

**Faculty of Medicine of Harvard University  
Curriculum Vitae**

**Date Prepared:** January 6, 2021

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**Place of Birth:** US

**Education**

05/2002	BA ( <i>summa cum laude</i> )	Biochemistry (Mark A Lemmon)	University of Pennsylvania
05/2002	MS	Biochemistry & Molecular Biophysics (Mark A Lemmon)	University of Pennsylvania
08/2003	M.Phil/Ph.M	Biochemistry (Sir Tom Blundell)	University of Cambridge
05/2010	MD, PhD ( <i>magna cum laude</i> )	Medicine/ Genetics (Stuart H Orkin)	Harvard Medical School/ Massachusetts Institute of Technology, Medical Scientist Training Program

**Postdoctoral Training**

10/08 - 09/09	Research Fellow	Molecular Hematology (Stuart H. Orkin)	Boston Children's Hospital
10/09 - 06/13	Visiting Scientist/Scholar	Human Genetics (Harvey F. Lodish and Eric S. Lander)	Broad Institute & Whitehead Institute for Biomedical Research
06/10 - 06/11	Intern	Pediatrics	Boston Children's Hospital
06/10 - 12/13	Clinical Fellow	Clinical Fellow in Pediatrics (EXT)	Harvard Medical School, Harvard Medical School
07/11 - 06/13	Resident	Pediatrics	Boston Children's Hospital
07/13 - 06/15	Clinical Fellow	Pediatric Hematology / Oncology	Boston Children's Hospital

## Faculty Academic Appointments

01/14 - 01/19	Assistant Professor	Pediatrics	Harvard Medical School
02/19 - Present	Associate Professor	Pediatrics	Harvard Medical School

## Appointments at Hospitals/Affiliated Institutions

### Current

07/15 - Present	Attending Physician	Pediatrics (Hematology/Oncology)	Boston Children's Hospital
07/15 - Present	Attending Physician	Pediatrics (Hematology/Oncology)	Dana-Farber Cancer Institute
07/15 - Present	Consulting Staff	Pediatrics	Beth Israel Deaconess Medical Center
07/15 - Present	Consulting Staff	Pediatrics	Brigham and Women's Hospital

## Other Professional Positions

2010 - Present	Member	American Academy of Pediatrics	
2013 - Present	Member	American Society of Hematology	
2013 - Present	Associate Member	Broad Institute of MIT and Harvard	
2015 - Present	Principal Faculty	Harvard Stem Cell Institute	Principal Faculty
2019 - 2020	Interim Chief, Division of Hematology/Oncology	Boston Children's Hospital	10%

## Major Administrative Leadership Positions

### Local

2016 - Present	Course Co-Director, BCMP 218/ HST 140: "Molecular Medicine"	Harvard Medical School
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## Committee Service

### Local

2015 - 2017	Institutional Biosafety Committee 2015 - 2017	Boston Children's Hospital Committee Member
2017 - Present	Institutional Precision Medicine Committee	Boston Children's Hospital

2017 - Present Committee Member

### International

2020 - Present International Common Disease Alliance Working Group  
2020 - Present Committee Member

### **Professional Societies**

2010 - Present American Academy of Pediatrics (AAP)

2013 - Present American Society of Hematology (ASH)

2016 - Present American Society of Human Genetics (ASHG)

2016 - Present International Society for Experimental Hematology

2016 - Present Society for Pediatric Research (SPR)

2016 - Present Elected Member

2017 - Present American Society of Hematology Scientific Committee on Red Cell Biology

2017 - Present Elected Member

2018 - Present American Society for Clinical Investigation (ASCI)

2018 - Present Elected Member

### **Editorial Activities**

#### Ad hoc Reviewer

*American Journal of Hematology*

*Blood*

*Blood Advances*

*Blood, Cells, Molecules, and Diseases*

*British Journal of Hematology*

*Cell*

*Cell Reports*

*Cell Stem Cell*

*eLife*

*Haematologica*

*Hemoglobin*

*Human Molecular Genetics*

*Journal of Clinical Investigation*

*Journal of Experimental Medicine*

*Nature*

*Nature Cell Biology*

*Nature Communications*

*Nature Genetics*

*Nature Medicine*

*Nature Reviews Genetics*

*PLOS Genetics*

*Proceedings of the National Academy of Sciences of the U.S.A.*

*Science*

*Science Translational Medicine*

*The American Journal of Human Genetics*

*The New England Journal of Medicine*

#### Other Editorial Roles

2019 - Present Editorial Board Member *Blood*

2019 - Present Editorial Board Member *Review Commons*

#### **Honors and Prizes**

2010	Henry Asbury Christian Award	Harvard Medical School	For notable scholarship in research by graduate of Harvard Medical School
2011	Children's Hospital Boston Alumni Association Travel Award	Children's Hospital Boston	Research/ clinical award
2012	Beth Israel Deaconess Medical Center Outstanding Resident Teaching Award	Beth Israel Deaconess Medical Center	Medical student teaching
2014	Basil O'Connor Scholar Award	March of Dimes Foundation	For young faculty members doing research relevant to birth defects
2014	Cohen Memorial	Children's Hospital of Philadelphia	An honorary lectureship

	Lectureship		where presentation was given at medical grand rounds at the Children's Hospital of Philadelphia
2014	Diamond Blackfan Anemia Achievement Award	Diamond Blackfan Anemia International Consensus Conference	Given to a single individual every two years for contributions to the Diamond Blackfan anemia field
2015	Rising Star Award	Boston Children's Hospital	
2015	Young Investigator Award	Society for Pediatric Research	
2016	New Investigator Award	International Society for Experimental Hematology	Given annually to a young investigator whose work has had a significant impact on the field of hematology research
2017	Outstanding New Member Award and Lecture	Society for Pediatric Research	
2017 - 2018	Faculty Teaching Honor Roll	Boston Combined Residency Program in Pediatrics	
2018	Gale and Ira Drukier Prize in Children's Health Research	Gale and Ira Drukier and the Department of Pediatrics at Weill Cornell Medicine	This prize honors an early-career pediatrician whose research has made important contributions towards improving the health of children and adolescents
2018 - Present	New York Stem Cell Foundation-Robertson Investigator	New York Stem Cell Foundation	
2019	Donald Seldin-Holly Smith Award for Pioneering Research	American Society of Clinical Investigation	
2019 - Present	Lodish Family Career Development Chair	Boston Children's Hospital	
2020	Visiting Professorship	The Aflac Cancer and Blood Disorders Center at Children's Healthcare of Atlanta and Emory University	

## **Report of Funded and Unfunded Projects**

### **Grants and Sponsored Research**

#### **Past**

- 2013 - 2018  
NCE 2018 -  
2019
- Translational and clinical studies targeting  $\gamma$ -globin modulation  
National Institutes Of Health (NIH), 5U01HL117720-05 (5 cycles)  
PD/PI, Direct Costs: \$236,679  
Current funding cycle began in 2017.  
The goal is to exploit the observation that individuals with propionic acidemia (PA) have enhanced HbF expression, in 3 specific aims, the PI proposes to 1) extend the observation that PA is associated specifically with HbF induction, 2) determine the unique metabolites that may be associated with said induction and 3) examine the effects of candidate metabolites on in vitro erythroid cell cultures. Specific globin gene expression, global gene expression, epigenetic modification of the beta globin locus, and a specific analysis of the effects of these metabolites on HDAC activity are proposed.
- 2013 - 2019  
NCE 2018 -  
2019
- Translational and clinical studies targeting  $\gamma$ -globin modulation  
NATIONAL HEART, LUNG, AND BLOOD INSTITUTE, 5U01HL117720-03 (5 cycles)  
PD/PI, Direct Costs: \$411,312  
Current funding cycle began in 2017.  
The goal is to exploit the observation that individuals with propionic acidemia (PA) have enhanced HbF expression, in 3 specific aims, the PI proposes to 1) extend the observation that PA is associated specifically with HbF induction, 2) determine the unique metabolites that may be associated with said induction and 3) examine the effects of candidate metabolites on in vitro erythroid cell cultures. Specific globin gene expression, global gene expression, epigenetic modification of the beta globin locus, and a specific analysis of the effects of these metabolites on HDAC activity are proposed.
- 2013 - 2016
- The Manton Center for Orphan Disease Research: Endowed Scholar Award  
Children'S Hospital Boston, N/A  
Investigator  
The goals of this award are to pursue studies of rare disorders of red blood cell production, including Diamond-Blackfan anemia and Cartilage Hair Hypoplasia
- 2013 - 2014
- Systematic Dissection of Human Genetic Variation in Erythropoiesis (Broad Funding Collaboration)  
Children'S Hospital Boston, N/A  
Investigator  
The goal of this grant is to utilize massively parallel sequencing resources at the Broad Institute to better define the basis of genetic variation in red blood cell traits.
- 2014 - 2016
- The Role of GATA1 in Diamond-Blackfan (Congenital Hypoplastic) Anemia  
March Of Dimes, N/A  
Investigator  
The aim of this grant is to understand how GATA1 plays a role in the pathogenesis of Diamond-Blackfan anemia and study at a mechanistic level how GATA1 mRNA translation is downregulated by ribosomal protein haploinsufficiency, a common genetic cause of Diamond-Blackfan anemia.

- 2014 - 2016 Identifying genome-wide association study-nominated regulators of erythropoiesis  
National Heart, Lung And Blood Institute (NHLBI), 1R21HL120791-01  
Investigator  
Collectively, aims will exploit erythroid trait GWAS results to identify important new regulators of erythropoiesis, a process of significant clinical and biological importance. More generally, this work has the potential to serve as a paradigm for how the power of GWAS can be exploited to increase our understanding of biology and medicine.
- 2014 - 2015 Defining GATA1 Transcriptional Alterations in Diamond-Blackfan Anemia  
Diamond Blackfan Anemia Foundation, N/A  
Investigator  
Per PI: Grant given for Supplies/Reagents only.
- 2014 - 2019 Systematic Genetic Dissection of Human Erythropoiesis  
National Institutes Of Health (NIH), 5R01DK103794-04 (5 cycles)  
PD/PI, Direct Costs: \$1,125,000  
Current funding cycle began in 2018.  
The goal of this grant is to uncover common regulatory patterns that underlie common genetic variation in red blood cell traits. This grant is focused on improving our understanding of how transcription factor binding and histone marks may be altered by human genetic variation with the focus being on variation of erythroid traits.
- 2015 - 2017 Enabling personalized cell therapy by making human HSCs in vitro (Broad Funding  
NCE 2017 - Collaboration)  
2018 Broad Institute  
Investigator  
The goal of this project is to use single cell RNA sequencing to examine and distinguish various hematopoietic progenitors arising from human pluripotent stem cells.
- 2016 - 2019 Identifying genome-wide association study-nominated regulators of erythropoiesis  
National Heart, Lung And Blood Institute (NHLBI), 5R33HL120791-04 (3 cycles)  
PD/PI, Direct Costs: \$972,831  
Current funding cycle began in 2018.  
Collectively, aims will exploit erythroid trait GWAS results to identify important new regulators of erythropoiesis, a process of significant clinical and biological importance. More generally, this work has the potential to serve as a paradigm for how the power of GWAS can be exploited to increase our understanding of biology and medicine.
- 2016 - 2018 Enabling personalized leukemia cell therapy by human hematopoietic stem cell  
NCE 2017 - generation  
Present DFCI, 9617550  
PI, Direct Costs: \$68,182  
Enabling personalized leukemia cell therapy by human hematopoietic stem cell generation
- 2017 - 2018 Dissecting the Genetic Architecture of Human Hematopoiesis  
American Society of Hematology  
PI, Direct Costs: \$5,000  
Erik Bao ASH HONORS Stipend
- 2017 - 2018 Dissecting the Genetic Architecture of Diamond Blackfan Anemia  
DIAMOND BLACKFAN ANEMIA FOUNDATION, N/A

PD/PI, Direct Costs: \$56,021

This grant is focused on completing analysis of a large exome sequencing study of patients with a diagnosis of Diamond-Blackfan anemia. We have exome sequencing data from a total of 450 patients with Diamond-Blackfan anemia

2017 - 2018 Lineage Tracing by Mitochondrial Mutation Tracking in Single Cells  
BROAD Institute  
PI, Direct Costs: \$50,000  
Lineage Tracing by Mitochondrial Mutation Tracking in Single Cells

### Current

2017 - 2019 Refining the Human Genetics of Fetal Hemoglobin Regulation  
Vertex Pharmaceuticals Incorporated, N/A (2 cycles)  
PD/PI, Direct Costs: \$406,675  
Current funding cycle began in 2017.  
This human genetic study intends to provide important insight into fetal hemoglobin (HbF) regulation, yield potential approaches for genome editing strategies, and point to novel targets for small molecule therapeutics to induce HbF in patients with sickle cell disease and B-thalassemia.

2018 - 2019 Defining regulators of immunity to acute infection using CRISPR screens (Pilot Project)  
HARVARD MEDICAL SCHOOL/NIH, U19  
Site PI, Direct Costs: \$40,937  
Inference of clonal immune cell populations via mitochondrial mutations and single-cell assays

2019 - 2020 Dissecting Germline Genetic Risk for Myeloproliferative Neoplasms  
Dana-Farber Cancer Institute, 9619501 (2 cycles)  
PD/PI, Direct Costs: \$260,000  
To utilize large population-based studies to delineate common and rare genetic variants that predispose individuals to myeloproliferative neoplasms (MPNs). To predict the gene targets of MPN risk variants and the stages of human hematopoiesis in which they act. To dissect the functional mechanisms by which MPN risk variants predispose to disease.

2019 - 2023 Genetic Studies of Human Hematopoiesis  
NEW YORK STEM CELL FOUNDATION, NYSCF-R-I48 (5 cycles)  
PD/PI, Direct Costs: \$1,500,000  
This grant uses genetic studies and follow-up analysis to further understand both normal and disordered human blood cell production.

2019 - 2020 Mapping enhancer function across differentiation of primary hematopoietic cells in vitro  
BROAD INSTITUTE, 4000670  
PD/PI, Direct Costs: \$50,000  
The goal is to carry out experiments in primary blood progenitor cells to examine how trait-associated genetic variation may impact cellular functions and provide a paradigm for moving from variant to function across a range of human diseases and traits.

2019 - 2020 Gene Therapy for Diamond-Blackfan Anemia  
Boston Children's Hospital Translational Research Program, TRP



PD/PI, Direct Costs: \$56,000

The goal of this grant is to help develop a gene therapy approach for Diamond-Blackfan anemia through the use of regulated GATA1 expression.

- 2020 - 2023 Dissecting Pathogenic Mechanisms of ETV6 Deregulation in Myelodysplastic Syndromes  
EVANSMDS, DRG2020  
PD/PI, Direct Costs: \$545,454  
The goal of this project is to dissect how germline variants in ETV6 predispose to myelodysplastic syndrome and how these mutations can cooperate with somatic mutations to promote progression in this disease

### **Training Grants and Mentored Trainee Grants**

- 2019 - 2024 Systematic Genetic Dissection of Human Erythropoiesis (Active)  
National Institutes Of Health (NIH), 2R01DK103794-06 (5 cycles)  
PD/PI, Direct Costs: \$1,125,000  
The goal of this grant is to uncover common regulatory patterns that underlie common genetic variation in red blood cell traits. This grant is focused on improving our understanding of how transcription factor binding and histone marks may be altered by human genetic variation with the focus being on variation of erythroid traits.
- 2019 - 2021 Dissecting Genetic Risk for Myeloproliferative Neoplasms (Active)  
MPN Research Foundation  
PD/PI, Direct Costs: \$100,000  
The goal of this project is to define the underlying biology of genetic loci that predispose individuals to acquiring MPNs or other clonal disorders
- 2020 - 2024 Next Generation Functional Genomics of Hematology Traits (Active)  
NIH / University of Washington, 1R01HL146500-01A1  
PD/PI, Direct Costs: \$299,250  
The goal of this collaborative grant is to perform further genetic studies of population-based variation in blood cell traits and functionally interrogate the impact of these variants in human hematopoiesis
- 2020 - 2021 Integrating Human Genetics and Single-Cell Functional Assays to Elucidate Mechanisms of Fetal Hemoglobin Regulation (Active)  
NIH/NIDDK, 1R56DK125234-01  
PD/PI  
The goal of this grant is to integrate information from human genetic studies with a single-cell functional assay to define how fetal hemoglobin regulation occurs
- 2020 - 2021 Dissecting Pathogenic Mechanisms of ETV6 Deregulation in Myeloidysplastic Syndromes (Pending)  
NIH/NCI  
PD/PI  
This grant seeks to dissect how germline variants in ETV6 predispose to myeloid malignancies and how these mutations can cooperate with the somatic mutations to promote disease progression

## **Report of Local Teaching and Training**

### **Teaching of Students in Courses**

2014	Hematology Course  2nd year HMS MD students	Harvard Medical School Boston, Massachusetts, United States 2 hour lecture and discussion session
2015	Hematology Course  2nd year HMS MD students	Harvard Medical School Boston, Massachusetts 2 hour lecture and discussion session
2015	Thalassemia: An Overview MD Students	Harvard Medical School 2 hour lecture and discussion session
2016	Hematology Course	Harvard Medical School Boston, Massachusetts 2 hour lecture and discussion session
2016 - Present	BCMP 218/ HST 140: "Molecular Medicine"	Harvard Medical School  Co-Director of Course for Fall Semester

### **Formal Teaching of Residents, Clinical Fellows and Research Fellows (post-docs)**

2012 - 2014	Uncertainty in clinical genetic testing  Residents at Boston Children's Hospital	Boston Children's Hospital Boston, Massachusetts, United States Resident Teaching
2013	HST 160: Genetics  1st year HST MD and PhD students	Local Teach and Training Boston, Massachusetts, United States 4 hour lecture and discussion session

### **Formally Mentored Harvard Medical, Dental, and Graduate Students**

2016 - 2019	Aaron Cheng, HMS class of 2020 Daily mentorship - Studied the genetic basis of rare blood disorders. Received HHMI Medical Research Fellowship in 2018.
2017 - 2020	Caleb A Lareau, HMS class of 2020 Daily mentorship - Studying the genetic basis of variation in human hematopoiesis using single cell genomic approaches.
2017 - Present	Erik L Bao, HMS class of 2021 Daily mentorship - Studies of genetic variation in human hematopoiesis and blood disorders. Received HHMI Medical Research Fellowship in 2018

- 2018 - Present Emory Werner, HMS class of 2021  
Daily mentorship - Ongoing studies of genetic variation impacting blood traits and polygenic selection in human history
- 2020 - Present Jorge Diego Martin Rufino, DMS class of 2023  
Daily mentorship - Functional genetic studies of human hematopoiesis.

### **Other Mentored Trainees and Faculty**

- 2011 - 2015 Leif Ludwig, Charite Universitätsmedizin-Berlin, Germany class of 2011 /  
*Career Stage:* Graduate  
*Accomplishments:* Daily mentorship – Supervision of MD-PhD student
- 2013 - 2015 Felix C Giani, Charite Universitätsmedizin-Berlin, Germany class of 2016 /  
*Career Stage:* Graduate  
*Accomplishments:* Daily mentorship – Supervision of MD student
- 2014 - 2017 Rajiv K Khajuria, Charite Universitätsmedizin-Berlin, Germany class of 2019 /  
*Career Stage:* Graduate  
*Accomplishments:* Daily mentorship – Supervision of MD student
- 2014 - 2019 Claudia Fiorini, PhD / Senior Scientist, AvroBio  
*Career Stage:* Research Scientist *Mentoring Role:* Mentor/PI *Accomplishments:* Functional and mechanistic studies of human hematopoiesis
- 2014 - 2020 Anindita Basak, PhD / Senior Scientist, Forma Therapeutics  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/PI *Accomplishments:* Studies of human fetal hemoglobin regulation.
- 2014 - Present Satish K. Nandakumar, PhD  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/PI *Accomplishments:* Studies of how common genetic variation impacts human hematopoiesis
- 2015 - 2018 Ah Ram Kim, PhD / Senior Scientist, Vertex Pharmaceuticals  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/PI *Accomplishments:* Studies of human variation in cell signaling pathways impacting hematopoiesis.
- 2016 - 2018 Sean K. McFarland, PhD / Senior Scientist, Tevard Biosciences  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/PI *Accomplishments:* Computational analysis of functional studies to assess how common genetic variation impacts human hematopoiesis.
- 2017 Beat Frey, MD / C.E.O. and Chief Physician, Blutspende Zurich  
*Career Stage:* Visiting Scientist (On Sabbatical) *Mentoring Role:* Mentor/PI  
*Accomplishments:* Genetic studies of human hematopoiesis
- 2017 - 2018 Laura Mateyka, University of Heidelberg class of 2018 /  
*Career Stage:* Graduate  
*Accomplishments:* Daily mentorship – Studied the mechanisms of variation in human hematopoiesis.
- 2017 - 2019 Kara Montbleau, Boston University class of 2019 /  
*Career Stage:* Graduate  
*Accomplishments:* Daily mentorship – Studying fetal hemoglobin regulation in humans. Received Samuel E. Lux, MD Fellowship in 2017.

- 2017 - 2020 Leif S. Ludwig, MD/PhD / Group Leader, Berlin Institute for Medical Systems Biology/ Max Delbrück Center for Molecular Medicine  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/PI *Accomplishments:* Developing approaches for lineage tracing in humans and studying how transcriptional regulation plays a role in human hematopoiesis.
- 2018 - 2019 Chantree Hantaweepant, MD / Hematologist, Siriraj Hospital/ Mahidol University  
*Career Stage:* Visiting Postdoctoral Fellow *Mentoring Role:* Mentor/ PI  
*Accomplishments:* Studies of genetic variation in beta-thalassemia.
- 2018 - Present Yong Shen, PhD  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/ PIU *Accomplishments:* Genetic studies of fetal hemoglobin regulation.
- 2018 - Present Richard A Voit, MD/PhD  
*Career Stage:* Postdoctoral Fellow/ Clinical Fellow *Mentoring Role:* Mentor/ PI  
*Accomplishments:* Genetic studies of human hematopoietic stem cell biology and disruption in bone marrow failure disorders.
- 2019 - 2020 Ayesha Ejaz, MD / Hematologist, Oxford University Hospital/ University of Oxford  
*Career Stage:* Visiting Postdoctoral Fellow *Mentoring Role:* Mentor/ PI  
*Accomplishments:* Genetic studies of fetal hemoglobin regulation.
- 2019 - Present L. Alexander Liggett, PhD  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/ PI *Accomplishments:* Impact of inherited variation on somatic mutation rate in clonal hematopoietic disorders.
- 2019 - Present Lara Wahlster, MD/PhD  
*Career Stage:* Postdoctoral Fellow/ Clinical Fellow *Mentoring Role:* Mentor/ PI  
*Accomplishments:* Genetic studies of rare high impact alleles predisposing to myeloid malignancies.
- 2020 - Present Liam Cato, M.B. Ch.B.  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/ PI *Accomplishments:* Inherited basis of myeloid malignancy predisposition.
- 2020 - Present Alexis Caulier, MD/PhD  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/ PI *Accomplishments:* Studies on genetic variation impacting hematopoiesis
- 2020 - Present Fulong Yu, PhD  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/ PI *Accomplishments:* Computational reconstruction of regulatory networks in human hematopoiesis
- 2020 - Present Jiawei Zhao, PhD  
*Career Stage:* Postdoctoral Fellow *Mentoring Role:* Mentor/ PI *Accomplishments:* Regulation of human hematopoiesis by transcriptional regulators implicated through genetics.

### Local Invited Presentations

Those presentations below sponsored by 3rd parties/outside entities are so noted and the sponsor(s) is identified

- 2011 Regulation of Fetal Hemoglobin / Oral Presentation

- Orphan Disease Symposium  
Boston Children's Hospital (Manton Center for Orphan Disease Research)
- 2013 Regulation of Fetal Hemoglobin / Oral Presentation  
Grand Rounds  
Boston Children's Hospital (Department of Medicine)
- 2015 Genetic Studies of Human Hematopoiesis / Oral Presentation  
Grand Rounds  
Medicine, Boston Children's Hospital
- 2015 New Insights into Diamond-Blackfan Anemia / Oral Presentation  
Grand Rounds  
Medicine, Boston Children's Hospital
- 2016 From GWAS Loci to Causal Nucleotides and Target Genes: Lessons from Human  
Hematopoiesis / Oral Presentation  
Common Disease Workshop  
Broad Institute
- 2016 Genetic Studies of Human Hematopoiesis / Oral Presentation  
Models of Disease Boot Camp for Fellows  
Harvard Catalyst Program
- 2016 What Patients Can Teach Us About Hematopoiesis / Oral Presentation  
Seminar Series  
Medicine, Boston Children's Hospital
- 2018 Genetic Studies of Human Hematopoiesis / Demonstration  
Transfusion Medicine Conference  
Harvard Medical School

**Report of Regional, National and International Invited Teaching and Presentations**

Those presentations below sponsored by 3rd parties/outside entities are so noted and the sponsor(s) is identified.

Regional

- 2014 Using human genetics to understand and development treatments for blood disorders /  
Oral Presentation  
Manton Center for Orphan Disease Research  
Boston Children's Hospital  
Boston, Massachusetts (Manton Center for Orphan Disease Research)
- 2018 Of Lineage & Legacy: Tracing Cellular Hierarchies in Intact Humans / Oral Presentation  
- Presenter  
Cardiovascular Institute Seminar Series  
University of Pennsylvania  
Philadelphia, Pennsylvania
- 2019 There Will Be Blood: Human Genetic Studies of Blood Cell Production in Health &  
Disease / Oral Presentation - Presenter  
Vertex Pharmaceuticals Science & Medicine Seminar  
Boston, Massachusetts

- 2019      There Will Be Blood: Genetic Studies of Human Hematopoiesis / Oral Presentation -  
Presenter  
Clinical Research Seminar, Rockefeller University  
New York, New York
- 2019      Of Lineage & Legacy: Tracing Cellular Hierarchies in Intact Humans / Oral Presentation  
- Presenter  
Human Genetics in NYC Meeting  
New York, New York
- 2019      Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
Bar Harbor, Maine
- 2019      What Can Patients Teach Us About Blood Cell Production? / Oral Presentation -  
Presenter  
Pediatric Hematology/ Oncology Grand Rounds, University of Michigan C.S. Mott  
Children's Hospital  
Ann Arbor, Michigan
- 2019      Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
The Ruth L. and David S. Gottesman Institute for Stem Cell and Regenerative Medicine  
Research, Albert Einstein College of Medicine    Bronx, NY, USA  
Bronx, New York
- 2019      Why Are Some Individuals Predisposed to Acquiring Blood Cancers? / Keynote/Plenary  
Address - Presenter  
Dana-Farber Cancer Institute Department of Pediatric Oncology Annual Retreat  
Dedham, Massachusetts
- 2019      Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
University of Virginia Center for Public Health Genomics - Genome Sciences Seminar  
Charlottesville, Virginia
- 2019      How Can We Learn from Patients About Blood Production / Oral Presentation -  
Presenter  
Boston Children's Hospital Division of Newborn Medicine Seminar Series  
Boston Children's Hospital  
Boston, Massachusetts
- 2019      Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
UT Southwestern Children's Medical Center Research Institute Seminar  
Dallas, Texas
- 2020      What Can We Learn From Patients About Fetal Hemoglobin Regulation? / Oral  
Presentation - Presenter  
The Aflac Cancer and Blood Disorders Center at Children's Healthcare of Atlanta and  
Emory University Seminar
- 2020      What Can We Learn From Patients About Fetal Hemoglobin Regulation? / Oral  
Presentation - Presenter  
National Heart, Lung, and Blood Institute - Hematology Conference  
Bethesda, Maryland
- 2020      What Can We Learn From Patients About Fetal Hemoglobin Regulation? / Oral

Presentation - Presenter  
 Children's Hospital of Michigan - Pediatric Grand Rounds  
 Detroit, Michigan

2020 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
 Massachusetts General Hospital Center for Genomic Medicine Seminar Series  
 Boston, Massachusetts

2020 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
 City College of New York Department of Biology Colloquium  
 New York, New York

National

2010 New insights into fetal hemoglobin regulation / Oral Presentation  
 Division of Hematology  
 Children's Hospital of Philadelphia  
 Philadelphia, Pennsylvania

2011 Regulation of erythropoiesis by cyclin D3 / Oral Presentation  
 Red Cell Club Meeting  
 Philadelphia, Pennsylvania

2013 Genetics of human erythropoiesis / Oral Presentation  
 Abramson Family Cancer Research Institute Seminar Series  
 University of Pennsylvania  
 Pennsylvania

2013 The role of GATA1 in rare and common human genetic variation in erythropoiesis / Oral  
 Presentation  
 Lindsley Kimball Research Institute Seminar Series  
 New York Blood Center  
 Pennsylvania

2014 Impaired hematopoiesis through altered translation / Lecture - Presenter  
 56th Annual Meeting of the American Society of Hematology  
 San Francisco, California

2014 Using human genetics to understand and development treatments for blood disorders /  
 Oral Presentation  
 Cohen Memorial Lectureship, Pediatric Grand Rounds  
 Children's Hospital of Philadelphia  
 Philadelphia, Pennsylvania

2014 Genetic studies of human hematopoiesis / Oral Presentation  
 Hematology Seminar Series  
 St. Jude Children's Research Hospital  
 Memphis, Tennessee

2015 Genetics Studies of Human Hematopoiesis / Oral Presentation  
 Harrington Discovery Institute  
 Case Western Reserve University  
 Cleveland, Ohio

2015 Human Genetic Studies of Blood Disorders: What Patients Have Taught Us about Blood

- Cell Production / Oral Presentation  
Society for Pediatric Research Annual Meeting  
Young Investigator Award Seminar  
San Diego, California (Young Investigator Award Seminar)
- 2015 Genetic studies of human erythropoiesis / Oral Presentation  
Special seminar  
Rockefeller University  
New York City, New York
- 2016 What Patients Can Teach Us About Hematopoiesis / Oral Presentation  
MSTP Program Annual Lecture  
Indiana University, Indianapolis  
Indianapolis, Indiana
- 2016 What Patients Can Teach Us About Hematopoiesis / Oral Presentation  
Seminar Series Division of Hematology/Oncology  
University of Michigan  
Ann Arbor, Michigan
- 2017 Genetic Insights into Human Erythropoiesis / Oral Presentation - Presenter  
2017 Red Cells Gordon Research Conference  
Newport, Rhode Island
- 2017 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
Charles Bronfman Institute for Personalized Medicine Seminar Series  
The Icahn School of Medicine  
Mount Sinai, New York
- 2017 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
Genetic Studies of Human Hematopoiesis/ Invited Talk Department of Molecular Biology  
& Genetics  
Department of Molecular Biology & Genetics, Johns Hopkins University School of  
Medicine, Baltimore  
Baltimore, Maryland
- 2017 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
Human Oncology and Pathogenesis Program  
Memorial Sloan Kettering Cancer Center  
New York, New York
- 2017 Ribosome Levels Selectively Regulate Translation and Lineage Commitment in Human  
Hematopoiesis / Oral Presentation - Presenter  
Myeloid Workshop, American Society of Hematology Annual Meeting  
American Society of Hematology  
Atlanta, Georgia
- 2017 There Will Be Blood: Genetic Studies of Human Blood Production and Disease / Lecture  
- Presenter  
Society for Pediatric Research 2017 Presidential Plenary Session  
San Francisco, California
- 2017 The Regulation of Fetal Hemoglobin Expression / Oral Presentation - Presenter  
Towards Transformative Therapies for Sickle Cell Disease



New York Academy of Sciences  
New York, New York

- 2018 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
Vanderbilt University Department of Molecular Physiology & Biophysics  
Vanderbilt University  
Nashville, Tennessee
- 2018 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
University of Massachusetts Medical Center Seminar Series  
Worcester, Massachusetts
- 2018 Gale & Ira Drukier Prize Lecture / Demonstration - Presenter  
Weill Cornell Medicine  
Weill Cornell Medicine  
New York, New York
- 2018 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
University of Chicago Comer Children's Hospital Seminar Series  
University of Chicago Comer Children's Hospital  
Chicago, Illinois
- 2018 Insights into Aplastic Anemia Treatment from Human Genetic Studies / Oral  
Presentation - Presenter  
North American Pediatric Aplastic Anemia Consortium Meeting  
Pittsburgh, Pennsylvania
- 2018 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
The Feinstein Institute for Medical Research/ Donald and Barbara Zucker School of  
Medicine at Hofstra/Northwell  
Manhasset, New York
- 2018 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
Division of Regenerative Medicine Seminar Series, University of California San Diego  
San Diego, California
- 2019 Dissecting Cellular Hierarchies in Intact Humans / Oral Presentation - Presenter  
NHLBI Single-Cell Omics Workshop  
Bethesda, Maryland

International

- 2010 The molecular basis of elevated fetal hemoglobin in trisomy 13 / Oral Presentation  
17th Conference on Hemoglobin Switching  
Oxford, Oxford, United Kingdom
- 2011 Targeted therapeutic approaches for fetal hemoglobin induction / Oral Presentation  
(Selected Oral Abstract)  
53rd Annual Meeting of the American Society of Hematology  
San Diego, California
- 2012 The regulation of fetal hemoglobin: from human genetics to therapeutic targets / Lecture  
American Association of Blood Banks Annual Meeting  
Boston, Massachusetts
- 2012 The genetics of Diamond-Blackfan anemia and human erythropoiesis / Oral

- Presentation  
Diamond-Blackfan anemia symposium  
Lund University  
Lund, Sweden
- 2013 The regulation of fetal hemoglobin / Oral Presentation  
Hemoglobinopathies Workshop  
San Francisco, California
- 2013 New insight into the pathogenesis of Diamond-Blackfan anemia / Oral Presentation  
(Selected Oral Abstract)  
Red Cell Gordon Conference at Proctor Academy in Andover  
New Hampshire
- 2014 Human genetics of fetal hemoglobin regulation / Oral Presentation (Selected Oral  
Abstract)  
19th Conference on Hemoglobin Switching  
Oxford, United Kingdom
- 2014 The role of GATA1 in Diamond-Blackfan anemia / Oral Presentation  
Diamond Blackfan anemia International Consensus Conference
- 2014 Molecular pathophysiology of Diamond-Blackfan anemia / Oral Presentation  
European DBA International Conference: "Building Global Bridges"  
Freiburg, Germany
- 2015 Genetic Studies of Dyserythropoietic and Hypoplastic Anemia / Oral Presentation  
Brazilian Congress of Hematology  
Hemotherapy, and Cell Therapy  
Sao Paulo, Brazil
- 2015 Progress in Understanding Fetal Hemoglobin Regulation / Oral Presentation  
Brazilian Congress of Hematology  
Hemotherapy, and Cell Therapy  
Sao Paulo, Brazil
- 2015 Limiting ribosome availability impairs human hematopoiesis / Oral Presentation  
Red Cell Gordon Conference  
Holderness School in Holderness  
New Hampshire
- 2015 Modulation of Pathways that Control HbF Synthesis / Oral Presentation  
Tenth Cooley's Anemia Symposium  
Chicago, Illinois
- 2016 Hematopoiesis In Natura: Insight From Human Variation in Blood Production / Oral  
Presentation  
2nd International Symposium on Erythrocyte Biology  
Chengdu, China
- 2016 Genetic Studies of Human Hematopoiesis / Oral Presentation  
GeneForum 2016  
Tartu, Estonia

- 2016 The Regulation of Fetal Hemoglobin / Oral Presentation  
Thai Society of Hematology 5th International Symposium  
Bangkok, Thailand
- 2017 Genetic Studies of Human Erythropoiesis / Oral Presentation - Presenter  
22nd Congress - European Hematology Association  
Madrid, Spain
- 2017 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
Human Cell Atlas Meeting  
Weizmann Institute of Science  
Rehovot, Israel
- 2017 Functional Interrogation of Common Genetic Variation Uncovers Regulators of Human Hematopoiesis / Oral Presentation - Presenter  
The Genomics of Common Diseases Conference 2017  
Cambridge, United Kingdom
- 2018 Ribosome Levels Selectively Regulate Translation and Lineage Commitment in Human Hematopoiesis / Oral Presentation - Presenter  
Diamond-Blackfan Anemia International Consensus Conference  
Atlanta, Georgia
- 2018 Functional Interrogation of Common Genetic Variation Provides Insights into Human Hematopoiesis / Oral Presentation - Presenter  
NIDDK Workshop: Towards a Functional Understanding of the Diabetic Genome  
Washington, DC, Maryland
- 2018 Emerging Roles for Translational Regulation in Human Erythropoiesis / Oral Presentation - Presenter  
NIDDK Workshop: Beyond Transcriptomics  
Washington, DC, Maryland
- 2018 Genetic Studies of Human Hematopoiesis / Oral Presentation - Presenter  
Swisstransfusion Annual Congress 2018  
Bern, Switzerland
- 2018 Control of Hemoglobin Switching by LIN28B-Mediated Regulation of BCL11A Translation / Oral Presentation - Presenter  
21st Hemoglobin Switching Conference  
Oxford, United Kingdom
- 2018 Moving from Variant to Function: Lessons from Genetic Studies of Human Blood Production / Oral Presentation - Presenter  
Big Data Institute & Wellcome Trust Center for Human Genetics Seminar Series  
Oxford, United Kingdom
- 2018 Of Lineage & Legacy: Tracing Cellular Hierarchies in Intact Humans / Oral Presentation - Presenter  
Weatherall Institute of Molecular Medicine, Molecular Haematology Unit Away Day  
Keynote Lecture  
Oxford, United Kingdom
- 2019 Translational Regulation in Human Hematopoiesis / Oral Presentation - Presenter  
Gordon Research Conference - 2019 Translation Machinery in Health and Disease

Galveston, Texas

- 2019 Of Lineage & Legacy: Tracing Cellular Hierarchies in Intact Humans / Oral Presentation - Presenter  
American Heart Association, Vascular Discovery 2019 Scientific Sessions  
Boston, Massachusetts
- 2019 What Underlies the Inherited Risk for Acquiring Blood Cancers? / Oral Presentation - Presenter  
Gordon Research Conference on Human Genetics and Genomics  
Waterville Valley, New Hampshire
- 2019 A Congenital Hemolytic Anemia Dissociates the Pleiotropic Functions of a Master Transcription Factor / Oral Presentation - Presenter  
Gordon Research Conference on Red Cells  
Newport, Rhode Island
- 2019 What Underlies the Inherited Risk for Acquiring Blood Cancers? / Oral Presentation - Presenter  
Blood Disorders: Models, Mechanisms and Therapies (Disease Models & Mechanisms Conference)  
Boston, Massachusetts
- 2019 Variant to Function: Using a multi-omics approach to understand genetic disease mechanisms / Oral Presentation - Presenter  
Nature Genetics Webcast  
New York, New York
- 2020 Why Do Some People Get Blood Cancers? / Oral Presentation - Presenter  
New York Stem Cell Foundation Innovators Retreat Remote Conference  
New York City, New York (New York Stem Cell Foundation)
- 2020 From Variant to Function: Lessons from Studies of Blood Cell Production / Oral Presentation - Presenter  
European Society of Human Genetics 2020 Virtual Meeting  
Vienna, Austria
- 2020 Genetic Studies of Human Hematopoiesis: Toward Lineage Tracing in Intact Humans / Oral Presentation - Presenter  
International Society of Experimental Hematology 2020 Virtual Meeting  
Chicago, Illinois
- 2020 Inherited Risk for Myeloproliferative Neoplasms / Oral Presentation - Presenter  
MPN Research Foundation Roundtable Conference (Virtual)  
Chicago, Illinois
- 2020 Diamond-Blackfan Anemia, Ribosomes, and the Regulation of Human Hematopoiesis / Oral Presentation - Presenter  
Translational Research E-Conference on Bone Marrow Failure Disorders  
Paris, France (European School of Hematology)
- 2020 From Maps to Mechanisms to Medicines for Blood Disorders / Oral Presentation - Presenter  
International Common Disease Alliance Virtual Scientific Plenary  
Boston, Massachusetts

2020            Insights into Hematopoiesis by Integrating Single-Cell Genomics and Human Genetics /  
Oral Presentation - Presenter  
American Society of Hematology Annual Meeting  
San Diego, California

### **Report of Clinical Activities and Innovations**

#### **Current Licensure and Board Certification**

2010 - 2013      Massachusetts Limited Medical License

2013 -  
Present            Massachusetts Full Medical License

2014 -  
Present            Pediatric Advanced Life Support

2014 -  
Present            Board Certification in General Pediatrics  
American Board of Pediatrics

2016 -  
Present            DEA License

2017 -  
Present            Board Certification in Pediatric Hematology/Oncology  
American Board of Pediatrics

#### **Practice Activities**

2015 - Present	Attending Physician	Pediatric Hematology/Oncology, Dana-Farber/Boston Children's Cancer and Blood Disorders Center	4 weeks inpatient service per year, 3 outpatient clinics per month
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### **Report of Technological and Other Scientific Innovations**

Modulation of BCL11A for treatment of hemoglobinopathies (2009 - Present)      US Patent, PCT/US09/56770, Filed September 14, 2009  
Describes the targeting of BCL11A as a therapeutic strategy to induce fetal hemoglobin based on work done with Dr. Stuart Orkin (co-inventor).

Modulation of SH2B3 to Improve Red Blood Cell Production From Stem Cells and/or Progenitor Cells (2015 - Present)      Patent - Awarded  
Describes the suppression of SH2B3 as a strategy to improve red blood cell production ex vivo from a variety of human stem and progenitor cell sources.

### **Report of Education of Patients and Service to the Community**

#### **Activities**

No activities below were sponsored by 3<sup>rd</sup> parties/outside entities

2012            Cooley's Anemia Foundation Patient-Family Conference / Guest Speaker  
Spoke about fetal hemoglobin induction to patients and families with

- thalassemia
- 2012 Diamond-Blackfan Anemia Foundation 2012 Camp Sunshine / Guest Speaker  
Spoke to patients and families with Diamond-Blackfan anemia about genetic testing
- 2013 Diamond-Blackfan Anemia Foundation 2013 Camp Sunshine / Guest Speaker  
Spoke to patients and families with Diamond-Blackfan anemia about new research developments based upon recent genetic findings.
- 2015 Diamond-Blackfan Anemia Foundation 2015 Camp Sunshine / Guest Speaker  
Spoke to patients and families with Diamond-Blackfan anemia about new research developments relevant to this anemia.
- 2017 Science for All Seasons Lecture at the Broad Institute / Invited Lecture  
Spoke to a general lay audience about the history of studies on blood cell production and the basis for our ongoing studies in the laboratory

## **Report of Scholarship**

### **Peer-Reviewed Scholarship in print or other media**

#### **Research Investigations**

1. Ferguson KM, Kavran JM, **Sankaran VG**, Fournier E, Isakoff SJ, Skolnik EY, Lemmon MA. Structural basis for discrimination of 3-phosphoinositides by pleckstrin homology domains. *Molecular cell*. 2000 Aug 1; 6(2): 373-84. PubMed PMID: 10983984.
2. **Sankaran VG**, Klein DE, Sachdeva MM, Lemmon MA. High-affinity binding of a FYVE domain to phosphatidylinositol 3-phosphate requires intact phospholipid but not FYVE domain oligomerization. *Biochemistry*. 2001 Jul 24; 40(29): 8581-7. PubMed PMID: 11456498.
3. Uda M, Galanello R, Sanna S, Lettre G, **Sankaran VG**, Chen W, Usala G, Busonero F, Maschio A, Albai G, Piras MG, Sestu N, Lai S, Dei M, Mulas A, Crisponi L, Naitza S, Asunis I, Deiana M, Nagaraja R, Perseu L, Satta S, Cipollina MD, Sollaino C, Moi P, Hirschhorn J, Orkin SH, Abecasis GR, Schlessinger D, Cao A. Genome-wide association study shows BCL11A associated with persistent fetal hemoglobin and amelioration of the phenotype of beta-thalassemia. *Proceedings of the National Academy of Sciences of the United States of America*. 2008 Feb 5; 105(5): 1620-5. PubMed PMID: 18245381; PubMed Central PMCID: PMC2234194.
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5. Walkley CR, Qudsi R, **Sankaran VG**, Perry JA, Gostissa M, Roth SI, Rodda SJ, Snay E, Dunning P, Fahey FH, Alt F, McMahon AP, Orkin SH. Conditional mouse osteosarcoma, dependent on p53 loss and potentiated by loss of Rb, mimics the human disease. *Genes & development*. 2008 Jun 15; 22(12): 1662-76. PubMed PMID: 18559481; PubMed Central PMCID: PMC2428063.

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7. **Sankaran VG**, Menne TF, Xu J, Akie TE, Lettre G, Van Handel B, Mikkola HK, Hirschhorn J, Cantor A, Orkin SH. Human fetal hemoglobin expression is regulated by the developmental stage-specific repressor BCL11A. *Science (New York, N.Y.)*. 2008 Dec 19; 322(5909): 1839-42. PubMed PMID: 19056937.
8. **Sankaran VG**, Xu J, Ragoczy T, Ippolito GC, Walkley CR, Maika SD, Fujiwara Y, Ito M, Groudine M, Bender MA, Tucker PW, Orkin SH. Developmental and species-divergent globin switching are driven by BCL11A. *Nature*. 2009 Aug 27; 460(7259): 1093-7. PubMed PMID: 19657335; PubMed Central PMCID: PMC3749913.
9. Xu J, **Sankaran VG**, Ni M, Menne TF, Puram RV, Kim W, Orkin SH. Transcriptional silencing of {gamma}-globin by BCL11A involves long-range interactions and cooperation with SOX6. *Genes & development*. 2010 Apr 15; 24(8): 783-98. PubMed PMID: 20395365; PubMed Central PMCID: PMC2854393.
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## **Narrative Report**

In my training and independent career, I have focused on integrating rigorous studies of human genetic variation with functional studies of human hematopoiesis. As a result, we have made important advances in our understanding of blood cell production in human health and disease. For example, our work led to the identification of BCL11A as a key regulator of fetal hemoglobin expression - a finding that has now resulted in a number of ongoing clinical trials. We have identified a number of rare alleles that cause blood diseases and have gained fundamental insights into pathogenic mechanisms and potential therapeutic avenues as a result. For instance, we have identified a key role for altered translation of the hematopoietic master transcription factor GATA1 in the pathogenesis of Diamond-Blackfan anemia, inspired by rare variant discoveries we have made. We have also helped to define thousands of common variants associated with a number of blood cell traits and diseases in population-based studies. We have not only performed new variant discoveries, but performed individual and higher-throughput functional follow up to provide new knowledge about human hematopoiesis. In addition, our laboratory has pioneered the development of cutting-edge technologies to enable improved studies of human biology, including the detection of somatic mitochondrial DNA mutations using single cell genomic tools for lineage tracing in humans.

Over the coming years, I seek to pursue two overarching goals. First, I would like to further advance our work that lies at the interface of the fields of human genetics and hematopoiesis, which we term "human hematopoietic genetics." Second, a number of students and fellows from our group have gone on to successful independent careers in academia and industry and I seek to continue to train the next generation of leaders who can further expand the work on human hematopoietic genetics.

Area of Excellence: Investigation

Significant Supporting Activities:

Clinical Expertise

Special Merit in Education