

# Stanford

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## Heather Byers, MD

Adjunct Clinical Assistant Professor, Pediatrics - Medical Genetics

### Bio

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#### LINKS

- Recurrent Pregnancy Loss Program: <http://med.stanford.edu/lathi>
- DSD Program: <https://www.stanfordchildrens.org/en/service/dsd>
- Stanford Clinical Genetics: <http://med.stanford.edu/medicalgenetics.html>

### Publications

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#### PUBLICATIONS

- **How geneticists think about Differences/Disorders of Sexual Development (DSD): A conversation.** *Journal of pediatric urology*  
Byers, H. M., Fossum, M., Wu, H.  
2020
- **Medical genetics education in the midst of the COVID-19 pandemic: Shared resources.** *American journal of medical genetics. Part A*  
Regier, D. S., Smith, W. E., Byers, H. M.  
2020
- **The Impact of Rapid Exome Sequencing on Medical Management of Critically Ill Children.** *The Journal of pediatrics*  
Freed, A. S., Clowes Candadai, S. V., Sikes, M. C., Thies, J. n., Byers, H. M., Dines, J. N., Ndugga-Kabuye, M. K., Smith, M. B., Fogus, K. n., Mefford, H. C., Lam, C. n., Adam, M. P., Sun, et al  
2020
- **Factor VIII and vWF deficiency in STT3A-CDG.** *Journal of inherited metabolic disease*  
Chang, I. J., Byers, H. M., Ng, B. G., Merritt, J. L., Gilmore, R., Shrimai, S., Wei, W., Zhang, Y., Blair, A. B., Freeze, H. H., Zhang, B., Lam, C.  
2018
- **Discordant sex between fetal screening and postnatal phenotype requires evaluation.** *Journal of perinatology : official journal of the California Perinatal Association*  
Byers, H. M., Neufeld-Kaiser, W., Chang, E. Y., Tsuchiya, K., Oehler, E. S., Adam, M. P.  
2018
- **Increasing the Participation of Pregnant Women in Clinical Trials.** *JAMA*  
Heyrana, K., Byers, H. M., Stratton, P.  
2018
- **Minimal mosaicism, maximal phenotype: Discordance between clinical and molecular findings in two patients with tuberous sclerosis.** *American journal of medical genetics. Part C, Seminars in medical genetics*  
Byers, H. M., Jensen, D. M., Glass, I. A., Bennett, J. T.  
2018
- **Postmortem Somatic Sequencing of Tumors From Patients With Suspected Lynch Syndrome Has Clinical Utility for Surviving Relatives.** *JCO precision oncology*  
Byers, H. M., Jacobson, A., McFaddin, A. S., Ussakli, C. H., Newlin, A., Stanich, P. P., More, S., Hamblett, A., Tait, J. F., Shirts, B., Pritchard, C. C., Konnick, E. Q., Lockwood, et al  
2018; 2

- **Expanding the phenotype of congenital central hypoventilation syndrome impacts management decisions.** *American journal of medical genetics. Part A*  
Byers, H. M., Chen, M., Gelfand, A. S., Ong, B., Jendras, M., Glass, I. A.  
2018
- **Acute liver failure in neonates with undiagnosed hereditary fructose intolerance due to exposure from widely available infant formulas** *MOLECULAR GENETICS AND METABOLISM*  
Li, H., Byers, H. M., Diaz-Kuan, A., Vos, M. B., Hall, P. L., Tortorelli, S., Singh, R., Wallenstein, M. B., Allain, M., Dimmock, D. P., Farrell, R. M., McCandless, S., Gambello, et al  
2018; 123 (4): 428–32
- **Postmortem Somatic Sequencing of Tumors From Patients With Suspected Lynch Syndrome Has Clinical Utility for Surviving Relatives** *JCO Precision Oncology*  
Byers, H. M., Jacobson, A., McFaddin, A. S., Ussakli, C. H., Newlin, A., Stanich, P. S., More, S., Hamblett, A., Tait, J., Shirts, B., Pritchard, C. C., Konnick, E. Q., Lockwood, et al  
2018; 2 (1): 1-7
- **Unexpected ethical dilemmas in sex assignment in 46,XY DSD due to 5-alpha reductase type 2 deficiency** *AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS*  
Byers, H. M., Mohnach, L. H., Fechner, P. Y., Chen, M., Thomas, I. H., Ramsdell, L. A., Shnorhavorian, M., McCauley, E. A., Oelschlager, A., Park, J. M., Sandberg, D. E., Adam, M. P., Keegan, et al  
2017; 175 (2): 260–67
- **Description of a new oncogenic mechanism for atypical teratoid rhabdoid tumors in patients with ring chromosome 22.** *American journal of medical genetics. Part A*  
Byers, H. M., Adam, M. P., Lacroix, A., Leary, S. E., Cole, B., Dobyns, W. B., Mefford, H. C.  
2017; 173 (1): 245-249
- **Dramatic Response After Lamotrigine in a Patient With Epileptic Encephalopathy and a De Novo CACNA1A Variant** *PEDIATRIC NEUROLOGY*  
Byers, H. M., Beatty, C. W., Hahn, S. H., Gospe, S. M.  
2016; 60: 79-82
- **Novel Report of Phosphoserine Phosphatase Deficiency in an Adult with Myeloneuropathy and Limb Contractures.** *JIMD reports*  
Byers, H. M., Bennett, R. L., Malouf, E. A., Weiss, M. D., Feng, J., Scott, C. R., Jayadev, S.  
2016; 30: 103-108
- **A proposed method to predict preterm birth using clinical data, standard maternal serum screening, and cholesterol** *AMERICAN JOURNAL OF OBSTETRICS AND GYNECOLOGY*  
Alleman, B. W., Smith, A. R., Byers, H. M., Bedell, B., Ryckman, K. K., Murray, J. C., Borowski, K. S.  
2013; 208 (6)
- **Variations in CRHR1 are associated with persistent pulmonary hypertension of the newborn** *PEDIATRIC RESEARCH*  
Byers, H. M., Dagle, J. M., Klein, J. M., Ryckman, K. K., McDonald, E. L., Murray, J. C., Borowski, K. S.  
2012; 71 (2): 162-167