

# Medical Scientist Training Program

## Publications 2012-2013

- Kim JY, **Ash RT**, Ceballos-Diaz C, Levites Y, Gold TE, Smirnakis SM, Janowsky JL. (2013). Viral transduction of the neonatal brain offers controllable transgene mosaicism for a range of neuroscience applications. *European Journal of Neuroscience*. [Epub Ahead of Print] PMID: 23347239
- Baker SA**, Chen L, Wilkins AD, Yu P, Lichtarge O, Zoghbi HY (2013). An AT-Hook Domain in MeCP2 Determines the Clinical Course of Rett Syndrome and Related Disorders. *Cell* 152(5):984-96. PMID: 23452848.
- Barry MA**, Bezek S, Serpa JA, Hotez PJ, Woc-Colburn L. (2012). Neglected infections of poverty in Texas and the rest of the United States: management and treatment options. *Clin Pharmacol Ther* 92, 170-181. PMID: 22760004
- Barry MA**, Weatherhead JE, Hotez PJ, Woc-Colburn L. (2013). Childhood Parasitic Infections Endemic to the United States. *Pediatr Clin North Am* In Press.
- Ajami NJ, **Barry MA**, Carrillo B, Muhaxhiri Z, Neill FH, Prasad BV, Opekun AR, Gilger MA, Graham DY, Atmar RL, Estes MK. (2012). Antibody responses to norovirus genogroup GI.1 and GII.4 proteases in volunteers administered Norwalk virus. *Clin Vaccine Immunol* 19, 1980-1983. PMID: 23035177
- Bayat V**, Thiffault I, Jaiswal M, Tétreault M, Donti T, Sasarman F, Bernard G, Demers-Lamarche J, Dicaire MJ, Mathieu J, Vanasse M, Bouchard JP, Rioux MF, Lourenco CM, Li Z, Haueter C, Shoubridge EA, Graham BH, Brais B, Bellen HJ. (2012). Mutations in the Mitochondrial Methionyl-tRNA Synthetase in *Drosophila melanogaster* and *Homo sapiens* Cause Neurodegeneration. *PLoS Biol* 10(3):e1001288. PMID: 22448145
- Jaiswal M, Sandoval H, Zhang K, **Bayat V**, Bellen HJ. (2012). Probing mechanisms that underlie human neurodegenerative diseases in *Drosophila*. *Annu Rev Genet* 2012;46:371-96. PMID: 22974305
- Xiong B, **Bayat V**, Jaiswal M, Zhang K, Sandoval H, Charng WL, Li T, David G, Duraine L, Lin YQ, Neely GG, Yamamoto S, Bellen HJ. (2012). Crag is a GEF for Rab11 required for rhodopsin trafficking and maintenance of adult photoreceptor cells. *PLoS Biol* 2012;10(12):e1001438. PMID: 23226104
- Yamamoto S, Charng WL, Rana NA, Kakuda S, Jaiswal M, **Bayat V**, Xiong B, Zhang K, Sandoval H, David G, Wang H, Haltiwanger RS, Bellen HJ. (2012). A mutation in EGF repeat-8 of Notch discriminates between Serrate/Jagged and Delta family ligands. *Science* 338(6111):1229-32. PMID: 23197537
- Boone PM**, Soens ZT, Campbell IM, Stankiewicz P, Cheung SW, Patel A, Beaudet AL, Plon SE, Shaw CA, McGuire AL, Lupski JR. (2013). Incidental copy-number variants identified by routine genome testing in a clinical population. *Genet Med* 15, 45-54. PMID: 22878507
- Ku BJ, Kim TH, Lee JH, **Buras ED**, White LD, Stevens RD, Ilkayeva OR, Bain JR, Newgard CB, DeMayo FJ, Jeong JW. (2012). Mig-6 plays a critical role in the regulation of cholesterol homeostasis and bile acid synthesis. *PLoS One* 7(8):e42915. PMID: 22912762
- Xie A, **Buras ED**, Xia J, Chen W. (2012). The emerging role of interleukin-21 in transplantation. *Journal of Clinical and Cellular Immunology* S9: 002
- Boone PM, Soens ZT, **Campbell IM**, Stankiewicz P, Cheung SW, Patel A, Beaudet AL, Plon SE, Shaw CA, McGuire AL, Lupski JR. (2012). Incidental copy-number variants identified by routine genome testing in a clinical population. *Genet Med* 15, 45-54. PMID: 22878507
- Chotewutmontri P, Reddick LE, McWilliams DR, **Campbell IM**, Bruce BD. (2012). Differential Transit Peptide Recognition during Preprotein Binding and Translocation into Flowering Plant Plastids. *Plant Cell* 24, 3040-3059. PMID: 22829148
- Campbell IM\***, Yatsenko SA\*, Hixson P, Reimschisel T, Thomas M, Wilson W, Dayal U, Wheless JW, Crunk A, Curry C, Parkinson N, Fishman L, Riviello JJ, Nowaczyk MJ, Zeeman S, Rosenfeld JA, Bejjani BA, Shaffer LG, Cheung SW, Lupski JR, Stankiewicz P, Scaglia F. (2012). Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A. *Genet Med* 14, 868-876. PMID: 22722545. \* These authors contributed equally to the manuscript.
- DeGrande S, Nixon D, Koval O, Curran JW, Wright P, Wang Q, Kashef F, **Chiang D**, Li N, Wehrens XH, Anderson ME, Hund TJ, Mohler PJ. (2012). CaMKII inhibition rescues proarrhythmic phenotypes in the model of human ankyrin-B syndrome. *Heart Rhythm* 9(12), 2034-41. PMID: 23059182

# Medical Scientist Training Program

## Publications 2012-2013

- Dixit S, Wang T, Manzano E, Yoo E, Lee JK, **Chiang DY**, Ryan N, Yechoor V, Wehrens XHT. (2013). Effects of CaMKII-Mediated Phosphorylation of Ryanodine Receptor Type 2 on Islet Calcium Handling, Insulin Secretion, and Glucose Tolerance. *PLoS One* .
- Wehrens XHT, **Chiang DY**, Li N. (2013). Chronic Exercise: A Contributing Factor to Atrial Fibrillation? *J Am Coll Cardiol*.
- Chow KH**, Naik S, Kakarla S, Brawley VS, Shaffer DR, Yi Z, Rainusso N, Wu MF, Liu H, Kew Y, Grossman RG, Powell S, Lee D, Ahmed N, Gottschalk S. (2012). T cells redirected against EphA2 for the immunotherapy of glioblastoma. *Molecular Therapy* . [Epub Ahead of Print] PMID: 23070117
- Connell PS**, Han RI, Grande-Allen KJ. (2012). Differentiating the aging of the mitral valve from human and canine myxomatous degeneration. *J Vet Cardiol* 14(1), 31-45. PMID: 22364720
- Eleswarapu SV** and Athanasiou KA. (2013). TRPV4 channel activation improves the tensile properties of self-assembled articular cartilage constructs. *Acta Biomaterialia* 9, 5554-61. PMID: 23128162
- Garcia I**, Huang L, Ung K, Arenkiel BR. (2012). Tracing synaptic connectivity onto embryonic stem cell-derived neurons. *Stem Cells* 30, 2140-2151. PMID: 22996827
- Garcia I**, Kim C, Arenkiel BR. (2012). Genetic strategies to investigate neuronal circuit properties using stem cell-derived neurons. *Front Cell Neurosci* 6, 59. PMID: 23264761
- Garcia I**, Kim C, Arenkiel BR. (2013). Revealing neuronal circuitry using stem cell-derived neurons. *Current Protoc. Stem Cell Biology. Curr Protoc Stem Cell Biol* . [Epub Ahead of Print]
- Huang L, **Garcia I**, Jen H, Arenkiel BR. (2013). Reciprocal connectivity between mitral cells and external plexiform layer interneurons in the mouse olfactory bulb. *Front Neural Circuits* . [Epub Ahead of Print]
- Gill BJ**, Gibbons DL, Roudsari LC, Saik JE, Rizvi ZH, Roybal JD, Kurie JM, West JL. (2012). A synthetic matrix with independently tunable biochemistry and mechanical properties to study epithelial morphogenesis and EMT in a lung adenocarcinoma model. *Cancer Res* 72, 6013-6023. PMID: 22952217
- Gould DJ**, Reece GP. (2012). Skin graft vascular maturation and remodeling: a multifractal approach to morphological quantification. *Microcirculation* (7):652-63. PMID: 22672367
- Gould DJ**, Salmans JA, Lassinger BK, Contreras A, Gutierrez C, Bonafas E, Liscum KR, Silberfein EJ. (2012). Factors associated with phyllodes tumor of the breast after core needle biopsy identifies fibroepithelial neoplasm. *J Surg Res* (1):299-303. PMID: 22524977
- Kemnade JO**, Seethammagari M, Narayanan P, Levitt JM, McCormick AA, Spencer DM. (2012). Off-the-shelf adenoviral-mediated immunotherapy via bicistronic expression of tumor antigen and iMyD88/CD40 adjuvant. *Molecular Therapy* 7, 1462-71. PMID: 22434138
- Garcia I, **Kim C**, Arenkiel BR. (2012). Genetic strategies to investigate neuronal circuit properties using stem cell-derived neurons. *Frontiers in Cellular Neuroscience* 6, 59. [Epub Ahead of Print] PMID: 23264761
- Garcia I, **Kim C**, Arenkiel BR. (2013). Revealing neuronal circuitry using stem cell-derived neurons. *Current Protoc. Stem Cell Biology. Curr Protoc Stem Cell Biol* . [Epub Ahead of Print]
- Lin AY**, Lunsford J, Bear AS, Young JK, Eckels P, Luo L, Foster AE, Drezek RA. (2013). High-density sub-100-nm peptide-gold nanoparticle complexes improve vaccine presentation by dendritic cells in vitro. *Nanoscale Res Lett* 8, 72. PMID: 23402570
- Wang Y\*, **Lu JT\***, Yu J, Gibbs RA, Yu F. (2013). An integrative variant analysis pipeline for accurate genotype/haplotype inference in population NGS data. *Genome Research* . [Epub Ahead of Print] PMID: 23296920. \* These authors contributed equally to the manuscript.
- Burrage LC, **Lu JT**, Liu D, Moss T, Gibbs RA, Schlesinger AE, Bacino CA, Campeau PM, Lee BH. (2013). Early Childhood Presentation of Czech Dysplasia. *Clinical Dysmorphology* . [Epub Ahead of Print]
- Campeau PM\*, **Lu JT\***, Dawson BC, Fokkema IF, Robertson SP, Gibbs RA, Lee BH. (2012). The KAT6B-related disorders Genitopatellar syndrome and Ohdo/SBBYS syndrome have distinct clinical features reflecting distinct molecular mechanisms. *Human Mutation* 33(11). PMID: 22715153. \* These authors contributed equally to the manuscript.

# Medical Scientist Training Program

## Publications 2012-2013

- Campeau PM\*, **Lu JT\***, Sule G, Jiang MM, Bae Y, Madan S, Högl W, Shaw NJ, Mumm S, Gibbs RA, Whyte MP, Lee BH. (2012). Whole Exome Sequencing Identifies Mutations in the Nucleoside Transporter Gene SLC29A3 in Dysosteosclerosis, a Form of Osteopetrosis. *Human Molecular Genetics* 21(22):4904-9. PMID: 22875837. \* These authors contributed equally to the manuscript.
- Laine CM\*, Joeng KS\*, Campeau PM\*, Kiviranta R\*, Tarkkonen K, Grover M, **Lu JT**, Pekkinen M, Wessman M, Heino TJ, Nieminen-Pihala V, Aronen M, Laine T, Kroger H, Cole WG, Lehesjoki AE, Nevarez L, Krakow D, Curry C, Cohn D, Gibbs RA, Lee BH, Makitie O. (2013). WNT1 Mutations in Early-onset Osteoporosis and Osteogenesis Imperfecta. *New England Journal of Medicine*. [Epub Ahead of Print]. \* These authors contributed equally to the manuscript.
- Shapiro JR, Lietman C, Grover M, **Lu JT**, Nagamani SC, Dawson BC, Baldridge DM, Bainbridge MN, Cohn DH, Blazo M, Roberts TT, Brennen FS, Wu Y, Gibbs RA, Melvin P, Campeau PM, Lee BH. (2013). Phenotypic variability of osteogenesis imperfecta type V caused by an IFITM5 mutation. *Journal of Bone Mineral Research*. [Epub Ahead of Print] PMID: 23408678
- The 1000 Genomes Project Consortium. (2012). An integrated map of genetic variation from 1092 human genome. *Nature* 491(7422):56-65. PMID: 23128226 (**Lu, JT**)
- Samaco RC, Mandel-Brehm C, **McGraw CM**, Shaw CA, McGill BE, Zoghbi HY. (2012). Crh and Oprm1 mediate anxiety-related behavior and social approach in a mouse model of MECP2 duplication syndrome. *Nat Genet* 44, 206-211. PMID: 22231481
- Samaco RC, **McGraw CM**, Ward CS, Sun Y, Neul JL, Zoghbi HY. (2012). Female Mecp2+/- mice display robust behavioral deficits on two different genetic backgrounds providing a framework for pre-clinical studies. *Human Molecular Genetics* 22, 96-109. PMID: 23026749
- Heo J, Reid T, Ruo L, Breitbach CJ, Rose S, Bloomston M, Cho M, Lim HY, Chung HC, Kim CW, Burke J, Lencioni R, Hickman T, Moon A, Lee YS, Kim MK, Daneshmand M, Dubois K, Longpre L, **Ngo M**, Rooney C, Bell JC, Rhee BG, Patt R, Hwang TH, Kirn DH. (2013). Randomized dose-finding clinical trial of oncolytic immunotherapeutic vaccinia JX-594 in liver cancer. *Nature Medicine*. [Epub Ahead of Print] PMID: 23396206
- Lapteva N, Durett AG, Sun J, Rollins LA, Huye LL, Fang J, Dandekar V, Mei Z, Jackson K, Vera J, Ando J, **Ngo M**, Coustan-Smith E, Campana D, Szmania S, Garg T, Moreno-Bost A, Vanrhee F, Gee AP, Rooney CM. (2012). Large-scale ex vivo expansion and characterization of natural killer cells for clinical applications. *Cytotherapy* 14(9):1131-43. PMID: 22900959
- Olivetti PR**, Noebels JL. (2012). Interneuron, interrupted: molecular pathogenesis of ARX mutations and X-linked infantile spasms. *Current Opinion in Neurobiology* 22(5):859-65. [Epub Ahead of Print] PMID: 22565167
- Musselman LP, Fink JL, **Ramachandran PV**, Patterson BW, Okunade AL, Maier E, Brent MR, Turk J, Thomas J Baranski. (2013). Role of fat body lipogenesis in protection against the effects of caloric overload in Drosophila. *Journal of Biological Chemistry*. [Epub Ahead of Print] PMID: 23355467
- Pendse J, **Ramachandran PV**, Na J, Narisu N, Fink JL, Cagan RS, Francis S Collins, Thomas J Baranski. (2013). A Drosophila functional evaluation of candidates from human genome-wide association studies of type 2 diabetes identifies tissue-specific roles for dHHEX. *BMC Genomics*. [Epub Ahead of Print] PMID: 87208711
- Bammes BE\*, **Rochat RH\***, Jakana J, Chen DH, Chiu W. (2012). Direct electron detection yields cryo-EM reconstructions at resolutions beyond 3/4 Nyquist frequency. *Journal of Structural Biology*. [Epub Ahead of Print] PMID: 22285189. \* These authors contributed equally to the manuscript.
- Chang J\*, Liu X\*, **Rochat RH\***, Baker ML\*, Chiu W. (2012). Reconstructing virus structures from nanometer to near-atomic resolutions with cryo-electron microscopy and tomography. *Adv Exp Med Biol* 726, 49-90. PMID: 22297510. \* These authors contributed equally to the manuscript.
- Schmid, M. F., Hecksel, C. W., **Rochat, RH**, Bhella, D., Chiu, W., Rixon, F. J. (2012). A tail-like assembly at the portal vertex in intact herpes simplex type-1 virions. *PLoS Pathogens*, 8(10), e1002961. PMID: 23055933
- Mazyar H, Van den Berg R, **Seilheimer R**, Ma WJ (2013) Independence is elusive: set size effects on encoding precision in visual search *Journal of Vision*, in press.

# Medical Scientist Training Program

## Publications 2012-2013

- Farmakiotis D, Chien KS, **Tong Shum TC**, Rodriguez-Barradas M, Musher DM. (2013). Photo quiz: to scan or not to scan? *Clinical infectious diseases : an official publication of the Infectious Diseases Society of America* 56(7):1052-3. PMID: 23463795.
- Spicer PP**, Kretlow JD, Young S, Jansen JA, Kasper FK, Mikos AG. (2012). Evaluation of bone regeneration using the rat critical size calvarial defect. *Nat Protoc* 7, 1918-29. PMID: 23018195
- Appel AA, Larson JC, Somo S, Zhong Z, **Spicer PP**, Kasper FK, Garson AB 3rd, Zysk AM, Mikos AG, Anastasio MA, Brey EM. (2012). Imaging of poly( $\alpha$ -hydroxy-ester) scaffolds with X-ray phase-contrast microcomputed tomography. *Tissue Eng Part C Methods* 18, 859-865. PMID: 22607529
- Wang L, Yoon DM, **Spicer PP**, Henslee AM, Scott DW, Wong ME, Kasper FK, Mikos AG. (2012). Characterization of porous polymethylmethacrylate space maintainers for craniofacial reconstruction. *J Biomed Mater Res B Appl Biomater*. [Epub Ahead of Print] PMID: 23359449
- Wat MJ**, Beck TF, Hernández-García A, Yu Z, Veenma D, Garcia M, Holder AM, Wat JJ, Chen Y, Mohila CA, Lally KP, Dickinson M, Tibboel D, de Klein A, Lee B, Scott DA. (2012). Mouse model reveals the role of SOX7 in the development of congenital diaphragmatic hernia associated with recurrent deletions of 8p23.1. *Human Molecular Genetics* 21, 4115-25. PMID: 22723016
- Fong EL\*, **Watson BM\***, Kasper FK, Mikos AG. (2012). Building bridges: leveraging interdisciplinary collaborations in the development of biomaterials to meet clinical needs. *Adv Mater* 24(36):4995-5013. PMID: 22821772. \* These authors contributed equally to the manuscript.
- Appukuttan B, McFarland TJ, Stempel A, Kassem JB, Hartzell M, Zhang Y, Bond D, West K, **Wilson R**, Stout A, Pan Y, Ilias H, Robertson K, Klein ML, Wilson D, Smith JR, Stout JT. (2012). The related transcriptional enhancer factor-1 isoform, TEAD4(216), can repress vascular endothelial growth factor expression in mammalian cells. *PLoS One* 7, e31260. [Epub Ahead of Print] PMID: 22761647
- Rossi L, Lin KK, Boles NC, **Yang L**, King KY, Jeong M, Mayle A, Goodell MA. (2012). Less is more: unveiling the functional core of hematopoietic stem cells through knockout mice. *Cell Stem Cell* 11(3):302-17. PMID: 22958929
- Ziats MN** and Rennert OM. (2013). Aberrant Expression of Long Noncoding RNAs in Autistic Brain. *J Mol Neurosci* 3, 589-593. PMID: 22949041
- Kriel A, Bittner AN, Kim SH, Liu K, Tehranchi AK, **Zou WY**, Rendon S, Chen R, Tu BP, Wang JD. (2012). Direct Regulation of GTP Homeostasis by (p)ppGpp: A Critical Component of Viability and Stress Resistance. *Molecular Cell* 48, 231-41. PMID: 22981860