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Mehawej, PhD

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CURRICULUM VITAE

I. PERSONAL EXPERIENCE

PERSONAL BACKGROUND

Title: Assistant Professor of Genetics

Institution: Gilbert and Rose-Marie Chagoury school of medicine; Lebanese American University, Byblos, Lebanon

Address: Byblos, Lebanon.

LANGUAGES

English (Reading: Excellent /Writing: Excellent /Verbal: Excellent)

Arabic (Reading: Excellent /Writing: Excellent /Verbal: Excellent)

French (Reading: Excellent /Writing: Excellent /Verbal: Excellent)

WORK EXPERIENCE AND ACADEMIC POSITIONS

September 2020 - Present
Byblos, Lebanon

Assistant Professor
Department of Human Genetics
Lebanese American University, Gilbert and Rose-Marie Chagoury school of medicine

September 2017 – August 2020
Beirut, Lebanon

Assistant Professor
Department of Human Genetics
Saint-Joseph University, School of Medicine

EDUCATIONAL BACKGROUND

March 2015 – May 2017
Boston, USA

Postdoctoral Fellowship in Immunogenetics
Harvard Medical School, Boston Children's Hospital
Mentor: Pr Raif Geha

2010 - 2013
Paris, France
Beirut, Lebanon

Doctor of Philosophy, PhD- Genetics
Very Honorable
International Dual Degree PhD (Cotutelle)
Saint-Joseph University and Paris Descartes University
Mentors: Pr Valérie Cormier-Daire
Pr Andre Megarbane

2008-2010
Beirut, Lebanon

Research Master's degree – Genomics and Health
Very Honorable
Lebanese University, Doctoral School of *Sciences and technologies*

2005-2008
Beirut, Lebanon

Bachelor degree – Biochemistry
Very Honorable
Lebanese University, Faculty of Sciences 2

1991-2005
Beirut, Lebanon

Lebanese Baccalaureate Certificate of Secondary Education- Life sciences
With Honors (Very Good)
Collège des Soeurs du Rosaire – Mountazah, Lebanon

ADMINISTRATIVE APPOINTMENTS

July 2017- August 2020
Beirut, Lebanon

Head of functional genetics unit
Department of Human Genetics
Saint-Joseph University, School of Medicine

TRAINING AND CERTIFICATES

December 2023
Lebanon

Public Speaking Course
Course organized by the CME/CPD office
Gilbert and Rose-Marie Chagoury school of medicine
Lebanese American University

October 2020
Online

Collaborative Institutional Training Initiative (CITI) training for biomedical researchers

December 2018
Belgium

Observership
Institute of Pathology And Genetics (IPG)

Topic: Next Generation Sequencing techniques

March 2015 – June 2017
Boston - USA

Post-doctoral Research Fellowship

Boston Children's Hospital, Harvard Meical School
Mentor: Pr Raif Geha
Topic: Immunogenetics

January 2014 – February 2015
Beirut, Lebanon

Post-doctoral Research Fellowship
Department of Human Genetics
Saint-Joseph University, School of Medicine
Mentor: Pr Andre Megarbane
Topic: Gene identification using Whole Exome Sequencing.

September 2010 – December 2013
Paris, France

PhD Fellowship
Research Unit on molecular and physiopathological bases of osteochondrodysplasia at INSERM Unit UMR1163,
Necker–Enfants Malades Hospital,
Mentor: Pr Valérie Cormier-Daire
Topic: Identification of genes involved in Skeletal Dysplasia.

February 2013 – December 2013
Orsay, France

Training
Genetics and Microbiology institute, CNRS UMR 8621, Orsay, France
Supervisor: Pr Agnes Delahodde
Topic: Functional studies in yeast

January 2012 – February 2012
Marseille, France.

Training
INSERM, UMR_S 910, Aix Marseille University, GMGF, 13385, Marseille, France.
Supervisor: Pr Valérie Delague
Topic: Whole Exome Sequencing analysis

January 2010 – December 2013
Beirut, Lebanon

Research Fellowship (Masters and PhD Studies)
Department of Human Genetics
Saint-Joseph University, School of Medicine
Mentor: Pr Andre Megarbane
Topic: Localization and Identification of genes involved in Mendelian diseases.

HONORS AND AWARDS

September 2023
Lebanon

The L'Oréal UNESCO For Women In Science Levant Young Talents award, provided by L'Oréal Foundation, L'Oreal Lebanon Management Committee, and the National Council for Scientific Research Lebanon.

August 2023
Washington, USA

The **American Society of Human Genetics' (ASHG's) Resource Limited Country Award 2023.**

December 2012
Paris, France

The scientific excellence award 2012, award given by the **Franco-Lebanese Medical Association**, Paris, France.

FELLOWSHIPS AND TRAVEL GRANTS

Date	Fellowship or travel award	Source
May 2022	Travel award to attend the Molecular Biology of Hearing and Deafness Meeting 2022, Iowa, USA	The University of Iowa, USA
October 2020	Acceptance as observer at the Clinical Immunology Society (CIS) diagnostic school 2020, Online	Clinical Immunology Society, USA
July 2019	Fellowship to attend the Clinical Immunology Society (CIS) diagnostic school 2019, Boston, USA	Clinical Immunology Society, USA
June 2019	Fellowship to attend the "Recent Advances in Rare Disease" (RARD) meeting 2019, Bogota, Colombia	Centogene, Germany.
November 2018	Fellowship to attend the Primary Immunodeficicencies Course, Reggio Calabria, Italy	Course organized by the Universal Scientific Education and Research Network (USERN) and the European School of Genetic Medicine

June 2013	Scholarship for young researchers – lecturers of Saint Joseph University	"Institut Français", Lebanese office
2012- 2013	PhD Travel scholarship (6 months)	« Agence Universitaire de la Francophonie", Middle East,
2011 –2012	PhD Travel scholarship (6 months)	« Agence Universitaire de la Francophonie", Middle East.

II. RESEARCH

ORCID ID: 0000-0002-7209-9359

Peer Reviewed Journal Publications

Items 1-44

1. Genetic Polymorphisms Involved in Bladder Cancer: A Global Review.

Kourie HR, Zouein J, Succar B, Mardirossian A, Ahmadieh N, Chouery E, **Mehawej C**, Jalkh N, Kattan J, Nemr E.

Oncol Rev. 2023 Nov 6;17:10603. doi: 10.3389/or.2023.10603. eCollection 2023.

PMID: 38025894 **Free PMC article.**

2. A homozygous frameshift variant expands the clinical spectrum of SAMD9 gene defects.

Mehawej C, Ibrahim M, Khalife L, Chouery E, El Hachem S, Sayad A, El Traboulsi A, Inati A, Megarbane A.

Clin Genet. 2023 Oct 13. doi: 10.1111/cge.14439. Online ahead of print.

PMID: 37830462

3. Analysis of G-quadruplex forming sequences in podocytes-marker genes and their potential roles in inherited glomerular diseases.

Saad M, **Mehawej C**, Faour WH.

Heliyon. 2023 Sep 15;9(9):e20233. doi: 10.1016/j.heliyon.2023.e20233. eCollection 2023 Sep.

PMID: 37809648 **Free PMC article.**

4. G-quadruplex forming sequences in the genes coding for cytochrome P450 enzymes and their potential roles in drug metabolism.

Saad M, Zhang R, Cucchiarini A, **Mehawej C**, Mergny JL, Mroueh M, Faour WH.

Biochimie. 2023 Sep 1:S0300-9084(23)00205-5. doi: 10.1016/j.biochi.2023.08.014. Online ahead of print.

PMID: 37660977

5. *CHAMP1*-Related Disorder: Sharing 20 Years of thorough Clinical Follow-Up and Review of the Literature.

Abi Raad S, Yazbeck Karam V, Chouery E, **Mehawej C,** Megarbane A.

Genes (Basel). 2023 Jul 28;14(8):1546. doi: 10.3390/genes14081546.

PMID: 37628598 **Free PMC article.** Review.

6. Report on a Case with Moreno-Nishimura-Schmidt Overgrowth Syndrome: A Clinically Delineated Disease Yet of an Unknown Origin!

Mehawej C, Chouery E, Al Hage Chehade G, Bejaoui Y, Mahfoud D, Gerges M, Delague V, El Hajj N, Megarbane A.

Mol Syndromol. 2023 Jun;14(3):219-224. doi: 10.1159/000527215. Epub 2023 Jan 17.

PMID: 37323196

7. Genetic profile of borderline ovarian tumors in the Lebanese population by whole-exome sequencing.

Feghaly I, Kourie H, Moubarak M, Chouery E, **Mehawej C,** Jalkh N, Atallah D.

Int J Gynaecol Obstet. 2023 Sep;162(3):1027-1032. doi: 10.1002/ijgo.14805. Epub 2023 Apr 25.

PMID: 37185951

8. POLD3 deficiency is associated with severe combined immunodeficiency, neurodevelopmental delay, and hearing impairment.

Mehawej C*, Chouery E, Azar-Atallah S, Shebaby W, Delague V, Mansour I, Mustapha M, Lefranc G, Megarbane A*.

*Co-corresponding authors.

Clin Immunol. 2023 Jun;251:109326. doi: 10.1016/j.clim.2023.109326. Epub 2023 Apr 6.

PMID: 37030525

This study received the American Society of Human Genetics award for resources-limited countries 2023.

9. Non-syndromic hypotrichosis: A report of two novel variants in the LSS gene.

El Hakim J, **Mehawej C,** Chouery E, Megarbane A, El-Feghaly J, El Khoury J.

Pediatr Dermatol. 2023 Apr 7. doi: 10.1111/pde.15320. Online ahead of print.

PMID: 37029088

10. *PCDH19* in Males: Are Hemizygous Variants Linked to Autism?

Chouery E, Makhlouf J, Daoud Khatoun W, **Mehawej C**, Megarbane A.

Genes (Basel). 2023 Feb 27;14(3):598. doi: 10.3390/genes14030598.

PMID: 36980870 **Free PMC article.**

11. Spondyloocular Syndrome: A Report of an Additional Family and Phenotypic Spectrum Delineation.

Chouery E, Karam R, Mrad YN, **Mehawej C**, Dib El Jalbout N, Bleik J, Mahfoud D, Megarbane A.

Genes (Basel). 2023 Feb 15;14(2):497. doi: 10.3390/genes14020497.

PMID: 36833424 **Free PMC article.** Review.

12. BHLHA9 homozygous duplication in a consanguineous family: A challenge for genetic counseling.

Chouery E, Tahan E, Karam R, Pharoun J, **Mehawej C**, Megarbane A.

Am J Med Genet A. 2023 Apr;191(4):923-929. doi: 10.1002/ajmg.a.63094. Epub 2022 Dec 24.

PMID: 36565049

13. The Middle East and North Africa Diagnosis and Management Guidelines for Inborn Errors of Immunity.

Baris S, Abolhassani H, Massaad MJ, Al-Nesf M, Chavoshzadeh Z, Keles S, Reisli I, Tahiat A, Shendi HM, Elaziz DA, Belaid B, Al Dhaheri F, Haskologlu S, Dogu F, Ben-Mustapha I, Sobh A, Galal N, Meshaal S, Elhawary R, El-Marsafy A, Alroqi FJ, Al-Saud B, Al-Ahmad M, Al Farsi T, Al Sukaiti N, Al-Tamemi S, **Mehawej C**, Dbaibo G, ElGhazali G, Kilic SS, Genel F, Kiykim A, Musabak U, Artac H, Guner SN, Boukari R, Djidjik R, Kechout N, Cagdas D, El-Sayed ZA, Karakoc-Aydiner E, Alzyoud R, Barbouche MR, Adeli M, Wakim RH, Reda SM, Ikinciogullari A, Ozen A, Bousfiha A, Al-Mousa H, Rezaei N, Al-Herz W, Geha RS.

J Allergy Clin Immunol Pract. 2023 Jan;11(1):158-180.e11. doi: 10.1016/j.jaip.2022.10.003. Epub 2022 Oct 17.

PMID: 36265766 Review.

14. Genetic susceptibility of bladder cancer in the Lebanese population.

Kourie HR, Succar B, Chouery E, **Mehawej C,** Ahmadieh N, Zouein J, Mardirossian A, Jalkh N, Sleilaty G, Kattan J, Nemr E.

BMC Med Genomics. 2022 Oct 17;15(1):217. doi: 10.1186/s12920-022-01372-z.

PMID: 36253817 **Free PMC article.** Review.

15. New complementary python codes to locate Single Nucleotide Polymorphisms (SNPs) and Overlapping G-Quadruplex Sequences (G4s).

Saad M, Shebaby M, **Mehawej C**, Faour W.

MethodsX. 2022 Oct 6;9:101875. doi: 10.1016/j.mex.2022.101875. eCollection 2022.

PMID: 36249933 **Free PMC article.**

16. NEK8-Associated Nephropathies: Do Autosomal Dominant Forms Exist?

Mehawej C, Chouery E, Ghabril R, Tokajian S, Megarbane A.

Nephron. 2023;147(3-4):229-233. doi: 10.1159/000526841. Epub 2022 Oct 10.

PMID: 36215968

This study was featured by the American Society of Nephrology (ASN) in ASN in the loop – May 2023.

17. Genetic predisposition to porto-sinusoidal vascular disorder: A functional genomic-based, multigenerational family study.

Shan J, Megarbane A, Chouchane A, Karthik D, Temanni R, Romero AR, Hua H, Pan C, Chen X, Subramanian M, Saad C, Mbarek H, **Mehawej C,** Chouery E, Abuaqel SW, Dömling A, Remadi S, Yaghi C, Li P, Chouchane L.

Hepatology. 2023 Feb 1;77(2):501-511. doi: 10.1002/hep.32735. Epub 2022 Sep 3.

PMID: 35989577 **Free PMC article.**

18. Identification by whole-exome sequencing of new single-nucleotide polymorphisms associated with molar-incisor hypomineralisation among the Lebanese population.

Elzein R, Abdel-Sater F, **Mehawej C**, Jalkh N, Ayoub F, Chouery E.

Eur Arch Paediatr Dent. 2022 Dec;23(6):919-928. doi: 10.1007/s40368-022-00738-2. Epub 2022 Aug 20.

PMID: 35986881

19. Early infantile epileptic encephalopathy related to NECAP1: Clinical delineation of the disease and review.

Chouery E, **Mehawej C**, Sabbagh S, Bleik J, Megarbane A.

Eur J Neurol. 2022 Aug;29(8):2486-2492. doi: 10.1111/ene.15424. Epub 2022 Jun 9.

PMID: 35638367

20. A novel homozygous variant in RNF170 causes hereditary spastic paraplegia: a case report and review of the literature.

Chouery E, **Mehawej C**, Megarbane A.

Neurogenetics. 2022 Apr;23(2):85-90. doi: 10.1007/s10048-022-00685-6. Epub 2022 Jan 18.

PMID: 35041108 Review.

21. A 20-year Clinical and Genetic Neuromuscular Cohort Analysis in Lebanon: An International Effort.

Megarbane A, Bizzari S, Deepthi A, Sabbagh S, Mansour H, Chouery E, Hmaimess G, Jabbour R, **Mehawej C**, Alame S, Hani A, Hasbini D, Ghanem I, Koussa S, Al-Ali MT, Obeid M, Talea DB, Lefranc G, Lévy N, Leturcq F, El Hayek S, Delague V, Urtizberea JA.

J Neuromuscul Dis. 2022;9(1):193-210. doi: 10.3233/JND-210652.

PMID: 34602496 **Free PMC article.**

22. Molecular pathogenesis of hereditary lung cancer: a literature review.

Rammal S, Kourie HR, Jalkh N, **Mehawej C**, Chouery E, Moujaess E, Dabar G.

Pharmacogenomics. 2021 Aug;22(12):791-803. doi: 10.2217/pgs-2020-0150. Epub 2021 Aug 19.

PMID: 34410147 Review.

23. Molecular profiling of basal cell carcinomas in young patients.

Abi Karam M, Kourie HR, Jalkh N, **Mehawej C**, Kesrouani C, Haddad FG, Feghaly I, Chouery E, Tomb R.

BMC Med Genomics. 2021 Jul 20;14(1):187. doi: 10.1186/s12920-021-01030-w.

PMID: 34284772 **Free PMC article.**

24. Consensus Middle East and North Africa Registry on Inborn Errors of Immunity.

Aghamohammadi A, Rezaei N, Yazdani R, Delavari S, Kutukculer N, Topyildiz E, Ozen A, Baris S, Karakoc-Aydiner E, Kilic SS, Kose H, Gulez N, Genel F, Reisli I, Djenouhat K, Tahiat A, Boukari R, Ladj S, Belbouab R, Ferhani Y, Belaid B, Djidjik R, Kechout N, Attal N, Saidani K, Barbouche R, Bousfiha A, Sobh A, Rizk R, Elnagdy MH, Al-Ahmed M, Al-Tamemi S, Nasrullayeva G, Adeli M, Al-Nesf M, Hassen A, **Mehawej C**, Irani C, Megarbane A, Quinn J; MENA-I. E. I. Study Group; Maródi L, Modell V, Modell F, Al-Herz W, Geha RS, Abolhassani H.

J Clin Immunol. 2021 Aug;41(6):1339-1351. doi: 10.1007/s10875-021-01053-z. Epub 2021 May 29.

PMID: 34052995 **Free PMC article.**

25. A family history of SCID and unrevealing WES: An approach to management and guidance of patients.

Mehawej C*, Khayat CD, Hamdan N, Chouery E, Platt CD.

*Corresponding author

Clin Immunol. 2020 Sep;218:108520. doi: 10.1016/j.clim.2020.108520. Epub 2020 Jul 4.

PMID: 32629161

26. ITK deficiency presenting as autoimmune lymphoproliferative syndrome.

Wallace JG, Alosaimi MF, Khayat CD, Jaber F, Almutairi A, Beaussant-Cohen S, Pinkus G, Fleming M, **Mehawej C**, Chou J, Geha RS.

J Allergy Clin Immunol. 2021 Feb;147(2):743-745.e1. doi: 10.1016/j.jaci.2020.06.019. Epub 2020 Jul 4.

PMID: 32628964 **Free PMC article.**

27. A homozygous stop gain mutation in BOD1 gene in a Lebanese patient with syndromic intellectual disability.

Hamdan N, **Mehawej C**[#], Sebaaly G, Jalkh N, Corbani S, Abou-Ghoch J, De Backer O, Chouery E.

[#] Equal contribution.

Clin Genet. 2020 Sep;98(3):288-292. doi: 10.1111/cge.13799.

PMID: 32578875

28. DNMT3B deficiency presenting as severe combined immune deficiency: A case report.

Mehawej C*, Khalife H, Hanna-Wakim R, Dbaibo G, Farra C.

*Corresponding author

Clin Immunol. 2020 Jun;215:108453. doi: 10.1016/j.clim.2020.108453. Epub 2020 Apr 30.

PMID: 32360517

29. Actionable Exomic Secondary Findings in 280 Lebanese Participants.

Jalkh N, **Mehawej C**, Chouery E.

Front Genet. 2020 Mar 13;11:208. doi: 10.3389/fgene.2020.00208. eCollection 2020.

PMID: 32231684 **Free PMC article.**

30. A Homozygous Splicing Mutation in PDE2A in a Family With Atypical Rett Syndrome.

Haidar Z, Jalkh N, Corbani S, Abou-Ghoch J, Fawaz A, **Mehawej C**[#], Chouery E.

[#] Equal contribution

Mov Disord. 2020 May;35(5):896-899. doi: 10.1002/mds.28023. Epub 2020 Mar 20.

PMID: 32196122 No abstract available.

31. Recessive marfanoid syndrome with herniation associated with a homozygous mutation in Fibulin-3.

Bizzari S, El-Bazzal L, Nair P, Younan A, Stora S, **Mehawej C**, El-Hayek S, Delague V, Mégarbané A.

Eur J Med Genet. 2020 May;63(5):103869. doi: 10.1016/j.ejmg.2020.103869. Epub 2020 Jan 30.

PMID: 32006683 Review.

32. The added value of WES reanalysis in the field of genetic diagnosis: lessons learned from 200 exomes in the Lebanese population.

Jalkh N, Corbani S, Haidar Z, Hamdan N, Farah E, Abou Ghoch J, Ghosn R, Salem N, Fawaz A, Djambas Khayat C, Rajab M, Mourani C, Moukarzel A, Rassi S, Gerbaka B, Mansour H, Baassiri M, Dagher R, Breich D, Mégarbané A, Desvignes JP, Delague V, **Mehawej C**, Chouery E.

BMC Med Genomics. 2019 Jan 21;12(1):11. doi: 10.1186/s12920-019-0474-y.

PMID: 30665423 **Free PMC article.**

33. First molecular study in Lebanese patients with Cockayne syndrome and report of a novel mutation in ERCC8 gene.

Chebly A, Corbani S, Abou Ghoch J, **Mehawej C,** Megarbane A, Chouery E.

BMC Med Genet. 2018 Sep 10;19(1):161. doi: 10.1186/s12881-018-0677-7.

PMID: 30200888 **Free PMC article.**

34. Homozygous mutation in ELMO2 may cause Ramon syndrome.

Mehawej C, Hoischen A, Farah RA, Marey I, David M, Stora S, Lachlan K, Brunner HG, Mégarbané A.

Clin Genet. 2018 Mar;93(3):703-706. doi: 10.1111/cge.13166. Epub 2018 Jan 25.

PMID: 29095483

35. "Fork and bracket" syndrome expands the spectrum of SBF1-related sensory motor polyneuropathies.

Romani M, **Mehawej C**, Mazza T, Mégarbané A, Valente EM.

Neurol Genet. 2016 Mar 3;2(2):e61. doi: 10.1212/NXG.0000000000000061. eCollection 2016 Apr.

PMID: 27123480 **Free PMC article.**

36. Neuroblastoma Amplified Sequence (NBAS) mutation in recurrent acute liver failure: Confirmatory report in a sibship with very early onset, osteoporosis and developmental delay.

Capo-Chichi JM, **Mehawej C,** Delague V, Caillaud C, Khneisser I, Hamdan FF, Michaud JL, Kibar Z, Mégarbané A.

Eur J Med Genet. 2015 Dec;58(12):637-41. doi: 10.1016/j.ejmg.2015.11.005. Epub 2015 Nov 11.

PMID: 26578240 Review.

37. Exome sequencing reveals a mutation in DMP1 in a family with familial sclerosing bone dysplasia.

Gannagé-Yared MH, Makrythanasis P, Chouery E, Sobacchi C, **Mehawej C**, Santoni FA, Guipponi M, Antonarakis SE, Hamamy H, Mégarbané A.

Bone. 2014 Nov;68:142-5. doi: 10.1016/j.bone.2014.08.014. Epub 2014 Aug 30.

PMID: 25180662

38. The impairment of MAGMAS function in human is responsible for a severe skeletal dysplasia.

Mehawej C, Delahodde A, Legeai-Mallet L, Delague V, Kaci N, Desvignes JP, Kibar Z, Capo-Chichi JM, Chouery E, Munnich A, Cormier-Daire V, Mégarbané A.

PLoS Genet. 2014 May 1;10(5):e1004311. doi: 10.1371/journal.pgen.1004311. eCollection 2014 May.

PMID: 24786642 **Free PMC article.**

This study was featured as a Research highlight by Nature Middle East. doi:10.1038/nmiddleeast.2014.121

39. A second family with autosomal recessive spondylometaphyseal dysplasia and early death.

Mégarbané A, **Mehawej C**, El Zahr A, Haddad S, Cormier-Daire V.

Am J Med Genet A. 2014 Apr;164A(4):1010-4. doi: 10.1002/ajmg.a.36372. Epub 2014 Jan 23.

PMID: 24458487

40. The identification of MAFB mutations in eight patients with multicentric carpo-tarsal osteolysis supports genetic homogeneity but clinical variability.

Mehawej C, Courcet JB, Baujat G, Mouy R, Gérard M, Landru I, Gosselin M, Koehrer P, Mousson C, Breton S, Quartier P, Le Merrer M, Faivre L, Cormier-Daire V.

Am J Med Genet A. 2013 Dec;161A(12):3023-9. doi: 10.1002/ajmg.a.36151. Epub 2013 Aug 16.

PMID: 23956186

41. Marfanoid habitus, inguinal hernia, advanced bone age, and distinctive facial features: a new collagenopathy?

Mégarbané A, Hanna N, Chouery E, Jalkh N, **Mehawej C**, Boileau C.

Am J Med Genet A. 2012 May;158A(5):1185-9. doi: 10.1002/ajmg.a.35279. Epub 2012 Apr 9.

PMID: 22489068

42. Identification of a novel causative mutation in the ROR2 gene in a Lebanese family with a mild form of recessive Robinow syndrome.

Mehawej C, Chouery E, Maalouf D, Baujat G, Le Merrer M, Cormier-Daire V, Mégarbané A.

Eur J Med Genet. 2012 Feb;55(2):103-8. doi: 10.1016/j.ejmg.2011.11.003. Epub 2011 Nov 27.

PMID: 22178368

43. Ambiguous genitalia, microcephaly, seizures, bone malformations, and early death: a distinct MCA/MR syndrome.

Mégarbané A, Chouery E, Mignon-Ravix C, El Sabbagh S, Corbani S, Ghoch JA, Jalkh N, **Mehawej C**, Lévy N, Villard L.

Am J Med Genet A. 2011 May;155A(5):1147-51. doi: 10.1002/ajmg.a.33938. Epub 2011 Apr 4.

PMID: 21465653

44. Molecular Study of Three Lebanese and Syrian Patients with Waardenburg Syndrome and Report of Novel Mutations in the EDNRB and MITF Genes.

Haddad NM, Ente D, Chouery E, Jalkh N, **Mehawej C**, Khoueir Z, Pingault V, Mégarbané A.

Mol Syndromol. 2011 Jan;1(4):169-175. doi: 10.1159/000322891. Epub 2011 Jan 10.

PMID: 21373256 **Free PMC article.**

Research Projects and grants

1- Principal investigator on the following peer-reviewed grants that received funding:

Year	Grant type, title and budget	Funding source
2023	Research grant Underlying new mechanisms of inborn errors of immunity 10000 euros	The L'Oréal UNESCO For Women In Science Levant Young Talents award
2022 – Present	Research Grant (2 consecutive years) Identification of the molecular bases of Inborn errors of immunity (IEI) `50 K USD	President's Intramural Research Fund (PIRF) grant, Lebanese American University
2018-2020	Research grant. Identification and Characterization of the molecular basis of Primary Immunodeficiencies in patients referred to the Medical Genetics Unit –USJ 29.8 K USD	Research Council of Saint-Joseph University
2017-2019	Validation of the molecular basis of a syndromic intellectual disability 17.9 K USD	Research Council of Saint-Joseph University

2- Co-PI on the following peer-reviewed research grant that received funding

Year	Grant type, title and budget	Funding source
2022 – Present	Research Grant	President's Intramural Research Fund (PIRF) grant, Lebanese American University

	Title: Identification of Genetic and Environmental Factors predispoing to Tinnitus in a cohort of Lebanese Patients (PI: Dr Eliane Chouery; Co-PI: Dr Cybel Mehawej) ~40 K USD	

Poster Presentation

1- **Mehawej C,** Chouery E, Ghabril R, Tokajian S, Megarbane A. NEK8-associated nephropathies: do autosomal dominant forms exist? June 2022. ESHG 2022, Vienna, Austria.

2- Chouery E, **Mehawej C,** Sabbagh S, Bleik J, Megarbane A. Early Infantile Epileptic Encephalopathy related to NECAP1; clinical delineation of the disease and review. June 2022. ESHG 2022, Vienna, Austria.

3- Hamdan N, De Backer O, Chouery E, **Mehawej C.** Uncovering BOD1 mutation effect and its correlation to syndromic intellectual disabilities. June 2022. ESHG 2022, Vienna, Austria.

4- **Mehawej C,** Chouery E, Megarbane A. Tailored WES data analysis and reanalysis in the Lebanese population and lessons learned. May 2022. MBHD meeting, Iowa city, USA.

5- Chouery E, Azaiez H, Mustapha M, **Mehawej C,** Megarbane A. Genetics of Age-Related Hearing Loss in the Lebanese Population: A First Step in this Fertile Landscape. May 2022. MBHD meeting, Iowa city, USA.

6- **Mehawej C.**, Khalife H., Hanna-Wakim R., Dbaibo G., Farra C. An atypical presentation of Immunodeficiency, Centromeric Instability, Facial anomalies syndrome. June 2020. ESHG, virtual conference (Interactive ePoster).

7- Haidar Z., Jalkh N., Corbani S, Abou-Ghoch J, Fawaz A, **Mehawej C**#, Chouery E#. A Homozygous Splicing Mutation in PDE2A in a Family With Atypical Rett Syndrome June 2020. ESHG, virtual conference (ePoster).

8- Irani C, Farah P, Daniel P, **Mehawej C.** The wolf in disguise: beyond humoral immunodeficiency. December 2019. World Allergy Conference, Lyon, France.

9- **Mehawej C.** A Challenging diagnosis of a Patient with Severe Combined Immunodeficiency. June 2019. RARD meeting. Bogota, Colombia.

10- El Kadddoum R, Eid R, Haddad F, Germanos M, **Mehawej C**, Raphael Kourie H, Kattan J. May 2019. The results of 3000 Fit: A prospective study led at Saint Joseph University of Beirut. ASCO Annual Meeting. Chicago, USA.

11- Cohen SB, Alosaimi M, Djambas Khayat C, **Mehawej C,** Chou J, Geha RS. October 2018. ITK deficiency presenting as Autoimmune Lymphoproliferative Syndrome. ESID conference. Lisbon, Portugal.

12- Mehawej C, Hoischen A, Farah RA, Marey I, David M, Stora S, Lachlan K, Brunner HG, Mégarbané A. January 2018. Homozygous Mutation in ELMO2 may cause Ramon syndrome. 9èmes Assises de Génétique. Nantes, France.

13- Mehawej C, Hoischen A, Farah RA, Marey I, David M, Stora S, Lachlan K, Brunner HG, Mégarbané A. January 2018. Homozygous Mutation in ELMO2 may cause Ramon syndrome. 7th Pan Arab Human Genetics Conference. Dubai.

14- Mehawej C, Massaad MJ, Kiykim A, Karakoc-Aydiner E, Baris S, Chou J, Geha RS. September 2016. IRAK1 mutation confers susceptibility to autoimmune manifestations. ESID conference 2016. Barcelona, Spain.

15- Gannagé-Yared M, Makrythanasis P, Chouery E, Sobacchi C, **Mehawej C**, Santoni F. A., Guipponi M, Antonarakis S. E, Hamamy H, Megarbane A. June 2015. Exome sequencing reveals a mutation in DMP1 in a family with sclerosing bone dysplasia. ESHG conference 2015. Glasgow, Scotland, UK.

16- Capo-Chichi JM, **Mehawej C**, Delague V, Caillaud C, Khneisser I, Hamdan FF, Michaud JL, Kibar Z, Megarbane A. June 2015. Exome sequencing reveals a mutation in Neuroblastoma Amplified Sequence gene in a family with early death, congenital fractures, dysmorphic features and hepatoc failure. ESHG conference 2015. Glasgow, Scotland, UK.

17- Mehawej C, Delahodde A, Legeai-Mallet L, Delague V, Kaci N, Desvignes JP, Kibar Z, Chouery E, Munnich A, Cormier-Daire V, Mégarbané A. June 2014. The impairment of MAGMAS function in human is responsible for a severe skeletal dysplasia. ESHG conference 2014. Milano, Italy.

18- Mehawej C, Courcet JB, Baujat G, Mouy R, Gérard M, Landru I, Gosselin M, Koehrer P, Mousson C, Breton S, Quartier P, Le Merrer M, Faivre L, Cormier-Daire V. June 2013. Multicentric carpo-tarsal osteolysis: clinical variability cannot be explained by genotype-phenotype correlations. ESHG conference 2013. Paris, France.

19- Mehawej C, Megarbane A, Cormier-Daire V. March 2013. « Identification de gènes impliqués dans des dysplasies osseuses dans des familles libanaises consanguines ». Doctoral forum of « Université Paris Descartes 2013 ».

Presentations and Invited Talks – International

1. **Mehawej C.** The molecular bases of Inborn Errors of Immunity: from bench to bedside. Division of Allergy and Immunology; **Boston Children's Hospital, Harvard Medical School**. Invited Talk scheduled for January 23rd 2024 (virtual event).

2. **Mehawej C.** The molecular bases of Inborn Errors of Immunity: from bench to bedside. Rosalind Franklin Rare Disease Grand Rounds; **NORD Center of Excellence for Rare Disorders at Boston Children's Hospital, Harvard Medical School**. Invited Talk scheduled for January 18th 2024 (virtual event).

3. **Mehawej C.** A Challenging diagnosis of a Patient with Severe Combined Immunodeficiency. July 2019. **Clinical Immunology Society (CIS) School. Boston, USA.**

4. **Mehawej C.** A Challenging diagnosis of a Patient with Severe Combined Immunodeficiency. June 2019. **Recent Advances in Rare Diseases (RARD) conference. Bogota, Colombia.**

5. **Mehawej C.** Approaches for novel gene discovery in genetic disorders. March 2019. Postgraduate seminars. **University of Cyprus. Nicosia, Cyprus**. (Invited talk)

6. **Mehawej C.** Identification de la base moléculaire d'un nouveau déficit immunitaire. August 2018. **Narilis Seminar. Namur, Belgium**. (Invited talk)

7. **Mehawej C,** Delahodde A, Legeai-Mallet L, Delague V, Kaci N, Desvignes JP, Kibar Z, Chouery E, Munnich A, Cormier-Daire V, Mégarbané A. November 2014. "The impairment of MAGMAS function in human is responsible for a severe skeletal dysplasia". **American Society of Human Genetics annual meeting. San Diego, California.**

8. **Mehawej C,** Delahodde A, Legeai-Mallet L, Delague V, Kaci N, Desvignes JP, Kibar Z, Chouery E, Munnich A, Cormier-Daire V, Mégarbané A. January 2014. "La protéine mitochondriale MAGMAS est impliquée dans une dysplasie squelettique sévère ». **7ème Assises de Génétique Humaine et Médicale, Bordeaux, France.**

Invited talks – Regional

1. **Mehawej C.** Invited to **deliver the Keynote address** highlighting my contributions to the field of human genetics especially after obtaining the L'Oréal-UNESCO for Women in Science Levant Young Talents Grant; at the Student Research Day organized by the **College of Health and Life Sciences at Hamad Bin Khalifa University.** Event will take place in Qatar on February 22nd 2024.
2. **Mehawej C.** Precision Medicine in Immune **Dysregulation** Diseases. December 2021. **The 9th Pan Arab Human Genetics Conference** organized by the Center for Arab Genomic Studies. Webinar.

Presentations and invited talks– National and Local

1. **Mehawej C.** Molecular diagnostics of rare genetic diseases: Challenges and Opportunities! March 4, 2023. *Rare Diseases Day, LAU Beirut Campus*.

2. **Mehawej C.** Artificial intelligence in Genetics. *LAU webinar. June 2021.*

3. **Mehawej C.,** Chouery E. and Megarbane A. Genetics in Healthcare. *LAU grand round. January 2021. Online webinar.*

4. **Mehawej C.** Personalized Medicine in the Field of Primary Immunodeficiencies. December 2020. 15th Annual meeting of the *Lebanese Society of Allergy and Immunology".* Webinar. (Invited talk)

5. **Mehawej C.** Multiplexing HPV E6-E7 mRNA and Cell Cycle Exact Classification of Cervical Pre-Cancer. January 2019. Staff meeting. *Abou Jaoude Hospital*. (Invited talk)

6. **Mehawej C.** Single Cell diagnostics. December 2018. *Saint-Jospeh Univesrity. Beirut, Lebanon.*

7. **Mehawej C**. "Dépistage des déficits immunitaires". May 2018. *5th edition of Printemps de la Faculté de Médecine. Beirut, Lebanon.* (Invited talk)

8. **Mehawej C**., Chebly A., Kourié H.R. August 2017. M.R.D in malignant hemtaology. 6th course of Hematology. *Bellevue Medical Center. Beirut, Lebanon.*

9. **Mehawej C.** "Practical Evaluation and Diagnosis of Primary Immunodeficiency. November 2017. *13th Annual meeting of the Lebanese Society of Allergy and Immunology". Beirut, Lebanon*. (Invited talk)

10. **Mehawej C.** "Approaches for novel gene discovery in genetic disorders". August 2016. Seminar. *Human Morphology- conference room D.T.S., American University of Beirut. Beirut, Lebanon*. (Invited talk)

III. TEACHING

Teaching Activities

June 2021 2020 - Present

Assistant Professor of Genetics
Discipline: Immunology
Medical students (year I)
Gilbert and Rose-Marie School of Medicine, *Lebanese American University*

September 2020- Present	Assistant Professor of Genetics **Discipine: Human Genetics Co-coordinator and instructor** Medical students (years I and II) *Gilbert and Rose-Marie School of Medicine*, *Lebanese American University*
September 2020 – Present	Assistant Professor of Genetics **Facilitator of Problem-Based Learning sessions** Medical students (years I and II) *Gilbert and Rose-Marie School of Medicine*, *Lebanese American University*
September 2020- Present	Assistant Professor of Genetics **OSCE examiner** Medical students (years I and II) *Gilbert and Rose-Marie School of Medicine*, *Lebanese American University*
2017 – August 2020	Assistant Professor **Courses: Immunogenetics and Functional Genetics** Medical students *School of Medicine*, *Saint-Joseph University*
2017- August 2020	Assistant Professor, **Course: Introduction to Functional Genetics** *Faculty of Dentistry, Saint-Joseph University*
2014 - 2015	Lecturer **Basic Genetics Course** *Midwifery School*, *School of Medicine*, *Saint-Joseph University.*

Mentoring Academic Trainees

2020- Present	**Advising and counseling medical students** (in their pre-clinical years). *Gilbert and Rose-Marie Chagoury school of medicine, LAU*
2023-Present	**Supervisor of Year-Long Project** **Students:** Robin Farah, Marianne Chaiban, Anthony Tannous, Michael Anthony Juvelekian, Hala El Feel, Romy Assaf, Tiya Berjawi, Nour Allakis **Title:** Assessing the knowledge and attitude related to newborn blood spot test among Lebanese health care students *Gilbert and Rose-Marie Chagoury school of medicine, LAU*
2022-2023	

Supervisor of Year-Long Project
Students: Jane Aoun, Ghady Baz, Joseph El-Haddad, Mohammad Hamzah, Genty Makhoul, Nadia Rifai, Khalid Trad, Karl Khalil
Title: Emergency contraception knowledge among Lebanese health care students *Gilbert and Rose-Marie Chagoury school of medicine, LAU*

2021- 2022

Supervisor of Year-Long Project
Students: Fatima Al Sayed, Judy Al Ghoul, Elise Asouad, Hussein Abedel Nabi, Ahmad Haj Hussein, Phillipe Saad, Hassan Soucair, Ramy Touma Sawaya
Title: Assessing the knowledge and attitude of the Lebanese population towards organ donation
Gilbert and Rose-Marie Chagoury school of medicine, LAU

2020- Present

Supervising medical students in preparing, writing and publishing case reports
Gilbert and Rose-Marie Chagoury school of medicine, LAU

2020- Present

Mentoring a PhD student, in collaboration with Dr Olivier de Backer (University of Namur) and Dr Eliane Chouery (LAUSOM)
Student name: Miss Nadine Hamdan
Project title: BOD1-associated syndromic intellectual disability: from the clinic to the mouse models
University of Namur, Belgium

2019-2020

Master's mentor and supervisor (Master's in Biological and Medical sciences)
Student name: Miss Gaelle Feghali
Project title: Investigation of the molecular bases of autoinflammatory diseases
Faculty of medicine, Saint-Joseph University of Beirut

2018-2019

Master's mentor and supervisor (Master's in Biological and Medical sciences)
Student name: Mr Walid Maacaron
Project title: Assessment, in the lebanese population, of two polymorphisms located in the IL1B gene and associated with increased susceptibility to lung cancer
Faculty of medicine, Saint-Joseph University of Beirut

Curricular Developments, Reviews and Academic Contributions

September 2023

Contribution to the design of the syllabus of the "Certificate in Genetics and Genomic Medicine" prepared by the Department of human genetics in collaboration with the CME/CPD office.
Targeted audience: Healthcare professionals including physicians, nurses, laboratory professionals, pharmacists, researchers, and public health professionals.

Gilbert and Rose-Marie School of Medicine, *Lebanese American University*

2022- Present **Coordinator of humanities at the LAUSOM**
Gilbert and Rose-Marie School of Medicine, *Lebanese American University*

September 2020 – Present **Co-coordination of the genetics discipline at the LAUSOM**
Gilbert and Rose-Marie School of Medicine, *Lebanese American University*

September 2020- Present **Contribution to the Research Certificate** (contribution to syllabus update and delivery of sessions), in collaboration between LAU Gilbert and Rose-Marie School of Medicine research committee and CME/CPD office
Targeted audience: LAU physicians and faculty
Gilbert and Rose-Marie School of Medicine, *Lebanese American University*

CONFERENCE DESIGN

September 2023 **Member in the scientific committee of the symposium "Update on Hereditary and Familial Cancer" symposium** that will be held on September 29, 2023, at LAU medical center -Rizk Hospital, in the presence of the Minister of Public Health and international renowned speakers.

February 2023 **Member of the organizing and scientific committee of the Rare Disease Day 2023,** event organized for the first time in Lebanon, by the LAUSOM department of human genetics in collaboration with the CME/CPD office. This event included: A morning activity with the patients affected with rare genetic diseases and their families (a short basketball game), a symposium including two international speakers and national speakers; in addition to a musical concert.

February 2021 **Member in the scientific committee of the webinar**: "COVID-19: Immune response towards SARS-CoV-2 infection and vaccine". This webinar hosted host 4 Internationally recognized speakers (from Harvard Medical School, Radboud university, Paris-Diderot University and University of Zurich); and was organized by the Department of Human Genetics – LAUSOM, in collaboration with the LAUSOM CME/CPD office and the Center for Arab Genomics Studies (UAE)

December 2021 **Member in the scientific committee of the webinar**: **"Genetics: Beyond Mendel's Laws"**. This webinar hosted 2 internationally recognized speakers (from University of Lausanne and University of Versailles Saint-Quentin-en-Yvelines); and was organized by the Department of Human Genetics – LAUSOM, in collaboration with the LAUSOM CME/CPD office and the Center for Arab Genomics Studies (UAE)

IV. SERVICES

Professional Societies Membership

2018 - 2020	European Society of Immunodeficiencies (ESID)
2019-2021	Clinical immunology society (CIS)
2022 – Present	Member of the academic committee at the arab association for genetic counselors
2023 – Present	American Society of Human Genetics (ASHG)

Reviewer/ Editor

2014- present **Reviewer in peer-reviewed journals including** *BMC medical genomics, Frontiers in genetics, International journal of medical research, American journal of medical genetics…*

Services to the Institution and to the School of Medicine

Services at the Lebanese American University:

- **Student's affair committee** (LAUSOM, Since 2023).
- **School Of Medicine Faculty Liaison (FL)** at LAU Library Liaison Program (LAU, since 2023). *The Library Liaison Program works to improve collection development, research needs, information literacy skills for faculty and students and any other service offered by the LAU Libraries.*
- **Assessment committee** (LAUSOM, since 2022): *This committee aims to design and revise assessment plans and to oversee assessment practices to maintain alignment with the SOM mission, education, and educational outcomes.*
- **Students disciplinary committee** (LAUSOM, since 2021): *This committee, chaired by the Dean of the LAUSOM, investigates complaints against students, when they occur; in order to make a determination and issue any appropriate sanction.*

- **Student promotion subcommittee** (LAUSOM, since 2020): *This subcommittee discusses the performance of students, the status of the borderline/failing students and possible requirements/recommendations or plans for these students.*

- **Program Evaluation Committee** (LAUSOM, since 2020): *This is a standing committee at the LAUSOM that aims to design and revise program evaluation plans and to oversee program evaluation practices in order to implement continual improvement of the curriculum.*

- **Exam committee**, MCQ Exam Review Subcommittee for Med II (LAUSOM, since 2020). *This committee reviews the MedII local exams prepared by the faculty members, performs english editing when needed, and ensures that the questions are formulated as per guidelines set by this committee.*

- **Examiner in the OSCEs** for year one and two medical students ((LAUSOM, since 2021).

- **Contribution to medical students' admission process as an interviewer** (LAUSOM, since 2020).

- **Contribution to students' recruitment at LAU** (Participation in the open days organized at LAU).

Services to the Medical Centers

2020- Present	In charge of the molecular diagnosis of patients referred to the department of Human Genetics. Gilbert and Rose-Marie Chagoury school of medicine, Lebanese American University.
2018-2020	Management of the neonatal screening for severe primary immunodeficiencies (TREC test) at the Medical Genetics Unit, Saint-Joseph University, Beirut, Lebanon
2019	Introduction and management of HPV testing by Flow cytometry at the Medical Genetics Unit, Saint Joseph University, Beirut
2017-2020	In charge of the molecular diagnosis of patients with primary immunodeficiencies. Medical Genetics Unit, Saint-Joseph University, Beirut, Lebanon

Services to the Community

2017- present Contribution to several public awareness campaigns related to rare genetic diseases, consanguinity, and inborn errors of immunity (interviews on several media platforms, oral presentations in meetings, presence and participation in medical staff meetings at several hospitals, etc.)

2018 Contribution to the implementation of the neonatal screening for severe inborn errors of immunity (TREC test) in Lebanon, in collaboration between Saint-Joseph University, American University fof Beorut, the Lebanese ministry of public health and iFight PID fund.

IV. CME & CPD ACTIVITIES

CME & CPD Development Activities

Scientific meetings and conferences:

2022: 20th Biennial meeting of the European Society for Immunodeficiencies (ESID), Gothenberg, Sweden; Hybrid meeting.
2022: European Society of Human Genetics (ESHG) annual conference, Vienna, Austria.
2022: Essentials skills in Medical Education and Professional Development. Lebanese American University Gilbert and Rose-Marie Chagoury School of medicine.
2021: 9th Pan Arab Human Genetics Conference, Dubai, online.
2021: Center for Arab Genomic Studies webinar series.
2020: Clinical Immunology School. Online meeting.
2020: 15th Annual meeting of the Lebanese Society of Allergy and Immunology. Webinar.
2019: Clinical Immunology School, July 2019, Boston. USA.
2019: Recent Advances in Rare Diseases meeting. Bogota, Colombia
2018: Primary Immunodeficiency Diseases (PID) Course, organized by Universal Scientific Education and Research Network (USERN) and European School of Genetic Medicine (ESGM), hosted by the Universita Mediterranea of Reggio Calabria, Reggio Calabria, Italy.
2018: European Society of Immunodeficiencies meeting, Lisbon, Portugal.
2018: European Society of Human Genetics (ESHG) annual conference, Milan, Italy.
2018: 7th Pan Arab Human Genetics Conference, Dubai
2017: 13th Annual meeting of the Lebanese Society of Allergy and Immunology, Beirut, Lebanon.
2016: European Society for Immunodeficiencies (ESID), Barcelona, Spain.
2014: American Society of Human Genetics (ASHG) annual conference 2014, San Diego, California.
2014: LEB'IN conference, "Challenges and Advances in Rare Genetic diseases: Impact on EU-Mediterranean Cooperation", 2014, Marseille, France.
2014: European Society of Human Genetics (ESHG) annual conference 2014, Milan, Italy.
2014: 7èmes Assises de Génétique Humaine et Médicale, Bordeaux, France.
2013: European Society of Human Genetics (ESHG) annual conference 2013, Paris, France.
2013: Doctoral forum of "Université Paris Descartes", 2013, Paris, France.
2012: Doctoral forum of "Université Paris Descartes", 2012, Paris, France.
2012: Clinical Genetics seminar of the « French reference center for rare diseases - Constitutional bone diseases», Paris, France.
2012: 6èmes Assises de Génétique Humaine et Médicale, 2012, Marseille, France.
2011: European Society of Human Genetics (ESHG) annual conference 2011, Amsterdam, Holland.

V. REFEREES

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