Human Genetics

Division Data Summary

Research and Training Details

Number of Faculty	**27**
Number of Joint Appointment Faculty	**3**
Number of Research Students	**2**
Number of Support Personnel	**116**
Direct Annual Grant Support	**$2,233,322**
Direct Annual Industry Support	**$684,113**
Peer Reviewed Publications	**74**

Clinical Activities and Training

Number of Clinical Staff	**25**
Number of Clinical Fellows	**5**
Number of Clinical Students	**30**
Inpatient Encounters	**513**
Outpatient Encounters	**5082**

Division Photo

Row 1: G Grabowski, G Zhang, M Myers
Row 2: T Smolarek, R Hopkin, A Barski, T Burrow, L Bao
Row 3: C Prows, S Zimmerman, E Schorry, K Komurov, D Neilson, C Atzinger, K Zhang
Row 4: L Martin, S Oh, I Sageser, S Theru Arumugam, B Tinkle, D Prows, R Stottmann, H Saal, M Pandey

Significant Publications

Mazzulli JR, Xu YH, Sun Y, Knight AL, McLean PJ, Caldwell GA, Sidransky E, Grabowski GA, Krainc D. **Gaucher disease glucocerebrosidase and alpha-synuclein form a bidirectional pathogenic loop in synucleinopathies**. *Cell*. 146(1):37-52. Jul 8, 2011.

Tinkle B. **Joint Hypermobility Handbook: A Guide for the Issues & Management of Ehlers-Danlos Syndrome Hypermobility Type and the Hypermobility Syndrome**. Greens Fork, IN: Left Paw Press; 2011.

The first two publications show the global involvement of the CNS by alpha-synuclein accumulation in Gaucher disease mouse models. They furthermore provide a potential mechanism for the relationship between excess glycosphingolipid deposition in Gaucher disease and neurodegenerative diseases.

Wang S, Li R, Fettermann A, Li Z, Qian Y, Liu Y, Wang X, Zhou A, Mo JQ, Yang L, Jiang P, Taschner A, Rossmanith W, Guan MX. **Maternally inherited essential hypertension is associated with the novel 4263A>G mutation in the mitochondrial tRNAIle gene in a large Han Chinese family**. *Circ Res*. 108(7):862-70. Apr 1, 2011.

This second edition provides a comprehensive evaluative approach to joint examinations for hypermobility. Significantly, this is the only such single source for this such physical examinations in a quantitative manner.

Xu YH, Sun Y, Ran H, Quinn B, Witte D, Grabowski GA. **Accumulation and distribution of alpha-synuclein and ubiquitin in the CNS of Gaucher disease mouse models**. *Mol Genet Metab*. 2011 Apr;102(4):436-47.

Mitochondrial dysfunction is being increasingly recognized as a component, if not causality of, many complex and maternally inherited/influenced diseases. This paper is the first to demonstrate a specific mitochondrial genomic mutation as directly linked to essential hypertension, a disease that affects millions world-wide.

Zhou X, Yan L, Prows DR, Yang R. **Generalized F accelerated failure time model for mapping survival trait loci**. *Genomics*. 97(6):379-85. Jun, 2011.

A significant problem in characterizing lethal treats is the quantitative analyses of survival. This approach provides a new and innovative solution to this vexing problem.

Division Highlights

Dr. Grabowski, Dr. Keddachi

Drs. Grabowski and Keddache, together with the Division of Biomedical Informatics, collaborated with the Institute for Blood Disorders and Cancer to establish two new National Centers at CCHMC, the Center of Excellence in Molecular Hematology (Dr. Y. Zhang, P.I.) and the Progenitor Cell Biology Consortium Characterization Core (Dr. P. Malik, P.I.). These two Centers provide support for the molecular and genetic characterization, and potential new therapies for major disorders of childhood.

Drs. Grabowski and Keddache have upgraded the genotyping and sequencing capabilities and analyses of the Genetic Variation and Gene Discover Core as part of the CCHMC effort to enhance Genomics. These are accomplished in close collaboration with the Division of Rheumatology and Dr. John Harley, and the Division of Biomedical informatics through joint hiring of new faculty, cross-platform development, and combined grant applications. Additional joint hires of faculty with Experimental Hematology and Allergy Research provide enhanced bioinformatic analytic capabilities for CCHMC.

Dr. K. Zhang

In addition to Dr. Zhang's research and translational work with Dr. Grom in Rheumatology, she and Dr. Keddache worked closely with Dr. Clint Joiner and Theodosia Kalfa to establish a clinical panel for testing Red Blood Cell Skeleton Disorders. This panel will provide a unique resource for CCHMC, national, and International patients with these types of disorders. Such approaches currently are being extended to the hemoglobinopathies. An additional collaborative program includes the Division of Pediatric Dermatology and is focused on the genomics of Epidermolysis Bullosa.

Dr. Keddache

Dr. Keddache is the lead of the Genetic Variation and Gene Discovery Core (GVGDC) of UC/CCHMC. During the past year, he has expanded the Investigator base of this laboratory to >350 that includes CCHMC, UC, regional, governmental, and international clients desiring state-of-the-art genomic characterization by SNP genotyping and Sanger and NextGen massively paralleled DNA and RNA sequencing. The GVGDC provides these services, but also collaboration, design, development, and initial analyses and consultations for large and small projects for UC/CCHMC investigators. Together with our quantitative genomics team (Drs. Lisa Martin, Ge Zhang, Siva Theru-Arumugam, and Sunghee Oh), investigators have interactive access to modern genomic analyses that is supplemented by faculty in bioinformatics and jointly appointed faculty in Experimental Hematology (Dr. K. Komurov).

Dr. Burrow

Dr. Burrow has established two major collaborative clinics at CCHMC. Together with Dr. Ton DeGraw and Barbara Hillinan, he established an expanding multidisciplinary neurometabolic clinic to service CCHMC, national, and international patients with such conditions. Similarly, he and Drs. K. Shah and A. Lucky have initiated a Dermatogenetics Clinic that focuses on patients with such conditions, their molecular diagnosis, and developing new therapies.

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

Carrie Atzinger, MS, Assistant Professor

Assoc. Director, The Genetic Counseling Program
Research Interests

Liming Bao, MD, PhD, Associate Professor

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

Research Interests Lysosomal storage diseases, particularly Gaucher disease; Inborn errors of metabolism, particularly focusing on development of evidence based medicine

Hong Du, PhD, 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, Professor

Research Interests Mechanisms of mitochondrial disorders, with a focus on maternally transmitted hearing loss and vision loss

Robert Hopkin, MD, Associate Professor

Director, Genetic Residency Programs
Research Interests Fabry Disease; Robin Sequence; 22q11 deletion; Neurofibromatosis; craniofacial genetics; chromosomal anomalies

Mehdi Keddache, PhD, Assistant Professor

Leader, Genomics Core
Research Interests DNA Sequencing / Genotyping Core/Linkage and Association analyses

Nancy Doan Leslie, MD, Professor

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

Lisa Martin, PhD, Associate Professor

Biostatistics and Epidemiology
Research Interests Focus on common complex diseases including obesity and heart malformations

Melanie Myers, PhD, MS, CGC, Associate Professor

Director, The Genetic Counseling Program
Research Interests Clinical utility of family health history and other genomic tools in health promotion

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

Manoj Pandey, PhD, Instructor
Research Interests Immunobiology of the lysosomal storage disease

Daniel R Prows, PhD, Associate 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, 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
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, Assistant Professor
Craniofacial Center
Research Interests Multidisciplinary management of individuals affected by craniofacial abnormalities

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

Teresa A Smolarek, PhD, Associate Professor
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, Associate Professor
Research Interests The pathological mechanisms of lysosomal storage diseases

Siva Theru Arumugam, PhD, Assistant Professor
Assistant Director, Molecular Genetics Laboratory
Research Interests Finding the genes for macular deterioration, Quantitative Genetics

Bradley T Tinkle, MD, Associate Professor
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

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

Ge Zhang, MD, PhD, Assistant Professor

Research Interests

Kejian Zhang, MD, Associate Professor

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

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

Research Interests Genetics of Hearing Loss

Kakajan Komurov, PhD, Assistant Professor

Exp. Hem. & Cancer Bio.

Stephanie Ware, MD, PhD, Associate Professor

Molecular Cardiovascular Biology

Research Interests Genetic disorders of cardiac structure and function

Clinical Staff Members

- Laurie Bailey, MS, Lysosomal Disease Center Coordinator;Clinical Trials Coordinator
- Judy Belli, RN, Certified Pediatric Nurse, Craniofacial Team
- Patricia Bender, RN, MSN, Craniofacial Team
- Lisa Berry, MS, Genetic Counselor for Lysosomal Disease Center
- Kathleen Collins, MS
- Susan Cordes, MS, Genetic Counselor
- Jennifer Glass, MS
- Carol Hetteburg, RN, MSN
- Judy Johnson, MS
- Sandy Kaiser, LPN
- Sara Knapke, MS, Hereditary Cancer Program
- Betty Leech, MS, VCFS
- Anne Lovell, RN, MSN, APN, Neurofibromatosis Clinic Nurse
- Erin Mundt, MS
- Kimberly Page, RD
- Cynthia Prows, MSN,CNS, Genetics Clinical Nurse Specialist
- Shelly Rudnick, MS
- Jodie Rueger, MS
- Kerry Shooner, MS
- Diana Smith, MS

- Christine Spaeth, MS, Fetal Care Genetic Counselor
- Kristen Sund, PhD
- Martha Walker, MS
- Connie Wehmeyer, RN, Nurse Coordinator for Lysosomal Disease Center
- Katie Wusik, MS

Trainees

- Sophia Bous, MD, PGY1, Pediatrics/Genetics Combined Residency
- Jaya George-Abraham, MD, PGY5, Medical Genetics Fellowship
- Melissa Maxwell-Stropes, PhD, Clinical Molecular Genetics Fellowship
- Haiying Meng, PhD, Clinical Cytogenetics Fellowship
- Stephanie Peters Santoro, MD, PGY2, Pediatrics/Genetics Combined Residency
- Carlos Prada, MD, PGY5, Pediatrics/Genetics Combined Residency
- Elizabeth Sellars, MD, PGY4, Pediatrics/Genetics Combined Residency
- Dipesh Tamakuwala, PhD, Clinical Cytogenetics Fellowship
- K. Nicole Weaver, MD, PGY2, Pediatrics/Genetics Combined Residency
- Wenying Zhang, MD, PhD, Clinical Molecular Genetics Fellowship

Significant Accomplishments

Gaucher disease linked with Parkinson's

Gregory Grabowski, MD, You-Hai Xu, PhD, and Ying Sun, PhD have collaborated with researchers in Boston and Ottawa to show a mechanistic link between Gaucher disease and Parkinson disease. Based on these results, additional ongoing studies provide a framework for the dissection of the pathogenesis and potential novel treatments of Parkinson disease and other common neurodegenerative diseases.

Progress in HLH research

Kejian Zhang, MD, MBA, has collaborated with the National Institutes of Health and Stanford University on studies that have delineated the phenotype and molecular genetic characteristics in the development of familial hemophagocytic lymphohistiocytosis (HLH) and associated lymphoproliferative syndromes. These studies expand basic knowledge about the pathogenic mechanisms of these diseases and point toward targets to improve their treatment outcomes.

Pulmonary arterial hypertension advancements

William Nichols, PhD, contributed to significant advancements in understanding the genetic basis of Parkinson's disease and pulmonary arterial hypertension (PAH). Major efforts focused on the establishment of the National Biological Sample and Data Repository for Pulmonary Arterial Hypertension in conjunction with investigators of the REVEAL registry. This national/international collaborative effort at Cincinnati Children's will provide basic and clinical data for the analyses of the pathogenic mechanisms and treatment of PAH.

Division Publications

1. Atzinger CL, Meyer RA, Khoury PR, Gao Z, Tinkle BT. **Cross-sectional and longitudinal assessment of aortic root dilation and valvular anomalies in hypermobile and classic Ehlers-Danlos syndrome**. *J Pediatr*. 2011; 158:826-830 e1.

2. Baye TM, Butsch Kovacic M, Biagini Myers JM, Martin LJ, Lindsey M, Patterson TL, He H, Ericksen MB, Gupta J, Tsoras AM, Lindsley A, Rothenberg ME, Wills-Karp M, Eissa NT, Borish L, Khurana Hershey GK. **Differences in Candidate Gene Association between European Ancestry and African American Asthmatic Children**. *PLoS One*. 2011; 6:e16522.
3. Baye TM, Martin LJ, Khurana Hershey GK. **Application of genetic/genomic approaches to allergic disorders**. *J Allergy Clin Immunol*. 2010; 126:425-36; quiz 437-8.
4. Bendik EM, Tinkle BT, Al-shuik E, Levin L, Martin A, Thaler R, Atzinger CL, Rueger J, Martin VT. **Joint hypermobility syndrome: A common clinical disorder associated with migraine in women**. *Cephalalgia*. 2011; 31:603-13.
5. Berry SA, Jurek AM, Anderson C, Bentler K. **The inborn errors of metabolism information system: A project of the Region 4 Genetics Collaborative Priority 2 Workgroup**. *Genet Med*. 2010; 12:S215-9.
6. Bestebroer J, Aerts PC, Rooijakkers SH, Pandey MK, Kohl J, van Strijp JA, de Haas CJ. **Functional basis for complement evasion by staphylococcal superantigen-like 7**. *Cell Microbiol*. 2010; 12:1506-16.
7. Burrow TA, Grabowski GA. **Velaglucerase alfa in the treatment of Gaucher disease type 1**. *Clin Invest*. 2011; 1:285-293.
8. Burrow TA, Grabowski GA. **Analyses of agalsidase alfa and agalsidase beta for the treatment of Fabry disease.**. *Fabry disease.* New York: Springer; 2010: 417-432. .
9. Burton BK, Guffon N, Roberts J, van der Ploeg AT, Jones SA. **Home treatment with intravenous enzyme replacement therapy with idursulfase for mucopolysaccharidosis type II - data from the Hunter Outcome Survey**. *Mol Genet Metab*. 2010; 101:123-9.
10. Calloway TJ, Martin LJ, Zhang X, Tandon A, Benson DW, Hinton RB. **Risk factors for aortic valve disease in bicuspid aortic valve: A family-based study**. *Am J Med Genet A*. 2011; 155:1015-20.
11. Cullen V, Sardi SP, Ng J, Xu YH, Sun Y, Tomlinson JJ, Kolodziej P, Kahn I, Saftig P, Woulfe J, Rochet JC, Glicksman MA, Cheng SH, Grabowski GA, Shihabuddin LS, Schlossmacher MG. **Acid beta-glucosidase mutants linked to gaucher disease, parkinson disease, and lewy body dementia alter alpha-synuclein processing**. *Ann. Neuro.*. 2011; 69:940-53.
12. Czosek RJ, Haaning A, Ware SM. **A mouse model of conduction system patterning abnormalities in heterotaxy syndrome**. *Pediatr Res*. 2010; 68:275-80.
13. Dalal P, Leslie ND, Lindor NM, Gilbert DL, Espay AJ. **Motor tics, stereotypies, and self-flagellation in primrose syndrome**. *Neurology*. 2010; 75:284-6.
14. Fakhro KA, Choi M, Ware SM, Belmont JW, Towbin JA, Lifton RP, Khokha MK, Brueckner M. **Rare copy number variations in congenital heart disease patients identify unique genes in left-right patterning**. *Proc Natl Acad Sci U S A*. 2011; 108:2915-20.
15. Filipovich AH, Zhang K, Snow AL, Marsh RA. **X-linked lymphoproliferative syndromes: brothers or distant cousins?**. *Blood*. 2010; 116:3398-408.
16. Fritsche LG, Lauer N, Hartmann A, Stippa S, Keilhauer CN, Oppermann M, Pandey MK, Kohl J, Zipfel PF, Weber BH, Skerka C. **An imbalance of human complement regulatory proteins CFHR1, CFHR3 and factor H influences risk for age-related macular degeneration (AMD)**. *Hum Mol Genet*. 2010; 19:4694-704.
17. Ginsburg OM, Kim-Sing C, Foulkes WD, Ghadirian P, Lynch HT, Sun P, Narod SA. **BRCA1 and BRCA2 families and the risk of skin cancer**. *Fam Cancer*. 2010; 9:489-93.
18. Grabowski G, Barnes, Burrow. **Prevalence and management of Gaucher disease**. *Pediatr. Health Med Thera* . 2011; 2:59.
19. Grabowski GA, Petsko, G.A., and Kolodny. (2010) **Gaucher Disease, In: The Metabolic and Molecular Bases of Inherited Disease, 9th ed.**. The McGraw-Hill Companies, Inc..
20. Guan MX. **Mitochondrial 12S rRNA mutations associated with aminoglycoside ototoxicity**.

Mitochondrion. 2011; 11:237-45.

21. Hinton RB, Michelfelder EC, Marino BS, Bove KE, Ware SM. **A fetus with hypertrophic cardiomyopathy, restrictive, and single-ventricle physiology, and a beta-myosin heavy chain mutation**. *J Pediatr*. 2010; 157:164-6.
22. Huang YJ, Bao LM, Wang JY, Huang M. **Association between the polymorphism of GNB3C825T gene and vasovagal syncope in children**. *J Chin Pediatrics*. 2010; 48:896-9.
23. Hummel TR, Jessen WJ, Miller SJ, Kluwe L, Mautner VF, Wallace MR, Lazaro C, Page GP, Worley PF, Aronow BJ, Schorry EK, Ratner N. **Gene expression analysis identifies potential biomarkers of neurofibromatosis type 1 including adrenomedullin**. *Clin Cancer Res*. 2010; 16:5048-57.
24. Huson SM, Acosta MT, Belzberg AJ, Bernards A, Chernoff J, Cichowski K, Gareth Evans D, Ferner RE, Giovannini M, Korf BR, Listernick R, North KN, Packer RJ, Parada LF, Peltonen J, Ramesh V, Reilly KM, Risner JW, Schorry EK, Upadhyaya M, Viskochil DH, Zhu Y, Hunter-Schaedle K, Giancotti FG. **Back to the future: proceedings from the 2010 NF Conference**. *Am J Med Genet A*. 2011; 155A:307-21.
25. Izumi K, Hahn A, Christ L, Curtis C, Neilson DE. **Familial 9q22.3 microduplication spanning PTCH1 causes short stature syndrome with mild intellectual disability and dysmorphic features**. *Am J Med Genet A*. 2011; 155:1384-9.
26. Ji YC, Liu XL, Zhao FX, Zhang JJ, Zhang Y, Zhou XT, Qu J, Guan MX. **[The mitochondrial ND5 T12338C mutation may be associated with Leber's hereditary optic neuropathy in two Chinese families]**. *Yi chuan = Hereditas / Zhongguo yi chuan xue hui bian ji*. 2011; 33:322-8.
27. Johnston JJ, Sapp JC, Turner JT, Amor D, Aftimos S, Aleck KA, Bocian M, Bodurtha JN, Cox GF, Curry CJ, Day R, Donnai D, Field M, Fujiwara I, Gabbett M, Gal M, Graham JM, Hedera P, Hennekam RC, Hersh JH, Hopkin RJ, Kayserili H, Kidd AM, Kimonis V, Lin AE, Lynch SA, Maisenbacher M, Mansour S, McGaughran J, Mehta L, Murphy H, Raygada M, Robin NH, Rope AF, Rosenbaum KN, Schaefer GB, Shealy A, Smith W, Soller M, Sommer A, Stalker HJ, Steiner B, Stephan MJ, Tilstra D, Tomkins S, Trapane P, Tsai AC, Van Allen MI, Vasudevan PC, Zabel B, Zunich J, Black GC, Biesecker LG. **Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations**. *Hum Mutat*. 2010; 31:1142-54.
28. Kaimal V, Chu Z, Mahller YY, Papahadjopoulos-Sternberg B, Cripe TP, Holland SK, Qi X. **Saposin C Coupled Lipid Nanovesicles Enable Cancer-Selective Optical and Magnetic Resonance Imaging**. *Mol Imaging Biol.*. 2010; ahead.
29. Karns R, Zhang G, Jeran N, Havas-Augustin D, Missoni S, Niu W, Indugula SR, Sun G, Durakovic Z, Narancic NS, Rudan P, Chakraborty R, Deka R. **Replication of genetic variants from genome-wide association studies with metabolic traits in an island population of the Adriatic coast of Croatia**. *Eur J Hum Genet*. 2011; 19:341-346.
30. Kokotas H, Grigoriadou M, Yang L, Lodahl M, Rendtorff ND, Gyftodimou Y, Korres GS, Ferekidou E, Kandiloros D, Korres S, Tranebjaerg L, Guan MX, Petersen MB. **Homoplasmy of the G7444A mtDNA and heterozygosity of the GJB2 c.35delG mutations in a family with hearing loss**. *Int J Pediatr Otorhinolaryngol.*. 2011; 75:89-94.
31. Kong WJ, Hu YJ, Wang Q, Wang Y, Han YC, Cheng HM, Kong W, Guan MX. **Corrigendum to "The effect of the mtDNA4834 deletion on hearing" [Biochem. Biophys. Res. Commun. 344 (2006) 425-430]**. *Biochem Biophys Res Commun.*. 2010; 398:791.
32. Leikauf GD, Concel VJ, Liu P, Bein K, Berndt A, Ganguly K, Jang AS, Brant KA, Dietsch M, Pope-Varsalona H, Dopico RA, Jr., Di YP, Li Q, Vuga LJ, Medvedovic M, Kaminski N, You M, Prows DR. **Haplotype association mapping of acute lung injury in mice implicates activin a receptor, type 1**. *Am J Respir Crit Care Med*. 2011; 183:1499-509.
33. Leikauf GD, Pope-Varsalona H, Concel VJ, Liu P, Bein K, Brant KA, Dopico RA, Di YP, Jang AS, Dietsch

M, Medvedovic M, Li Q, Vuga LJ, Kaminski N, You M, Prows DR. **Functional genomics of chlorine-induced acute lung injury in mice**. *Proc Am Thorac Soc*. 2010; 7:294-6.

34. Liu XL, Zhou X, Zhou J, Zhao F, Zhang J, Li C, Ji Y, Zhang Y, Wei QP, Sun YH, Yang L, Lin B, Yuan Y, Li Y, Qu J, Guan MX. **Leber's hereditary optic neuropathy is associated with the T12338C mutation in mitochondrial ND5 gene in six Han Chinese families**. *Ophthalmology*. 2011; 118:978-85.
35. Lu K, Zhao G, Lu H, Zhao S, Song Y, Qi X, Hou Y. **Toll-like receptor 4 can recognize SapC-DOPS to stimulate macrophages to express several cytokines**. *Inflamm Res.*. 2011; 60:153-61.
36. Marsh RA, Madden L, Kitchen BJ, Mody R, McClimon B, Jordan MB, Bleesing JJ, Zhang K, Filipovich AH. **XIAP deficiency: a unique primary immunodeficiency best classified as X-linked familial hemophagocytic lymphohistiocytosis and not as X-linked lymphoproliferative disease**. *Blood*. 2010; 116:1079-82.
37. Marsh RA, Satake N, Biroschak J, Jacobs T, Johnson J, Jordan MB, Bleesing JJ, Filipovich AH, Zhang K. **STX11 mutations and clinical phenotypes of familial hemophagocytic lymphohistiocytosis in North America**. *Pediatr Blood Cancer*. 2010; 55:134-40.
38. Martin LJ, Woo JG, Morrison JA. **Evidence of shared genetic effects between pre- and postobesity epidemic BMI levels**. *Obesity (Silver Spring)*. 2010; 18:1378-82.
39. Mehta PA, Harris RE, Davies SM, Kim MO, Mueller R, Lampkin B, Mo J, Myers K, Smolarek TA. **Numerical chromosomal changes and risk of development of myelodysplastic syndrome--acute myeloid leukemia in patients with Fanconi anemia**. *Cancer Genet Cytogenet*. 2010; 203:180-6.
40. Mendelsohn NJ, Harmatz P, Bodamer O, Burton BK, Giugliani R, Jones SA, Lampe C, Malm G, Steiner RD, Parini R. **Importance of surgical history in diagnosing mucopolysaccharidosis type II (Hunter syndrome): data from the Hunter Outcome Survey**. *Genet Med*. 2010; 12:816-22.
41. Metcalfe K, Lubinski J, Lynch HT, Ghadirian P, Foulkes WD, Kim-Sing C, Neuhausen S, Tung N, Rosen B, Gronwald J, Ainsworth P, Sweet K, Eisen A, Sun P, Narod SA. **Family history of cancer and cancer risks in women with BRCA1 or BRCA2 mutations**. *J Natl Cancer Inst*. 2010; 102:1874-8.
42. Neilson DE. **The interplay of infection and genetics in acute necrotizing encephalopathy**. *Curr Opin Pediatr.*. 2010; 22:751-7.
43. Piao Z, Zhou X, Yan L, Guo Y, Yang R, Luo Z, Prows DR. **Statistical optimization of parametric accelerated failure time model for mapping survival trait loci**. *Theor Appl Genet*. 2011; 122:855-63.
44. Prada CE, Zarate YA, Hagenbuch S, Lovell A, Schorry EK, Hopkin RJ. **Lethal presentation of neurofibromatosis and Noonan syndrome**. *Am J Med Genet A*. 2011; 155:1360-6.
45. Prows C. **Hereditary influences on health promotion of the child and family**. *Wong's Nursing Care of Infants and Children.* St. Louis: Elsevier-Mosby; 2011: 78-116. .
46. Qu J, Wang Y, Tong Y, Zhou X, Zhao F, Yang L, Zhang S, Zhang J, West CE, Guan MX. **Leber's hereditary optic neuropathy affects only female matrilineal relatives in two Chinese families**. *Invest Ophthalmol Vis Sci.*. 2010; 51:4906-12.
47. Qu P, Yan C, Du H. **Matrix metalloproteinase 12 overexpression in myeloid lineage cells plays a key role in modulating myelopoiesis, immune suppression, and lung tumorigenesis**. *Blood*. 2011; 117:4476-89.
48. Rieley MB, Stevenson DA, Viskochil DH, Tinkle BT, Martin LJ, Schorry EK. **Variable expression of neurofibromatosis 1 in monozygotic twins**. *Am J Med Genet A*. 2011; 155A:478-85.
49. Sellars EA, Zimmerman SL, Smolarek T, Hopkin RJ. **Ventricular noncompaction and absent thumbs in a newborn with tetrasomy 5q35.2-5q35.3: An association with Hunter-McAlpine syndrome?**. *Am J Med Genet A* . 2011; 155:1409-13.
50. Shen Z, Zheng J, Chen B, Peng G, Zhang T, Gong S, Zhu Y, Zhang C, Li R, Yang L, Zhou J, Cai T, Jin L, Lu J, Guan MX. **Frequency and spectrum of mitochondrial 12S rRNA variants in 440 Han Chinese**

hearing impaired pediatric subjects from two otology clinics. *J Transl Med.*. 2011; 9:4.

51. Sherrill JD, Gao PS, Stucke EM, Blanchard C, Collins MH, Putnam PE, Franciosi JP, Kushner JP, Abonia JP, Assa'ad AH, Kovacic MB, Biagini Myers JM, Bochner BS, He H, Hershey GK, Martin LJ, Rothenberg ME. **Variants of thymic stromal lymphopoietin and its receptor associate with eosinophilic esophagitis**. *J Allergy Clin Immunol*. 2010; 126:160-5 e3.
52. Slaughter JL, Meinzen-Derr J, Rose SR, Leslie ND, Chandrasekar R, Linard SM, Akinbi HT. **The effects of gestational age and birth weight on false-positive newborn-screening rates**. *Pediatrics*. 2010; 126:910-6.
53. Smolarek TA, Sund K, Bao L, Rudnick S, Zimmerman S. **SNP Microarray Analysis of Hematologic and Solid Tumor Samples Results from a Validation Cohort**. *J Mol Diagn.*. 2010; 12:867-867.
54. Sumegi J, Barnes MG, Nestheide SV, Molleran-Lee S, Villanueva J, Zhang K, Risma KA, Grom AA, Filipovich AH. **Gene expression profiling of peripheral blood mononuclear cells from children with active hemophagocytic lymphohistiocytosis**. *Blood*. 2011; 117:e151-60.
55. Sun GH, Samy RN, Tinkle BT, Cornelius RS, Brown DK. **Craniometaphyseal dysplasia-induced hearing loss**. *Otol Neurotol*. 2011; 32:e9-10.
56. Sun Y, Grabowski GA. **Impaired autophagosomes and lysosomes in neuronopathic Gaucher disease**. *Autophagy*. 2010; 6:648 - 649.
57. Sun Y, Ran H, Liou B, Quinn B, Zamzow M, Zhang W, Bielawski J, Kitatani K, Setchell KD, Hannun YA, Grabowski GA. **Isofagomine in vivo effects in a neuronopathic Gaucher disease mouse**. *PloS one*. 2011; 6:e19037.
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Grants, Contracts, and Industry Agreements

Grant and Contract Awards		Annual Direct / Project Period Direct
GRABOWSKI, G		
Cincinnati Regional Genetics Center		
Health Resources & Services Admin(Ohio Department of Health)		
	07/01/04-06/30/11	$383,500
Studies of Gaucher Disease: A Prototype Lipidosis		
National Institutes of Health		
R01 DK 036729	09/27/07-08/31/12	$343,499
Studies of Gaucher Disease: A Prototype Lipidosis		
National Institutes of Health		
R01 DK 036729	9/1/10-8/31/12	$8,000
Cincinnati Center for Excellence in Molecular Hematology		
National Institutes of Health		
P30 DK 090971	9/30/11-6/30/15	$63,001
Lysosomal Disease Network: Epidemiology and Natural History of Wolman Disease and Cholesteryl Ester Storage Disease		
National Institutes of Health(University of Minnesota)		
U54 NS 065768	09/30/09-08/31/11	$40,000
Digestive Health Center: Bench to Bedside Research in Pediatric Digestive Health (Sequencing Core)		
National Institutes of Health		
P30 DK 078392	08/01/07-05/31/12	$21,466

GUAN, M

Nuclear Modifier Genes for Maternally Inherited Deafness
National Institutes of Health
R01 DC 007696 07/01/07-06/30/11 $207,641

NICHOLS, W

Parkinson Disease Collaborative Study of Genetic Linkage
National Institutes of Health(Indiana University)
R56 NS 037167 08/15/10-08/14/12 $100,000

Genetic Analysis of Murine Chronic Hypoxia-Induced Pulmonary Hypertension
National Institutes of Health
R01 HL 102107 04/01/10-03/31/14 $434,629

PROWS, D

Genetic Analysis of Hyperoxia Induced Acute lung Injury
National Institutes of Health
R01 AI 075562 05/01/09-04/30/13 $257,548

SCHORRY, E

NF Consortium Development Operation PK Ctr STOPN
Department of Defense Army(University of Alabama-Birmingham)
07/01/08-06/30/11 $65,740

Genetic & Epigenetic Differences in Monozygotic Twins with NF1
Department of Defense Army
09/15/10-09/14/12 $82,655

A Randomized Placebo-Controlled Study of Lovastatin
Department of Defense Army(University of Alabama-Birmingham)
10/01/10-02/28/11 $38,461

STOPN mTOR Protocol
Department of Defense Army(University of Alabama-Birmingham)
07/01/10-03/31/12 $88,461

Neurofibromatosis Consortium Development Operation
Department of Defense Army(University of Alabama-Birmingham)
07/01/07-03/31/12 $26,607

KNAPKE, S

Hereditary Breast & Ovarian Cancer Syndrome: Life After a BRCA Mutation
Komen Breast Cancer Foundation
4/1/11-3/31/12 $47,832

KEDDACHE, M

Digestive Health Center, Bench-to-Bedside Research in Pediatrics (Sequencing Core)
National Institutes of Health
P30 DK 078392 8/1/07-5/31/12 $21,466

ZHANG, G

Center for Environmental Genetics
National Institutes of Health (University of Cincinnati)
P30 ES 006096 10/1/10-3/31/11 $2,816

Current Year Direct **$2,233,322**

Industry Contracts

BURROW, A

Genzyme Corporation $125,074

GRABOWSKI, G		
Genzyme Corporation		$15,946
Shire Human Genetic Therapies		$315,871
HOPKIN, R		
Genzyme Corporation		$83,274
Amicus Therapeutics, Inc.		$8,426
LESLIE, N		
BioMarin		$1,925
Genzyme Corporation		$39,847
QI, X		
Bexion Pharmaceticals		$49,761
Sisene SAS		$43,989
	Current Year Direct Receipts	**$684,113**
	Total	**$2,917,435**