Icahn School of Medicine at Mount Sinai

Kravis Children's Hospital
Department of Pediatrics
The Mindich Child Health and Development Institute

Child Health Research Directory

SPRING 2015

The Jack and Lucy Clark Department of Pediatrics

Lisa M. Satlin, M.D., Chair

Mindich Child Health and Development Institute

Bruce D. Gelb., M.D., Director

Department of Preventive Medicine

Philip J. Landrigan, M.D., M.Sc., D.I.H., Chair

Rachel A. Annunziato, Ph.D.
Associate Professor of Pediatrics (Behavioral and Developmental Pediatrics)

Lab/Location: Annenberg 4-51
Email:
rachel.annunziato@mssm.edu

Research Interests: Dr. Annunziato's research focuses primarily on the psychosocial needs of medically ill children and adults. She is interested in the transition to adulthood for adolescents with a medical illness. Her research in this area aims to develop interventions to improve medical and mental health outcomes when patients are shifted from pediatric to adult oriented settings.

Type of Research: Clinical/Translational

Publications:

Annunziato RA, Freiberger D, Martin K, Helcer J, Fitzgerald C, Lefkowitz D. An empirically-based practice perspective on the transition to adulthood for solid-organ transplant recipients. *Pediatr Transplant.* 2014. *[Epub ahead of print].*

Annunziato RA, Rubes M, Caso N, Dillon M, Sicherer SH, Shemesh E. Allocation of food allergy responsibilities and its correlates for children and adolescents. *J Health Psychol.* 2014. *[Epub ahead of print].*

Annunziato RA, Parbhakar M, Helcer J, Kapoor K, Henkel K, Arnon R. (2014). Strategies for measuring quality of life among pediatric solid-organ transplant recipients. *Prog Transplant.* 2014; 24(3): 247-256.

Annunziato RA, Baisley MC, Arrato N, Barton C, Henderling F, Arnon R, Kerkar N. Strangers headed to a strange land? Utilization of a transition coordinator to improve transfer from pediatric to adult service. *J Pediatr*. 2013; 163(6): 1628-1633.

Kerkar N, D'Urso C, Van Nostrand K, Kochin I, Gault A, Suchy F, Miloh T, Arnon R, **Annunziato RA.** Psychosocial outcomes for children with NAFLD over time and compared to obese children without NAFLD. *J Gastroenterol Hepatol.* 2012; 56(1): 77-82.

Manish Arora, B.D.S., M.P.H., Ph.D.

Assistant Professor of Preventive Medicine, and Dentistry
Director, Exposure Biology Laboratory

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Atran 3-02

Email: manish.arora@mssm.edu

Research Interests: Dr. Arora conducts laboratory and epidemiologic studies on environmental health. His team has developed a biomarker of fetal exposure to environmental chemicals using deciduous and permanent teeth. He also studies the environmental determinants of oral health.

Type of Research: Clinical/Translational

Publications:

Austin C, Smith TM, Bradman A, Hinde K, Joannes-Boyau R, Bishop D, Hare DJ, Doble P, Eskenazi B, **Arora M**. Barium distributions in teeth reveal early-life dietary transitions in primates. *Nature*. 2013 Jun 13;498(7453):216-9.

Arora M, Austin C. Teeth as a biomarker of past chemical exposure. *Curr Opin Pediatr*. 2013. Apr;25(2):261-7.

Hare D, Austin C, Doble P, **Arora M**. Elemental bio-imaging of trace elements in teeth using laser ablation-inductively coupled plasma-mass spectrometry. *J Dent*. 2011. May;39(5):397-403.

Arora M, Weuve J, Fall K, Pedersen N, Mucci LA. An exploration of shared genetic risk factors between periodontal disease and cancers: a prospective co-twin study. *Am J Epidemiol. 2009;* 171(2):253-9

Arora M, Weuve J, Weiskopf M, Nie H, Sparrow D, Garcia RI, Hu H. Cumulative lead exposure and tooth loss in older men: Normative Aging Study. *Environ Health Perspect*. 2009; 117(10):1531-4

Margaret Baron, M.D., Ph.D.

Professor of Medicine (Hematology and Medical Oncology), Developmental and Regenerative Biology, and Oncological Sciences

Institute Affiliations:
Tisch Cancer Institute;
Black Family Stem Cell Institute

Lab/Location: Annenberg 24-68

Email: margaret.baron@mssm.edu

Research Interests: The research in the Baron lab combines embryology and stem cell biology, with a focus on hematopoietic development. The lab has a longstanding interest in embryonic hematopoiesis and more recently has been studying mechanisms regulating the development and maturation of adult-type (definitive) erythroid progenitors.

Type of Research: Basic/Translational

Publications:

Zhang, H., Nieves, J.L., Fraser, S.T., Isern, J., Douvaras, P., Papatsenko, D., D'Souza, S., Lemischka, I.R., Dyer, M.A., and **Baron, M.H**. Expression of Podocalyxin separates the hematopoietic and vascular potentials of mouse ES cell-derived mesoderm. *Stem Cells.* 2014; 32(1): 191-203.

Vacaru, A., Isern, J., Fraser, S.T., and **Baron, M.H.** Analysis of primitive erythroid cell proliferation and enucleation using a cyan fluorescent reporter in transgenic mice. *Genesis.* 2013; 51(11): 751-762.

Baron, M.H. Early Embryonic Erythropoiesis ... Not so Primitive After All. *Stem Cells.* 2013; 31: 849-856.

Isern, J., He, Z., Fraser, S.T. Nowotschin, S., Ferrer-Vaquer, A., Moore, R., Hadjantonakis, A.-K., Schulz, V., Tuck, D., Gallagher, P.G., and **Baron, M.H.** Single Lineage Transcriptome Analysis Reveals Key Regulatory Pathways in Primitive Erythroid Progenitors in the Mouse Embryo. *Blood. 2011;* 117: 4924-4934.

Isern, J., Fraser, S.T., He, Z., Zhang, H., and **Baron, M.H.** Dose-dependent regulation of primitive erythroid maturation and identity by the transcription factor Eklf. *Blood. 2010;* 116: 3972-3980.

Keith Benkov, M.D.

Associate Professor of Pediatrics (Gastroenterology) and Division Chief of Gastroenterology

Medical Director, Children's Inflammatory Bowel Disease Center

Lab/Location:
5 E. 98th Street, 10th Floor

Email: keith.benkov@mssm.edu

Research Interests: Dr. Benkov investigates the outcomes of children and adolescents with inflammatory bowel disease, based on a large database of over 1500 patients seen in the last 14 years at the Children's IBD Center. His particular interests include the genetic aspects of Crohn's disease and the unique clinical phenotype on young children presenting with inflammatory bowel disease.

Type of Research: Clinical/Translational

Publications:

Masia Warner C, Reigada LC, Fisher PH, Saborsky AL, **Benkov KJ**. CBT for anxiety and associated somatic complaints in pediatric medical settings: an open pilot study. *J Clin Psychol Med Settings.* 2009 16:169-77.

Colletti RB, Baldassano RN, Milov DE, Margolis PA, Bousvaros A, Crandall WV, Crissinger KD, D'Amico MA, Day AS, Denson LA, Dubinsky M, Ebach DR, Hoffenberg EJ, Kader HA, Keljo DJ, Leibowitz IH, Mamula P, Pfefferkorn MD, Qureshi MA. Pediatric IBD Network for Research and Improvement. Variation in care in pediatric Crohn's disease. *J Pediatr Gastroenterol Nutr*. 2009 49:297-303.

Dunkin D, **Benkov KJ**, Rosenberg HK. Duodenal and rectal hematomas complicating endoscopic biopsy: use of sonography in pediatrics. *J Ultrasound Med*. 2009 28:1575-80.

Paul T, Birnbaum A, Pal D, Pittman N, Ceballos C, LeLeiko N, **Benkov K**. Distinct phenotype of early childhood inflammatory bowel disease. *J Clin Gastroenterol*. 2006 40:583-6.

Tomer G, Ceballos C, Concepcion E, **Benkov KJ**, Nod2/card15 variants are associated with lower weight at diagnosis in children with Crohn's disease. *Am J Gastroenterol.* 2003 Nov;98(11):2479-84.

M. Cecilia Berin, Ph.D.

Associate Professor of Pediatrics (Allergy and Immunology)

Institute Affiliations:
Immunology Institute; Mindich Child Health and Development Institute

Lab/Location: Hess CSM 5-301

Email: cecilia.berin@mssm.edu

Research Interests: Dr. Berin studies mechanisms of tolerance and allergy to foods. Using mouse models of food allergy as well as samples from patients enrolled in clinical trials, her goal is to understand how allergic sensitization to foods occurs, and to identify ways to manipulate the immune response to re-establish immune tolerance.

Type of Research: Basic/Translational

Publications:

Tordesillas L, Goswami R, Benedé S, Grishina G, Dunkin D, Järvinen KM, Maleki SJ, Sampson HA, **Berin MC**. Skin exposure promotes a Th2-dependent sensitization to peanut allergens. *J Clin Invest*. 2014; 124:4965-75.

Järvinen KM, Westfall JE, Seppo MS, James AK, Tsuang AJ, Feustel PJ, Sampson HA, **Berin C**. Role of maternal elimination diets and human milk IgA in the development of cow's milk allergy in the infants. *Clin Exp Allergy*. 2014; 44:69-78.

Berin MC, Sampson HA. Food allergy: an enigmatic epidemic. *Trends Immunol*. 2013; 34:390-7.

Shan M, Gentile M, Yeiser JR, Walland AC, Bornstein VU, Chen K, He B, Cassis L, Bigas A, Cols M, Comerma L, Huang B, Blander JM, Xiong H, Mayer L, **Berin C**, Augenlicht LH, Velcich A, Cerutti A. Mucus enhances gut homeostasis and oral tolerance by delivering immunoregulatory signals. *Science*. 2013; 342:447-53.

Leonard SA, Martos G, Wang W, Nowak-Wegrzyn A, **Berin MC**. Oral immunotherapy induces local protective mechanisms in the gastrointestinal mucosa. *J Allergy Clin Immunol.* 2012; 129:1579-1587.e1.

James J. Bieker, Ph.D.

Professor of Developmental and Regenerative Biology

Institute Affiliations: Mindich Child Health and Development Institute; Black Family Stem Cell Institute; Tisch Cancer Center

Lab/Location: Annenberg, 25-84B

Email: james.bieker@mssm.edu

Research Interests: We are actively pursuing the mechanism of action of the EKLF (KLF1) transcription factor using biochemical, molecular, cellular, and developmental approaches. Our focus is on illuminating EKLF-directed transcriptional and epigenetic controls that lead to regulated erythroid gene expression, and in determining whether mutations are causative for aberrant or malignant hematology.

Type of Research: Basic/Translational

Publications:

Yien YY, **Bieker JJ**. EKLF/KLF1, a tissue-restricted integrator of transcriptional control, chromatin remodeling, and lineage determination. *Molecular and Cellular Biology.* 2013 Jan; 33(1): 4-13.

Siatecka M, **Bieker JJ**. The multifunctional role of EKLF/KLF1 during erythropoiesis. *Blood.* 2011 Aug; 118(8): 2044-2054.

L. Xue, M Galdass, MN Gnanapragasam, D Manwani, **JJ. Bieke.** Extrinsic and intrinsic control by EKLF (KLF1) within a specialized erythroid niche. *Development.* 2014; 141: 2245-2254.

Bieker JJ. Putting a finger on the switch. **Nature Genetics**. 2010 Sep; 42(9): 733-734.

Siatecka M, Sahr KE, Andersen SG, Mezei M, **Bieker JJ**, Peters LL. Severe anemia in the Nan mutant mouse caused by sequence-selective disruption of erythroid Kruppel-like factor. *Proceedings of the National Academy of Sciences of the United States of America.* 2010 Aug; 107(34): 15151-56.

Dusan Bogunovic, Ph.D.

Assistant Professor, Department of Microbiology

Lab/Location: Annenberg 16-10

Email:
dusan.bogunovic@mssm.edu

Research Interests: Dr. Bogunovic's research focuses on human immunogenetics. He studies individuals with severe clinical presentations of infections usually causing mild or no clinical disease. The hypothesis of the lab is that inter-individual variability in susceptibility to infectious agents can also be explained by the immune genetic composition of the host.

Type of Research Basic/Translational

Publications:

Bogunovic D#, Zhang X# , Payelle-Brogard B#, François-Newton V#, Yuan C, Sanal O, Mansouri D, *et.al* Free intracellular ISG15 is an IFN-α/β-inducible negative regulator of IFN-α/β amplification that prevents IFN-α/β-mediated auto-inflammation in humans. *Nature*. 2014 Oct; #denotes equal contribution.

Bogunovic D, Byun M, Durfee LA, Abhyankar A, *et al.* Mycobacterial disease and impaired IFN-γ immunity in humans with inherited ISG15 deficiency. *Science*. 2012 Sep; 337(6102).

Bogunovic D, Manches O, Godefroy E, Yewdall A, Gallois A, *et al.* TLR4 engagement during TLR3-induced proinflammatory signaling in dendritic cells promotes IL-10-mediated suppression of antitumor immunity. *Cancer Research.* 2011 Aug; 71(16).

Bogunovic D, O'Neill DW, Belitskaya-Levy I, Vacic V, *et al*. Immune profile and mitotic index of metastatic melanoma lesions enhance clinical staging in predicting patient survival. *Proceedings of the National Academy of Sciences of the United States of America*. 2009 Dec; 106(48).

Bogunovic D, Boisson-Dupuis S, Casanova JL. ISG15: leading a double life as secreted molecule [review]. *Experimental & Molecular Medicine.* 2013; 45.

Erwin P. Bottinger, M.D.

Professor of Medicine (Nephrology) and Pharmacology and Systems Therapeutics

Institute Affiliation:
Charles Bronfman Institute For Personalized Medicine (Director)

Lab/Location: Annenberg 18-16

Email: erwin.bottinger@mssm.edu

Research Interests: Dr. Bottinger is interested in understanding the genetic, environmental and clinical factors that underlie differences in risk for common complex disease, including diabetes and kidney disease. He is using electronic medical records and genomics to advance gene-based information in clinical practice.

Type of Research: Basic, Clinical/Translational

Publications:

Gottesman O, Scott SA, Ellis SB, Overby CL, Ludtke A, Hulot JS, Hall J, Chatani K, Myers K, Kannry JL, **Bottinger EP**. The CLIPMERGE PGx Program: Clinical Implementation of Personalized Medicine Through Electronic Health Records and Genomics-Pharmacogenomics. *Clin Pharmacol Ther*. 2013;94:214

Starren JS, Williams MC, and **Bottinger EP**. Crossing The Omics Chasm: A Time For Omic Ancillary Systems. *JAMA*, 2013; Mar 14:1-2

Gottesman O, Kuivaniemi H, Tromp G...**Böttinger EP**, Williams MS. The Electronic Medical Records and Genomics (eMERGE) Network: past, present, and future. *Genet Med*. 2013; Jun 6.

Keri L. Monda, Gary K. Chen, Kira C. Taylor, Cameron Palmer, Todd L. Edwards, ...Omri Gottesman, ... Vaneet Lotay, ...Rajiv Nadukuru, ...**Erwin P. Bottinger**, ...Alex P. Reiner, Joel N. Hirschhorn, Ruth JF Loos, Kari E. North, Christopher A. Haiman. A Meta-Analysis Identifies New Loci Associated with Body Mass index in Individuals of African Ancestry. *Nature Genet*. 2013; Jun;45(6):690-6

Tayo BO, Teil M, Tong L, Qin H, Khitrov G, Zhang W, Song Q, Gottesman O, Zhu X, Pereira AC, Cooper RS, **Bottinger EP**. Genetic background of patients from a university medical center in Manhattan: implications for personalized medicine. *PLoS One*. 2011; May 4;6(5):e19166.

Brian D. Brown, Ph.D.

Associate Professor of Genetics and Genomic Sciences

Institute Affiliation:
Mindich Child Health and Development Institute; Immunology Institute; Diabetes, Obesity and Metabolism Institute

Lab/Location:
Hess CSM, 5th S-117

Email: brian.brown@mssm.edu

Research Interests: Dr. Brown is deciphering the molecular networks that regulate the immune system, and exploiting this information to develop strategies that can enhance or subdue immune responses. The goal of his work is to develop a vaccine that can educate the immune system to prevent or reverse autoimmune diseases, such as type I diabetes.

Type of Research: Basic/Translational

Publications:

Israelow B^, Mullokandov G^, Agudo J, Sourisseau M, Bashir A, **Brown BD*** and Evans MJ*. Hepatitis C virus genetics affects miR-122 requirements and response to miR-122 inhibitors. *Nature Communications*. ^equal contribution, *co-corresponding. *In Press.*

Nair-Gupta P, Baccarini A, Tung N, Fabian F, Florey O, Huang Y, Banerjee M, Overholtzer M, Tampé R, **Brown BD**, Whiteheart SW, and Blander JM. TLRs Control Phagosomal MHC-I Delivery from Endosomal Recycling Compartments to Aid Crosspresentation in Dendritic Cells. *Cell.* 2014 Jul 31;158(3):506-21.

Agudo JA, Ruzo A, Tung N, Salmon H, Leboeuf M, Hashimoto D, Becker C, Garrett-Sinha LA, Baccarini A, Merad M, **Brown BD**. The microRNA-126/VEGFR2 axis controls the innate response to pathogen-associated nucleic acids. *Nature Immunology.* 2014 Jan;15(1):54-62.

Mullokandov G, Baccarini A, Ruzo A, Jayaprakash AD, Tung N, Israelow B, Evans MJ, Sachidanandam R, **Brown BD**. High-throughput assessment of microRNA activity and function using microRNA sensor and decoy libraries. *Nature Methods*. 2012 Jul 1;9(8):840-6.

Miller JC, **Brown BD**, Shay T, Gautier EL, Jojic V, Cohain A, Pandey G, Leboeuf M, Elpek KG, Helft J, Hashimoto D, Chow A, Price J, Greter M, Bogunovic M, Bellemare-Pelletier A, Frenette PS, Randolph GJ, Turley SJ, Merad M. Deciphering the transcriptional network of the dendritic cell lineage. *Nature Immunology.* 2012 Sep; 13(9).

Supinda Bunyavanich, M.D., M.P.H.

Assistant Professor of Pediatrics (Allergy and Immunology) and Genetics and Genomic Sciences

Institute Affiliations:
Jaffe Food Allergy Institute; Mindich Child Health and Development Institute; Icahn Institute for Multiscale Biology

Lab/Location: Annenberg 17-90

Email: supinda.bunyavanich@mssm.edu

Research Interests: Dr. Bunyavanich investigates the genetics and genomics of asthma and allergic diseases. She applies genetic and genome-wide approaches to population-based studies to better understand atopic disorders.

Type of Research: Clinical/Translational

Publications:

Bunyavanich S, Celedon JC. Use of inhaled corticosteroids among Hispanics in the United States. *Annals of the American Thoracic Society.* (Accepted Jan 7, 2015.)

Bunyavanich S, Schadt EE. Systems Biology of Asthma and Allergic Diseases: A Multiscale Approach. *Journal of Allergy and Clinical Immunology.* 2015; Jan;135(1):31-42.

Bunyavanich S, Schadt EE, Himes BE, Lasky-Su J, Qiu W, ...,et al. Integrated Genome-wide Association, Coexpression Network, and Expression Single Nucleotide Polymorphism Analysis Identifies Novel Pathway in Allergic Rhinitis. *BMC Medical Genomics*. 2014 Aug 2;7(1):48.

Bunyavanich S, Rifas-Shiman SL, Platts-Mills TA, Workman L, Sordillo JE, Gillman MW, Gold DR, Litonjua AA. Peanut allergy prevalence among school-age children in a US cohort not selected for any disease. *Journal of Allergy and Clinical Immunology*. 2014 Sep; 134(3):753-5. (Epub 2014 Jul 30.)

Bunyavanich S, Rifas-Shiman SL, Platts-Mills TA, Workman L, Sordillo JE, Camargo CA, Gillman MW, Gold DR, Litonjua AA. Peanut, milk, and wheat intake during pregnancy is associated with reduced allergy and asthma in children. *Journal of Allergy and Clinical Immunology*. 2014 May;133(5):1373-82. (Epub 2014 Feb 9)

Joseph Buxbaum, Ph.D.

Vice Chair for Research and Mentoring, Department of Psychiatry; Director, Seaver Autism Center for Research and Treatment; Professor of Psychiatry, Neuroscience, and Genetics and Genomic Sciences

Institute Affiliations: Mindich Child Health and Development Institute; Friedman Brain Institute

Lab/Location: Annenberg 22-24

Email: joseph.buxbaum@mssm.edu

Research Interests: Dr. Buxbaum is interested in understanding the causes of childhood onset psychiatric disorders so that he can develop new treatments. He leads a very extensive program in autism including genetics, model systems, clinical research, and treatment research.

Type of Research: Basic/Translational

Publications:

Ionita-Laza I, Capanu M, De Rubeis S, McCallum K, **Buxbaum JD**. Identification of Rare Causal Variants in Sequence-Based Studies: Methods and Applications to VPS13B, a Gene Involved in Cohen Syndrome and Autism. *PLoS Genet*. 2014; Dec 11;10(12):e1004729.(eCollection 2014 Dec.)

De Rubeis S, He X, Goldberg AP, Poultney CS, ... **Buxbaum JD**. Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*. 2014; Nov 13;515(7526):209-15. (Epub 2014 Oct 29.)

Samocha KE, Robinson EB, Sanders SJ, ... **Buxbaum JD**, Cook EH Jr, Gibbs RA, Schellenberg GD, Sutcliffe JS, Devlin B, Roeder K, Neale BM, Daly MJ. A framework for the interpretation of de novo mutation in human disease. *Nat Genet*. 2014; Sep;46(9):944-50. [Epub 2014 Aug 3.]

Gaugler T, Klei L, Sanders SJ, ... **Buxbaum JD**. Most genetic risk for autism resides with common variation. *Nat Genet*. 2014; Aug;46(8):881-5. [Epub 2014 Jul 20.]

Charles R, Sakurai T, Takahashi N, Elder GA, Gama Sosa MA, Young LJ, **Buxbaum JD**. Introduction of the human AVPR1A gene substantially alters brain receptor expression patterns and enhances aspects of social behavior in transgenic mice. *Dis Model Mech*. 2014 Aug;7(8):1013-22. [Epub 2014 Jun 12.]

Ross L. Cagan, Ph.D.

Professor of Developmental and Regenerative Biology and Associate Dean of the Graduate School of Biological Sciences

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Annenberg 25-40

Email: ross.cagan@mssm.edu

Research Interests: The Cagan lab uses Drosophila to explore cancer and diabetes with the goal of developing drug therapeutics designed to account for whole animal complexity. Using current human sequencing efforts, he has developed multigenic cancer models designed to capture the complexity observed in human disease. In addition, the lab has developed robotics-based methods for screening whole flies in a semi-high throughput manner, allowing for the development of novel drugs that target multiple pathways.

Type of Research: Basic/Translational

Publications:

Hirabayashi S, Baranski TJ, **Cagan RL**. Transformed Drosophila Cells Evade Diet-Mediated Insulin Resistance through Wingless Signaling. *Cell*. 2013 Aug 1;154(3):664-75.

Dar AC, Das T, Shokat KM, and **Cagan R.** Chemical Genetic Discovery of Targets and Anti-targets for Polypharmacological Treatment of Cancer. *Nature*. 2012 Jun 6;486(7401):80-4.

Larson D., Johnson R., Swat M., Cordero J., Glazier J. and **Cagan R.** Computer Simulation of Cellular Patterning Within the Drosophila Pupal Eye. *PLoS Comput.* 2010; 6:e1000841.

Vidal, M., Larson, D. Read, R., and **Cagan, R.** Drosophila Csk regulates oncogenic growth through multiple mechanisms. *Dev Cell*. 2006;10(1):33-44.

Read, R., Goodfellow, P., Mardis, E., Novack, N., and **Cagan, R**. A Drosophila model of Multiple Endocrine Neoplasia Type 2. *Genetics.* 2005;171, 1057-81.

Chen-Leng Cai, Ph.D.

Assistant Professor of Developmental and Regenerative Biology

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Hess CSM 7-105

Email: chenleng.cai@mssm.edu

Research Interests: Dr. Cai has a strong interest in elucidating the molecular pathways controlling mammalian heart development, disease and regeneration. His research uses genetic mouse models to uncover the networking of the key transcription factors during early cardiac induction and development, and how different cardiac precursors coordinate to contribute to the heart formation.

Type of Research: Basic/Translational

Publications:

Zhang L, Nomura-Kitabayashi A, Sultana N, Cai W, Cai X, Moon AM, **Cai CL**, Mesodermal Nkx2.5 is necessary and sufficient for early second heart field development. *Dev Biol.* 2014 Jun 1;390(1):68-79.

Cai X, Zhang W, Hu J, Zhang L, Sultana N, Wu B, Cai W, Zhou B, **Cai CL**, Tbx20 acts upstream of Wnt signaling to regulate endocardial cushion formation and valve remodeling during mouse cardiogenesis. *Development.* 2013 Aug;140(15):3176-87.

Jianyun Yan, Jinshu Xu, Xiaoqiang Cai, Lu Zhang, Nishat Sultana, Jun Hu, Jun Li, Pin-Xian Xu, **Chen-Leng Cai**, Smad4 regulates ureteral smooth muscle cell differentiation during mouse embryogenesis, *PLOS ONE*. In press.

Xu J, Nie X, Cai X, **Cai CL**, Xu PX, Tbx18 is essential for normal development of vasculature network and glomerular mesangium in the mammalian kidney. *Dev Biol.* 2014 Jul 1;391(1):17-31

Zhang H, von Gise A, Liu Q, Hu T, Tian X, He L, Pu W, Huang X, He L, **Cai CL**, Camargo FD, Pu WT, Zhou B. Yap1 is required for endothelial to mesenchymal transition of the atrioventricular cushion. *J Biol Chem*. 2014 May 15.

Patrizia Casaccia, M.D,. Ph.D.

Professor of Neuroscience, Genetics and Genomics, Neurology; Chief Center of Excellence for Myelin Repair

Institute Affiliation:
Mindich Child Health and Development Institute

Lab/Location: Icahn 10th Floor, Room 70F

Email: patrizia.casaccia@mssm.edu

Research Interests: Dr. Casaccia's lab adopts state-of-the art molecular and cellular techniques to define key questions related to environment/gene interaction in physiological brain development and in pathological conditions. Her research addresses mechanisms relevant to pathogenesis and treatment of neurodevelopmental disorders characterized by impaired myelin formation as detected in premature babies, genetic disorders, traumatic brain injury.

Type of Research: Basic and Clinical Translational

Publications:

Liu, J., Magri, L., Zhang, F., Marsh NO., Albrecht, S., Huynh, J.L., Kaur, J, Wu, Q., Kuhlmann T, Zhang, W., Slesinger PA, **Casaccia P.** Chromatin landscape defined by repressive histone methylation during oligodendrocyte differentiation. *J. Neuroscience.* 2015; in press.

Gacias M, Gerona-Navarro G, Plotnikov AN, Zhang G, Zeng L, Kaur J, Moy G, Rusinova E, Rodriguez Y, Matikainen B, Vincek A, Joshua J, **Casaccia P**, Zhou MM. Selective Chemical Modulation of Gene Transcription Favors Oligodendrocyte Lineage Progression. *Chem Biol.* 2014 Jun 18;pii: S1074-5521(14)00179-3. [Epub ahead of print]

Magri. L., Swiss VA**,** Jablonska B., Lei, L., Pedre X., Walsh, M., w. Zhang., Gallo V., Canoll, P., **Casaccia P.** (E2F1 co-regulates cell cycle genes and chromatin components during the transition of oligodendrocyte progenitors from proliferation to differentiation. *J. Neurosci.* 2014; 22;34(4):1481-93.

Liu J., Dietz K, DeLoyht JM, Pedr X., Kelkar, D., Kaur, J., Vialou V., Lobo MK, Dietz, D., Nestler E., Dupree J., **Casaccia P.** Impaired adult myelination in the prefrontal cortex of socially isolated mice. *Nature Neuroscience.* 2012; 15:1621-1623

He Y, Kim JY, Dupree J, Tewari A, Melendez-Vasquez C, Svaren J, **Casaccia P**. Yy1 as a molecular link between neuregulin and transcriptional modulation of peripheral myelination. *Nat Neurosci.* 2010; Dec;13(12):1472-80.

Mirna Chehade, M.D., M.P.H.

Associate Professor of Pediatrics (Allergy and Immunology) and Medicine (Gastroenterology)

Director, Mount Sinai Center for Eosinophilic Disorders, Jaffe Food Allergy Institute

Institute Affiliation:
Jaffe Food Allergy Institute

Lab/Location: Annenberg 17-90, Icahn 11th Floor

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Research Interests: Dr. Chehade's research is focused on allergic eosinophilic gastrointestinal disorders, including eosinophilic esophagitis and eosinophilic gastroenteritis. She studies clinical outcomes of therapies as well as the immunopathogenesis of these diseases, and is examining for non-invasive blood and urine biomarkers for these diseases.

Type of Research: Clinical/Translational

Publications:

Chehade M, Aceves SS, Furuta GT, Fleischer DM. Food Allergy and Eosinophilic Esophagitis: What Do We Do? *J Allergy Clin Immunol in Practice.* 2015; 3:25-32.

Kottyan LC et al. Identification of genome-wide susceptibility loci for eosinophilic esophagitis elucidates tissue-specificity of this allergic disease. *Nature Genetics.* 2014; 46:895-900.

Schoepfer AM, et al. Development and validation of a symptom-based activity index for adults with eosinophilic esophagitis. *Gastroenterology.* 2014; 147:1255-1266.

Ko HM, Morotti RA, Yershov O, **Chehade M**. Eosinophilic Gastritis in Children: Clinicopathological Correlation, Disease Course and Response to Therapy. *American Journal of Gastroenterology.* 2014; 109: 1277-85.

Lieberman JA, Morotti RA, Konstantinou GN, Yershov O, **Chehade M**. Dietary therapy can reverse esophageal subepithelial fibrosis in patients with eosinophilic esophagitis: A historical cohort. *Allergy.* 2012; 67:1299-307.

Jia Chen, ScD

Professor, Departments of Preventive Medicine, Pediatrics, Hematology and Medical Oncology, and Oncological Sciences

Institute Affiliation: Mindich Child Health and Development Institute

Institute Affiliation: Institute of Translational Epidemiology

Lab/Location: Annenberg 21-94

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Research Interests: Dr. Chen's lab uses a molecular epidemiology approach to understand complex interactions between the environment and genome/epigenome in human diseases. Her lab performs functional epigenetic analyses in population studies to elucidate disease mechanisms and to identify/validate biomarkers for disease risk and prognosis.

Type of Research: Basic/Translational

Publications:

Xu X, Gammon MD, Hernandez-Vargas H, Herceg Z. Wetmur JG, Teitelbaum SL, Bradshaw PT, Neugut AI, Santella RM, **Chen J.** DNA Methylation in Peripheral Blood Measured by LUMA is Associated with Breast Cancer in a Population-based Study. *FASEB J*. 2012; 26(6):2657-66.

Lambertini L, Marsit CJ, Sharma P, Maccani MA, Ma Y, Gagne L, Padbury JF, Hu J, **Chen J**. Imprinted Gene Expression in Fetal Growth and Development. *Placenta.* 2012; 33(6):480-6.

Marsit CJ, Lambertini L, Maccani MA, Koestler D, Houseman EA, Gagne L, Padbury JF, Lester BM, **Chen J.** Imprinted Gene Expression in the Placenta is Associated with Infant Neurobehavioral Outcomes. *J Pediatr*. 2012;160(5):854-860.

Diplas AI, Hu J, Lee MJ, Ma YY, Lee LY, Lambertini L, **Chen J** and Wetmur JG. Demonstration of all-or-none loss of imprinting in mRNA expression in single cells, *Nucleic Acid Res*., 2009 Sep 18. [Epub ahead of print]

Diplas AI, Lambertini L, Lee MJ, Sperling R, Lee YL, Wetmur J, **Chen J**. Differential expression of imprinted genes in normal and IUGR human placentas. *Epigenetics*. 2009 May 14;4(4) 235-40.

Jaime Chu, M.D.

Assistant Professor of Pediatrics (Hepatology)

Institute Affiliations: Recanati-Miller Transplant Institute; Mindich Child Health and Development Institute

Lab/Location: Annenberg 25-34

Email: jaime.chu@mssm.edu

Research Interests: Dr. Chu's research is focused on investigating a novel intersection of p53 and congenital disorders of glycosylation (CDG). Children with CDG have defects in N-glycosylation and present with debilitating, multi-systemic disease, including liver and gastrointestinal diseases. Her research aims to elucidate a new metabolic role for p53 with potential therapeutic targets for CDG.

Type of Research: Basic/Translational

Publications:

Rosen D, Thung SN, Sheflin-Findling S, Lai J, Rosen A, Arnon R, **Chu J**. IgG4 Sclerosing Cholangitis in a Pediatric Patient. *Semin Liver Dis.* 2015; 35:88-92.

Chu J, Mir A, Gao N, Rosa S, Monson C, Sharma V, Steet R, Freeze HH, Lehrman MA, Sadler KC. A zebrafish model of congenital disorders of glycosylation with phosphomannose isomerase deficiency reveals an early opportunity for corrective mannose supplementation. *Dis Model Mech.* 2013; 6(1):95-105.

Arnon R, Annunziato RA, Willis A, Parbhakar M, **Chu J**, Kerkar N, Shneider BL. Liver transplantation for children with biliary atresia in the pediatric end-stage liver disease era: The role of insurance status. *Liver Transpl.* 2013; 19(5):543-50.

Rosen D, **Chu J**, Morotti R, Levanon D, Rose S, Lee S, Arnon R. Hepatitis C Virus-Autoimmune Hepatitis Overlap Syndrome in an Adolescent: Case Report and Review of the Literature. *J Pediatr Gastroenterol Nutr* 2013; Dec 16. [Epub ahead of print]

Chu J, Loughlin EA, Gaur NA, Senbanerjee S, Jacob V, Monson C, Kent B, Oranu A, Ding Y, Ukomadu C, Sadler KC. UHRF1 phosphorylation by Cyclin A2/CDK2 is required for zebrafish embryogenesis. *Mol Biol Cell.* 2012;23(1):59-70.

Barbara Coffey, M.D., M.S.

Professor of Psychiatry, Chief of the Tics and Tourette's Clinical and Research Program

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: 1240 Park Avenue, 1-3

Email: barbara.coffey@mssm.edu

Research Interests: Dr. Coffey has focused on the clinical course, comorbidity, phenomenology, and treatment of Tourette Disorder.

Type of Research: Clinical/Translational

Publications:

Samar SM, Moyano MB, Braña-Berríos M, Irazoqui G, Matos A, Kichic R, Gellatly R, Ibanez-Gomez L, Zwilling AL, Petkova E, **Coffey BJ**. Children and adolescents with Tourette's disorder in the USA versus Argentina: behavioral differences may reflect cultural factors. *European Child and Adolescent Psychiatry.* 2013; 22 (11):701-707.

Lebowitz ER, Motlagh MG, Katsovich L, King RA, Lombroso PJ, Grantz H, Lin H, Bentley MJ, Gilbert DL, Singer HS, **Coffey BJ**, Kurlan RM, Leckman JF. Tourette syndrome in youth with and without obsessive compulsive disorder and attention deficit hyperactivity disorder. *European Child & Adolescent Psychiatry*. 2012 Aug; 21(8).

Gabbay V, Babb JS, Klein RG, Panzer AM, Katz Y, Alonso CM, Petkova E, Wang J, **Coffey BJ**. A double-blind, placebo-controlled trial of ω-3 fatty acids in Tourette's disorder. *Pediatrics*. 2012 Jun; 129(6).

Kurlan R, Crespi G, **Coffey B**, Mueller-Vahl K, Koval S, Wunderlich G. A multicenter randomized placebo-controlled clinical trial of pramipexole for Tourette's syndrome. *Movement Disorders: Official Journal of the Movement Disorder Society.* 2012 May; 27(6).

Coffey B, Rapoport J. Guest Editors, Special issue: Tourette's disorder and obsessive compulsive disorder. *Journal of Child and Adolescent Psychopharmacology.* 2010; 20(4): 235-236.

Charlotte Cunningham-Rundles, M.D., Ph.D.

Professor of Immunology, Medicine and Pediatrics
Program Director, Allergy Immunology Fellowship

Lab/Location: Icahn 1120

Email:
charlotte.cunningham-rundles@mssm.edu

Research Interests: Dr. Cunningham-Rundles is interested in the pathogenesis, characterization and treatment of primary human immune deficiency diseases. Her lab investigates the genetics and functions of B cells in primary immune defects and autoimmunity.

Type of Research: Basic/Translational

Publications:

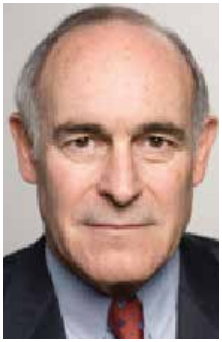
Maglione PJ, Simchoni N, Black S, Radigan L, Overbey JR, Bagiella E, Bussel JB, Bossuyt X, Casanova JL, Meyts I, Cerutti A, Picard C, **Cunningham-Rundles C.** IRAK-4 and MyD88 deficiencies impair IgM responses against T-independent bacterial antigens. *Blood.* 2014; Dec 4;124(24):3561-71.

Keller M, Glessner J, Resnick E, Perez E, Chapel H, Lucas M, Sullivan KE, **Cunningham-Rundles C,** Orange JS, Hakonarson H. Burden of copy number variation in common variable immunodeficiency. *Clin Exp Immunol.* 2014; Jul;177(1):269-71.

Park J, Munagala I, Xu H, Blankenship D, Maffucci P, Chaussabel D, Banchereau J, Pascual V, **Cunningham-Rundles C.** Interferon signature in the blood in inflammatory common variable immune deficiency. *PLoS One*. 2013; Sep 17;8(9).

Romberg N, Chamberlain N, Saadoun D, Gentile M, Kinnunen T, Ng YS, Virdee M, Menard L, Cantaert T, Morbach H, Rachid R, Martinez-Pomar N, Matamoros N, Geha R, Grimbacher B, Cerutti A, **Cunningham-Rundles C**, Meffre E. CVID-associated TACI mutations affect autoreactive B cell selection and activation. *J Clin Invest*. 2013; Oct 1;123(10):4283-93.

Martinez-Gallo M, Radigan L, Almejún MB, Martínez-Pomar N, Matamoros N, **Cunningham-Rundles C.** TACI mutations and impaired B-cell function in subjects with CVID and healthy heterozygotes. *J Allergy Clin Immunol.* 2013; Feb;131(2):468-76. Dec 11.



Robert J. Desnick, Ph.D., M.D.

Professor and Chair Emeritus of Genetics and Genomic Sciences; Dean for Genetic and Genomic Medicine

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Email: robert.desnick@mssm.edu

Research Interests: Dr. Desnick's research interests include genomics, gene discovery, pharmacogenetics, and inborn errors of metabolism. His translational research includes drug development for treatment of genetic diseases.

Type of Research: Basic, Clinical/Translational

Publications:

Yasuda, M., Gan, L., Chen, B., Kadirvel, S., Yu, C., Phillips, J.D., New, M.I., Liebow, A., Fitzgerald, K., Querbes, W., and **Desnick, R.J.**: RNAi-mediated silencing of hepatic Alas1 effectively prevents and treats the induced acute attacks in acute intermittent porphyria mice. *Proc. Natl. Acad. Sci.* [Epub May 12, 2014]

Bonkovsky, H.L., Maddukuri, V.C., Yazici, C., Anderson, K.E., Bissell, D.M., Bloomer, J.R., Phillips, J.D., Naik, H., Peter, I., Baillargeon, G., Bossi, K., Gandolfo, L., Light, C., Bishop, D., and **Desnick, R.J.**: Acute Porphyrias in the USA: Features of 108 Subjects from Porphyrias Consortium. *Am J. Med.* [Epub July 9, 2014]

Scott, S.A., Tan, Q., Baber, U., Yang, Y., Martis, S, Bander, J., Kornreich, R., Hulot, J.S., **Desnick, R.J.**: An Allele Specific PCR System for Rapid Detection and Discrimination of the CYP2C19∗4A, ∗4B, and ∗17 Alleles: Implications for Clopidogrel Response Testing. *J. Mol. Diagn.* 2013; 15:783-789. [Epub September 4, 2013]

Kimmel, S.E., French, B., Kasner, ...**Desnick, R.J**., Ortel, T.L., Billett, H.H., Pendleton, R.C., Geller, N.L., Halperin, J.L., Goldhaber, S.Z., Caldwell, M.D., Califf, R.M., Ellenberg, J.H.: A Randomized Trial of a Pharmacogenetic Algorithm versus a Clinical Algorithm for Warfarin Dosing. *New Eng J Med.* 369:2283-2293, 2013. [Epub November 19, 2013]

Shalata, A., Ramirez, M.C., **Desnick, R.J**., ... Martignetti, J.M.: Morbid Obesity Resulting from Inactivation of the Ciliary Protein CEP19 in Humans and Mice. *Am. J. Hum. Genet.* [Epub November 21, 2013]

Angela Diaz, M.D., M.P.H.

Professor of Pediatrics (Adolescent Medicine) and Preventive Medicine

Lab/Location: 320 East 94th St.

Email: angela.diaz@mssm.edu

Research Interests: Dr. Diaz's research interests focus on adolescent related issues, including cervical, anal and oral HPV persistence and risk factors among adolescent girls, the impact of physical and sexual abuse on the health and mental well being of adolescents, adolescent disclosure of abuse in primary care settings and sexual and reproductive health.

Type of Research: Clinical/Translational

Publications:

Diaz A., Clayton, E.W, Simon, P. Confronting Commercial Sexual Exploitation and Sex Trafficking of Minors. *JAMA Pediatrics*. 2015; 168(9) 791-792.

Bachorik, A, Friedman, J, Nucci-Sack, A, Horowitz, C, **Diaz A**. Adolescent and young adult women's knowledge of and attitudes toward etonogestrel implants. *Journal of Pediatric and Adolescent Gynecology.* August 2014; In press. (Accepted manuscript)

Diaz A., Petersen, A. Institute of Medicine Report: New Directions in Child Abuse and Neglect Research. *JAMA Pediatrics.* 2014; 168(2): 101-102.

Yavorsky, R, Hollman, D, Steever, J, Soghomonian, C, **Diaz A**., Strickler, H, Schlect, N, Burk, R, Ochner, C. Prevalence of Sexually Transmitted Infections in At-Risk Adolescent Females at a Comprehensive Stand-Alone Adolescent Health Center in New York City. *Clin Pediatr (Phila).* 2014; May 7;53(9):890-895.

Braun-Corville, D, Schlect, N, Burk, R, Strickler, H, Rojas, M, Lorde-Rollins, E, Nucci-Sack, A, Hollman, D, Linares, O, **Diaz A**., Strategies for Conducting Adolescent Health Research in the Clinical Setting: The Mount Sinai Adolescent Health Center HPV Experience. *Journal of Pediatric and Adolescent Gynecology.* August 2013. October 2014; 27(5) e103-e108.

Nicole C. Dubois, Ph.D.

Assistant Professor
of Developmental and
Regenerative Biology

Institute Affiliations: Mindich Child Health and Development Institute; Black Family Stem Cell Institute

Lab/Location: Hess CSM, 8th Floor

Email: nicole.dubois@mssm.edu

Research Interests: The research in the Dubois lab focuses on understanding development and disease of the human heart using the pluripotent stem cell model. Specifically, the lab is interested in investigating the molecular mechanisms directing lineage specification with the aim to translate our knowledge to better understand human congenital heart disease.

Type of Research: Basic/Translational

Publications:

El-Mounayri O, Mihic A, Shikatani A, Gagliardi M, **Dubois NC**, DaCosta R, Li RK, Keller G, Husain M. Serum-free differentiation of functional human coronary-like vascular smooth muscle cells from embryonic stem cells. *Cardiovasc Res*. 2013. Apr 1;98(1):125-35.

Dubois NC, Craft A, Sharma P, Elliott D, Stanley EG, Elefanty AG, Gramolini A, Keller G. A novel cell surface marker for the isolation of human pluripotent stem cell-derived cardiomyocytes. *Nat Biotechnol*. 2011. Oct 23;29(11):1011-8.

Kattman SJ, Witty AD, Gagliardi M, **Dubois NC**, Niapour M, Hotta A, Ellis J, Keller G. Stage-specific optimization of activin/nodal and BMP signaling promotes cardiac differentiation of mouse and human pluripotent stem cell lines. *Cell Stem Cell*. 2011. Feb 4;8(2):228-40.

David Dunkin, M.D.

Assistant Professor of Pediatrics (Gastroenterology and Nutrition)

Institute Affiliation: Mindich Child Health and Development Institute

Lab: Annenberg 17-46

Email:david.dunkin@mssm.edu

Research Interests: Dr. Dunkin is interested in understanding the mechanism by which the human body develops or fails to develop tolerance to foreign antigens including food and intestinal flora that leads to diseases such as allergies and inflammatory bowel disease. Using animal models, he is investigating the mechanisms of epicutaneous exposure leading to the induction of tolerance to antigens. The goal is to apply this knowledge towards the treatment of diseases including food allergies and IBD. In addition, Dr. Dunkin and his collaborators in the Center for Chinese Medicine are investigating the use of Chinese herbal therapies in both murine models and in humans for the treatment of IBD.

Type of Research: Basic/Translational

Publications:

Dunkin D, Mehandru S and Colombel J. Immune Cell Therapy in IBD. *Digestive Diseases*. December 2014; 32 Suppl 1:61-66.

Laitman LE, Mathern DR, Hovhannisyan Z, **Dunkin D**, Farsio S, Iuga A, Roda G, Dahan S. Epithelial Notch-1 Signaling Associated with a Regulatory T cell Signature: A Role for the Epithelial Barrier. *Mucosal Immunology*. January 2014.

Srivastava KD, **Dunkin D**, Liu C, Yang N, Miller RL, Sampson H, Li XM. Herbal formula ASHMI suppresses neutrophil predominant airway inflammation in a ragweed sensitized murine asthma model. *Annals of Allergy, Asthma & Immunology.* April 2014; 112(4): 339-347.

Ceballos C, Bao K, **Dunkin DS**, Song Y, Li XM, Benkov K: Complementary and Alternative Medicine use at a Single Pediatric Inflammatory Bowel Disease Center. *Gastroenterol Nurs.* 2014; 37(4).

Kirsten Sadler Edepli, Ph.D.

Associate Professor of Medicine (Division of Liver Diseases) and Developmental and Regenerative Biology

Institute Affiliations: Tisch Cancer Institute; Mindich Child Health and Development Institute

Lab/Location: Annenberg 25-30

Email: kirsten.edepli@mssm.edu

Research Interests: Dr. Edepli's research focuses on the epigenetic contribution to liver development and liver cancer and the cellular basis for fatty liver disease.

Type of Research: Basic/Translational

Publications:

Mudbhary, R., Hoshida, Y., Chernyavskaya, Y., Villanova, N., Jacob, V., Fiel, I., Chen, X., Kojima, K., Thung, S., Bronson, R.T., Lachenmayer, A., Alsinet, C., Desai, A., Senbanerjee, S., Ukomadu, C., Llovet, J.M., **Sadler, K.C.** Overexpression of UHRF1 drives DNA hypomethylation and hepatocellular carcinoma. *Cancer Cell.* 2014; 2:1-14.

Vacaru, A.M., Di Narzo, A.F., Howarth, D.L., Tsedensodnom, O., Imrie, D., Cinaroglu, A., Amin, S., Hao, K., **Sadler, K.C.** Molecularly defined unfolded protein response subclasses have distinct correlations with fatty liver disease in zebrafish. *Dis Model Mech.* 2014; 7:823-835.

Vacaru, A.M., Unlu, G., Spitzner, M., Mione, M., Knapik, E.W., **Sadler, K.C.** In vivo cell biology in zebrafish - providing insights into vertebrate development and disease. *J Cell Sci.* 2014; 127:485-495.

Tsedensodnom, O., Vacaru, A.M., Howarth, D.L., Yin, C., **Sadler, K.C.** Ethanol metabolism and oxidative stress are required for unfolded protein response activation and steatosis in alcoholic liver disease. *Dis Model Mech.* 2013; 6:1213-1226.

Chu, J., Mir, A., Gao, N., Rosa, S., Monson, C., Sharma, V., Steet, R., Freeze, H.H., Lehrman, M.A., **Sadler, K.C.** A zebrafish model of congenital disorders of glycosylation with phosphomannose isomerase deficiency reveals an early opportunity for corrective mannose supplementation. *Dis Model Mech*. 2013; 6:95-105.

Vilma Gabbay, M.D., M.S.

Associate Professor of Psychiatry and Neuroscience
Chief, Pediatric Mood and Anxiety Disorders Program

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Behavioral Science Unit, 1240 Park Ave (Lab Entrance: 96th Street)

Email: vilma.gabbay@mssm.edu

Research Interests: Dr. Gabbay studies the neurological and immunological mechanisms that contribute to the development and maintenance of mood disorders in youth. Her research efforts utilize an array of sophisticated, cutting-edge techniques, including functional magnetic resonance (MR) imaging, MR spectroscopy, immunological and genetic assays, and comprehensive clinical evaluations.

Type of Research: Clinical/Translational

Publications:

Gabbay V, Johnson AR, Alonso C, Evans L, Babb J, Klein R. Anhedonia but not irritability is associated with illness severity outcomes in adolescent major depression. *Child and Adolescent Psychopharmacology.* 2015; in press.

Gabbay V, Ely B, Li Q, Bangaru S, Panzer A, Alonso C, Castellanos X, Milham M. Striatum-Based Circuitry of Adolescent Depression and Anhedonia. *J Am Acad Child Adolesc Psychiatry.* 2013; 52 (6): 628-641.

Gabbay V, Mao X, Klein RG, Ely BA, Babb JS, Panzer A, Alonso C, Shungu DC. Anterior Cingulate Cortex γ-Aminobutyric Acid in Depressed Adolescents: Relationship to Anhedonia. *Arch Gen Psychiatry*. 2012; 69(2): 139-149.

Gabbay V, Liebes L, Katz Y, Liu S, Mendoza S, Babb JS, Alonso CM, Gonen O. The kynurenine pathway in adolescent depression: Preliminary findings from a proton MR spectroscopy study. *Prog Neuro-psychopharmacol Biol Psychiatry*. 2010; 34(1): 37-44.

Gabbay V, Klein RG, Katz Y, Mendoza S, Guttman LE, Alonso CM, Babb JS, Hirsch GS, Liebes L. The possible role of the kynurenine pathway in adolescent depression with melancholic features. *J Child Psychol and Psychiatry*. 2010; 51(8): 935-943.

Maida P. Galvez, M.D., M.P.H.

Associate Professor of Preventive Medicine and Pediatrics

Lab/Location: 17 East 102nd St., 2nd Floor
Email: maida.galvez@mssm.edu

Research Interests: Dr. Galvez's research is focused on neighborhood factors in the urban built environment and environmental endocrine disruptor exposures and their impact on children's growth and development.

Type of Research: Clinical/Translational

Publications:

Deierlein AL, **Galvez MP**, Yen IH, Pinney SM, Biro FM, Kushi LH, Teitelbaum S, Wolff MS. Local food environments are associated with girls' energy, sugar-sweetened beverage and snack-food intakes. *Public Health Nutr.* 2014;May 12:1-7.

Galvez MP, McGovern K, Knuff C, Resnick S, Brenner B, Teitelbaum SL, Wolff MS. Associations between neighborhood resources and physical activity in inner-city minority children. *Acad Pediatr*. 2013 Jan-Feb;13(1):20-6.

Teitelbaum SL, Mervish N, Moshier EL, Vangeepuram N, **Galvez MP,** Calafat AM, Silva MJ, Brenner BL, Wolff MS. Associations between Phthalate Metabolite Urinary Concentrations and Body Size Measures in New York City Children. *Environ Res*. 2012 Jan;112:186-93.

Biro FM, **Galvez MP**, Greenspan LC, Succop PA, Vangeepuram N, Pinney SM, Teitelbaum S, Windham GC, Kushi LH, Wolff MS. Pubertal Assessment Method and Baseline Characteristics in a Mixed Longitudinal Study of Girls. *Pediatrics*. 2010 Sept;126(3):e 583-90.

Galvez, MP, Pearl M. and Yen I. Childhood Obesity and the Built Environment. *Current Opinions in Pediatrics*. 2010 Apr;22(2):202-7.

Adolfo Garcia-Ocana, Ph.D.

Professor of Medicine (Endocrinology, Diabetes and Bone Diseases)

Institute Affiliations: Mindich Child Health and Development Institute; Diabetes, Obesity and Metabolism Institute

Lab/Location: Atran 5-02

Email: adolfo.g.ocana@mssm.edu

Research Interests: Dr. Garcia-Ocaña's research focuses on tissue regeneration, growth factors and intracellular signaling. More specifically, his group is analyzing the therapeutic potential of growth promoting agents to induce pancreatic beta cell regeneration and in diabetes.

Type of Research: Basic

Publications:

Ljubicic S, Polak K, Fu A, Wiwczar J, Szlyk B, Chang Y, Alvarez-Perez JC, Bird GH, Walensky LD, **Garcia-Ocaña A**, Danial NN. *Cell Reports.* 2015;10,497–504.

Ernst S., Alvarez-Perez J.C., Demerci C., Casinelli G.P., Mellado-Gil J.M.D., Vasavada R.C., **Garcia-Ocana A.**. Hepatocyte Growth Factor/c-Met signaling is required for ß-cell regeneration. *Diabetes.* 2014; 63:216-223.

Bernal-Mizrachi E., Kulkarni R., Scott D.K., Mauvais-Jarvis F., Stewart A.F. and **Garcia-Ocaña A.**. Human ß-Cell Proliferation and Intracellular Signaling Part 2: Driving in the Dark without a Roadmap. *Diabetes.* 2014; 63:819-831.

Y Watanabe, S Singamsetty, B Zou, L Guo, D Stefanovski, LC Alonso, **A. Garcia-Ocana**, CP O'Donnell, BJ McVerry. Exogenous glucose administration impairs glucose tolerance and pancreatic insulin secretion during acute sepsis in non-diabetic mice. *PLOS One.* 2013; 8(6):e67716.

Kulkarni R, Bernal-Mizrachi E, **Garcia-Ocaña A**, Stewart AF. Human ß-Cell Proliferation and Intracellular Signaling: Driving in the Dark without a Roadmap. *Diabetes.* 2012; 61: 2205-2213.

Bruce D. Gelb, M.D.

Professor of Pediatrics (Cardiology) and Genetics & Genomic Sciences

Institute Affiliation: Mindich Child Health and Development Institute

Lab: Hess CSM 8-301

Email: bruce.gelb@mssm.edu

Research Interests: Dr. Gelb is interested in uncovering and then understanding the genetic causes of congenital heart defects (CHD). Using state-of-the-art genomic approaches, his research group is studying Mendelian and complex traits with CHD, and then models disease genes in cells and animals.

Type of Research: Basic/Translational

Publications:

Dhandapany, PS, Razzaque MA, Muthusami U, Kunnoth S, Edwards JJ, Mulero-Navara S, Riess I, Pardo S, Sheng J, Ravni DS, Rani B, Govindaraj P, Flex E, Yokota T, Furutani M, Nishizawa T, Nakanishi T, Robbins J, Limongelli G, Hajjar RJ, Lebeche D, Bahl A, Khullar M, Rathinavel A, Sadler KC, Tartaglia M, Matsuoka R, Thangaraj K,* **Gelb BD**.* *RAF1* mutations in childhood-onset dilated cardiomyopathy. *Nature Genetics.* 2014; 46:635-639. * denotes co-senior authorship.

Zaidi S, Choi M, Brueckner M,* Chung WK,* **Gelb BD**,* Goldmuntz E,* Seidman CE,* Lifton RP.* Increased frequency of *de novo* mutations in histone modifying genes in congenital heart disease. *Nature.* 2013; 498:220-223. * denotes equal contribution.

Carvajal-Vergara X, Sevilla A, D'Souza SL...**Gelb BD***, Lemischka I*. Patient-specific induced pluripotent stem cell derived models of LEOPARD syndrome. *Nature.* 2010; 465:808-812. * denotes equal contribution.

Pandit B, Sarkozy A, Pennacchio LA...**Gelb BD**. Gain-of-function *RAF1* mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. *Nature Genet.* 2007; 39:1007-1012.

Tartaglia M, Mehler EL, Goldberg R, Zampino G, Brunner HG, Kremer H, van der Burgt I, Crosby AH, Ion A, Jeery S, Kalidas K, Patton MA, Kucherlapati RS, **Gelb BD**. Mutations in *PTPN11*, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. *Nature Genet*. 2001; 29:465-4680.

Chris Gennings, Ph.D.

Professor, Department of Preventive Medicine

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: CAM D3-134

Email: chris.gennings@mssm.edu

Research Interests: Dr. Gennings' research focuses on development of novel biostatistical methods for designing and analyzing studies of mixtures, including environmental chemical mixtures and nutrients. An example includes the development of a method to estimate the "bad actors" in a mixture that are most related to a health outcome.

Type of Research Basic/Translational

Publications:

Carrico CK, **Gennings C**, Wheeler DC, Factor-Litvak P. Characterization of Weighted Quantile Sum Regression for Highly Correlated Data in a Risk Analysis Setting. *Journal of Agricultural, Biological and Environmental Statistics.* 2014; In press.

Yorita Christensen, KL, Carrico CK, Sanyal AJ, **Gennings, C**. Multiple classes of environmental chemicals are associated with liver disease: NHANES 2003-04. *International Journal of Hygiene and Environmental Health.* 2013; 216(6):703-9.

Marshall, S, **Gennings, C,** Teuschler, LK, Stork, LG, Tornero-Velez, R, Crofton, KM, Rice, GE. An empirical approach to sufficient similarity: Combining Exposure Data and Mixtures Toxicology Data. *Risk Analysis*. 2013; Epub ahead of print Feb 11 .

Gennings C, Carrico C, Factor-Litvak P, Cirillo PM, Cohn BA. A cohort study evaluation of maternal PCB exposure related to time to pregnancy in daughters. *Environmental Health*. 2013; 12:66

Carr CK, Watkins AM, Wolf CJ, Abbott BD, Lau C, **Gennings C** Testing for departures from additivity in mixtures of perfluoroalkyl acids (PFAAs). *Toxicology*. 2013; 306: 169-75.

Valerie Gouon-Evans, PharmD., Ph.D.

Assistant Professor
Department of Developmental and Regenerative Biology
Department of Medicine, Division of Liver diseases

Institute Affiliations: Mindich Child Health and Development Institute; Black Family Stem Cell Institute; Tisch Cancer Institute

Lab/Location: Atran Rm 7-10F

Email: valerie.gouon-evans@mssm.edu

Research Interests:
My research focuses on liver development, regeneration and cancer using the pluripotent stem cell (PSC) differentiation system as well as mouse models and human specimens. Additionally, we are investigating the ultimate utility of human PSC-derived hepatic cells for cell therapy in liver diseases.

Type of Research: Basic/Translational

Publications:

Goldman, O.*, S. Han*, M. Sourrisseau, N. Dziedzic, W. Hamou, B. Corneo, S. D'Souza, T. Sato, D.N. Kotton, K.D. Bissig, T. Kalir, A. Jacobs, T. Evans, M.J. Evans, and **V. Gouon-Evans.** KDR Identifies a Conserved Human and Murine Hepatic Progenitor and Instructs Early Liver Development. *Cell Stem Cell*. 2013; 12:748-760.

Marion Sourisseau, M.*, Goldman, O.*, He, W., Gori, J. L., Kiem, H., **Gouon-Evans, V.,**** Evans, M.J.** Pigtail macaque induced pluripotent stem cell-derived hepatocytes support hepatitis C virus infection. *Gastroenterology*. 2013; 145(5)966-969 e7.

Goldman O*, Han S*, **Gouon-Evans V.** Liver progenitor cell and KDR. *Cell Cycle*. 2014; 13(7)1051-1052. (selected for cover page)

Gouon-Evans V. The Race for Regeneration: Pluripotent-Stem-Cell-Derived 3D Kidney Structures. *Cell Stem Cell*. 2014; In Press.

5. Goldman, O.*, S. Han*, W. Hamou, V. Jodon de Villeroche, G. Uzan, H. Lickert, **V. Gouon-Evans**. Endoderm generates endothelial cells during liver development. *Stem Cell Reports*. 2014; 3:556-565.

Dorothy E. Grice, M.D.

Professor of Psychiatry;
Director, OCD and Related Disorders Clinical and Research Program;
Associate Director, Tics and Tourette's Clinical and Research Program

Institute Affiliations: Friedman Brain Institute; Mindich Child Health and Development Institute

Lab/Location: 1425 Madison Avenue

Email: dorothy.grice@mssm.edu

Research Interests: Dr. Grice focuses on phenomenology, genetics and biology of tic disorders, OCD and autism. Specific programs include an international epidemiological study of genetic and environmental risks for OCD and tic disorders (Denmark and Sweden), genetic studies of tic disorders and OCD, and the effect of antipsychotic medication on the microbiome.

Type of Research: Clinical/Translational

Publications:

Browne HA, Hansen SN, Buxbaum JD, ... **Grice DE**. Familial clustering of tic disorders and obsessive-compulsive disorder. *JAMA Psychiatry*. 2015; in press.

Chaste P, Klei L, Sanders SJ, ... **Grice DE**, Ledbetter DH, Mane SM, Martin DM, Morrow EM, Walsh CA, Sutcliffe JS, Lese Martin C, Beaudet AL, Lord C, State MW, Cook EH Jr, Devlin B. A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity? *Biol Psychiatry.* 2014; Sep 30.

Dietrich A, Fernandez TV, King RA, State MW, Tischfield JA, Hoekstra PJ, Heiman GA, the Tic Genetics Collaborative Group. The Tourette International Collaborative Genetics (TIC Genetics) study, finding the genes causing Tourette syndrome: objectives and methods. *European Child & Adolescent Psychiatry*. 2014; Apr 26.

Cross-Disorder Group of the Psychiatric Genomics Consortium. Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics.* 2013 Sep; 45(9):984-94.

Peter Heeger, M.D.

Professor of Medicine (Nephrology)

Institute Affiliations:
Immunology Institute; Recanati Miller Transplant Institute (Director of Research)

Lab/Location: Annenberg 21-32

Email: peter.heeger@mssm.edu

Research Interests: The focus of the Heeger research program is transplantation immunology and complement biology. The lab uses mouse models to identify mechanisms of immune injury and tolerance and then apply the findings to develop monitoring and treatment strategies in human transplant recipients with the long term goal of prolonging allograft survival and improving patient health.

Type of Research: Basic/Translational

Publications:

Wing-hong Kwan, William van der Touw, Estela Paz-Artal, Ming O. Li, **Peter S. Heeger**. Signaling through C5a Receptor and C3a Receptor Modulates Function of Natural Regulatory T cells in Mice. *J Exp Med.* 2013; 210: 257-68.

Jessica Reid-Adam, Nan Yang, Ying Song, Paolo Cravedi, Xiu-Min Li, **Peter Heeger**. Immunosuppressive Effects of the Traditional Chinese Herb Qu Mai on Human Alloreactive T Cells Amer. *J Transplantation.* 2013; 13:1159-67.

Donald E. Hricik, Emilio D. Poggio, Kenneth J. Woodside, Naragaju Sarabu, Edmund Q. Sanchez, James A. Schulak, Aparna Padiyar, **Peter S. Heeger**, Joshua J. Augustine Effects of Cellular Sensitization and Donor Age on Acute Rejection and Graft Function after Deceased Donor Kidney Transplantation. *Transplantation.* 2013; 95:1254-8.

Paolo Cravedi*, Joaquin Manrique*, Katherine E. Hanlon, Jessica Reid-Adam, Joshua Brody, Praeophayom Prathuangsuk, Anita Mehrotra, **Peter S. Heeger** (*co-first authors) Immunosuppressive Effects of Erythropoietin on Human Alloreactive T cells. *J Am Soc Nephrology.* 2014; 25:2003-15.

Paolo Cravedi and **Peter S. Heeger.** Complement as a multifaceted modulator of kidney transplant injury. *The Journal of Clinical Investigation.* 2014; 124:2348-2354.

Tom Hildebrandt, Psy.D.

Assistant Professor of Psychiatry; Director, Eating and Weight Disorders Program

Lab/Location:
1425 Madison Ave, 6th Floor

Email:
tom.hildebrandt@mssm.edu

Research Interests: Dr. Hildebrandt studies the neuroendocrinology of eating and weight disorders and anabolic-androgenic steroid use, with special interest in disruption of gonadal and appetite hormones and their influences on emotional and eating disturbances. He uses the identification of these neurobiological mechanisms to guide the development of behavioral and family based treatments for adolescents and adults.

Type of Research: Clinical/Translational

Publications:

Kilpela L A, Hill K, Kelly M C, Elmquist J, Ottoson P, Keith D, **Hildebrandt T**, & Becker C B. Reducing eating disorder risk factors: A controlled investigation of a blended task-shifting/train-the-trainer approach to dissemination and implementation. *Behaviour Research and Therapy*. 2015; in press.

Sysko R, **Hildebrandt T**, Kaplan S, Devlin M J, Brewer S K, Zitsman J L, Walsh B T. Predictors of follow-up visit adherence among adolescents receiving laparoscopic adjustable gastric banding. *Surg Obes Rel Dis.* 2015; in press.

Hildebrandt T, Bacow T, Flores A, & Greif R. Exposure-based family therapy (FBT-E): An open case series of a new treatment for anorexia nervosa. *Cog & Beh Prac*. 2015; in press.

*DeBar L L, Wilson G T, Yarborough B J, Burns B, Oyler B, **Hildebrandt T**, ... Striegel R H, et al. Cognitive behavioral treatment for recurrent binge eating in adolescent girls: a pilot trial. *Cog & Beh Prac.* 2013; 20(2): 147-161. [Available on May 1, 2014]

Hildebrandt, T., Alfano, L., Tricamo, M., & Pfaff, D. Conceptualizing the role of estrogens in the development and maintenance of bulimia nervosa. *Clin Psych Rev.* 2010;30,665-668.

Megan K. Horton, Ph.D., M.P.H.

Assistant Professor of Preventive Medicine

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: CAM 3rd Floor

Email: megan.horton@mssm.edu

Research Interests: Dr. Horton's research focuses on understanding the mechanisms through which prenatal and early childhood exposure to environmental toxicants adversely affect children's health.

Type of Research: Basic/Translational

Publications:

Horton MK, Bouselman S, Jones R, Sjodin A, Liu X, Whyatt R, Wapner R, Factor-Litvak P. Predictors of serum concentrations of polybrominated flame retardants among health pregnant women in an urban environment; a cross sectional study. *Env Health.* 2013; 12:23.

Horton MK, Kahn L, Perera F, Barr D, Rauh V. Does the home environment and the sex of the child modify the adverse effects of prenatal exposure to chlorpyrifos on child working memory? *Neurotoxicol Teratol.* 2013; 34(5): 534-41.

Rauh VS, Perera F, **Horton M**, Whyatt R, Bansal R, Hao X, Barr D, Slotkin T, Peterson B. Brain anomalies in children exposed prenatally to a common organophosphate pesticide. *Proc Natl Acad Sci.* 2012; 109(20): 7871-6.

Horton, MK, Rundle A, Camann D, Barr D, Rauh V, Whyatt RM. Impact of prenatal exposure to piperonyl butoxide and permethrin on 36-month neurodevelopment. *Pediatrics.* 2012; 127(3):e699-706.

Rauh V, Arunadajai S, **Horton M**, Perera F, Hoepner L, Barr D, Whyatt R. 7-year Neurodevelopmental Scores and Prenatal Exposure to Chlorpyrifos, a Common Agricultural Pesticide. *Environ Health Perspect.* 2011; 119(8):1196-201.

Ethylin Wang Jabs, M.D.

Professor of Genetics & Genomic Sciences, Pediatrics, and Developmental and Regenerative Biology

Lab/Location: Icahn 14-76

Email: ethylin.jabs@mssm.edu

Research Interests: Dr. Jabs has a strong interest in understanding the genetic basis of birth defects. Her research is focused on craniofacial disorders including craniosynostosis and cleft lip and palate. Her group uses developmental biology and "omic" approaches to study the pathogenetic mechanisms, signaling pathways and networks involved in developmental processes. Based on these findings, therapeutic strategies are being tested in animals models.

Type of Research: Basic/Translational

Publications:

Justice CM, Yagnik G, Kim Y, Peter I, **Jabs EW**, *et al.* 28 more authors. A genome-wide association study identifies susceptibility loci for non-syndromic sagittal craniosynostosis near BMP2 and within BBS9. *Nature Genetics.* 2012; 44:1360-4.

Wang Y, Zhou X, Oberoi K, Phelps R, Couwenhoven R, Sun M, Rezza A, Holmes G, Percival CJ, Friedenthal J, Krejci P, Richtsmeier JT, Huso DL, Rendl M, **Jabs EW**. p38 inhibition ameliorates skin and skull abnormalities in FGFR2 Beare-Stevenson mice. *J Clin Invest.* 2012; 122:2153-64.

Webb BD, Shaaban S, *et al*...**Jabs EW**. HOXB1 founder mutation in humans recapitulates the phenotype of Hoxb1$^{-/-}$ mice. *Am J Hum Genet.* 2012; 91:171-9.

Ng SB, Buckingham KJ, Lee C, Bigham AW, Tabor HK, Dent KM, Huff CD, Shannon PT, **Jabs EW**, Nickerson DA, Shendure J, Bamshad MJ. Exome sequencing identifies the cause of a Mendelian disorder. *Nature Genetics.* 2010; 42:30-5.

Beaty TH, Murray JC, ...**Jabs EW**, *et al.* 49 more authors. A genome-wide association study of cleft lip with and without cleft palate identifies risk variants near MAFB and ABCA4. *Nature Genetics.* 2010; 42:525-9.

Lawrence C. Kleinman, M.D., M.P.H.

Professor of Pediatrics and Population Health Science and Policy (Vice Chair for Research and Education)

Institute Affiliation: Institute for Translational Epidemiology

Lab/Location: Icahn, 3rd Floor, Suite L3-50

Email: lawrence.kleinman@mssm.edu

Research Interests: Dr. Kleinman is a health services researcher and methodologist who has developed innovative analytical approaches using quantitative and qualitative data and is known for his work in measuring and improving the quality of health care. His research frequently includes community engagement. He directs the Collaboration for Advancing Pediatric Quality Measures (CAPQuaM), an AHRQ-CMS Center of Excellence in the federal Pediatric Quality Measures Program.

Type of Research: Clinical/Translational

Publications:

Rabin Fastman B, Howell EA, Holzman I, **Kleinman LC**. Current Perspectives on Temperature Management and Hypothermia in Low Birth Weight Infants. *Newborn & Infant Nursing Reviews.* 2014;14:50-55.

Shemesh E, Howell EA, Annunziato R, **Kleinman LC**. Racial and economic disparities in transplant outcomes: the not-so-hidden morbidities. *Liver Transplantation*. 2014;20:4-6.

Byron SC, Gardner WP, **Kleinman LC**, Mangione-Smith R, Moon JH, Sachdeva R, Schuster MA, Freed G, Smith G, Scholle SH. Developing Measures for Pediatric Quality: Methods and Experiences of the CHIPRA Pediatric Quality Measures Program Grantees. *Academic Pediatrics.* 2014;14:S27-32.

Norton EC, Carroll N, Miller MM, Coyne K, Wang J, **Kleinman LC**. Computing Risk Ratios from Data with Complex Survey Design. *Health Services & Outcomes Research Methodology*. 2014;14:3-14.

Kleinman LC, Dougherty D. Assessing Quality Improvement in Health Care: Theory for Practice. *Pediatrics*. 2013; Suppl1:S110-119.

Alex Kolevzon, M.D.

Associate Professor of Psychiatry and Pediatrics; Clinical Director, Seaver Autism Center for Research and Treatment

Institute Affiliation: Friedman Brain Institute

Lab/Location: Icahn, 4-32

Email: alexander.kolevzon@mssm.edu

Research Interests: Dr. Kolevzon's research is focused on developing new pharmacological treatments in autism spectrum disorders. He collaborates with basic scientists at the Seaver Autism Center where genetic discovery in autism leads to the use of model systems and testing targeted molecular therapeutics. Dr. Kolevzon leads the clinical research team to then study these compounds in clinical trials.

Type of Research: Clinical/Translational

Publications:

Kolevzon A, Bush L, Want AT, Halpern L, Frank Y, Grodberg D, Rapaport R, Tavassoli T, Chaplin W, Soorya L, Buxbaum JD. A Pilot Controlled Trial of Insulin-Like Growth Factor-1 in Children with Phelan McDermid Syndrome. *Mol Autism*. 2014; 5:54.

Kolevzon A, Angarita B, Bush L, Wang AT, Frank Y, Yang A, Rapaport R, Saland J, Srivastava S, Farrell F, Edelmann L, Buxbaum JD. Phelan-McDermid Syndrome: A Review of the Literature and Practice Parameters for Medical Assessment and Monitoring. *J Neurodev Disord*. 2014; 6:39.

Kolevzon A, Lim T, Schmeidler J, Martello T, Cook Jr EH, Silverman JM. Self-injury in autism spectrum disorder: An effect of serotonin transporter gene promoter variants. *Psychiatry Res*. 2014; Oct 6;220(3):987-990.

Soorya L, **Kolevzon A**, Zweifach J, Lim T, Dobry Y, Schwartz L, Frank Y, Wang AT, Cai G, Parkhomenko E, Halpern D, Grodberg D, Angarita B, Willner JP, Yang A, Canitano R, Chaplin W, Betancur C, Buxbaum JD. Prospective investigation of autism and genotype-phenotype correlations in 22q13 deletion syndrome and SHANK3 deficiency. *Mol Autism.* 2013 Jun 11;4(1):18.

Grodberg D, Weinger P, **Kolevzon A**, Soorya L, Buxbaum J. The Autism Mental Status Examination (AMSE): Development of a Brief Autism-Focused Exam. *J Aut Dev Dis.* 2012 Mar;42(3):455-9.

Robert S. Krauss, Ph.D.

Professor of Developmental and Regenerative Biology and Oncological Sciences

Institute Affiliations:
Black Family Stem Cell Institute; Tisch Cancer Institute; Mindich Child Health and Development Institute

Lab: Annenberg 25-70

Email: robert.krauss@mssm.edu

Research Interests: Dr. Krauss, a cell and developmental biologist, is interested in mechanisms of cell adhesion and signal transduction during fetal development and how when these processes go awry it contributes to birth defects and disease. One area of focus is the role of the Hedgehog pathway in midline patterning, defects in which cause the common and devastating birth defect holoprosencephaly.

Type of Research: Basic/Translational

Publications:

Hong, M. and **R.S. Krauss**. (2013). Rescue of holoprosencephaly in fetal alcohol-exposed *Cdon* mutant mice by reduced gene dosage of *Ptch1*. *PLOS ONE* 8(11): e79269.

Romer AI, Singh J, Rattan S, **Krauss RS.** Smooth muscle fascicular reorientation is required for esophageal morphogenesis and dependent on Cdo. *J. Cell Biol.* 2013; 201: 309-323.

Hong M, **Krauss RS.** Cdon mutation and fetal ethanol exposure synergize to produce midline signaling defects and holoprosencephaly spectrum disorders in mice. *PLoS Genet.* 2012; 8(10): e1002999.

Bae GU, Domené S, Roessler E, Schachter K, Kang JS, Muenke M, **Krauss RS.** Mutations in CDON, encoding a hedgehog receptor, result in holoprosencephaly and defective interactions with other hedgehog receptors. *Amer. J. Hum. Genet.* 2011; 89: 231-240.

Allen BL, Song JY, Izzi L, Althaus IW, Kang JS, Charron F, **Krauss RS**, McMahon AP. Overlapping roles and collective requirement for the co-receptors Gas1, Cdo and Boc in Shh pathway function. *Dev Cell*. 2011; 20: 775-787.

Luca Lambertini, Ph.D.

Assistant Professor of Preventive Medicine and Obstetrics, Gynecology and Reproductive Science

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location:
Annenberg 21-94/CAM D4-123

Email: luca.lambertini@mssm.edu

Research Interests: Dr. Lambertini's research is focused on the identification and characterization of biomarkers of improper fetal development leading to the manifestation of chronic and developmental disorders in children. Dr. Lambertini co-directs the Mount Sinai Pregnancy Biobank (MSPB), a tissue bank that collects and makes available placenta and umbilical cord blood samples linked to relevant clinical information for research on pregnancy outcomes and child health.

Type of Research: Basic/Translational

Publications

Jayaprakash AD, Benson EK, Gone S, Liang R, Shim J, **Lambertini L**, Toloue MM, Wigler M, Aaronson SA, Sachidanandam R. Stable heteroplasmy at the single-cell level is facilitated by intercellular exchange of mtDNA. *Nucleic Acids Res.* 2015. [Epub ahead of print]

Kappil M, **Lambertini L**, Chen J. Environmental Influences on Genomic Imprinting. *Current Environ Health Rep.* 2015; in press

Lambertini L. Genomic imprinting: sensing the environment and driving the fetal growth. *Curr Opin Pediatr*. 26 (2): 237-42; 2014.

Chen J, Rialdi A, Li Q, Mystal E, Ly J, Finik J, **Lambertini L**, Nomura Y. Influences of Maternal Stress during Pregnancy on the Epi/genome: Comparison of Placenta and Umbilical Cord Blood. *Depress Anxiety.* 2014, 3 (2): 1-6; 2014.

Nomura Y*, **Lambertini L*,** Rialdi A, ...et al. Global methylation in the placenta and umbilical cord blood from pregnancies with maternal gestational diabetes, preeclampsia, and obesity. *Reprod Sci.* 21(1): 131-7; 2014. * Equal contribution.

Philip J. Landrigan, M.D., M.Sc., D.I.H.

Professor and Chair of Preventive Medicine, Professor of Pediatrics

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: CAM 3 West
Email: phil.landrigan@mssm.edu

Research Interests: Dr. Landrigan has a strong interest in discovering how environmental exposures in prenatal and early postnatal life – in particular, to toxic chemicals – influence health and development in children and across the entire human lifespan. He has studied the developmental toxicity of heavy metals, pesticides and endocrine disruptors. He utilizes the power of prospective, birth cohort, epidemiologic studies to elucidate the neurobehavioral consequences of early-life exposures to toxic chemicals.

Type of Research: Clinical/Translational

Publications:

Grandjean P, **Landrigan PJ**. Neurobehavioural effects of developmental toxicity. *Lancet Neurol* 2014; 13:330-338.

Landrigan PJ, Etzel RA (Editors). *Textbook of Children's Environmental Health.* London: Oxford University Press, 2013.

Landrigan PJ, Lambertini L, Birnbaum LS. A Research Strategy to Discover the Environmental Causes of Autism and Neurodevelopmental Disabilities. *Environ Health Perspect.* 2012 Jul; 120(7): a258-60.

Wisnivesky JP, Teitelbaum S, Todd A, Boffetta P, Crane M, Crowley L, Dellenbaugh C, Harrison D, Herbert R, Hyun K, Jeon Y, Kaplan J, Katz C, Levin S, Luft B, Markowitz S, Moline J, Osbay F, Pietrzak R, Shapiro M, Sharma V, Skloot G, Southwick S, Stevenson L, Udasin I, Wallenstein S, **Landrigan PJ**. Persistence of Multiple Illness in September 11 Rescue Workers. *Lancet.* 2011;378: 888–897.

Landrigan PJ, Goldman LR. Children's Vulnerability to Toxic Chemicals: A Challenge and Opportunity to Strengthen Health and Environmental Policy. *Health Affairs.* 2011;30(5):1-10.

Xiu-Min Li, M.D., M.S.

Professor of Pediatrics (Allergy and Immunology)

Institute Affiliations:
Jaffe Food Allergy Institute; Mindich Child Health and Development Institute

Lab/Location: Altenburg 17-25 and 17-80

Email: xiu-min.li@mssm.edu

Research Interests: Dr. Li's research focuses on understanding the mechanisms underlying the pathogenesis of allergic diseases including food allergy and allergic asthma, and on developing novel therapies for these diseases, including immunomodulators, natural products, and novel active compounds isolated from natural products.

Type of Research: Basic/Translational

Publications:

Song Y. Liu C, Hui Y, Srivastava K, Zhou Z, Chen J, Miller RL, Finkelman FD, and **Li X-M.** Maternal allergy increases susceptibility to offspring allergy in association with Th2 biased epigenetic alterations in a mouse model of peanut allergy. *J Allergy Clin Immunol.* 2014; Oct 16;134(6):1339-1345. [Epub ahead of print].

Yang N, Liang B, Srivastava K, Zeng J, Zhan J, Brown L, Sampson H, Goldfarb J, Emala C, **Li XM**. The Sophora flavescens flavonoid compound trifolirhizin inhibits acetylcholine induced airway smooth muscle contraction. *Phytochemistry.* 2013; Nov;95:259-67.

Lisann L, Song Y, Wang J, Ehrlich P, Maitland A and **X-M Li.** Successful prevention of extremely frequent and severe food anaphylaxis in three children by combined traditional Chinese medicine therapy. *Allergy, Asthma and clinical immunology. Allergy Asthma Clin Immunol*. 2014; Dec 20;10(1):66.

Srivastava K, Bardina L, Sampson H, **X-M Li**. Efficacy and immunological actions of FAHF-2 in a murine model of multiple food allergies. *Ann Allergy Asthma Immunol*. 2012; May;108(5):351-358.e1.

Patil P, Wang J, Song Y, Noone S, Yang N, Wallenstein S, Sampson HA, and **Li X-M**. Clinical safety of FAHF-2, and inhibitory effect on basophils from patients with food allergy – extended phase I study. *J Allergy Clin Immunol*. 2011; Dec;128(6):1259-1265.

Ruth J.F. Loos, Ph.D.

Professor of Preventive Medicine; Director, Genetics of Obesity and Related Metabolic Traits Program

Institution Affiliation: Mindich Child Health and Development Institute

Lab/Location: Annenberg 18-80

Email: ruth.loos@mssm.edu

Research Interests: Dr. Loos' interests focus on the identification of genetic loci contributing to risk of obesity and related metabolic traits. She has led several large-scale gene-discovery efforts for obesity-related traits and has contributed to similar efforts for metabolic traits. Increasingly, her gene discovery work focuses on the identification of low-frequency variants through the implementation exomechip and sequencing projects, in particular in non-white ancestry populations. Her work also assesses the predictive value of established genetic loci and their interaction with lifestyle factors.

Type of Research: Translational

Publications:

Winkler TW, Day FR, Croteau-Chonka DC...Kutalik Z, Heid IM, **Loos RJ**. Quality control and conduct of genome-wide association meta-analyses. Genetic Investigation of Anthropometric Traits (GIANT) Consortium. *Nat Protoc.* 2014 May;9(5):1192-212.

TG and HDL Working Group of the Exome Sequencing Project, National Heart, Lung, and Blood Institute, Crosby J, Peloso GM, ... **Loos RJ,** ... Kathiresan S. Loss-of-function mutations in APOC3, triglycerides, and coronary disease. N Engl J Med. 2014 Jul 3;371(1):22-31. (Epub 2014 Jun 18).

Wood AR, Esko T, Yang J, Vedantam S, Pers TH, Gustafsson S, ... **Loos RJ**, ... Frayling TM. Defining the role of common variation in the genomic and biological architecture of adult human height. Nat Genet. 2014 Nov;46(11):1173-86.

Monda KL, Chen GK, Taylor KC, ... **Loos RJ**, North KE, Haiman CA. A meta-analysis identifies new loci associated with body mass index in individuals of African ancestry. *Nat Genet*. 2013

Berndt SI, Gustafsson S, Mägi R, ... North KE, **Loos RJ**, Ingelsson E. Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. *Nat Genet*. 2013; 45(5):501-12.

Roberto Lucchini, M.D.

Director, Division of Occupational and Environmental Medicine
Professor of Preventive Medicine

Lab/Location: CAM D3-112

Email:
roberto.lucchini@mssm.edu

Research Interests: Dr. Lucchini's research focuses on neurotoxic effects from occupational and environmental exposure to chemicals including manganese, lead, mercury, PCB, solvents, pesticides. A variety of motor, cognitive, sensory and endocrine outcomes are related to the exposure in different age groups including children, workers, and elderly.

Type of Research: Clinical/Translational

Publications:

Zoni S, **Lucchini RG**. Manganese exposure: cognitive, motor and behavioral effects on children: a review of recent findings. *Curr Opin Pediatr*. 2013; Apr;25(2):255-60.

Zoni S, Bonetti G, **Lucchini R**. Olfactory functions at the intersection between environmental exposure to manganese and parkinsonism. *J Trace Elem Med Biol*. 2012; 26(2-3):179-82.

Lucchini RG, Zoni S, Guazzetti S, Bontempi E, Micheletti S, Broberg K, Parrinello G, Smith DR. Inverse association of intellectual function with very low blood lead but not with manganese exposure in Italian adolescents. *Environ Res*. 2012 Oct;118:65-71.

Lucchini RG, Guazzetti S, Zoni S, Donna F, Peter SA, Zacco A, Bontempi E, Salmistraro M, Zimmerman NJ, Smith DR. Tremor, olfactory and motor changes in Italian adolescents exposed to historical ferro-manganese emission. *Neurotoxicology.* 2012; 33(4):687-96.

Rentschler G, Covolo L, Ahmadi Haddad A, **Lucchini RG**, Zoni S, Broberg K. ATP13A2 (PARK9) polymorphisms influence the neurotoxic effects of manganese. *Neurotoxicology*. 2012 Aug;33(4):697-702.

Madhan Masilamani, Ph.D.

Assistant Professor of Pediatrics (Allergy and Immunology)

Institute Affiliations: Jaffe Food Allergy Institute; Mindich Child Health and Development Institute

Lab/Location: Annenberg 17-40

Email: madhan.masilamani@mssm.edu

Research Interests: Dr. Masilamani is interested in finding novel ways to treat food allergy. His current research focus is on the effects of anti-inflammatory phytochemicals on allergic sensitization and anaphylaxis to food in animal models. He is also involved in the development of T cell based immunotherapy for peanut allergy.

Type of Research: Basic/Translational

Publications:

Pascal M, Konstantinou G, **Masilamani M**, Lieberman J, Sampson H. In silico prediction of Ara h 2 T cell epitopes in peanut allergic children. *Clin Exp Allergy.* 2013; 43(1):116-27.

Ford LS, Bloom KA, Nowak-Wegrzyn AH, Shreffler WG, **Masilamani M,** Sampson HA. Basophil reactivity, wheal size, and immunoglobulin levels distinguish degree of cow's milk tolerance. J *Allergy and Clin Immunol*. 2013 Jan;131(1):180-6.e1-3.

Wei J, Bhatt S, Chang LM, Sampson HA, **Masilamani M**. Isoflavones, genistein and daidzein, regulate mucosal immune response by suppressing dendritic cell function. *PLoS One.* 2012;7(10):e47979.

Masilamani M, Wei J, Sampson HA. Regulation of the immune response by soybean isoflavones. *Immunol Res.* 2012 Dec;54(1-3):95-110.

Masilamani M, Wei J, Bhatt S, Paul M, Yakir S, Sampson HA. Soybean isoflavones regulate dendritic cell function and suppress allergic sensitization to peanut. *J Allergy and Clin Immunol.* 2011, Dec;128(6):1242-1250.

Hirofumi Morishita, M.D. Ph.D.

Assistant Professor of Psychiatry, Neuroscience, and Ophthalmology

Institute Affiliations: Mindich Child Health and Development Institute; Friedman Brain Institute

Lab/Location: Hess CSM 9-113

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Research Interests:
Dr. Morishita's research focuses on understanding the mechanisms of experience-dependent brain plasticity during developmental critical periods. By combining molecular, circuit, and systems level methodologies, his research aims to understand pathophysiology and therapeutic intervention for amblyopia and other neurodevelopmental disorders such as schizophrenia.

Type of Research: Basic/Translational

Publications:

Demars MP, **Morishita H**. Cortical parvalbumin and somatostatin GABA neurons express distinct endogenous modulators of nicotinic acetylcholine receptors. *Mol Brain*. 2014;7:75.

Nabel EM, **Morishita H**. Regulating critical period plasticity: insight from the visual system to fear circuitry for therapeutic interventions. *Front Psychiatry*. 2013;4:146. Review.

Cabungcal JH, Steullet P, **Morishita H**, Kraftsik R, Cuenod M, Hensch TK, Do KQ. Perineuronal nets protect fast-spiking interneurons against oxidative stress. *Proc Natl Acad Sci USA*. 2013 May 28;110(22):9130-5

Morishita H, Miwa JM, Heintz N, Hensch TK. Lynx1, a cholinergic brake, limits plasticity in adult visual cortex. *Science.* 2010 Nov 26;330(6008):1238-40.

Morishita H, Hensch TK. Critical period revisited: impact on vision. *Curr Opin Neurobiol.* 2008 Feb;18(1):101-7.

Eric Nestler, M.D.

Professor and Chair of Neuroscience; Professor of Pharmacology and Systems Therapeutics, and Psychiatry

Institute Affiliation: Friedman Brain Institute (Director)

Lab/Location: Annenberg 21-32

Email: eric.nestler@mssm.edu

Research Interests: Dr. Nestler's research focuses on identifying the neurobiological basis of drug addiction and depression in rodent models. He studies the molecular and cellular changes that occur in regions of the brain important for reward and motivation in response to chronic administration of a drug of abuse or chronic exposure to stress. He is particularly interested in long-lasting changes that are mediated via alterations in gene expression and chromatin remodeling. The result of his research will guide future efforts toward the development of more effective treatments for addiction and depression.

Type of Research: Basic/Translational

Publications:

Dietz, D.M., Sun, H.S., Lobo, M.K., **...Nestler, E.J.** Essential for Rac1 in cocaine-induced structure plasticity of nucleus accumbens neurons. *Nature Neurosci.* 2012; 15:891-896.

Robison, A.J. and **Nestler, E.J**. Transcriptional and epigenetic mechanisms of addiction. *Nature Rev. Neurosci.* 2011;12:623-637.

Mazei-Robison, M.S., Koo, J.W., Friedman, A. **...Nestler, E.J.** Role for mTOR signaling and neuronal activity in morphine-induced adaptations in ventral tegmental areadopamine neurons. *Neuron* 2011; 72:977-990.

Maze, I., Covington, H.E. III, Dietz, D.M., **...Nestler, E.J.** Essential role of the histone methyltransferase G9a in cocaine-induced plasticity. *Science* 2010; 327:213-216.

Lobo, M.K., Covington, H.E. III, Chaudhury, D. **...Nestler, E.J.** Cell type specific loss of BDNF signaling mimics optogenetic control of cocaine reward. *Science* 2010; 330:385-390.

Maria I. New, M.D.

Professor of Pediatrics (Endocrinology) and Genetics & Genomic Sciences

Director, Adrenal Steroid Disorders Program

Lab/Location: Terrence Cardinal Cooke 419 Annex

Email: maria.new@mssm.edu

Research Interests: Dr. New is interested in discovering and then understanding the genetic causes of adrenal steroid disorders, including congenital adrenal hyperplasia (CAH). Her research emphasizes genotype/phenotype correlation and prenatal diagnosis and treatment.

Type of Research: Clinical/Translational

Publications:

Shozeb Haider, Barira Islam, Valentina D'Atri... **Maria I. New.** Structure-Phenotype Correlations of Human CYP21A2 Mutations in Congenital Adrenal Hyperplasia. *Proc Natl Acad Sci U S A*. 2013; Feb 12;110(7):2605-10. Epub 2013 Jan 28.

Maria I. New, Moolamannil Abraham, Brian Gonzalez, Miroslav Dumic, Maryam Razzaghy Azar, David Chitayat, Li Sun, Mone Zaidi, Robert Wilson and Tony Yuen. Genotype-Phenotype Correlation in 1,507 Families with Congenital Adrenal Hyperplasia owing to 21-Hydroxylase Deficiency. *Proc Natl Acad Sci U S A*. 2013;Feb 12;110(7):2611-6.

Sun L, Zhu LL, Lu P... **New MI**, Davies TF, Zaidi M. Genetic confirmation for a central role for TNFα in the direct action of thyroid stimulating hormone on the skeleton. *Proc Natl Acad Sci USA.* 2013;110:9891-6.

Ahmed M. Khattab, Cedric H.L. Shackleton, Beverly A. Hughes, Jayesh B. Bodalia, **Maria I. New**. Remission of hypertension and electrolyte abnormalities following renal transplantation in a patient with apparent mineralcorticoid excess well documented throughout childhood. *J Ped Endocrinol Metab*. 2013;Aug; 0;0,1-5.

New MI, Tong YK, Yuen T, Jiang P, Pina C, Chan KC, Khattab A, Liao JW, Yau M, Kim SM, Chiu WK, Sun L, Zaidi M, Lo YM. Noninvasive Prenatal Diagnosis of Congenital Adrenal Hyperplasia Using Cell-Free Fetal DNA in Maternal Plasma. *Journal of Clinical Endocrinology and Metabolism*. 2014 Jun;99(6):E1022-30.

Jeffrey H. Newcorn, M.D.

Associate Professor of Psychiatry (Child and Adolescent Psychiatry) and Pediatrics
Director, Division of ADHD and Learning Disorders;
Director, Pediatric Psychopharmacology

Institute Affiliation: Friedman Brain Institute

Lab/Location:
19 East 98th St. 5th Floor

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Research Interests: Dr. Newcorn studies the neurobiological basis of attention-deficit/hyperactivity disorder (ADHD), and clinical efficacy and mechanism of action of stimulant and non-stimulant medications. He conducts clinical treatment studies which include neuroimaging and genetic biomarkers of response.

Type of Research: Clinical/Translational

Publications:

Bédard AC, Stein MA, Halperin JM, Krone B, Rajwan E, **Newcorn JH**. Differential impact of methylphenidate and atomoxetine on sustained attention in youth with attention-deficit/hyperactivity disorder. *J Child Psychol Psychiatry*. 2015 Jan;56(1):40-8. Epub 2014 Jun 19. [PubMed - in process]

Bédard AC, **Newcorn JH**, Clerkin SM, Krone B, Fan J, Halperin JM, Schulz KP. Reduced prefrontal efficiency for visuospatial working memory in attention-deficit/hyperactivity disorder. *J Am Acad Child Adolesc Psychiatry*. 2014 Sep;53(9):1020-1030.e6. [Epub 2014 Jun 19.]

Newcorn JH, Stein MA, Childress AC, Youcha S, White C, Enright G, Rubin J. Randomized, double-blind trial of guanfacine extended release in children with attention-deficit/hyperactivity disorder: morning or evening administration. *J Am Acad Child Adolesc Psychiatry* 2013 Sep;52(9):921-30.

Schulz KP, Clerkin SM, Fan J, Halperin JM, **Newcorn JH**. Guanfacine modulates the influence of emotional cues on prefrontal cortex activation for cognitive control. *Psychopharmacology* (Berl) 2013 Mar;226(2):261-71

Schulz , KP, Fan, J, Bédard, A-C, Clerkin, S, Ivanov, I, Tang, C, Halperin, J, **Newcorn, JH**. Common and Unique Therapeutic Mechanisms of Stimulant and Non-Stimulant Treatments for ADHD. *Arch Gen Psychiatry* 2012;69(9):952-961.

Anna Nowak-Wegrzyn, M.D.

Associate Professor of Pediatrics (Allergy and Immunology)

Institute Affiliations: Jaffe Food Allergy Institute; Mindich Child Health and Development Institute

Lab/Location: Jaffe Food Allergy Institute

Email: anna.nowak-wegrzyn@mssm.edu

Research Interests: Dr. Nowak-Wegrzyn has a special interest in food allergy. Her research focuses on egg and milk allergy treatment and pathophysiology of food protein-induced enterocolitis syndrome (FPIES).

Type of Research: Clinical/Translational

Publications:

Caubet JC, Ford LS, Sickles L, Järvinen KM, Sicherer SH, Sampson HA, **Nowak-Wegrzyn A**. Clinical features and resolution of food protein-induced enterocolitis syndrome: 10-year experience. *J Allergy Clin Immunol.* 2014 Aug;134(2):382-9.

Bloom KA, Huang FR, Bencharitiwong R, Bardina L, Ross A, Sampson HA, **Nowak-Wegrzyn A**. Effect of Heat Treatment on Milk and Egg Proteins Allergenicity. *Pediatr Allergy Immunol*. 2014 Sep 24.

Konstantinou GN, Ramon B, Grishin A, Caubet JC, Bardina L, Sicherer SH, Sampson HA, **Nowak-Wegrzyn A.** The role of casein-specific IgA and TGF-β in children with Food Protein-Induced Enterocolitis Syndrome to milk. *Pediatr Allergy Immunol.* 2014 Oct 6.

Nowak-Wegrzyn A, Albin S. Oral immunotherapy for food allergy: mechanisms and role in management. *Clin Exp Allergy*. 2014 Jul 31.

Sampson HA, Aceves S, Bock SA, James J, Jones S, Lang D, Nadeau K, **Nowak-Wegrzyn** A, Oppenheimer J, Perry TT, Randolph C, Sicherer SH, Simon RA, Vickery BP, Wood R. Food allergy: A practice parameter update-2014. *J Allergy Clin Immunol.* 2014 Aug 28. pii: S0091-6749(14)00672-1. [Epub ahead of print]

Coro Paisán-Ruiz, Ph.D.

Assistant Professor of Neurology, Genetics and Genomic Sciences, and Psychiatry

Institute Affiliations: Mindich Child Health and Development Institute; Friedman Brain Institute

Lab/Location: Annenberg 22-38

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Research Interests: Dr. Paisan-Ruiz's laboratory focuses on elucidating and understanding the molecular basis underlying and contributing to movement disorders, such as Parkinson's disease, atypical parkinsonism, and essential tremor. By collaborating with internationally recognized physicians and employing state-of-art molecular techniques, her research team has identified the first pathogenic mutations in genes underlying Mendelian forms of movement disorders.

Type of Research: Basic/Translational

Publications:

Krebs KE*, Karkheiran S*, Powell JC, Cao M, Makarov V, et al. The Sac1 domain of *SYNJ1* identified mutated in a family with early-onset progressive parkinsonism with generalized seizures. Rapid communication. *Hum Mutat.* 2013 Sep; 34(9): 1200-12007.

Martí-Massó JF, Bergareche A, Makarov V, Ruiz-Martinez J, Gorostidi A, et al. Identification of a nonsense *ACMSD* mutation (p.W26X) in familial cortical myoclonic tremor with epilepsy and parkinsonism. *J Mol Med.* 2013 Dec; 91(12): 1399-1406.

Karkheiran S, Krebs CE, Darvish H, Asadian M, Shahidi GA, and **Paisán-Ruiz C**. Variable phenotypic expression in families with early-onset Parkinsonism due to *PRKN* mutations. *J. Neurol.* 2014 Jun;261(6):1223-1226.

Deik A, Johannes B, Rucker JC, Sánchez E, Brodie SE, et al. Boucher-Neuhäuser syndrome due to *PNPLA6* mutations is an etiology of late onset ataxia**.** *J. Neurol.* 2014 Dec; 261(12): 2411-2423.

Dalila Pinto, Ph.D.

Assistant Professor of Psychiatry, and Genetics and Genomic Sciences

Institution Affiliations: Mindich Child Health and Development Institute; Friedman Brain Institute; Seaver Center; Icahn Institute for Multiscale Biology

Lab/Location: Hess CSM 8-115

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Research Interests: Dr. Pinto's laboratory focuses on identifying risk factors and pathways involved in neurodevelopmental disorders (including autism, epilepsy, intellectual disability, and Rett syndrome-like). By using a combination of innovative high-throughput experimental and bioinformatics approaches, her lab maps and characterizes various forms of genetic variation (deletions, duplications, complex rearrangements, and point-mutations) that are further integrated with coding and non-coding gene expression, epigenetics and clinical data to shed light on the mechanisms underlying these disorders.

Type of Research: Basic/Translational

Publications:

Pinto D, Delaby E, Merico D, et al. Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *Am J Hum Genet* 2014 May 1;94(5):677-94.

Anney R, Klei L, **Pinto D**, et al. Individual common variants exert weak effects on risk for autism spectrum disorders. *Hum Mol Genet* 2012 Nov 1;21(21):4781-92.

Pinto D, Darvishi K, Shi X, et al. Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. *Nature Biotech* 2011 May 8;29(6):512-20.

Pinto D, Pagnamenta AT, Klei L, et al. Functional impact of global rare copy number variation in autism spectrum disorder. *Nature* 2010 Jul 15;466(7304):368-72.

Conrad DF*, **Pinto D***, Redon R, et al. Origins and functional impact of copy number variation in the human genome. *Nature*. 2010 Apr 1;464(7289):704-12.

Francesco Ramirez, D.Sc.

Professor of Pharmacology and Systems Therapeutics, Medicine (Cardiology) and Orthopaedics

Lab/Location:
Annenberg 19-64A

Email:
francesco.ramirez@mssm.edu

Research Interests: Dr. Ramirez is a molecular geneticist with a long standing interest in heritable disorders of connective tissue. Current work focuses on elucidating the pathogenesis of cardiovascular and musculoskeletal abnormalities in mouse models of Marfan syndrome, and on identifying new therapeutic means to mitigate progression of these systemic manifestations.

Type of Research: Basic/Translational

Publications:

Cook JR, Carta L, Benard L, Chemaly ER, Chiu E, Rao SK, Hampton TG, Yurchenco P, GenTAC Registry Consortium, Costa KD, Hajjar RJ, **Ramirez F**. Abnormal muscle mechanosignaling triggers cardiomyopathy in mice with Marfan syndrome. *J. Clin Invest*. 2014; 124:1329-1339.

Cook, J.R., Smaldone S., Cozzolino C., del Solar M., Lee-Arteaga S., Nistala H. and **Ramirez F**. Generation of Fbn1 conditional null mice implicates microfibrils in osteoprogenitor recruitment. *Genesis* 2012; 50:635-641.

Nistala H., Lee-Arteaga S., Carta L., Cook J.R., Smaldone S., Siciliano G., Rifkin A.N., Dietz H.C., Rifkin D.B., and **Ramirez F**. Differential effects of alendronate and losartan therapy on osteopenia and aortic aneurysm in mice with severe Marfan syndrome. *Hum. Mol. Genet.* 2010; 19:4790-4798.

Nistala H., Lee-Arteaga S., Smaldone S., Siciliano G., Ono R., Sengle G., Arteaga-Solis E., Levasseur R, Ducy P., Sakai L.Y., Karsenty G., and **Ramirez F**. Fibrillin-1 and -2 differentially modulate endogenous TGFβ and BMP bioavailability during bone formation. *J. Cell Biol.* 2010; 190:1107-1121.

Ramirez, F. and Rifkin D.B. Extracellular microfibrils: contextual platforms for TGFb and BMP signaling. *Curr. Opin. Cell Biol.* 2009; 21:616-622.

Robert Rapaport, M.D.

Professor of Pediatrics and Director of the Division of Pediatric Endocrinology and Diabetes

Lab/Location:
Annenberg 4 Room 4-81

Email:
robert.rapaport@mountsinai.org

Research Interests: Dr. Rapaport's research interests are focused on three main areas: growth, growth hormone treatment and metabolic aspects of children born small for gestational age, neonatal thyroid disease and pubertal disorders.

Type of Research: Clinical/Translational

Publications:

Klein M, Iazzetti L, Speiser P, Carey D, Shelov S, Accacha S, Fennoy I, Rosenbaum M, **Rapaport R**. Alanine transferase: an independent indicator of adiposity related comorbidity risk in youth. *J Diabetes*. 2014; Sept 30. [Epub ahead of print].

Rashid N, Saenger P, Wu YL, Woehling H, Frankel M, Lifshitz F, Muenzberg M, **Rapaport R**. Switching to Omnitrope from other recombinant human growth hormone therapies: a retrospective study in an integrated healthcare system. *Biol Ther*. 2014; Aug 6. [Epub ahead of print].

Graber E, Regelmann MO, Annunziato R, Machac J, **Rapaport R**. The role of I imaging in the evaluation of infants with mild congenital hypothyroidism. *Horm Res Paediatr*. 2014; 16. [Epub ahead of print]

Regelmann MO, **Rapaport R**. Growth hormone treatment in patients with hypochondroplasia. *Horm Res Paediatr*. 2014; 16:35-354.

Avi Reichenberg, Ph.D.

Professor of Psychiatry and Preventative Medicine

Institution Affiliations: Seaver Center for Autism; Mindich Child Health and Development Institute

Lab/Location: CAM West Tower D5-143

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Research Interests: Dr. Reichenberg's research group focuses on the role of environmental and familial factors in the etiology of developmental and psychotic disorders. His work includes human population-based studies, molecular genetic and epigenetic, bioinformatic methods and animal models. The goal is to gain better insight into the causes of psychiatric disorders, particularly autism and schizophrenia.

Type of Research: Basic/Translational/Clinical

Publications:

Sandin S, Nygren KG, Iliadou A, Hultman CM, **Reichenberg A**. Autism and mental retardation among offspring born after in vitro fertilization. *JAMA*. 2013 Jul 3;310(1):75-84.

Frans EM, Sandin S, **Reichenberg A**, Långström N, Lichtenstein P, McGrath JJ, Hultman CM. Autism risk across generations: a population-based study of advancing grandpaternal and paternal age. *JAMA Psychiatry*. 2013 May;70(5):516-21.

Meier MH, Caspi A, **Reichenberg A**, Keefe RS, Fisher HL, Harrington H, Houts R, Poulton R, Moffitt TE. Neuropsychological Decline in Schizophrenia From the Premorbid to the Postonset Period: Evidence From a Population-Representative Longitudinal Study. *Am J Psychiatry*. 2013 Sep 13.

Smith RG, **Reichenberg A**, Kember RL, Buxbaum JD, Schalkwyk LC, Fernandes C, Mill J. Advanced paternal age is associated with altered DNA methylation at brain-expressed imprinted loci in inbred mice: implications for neuropsychiatric disease. *Mol Psychiatry*. 2013 Jun;18(6):635-6.

Nosarti C, **Reichenberg A**, Murray RM, Cnattingius S, Lambe MP, Yin L, MacCabe J, Rifkin L, Hultman CM. Preterm birth and psychiatric disorders in young adult life. *Arch Gen Psychiatry*. 2012 Jun;69(6):E1-8.

Michael Rendl, M.D.

Associate Professor, Department of Developmental and Regenerative Biology, Department of Dermatology

Institute Affiliations: Black Family Stem Cell Institute; Mindich Child Health and Development Institute

Lab/Location: Atran 7-10C

Email: michael.rendl@mssm.edu

Research Interests: Dr. Rendl's lab studies the function of stem cell niches. They utilize genetic mouse models for embryonic hair follicle formation and adult hair regeneration to uncover how Dermal Papilla niche cells instruct hair follicle stem cells. Insights from these studies will provide a platform for developing hair regenerative therapies.

Type of Research: Basic/ Translational

Publications:

Tsai SY, Sennett R, Rezza A, Clavel C, Grisanti L, Zemla R, Najam S, **Rendl M**. Wnt/β-catenin signaling in dermal condensates is required for hair follicle formation. *Developmental biology.* 2014 Jan; 385(2).

Grisanti L, Rezza A, Clavel C, Sennett R, **Rendl M**. Enpp2/Autotaxin in dermal papilla precursors is dispensable for hair follicle morphogenesis. *The Journal of investigative dermatology.* 2013 Oct; 133(10).

Clavel C, Grisanti L, Zemla R, Rezza A, Barros R, Sennett R, Mazloom AR, Chung CY, Cai X, Cai CL, Pevny L, Nicolis S, Ma'ayan A, **Rendl M**. Sox2 in the dermal papilla niche controls hair growth by fine-tuning BMP signaling in differentiating hair shaft progenitors. *Developmental cell.* 2012 Nov; 23(5).

Grisanti L, Clavel C, Cai X, Rezza A, Tsai SY, Sennett R, Mumau M, Cai CL, **Rendl M**. Tbx18 targets dermal condensates for labeling, isolation, and gene ablation during embryonic hair follicle formation. *The Journal of investigative dermatology.* 2013 Feb; 133(2).

Sennett R, **Rendl M**. Mesenchymal-epithelial interactions during hair follicle morphogenesis and cycling [review]. *Seminars in cell & developmental biology.* 2012 Oct; 23(8).

Laura Rodriguez-Murillo, Ph.D.
Assistant Professor of Pediatrics and Genetics and Genomic Sciences/ Pediatric and Genetics and Genomic Sciences Department

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Hess CSM 8-108

Email: laura.rodriguezmurillo@mssm.edu

Research Interests: Dr. Murillo's research is on investigating the role of genetic variation on the etiology of complex disorders. Her main focus is in the genetics of congenital heart disease, applying methods in genetics and genomics to array and sequencing data to study plausible candidate genes and their interactions that may cause congenital heart disease.

Type of Research: Basic/Translational

Publications:

T. Karayannis, E. Au, J. C. Patel, ... **Rodriguez- Murillo L.**, N. C. Roy, J. A. Gogos, B. Rudy, M. E. Rice, M. Karayiorgou, H. Hakonarson, B. Keren, G. Huguet, T. Bourgeron, C. Hoeffer, R. W. Tsien, E. Peles & G. Fishell. Cntnap4 differentially contributes to GABAergic and dopaminergic synaptic transmission. *Nature*. 2014; Jul 10;511(7508):236-40.

Backenroth D., Homsy J., **Rodriguez-Murillo L.**, Glessner J., Lin E., Brueckner M., Lifton R., Goldmuntz E., Chung, W., Shen Y. Detecting rare copy number variants from whole exome sequencing data. *Nucleic Acids Res.* 2014; Jul;42(12):e97.

Rodriguez-Murillo L., Xu B., Roos J.L., Abecasis G.R., Gogos J.A., Karayiorgou M. Fine mapping on chromosome 13q32-34 and brain expression analysis implicates MYO16 in schizophrenia. Neuropsychopharmacology. 2014 Mar;39(4):934-43.

Rodriguez-Murillo L., Subaran R., Stewart W.C.L., Sreemanta P., Marathe S., Barst R.J., Chung W.K., Greenberg D.A. Novel loci interacting epistatically with Bone Morphogenetic Protein Receptor 2 cause familial pulmonary arterial hypertension. Journal of Heart and Lung Transplantation. 2010; 29(2): 174-180.

Xu B., Woodroffe A., **Rodriguez-Murillo L.**, Roos J.L., van Rensburg E.J., Abecasis G.R., Gogos J.A., Karayiorgou M. Elucidating the genetic architecture of familial schizophrenia using rare copy number variant and linkage scans. Proceedings of the National Academy of Sciences USA. 2009; 106(39): 16746-16751.

Jeffrey M. Saland, M.D., M.S.C.R.
Associate Professor of Pediatrics (Nephrology) and Division Chief of Nephrology

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Annenberg 14-22

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Research Interests:
Dr. Saland participates in local and multicenter clinical studies of chronic kidney disease. He has focused on disorders of lipoprotein metabolism and cardiovascular complications in children with CKD. Dr. Saland has also been active in developing treatment for children with atypical hemolytic uremic syndrome.

Type of Research: Clinical/Translational

Publications:

Sarnak, M.J., Bloom, R., Muntner, P., Rahman, M., **Saland, J.M.**, Wilson, P.W., and Fried, L. KDOQI US Commentary on the 2013 KDIGO Clinical Practice Guideline for Lipid Management in CKD. *Am J Kidney Dis*. 2014; Nov 18. [Epub ahead of print.]

Matloff, R.G., Arnon, R., and **Saland, J.M.** The kidney in pediatric liver transplantation: An updated perspective. *Pediatr Transplant.* 2012; 16:818-828.

Brady, T.M., Schneider, M.F., Flynn, J.T., Cox, C., Samuels, J., **Saland, J.**, White, C.T., Furth, S., Warady, B.A., and Mitsnefes, M. 2012. Carotid Intima-Media Thickness in Children with CKD: Results from the CKiD Study. *Clin J Am Soc Nephrol*. 2012; Dec 7(12):1930-7. [Epub 2012 Sep 13.]

Saland, J.M., Pierce, C.B., Mitsnefes, M.M., Flynn, J.T., Goebel, J., Kupferman, J.C., Warady, B.A., and Furth, S.L. Dyslipidemia in children with chronic kidney disease. *Kidney Int*. 2010; Dec;78(11):1154-63. [Epub 2010 Aug 25.]

Saland, J.M., Ruggenenti, P., and Remuzzi, G. Liver-kidney transplantation to cure atypical hemolytic uremic syndrome. *J Am Soc Nephrol*. 2009; 20:940-949.

Hugh A. Sampson, M.D.

Professor of Pediatrics (Allergy and Immunology)
Dean for Translational Biomedical Sciences

Institute Affiliations: Jaffe Food Allergy Institute (Director); Mindich Child Health and Development Institute

Lab/Location: Annenberg 17-40; 17-46; 17-60

Email: hugh.sampson@mssm.edu

Research Interests: Dr. Sampson's research interests have focused on food allergic disorders including work on the immuno-pathogenic role of food hypersensitivity in atopic dermatitis, the pathogenesis of food-induced anaphylaxis, characterization of food-induced gastrointestinal hypersensitivities, molecular characterization of food allergens, and more recently immunotherapeutic strategies for treating food allergies.

Type of Research: Clinical/Translational

Publications:

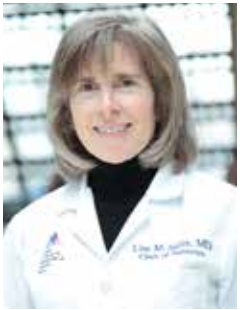
Sicherer SH, Wood RA, Vickery BP, Jones SM, Liu AH, Fleischer DM, Dawson P, Mayer L, Burks AW, Grishin A, Stablein D, **Sampson HA**. The natural history of egg allergy in an observational cohort. *J Allergy Clin Immunol.* 2014; 133:492-499.

Tordesillas L, Goswami R, Benedé S, Grishina G, Dunkin D, Järvinen KM, Maleki SJ, **Sampson HA**, Berin MC. Skin exposure promotes a Th2-dependent sensitization to peanut allergens. *J Clin Invest.* 2014; 124(11):4965-75.

Pascal M, Konstantinou GN, Masilamani M, Lieberman J, **Sampson HA**. In silico prediction of Ara h 2 T cell epitopes in peanut-allergic children. *Clin Exp Allergy*. 2013 Jan;43(1):116-27.

Wood RA, Sicherer SH, Burks AW, Grishin A, Henning AK, Lindblad R, Stablein D, **Sampson HA**. A phase 1 study of heat/phenol-killed, E. coli-encapsulated, recombinant modified peanut proteins Ara h 1, Ara h 2, and Ara h 3 (EMP-123) for the treatment of peanut allergy. *Allergy*. 2013 Jun;68(6):803-8.

Burks AW, Jones SM, Wood RA, Fleischer DM, Sicherer SH, Lindblad RW, Stablein D, Henning AK, Vickery BP, Liu AH, Scurlock AM, Shreffler WG, Plaut M, **Sampson HA**; Consortium of Food Allergy Research (CoFAR). Oral immunotherapy for treatment of egg allergy in children. *N Engl J Med*. 2012;Jul 19;367(3):233-43.



Lisa M. Satlin, M.D.

Professor (Nephrology) and Chair of Pediatrics

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Annenberg 14-18, -19

Email: lisa.satlin@mssm.edu

Research Interests:
Dr. Satlin's developmental physiology lab focuses on exploring the molecular mechanisms underlying the ability of the maturing kidney to adjust sodium and potassium balance during periods of somatic growth, and the role of variations in urinary flow rate in the mechanoregulation of ion channels and transport proteins in the kidney in health and disease.

Type of Research: Basic/Translational

Publications:

Wei Y, Liao Y, Zavilowitz B, Ren J, Liu W, Chan P, Rohatgi R, Estilo G, Jackson EK, Wang W, and **Satlin LM**. Angiotensin type 2 receptor regulates ROMK-like K channel activity in renal cortical collecting duct during high dietary potassium adaptation. *Am. J. Physiol. Renal Physiol.* 2014;307:F833-43.

Carrisoza-Gaytán R, Liu Y, Flores D, Else D, Lee HG, Rhodes G, Sandoval R, Kleyman T, Lee F, Molitoris B, **Satlin LM***, Rohatgi R*. Effects of biomechanical forces on signaling in the cortical collecting duct (CCD). *Am. .J Physiol. Renal Physiol.* 2014;307:F195-204 (*, contributed equally to this work)

Wang Z, Subramanya A, **Satlin LM**, Pastor-Soler N, Carattino M, Kleyman T. Regulation of large conductance Ca^{2+}- activated K^+ channels by WNK4 kinase. *Am. J. Physiol. Cell Physiol.* 2013;305:C846-53.

Flores D, Liu Y, Liu W, **Satlin LM**, and Rohatgi R. Flow induced prostaglandin E2 release regulates Na and K transport in the collecting duct. *Am. J. Physiol. Renal Physiol.* 303: F632-8, 2012 (named as *2012 AJP: Renal Paper of the Year*)

Rbaibi Y, Cui S, Mo D, Carattino M, Rohatgi R, **Satlin LM**, Szalinski CM, Swanhart LM, Fölsch H, Hukriede NA, Weisz OA. OCRL1 modulates cilia length in renal epithelial cells. *Traffic.* 2012;13:1295-305.

Kurt P. Schulz, Ph.D.

Assistant Professor of Psychiatry

Lab/Location: 19 East 98th St., 5th Floor

Email: kurt.schulz@mssm.edu

Research Interests: Dr. Schulz uses behavioral, neuropsychological, neuroendocrine and functional imaging techniques to study the development of cognitive processes in children with attention-deficit/hyperactivity disorder (ADHD) and other disruptive behavioral disorders. His current research interests include neurobiological mechanisms of ADHD treatments and ancillary emotion problems.

Type of Research: Clinical/Translational

Publications:

Clerkin, S.M., **Schulz, K.P.**, Berwid, O.G., Fan, J., Newcorn, J.H., Tang, C.Y., & Halperin, J.M. Presence of response preparation deficits despite remission of attention-deficit/ hyperactivity disorder symptoms in adulthood. *Am J Psychiatry.* 2013;170, 1011-1019.

Schulz, K.P., Clerkin, S.M., Fan, J., Halperin, J.M., & Newcorn, J.H. Guanfacine modulates the influence of emotional cues on prefrontal cortex activation for cognitive control. *Psychopharmacology.* 2013;*226,* 261-271.

Schulz, K.P., Fan, J., Bédard, A.C., Clerkin, S.M., Ivanov, I., Tang, C.Y., Halperin, J.M., & Newcorn, J.H. Common and unique therapeutic mechanisms of stimulant and non-stimulant treatments for attention-deficit/hyperactivity disorder. *Arch Gen Psychiatry.* 2012;69, 952-961.

Schulz, K.P., Clerkin, S.M., Halperin, J.M., Newcorn, J.H., Tang, C.Y., & Fan, J. Dissociable neural effects of stimulus valence and preceding context during the inhibition of responses to emotional faces. *Hum Brain Map.* 2009;30, 2821-2833.

Clerkin, S.M., **Schulz, K.P.**, Halperin, J.M., Newcorn, J.H., Ivanov, I., Tang, C.Y., & Fan, J. Guanfacine potentiates the activation of prefrontal cortex evoked by warning cues. *Biol Psychiatry.* 2009;66, 307-312.

Donald Scott, Ph.D.

Professor of Medicine (Endocrinology)

Institute Affiliation: Mindich Child Health and Development Institute; Obesity, Diabetes and Metabolism Institute

Lab/Location: Atran 5-17

Email: donald.scott@mssm.edu

Research Interests: Dr. Scott has a longstanding interest in how nutrients change cellular phenotype. Dr. Scott has focused on two transcription factors, ChREBP and Myc. The Myc/ChREBP relationshio reflects fundamental cellular adaptations to varying metabolic environments, and is applicable to a wide range of diseases, including diabetes, cancer, cardiovascular disease and aging.

Type of Research: Basic/Translational

Publications:

Stewart, A.F., Wang, P., Fiaschi-Taesch, N., Vasavada, R., **Scott, D.K**., and Garcia-Ocana, A. Diabetes Mellitus-advances and challenges in human β-cell proliferation. *Nat Rev Endo*. 2015. (in press)

Wang, P, Felsenfeld, DP, Liu, H., BA, Sivendran, S., Bender A., Sanchez, R. **Scott, D.K**, Stewart, A.F. Induction of Rodent and Human Pancreatic Beta Cell Replication By Inhibitors of Dual Specificity, Tyrosine-Regulated Kinase 1a. *Nature Medicine*. 2015. (in press)

Edmunds, L.R., Sharma, L., Kang, A., Lu, J., Vockley, J., Basu, S., Uppala, R., Goetzman, E.S., Beck, M., **Scott, D.K.,** and Prochownik, E.V. c-Myc programs fatty acid metabolism and dictates acetyl CoA abundance and fate. *J. Biol. Chem.* 2014;289:25382-25392.

Metukuri, M.R., Zhang, P., Alonso, L.C., Takane, K., Gramignoli, R., Strom, S.C., O'Doherty, R.M., Stewart, A.F., Vasavada, R.C., Garcia-Ocana, A., **Scott, D.K.**. ChREBP mediates glucose-stimulated pancreatic beta cell proliferation. *Diabetes*. 2012;61:2004-2015.

Zhang, P, Metukuri, M.R., Bindom, S.M., Prochownik, E.V., O'Doherty, R.M., **Scott, D.K**. c-Myc is required for the ChREBP-dependent activation of glucose-responsive genes. *Mol. Endocrinol. 201;*24:1274-1286.

Andrew Sharp, Ph.D.

Associate Professor of Genetics and Genomic Sciences

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Hess CSM 8-301

Email: andrew.sharp@mssm.edu

Research Interests: The Sharp lab is an integrated research environment combining both experimental and bioinformatic approaches. Dr. Sharp's research uses genomic approaches to perform basic studies of human genome function, and a wide variety of diseases, including congenital disorders such as cleft lip/palate and spina bifida, and chromosomal disorders such as translocations and microdeletions.

Type of Research: Basic/Translational

Publications:

Garg P, Ludwig KU, Böhmer AC...**Sharp AJ**. Genome-wide analysis of parent-of-origin effects in non-syndromic orofacial clefts. *Eur J Hum Genet*. 2013; in press

Borel C, Cheung F, Stewart H...**Sharp AJ**. Evaluation of PRDM9 variation as a risk factor for recurrent genomic disorders and chromosomal non-disjunction. *Human Genet*. 2012, 131:1519-1524

Sharp AJ, Migliavacca E, Dupre Y, Stathaki E, Sailani MR, Mackay D, Robinson DO, Cobellis G, Cobellis L, Brunner H, Steiner B, Antonarakis SE. Methylation profiling in cases with uniparental disomy identifies novel differentially methylated regions on chromosome 15. *Genome Res*. 2010, 20:1271-8

Mefford HC*, **Sharp AJ***, Baker C, Itsara A, Jiang Z, Buysse K, et al. Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New Engl J Med*. 2008; 359:1685-1699. *Contributed equally to this work

Sharp AJ, Mefford HC, Li KE, Broomer AJ, Wang Y, Xiao C, Barbacioru C, Skinner C, Baker C, Stevenson RE, Schroer R, Novara F, De Gregori M, Ciccone R, Gimelli G, Dalla Bernardina B, Torniero C, Giorda R, Regan R, Murday V, Mansour S, Fichera M, Castiglia L, Failla P, Cooper GM, Knight SJL, Romano C, Zuffardi O, Chen C, Schwartz C, Eichler EE. A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. *Nature Genet*. 2008, 40:322-8.

Eyal Shemesh, M.D.

Associate Professor of Pediatrics and Psychiatry; Division Chief of Developmental and Behavioral Pediatrics

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Annenberg 4-54

Email: eyal.shemesh@mssm.edu

Research Interests: Dr. Shemesh, who trained as both a pediatrician and a child psychiatrist, studies ways to assess and then improve the well-being (especially emotional well-being) of children and adults who suffer from medical illnesses and their families, with a particular focus on traumatic stress reactions and nonadherence to medical regimens.

Type of Research: Clinical/Translational

Publications:

Shemesh E, Annunziato RA, Ambrose MA, Ravid NL, Mullarkey C, Rubes M, Chuang K, Sicherer M, Sicherer SH. Child and parental reports of bullying in a consecutive sample of children with food allergy. *Pediatrics*. 2013 Jan;131(1):e10-7.

Shemesh E, Annunziato RA, Weatherley BD, Cotter G, Feaganes JR, Santra M, Yehuda R, Rubinstein D. A randomized controlled trial of the safety and promise of cognitive-behavioral therapy using imaginal exposure in patients with posttraumatic stress disorder resulting from cardiovascular illness. *J Clin Psychiatry*. 2011;Feb;72(2):168-74.

Shemesh E., Annunziato RA, Rubinstein D, Sultan S, Malhotra J, Santra M, Weatherley BD, Feaganes JR, Cotter G, Yehuda R. Screening for Depression and Suicidality in patients with Cardiovascular Illnesses. *Am J Cardiol.* 2009;Nov 1;104(9):1194-7.

Shemesh E, Yehuda R, Rockmore L, Shneider BL, Emre S, Bartell AS, Schmeidler J, Annunziato RA, Stuber ML, Newcorn JH. Assessment of Depression in Medically Ill Children Presenting to Pediatric Specialty Clinics. *J Am Acad Child Adolesc Psychiatry*. 2005;Dec;44(12):1249-1257.

Shemesh E, Shneider BL, Savitzky JL, Arnott L, Gondolesi G, Krieger N, Kerkar N, Magid M, Stuber ML, Schmeidler J, Yehuda R, Emre S. Medication adherence in pediatric and adolescent liver transplant Recipients. *Pediatrics*. 2004;Apr;113(4):825-32.

Scott H. Sicherer, M.D.

Professor of Pediatrics (Allergy and Immunology) and Division Chief of Allergy and Clinical Immunology; Medical Director, Clinical Research Center

Institute Affiliation: Jaffe Food Allergy Institute, Mindich Child Health and Development Institute

Lab/Location: Annenberg 17-90

Email:scott.sicherer@mssm.edu

Research Interests: Dr. Sicherer has a strong interest in food allergy with research focusing on epidemiology, psychosocial issues, prevention, modalities to educate physicians and parents about food allergy, daily management, natural history, and novel therapeutics.

Type of Research: Clinical/Translational

Publications:

Wood RA*, **Sicherer SH***[*Shared First author]**,** Vickery BP, Jones SM, Liu AH, Fleischer DM, Henning AK, Mayer L, Burks AW, Grishin A, Stablein D, Sampson HA. The natural history of milk allergy in an observational cohort. J Allergy Clin Immunol. *J Allergy Clin Immunol*. 2013 Mar;131(3):805-12.

Wood RA*, **Sicherer SH*[**Shared First author], Burks AW, Grishin A, Henning AK, Lindblad R, Stablein D, Sampson HA. A phase 1 study of heat/phenol-killed, E. coli-encapsulated, recombinant modified peanut proteins Ara h 1, Ara h 2, and Ara h 3 (EMP-123) for the treatment of peanut allergy. *Allergy*. 2013 Jun;68(6):803-8

Shemesh E, Annunziato RA, Ambrose MA, Ravid NL, Mullarkey C, Rubes M, Chuang K, Sicherer M, **Sicherer SH**. Child and parental reports of bullying in a consecutive sample of children with food allergy. *Pediatrics*. 2013 Jan;131(1):e10-7

Sicherer SH, Vargas PA, Groetch ME, Christie L, Carlisle SK, Noone S, Jones SM. Development and Validation of Educational Materials for Food Allergy. *J Pediatr*. 2012 Apr;160(4):651-6.

Fleischer DM, Perry TT, Atkins D, Wood RA, Burks AW, Jones SM, Henning AK, Stablein D, Sampson HA, **Sicherer SH**. Allergic reactions to foods in preschool-aged children in a prospective observational food allergy study. *Pediatrics*. 2012 Jul;130(1):e25-32

Mary V. Solanto, Ph.D.

Associate Professor of Psychiatry in Child Psychiatry
Director, ADHD Center

Lab/Location: 19 East 98th St.; Suite 5-D

Email: mary.solanto@mssm.edu

Research Interests: Dr. Solanto's work has focused on the diagnosis and neuropsychology of ADHD in children and adults, the effects of stimulant medication on cognitive and behavioral functioning, and the mechanisms of action of treatment. She innovated a cognitive-behavioral treatment for adult ADHD, and currently also has a particular interest in interventions to facilitate adherence to medication treatment in children with ADHD.

Type of Research: Clinical/Translational

Publications:

Willcut E, Nigg JT, Pennington BF, **Solanto MV**, Rohde LA, Tannock R, Loo SK, Carlson CL, McBurnett K. Validity of DSM-IV attention–deficit/hyperactivity disorder symptom dimensions and subtypes. *J Abnorm Psychol.* 2012; epub ahead of print

Solanto MV, Marks DM, Wasserstein J, Mitchell K. Diagnosis of ADHD in Adults: What is the Appropriate DSM-5 Symptom Threshold for Hyperactivity-Impulsivity? *J Atten Disord.* 2012, epub ahead of print.

Solanto MV, Marks DJ, Wasserstein J, Mitchell K, Abikoff H, Alvir JM, Kofman MD. Efficacy of meta-cognitive therapy (MCT) for adult ADHD. *Am J Psychiatry*. 167(8): 958-968.

Solanto MV, Newcorn JN, Gilbert SN, Vail L, Raj A, Ivanov I, Lara R. Stimulant drug response in ADHD, Predominantly Inattentive subtype. *J Child Adolesc Psychopharmacol.* 2009; 19(6): 663-671.

Solanto MV, Gilbert SN, Raj A, Zhu J, Pope-Boyd S, Stepak B, Vail L, Newcorn JH. Neurocognitive functioning in AD/HD, Predominantly Inattentive Subtype. *J Abnorm Child Psychol.* 2007; 35(5): 729-744.

Philippe M. Soriano, Ph.D.
Professor of Developmental and Regenerative Biology and Oncological Sciences

Institute Affiliation:
Tisch Cancer Institute (Associate Director)

Lab/Location:
Annenberg 25-70

Email: philippe.soriano@mssm.edu

Research Interests: Dr. Soriano is a developmental biologist who studies growth factor signaling pathways that have important roles in mouse craniofacial development and in stem cells of the early embryo. His laboratory uses state-of-the-art molecular genetic approaches in the mouse to address the general question of how biological specificity is acquired upon engagement of growth factor signaling.

Type of Research: Basic

Publications:

Vasudevan, H.N. and **Soriano, P.** SRF regulates craniofacial development through selective recruitment of MRTF cofactors by PDGF signaling. *Dev. Cell.* 2014; 31;332-344.

Fantauzzo, K.A. and **Soriano, P.** PI3K-mediated PDGFRα signaling regulates survival and proliferation in skeletal development through p53-dependent intracellular pathways. *Genes Dev.* 2014; 28:1005-1017.

He, F. and **Soriano, P.** A critical role for PDGFRa signaling in medial nasal process development. *PLoS Gen.* 2013; 9, e1003851.

Olson, L.E. and **Soriano, P.** PDGFRb Signaling Regulates Mural Cell Plasticity and Inhibits Fat Development. *Dev. Cell.* 2011; 20:815-826.

Bush, J.O. and **Soriano, P.** Ephrin-B1 forward signaling regulates craniofacial morphogenesis by controlling cell proliferation across Eph-ephrin boundaries. *Genes Dev.* 2010; 24:2068-2080.

Mihaela Stefan, Ph.D.
Assistant Professor of Medicine, Endocrinology, Diabetes and Bone Disease

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location:
Annenberg Rm 18-20

Phone: 212-241-1728

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Research Interests: Dr. Stefan's research focuses on how environmental-induced epigenetic changes affect gene expression and contribute to the development of autoimmune diseases (AID), identification of AID causative gene variants, finding the mechanisms by which these variants trigger the diseases and, identification of cell-type specific functional epigenetic features associated with autoimmunity.

Type of Research: Basic/Translational

Publications:

Genetic-epigenetic dysregulation of thymic TSH receptor gene expression triggers thyroid autoimmunity. **Stefan M**, Wei C, Lombardi A, Li CW, Concepcion ES, Inabnet WB 3rd, Owen R, Zhang W, Tomer Y. *Proc Natl Acad Sci. U S A.* 2014;111(34):12562-7.

Stefan M, Zhang W, Concepcion E, Yi Z, Tomer Y. DNA methylation profiles in type 1 diabetes twins point to strong epigenetic effects on etiology. *J Autoimmun.* 2014; 50:33-7.

Hasham A, Zhang W, Lotay V, Haggerty S, **Stefan M**, Concepcion E, Dieterich DT, Tomer Y. Genetic analysis of interferon induced thyroiditis (IIT): Evidence for a key role for MHC and apoptosis related genes and pathways. *J Autoimmun.* 2013; 44:61-70.

Tomer Y, Hasham A, Davies TF, **Stefan M**, Concepcion E, Keddache M, Greenberg AD, Fine mapping of loci linked to autoimmune thyroid disease identifies novel susceptibility genes. *J Clin Endocrinol Metab.* 2013; 98(1):E144-52.

Henson BJ, Zhu W, Hardaway K, Wetzel JL, **Stefan M**, Albers KM, Nicholls RD, Transcriptional and post-transcriptional regulation of *SPAST*, the gene most frequently mutated in hereditary spastic paraplegia. *PLoS ONE*. 2012; 7(5): e36505.

Cheryl R. Stein, Ph.D.

Assistant Professor of Preventive Medicine

Lab/Location: CAM D2-135

Email: cheryl.stein@mssm.edu

Research Interests: Dr. Stein is a perinatal epidemiologist and her research focus is environmental exposures during pregnancy and their effect on pregnancy health, birth outcomes, and child development.

Type of Research: Clinical/Translational

Publications:

Stein CR, Savitz DA, Elston B, Thorpe PG, Gilboa SM. Perfluorooctanoate Exposure and Major Birth Defects. *Reproductive Toxicology*. 2014;47:15-20.

Stein CR, Savitz DA, Bellinger DC. Perfluorooctanoate Exposure in a Highly Exposed Community and Parent and Teacher Reports of Behavior in 6 – 12 Year Old Children*. Paediatric and Perinatal Epidemiology.* 2014;28(2):146–156. (PMC Journal – In Process.)

Darrow LA, **Stein CR,** Steenland NK. Serum Perfluorooctanoic Acid and Perfluorooctane Sulfonate Concentrations and Birth Outcomes in the Mid-Ohio Valley, 2005-2010. *Environmental Health Perspectives.* 2013;121(10):1207–1213.

Stein CR, Savitz DA, Bellinger DC. Perfluorooctanoate exposure and neuropsychological outcomes in 6 – 12 year old children. *Epidemiology*. 2013;24(4)590-599.

Barría MI, Garrido JL, **Stein CR**, Scher E, Ge Y, Engel SM, Kraus TA, Banach D, Moran TM. Localized mucosal response to intranasal live attenuated influenza vaccine in adults. *Journal of Infectious Diseases*. 2013;207(1)115-124.

Andrew F. Stewart, M.D.

Irene and Dr. Arthur M. Fishberg Professor of Medicine

Institute Affiliation: Diabetes, Obesity and Metabolism Institute (Director)

Lab/Location: Atran 5

Email: andrew.stewart@mssm.edu

Research Interests: The Stewart lab works on induction of proliferation and expansion of human pancreatic beta cells for regenerative and replacement therapies for Types 1 and 2 diabetes mellitus.

Type of Research: Basic

Publications:

Fiaschi-Taesch N, Kleinberger JW, Salim F, Troxell R, Cox AE, Takane KK, Scott DK, **Stewart AF**. Developing A Human Pancreatic Beta Cell G1/S Molecule Atlas. *Diabetes* 2013; 62:2450-59.

Fiaschi-Taesch NM, Kleinberger JW, Salim F, Troxell R, Cox AE, Takane KK, Srinivas H, Scott DK, **Stewart AF**. Cytoplasmic-Nuclear Trafficking of G1/S Cell Cycle Molecules and Adult Human Beta Cell Replication: A Revised Model of Human Beta Cell G1/S Control. *Diabetes* 2013; 62:2460-70.

Takane KK, Kleinberger J, Salim F, Fiaschi-Taesch NM, Scott DK, **Stewart AF**. Regulated and reversible induction of adult human beta cell replication. *Diabetes* 2012; 61:418-24.

Metukuri MR, Zhang P, Basantani MK, Chin C, Stamateris RE, Alonso LC, Takane KK, Gramignoli R, Strom SC, O'Doherty RM, **Stewart AF**, Vasavada RC, Garcia-Ocaña A, Scott DK. ChREBP mediates glucose-stimulated pancreatic beta cell proliferation. *Diabetes* 2012; 61:2004-15.

Karslioglu E, Kleinberger J, Salim F, Cox A, Takane KK, Donald K. Scott DK, **Stewart AF**. cMyc is the principal upstream driver of beta cell proliferation in rat insulinoma cell lines and Is an effective mediator of human beta cell replication. *Mol Endocrinology* 2011;25:1760-72.

Annemarie Stroustrup, M.D., M.P.H.

Assistant Professor of Pediatrics (Newborn Medicine) and Preventive Medicine

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location:
1184 5th Avenue, P6-336

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Research Interests: Dr. Stroustrup's research focuses on modifiable causes of adverse outcomes in preterm and/or low birth weight neonates. She is particularly interested in understanding the impact of the hospital environment neurodevelopmental outcomes of NICU graduates.

Type of Research: Clinical/Translational

Publications:

Santos J, Pearce SE, **Stroustrup A**. Impact of Hospital-Based Environmental Exposures on Neurodevelopmental Outcomes of Preterm Infants. *Curr Opin Pediatr* 2015; *in press.*

Webb BD, Scharf RJ, Spear EA, Edelmann LJ, **Stroustrup A**. Evaluation of the Affymetrix CytoScan® Dx Assay for developmental delay. *Expert Rev Mol Diagn* 2015; *in press*.

Egbe A, Uppu S, **Stroustrup A**, Lee S, Ho D, Srivastava S. Incidence and sociodemographics of specific congenital heart diseases in the United States of America: an evaluation of hospital discharge diagnoses. *Pediatr Cardiol* 2014; 35(6):975-82.

Pierce LM, Raab EL, Holzman IR, Ginsburg RN, Brodie SE, **Stroustrup A**. Importance of birth weight as a risk factor for severe retinopathy of prematurity when gestational age is 30 or more weeks. *Am J Ophthalmol* 2014;157(6):1227-1230.

Stroustrup A, Plafkin C, Savitz D. Impact of physician awareness on diagnosis of fetomaternal hemorrhage. *Neonatology* 2014;105:250-255.

Shanna H. Swan, Ph.D.

Professor, Department of Preventive Medicine

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: CAM West Tower D3-135

Email: shanna.swan@mssm.edu

Research Interests: Dr. Shanna Swan is an Environmental and Reproductive Epidemiologist. Her research group examines the impact of environmental exposures on reproductive health and neurodevelopment in multi-center pregnancy cohort studies that include over 1,000 mothers and their children. This research is focused on identifying sex-differences in environmental effects, development and disease.

Type of Research: Clinical/Translational

Publications:

Bornehag C-G, Carlstedt F, Jönsson B, Lindh CH, Jensen TK, Bodin A, Jonsson C, Janson S, **Swan SH**. First trimester exposure to di-isononyl phthalate (DiNP) is associated with shorter anogenital distance in Swedish boys. *Environ Health Persect*. 2014; November.

Evans SF, Kobrosly RW, Barrett ES, Thurston SW, Calafat AM, Weiss B, Stahlhut R, Yolton K, **Swan SH**. Prenatal Bisphenol A Exposure and maternally reported behavior in boys and girls. *NeuroToxicology*. 2014; 45: 91-99.

Jensen TK, **Swan SH**, Jørgensen N, Toppari J, Redmon B, Punab M, Drobnis EZ, Haugen TB, Zilaitiene B, Sparks AE, Irvine DS, Wang C, Jouannet P, Brazil C, Paasch U, Salzbrunn A, Skakkebæk NE, Andersson AM. Alcohol and male reproductive health: a cross-sectional study of 8344 healthy men from Europe and the USA. *Hum Reprod.* 2014.

Sathyanarayana S, Barrett E, Butts S, Wang C, **Swan SH**. Phthalate exposure and reproductive hormone concentrations in pregnancy. *Reproduction.* 2014; Mar 2;147(4):401-9.

Barrett ES, Parlett LE, Redmon JB, **Swan SH**. Evidence for sexually dimorphic associations between maternal characteristics and anogenital distance, a marker of reproductive development. *Am J Epidemiol.* 2014; Jan 1;179(1):57-66.

Yaron Tomer, M.D.

Professor and Chief, Division of Endocrinology, Diabetes and Bone Disease

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Annenberg 18-20

E-mail: yaron.tomer@mssm.edu

Research Interests: Dr. Tomer's research focuses on the immunogenetic, epigenetic, and environmental mechanisms underlying the development thyroid autoimmunity, and type 1 diabetes. Specific areas of research include: analyzing peptide presentation by MHC II in thyroid autoimmunity and type 1 diabetes and developing novel strategies to block peptide presentation, mapping and analyzing susceptibility genes for autoimmune thyroiditis and diabetes, studies on the role of infection and genetic-epigenetic interactions in the development of autoimmune thyroiditis.

Type of Research: Basic/Translational

Publications:

Stefan M, Wei C, Lombardi A, Li CW, Concepcion E, Inabnet III WB, Owen R, Zhang W, **Tomer Y**. Genetic-epigenetic dysregulation of thymic TSH receptor gene expression triggers thyroid autoimmunity. *Proc Natl Acad Sci USA*. 2014; 111: 12562-12567.

Stefan M, Zhang W, Concepcion E, Yi Z, **Tomer Y**. DNA methylation profiles in type 1 diabetes twins point to strong epigenetic effects on etiology. *J Autoimmun.* 2014; 50: 33-37.

Tomer Y, Hasham A, Davies TF, Stefan M, Concepcion E, Keddache M, Greenberg DA. Fine mapping of loci linked to autoimmune thyroid disease identifies novel susceptibility genes. *J Clin Endocrinol Metab.* 2013; 98: E144-152.

Huber AK, Finkelman FD, Li CW, Concepcion E, Smith E, Jacobson E, Latif R, Keddache M, Zhang W, **Tomer Y**. Genetically driven target tissue overexpression of CD40: A novel mechanism in autoimmune disease. *J Immunol.* 2012; 189: 3043-3053.

Menconi F, Osman R, Monti MC, Greenberg DA, Concepcion ES, **Tomer Y**. Shared molecular amino acid signature in the HLA-DR peptide binding pocket predisposes to both autoimmune diabetes and thyroiditis. *Proc Natl Acad Sci USA.* 2010; 107: 16899-16903.

Rupangi Vasavada, Ph.D.

Associate Professor of Medicine (Endocrinology, Diabetes and Bone Disease)

Institute Affiliation: Diabetes, Obesity and Metabolism Institute

Lab/Location: Atran 5-02

Email: rupangi.vasavada@mssm.edu

Research Interests: Dr. Vasavada's research has focused on understanding the pathways and mechanisms that regulate pancreatic beta cell growth, survival, and function, in normal beta cell physiology and in the pathophysiological settings of diabetes and islet transplantation, specifically centered on the role of growth factors and the signaling and molecular pathways through which they mediate their effects.

Type of Research: Basic/Translational

Publications:

Alvarez-Perez JC, Rosa TC, Casinelli GP, Valle SR, Lakshmipathi J, Rosselot C, Rausell-Palamos F, **Vasavada RC**, Garcia-Ocaña A. Hepatocyte growth factor (HGF) ameliorates hyperglycemia and corrects ß-cell mass in IRS2 deficient mice. *Molecular Endocrinology.* 2014; 28:2038-2048.

Mozar A*, Guthalu NK*, Pollack I, Fenutria R, **Vasavada RC**. The role of PTHrP in pancreatic beta cells and implications for diabetes pathophysiology and treatment. *Clinical Reviews in Bone and Mineral Metabolism.* 2014; 12:165-177.

Alvarez-Perez JC, Ernst S, Demirci C, Casinelli GP, Mellado-Gil JM, Rausell-Palamos F, **Vasavada RC**, Garcia-Ocaña A. Hepatocyte Growth Factor/c-Met Signaling Is Required for ß-Cell Regeneration. *Diabetes.* 2014; 63:216-223.

Guthalu NK, Mozar A, Otero A, Chin C, Garcia-Ocaña A, **Vasavada RC**. Lactogens protect rodent and human beta cells against glucolipotoxicity-induced cell death through Jak2/Stat5 signaling. *Diabetologia.* 2012; 55:1721-1732.

Williams K, Abanquah D, Joshi-Gokhale S, Lin H, Guthalu NK, Zhang XY, Bisello A, Stewart AF, Garcia-Ocaña A, **Vasavada RC**. Systemic administration of parathyroid hormone-related peptide 1-36 stimulates endogenous beta cell growth while preserving function in adult mice. *Diabetologia.* 2011; 54:2867-2877.

Alfin G. Vicencio, M.D.

Associate Professor of Pediatrics (Pulmonology) and Chief of the Division of Pulmonology

Lab/Location: 5 East 98th Street, 10th Floor

Email: alfin.vicencio@mssm.edu

Research Interests: Dr. Vicencio is investigating sub-clinical fungal infection as a potential cause of severe, refractory asthma. Together with collaborators, he is studying deficiencies in airway immune factors that could potentially contribute to infection, and is also analyzing fluid collected from the lower airways of children for molecular evidence of infection.

Type of Research: Clinical/Translational

Publications:

Vicencio AG, Santiago MT, Tsirilakis K, Stone A, Worgall S, Foley EA, Bush D, and Goldman DL. Fungal sensitization in childhood persistent asthma is associated with disease severity. *Pediatr Pulmonol*. 2013; Feb 8. doi: 10.1002/ppul.22779. [Epub ahead of print]

Goldman DL, Li X, Tsirilakis K, Andrade C, Casadevall A and **Vicencio AG**. Increased chitinase expression and fungal specific antibodies in the bronchoalveolar lavage fluid of asthmatic children. *Clin Exp Allergy.* 2012;42(4):523-30.

Patel K, **Vicencio AG**, Salva P, Tsirilakis K, Du Z and Webley WC. Chlamydia pneumoniae is associated with elevated IL-8 and airway neutrophilia in children with refractory asthma. *Ped Infect Dis J*. 2010;December;29(12):1093-8.

Vicencio AG, Chupp GL, Tsirilakis K, Kessel A, Nandalike K, Veler H, Kipperman S, Young MC and Goldman DL. CHIT1 mutations: A genetic risk factor for severe asthma with fungal sensitization *Pediatrics* 2010;Oct;126(4):e982-5.

Vicencio AG, Narain S, Du Z, Zeng WY, Ritch J, Casadevall A and Goldman DL. Pulmonary cryptococcosis induces chitinase in the rat. *Respir Res.* 2008;May 15; 9:40.

Martin J. Walsh, Ph.D.

Associate Professor of Pediatrics (Gastroenterology) and Structural and Chemical Biology

Institute Affiliations: Tisch Cancer Institute; Mindich Child Health and Development Institute

Lab/Location: Annenberg 14-30A

Email: martin.walsh@mssm.edu

Research Interests: Dr. Walsh's area of interest is in chromatin biology of human disease and development. The focus of the laboratory is to investigate the transcriptional regulatory networks that are associated cancer and cystic fibrosis.

Type of Research: Basic/Translational

Publications:

Sancho, A., Li, S., Paul, T., Zhang, F., Aguilo, F., Vashisht, Balasubramaniyan, N., LeLeiko, N. S., Suchy, F. J., Wohlschlegel, J. A., Zhang, W., and **Walsh, M.J**., (2015) CHD6 Regulates the topological arrangement of the *CFTR* locus. *Human Molecular Genetics*. 2015; in press.

Aguilo, F., Li, S., Balasubramaniyan, N., Zhang, F., Sancho-Medina, A., Benko, S., Chen, C.-H., Zhou, F., Qian, C., Wohlschlegel, J. A., Zhang, W., Suchy, F. and **Walsh, M. J.** Regulation of enhancer-associated RNA function by PGC1α. *Molecular Cell.* 2015; in press.

Aguilo, F., De Cecilia, S., and **Walsh, M. J.** Novel insight into the long ncRNA ANRIL and polycomb in human cancers and cardiovascular disease. Eds. Leonard Lipovitch and Kevin Morris. Current Topics in Microbiology and Immunology: Long non-coding RNAs in Human Health and Disease. *Springer International.* 2015; in press.

Virginia Walther, M.S.W., L.C.S.W.

Associate Director of Social Work for Women's and Children's Health

Lab/Location: 1184 Fifth Avenue, 6th Floor
Email: virginia.walther@mountsinai.org

Research Interests: Ms. Walther is a social worker whose research interests are related to the role high risk psychosocial factors play in adherence to care in chronically ill populations of children and their families and the roles social workers can play in mitigating the impacts of illness in families. She is also interested in transition of care for adolescents with chronic illnesses who are aging into adulthood as well as utilization of pediatric palliative care programs. Other research interests are concerned with family violence and its treatment.

Type of Research: Clinical/Translational

Publications:

Millery, M., Vazquez, S. **Walther, V.**, Humphrey, N., Schlecht, J. and VanDevanter, N. Pregnancies in perinatally HIV infected young women and implications for care and service programs. *J Assoc Nurses AIDS Care.* 2012; 23:41-51.

Schreiner-Engel, P., **Walther, V**., Mindes, J., Lynch, L., Berkowitz, R. First-trimester multifetal pregnancy reduction: acute and persistent psychological reactions. *Am J Obstet Gynecol.* 1995; 172:541-547.

Mailick, M., Holden, G. **Walther, V.** Coping with childhood asthma. *Health Soc Work.* 1994; (19):103-112.

Greenfeld, D. and **Walther V**. Psychological considerations in multifetal pregnancy reduction. *Infertil Reprod Med Clin N America.* 1993; 4:535-544.

Greenfield, D. and **Walther V**. Psychological aspects of recurrent pregnancy loss. *Infertil Reprod Med Clin N America.* 1991; 2:235-247.

Julie Wang, M.D.

Associate Professor of Pediatrics (Allergy and Immunology)

Institute Affiliations: Jaffe Food Allergy Institute; Mindich Child Health and Development Institute

Lab/Location: Annenberg 17

Email: julie.wang@mssm.edu

Research Interests: Dr. Wang's research interests include Chinese herbal medicine for the treatment of food allergy, diagnostic issues in food allergy, and food allergy in the inner city.

Type of Research: Clinical/Translational

Publications:

Taylor-Black S, Mehta H, Weiderpass E, Boffetta P, Sicherer SH, **Wang J**. Prevalence of food allergy in New York City (NYC) school children. *Ann Allergy Asthma Immunol.* 2014; 112(6):554-556.e1.

Mehta H, Ramesh M, Feuille E, Groetch M, **Wang J**. Growth Comparison in Children with and without Food Allergies in 2 Different Demographic Populations. *J Pediatr.* 2014; 165(4):842-8.

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Patil SP, **Wang J**, Noone S, Yang N, Sampson HA, Li XM. Clinical safety of FAHF-2, and inhibitory effect on basophils from patients with food allergy – extended phase I study. *J Allergy Clin Immunol* 2011; 128:1259-1265.

Wang J, Patil SP, Yang N, Ko J, Lee J, Noone S, Sampson HA, Li XM. Safety, tolerability, and immunologic effects of a food allergy herbal formula (FAHF-2) in food allergic individuals: a randomized, double-blind, placebo-controlled, dose escalation phase I study. *Ann Allergy Asthma Immunol* 2010; 105:75-84.

Birte Wistinghausen, M.D.

Assistant Professor of Pediatrics and Medical Director (Hematology/Oncology)

Institute Affiliations: TIsch Cancer Center; Mindich Child Health and Development Institute; Institute of Medical Education

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Research Interests: Dr. Wistinghausen is interested in the prevention and treatment of post-transplant lymphoproliferative disease (PTLD) in children who have received a solid organ transplant and in the role EBV virus plays in causing this disease. She is also working on defining the biology of and optimal treatment for B-lymphoblastic lymphoma. Lastly, she is interested in outcomes of survivors of childhood cancer.

Type of Research: Clinical/Translational

Publications:

Shiramizu B, **Wistinghausen B,** Hayashi R. Lymphoproliferative Disorders and Malignancies related to Immunodeficiencies in Pizzo and Poplack, *Principles and Practice of Pediatric Oncology,* Seventh Edition, 2014; submitted.

Wistinghausen B, Gross TG, Bollard CM. Post-transplant Lymphoproliferative Disease. *Pediatric Hematology-Oncology*. 2013; 3-:520-531.

Jossen J, Chu J, Hotchkiss H, **Wistinghausen B**, Iyer K, Magid M, Kamath A, Roayaie S, Arnon R. Epstein-Barr virus-associated smooth muscle tumors in children following solid organ transplantation: A review. *Pediatr Transplant.* 2015; Jan 9. [Epub ahead of print]

Weintraub L, Weiner C, Miloh T, Tomaino J, Joashi U, Benchimol C, Strauchen J, **Wistinghausen B.** Identifying Predictive Factors for Post-Transplant Lymphoproliferative Disease (PTLD) in Pediatric Solid Organ Trasplant (SOT) Recipients with Epstein-Barr Virus (EBV) viremia. *J Pediatr Hematol Oncol*. 2014; May 29. [Epub ahead of print]

Weiner C, Weintraub L, **Wistinghausen B,** Arnon R, Kerkar N, Miloh T. Graft rejection in pediatric liver transplant patients with EBV viremia and PTLD. *Pediatric Transplantation.* 2012; Aug; 16(5):458-464

Mary S. Wolff, Ph.D.

Professor of Preventive Medicine/Oncological Sciences

Lab/Location: CAM D3-109

Email: mary.wolff@mssm.edu

Research Interests: Dr. Wolff's research focuses on environmental exposures, chiefly hormonally active agents in early life, and their relationships with child development, including neurobehavior, somatic growth, and pubertal timing as well as mechanisms of action.

Type of Research: Basic/Translational

Publications:

Wolff MS, Teitelbaum SL, McGovern K, Windham GC, Pinney SM, Galvez M, Calafat AM, Kushi LH, Biro FM, the Breast Cancer and Environment Research Program. Phthalate Exposure and Pubertal Development in a Longitudinal Study of U.S. Girls. *Human Reprod*. 2014; Jul;29(7):1558-66.

Mervish N, McGovern K, Teitelbaum S, Pinney S, Windham GC, Biro, Kushi LH, Silva M, Ye X, Calafat A, **Wolff M**. Dietary Predictors of Urinary Biomarkers in young girls, BCERP, 2004-7. *Environ Res*. 2014; Aug;133:12-9.

Deierlein AL, Galvez MP, Yen HP, Pinney SM, Biro FM, Kushi LH, Teitelbaum SL, **Wolff MS**. Local food environments are associated with girls' energy, sugar sweetened beverage and snack-food intakes. *Public Health Nutr.* 2014; May 12:1-7. PMC in progress

Furlong MA, Engel SM, Barr DB, **Wolff MS**. Prenatal exposure to organophosphate pesticides and reciprocal social behavior in childhood. *Environment Int*. 2014; Sep:70: 125–31.

Hong CC, Pajak A, Teitelbaum SL, Vangeepuram N, Galvez M, Pinney SM, Windham G, Kushi LH, Biro FM, **Wolff MS.** The Breast Cancer Environment Research Program; Younger pubertal age is associated with allergy and other atopic conditions in girls. *Pediatr Allergy Immunol*. 2014; Dec; 25(8);773-80. PMC in progress.

Robert O. Wright, M.D., M.P.H.

Professor of Preventive Medicine and Pediatrics

Director, Division of Environmental Health, Preventive Medicine

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Atran 3-02

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Research Interests: Dr. Wright conducts epidemiologic studies of children's environmental health, focused primarily on neurodevelopment and fetal growth. His work incorporates molecular biomarkers of effect with measures of toxic chemical exposure, social environment and nutrition.

Type of Research: Clinical/Translational

Publications:

Braun JM, Wright RJ, Just AC, Power MC, Tamayo Y Ortiz M, Schnaas L, Hu H, **Wright RO**, Tellez-Rojo MM Relationships between lead biomarkers and diurnal salivary cortisol indices in pregnant women from Mexico City: a cross-sectional study. E*nviron Health.* 2014;Jun 10;13(1):50.

Arora M, Austin C, Sarrafpour B, Hernández-Ávila M, Hu H, **Wright RO**, Tellez-Rojo MM.Determining prenatal, early childhood and cumulative long-term lead exposure using micro-spatial deciduous dentine levels. *PLoS One.* 2014; May 19;9(5):e97805. eCollection 2014.

Karwowski MP, Just AC, Bellinger DC, Jim R, Hatley EL, Ettinger AS, Hu H, **Wright RO**. Maternal iron metabolism gene variants modify umbilical cord blood lead levels by gene-environment interaction: a birth cohort study. *Environ Health.* 2014; Oct 6;13:77.

Lakshmanan A*, Chiu YHM*, Coull BA, Just AC, Maxwell SL, Schwartz J, Gryparis A, Kloog I, Wright RJ, **Wright RO**. Associations between Prenatal traffic-related air pollution exposure and birth weight: modification by sex and maternal pre-pregnancy body mass index. *Environ Res.* 2014; Mar;71(3):201-7. Epub 2013.

Bobb JF, Valeri L, Claus Henn B, Christiani DC, **Wright RO**, Mazumdar M, Godleski JJ, Coull BA. Bayesian kernel machine regression for estimating the health effects of multi-pollutant mixtures. *Biostatistics*. 2014;Dec 22.

Rosalind J. Wright, M.D., M.P.H.

Professor of Pediatrics (Pulmonology) and Preventive Medicine

Vice-Chair, Clinical & Translational Research, Department of Pediatrics

Institute Affiliation: Mindich Child Health and Development Institute

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Research Interests: Dr. Wright has a primary interest in early life (prenatal and early childhood) predictors of developmental diseases including asthma, obesity, neurobehavioral development, and lung growth and development. A particular focus has been on population-based studies considering the role of both social (e.g., psychosocial stress) and physical (e.g., air pollution, allergens) environmental factors in explaining health disparities among urban, ethnic minority populations.

Type of Research: Clinical/Translational

Publications:

Brunst KJ, Enlow MB, Kannan S, Carroll KN, Coull BA, **Wright RJ.** Effects of prenatal social stress and maternal dietary fatty acid ratio on infant temperament: does race matter? *Epidemiology.* (Sunnyvale) 2014; 4(4). Pii: 1000167

Bosquet Enlow M, King L, Schreier HM, Howard JM, Rosenfield D, Ritz T, **Wright RJ.** Maternal sensitivity and infant autonomic and endocrine stress responses. *Early Hum Dev.* 2014; 90(7):377-85.

Chiu YH, Coull BA, Sternthal MJ, Kloog I, Schwartz J, Cohen S, **Wright RJ.** Effects of prenatal community violence and ambient air pollution on childhood wheeze in an urban population. *J Allergy Clin Immunol.* 2014; 133(3):713-22.

Lakshmanan A*, Chiu YHM*, Coull BA, Just AC, Maxwell SL, Schwartz J, Gryparis A, Kloog I, **Wright RJ**, Wright RO. (*co-first authors) Associations between prenatal traffic-related air pollution exposure and birth weight: modification by sex and maternal pre-pregnancy body mass index. *Environ Res.* [Epub ahead of print]

Chiu Y-HM, Coull BA, Anderson S, Barber R. Wright RO, **Wright RJ.** Associations between traffic related black carbon exposure and attention in a prospective birth cohort of urban children. *Environ Health Perspect.* 2013; 12:1859-64.

Yong Zhao, Ph.D. M.D.
Assistant Professor of Genetics and Genomic Sciences

Institute Affiliation: Mindich Child Health and Development Institute

Lab/Location: Hess CSM 7-202

Email: yong.zhao@mssm.edu

Research Interests: Dr. Zhao is interested in how genetic and epigenetic programs regulate cardiac development. Using mouse genetics, ES cells differentiation in vitro, and other cutting edge technologies, his lab is beginning to understand the roles of transcription factors and epigenetic regulators in regulating heart development and in pathogenesis. It is anticipated that his studies will improve the treatment of human heart disease.

Type of Research: Basic/Translational

Publications:

Wei Y, Peng S, Wu M, Sachidanandam R, Tu Z, Zhang S, Falce C, Sobie EA, Lebeche D, **Zhao Y**. Multifaceted roles of miR-1s in repressing the fetal gene program in the heart. *Cell Research.* 2014 Mar;24(3):278-92.

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Wu M, Peng S, Yang J, Tu Z, Cai X, Cai CL, Wang Z, **Zhao Y**. Baf250a orchestrates an epigenetic pathway to repress the Nkx2.5-directed contractile cardiomyocyte program in the sinoatrial node. *Cell Res.* 2014; 24:1201-1213

Yang F, Zhou L, Wang Q, You X, Li Y, **Zhao Y**, Han X, Chang Z, He X, Cheng C, Wu C, Wang WJ, Hu FY, Zhao T, Li Y, Zhao M, Zheng GY, Dong J, Fan C, Yang J, Meng X, Zhang Y, Zhu X, Xiong J, Tian XL, Cao H. NEXN inhibits GATA4 and leads to atrial septal defects in mice and humans. *Cardiovasc Res.* 2014; 103:228-37.

Josowitz R, Lu J, Falce C, D'Souza SL, Wu M, Cohen N, Dubois NC, **Zhao Y**, Sobie EA, Fishman GI, Gelb BD. Identification and purification of human induced pluripotent stem cell-derived atrial-like cardiomyocytes based on sarcolipin expression. *PLoS One.* 2014; 9:e101316.

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