Cincinnati Children's
change the outcome

Human Genetics

Division Photo

First Row: E. Schorry, G. Grabowski, H. Saal, K. Zhang, N. Leslie; Second Row: N. Warren, T. Smolarek, H. Du, L. Martin, S. Ware, I. Sageser, Y. Sun, D. Prows, S. Zimmerman, T.A. Burrow, Y.H. Xu; Third Row: M.G. Guan, L. Bao, R. Hopkin, X. Qi

Division Data Summary

Research and Training Details	
Number of Faculty	23
Number of Joint Appointment Faculty	4
Number of Research Fellows	3
Number of Support Personnel	125
Direct Annual Grant Support	$2,240,815
Direct Annual Industry Support	$548,871
Peer Reviewed Publications	65
Clinical Activities and Training	
Number of Clinical Staff	26
Number of Clinical Fellows	2
Number of Clinical Students	9
Number of Other Students	7
Inpatient Encounters	312
Outpatient Encounters	4,893

Significant Publications

Li. R., Liu, Y., Li, Z., Yang, L., Wang, S., Guan, MX.: Failures in mitochondrial tRNAGln metabolism caused by the novel 4401A>G mutation are involved in essential hypertension in a Han Chinese Family. Hypertension. 2009 Aug;54(2):329-337.

First mitochondrial mutations in essential hypertension.

Du, H., Cameron, T.L., Garger, S.J., Pogue, G.P., Hamm, L.A., White, E., Hanley, K.M., Grabowski, G.A.: Wolman disease/cholesteryl ester storage disease:efficacy of plant-produced human lysosomal acid lipase in mice. J Lipid Research. 2008;49:1646-1657.

This paper described pharmacokinetics/pharmacodynamics, therapeutic effect of plant-produced human lysosomal acid lipase (LAL) to treat Wolman disease and cholesteryl ester storage disease in preclinical mouse model. It provides a strong foundation to pursue the next step to initiate clinical trials of LAL enzyme therapy to treat cholesteryl ester storage disease.

Zhang, K., Biroschak, J., Glass, D.N., Thompson, S., Finkel, T., Passo, M.H., Filipovich, A., Grom, A.A.: Macrophage Activation Syndrome in patients with Systemic Juvenile Idiopathic Arthritis Is Associated with MUNC13-4 Gene Polymorphisms. Arthritis and Rheumatism. 2008 Sep;58(9):2892-2896.

It is the first peer reviewed publication that identified a potential genetic cause of Macrophage Activation Syndrome in patients with Systemic Juvenile Idiopathic Arthritis.

Neilson, D.E., Adams, M.D., Orr, C.M., Schelling, D.K., Eiben, R.M., Kerr, D.S., Anderson, J.: Infection triggered familial or recurrent cases of acute necrotizing encephalopathy caused by mutations in a component of the nuclear pore, RANBP2. Am J Hum Genet. 2009;84:44-51.

This paper describes the first mutations in the RANBP2 gene that are causative for a human disease.

Hopkin, R.J., Bissler, J., Banikazemi, M., Clarke, L., Eng, C.M., Germain, D.P., Lemay, R., Tylki-Symanska, A., Wilcox, W.R.: Characterization of Fabry disease in 352 pediatric patients in the Fabry Registry. 2008 Nov;64(5):550-555.

This is the largest series of pediatric patients with Fabry disease published to date. We were able to demonstrate an affect of Fabry disease on growth in children and to document the low incidence of active renal and cardiac dysfunction in this population. This contributed directly to the development of recommended monitoring guidelines for children with Fabry disease that have just been released.

Division Highlights

Daniel R. Prows, PhD

Our large-scale project assessing the genetic components of ozone-induced acute lung injury survival has made significant gains in the past year. We have initiated reciprocal cogenic lines for *Aliq2* (chromosome 13), a major region of linkage, and continue to further refine this region, along with *Aliq1* (on chromosome 11) by generating subcogenic lines, which contain sub-segments of the larger validated areas on chromosomes 11 and 13.

Elizabeth K. Schorry, MD

The natural history of spinal abnormalities in NF1 was evaluated in 30 patients from our center and have contributed significantly to the development and implementation of an NIH project.

William C. Nichols, PhD

We have continued our studies to identify novel genes for pulmonary hypertension (PH) through the use of mouse models exposed to chronic hypoxia. A major focus is a strain survey to identify differences in susceptibility to hypoxia-induced PH in common inbred strains. Most notably, we have idenitified the PL/J strain as being extremely sensitive to large increases in both right ventricular systolic pressure and RV/LV+S after chronic hypoxia. A total of 18 strains have now been surveyed. Strains have been identified which can serve as models for genetic mapping of loci contributing to hypoxia-induced PH. We are in the process of analyzing F1 animals for four different genetic models to determine any parent of origin effects and those models most appropriate for the genetic mapping studies.

You-hai Xu, MD, PhD

Global and macrophage activation gene expressions in Gaucher mice. To understand the molecular pathogenesis of GD, developmental global gene expression was examined by microarray analyses of total mRNAs from lung and liver of GCase point-mutated V394L/V394L (4L) and D409V/null (9V/null) mice. 2-3% of total 45,101 proble sets were determined as significantly differentially expressed. Among them, 13 INFy regulated pro-inflammatory and 11 IL-4 regulated anti-inflammatory cytokine/mediator genes were consistently altered during disease progress (12 to 18 weeks) in a tissue (lung > liver) and mutation type (9V/null>4L) correlated manner. A cascade of cross-interactive gene network was built using macrophage activation genes as a core network and substantial numbers of genes (411 in 9V/null lung and 237 in 4L lung) were cross-interacted, which indicates altered global and macrophage activation gene expressions in Gaucher mice.

Nancy D. Leslie, MD

Newborn screening for Duchenne Muscular Dystrophy is being expanded throughout Ohio. Our group is charged with finding ways to educate and provide the opportunity for consent in a setting removed from the busy peripartum period.

Ying Sun, PhD

Evaluated *ex vivo* and *in vivo* effect of LTCC blocker in Gaucher model. LTCC blocker, diltiazem, enhanced GCase protein, activity, and lysosomal trafficking in GCase mutant fibroblast cells. It increased GCase protein level but no activity *in vivo*, which limited its clinical application.

Division Collaboration

Collaboration with Neurology

Collaborating Faculty: deGrauw, T

Dr. T. Andrew Burrow is currently working with Dr. Ton deGrauw in Neurology to establish a neurometabolic clinic. This will be particularly significant as it will improve the care of children with these diseases.

Collaboration with Cardiology

Collaborating Faculty: Benson, W; Hinton, R

Dr. Lisa Martin is a co-investigator of a P50 and K24 grant in which Dr. Benson is looking for genes related to bicuspid aortic valves and related heart malformations. They have identified 1 locus significantly linked to BAV and 2 other loci with promising evidence. They are currently attempting to narrow the region of interest. Dr. Martin is also a co-investigator of a K23 held by Dr. Hinton which is looking for genes related to hypoplastic left heart. In the last year, they have published the first linkage study of HLHS and have identified 2 promising regions of linkage. Given these promising findings they have submitted a grant in response to an RFA which could continue this interesting line of work.

Collaboration with Asthma Research

Collaborating Faculty: Hershey, N

With the departure of Dr. Todd Nick, Dr. Lisa Martin has assumed the responsibility of the statistical support of Dr. Hershey's U01 on the genetics of asthma. They have made tremendous progress and have identified several SNPs in genes highly associated with pediatric asthma.

Collaboration with Allergy

Collaborating Faculty: Rothenberg, M

For the last year, Dr. Lisa Martin has been collaborating with Dr. Rothenberg and his group on the genetics of eosinophillic esophagitis. Dr. Martin has been running analyses of the genome wide association data generated at CHOP and they are currently drafting a manuscript of their findings. They have also submitted a grant to the DOD to expand their analyses.

Collaboration with Endocrinology

Collaborating Faculty: Dolan, L

Dr. Lisa Martin is working with several of Dr. Dolan's fellows on research projects related to obesity genetics. They are currently in the analysis phase of the project.

Collaboration with Neurology

Collaborating Faculty: Molly, C

Dr. Lisa Martin is working with Dr. Molloy on the genetics of autism as part of a grant from Austism Speaks. Dr. Martin is performing all the statistical analysis including a genome wide association.

Collaboration with Nephrology

Collaborating Faculty: Hooper, D

Dr. Lisa Martin is working with Dr. Hooper on a project looking for genetic variants which are associated with Mycophenolate Mofetil Related Complications in Pediatric Transplant Recipients. Dr. Martin is working with Dr. Hooper on the statistical design and will perform the analyses.

Collaboration with Neurology

Collaborating Faculty: Gilbert, D

Dr. Robert Hopkin is collaborating with Dr. Gilbert regarding the Transcranial Magnetic Stimulation in patients with NF1. This is being done to better characterize the brain function of children with NF1. There is CCHMC grant funding to support this preliminary project. They hope to move on to a larger project based on the results of this study. Comparisons will be made with general population controls and children with attention deficit disorder. This technique has not been used for patients with NF1 in the past.

Collaboration with Cardiology

Collaborating Faculty:

Dr. Robert Hopkin has also had a small role in an ongoing collaboration with the Division of Cardiology in the SVR project. This project is comparing two different surgical techniques for management of hypoplastic left heart. Dr. Hopkin's role is to evaluate the possible syndromes or genetic conditions.

Collaboration with Immunobiology

Collaborating Faculty: Wills-Karp, M; Inoue, Y

Dr. Ying Sun has been collaborating with the Division of Immunobiology to characterize the macrophages in Gaucher disease mouse models.

Collaboration with Neurology

Collaborating Faculty: Williams, MT; Skelton, MR; Vorhees, CV

Dr. Ying Sun has been collaborating with Neurology regarding the neurobehavioral assessment of saposin deficiency and neuronopathic type Gaucher disease models.

Collaboration with Hematology Oncology

Collaborating Faculty: Geller, J

Dr. Nancy Leslie and Dr. James Geller have launched a Pediatric Cancer Predisposition Clinic together. This provides a referral and follow up home for individuals who are at risk for cancer but do not yet have it. In addition, the interaction has spurred several potential research opportunities, currently being pursued along with colleagues in cytogenetics.

Collaboration with Neurology

Collaborating Faculty: Rose, D

Dr. Nancy Leslie is working with Dr. Douglas Rose to look at MEG findings in individuals with Angelman syndrome, and to correlate this interesting neurophenotype with the molecular basis of Angelman, including haploinsufficiency at GABR3.

Collaboration with Cardiology

Collaborating Faculty: Strauss, A

Dr. Nancy Leslie has collaborated with Dr. Arnold Strauss to develop analytical tools to support a clinical trial of bezafibrate in patients with fatty acid oxidation disorders.

Collaboration with Immunobiology

Collaborating Faculty: Wills-Karp, M

Dr. You-hai Xu is working on a collaborative study with Dr. Marsha Wills-Karp on lung macrophage activation in D409V/null Gaucher mice.

Collaboration with Hematology Oncology; Experimental Hematology; Orthopaedic Surgery

Collaborating Faculty: Perentesis, J; Weiss, B; Ratner, N; Crawford, A

Dr. Elizabeth Schorry has been collaborating with multiple divisions due to the multidisciplinary nature of our Neurofibromatosis Center. Drs. John Perentesis and Brian Weiss are also members of the NF Consortium, and have played major roles in the development and implementation of the drug trials for plexiform neurofibromas, optic nerve gliomas, and MPNST. Our research efforts are greatly enhanced by input from Dr. Nancy Ratner who provides basic science expertise as the basis for our clinical trials. She also interacts closely with Dr. Alvin Crawford on clinical studies involving bone complications of NF1.

Collaboration with Developmental and Behavioral Pediatrics; Pediatric Otolaryngology; Biostatistics; Molecular and Developmental Biology

Collaborating Faculty: Oppenheimer, S; Parrish, R; Greinwald, J; Martin, L; Tabangin, M; Benson, W; Ratner, N

Nancy Warren has collaborated with faculty in multiple divisions to launch several genetic counseling program thesis projects.

Collaboration with Pastoral Care

Collaborating Faculty:

Nancy Warren is also working with the Department of Pastoral Care on her JEMF project which focuses on enhancing cultural and linguistic competence in genetic counseling.

Collaboration with Pulmonary Biology

Collaborating Faculty: Le Cras, T; Akinbi, H; Akeson, A

Dr. William Nichols has been working with Dr. Tim Le Cras on the development of pulmonary hypertension after exposure to house dust mite allergen in mice. He has also been working with Drs. Akinbi and Akeson regarding lung development in mice under hypoxic conditions.

Collaboration with Nephrology

Collaborating Faculty: Patterson, L

Dr. William Nichols has been working with Dr. Larry Patterson regarding kidney development in mice under hypoxic conditions.

Collaboration with Experimental Biology and Cancer Biology

Collaborating Faculty: Malik, P

Dr. William Nichols has collaborated with Dr. Punam Malik regarding the development of pulmonary hypertension in sickle cell mice.

Collaboration with Hematology Oncology

Collaborating Faculty: Filipovich, AH; Bleesing, J; Marsh, R

Dr. Kejian Zhang has partnered with the Program of Immunodeficiency in the Division of Hematology/Oncology to establish the Diagnostic Center for Heritable Immunodeficiencies. This is now a nationally recognized clinical diagnostic center.

Collaboration with Pediatric Otolaryngology

Collaborating Faculty: Greinwald, J

Dr. Kejian Zhang has partnered with Dr. John Greinwald in the development of genetic testing for hearing loss.

Collaboration with Neurology; Clinical Pharmacology

Collaborating Faculty: Glauser, TA; Vinks, AS

Dr. Kejian Zhang has partnered with Drs. Glauser and Vinks to provide genetic testing to assess drug metabolism.

Collaboration with Hematology Oncology

Collaborating Faculty: Davies, S; Bleesing, J

Dr. Kejian Zhang is in collaboration with Drs. Davies and Bleesing to develop molecular testing for bone marrow engraftment monitoring. Most recently, they introduced sub-cell-type BME assay, which allows physicians to look at the cell engraftment at different cell populations.

Collaboration with Hematology Oncology

Collaborating Faculty: Gruppo, R

Dr. Kejian Zhang is working with Dr. Gruppo regarding molecular testing for thrombosis.

Collaboration with Pediatric Gastroenterology, Hepatology and Nutrition

Collaborating Faculty: Bezerra, JA

Dr. Kejian Zhang is in collaboration with Dr. Bezerra to introduce the "The Jaundice Chip," a microarray based molecular test for children and adults with heritable liver diseases.

Collaboration with Neurology

Collaborating Faculty: Gilbert, D

Drs. Derek Neilson and Nancy Leslie have collaborated with Dr. Don Gilbert regarding the mutation discovery process for the neurologic disorder, Hereditary Spastic Paraplegia and Dystonia.

Collaboration with Neurology

Collaborating Faculty: Glauser, TA

Mehdi Keddache and Dr. Tracy Glauser are working on publishing the results of the five year CAE study.

Collaboration with Pulmonary Biology

Collaborating Faculty: Glasser, S

Dr. Daniel Prows has made considerable progress on a collaborative project with Dr. Steve Glasser over this past year. They have completed the genetic purification of two lines of surfactant protein-C (SP-C) "knockout mice" on the 129S6/SvEvTac and FVB/NJ inbred strains.

Collaboration with Mayerson Center for Safe and Healthy Children

Collaborating Faculty: Shapiro, B; Makoroff, K; Putnam, FW

Dr. Bradley Tinkle has collaborated with Drs. Shapiro, Makoroff and Putnam to screen for bone fragility issues in those infants/young children with "unexplained" multiple fractures.

Collaboration with Orthopaedics

Collaborating Faculty: Jain, V; Do, T

Dr. Bradley Tinkle has collaborated with Drs. Jain and To to better coordinate the management of skeletal dysplasias.

Collaboration with Pediatric and Thoracic Surgery

Collaborating Faculty: Crawford, K

Dr. Bradley Tinkle is participating in the Spine Center with Dr. Kelly Crawford to screen for, discuss, and help manage patients with spinal disorders as part of an underlying syndromic etiology.

Collaboration with Cardiology

Collaborating Faculty: Meyer, R

Dr. Bradley Tinkle is working with Dr. Meyer in the Marfan/EDS clinic held in Cardiology to carefully assess and monitor aortic dilatation commonly seen in this patient population.

Collaboration with Fetal Care Center

Collaborating Faculty: Crombleholme, T

Dr. Bradley Tinkle and Dr. Timothy Crombleholme provide fetal diagnostic services and expectant management.

Collaboration with Pulmonary Medicine

Collaborating Faculty: Ednick, M; Simakajornboon, N

Dr. Bradley Tinkle and Drs. Ednick and Simakajornboon analyzed a cluster of achondroplasia patients to better describe the altered sleep architecture. This resulted in three scientific posters and a manuscript in press.

Collaboration with Pulmonary Medicine

Collaborating Faculty: Chini, B

Iris Sageser and Dr. Barbara Chini are now collaborating together in our Craniofacial Center. Dr. Chini is a participant in the Craniofacial Center Registry and will evaluate treatment outcomes.

Collaboration with Molecular Cardiobiology

Collaborating Faculty: Hinton, R

Dr. Teresa Smolarek and Dr. Robert Hinton have collaborated on the characterization of chromosomal breakpoints.

Collaboration with Molecular Cardiobiology

Collaborating Faculty: Ware, S

Drs. Teresa Smolarek and Stephanie Ware are using the SNP microarray to identify DNA copy number changes in patients with heterotaxy.

Collaboration with Hematology Oncology

Collaborating Faculty: Mehta, P

Dr. Teresa Smolarek is working with Dr. Parinda Mehta regarding chromosome abnormalities in Fanconi Anemia.

Collaboration with Neonatology and Pulmonary Biology

Collaborating Faculty: Suzuki, T; Trapnell. B

Dr. Sarah Zimmerman is working with Drs. Suzuki and Trapnell on a research project that resulted in a paper published in The Journal of Experimental Medicine.

Collaboration with Cardiology

Collaborating Faculty: Hinton, R

Dr. Sarah Zimmerman has worked with Dr. Hinton on two different projects. The first project was to see if there were any chromosome abnormalities in a patient and father using SNP array technology and then characterize the breakpoints if one was identified. The second project was to determine if the father, which is a carrier of a balanced translocation had small deletion or duplications near the breakpoints to determine if there were any important genes within this region.

Faculty Members

Gregory Grabowski, MD, Professor ; *The A. Graeme Mitchell Chair in Human Genetics; Director, Division of Human Genetics; Professor of Pediatrics and Molecular Genetics, Biochemistry and Microbiology*
Research Interests: Molecular pathogenesis and therapy of human genetic disease

Liming Bao, MD, PhD, Associate Professor Clinical ; *Associate Director, Clinical Cytogenetics Laboratory*
Research Interests: Genetic biomarkers of hematological diseases and their underlying pathogenesis and clinical relevance

T. Andrew Burrow, MD, Assistant Professor Clinical
Research Interests: Lysosomal storage diseases, particularly Gaucher disease; Inborn errors of metabolism, particularly focusing on development of evidence based medicine

Hong Du, PhD, Research Associate Professor
Research Interests: Understanding the molecular mechanisms of genetic disorders of lipid metabolism and the development of therapeutic treatments for these disorders

Min-Xin Guan, PhD, Associate Professor
Research Interests: Mechanisms of mitochondrial disorders, with a focus on maternally transmitted hearing loss and vision loss

Robert Hopkin, MD, Associate Professor Clinical ; *Director, Genetic Residency Programs*
Research Interests: Fabry Disease; Robin Sequence; 22q11 deletion; Neurofibromatosis; craniofacial genetics; chromosomal anomalies

Mehdi Keddache, MS, Research Instructor ; *Leader, Genomics Core*
Research Interests: DNA Sequencing / Genotyping Core

Nancy Doan Leslie, MD, Professor Clinical ; *Director, Biochemical Genetics Laboratory; Director, Medical Biochemical Genetics Fellowship; Program Director, Laboratory Fellowships*
Research Interests: Inborn errors of metabolism, with an emphasis on long term outcome in PKU and in the molecular biology of galactosemia

Derek Neilson, MD, Assistant Professor
Research Interests: The genetic contribution to acute necrotizing encephalopathy, a disorder in which children are predisposed to devastating neurologic injury following common infections

William Nichols, PhD, Associate Professor ; *Chairman, DHG Research Review Committee*
Research Interests: The identification of genetic variants contributing to disease susceptibility

Cindy Prows, MSN, CNS, RN, FAAN, Adjunct Associate Professor
Research Interests: Developing and testing education models to teach nurses about genetics; family responses to genetic information, technology and services

Daniel R Prows, PhD, Assistant Professor
Research Interests: Mouse models of complex human diseases, with specific interest in mouse models of acute lung injury; use of quantitative trait locus analysis to identify regions linked to complex traits

Xiaoyang Qi, PhD, Research Associate Professor
Research Interests: Translational research focused on the role of saposin C in multivesicular body bioformation and neuropathogenesis and the development of saposin C-containing nanovesicles as a novel anticancer agent

Howard Saal, MD, Professor Clinical ; *Director, Clincal Genetics; Medical Director, Cytogenetics Laboratory; Director, Cincinnati Children's Craniofacial Center*
Research Interests: The natural history of genetic disorders, especially as they relate to craniofacial disorders; developing treatment and management protocols for craniofacial disorders

Iris Sageser, RDH, MS, Field Service Assistant Professor ; *Craniofacial Center*
Research Interests: Multidisciplinary management of individuals affected by craniofacial abnormalities

Elizabeth K Schorry, MD, Associate Professor Clinical
Research Interests: Psychosocial and orthopedic aspects of neurofribromatosis; Clinical drug trials for NF1

Teresa A Smolarek, PhD, Associate Professor Clinical ; *Director, Clinical Cytogenetics Laboratory; Director, Clinical Cytogenetics Fellowship Program*
Research Interests: Application of SNP microarrays to determine constitutional and acquired DNA copy number changes; the genetic basis of pulmonary lymphagioleiomyomatosis

Ying Sun, PhD, Research Associate Professor
Research Interests: The pathological mechanisms of lysosomal storage diseases

Bradley T Tinkle, MD, Assistant Professor Clinical ; *Clinical Geneticist; Assistant Director, Molecular Genetics Laboratory; Director, Skeletal Dysplasia Center; Co-Director, Marfan/Ehlers-Danlos Syndromes Clinic*
Research Interests: Natural history of connective tissue disorders and outcome studies of various clinical interventions

Nancy Warren, MS, Adjunct Associate Professor ; *Program Director, Genetic Counseling Graduate Program*
Research Interests: Genetic counseling education, professional development and cultural competence

You-hai Xu, MD, PhD, Research Assistant Professor
Research Interests: Molecular and pathophysiological mechanisms of Gaucher Disease, particularly of neuronopathic Gaucher Disease

Kejian Zhang, MD, Assistant Professor Clinical ; *Director, Molecular Genetics Laboratory*
Research Interests: Molecular defects and molecular diagnosis of primary immunodeficiency diseases; Genetic aspects of predictive personalized medicine, eg., Pharmacogenetics; Fanconi Anemia SeqChip development

Sarah Zimmerman, PhD, Assistant Professor Clinical ; *Assistant Director, Clinical Cytogenetics Laboratory*
Research Interests: Genome-wide analysis for chromosome aberrations in congenital disorders and cancer using molecular cytogenetic techniques

Joint Appointment Faculty Members

John Greinwald, MD, Associate Professor
Otolaryngology
Genetics of Hearing Loss

Lisa Martin, PhD, Research Associate Professor
Biostatistics and Epidemiology
Focus on common complex diseases including obesity and heart malformations

Melanie Myers, PhD, MS, CGC, Adjunct Assistant Professor
Genetics Counseling Program, College of Medicine
Clinical utility of family health history and other genomic tools in health promotion

Stephanie Ware, MD, PhD, Assistant Professor
Molecular Cardiovascular Biology
Genetic disorders of cardiac structure and function

Clinical Staff Members

- **Erin Acra, MS**
- **Carrie Atzinger, MS**
- **Laurie Bailey, MS**
- **Angela Bedard, MS**
- **Judy Belli, RN**
- **Patricia Bender, RN, MSN**
- **Lisa Berry, MS**
- **Kathleen Collins, MS**
- **Kimberly Edge, MS**
- **Carol Hetteburg, RN, MSN**
- **Judy Johnson, MS**
- **Sandy Kaiser, LPN**
- **Sara Knapke, MS**
- **Anne Lovell, RN, MSN, APN**
- **Erin Miller, MS**
- **Elizabeth Peach, MS**
- **Jodie Rueger, MS**
- **Jennifer Ruschman, SCM**
- **Kerry Shooner, MS**
- **Christine Spaeth, MS**
- **Martha Walker, MS**
- **Connie Wehmeyer, RN**
- **Katie Wusik, MS**

Trainees

- **Jaya George-Abraham, MD,** PGY4, Medical Genetics Fellowship
- **Stephanie Peters Santoro, MD,** PGY1, Pediatrics/Genetics Combined Residency
- **Carlos Prada, MD,** PGY3, Pediatrics/Genetics Combined Residency
- **Elizabeth Sellars, MD,** PGY1, Pediatrics/Genetics Combined Residency
- **Dipesh Tamakuwala, PhD,** Clinical Cytogenetics Fellowship
- **K. Nicole Weaver, MD,** PGY1, Pediatrics/Genetics Combined Residency
- **Wenying Zhang, MD, PhD,** Clinical Molecular Genetics Fellowship

Significant Accomplishments

Recruitment Success

Dr. Robert Hopkin, Director of the Medical Genetics Residency Program, succeeded in recruiting three residency postions, one straight medical genetics resident and two combined pediatrics/medical genetics residents. This was in a year when nationally, only one other combined pediatric/medical genetics resident matched a single resident.

Gaucher Disease

Drs. Gregory Grabowski and Ying Sun used a newly developed neuronopathic model of Gaucher disease to evaluate for the first time, pharmacological chaperones for the treatment of malfolded lysosomal enzymes *in vivo*. The results show that selected chaperones increased the Gaucher disease protein and activity, suppressed the proinflammation, and extended life span, but had no effect on substrate accumulation.

Dr.William Nichols extended the association of variants in the Gaucher disease gene and susceptibility to Parkinson disease. Two polymorphisms variants lower the age of disease onset in PD patients heterozygous for such variants. Additional associations of parkin gene variant heterozygosity and PD susceptibility were shown for an exonic deletion/duplication of the parkin locus and increased susceptibility to PD compared to those heterozygous for a non-synonymous variant.

Acute Necrotizing Encephalopathy

Dr. Derek Neilson, a newly recruited junior faculty, identified mutations in the RANBP2 in over a dozen families with acute necrotizing encephalopathy (ANE). This has established the first genetic cause for this childhood disorder and may hold insights applicable to other neurodegenerative diseases.

Division Publications

1. Miller E, Warren NS. (2009) The impact of a career ladder in genetic counseling on professional development. Perspect Genet Couns. Chicago, National Society of Genetic Counselors. 30:
2. Prows CA. "New models for service delivery: Cincinnati Children's Hospital Medical Center." *Innovations in service delivery in the age of genomics: workshop summary.* Washington, DC: National Academies Press; 2009: 27-31.
3. Wallace J, Baugh C, Cornett S, Hood B, Prows C, Ryan N, Warren N, Au M, Brewster R, Brown MK, Glandorf K, Jarrell J, Sorrell J, Walters J, Myers M. A family history demonstration project among women in an urban Appalachian community. *Prog Commun Health Partnerships Res Educ Action.* 2009; 3: 155-163.
4. Leslie ND, Tinkle BT, Strauss AW, Shooner K, Zhang K. (2009) Very long chain acyl-coenzyme A dehydrogenase deficiency. GeneReviews. Seattle, WA, University of Washington.
5. Black JH, Braverman AC, Byers P, Oderich G, Sundt T, Tinkle B, Wyse P. (2009) **Vascular type: medical resource guide.** Los Angeles, CA, Ehlers-Danlos National Foundation.
6. Ding Y, Li Y, You J, Yang L, Chen B, Lu J, Guan MX. Mitochondrial tRNA(Glu) A14693G variant may modulate the phenotypic manifestation of deafness-associated 12S rRNA A1555G mutation in a Han Chinese family. *J Genet Genomics.* 2009; 36: 241-50.
7. Mistry PK, Weinreb NJ, Brady RO, Grabowski GA. Gaucher disease: resetting the clinical and scientific agenda. *Am J Hematol.* 2009; 84: 205-7.
8. Stevenson DA, Viskochil DH, Carey JC, Slater H, Murray M, Sheng X, D'Astous J, Hanson H, Schorry E, Moyer-Mileur LJ. Tibial geometry in individuals with neurofibromatosis type 1 without anterolateral bowing of the lower leg using peripheral quantitative computed tomography. *Bone.* 2009; 44: 585-9.
9. Warren NS, Ormond KE. Diversity in genetic counseling: past, present and future. *J Genet Couns.* 2009; 18: 197-

9.
10. Grabowski GA, Kacena K, Cole JA, Hollak CE, Zhang L, Yee J, Mistry PK, Zimran A, Charrow J, vom Dahl S. Dose-response relationships for enzyme replacement therapy with imiglucerase/alglucerase in patients with Gaucher disease type 1. *Genet Med.* 2009; 11: 92-100.
11. Wang S, Ware SM. Use of FOXJ1CreER2T mice for inducible deletion of embryonic node gene expression. *Genesis.* 2009; 47: 132-6.
12. Whitley CB, Barranger JA, Eng CM, Davidson BL, Grabowski GA, Kohler B, Muenzer J, Murray GJ, Pastores GM, Patel SK, Shapiro EG, Steiner RD, Walkley SU, Wedehase BA, Wilcox WR. Lysosomal Disease Network's "WORLD Symposium 2009". Introduction. *Mol Genet Metab.* 2009; 96: S3-5.
13. Qu P, Du H, Li Y, Yan C. Myeloid-specific expression of Api6/AIM/Sp alpha induces systemic inflammation and adenocarcinoma in the lung. *J Immunol.* 2009; 182: 1648-59.
14. Gilbert DL, Leslie EJ, Keddache M, Leslie ND. A novel hereditary spastic paraplegia with dystonia linked to chromosome 2q24-2q31. *Mov Disord.* 2009; 24: 364-70.
15. Neilson DE, Adams MD, Orr CM, Schelling DK, Eiben RM, Kerr DS, Anderson J, Bassuk AG, Bye AM, Childs AM, Clarke A, Crow YJ, Di Rocco M, Dohna-Schwake C, Dueckers G, Fasano AE, Gika AD, Gionnis D, Gorman MP, Grattan-Smith PJ, Hackenberg A, Kuster A, Lentschig MG, Lopez-Laso E, Marco EJ, Mastroyianni S, Perrier J, Schmitt-Mechelke T, Servidei S, Skardoutsou A, Uldall P, van der Knaap MS, Goglin KC, Tefft DL, Aubin C, de Jager P, Hafler D, Warman ML. Infection-triggered familial or recurrent cases of acute necrotizing encephalopathy caused by mutations in a component of the nuclear pore, RANBP2. *Am J Hum Genet.* 2009; 84: 44-51.
16. Pankratz N, Wilk JB, Latourelle JC, DeStefano AL, Halter C, Pugh EW, Doheny KF, Gusella JF, Nichols WC, Foroud T, Myers RH. Genomewide association study for susceptibility genes contributing to familial Parkinson disease. *Hum Genet.* 2009; 124: 593-605.
17. Nichols WC, Pankratz N, Marek DK, Pauciulo MW, Elsaesser VE, Halter CA, Rudolph A, Wojcieszek J, Pfeiffer RF, Foroud T. Mutations in GBA are associated with familial Parkinson disease susceptibility and age at onset. *Neurology.* 2009; 72: 310-6.
18. Ahmed F, Osman N, Lucas F, Neff G, Smolarek T, Bennett JM, Komrokji RS. Therapy related CMML: a case report and review of the literature. *Int J Hematol.* 2009; 89: 699-703.
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Grants, Contracts, and Industry Agreements

Grant and Contract Awards — Annual Direct / Project Period Direct

DU, H

PPARgamma in the Lung
National Institutes of Health
R01 HL 087001 — 06/09/08 - 05/31/13 — $250,000 / $1,250,000

GRABOWSKI, G

Cincinnati Regional Genetics Center
Ohio Department of Health
31-3-001-1GS0209 — 07/01/08 - 06/30/09 — $383,500 / $383,500

Studies of Gaucher Disease
National Institutes of Health

R01 DK 036729 09/27/07 - 08/31/12 $276,902 / $1,449,180

Studies of Gaucher Disease
National Institutes of Health
R01 DK 036729 (supplement) 06/15/09 - 12/31/09 $5,000 / $5,000

Therapy of Neuronopathic Gaucher Disease
National Institutes of Health
R21 HD 059823 01/10/09 - 12/31/10 $100,000 / $275,000

Therapy of Gaucher Disease: In Vivo Enhancement
National Institutes of Health
R21 NS 064352 05/15/09 - 04/30/11 $126,883 / $275,000

Digestive Health Center: Bench to Bedside Research in Pediatric Digestive Disease
National Institutes of Health
104571 08/01/07 - 05/31/12 $21,243 / $106,215

GUAN, M

Nuclear Modifier Genes for Maternally Inherited Deafness
National Institutes of Health
R01 DC 007696 07/01/07 - 06/30/11 $209,738 / $850,000

Nuclear Modifier Genes for Maternally Inherited Deafness (Summer Supplement)
National Institutes of Health
R01 DC 007696 06/01/09 - 10/31/10 $10,000 / $10,000

KNAPKE, S

Hereditary Breast Cancer: BRCA and Beyond
Komen Breast Cancer Foundation
04/01/09 - 03/31/10 $47,045 / $47,045

NEILSON, D

Genetic Mechanism of Acute Necrotizing Encaphalopathy
National Institutes of Health
K08 NS 050331 09/01/08 - 06/30/10 $156,412 / $250,954

PROWS, D.

Genetic Analysis of Hyperoxia-Induced Acute Lung Injury
National Institutes of Health
R01 HL 075562 05/01/09 - 04/30/13 $295,265 / $1,116,554

QI, X

SapC-DOPS Nanovesicles for Treating Glioblastoma Multiform
National Institutes of Health (Bexion Pharmaceuticals)
R43 CA 136017 09/25/08 - 06/24/09 $97,200 / $97,200

A Novel Biotherapeutic Treatment for Pancreatic Cancer
National Institutes of Health (Bexion Pharmaceuticals)
R43 CA 117283 09/25/08 - 06/24/09 $32,649 / $32,649

RUSCHMAN, J

Puberty and Cancer Initiation: Environment Diet and Obesity
National Institutes of Health (University of Cincinnati)
U01 ES 012770 08/01/08 - 07/31/10 $4,210 / $17,461

SCHORRY, E

Spinal Abnormalities in Neurofibromatosis Type 1
National Institutes of Health (University of Utah)
R01 NS 050509 07/01/06 - 03/31/11 $46,168 / $333,163

Neurofibromatosis Consortium Development Operation
Department of Defense - Army (University of Alabama at Birmingham)
W81XWH-05-1-0615 07/01/07 - 03/31/10 $24,754 / $104,791

Neurofibromatosis Consortium Development PK Center STOPN
Department of Defense - Army (University of Alabama at Birmingham)
W81XWH-05-1-0615 07/01/08 - 06/30/11 $67,840 / $271,360

Neurofibromatosis Consortium Development STOPn Protocol - Laboratory
Department of Defense - Army (University of Alabama at Birmingham)
W81XWH-05-1-0615 07/01/08 - 06/30/11 $66,283 / $265,132

LESLIE, N

Implementing Newborn Screening for Duchenne Muscular Dystrophy in the Community
National Institutes of Health
104471 09/01/07 - 08/31/10 $19,723 / $62,028

Current Year Direct **$2,240,815**

Industry Contracts

Grabowski
Genzyme Corporation $ 59,565
Shire Human Genetic Therapies $ 413,608

Hopkin
Genzyme Corporation $ 39,971

Leslie
Amicus Therapeutics, Inc. $ 40,265
Genzyme Corporation $ 40,425

Qi
Bexion Pharmaceticals $ 14,602

Current Year Direct Receipts **$548,871**

Funded Collaborative Efforts

ZHANG, K

Macrophage Activation Syndrome Biomarkers in Systemic Juvenile Idiopathic Arthritis
National Institutes of Health
GROM, A 08/18/08 - 07/31/13 5 %

Total 2,789,686