Scott M. Blackman, M.D., Ph.D.
CURRICULUM VITAE

1/23/2015

Current Appointment:

2009-present Assistant Professor, Department of Pediatrics,
Johns Hopkins University School of Medicine

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EDUCATION AND TRAINING:

1992	A.B.	Princeton University (Chemistry, *cum laude*)
1998	Ph.D.	Vanderbilt University School of Medicine (Molecular Physiology and Biophysics)
2000	M.D.	Vanderbilt University School of Medicine
2001-2004		Internship and Residency, Pediatrics, Johns Hopkins University School of Medicine
2004-2007		Fellowship, Pediatric Endocrinology, Johns Hopkins University School of Medicine

PROFESSIONAL EXPERIENCE:

1991	Research Assistant NIH-NIAID, Laboratory for Clinical Investigation P.I.: S. Strauss M.D.
1992-2000	Medical Scientist Training Program Vanderbilt University School of Medicine, Nashville, TN Dissertation advisor: Albert H. Beth, Ph.D.
2000-2001	Postdoctoral Research Associate Brown University School of Medicine, Providence, RI P.I.: Dale Mierke, M.D.
2001-2004	Pediatric Intern and Resident Johns Hopkins University School of Medicine
2004-2007	Pediatric Endocrine Fellow Johns Hopkins University School of Medicine Research mentor: Garry R. Cutting, M.D.
2007-2009	Instructor, Division of Pediatric Endocrinology Department of Pediatrics Johns Hopkins University School of Medicine
2009-present	Assistant Professor, Division of Pediatric Endocrinology Department of Pediatrics Johns Hopkins University School of Medicine

RESEARCH ACTIVITIES

Publications:

1. Niswender, K. D., **Blackman, S. M.**, Rohde, L., Magnuson, M. A., and Piston, D. W. Quantitative imaging of green fluorescent protein in cultured cells: comparison of microscopic techniques, use in fusion proteins and detection limits. *J. Microsc.* 1995; 180: 109-116. (PMID: 8537958)
2. **Blackman, S. M.**, Cobb, C. E., Beth, A. H., and Piston, D. W. The orientation of eosin-5-maleimide on human erythrocyte band 3measured by fluorescence polarization microscopy. *Biophys. J.* 1996; 71: 194-208. (PMID: 8804603, [full text]).
3. **Blackman, S. M.**, Piston, D. W., and Beth, A. H. Oligomeric state of human erythrocyte band 3 measured by fluorescence resonance energy homotransfer. *Biophys. J.* 1998; **75:** 1117-1130. (PMID: 9675213, [full text])
4. **Blackman, S. M.**, Hustedt, E. J., Cobb, C. E., and Beth, A. H. Flexibility of the cytoplasmic domain of the anion exchange protein, band 3, in human erythrocytes. *Biophys. J.* 2001; 81: 3363-3376. (PMID: 11720999, [full text])
5. Piserchio, A., Pellegrini, M., Mehta, S., **Blackman, S. M.**, Garcia, E. P., Marshall, J., and Mierke, D. F. The PDZ1 Domain of SAP90: Characterization of Structure and Binding. *J. Biol. Chem.* 2002; 277: 6967-6973. (PMID: 11744724, [full text])
6. **Blackman S. M.**, Deering-Brose R., McWilliams R., Naughton K., Coleman B., Lai T., Algire M., Beck S., Hoover-Fong J., Hamosh A., Fallin M. D., West, K., Arking, D. E., Chakravarti, A., and Cutting G. R.. Relative Contribution of Genetic and Non-genetic Modifiers to Intestinal Obstruction in Cystic Fibrosis. *Gastroenterology* 2006; 231(4): 1030-1039. (PMID: 17030173, [full text]).
7. Vanscoy, L. L., **Blackman, S. M.**, Collaco, J. M., Bowers, A., Lai, T., Naughton, K., Algire, M., McWilliams, R., Beck, S., Hoover-Fong, J., Hamosh, A., Cutler, D., Cutting, G.R. Heritability of Lung Disease Severity in Cystic Fibrosis. *Am. J. Respir. Crit. Care Med.* 2007; **175**(10): 1036-43 (PMID: 17332481, [full text]).
8. Collaco, J.M., Vanscoy, L., Bremer, L., McDougal, K., **Blackman S. M.**, Bowers, A., Naughton K., Jennings, J., Ellen, J., and Cutting G. R.. Interactions between Secondhand Smoke and Genes that Affect Cystic Fibrosis Lung Disease. *JAMA*. 2008; 299(4): 417-424 (PMID: 18230779, [full text]).
9. Bremer L.A., **Blackman S. M.**, Vanscoy L.L., McDougal K.E., Bowers A., Naughton K.M., Cutler D.J., Cutting G.R.. Interaction between a novel TGFB1 haplotype and CFTR genotype is associated with improved lung function in cystic fibrosis. *Hum Mol Genet.* 2008 Jul 15;17(14):2228-37. Epub 2008 Apr 17. (PMID: 18424453, [full text])
10. **Blackman S. M.**, Hsu S., Vanscoy L. L., Collaco J.M., Ritter S.E., Naughton K., Cutting G.R. Genetic modifiers play a substantial role in diabetes complicating cystic fibrosis. *J Clin Endocrinol Metab.* 2009 Apr;94(4):1302-9. Epub 2009 Jan 6. (PMID: 19126627, [full text])
11. **Blackman S.M.**, Hsu S., Ritter S.E., Naughton K.M., Wright F.A., Drumm M.L., Knowles M.R., Cutting G.R. A susceptibility gene for type 2 diabetes confers substantial risk for diabetes complicating cystic fibrosis. *Diabetologia*. 2009 Sep;52(9):1858-65. Epub 2009 Jul 8. (PMID: 19585101, [full text])

12. Dorfman R, Li W, Sun L, Lin F, Wang Y, Sandford A, Paré PD, McKay K, Kayserova H, Piskackova T, Macek M, Czerska K, Sands D, Tiddens H, Margarit S, Repetto G, Sontag MK, Accurso FJ, **Blackman S**, Cutting GR, Tsui LC, Corey M, Durie P, Zielenski J, Strug LJ. Modifier gene study of meconium ileus in cystic fibrosis: statistical considerations and gene mapping results. *Hum Genet*. 2009 Dec;126(6):763-78. doi: 10.1007/s00439-009-0724-8. PubMed PMID: 19662435; PubMed Central PMCID: PMC2888886.
13. McDougal KE, Green DM, Vanscoy LL, Fallin MD, Grow M, Cheng S, **Blackman SM**, Collaco JM, Henderson LB, Naughton K, Cutting GR. Use of a modeling framework to evaluate the effect of a modifier gene (MBL2) on variation in cystic fibrosis. *Eur J Hum Genet.* 2010 Jun;18(6):680-4. Epub 2010 Jan 13. (PMID: 20068595, [full text]).
14. Collaco JM, **Blackman SM**, McGready J, Naughton KM, Cutting GR. Quantification of the Relative Contribution of Environmental and Genetic Factors to Variation in Cystic Fibrosis Lung Function. *J Pediatr*. Epub 2010 Jun 25. (PMID: 20580019, [full text]).
15. Li W, Sun L, Corey M, Zou F, Lee S, Cojocaru A, Taylor C, **Blackman S**, Stephenson A, Sandford A, Dorfman R, Drumm M, Cutting G, Knowles M, Durie P, Wright F, Strug L. Understanding the population structure of North American patients with cystic fibrosis. *Clin Genet.* Epub 2010 Jul 2. (PMID: 20681990, [full text]).
16. Green DM, McDougal KE, **Blackman SM**, Sosnay PR, Henderson LB, Naughton KM, Collaco JM, Cutting GR. Mutations that permit residual CFTR function delay acquisition of multiple respiratory pathogens in CF patients. *Respir Res*. 2010 Oct 8;11:140. (PMID: 20932301, [full text]).
17. Taylor C, Commander CW, Collaco JM, Strug LJ, Li W, Wright FA, Webel AD, Pace RG, Stonebraker JR, Naughton K, Dorfman R, Sandford A, **Blackman SM**, Berthiaume Y, Paré P, Drumm ML, Zielenski J, Durie P, Cutting GR, Knowles MR, Corey M. A novel lung disease phenotype adjusted for mortality attrition for cystic fibrosis Genetic modifier studies. *Pediatr Pulmonol*. Epub 2011 Apr 1. (PMID: 21462361 [full text]).
18. Wright FA, Strug LJ, Doshi VK, Commander CW, **Blackman SM**, Sun L, Berthiaume Y, Cutler D, Cojocaru A, Collaco JM, Corey M, Dorfman R, Goddard K, Green D, Kent JW Jr, Lange EM, Lee S, Li W, Luo J, Mayhew GM, Naughton KM, Pace RG, Paré P, Rommens JM, Sandford A, Stonebraker JR, Sun W, Taylor C, Vanscoy LL, Zou F, Blangero J, Zielenski J, O'Neal WK, Drumm ML, Durie PR, Knowles MR, Cutting GR. Genome-wide association and linkage identify modifier loci of lung disease severity in cystic fibrosis at 11p13 and 20q13.2. *Nat Genet*. 2011 Jun;43(6):539-46. Epub 2011 May 22. (PMID: 21602797, [full text]).
19. Henderson LB, Doshi VK, **Blackman SM**, Naughton KM, Pace RG, Moskovitz J, Knowles MR, Durie PR, Drumm ML, Cutting GR. Variation in MSRA modifies risk of neonatal intestinal obstruction in cystic fibrosis. *PLoS Genet.* 2012 Mar;8(3):e1002580. Epub 2012 Mar 15. PubMed PMID: 22438829; PubMed Central PMCID: PMC3305406.
20. Green DM, Collaco JM, McDougal KE, Naughton KM, **Blackman SM**, Cutting GR. Heritability of respiratory infection with pseudomonas aeruginosa in cystic fibrosis. *J. Pediatr.* 2012 Aug;161(2):290-5.e1. Epub 2012 Feb 23. PubMed PMID: 22364820.
21. Sun L, Rommens JM, Corvol H, Li W, Li X, Chiang TA, Lin F, Dorfman R, Busson PF, Parekh RV, Zelenika D, **Blackman SM**, Corey M, Doshi VK, Henderson L, Naughton KM, O'Neal WK, Pace RG, Stonebraker JR, Wood SD, Wright FA, Zielenski J, Clement A, Drumm ML, Boëlle PY, Cutting GR, Knowles MR, Durie PR, Strug LJ. Multiple apical plasma membrane constituents are associated with susceptibility to meconium ileus in individuals with cystic fibrosis. *Nat Genet*. 2012 May;44(5):562-9. PubMed PMID: 22466613; PubMed Central PMCID: PMC3371103.

22. Bradley GM*, **Blackman SM***, Watson CP, Doshi VK, Cutting GR. Genetic modifiers of nutritional status in cystic fibrosis. Am J Clin Nutr. 2012 Dec;96(6):1299-308. doi: 10.3945/ajcn.112.043406. Epub 2012 Nov 7. PubMed PMID: 23134884,); PubMed Central PMCID: PMC3497925. (*equal contributors).
23. **Blackman SM**, Commander CW, Watson C, Arcara KM, Strug LJ, Stonebraker JR, Wright FA, Rommens JM, Sun L, Pace RG, Norris SA, Durie PR, Drumm ML, Knowles MR, Cutting GR. Genetic modifiers of cystic fibrosis-related diabetes. *Diabetes*. 2013 Oct;62(10):3627-35. doi: 10.2337/db13-0510. Epub 2013 May 13. PubMed PMID: 23670970; PubMed Central PMCID: PMC3781476. [**Published with commentary**: Meyre D, Pare G. Genetic dissection of diabetes: facing the giant. *Diabetes.* 2013 Oct;62(10):3338-40. doi: 10.2337/db13-1154. PubMed PMID: 24065794; PubMed Central PMCID: PMC3781438.]
24. **Blackman SM,** Raghinaru D, Adi S, Simmons JH, Ebner-Lyon L, Chase HP, Tamborlane WV, Schatz DA, Block JM, Litton JC, Raman V, Foster NC, Kollman CR, DuBose SN, Miller KM, Beck RW, DiMeglio LA. Insulin pump use in young children in the T1D Exchange clinic registry is associated with lower hemoglobin A1c levels than injection therapy. *Pediatr Diabetes.* 2014 Dec;15(8):564-72. doi: 10.1111/pedi.12121. Epub 2014 Feb 4. PubMed PMID: 24494980.
25. Morrow CB, Raraigh KS, Green DM, **Blackman SM**, Cutting GR, Collaco JM. Cat and dog exposure and respiratory morbidities in cystic fibrosis. *J Pediatr*. 2014 Oct;165(4):830-5.e2. doi: 10.1016/j.jpeds.2014.05.046. Epub 2014 Jul 12. PubMed PMID: 25027361; PubMed Central PMCID: PMC4177281.
26. Collaco JM, **Blackman SM,** Raraigh KS, Morrow CB, Cutting GR, Paranjape SM. Self-reported exercise and longitudinal outcomes in cystic fibrosis: a retrospective cohort study. *BMC Pulm Med*. 2014 Oct 6;14:159. doi: 10.1186/1471-2466-14-159. PubMed PMID: 25287419; PubMed Central PMCID: PMC4195986.
27. Lewis C, **Blackman SM**, Nelson A, Oberdorfer E, Wells D, Dunitz J, Thomas W, Moran A. Diabetes-Related Mortality in Adults with Cystic Fibrosis: Role of Genotype and Gender. *Am J Respir Crit Care Med.* 2014 Dec 5. [Epub ahead of print] PubMed PMID: 25479583.

Platform Presentations:

1. **Blackman S.M.,** Cobb C.E., Beth A.H. 8th Fisher Winternational Symposium on Cellular and Molecular Biology, April, 1998, “New Insights into the Oligomeric Structure of Band 3 In Situ” Banff, Alberta, Canada.
2. **Blackman S.M.,** Piserchio A, Mierke DM, 3rd Harvard-Brown-Padova Workshop on Parathyroid Hormone-Receptor Bimolecular Interactions, September 2000, “Two against One: Structural comparison of the receptors for parathyroid hormone” Le Château de Corbère, France.
3. **Blackman S. M.**, Deering-Brose R., Naughton K., Coleman B., Lai T., Algire M., Beck, S., McWilliams R., Hoover-Fong J., Hamosh A., Fallin M. D., Cutting G. R.. (2005) Modifier genes are responsible for meconium ileus in cystic fibrosis patients, but a major modifier gene is not located on chromosome 19q13. North American Cystic Fibrosis Conference 2005, Baltimore, MD. *Pediatric Pulmonology* 40(S28): 247. Platform presentation.
4. **Blackman, S.M.**, Hsu S., Ritter S. E., Naughton, K. M., Bowers, A., and Cutting, G.R. TCF7L2, a risk gene for type 2 diabetes, is associated with cystic fibrosis-related diabetes. North American Cystic Fibrosis Conference 2007, Anaheim, CA. *Pediatric Pulmonology* 42(S30): 265. Platform presentation.

5. **Blackman, S.M.**, Vanscoy, L.L., Collaco, J.M., Naughton, K. M., and Cutting, G.R. Variability in body mass index in cystic fibrosis is determined partly by a genetic locus on chromosome 5. North American Cystic Fibrosis Conference 2008, Orlando, CA. *Pediatric Pulmonology* 43(S31): 271. Platform presentation.
6. **Blackman, S.M.**, Hsu S., Ritter S.E., Naughton K.M., Drumm M.L., Knowles M.R., Cutting G.R. A Susceptibility Gene for Type 2 Diabetes Is a Genetic Modifier of Diabetes Complicating Cystic Fibrosis. Eastern Society for Pediatric Research, Philadelphia, PA, March 2009. Platform presentation (Plenary session), *awarded the 2009 ESPR Young Investigator (Faculty) award*.
7. **Blackman, S.M.**, Hsu S., Ritter S.E., Naughton K.M., Drumm M.L., Knowles M.R., Cutting G.R. A Susceptibility Gene for Type 2 Diabetes Is a Genetic Modifier of Diabetes Complicating Cystic Fibrosis. 2009 Pediatric Academic Societies Annual Meeting, Baltimore, MD, May 2009. Platform presentation.
8. **Blackman, S.M.,** Ritter S.E., Webel, A., Pace R., Norris S., Naughton K.M., Drumm M.L., Knowles M.R., Cutting G.R. Initial genome-wide testing of common variants for association with CF-related diabetes. North American Cystic Fibrosis Conference 2009, Minneapolis, MN. *Pediatric Pulmonology* 44(S32): 267. Platform presentation.
9. **Blackman, S.M.,** Ritter S.E., Webel, A., Pace R., Norris S., Naughton K.M., Drumm M.L., Knowles M.R., Cutting G.R. Genome-wide analysis identifies alpha-2 catenin (CTNNA2) as a risk factor for diabetes complicating cystic fibrosis (CF). American Society of Human Genetics 2009, Honolulu, HI. Platform presentation.
10. **Blackman, S.M.,** Ritter S.E., Webel, A., Pace R., Norris S., Naughton K.M., Drumm M.L., Knowles M.R., Cutting G.R. Genome-wide analysis identifies alpha-2 catenin (CTNNA2) as a risk factor for diabetes complicating cystic fibrosis (CF). 2010 Pediatric Academic Societies Annual Meeting, Vancouver, Canada. Platform presentation.
11. **Blackman SM**, Commander CW, Drumm ML, Durie PR, Knowles MR, Cutting GR, North American CF Modifier Consortium. S9.1: CF-related diabetes and type 2 diabetes: Different on the outside, similar on the inside? North American Cystic Fibrosis Conference 2010, Anaheim, CA. *Pediatric Pulmonology* 45(S34): 149-151. Platform presentation.
12. **Blackman, S.M.** S8.3: Genetic Susceptibility for CF-related Diabetes Identified by Genome-Wide Association: The International Gene Modifier Consortium. North American Cystic Fibrosis Conference 2011, Anaheim, CA. *Pediatric Pulmonology* 46(S34): 145-146. Platform presentation.
13. **Blackman, S.M.**, Collaco, J.M., Cutting, G.R. Genes Other than CFTR Affect Sweat Chloride in Cystic Fibrosis. North American Cystic Fibrosis Conference 2012, Orlando, FL. *Pediatric Pulmonology* 47(S35): 277. Platform presentation.
14. **Blackman S.M.,** Raghinaru D., Adi S., Beck R., Block J., Chase H.P., DiMeglio, L., DuBose, S., Ebner-Lyon, L., Litton, J., Miller, K., Raman, V., Tamborlane, W., & Schatz, D. for the T1D Exchange Clinic Network. "Insulin pump use in 1182 young children in the T1D Exchange clinic registry is associated with lower hemoglobin in A1c levels than injection therapy." Diabetes Technology Society 2012, Bethesda, MD. *J Diabetes Sci Technol.* 2013; 7:561**.** Platform presentation (highest rated abstract).
15. **Blackman S.M.**, Commander C.W., Watson C., Arcara K, Strug, L.J., Stonebraker J.R., Wright F.A., Rommens J.M., Sun L., Pace R.G., Norris S., Durie P., Drumm M.L., Knowles M.R., Cutting G.R. "Genome-wide association studies for type 2 diabetes and CFRD reveal common risk loci." North American Cystic Fibrosis Conference 2013, Salt Lake City, UT. *Pediatric Pulmonology* 48(S36): 263. Platform presentation.

16. Jonas A., Collaco J. M., **Blackman S.M.** "Impaired fasting glucose is followed by more rapid decline in lung function in cystic fibrosis." North American Cystic Fibrosis Conference 2013, Salt Lake City, UT. *Pediatric Pulmonology* 48(S36): 418. Platform presentation (by research mentee).

Extramural Sponsorship:

Current:

Research Scholars Award in CF (PI- Blackman) 1/1/2015-12/30/2017
Gilead Sciences
Genetic architecture of CF-related diabetes: How much CFTR function is needed and what other genes are involved?
The major goals of this project are to determine how much CFTR function is needed to reduce the risk of CFRD (to aid development of CFTR-directed medications); and to identify additional non-CFTR genes involved in the development of CFRD (which can provide new therapeutic targets).

P30DK079637 Diabetes RTC P&F (PI- Blackman) 5/1/2013-4/30/2015
NIH/NIDDK
Cellular senescence in cystic fibrosis-related diabetes
The major goal of this project is to measure markers of cellular senescence in individuals with CF with and without CFRD, including telomere length and activation of CDKN2A, a type 2 diabetes susceptibility gene which we identified to be a modifier of CFRD.

Type 1 Diabetes T1D Exchange Study (PI- Blackman) 4/26/2011-12/31/2014
JAEB center / Helmsley Trust
The Type 1 Diabetes T1D Exchange
The major goal of this project is to improve the care and treatment of patients with type 1 diabetes. We are collecting comprehensive clinical and phenotypic data from approximately 50,000 patients in the U.S. with type 1 diabetes. Future studies will be planned based on recruitment from this parent study.

Previous:

1 K23 DK083551-01 (PI- Blackman) 7/1/2009-6/30/2014 NCE 12/30/2014
NIH/NIDDK
Genetic Modifiers of Diabetes in Cystic Fibrosis
The major goal of this project is to identify and characterize susceptibility genes for diabetes in patients with cystic fibrosis and to assess their effect in type 2 diabetes.

LWPES Clinical Scholar Award (PI- Blackman) 7/1/2009-6/30/2013
LWPES
Genetic Modifiers of Diabetes in Cystic Fibrosis
The major goal of this project is to identify genetic modifiers which affect risk for diabetes in cystic fibrosis. Funding from this source is intended to support fine mapping of loci identified by the parent study, a genome-wide association study. The PI does not receive salary support from this award; calendar months shown are for oversight of lab tech.

Sanofi TEEN Study (PI- Blackman) 7/1/2012-5/31/2013
Sanofi/Aventis Pharmaceuticals
TEENS: Glycemic control and quality of life in children, adolescents and young adults with type-1 diabetes mellitus described in a world-wide cross-sectional study in 2012: Impact of age-patient-related, treatment-related, behavior and structure of care-related variables
The major goal of this project is to identify clinical and behavioral predictors of hemoglobin A1c in people with type 1 diabetes.

R025-CR07 Research Dev Program (Blackman, Co-I) 7/1/2007-6/30/2009
U.S. Cystic Fibrosis Foundation
Molecular Defects in Cystic Fibrosis: Genetic Modifiers of Cystic Fibrosis Related Diabetes
The major goal of this project is to identify modifier genes which affect the risk for diabetes in patients with cystic fibrosis.

1 F32 DK076446-01 (PI- Blackman) 7/1/2006 – 6/30/2008
NIH/NIDDK
Modifier genes in CF-related diabetes and meconium ileus
The major goal of this project is to identify modifier genes which affect the risk for diabetes and for meconium ileus in patients with cystic fibrosis

DNA Resequencing & Genotyping Service (Cutting) 3/27/2006
NIH/NHLBI
P.I.: G. Cutting; Co-Investigator: S. Blackman
Genetic modifiers of cystic fibrosis: Sibling Study
The goal of this project is to identify genes that modify CF phenotypes by analysis of twin and sibling pairs affected with CF.

Research Program:

1994-1998 Doctoral dissertation, Vanderbilt University School of Medicine (P.I.: Albert H. Beth, Ph.D.). "Dynamics and Self-Association of the Erythrocyte Anion Exchange Protein, Band 3, *in situ*." Goal: to measure the oligomeric state and tether flexibility of the erythrocyte membrane protein, band 3 (a.k.a. *AE1*, *SLC4A1*), the primary attachment site to the membrane skeleton.

- Developed the theoretical framework and data acquisition methodology to measure the orientation of a commonly used chromophore covalently bound to band 3 via polarized fluorescence confocal microscopy, enabling strong constraints to be placed on band 3 rotational dynamics (Blackman et al., 1995)
- Discovered and characterized fluorescence resonance energy homotransfer in this system, giving an independent test of the oligomeric state of band 3 (Blackman et al., 1998)
- Measured the flexibility of the skeletal tether, unifying results from optical and magnetic resonance spectroscopies into a consistent model of restricted-amplitude flexibility (Blackman et al., 2001).

2001 Postdoctoral fellow, Brown University School of Medicine (P.I.: Dale F. Mierke, Ph.D.). Goal: to characterize the determinants for binding of the PDZ1 domain of a postsynaptic density protein, SAP90, to its target via multidimensional NMR.

- Developed a fluorescence anisotropy-based assay for measurement of real-time binding of the PDZ1 domain of SAP90 to a fluorescently labeled peptide target (Piserchio et al., 2002).

2005-2007 Pediatric Endocrine Fellow, Johns Hopkins University School of Medicine. Goal: to test whether modifier genes play a substantial role in meconium ileus and glucose homeostasis (CF-related diabetes).

- Investigated the genetics of neonatal and postnatal intestinal obstruction (meconium ileus and distal intestinal obstruction syndrome) as important factors in weight and nutritional status. Ruled out a significant role for a previously identified modifier locus and identified new genetic loci possibly containing risk genes (Blackman et al., 2006).
- Quantitated the genetic heritability of CF-related diabetes (CFRD) and demonstrated for the first time that genetic modifiers are the primary cause for CFRD (Blackman et al., 2009).
- Demonstrated a correlation between family history of type 2 diabetes and CFRD, suggesting sharing of risk factors.

2008-present Pediatric Endocrine Faculty, Johns Hopkins University School of Medicine. Goal: to identify genes involved in regulation of glucose homeostasis and body weight by studying individuals with cystic fibrosis, a group at high risk for dysregulation of these pathways.

- Expanded on previous studies to include all 3 large North American CF genetic studies. Using these resources, we identified the first genetic modifier for CFRD as *TCF7L2*, a susceptibility gene for type 2 diabetes (Blackman et al., 2010).
- Identified 3 additional T2D susceptibility genes and one non-T2D susceptibility gene (*SLC26A9*) involved in CFRD (manuscript in preparation). SLC26A9 is a chloride/bicarbonate channel expressed in lung, stomach, kidney, and other tissues and has been shown to interact with CFTR.
- Current work in this project is aimed at understanding the mechanisms of action for SNPs controlling risk of CFRD. SNPs which are associated with CFRD either affect CFRD pathogenesis (i.e., are causal), or lie on the same segment of DNA as the untyped causal allele (due to linkage disequilibrium). We are characterizing the tissue expression and splicing of the *SCL26A9* gene to identify the effects of CFRD risk SNPs on gene expression.
- Using high-throughput DNA sequencing we will test for rarer or unique variants in *SLC26A9* and other genes of interest. A pilot study has demonstrated the ability to obtain high-quality sequence information in small numbers of subjects. The next phase is to study groups of families (~800 sibling pairs), in whom inheritance patterns allow for improved quality control and incorporation of linkage information. The subsequent phase will involve the remainder of ~7000 generally unrelated CF subjects available within the International CF Modifier Consortium.
- A second GWAS for CFRD is in progress using patients in the International CF Modifier Consortium. So far genotypes for >4000 individuals are being typed which will allow for confirmation/replication of loci in the first CFRD GWAS, and will increase the sensitivity for detecting novel loci associated with CFRD.
- Using data from >70,000 individuals worldwide with CF, the risk of CFRD will be determined as a function of specific rare and common CF-causing mutations in CFTR (while simultaneously adjusting for other known risk factors for CFRD). This project will determine whether low levels of CFTR function may reduce the risk of CFRD, telling us whether even partial restoration of CFTR function achievable with available CFTR-directed medications may be expected to improve diabetes outcomes in CF.
- We have previously shown nutritional status in CF (quantified by body mass index, BMI) to be determined largely by genetic factors; the same was also true for height and weight in CF. We also identified quantitative trait loci via genome-wide linkage for BMI

(Bradley et al., 2012). We are now performing a genome-wide association study to identify specific risk genes which contribute to this genetic propensity.
- The impact of pre-diabetes (impaired fasting glucose, impaired glucose tolerance) and newly diagnosed CFRD on health in CF is being investigated through the use of CFF Patient Registry data.
- Non-CF-related projects: the T1D Exchange is a network of 80 centers treating people with type 1 diabetes. Using data from >25,000 people with T1D, we are investigating the effects of insulin pump use in children under age 6 with T1D.

EDUCATIONAL ACTIVITIES

Educational Publications:

1. **Blackman, S. M.** Dynamics and Self-Association of the Erythrocyte Anion Exchange Protein, Band 3, *in situ*. Ph.D. Dissertation, Vanderbilt University (1998).

2. **Blackman, S.M.**, and Cooke, D.W. (2013) Diabetes. In: Lennarz, W.J. and Lane, M.D. (eds.) *The Encyclopedia of Biological Chemistry*, Vol. 3, pp. 649-658. Waltham, MA: Academic Press.

Teaching:

Medical student education:

Instructor, Endocrinology section, JHU SOM Genes to Society curriculum (2nd year course), December 2011, December 2012, December 2013.

Clinical instruction:

Inpatient Pediatric Endocrine Service (teaching fellows, residents, and medical students), 4.5 weeks/year, 2007-2009; 10 weeks/year 2010-present.
Pediatric Endocrine Outpatient Clinic (teaching fellows, residents, medical students), 2007-present
Preceptor, Pediatric Endocrine Fellows' Clinic patients, 2007-present

Research mentoring:

Andrea Jonas Medical Student Mentor for 2011 Summer Research Opportunity (Jun-Aug 2011). The goal of this project is to assess the effect of impaired fasting glucose and impaired glucose tolerance in clinical outcomes in CF (BMI, lung function, rates of developing CFRD). *The project is ongoing under Dr. Blackman's supervision. Ms.Jonas gave a platform presentation at the 2013 North American CF Conference in Salt Lake City, UT.*

Monica Majumdar Diabetes Research & Training Center, Summer Medical Student Research mentor (Jun-Aug 2011). The goal of this project is to assess the effects of insulin treatment on nutritional status and lung function in people with CF who were recently diagnosed with diabetes. This retrospective study used data from the CFRD study and Twin-Sibling study.

Ainsley Adams Diabetes Research & Training Center, Summer Medical Student Research mentor (Jun-Aug 2012). The goal of this project is to assess the effects of specific CFTR mutations on rates of CFRD using data from the CFTR2 project. Understanding how

variants at CFTR contribute to CFRD risk will help understand the mechanism of CFRD development, stratify risk for future CFRD development, and increase the power of CFRD genetic studies by reducing the variability arising from CFTR.

Brianna Vecchio Ph.D. Thesis Committee (Graduate program in Cellular & Molecular Medicine)

Kristin Arcara, M.D. Scholarship Oversight Committee (Fellowship in Pediatric Endocrinology)

Anish Shah Diabetes Research & Training Center, Summer Medical Student Research mentor (Jun-Aug 2013). The goal of this project is to identify loci linked to onset of CF-related diabetes by genome-wide linkage using data from the CF Twin and Sibling and CFRD studies.

Selected CME and other presentations:

1. Pediatric Grand Rounds, 5/13/2009, “Diabetes, Genes, and Cystic Fibrosis”
2. Pediatric Endocrinology for the Primary Care Clinician (CME course), 6/20/2009, “Type 2 Diabetes”
3. Child and Adolescent Psychiatry Clinical Conference, 3/17/2010, “Antipsychotic medications, pre-diabetes, and diabetes”
4. Division of Neonatology, 4/1/2010, “Thyroid disease in the neonate”
5. Pediatric Endocrinology for the Primary Care Clinician (CME course), 6/19/2010, “Type 2 Diabetes”
6. Diabetes short course (CME Course), North American CF Conference, 10/20/2010
7. Seminars in Endocrinology, 3/2/2011, “Methods in Human Genetics in Endocrine Research”
8. Johns Hopkins Pediatric Trends 2011 (CME course), 4/12/2011, “Type 2 Diabetes in Children”
9. Pediatric Endocrinology for the Primary Care Clinician (CME course), 6/17/2011, “Type 2 Diabetes”
10. Child and Adolescent Psychiatry Clinical Conference, February 2012, “Endocrine physiology and pathophysiology for the child and adolescent psychiatrist”
11. Division of Neonatology, March 2012, “Thyroid disease in the neonate”
12. Johns Hopkins Hospital Endocrine Grand Rounds, 1/09/13, “Genetic Modifiers of Cystic Fibrosis-Related Diabetes Include a CFTR-Interacting Protein and a Marker of Cellular Senescence.”

13. Pediatric Pulmonology Seminar, 2/26/13, “CF-related diabetes and nutritional status in CF.”

14. Pediatric Resident Research Seminar, 1/9/2014, “Susceptibility genes for CF-related diabetes and type 2 diabetes.”

15. Division of Endocrinology, 3/5/2014, “Misconceptions about CFRD"

16. Division of Neonatology, April 25, 2014, “Thyroid disease in the neonate.”

17. Johns Hopkins Hospital Scientific Abstracts Grand Rounds, 4/30/14, “Impaired fasting glucose is followed by more rapid decline in lung function in cystic fibrosis.”

18. Pediatric Case Conference, 8/29/2014, “11 y.o. with recurrent hypoglycemia.”

19. Johns Hopkins Hospital Endocrine Grand Rounds, 12/17/14, “Cystic Fibrosis-related diabetes in the genome era.”

20. Pediatric Resident Teaching Conference, 1/6/15, “Recognition and management of disorders of puberty”

Editorial Activities:

Journal Editorial Board:

Journal of Cystic Fibrosis (2013-)

Journal Peer Reviewer:

Journal of Clinical Endocrinology and Metabolism
Diabetes Care
Journal of Pediatrics
Diabetes Research and Clinical Practice
Journal of Cystic Fibrosis
Medicine

Grant Reviews/Study Sections:

Baltimore DRTC reviewer, 2011 and 2012 cycles
CF Foundation, Clinical Research grant reviewer, Bethesda, MD (2012, 2013, 2014)
NIDDK R01 reviewer (study section), March 2012
NIDDK R01 reviewer (study section), July 2014
NIDDK P30 reviewer (study section), Bethesda, MD, November 2014

CLINICAL ACTIVITIES

Certification

2000 USMLE Steps 1, 2, and 3 passed
2004 Board Certification, American Board of Pediatrics (#080245, exp. 12/31/2016)
2007 State of Maryland Medical License (D0065984, exp. 9/30/2016)
2007 Board Certification, Pediatric Endocrinology (exp. 12/31/2016)

Clinical (Service) Responsibilities

July 2007-Dec 2009	Pediatric Endocrinology Inpatient Service, Attending, 1 month/year
January 2010-present	Pediatric Endocrinology Inpatient Service, Attending, 2 months/year
July 2007-present	Pediatric Endocrinology Clinic, Attending, 3-4 half-day clinics/month

Clinical Program Building:

July 2007-present	Founder and Director, Johns Hopkins Cystic Fibrosis-related diabetes clinic
April 2010-present	JHU PI for T1D Exchange (type 1 diabetes patient registry and clinical trials umbrella study)
September 2010-October 2011	CFRD clinic selected for pilot program in Quality Improvement for CF-related diabetes (Pediatric Endocrine Society / CFF Foundation)
July 2014-present	Pediatric Endocrinology Division QI project leader

ORGANIZATIONAL ACTIVITIES

December 2007-	Faculty Interviewer, Pediatric Residency
April 2009	Residency Career Planning Event (representative of Endocrinology)
2008-2010	POE sponsor for diluted insulin order set
2010-2011	LWPES/NACF CF-related diabetes LLC (joint Endocrine/Pulmonary Quality Improvement committee)
2012-	Committee Chairman, T1D Exchange Study, Insulin Pumps in Children

Conference Organizer, Session Leader, North American Cystic Fibrosis Conference, October 2008 (Orlando, FL)

Conference Organizer, Session Leader, North American Cystic Fibrosis Conference, October 2009 (Minneapolis, MN)

Conference Organizer, Session Leader, North American Cystic Fibrosis Conference, October 2010 (Baltimore, MD)

Roundtable moderator, North American Cystic Fibrosis Conference, October 2011 (San Diego, CA)

Program Planning Committee, North American Cystic Fibrosis Conference, December 2012-present; Session leader for Symposium and for Workshop (Salt Lake City, October 2013).

Diabetes Core Working Group, Advancing Pediatric Therapeutics, NICHD (Obstetric and Pediatric Pharmacology and Therapeutics Branch (OPPTB)) (Jun 2013-present)

Pediatric Resident Research Day, Faculty Judge (2013, 2014)

Program Planning Committee, North American Cystic Fibrosis Conference, (Atlanta, October 2014); Workshop session leader, Roundtable Moderator.

T1D Exchange, Pediatric Study Committee (2014)

ACGME CLER site visit, pediatric endocrine faculty representative, 11/12/2014

Professional Societies:

Endocrine Society, 2005-present
American Society of Human Genetics, 2005-present
Lawson Wilkins Pediatric Endocrine Society, 2004-present
American Diabetes Association, 2012-present
Sigma Xi, 2000-present
Society for Pediatric Research, 2012-present

RECOGNITION

Awards:

National Merit Scholar, 1988
Baccalaureate awarded with honors in Chemistry, 1992
Robert Thornton McCay Prize in Physical Chemistry, Princeton University, 1992
Lawson Wilkins Pediatric Endocrine Society Research Fellowship Award, 2006.
Endocrine Society Genentech Clinical Fellows Travel Grant, June 2007
Lawson Wilkins Pediatric Endocrine Society Clinical Scholars Award, 2009
ESPR Young Investigator Award (Faculty), March 2009
Castle Connelly/U.S. News Top Doctor, 2011, 2012, 2013, 2014
Society for Pediatric Research, March 2012
Gilead Sciences Research Scholar Award in Cystic Fibrosis, October 2014

Invited talks:

1. Symposium Speaker, Red Cell Club Annual Meeting, October 1998, "Oligomeric state of human erythrocyte band 3 measured by fluorescence resonance energy homotransfer" Philadelphia, PA.

2. Symposium Speaker, 32nd European CF Conference, Brest, France, June 12, 2009, "The North American Consortium for CF Genetic Modifiers"

3. Invited speaker, UNC Chapel Hill, Genetics division, "Shared genetic susceptibility for cystic fibrosis-related diabetes and type 2 diabetes," September 15, 2010.

4. Symposium Speaker, North American Cystic Fibrosis Conference, Baltimore, MD, October 22, 2010, "CFRD and type 2 diabetes: Different on the outside, similar on the inside?"

5. Invited speaker, University of Iowa, Genetics division, "Shared genetic susceptibility for cystic fibrosis-related diabetes and type 2 diabetes," February 1, 2011.

6. Award lecture, Pediatric Academic Societies, Denver, CO, April 30, 2011, "Genetic Modifiers of Cystic Fibrosis-related Diabetes"

7. NIDDK Workshop on Cystic Fibrosis-related Diabetes, May 25-26, 2011. "The purpose of this workshop is to develop a research agenda for the NIDDK for both basic and clinical research on this topic."

8. Symposium Speaker, North American Cystic Fibrosis Conference, San Diego, CA, November 4, 2011, “Genetic susceptibility for CF-related diabetes identified by genome-wide association: the International Gene Modifier Consortium”

9. Invited speaker, University of Pittsburgh, Richard King Mellon Institute for Pediatric Research, August 7, 2012, “Genetic Modifiers of Cystic Fibrosis-related Diabetes”

10. Invited speaker, NICHD, Endocrine Grand Rounds, September 21, 2012, “Genetic Modifiers of Cystic Fibrosis-related Diabetes”

11. Invited speaker, University of Washington and Seattle Children’s Hospital, March 13, 2013.

12. Invited speaker, Children’s Hospital of Philadelphia, Endocrinology Grand Rounds, April 17, 2013.

13. Invited speaker, Vanderbilt University School of Medicine, Endocrinology Grand Rounds, October 9, 2013.

14. Invited speaker, Seattle Children’s Hospital, Division of Pediatric Endocrinology, 10/20/2014.