

National Birth Defects Prevention Study
Genetics/Biospecimen-Related Projects

Published/In Press Manuscripts

2025

1. Carter, T. C., Kay, D. M., Pangilinan, F., Almli, L. M., Jenkins, M. M., Blue, E. E., Sok, P., White, J. J., Cunniff, C. M., Agopian, A. J., Bamshad, M. J., Botto, L. D., Brody, L. C., Gucsavas-Calikoglu, M., Chong, J. X., Gomez-Acevedo, H., Lupo, P. J., Moore, C. A., Nembhard, W. N., . . . University of Washington Center for Mendelian Genomics, NISC Comparative Sequencing Program, the National Birth Defects Prevention Study. (2025). Exome Sequencing to Identify Novel Susceptibility Genes for Nonsyndromic Split-Hand/Ft Malformation: A Report From the National Birth Defects Prevention Study. *Birth Defects Res*, 117(5), e2472. <https://doi.org/10.1002/bdr2.2472>

2024

2. Blue, E. E., Moore, K. J., North, K. E., Desrosiers, T. A., Carmichael, S. L., White, J. J., Chong, J. X., Bamshad, M. J., Jenkins, M. M., Almli, L. M., Brody, L. C., Freedman, S. F., Reefhuis, J., Romitti, P. A., Shaw, G. M., Werler, M., Kay, D. M., Browne, M. L., Feldkamp, M. L., National Institutes of Health Intramural Sequencing Center, University of Washington Center for Mendelian Genomics, National Birth Defects Prevention Study. (2024). Exome sequencing identifies novel genes underlying primary congenital glaucoma in the National Birth Defects Prevention Study. *Birth Defects Research*, 116(7), e2384. <https://doi.org/https://doi.org/10.1002/bdr2.2384>
3. Nicoletti, P., Zafer, S., Matok, L., Irron, I., Patrick, M., Haklai, R., Evangelista, J. E., Marino, G. B., Ma'ayan, A., Sewda, A., Holmes, G., Britton, S. R., Lee, W. J., Wu, M., Ru, Y., Arnaud, E., Botto, L., Brody, L. C., Byren, J. C., . . . Peter, I. (2024). Regulatory elements in SEM1-DLX5-DLX6 (7q21.3) locus contribute to genetic control of coronal nonsyndromic craniosynostosis and bone density-related traits. *Genet Med Open*, 2. <https://doi.org/10.1016/j.gimo.2024.101851>

2023

4. Huang, M., Lyu, C., Liu, N., Nembhard, W. N., Witte, J. S., Hobbs, C. A., & Li, M. (2023). A gene-based association test of interactions for maternal-fetal genotypes identifies genes associated with nonsyndromic congenital heart defects. *Genet Epidemiol*. <https://doi.org/10.1002/gepi.22533>
5. Blue, E. E., White, J. J., Dush, M. K., Gordon, W. W., Wyatt, B. H., White, P., Marvin, C. T., Helle, E., Ojala, T., Priest, J. R., Jenkins, M. M., Almli, L. M., Reefhuis, J., Pangilinan, F., Brody, L. C., McBride, K. L., Garg, V., Shaw, G. M., Romitti, P. A., . . . Bamshad, M. J. (2023). Rare variants in CAPN2 increase risk for isolated hypoplastic left

heart syndrome. *HGG Adv*, 4(4), 100232. <https://doi.org/10.1016/j.xhgg.2023.100232>

6. Sok, P., Sabo, A., Almli, L. M., Jenkins, M. M., Nembhard, W. N., Agopian, A. J., Bamshad, M. J., Blue, E. E., Brody, L. C., Brown, A. L., Browne, M. L., Canfield, M. A., Carmichael, S. L., Chong, J. X., Dugan-Perez, S., Feldkamp, M. L., Finnell, R. H., Gibbs, R. A., Kay, D. M., . . . University of Washington Center for Mendelian Genomics, NISC Comparative Sequencing Program, the National Birth Defects Prevention Study. (2023). Exome-wide assessment of isolated biliary atresia: A report from the National Birth Defects Prevention Study using child-parent trios and a case-control design to identify novel rare variants. *Am J Med Genet A*, 191(6), 1546-1556. <https://doi.org/10.1002/ajmg.a.63185>
7. Webber, D. M., Li, M., MacLeod, S. L., Tang, X., Levy, J. W., Karim, M. A., Erickson, S. W., Hobbs, C. A., & The National Birth Defects Prevention Study. (2023). Gene-Folic Acid Interactions and Risk of Conotruncal Heart Defects: Results from the National Birth Defects Prevention Study. *Genes (Basel)*, 14(1). <https://doi.org/10.3390/genes14010180>

2022

8. Hsu, P. C., Maity, S., Patel, J., Lupo, P. J., & Nembhard, W. N. (2022). Metabolomics Signatures and Subsequent Maternal Health among Mothers with a Congenital Heart Defect-Affected Pregnancy. *Metabolites*, 12(2), 100. <https://doi.org/10.3390/metabo12020100>
9. Li, J., Yang, W., Wang, Y. J., Ma, C., Curry, C. J., McGoldrick, D., Nickerson, D. A., Chong, J. X., Blue, E. E., Mullikin, J. C., Reefhuis, J., Nembhard, W. N., Romitti, P. A., Werler, M. M., Browne, M. L., Olshan, A. F., Finnell, R. H., Feldkamp, M. L., Pangilinan, F., . . . Shaw, G. M. (2022). Exome sequencing identifies genetic variants in anophthalmia and microphthalmia. *Am J Med Genet A*, 188(8), 2376-2388. <https://doi.org/10.1002/ajmg.a.62874>
10. Wong, E. C., Fisher, S. C., Feldkamp, M. L., Romitti, P. A., Nestoridi, E., & Desrosiers, T. A. (2022). Factors associated with maternal consent for use of residual newborn bloodspots in the National Birth Defects Prevention Study. *Birth Defects Res*. <https://doi.org/10.1002/bdr2.1991>
11. Pitsava, G., Feldkamp, M. L., Pankratz, N., Lane, J., Kay, D. M., Conway, K. M., Hobbs, C., Shaw, G. M., Reefhuis, J., Jenkins, M. M., Almli, L. M., Moore, C., Werler, M., Browne, M. L., Cunniff, C., Olshan, A. F., Pangilinan, F., Brody, L. C., Sicko, R. J., . . . Mills, J. L. (2022). Exome sequencing identifies variants in infants with sacral agenesis. *Birth Defects Res*, 114(7), 215-227. <https://doi.org/10.1002/bdr2.1987>
12. Rashkin, S. R., Cleves, M., Shaw, G. M., Nembhard, W. N., Nestoridi, E., Jenkins, M. M., Romitti, P. A., Lou, X. Y., Browne, M. L., Mitchell, L. E., Olshan, A. F., Lomangino, K., Bhattacharyya, S., Witte, J. S., & Hobbs, C. A. (2022). A genome-wide

association study of obstructive heart defects among participants in the National Birth Defects Prevention Study. *Am J Med Genet A*. <https://doi.org/10.1002/ajmg.a.62759>

2021

13. Patel, J., Bircan, E., Tang, X., Orloff, M., Hobbs, C. A., Browne, M. L., Botto, L. D., Finnell, R. H., Jenkins, M. M., Olshan, A., Romitti, P. A., Shaw, G. M., Werler, M. M., Li, J., Nembhard, W. N., & the National Birth Defects Prevention Study. (2021). Paternal genetic variants and risk of obstructive heart defects: A parent-of-origin approach. *PLOS Genetics*, 17(3), e1009413. <https://doi.org/10.1016/j.jpeds.2016.05.066>
14. Pitsava, G., Feldkamp, M. L., Pankratz, N., Lane, J., Kay, D. M., Conway, K. M., Shaw, G. M., Reefhuis, J., Jenkins, M. M., Almli, L. M., Olshan, A. F., Pangilinan, F., Brody, L. C., Sicko, R. J., Hobbs, C. A., Bamshad, M., McGoldrick, D., Nickerson, D. A., Finnell, R. H., . . . Mills, J. L. (2021). Exome sequencing of child-parent trios with bladder exstrophy: Findings in 26 children. *Am J Med Genet A*, 185(10), 3028-3041. <https://doi.org/10.1002/ajmg.a.62439>

2020

15. Lyu, C., Webber, D. M., MacLeod, S. L., Hobbs, C. A., Li, M., & National Birth Defects Prevention, S. (2020). Gene-by-gene interactions associated with the risk of conotruncal heart defects. *Mol Genet Genomic Med*, 8(1), e1010. <https://doi.org/10.1002/mgg3.1010>

2019

16. Hoang, T. T., Lei, Y., Mitchell, L. E., Sharma, S. V., Swartz, M. D., Waller, D. K., Finnell, R. H., Benjamin, R. H., Browne, M. L., Canfield, M. A., Lupo, P. J., McKenzie, P., Shaw, G., Agopian, A. J., & National Birth Defects Prevention Study. (2019). Maternal Lactase Polymorphism (rs4988235) Is Associated with Neural Tube Defects in Offspring in the National Birth Defects Prevention Study. *J Nutr*, 149(2), 295-303. <https://doi.org/10.1093/jn/nxy246>
17. Hoang, T. T., Lei, Y., Mitchell, L. E., Sharma, S. V., Swartz, M. D., Waller, D. K., . . . National Birth Defects Prevention Study. (2019). Maternal genetic markers for risk of celiac disease and their potential association with neural tube defects in offspring. *Mol Genet Genomic Med*, 7(6), <https://doi.org/10.1002/mgg3.688>
18. Jenkins, M. M., Almli, L. M., Pangilinan, F., Chong, J. X., Blue, E. E., Shapira, S. K., . . . National Birth Defects Prevention Study. (2019). Exome sequencing of family trios from the National Birth Defects Prevention Study: Tapping into a rich resource of genetic and environmental data. *Birth Defects Res*, 111(20), <https://doi.org/10.1002/bdr2.1554>

2018

19. Nembhard, W. N., Tang, X., Li, J., MacLeod, S. L., Levy, J., Schaefer, G. B., . . . National Birth Defects Prevention Study. (2018). A parent-of-origin analysis of paternal

genetic variants and increased risk of conotruncal heart defects. *Am J Med Genet A*, 176(3), 609-617. <https://doi.org/10.1002/ajmg.a.38611>

20. Tang, X., Eberhart, J. K., Cleves, M. A., Li, J., Li, M., MacLeod, S., Nembhard, W. N., & Hobbs, C. A. (2018). PDGFRA gene, maternal binge drinking and obstructive heart defects. *Sci Rep*, 8(1), 11083. <https://doi.org/10.1038/s41598-018-29160-9>

2017

21. Jenkins, M. M., Reefhuis, J., Herring, A. H., & Honein, M. A. (2017). Impact of sample collection participation on the validity of estimated measures of association in the National Birth Defects Prevention Study when assessing gene-environment interactions. *Genet Epidemiol*, 41(8), 834-843. <https://doi.org/10.1002/gepi.22088>
22. Moreno Uribe, L. M., Fomina, T., Munger, R. G., Romitti, P. A., Jenkins, M. M., Gjessing, H. K., . . . Wehby, G. L. (2017). A Population-Based Study of Effects of Genetic Loci on Orofacial Clefts. *J Dent Res*, 96(11), 1322-1329. <https://doi.org/10.1177/0022034517716914>
23. Nembhard, W. N., Tang, X., Hu, Z., MacLeod, S., Stowe, Z., Webber, D., & National Birth Defects Prevention, S. (2017). Maternal and infant genetic variants, maternal periconceptional use of selective serotonin reuptake inhibitors, and risk of congenital heart defects in offspring: population based study. *BMJ*, 356, j832. <https://doi.org/10.1136/bmj.j832>

2016

24. Li, M., Li, J., He, Z., Lu, Q., Witte, J. S., Macleod, S. L., . . . National Birth Defects Prevention Study. (2016). Testing Allele Transmission of an SNP Set Using a Family-Based Generalized Genetic Random Field Method. *Genet Epidemiol*, 40(4), 341-351. <https://doi.org/10.1002/gepi.21970>
25. Li, M., Li, J., Wei, C., Lu, Q., Tang, X., Erickson, S. W., . . . Hobbs, C. A. (2016). A Three-Way Interaction among Maternal and Fetal Variants Contributing to Congenital Heart Defects. *Ann Hum Genet*, 80(1), 20-31. <https://doi.org/10.1111/ahg.12139>
26. Patel, P. M., Momany, A. M., Schaa, K. L., Romitti, P. A., Druschel, C., Cooper, M. E., . . . Dagle, J. M. (2016). Genetic Modifiers of Patent Ductus Arteriosus in Term Infants. *J Pediatr*, 176, 57-61 e51. <https://doi.org/10.1016/j.jpeds.2016.05.066>

2015

27. Tai C. G., Graff R. E., Liu J., Passarelli M. N., Mefford J. A., Shaw G. M., Hoffmann T. J., Witte J. S. (2015) Detecting gene-environment interactions in human birth defects: Study designs and statistical methods. *Birth Defects Res A Clin Mol Teratol*, 103(8), 692-702. <https://doi.org/10.1002/bdra.23382>

28. Tang, X., Cleves, M. A., Nick, T. G., Li, M., MacLeod, S. L., Erickson, S. W., . . . National Birth Defects Prevention Study. (2015). Obstructive heart defects associated with candidate genes, maternal obesity, and folic acid supplementation. *Am J Med Genet A*, 167(6), 1231-1242. <https://doi.org/10.1002/ajmg.a.36867>
29. Tang, X., Hobbs, C. A., Cleves, M. A., Erickson, S. W., MacLeod, S. L., Malik, S., & National Birth Defects Prevention Study. (2015). Genetic variation affects congenital heart defect susceptibility in offspring exposed to maternal tobacco use. *Birth Defects Res A Clin Mol Teratol*, 103(10), 834-842. <https://doi.org/10.1002/bdra.23370>

2014

30. Glidewell, J., Reefhuis, J., Rasmussen, S. A., Woomert, A., Hobbs, C., Romitti, P. A., & Crider, K. S. (2014). Factors affecting maternal participation in the genetic component of the National Birth Defects Prevention Study-United States, 1997-2007. *Genet Med*, 16(4), 329-337. <https://doi.org/10.1038/gim.2013.143>
31. Hobbs, C. A., Cleves, M. A., Macleod, S. L., Erickson, S. W., Tang, X., Li, J., . . . National Birth Defects Prevention Study. (2014). Conotruncal heart defects and common variants in maternal and fetal genes in folate, homocysteine, and transsulfuration pathways. *Birth Defects Res A Clin Mol Teratol*, 100(2), 116-126. <https://doi.org/10.1002/bdra.23225>
32. Jenkins, M. M., Reefhuis, J., Gallagher, M. L., Mulle, J. G., Hoffmann, T. J., Koontz, D. A., . . . National Birth Defects Prevention Study. (2014). Maternal smoking, xenobiotic metabolizing enzyme gene variants, and gastroschisis risk. *Am J Med Genet A*, 164A(6), 1454-1463. <https://doi.org/10.1002/ajmg.a.36478>
33. Li, M., Cleves, M. A., Mallick, H., Erickson, S. W., Tang, X., Nick, T. G., . . . National Birth Defect Prevention Study. (2014). A genetic association study detects haplotypes associated with obstructive heart defects. *Hum Genet*, 133(9), 1127-1138. <https://doi.org/10.1007/s00439-014-1453-1>
34. Li, M., Erickson, S. W., Hobbs, C. A., Li, J., Tang, X., Nick, T. G., . . . National Birth Defect Prevention Study. (2014). Detecting maternal-fetal genotype interactions associated with conotruncal heart defects: a haplotype-based analysis with penalized logistic regression. *Genet Epidemiol*, 38(3), 198-208. <https://doi.org/10.1002/gepi.21793>
35. Lupo, P. J., Mitchell, L. E., Canfield, M. A., Shaw, G. M., Olshan, A. F., Finnell, R. H., . . . National Birth Defects Prevention Study. (2014). Maternal-fetal metabolic gene-gene interactions and risk of neural tube defects. *Mol Genet Metab*, 111(1), 46-51. <https://doi.org/10.1016/j.ymgme.2013.11.004>
36. Tang, X., Nick, T. G., Cleves, M. A., Erickson, S. W., Li, M., Li, J., . . . Hobbs, C. A. (2014). Maternal obesity and tobacco use modify the impact of genetic variants on the

occurrence of conotruncal heart defects. *PLoS One*, 9(9), e108903.

<https://doi.org/10.1371/journal.pone.0108903>

2012

37. Lupo, P. J., Canfield, M. A., Chapa, C., Lu, W., Agopian, A. J., Mitchell, L. E., . . . Zhu, H. (2012). Diabetes and obesity-related genes and the risk of neural tube defects in the National Birth Defects Prevention Study. *Am J Epidemiol*, 176(12), 1101-1109. <https://doi.org/10.1093/aje/kws190>
38. Lupo, P. J., Chapa, C., Noursome, D., Duhon, C., Canfield, M. A., Shaw, G. M., . . . National Birth Defects Prevention Study. (2012). A GCH1 haplotype and risk of neural tube defects in the National Birth Defects Prevention Study. *Mol Genet Metab*, 107(3), 592-595. <https://doi.org/10.1016/j.ymgme.2012.09.020>
39. Erickson, S. W., MacLeod, S. L., & Hobbs, C. A. (2012). Cheek swabs, SNP chips, and CNVs: Assessing the quality of copy number variant calls generated with subject-collected mail-in buccal brush DNA samples on a high-density genotyping microarray. *BMC Medical Genetics*, 13(1), 51. <https://doi.org/10.1186/1471-2350-13-51>

2011

40. Jenkins, M. M., Reed-Gross, E., Barfield, W. D., Prue, C. E., Gallagher, M. L., Rasmussen, S. A., & Honein, M. A. (2011). Qualitative assessment of study materials and communication strategies used in studies that include DNA collection. *Am J Med Genet A*, 155A(11), 2721-2731. <https://doi.org/10.1002/ajmg.a.34263>
41. Olshan, A. F., Hobbs, C. A., & Shaw, G. M. (2011). Discovery of genetic susceptibility factors for human birth defects: an opportunity for a National Agenda. *Am J Med Genet A*, 155A(8), 1794-1797. <https://doi.org/10.1002/ajmg.a.34103>
42. Cleves, M.A., Hobbs, C.A., Zhao, W., Krakowiak, P.A., MacLeod, S.L. (2011). Association between selected folate pathway polymorphisms and nonsyndromic limb reduction defects: A case-parental analysis. *Paediatric and Perinatal Epidemiology* 25(2), 124-134. <https://doi.org/10.1111/j.1365-3016.2010.01160.x>

2010

43. Schmidt, R. J., Romitti, P. A., Burns, T. L., Murray, J. C., Browne, M. L., Druschel, C. M., Olney, R. S., & National Birth Defects Prevention Study. (2010). Caffeine, selected metabolic gene variants, and risk for neural tube defects. *Birth Defects Res A Clin Mol Teratol*, 88(7), 560-569. <https://doi.org/10.1002/bdra.20681>

2009

44. Jenkins, M. M., Reed-Gross, E., Rasmussen, S. A., Barfield, W. D., Prue, C. E., Gallagher, M. L., & Honein, M. A. (2009). Maternal attitudes toward DNA collection for gene-environment studies: a qualitative research study. *Am J Med Genet A*, 149A(11),

2378-2386. <https://doi.org/10.1002/ajmg.a.33043>

2008

45. Jenkins, M. M., Rasmussen, S. A., Moore, C. A., & Honein, M. A. (2008). Ethical issues raised by incorporation of genetics into the National Birth Defects Prevention Study. *Am J Med Genet C Semin Med Genet*, 148C(1), 40-46. <https://doi.org/10.1002/ajmg.c.30157>