



Jonathan A. Epstein, MD

William Wikoff Smith Professor of Medicine
Chair, Department of Cell and Developmental Biology
Scientific Director, Penn Cardiovascular Institute

[E-Mail](#)

Hospital of the University of Pennsylvania
Cardiovascular Medicine Division
Molecular Cardiology Laboratory
954 Biomedical Research Building II/III
421 Curie Boulevard
Philadelphia, PA 19104

[Appointments](#)

Contact: (215) 898-8731

Clinical Specialty:

- Cardiovascular Medicine

Practice Location:

- Hospital of the University of Pennsylvania

Education:

- MD: Harvard Medical School, Boston, MA
- Residency: Brigham and Women's Hospital, Boston, MA
- Fellowship: Harvard Medical School, Boston, MA
- Postdoctoral Fellowship: Harvard Medical School, Boston, MA

Research Interest:

- Congenital Heart Disease Research
- Genetics
- Molecular Biology
- Stem Cell Biology

Selected Publications:

1. Chen, F., Kook, H., Milewski, R., Gitler, A., Lu, M.M., Li, J., Nazarian, R., Schnepf, R., Kuangyu, J., Biben, C., Runke, G., Mackay, J., Novotny, J., Schwartz, R., Harvey, R., Mullins, M, Epstein, J.A.: Hop in an unusual homeobox gene that modulates cardiac development. *Cell* 110(6): 713-723, 2002.

2. Kochilas, L., Merscher-Gomez, S., Lu, M.M., Potluri, V., Liao, J., Kucherlapati, R., Morrow, B. and Epstein, J.A.: The role of neural crest during cardiac development in a mouse model of DiGeorge syndrome. *Developmental Biology* 251(1): 157-166, 2002.
3. Gitler, A.D., Zhu, Y., Ismat, F.A., Lu, M.M., Yamauchi, Y., Parada, L.P., Epstein, J.A.: Nf1 has an essential role in endothelial cells. *Nature Genetics* 33(1): 75-79, 2003.
4. Kook, H., Lepore, J., Gitler, A.D., Lu, M.M., Yung, W., Mackay, J., Zhou, R., Ferrari, V., Gruber, P. and Epstein, J.A.: Cardiac hypertrophy and histone deacetylase-dependent transcriptional repression mediated by the atypical homeodomain protein Hop, *Journal of Clinical Investigation* 112(6): 863-71, 2003.
5. Lang, D. and Epstein, J.A.: Sox10 and Pax3 physically interact to mediate activation of a conserved c-RET enhancer. *Human Molecular Genetics* 12(8): 937-945, 2003.
6. Lang, D., Brown, C.B., Milewski, R., Jiang, Q.J., Lu, M.M., and Epstein, J.A.: Distinct enhancers regulate neural expression of Pax7. *Genomics* 82(5): 553-560, 2003.
7. Brown, C.B., Wenning, J.M., Lu, M.M., Epstein, D.J., Meyers, E.N., Epstein, J.A.: Cre mediated excision of Fgf8 in the Tbx1 expression domain reveals a critical role for Fgf8 in cardiovascular development in the mouse. *Developmental Biology* 267(1): 190-202, 2004.
8. Gitler, A.D., Lu, M.M., Epstein, J.A.: Plexin D1 and semaphorin signaling are required in endothelial cells for cardiovascular development. *Developmental Cell* 7(14): 107-116, 2004.
9. Kaartinen, V., Dudas, M., Nagy, A., Sridurongrit, S., Lu, M.M. and Epstein, J.A.: Cardiac outflow tract defects in mice lacking Alk2 in neural crest cells. *Development* 131(14): 3481-3490, 2004.
10. Milewski, R., Chi, N.C., Li, J., Brown, C., Lu, M.M., Epstein, J.A.: Identification of minimal enhancer elements sufficient for Pax3 expression in neural crest and implication of TEAD-2 as a regulator of Pax3. *Development* 131(4): 829-837, 2004.
11. Torres-Vasquez, J., Gitler, A.D., Fraser, S.D., Berk, J.A., Pham, V.N., Fishman, M.C. Childs, S., Epstein, J.A., Weinstein, B.M.: Semaphorin-plexin signaling guides patterning of the developing vasculature. *Developmental Cell* 7(14): 117-123, 2004. Notes: co-senior author.
12. Lang, D., Lu, M.M., Huang, L., Engleka, K.A., Zhang, M., Chu, E.Y., Lipner, S., Skoutlchi, A., Millar, S., Epstein, J.A.: Pax3 functions at a nodal point in melanocyte stem cell differentiation. *Nature* 433: 884-887, 2005.
13. Brown, C.B., Engleka, K.A., Wenning, J., Lu, M.M., Epstein, J.A.: Identification of a hypaxial somite enhancer element regulating Pax3 expression in migrating myoblasts and characterization of hypaxial muscle Cre transgenic mice. *Genesis* 41 (4): 202-209, 2005.
14. Stoller, J.Z., Epstein, J.A.: Identification of a novel nuclear localization signal in Tbx1 that is deleted in DiGeorge syndrome patients harboring the 1223delC mutation. *Human Molecular Genetics* 14(7): 885-892, 2005.

15. Engleka, K.A., Gitler, A.D., Zhang, M., Zhou, D.D., High, F.A., Epstein, J.A.: Insertion of Cre into the Pax3 locus creates a new allele of Splotch and identifies unexpected Pax3 derivatives. *Developmental Biology* 280(2), 396-406, 2005.
16. Li, J., Zhu, X., Chen, M., Cheng, L., Zhou, D., Lu, M.M., Du, K., Epstein, J.A., Parmacek, M.S.: MRTF-B is required in cardiac neural crest for smooth muscle differentiation and cardiovascular development. *Proceedings of the National Academy of Sciences of the United States of America* 102(25), 8916-8921.
17. Stoller, J.Z., Epstein, J.A.: Cardiac neural crest. *Seminars in Cell and Developmental Biology* 16 (6), 704-15, 2005.
18. Liu, S., Liu, F, Schnieder, A.E., St. Amand, T., Epstein, J.A., Gutstein, D.E.: Distinct cardiac malformations caused by absence of connexin43 in the neural crest and in the non-crest neural tube. *Development* 2006, 133(10), 2063-73.
19. Nowotschin S., Liao J., Gage P.J., Epstein J.A., Campione M., Morrow B.E.: Related Articles Tbx1 affects asymmetric cardiac morphogenesis by regulating Pitx2 in the secondary heart field. *Development* 2006 Apr;133(8):1565-73.
20. Yin Z., Gonzales L., Kolla V., Rath N., Zhang Y., Lu M.M., Kimura S., Ballard P.L., Beers M.F., Epstein J.A., Morrisey E.E.: Hop functions downstream of Nkx2.1 and GATA6 to mediate HDAC-dependent negative regulation of pulmonary gene expression. *Am J Physiol Lung Cell Mol Physiol.* 291:L191-199, 2006.
21. Kook, H., Yung, W.W., Simpson, R.J., Kee, H.J., Shin, S., Lowry, J., Loughlin, F.E., Epstein, J.A., Mackay, J.P.: Analysis of the structure and function of the transcriptional co-regulator HOP. *Biochemistry*, 45: 10584-90, 2006.
22. Ismat F.A., Xu J., Lu M.M., Epstein J.A.: The neurofibromin GAP-related domain rescues endothelial but not neural crest development in Nf1 mice. *J Clin Invest*, 116: 2378-84, 2006.
23. Hannenhalli, S., Putt, M.E., Gilmore, J.M., Wang, J., Parmacek, M.S., Epstein, J.A., Morrisey, E.E., Margulies, K.B., Cappola, T.A.: Transcriptional genomics associates FOX transcription factors with human heart failure. *Circulation*, 114: 1269-76, 2006.
24. Luo, Y., High, F.A., Epstein, J.A., Radice, G.L.: N-cadherin is required for neural crest remodeling of the cardiac outflow tract. *Dev Biol.* 2006 Nov 15;299(2):517-28.
25. High, F.A., Zhang, M., Proweller, A., Tu, L.L., Parmacek, M.S., Pear, W.S., Epstein, J.A. : An essential role for Notch in neural crest during cardiovascular development and smooth muscle differentiation. *J Clin Invest.* 2007 February 1;117(2): 353-363.
26. Trivedi, C.M., Luo, Y., Zhan, Y., Zhang, M., Zhu, W., Wang, T., Floss, T., Goettlicher, M., Noppinger, P.R., Wurst, W., Ferrari, V.A., Abrams, C.S., Gruber, P.J., Epstein, J.A.: Hdac2 regulates the cardiac hypertrophic response by modulating Gsk3beta; activity. *Nat Med.* 2007 Mar;13(3):324-331.