

Publications

Neural Tube Defects

Soldano KL, Garrett ME, Cope HL, Rusnak M, Ellis NJ, Dunlap KL, Speer MC, Gregory SG, Ashley-Koch AE. (2013) Genetic association analyses of nitric oxide synthase genes and neural tube defects vary by phenotype. [Birth Defects Res \(Part B\) 98:365-373](#).

Cope H, McMahon K, Heise E, Eubanks S, Garrett M, Gregory S, Ashley-Koch A. (2013) [Outcome and life satisfaction of adults with myelomeningocele](#). Disability and Health J 6:236-243.

Krupp DR, Xu P, Thomas S, Dellinger A, Etchever HC, Vekemans M, Gilbert JR, Speer MC, Ashley-Koch AE, Gregory SG. (2012) Transcriptome profiling of gene involved in neural tube closure during human embryonic development using long serial analysis of gene expression (Long-SAGE). [Birth Defects Res \(Part A\) 94:683-692](#).

Deak KL, Siegel DG, George TM, Gregory S, Ashley-Koch A, Speer MC. *Further evidence for a maternal genetic effect and a sex-influenced effect contributing to risk for human neural tube defects*. (2008) [Birth Defects Res 82:662-669](#).

Torres OA, Palencia E, Lopez de Pratdesaba L, Grajeda R Fuentes M, Speer MC, Merrill AH Jr, O'Donnell K, Bacon CW, Glenn AE, Riley RT. *Estimated fumonisin exposure in Guatemala is greatest in consumers of lowland maize*. (2007) [Journal of Nutrition. 137\(12\):2723-9](#).

Rampersaud E., Morris RW, Weinberg CR, Speer MC, Martin ER. *Power calculations of likelihood ratio tests for genetic risks, maternal effects and parent-of-origin (POO) effects in the presence of missing parental genotypes when unaffected siblings are available*. [Genet Epidemiol.2007 Jan;31\(1\):18-30](#).

Cotten CM, Ginsburg GS, Goldberg RN, Speer MC. *Genomic analyses in neonatology*. [Journal of Pediatrics, 2006 Jun;148\(6\):720-726](#).

Stamm DS, Rampersaud E, Slifer SH, Mehlretter L, Siegel DG, Xie J, Hu-Lince D, Craig DW, Stephan DA, George TM, Gilbert JR, Speer MC and the NTD Collaborative Group. *High-density single nucleotide polymorphism screen in a large multiplex neural tube defect family refines linkage to loci at 7p21.1-pter and 2q33.1-q35*. [Birth Defects Research \(Part A\): Clinical and Molecular Teratology, 2006 Jun; 76\(6\):499-505](#).

Boyles A.L., Billups A.V., Deak K.L., Siegel D.G., Mehlretter L., Slifer S.H., Gassuk A.G., Kessler J.A., Reed M.C., Nijhout H.F., George T.M., Enterline D.S., Speer, M.C. and the NTD CollaborativeGroup. *Neural Tube Defects and folate pathway genes: family-based association tests of gene-gene and gene-environment interactions*, [Environmental Health Perspectives, 2006 Oct; 114\(10\):1547-52](#).

Boyles AL, Scott WK, Martin ER, Schmidt S, Li Y-J, Ashley-Koch A, Bass MP, Schmidt M, Pericak-Vance MA, Speer MC, Hauser ER. *Linkage disequilibrium inflates Type I error rates in multipoint linkage analysis when parental genotypes are missing*. [Human Heredity, 59: 200-227 \(2005\)](#).

Vieira A, Speer MC, Dickerson M, Murray J. *Studies of reduced folate carrier 1 (RFC1) A80G and 5,10-methylenetetrahydrofolate reductase (MTHFR) C677T polymorphisms with neural tube and orofacial cleft defects*. [American Journal of Medical Genetics, 135A:220-223, \(2005\)](#).

Detrait ER, George TM, Etchevers HC, Gilbert JR, Vekemans Michel, Speer MC. *Human Neural Tube Defects: Developmental Biology, Epidemiology, and Genetics*. [Neurotoxicology and Teratology, 27:515-524, 2005.](#)

Rampersaud E, Bassuk AG, Enterline DS, George TM, Siegel DG, Melvin EC, Aben J, Allen J, Aylsworth A, Brei T, Bodurtha J, Buran C, Floyd LE, Hammock P, Iskander B, Ito J, Kessler JA., Lasarsky N, Mack P, Mackey J, McLone D, Meeropol E, Mehlretter L, Mitchell LE, Oakes WJ, Nye JS, Powell C, Sawin K, Stevenson R, Walker M, West SG, Worley G, Gilbert JR, and Speer MC. (2005) *Whole genome-wide linkage screen for Neural Tube Defects reveals regions of interest on chromosomes 7 and 10*. [J Med Genet 42:940-946, 2005.](#)

Deak KL, Boyles AL, Etchevers HC, Melvin EC., Siegel DG, Graham FL, Slifer SH, Enterline DS, George TM, Vekemans M, McClay D, Bassuk AG, Kessler JA, Linney E, Gilbert JR, Speer MC. and NTD Collaborative Group. *SNPs in the neural cell adhesion molecule 1 gene (NCAM1) may be associated with human neural tube defects*. Human Genetics, May 10, 2005.

Boyles AL, Hammock P and Speer MC. *Candidate Gene Analysis in Human Neural Tube Defects*. [American Journal of Medical Genetics Part C \(Semin. Med. Genet.\)135C:9-23, 2005.](#)

Sebold CD, Melvin EC, Siegel DG, Mehlretter L, Enterline DS, Nye JS, Kessler J, Bassuk A, Speer MC., George TM, and the NTD Collaborative Group. *Recurrence risks for neural tube defects in siblings of patients with lipomyelomeningocele*. [Genet Med 7\(1\):64-67, 2005.](#)

Benz LP, Swift FE, Graham FL, Enterline DS, Melvin EC, Hammock P, Gilbert JR, Speer MC, Bassuk AG, Kessler JA, Timothy M, and the NTD Collaborative Group. *TERC is not a major gene in Human Neural Tube Defects*. [Clinical and Molecular Teratology, 70:531-533, 2004.](#)

Rampersaud E., Brusato C., Metcalf K., Melvin E, Speer MC and NTD Collaborative Group. *No evidence for heterozygote advantage at MTHFR in patients with lumbosacral myelomeningocele or their relatives*. [Genetics in Medicine. 6\(1\):69-70, January/February 2004.](#)

Rampersaud E, Melvin EC, Siegel D, Mehlretter L, Dickerson ME, George TM, Enterline D, Nye JS, Speer MC, The NTD Collaborative Group. *Updated investigations of the role of methylenetetrahydrofolate reductase in human neural tube defects*. [Clin Genet 63: 210-214, 2003.](#)

Speer MC, Melvin EC, Viles KD, Bauer KA, Rampersaud E, Drake C, George TM, Enterline DS, Mackey JF, Worley G, Gilbert JR and Nye JS. *T locus shows no evidence for linkage disequilibrium or mutation in American Caucasian neural tube defect families*. [Am J Med Genet 110:215-218, 2002.](#)

Bauer KA, George TM, Enterline DS, Melvin EC, Siegel DG, Samal S, Hauser MA, Nye JS, Speer MC, the NTD Collaborative Group. *Mutations in the Gene Encoding Noggin are not Causative in Human Neural Tube Defects*. [J Neurogenet 16:65-71, 2002.](#)

Vieira AR, Trembath D, Vandyke DC, Murray JC, Marker S, Lerner G, Bonner E, Speer M. *Studies with His475Tyr Glutamate Carboxipeptidase II Polymorphism and Neural Tube Defects*. [Am J Med Genetics 111:218-219, 2002.](#)

Thomas AQ, Lane K, Phillips III J, Prince M, Markin C, Speer M, Schwartz DA, Gaddipati R, Marney A, Johnson J, Roberts R, Haines J, Stahlman M. *Heterozygosity for a Surfactant Protein C Gene Mutation Associated with Usual*

Interstitial Pneumonitis and Cellular Nonspecific Interstitial Pneumonitis in One Kindred. [Am J Crit Care Med](#) 165:1322-1328, 2002.

George TM, Speer MC, and the NTD Collaborative Group. *Genetic and embryological approaches to studies of neural tube defects: A critical review.* [Neurol Res](#) 22:117-122, 2000.

Melvin EC, George TM, Worley G, Franklin A, Mackey J, Viles K, Shah N, Drake CR, Enterline DS, McLone D, Nye J, Oakes WJ, McLaughlin C, Walker ML, Peterson P, Brei T, Buran C, Aben J, Ohm B, Bermans I, Qumsiyeh M, Vance J, Pericak-Vance MA, Speer MC. NTD Collaborative Group. *Genetic studies in neural tube defects.* [Ped Neurosurg](#) 32(1):1-9, 2000.

Speer MC, Nye J, McLone D, Worley G, Melvin EC, Viles KD, Franklin A, Drake C, Mackey J, George TM & NTD Collaborative Group. *Possible interaction of genotypes at cystathionine β -synthase and methylenetetrahydrofolate reductase (MTHFR) in neural tube defects.* [Clinical Genetics](#), 56(2):142-144, 1999.

Speer MC, Worley G, Mackey JF, Melvin E, Oakes WJ, George TM and the NTD Collaborative Group. *The thermolabile variant of methylenetetrahydrofolate reductase (MTHFR) is not a major risk factor for neural tube defect in American Caucasians.* [Neurogenetics](#), 1:149-150, 1997.