



Icahn School
of Medicine at
Mount
Sinai

*The Mindich
Child Health and
Development Institute*

MCHDI Developmental Outcomes

Fall 2020

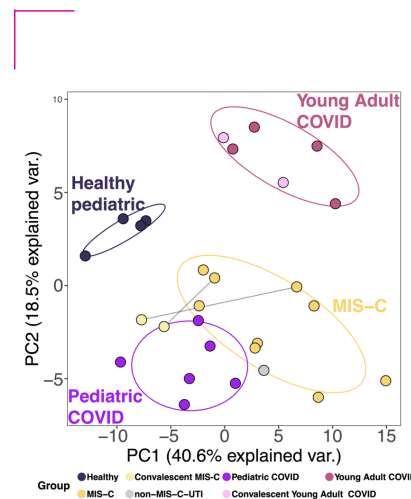
Research Advancements: COVID-19

MCHDI Faculty Lead Systems-Level Investigation of the Multisystem Inflammatory Syndrome in Children (MIS-C)

When the COVID-19 pandemic first emerged, the demographics of the hospitalized suggested that children were spared from severe disease. However a month into regions with ongoing epidemics, clinicians began to admit children presenting with a novel pediatric condition, similar to Kawasaki disease, characterized by multiorgan dysfunction and severe inflammation.

A multi-departmental collaborative effort led by Dusan Bogunovic, PhD, a MCHDI faculty member and the Director of the Center of Inborn errors of Immunity, took a systems-level approach to map the clinical and immune features of the pediatric syndrome. The team discovered an immune signature unique to MIS-C, defined by elevations in inflammatory molecules and aberrant activation of immune cells in patient's blood. Furthermore, the team also detected auto-antibodies targeting the organ systems central to the pathology of MIS-C.

"We anticipate MIS-C recurrence will be rare, as is the case with Kawasaki disease. However, the presence of autoantibodies, which can attack the host is of concern. We postulate that these autoantibodies may unleash an immune cell-driven attack against host tissues. If true, this may pose a risk for children when a vaccine becomes available," says Dr. Bogunovic. "And although further studies into the causes of MIS-C are needed and we are actively pursuing them, good news is that MIS-C is now successfully treated."



Title: A unique secreted inflammatory cytokine signature distinguishes MIS-C

Legend: Cytokine profiling of plasma from MIS-C patients (N = 9), pediatric COVID-19 patients (N = 6), active young adult COVID-19 patients (N = 4), convalescent young adult COVID-19 (N = 2), age matched healthy pediatric controls (N = 4), and convalescent (recovered) MIS-C patients (N = 2). Principal component analysis of subjects at the first time point sampled. Points are colored by sample group classification. Ellipses reflect a 68% confidence interval around the colored group centroid.

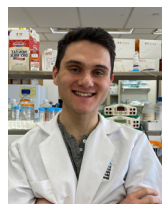
In addition to more finely understanding the causes of MIS-C, Dr. Bogunovic's lab is also developing diagnostics and screening measures for MIS-C. "Our analysis lays the foundation for future investigations into MIS-C. In our lab, our next focus will be learning how to recognize this syndrome and identify ways to prevent it. This work will in turn pave the way for a much needed prevention algorithm," says Roosheel Patel, a PhD candidate who jointly led this study with Conor Gruber, an MD/PhD candidate, both of whom are students in Bogunovic Lab.

Ultimately, this work represents the first in-depth immune profiling of MIS-C, and now published in *Cell* on September 14th, 2020, will act as a resource for clinicians studying the rare pediatric condition.



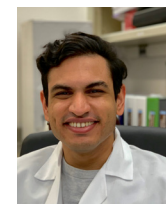
Dusan Bogunovic, PhD

Dusan Bogunovic, PhD, Associate Professor
Director, Center for Inborn Errors of Immunity
Precision Immunology Institute, Mindich Child
Health and Development Institute
Department of Microbiology, Pediatrics, and
Oncological Sciences



Conor Gruber

MD/PhD candidate



Roosheel Patel

PhD candidate

Genetic Impairments in Heart Cells Alter Infectivity of SARS-CoV-2

There is a longstanding association between viral infections and development of both acute (myocarditis) and chronic (cardiomyopathy) heart dysfunction. Pre-existing heart disease increases risk of infection with SARS-CoV-2 virus and leads to more severe outcomes in COVID-19. Several recent studies, including from Mount Sinai, have identified myocardial injury in around 30% of COVID-19 patients but thus far, no mechanisms explaining this heightened susceptibility to cardiac involvement have been reported. MCHDI and Cardiovascular Institute researcher Amy Kontorovich, MD, PhD and SUNY Upstate collaborator Iwona Koenig, PhD are studying genetic factors of the human heart that modulate SARS-CoV-2 infection.

SARS-CoV-2 at a multiplicity of infection of 0.1 and viral replication was measured at 12, 24, 48 and 72 hours by TCID50 assay.

SARS-CoV-2 replicated robustly in human heart cells and did not cause discernable cell death until after 48 hours. Compared to other cardiotropic viruses that Drs. Kontorovich and Koenig study, this represents a slower and more sustained infection that is less toxic, allowing the cells to survive longer to manufacture more infectious viral particles. Genetic alterations in structural cardiac genes were associated with less robust viral replication, with infectivity being lower in DMD-mutated and lowest in DSC2-mutated cells. This pattern matched observed differences in

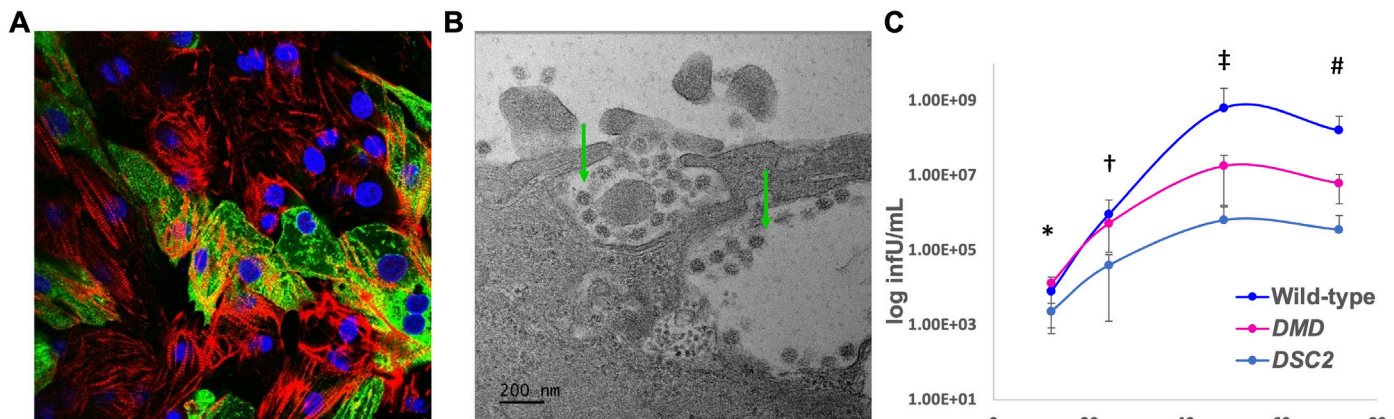


Figure 1. A) SARS-CoV-2 is cardiotropic, as evidenced by viral uptake (green, Nucleocapsid protein) in representative wild-type (WT) hiPSC-CM (red, troponin T) cells (blue, nuclei) at 48 hours post infection (h.p.i); scale bar = 25 μ m. B) SARS-CoV-2 viral particles (green arrows) can be seen disseminating from WT hiPSC-CM on transmission electron microscopy. C) SARS-CoV-2 replication (measured as infectious units per mL, infU/mL) within hiPSC-CM is blunted in the context of loss-of-function mutations in key structural cardiomyocyte genes. * *DMD vs. DSC2* $p=0.01$; † *WT vs. DSC2* $p<0.01$, *DMD vs. DSC2* $p=0.02$; ‡ *WT vs. DSC2* $p=0.02$, *DMD vs. DSC2* $p=0.03$; # *WT vs. DSC2* $p<0.01$, *DMD vs. DSC2* $p=0.02$.

The *in vitro* platform used for this research is based on human induced pluripotent stem cells (hiPSC). Using gene editing through clustered regularly interspaced short palindromic repeats (CRISPR) technology, hiPSC are selectively mutated and then differentiated to heart cells or cardiomyocytes (hiPSC-CM). Dr. Kontorovich's team previously showed that healthy and mutated hiPSC-CM can be infected with cardiotropic viruses to model myocarditis. Now, they are interested in understanding how impairments in genes that regulate heart function affect the virulence of SARS-CoV-2 after cardiac infection.

In these experiments, hiPSC from a healthy control were CRISPR-edited to induce loss-of-function mutations in genes related to common forms of genetic cardiomyopathy, dilated (*DMD* gene) and arrhythmogenic (*DSC2* gene). These CRISPR-edited cells are genomically identical to the original healthy cells except for the single induced mutation, allowing the team to hone in on specific effects from the genes of interest. Healthy control (wild-type), *DMD*-mutated and *DSC2*-mutated hiPSC-CM were infected with

expression of the SARS-CoV-2 receptor, angiotensin converting enzyme 2 (ACE2); the wild-type hiPSC-CM had the highest ACE2 expression level, followed by the DMD and then DSC2 mutants. Genetically-mediated structural impairments in hiPSC-CM may directly impact expression of ACE2 thereby reducing the viral "dose" that the cells are exposed to, and may also hinder the host's cellular machinery needed for replication and release of new viral particles. A slower and prolonged infection may stall the systemic immune response, promoting viral persistence and ultimately increased inflammation and tissue injury. Drs. Kontorovich and Koenig are now investigating the innate immune properties of healthy and genetically-altered hiPSC-CM that impact uptake and replication of SARS-CoV-2. A better understanding of host genetic susceptibilities in COVID-19 may one day lead to new strategies for protecting the heart from infection.

Amy Kontorovich, MD, PhD
Assistant Professor, Medicine



Trainee Pilot Projects: 2020 Awardees

Project Title: The Role of Nrf2 in Expanding Neonatal Pancreatic-β cell Mass

Investigator: Sharon Baumel-Alterzon, Postdoctoral fellow, Diabetes, Obesity and Metabolism Institute

Primary Mentor: Donald K. Scott, PhD, Professor of Medicine at the Diabetes, Obesity and Metabolism Institute, Mindich Child Health and Development Institute

Secondary Mentor: Adolfo Garcia-Ocaña, PhD, Professor of Medicine, Diabetes, Obesity and Metabolism Institute, Mindich Child Health and Development Institute

Abstract: Type 1 and type 2 diabetic patients suffer from insufficient functional β-cell mass. Moreover, the sharp increase in the annual incidence of both type 1 (1.8%) and type 2 (4.8%) diabetes in the pediatric population in the last few decades, highlights the urgent need for discovering the mechanisms that regulate functional β-cell mass. β-cell proliferation, which reaches its maximal peak during postnatal stages, is the major contributor for increasing β-cell mass in early postnatal development. Nrf2 is a transcription factor that plays an important role against oxidative stress. Several mutations in Nrf2 are associated with diabetes. We recently discovered that

Nrf2 is necessary for β-cell proliferation, expansion and protection against oxidative stress in adult mice in situations of over-nutrition. Importantly, Nrf2 gain-of-function is sufficient to stimulate β-cell expansion in adult mice fed a normal diet and to increase human β-cell proliferation. Taken together, these results suggest that loss of Nrf2 function may play a role in the development of diabetes. Importantly, Nrf2 levels are increased in β-cells at early postnatal days, concomitant with accelerated β-cell proliferation. In this proposal, we aim to uncover the role of Nrf2 in β-cell proliferation, survival and mass expansion in early postnatal ages, which may contribute to the maintenance of normal β-cell mass later in life. Outcomes from these studies will provide better understanding of the mechanisms leading to inadequate β-cell mass expansion in young children that may enhance their risk for developing diabetes. Our studies may also identify potential new therapeutic targets for the treatment of the disease.

Sharon Baumel-Alterzon, PhD

Postdoctoral Fellow, Diabetes, Obesity and Metabolism Institute



Project Title: The Distinct Role of Allergen-Specific T Follicular Helper Cells in IgE-Mediated and Non-IgE-Mediated Cow's Milk Allergy

Investigator: Daniel Lozano-Ojalvo, PhD, Postdoctoral fellow, Precision Immunology Institute

Primary Mentor: M. Cecilia Berin, PhD, Professor, Department of Pediatrics

Secondary Mentors: Maria Lafaille, PhD, Associate Professor, Department of Pediatrics and David Dunkin, MD, Assistant Professor, Department of Pediatrics

Abstract: The prevalence of allergic diseases triggered by milk has significantly increased in children in the last decades. Cow's milk allergy is an immediate hypersensitivity reaction orchestrated by specific IgE. On the other hand, cow's milk is also the most common trigger of the eosinophilic esophagitis (EoE), a non-IgE-mediated food allergy that has been strongly related to the generation of specific IgG4. T follicular helper (Tfh) cells are essential in both milk-driven allergic diseases based on the necessary cross-talk existing between milk-specific Tfh and B cells for the generation of a humoral response. Since IgE-mediated milk allergy and EoE (IgG4-related) show different clinical manifestations but triggered by the same allergens of cow's

milk, we hypothesized that milk-specific Tfh2 cells are critical in the development and maintenance of both diseases, and a distinct functional profile of these cells determines B cell class-switching. We have previously identified and characterized milk-specific Th2 effector cells in peripheral blood of milk-allergic and EoE patients based on the expression of activation markers. In addition, we have been reported a population of allergen-specific Tfh cell in peripheral blood from peanut allergic patients, which showed a Tfh2 phenotype (CRTH2+CXCR5+) and function (IL4+IL13+). The main goal of this project is to identify and determine the functional characteristics of allergen-specific Tfh2 cells in IgE-mediated milk allergy and EoE. Here, we propose to use a similar approach to identify and characterize milk-specific Tfh2 cells in a selected cohort of IgE-mediated allergic patients and EoE-diagnosed individuals with high levels of milk-specific IgG4. In this pilot proposal, we will use an already optimized panel for Cytex™ AURORA, which includes 29 parameters combining surface activation markers (identification of allergen-specific T cells) and functional profiling markers (cytokine and chemokine expression). The phenotypical profile of milk-specific Tfh2 cells will be then correlated circulating levels of milk-specific IgE and IgG4 in both allergic diseases. This project will benefit children's health in two ways. First, the results of this pilot grant will determine if milk-specific Tfh2 are involved in the pathogenesis of EoE, which we anticipate will reveal novel pathways that could be targeted with biologics. Secondly, this pilot grant is the first step in our ultimate goal of study of oral tolerance induction to food antigens in pediatric patients.



Daniel Lozano-Ojalvo, PhD

Postdoctoral Fellow, Precision Immunology Institute

Trainee Leadership Committee: Introducing the New Trainee Incubator Series

The MCHDI trainee leadership committee (TLC) opened its doors in the Fall of 2016 to help brainstorm ways to provide more resources and support for trainees. The MCHDI TLC takes honor in representing trainees from multiple research areas. The TLC has been leading workshops and hosting social events to facilitate the interaction between trainees and faculty members, especially during these challenging times.

This year the TLC welcomes as new members David Gonzalez, MD/PhD student and Dr. Silvia De Rubeis, Assistant Professor in the Department of Psychiatry who will provide additional guidance to the TLC as a faculty mentor. Together with Dr. De Rubeis, we are excited to announce a new MCHDI Incubator Series entirely dedicated to trainees. This is modeled after the successful MCHDI Faculty Incubator Series, where faculty discuss new ideas for projects and grant applications. The series will give the trainees the opportunity to provide and receive peer advice on new project ideas, upcoming job talks or presentations at major conferences, and fellowship applications. With this new series, we aim at helping trainees to best define their question(s), refine the scope of their project, and craft

successful applications. This series will be particularly useful for late stage post-docs who are preparing for upcoming faculty interviews, NIH grant applications, or PhD students preparing applications for fellowships. The TLC also organizes and promotes trainee social events. This year, we hosted one online happy hour full of interactive games where two winners received prizes. The TLC helps in organizing workshops and seminar for the Child Health Seminar series, hosted by the Pediatrics Department and co-sponsored by the MCHDI and co-chaired by Drs. Shelley Liu and Rebecca Trachtman. The TLC will be hosting two seminars next year, 1/19 and 3/16.

The TLC would like to thank last year's CHRS co-chairs, Drs. Shelley Liu and Alan Groves, MD and previous TLC members, including Oscar Rodriguez and Jennie Altman, PhD for all their hard work and dedication. Lastly, we would also like to thank Carolina Cappi, PhD, David Gonzalez, Xueying Zhang, PhD and Silvia De Rubeis, PhD for their efforts in leading the committee! We look forward to another great year ahead.



Carolina Cappi, PhD
Postdoctoral Fellow
Department of Psychiatry



David Gonzalez
MD, PhD Candidate
Department of Cell, Development
and Regenerative Biology



Xueying Zhang, PhD
Postdoctoral Fellow
Department of Environmental
Medicine and Public Health



Silvia De Rubeis, PhD
Assistant Professor
Department of Psychiatry

Save the Dates

Child Health Research Seminars

**Dates: January 19, 2021
March 16, 2021**

Time: 12:00 - 1:00 PM

New Extramural Faculty

Ernest Turro, PhD

Ernest Turro, PhD is an Associate Professor in the Department of Genetics and Genomic Sciences and the Arthur J. and Nellie Z. Cohen Chair. He is a biostatistician with 14 years of research experience in genomics and molecular diagnostics.

He has developed Bayesian statistical association methods for identifying the genetic determinants of rare diseases and he has applied them to a wide range of disorders, with an emphasis on hematological disorders and immune deficiencies. His work has implicated several genes in rare diseases for the first time and it has broadened understanding of the etiological roles of several others. Concurrently, Dr. Turro has worked on a diagnostic high-throughput sequencing platform that has been used to test thousands of patients with bleeding and platelet disorders, including patients with hereditary hemorrhagic telangiectasia.

Dr. Turro has also researched the interplay between genetic variation in the mitochondrial and nuclear genomes. This research found evidence that the mitochondrial genome may adapt to the cell nucleus. He has also helped repudiate a recent study questioning the dogma that mitochondrial DNA in humans is exclusively transmitted along the maternal lineage.



Ernest Turro, PhD

Associate Professor, Genetics and Genomic Sciences

Key Publications:

- Turro E, et al. Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*. 2020; 583:96–102.
- Wei W, ..., Turro E*, Chinnery PF*. Germline selection shapes human mitochondrial DNA diversity. *Science*. 2019; 364(6442).
- Greene E, NIH BioResource, Richardson S, Turro E. A fast association test for identifying pathogenic variants involved in rare diseases. *The American Journal of Human Genetics*. 2017; 101:104–11.
- Greene E, NIH BioResource, Richardson S, Turro E. Phenotype similarity regression for identifying the genetic determinants of rare diseases. *The American Journal of Human Genetics*. 2016; 98:490–499.
- Turro E, et al. Haplotype and isoform specific expression estimation using multi-mapping RNA-seq reads. *Genome Biology*. 2011; 12:R13.

Previously, Dr. Turro developed a number of influential statistical methods for modelling gene expression data. He developed the first statistical method for modelling isoform specific expression using microarray data. Later, he developed the first statistical methods for modelling haplotype and isoform specific expression using RNA sequencing data.

Dr. Turro has authored 60 peer-reviewed articles (10 as first author, 11 as last author), including work as a senior author in *Science*, *Nature*, *Blood* and the *American Journal of Human Genetics*, amongst others.

New Intramural Faculty

Yuval Itan, PhD

Yuval Itan, PhD is an Assistant Professor in the Department of Genetics and Genomic Sciences, and a core member of The Charles Bronfman Institute for Personalized Medicine, at the Icahn School of Medicine at Mount Sinai in New York City. He has obtained a BSc in computational biology from Bar-Ilan University, PhD in human evolutionary genetics from University College London, followed by postdoctoral research in human disease genomics at the Rockefeller University. Yuval has been extensively investigating the genetics that underlie human diseases, and has been developing computational methods to predict the functional consequence of human genetic variants in next generation sequencing data of patients, some examples include: (1) the human gene connectome (HGC) to prioritize disease-causing gene candidates by biological distance in protein-protein interactomic networks; (2) the gene damage index (GDI) to estimate the mutational damage of human genes in the general population; and (3) the mutation significance cutoff (MSC) to provide gene-based deleteriousness scores for accurate predictions. His current work includes: (1) developing a machine learning classifier to differentiate gain-of-function from loss-of-function mutations; (2) developing a deep learning classifier to detect pathogenic mutations based on disease groups; and (3) combining state-of-the-art with cutting edge methods in case-control gene burden association studies to detect novel disease-causing



Yuval Itan, PhD

Assistant Professor, Genetics and Genomic Sciences

Key Publications:

- Sevim Bayrak C, Zhang P, Tristani-Firouzi M, Gelb BD, Itan Y. De novo variants in exomes of congenital heart disease patients identify risk genes and pathways. *Genome Med*. 2020 Jan 15;12(1):9.
- Zhang P, Boisson B, Stenson PD, Cooper DN, Casanova JL, Abel L, Itan Y. SeqTailor: a user-friendly webserver for the extraction of DNA or protein sequences from next-generation sequencing data. *Nucleic Acids Res*. 2019 Jul 2;47(W1):W623-W631.
- Maffucci P, Bigio B, Rapaport F, Cobat A, Borghesi A, Lopez M, Patin E, Bolze A, Shang L, Bendavid M, Scott EM, Stenson PD, Cunningham-Rundles C, Cooper DN, Gleeson JG, Fellay J, Quintana-Murci L, Casanova JL, Abel L, Boisson B, Itan Y. Blacklisting variants common in private cohorts but not in public databases optimizes human exome analysis. *Proc Natl Acad Sci USA*. 2019 Jan 15;116(3):950-959.
- Zhang P, Bigio B, Rapaport F, Zhang SY, Casanova JL, Abel L, Boisson B, Itan Y. PopViz: a webserver for visualizing minor allele frequencies and damage prediction scores of human genetic variations. *Bioinformatics*. 2018 Dec 15;34(24):4307-4309.
- Itan Y, Shang L, Boisson B, Ciancanelli MJ, Markle JG, Martinez-Barricarte R, Scott E, Shah I, Stenson PD, Gleeson J, Cooper DN, Quintana-Murci L, Zhang SY, Abel L, Casanova JL. The mutation significance cutoff: gene-level thresholds for variant predictions. *Nat Methods*. 2016 Feb;13(2):109-10.

mutations and genes in various patient cohorts such as inflammatory bowel disease, congenital heart disease, COVID-19 and more.

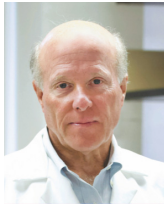
New Intramural Faculty

Andrew F. Stewart, MD

Andrew F. Stewart, MD is the Director, Diabetes Obesity and Metabolism Institute and Irene and Dr. Arthur M Fishberg Professor of Medicine. Dr. Stewart received his BS from Trinity College, and his M.D. from Columbia University. He was a postdoc at Yale, where he rose to tenured Professor. He served as Chief of Endocrinology at the University of Pittsburgh before moving to Mount Sinai in 2012.

His group was the first to induce robust replication of human insulin-producing beta cells. In 2015, they discovered the first drugs able to induce human beta cell replication, findings that have been reproduced around the world in pharma and academia. In 2017 and 2020, they defined the genomic pathways underlying beta cell expansion and insulin over-secretion in human insulinomas: a “wiring diagram” for human beta cell regenerative drug discovery. In 2019 and 2020, they reported that combination treatment with harmine and TGF- β inhibitors or GLP1 receptor agonists dramatically increases human beta cell proliferation. The work has clear translational implications for Types 1 as well as Type 2 diabetes, both of which result from an absolute or relative deficiency of insulin-producing beta cells.

He has published more than 250 papers in the NEJM, Science, Science Translational Medicine, Cell Metabolism, Nature Medicine, Nature Communications, JCI, PNAS, and others. He has had continuous NIH grant support for the past 40 years. He served as Councilor and Secretary-Treasurer of the Endocrine Society, and was the 2008 recipient of the Endocrine Society’s Gerald Aurbach Award for outstanding scientific achievement. He served as the Chair of the Endocrine Society Meeting in 1998 and American Diabetes Association Annual Meetings for 2010 and 2011.



Andrew F. Stewart, MD

Director, Diabetes Obesity and Metabolism Institute
Irene and Dr. Arthur M Fishberg Professor, Medicine

Key Publications:

Wang P, Felsenfeld DP, Liu H, Sivendran S, Bender A, Kumar A, Alvarez-Perez JC, Garcia-Ocaña A, Sanchez R, Scott DK, Stewart AF. [A high-throughput chemical screen reveals that harmine-mediated inhibition of DYRK1A increases human pancreatic beta cell replication.](#) *Nat Med.* 2015 Apr;21(4):383-8.

Wang P, Karakose E, Liu H, Swartz E, Aceffi C, Zlatanic V, Wilson J, Argmann C, Scott DK, Garcia-Ocaña A, Stewart AF. [Combined Inhibition of DYRK1A, SMAD and Trithorax Pathways Synergizes to Induce Robust Replication in Adult Human Beta Cells.](#) *Cell Metab.* 2019 Mar 5;29(3):638-652.e5.

Wang H, Bender A, Wang P, Karakose E, Inabnet WB, Libutti SK, Arnold A, Lambertini L, Stang MT, Chen H, MD, Kasai Y, Mahajan M, Kinoshita Y, Fernandez-Ranvier G, Becker TC, Takane KK, Walker LA, Saul S, Chen R, PhD, Scott DK, Ferrer J, Antipin Y, Donovan M, Uzilov AV, Reva B, PhD, Schadt EE, Losic B, Argmann C, Stewart AF. [Insights into Beta Cell Regeneration for Diabetes via Integration of Molecular Landscapes in Human Insulinomas.](#) *Nat Commun.* 2017 Oct 3;8(1):767.

Aceffi C, Wang P, Karakose E, Manning Fox JE, González BJ, Liu H, Swartz E, Kumar K, MacDonald PE, Sanchez R, DeVita R, Homann D, Egli D, Garcia-Ocaña A, Scott DK, Stewart AF. [Conversion of GLP1 Receptor Agonist Diabetes Drugs into Potent Human Beta Cell Regenerative Drugs By Combination with DYRK1A Inhibitors.](#) *Sci Transl Med.* 2020 Feb 12;12(530):eaaw9996.

Karakose E, Wang H, Inabnet W, Thakker RV, Libutti S MD, Fernandez-Ranvier G, Suh H, Stevenson M, Kinoshita Y, Donovan M, Antepin Y, Wang P, Uzilov A, Argmann C, Schadt EE, Stewart AF, Scott DK, Lambertini L. [Aberrant Methylation of the 11p15 Imprinted Region Underlies Abnormal Chromatin Arrangement, Insulin Expression and Cell Proliferation in Human Insulinomas.](#) *Nat Commun.* 2020;11, 5210.

Grants, Awards/Honors, Publications

Faculty Grants

Dusan Bogunovic, PhD, NIAID, R01, “Inborn Errors of Immunity Leading to Autoinflammatory Syndromes”

Dusan Bogunovic, PhD, NIAID, R01, “Role of SARS-CoV-2-mediated Type I IFN antagonism in individuals with Down Syndrome”

Dusan Bogunovic, PhD, and Brad Rosenberg, MD, PhD, NIAID, R01, “Next Generation Resolution of Antiviral Gene Networks”

Adolfo Garcia-Ocana, PhD and Donald K. Scott, PhD, NIDDK, R01, “Myc Physiology in the Pancreatic Beta Cell”

Adolfo Garcia-Ocana, PhD, Robert J. DeVita, PhD, and Andrew F. Stewart, MD, NIDDK, R01, “Biological and Medicinal Chemistry Approaches to Human Beta Cell Regeneration”

Hirofumi Morishita, MD, PhD, NIH/NEI, R01, “Nicotinic Modulation of Deep Layer Inhibitory Neurons for Visual Cortical Plasticity”

Andrew J. Sharp, PhD, NIH/NIA, R03, “Investigating Tandem Repeat Variation as a Cause of Alzheimer’s Disease From Exome Sequencing Data”

Andrew J. Sharp, PhD, NIH/NIA, R01, “Pilot Investigation of Tandem Repeat Variation as a Cause of Alzheimer’s Disease Using Whole Genome Sequencing”

Andrew J. Sharp, PhD, American Parkinson’s Disease Association, “Identifying Novel Repeat Expansions as a Cause of Parkinson’s Disease”

Faculty Honors/Awards

Dalila C. Pinto, PhD, World Conference on Psychiatric Genetics, October 20, 2020, Co-Chair and Speaker of Symposium “PsychENCODE Updates: Discovery of Functional Elements for Psychiatric GWAS Signals”

Dalila C. Pinto, PhD, 25rd Biennial Meeting of the International Society for Developmental Neuroscience, Chair and Speaker of Symposium “Autism Spectrum Disorders”

Hirofumi Morishita MD, PhD, One Mind Bipolar Research Award

Trainee Honors/Awards

Xueying Zhang, PhD, National Public Radio, Invited Speaker, “New Study Points To Invisible Killer Of Infants”

Publications

- Patil SU, **Bunyavanich S**, **Berin MC**. Emerging food allergy biomarkers. *J Allergy Clin Immunol Pract*. 2020 Sep;8(8):2516-24.
- Mansoor A, Mansoor MO, Patel JL, Zhao S, Natkunam Y, **Bieker JJ**. *Klf1/eklf* expression in acute leukemia is correlated with chromosomal abnormalities. *Blood Cells Mol Dis*. 2020 Jul;83:102434.
- Malle L, Gao C, Hur C, Truong HQ, Bouvier NM, Percha B, ... **Bogunovic D**. Individuals with Down syndrome hospitalized with COVID-19 have more severe disease. *Genet Med*. 2020 Oct 16. [Online ahead of print]
- Gruber CN, Calis JJA, Buta S, Evrony G, Martin JC, Uhl SA, ..., **Dunkin D**, ... **Webb BD**, **Saland JM**, ... **Gelb BD**, **Bogunovic D**. Complex autoinflammatory syndrome unveils fundamental principles of jak1 kinase transcriptional and biochemical function. *Immunity*. 2020 Sep 15;53(3):672-84.e11.
- Gruber CN, Patel RS, **Trachtman R**, Lepow L, Amanat F, Krammer F, **Wilson KM**, ... **Gelb BD**, ... **Bogunovic D**. Mapping systemic inflammation and antibody responses in multisystem inflammatory syndrome in children (mis-c). *Cell*. 2020 Sep 14.
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- Kunkle BW, Schmidt M, Klein HU, Naj AC, Hamilton-Nelson KL, Larson EB, ... **Buxbaum JD**, ... Kukull WA. Novel alzheimer disease risk loci and pathways in african american individuals using the african genome resources panel: A meta-analysis. *JAMA Neurol*. 2020 Oct 19.
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- Jin H, Moss R, Reed JC, Hertzberg E, Cruz MR, Akkoyun E, ... **Bogunovic D**, **Cunningham-Rundles C**. *Ifn-γ* receptor 2 deficiency initial mimicry of multisystem inflammatory syndrome in children (mis-c). *J Allergy Clin Immunol Pract*. 2020 Sep 8.
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