Reham Khalaf-Nazzal, MD, PhD
Associate Professor of Neurosciences & Human Genetics
Head of Physiology and Pharmacology Department
Faculty of Medicine, Arab American University/Jenin
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REHAM KHALAF-NAZZAL, M.D. PhD

PROFESSIONAL SUMMARY

I lead the 'Stories of Hope, Stories from Palestine' genomics project, focusing on rare diseases in Palestinian communities. The project aims to improve diagnostics and clinical care by translating research findings into improved diagnostic provision and clinical care for affected communities.

My team compiles a database of founder gene mutations used in diagnostics, carrier testing, and newborn screening. This multidisciplinary approach advances scientific knowledge of hereditary and neurodevelopmental disorders, benefiting patients and families through innovative research and clinical management. By integrating research findings into educational curricula, I am to foster evidence-based practice, and critical thinking among medical students, while continuously refining my skills in pedagogical approaches to enhance student engagement and inspire future medical professionals.

EMPLOYMENT

ASSOCIATE PROFESSOR IN NEUROSCIENCES, HUMAN PHYSIOLOGY, AND MEDICAL GENETICS
Faculty of Medicine, Arab American University in Jenin
From May 2023 to present

HEAD OF PHYSIOLOGY AND PHARMACOLOGY DEPARTMENT
Faculty of Medicine, Arab American University in Jenin
From August 2020 to present

ASSISTANT PROFESSOR IN NEUROSCIENCES, HUMAN PHYSIOLOGY, AND MEDICAL GENETICS.
Faculty of Medicine, Arab American University in Jenin
From August 2020 to April 2023

ASSISTANT PROFESSOR IN NEUROSCIENCES, HUMAN PHYSIOLOGY, AND MEDICAL GENETICS.
Faculty of Dentistry, Arab American University in Jenin
From October 2017 to July 2020

ASSISTANT PROFESSOR IN NEUROSCIENCES, HUMAN PHYSIOLOGY, AND MEDICAL GENETICS.
Faculty of Medicine and Health Sciences, An-Najah National University, Nablus
From January 2013 to September 2017

POSTDOCTORAL RESEARCHER IN NEUROSCIENCES AND NEUROGENETICS
Fiona Francis laboratory, institute du Fer a Moulin, Paris, France
From October 2012 to December 2012

RESIDENT DOCTOR , SPECIALIZED ARAB HOSPITAL, NABLUS, MAKASSED ISLAMIC CHARITABLE HOSPITAL, EAST JERUSALEM, AND MARTYR KHALIL SULAIMAN PUBLIC HOSPITAL IN JENIN

Rotated through various medical specialties, including pediatrics, internal medicine, general surgery, OBGYN, ophthalmology, among others; to fulfill requirements for general practitioner training.

Provided comprehensive medical care under supervision, gaining practical experience in diverse clinical settings and patient populations.

Developed proficiency in diagnosing and managing a wide range of medical conditions, enhancing clinical skills and readiness for independent practice in outpatient and emergency settings

From August 2005 to August 2007

CLINICAL AND RESEARCH TEACHING ASSISTANT, AN-NAJAH NATIONAL UNIVERSITY MEDICAL SCHOOL

From September 2007 to September

EDUCATION

2009-2012

PH.D. IN GENETICS AND DEVELOPMENTAL NEUROSCIENCES. UNIVERSITÉ PIERRE ET MARIE CURIE (SORBONNE UNIVERSITY), FRANCE. MENTION THE HIGHEST HONOR.

Mention the highest honor.

My PhD work focused on understanding molecular and cellular mechanisms underlying cortical development and brain malformations. Using in vivo and in vitro techniques, I investigated the pathophysiological processes contributing to heterotopia, across cellular, molecular, and structural levels.

2008-2009

FOREIGN STUDENT IN MASTER 2 (DEROGATION DU STAGE, MASTER BIOLOGIE INTÉGRATIVE ET PHYSIOLOGIE BIP). UNIVERSITÉ PIERRE ET MARIE CURIE (SORBONNE UNIVERSITY), FRANCE.

1999-2005

MEDICAL DEGREE IN GENERAL MEDICINE & SURGERY, AL-QUDS UNIVERSITY, PALESTINE.

Mention very good; 81.5%.

1998-1999

HIGH SCHOOL (TAWJIHI), SCIENTIFIC BRANCH

Mention excellent (97.8%)

PUBLICATIONS

Khalaf-Nazzal R., Dweikat I., Ubeyratna N., Fasham J., Alawneh M., Leslie J., Maree M., Gunning A., Zayed D., Voutsina N., Mcgavin L., Sawafta R., Owens M., Baker W., Turnpenny P., Al-Hijawi F., Baple E., Crosby A., Rawlins L. *TECPR2*-related hereditary sensory and autonomic neuropathy in two siblings from Palestine.

Accepted on February 2024, DOI: 10.1002/ajmg.a.63579.

Fasham J, Huebner AK, Liebmann L, **Khalaf-Nazzal R**, Maroofian R, Kryeziu N, Wortmann SB, Leslie JS, Ubeyratna N, Mancini GMS, van Slegtenhorst M, Wilke M, Haack TB, Shamseldin H, Gleeson JG, Almuhaizea M, Dweikat I, Abu-Libdeh B, Daana M, Zaki MS, Wakeling MN, McGavin L, Turnpenny PD, Alkuraya FS, Houlden H, Schlattmann P, Kaila K, Crosby AH, Baple EL, Hübner CA. SLC4A10 mutation causes a neurological disorder associated with impaired GABAergic transmission. Brain. 2023 Jul 17:awad235. doi: 10.1093/brain/awad235. PMID: 37459438.

Dweikat I. & **Khalaf-Nazzal R**. Clinical heterogeneity of hyperornithinemia-hyperammonemia-homocitrullinuria syndrome in thirteen palestinian patients and report of a novel variant in the SLC25A15 gene. Front Genet. 2022 Nov 24;13:1004598. DOI: 10.3389/fgene.2022.1004598.

Khalaf-Nazzal R., Fasham, J., Inskeep, K. A., Blizzard, L. E., Leslie, J. S., Wakeling, M. N., Ubeyratna, N., Mitani, T., Griffith, J. L., Baker, W., Al-Hijawi, F., Keough, K. C., Gezdirici, A., Pena, L., Spaeth, C. G., Turnpenny, P. D., Walsh, J. R., Ray, R., Neilson, A., Kouranova, E., … Baple, E. L. (2022). Bi-allelic CAMSAP1 variants cause a clinically recognizable neuronal migration disorder. American journal of human genetics, 109(11), 2068–2079. https://doi.org/10.1016/j.ajhg.2022.09.012

Khalaf-Nazzal R, Dweikat I, Maree M, Alawneh M, Barahmeh M, Doulani RT, Qrareya M, Qadi M, Dudin A. Prevalent MLC1 mutation causing autosomal recessive megalencephalic leukoencephalopathy in consanguineous Palestinian families. Brain Dev. 2022 Aug;44(7):454-461. doi: 10.1016/j.braindev.2022.03.009. Epub 2022 Apr 16. PMID: 35440380.

Stouffer, M. A., **Khalaf-Nazzal, R.,** Cifuentes-Diaz, C., Albertini, G., Bandet, E., Grannec, G., Lavilla, V., Deleuze, J. F., Olaso, R., Nosten-Bertrand, M., & Francis, F. (2022). Doublecortin mutation leads to persistent defects in the Golgi apparatus and mitochondria in adult hippocampal pyramidal cells. Neurobiology of disease, 168, 105702. https://doi.org/10.1016/j.nbd.2022.105702

Lin, S., Fasham, J., Al-Hijawi, F., Qutob, N., Gunning, A., Leslie, J. S., McGavin, L., Ubeyratna, N., Baker, W., Zeid, R., Turnpenny, P. D., Crosby, A. H., Baple, E. L., & **Khalaf-Nazzal, R.** (2021). Consolidating biallelic SDHD variants as a cause of mitochondrial complex II deficiency. European journal of human genetics: EJHG, 29(10), 1570–1576. https://doi.org/10.1038/s41431-021-00887-w

Khalaf-Nazzal, R.; Fasham, J.; Ubeyratna, N.; Evans, D.J.; Leslie, J.S.; Warner, T.T.; Al-Hijawi, F.; Alshaer, S.; Baker, W.; Turnpenny, P.D.; Baple, E.L.; Crosby, A.H. Final Exon Frameshift Biallelic PTPN23 Variants Are Associated with Microcephalic Complex Hereditary Spastic Paraplegia. Brain Sci. 2021, 11, 614. https://doi.org/10.3390/brainsci11050614

Giménez-Mascarell P, Oyenarte I, Hardy S, Breiderhoff T, Stuiver M, Kostantin E, Diercks T, Pey AL, Ereño-Orbea J, Martínez-Chantar ML, **Khalaf-Nazzal R**, Claverie-Martin F, Müller D, Tremblay ML, Martínez-Cruz LA. Structural Basis of the Oncogenic Interaction of Phosphatase PRL-1 with the Magnesium Transporter CNNM2. J Biol Chem. 2017 ;292(3):786-801.

Khalaf-Nazzal R, Stouffer MA, Olaso R, Muresan L, Roumegous A, Lavilla V, Carpentier W, Moutkine I, Dumont S, Albaud B, Cagnard N, Roest Crollius H, Francis F. Early born neurons are abnormally positioned in the doublecortin knockout hippocampus. Hum Mol Genet. 2017;26(1):90-108.

Dweikat IM, Damsah N, **Khalaf R**. A distinct phenotype of childhood leukodystrophy presenting as absence seizure. J Pediatr Neurosci. 2014 Jan;9(1):63-5. doi: 10.4103/1817-1745.131492.

Hamelin S, Pouyatos B, **Khalaf-Nazzal R**, Chabrol T, Francis F, David O, Depaulis A. Long-term modifications of epileptogenesis and hippocampal rhythms after prolonged hyperthermic seizures in the mouse. Neurobiol Dis. 2014;69:156-68.

Khalaf-Nazzal R, Francis F. Hippocampal development - Old and new findings. Neuroscience. 2013;248C:225-242.

Khalaf-Nazzal R; Elodie Bruel-Jungerman; Jean-Paul Rio; Jocelyne Bureau; Theano Irinopoulou; Iffat Sumia; Audrey Roumegous; Elodie Martin; Robert Olaso; Carlos Parras; Carmen Cifuentes-Diaz; Fiona Francis Organelle and cellular abnormalities associated with hippocampal heterotopia in neonatal doublecortin knockout mice. PLoS One. 2013;8(9)

Bahi-Buisson, N., Souville, I., Fourniol, F., Toussaint, A., Moores, C., **Khalaf-Nazzal, R**, Hully, M, Poirier, K, Leger, PL., Elie, C, Boddaert, N, SBH-LIS European consortium, Chelly, J, Beldjord, C., Francis, F. New insights into genotype-phenotype correlations for the DCX-related lissencephaly spectrum. Brain (2013) 136 (1): 223-244.

PROFESSIONAL ACHIEVEMENTS

AWARDS AND TRAVEL GRANTS

Arab American University Award For Excellence In Scientific Research, 2023.

Awarded research publication: Clinical heterogeneity of hyperornithinemia-hyperammonemia-homocitrullinuria syndrome in thirteen palestinian patients and report of a novel variant in the *SLC25A15* gene

The Academy of Medical Sciences travel award/ Daniel Turnberg UK/Middle East Travel Fellowship.

Award theme: Studying Neurodevelopmental Disorders Among Palestinians.

Research prize for emerging research in developing countries, French Foundation for research in Epilepsy FFRE.

Project title 'Molecular causes of epilepsy syndromes among the Palestinian population-towards constructing a database of epilepsy genes and mutations for future development of genetic diagnosis services'

Zamala Program-Taawon travel grant to perform two-month training in Clinical and Laboratory Genetics at Exeter University Medical School, 2017.

Best Poster 3rd prize, Middle East Molecular Biology Conference, AbuDhabi, 2017

Research: Prevalent MLC1 founder mutation causing autosomal recessive Megalencephalic Leukoencephalopathy in consanguineous Palestinian families.

Combined Palestinian-French government scholarship to perform a PhD in developmental neuroscience and neurogenetics. 2008-2012

Two grants to attend residential training course in Pediatric Rheumatology and Neonatal Intensive Care, Gaslini International, Italy. September 2006.

CISEPO International Child Elective on Nutritional Health travel grant, University of Toronto, Canada, July 18th- August 12th 2005

Palestinian government grant, 6 weeks training course at Sheffield Kidney Institute, Northern General Hospital, University of Sheffield, UK. August-September/2004

RESEARCH GRANTS

Unity and Diversity in Nature and Society, Palestine Institute for Biodiversity and Sustainability fund, December 2020, 4600 USDs, project title 'The Design, Validation and Community-based Application of a Custom-Designed Targeted Next-Generation Sequencing Panel for the Screening of Rare Hereditary Diseases in Palestinian Communities.'

Zamala Program-Taawon professional hosting program, establishing first rare diseases Consortium in Palestine, April 2019. 10000 USDs.

Molecular causes of ultra-rare syndromic retinal degeneration, obesity, and diabetes mellitus in consanguineous Palestinian families, AAUP research grant of 7000 JOD, 2018

French-Palestinian project, Al-Maqdisi programme 2012 'Cortical development and genetic causes of brain malformations among Palestinian families'. 19000 euros

Cortical development and molecular causes of brain malformations among Palestinia, population. 5000 JD funded by An-Najah National University, 2012.

SKILLS

ACADEMIC SKILLS:

Course Design and Development:

Directed the development of several courses for Medicine students including Neuroscience course module (3rd Year), Biology and Cell Biology:, General Human Physiology, Molecular Biology, and Medical Genetics.

Active Learning Strategies:

Problem-Based Learning (PBL) to facilitate student-centered learning by presenting real-world clinical scenarios. Encourage students to analyze, discuss, and solve problems collaboratively. Encourage students to explore diverse perspectives

Technology Integration and E-Learning Platforms:

Utilize online platforms for course materials, quizzes, and interactive modules. Promote self-directed learning.

Assessment Strategies:

Including formative assessment with regular quizzes, case discussions, and self-assessment. Monitor student progress and identify areas for improvement, and summative assessment delivering fair and comprehensive exams to evaluate overall knowledge and

clinical competences, in line with the relevant course learning outcomes, and Bachelor degree in General Medicine and Surgery program learning and outcome.

Objective Structured Clinical Examinations (OSCEs):

Assess clinical skills in a standardized manner.

RESEARCH ETHICS:

Demonstrated ability to lead and guide committee members in ethical decision-making processes. Experience in facilitating discussions, reaching consensus, and making informed judgments. In-depth understanding of ethical principles, regulations, and guidelines. Ability to apply ethical frameworks to diverse research scenarios. Thoroughly review study applications, protocols, and related documents.

Identify potential ethical concerns and propose solutions. Promote fairness and equal treatment regardless of gender, race, disability, or other factors. Uphold diversity and inclusion principles.

RESEARCH SUPERVISION SKILLS:

Demonstrated ability to lead and mentor students in conducting clinically relevant research projects, contributing to the advancement of knowledge in medical genetics and neurodevelopmental disorders. With special expertise in project design, methodological expertise, clinical integration, interdisciplinary collaboration, student mentorship, and effective communication.

CLINICAL AND LABORATORY GENETICS SKILLS

Clinical assessment and dysmorphology. Pedigree analysis and determination of mode of inheritance. Determining sequencing strategy considering factors like cost, coverage, and clinical relevance. Analyzing Next Generation Sequencing (NGS) data: Identifying genetic variants, assessing their pathogenicity, and linking them to clinical phenotypes.

Laboratory techniques necessary for segregation and analysis. Functional interpretation of genomic variants Understanding the impact of genetic variants on protein function. Genomic results reporting and family counseling. Patient registry and database management. Coordination with multidisciplinary team members.

CELLULAR AND MOLECULAR BIOLOGY RESEARCH SKILLS:

Laboratory techniques: cell culture. Nucleic acids techniques. In utero electroporation. Data analysis. Gene expression analysis and transcrptomics. Research design and planning. Scientific writing and communication. Laboratory safety and best laboratory practices

CONTINUOUS EDUCATION COURSE DESIGN:

Co-developed the ‘Fundamentals of Genomic Data Interpretation in Clinical Genomics’ continuing medical education course in collaboration with colleagues from University of Exeter Medical School

CONTINUOUS EDUCATION COURSES

TEACHING THE TEACHERS FOR HEALTH SCIENCES EDUCATORS, INTERNATIONAL MEDICAL EDUCATION TRUST 2000 PALESTINE

Dates 11/2023-12/2023

OSCE TRAINING CONTINUING PROFESSIONAL DEVELOPMENT DEPARTMENT, LEEDS INSTITUTE OF MEDICAL EDUCATION

Dates 07/2012- 10/2021

LEENA PELTON SCHOOL OF HUMAN GENOMICS, WELLCOME TRUST SCIENTIFIC CONFERENCES, CAMBRIDGE

Dates 19-23/08/2012

HUMAN GENETICS COURSE, UPMC, PARIS

Dates 13/03/2012, 27/03/2012, and 03/04/2012

ANIMAL EXPERIMENTATION COURSE, LEVEL 1, UPMC, PARIS

Dates 24/01/2012-08/02/2012

NATIONAL & INTERNATIONAL RELATIONS

Developer, collaboration between Arab American University Health Sciences and Paris-Sorbonne University, Paris, France

Developer, collaboration between Arab American University Health Sciences and Exeter Medical School, Exeter, UK.

Collaborator, GREGoR consortium for genomic research in rare diseases

Arab American University representative member, Islamic World Educational, Scientific and Cultural Organization (ISESCO) health consortium

Arab American University representative member, Scientists for Palestine S4P

Member of 'Undiagnosed Diseases Network International' UDNI network, March 2023- present

Member, ClinGen Gene Curation Panel

Advisory board member, Medical & Health Sciences Division- Palestine Academy for Science & Technology MHSD, December 2022- present

COMMUNITY SERVICE

Member of Alstrom Global patient support group, and liaison point between Alstrom patients in Palestine, and Alstrom Global and UK support groups.

Developer: Community-based genetics diagnosis services in Palestine, towards establishing a nation-wide genetics consortium. In this project, we perform field visits to communities affected with rare diseases, providing free genetic testing for patients and families in need, and educate communities about the identified pathogenic founder variants.

Member, Board of Directors, Korea Jenin Youth Center, Jenin Municipality

LANGUAGES

ARABIC

Native language

ENGLISH

Excellent reading, writing, and conversation

FRENCH

Good reading and conversation, Fair in writing

REFERENCES

PROFESSOR PETER TURNPENNY

Consultant Clinical Geneticist & Clinical Professor, University of Exeter Medical School
Clinical Genetics | Royal Devon University Healthcare NHS Foundation Trust | Exeter EX1 2ED | UK
Secretary, UEMS (Union Europeenne des Medecins Specialistes)—Section of Medical Genetics
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DR. IMAD DWEIKAT

Associate Professor of Pediatrics & Metabolic Diseases
Head of Metabolic Laboratory
Consultant Metabolic Specialist, and Pediatrician
Arab American University - Palestine
Tel: +970 (4) 2418888 | email: imad.dweikat@aaup.edu

PROFESSOR FIONA FRANCIS

DR1 CNRS (PhD, HDR)
Director, Institut du Fer à Moulin INSERM UMR-S 1270
Sorbonne University. F-75005 Paris, France
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PROFESSOR HAYNES R. MILLER

Professor Emeritus of Mathematics
Department of Mathematics. Massachusetts Institute of Technology. Cambridge, MA 02139
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