants with body wall complex with and without limb deficiency. *Am J Med Genet* 1997; 73:170-175.
- 115.** Martínez-Frías ML. Epidemiological characteristics of amniotic band sequence (ABS) and body wall complex (BWC): Are they two different entities? *Am J Med Genet* 1997; 73:176-179.
- 116.** Martínez-Frías ML, Arroyo I, Bermejo E, Espinosa J, García MJ. Severe limb deficiencies, vertebral hypersegmentation, and mirror polydactyly: Two additional cases that expand the phenotype to a more generalized effect on blastogenesis. *Am J Med Genet* 1997; 73:205-209.
- 117.** Martínez-Frías ML, Bermejo E. Major congenital malformations in Down Syndrome (Letter to the Editor). *Am J Med Genet* 1997; 73:91.
- 118.** Martínez-Frías ML, Bermejo E, Aparicio P, Blanco M, Burón E, Cuevas L, Espinosa MJ, Fondevilla J, Gallo M, Hernández F, Marco JJ, Martínez S, Morales MC, Mújica I, Paisán L, Valdivia L. Amelia: Analysis of its epidemiological and clinical characteristics. *Am J Med Genet* 1997; 73:189-193.
- 119.** Martínez-Frías ML, Frías JL. Are blastogenetic anomalies sporadic? *Am J Med Genet* 1997; 68:381-385.
- 120.** Martínez-Frías ML, Frías JL. Primary developmental field III: Clinical and epidemiological study of blastogenetic anomalies and their relationship to different MCA patterns. *Am J Med Genet* 1997; 70:11-15.
- 121.** Martínez-Frías ML, Frías JL, Bermejo E, Rodríguez-Pinilla E, Urioste M. Epidemiological analysis of the Schisis association in the Spanish Registry of Congenital Malformations. *Am J Med Genet* 1997; 70:16-23.
- 122.** Martínez-Frías ML, Rodríguez-Pinilla E, Bermejo E. Correlation between drug exposure and major malformations (Letter to the Editor). *Am J Med Genet* 1997; 70:99.
- 123.** Martínez-Frías ML, Rodríguez-Pinilla E, Prieto L. Prenatal exposure to salicylates and gastroschisis: A case-control study. *Teratology* 1997; 56:241-243.
- 124.** Urioste M, Rodríguez JI, Bofarull JM, Torán N, Ferrer C, Villa A. Giant-cell chondrodysplasia in a male infant with clinical and radiological findings resembling the Piepkorn type of lethal osteochondrodysplasia. *Am J Med Genet* 1997; 68:342-346.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

Año 1998

- 125.** Bermejo E, Martínez-Frías ML. Congenital eye malformations: Clinical-epidemiological analysis of 1,124,654 consecutive births in Spain. *Am J Med Genet* 1998; 75:497-504.
- 126.** Bermejo E, Martínez-Frías ML. Clinical-epidemiological aspects of Down syndrome in Spain. *Ital J Intellect Impair / Riv Ital Disturbo Intellet* 1998;11:23-30.
- 127.** Lorda-Sánchez I, Prieto L, Rodríguez-Pinilla E, Martínez-Frías ML. Increased parental age and number of pregnancies in Klippel-Trenaunay-Weber syndrome. *Ann Hum Genet* 1998; 62:235-239.
- 128.** Martínez-Frías ML, Bermejo E, Prieto L. Maternal occupation in agriculture during pregnancy and congenital anomalies: A case-control study. *Int J Risk & Saf Med* 1998; 11:217-224.
- 129.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Frías JL. Congenital anomalies in the offspring of mothers with a bicornuate uterus. *Pediatrics* 1998; 101,4,e10.
URL:<http://www.pediatrics.org/cgi/content/full/101/4/e10>.
- 130.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto L. Case-control study on occupational exposure to anesthetic gases during pregnancy. *Int J Risk & Saf Med* 1998; 11:225-231.
- 131.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto L, Frías JL. Epidemiological analysis of outcomes of pregnancy in gestational diabetic mothers. *Am J Med Genet* 1998; 78:140-145.
- 132.** Martínez-Frías ML, Frías JL, Opitz JM. Errors of morphogenesis and developmental field theory. *Am J Med Genet* 1998; 76:291-296.
- 133.** Martínez-Frías ML, García A, Bermejo E. Cyclopia and sirenomelia in a liveborn infant. *J of Med Genet* 1998; 35:263-264.
- 134.** Martínez-Frías ML, Rodríguez-Pinilla E, Bermejo E, Blanco M. Prenatal exposure to Penicillamine and oral clefts: Case Report (Letter to the Editor). *Am J Med Genet* 1998; 76:274-275.
- 135.** Martínez-Frías ML, Rodríguez-Pinilla E, Bermejo E, Prieto L. Prenatal exposure to sex hormones: A case-control study. *Teratology* 1998; 57:8-12.
- 136.** Martínez-Frías ML, Rodríguez-Pinilla E, Bermejo E, Prieto L. Reply to Dr. Li: "Prenatal Exposure to Sex Hormones: A Case-Control Study" (Letter to the Editor). *Teratology* 1998; 58:29.
- 137.** Martínez-Frías ML, Sanchis A, Aparicio P, Blanco M, García MJ, Gómez-Ullate J, Félix V, Huertas H, Jiménez N, López JA, Marco JJ, Martín M, Palacios G, Romero D, Vázquez MS. Description of the characteristics of cases with noncontiguous neural tube defects identified in a series of consecutive births. *Teratology* 1998; 57:13-16.
- 138.** Rodríguez-Pinilla E, Martínez-Frías ML. Corticosteroids during pregnancy and oral clefts: A case-control study. *Teratology* 1998; 58:2-5.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 139.** Urioste M, Rosa A. Anencephaly and faciocranioschisis: Evidence of complete failure of closure 3 of the neural tube in humans. *Am J Med Genet* 1998; 75:4-6.

Año 1999

- 140.** Martínez-Frías ML, Bermejo E, Frías JL. Analysis of deformations in 26,810 consecutive infants with congenital defect. *Am J Med Genet* 1999; 84:365-368.
- 141.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Frías JL. Maternal and Fetal Factors related to abnormal amniotic fluid. *Journal Perinatology* 1999; 19(7):514-520.
- 142.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto L. Exploratory case-control study on maternal occupation as cook during pregnancy and congenital defects. *Environ Epidemiol Toxicol* 1999; 1:148-152.
- 143.** Martínez-Frías ML, Czeizel AE, Rodríguez-Pinilla E, Bermejo E. Smoking during pregnancy and Poland Sequence: Results of a population-based registry and a case-control registry. *Teratology* 1999; 59:35-38.
- 144.** Martínez-Frías ML, Frías JL. VACTERL as primary, polytopic developmental field defects. *Am J Med Genet* 1999; 83:13-16.
- 145.** Martínez-Frías ML, Rodríguez-Pinilla E. First-trimester exposure to topical Tretinoin: Its safety is not warranted (Letter to the Editor). *Teratology* 1999; 60:5.
- 146.** Martínez-Frías ML, Rodríguez-Pinilla E. Problems of using data from Teratology Information Services (TIS) to identify putative teratogens (Letter to the Editor). *Teratology* 1999; 60:54-55.
- 147.** Prieto L, Martínez-Frías ML. Case-control studies using only malformed infants: Are we interpreting the results correctly? (Letter to the Editor). *Teratology* 1999; 60:1-2.

Año 2000

- 148.** Blatter BM, Roeleveld N, Bermejo E, Martínez-Frías ML, Siffel C, Czeizel AE. Spina bifida and parental occupation: Results from three malformation monitoring programs in Europe. *Eur J Epidemiol* 2000; 16:343-351.
- 149.** Martínez-Frías ML. Response to 'what kind of controls to use in case control studies of malformed infants: Recall bias versus "teratogen nonspecificity" bias'. (Letter to the Editor). *Teratology* 2000; 62:372.
- 150.** Martínez-Frías ML, Bermejo E, Frías JL. Pathogenetic classification of a series of 27,145 consecutive infants with congenital defects. *Am J Med Genet* 2000; 90:246-249.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 151.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto L. Case-control study of maternal occupation as hairdresser during pregnancy and congenital defects. *Environ Epidemiol Toxicol* 2000; 2:20-23.
- 152.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E. Body stalk defects, body wall defects, amniotic bands with and without body wall defects, and gastroschisis: Comparative epidemiology. *Am J Med Genet* 2000; 92:13-18.
- 153.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E. Anal atresia, vertebral, genital, and urinary tract anomalies: A primary polytopic developmental field defect identified through an epidemiological analysis of associations. *Am J Med Genet* 2000; 95:169-173.
- 154.** Martínez-Frías ML, Castilla EE, Bermejo E, Prieto L, Orioli IM. Isolated small intestinal atresias in Latin America and Spain: Epidemiological analysis. *Am J Med Genet* 2000; 93:355-359.
- 155.** Martínez-Frías ML, Villa A, Acero de Pablo R, Ayala A, Calvo MJ, Bermejo E, Rodríguez L. Limb deficiencies in infants with trisomy 13. *Am J Med Genet* 2000; 93:339-341.
- 156.** Prieto L, Martínez-Frías ML. Response to “Case-control studies using only malformed infants who were prenatally exposed to drugs. What do the results mean?” (Letter to the Editor). *Teratology* 2000; 62:8-9.
- 157.** Prieto L, Martínez-Frías ML. Response to “What kind of controls to use in case control studies of malformed infants: recall bias versus ‘teratogen nonspecificity’ bias”. (Letter to the Editor). *Teratology* 2000; 62:372.
- 158.** Rodríguez-Pinilla E, Arroyo I, Fondevilla J, García MJ, Martínez-Frías ML. Prenatal exposure to Valproic Acid during pregnancy and limb deficiencies: A case-control study. *Am J Med Genet* 2000; 90:376-381.
- 159.** Rodríguez L, Sanchis A, Villa A, Cánovas A, Peris S, Estivalis M, Pons S, Martínez-Frías ML. Ring chromosome 7 and sacral agenesis. *Am J Med Genet* 2000; 94:52-58.
- 160.** Rosano A, Botto LD, Olney RS, Khoury MJ, Ritvanen A, Goujard J, Stoll C, Cocchi G, Merlob P, Mutchinick O, Cornel MC, Castilla EE, Martínez-Frías ML, Zampino G, Erickson JD, Mastroiacovo P. Limb defects associated with major congenital anomalies: Clinical and epidemiological study from the International Clearinghouse for Birth Defects Monitoring Systems. *Am J Med Genet* 2000; 93:110-116.
- 161.** Villa A, Galán Gómez E, Rodríguez L, Hernández Rastrollo R, Martínez Tallo ME, Martínez-Frías ML. Interstitial tandem duplication of 6p: A case with partial trisomy (6)(p12p21.3). *Am J Med Genet* 2000; 90:369-375.

Año 2001

- 162.** Martínez-Frías ML. Heterotaxia as an outcome of maternal diabetes: An epidemiological study. *Am J Med Genet* 2001; 99:142-146.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 163.** Martínez-Frías ML. Editorial Comment: Approaches to the analysis of infants with multiple congenital anomalies. *Am J Med Genet* 2001; 101:33-35.
- 164.** Martínez-Frías ML, Bermejo E, Frías JL. The VACTERL association: Lessons from the Sonic hedgehog pathway. (Letter to the Editor). *Clin Genet* 2001; 60:397-398.
- 165.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Frías JL. Exstrophy of the cloaca and exstrophy of the bladder: Two different expressions of a primary developmental field defect. *Am J Med Genet* 2001; 99:261-269.
- 166.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto L. Periconcepcional exposure to contraceptive pills and risk for Down syndrome. *Journal of Perinatology* 2001; 21:288-292.
- 167.** Martínez-Frías ML, García Mazario MJ, Feito Caldas C, Conejero Gallego MP, Bermejo E, Rodríguez-Pinilla E. High maternal fever during gestation and severe congenital limb disruptions. *Am J Med Genet* 2001; 98:201-203.
- 168.** Martínez-Frías ML, Rodríguez-Pinilla E. Epidemiologic analysis of prenatal exposure to cough medicines containing dextromethorphan: No evidence of human teratogenicity. *Teratology* 2001; 63:38-41.
- 169.** Martínez-Frías ML, Rodríguez L, Bermejo E, López F, Rodríguez-Pinilla E. It is necessary to perform high-resolution band chromosomes in any child with malformations, before making a diagnosis or establishing a possible relationship with any risk factor. (Letter to the Editor). *Am J Med Genet* 2001; 101:80.
- 170.** Stoll C, Rosano A, Botto LD, Erickson D, Khoury MJ, Olney RS, Castilla EE, Cocchi G, Cornel MC, Goujard J, Bermejo E, Merlob P, Mutchinick O, Ritvanen A, Zampino G, Mastroiacovo P. On the symmetry of limb deficiencies among children with multiple congenital anomalies. *Ann Génét* 2001; 44:19-24.

Año 2002

- 171.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Frías JL. Reply to the research letter by Bohring-"OEIS Complex, VATER, and the ongoing difficulties in terminology and delineation". (Letter to the Editor). *Am J Med Genet* 2002; 107:77.
- 172.** Martínez-Frías ML, Rodríguez-Pinilla E, Bermejo E, Prieto L. Epidemiological evidence that Maternal Diabetes does not appear to increase the risk for Down Syndrome. *Am J Med Genet* 2002; 112:335-337.
- 173.** Martínez-Frías ML, Rodríguez L, López F, Bermejo E, Rodríguez-Pinilla E. Nowadays it is preceptive to perform chromosomal studies with high resolution G-bands and FISH techniques when necessary. (Letter to the Editor). *Am J Med Genet* 2002; 108:254.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 174.** Rodríguez L, Cuadrado Pérez I, Herrera Montes J, Lorente Jarreño ML, López Grondona F, Martínez-Frías ML. Terminal deletion of the chromosome 7(q36-qter) in an infant with sacral agenesis and anterior myelomeningocele. *Am J Med Genet* 2002; 110:73-77.
- 175.** Rodríguez L, López F, Paisán L, Portugués De La Red M del M, Ruiz AM, Blanco M, Antelo Cortizas J, Martínez-Frías ML. Pure partial trisomy 7q: Two new patients and review. *Am J Med Genet* 2002; 113:218-224.
- 176.** Wang R, Martínez-Frías ML, Graham JM. Infants of diabetic mothers are at increased risk for the oculo-auriculo-vertebral sequence: A case-based and case-control approach. *J Pediatrics* 2002; 141:611-617.

Año 2003

- 177.** Arroyo Carrera I, Pitarch V, García MJ, Barrio AR, Martínez-Frías ML. Unusual congenital abdominal wall defect and review. *Am J Med Genet* 2003; 119A:211-213.
- 178.** Frías JP, Martínez-Frías ML, Frías PA, Frías JL. Obesity increases the risk of congenital heart defects in women with gestational diabetes mellitus. *Diabetologia* 2003; 46: A66-184.
- 179.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E. Incidence of Smith-Lemli-Opitz syndrome in Ontario, Canada. (Letter to the Editor). *Am J Med Genet* 2003; 116A:101.
- 180.** Martínez-Frías ML, Rodríguez-Pinilla E, Bermejo E, Prieto L. Response to Dr. Narchi's comments on "Epidemiological evidence that maternal diabetes does not appear to increase the risk for Down syndrome". (Letter to the Editor). *Am J Med Genet* 2003; 121A:191-192.
- 181.** Rodríguez L, Martínez Guardia N, Herens C, Jamar M, Verloes A, López F, Santos Muñoz J, Martínez-Frías ML. Subtle trisomy 12q24.3 and subtle monosomy 22q13.3: Three new cases and review. *Am J Med Genet* 2003; 122A:119-124.
- 182.** Wilcken B, Bamforth F, Li Z, Zhu H, Ritvanen S, Redlund M, Stoll C, Alembik Y, Dott B, Czeizel AE, Gelman-Kohan Z, Scarano G, Bianca S, Ettore G, Tenconi R, Bellato S, Scala I, Mutchinick OM, López MA, De Walle H, Hofstra R, Joutchenko L, Kavteladze L, Bermejo E, Martínez-Frías ML, Gallagher M, Erickson JD, Vollset SE, Mastroiacovo P, Andria G, Botto LD. Geographical and ethnic variation of the 677C>T allele of 5,10 methylenetetrahydrofolate reductase (MTHFR): Findings from over 7000 newborns from 16 areas world wide. *J Med Genet* 2003; 40:619-625.

Año 2004

- 183.** Galán-Gómez E, Carbonell-Pérez JM, Cardesa-García JJ, Val-Sánchez de León JM, Campo-Sampedro FM, Martínez-Frías ML, Frías JL. A diagnostic conundrum: Two siblings with features overlapping the Kabuki and Malpuech Syndromes. A new MCA Syndrome? *Am J Med Genet* 2004; 125A:306-309.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 184.** Kroes HY, Olney RS, Rosano A, Liu Y, Castilla EE, Cocchi G, De Vigan C, Martínez-Frías ML, Mastroiacovo P, Merlob P, Mutchinick O, Ritvanen A, Stoll C, van Essen AJ, Cobben JM, Cornel MC. Renal defects and limb deficiencies in 197 infants: It is possible to define the “Acrorenal syndrome”? *Am J Med Genet* 2004; 129A:149-155.
- 185.** Martínez-Frías ML. Editorial Comment: Segmentation anomalies of the vertebrae and ribs: One expression of the primary developmental field. *Am J Med Genet* 2004; 128A:127-131.
- 186.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Frías JL. Risk for congenital anomalies associated with different sporadic and daily doses of alcohol consumption during pregnancy: A case-control study. *Birth Defects Res (Part A): Clin Mol Teratol* 2004; 70:194-200.
- 187.** Martínez-Frías ML, Prieto D, Prieto L, Bermejo E, Rodríguez-Pinilla E, Cuevas L. Secular decreasing trend of the frequency of hypospadias among newborn male infants in Spain. *Birth Defects Res (Part A): Clin Mol Teratol* 2004; 70:75-81.
- 188.** Martínez-Frías ML, Rodríguez L, López-Grondona F, Bermejo E, Rodríguez-Pinilla E, Frías JL. Frequency of neural tube defects and Down syndrome in the same sibship: Analysis of the Spanish Ongoing case-control study. (Letter to the Editor). *Am J Med Genet* 2004; 126A:430-431.

Año 2005

- 189.** Bermejo E, Félix V, Lapunzina P, Galán E, Soler V, Delicado A, Pantoja A, Márquez MD, García M, Mora E, Cuevas L, Ureta A, López-Pajares I, Martínez-Frías ML. Craniofacial dyssynostosis: Description of the first four Spanish cases and review. *Am J Med Genet* 2005; 132A:41-48.
- 190.** Bermejo E, Lapunzina P, Galán E, Félix V, Soler V, Martínez-Frías ML. Correspondence: New findings in craniofacial dyssynostosis. *Am J Med Genet* 2005; 134A:344-345.
- 191.** Martínez-Frías ML. Correspondence: The real earliest historical evidence of Down syndrome. *Am J Med Genet* 2005; 132A:231.
- 192.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto D, Prieto L. Correspondence: MTHFR 677C-T Polymorphism is not excluded as maternal risk for Down syndrome among Turkish women. *Am J Med Genet* 2005; 134A:461.
- 193.** Martínez-Frías ML, Frías JP, Bermejo E, Rodríguez-Pinilla E, Prieto L, Frías JL. Pre-gestational maternal body mass index predicts an increased risk of congenital malformations in infants of mothers with gestational diabetes. *Diabetic Medicine* 2005; 22:775-781.
- 194.** Martínez-Frías ML, Fernández Toral J, López-Grondona F, Mendioroz J, Bermejo E. Clinical Report: Growth deficiency, facial anomalies, and brachydactyly (Frías syndrome): A second family. *Am J Med Genet* 2005; 137A:288-291.
- 195.** Mendioroz J, Fernández-Toral J, Suárez E, López-Grondona F, Kjaer KW, Bermejo E, Martínez-Frías ML. Clinical Report: Sensorineural deafness, abnormal genitalia, synostosis of metacarpals and metatarsals 4 and 5, and mental retardation: Description of a second patient and exclusion of HOXD13. *Am J Med Genet* 2005; 135A:211-213.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 196.** Pogue R, Ehtesham N, Repetto GM, Carrero-Valenzuela R, Bazán de Casella C, Pintos de Pons S, Martínez-Frías ML, Heuertz S, Cormier-Daire V, Cohn DH. Research letter: Probable identity-by-descent for a mutation in the Dyggve-Melchior-Clausen/Smith-McCort dysplasia (Dymeclin) gene among patients from Guam, Chile, Argentina and Spain. *Am J Med Genet* 2005; 138A:75-78.
- 197.** Rodríguez L, Starke H, Martínez Guardia N, Tönnies H, Neitzel H, Kozłowski P, Mazauric ML, Heller A, López Grondona F, Mansilla E, Santos Muñoz MJ, Liehr T, Martínez-Frías ML. Three new cases with a supernumerary ring chromosome 1. *Clin Dysmorphol* 2005; 14:169-175.
- 198.** Rodríguez L, Zollino M, Climent S, Mansilla E, López-Grondona F, Martínez-Fernández ML, Murdolo M, Martínez-Frías ML. Clinical Report: The new Wolf-Hirschhorn syndrome critical region (WHSCR-2): A description of a second case. *Am J Med Genet* 2005; 136A:175-178.
- 199.** Sanchis A, Cerveró L, Bataller A, Tortajada JL, Huguet J, Crow YJ, Au M, Higuete LJ, Martínez-Frías ML. Genetics syndromes mimic congenital infections. *J Pediatr* 2005; 146:701-705.

Año 2006

- 200.** Ali M, Highet LJ, Lacombe D, Goizet C, King MD, Tacke U, van der Knaap MS, Lagae L, Rittey C, Brunner HG, von Bokhoven H, Hamel B, Oade YA, Sanchis A, Desguerre I, Cau D, Mathieu N, Moutard ML, Lebon P, Kumar D, Jackson AP, Crow YJ. A second locus for Aicardi-Goutières syndrome at chromosome 13q14-21. *J Med Genet* 2006; 43:444-450.
- 201.** Bermejo E, Mendioroz J, Cuevas L, Martínez-Frías ML. The incidence of gastroschisis: Is also increasing in Spain particularly among babies of young mothers. (Letter to the Editor). *BMJ* 2006; 332,7538:424.
- 202.** Crow YJ, Leitch A, Hayward BE, Garner A, Parmar R, Griffith E, Ali M, Semple C, Aicardi J, Babul-Hirji R, Baumann C, Baxter P, Bertini E, Chandler KE, Chitayat D, Cau D, Déry C, Fazzi E, Goizet C, King MD, Klepper J, Lacombe D, Lanzi G, Lyall H, Martínez-Frías ML, Mathieu M, McKeown C, Monier A, Oade Y, Quarrell OW, Rittey CD, Curtis Rogers R, Sanchis A, Stephenson JBP, Tacke U, Till M, Tolmie JL, Tomlin P, Voit T, Weschke B, Geoffrey Woods C, Lebon P, Bonthron DT, Ponting CP, Jackson AP. Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-Goutières syndrome and mimic congenital viral brain infection. *Nature Genetics* 2006; 38:910-916.
- 203.** Liehr T, Mrasek K, Weise A, Dufke A, Rodríguez L, Martínez Guardia N, Sanchis A, Vermeesch JR, Ramel C, Polityko A, Haas OA, Anderson J, Claussen U, Von Eggeling F, Starke H. Small supernumerary marker chromosomes-progress towards a genotype-phenotype correlation. *Cytogenet Genome Res* 2006; 112:23-34.
- 204.** Martínez-Frías ML. Folic acid: a public-health challenge. (Letter to the Editor). *Lancet* 2006 Jun 24; 367(9528):2057.
- 205.** Martínez-Frías ML, Bermejo E. Do we have enough evidences to consider that infertility treatments may not be causally related with congenital anomalies in newborn infants? (Letter to the Editor). *BMJ* (17 Noviembre 2006). <http://www.bmj.com/cgi/eletters/333/7570/665>.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 206.** Martínez-Frías ML, Pérez B, Desviat LR, Castro M, Leal F, Rodríguez L, Mansilla E, Martínez-Fernández ML, Bermejo E, Rodríguez-Pinilla E, Prieto D, Ugarte M and ECEMC Working Group. Maternal polymorphisms 677C-T and 1298A-C of MTHFR, and 66A-G MTRR genes: Is there any relationship between polymorphisms of the folate pathway, maternal homocysteine levels, and the risk for having a child with Down syndrome? *Am J Med Genet Part A* 2006; 140A:987-997.
- 207.** Meijer WM, Cornel MC, Dolk H, de Walle HEK, Armstrong NC, de Jong-van den Berg LTW, EUROCAT Working Group (Martínez-Frías ML y Bermejo E como miembros del Eurocat Working Group). The potential of the European network of congenital anomaly registers (EUROCAT) for drug safety surveillance: a descriptive study. *Pharmacoepidemiol Drug Saf* 2006; 15:675-682.

Año 2007

- 208.** Chabchoub E, Rodríguez L, Galán E, Mansilla E, Martínez-Fernández ML, Martínez-Frías ML, Fryns JP, Vermeesch JR. Molecular characterisation of a mosaicism with a complex chromosome rearrangement: evidence for coincident chromosome healing by telomere capture and neo-telomere formation. *J Med Genet* 2007; 44:250-256.
- 209.** Frías JL, Frías JP, Frías PA, Martínez-Frías ML. Infrequently studied congenital anomalies as clues to the diagnosis of maternal diabetes mellitus. *Am J Med Genet Part A* 2007; 143A:2904-2909.
- 210.** Frutos de CA, Vega S, Manzanares M, Flores JM, Huertas H, Martínez-Frías ML, Nieto MA. Snail1 Is a Transcriptional Effector of FGFR3 Signaling during Chondrogenesis and Achondroplasias. *Dev Cell* 2007; 13:872-883.
- 211.** Galán-Gómez E, Blesa Sánchez E, Arias-Castro S, Cardesa-García JJ. Intrauterine growth retardation, duodenal and extrahepatic biliary atresia, hypoplastic pancreas and other intestinal anomalies: further evidence of the Martínez-Frías syndrome. *Eur J Med Gene* 2007; 50:144-148.
- 212.** Martínez-Frías ML. Postmarketing analysis of medicines: Methodology and value of the Spanish case-control study and surveillance system in preventing birth defects. *Drug Safety* 2007; 30(4):307-316.
- 213.** Martínez-Frías ML. Comments on the "Systematic review of the fetal effects of prenatal binge-drinking" paper by Henders (*JECH* 2007; 61:1069-1073). *JECH* (13 Diciembre).
<http://jech.bmj.com/cgi/eletters/61/12/1069>.
- 214.** 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, McDonnell 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 Part A* 2007; 143A:660-671.
- 215.** Rodríguez L, Liehr T, Mrasek K, Mansilla E, Martínez-Fernández ML, García A, Martínez-Frías ML. Clinical Report: Small supernumerary chromosome marker generating complete and pure trisomy 18p, characterized by molecular cytogenetic techniques and review. *Am J Med Genet Part A* 2007; 143A:2727-2732.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 216.** Rodríguez L, Zollino M, Mansilla E, Martínez-Fernández ML, Pérez P, Murdolo M, Martínez-Frías ML. Clinical Report: The first 4p euchromatic variant in a healthy carrier having an unusual reproductive history. *Am J Med Genet Part A* 2007; 143A:995-998.
- 217.** Rouhani P, Fleming LE, Frías J, Martínez-Frías ML, Bermejo E, Mendioroz J. Pilot study of socioeconomic class, nutrition and birth defect in Spain. *Matern Child Health J* 2007; 11:403-405.
- 218.** Tönnies H, Pietrzak J, Bocian E, Macdermont K, Kuechler A, Belitz B, Trautmann U, Schmidt A, Schulze B, Rodríguez L, Binkert F, Yardin C, Kosyakova N, Volleth M, Mkrtychyan H, Schreyer I, von Eggeling F, Weise A, Mrasek K, Liehr T. New immortalized cell lines of patients with small supernumerary marker chromosome (sSMC): Towards the establishment of a cell bank. *J Histochem Cytochem* 2007; 55(6):651-660.
- 219.** Zollino M, Lecce R, Murdolo M, Orteschi D, Marangi G, Selicorni A, Midro A, Sorge G, Zampino G, Memo L, Battaglia D, Petersen M, Pandelia E, Gyftodimou Y, Faravelli F, Tenconi R, Garavelli L, Mazzanti L, Fischetto R, Cavalli P, Savasta S, Rodríguez L, Neri G. Wolf-Hirschhorn syndrome-associated chromosome changes are not mediated by olfactory receptor gene clusters nor by inversion polymorphism on 4p16. *Hum Genet* 2007; 122: 423-430.

Año 2008

- 220.** Bonaglia MC, Ciccone R, Gimelli G, Gimelli S, Marelli S, Verheij J, Giorda R, Grasso R, Borgatti R, Pagone F, Rodríguez L, Martínez-Frías ML, van Ravenswaaij C, Zuffardi O. Detailed phenotype-genotype study in five patients with chromosome 6q16 deletion: narrowing the critical region for Prader-Willi-like phenotype. *Eur J Hum Genet* 2008; 16:1443-1449. Epub 2008 Jul 23.
- 221.** Cotarelo RP, Valero MC, Prados B, Peña A, Rodríguez L, Fano O, Marco JJ, Martínez-Frías ML, Cruces J. Two new patients bearing mutations in the fukutin gene confirm the relevance of this gene in Walker-Warburg syndrome. *Clin Genet* 2008; 73:139-145.
- 222.** Martínez-Frías ML. The biochemical structure and function of methylenetetrahydrofolate reductase provide the rationale to interpret the epidemiological results on the risk for infants with Down syndrome. *Am J Med Genet Part A* 2008; 146A:1477-1482.
- 223.** Martínez-Frías ML. Genetic Drift: The Balance of Nature: Reflections on the physics and mathematics structure of the living world and the human genome. *Am J Med Genet Part A* 2008; 146A:1781-1787.
- 224.** Martínez-Frías ML, Grupo de trabajo del ECEMC. Epidemiological association between isolated skin marks in newborn infants and single umbilical artery (SUA). Does it have biological plausibility? *Am J Med Genet Part A* 2008; 146A:26-34.
- 225.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto D, y Grupo de Trabajo del ECEMC. 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 Part A* 2008; 146A:15-25.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 226.** Martínez-Frías ML, Rodríguez-Pinilla E. Problem of using cases with genetic anomalies as a reference group in case-control studies on drug use and birth defects. *Birth Defects Res (Part A): Clin Mol Teratol* 2008; 82:173-174 (author reply 175).
- 227.** 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; 45,5:525-532.
- 228.** Rodríguez L, Liehr T, Martínez-Fernández ML, Lara A, Torres A, Martínez-Frías ML. A new small supernumerary marker chromosome, generating mosaic pure trisomy 16q11.1–q12.1 in a healthy man. *Mol Cytogenet* 2008; 1:4.
- 229.** Rodríguez L, Martínez-Fernández ML, Mansilla E, Mendioroz J, Arteaga RM, Toral JF, Guardia NM, García A, Centeno F, Pantoja J, Jovani C, Martínez-Frías ML. Screening for subtelomeric chromosome alteration in a consecutive series of newborns with congenital defects. *Clin Dysmorphol* 2008; 17:5-12.
- 230.** Rodríguez L, Niebuhr E, García A, Martínez-Fernández ML, Peña Segura JL. Research Letter: Be careful with familial unbalanced chromosome abnormalities!. *Am J Med Genet Part A* 2008; 146A:2005-2007.
- 231.** Rodríguez L, Diego-Alvarez D, Lorda-Sanchez I, Gallardo FL, Martínez-Fernández ML, Arroyo-Muñoz ME, Martínez-Frías ML. A small and active ring X chromosome in a female with features of Kabuki syndrome. *Am J Med Genet Part A* 2008; 146A:2816-2821.
- 232.** Rodríguez-Pinilla E, Mejías C, Prieto-Merino D, Fernández P, Martínez-Frías ML; ECEMC Working Group. Risk of hypospadias in newborn infants exposed to valproic acid during the first trimester of pregnancy: a case-control study in Spain. *Drug Saf* 2008; 31(6):537-543.
- 233.** Trifonov V, Fluri S, Binkert F, Nandini A, Anderson J, Rodríguez L, Gross M, Kosyakova N, Mkrtychyan H, Ewers E, Reich D, Weise A, Liehr T. Complex rearranged small supernumerary marker chromosomes (sSMC), three new cases; evidence for an underestimated entity? *Mol Cytogenet* 2008; 1:6.
- 234.** Weber-Schoendorfer C, Hannemann D, Meister R, Eléfant E, Cuppers-Maarschalkerweerd B, Arnon J, Vial T, Rodríguez-Pinilla E, Clementi M, Robert-Gnansia E, De Santis M, Malm H, Dolivo A, Schaefer C. The safety of calcium channel blockers during pregnancy: a prospective, multicenter, observational study. *Reprod Toxicol* 2008; 26:24-30.

Año 2009

- 235.** Alías L, Bernal S, Fuentes-Prior P, Barceló MJ, Also E, Martínez-Hernández R, Rodríguez-Alvarez FJ, Martín Y, Aller E, Grau E, Peciña A, Antiñolo G, Galán E, Rosa AL, Fernández-Burriel M, Borrego S, Millán JM, Hernández-Chico C, Baiget M, Tizzano EF. Mutation update of spinal muscular atrophy in Spain: molecular characterization of 745 unrelated patients and identification of four novel mutations in the SMN1 gene. *Hum Genet* 2009; 125:29-39.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 236.** Bermejo E, Mendioroz J, Cuevas L, Martínez-Frías ML. Congenital Defects Associated with Congenital Diaphragmatic Hernia (CDH): Multivariate Analysis on a Large Series of Newborn Infants in Spain. *Birth Defects Res (Part A): Clin Mol Teratol* 2009; 85:240.
- 237.** Martínez-Frías ML and the ECEMC Working Group. Epidemiology of acephalus/acardius monozygotic twins: New insights into an epigenetic causal hypothesis. *Am J Med Genet Part A* 2009; 149A:640-649.
- 238.** 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 Ped Surg* 2009; 44:811-820.
- 239.** Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E and ECEMC Working Group. Letter to the Editor: Human genetics selection for the MTHFR 677>T polymorfism: A leap in the dark. 2009 February. Acceso: <http://www.biomedcentral.com/1471-2350/9/104/comments>
- 240.** Martínez-Frías ML. Conjoined twins presenting with different sex: Description of a second case that truly represents the earliest historical evidence in humans. *Am J Med Genet Part A* 2009; 149A:1595-1596.
- 241.** Martínez-Frías ML. Correspondence to the authors: Topiramate in pregnancy: Preliminary experience from the UK Epilepsy and Pregnancy Register. *Neurology* 2009; 72,23:2054-2055.
- 242.** Rodríguez L, Martínez-Fernández ML, Aceña MI, López Mendoza S, Martín Fumero L, Rodríguez de Alba M, Gallego-Merlo J, Martínez-Frías ML. Dicentric inverted duplication of entire 4p arm with no apparent deletion and internal placing of the (-TTAGGG-)n sequence: Description of the first patient. *Am J Med Genet Part A* 2009; 149A:1058-1061.
- 243.** Valencia M, Lapunzina P, Lim D, Zannolli R, Bartholdi D, Wollnik B, Al-Ajlouni O, Eid SS, Cox H, Buoni S, Hayek J, Martínez-Frías ML, Antonio PA, Temtamy S, Aglan M, Goodship JA, Ruiz-Perez VL. Widening the mutation spectrum of EVC and EVC2: ectopic expression of Weyer variants in NIH 3T3 fibroblasts disrupts hedgehog signaling. *Hum Mutat* 2009; 30,12:1667-1675.

Año 2010

- 244.** Bermejo E, Martínez-Frías ML. Prevention, diagnosis and services. *Adv Exp Med Biol* 2010; 686:55-75.
- 245.** Bermejo E, Martínez-Frías ML. Prevention, Diagnosis and Services. En el Libro: “Rare Diseases Epidemiology Book”. *Advances in Experimental Medicine and Biology Series*. Ed. M. Posada and SC Groft. Springer Science & Busines Media B.V. Dordrecht, Heidelberg, London, New York, 2010, pp. 55-76 (ISBN: 978-90-481-9484-1; e-ISBN: 978-90-481-9485-8).
- 246.** Fernández-Toral J, Rodríguez L, Plasencia A, Martínez-Frías ML, Ewers E, Hamid AB, Ziegler M, Liehr T. Four small supernumerary marker chromosomes derived from chromosomes 6, 8, 11 and 12 in a patient with minimal clinical abnormalities: a case report. *J Med Case Reports* 2010; 4:239.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 247.** MacDonald AH, Rodríguez L, Aceña I, Martínez-Fernández ML, Sánchez-Izquierdo D, Zuazo E, Martínez-Frías ML. Subtelomeric deletion of 12p: Description of a third case and review. *Am J Med Genet Part A* 2010; 152A:1561-1566.
- 248.** Martínez-Frías ML. Can our understanding of epigenetics assist with primary prevention of congenital defects? *J Med Genet* 2010; 47:73-80.
- 249.** Martínez-Frías ML, de Frutos CA, Bermejo E, ECEMC Working Group, Nieto MA. Review of the recently defined molecular mechanisms underlying Thanatophoric Dysplasia and their potential therapeutic implications for Achondroplasia. *Am J Med Genet Part A* 2010; 152A:245-255.
- 250.** Orrico A, Galli L, Faivre L, Clayton-Smith J, Azzarello-Burri SM, Hertz JM, Jacquemont S, Taurisano R, Arroyo Carrera I, Tarantino E, Devriendt K, Melis D, Thelle T, Meinhardt U, Sorrentino V. Aarskog-Scott syndrome: clinical update and report of nine novel mutations of the FGD1 gene. *Am J Med Genet Part A* 2010; 152A:313-318.
- 251.** Romanelli V, Belinchón A, Benito-Sanz S, Martínez-Glez V, Gracia-Bouthelie R, Heath KE, Campos-Barros A, García-Miñaur S, Fernández L, Meneses H, López-Siguero JP, Guillén-Navarro E, Gómez-Puertas P, Wesselink J-J, Mercado G, Esteban-Marfil V, Palomo R, Mena R, Sánchez A, del Campo M, Lapunzina P. *CDKN1C* (*p57^{Kip2}*) analysis in Beckwith-Wiedemann syndrome (BWS) patients: Genotype-phenotype correlations, novel mutations, and polymorphisms. *Am J Med Genet Part A* 2010; 152A:1390-1397.

Año 2011

- 252.** Arroyo Carrera I, García García MJ, Izquierdo Martín A, Martín Fernández R, Lapunzina Badía P, Orrico A. First reported splice site mutation (c.1935+3A>C) of the FGD1 gene in a patient with Aarskog-Scott syndrome. *Eur J Hum Genet* 2011; 19 Suppl 2:453. (Abstract de la European Human Genetics Conference 2011. Amsterdam, 2011)
- 253.** 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 Part C (Semin Med Genet)* 2011; 157:288-304.
- 254.** 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 Part C (Semin Med Genet)* 2011; 157:305-320.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 255.** Bonaglia MC, Giorda R, Beri S, De Agostini C, Novara F, Fichera M, Grillo L, Galesi O, Vetro A, Ciccone R, Bonati MT, Giglio S, Guerrini R, Osimani S, Marelli S, Zucca C, Grasso R, Borgatti R, Mani E, Motta C, Molteni M, Romano C, Greco D, Reitano S, Baroncini A, Lapi E, Cecconi A, Arrigo G, Patricelli MG, Pantaleoni C, D'Arrigo S, Riva D, Sciacca F, Dalla Bernardina B, Zoccante L, Darra F, Termine C, Maserati E, Bigoni S, Priolo E, Bottani A, Gimelli S, Bena F, Brusco A, di Gregorio E, Bagnasco I, Giussani U, Nitsch L, Politi P, Martínez-Frías ML, Martínez-Fernández ML, Martínez Guardia N, Bremer A, Anderlid BM, Zuffardi O. Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. *PLoS Genet* 2011 Jul;7(7):e1002173. Epub 2011 Jul 14.
- 256.** Botto LD, Feldkamp ML, Amar E, Carey JC, Castilla EE, Clementi M, Cocchi G, de Walle HEK, Halliday J, Leoncini E, Li Z, Lowry RB, Marengo LK, Martínez-Frías ML, Merlob P, Morgan M, Muñoz LL, Rissmann A, Ritvanen A, Scarano G, Mastroiacovo P. Acardia: Epidemiologic findings and literature review from the International Clearinghouse of Birth Defects Surveillance and Research. *Am J Med Genet Part C (Semin Med Genet)* 2011; 157:262-273.
- 257.** Feldkamp ML, Botto LD, Amar E, Bakker MK, Bermejo-Sánchez E, Bianca S, Canfield MA, Castilla EE, Clementi M, Csáky-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 of Birth Defects Surveillance and Research. *Am J Med Genet Part C (Semin Med Genet)* 2011; 157:333-343.
- 258.** Garrido-Allepuz C, Haro E, González-Lamuño D, Martínez-Frías ML, Bertocchini F, Ros MA. A clinical and experimental overview of sirenomelia: insight into the mechanisms of congenital limb malformations. *Dis Model Mech* 2011; 4:289-299.
- 259.** Greenlees R, Neville A, Addor MC, Amar E, Arriola L, Bakker M, Barisic I, Boyd PA, Calzolari E, Doray B, Draper E, Emil Vollset S, Garne E, Gatt M, Haeusler M, Kallen K, Khoshnood B, Latos-Bielenska A, Martínez-Frías ML, Materna-Kiryluk A, Matias Dias C, McDonnell B, Mullaney C, Nelen V, O'Mahony M, Pierini A, Queisser-Luft A, Randrianaivo-Ranjatoélina H, Rankin J, Rissmann A, Ritvanen A, Salvador J, Sipek A, Tucker D, Verellen-Dumoulin C, Wellesley D, Wertelecki W. Paper 6: EUROCAT member registries: Organization and activities. *Birth Defects Res (Part A): Clin Mol Teratol* 2011; 91:S51-S100.
- 260.** Martínez-Frías ML, Egiés 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 Part A* 2011; 155:197-202.
- 261.** Mutchinick OM, Luna-Muñoz L, Amar E, Bakker MK, Clementi M, Cocchi G, da Graça Dutra M, Feldkamp M, Landau D, Leoncini E, Li Z, Lowry B, Marengo LK, Martínez-Frías ML, Mastroiacovo P, Métneki J, Morgan M, Pierini A, Rissman A, Ritvanen A, Scarano G, Siffel C, Szabova E, Arteaga-Vázquez J. Conjoined twins: A worldwide collaborative epidemiological study of the International Clearinghouse for Birth Defects Surveillance and Research. *Am J Med Genet Part C (Semin Med Genet)* 2011; 157:274-287.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 262.** 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 Part C (Semin Med Genet)* 2011; 157:344-357.
- 263.** Orioli IM, Amar E, Arteaga-Vázquez J, Bakker MK, Bianca S, Botto LD, Clementi M, Correa A, Csáky-Szunyogh M, Leoncini E, López-Camelo JS, Li Z, Lowry RB, Marengo L, Martínez-Frías ML, Mastroiacovo P, Morgan M, Pierini A, Ritvanen A, Scarano G, Szabova E, Castilla EE. Sirenomelia: An epidemiologic study in a large dataset from the International Clearinghouse of Birth Defects Surveillance and Research, and literature review. *Am J Med Genet Part C (Semin Med Genet)* 2011; 157:358-373.
- 264.** Palomares M, Delicado A, Mansilla E, de Torres ML, Vallespín E, Fernández L, Martínez-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; 89:295-301.
- 265.** Romanelli V, Meneses HN, Fernández L, Martínez-Glez V, Gracia-Bouthelier R, Fraga M, Guillén E, Nevado J, Gean E, Martorell L, Esteban Marfil V, García-Miñaur S, Lapunzina P. Beckwith-Wiedemann syndrome and uniparental disomy 11p: fine mapping of the recombination breakpoints and evaluation of several techniques. *Eur J Hum Genet* 2011; 19(4):416-421.
- 266.** 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 Part C (Semin Med Genet)* 2011; 157:321-332.

Año 2012

- 267.** Carrera IA, Matthijs G, Perez B, Cerdá CP. DPAGT1-CDG: report of a patient with fetal hypokinesia phenotype. *Am J Med Genet A*. 2012; 158A,8:2027-30.
- 268.** Martínez-Frías ML. Assessing pre-implantation embryo development in mice provides a rationale for understanding potential adverse effects of ART and PGD procedures. *Am J Med Genet Part A* 2012; 158A:2526-2533.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

Año 2013

- 269.** 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 Part A* 2013; 161A:2281-2290.
- 270.** Girón Vallejo O, Benítez Sánchez MC, Salcedo Cánovas C, Díez Ontiveros J, Ruiz Jiménez JJ, Bermejo-Sánchez E, Martínez-Frías ML. Patient with disorganization syndrome: Surgical procedures, Pathology, and potential causes. *Birth Defects Res (Part A): Clin Mol Teratol.* 2013; 97:781-785.
- 271.** Martínez-Frías ML, Martínez Fernández ML. A highly specific coding system for structural chromosomal alterations. *Am J Med Genet Part A* 2013; 161A:732-736.

Año 2014

- 272.** 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 Part A* 2014; 164A:338-345.
- 273.** 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 Part A* 2014; 164A:639-647.
- 274.** Taruscio D, Arriola L, Baldi F, Barisic I, Bermejo-Sánchez E, Bianchi F, Calzolari E, Carbone P, Curran R, Garne E, Gatt M, Latos-Bieleńska A, Khoshnood B, Irgens L, Mantovani A, Martínez-Frías ML, Neville A, Reißmann 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; 17:115-123.

Año 2015

- 275.** 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; 167A:1243-1251.
- 276.** Bermúdez L, García-Vicent C, López J, Torró MI, Lurbe E. Assessment of ten trace elements in umbilical cord blood and maternal blood: association with birth weight. *J Transl Med.* 2015; 13:291. doi: 10.1186/s12967-015-0654-2.
- 277.** Faus-Pérez A, Sanchis-Calvo A, Codoñer-Franch P. Ciliopathies: An Update. *Pediatr Res Int J.* 2015. DOI: 10.5171/2015.935983.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 278.** 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, Tift 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; 97:99-110.
- 279.** 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 Part A* 2015; 167A:2034-2041.
- 280.** Martínez F, Marín-Reina P, Sanchis-Calvo A, Perez-Aytés A, Oltra S, Roselló M, Mayo S, Monfort S, Pantoja J, Orellana C. Novel mutations of NFIX gene causing Marshall-Smith syndrome or Sotos-like syndrome: one gene, two phenotypes. *Pediatr Res* 2015; 78(5):533-539. doi: 10.1038/pr.2015.135. Epub 2015 Jul 22.

Año 2016

- 281.** Carrera IA, de Zaldívar MS, Martín R, Begemann M, Soellner L, Eggermann T. Microdeletions of the 7q32.2 imprinted region are associated with Silver-Russell syndrome features. *Am J Med Genet A*. 2016,170,3:743-9.
- 282.** 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.

Año 2017

- 283.** Bermejo-Sánchez E, Posada de la Paz M. Congenital anomalies: Cluster detection and Investigation. *Adv Exp Med Biol*. 2017; 1031:535-557.
- 284.** Campos-Sanchez E, Deleyto-Seldas N, Dominguez V, Carrillo-de-Santa-Pau E, Ura K, Rocha PP, Kim J, Aljoufi A, Esteve-Codina A, Dabad M, Gut M, Heyn H, Kaneda Y, Nimura K, Skok JA, Martínez-Frías ML, Cobaleda C. Wolf-Hirschhorn Syndrome Candidate 1 Is Necessary for Correct Hematopoietic and B Cell Development. *Cell Rep*. 2017; 19:1586-1601. doi:10.1016/j.celrep.2017.04.069.
- 285.** Polo-Antúnez A, Arroyo-Carrera I. Severe Neurological Phenotype in a Girl with Xp22.31 Triplication. *Mol Syndromol*. 2017; 8:219-223. doi: 10.1159/000475795.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

Año 2018

- 286.** 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, 15, 1715; doi:10.3390/ijerph15081715.
- 287.** Bermejo-Sánchez E, Botto LD, Fedkamp ML, Groisman B, Mastroiacovo P. Value of sharing and networking among birth defects surveillance programs: an ICBDSR perspective. *Journal Community Genetics*, 2018; 9:411-415. doi: 10.1007/s12687-018-0387-z.
- 288.** 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, 15, 1746; doi:10.3390/ijerph15081746.

Año 2019

- 289.** Bakker MK, Kancherla V, Canfield MA, Bermejo-Sánchez E, Cragan JD, Dastgiri S, De Walle HEK, Feldkamp ML, Groisman B, Gatt M, Hurtado-Villa P, Kallen K, Landau D, Lelong N, López 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.
- 290.** Goel N, Morris JK, Tucker D, K. de Walle HE, Bakker MK, Kancherla V, Marengo L, Canfield MA, Kallen K, Lelong N, Camelo JL, Stallings EB, Jones AM, Nance A, Huynh M-P, Martínez-Fernández ML, Sipek A, Pierini A, Nembhard WN, Goetz D, Rissmann A, Groisman B, Luna-Muñoz L, Szabova E, Lapchenko S, Zarante I, Hurtado-Villa P, Martínez LE, Tagliabue G, Landau D, Gatt M, Dastgiri S, Morgan M. Trisomy 13 and 18 – Prevalence and mortality – A multi-registry population based analysis. *Am J Med Genet A*. 2019; 179(12):2382-2392. doi: 10.1002/ajmg.a.61365.
- 291.** Groisman B, Bermejo-Sánchez E, Romitti PA, Botto LD, Feldkamp ML, Walai SR, Mastroiacovo P. Join World Birth Defects Day. *Pediatr Res*. 2019; 86:3-4. <https://doi.org/10.1038/s41390-019-0392-x>.
- 292.** Llamosas-Falcón L, Bermejo-Sánchez E, Sánchez-Días 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. <https://doi.org/10.1186/s13023-019-1056-y>.
- 293.** Romero-Rodríguez E, Cuevas L, Simón L, ECEMC Peripheral Group, Bermejo-Sánchez E, Galán I. Changes in alcohol intake during pregnancy in Spain, 1980-2014. *Alcohol Clin Exp Res*. 2019; 43(11):2367-2373. doi: 10.1111/acer.14193.
- 294.** 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 Sanità*. 2019; 55(3):258-264. doi: 10.4415/ANN_19_03_11.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

295. 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, Fernández 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; 76(4):482-490. doi: 10.1016/j.eururo.2019.06.027.

Año 2020

296. 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-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; 41(9):1615-1628. doi: 10.1002/humu.24067.

297. Nembhard WN, Bergman JEH, Politis MD, Arteaga-Vázquez J, Bermejo-Sánchez E, Canfield MA, Cragan JD, Dastgiri S, de Walle HEK, Feldkamp ML, Nance A, Gatt M, Groisman B, Hurtado-Villa P, Kallén K, Landau D, Lelong N, Lopez-Camelo J, Martinez L, Morgan M, Pierini A, Rissmann A, Šípek A, Szabova E, Tagliabue G, Wertelecki W, Zarante I, Bakker MK, Kancherla V, Mastroiacovo P. A multi-country study of prevalence and early childhood mortality among children with omphalocele. *Birth Defects Res.* 2020; 112:1787-1801. doi: 10.1002/bdr2.1822. Epub 2020 Oct 17.

298. Palencia-Campos A, Martínez-Fernández ML, Altunoglu U, Soto-Bielicka P, Torres A, Marín P, Aller E, Sentürk L, Berköz Ö, Yildiran 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, Rossell 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 pathogenetic 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.

299. Tenorio J, Alarcón P, Arias P, Dapía I, García-Miñaur S, Palomares Bralo M, Campistol J, Climent S, Valenzuela I, Ramos S, Monseny AM, Grondona FL, Botet J, Serrano M, Solís M, Santos-Simarro F, Álvarez S, Teixidó-Tura G, Fernández Jaén A, Gordo G, Bardón Rivera MB, Nevado J, Hernández A, Cigudosa JC, Ruiz-Pérez VL, Tizzano EF; SOGRI Consortium, Lapunzina P. Further delineation of neuropsychiatric findings in Tatton-Brown-Rahman syndrome due to disease-causing variants in *DNMT3A*: seven new patients. *Eur J Hum Genet.* 2020; 28(4):469-479. doi: 10.1038/s41431-019-0485-3.

300. Urreizti R, Lopez-Martin E, Martinez-Monseny A, Pujadas M, Castilla-Vallmanya L, Pérez-Jurado LA, Serrano M, Natera-de Benito D, Martínez-Delgado B, Posada-de-la-Paz M, Alonso J, Marin-Reina P, O'Callaghan M, Grinberg D, Bermejo-Sánchez E, Balcells S. Five new cases of syndromic intellectual disability due to *KAT6A* mutations: widening the molecular and clinical spectrum. *Orphanet J Rare Dis.* 2020 Feb 10;15(1):44. doi: 10.1186/s13023-020-1317-9.

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

Año 2021

- 301.** Bell JC, Baynam G, Bergman JEH, Bermejo-Sánchez E, Botto LD, Canfield MA, Dastgiri S, Gatt M, Groisman B, Hurtado-Villa P, Kallen K, Khoshnood B, Konrad V, Landau D, Lopez-Camelo JS, Martinez L, Morgan M, Mutchinick OM, Nance AE, Nembhard W, Pierini A, Rissmann A, Shan X, Sipek A, Szabova E, Tagliabue G, Yevtushok LS, Zarante I, Nassar N. Survival of infants born with esophageal atresia among 24 international birth defects surveillance programs. *Birth Defects Res* 2021;113,12:945-957. doi: 10.1002/bdr2.1891.
- 302.** 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 Jr. A, Stevens S, Tucker D, Verellen-Dumoulin, de Walle HEK, Wellesley D, Wertelecki W, Bermejo-Sanchez E. Prevention of neural tube defects in Europe: A public health failure. *Front Pediatric* 9:647038. doi: 10.3389/fped.2021.647038.
- 303.** Nevado J, Bel-Fenellós C, Sandoval-Talamantes Ak, Hernández A, Biencinto-López C, Martínez-Fernández ML, Barrúz P, Santos-Simarro F, Mori-Álvarez MA, Mansilla E, García-Santiago FA, Valcorba I, Sáenz-Rico B, Martínez-Frías ML, Lapunzina P. Deep Phenotyping and Genetic Characterization of a Cohort of 70 Individuals With 5p Minus Syndrome. *Front Genet* 2021;12:645595. doi: 10.3389/fgene.2021.645595.
- 304.** Politis MD, Bermejo-Sánchez E, Canfield MA, Contiero P, Cragan JD, Dastgiri S, de Walle HEK, Feldkamp ML, Nance N, Groisman B, Gatt M, Benavides-Lara A, Hurtado-Villa P, Kallén K, Landau D, Lelong N, Lopez-Camelo J, Martinez L, Morgan M, Mutchinick OM, Pierini A, Rissmann A, Šípek A, Szabova E, Wertelecki W, Zarante I, Bakker MK, Kancherla V, Mastroiacovo P, Nembhard WN, International Clearinghouse for Birth Defects Surveillance and Research. Prevalence and mortality in children with congenital diaphragmatic hernia: a multicountry study. *Ann Epidemiol* 2021; 56:61-69.e3. doi: 10.1016/j.annepidem.2020.11.007.

Año 2022

- 305.** Gili JA, López-Camelo JS, Nembhard WN, Bakker M, de Walle HEK, Stallings EB, Kancherla V, ECEMC Peripheral Group, Contiero P, Dastgiri S, Feldkamp ML, Nance A, Gatt M, Martínez L, Canessa MA, Groisman B, Hurtado-Villa P, Källén K, Landau D, Lelong N, Morgan M, Arteaga-Vázquez J, Pierini A, Rissmann A, Sipek A, Szabova E, Wertelecki W, Zarante I, Canfield MA, Mastroiacovo P. Analysis of early neonatal case fatality rate among newborns with congenital hydrocephalus, a 2000-2014 multi-country registry-based study. *Birth Defects Res* 2022; 114,12:631-644.
- 306.** Kancherla V, Tandaki L, Sundar M, Lux A, Bakker MK, Bergman JE, Bermejo-Sánchez E, Canfield MA, Feldkamp ML, Groisman B, Hurtado-Villa P, Källén K, Landau D, Lelong N, Lopez-Camelo J, Mastroiacovo P, Morgan M, Mutchinick OM, Nance AE, Nembhard WN, Pierini A, Šípek A, Stallings EB, Szabova E, Wertelecki W, Zarante I, Rissmann A. A Multicountry Analysis of Prevalence and Mortality among Neonates and Children with Bladder Exstrophy. <<https://pubmed.ncbi.nlm.nih.gov/35644130/>> *Am J Perinatol* 2022 May 29. doi: 10.1055/s-0042-1748318. Online ahead of print. PMID: 35644130

PUBLICACIONES DEL ECEMC: PUBLICACIONES EN INGLÉS

- 307.** Salinas-Vilca A, Cuevas L, ECEMC Peripheral Group, Bermejo-Sánchez E, Galán I. Smoking during pregnancy: changes and associated risk factors in Spain, 1980-2016. *J Public Health (Oxf.)* 2022; 44,2:438-446.

Año 2023

- 308.** Goldrick NMc, Revie G, Groisman B, Hurtado-Villa P, Sipek A, Khoshnood B, Rissmann A, Dastgiri S, Landau D, Tagliabue G, Pierini A, Gatt M, Mutchinick OM, Martínez L, de Walle HEK, Szabova E, Lopez Camelo J, ECEMC Peripheral Group, Källén K, Morgan M, Wertelecki W, Nance A, Stallings EB, Nembhard WN, Mossey P. A multi-program analysis of cleft lip with cleft palate prevalence and mortality using data from 22 International Clearinghouse for Birth Defects Surveillance and Research programs, 1974-2014. *Birth Defects Res* 2023;115,10:980-997.
- 309.** González Álvarez LF, Tenorio-Castaño J, Poletta FA, Santos-Simarro F, Arias P, Gallego N, Orioli IM, Mundlos S, Castilla EE, Martínez-Glez V, Martínez-Frías ML, Ruiz-Pérez VL, Nevado J, Lapunzina P. A large, ten-generation family with autosomal dominant preaxial polydactyly/triphalangeal thumb: Historical, clinical, genealogical, and molecular studies. *Am J Med Genet A* 2023; 191,1:100-107.
- 310.** Kancherla V, Sundar M, Lucita T, Lux A, Bakker MK, Bergman JE, Bermejo-Sánchez E, Canfield MA, Dastgiri S, Feldkamp ML, Gatt M, Groisman B, Hurtado-Villa P, Kallen K, Landau D, Lelong N, Lopez-Camelo J, Martinez LE, Mastroiacovo P, Morgan M, Mutchinick OM, Nance AE, Nembhard WN, Pierini A, Sipek A, Stallings EB, Szabova E, Tagliabue G, Wertelecki W, Zarante I, Rissmann A. Prevalence and mortality among children with anorectal malformation: A multi-country analysis. <<https://pubmed.ncbi.nlm.nih.gov/36401554/>> *Birth Defects Res* 2023; 115,3:390-404.
- 311.** Kleven DL, Mai CT, Bermejo-Sánchez E, Groisman B, Walani S, Peck J, Cosentino V, Botto LD, Zezza S, Romitti PA, Mastroiacovo P. Using a health observance event to raise awareness: An assessment of World Birth Defects Day. <<https://pubmed.ncbi.nlm.nih.gov/37306055/>> *Birth Defects Res.* 2023;115,12:1140-1150.
- 312.** Onken M, Lohse L, Coulm B, Beghin D, Richardson JL, Bermejo-Sánchez E, Aguilera C, Bosch M, Cassina M, Chouchana L, De Santis M, Duman MK, Gören MZ, Johnson D, Bera APJ, Kaplan YC, Kennedy D, Kwok S, Lacroix I, Lepelley M, Pistelli A, Schaefer C, Te Winkel B, Uysal N, Winterfeld U, Yakuwa N, Diav-Citrin O, Vial T, Dathe K. Effects of maternal modafinil treatment on fetal development and neonatal growth parameters - a multicenter case series of the European Network of Teratology Information Services (ENITS). <<https://pubmed.ncbi.nlm.nih.gov/38110225/>> *Acta Psychiatr Scand* 2023; Dec 18. doi: 10.1111/acps.13643. Online ahead of print.