BIOGRAPHICAL SKETCH

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NAME	POSITION TITLE
Phillips, John A.	Professor of Pediatrics, Biochemistry and Medicine
eRA COMMONS USER NAME (credential, e.g., agency login) phillija	

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	YEAR(s)	FIELD OF STUDY
University of North Carolina at Chapel Hill	None	1962-1965	Chemistry
Bowman Gray School of Medicine	M.D.	1965-1969	Medicine

A. Positions and Honors

Positions and Employment

1969-71 Intern and Jr. Resident Pediatrics, Children's Hospital Medical Center, Boston, MA
1971-73 Lt. Comdr., USNR (MC) Naval Regional Medical Center, Jacksonville, FL
1973-75 Sr. and Chief Resident, Pediatrics, Children's Hospital Medical Center, Boston, MA
1975 Sidney Farber Award, Children's Hospital Medical Center, Boston, MA
1975-77 Research Fellow in Pediatric Genetics, Johns Hopkins Univ. Schl of Medicine, Baltimore, MD
1981- Member, Society for Pediatric Research
1978-82 Assistant Professor of Pediatrics, Johns Hopkins University School of Medicine, Baltimore, MD
1982-84 Associate Professor of Pediatrics, Johns Hopkins University School of Medicine, Baltimore, MD
1984- Professor of Pediatrics and Biochemistry; Director, Division of Genetics, Vanderbilt University School of Medicine, Nashville, TN
1984 E. Mead Johnson Award for Research in Pediatric Research
1984-88 Member, Board of Scientific Counselors, NICHD
1985- Member, American Society for Clinical Investigation
1992- David T. Karzon Professor of Pediatrics, Vanderbilt University School of Medicine, Nashville, TN
1994-00 Member, Board of Directors, American College of Medical Genetics
1994-08 Elected by Peers for inclusion in Best Doctors in America
1999- Member Executive Board, Vanderbilt Program in Human Genetics
1999- Adjunct Professor of Microbiology, Meharry Medical College, Nashville, TN
2001-06 Director, Genetics Training Program (T32, GM62758)
2001-07 GCRC Director of Genetics, Vanderbilt General Clinical Research Center
2002 Harvie Branscomb Distinguished Professor Award, Vanderbilt University School of Medicine
2002-08 Selected as one of “America’s Top Pediatricians”
2003-08 Co-Investigator and CORE Lab Director PPH PPG (HL072058)
2006-08 Course Director, Genetics (IDIS5040) Course, Vanderbilt University School of Medicine
2005- Co-Director, American College Medical Genetics Board Review Course

B. Selected peer-reviewed publications (in chronological order)

Scott, AF, Phillips, JA III and Migeon, BR: DNA restriction endonuclease analysis for the localization of the human delta and beta globin genes on chromosome 11. Proc. Natl. Acad. Sci. USA 76:4563-4565, 1979.

Phillips, JA III, Hjelle, BL, Seeburg, PH and Zachmann, M: A molecular basis for familial isolated growth hormone deficiency. Proc. Natl. Acad. Sci. USA 78:6372-6375, 1981.

Stokes, DC, Phillips, JA III, Leonard, CO, Dorst, JP, Kopits, SE, Trojak, JE and Brown, DL: Respiratory complications of achondroplasia. J. Pediat. 102:534-541, 1983.

Boehm, CD, Antonarakis, SE, Phillips, JA III, Stetten, G, and Kazazian, HH Jr: Prenatal diagnosis using DNA polymorphisms: Report on 95 pregnancies at risk for sickle-cell disease or ß-thalassemia. New Engl. J. Med. 308:1054-1058, 1983.

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Vnencak-Jones, CL, Phillips, JA, III, Chen, EY, and Seeburg, PH: Molecular basis of human growth hormone gene deletions. Proc. Natl. Acad. Sci (USA) 85:5615-5619, 1988.

Vnencak-Jones, CL and Phillips, JA III: Non Alu sequences constitute hot spots for human growth hormone gene deletions. Science 250:1745-1748, 1990.

Campbell, PW III, Phillips, JA III, Krishnamani, MRS, Maness, KJ and Hazinski, TA: Cystic Fibrosis: Relationship between clinical status and F508. J. Peds. 118:239-241, 1991.

Campbell, PW III, Parker, RA, Roberts, BT, Krishnamani, MRS and Phillips, JA III: Association of poor clinical status and heavy exposure to tobacco smoke in cystic fibrosis patients homozygous for the F508 deletion. J. Peds. 120: 261-264, 1992.

The cystic fibrosis genotype-phenotype consortium: Correlation between genotype and phenotype in cystic fibrosis: Analysis of seven common mutations. N. Engl. J. Med. 329:1308-1313, 1993.

Cogan, JD, Ramel, B, Lehto, M, Phillips, JA III, Prince, M, Blizzard, M, deRavel, TJL, Brammert, M and Groop, L: A recurring dominant-negative mutation causes autosomal dominant growth hormone deficiency. J. Clin. Endocrinol. & Metab. 80:3591-3595, 1995.

Clayton, EW, Hannig, VL, Pfotenhauer, JP, Parker, RA, Campbell, PW III and Phillips, JA III: Teaching about cystic fibrosis carrier screening using written and video information. Am. J. Hum. Genet. 57:171-181, 1995.

Campbell, PW III, Phillips, JA III, Heidecker, GJ, Krishnamani, MRS, Zahorchak, R and Stull, TL: Detection of *Pseudomonas (Burkholderia) cepacia* using species-specific PCR. Pediatric Pulmonology 20:44-49, 1995.

Clayton, EW, Hannig, VL, Pfotenhauer, JP, Parker, RA, Campbell, PW III & Phillips, JA III: Lack of interest by nonpregnant couples in population based CF carrier screening. Am. J. Hum. Genet. 58:617-627, 1996.

Nichols, WC, Koller, DL, Slovis, B, Foroud, T, Terry, VH, Arnold, ND, Siemieniak, DR, Wheeler, L, Phillips, JA III, Newman, JH, Conneally, PM, Ginsburg, D and Loyd, JE: Localization of the gene for familial primary pulmonary hypertension to chromosome 2q31-32. Nature Genetics 15:277-280, 1997.

Cogan, JD, Prince, MA, Lekhakula, S, Bundey, S, Futrakul, A, McCarthy, EMS and Phillips, JA III: A novel mechanism of aberrant pre-mRNA splicing in humans. Hum. Molec. Genetics 6:909-912, 1997.

Wu, W, Cogan, JD, Pfaffle, RW, Dasen, JS, Frisch, H, O'Connell, SM, Flynn, SE, Brown, MR, Mullis, PE, Parks, JS, Phillips, JA III and Rosenfeld, MG: Mutations in PROP1 cause familial combined pituitary hormone deficiency. Nature Genetics 18:147-149, 1998.

McCarthy, EMS and Phillips, JA III: Characterization of an intron splice enhancer that regulates alternative splicing of GH pre-mRNA. Human Molec. Genetics 7:1491-1496, 1998.

Salvatori, R, Gondo, RG, de Aguirar Oliveira, MH, Phillips, JA III, Souza, AH, Hayashida, CY, Toledo, SP, Conceicao, MM, Prince, M, Baumann, G and Levine, MA: Familial isolated growth hormone deficiency due to a novel mutation in the GHRHR. J. Clin. Endo. Metab. 84:917-923, 1999.

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Newman, JH, Wheeler, L, Lane, KB, Loyd, E, Gaddipati, R, Phillips, JA III and Loyd, JE: Mutation in the gene for bone morphogenetic protein receptor II as a cause of primary pulmonary hypertension in a large kindred. N. Engl. J. Med. 345:319-324, 2001.

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Thomas, AQ, Lane, K, Phillips, JA III, Prince, M., Markin, C, Gaddipati, R, Marney, A, Johnson, J, Haines, J, Stahlman, M and Loyd, JE: Heterozygosity for a surfactant protein C mutation causes familial usual interstitial pneuomonitis in adults and cellular interstitial pneumonitis in children. Am J Respir Crit Care Med 165: 1322-1328, 2002.

Runo, JR, Vnencak-Jones, CL, Prince, M, Loyd, JE, Wheeler, L, Robbins, IM, Lane, KB, Newman, JH, Johnson, J, Nichols, WC and Phillips, JA III: Pulmonary veno-occlusive disease caused by an inherited mutation in the bone morphogenetic protein receptor2. Am J Respir Crit Care Med 167: 889-894, 2003.

McGuinness, L, Magoulas, C, Sesay, AK, Mathers, K, Carmignac, D, Manneville, J-B, Christian, H, Phillips, J.A. III and Robinson, ICAF: Autosomal dominant growth hormone deficiency disrupts secretory vesicles in vitro and in vivo in transgenic mice. Endocrinol 144: 720-731, 2003.

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Cogan, JD, Vnencak-Jones, CL, Phillips, JA III, Lane, KB, Wheeler, LA, Robbins, IM, Garrison, G, Ledges, LK and Loyd, JE: Gross BMPR2 gene rearrangements constitute a new cause for primary pulmonary hypertension. Genetics in Medicine 7:169-174, 2005.

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Potter, A, Phillips, JA III and Rimoin, DL: Genetic Disorders of the Pituitary Gland: In Principles and Practice of Medical Genetics, Rimoin, DL, Connor, JM, Pyeritz, RE and Korf, BR (eds), Fifth Edition, Elsevier Inc, in press, 2006.

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Cogan, JD, Pauciulo, MW, Batchman, AP, Prince, MA, Robbins, IM, Hedges, LK, Stanton, KC, Wheeler, LA, Phillips, JA III, Loyd, JE and Nichols, WC: High frequency of BMPR2 exonic deletions/duplications in familial arterial hypertension. Am J Resp Crit Care Med 174:590-8, 2006.

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C. Research Support

Ongoing Research Support

5R01 DK35592-20 (Phillips) 04/01/2005-04/30/2010
NIH/NIDDK
GH Alternative Splicing: Mechanisms and Disease
This is a study of a new type of enhancer sequences that regulate alternative splicing of gene transcripts. The interactions between these enhancers and SR proteins to achieve regulation of normal splicing patterns is being studied to determine the underlying mechanisms that regulate alternative splicing.

N/A (Phillips) 07/01/2008-06/30/2009
State of Tennessee
The major goals of this project seek to reduce the frequency and burden of genetic disorders in the middle Tennessee and adjoining regions by newborn screening, early diagnosis, treatment and education of affected individuals, their relatives and those at risk. Methods used include biochemical, clinical and cytogenetic and molecular studies to identify affected individuals.

1P01 HL072058-05 (Loyd) 08/04/2003-07/31/2009
NIH/NHLB
Genetic and Environmental Pathogenesis of PPH
Our target goals are to identify the modifying genes and environmental features that regulate the clinical expression of mutations in BMPR2; to develop understanding about how BMPR2 mutations result in disease; and to identify the undiscovered mutations which cause PPH.

1 U54 RR 023499-01A1 (Bernard) 09/30/2007 - 09/29/2012
NIH/NCRR
The Vanderbilt Institute for Clinical and Translational Research (VICTR)
This program constitutes an institute which permits Vanderbilt to forge a uniquely transformative, novel, self-evaluated, integrative academic home for clinical and translational science, where both research and researchers flourish. This program will be organized and designed to focus on both bench to bedside and bedside to practice translation. Vanderbilt's institute will complement and interact with the existing NIH funded centers and industry. The program incorporates a

Completed Research Support

2M01 RR00095 (Balser) 12/1/2002-11/30/2007
NIH/NCRR
General Clinical Research Center – Genetics Core
The goal of this study is to provide information about and access to current methods of genetic analysis to assist GCRC investigators in their studies.