

Division of Human Genetics

DIVISION PROFILE	
Number of Faculty	20
Number of Joint Appointment Faculty	6
Number of Fellows	
Clinical Fellows	4
Research Fellows	4
Number of Graduate Students	16
Number of Other Students (full and part-time)	3
Number of Support Personnel	120
Annual Total Grant Support (direct)	\$3,006,178
Annual Total Industry Contracts (direct)	\$373,760
Number of Peer Reviewed Publications	84
Patient Encounters	
Outpatient	3,959
Inpatient	312

FACULTY LISTING

Gregory A. Grabowski, MD, Professor, Division Director, The A. Graeme Mitchell Chair in Human Genetics; Head, Medical Genetics Fellowship Programs; Director, STAR Lysosomal Disease Center

Liming Bao, MD, PhD, Assistant Professor Clinical, Associate Director, Cytogenetics Laboratory

Hong Du, PhD, Research Assistant Professor

Min-Xin Guan, PhD, Associate Professor, Mitochondrial Genetics Research

Robert Hopkin, MD, Assistant Professor Clinical, Head, Toledo Children's Outreach Genetics Program; Director, Medical Genetics Residency Program

Mehdi Keddache, MS, Research Scientist, Leader, DNA Sequencing/Genotyping Core

Nancy Leslie, MD, Associate Professor Clinical, Director, Clinical Operations; Director, Metabolic Service

William Nichols, PhD, Associate Professor, Co-Chair, Research Review Committee

Cindy Prows, MSN, CNS, Adjunct Associate Professor, College of Nursing, Head, Nursing Genetics Education

Daniel Prows, PhD, Assistant Professor, Quantitative Mouse Genetics

Xiaoyang Qi, PhD, Assistant Professor

Howard Saal, MD, Professor, Senior Associate Director, Cytogenetics Laboratory; Director, Clinical Services

Elizabeth Schorry, MD, Associate Professor Clinical, Head, Neurofibromatosis Clinics

Teresa Smolarek, PhD, Assistant Professor Clinical, Assistant Director, Cytogenetics Laboratory

Ying Sun, PhD, Research Assistant Professor

Bradley Tinkle, MD, PhD, Assistant Professor Clinical, Assistant Director, Molecular Genetics Laboratory

Nancy Steinberg Warren, MS, Adjunct Associate Professor, College of Applied Health Sciences, Director, Genetic Counseling Graduate Program

Richard Wenstrup, MD, Professor, Director, Divisional Laboratory Operations; Director, Molecular Genetics Laboratory; Head, Connective Tissue Clinic

You-Hai Xu, MD, PhD, Research Assistant Professor

Kejian Zhang, MD, MBA, Assistant Professor Clinical, Associate Director, Molecular Genetics Laboratory

FACULTY JOINT APPOINTMENT LISTING

John Greinwald, MD, Associate Professor, Pediatric Otolaryngology Research

Lisa Martin, PhD, Research Assistant Professor, Center for Epidemiology and Biostatistics

Melanie Myers, PhD, MS, CGC, Assistant Professor of Clinical, College of Allied Health Sciences, Assistant Director & Clinical Coordinator, Genetic Counseling Program

Todd Nick, PhD, Professor, Center for Epidemiology and Biostatistics

Ning Wang, PhD, Research Assistant Professor, Allergy and Immunology

OVERVIEW

The Division and Program for Human Genetics (DPHG) has four major areas: clinical, laboratory services, teaching, and research. The divisional goals are directed to the immediate and long term provision of state-of-the-art medical care and treatment for individuals and families afflicted with heritable disorders.



Left to Right: (1st row) K. Zhang, G. Grabowski, N. Leslie, N. Wang, M. Myers (2nd row) N. Warren, L. Martin, Y-H Xu, R. Hopkin (3rd row) Y. Sun, C. Prows, B. Tinkle (4th row) T. Nick, H. Saal (5th row) D. Prows, R. Wenstrup, J. Greinwald

Clinically, the DPHG provides regional and national referral for comprehensive diagnostic, counseling, treatment and management services through multi-disciplinary teams. These include board certified medical geneticists, clinical cytogeneticists, clinical molecular geneticists, clinical metabolic/biochemical disease specialists, genetic counselors, genetic nurse specialists, as well as other medical care professionals. Speciality clinics within the DPHG include Pediatric and Adult Neurofibromatosis, Heredity Connective Tissue, Metabolic Diseases, Velo-Pharyngeal Insufficiency, Prenatal, Craniofacial (Multi-disciplinary, and Infants and Toddlers), Tuberous Sclerosis (with Neurology), Hereditary Cancer, and STAR Lysosomal Storage Disease Center.

Training within the DPHG focuses on MD and PhD post-doctoral fellows and genetic counselors (MS Program) attaining certification by the American Boards of Medical Genetics and Genetic Counseling, respectively. The division houses an ABMG approved site for residency training in Medical Genetics and Medical Genetics subspecialty Fellowship Programs in Biochemical Genetics, Cytogenetics and Molecular Genetics. The DPHG hosts a unique program for nurses directed to increasing genetic awareness and content in RN preparatory programs. Pre-doctoral and post-doctoral training focuses on developing physician/scientists with independent research careers.

Research activities focus on:

1. defining the genetic and gene-environment influences on human diseases
2. improving tests for cytogenetic and molecular diagnosis
3. elucidating the cellular, biochemical and molecular pathogenesis of heritable diseases
4. developing effective gene-based therapeutic strategies for affected families with emphasis on prototypical lysosomal storage diseases and connective tissue disorders

The long term objectives of the DPHG are to enhance diagnostic, clinical and training environments, to apply molecular technologies to the elucidation of disease mechanisms, and to develop treatments for heritable diseases and inherited disease susceptibilities; i.e., interventional genetics.

HIGHLIGHTS

We continue our efforts to enhance the clinical, teaching, and research goals involving individuals and their families afflicted with heritable disorders. Clinically, the numbers of patients evaluated and provided care continues to expand. In addition, innovative programs to provide genetic care to underserved populations in the Cincinnati Region as well as outreach programs continue to be successful. These programs complement our expanding outreach programs in the Cincinnati Region and the Toledo Children's Hospital. The laboratory services including Cytogenetics, Molecular, and Metabolic Diagnostic Laboratories have continued to grow and serve CCHMC, UC, the Cincinnati area and the State of Ohio. Concomitant with these programs has been an expansion of clinical activities in Medical Genetics at CCHMC and UC.

The educational/training and service programs continue to grow and include the Genetic Counseling Program, the Nurse Genetic Program (Distance Learning), Genetics Fellowships, and the Medical Genetics Residency Program. The Distance Learning Programs include two web and interactive modules and a set of 14 curriculum models (C. Prows). The Genetic Counseling Program has placed emphasis on minority recruitment (N. Warren). The division has taken a leadership role in the unique Bang Bao China Scholar Exchange Program (Bao). A unique collaborative service, the pharmacogenetics service (GPS) has integrated genetic variation in drug metabolizing enzymes with clinical care and therapeutic outcome. Working collaboratively, Drs. Wenstrup (DPHG), Glauser (Neurology), Vinks (Clinical Trials) and Pestian (Bioinformatics) have developed accessible and generalized systems for personalization of drug dosing based on an individual's genetic constitution.

Clinical and basic research activities are directed to interventional genetics and the genetic/molecular bases of disease. Clinical research focused on expansion of therapeutic approaches through the CCHMC STAR Lysosomal Disease Center, including new enzyme, pharmacologic chaperone, adjunctive therapies, and implementation of testing for acyl-carnitines and lysosomal diseases (Leslie). Drs. Grabowski, Hopkin, and Leslie have headed programs for new therapies for Gaucher, Fabry, and Pompe, Mucopolysaccharidosis type I (Hurler Syndrome) and Niemann-Pick B diseases, respectively, using enzyme therapy and/or bone marrow transplantation (with Dr. Stella Davies), and contributions to large scale international databases for these diseases. Dr. Bao continues international studies of mutagenic agents in cancer. Dr. Schorry is an integral part of studies of bone disease and therapeutics for large Neurofibromatosis-1 populations.

Basic research has focused on the genetic/molecular basis and treatment of human diseases. 1) Identification of modifier genes of mitochondrial gene function and their role in disease susceptibility, particularly deafness and vision (Guan). 2) Critical role of collagen type V in the initiation of collagen formation and the delineation of bone cell ontology using novel mouse models (Wenstrup). 3) Identification of novel genetic variations in selected genes as contributing to Parkinson's disease, pulmonary hypertension, spheroid body myopathy and good health (Nichols). 4) Localization of genes predisposing to lung toxicity by environmental agents and linkage studies for hypertrophic cardiomyopathy (Prows). 5) The use of small lysosomal proteins, saposins, as antineoplastic agents based on their structural organizations (Qi). 6) Identification of shared pro-inflammatory signature pathways in the pathogenesis and propagation of sphingolipid disorders, and molecular and genetic therapies using mouse models of Gaucher disease and saposin deficiencies (Grabowski, Sun, Xu). 7) Characterization of acid lipase's role in the pathogenesis of emphysema mediated by PPAR-gamma (Du). 8) Pre-clinical mouse models for the development of pharmacological chaperone (Gaucher) and enzyme therapies for NASH and atherosclerosis (Grabowski/Du). The DPHG also leads the

DNA sequencing and genotyping core for the Computational Medicine Center of CCHMC and UC (Grabowski and Keddache).

TRAINING

Yuri Zarate, MD	PL-III	Universidad Industrial de Santander, Colombia
Andrew Burrow, MD	PL-IV	University of Arkansas School of Medicine
Jillene Kogan, MD, PhD	PL-V	University of Illinois School of Medicine
Carlos Prada, MD	Intern	Universidad Industrial de Santander, Colombia
Sonya Barnes, PhD		University of Cincinnati
Zhengtao Chu, MD		Institute of Medical Biology, Peking Union Medical College, China
Yaping Qian, PhD		Union Medical College, China
Bradley Tinkle, MD, PhD		Pediatric/Genetics Residency at CCHMC

GRANTS, CONTRACTS AND INDUSTRY AGREEMENTS

Grant and Contract Awards Annual Direct/Project Period Direct

Bao, L		
Studies of Benzene Toxicity American Petroleum Institute (University of Colorado Health Sciences Center subcontract)	11/01/01 – 12/31/06	\$73,400/\$290,332
Du, H		
Nuclear Receptor Co-Activators in the Lung National Institutes of Health (Indiana University subcontract)	R01 HL 067862 02/01/06 – 06/30/07	\$26,187/\$61,489
Genetic Control of SP-B Gene Expression in the Lung National Institutes of Health (Indiana University – Purdue University Indiana subcontract)	R01 HL 061803 03/06/06 – 06/30/07	\$26,187/\$61,489
Everett, J		
Puberty and Cancer Initiation Environment, Diet and Obesity National Institutes of Health (University of Cincinnati subcontract)	U01 ES 012770 09/29/03 – 07/31/06	\$3,202/\$9,249
Gamm, J		
Breast Cancer Education, Outreach and Follow-up Susan B Komen Breast Cancer Foundation	04/01/06 – 03/31/07	\$55,509
Grabowski, G		
Cincinnati Regional Genetics Center Project Ohio Department of Health	S-31-3-001-1-AU 07/01/05 – 06/30/06	\$287,000
Studies of Prosaposin's Physiologic Role National Institutes of Health	R01 NS 036681 12/1/02 – 11/30/07	\$291,088/\$1,476,953
Biomedical Research and Technology Transfer CMC Ohio Department of Development	12/15/03 - 6/15/07	\$392,938
Guan, M		
Molecular Mechanisms of Aminoglycoside Ototoxicity National Institutes of Health	R01 DC 005230 09/15/02 – 08/31/07	\$250,000/\$1,250,000

Leslie, N		
Early Screening and Diagnosis of Duchenne Muscular Dystrophy Centers for Disease Control (Columbus subcontract) U50 DD 000030-01	09/30/04 – 09/29/07	\$12,456/\$49,545
Nichols, W		
Cloning of Familial Primary Pulmonary Hypertension Gene National Institutes of Health R01 HL 061997	08/01/03 – 07/31/07	\$200,000/\$800,000
Parkinson Disease Collaborative Study of Genetic Linkage National Institutes of Health (Indiana University-Purdue University Indianapolis subcontract) R01 NS 037167	02/04/04 – 01/31/09	\$209,902/\$1,239,388
Project II: Genetic Modifiers of Murine Pulmonary Hypertension National Institutes of Health (Vanderbilt University subcontract) P01 HL 072058	08/04/03 – 07/31/08	\$295,518/\$1,455,866
Genetics of Pulmonary Hypertension in Mice National Institutes of Health (University of Colorado subcontract) R21 HL 079315	01/07/05 – 12/31/06	\$16,765/\$24,368
Prows, D		
Genetic Analysis of Hyperoxia-Induced Acute Lung Injury National Institutes of Health R01 HL 075562	12/15/03 – 11/30/07	\$244,125/\$994,125
Mechanisms of Tropomyosin Induced Hypertrophy National Institutes of Health (University of Cincinnati subcontract) R01 HL 071952	08/01/03 – 06/30/08	\$62,332/\$318,339
Regulation of Respiratory Epithelial Cell Homeostasis – Project 2 National Institutes of Health P01 HL 061646	08/10/04 – 06/30/09	\$191,667
Schorry, E		
Multicenter Study of Tibial Dysplasia in Neurofibromatosis Type I Patients Shriners Hospitals for Children	01/01/04 – 12/31/07	\$6,927/\$27,197
Cincinnati Neurofibromatosis Center Consortium Department of Defense W81XWH-06-1-0023	10/15/05 – 10/15/06	\$20,045
Neurofibromatosis Consortium Development Operations Center Department of Defense (University of Alabama at Birmingham subcontract) W81XWH-05-1-0615	01/01/06 – 09/30/06	\$13,000
Wenstrup, R		
Use of Hammerhead Ribozymes in an Animal Model of Osteogenesis Imperfecta National Institutes of Health R01 AR 048347	04/01/03 – 03/31/08	\$327,930/\$1,660,195
Current Year Direct		\$3,006,178
Industry Contracts		
Grabowski, G		
Genzyme Corporation		\$106,837
Large Scale Biology Corporation		\$80,801
Wenstrup, R		
Novartis		\$47,730
Genzyme Corporation		\$9,240

Leslie, N Genzyme Corporation	\$118,950
Hopkin, R Genzyme Corporation	\$10,202
Current Year Direct Receipts	\$373,760
TOTAL	\$3,379,938

PUBLICATIONS

1. Bao L, Schorry EK. A girl with partial trisomy 12q24.31 inherited from her father and a possible novel syndrome transmitted from her mother. *Am J Med Genet A* 2005;138(4):361-4.
2. Irons RD, Lv L, Gross SA, Ye X, Bao L, Wang XQ, Ryder J, Armstrong TW, Zhou Y, Miao L, Le AT, Kerzic PJ, Ni W, Fu H. Chronic exposure to benzene results in a unique form of dysplasia. *Leuk Res* 2005;29(12):1371-80.
3. Mo J, Lampkin B, Perentesis J, Poole L, Bao L. Translocation (8;18;16)(p11;q21;p13). A new variant of t(8;16)(p11;p13) in acute monoblastic leukemia: case report and review of the literature. *Cancer Genet Cytogenet* 2006;165(1):75-8.
4. Burrow TA, Hopkin RJ, Bove KE, Miles L, Wong BL, Choudhary A, Bali D, Li SC, Chen YT. Non-lethal congenital hypotonia due to glycogen storage disease type IV. *Am J Med Genet A* 2006;140(8):878-82.
5. Castoreno AB, Wang Y, Stockinger W, Jarzylo LA, Du H, Pagnon JC, Shieh EC, Nohturfft A. Transcriptional regulation of phagocytosis-induced membrane biogenesis by sterol regulatory element binding proteins. *Proc Natl Acad Sci U S A* 2005;102(37):13129-34.
6. Du H, Levine M, Ganesa C, Witte DP, Cole ES, Grabowski GA. The role of mannosylated enzyme and the mannose receptor in enzyme replacement therapy. *Am J Hum Genet* 2005;77(6):1061-74.
7. Lian X, Yan C, Qin Y, Knox L, Li T, Du H. Neutral lipids and peroxisome proliferator-activated receptor-g control pulmonary gene expression and inflammation-triggered pathogenesis in lysosomal acid lipase knockout mice. *Am J Pathol* 2005;167(3):813-21.
8. Grabowski GA. Recent clinical progress in Gaucher disease. *Curr Opin Pediatr* 2005;17(4):519-24.
9. Grabowski GA, Kazimierczuk A, Liou B. Cell biology and biochemistry of acid beta-glucosidase: the Gaucher Disease enzyme. In: Futerman AH, Zimran A, editors. *Gaucher disease*. Boca Raton: CRC/Taylor & Francis; 2006. p. 44-66.
10. Liou B, Kazimierczuk A, Zhang M, Scott CR, Hegde RS, Grabowski GA. Analyses of variant acid b-glucosidases: effects of Gaucher disease mutations. *J Biol Chem* 2006;281(7):4242-53.
11. McEachern KA, Nietupski JB, Chuang WL, Armentano D, Johnson J, Hutto E, Grabowski GA, Cheng SH, Marshall J. AAV8-mediated expression of glucocerebrosidase ameliorates the storage pathology in the visceral organs of a mouse model of Gaucher disease. *J Gene Med* 2006;8(6):719-29.
12. Weinreb NJ, Barranger JA, Charrow J, Grabowski GA, Mankin HJ, Mistry P. Guidance on the use of miglustat for treating patients with type 1 Gaucher disease. *Am J Hematol* 2005;80(3):223-9.
13. Tang NL, Zhang W, Grabowski GA, To KF, Choy FY, Ma SL, Shi HP. Novel mutations in type 2 Gaucher disease in Chinese and their functional characterization by heterologous expression. *Hum Mutat* 2005;26(1):59-60.
14. Tylki-Szymanska A, Keddache M, Grabowski GA. Characterization of neuronopathic Gaucher disease among ethnic Poles. *Genet Med* 2006;8(1):8-15.
15. Dai P, Liu X, Han D, Qian Y, Huang D, Yuan H, Li W, Yu F, Zhang R, Lin H, He Y, Yu Y, Sun Q, Qin H, Li R, Zhang X, Kang D, Cao J, Young WY, Guan MX. Extremely low penetrance of deafness associated

- with the mitochondrial 12S rRNA mutation in 16 Chinese families: implication for early detection and prevention of deafness. *Biochem Biophys Res Commun* 2006;340(1):194-9.
16. Guan MX. Prevalence of mitochondrial 12S rRNA mutations associated with aminoglycoside ototoxicity. *Volta Rev* 2005;105:211-237.
 17. Kong WJ, Hu YJ, Wang Q, Wang Y, Han YC, Cheng HM, Kong W, Guan MX. The effect of the mtDNA4834 deletion on hearing. *Biochem Biophys Res Commun* 2006;344(1):425-30.
 18. Lu JX, Peng Y, Meng ZF, Jin LQ, Lu YS, Guan MX. Rational design of an EGF-IL18 fusion protein: implication for developing tumor therapeutics. *Biochem Biophys Res Commun* 2005;334(1):157-61.
 19. Qian Y, Zhou X, Hu Y, Tong Y, Li R, Lu F, Yang H, Mo JQ, Qu J, Guan MX. Clinical evaluation and mitochondrial DNA sequence analysis in three Chinese families with Leber's hereditary optic neuropathy. *Biochem Biophys Res Commun* 2005;332(2):614-21.
 20. Qu J, Li R, Zhou X, Tong Y, Lu F, Qian Y, Hu Y, Mo JQ, West CE, Guan MX. The novel A4435G mutation in the mitochondrial tRNAMet may modulate the phenotypic expression of the LHON-associated ND4 G11778A mutation. *Invest Ophthalmol Vis Sci* 2006;47(2):475-83.
 21. Wang Q, Li QZ, Han D, Zhao Y, Zhao L, Qian Y, Yuan H, Li R, Zhai S, Young WY, Guan MX. Clinical and molecular analysis of a four-generation Chinese family with aminoglycoside-induced and nonsyndromic hearing loss associated with the mitochondrial 12S rRNA C1494T mutation. *Biochem Biophys Res Commun* 2006;340(2):583-8.
 22. Yan Q, Bykhovskaya Y, Li R, Mengesha E, Shohat M, Estivill X, Fischel-Ghodsian N, Guan MX. Human TRMU encoding the mitochondrial 5-methylaminomethyl-2-thiouridylate-methyltransferase is a putative nuclear modifier gene for the phenotypic expression of the deafness-associated 12S rRNA mutations. *Biochem Biophys Res Commun* 2006;342(4):1130-6.
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 24. Zhao L, Wang Q, Qian Y, Li R, Cao J, Hart LC, Zhai S, Han D, Young WY, Guan MX. Clinical evaluation and mitochondrial DNA sequence analysis in two Chinese families with aminoglycoside-induced and non-syndromic hearing loss. *Biochem Biophys Res Commun* 2005;336(3):967-73.
 25. Zhou X, Wei Q, Yang L, Tong Y, Zhao F, Lu C, Qian Y, Sun Y, Lu F, Qu J, Guan MX. Leber's hereditary optic neuropathy is associated with the mitochondrial ND4 G11696A mutation in five Chinese families. *Biochem Biophys Res Commun* 2006;340(1):69-75.
 26. Zhu Y, Qian Y, Tang X, Wang J, Yang L, Liao Z, Li R, Ji J, Li Z, Chen J, Choo DI, Lu J, Guan MX. Aminoglycoside-induced and non-syndromic hearing loss is associated with the G7444A mutation in the mitochondrial COI/tRNASer(UCN) genes in two Chinese families. *Biochem Biophys Res Commun* 2006;342(3):843-50.
 27. Cunningham ML, Seto ML, Hing AV, Bull MJ, Hopkin RJ, Leppig KA. Cleidocranial dysplasia with severe parietal bone dysplasia: C-terminal RUNX2 mutations. *Birth Defects Res A Clin Mol Teratol* 2006;76(2):78-85.
 28. Street NJ, Yi MS, Bailey LA, Hopkin RJ. Comparison of health-related quality of life between heterozygous women with Fabry disease, a healthy control population, and patients with other chronic disease. *Genet Med* 2006;8(6):346-53.
 29. White DR, Giambra BK, Hopkin RJ, Daines CL, Rutter MJ. Aspiration in children with CHARGE syndrome. *Int J Pediatr Otorhinolaryngol* 2005;69(9):1205-9.
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33. Martin LJ, Avery CL, Williams JT, North KE. Quantitative trait locus-specific genotype x alcoholism interaction on linkage for evoked electroencephalogram oscillations. *BMC Genet* 2005;6 Suppl 1:S123.
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36. Molloy CA, Keddache M, Martin LJ. Evidence for linkage on 21q and 7q in a subset of autism characterized by developmental regression. *Mol Psychiatry* 2005;10(8):741-6.
37. Mosher MJ, Martin LJ, Cupples LA, Yang Q, Dyer TD, Williams JT, North KE. Genotype-by-sex interaction in the regulation of high-density lipoprotein: the Framingham Heart Study. *Hum Biol* 2005;77(6):773-93.
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45. Zhang B, McGee B, Yamaoka JS, Guglielmone H, Downes KA, Minoldo S, Jarchum G, Peyvandi F, de Bosch NB, Ruiz-Saez A, Chatelain B, Olpinski M, Bockenstedt P, Sperl W, Kaufman RJ, Nichols WC, Tuddenham EG, Ginsburg D. Combined deficiency of factor V and factor VIII is due to mutations in either LMAN1 or MCFD2. *Blood* 2006;107(5):1903-7.
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