Said El Shamieh, PhD, HDR



Human and Statistical Geneticist
Lebanese-French, 41 years
H-index: 25 | i10-index: 49 | 83 peer-reviewed publications | 1,850 citations
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EDUCATION

2026: Habilitation à Diriger des Recherches (HDR), Health Section, *Université Sorbonne*, Paris, France.

2009–2012: PhD in Health and Life Sciences, *Université de Lorraine*, Nancy, France.

2008–2009: M.Sc. in Molecular, Structural, and Cellular Biology, *Université Henri Poincaré*, Nancy, France.

2007–2008: Master's 1 in Biochemistry and Molecular Biology, *Université Henri Poincaré*, Nancy, France.

2003–2007: Bachelor's degree in Laboratory Sciences, Lebanese University, Lebanon.

PROFESSIONAL EXPERIENCE

2025–Present: Professor of Human Statistical Genetics, Faculty of Health Sciences, Beirut Arab University, Beirut, Lebanon.

June 2024–Present: Adjunct Researcher in Human Statistical Genetics, Audo-Zeitz team, Institut de la Vision, Université Sorbonne, Paris, France.

2019–2025: Associate Professor of Human Statistical Genetics, Faculty of Health Sciences, Beirut Arab University, Beirut, Lebanon.

2016–2019: Assistant Professor of Human and Statistical Genetics, Faculty of Health Sciences, Beirut Arab University, Beirut, Lebanon.

2015–2016: Part-time Assistant Professor, Faculty of Sciences, Beirut Arab University, Beirut, Lebanon.

2012–2014: Postdoctoral Fellow, Audo-Zeitz team, Institut de la Vision, Université Sorbonne, Paris, France.

2009–2012: Graduate Teaching, Faculty of Pharmacy, Université de Lorraine, Nancy, France.

SCIENTIFIC LEADERSHIP AND ADVISORY ROLES

2024–2025: Strategic Partner, Center of Arab Genomic Studies, UAE.

Supports regional efforts to document, analyze, and reduce the burden of genetic disorders in Arab populations.

2022–Present: Associate Editor, BMC Medical Genomics (Springer Nature) and Frontiers in Bioscience-Landmark (IMR Press).

2021–2024: Team Leader and Steering Committee Member, Erasmus+ CBHE grant: "Online Pharmacogenomics and Personalized Medicine" (OPPM; 1 Million Euros).

Led work packages and oversaw the design and accreditation of the first postgraduate program in Pharmacogenomics and Personalized Medicine in the MENA region.

2020–2021: Scientific Consultant, Digitsole® Company, Nancy, France.

Advised on the integration of biometric and genomic data for personalized digital medicine.

AWARDS AND HONORS

2026: Winner of the Beirut Arab University Research Award, Beirut Arab University, Beirut, Lebanon.

2024: Rare Disease Science Communication Award (3billion Inc., Seoul, South Korea) for ABCA4-associated inherited retinal diseases in Lebanon.

2024–2025: Selected as Strategic Partner, Center of Arab Genomic Studies (CAGS), UAE.

2009–2012: Région Lorraine Doctoral Research Grant, France (29,000 EUR, 3 years).

PUBLICATIONS

83 International Peer-reviewed Publications

1: Colombo L, Maltese PE, Castori M, El Shamieh S, Zeitz C, Audo I, Zulian A, Marinelli C, Benedetti S, Costantini A, Bressan S, Percio M, Ferri P, Abeshi A, Bertelli M, Rossetti L. Molecular Epidemiology in 591 Italian Probands With Nonsyndromic Retinitis Pigmentosa and Usher Syndrome. Invest Ophthalmol Vis Sci. 2021 Feb 1;62(2):13. doi: 10.1167/iovs.62.2.13. PMID: 33576794.

2: Ibrahim M, Jaffal L, Assi A, Helou C, El Shamieh S. ABCA4-related retinopathies in Lebanon. Heliyon. 2024 Apr 26;10(9):e30304. doi: 10.1016/j.heliyon.2024.e30304. PMID: 38694055; PMCID: PMC11061736.

3: Jaffal L, Mrad Z, Ibrahim M, Salami A, Audo I, Zeitz C, El Shamieh S. The research output of rod-cone dystrophy genetics. Orphanet J Rare Dis. 2022 Apr 23;17(1):175. doi: 10.1186/s13023-022-02318-5. PMID: 35461258.

4: Audo I, Mohand-Said S, Boulanger-Scemama E, Zanlonghi X, Condroyer C, Démontant V, Boyard F, Antonio A, Méjécase C, El Shamieh S, Sahel JA, Zeitz C. MERTK mutation update in inherited retinal diseases. Hum Mutat. 2018 Jul;39(7):887-913. doi: 10.1002/humu.23431. Epub 2018 May 23. PMID: 29659094.

5: El Shamieh S, Maltese PE. Editorial: The genetics of inherited retinal diseases in understudied ethnic groups: Novel associations, challenges, and perspectives. Front Genet. 2022 Aug 23;13:990782. doi: 10.3389/fgene.2022.990782. PMID: 36081992; PMCID: PMC9445133.

6: El Ghoch M, El Shamieh S. Is There a Link Between Nutrition, Genetics, and Cardiovascular Disease? J Cardiovasc Dev Dis. 2020 Aug 27;7(3):33. doi: 10.3390/jcdd7030033. PMID: 32867398; PMCID: PMC7570096.

7: El Shamieh S, Chebly A. Trends in scientific publishing: does quantity compromise quality in life sciences and medicine? Syst Rev. 2024 Oct 1;13(1):249. doi: 10.1186/s13643-024-02668-0. PMID: 39354621; PMCID: PMC11443745.

8: Zeitz C, Navarro J, Azizzadeh Pormehr L, Méjécase C, Neves LM, Letellier C, Condroyer C, Albadri S, Amprou A, Antonio A, Ben-Yacoub T, Wohlschlegel J, Andrieu C, Serafini M, Bianco L, Antropoli A, Nassisi M, El Shamieh S, Chantot- Bastaraud S, Mohand-Saïd S, Smirnov V, Sahel JA, Del Bene F, Audo I. Variants in UBAP1L lead to autosomal recessive rod-cone and cone-rod dystrophy. Genet Med. 2024 Jun;26(6):101081. doi: 10.1016/j.gim.2024.101081. Epub 2024 Jan 28. PMID: 38293907.

9: Ndiaye NC, Azimi Nehzad M, El Shamieh S, Stathopoulou MG, Visvikis-Siest S. Cardiovascular diseases and genome-wide association studies. Clin Chim Acta. 2011 Sep 18;412(19-20):1697-701. doi: 10.1016/j.cca.2011.05.035. Epub 2011 Jun 7. PMID: 21672533.

10: Khachab Y, El Shamieh S, Sokhn ES. Gram-negative bacterial colonization in the gut: Isolation, characterization, and identification of resistance mechanisms. J Infect Public Health. 2024 Oct;17(10):102535. doi: 10.1016/j.jiph.2024.102535. Epub 2024 Aug 30. PMID: 39216133.

11: Zeitz C, Nassisi M, Laurent-Coriat C, Andrieu C, Boyard F, Condroyer C, Démontant V, Antonio A, Lancelot ME, Frederiksen H, Kloeckener-Gruissem B, El- Shamieh S, Zanlonghi X, Meunier I, Roux AF, Mohand-Saïd S, Sahel JA, Audo I. CHM mutation spectrum and disease: An update at the time of human therapeutic trials. Hum Mutat. 2021 Apr;42(4):323-341. doi: 10.1002/humu.24174. Epub 2021 Feb 19. PMID: 33538369.

12: Jaffal L, Ibrahim M, El Shamieh S. Analysis of rod-cone dystrophy genes reveals unique mutational patterns. BMJ Open Sci. 2022 Dec 13;6(1):e100291. doi: 10.1136/bmjos-2022-100291. PMID: 36618607; PMCID: PMC9812813.

13: El Shamieh S, Visvikis-Siest S. Genetic biomarkers of hypertension and future challenges integrating epigenomics. Clin Chim Acta. 2012 Dec 24;414:259-65. doi: 10.1016/j.cca.2012.09.018. Epub 2012 Sep 23. PMID: 23010416.

14: Jaffal L, Joumaa H, Mrad Z, Zeitz C, Audo I, El Shamieh S. The genetics of rod-cone dystrophy in Arab countries: a systematic review. Eur J Hum Genet. 2021 Jun;29(6):897-910. doi: 10.1038/s41431-020-00754-0. Epub 2020 Nov 13. PMID: 33188265; PMCID: PMC8187393.

15: El Shamieh S, Zgheib NK. Pharmacogenetics in developing countries and low resource environments. Hum Genet. 2022 Jun;141(6):1159-1164. doi: 10.1007/s00439-021-02260-9. Epub 2021 Feb 9. PMID: 33564904.

16: Bissar N, Kassir R, Salami A, El Shamieh S. Association of immunity-related gene SNPs with Alzheimer's disease. Exp Biol Med (Maywood). 2024 Nov 22;249:10303. doi: 10.3389/ebm.2024.10303. PMID: 39651329; PMCID: PMC11620869.

17: Banjak M, Noureldine J, Mousawi Z, Nehme J, Jaffal L, El Shamieh S. Systematic review of genotype-phenotype associations in CRX-associated retinal dystrophies. BMJ Open Ophthalmol. 2025 Mar 25;10(1):e002030. doi: 10.1136/bmjophth-2024-002030. PMID: 40132901; PMCID: PMC11938251.

18: Daher A, Banjak M, Noureldine J, Nehme J, El Shamieh S. Genotype-phenotype associations in CRB1 bi-allelic patients: a novel mutation, a systematic review and meta-analysis. BMC Ophthalmol. 2024 Apr 15;24(1):167. doi: 10.1186/s12886-024-03419-4. PMID: 38622537; PMCID: PMC11017593.

19: Bianco L, Navarro J, Michiels C, Sangermano R, Condroyer C, Antonio A, Antropoli A, Andrieu C, Place EM, Pierce EA, El Shamieh S, Smirnov V, Kalatzis V, Mansard L, Roux AF, Bocquet B, Sahel JA, Meunier I, Bujakowska KM, Audo I, Zeitz C. Identification of IDH3G, encoding the gamma subunit of mitochondrial isocitrate dehydrogenase, as a novel candidate gene for X-linked retinitis pigmentosa. Genet Med. 2025 Jun;27(6):101418. doi: 10.1016/j.gim.2025.101418. Epub 2025 Mar 19. PMID: 40119724.

20: Fakhoury HMA, El Shamieh S, Rifai A, Tamim H, Fakhoury R. Vitamin D Related Gene Polymorphisms and Cholesterol Levels in a Mediterranean Population. JCardiovasc Dev Dis. 2022 Mar 27;9(4):102. doi: 10.3390/jcdd9040102. PMID: 35448078; PMCID: PMC9028837.

21: Maltese PE, Colombo L, Martella S, Rossetti L, El Shamieh S, Sinibaldi L, Passarelli C, Coppè AM, Buzzonetti L, Falsini B, Chiurazzi P, Placidi G, Tanzi B, Bertelli M, Iarossi G. Genetics of Inherited Retinal Diseases in Understudied Ethnic Groups in Italian Hospitals. Front Genet. 2022 Jun 28;13:914345. doi: 10.3389/fgene.2022.914345. PMID: 35836572; PMCID: PMC9274138.

22: Salem-Sokhn E, Salami A, Fawaz M, Eid AH, El Shamieh S. Helicobacter Pylori Interacts with Serum Vitamin D to Influence Hypertension. Curr Aging Sci. 2021;14(1):26-31. doi: 10.2174/1874609813666200925104248. PMID: 32981515.

23: Rancier M, Zaaber I, Stathopoulou MG, Chatelin J, Saleh A, Marmouch H, El Shamieh S, Masson C, Murray H, Lamont J, Fitzgerald P, Mahjoub S, Said K, Tensaout BB, Mestiri S, Visvikis-Siest S. Pro- and anti-angiogenic VEGF mRNAs in autoimmune thyroid diseases. Autoimmunity. 2016 Sep;49(6):366-372. doi: 10.1080/08916934.2016.1199019. Epub 2016 Aug 5. PMID: 27494076.

24: Nivet-Antoine V, Labat C, El Shamieh S, Dulcire X, Cottart CH, Beaudeux JL, Zannad F, Visvikis-Siest S, Benetos A. Relationship between catalase haplotype and arterial aging. Atherosclerosis. 2013 Mar;227(1):100-5. doi: 10.1016/j.atherosclerosis.2012.12.015. Epub 2013 Jan 8. PMID: 23340375.

25: Méjécase C, Hummel A, Mohand-Saïd S, Andrieu C, El Shamieh S, Antonio A, Condroyer C, Boyard F, Foussard M, Blanchard S, Letexier M, Saraiva JP, Sahel JA, Zeitz C, Audo I. Whole exome sequencing resolves complex phenotype and identifies CC2D2A mutations underlying non-syndromic rod-cone dystrophy. Clin Genet. 2019 Feb;95(2):329-333. doi: 10.1111/cge.13453. Epub 2018 Nov 4. PMID: 30267408.

26: Kikuchi S, Kameya S, Gocho K, El Shamieh S, Akeo K, Sugawara Y, Yamaki K, Zeitz C, Audo I, Takahashi H. Cone dystrophy in patient with homozygous RP1L1 mutation. Biomed Res Int. 2015;2015:545243. doi: 10.1155/2015/545243. Epub 2015 Jan 29. PMID: 25692141; PMCID: PMC4322316.

27: Naja K, El Shamieh S, Fakhoury R. rs622342A>C in SLC22A1 is associated with metformin pharmacokinetics and glycemic response. Drug Metab Pharmacokinet. 2020 Feb;35(1):160-164. doi: 10.1016/j.dmpk.2019.10.007. Epub 2019 Nov 1. PMID:31974043.

28: El Shamieh S, Costanian C, Kassir R, Visvkis-Siest S, Bissar-Tadmouri N. APOE genotypes in Lebanon: distribution and association with hypercholesterolemia and Alzheimer's disease. Per Med. 2019 Jan;16(1):15-23. doi: 10.2217/pme-2018-0067. Epub 2018 Nov 20. PMID: 30457419.

29: Masri I, Salami A, El Shamieh S, Bissar-Tadmouri N. rs3851179G>A in PICALM is Protective Against Alzheimer's Disease in Five Different Countries Surrounding the Mediterranean. Curr Aging Sci. 2020;13(2):162-168. doi: 10.2174/1874609812666191019143237. PMID: 31648652.

30: Jaffal L, Joumaa WH, Assi A, Helou C, Condroyer C, El Dor M, Cherfan G, Zeitz C, Audo I, Zibara K, El Shamieh S. Novel Missense Mutations in BEST1 Are Associated with Bestrophinopathies in Lebanese Patients. Genes (Basel). 2019 Feb 18;10(2):151. doi: 10.3390/genes10020151. PMID: 30781664;PMCID: PMC6409913.

31: Chedid P, Salami A, Ibrahim M, Visvikis-Siest S, El Shamieh S. The association of vascular endothelial growth factor related SNPs and circulating iron levels might depend on body mass index. Front Biosci (Landmark Ed). 2022 Jan 18;27(1):27. doi: 10.31083/j.fbl2701027. PMID: 35090332.

32: Chedid P, Salami A, El Shamieh S. The Association of rs1898830 in Toll- Like Receptor 2 with Lipids and Blood Pressure. J Cardiovasc Dev Dis. 2020 Jul 8;7(3):24. doi: 10.3390/jcdd7030024. PMID: 32650372; PMCID: PMC7569770.

33: Neuillé M, El Shamieh S, Orhan E, Michiels C, Antonio A, Lancelot ME, Condroyer C, Bujakowska K, Poch O, Sahel JA, Audo I, Zeitz C. Lrit3 deficient mouse (nob6): a novel model of complete congenital stationary night blindness (cCSNB). PLoS One. 2014 Mar 5;9(3):e90342. doi: 10.1371/journal.pone.0090342. PMID: 24598786; PMCID: PMC3943948.

34: Smirnov VM, Nassisi M, Solis Hernandez C, Méjécase C, El Shamieh S, Condroyer C, Antonio A, Meunier I, Andrieu C, Defoort-Dhellemmes S, Mohand-Said S, Sahel JA, Audo I, Zeitz C. Retinal Phenotype of Patients With Isolated Retinal Degeneration Due to CLN3 Pathogenic Variants in a French Retinitis Pigmentosa Cohort. JAMA Ophthalmol. 2021 Mar 1;139(3):278-291. doi:10.1001/jamaophthalmol.2020.6089. Erratum in: JAMA Ophthalmol. 2021 Dec 1;139(12):1324.

35: Nzietchueng R, El Shamieh S, Benachour H, Labat C, Herbeth B, Ndiaye NC, Masson C, Visvikis-Siest S, Benetos A. Klotho KL-VS genotype is involved in blood pressure regulation. Clin Chim Acta. 2011 Sep 18;412(19-20):1773-7. doi: 10.1016/j.cca.2011.05.032. Epub 2011 Jun 1. PMID: 21663735.

36: Jaffal L, Akhdar H, Joumaa H, Ibrahim M, Chhouri Z, Assi A, Helou C, Lee H, Seo GH, Joumaa WH, El Shamieh S. Novel Missense and Splice Site Mutations in USH2A, CDH23, PCDH15, and ADGRV1 Are Associated With Usher Syndrome in Lebanon. Front Genet. 2022 May 16;13:864228. doi: 10.3389/fgene.2022.864228. PMID: 35651951; PMCID: PMC9149366.

37: Mangino M, Hwang SJ, Spector TD, Hunt SC, Kimura M, Fitzpatrick AL, Christiansen L, Petersen I, Elbers CC, Harris T, Chen W, Srinivasan SR, Kark JD, Benetos A, El Shamieh S, Visvikis-Siest S, Christensen K, Berenson GS, Valdes AM, Viñuela A, Garcia M, Arnett DK, Broeckel U, Province MA, Pankow JS, Kammerer C, Liu Y, Nalls M, Tishkoff S, Thomas F, Ziv E, Psaty BM, Bis JC, Rotter JI, Taylor KD, Smith E, Schork NJ, Levy D, Aviv A. Genome-wide meta-analysis points to CTC1 and ZNF676 as genes regulating telomere homeostasis in humans. Hum Mol Genet. 2012 Dec 15;21(24):5385-94. doi: 10.1093/hmg/dds382. Epub 2012 Sep 21. PMID: 23001564; PMCID: PMC3510758.

38: Stathopoulou MG, Monteiro P, Shahabi P, Peñas-Lledó E, El Shamieh S, Silva Santos L, Thilly N, Siest G, Llerena A, Visvikis-Siest S. Newly identified synergy between clopidogrel and calcium-channel blockers for blood pressure regulation possibly involves CYP2C19 rs4244285. Int J Cardiol. 2013 Oct 3;168(3):3057-8. doi: 10.1016/j.ijcard.2013.04.097. Epub 2013 May 3. PMID: 23643422.

39: Méjécase C, Mohand-Saïd S, El Shamieh S, Antonio A, Condroyer C, Blanchard S, Letexier M, Saraiva JP, Sahel JA, Audo I, Zeitz C. A novel nonsense variant in REEP6 is involved in a sporadic rod-cone dystrophy case. Clin Genet. 2018 Mar;93(3):707-711. doi: 10.1111/cge.13171. PMID: 29120066.

40: Jaffal L, Joumaa WH, Assi A, Helou C, Cherfan G, Zibara K, Audo I, Zeitz C, El Shamieh S. Next Generation Sequencing Identifies Five Novel Mutations in Lebanese Patients with Bardet-Biedl and Usher Syndromes. Genes (Basel). 2019 Dec 16;10(12):1047. doi: 10.3390/genes10121047. PMID: 31888296; PMCID: PMC6947157.

41: El Shamieh S, Neuillé M, Terray A, Orhan E, Condroyer C, Démontant V, Michiels C, Antonio A, Boyard F, Lancelot ME, Letexier M, Saraiva JP, Léveillard T, Mohand-Saïd S, Goureau O, Sahel JA, Zeitz C, Audo I. Whole-exome sequencing identifies KIZ as a ciliary gene associated with autosomal-recessive rod-cone dystrophy. Am J Hum Genet. 2014 Apr 3;94(4):625-33. doi:10.1016/j.ajhg.2014.03.005. Epub 2014 Mar 27. PMID: 24680887; PMCID: PMC3980423.

42: Audo I, Bujakowska K, Orhan E, El Shamieh S, Sennlaub F, Guillonneau X, Antonio A, Michiels C, Lancelot ME, Letexier M, Saraiva JP, Nguyen H, Luu TD, Léveillard T, Poch O, Dollfus H, Paques M, Goureau O, Mohand-Saïd S, Bhattacharya SS, Sahel JA, Zeitz C. The familial dementia gene revisited: a missense mutation revealed by whole-exome sequencing identifies ITM2B as a candidate gene underlying a novel autosomal dominant retinal dystrophy in a large family. Hum Mol Genet. 2014 Jan 15;23(2):491-501. doi: 10.1093/hmg/ddt439. Epub 2013 Sep 10. PMID: 24026677.

43: Salami A, El Shamieh S. Association between SNPs of Circulating Vascular Endothelial Growth Factor Levels, Hypercholesterolemia and Metabolic Syndrome. Medicina (Kaunas). 2019 Aug 11;55(8):464. doi: 10.3390/medicina55080464. PMID: 31405227; PMCID: PMC6723464.

44: Salami A, Costanian C, El Shamieh S. rs2569190A>G in CD14 is Independently Associated with Hypercholesterolemia: A Brief Report. J Cardiovasc Dev Dis. 2019 Oct 30;6(4):37. doi: 10.3390/jcdd6040037. PMID: 31671579; PMCID:PMC6955813.

45: Froguel P, Ndiaye NC, Bonnefond A, Bouatia-Naji N, Dechaume A, Siest G, Herbeth B, Falchi M, Bottolo L, Guéant-Rodriguez RM, Lecoeur C, Langlois MR, Labrune Y, Ruokonen A, El Shamieh S, Stathopoulou MG, Morandi A, Maffeis C, Meyre D, Delanghe JR, Jacobson P, Sjöström L, Carlsson LM, Walley A, Elliott P, Jarvelin MR, Dedoussis GV, Visvikis-Siest S. A genome-wide association study identifies rs2000999 as a strong genetic determinant of circulating haptoglobin levels. PLoS One. 2012;7(3):e32327. doi: 10.1371/journal.pone.0032327. Epub 2012 Mar 5. PMID: 22403646; PMCID: PMC3293812.

46: Alghalyini B, El Shamieh S, Salami A, Visvikis Siest S, Fakhoury HM, Fakhoury R. Effect of SLCO1B1 gene polymorphisms and vitamin D on statin-induced myopathy. Drug Metab Pers Ther. 2018 Mar 28;33(1):41-47. doi: 10.1515/dmpt-2017-0030. PMID: 29420305.

47: El Shamieh S, Salami A, Fawaz M, Jounblat R, Waked M, Fakhoury R. rs6837671A>G in FAM13A Is a Trans-Ethnic Genetic Variant Interacting with Vitamin D Levels to Affect Chronic Obstructive Pulmonary Disease. J Pers Med. 2021 Jan 30;11(2):84. doi: 10.3390/jpm11020084. PMID: 33573279; PMCID: PMC7912529.

48: Orhan E, Prézeau L, El Shamieh S, Bujakowska KM, Michiels C, Zagar Y, Vol C, Bhattacharya SS, Sahel JA, Sennlaub F, Audo I, Zeitz C. Further insights into GPR179: expression, localization, and associated pathogenic mechanisms leading to complete congenital stationary night blindness. Invest Ophthalmol Vis Sci. 2013 Dec 9;54(13):8041-50. doi: 10.1167/iovs.13-12610. PMID: 24222301.

49: Moussa S, Saleh F, El Shamieh S, Assi T, Othman A, Farhat F. Detection of PIK3R1 (L449S) Mutation in a Patient with Ovarian Cancer: A Case Report. Case Rep Oncol. 2020 Feb 20;13(1):188-192. doi: 10.1159/000505723. PMID: 32231543; PMCID: PMC7098360.

50: El-Gowilly SM, Metwaly HA, Makhlouf D, Elmansoury N, Abuiessa SA, Sorour AA, Abdelgalil MH, Fawaz M, Abushady AM, Gamaleldin M, Abdelghany TM, Fakhoury R, Abdelhady R, Ghanim AM, Shehata S, Kamal M, Bahy R, Haroon SA, Manolopoulos VG, Cascorbi I, Daly A, Abdelkader NF, El Shamieh S, Nagy M, Wahid A. Analysis of the current situation of pharmacogenomics in terms of educational and healthcare needs in Egypt and Lebanon. Pharmacogenomics. 2024;25(10-11):429-440. doi: 10.1080/14622416.2024.2403967. Epub 2024 Oct 9. PMID: 39382016; PMCID: PMC11492645.

51: El Shamieh S, Boulanger-Scemama E, Lancelot ME, Antonio A, Démontant V, Condroyer C, Letexier M, Saraiva JP, Mohand-Saïd S, Sahel JA, Audo I, Zeitz C. Targeted next generation sequencing identifies novel mutations in RP1 as a relatively common cause of autosomal recessive rod-cone dystrophy. Biomed Res Int. 2015;2015:485624. doi: 10.1155/2015/485624. Epub 2015 Jan 6. PMID: 25692139; PMCID: PMC4307388.

52: El Shamieh S, Saleh F, Moussa S, Kattan J, Farhat F. RICTOR gene amplification is correlated with metastasis and therapeutic resistance in triple-negative breast cancer. Pharmacogenomics. 2018 Jun 1;19(9):757-760. doi: 10.2217/pgs-2018-0019. Epub 2018 May 23. PMID: 29790419.

53: El Shamieh S, Ndiaye NC, Stathopoulou MG, Murray HA, Masson C, Lamont JV, Fitzgerald P, Benetos A, Visvikis-Siest S. Functional epistatic interaction between rs6046G>A in F7 and rs5355C>T in SELE modifies systolic blood pressure levels. PLoS One. 2012;7(7):e40777. doi: 10.1371/journal.pone.0040777. Epub 2012 Jul 18. PMID: 22815813; PMCID: PMC3399862.

54: Gorenjak V, Vance DR, Petrelis AM, Stathopoulou MG, Dadé S, El Shamieh S, Murray H, Masson C, Lamont J, Fitzgerald P, Visvikis-Siest S. Peripheral blood mononuclear cells extracts VEGF protein levels and VEGF mRNA: Associations with inflammatory molecules in a healthy population. PLoS One. 2019 Aug 16;14(8):e0220902. doi: 10.1371/journal.pone.0220902. Erratum in: PLoS One. 2019 Oct 24;14(10):e0224591. doi: 10.1371/journal.pone.0224591. PMID: 31419243;

55: Boulanger-Scemama E, Mohand-Saïd S, El Shamieh S, Démontant V, Condroyer C, Antonio A, Michiels C, Boyard F, Saraiva JP, Letexier M, Sahel JA, Zeitz C, Audo I. Phenotype Analysis of Retinal Dystrophies in Light of the Underlying Genetic Defects: Application to Cone and Cone-Rod Dystrophies. Int J Mol Sci. 2019 Sep 30;20(19):4854. doi: 10.3390/ijms20194854. PMID: 31574917; PMCID: PMC6801687.

56: Siest G, Nezhad MA, Bagrel D, El Shamieh S, Lambert D, Ndiaye NC, Shahabi P, Visvikis-Siest S. Functional genomics towards personalized healthcare and systems medicine. Per Med. 2011 May;8(3):227-242. doi: 10.2217/pme.10.89. PMID: 29783520.

57: Assaad S, Costanian C, Jaffal L, Tannous F, Stathopoulou MG, El Shamieh S. Association of TLR4 Polymorphisms, Expression, and Vitamin D with Helicobacter pylori Infection. J Pers Med. 2019 Jan 11;9(1):2. doi: 10.3390/jpm9010002. PMID: 30641993; PMCID: PMC6463146.

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GRANTS

2024–Present: Principal Investigator, Intramural Fund, Beirut Arab University (10,000 USD, 1 year). This grant supports an institutional research initiative to advance genomic diagnostics for inherited retinal diseases. It involves targeted sequencing and phenotype correlation studies using patient cohorts from underserved communities.

2024–Present: Co-investigator, AlFaisal University (12,000 USD, 24 months). The project investigates rare ophthalmic disorders in Arab populations using whole-exome sequencing and functional annotation. My role includes variant curation, pathogenicity assessment, and capacity building for local genomic data analysis.

2024: SSHN Mobility Grant, French Institute, French Embassy in Lebanon (3,000 USD). This mobility grant facilitated scientific exchange and collaborative visits to research units in France, enhancing knowledge transfer and strengthening bilateral projects in vision genetics and pharmacogenomics.

2021–2024: Partner and Work Package Leader, Erasmus+ OPPM Project (100,000 USD, 3 years). As a steering committee member and WP10.3 leader, I coordinated management-related activities, led curricular development, and contributed to the accreditation of the first master's program in Pharmacogenomics and Precision Medicine in the MENA region.

2021: Principal Investigator, 3Billion Inc., Seoul, South Korea (35,000 USD, 8 months). This industry-academic collaboration aimed to assess the clinical utility of whole-exome sequencing in Lebanese patients with suspected inherited retinal disorders. Results informed variant interpretation frameworks and highlighted regional mutational spectra.

2018–2020: Principal Investigator, CNRS–Beirut Arab University Joint Grant (18,500 USD, 2 years). The project explored novel genotype-phenotype correlations in autosomal recessive retinopathies. It led to high-impact publications and reinforced national research capacity in molecular diagnostics.

2018–2020: Co-investigator, AlFaisal University (13,000 USD, 18 months). This grant supported cross-institutional data sharing and analysis of exome datasets from Middle Eastern cohorts. I contributed to the prioritization of rare variants and manuscript preparation.

2018–2020: Co-investigator, Lebanese University President’s Research Grant (16,000 USD, 2 years). The research focused on uncovering genetic factors underlying complex cardiovascular and ophthalmic traits in the Lebanese population. My contribution centered on bioinformatics and translational interpretation.

2016–2018: Principal Investigator, Lebanese University President’s Research Grant (15,000 USD, 2 years). This project initiated the first structured biobank of Lebanese families with inherited eye diseases, laying the groundwork for downstream genetic and pharmacogenomic research.

2016–2018: Co-investigator, Lebanese University President's Research Grant (12,000 USD, 2 years). I contributed to a collaborative project investigating the genetic basis of metabolic traits and their interaction with environmental risk factors, employing SNP-based association models.

2009–2012: Région Lorraine Doctoral Research Grant, France (29,000 EUR, 3 years). Awarded during my PhD training, this grant supported original research on statistical genomics and gene-environment interactions. It enabled extensive methodological development and multiple peer-reviewed outputs.

MENTORSHIP OF GRADUATE STUDENTS

PhD Thesis:

2024-Present: Main Supervisor of the PhD thesis in Biology: Uncovering Novel Genotype-Phenotype Associations in Inherited Retinal Diseases through Whole-Exome Sequencing and Functional Characterization. Mrs. Zahraa Mousawi, Beirut Arab University, Faculty of Sciences.

2016-2020: Main supervisor of the PhD thesis in Biology: Next Generation Sequencing for Lebanese Individuals with Inherited Retinal Diseases. Mrs. Lama Jaffal, Beirut Arab University, Faculty of Sciences.

2018- 2020: Co-supervisor of the PhD thesis in Biology: Analysis of CYP2C9 and (OCT1) polymorphisms in Lebanese patients with type 2 diabetes mellitus: influence on metformin/ glibenclamide therapy. Mr Khaled Naja, Beirut Arab University, Faculty of Sciences.

Master's Thesis:

2025: Vitamin D role and innate immune gene expression in Helicobacter pylori infection. Ms. Amara Ajib, Faculty of Health Sciences, Beirut Arab University.

2025: Association of Vitamin D with COVID-19 infection. Ms. Ferdos Misilmani, Faculty of Health Sciences, Beirut Arab University.

2025: Assessment of CCL20 and DEFA1-3 Gene Expression in COVID-19 Infection. Ms. Dima Maarabouni, Faculty of Health Sciences, Beirut Arab University.

2023: The study of the association of FTO rs9939609 and Type 2 Diabetes Mellitus, and the possible effect of Mediterranean Diet. Ms. Zeina El Kheir, M.Sc. in Nutrition and Dietetics, Faculty of Health Sciences, Beirut Arab University. (Co-supervised with Prof. Rajaa Fakhoury.)

2020: Investigating the Genetics of Rod-Cone Dystrophy in One Lebanese Family using Whole Exome Sequencing. Mrs Zamzam Mrad, Investigating the Genetics of Rod-Cone Dystrophy in One Lebanese Family using Whole Exome Sequencing.

2020 : Le séquençage de l'exome entier pour identifier la cause du syndrome d'USHER et de Rétinite pigmentaire dans deux familles Libanaises. Ms Hawraa Joumaa, Master 2 in Genomics and Health, Faculty of Sciences, Lebanese University.

2020–2021: Halal Authentication of Meat Species in Processed Canned Meat Commercialized in the Lebanese Market. Ms. Rayane Mohamad Jamal Damer, M.Sc. in Food Analysis and Safety, Faculty of Health Sciences, Beirut Arab University. (Co-supervised with Dr. Nada El Darra.)

2019–2020: Halal Authentication of Gelatine in Confectioneries Commercialized in Lebanese Market. Ms. Safaa Moustafa Osta, M.Sc. in Food Analysis and Safety, Faculty of Health Sciences, Beirut Arab University. (Co-supervised with Dr. Nada El Darra.)

2019: Investigating the Genetics of Usher Syndrome in Two Lebanese Families Using Whole Exome Sequencing. Ms. Zahraa Chhouri, Master 2 in Genomics and Health, Faculty of Sciences, Lebanese University.

2019: Investigating the Genetics of Rod Cone Dystrophy in Two Lebanese Families Using Whole Exome Sequencing. Ms. Mariam Ibrahim, Master 2 Physiology Epigenetics Development Cell Differentiation, Université Grenoble Alpes and Lebanese University.

2018: Mutation Analysis of Bestrophin1 (BEST1) gene in a Lebanese Family with Best Vitelliform Macular Dystrophy. Ms. Malak Banjak, Investigating the Genetics of Rod-Cone Dystrophy in One Lebanese Family using Whole Exome Sequencing.

2017: Epidemiological study of acute gastroenteritis among hospitalized children. Ms. Lamis Salloum, Master 2 Physiology Epigenetics Development Cell Differentiation, Université Grenoble Alpes and Lebanese University.

2017: Targeted Next Generation Sequencing for Two Lebanese Individuals with Rod Cone Dystrophy. Ms. Shirin Komeiha, Master 2 Physiology Epigenetics Development Cell Differentiation, Université Grenoble Alpes and Lebanese University.

COMMITTEES

2021-Present: Chair of the organization committee of BAU Webinar: The COVID-19 Pandemic in the Lebanese Setting: The Implications on Health & Wellbeing, January 27, 2021.

2019-Present: Member of the University research committee, Beirut Arab University, Beirut, Lebanon.

2017-Present: Member of the Lebanese colloquium committee for medical laboratory technology exams.

2016-Present: Chair of the research committee, Faculty of Health Sciences, Beirut Arab University.

CONFERENCES & WORKSHOPS

Invited Oral presentations:

2025: Inherited Retinal Diseases From Gene Discovery to Gene Therapy: Filling the gap between neurodevelopment and neurodegeneration. DAAD-Nile University, Egypt.

2025: Copy-number variations significantly contribute to the pathogenicity of inherited retinal diseases in Lebanon, International Society for Genetic Eye Diseases & Retinoblastoma (ISGEDR), Oslo, Norway.

2025: Genetics of Inherited Retinal Diseases (IRDs): From Gene Discovery to Gene Therapy, LAU Rare Diseases Day, March 8, 2025.

2022: Pharmacogenetics in Developing Countries: gaps, challenges, genetic diversity, and perspectives. The Industry Pharmacogenomics Working Group (I-PWG). Webex meeting.

2022: Three courses, human genetics, Human genomics, and genomics of infectious diseases, will be presented at the OPPM (Online pharmacogenomics and personalized medicine postgraduate program) all-partners' meeting in Oviedo, Spain.

2018: Genetic and protein profiling of cancer tumors, a first step towards personalized therapy, 9th International Santorini Conference" Systems medicine and Personalized Health and Therapy", Santorini, Greece.

2016: Cancer: A Role of Single Nucleotide Polymorphisms and Mutations, Workshop 'New trends in Cancer Research', Beirut Arab University, Faculty of Sciences, Lebanon.

2014: eMethSNPs; a hypothesis to be tested in the post-GWAS era, UniGR-Workshop Systems Biology, Epigenetics & Systems Analysis, Saarbrücken, Germany.

2014: Whole-Exome Sequencing Identifies KIZ as a Ciliary Gene Associated with Autosomal-Recessive Rod-Cone Dystrophy, Fifth annual Young Researchers in Life Sciences conference, Paris, France.

2013: Next-generation sequencing to determine the prevalence of gene mutations underlying rod-cone dystrophies, International Society for Genetic Eye Diseases & Retinoblastoma, Ghent, Belgium.

2012: eMethSNPs: a hypothesis to be tested in the post-GWAS era, 6th Santorini International Conference, Santorini, Greece.

2010: Human FPR1 C32T SNP interacts with age and is associated with blood pressure levels, 4th International Meeting of the French Society of Hypertension, Paris, France.

B- Poster presentations:

2026: Bridging the diagnostic gap in inherited retinal dystrophies using long-read sequencing, the Hybrid conference of the European Society of Human Genetics on Monday, Sweden.

2018: Mutated RB1 may promote chemo-resistance to palbociclib in ovarian cancer, 9th Santorini International Conference, Santorini, Greece.

2018: APOE genotypes in Lebanon: Distribution and association with hypercholesterolemia and Alzheimer's disease, 9th Santorini International Conference, Santorini, Greece.

2017: Mutation Analysis of the BEST1 Gene in a Lebanese Family with Vitelliform Macular Dystrophy Reveals a Novel Mutation with Incomplete Penetrance, American Academy of Ophthalmology, New Orleans, Louisiana, USA.

2014: Targeted next-generation sequencing identifies mutations in RP1 as a relatively common cause of autosomal recessive rod-cone dystrophy, ARVO conference, Orlando, Florida, USA.

2011: Klotho Kl-Vs Genotype Is Involved in Blood Pressure Regulation, The International Meeting of the French Society of Hypertension, Paris, France.

2010: Human FPR1 C32T SNP interacts with age and is associated with blood pressure levels, 4th International Meeting of the French Society of Hypertension, The Santorini International Conference, Santorini, Greece.

AD-HOC REVIEWER

I serve as a reviewer for; IOVS, Ophthalmic Genetics (Taylor & Francis), Molecular Vision, (Emory University), Clinica Chimica Acta (Elsevier), International Journal of Molecular Sciences (MDPI), BMC Medical Genetics (Springer Nature), Translational Vision Science and Technology (ARVO), British Journal of Ophthalmology (BMJ journals).

ACADEMIC JURY MEMBER (PhD and Master's)

I have served as an examiner for more than 30 theses (PhD and master's) at Beirut Arab University and other Lebanese institutions (Lebanese University, Université Saint Joseph, University of Balamand, Lebanese German University) since 2014, spanning fields such as human genetics, epigenetics, and molecular biology.

SKILLS & EXPERTISE

Research Methodologies:

Whole-exome sequencing (WES); targeted next-generation sequencing (NGS); long-read sequencing; copy number variation (CNV) analysis; Sanger sequencing; genotype-phenotype correlation; systematic review and meta-analysis; genome-wide association studies (GWAS); functional characterization of variants.

Bioinformatics and Data Analysis:

Variant calling and annotation (GATK, ANNOVAR, VEP); ClinVar and ACMG/AMP variant classification; population genetics databases (gnomAD, ExAC); statistical genetics (R, Python, PLINK); bibliometric analysis.

Clinical Genetics:

Genetic testing and counseling for inherited retinal diseases; pharmacogenomic testing interpretation; ACMG/AMP variant pathogenicity assessment; clinical report writing.

Languages:

Arabic (native), French (fluent), English (fluent).

TEACHING ACTIVITIES

Beirut Arab University, Faculty of Health Sciences:

- Human Genetics (undergraduate and graduate levels)
- Medical Genetics
- Human Genomics
- Genomics of Infectious Diseases
- Molecular Biology
- Biostatistics
- Research Methodology

Erasmus+ OPPM Program (2021–2024):

- Human Genetics (online postgraduate module)
- Human Genomics (online postgraduate module)
- Genomics of Infectious Diseases (online postgraduate module)

Université de Lorraine, Faculty of Pharmacy (2009–2012):

- Graduate teaching assistant in Genetics and Biochemistry

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