Genetic Disorders of Mucociliary Clearance

Book Chapters

1. Knowles MR, Morillas HN, Leigh M, Zariwala M. Primary ciliary dyskinesia. In: Rounds S, ed. *Molecular Basis of Lung Disease, Insights from Rare Lung Disorders*. Totowa, NJ: Humana Press; 2010.

Abstracts Presented at Conferences

1. Olin J, Sagel S, Knowles M. Success (vs diagnostic yield?) of nasal scrape biopsies in diagnosing primary ciliary dyskinesia. Paper presented at: American Thoracic Society Conference; April, 2009; San Diego, CA.
2. Lie H, Zariwala M, Puffenberger E, Strauss K, Bowcock A, Carson J, Leigh M, Knowles M, Ferkol T. The genetic basis of primary ciliary dyskinesia in Amish communities. Paper presented at: American Thoracic Society Conference; April, 2009; San Diego, CA.
3. Dell S, Dupruis A, Knowles M, Quittner A, Leigh M. Impaired Health-Related, Quality of Life (HRQQL) in Children with Primary Ciliary Dyskinesia (PCD). Paper presented at: American Thoracic Society conference 2009; April, 2009; San Diego, CA.
4. Chawla K, Hazucha M, Dell S, Ferkol T, Sagel S, Rosenfeld M, Baker B, David S, Knowles M, Leigh M. A Multi-Center, Longitudinal Study of Nasal Nitric Oxide in Children with Primary Ciliary Dyskinesia. Paper presented at: American Thoracic Society2010; New Orleans, LA.
5. Radhakrishnan D, Leigh M, Knowles M, Carson J, Metijan H, Cutz E, Wilkes D, Dell S. A comparison of two methods to detect classic ciliary ultrastructural defects in a population of children and suspected primary ciliary dyskinesia. Paper presented at: American Thoracic Society2010; New Orleans, LA
6. Kureshi S, Nakhleh N, Seton M, Francis R, Chatterjee B, Sami I, Kuehl K, Olivier K, Jonas R, Tian X, Leigh M, Knowles M, Leatherbury L, Lo C. Nasal nitric oxide & ciliary function in patients with non-heterotaxy congenital heart disease. Paper presented at: American Thoracic Society2010; New Orleans, LA.
7. LaVange L, Stewart D, Thomashow B, Olivier K, Knowles M, Daley C, Barker A. The Bronchiectasis Research Registry: a resource for collaborative research in non-cystic fibrosis bronchiectasis. Paper presented at: American Thoracic Society2010; New Orleans, LA.
8. Olivier K, O'Connell M, Holland S, Knowles M. Mucosal defense abnormalities in idiopathic bronchiectasis associated with nontuberculous mycobacteria. Paper presented at: American Thoracic Society2010; New Orleans, LA.
9. Shapiro A, Davis S, Olivier K, Ferkol T, Dell S, Sagel S, Rosenfeld M, Milla C, Atkinson J, Knowles M, Leigh M. Clinical symptoms associated with primary ciliary dyskinesia-results of a multi-centered study. Paper presented at: American Thoracic Society; 2010, 2010; New Orleans, LA.
10. Pittman J. Bronchodilator responsiveness by infant pulmonary function testing. Paper presented at: American Thoracic Society International Conference; May, 2010; New Orleans.
11. Zariwala MA, Leigh M, Ostrowski LE, Davis SD, Berg J, Huang L, Yin W, Carson JL, Hazucha MJ, Turner EH, MacKenzie A, Bamshad M, Nickerson DA, Schendure J, Knowles M, Genetic Disorders

of Mucociliary Clearance Consortium. Exome sequencing to identify genetic causes of primary ciliary dyskinesia with outer dynein arms defects. (Abstract 1071T). Paper presented at: 12th International Congress of Human Genetics/61st Annual Meeting of The American Society of Human Genetics; October 13, 2011; Montreal, Canada.

12. Leigh MW, Shapiro AJ, Pittman JE, Davis SD, Lee H, Krischer J, Ferkol TW, Atkinson JJ, Sagel SD, Rosenfield M, Dell SD, Milla C, Olivier KN, Knowles MR. Definition of clinical criteria for diagnosis of primary ciliary dyskinesia. Paper presented at: ATS (American Thoracic Society) 2012; May 18-23, 2012; San Francisco.

Journal Articles

1. Fliegauf M, Olbrich H, Horvath J, Wildhaber JH, Zariwala MA, Kennedy M, Knowles MR, Omran H. Mislocalization of DNAH5 and DNAH9 in respiratory cells from patients with primary ciliary dyskinesia. *Am. J. Respir. Crit. Care Med.* Jun 15 2005;171(12):1343-1349. PMID: 15750039, PMCID: PMC2718478

2. Kennedy MP, Omran H, Leigh MW, Dell S, Morgan L, Molina PL, Robinson BV, Minnix SL, Olbrich H, Severin T, Ahrens P, Lange L, Morillas HN, Noone PG, Zariwala MA, Knowles MR. Congenital heart disease and other heterotaxic defects in a large cohort of patients with primary ciliary dyskinesia. *Circulation.* Jun 5 2007;115(22):2814-2821. PMID: 17515466

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4. Leigh MW, Zariwala MA, Knowles MR. Primary ciliary dyskinesia: improving the diagnostic approach. *Curr. Opin. Pediatr.* Jun 2009;21(3):320-325. PMID: 19300264, PMCID: PMC3665363

5. Leigh MW, Pittman JE, Carson JL, Ferkol TW, Dell SD, Davis SD, Knowles MR, Zariwala MA. Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome. *Genet. Med.* Jul 2009;11(7):473-487. PMID: 19606528, PMCID: PMC3739704

6. Loges NT, Olbrich H, Becker-Heck A, Haffner K, Heer A, Reinhard C, Schmidts M, Kispert A, Zariwala MA, Leigh MW, Knowles MR, Zentgraf H, Seithe H, Nurnberg G, Nurnberg P, Reinhardt R, Omran H. Deletions and point mutations of LRRC50 cause primary ciliary dyskinesia due to dynein arm defects. *Am. J. Hum. Genet.* Dec 2009;85(6):883-889. PMID: 19944400, PMCID: PMC2795801

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9. Berg JS, Evans JP, Leigh MW, Omran H, Bizon C, Mane K, Knowles MR, Weck KE, Zariwala MA. Next generation massively parallel sequencing of targeted exomes to identify genetic mutations in primary ciliary dyskinesia: implications for application to clinical testing. *Genet. Med.* Mar 2011;13(3):218-229. PMID: 21270641, PMCID: PMC3755008

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11. Leigh MW, O'Callaghan C, Knowles MR. The challenges of diagnosing primary ciliary dyskinesia. *Proc. Am. Thorac. Soc.* Sep 2011;8(5):434-437. PMID: 21926395, PMCID: PMC3209576

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19. Leigh M, Chawla KK, Baker BR, Hazucha MJ, Brown DE, LaVange L, Horton BJ, Qaqish BF, Carson JL, Davis SD, Dell SD, Ferkol TW, Atkinson JJ, Olivier KN, Sagel SD, Rosenfeld C, Milla C, Zariwala MA, Knowles M. For the Genetic Diseases of Mucociliary Clearance Consortium. Standardization of nasal nitric oxide as screening test for primary ciliary dyskinesia. *Am. J. Respir. Crit. Care Med.* 2012;185. PMID: 21926395, PMCID: PMC3209576

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Knowles MR, Brody SL, Dutcher SK, Ferkol TW. Whole-exome capture and sequencing identifies HEATR2 mutation as a cause of primary ciliary dyskinesia. *Am. J. Hum. Genet.* Oct 5 2012;91(4):685-693. PMID: 23040496, PMCID: PMC3484505

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