

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.

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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.

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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.

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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

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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.
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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.
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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.

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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.

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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.
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56. Martínez-Frías ML, Rodríguez-Pinilla E. Folic acid supplementation and neural tube defects (Letter to the Editor). *Lancet* 1992; 340:620.
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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.
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- 63.** Rodríguez JI, Rodríguez-Peralto JL, Muro M, Urioste M, Palacios J. Anencephaly and limb deficiencies. *Am J Med Genet* 1992; 44:66-71.

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- 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.
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- 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.

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- 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.
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- 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.
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- 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.
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- 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 aphalangia, 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.

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- 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.
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- 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.

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- 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.
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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.
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- 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.
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