

EMPLOYMENT AND PROFESSIONAL EXPERIENCE

03/23/15 – present **Assistant Professor/Research**, Department of Human Genetics & South Texas Diabetes and Obesity Institute (STDOI), University of Texas Rio Grande Valley (UTRGV) School of Medicine.

At STDOI, my focus of research is to develop iPSC-based methodologies and disease models for assessing genes influencing human common complex diseases particularly, where assessment of genetic influence is difficult due to availability of relevant tissue types, environmental factors and medical interventions.

At STDOI we have a rich resource of lymphoblastoid cell lines (LCLs) established using the peripheral blood mononuclear cells (PBMCs) collected from more than 1400 Mexican American participants of our San Antonio Family Heart Study (SAFHS). Whole genome sequence data and extensive phenotype data for common complex human diseases are available for most of these SAFHS participants. Our large, well characterized LCL resource provides a unique opportunity to generate iPSCs from any of these individuals. I have optimized an efficient LCL to iPSC reprogramming method, achieving 100% reprogramming success and high reprogramming efficiency (~50-200 colonies/million nucleofected cells). Using this optimized methodology, I have generated > 190 iPSC and >185 NSC lines, which are being used in various ongoing disease modeling and gene identification studies.

I used iPSC generated NSCs to model the effect of a rare genetic variant in the γ -adducine (ADD3) gene on the integrity of adherens junctions in neural stem cells. This non-synonymous variant was identified in our SAFHS cohort and shows highly significant association with mental depression. The 3D neurosphere generated from iPSC derived NSCs of 11 cases carrying the rare variant and 12 normal controls were analyzed for various adherens junction related phenotypes.

In another ongoing project “Novel Approaches to Understanding the Role of Genetics in Parkinson’s disease (PD)” we used state-of-the-art iPSC technology to make neural stem cells (NSCs) from existing PD specific blood cell lines. Using genome wide RNA sequencing of the neural stem cells, I discovered an expression phenotype, which suggests a strong developmental component to sporadic PD. The discovered expression phenotype influences stem cell proliferation and neural crest cell development. The role of this phenotype in the maintenance of Isthmus and in the development of the dopaminergic system is being studied in iPSC generated ventral neuroepithelium, brain organoids, and dopaminergic neurons.

I have developed an iPSC based hepatic culture system consisting of mature hepatocytes and biliary/cholangiocytic structures, which better recapitulate the developmental and functional characteristics of the liver. This culture system is being used to model fat induced hepatic steatosis in our laboratory.

I have also used the reprogrammed iPSC lines to generate functional cardiomyocytes, pancreatic β cells, adipocytes and cerebral organoids to model the cardiomyopathies, metabolic and brain related disorders.

Apart from my current research in developing iPSC methodology in our laboratory, I have also had a major role in several large-scale human genetics projects at our institute. I have been largely involved in the molecular aspect of identifying genes influencing human cardiovascular health and other age-related metabolic diseases.

02/01/09 – 03/22/15 Staff Scientist I, Texas Biomedical Research Institute, San Antonio, TX, USA

While working at Texas Biomed, I was largely involved in the molecular aspect of identifying genes influencing human cardiovascular health and other age-related metabolic diseases. The brief outline of each of the major research projects I worked on is as follows:

Genetics of Atherosclerosis in Mexican Americans: Atherosclerosis is one of the leading causes of morbidity and mortality worldwide. Recently, oxidative stress has been shown to be a significant contributor to atherosclerosis. Plasma total antioxidant status (TAS) is a novel summary measure of an individual's ability to accommodate oxidative stress. While there is strong evidence that genetic factors play a major role in determination of plasma TAS levels, specific genes are yet to be identified. I was responsible for the molecular methods to identify potential functional regulatory variants in novel candidate genes influencing TAS levels, in Mexican American individuals from the San Antonio Family Heart Study (SAFHS).

Telomere Length Variation, its Genetic Regulation and Maintenance Mechanisms: Telomeres are terminal chromosomal structures. Given their age and disease related reduction, in association with p53 activation, they have emerged as prime instigators of a functional decline of tissue stem cells and also adversely affect renewal and bio-energetic support in diverse tissues, influencing ageing and age-related diseases. I performed telomere length assays and analysis in several of the large family-based studies at Texas Biomed.

Nuclear Gene Response to Depletion and Repopulation of Mitochondrial DNA in Human cells: A new era in mitochondrial research has emerged that concerns the role of mitochondria in intracellular signaling - a process that is likely to have far-reaching implications in development, aging, disease, and environmental adaptation. I employed a novel approach of generating various functional states of mitochondria by depleting and repopulating mitochondrial DNA experimentally in human cell lines. The genome wide transcriptome data generated from these cells is being analyzed to evaluate transcriptomic and functional changes that take place at the cellular level consequent to the change in mitochondria functional state.

Novel Approaches to Understanding the Role of Genetics in Parkinson's disease (PD): To identify novel neuronal biomarkers relevant for PD risk and the neuronal functional pathways that are most important in PD, we used induced pluripotent stem cell (iPSC) technology to make dopaminergic neurons from existing PD specific blood cell lines. I was responsible for generating iPSCs, their differentiation into dopaminergic neurons and performing functional assays relevant to understanding the pathophysiology of PD.

I have also had a major role in several large-scale human genetics projects at Texas Biomed. I performed whole genome genotyping on more than 15,000 human samples and whole genome gene expression on more than 3,000 samples from various studies. I was also responsible for critical aspects of quality control in our next generation sequencing efforts and served as the main molecular trouble-shooter for keeping our innovative research programs moving forward.

12/05/01 – 12/26/08 Assistant Anthropologist (Physical), Group 'B' (Gazetted) at
Anthropological Survey of India, Govt. of India, Southern Regional
Centre, Mysore

While working in Anthropological Survey of India I developed a Molecular Anthropology Facility at the Southern Regional Centre, Mysore and worked on the following research projects.

Principal Investigator: “*Genetic Structure, Health Profile and phylogeny of Tribal Groups of Southern, Karnataka*”.

Collected blood samples and epidemiological data among three scheduled tribes inhabiting southern districts of Karnataka, India. Analyzed collected blood samples and data to evaluate genetic structure, phylogenetic interrelationships and health profile of the studied tribes.

Co-Principal Investigator: “*DNA polymorphism in contemporary Indian populations and ancient Human skeleton remains: Human Evolution and Peopling of India*”.

In this collective effort of Anthropological Survey of India, my colleagues and I sequenced 1000 complete and ~2000 partial mitochondrial genomes collected from over 32 relic tribal populations of India. The research evaluating the early dispersal of modern humans from their African origins and phylogeny of southern Asian populations has been published in peer reviewed Journals in the form of five research papers.

Co-Investigator: “Role of *FLT3* and *NPM1* mutations in acute myeloid leukemia”. A collaborative project with Osmania University, Hyderabad, India.

Co-Investigator: “A validation study of type 2 diabetes-related variants of the *TCF7L2*, *HHEX*, *KCNJ11*, and *ADIPOQ* genes in an endogamous ethnic group of India”. A collaborative project with Department of Anthropology, University of Delhi, Delhi, India.

Research Coordinator: Ph.D. assistance program of Anthropological Survey of India. I coordinated molecular genetics analysis of the sample for following Ph.D. research programs.

- (a) Angiotensinogen Gene - M268T variant in Indian Populations and predisposition to essential hypertension, obesity and hyperlipidemia.
- (b) Mitochondrial genome qualitative and quantitative variations and Type 2 Diabetes.”
- (c) *ADIPOQ* and *TCF7L2* Gene Polymorphisms among southern Indians and their implications to Type 2 Diabetes.

12/23/99 – 12/01/01 Scientific Assistant (Biology) at Forensic Science Laboratory, Govt. of
NCT of Delhi, New Delhi.

Performed documentation and serological/biological/DNA finger printing analysis of crime exhibits of various medico-legal cases.

Actively contributed towards the establishment of a DNA finger printing (DNA profiling) facility in the Laboratory.

PUBLICATIONS

Original publications in peer-reviewed Journals

Kumar S, Curran JE, Espinosa EC, Glahn DC, Blangero J. Highly efficient induced pluripotent stem cell reprogramming of cryopreserved Lymphoblastoid cell lines. *J. Biol. Methods*, Submitted

Kumar S, Espinosa EC, Leandro AC, Curran JE, Blangero J. MicroRNA and mRNA interactions in induced pluripotent stem cell reprogramming of lymphoblastoid cell lines. *American Journal of Stem Cells. Am J Stem Cells*. 2019; 8(2):28-37.

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Blackburn NB, Michael LF, Meikle PJ, Peralta JM, Mosior M, McAhren S, Bui HH, Bellinger MA, Giles C, **Kumar S**, Leandro AC, Almeida M, Weir JM, Mahaney MC, Dyer TD, Almasy L, VandeBerg JL, Williams-Blangero S, Glahn DC, Duggirala R, Kowala M, Blangero J, Curran JE. Rare DEGS1 variant significantly alters de novo ceramide synthesis pathway. *J Lipid Res*. 2019; 60(9): 1630-1639. doi: 10.1194/jlr.P094433. PubMed PMID: 31227640.

Johnson MP, Keyho R, Blackburn NB, Laston S, **Kumar S**, Peralta J, Thapa SS, Towne B, Subedi J, Blangero J, Williams-Blangero S. Glycated serum protein genetics and pleiotropy with cardiometabolic risk factors. *J Diabetes Res*. 2019; 2310235. doi: 10.1155/2019/2310235. eCollection 2019

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Kumar S. Book Review [Review of the book Human Growth: Assessment and Interpretation by AF Roche and SS Sun (formerly Guo). Cambridge University Press, Cambridge (2005)]. J Hum Ecol. 2005; 17 (1): 67-69.

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CONFERENCE PRESENTATIONS

Kumar S, Curran J, Peralta JM, Leandro AC, Lehman DM, Glahn DC, Blangero J. (2019) Identifying the role of genetics and neurodevelopment in sporadic late onset Parkinson's disease. (Podium presentation; Program #256; session #72: Integrated Genomics and Transcriptomics in Parkinson's Disease. Accepted for presentation at the 2019 Annual Meeting of The American Society of Human Genetics, October 18, 2019, Houston, TX. 77010.

Kumar S, Blackburn, NB, Leandro AC, Leandro M, Peralta JM, Blangero J, Curran JE. (2019) Human iPSC-derived hepatocytes reveal the functional consequences of an Hispanic rare sequence variant in the DEGS1 gene; (Flash Talk and Poster Presentation # FSO-FT3 & FSO-30). Presented at the UTRGV School of Medicine 3rd Annual Research Symposium 2019 – Health Disparities: Community Engagement, September 14, 2019, McAllen Convention Center, McAllen, Texas.

Kumar S, Curran JE, Espinosa EC, Lehman DM, Duggirala R, Glahn DC, Blangero J. (2018) Utility of iPSC generated NSCs in modeling complex neurological disorders: from candidate gene prioritization to gene validation; (Podium Presentation; Plenary Session #3 - Translational Science). Presented at the UTRGV School of Medicine Research Symposium – Health Disparities 2018: Closing the Gap, September 15, 2018, McAllen Convention Center, McAllen, Texas.

Kumar S, Curran J, Lehman DM, Duggirala R, Glahn D, Blangero J. (2017) Parkinson's disease gene identification using differential gene expression analysis of iPSC generated neural stem cells; (Podium presentation; Program #145; session #34: Genetic architecture of neurological traits). Presented at the 2017 Annual Meeting of The American Society of Human Genetics, October 19, 2017, Orlando, FL. 2017.

Kumar S, Curran J, Blangero J. (2016, February) Role of microRNA in LCL to iPSC reprogramming. Presented at the HUGO's Human Genome Meeting 2016 (Podium presentation; HGM16-ABS-1084), Houston Texas.

Kumar S, Curran J, Blangero J. (2014, October) microRNA profiling of human lymphoblastoid, iPSC and neural stem cell lines shows overlapping but distinct expression patterns. Presented at The American Society of Human Genetics Annual Meeting (ASHG14-537T), San Diego, CA.

Kumar S, Bellis C, Johnson MP, Goring HHH, Dyer TD, Blangero J, Curran JE. (2012, November) Experimental depletion and regeneration of human mitochondrial DNA to investigate its role in nuclear gene regulation. Presented at The American Society of Human Genetics Annual Meeting (ASHG12-533F), San Francisco, CA.

TEACHING

2019

Courses taught

MS1 course: Molecules to Medicine -	Bioinformatics-Part I Bioinformatics-Part II
MS1 course: Molecules to Medicine -	Stem cell Basics Stem cell applications
MS1 course: Molecules to Medicine -	Cell Signaling Basic Signal Transduction Pathways-Short Term Cellular Response Signal Transduction Pathways-Longer Term Cellular Response

Research mentoring and staff and student trainings

Mentored one MS1 scholarly (MEDI-8127) research project in 2019: titled “Develop an iPSC derived hepatic cell culture based *in-vitro* model to study nonalcoholic fatty liver disease (NAFLD).” Student Name: Daniel Nwosuocha, MS1

Mentored/Trained Ms. Erica De Leon, Research Associate I in the Department of Human genetics and STDOI, in LCL culture, iPSC reprogramming, differentiation of iPSCs into disease target cells and in her research on “Transcriptomic and functional profiles of iPSC generated cardiomyocytes”.

Trained Ms. Laura Valdez, Graduate Student in the Molecular Science Laboratory, Department of Biomedical Sciences, UTRGV SOM in iPSC culture and cryopreservation

2018

Courses taught

MS1 course: Molecules to Medicine -	Bioinformatics-Part I Bioinformatics-Part II
MS1 course: Molecules to Medicine -	Stem cell Basics and applications

Research mentoring and staff and student trainings

Mentored/Trained Ms. Erika Espinosa, Research Associate I, Research Associate I in the Department of Human genetics and STDOI, in cardiomyocyte and hepatic cell differentiation from iPSCs, DNA and RNA extraction, genome wide ATAC sequencing and RNA sequencing technologies and mentioned her research on “Generation of functional cardiomyocytes from cryopreserved LCLs using iPSC technology”.

2017

Courses taught

PROFESSIONAL AFFILIATIONS/MEMBERSHIPS

The American Society of Human Genetics (ASHG).

The Human Genome Organization (HUGO).

American Heart Association/American Stroke Association member.

The Indian Society for Human Ecology, Department of Anthropology, University of Delhi, Delhi, India.