

CURRICULUM VITAE

NAME: Quasar Saleem Padiath
BUSINESS ADDRESS: Department of Human Genetics
Graduate School of Public Health
University of Pittsburgh
130 DeSoto Street
3135 Parran Hall
Pittsburgh, PA 15261
Phone: 412-624-7203
Fax: 412-624-3020
Email: qpadiath@pitt.edu

EDUCATION AND TRAINING

Under-Graduate

1990-1995	Kilpauk Medical College, Dr. MGR Medical University, Madras, India	MBBS	Bachelor of Medicine and Surgery
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Graduate

1996-2001	Indian Institute of Science, Bangalore, India	PhD	Human Genetics
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Post-Graduate

2001-2002	National Center for Biological Sciences, Bangalore, India	Visiting Fellow	Molecular Genetics
2002-2003	Dept. of Genetics, Stanford University, CA.	Post-Doctoral Fellow	Molecular Genetics Prof. Uta Francke
2003-2007	Dept. of Neurology, University of California, San Francisco, CA.	Post-Doctoral Fellow	Molecular Genetics Prof. Ying-Hui Fu

APPOINTMENTS AND POSITIONS

Academic

2007-2010	Research Associate	Lab of Prof. Ying-Hui Fu, Dept. of Neurology, University of California, San Francisco, CA, USA
Dec., 2010 to June, 2011	Visiting Assistant Professor	Dept. of Human Genetics, University of Pittsburgh, Graduate School of Public Health. Pittsburgh, PA
June, 2011- Dec., 2017	Assistant Professor (tenure stream)	Dept. of Human Genetics, University of Pittsburgh, Graduate School of Public Health. Pittsburgh, PA
2013-Present	Affiliate member	Pittsburgh Institute of Neurodegenerative Diseases (PIND)
2015-Present	Member	Multiple Sclerosis Center, Dept. of Neurology, University of Pittsburgh
2015-Present	Graduate Faculty	Graduate School of Public Health, University of Pittsburgh
2015-Present	Member	Center for Neuroscience, University of Pittsburgh (CNUP)
2015-Present	Member	Medical Scientists Training Program (MSTP)
2017-2018	Assistant Professor (Secondary appointment)	Dept. of Neurobiology, University of Pittsburgh.
2018-Present	Associate Professor	Dept. of Human Genetics, University of Pittsburgh, Graduate School of Public Health.
2018-Present	Associate Professor (Secondary appointment)	Dept. of Neurobiology, University of Pittsburgh.

MEMBERSHIP IN PROFESSIONAL AND SCIENTIFIC SOCIETIES

2015 - to date	Society for Neuroscience
2005 - to date	American Society of Human Genetics

AWARDS AND HONORS

Year	Award
2022	Nominated for the Craig Award for Teaching Excellence, Graduate School of Public Health, University of Pittsburgh.

- 2017 Selected as Senior Vice Chancellor seminar series speaker, University of Pittsburgh
- 2016 Nominated for the Craig Award for Teaching Excellence, Graduate School of Public Health, University of Pittsburgh.
- 2012 Samuel and Emma Winters Foundation Research award.
- 2007 Larry L. Hillblom Foundation for Aging Research Fellowship.
- 2003 Dean's Post Doctoral Fellowship, Dept. of Genetics, Stanford University, CA.
- 2002 C V Hanumantha Rao Medal for the best thesis in the Faculty of Science, Indian Institute of Science, Bangalore, India.
- 2002 Associate of the Indian Academy of Sciences.
- 2001 Indian National Science Academy (INSA) medal for young scientists, one of the most prestigious national awards for young scientists in India, for graduate work.
- 2001 Selected for oral presentation at 9th World Congress on Psychiatric Genetics, St. Louis, MO.
- 2000 Selected for oral presentation at "An Interactive Meet on Genomics Research in the New Millennium: Emerging Concepts, Paradigms and Technologies held at the Centre for Biochemical Technology, Delhi, India.
- 1999 FAOBMB young scientist fellowship to attend the 14th annual FAOBMB meeting at Dunedin, New Zealand.
- 1990 Selected for the MBBS course through the All India Pre Medical/Pre Dental Examination (Only 1500 candidates are selected out of ~ 200,000 applicants).

PROFESSIONAL ACTIVITIES
TEACHING

Courses Taught

Years Taught	Course Number: Title	Hours of Lecture each semester, credits Average Enrollment	Role in course Primary/Coordinator
2022	HUGEN2032: Genetic Techniques	34 hours, 2 credits, 13 students	Course Director
2022	HUGEN2060: Chromosomes	25 hours, 2 credits, 33 students	Course Director
2021	MSNBIO 2101 / NROSCI 2101: Cell and Molecular Neurobiology	2 hours, 3 credits, 15 students	Instructor

2021	MED 5115 Genetics - Human Genetics	4.5 hours, 150 students	Instructor
2021	HUGEN 2040 : Molecular Basis of Human Inherited Disease	2 hours, 3 credits, 50 students	Instructor
2021	MSNBIO 2112 - 1010(24614)- Neurobiology of Disease	2 hours, 2 credits, 12 students	Instructor
2021	HUGEN2090: Biochemical and Molecular Genetics of Complex Diseases	4 hours, 3 credits, 52 students	Instructor
2021	HUGEN2032: Genetic Techniques	34 hours, 2 credits, 13 students	Course Director
2020	MSNBIO 2101 / NROSCI 2101: Cell and Molecular Neurobiology	2 hours, 3 credits, 15 students	Instructor
2020	HUGEN 2040 : Molecular Basis of Human Inherited Disease	2 hours, 3 credits, 50 students	Instructor
2020	MED 5115 Genetics - Human Genetics	4.5 hours, 150 students	Instructor
2020	HUGEN2034: Biochemical and Molecular Genetics of Complex Diseases	3 hours, 3 credits, 26 students	Instructor
2020	HUGEN2032: Genetic Techniques	34 hours, 2 credits, 12 students	Course Director
2019	HuGen 2011: Scientific Writing	1 credit, 2 students	Reviewer
2019	MED 5115 Genetics - Human Genetics	4.5 hours, 150 students	Instructor

2019	HUGEN2034: Biochemical and Molecular Genetics of Complex Diseases	3 hours, 3 credits, 26 students	Instructor
2019	HUGEN 2040 : Molecular Basis of Human Inherited Disease	2 hours, 3 credits, 15-20 students	Instructor
2019	HUGEN2032: Genetic Techniques	20 hours, 2 credits, 12 students	Course Director
2019	MSNBIO 2112 - 1010(24614)- Neurobiology of Disease	2 hours, 2 credits, 12 students	Instructor
2018	HUGEN 2040 : Molecular Basis of Human Inherited Disease	2 hours, 3 credits, 15-20 students	Instructor
2018	MED 5115 Genetics - Human Genetics	4.5 hours, 150 students	Instructor
2018	MSNBIO 2101 / NROSCI 2101: Cell and Molecular Neurobiology	2 hours, 3 credits, 17 students	Lecturer
2018	HUGEN2034: Biochemical and Molecular Genetics of Complex Diseases	3 hours, 3 credits, 26 students	Instructor
2018	HUGEN2032: Genetic Techniques	8 hours, 2 credits, 12 students	Course Director
2017	HUGEN 2040 : Molecular Basis of Human Inherited Disease	7.5 hours, 3 credits, 15-20 students	Course Co-Director
2017	MED 5115 Genetics - Human Genetics	4.5 hours, 150 students	Instructor
2017	MSNBIO 2101 / NROSCI 2101: Cell and Molecular Neurobiology	2 hours, 3 credits, 17 students	Lecturer

2017	HUGEN2034: Biochemical and Molecular Genetics of Complex Diseases	3 hours, 3 credits, 26 students	Instructor
2017	HUGEN2032: Genetic Techniques	8 hours, 2 credits, 12 students	Course Director
2017	MSNBIO 2112: Neurobiology of Disease	2 hours, 2 credits, 12 students	Instructor
2016	MED 5115 Genetics - Human Genetics	3 hours, 150 students	Instructor
2016	CNUP Summer undergraduate program	1 hour, 12 students	Instructor
2016	MSNBIO 2112: Neurobiology of Disease	2 hours, 2 credits, 15 students	Instructor
2013-2016	HUGEN 2040 : Molecular Basis of Human Inherited Disease	4.5 hours, 3 credits, 15-20 students	Instructor
2011	INTBP 2290: Scientific Ethics	8 hours, 1 credit, 10 students	Moderator for Breakout Sessions

Major Advisor for Graduate Student Essays, Theses, and Dissertations

Name of Student	Degree Awarded, Year	Type of Document and Title
Virali Patel	BS- Neuroscience Honors Thesis, 2021	Characterization of Transgenic Mice Used to Study TUBB4A Mutations Causing Hypomyelination and Atrophy of the Basal Ganglia and Cerebellum
Michael Gosky	MS-GC, 2021	Assessment and Distribution of Online Educational Materials Regarding Autosomal Dominant Leukodystrophy
Bruce Nmezi	PhD, 2019	The Role of Lamin B1 in the Organization of the Nuclear Envelope and Myelin Regulation in Development and Disease
Talia Oranburg	PhD, Joined 2018	To be decided

Anushe Munir

Masters, 2020

Characterization of adult onset
Lamin B1 depletion in an inducible
knockout mouse model

Service on Masters or Doctoral Committees

Dates Served	Name of Student	Degree Awarded	Title of Dissertation/Essay
2021	Roberta Reis	PhD, IDM	HIV-1 Neuropathogenesis: Role of Neurogranin and its regulation by viral and host cellular factors
2020	Anushe Munir	Masters in Human Genetics	Characterization of adult onset Lamin B1 depletion in an inducible knockout mouse model
2020	Nandini Rmesh	PhD, Human Genetics	Investigating the roles of Matrin-3 in Amyotrophic Lateral Sclerosis and Distal Myopathy
2020	Lingling Chen	Masters in Human Genetics	Investigating Genetic Interactions Between Two Key Meiotic Genes <i>xnd-1</i> and <i>him-17</i> , in <i>C. elegans</i>
2019	Avani Ahuja	Masters in Human Genetics	Glioma Cancer Immunosuppressive Microenvironment
2018	Ann Piccirillo	PhD	A Role For Lysophosphatidyl choline Transporter <i>Mfsd2a</i> In Cd8+ T Cell Memory And Secondary Response To Infection
2017	Ruhee Jain	Master of Public Health	Understanding The Role Of Astrocytes In HIV-1 Neuropathogenesis: Novel Strategies To Target Astrocyte Function And Hiv-1 Associated Neurocognitive Disorder
2017	Martin Requena	MS, Human Genetics	A Cell Culture Model Of <i>Mrps2</i> -Related Cutis Laxa
2017	Ian Casci	PhD, Human Genetics	Identifying Novel Modifiers of FUS-Associated Toxicity in a <i>Drosophila</i>

2016	Marisa Adhikusuma	MS, Neuroscience	Model of Amyotrophic Lateral Sclerosis ERK 1/2 Activation in Pre-Existing Oligodendrocytes of Adult Mice Drives New Myelin Synthesis and Enhanced CNS Function
2016	Anne Piccirillo	MS, Human Genetics	The role of the lipid transporter MFSD2A in innate and adaptive effector T cells
2016	Briana Heath	PhD, Human Genetics	MT1 Receptor Mediated Neuroprotection in R6/2 Mouse Model of Huntington's Disease
2015	Priya Mittal	PhD, Human Genetics	The role of mediator complex subunit 12 (med12) in the murine reproductive tract
2014	Chi-Ting Su	PhD, Human Genetics	Molecular Mechanisms of LTBP4 related Cutis Laxa

Thesis examiner for International students

Dates Served	Name of Student	Degree Institution	Title of Dissertation/Essay
2017	Anshika Goenka	PhD Indian Institute of Technology, Kanpur, India	Role of human non-coding satellite-III transcripts in cellular stress response

Service on Comprehensive or Qualifying Examination Committees

Dates Served	Student Population	Type of Exam (Qualifying/Comprehensive)
2022	PhD (Shubnita Singh) Human Genetics	Comprehensive
2022	PhD (Jia Qi Zhang) Human Genetics	Comprehensive
2022	PhD (Wafaa Albalawy) Human Genetics	Qualifying
2022	PhD (Ally DePuyt) IDM	Qualifying
2022	Masters (Jocelyn Taddonio) IDM	Qualifying

2021	PhD (Shubnita Singh) Human Genetics	Qualifying
2021	PhD (Tyler Fortuna) (ISB graduate program)	Comprehensive
2021	PhD (Jia Qi Cheng Zhang) Human Genetics	Qualifying
2020	PhD (Maddy Svharz) IDM	Qualifying
2020	PhD (Talia Oranburg) Human Genetics	Qualifying
2020	PhD (Annie Arockiaraj) Human Genetics	Qualifying
2020	PhD (Tyler Fortuna) (ISB graduate program)	Qualifying
2020	PhD (Sydney Lamerand) CNUP program	Qualifying
2020	Masters (Anushe Munir) Human Genetics	Comprehensive
2020	Masters (Lingling Chen) Human Genetics	Comprehensive
2019	Masters (Avani Ahuja) Human Genetics	Comprehensive
2019	Masters (Hallie Goldstein) Human Genetics	Comprehensive
2018	PhD (Teresa Capasso) Human Genetics	Qualifying
2019	PhD (Nandini Ramesh) Human Genetics	Comprehensive
2017	PhD (Roberta Reis) IDM	Comprehensive
2017	PhD (Nandini Ramesh) Human Genetics	Qualifying
2017	PhD (Anne Piccirillo) Human Genetics	Comprehensive
2016	PhD (Nandini Ramesh) Human Genetics	Qualifying
2016	PhD (Bruce Nmezi) Human Genetics	Qualifying
2016	PhD (Nandini Ramesh) Human Genetics	Qualifying

2015	PhD (Ian Casci) Human Genetics	Comprehensive
2015	MS (Andrew Shinsheimer) Human Genetics	Comprehensive
2014	PhD (Priya Mittal) Human Genetics	Comprehensive
2014	PhD (Subhara Raveendran) Human Genetics	Comprehensive
2013	PhD (Brianna Heath) Human Genetics	Comprehensive
2012	PhD (Chi-Ting Su) Human Genetics	Qualifying

Supervision of Post-Doctoral Students, Residents, and Fellows

Dates Supervised	Name of Student	Position of Student
August 2020-Present	Guillermo Rodriguez Bey	Research Assistant Professor
Jan. 2021-Present	Shanshan Song	Instructor, Dept. of Neurology
Jan., 2015 – August, 2020	Guillermo Rodriguez Bey	Post-Doctoral fellow
August 2017 - 2019	Yuming Jia	Medical student (Tsinghua exchange student program)
August 2019 – November 2021	Bruce Nmezi	Post-Doctoral Fellow
August-2011 to Sept. 2014	Harshvardhan Rolyan	Post-Doctoral fellow

Mentoring of Graduate Students in Field Placements

Dates	Name of Student	Degree/Program Description	Field Site
Jan. 2022-August 2022	Sheha Joisha	Undergraduate	Lab rotation
Jan. 2021-Jan. 2022	Shruthi Subramaniam	Masters	Lab rotation
Jan., 2020-June, 2020	Sydney Lamerand	CNUP-Phd program	Lab rotation
October – December 2012	Andrew Shinsheimer	Masters	Lab rotation
October – December 2012	Ahmed Basudan	PhD	Lab Rotation

Other Teaching and Training

Dates	Teaching Activity	Program/Description
January, 2018- Present	Supervision of Lab work: Virali Patel	Summer intern
June 2018- August 2018	Supervision of Lab work: Lenia Holt	Summer intern – High School student mentorship program,
June 2018- August 2018	Supervision of Lab work: Heba Alsahlani	Summer intern – CNUP program
May -August 2017	Supervision of Lab work: Jaimin Patel	Summer intern
May -August 2016	Supervision of Lab work: Jessica Keller	CNUP Summer fellowship
August 2014- 2016	Supervision of Lab work: Kim Dinh	Lab rotation
August 2014- 2016	Supervision of Lab work: Nisha Nanavaty	Lab rotation
Jan – April 2014	Supervision of fellowship project: Chelsea King	Breckenridge fellowship
May– August 2013	Supervision of Lab work: Chelsea King	University of Pittsburgh – Summer Edge Program
June - November 2011	Supervision of Lab work: Laura Kropp	Masters in Public Health Genetics
June - November 2011	Supervision of Lab work: Radhika Joshi	Masters in Public Health Genetics

RESEARCH

Grants and Contracts Received

Current

Years Inclusive	Grant and/or Contract Number and Title	Source	Total Direct Costs	Role	% Effort
2021-2026	R01AR078872-01A1 -Therapeutic genetics and disease modeling in LAMA2-CMD	NIAMSD (NIH)	\$42,000	Co-I	5%

2020-2021	R01NS095884S1-Elucidating mechanisms involved in Lamin B1 mediated demyelination – Alzheimer’s Disease supplement	NINDS (NIH)	\$159,086	PI	Concurrent with parent RO1
2018 –2022 (NCE)	R21 NS104384-01A1-Exploring Antisense Oligonucleotides as a potential therapy for Autosomal Dominant Leukodystrophy	NINDS (NIH)	\$710,833	PI	15%
2018 –2022 (NCE)	R33NS106087- High-Content Screening for Modulators of Lamin B1 as a Therapeutic Target in Autosomal Dominant Leukodystrophy	NINDS (NIH)	\$750,000	Co-PI	15%
2016-2019	R01NS095884S1-Elucidating mechanisms involved in Lamin B1 mediated demyelination (Administrative Supplement)	NINDS (NIH)	\$121, 851	PI	Concurrent with parent RO1
2016-2021 (NCE)	R01NS095884 - Elucidating mechanisms involved in Lamin B1 mediated demyelination	NINDS (NIH)	\$1.14 million	PI	15%

Completed Years Inclusive	Grant and/or Contract Number and Title	Source	Total Direct Costs	Role	% Effort
2017 –2021	962820-RSUB- Frontiers in Leukodystrophy Initiative (FrontLINE)	Commonwealth of Pennsylvania (Children’s Hospital of Philadelphia-	\$181,365	Co-I	5%
2018-2020	ELA 2017-02112- Antisense oligonucleotide therapy for Autosomal Dominant Leukodystrophy	European Leukodystrophy Association (ELA)	\$221,988	PI	15% (Cost Shared)
2017-2018	Identification of potential drug therapies for Autosomal Dominant Leukodystrophy	United Leukodys-trophy Foundation	\$30,000	PI	5%

2014-2017	RG 5045A1/1; Studying the role of the nuclear lamina in age dependent demyelination	National Multiple Sclerosis Society	\$329,436	PI	25%
2015-2016	Contract-Ref No. 0050117 – Characterization of the mutation underlying UDP-10602	NHGRI (NIH)	\$24,000	PI	5% (Cost Shared)
2013-2016	NIH1R21AG046897-01; Studying the role of the nuclear lamina in age dependent demyelination.	NIA (NIH)	\$275,000	PI	25%
2012-2013	Clinical and Translational Science Institute (CTSI)-University of Pittsburgh, Characterization of a mouse model for Autosomal Dominant Leukodystrophy	CTSI, University of Pittsburgh	\$12,500	PI	5%
2012-2014	University of Pittsburgh, Central Research Development Fund (CRDF) - The transcription factor erect wing as a novel regulator of circadian rhythms	University of Pittsburgh	\$15,000	PI	5%
2012-2013	Clinical and Translational Science Institute (CTSI) - Generation of a zebrafish model for the demyelinating disease Autosomal Dominant Leukodystrophy (ADLD)	University of Pittsburgh	\$25,000	PI	10%
2012-2014	Characterization of the transcription factor Erect Wing as a novel regulator of circadian rhythms	Winters Foundation	\$9,000	PI	5%
2011-2012	Aging Institute Pilot Funding Program - Elucidating the role of Lamin B1 in aging dependent demyelination	University of Pittsburgh Institute for Aging	\$20,000	PI	10%
2007-2010	Fellowship Grant	Larry Hilblom Foundation	\$180,000	PI	100%

Invited Lectureships and Major Seminars Related to Your Research

Date	Title of Presentation	Venue
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7-1-2022	Insights into cell type specificity in ADLD: Disease mechanisms and treatment strategies – Keynote speaker	1 st ADLD Conference, University of Bologna
6-8-2022	Regulation of Lamin B1 expression in Autosomal Dominant Leukodystrophy	Intermediate Filament Gordon Research Conference, Mt. Snow, Vermont
10-19-2021	Autosomal Dominant Leukodystrophy (ADLD): An Overview	ADLD Center (Virtual webinar)
12-15-2020	Pathological Mechanisms in Autosomal Dominant Leukodystrophy	Kasturba Medical College, Manipal Academy of Higher Education, India
6-25-2020	Autosomal Dominant Leukodystrophy	United Leukodystrophy Foundation (ULF) annual meeting – Virtual webinar
5-21-2020	Pathological Mechanisms in Autosomal Dominant Leukodystrophy	Dept. of Neuropathology, School of Medicine, University of Pittsburgh
12-2-2019	Therapeutic approaches for autosomal dominant leukodystrophy	European Leukodystrophy Association (ELA), Luxembourg
10-17-2019	The spatial organization of the nuclear lamina	Science 2019, University of Pittsburgh
12-6-2018	The organization of the nuclear lamina and its role in disease	Conference on Genome Architecture and Cell Fate Regulation, University of Hyderabad, India
8-8-2018	The nuclear lamina and the regulation of cellular function, development and disease	Center for Cellular and Molecular Biology (CCMB), Hyderabad, India
7-31-2018	The nuclear lamina and the regulation of cellular function, development and disease	Indian Institute of Science Education and Research (IISER), Pune, India
10-26-2018	TUBB4A mutations result in specific neuronal and oligodendrocyte defects that closely match clinically distinct phenotypes	HABC Meeting, Children’s Hospital of Philadelphia (CHOP), Philadelphia, USA
9-11-2018	Leukodystrophies: From the clinic to the lab (and back)	Neurogenetics case conference, Rangos Research Center, Children’s Hospital of Pittsburgh (CHP), Pittsburgh, USA
6-23-2018	Autosomal Dominant Leukodystrophy: From mechanisms to potential therapeutic approaches	United Leukodystrophy Foundation, Annual International Conference, Charlotte, NC.
6-19-2018	Generation of inducible HABC mouse models’	Children’s Hospital of Philadelphia, Dept. of Neurology, University Of Pennsylvania, Philadelphia, PA.
4-17-2018	A model for the spatial organization of the Nuclear Lamina	EMBO workshop on Nuclear Mechanogenomics, Singapore

3-15-2018	Genomic architecture influences the disease phenotype in Autosomal Dominant Leukodystrophy (ADLD)	Clinical Genomics Case Conference, Magee-Women's Research Institute, University of Pittsburgh, Pittsburgh, PA.
9-15-2017	The Nuclear Envelope and Regulation of Central Nervous System Function and Disease	Senior Vice Chancellor's lecture- University of Pittsburgh, Pittsburgh, PA
6-5-2017	The Role of the Nuclear Lamina in Disease and Development	Nemours Biomedical Research, Alfred I duPont Hospital for Children, Wilmington, DE
3-21-2017	Cell Specific Syndromes Caused by TUBB4A Mutations (Selected to be part of Presidential Symposium)	American Society for Neurochemistry Annual Conference, Little Rock, AK
11-18-2016	The Role of the Nuclear Lamina in Development and Disease	Department of Anatomy and Neurobiology, VCU, Richmond, VA.
10-17-2016	Glial mechanisms in Tubulin mediated Leukodystrophies	Children's Hospital of Philadelphia, Dept. of Neurology, University Of Pennsylvania, Philadelphia, PA.
10-11-2016	The role of the nuclear lamina in the CNS	Dept. of Neurobiology, University of Pittsburgh, Pittsburgh, PA
7-28-2016	Mechanisms linking Lamin B1 and Autosomal Dominant Leukodystrophy	United Leukodystrophy Foundation, Annual International Conference, Omaha, NE.
6-12-2016	Lamin B1 and ADLD – Linking nuclear structure and myelin regulation	Gordon research Conference – Intermediate Filaments – Stowe, VT.
11-24-2015	The role of the nuclear lamina disease and development	Magee-Women's Research Institute's Work-in-Progress (WIP) Conference & Research Seminar Series, University of Pittsburgh, Pittsburgh, PA.
10-15-2015	The role of the nuclear lamina disease and development	Dept. of Environmental and Occupational Health, University of Pittsburgh, Pittsburgh, PA.
8-4-2015	The role of the nuclear lamina disease and development	Human Genetics Seminar Series, Dept. of Human Genetics, University of Pittsburgh, Pittsburgh, PA.
11/19/2014	Understanding the role of the Nuclear Lamina in Myelin Regulation and Demyelination	Western PA Multiple Sclerosis Society, Pittsburgh, Pittsburgh, PA.

10/1/2014	Glial mechanism in TUBB4A mediated hypomyelination	Children's National Hospital, Washington DC.
5/1/2014	Modeling Human Circadian variants in the fruit fly, <i>Drosophila melanogaster</i>	Multidisciplinary Sleep Conference, Division of Pulmonary, Allergy and Critical Care Medicine, University of Pittsburgh, Pittsburgh, PA.
4/4/2014	The Role of the 5q23 duplications in Autosomal Dominant Leukodystrophy	Microarray conference, Magee Women's Hospital, Pittsburgh, PA.
9/11/2013	The role of the nuclear lamina in development, aging and disease	Pittsburgh Institute of Neurodegenerative diseases, Dept. of Neurology, University of Pittsburgh, Pittsburgh, PA.
4-16-2012	The Role of the Nuclear Lamina in Development and Disease	Dept. of Infectious diseases and Microbiology, University of Pittsburgh, Pittsburgh, PA.
12-6-2011	Generation of a mouse model for Autosomal dominant leukodystrophy	Clinical and Translational Science Institute- University of Pittsburgh, Pittsburgh, PA.
7-12-2011	Elucidating the role of Lamin B1 in Autosomal Dominant Leukodystrophy	United Leukodystrophy Foundation, DeKalb, Il.
06-10-2011	The Role of the Nuclear Lamina in Demyelination, Disease and Aging	Basic Biology Of Aging Meeting, Dept. of Microbiology and Molecular Genetics, University of Pittsburgh, Pittsburgh, PA
03-18-2011	The Role of the Nuclear Lamina in Demyelination, Disease and Aging	Human Genetics Seminar Series, Dept. of Human Genetics, University of Pittsburgh, Pittsburgh, PA.
02-10-2011	The genetics of Adult onset demyelinating disorders	Dept. of Neurology, University of Pittsburgh Medical Center, Pittsburgh, PA.
08-05-2010	The Role of the Nuclear Lamina in Demyelination, Disease and Aging	Center for DNA Fingerprinting and Diagnostics (CDFD), Hyderabad, India.
06-25-2009	The Role of the Nuclear Lamina in Demyelination, Disease and Aging	Dept. of Human Genetics, University of Pittsburgh, PA.

06-11-2009	The genetics of Autosomal Dominant Leukodystrophy	Center for Cellular and Molecular Biology, Hyderabad, India.
06-04-2009	The Nuclear Lamina and Demyelination	Institute for Genomics and Integrative Biology, Delhi, India.
05-28-2009	The genetics of Adult onset demyelinating disorders	Dept. of Molecular and Cellular Biology, Indian Institute of Science, Bangalore, India.
05-20-2009	The Role of the Nuclear Lamina in Demyelination, Disease and Aging	National Center For Biological Sciences, Bangalore, India.
09-14-2007	The genetics of Adult onset demyelinating disorders	University of Texas Southwestern Medical Center, TX.
4-17-2006	Functional Consequences of A <i>CK1δ</i> Mutation Causing Familial Advanced Sleep Phase Syndrome	Dept. of Psychiatry, National Institute of Mental Health and Neurosciences, Bangalore, India.
4-18-2006	The role of the nuclear lamina in disease development and aging	National Center for Biological Sciences, Bangalore, India.
10-10-2006	When too much is too little – Lamin B1 duplications in Autosomal Dominant Leukodystrophy	Dept. of Genetics, Stanford University, CA
06-02-2005	Functional Consequences of A <i>CK1δ</i> Mutation Causing Familial Advanced Sleep Phase Syndrome	Fishman Symposium, Dept. of Neurology, University of California, San Francisco, USA.
5-13-2002	Trinucleotide repeats in Neurodegenerative and Neuropsychiatric disorders	Division of Biological Sciences, Tata Institute of Fundamental Research, Mumbai, India.
6-15-2002	Trinucleotide repeats in Neurodegenerative and Neuropsychiatric disorders	National Center for Biological Sciences, Bangalore, India.
2-24-2000	Chromosome 22 susceptibility loci for Bipolar Disorder and Schizophrenia	An Interactive Meet on Genomics Research in the New Millennium, Center for Biochemical Technology,

PUBLICATIONS

1. Refereed Articles

1. scMAPA: Identification of cell-type-specific alternative polyadenylation in complex tissues. Yulong Bai, Yidi Qin, Zhenjiang Fan, Robert M Morrison, KyongNyon Nam, Hassane Zarour, Radosveta Koldamova, **Quasar Saleem Padiath**, Soyeon Kim, Hyun Jung Park. *Gigascience*, 2022.
2. LMNB1 Duplication-Mediated Autosomal Dominant Adult-Onset Leukodystrophy in an Indian Family. Bijarnia-Mahay S, Roy G, **Padiath QS**, Saxena R, Verma IC. *Annals of Indian Academy of Neurology*, 2021, 24(3):413-416.
3. Deletion of conserved non-coding sequences downstream from NKX2-1: A novel disease-causing mechanism for benign hereditary chorea. Liao J, Coffman KA, Locker J, **Padiath QS**, Nmezi B, Filipink RA, Hu J, Sathanoori M, Madan-Khetarpal S, McGuire M, Schreiber A, Moran R, Friedman N, Hoffner L, Rajkovic A, Yatsenko SA, Surti U. *Mol Genet Genomic Med*. 2021 PMID: 33666368
4. Cardiac phenotype in *ATPIA3*-related syndromes: A multicenter cohort study. Balestrini S, Mikati MA, Álvarez-García-Rovés R, Carboni M, Hunanyan AS, Kherallah B, McLean M, Prange L, De Grandis E, Gagliardi A, Pisciotta L, Stagnaro M, Veneselli E, Campistol J, Fons C, Pias-Peleiteiro L, Brashear A, Miller C, Samões R, Brankovic V, **Padiath QS**, Potic A, Pilch J, Vezyroglou A, Bye AME, Davis AM, Ryan MM, Semsarian C, Hollingsworth G, Scheffer IE, Granata T, Nardocci N, Ragona F, Arzimanoglou A, Panagiotakaki E, Carrilho I, Zucca C, Novy J, Dzieżyc K, Parowicz M, Mazurkiewicz-Beldzińska M, Weckhuysen S, Pons R, Groppa S, Sinden DS, Pitt GS, Tinker A, Ashworth M, Michalak Z, Thom M, Cross JH, Vavassori R, Kaski JP, Sisodiya SM. *Neurology*. 2020 PMID: 32913013
5. TUBB4A mutations result in both glial and neuronal degeneration in an H-ABC leukodystrophy mouse model. Sase S, Almad AA, Boecker CA, Guedes-Dias P, Li JJ, Takanohashi A, Patel A, McCaffrey T, Patel H, Sirdeshpande D, Curiel J, Shih-Hwa Liu J, **Padiath Q**, Holzbaur EL, Scherer SS, Vanderver A. *Elife*. 2020 PMID: 32463361
6. Development and Optimization of a High-Content Analysis Platform to Identify Suppressors of Lamin B1 Overexpression as a Therapeutic Strategy for Autosomal Dominant Leukodystrophy. Nmezi B, Vollmer LL, Shun TY, Gough A, Rolyan H, Liu F, Jia Y, **Padiath QS**, Vogt A. *SLAS Discov*. 2020 Apr 30. PMID:32349647
7. Conditional depletion of Fus in oligodendrocytes leads to motor hyperactivity and increased myelin deposition associated with Akt and cholesterol activation. Guzman KM, Brink LE, Rodriguez-Bey G, Bodnar RJ, Kuang L, Xing B, Sullivan M, Park HJ, Koppes E, Zhu H, **Padiath Q**, Cambi F. *Glia*. 2020 Mar 18. PMID:32187401
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2. Books and Book Chapters

1. Raininko R, Gosky M, Padiath Q* LMNB1-Related Autosomal Dominant Leukodystrophy. 2021 (In press). In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2021.
2. Nahhas N, Sabet Rasekh P, Vanderver A, **Padiath Q***. Autosomal Dominant Leukodystrophy with Autonomic Disease. 2016 Jan 7. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016.

SERVICE (Professionally Related)

University/Institute of Higher Learning

Years	Committee	Position
2022	IDM Chair search committee	Member
2022	IDM Faculty recruitment committee	Member
2021- Present	Graduate School of Public Health - Faculty, Promotion and Tenure Committee (FAPTC)	Member
2015-Present	Admissions Committee - Dept. of Human Genetics	Member
2021	Social Justice Action Committee Work Group	Member
2021	Dean's day Poster Judging Committee	Organizer
2021	Dept. chair search committee, Dept. of Infectious Diseases	Member

2021	Faculty search committee, Dept. of Epidemiology	Member
2018- 2021	Office/facilities committee – Dept. of Human Genetics	Member
2017-2021	Human Genetics Spring seminar series and CC Li lecture	Organizer
2015-2021	Graduate School of Public Health, School - Planning and Budgeting Policies Committee (PBPC)	Member
2012-2018	Dean’s day Poster Judging Committee	Organizer
2014	Faculty search committee – Dept. of Human Genetics	Member
2013	Faculty search committee – Dept. of Human Genetics	Member
2012	Strategic planning committee– Dept. of Human Genetics	Member
2012	IDM Annual Research Day Poster Judging Committee	Member

Manuscript and Other Document/Publication Review

Dates	Journal Title
2022	Frontiers in Cell and Developmental Biology
2022	Cellular and Molecular Life Sciences
2021	Nature Materials
2021	Clinical Genetics
2020	Biochimie
2020	Cells
2020	Journal of Pediatric Genetics
2020	Cellular and Molecular Life Sciences
2019	Schizophrenia Bulletin
2019	Pediatric Neurology
2019	Neurochemical Research
2019	Nature Materials
2019	Journal of Neuroscience
2018	Orphanet Journal of Rare Diseases
2018	Pediatric Neurology
2017	Molecular Therapy
2017	Scientific Reports (Nature publications)
2017	BMC Pediatrics

2017	Neurogenetics
2017	Orphanet Journal of Rare Diseases
2016	Molecular Genetics and Metabolism Reports
2016	Genetics Home Reference (online content), National Library of Medicine, NIH
2015	Autonomic Neuroscience: Basic and Clinical
2015	Circulation-Cardiovascular Genetics
2015	British Journal of Dermatology
2015	Human Molecular Genetics
2015	Journal of the European Academy of Dermatology and Venereology
2014	Journal of Child Neurology
2013	Journal of Neurology
2012	PLoS One
2011	Molecular and Cellular Biochemistry
2011	Hemoglobin
2008	Journal of Genetics
2007	Journal of Neurogenetics
2001	Current Science

Editorial Boards

Dates	Journal Title
2021 to 2022	Frontiers in Cell and Developmental Biology (Review editor in Nuclear Organization and Dynamics)
2020 to Present	Cells
2020 to Present	Scientific Reports
2019 to Present	PLOS One

Study Sections, Review Panels, and Related Advisory Boards

Date	Position	Organization and Nature of Activity
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April, 2022	Ad Hoc Grant Reviewer	Reviewer: Competitive Medical Research Fund Grant, The University of Pittsburgh Office of Research, Health Sciences
April, 2021	Ad Hoc Grant Reviewer	Reviewer: Competitive Medical Research Fund Grant, The University of Pittsburgh Office of Research, Health Sciences
June, 2020	Ad Hoc Grant Reviewer	Cellular and Molecular Biology of Glia (CMBG) Study Section, NIH
April, 2020	Ad Hoc Grant Reviewer	Reviewer: Competitive Medical Research Fund Grant, The University of Pittsburgh Office of Research, Health Sciences
April, 2019	Ad Hoc Grant Reviewer	Reviewer: Competitive Medical Research Fund Grant, The University of Pittsburgh Office of Research, Health Sciences
Feb, 2019	Ad Hoc Grant Reviewer	Cellular and Molecular Biology of Glia (CMBG) Study Section, NIH
Feb, 2018	Ad Hoc Grant Reviewer	Cellular and Molecular Biology of Glia (CMBG) Study Section, NIH
June, 2016	Ad Hoc Grant Reviewer	Cellular and Molecular Biology of Glia (CMBG) Study Section, NIH
Feb, 2016	Ad Hoc Grant Reviewer	Cellular and Molecular Biology of Glia (CMBG) Study Section, NIH
2015	Ad Hoc Grant Reviewer	European Leukodystrophy Association (ELA)
2012	Ad Hoc Grant Reviewer	CTSI, University of Pittsburgh
2011	Ad Hoc Grant Reviewer	Western Psychiatric Institute and Clinic
2011	Ad Hoc Grant Reviewer	CTSI, University of Pittsburgh

Non-Governmental and Community-Based Organizations

Date	Organization	Nature of Activity
2020-Present	ADLD Center – Patient support group	Scientific advisory board member
2016-Present	Autosomal Dominant Leukodystrophy (ADLD) Patient support group	Helped initiate and organize this support group for ADLD patients
2014-Present	National Multiple Sclerosis Society, Western Pennsylvania Chapter	Education of patients and members of the lay public on research into demyelinating disorders.