

## Rare Lung Diseases Consortium

### Book Chapters

1. Krischer JP. Clinical trials for rare lung diseases. In: McCormack FX, Panos RJ, Trapnell BC, eds. *Molecular Basis of Pulmonary Disease*. 1st ed: Springer; 2010.

### Journal Articles

1. Whitsett JA, Wert SE, Trapnell BC. Genetic disorders influencing lung formation and function at birth. *Hum. Mol. Genet.* Oct 1 2004;13 Spec No 2:R207-215. PMID: 15358727
2. Uchida K, Beck DC, Yamamoto T, Berclaz PY, Abe S, Staudt MK, Carey BC, Filippi MD, Wert SE, Denson LA, Puchalski JT, Hauck DM, Trapnell BC. GM-CSF autoantibodies and neutrophil dysfunction in pulmonary alveolar proteinosis. *N. Engl. J. Med.* Feb 8 2007;356(6):567-579. PMID: 17287477
3. Juvet SC, McCormack FX, Kwiatkowski DJ, Downey GP. Molecular pathogenesis of lymphangioliomyomatosis: lessons learned from orphans. *Am. J. Respir. Cell Mol. Biol.* Apr 2007;36(4):398-408. PMID: 17099139, PMCID: PMC2176113
4. Deterding R. Evaluating infants and children with interstitial lung disease. *Semin. Respir. Crit. Care Med.* Jun 2007;28(3):333-341. PMID: 17562503
5. Trotta BM, Stolin AV, Williams MB, Gay SB, Brody AS, Altes TA. Characterization of the relation between CT technical parameters and accuracy of quantification of lung attenuation on quantitative chest CT. *AJR. American Journal of Roentgenology.* Jun 2007;188(6):1683-1690. PMID: 17515394
6. Deutsch GH, Young LR, Deterding RR, Fan LL, Dell SD, Bean JA, Brody AS, Nogee LM, Trapnell BC, Langston C, Albright EA, Askin FB, Baker P, Chou PM, Cool CM, Coventry SC, Cutz E, Davis MM, Dishop MK, Galambos C, Patterson K, Travis WD, Wert SE, White FV. Diffuse lung disease in young children: application of a novel classification scheme. *Am. J. Respir. Crit. Care Med.* Dec 1 2007;176(11):1120-1128. PMID: 17885266, PMCID: PMC2176101
7. Young LR, Inoue Y, McCormack FX. Diagnostic potential of serum VEGF-D for lymphangioliomyomatosis. *N. Engl. J. Med.* Jan 10 2008;358(2):199-200. PMID: 18184970, PMCID: PMC3804557
8. Inoue Y, Trapnell BC, Tazawa R, Arai T, Takada T, Hizawa N, Kasahara Y, Tatsumi K, Hojo M, Ichiwata T, Tanaka N, Yamaguchi E, Eda R, Oishi K, Tsuchihashi Y, Kaneko C, Nukiwa T, Sakatani M, Krischer JP, Nakata K. Characteristics of a large cohort of patients with autoimmune pulmonary alveolar proteinosis in Japan. *Am. J. Respir. Crit. Care Med.* Apr 1 2008;177(7):752-762. PMID: 18202348, PMCID: PMC2720118
9. Suzuki T, Sakagami T, Rubin BK, Nogee LM, Wood RE, Zimmerman SL, Smolarek T, Dishop MK, Wert SE, Whitsett JA, Grabowski G, Carey BC, Stevens C, van der Loo JC, Trapnell BC. Familial pulmonary alveolar proteinosis caused by mutations in CSF2RA. *J. Exp. Med.* Nov 24 2008;205(12):2703-2710. PMID: 18955570, PMCID: PMC2585845
10. Schmithorst VJ, Altes TA, Young LR, Franz DN, Bissler JJ, McCormack FX, Dardzinski BJ, Brody AS. Automated algorithm for quantifying the extent of cystic change on volumetric chest CT: initial

results in Lymphangiomyomatosis. *AJR. American Journal of Roentgenology*. Apr 2009;192(4):1037-1044. PMID: 19304711

11. Trapnell BC, Carey BC, Uchida K, Suzuki T. Pulmonary alveolar proteinosis, a primary immunodeficiency of impaired GM-CSF stimulation of macrophages. *Curr. Opin. Immunol.* Oct 2009;21(5):514-521. PMID: 19796925, PMCID: PMC2779868
12. Tazawa R, Trapnell BC, Inoue Y, Arai T, Takada T, Nasuhara Y, Hizawa N, Kasahara Y, Tatsumi K, Hojo M, Ishii H, Yokoba M, Tanaka N, Yamaguchi E, Eda R, Tsuchihashi Y, Morimoto K, Akira M, Terada M, Otsuka J, Ebina M, Kaneko C, Nukiwa T, Krischer JP, Akazawa K, Nakata K. Inhaled granulocyte/macrophage-colony stimulating factor as therapy for pulmonary alveolar proteinosis. *Am. J. Respir. Crit. Care Med.* Jun 15 2010;181(12):1345-1354. PMID: 20167854, PMCID: PMC2894410
13. Ingelfinger JR, Drazen JM. Patient organizations and research on rare diseases. *N. Engl. J. Med.* Apr 28 2011;364(17):1670-1671. PMID: 21410388
14. McCormack FX, Inoue Y, Moss J, et al. Efficacy and safety of sirolimus in lymphangiomyomatosis. *N. Engl. J. Med.* Apr 28 2011;364(17):1595-1606. PMID: 21410393, PMCID: PMC3118601
15. Rosenbaum L. How Much Would You Give to Save a Dying Bird? Patient Advocacy and Biomedical Research. *N. Engl. J. Med.* 2012;367(18):1755-1759. PMID: 23113489
16. Suzuki T, Arumugam P, Sakagami T, et al. Pulmonary macrophage transplantation therapy. *Nature.* Oct 23 2014;514(7523):450-454. PMID: 25274301, PMCID: PMC4236859
17. Trapnell BC. A lymphocyte-mediated cause of secondary PAP. *Blood.* Jan 8 2015;125(2):215-216. PMID: 25573969
18. Fan LL, Dishop MK, Galambos C, et al. Diffuse Lung Disease in Biopsied Children 2 to 18 Years of Age. Application of the chILD Classification Scheme. *Annals of the American Thoracic Society.* Oct 2015;12(10):1498-1505. PMID: 26291470, PMCID: PMC4627419
19. Saito A, Nikolaidis NM, Amlal H, et al. Modeling pulmonary alveolar microlithiasis by epithelial deletion of the Npt2b sodium phosphate cotransporter reveals putative biomarkers and strategies for treatment. *Sci Transl Med.* Nov 11 2015;7(313):313ra181. PMID: 26560359, PMCID: PMC4764987
20. Argula RG, Kokosi M, Lo P, et al. A Novel Quantitative Computed Tomographic Analysis Suggests How Sirolimus Stabilizes Progressive Air Trapping in Lymphangiomyomatosis. *Annals of the American Thoracic Society.* 2016;13(3):342-349. PMID: 26799509
21. Campo I, Luisetti M, Griese M, et al. Whole lung lavage therapy for pulmonary alveolar proteinosis: a global survey of current practices and procedures. *Orphanet J Rare Dis.* 2016;11(1):115. PMID: 27577926, PMCID: PMC5006612
22. El-Chemaly S, Young LR. Hermansky-Pudlak Syndrome. *Clin Chest Med.* 2016;37(3):505-511. PMID: 27514596, PMCID: PMC4987498
23. McCormack FX, Gupta N, Finlay GR, et al. Official American Thoracic Society/Japanese Respiratory Society Clinical Practice Guidelines: Lymphangiomyomatosis Diagnosis and Management. *Am J Respir Crit Care Med.* 2016;194(6):748-761. PMID: 27628078

24. O'Brien KJ, Lozier J, Cullinane AR, et al. Identification of a novel mutation in HPS6 in a patient with hemophilia B and oculocutaneous albinism. *Mol Genet Metab.* 2016;119(3):284-287. PMID: 27641950, PMCID: PMC5083180
25. Sahin M, Henske EP, Manning BD, et al. Advances and Future Directions for Tuberous Sclerosis Complex Research: Recommendations From the 2015 Strategic Planning Conference. *Pediatr. Neurol.* 2016;60:1-12. PMID: 27267556, PMCID: PMC4921275
26. Vicary GW, Vergne Y, Santiago-Cornier A, Young LR, Roman J. Pulmonary Fibrosis in Hermansky-Pudlak Syndrome. *Annals of the American Thoracic Society.* 2016;13(10):1839-1846. PMID: 27529121, PMCID: PMC5466158
27. Young LR, Trapnell BC, Mandl KD, Swarr DT, Wambach JA, Blaisdell CJ. Accelerating Scientific Advancement for Pediatric Rare Lung Disease Research. Report from a National Institutes of Health-NHLBI Workshop, September 3 and 4, 2015. *Annals of the American Thoracic Society.* 2016;13(12):385-393. PMID: 27925785, PMCID: PMC5291498
28. Cooley J, Lee YCG, Gupta N. Spontaneous pneumothorax in diffuse cystic lung diseases. *Curr Opin Pulm Med.* 2017;23(4):323-333. PMID: 28590337
29. Gaskill CF, Carrier EJ, Kropski JA, et al. Disruption of lineage specification in adult pulmonary mesenchymal progenitor cells promotes microvascular dysfunction. *J Clin Invest.* 2017;127(6):2262-2276. PMID: 28463231
30. Gupta N, Langenderfer D, McCormack FX, Schauer DP, Eckman MH. Chest Computed Tomographic Image Screening for Cystic Lung Diseases in Patients with Spontaneous Pneumothorax Is Cost Effective. *Annals of the American Thoracic Society.* 2017;14(1):17-25. PMID: 27737563, PMCID: PMC5461992
31. Kropski JA, Reiss S, Markin C, et al. Rare Genetic Variants in PARN Are Associated with Pulmonary Fibrosis in Families. *Am J Respir Crit Care Med.* 2017;196(11):1481-1484. PMID: 28414520, PMCID: PMC5736978
32. Kropski JA, Young LR, Cogan JD, et al. Genetic Evaluation and Testing of Patients and Families with Idiopathic Pulmonary Fibrosis. *Am J Respir Crit Care Med.* 2017;195(11):1423-1428. PMID: 27786550, PMCID: PMC5470751
33. Liu H, Jakubzick C, Osterburg AR, et al. Dendritic Cell Trafficking and Function in Rare Lung Diseases. *Am J Respir Cell Mol Biol.* 2017;57(4):393-402. PMID: 28586276, PMCID: PMC5650088
34. McCormack FX, Gupta N, Finlay GR, et al. Reply: The ATS/JRS Guidelines on Lymphangioliomyomatosis: Filling in the Gaps. *Am J Respir Crit Care Med.* 2017;196(5):660-661. PMID: 28288285
35. Sivley RM, Sheehan JH, Kropski JA, et al. Three-dimensional spatial analysis of missense variants in RTEL1 identifies pathogenic variants in patients with Familial Interstitial Pneumonia. *BMC Bioinformatics.* 2018;19(1):18. PMID: 29361909