Martina Brueckner, MD

Education:

B.S. (Chemistry)	University of Virginia, Charlottesville, Virginia	05-1980
M.D.	University of Virginia School of Medicine	05-1984

Academic Appointments:

06-1984-06-85	Pediatric Intern, University of Pittsburgh, School of Medicine
07-1985-06-87	Pediatric Resident, University of Pittsburgh, School of Medicine
07-1987-06-90	Pediatric Cardiology Fellow, Yale School of Medicine
07-1990-06-95	Assistant Professor of Pediatrics, Division of Cardiology, Yale School of Medicine
07-1995-06-99	Associate Research Scientist, Pediatrics, Division of Cardiology, Yale School of Medicine
07-1999-06-2000	Assistant Professor of Pediatrics, Division of Cardiology, Yale School of Medicine
07-2001-06-2005	Associate Professor of Pediatrics, Division of Cardiology, Yale School of Medicine
07-2005-06-2015	Associate Professor of Pediatrics (tenure), Division of Cardiology, Yale School of Medicine
07-2009-06-2015	Associate Professor of Genetics, Yale School of Medicine
07-2015-current	Professor of Pediatrics and Genetics, Yale School of Medicine

Board Certification:

American Board of Pediatrics	6-1989
American Board of Pediatrics, Sub-board in Pediatric Cardiology	12-1991, 6-1998, 11-2005, 12-2012, 12-2017

Professional Honors and Recognition:

2019	NHLBI Outstanding Investigator Award
2019	American Pediatric Society, member
2000-2003	American Heart Association Heilbrunn Fellow in Cardiovascular Research
2001	14th Patrick John Niland Memorial Lecturer, U. Mich.
2000	Gailani Faculty Award, Yale University School of Medicine
1993	American Heart Association, Established Investigator Award
1992-1995	Syntex Scholars Award for Cardiovascular Research
1990-1995	National Institutes of Health Physician Scientist award
1990-1995	American Heart Association Clinician Scientist Award

Invited Speaking Engagements (2009-current; national and international audience):

Invited Speaker, Dartmouth University Dept. of Biological Sciences, Hanover NH 2009
Discussion leader and elected chair, Gordon Conference on Cilia, IlCiocco, Italy 2009
Invited speaker, IGC workshop “Biophysical Mechanisms of Development”, Lisbon, Portugal 2009
Invited speaker, Boston Children’s Hospital Cardiovascular seminar series, Boston, MA 2010

Invited speaker, ASM, Portland, Or 2010
Vice-chair, Gordon Conference on Cilia and Mucociliary Interactions, Ventura, CA 2011
Invited Speaker, Emory University 15th annual BCMB Symposium, Atlanta, GA 2011
Invited Speaker, Symposium on Biology of Cilia, Albert Einstein College of Medicine, 2011
Invited Speaker, University of Pittsburgh Dept. of Genetics, 2012
Invited Speaker, Morehouse College, Atlanta GA, 2012
Chair, Gordon Conference on Cilia and Mucociliary Interactions, Il Ciocco, Italy 2013
Invited Speaker, European Society for Cardiology, Amsterdam, NE 2013
Invited Speaker, Cardiovascular Developmental Biology Center Symposium, MUSC, 2014
Session organizer and Speaker, SPR State-of-the-Art Plenary, Vancouver, BC 2014
Keynote Speaker, Institute Pasteur "Cilia 2014", Paris FR 2014
Invited Session leader, Gordon Conference on Cilia and Mucociliary Interactions, TX, 2015
Invited Speaker, FASEB Cilia meeting, CO, 2015
Invited Session Chair, Gordon Conference on Cilia and Mucociliary Interactions, TX, 2017
Invited Speaker, Fetal Cardiology Symposium, University of Maryland 2017
Invited Speaker and session chair, FASEB Cilia meeting, AZ, 2017
Invited Speaker, European Society of Cardiology Cardiac Development meeting, Padua, Italy, 2017
Invited Speaker, Medical University of South Carolina, SC, 2018
Keynote Speaker, Gordon Conference on Cilia and Mucociliary Interactions,IlCiocco, Italy, 2019
Invited Speaker, Johns Hopkins Cardioscience, Baltimore, MD, 2019
Invited Speaker, AHA, Philadelphia, PA 2019
Cazden Memorial Lecture, OHSU Department of Pediatrics, Portland, OR,2019
Pediatric Grand Rounds, Rady Children's Hospital, UCSD, San Diego, CA 3-2020 (deferred)
Invited Speaker, Company of Biologists "The Biology and Physics of LR Patterning", Buxted Park, UK, 2021

Professional Service:

Peer review groups:

1994-1998; 2015	American Heart Association. Lung and Development Study Group
2010, 2016	Ad-hoc reviewer for NIH CDD study section
2017	NIH DEV1 study section
2019	NIH-NHLBI R35 Study Section
2020	NIH-NHLBI F31 Study Section

Journal Service:

1999-2007	*Circulation*, Editorial board
2000-curent	Manuscript reviews including *Nature, PLOS, Science, Development, Dev Biol, PNAS*

Professional Organization Service:

2014-2015, 2017	PCGC Steering committee chair
2011-2013	Chair, Gordon Conference on Cilia and Mucociliary interactions
2009-2011	American Heart Association, Founders Affiliate Research Committee
2017-2018	AHA working group on genetic basis for congenital heart disease
2020	Session organizer, World Congress for Pediatric Cardiology and Cardiac Surgery

Yale University Service:

2005-current	Yale University School of Medicine, Executive admissions committee
2013-current	Yale University School of Medicine, Curriculum com., embryology
2013	Chair, Yale Department of Pediatrics Committee on Basic Research
2015-current	Yale genome editing core advisory board
2016-curent	Appointments and Promotions, Yale School of Medicine Department of Pediatrics

Public Service:
Mentor for High School Students (Jo Michelle Corales, Josie Lee)

Clinical Activities

Attending Physician, Pediatric Cardiology Inpatient Service
Responsible for care of pediatric cardiology and pediatric cardiac surgery patients at the Yale-New Haven Children's Hospital in the PICU, NBSCU and the general inpatient floors. Responsible for pediatric cardiology consults.

Attending Physician, Pediatric Cardiology Outpatient Clinic
Responsible for evaluation and follow-up of full range of pediatric cardiology patients.

Attending Physician and Founder, Yale Cardiac Genetics Clinic
Responsible for a joint genetics-cardiology clinic that provides comprehensive diagnostic evaluation and follow-up care for patients with genetic-cardiovascular disease.

Bibliography:

1. Peer-reviewed original Articles:

1. **M.Brueckner**, P.D'Eustachio and A.L.Horwich. Linkage mapping of a mouse gene, iv, that controls left-right asymmetry of the heart and viscera. PMID: 2740340 *Proc. Natl. Acad. Sci. USA,* 86:5035-5038 (1989).

2. J.McGrath, A.Horwich, and **M.Brueckner**. Duplication/Deficiency mapping of Iv, a gene which determines normal left:right body axis asymmetry in the mouse. PMID: 1427890 *Genomics,* 14:643-648 (1992).

3. J.Chang, **M.Brueckner**, and R.J.Touloukian. Intestinal rotation and fixation abnormalities in the heterotaxia syndrome: early detection and management. PMID: 8263687 *J.Peds.Surg*, 28:1281-1285 (1993).

4. D. Rounds, **M.Brueckner** and D.Ward. Isolation of Murine Telomere-proximal sequences by affinity capture and PCR. PMID: 8575753 *Genomics,* 29:616-622 (1995).

5. D.M.Supp, S.S.Potter, D.P.Witte, and **M.Brueckner**. Mutation of an axonemal dynein affects left-right asymmetry in inversus viscerum mice. PMID: 9353118 *Nature*, 389:963-966 (1997).

6. T.Pehlivan, B.Pober, **M.Brueckner**, S.Garrett, R.Slaugh, R.VanRheeden, D.B. Wilson, M.S. Watson, and A.V. Hing. T. GATA4 Haploinsufficiency in Patients with Interstitial Deletion of Chromosome Region 8p23.1 and Congenital Heart Disease. PMID: 10096597 *American Journal of Medical Genetics*, 83:201-206 (1999).

7. D.M. Supp*, **M. Brueckner***, M.R. Kuehn, D.P. Witte, L.A. Lowe, J. McGrath, J. Corrales, and S.S. Potter. Targeted deletion of the ATP binding domain of left-right dynein confirms its role in specifying development of left-right asymmetries. PMID: 10556073 *Development,* 126: 5495-5504 (1999).

8. J. J Essner, K.J.Vogan, M.K.Wagner, C.J.Tabin, H.J.Yost, and **M.Brueckner**. Conserved function for embryonic nodal cilia. PMID: 12097899 *Nature*, 418:37-38 (2002)

9. J.McGrath, S.Somlo, S.Makova, X.Tian and **M.Brueckner**. Two populations of node monocilia initiate left-right asymmetry in the mouse. PMID: 12859898 *Cell,* 114:61-73 (2003)

10. H.Hallaq, E.Pinter,J.Enisco,J.McGrath,C.Zeiss, **M.Brueckner**, J.Madri, H.C.Jacobs, C.M.Wilson, H.Vasavada,X.Jiang, and C.W.Bogue. A null mutation of Hhex results in abnormal cardiac development, defective vasculogenesis and elevated VEGFA levels. PMID: 15459110 *Development*, 131:5197-5209 (2004)

11. A.F.Cooper, K.P.Yu, **M.Brueckner**, J.M.McGrath, A.E.Bale. Cardiac and CNS defects ia a mouse with targeted disruption of suppressor of fused. PMID: 16155214 *Development*, 132:4407-4417 (2005)

12. D.A.McDermott, M.A. Bressan, J.He, J.S.Lee, S.Aftimos, **M.Brueckner**, F.Gilbert, G.E.Graham, J.W. Innis, M.E.M. Pierpont, A.Raas-Rothschild, A.L.Shanske, W.E.Smith, R.H.Spencer, M.G.St. John-Sutton, L.van Maldergem, D.J.Waggoner, C.T.Basson. TBX5 Genetic Testing Validates Strict Clinical Criteria for Holt-Oram Syndrome. PMID: 16183809 *Pediatric Research*, 58(5):981-6, (2005)

13. E.E.Davis, **M.Brueckner**, N.Katsanis. The Emerging Complexity of the Vertebrate Cilium: New Functional Roles for an Ancient Organelle. PMID: 16824949 *Developmental Cell*, 11:9-19 (2006)

14. B.Weeks, D.B.Tashjian, **M.Brueckner**, R.J.Touluokian. Outcomes following Ladd Procedure for Intestinal Malrotation with Heterotaxia. PMID: 17336193 *J.Peds.Surg*, 42:528-31 (2007)

15. G.N. Gathungu, D.A. Pashankar, C.D. Sarita-Reyes, E. Zambrano, M. Reyes-Mugica, **M. Brueckner**, P.K. Mistry, S.Z. Husain. Microvillus inclusion disease associated with coarctation of the aorta and bicuspid aortic valve. PMID: 18277898 *J Clin Gastroenterol*, 42(4):400-3 (2008)

16. J.Slough, L.Cooney, **M.Brueckner**. Monocilia in the embryonic mouse heart suggest a direct role for cilia in cardiac morphogenesis. PMID: 18729223 *Developmental Dynamics,* 237:2304-2314 (2008)

17. K.A.Fakhro, M.Choi, S.M.Ware, J.W.Belmont, J.A.Towbin, R.P.Lifton, M.K.Khokha, **M.Brueckner**. Rare copy number variations in congenital heart disease patients identify unique genes in left-right patterning. PMID: 21282601 *PNAS*, 108(7):2915-20 (2011)

18. B. Bisgrove, S. Makova, H.J.Yost, **M.Brueckner**. RFX2 is essential for development of LR asymmetry and identifies a population of organizer cells sufficient for effective nodal flow. PMID: 22233545 *Developmental Biology*, 363(1) 166-178 (2012)

19. Pediatric Cardiac Genomics Consortium, B. Gelb, **M. Brueckner**, W. Chung, E. Goldmuntz, J. Kaltman, J.P. Kaski, R. Kim, J. Kline, L. Mercer-Rosa, G. Porter, A. Roberts, E. Rosenberg, H. Seiden, C. Seidman, L. Sleeper, S. Tennstedt, C. Schramm, K. Burns, G. Pearson. The congenital heart disease genetic network study: rationale, design and early results. PMID: 23410879 *Circ Res,* 112(4):698-706 (2013)

20. S. Zaidi, M. Choi, H. Wakimoto, L. Ma, J. Jiang, J.D. Overton, A. Romano-Adesman, R.D. Bjornson, R.E. Breitbart, K.K. Brown, N.J. Carriero, Y.H. Cheung, J. Deanfield, S. Depalma, K.A. Fakhro , J. Glessner, H. Hakonarson, M.J. Italia, J. Kaltman, J. Kaski, R. Kim, J.K. Kline, T. Lee, J. Leipzig, A. Lopez, S.M. Mane, L.E. Mitchell, J.W. Newburger, M. Parfeno, I. Pe'er, G. Porter, A.E. Roberts, R. Sachidanandam, S.J. Sanders, H.S. Seiden, M.W. State, S. Subramanian, I.R. Tikhonova, W. Wang, D. Warburton, P.S. White, I.A. Williams, H. Zhao, J.G. Seidman, **M. Brueckner (co-corresponding author)**, W.K. Chung, B.D. Gelb, E. Goldmuntz, C.E. Seidman, R.P. Lifton. De novo mutations in histone-modifying genes in congenital heart disease. PMID: 23665959 *Nature*, 498:220-223(2013)

21. M. Boskovski, S. Yuan, N.B. Pedersen, C.K. Goth, M. Makova, H. Clausen, **M. Brueckner**, M.K. Khokha. The heterotaxy gene, GALNT11, glycosylates Notch to orchestrate cilia type and laterality. PMID: 24226769 *Nature*, 504:456-459 (2013)

22. D. Backenroth, J. Homsy, L.R. Murillo, J. Glessner, E. Lin, **M. Brueckner**, R.P. Lifton, E. Goldmuntz, W.K. Chung, Y. Shen. CANOES: detecting rare copy number variants from whole exome sequencing data. PMID: 24771342 *Nucleic Acids Res*, 42(12)e97 (2014)

23. J.T. Glessner, A.G. Bick, K. Ito, J. Homsy, L. Rodriguez-Murillo, M. Fromer, E. Mazaika, B. Vardarajan, M. Italia, J. Leipzig, S.R. DePalma, R. Golhar, S.J. Sanders, B. Yamron, M. Ronemus, I. Iossifov, A.J. Willsey, M.W. State, J.R. Kaltman, P.S. White, Y. Shen, D. Warburton, **M. Brueckner**, C. Seidman, E. Goldmuntz, B.D. Gelb, R. Lifton, J. Seidman, H. Hakonarson, W.K. Chung. Increased frequency of de-novo copy number variations in congenital heart disease by integrative analysis of SNP array and exome sequence data. PMID: 25205790 *Circulation Research* 115(10):884-96 (2014)

24. S. Yuan, L. Zhao, **M. Brueckner*,** Z. Sun*. Intraciliary calcium waves initiate vertebrate left-right asymmetry. PMID: 25660539 *Current Biology* 2:25(5):555-67 (2015)

25. S.J. Endicott, B. Basu, M.K. Khokha, **M. Brueckner**. The NIMA-like kinase Nek2 is a key switch balancing ciliogenesis and resorption in the development of left-right asymmetry. PMID: 26493400 *Development*, 142(23):4068-79 (2015)

26. J. Homsy, S. Zaidi, Y. Shen, J.S. Ware, K.E. Samocha, K.J. Karczewski, S.R. DePalma, D. McKean, H. Wakimoto, J. Gorham, S.C. Jin, J. Deanfield, A. Giardini, G.A. Porter Jr, R. Kim, K. Bilguvar, F. López-Giráldez, I. Tikhonova, S. Mane, A. Romano-Adesman, H. Qi, B. Vardarajan, L. Ma, M. Daly, A.E. Roberts, M.W. Russell, S. Mital, J.W. Newburger, J.W. Gaynor, R.E. Breitbart, I. Iossifov, M. Ronemus, S.J. Sanders, J.R. Kaltman, J.G. Seidman, **M. Brueckner**, B.D. Gelb, E. Goldmuntz, R.P. Lifton, C.E. Seidman, W.K. Chung. De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. PMID: 26785492 *Science* 350(6265):1262-6 (2015).

27. G.W. Dougherty, N.T. Loges, J.A. Klinkenbusch, H. Olbrich, P. Pennekamp, T. Menchen, J. Raidt, J. Wallmeier, C. Werner, C. Westermann, C. Ruckert, V. Mirra, R. Hjeij, Y. Memari, R. Durbin, A. Kolb-Kokocinski, K. Praveen, M.A. Kashef, S. Kashef, F. Eghtedari, K. Häffner, P. Valmari, G. Baktai, M. Aviram, L. Bentur, I. Amirav, E.E. Davis, N. Katsanis, **M. Brueckner**, A. Shaposhnykov, G. Pigino, B. Dworniczak, H. Omran. DNAH11 Localization in the Proximal Region of Respiratory Cilia Defines Distinct Outer Arm Dynein Complexes. PMID: 26909801 *Am J Respir Cell Mol Biol*. EPub ahead of print (2016)

28. S. Yuan, **M. Brueckner**. Visualization and manipulation of cilia and intraciliary calcium in the zebrafish left-right organizer. PMID: 27514920 *Methods in Molecular Biology* 1454:123-47 (2016)

29. D.M. McKean, J. Homsy, H. Wakimoto, N. Patel, J. Gorham, S.R. DePalma, J.S. Ware, S. Zaidi, W. Ma, N. Patel, R.P. Lifton, W.K. Chung, R. Kim, Y. Shen, **M. Brueckner**, E. Goldmuntz, A.J. Sharp, C.E. Seidman, B.D. Gelb, J.G. Seidman. Loss of RNA expression and allele-specific expression associated with congenital heart disease. PMID: 27670201 *Nature Communications* 27:7:12824 (2016)

30. S.C. Jin, J. Homsy, S. Zaidi, Q. Lu, S. Morton, S.R. DePalma, X. Zeng, H. Qi, W. Chang, M.C. Sierant, W.C. Hung, S. Haider, J. Zhang, J. Knight, R.D. Bjornson, C. Castaldi, I.R. Tikhonova, K. Bilguvar, S.M. Mane, S.J. Sanders, S. Mital, M.W. Russell, J.W. Gaynor, J. Deanfield, A. Giardini, G.A. Porter Jr, D. Srivastava, C.W. Lo, Y. Shen, W.S. Watkins, M. Yandell, H.J. Yost , M. Tristani-Firouzi, J.W. Newburger, A.E. Roberts, R. Kim, H. Zhao, J.R. Kaltman, E. Goldmuntz, W.K. Chung, J.G. Seidman, B.D. Gelb, C.E. Seidman, R.P. Lifton, **M. Brueckner**. Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. PMID: 28991257 *Nature Genetics* 49(11):1593-1601 (2017)

31. K.B. Manheimer, N. Patel, F. Richter, J. Gorham, A.C. Tai, J. Homsy, M.T. Boskovski, M. Parfenov , E. Goldmuntz, W.K. Chung, **M. Brueckner**, M. Tristani-Firouzi, D. Srivastava, J.G. Seidman, C.E. Seidman, B.D. Gelb, A.J. Sharp. Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. PMID

29527824 *Hum Mutat.* 39(6): 870-881. doi: 0.1002/humu.23419. Epub 2018 Mar 22.(2018)

32. K.B. Manheimer, F. Richter, L.J. Edelmann, S.L. D'Souza, L. Shi, Y. Shen, J. Homsy, M.T. Boskovski, A.C. Tai, J. Gorham, C. Yasso, E. Goldmuntz, M. **Brueckner**, R.P. Lifton, W.K. Chung, C.E. Seidman, J.G. Seidman, B.D. Gelb. Robust identification of mosaic variants in congenital heart disease. PMID 29417219 *Hum Genet.* 2018 Feb;137(2):183-193. doi: 10.1007/s00439-018-1871-6. (2018)

33. T.T. Hoang, E. Goldmuntz, A.E. Roberts, W.K. Chung, J.K. Kline, J.E. Deanfield, A. Giardini, A. Aleman, B.D. Gelb, M. Mac Neal, G.A. Porter Jr. R. Kim, **M. Brueckner**, R.P. Lifton, S. Edman, S. Woyciechowski, L.E. Mitchell, A.J. Agopian. The Congenital Heart Disease Genetic Network Study: Cohort Description. PMID: 29351346 *PLoS One* 13:e0191319 (2018)

34. S.J. Endicott and **M. Brueckner**. NUP98 sets the Size-Exclusion Diffusion Limit through the Ciliary base. PMID: 29731308 *Current Biology* 21;28(10):1643-1650 doi: 10.1016/j.cub.2018.04.014 (2018)

35. M.E. Pierpont, **M. Brueckner**, W.K. Chung, PhD; V. Garg, R.V. Lacro, A.L. McGuire, S. Mital, J.R. Priest, W.T. Pu, A. Roberts, S.M. Ware, B.D. Gelb, M.W. Russell, on behalf of the American Heart Association Council on Cardiovascular Disease in the Young, Council on Cardiovascular and Stroke Nursing, and Council on Functional Genomics and Translational Biology. AHA Scientific Statement Genetic Basis for Congenital Heart Disease: Revisited A Scientific Statement from the American Heart Association. PMID: 30571578 *Circulation* 138(21):e653-e711 (2018)

36. A. Robson, S.Z.Makova**,** S.Barish, S.Zaidi, S.Mehta, J.Dorzd, S.C.Jin, B.D.Gelb, C.E.Seidman, W.K.Chung, R.P.Lifton, M.K.Khokha, and **M.Brueckner.** Histone H2B monoubiquitination regulates heart development via epigenetic control of cilia motility. PMID 31235600 *Proc Natl Acad Sci USA* 116(28):14049-14054 doi: 10.1073/pnas.1808341116 (2019).

37. W.S.Watkins, E.J.Hernandez, S.Wesolowski, B.W.Bisgrove, R.T.Sunderland, E.Lin, G.Lemmon, B.L.Demarest, T.A.Miller, D.Bernstein, **M.Brueckner**, W.K.Chung, B.D.Gelb, E.Goldmuntz, J.W.Newburger, C.E.Seidman, Y.She, H.J.Yost, M.Yandell, M.Tristani-Firouzi. De-novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. PMID 31624253 *Nature Communications* 10:4722 (2019)

38. Boskovski MT, Homsy J, Nathan M, Sleeper LA, Morton S, Manheimer KB, Tai A, Gorham J, Lewis M, Swartz M, Alfieris GM, Bacha EA, Karimi M, Meyer D, Nguyen K, Bernstein D, Romano-Adesman A, Porter GA Jr, Goldmuntz E, Chung WK, Srivastava D, Kaltman JR, Tristani-Firouzi M, Lifton R, Roberts AE, Gaynor JW, Gelb BD, Kim R, Seidman JG, Brueckner M, Mayer JE Jr, Newburger JW, Seidman CE. De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. PMID: 32812804 *Circ Genom Precis Med*. (2020)

39. Hsieh A, Morton SU, Willcox JAL, Gorham JM, Tai AC, Qi H, DePalma S, McKean D, Griffin E, Manheimer KB, Bernstein D, Kim RW, Newburger JW, Porter GA Jr, Srivastava D, Tristani-Firouzi M, Brueckner M, Lifton RP, Goldmuntz E, Gelb BD, Chung WK, Seidman CE, Seidman JG, Shen Y. EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. PMID: 32349777 *Genome Med* (2020)

40. Edwards JJ, Rouillard AD, Fernandez NF, Wang Z, Lachmann A, Shankaran SS, Bisgrove BW, Demarest B, Turan N, Srivastava D, Bernstein D, Deanfield J, Giardini A, Porter G, Kim R, Roberts AE, Newburger JW, Goldmuntz E, Brueckner M, Lifton RP, Seidman CE, Chung WK, Tristani-Firouzi M, Yost HJ, Ma'ayan A, Gelb BD. Systems Analysis Implicates WAVE2 Complex in the Pathogenesis of Developmental Left-Sided Obstructive Heart Defects. PMID: 32368696 *JACC Basic Transl Sci.* (2020)

41. Genomic analyses implicate noncoding de novo variants in congenital heart disease. Richter F, Morton SU, Kim SW, Kitaygorodsky A, Wasson LK, Chen KM, Zhou J, Qi H, Patel N, DePalma SR, Parfenov M, Homsy J, Gorham JM, Manheimer KB, Velinder M, Farrell A, Marth G, Schadt EE, Kaltman JR, Newburger JW, Giardini A, Goldmuntz E, Brueckner M, Kim R, Porter GA Jr, Bernstein D, Chung WK, Srivastava D, Tristani-Firouzi M, Troyanskaya OG, Dickel DE, Shen Y, Seidman JG, Seidman CE, Gelb BD. PMID: 32601476 *Nat Genet*. (2020)

2. Reviews:

42. **M.Brueckner**, P.D'Eustachio, J.McGrath and A.L.Horwich. Establishment of Left-Right Asymmetry: Genetically Distinct Steps are Involved. In: Biological Asymmetry and Handedness, PMID: 1802643 *Ciba Foundation Symposium*, #162 :202-218 (1991).

43. **M.Brueckner**. The Genetics of Congenital Heart Disease. *Current Opinion in Cardiology,* 8:91-97 (1993).

44. Z.Bulbul, D.Rosenthal, and **M.Brueckner**. Genetic Aspects of Heart Disease in the Newborn. PMID: 8327904 *Seminars in Perinatology*, 17:61-75 (1993).

45. A. Horwich, **M. Brueckner**. Left, right and without a cue. PMID: 8298636 *Nature Genetics* 5(4):321-2 (1993)

46. P.N.Bowers, **M.Brueckner** and H.J.Yost. The Genetics of Left-Right Development and Heterotaxia. PMID: 9090782 *Seminars in Perinatology*, 20:577-588 (1996).

47. D.M.Supp, **M.Brueckner**, and S.S.Potter. Handed Asymmetry in the Mouse: Understanding how things go right (or left) by studying how they go wrong. PMID: 9572117 *Seminars in Cell and Developmental Biology*, 9:77-87 (1998).

48. D.M.Supp, S.S.Potter, J.McGrath, and **M.Brueckner**. Motor proteins and the development of left-right asymmetry. In: Etiology and Morphogenesis of Congenital Heart Disease, Clark and Takao, Eds. Futura Press (1999).

49. D.M.Supp, S.S.Potter, and **M.Brueckner**. Molecular motors: The driving force behind left-right development. PMID: 10652513 *Trends in Cell Biology*, (2000).

50. H.S.Schneider and **M.Brueckner**. Of mice and men: dissecting the genetic pathway that controls LR asymmetry in mice and humans. PMID: 11376437 *Am.J. Med. Gen.*, 97:258-70 (2001)

51. **M.Brueckner**. Cilia propel the embryo in the right direction. PMID: 11471157 *Am. J. Med. Genet.*, 101:339-44 (2001)

52. J.McGrath, and **M.Brueckner**. Cilia are at the heart of vertebrate left-right asymmetry. PMID: 12888012 *Curr. Opin. Genet. Dev.*, 13:385-92 (2003)

53. **M.Brueckner**. Heterotaxia, Congenital Heart Disease and Primary Ciliary Dyskinesia. PMID: 17548739 *Circulation*, 115:2793-5 (2007)

54. B.Basu, **M.Brueckner**. Cilia: multifunctional organelles at the center of vertebrate left-right asymmetry. PMID: 19147005 *Curr.Topics in Dev. Biol.*, 85:151-174 (2008).

55. M.T. Boskovski, **M.Brueckner**. The developmental biology and genetics underlying human heterotaxy, in: Hemodynamics and Cardiology, Neonatology Questions and Controversies, 2e, Elsevier Pub. 2011

56. S. Yuan, S. Zaidi, and **M. Brueckner**, Congenital heart disease, emerging themes linking genetics and development. PMID: 23790954 *Curr Op Gen Dev*, 23:352-359(2013)

57. S. Zaidi, **M. Brueckner**, Genetics and Genomics of Congenital Heart Disease. PMID 28302740 *Circulation Research* (2017)

58. M.A. Simmons, M. Brueckner, The genetics of congenital heart disease… understanding and improving long-term outcomes in congenital heart disease: a review for the general cardiologist and primary care physician. PMID: 28872494 *Current Opinion in Pediatrics* (2017)

3. Invited Editorials and Commentaries:

59. **M.Brueckner**. What comes first: The structure or the egg? Ross Granville Harrison on the origin of embryonic polarity. PMID: 15229864 *JEZ*, 301A(7):549-550 (2004)

60. B.Basu and **M.Brueckner**. Fibroblast "cilia growth" factor in the development of left-right asymmetry. PMID: 19386257 *DevCell,* 16:489-90 (2009)

61. **M.Brueckner**. Impact of genetic diagnosis on clinical management of patients with CHD: cilia point the way. PMID: 22499951 *Circulation,* 125(18)2178-2180 (2012)

62. S.Yuan and **M.Brueckner**. Left-Right Asymetry: Myosin 1D at the Center. PMID: 29738734 Current Biology, 28(9):R567-R569 (2018)

Grant Support:

1. Active

R35HL145249
M. Brueckner, PI
Direct costs $447,000/year (50% effort, M. Brueckner)
12/01/2018 – 11/30/2025
NIH-NHLBI
Cilia in heart development

2UO1HL098162-12
M. Brueckner, PI (MPI)
Direct costs $275,000.00
08/01/2020 – 07/31/2025
NHLBI/NIH

3. Completed (past five years)

Congenital Heart Disease Genetics and Clinical Outcomes

2UM1HL098162-07
M. Brueckner, PI (MPI)
Direct costs $291,000 year 1 (25% effort, M. Brueckner)
NIH
Genetics and genomics of congenital heart disease and associated neurodevelopmental abnormalities

1R01HL125885-01A1
M. Brueckner, PI, (MPI)
Direct costs $361,634 year 1 (20% effort, M. Brueckner)
NIH
Intraciliary calcium signaling directs cardiac left-right asymmetry

M. Brueckner, PI
Direct costs $70,000/year (1% effort, M.Brueckner)
ATS/PCD Foundation
Congenital Heart Disease Genes as an approach to identify novel PCD genes"

1R01HL124402-01A1
M.Brueckner, (MPI)

04/01/15-03/31/19
direct costs $370,492 year 1 (20% effort, M.Brueckner)
NIH/NHLBI
New mechanisms of heterotaxy and congenital heart disease: nucleoporins at cilia

1R21HL120783-01
M.K.Khokha, PI
03/10/2014 – 02/29/2016
direct costs $137,460 year 1
NIH/NHLBI
Cardiac phenotyping of CHD candidate genes in *Xenopus* 1 UO1

1HL098162-05
M. Brueckner, PI (MPI)
09/01/09-08/31/15
direct costs $469,219 year 5, (25% effort M. Brueckner; total direct costs $2,192,068)
NIH/NHLBI
Genetic determinants of human heterotaxy and aortic arch malformation

PCGC Steering committee chair
NIH/New England Research Institute
06-01-14 – 07-31-15
5% effort

1R01 HL093280-01A1
M. Brueckner, PI
04/01/09 – 03/31/14
direct costs $250,000/year, (total direct costs $1,250,000)
NIH/NHLBI
Cilia in cardiac morphogenesis