# Dr. Sarah A. Pendergrass

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## **Education:**

Ph.D. in Genetics, Dartmouth College, Hanover, NH	2009
M.S. in Engineering, Thayer School of Engineering, Hanover, NH	2004
B.A. in Physics, Smith College, Northampton, MA	2001

### **Research Experience:**

Research Faculty, Center for System Genomics, Department of Biochemistry and	2011-
Molecular Biology	2014
Lab of Dr. Marvlyn Ritchie	

Research Interests: Development of novel computational and data visualization approaches for data analysis and display of large high-throughput datasets. Characterization of the relationship between genetic variation, phenotypic outcome, and common complex disease, within the framework of ancestry and environment, including Phenome-Wide Association Study (PheWAS) projects. Analysis has included data from the Epidemiologic Architecture for Genes Linked to Environment (EAGLE) project, as well as data from the Population Architecture Using Genomics and Epidemiology (PAGE) consortium, the Electronic Medical Records and Genomics (eMERGE) network, the Pharmacogenomics Research Network (PGRN), and the Aids Clinical Trials Group (ACTG).

Postdoctoral Research Fellow, Center for Human Genetics Research, Vanderbilt University, Nashville TN Advisors: Dr. Marylyn Ritchie, Dr. Dana Crawford

Ph.D. research, Dartmouth College, Hanover, NH Advisors: Dr. Michael Whitfield, Dr. Jason Moore Thesis: "Gene expression subsets and biomarkers in the genome-wide expression profiles of systemic sclerosis"

M.S. research, Thayer School of Engineering, Dartmouth College, Hanover, NH Advisors: Dr. Paul Meany, Dr. Marvin Doyley Thesis: "Microwave imaging device design and improvement related to dielectric property measurement and experimentation"

#### **Awards and Grants:**

Huck Institutes of Science, Cyberhealth Innovation Seed Funds: iPhenoGram: Exploring Genetic Associations Interactively across Chromosomes for Multiple Organisms and Multiple Scales

Associate Faculty Member of the Faculty of 1000 Prime (http://f1000.com/prime)

Genome Technology 8th Annual Young Investigator, December 13, 2013

Young Investigator Award, 10th International Workshop on Scleroderma, 2008

Keystone Symposia Scholarship Recipient, 2008

NIAMS Autoimmunity and Connective Tissue Training Grant 2006 - 2008

#### **Teaching/Mentoring Experience:**

Mentoring graduate students in the lab of Dr. Marylyn Ritchie, 2011-2014

Taught bioinformatics courses at the Centers for Disease Control and Prevention (CDC), September 30-October 3, 2014

Co-taught tutorial titled "Detecting And Characterizing Pleiotropy: New Methods For Uncovering The Connection Between The Complexity Of Genomic Architecture And Multiple Phenotypes" at the Pacific Symposium on Biocomputing, January 4, 2014

Taught course titled "Next Generation Sequencing: Applications for Infectious Disease" at the CDC, February 28, 2012

Taught data visualization module for the "Comparison of Analytical Methods for Genetic Association Studies" course at the CDC, February 25, 2011

Participant in Dartmouth Center for Learning seminars for teacher training, mentoring, and professional development 2006 - 2008

Teaching Assistant, Course: Molecular and Computational Genomics, 2006

Teaching Assistant, Course: Introduction to Genetics, 2006

#### Skills:

Bioinformatics, Biostatistics, Data Visualization Development, Genomic Analysis, Programming, Numerical Methods

## **Memberships:**

American Society of Human Genetics

International Genetic Epidemiology Society

## **Event Organization:**

Co-leading a peer-reviewed paper session at the Pacific Symposium on Biocomputing, titled "Characterizing the Importance of Environmental Exposures, Interactions between the Environment and Genetic Architecture, and Genetic Interactions: New Methods for Understanding the Etiology of Complex Traits and Disease", January, 2015

Organized and led a three hour Data Visualization Workshop for the Bioinformatics and Genomics Retreat for the Huck Institutes of the Life Sciences at the Pennsylvania State University, September 13, 2014

Bioinformatic Track Program Committee, ACM Conference on Bioinformatics, Computational Biology and Health, September 20-23, 2014

Co-led a peer-reviewed paper session at the Pacific Symposium on Biocomputing, titled "Detecting And Characterizing Pleiotropy: New Methods For Uncovering The Connection Between The Complexity Of Genomic Architecture And Multiple Phenotypes", January 6, 2014

Co-led a workshop at the Pacific Symposium on Biocomputing, titled "Uncovering the Etiology of Autism Spectrum Disorders: Genomics, Bioinformatics, Environment, Data Collection and Exploration, and Future Possibilities", January 4, 2014

### **Invited Presentations:**

"Biofilter 2.0 for Advanced Predictive Model Development, Testing, and Hypothesis Generation using Expert Domain Knowledge Resources" American Medical Informatics Association – Translational Bioinformatics, San Francisco, CA, June 7, 2014

"Visualizing Multiple Types of Genomic Information Across Chromosomes With PhenoGram" American Medical Informatics Association – Translational Bioinformatics, San Francisco, CA, June 7, 2014

"Adding Value to Large Genomic Epidemiology Studies: Phenome Wide Association Studies for Exploring the Relationship Between the Phenome and Genome" American Association for Cancer Research, June 5, 2014 "Phenome Wide Association Study (PheWAS) for Detection of Pleiotropy within the Population Architecture using Genomics and Epidemiology (PAGE) Network " Translational Bioinformatics Conference, Seoul, Korea, October 4, 2013

"Elucidating the Genetic Architecture of Complex Disorders: Challenges and New Approaches" Drexel University, Philadelphia PA, May 22, 2013 "Visually integrating and exploring high throughput results using PheWAS-view, PhenoGram, and Synthesis-View" PGRN Statistical Analysis Resource, December 4, 2012

"Visually Integrating and Exploring High Throughput Phenome-Wide Association (PheWAS) Results Using PheWAS-view and PhenoGram" Annual Meeting of the American Society of Human Genetics, San Francisco, November 8, 2012

"Identification of Pleiotropy with a Phenome-Wide Association Study (PheWAS) using two National Health and Nutrition Examination Surveys (NHANES)" Translational Bioinformatics Conference, JeJu Island, Korea, October 15, 2012

"Challenges and Approaches for Elucidating the Genetic Architecture of Complex Disorders" Environmental Epidemiology Network Meeting, International Meeting for Autism Research, Toronto Canada, May 17, 2012

"A Phenome-wide Exploration of Genotype-Phenotype Associations and Pleiotropy using Metabochip in the PAGE Study" Keystone Symposium Complex Traits: Genomics and Computational Approaches, Breckenridge, Colorado, February 21, 2012

"A Phenome-wide Exploration of Novel Genotype-Phenotype Associations and Pleiotropy using Metabochip in the PAGE Network" Annual Meeting of the International Genetic Epidemiology Society, Heidelberg, Germany, September 20, 2011

"Visual Integration of Results Using Synthesis-View" Gene Environment Association Studies (GENEVA) Steering Committee Meeting, Washington D.C., January 18, 2011

"Visual Integration of Results from a Large DNA Biobank (BioVU) using Synthesis-View" Pacific Symposium on Biocomputing, Big Island, Hawaii, January 5, 2011

"Phenotype-Wide Association Study (PheWAS) for Detection of Pleiotropy within the Multi-Ethnic Cohorts of the Population Architecture Using Genomics and Epidemiology (PAGE) network"

Annual Meeting of the American Society of Human Genetics, Washington D.C., November 2, 2010

"Phenotype-Wide Association Study (PheWAS) for Exploration of Novel SNP and Phenotype Relationships within PAGE" 2010 Annual Meeting of the International Genetic Epidemiology Society, Boston, MA, October 10, 2010

## Software available at http://visualization.ritchielab.psu.edu/

*PhenoGram:* For visualizing information across human chromosomes, model organism visualization in beta

*Synthesis-View:* Through the use of stacked data-tracks, information on SNP genomic locations, presence of the SNP in a specific study or analysis, as well as related information such as genetic effect size and summary phenotype information, are available for plotting according to user preference

**PheWAS-View:** PheWAS-View creates visual summaries of the SNP, gene, phenotype, and association information resulting from PheWAS studies

## **Peer-reviewed Publications:**

- 1. **Pendergrass S.A.**, Ritchie M.D. Phenome-Wide Association Studies: Leveraging Comprehensive Phenotypic and Genotypic Data for Discovery. *Accepted Current Genetic Medicine Reports*
- 2. Chhibber A, Kroetz D, Tantisira KG, McGeachie M, Cheng C, Plenge R, Stahl E, Sadee W, Ritchie MD, **Pendergrass SA**. Genomic Architecture of Pharmacological Efficacy and Adverse Events. *Accepted Pharmacogenomics*
- 3. Ritchie MD, Holzinger ER, Li R, **Pendergrass SA**, Kim D. Systems Genomics analyses and Data Integration for exploring the genetic architecture of complex traits. *Accepted Nature Reviews Genetics*
- 4. Hall MA, Verma A, Brown-Gentry KD, Goodloe R, Boston J, Wilson S, McClellan B, Sutcliffe C, Dilks HH, Gillani NB, Jin H, Mayo P, Allen M, Schnetz-Boutaud N, Crawford DC, Ritchie MD, **Pendergrass SA**. Detection of Pleiotropy Through a Phenome-Wide Association Study (PheWAS) of Epidemiologic Data as Part of the Environmental Architecture for Genes Linked to Environment (EAGLE) Study. *In Press PLoS Genetics*
- 5. Barrie ES, Weinshenker D, Verma A, **Pendergrass SA**, Lange LA, Ritchie MD, Wilson JG, Kuivaniemi H, Tromp G, Carey DJ, Gerhard GS, Cubells JF, Sadee W. Regulatory polymorphisms in DBH affect peripheral gene expression and sympathetic phenotypes. *In Press Circulation Research*

- Kim D, Li R, Dudek SM, Frase AT, Pendergrass SA, Ritchie MD. Knowledge-driven genomic interactions: an application in ovarian cancer. BioData Min. 2014;7:20. PMID: 25214892
- Cronin RM, Field JR, Bradford Y, Shaffer CM, Carroll RJ, Mosley JD, Bastarache L, Edwards TL, Hebbring SJ, Lin S, Hindorff LA, Crane PK, Pendergrass SA, Ritchie MD, Crawford DC, Pathak J, Bielinski SJ, Carrell DS, Crosslin DR, Ledbetter DH, Carey DJ, Tromp G, Williams MS, Larson EB, Jarvik GP, Peissig PL, Brilliant MH, McCarty CA, Chute CG, Kullo IJ, Bottinger E, Chisholm R, Smith ME, Roden DM, Denny JC. Phenome-wide association studies demonstrating pleiotropy of genetic variants within FTO with and without adjustment for body mass index. Front Genet. 2014;5:250. PMID: 25177340
- 8. Kraja AT, Chasman DI, North KE, Reiner AP, Yanek LR, Kilpeläinen TO, Smith JA, Dehghan A, Dupuis J, Johnson AD, Feitosa MF, Tekola-Ayele F, Chu AY, Nolte IM, Dastani Z, Morris A, Pendergrass SA, Sun YV, Ritchie MD, Vaez A, Lin H, Ligthart S, Marullo L, Rohde R, Shao Y, Ziegler MA, Im HK, Cross Consortia Pleiotropy (XC-Pleiotropy) Group, the Cohorts for Heart and, Aging Research in Genetic Epidemiology (CHARGE), the Genetic Investigation of Anthropometric Traits (GIANT) Consortium, the Global Lipids Genetics Consortium (GLGC), the Meta-Analyses of Glucose, Insulinrelated traits Consortium (MAGIC), the Global BPgen (GBPG) Consortium, The ADIPOGen Consortium, the Women's Genome Health Study (WGHS), the Howard University Family Study (HUFS), Schnabel RB, Jørgensen T, Jørgensen ME, Hansen T, Pedersen O, Stolk RP, Snieder H, Hofman A, Uitterlinden AG, Franco OH, Ikram MA, Richards JB, Rotimi C, Wilson JG, Lange L, Ganesh SK, Nalls M, Rasmussen-Torvik LJ, Pankow JS, Coresh J, Tang W, Linda Kao WH, Boerwinkle E, Morrison AC, Ridker PM, Becker DM, Rotter JI, Kardia SLR, Loos RJF, Larson MG, Hsu Y-H, Province MA, Tracy R, Voight BF, Vaidya D, O'Donnell CJ, Benjamin EJ, Alizadeh BZ, Prokopenko I, Meigs JB, Borecki IB. Pleiotropic genes for metabolic syndrome and inflammation. Mol Genet Metab. 2014 May 9; PMID: 24981077
- Ciesielski TH, Pendergrass SA, White MJ, Kodaman N, Sobota R, Huang M, Bartlett J, Li J, Pan Q, Gui J, Selleck SB, Amos CI, Ritchie MD, Moore JH, Williams SM. Diverse convergent evidence in the genetic analysis of complex disease: coordinating omic, informatic, and experimental evidence to better identify and validate risk factors. BioData Mining. 2014 Jun 30;7(1):10.
- Mitchell SL, Hall JB, Goodloe RJ, Boston J, Farber-Eger E, Pendergrass SA, Bush WS, Crawford DC. Investigating the relationship between mitochondrial genetic variation and cardiovascular-related traits to develop a framework for mitochondrial phenome-wide association studies. BioData Mining. 2014 Apr 15;7(1):6. PMID: 24731735
- 11. Kocarnik JM, Pendergrass SA, Carty CL, Pankow JS, Schumacher FR, Cheng I, Durda P, Ambite JL, Deelman E, Cook NR, Liu S, Wactawski-Wende J, Hutter C, Brown-Gentry K, Wilson S, Best LG, Pankratz N, Hong C-P, Cole SA, Voruganti VS, Bůžkova P, Jorgensen NW, Jenny NS, Wilkens LR, Haiman CA, Kolonel LN, Lacroix A, North K, Jackson R, Le Marchand L, Hindorff LA, Crawford DC, Gross M, Peters U.

Multiancestral analysis of inflammation-related genetic variants and C-reactive protein in the population architecture using genomics and epidemiology study. Circ Cardiovasc Genet. 2014 Apr 1;7(2):178–188. PMID: 24622110

- 12. Chhibber A, Mefford J, Stahl EA, Pendergrass SA, Baldwin RM, Owzar K, Li M, Winer EP, Hudis CA, Zembutsu H, Kubo M, Nakamura Y, McLeod HL, Ratain MJ, Shulman LN, Ritchie MD, Plenge RM, Witte JS, Kroetz DL. Polygenic inheritance of paclitaxel-induced sensory peripheral neuropathy driven by axon outgrowth gene sets in CALGB 40101 (Alliance). Pharmacogenomics J. 2014 Feb 11; PMID: 24513692
- Mitchell SL, Goodloe R, Brown-Gentry K, Pendergrass SA, Murdock DG, Crawford DC. Characterization of mitochondrial haplogroups in a large population-based sample from the United States. Hum Genet. 2014 Feb 1; PMID: 24488180
- 14. **Pendergrass S**, Girirajan S, Selleck S. Uncovering the etiology of autism spectrum disorders: genomics, bioinformatics, environment, data collection and exploration, and future possibilities. Pac Symp Biocomput. 2014;19:422–426. PMID: 24297568
- 15. Hall MA, Dudek SM, Goodloe R, Crawford DC, Pendergrass SA, Peissig P, Brilliant M, McCarty CA, Ritchie MD. Environment-wide association study (EWAS) for type 2 diabetes in the Marshfield Personalized Medicine Research Project Biobank. Pac Symp Biocomput. 2014;200–211. PMID: 24297547
- 16. Tyler AL, Crawford DC, Pendergrass SA. Detecting and characterizing pleiotropy: new methods for uncovering the connection between the complexity of genomic architecture and multiple phenotypes- session introduction. Pac Symp Biocomput. 2014;19:183–187. PMID: 24297545
- 17. Moore CB, Wallace JR, Wolfe DJ, Frase AT, Pendergrass SA, Weiss KM, Ritchie MD. Low frequency variants, collapsed based on biological knowledge, uncover complexity of population stratification in 1000 genomes project data. PLoS Genet. 2013 Dec;9(12):e1003959. PMID: 24385916
- Pendergrass SA, Frase A, Wallace J, Wolfe D, Katiyar N, Moore C, Ritchie MD. Genomic analyses with biofilter 2.0: knowledge driven filtering, annotation, and model development. BioData Mining. 2013 Dec 30;6(1):25. PMID: 24378202
- 19. Denny JC, Bastarache L, Ritchie MD, Carroll RJ, Zink R, Mosley JD, Field JR, Pulley JM, Ramirez AH, Bowton E, Basford MA, Carrell DS, Peissig PL, Kho AN, Pacheco JA, Rasmussen LV, Crosslin DR, Crane PK, Pathak J, Bielinski SJ, Pendergrass SA, Xu H, Hindorff LA, Li R, Manolio TA, Chute CG, Chisholm RL, Larson EB, Jarvik GP, Brilliant MH, McCarty CA, Kullo IJ, Haines JL, Crawford DC, Masys DR, Roden DM. Systematic comparison of phenome-wide association study of electronic medical record data and genome-wide association study data. Nat Biotechnol. 2013 Dec;31(12):1102–1110. PMID: 24270849

- Holzinger ER, Dudek SM, Frase AT, Pendergrass SA, Ritchie MD. ATHENA: the analysis tool for heritable and environmental network associations. Bioinformatics. 2013 Oct 27; PMID: 24149050
- 21. Wolfe D, Dudek S, Ritchie MD, **Pendergrass SA**. Visualizing genomic information across chromosomes with PhenoGram. BioData Min. 2013;6(1):18. PMID: 24131735
- 22. Fesinmeyer MD, Meigs JB, North KE, Schumacher FR, B Ková P, Franceschini N, Haessler J, Goodloe R, Spencer KL, Voruganti VS, Howard BV, Jackson R, Kolonel LN, Liu S, Manson JE, Monroe KR, Mukamal K, Dilks HH, Pendergrass SA, Nato A, Wan P, Wilkens LR, Marchand LL, Ambite JL, Buyske S, Florez JC, Crawford DC, Hindorff LA, Haiman CA, Peters U, Pankow JS. Genetic variants associated with fasting glucose and insulin concentrations in an ethnically diverse population: results from the Population Architecture using Genomics and Epidemiology (PAGE) study. BMC Med Genet. 2013 Sep 25;14(1):98. PMID: 24063630
- 23. Moore CB, Wallace JR, Frase AT, Pendergrass SA, Ritchie MD. BioBin: a bioinformatics tool for automating the binning of rare variants using publicly available biological knowledge. BMC Med Genomics. 2013;6 Suppl 2:S6. PMID: 23819467
- 24. Park SL, Cheng I, Pendergrass SA, Kucharska-Newton AM, Lim U, Ambite JL, Caberto CP, Monroe KR, Schumacher F, Hindorff LA, Oetjens MT, Wilson S, Goodloe RJ, Love S-A, Henderson BE, Kolonel LN, Haiman CA, Crawford DC, North KE, Heiss G, Ritchie MD, Wilkens LR, Le Marchand L. Association of the FTO Obesity Risk Variant rs8050136 With Percentage of Energy Intake From Fat in Multiple Racial/Ethnic Populations: The PAGE Study. Am J Epidemiol. 2013 Jul 2; PMID: 23820787
- 25. McGeachie MJ, Stahl EA, Himes BE, Pendergrass SA, Lima JJ, Irvin CG, Peters SP, Ritchie MD, Plenge RM, Tantisira KG. Polygenic heritability estimates in pharmacogenetics: focus on asthma and related phenotypes. Pharmacogenet Genomics. 2013 Jun;23(6):324–328. PMID: 23532052
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- Moore CB, Wallace JR, Frase AT, Pendergrass SA, Ritchie MD. Using biobin to explore rare variant population stratification. Pac Symp Biocomput. 2013;332–343. PMID: 23424138
- 28. **Pendergrass SA**, Verma SS, Holzinger ER, Moore CB, Wallace J, Dudek SM, Huggins W, Kitchner T, Waudby C, Berg R, McCarty CA, Ritchie MD. Next-generation analysis of cataracts: determining knowledge driven gene-gene interactions using Biofilter, and

gene-environment interactions using the PhenX Toolkit. Pac Symp Biocomput. 2013;147–158. PMID: 23424120

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- 30. **Pendergrass SA**, Dudek SM, Crawford DC, Ritchie MD. Visually integrating and exploring high throughput Phenome-Wide Association Study (PheWAS) results using PheWAS-View. BioData Min. 2012;5(1):5. PMID: 22682510
- 31. **Pendergrass SA**, Lemaire R, Francis IP, Mahoney JM, Lafyatis R, Whitfield ML. Intrinsic gene expression subsets of diffuse cutaneous systemic sclerosis are stable in serial skin biopsies. The Journal of investigative dermatology. 2012 May;132:1363–73.
- 32. Haiman CA, Fesinmeyer MD, Spencer KL, Buzková P, Voruganti VS, Wan P, Haessler J, Franceschini N, Monroe KR, Howard BV, Jackson RD, Florez JC, Kolonel LN, Buyske S, Goodloe RJ, Liu S, Manson JE, Meigs JB, Waters K, Mukamal KJ, Pendergrass SA, Shrader P, Wilkens LR, Hindorff LA, Ambite JL, North KE, Peters U, Crawford DC, Le Marchand L, Pankow JS. Consistent directions of effect for established type 2 diabetes risk variants across populations: the population architecture using Genomics and Epidemiology (PAGE) Consortium. Diabetes. 2012 Jun;61(6):1642–1647. PMID: 22474029
- 33. Buchanan CC, Wallace JR, Frase AT, Torstenson ES, Pendergrass SA, Ritchie MD. A Biologically Informed Method for Detecting Associations with Rare Variants. In: Giacobini M, Vanneschi L, Bush WS, editors. Evolutionary Computation, Machine Learning and Data Mining in Bioinformatics 10th European Conference, EvoBIO 2012, Málaga, Spain, April 11-13, 2012 Proceedings. 1st ed. New York: Springer; 2012.
- 34. Avery CL, He Q, North KE, Ambite JL, Boerwinkle E, Fornage M, Hindorff LA, Kooperberg C, Meigs JB, Pankow JS, Pendergrass SA, Psaty BM, Ritchie MD, Rotter JI, Taylor KD, Wilkens LR, Heiss G, Lin DY. A Phenomics-Based Strategy Identifies Loci on APOC1, BRAP, and PLCG1 Associated with Metabolic Syndrome Phenotype Domains. Gibson G, editor. PLoS Genetics. 2011 Oct 13;7(10):e1002322.
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- 37. Christmann RB, Hayes E, Pendergrass S, Padilla C, Farina G, Affandi AJ, Whitfield ML, Farber HW, Lafyatis R. Interferon and alternative activation of monocyte/macrophages in systemic sclerosis-associated pulmonary arterial hypertension. Arthritis and rheumatism. 2011 Jun;63:1718–28.
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- Pendergrass S, Dudek SM, Roden DM, Crawford DC, Ritchie MD. Visual integration of results from a large DNA biobank (BioVU) using synthesis-view. Pac Symp Biocomput. 2011;265–275. PMID: 21121054
- 40. **Pendergrass SA**, Dudek SM, Crawford DC, Ritchie MD. Synthesis-View: visualization and interpretation of SNP association results for multi-cohort, multi-phenotype data and meta-analysis. BioData Min. 2010;3:10. PMID: 21162740
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- 43. Chandriani S, Frengen E, Cowling VH, Pendergrass SA, Perou CM, Whitfield ML, Cole MD. A core MYC gene expression signature is prominent in basal-like breast cancer but only partially overlaps the core serum response. PloS one. 2009;4:e6693.
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- Townley-Tilson WH, Pendergrass SA, Marzluff WF, Whitfield ML. Genome-wide analysis of mRNAs bound to the histone stem-loop binding protein. RNA. 2006 Oct;12:1853–67.
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- 49. Meaney PM, **Pendergrass SA**, Fanning MW, Paulsen KD. Importance of using a reduced contrast coupling medium in 2D microwave breast imaging. Journal of electromagnetic waves and applications. 2003;17:333–355.
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### References

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