The University of Jordan School of Medicine

Hatem El-Shanti, MD
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July 29, 2017

I. EDUCATIONAL AND PROFESSIONAL HISTORY

A. List of Institutions Attended

1977-1983	Cairo University, School of Medicine, Cairo, Egypt; Medicine; Bachelor of Medicine & Surgery (M.B., B.Ch.), November 1983
1984-1985	Cairo University Hospitals, Cairo, Egypt; House Officer, rotating internship
1987-1989	Indiana University, Department of Medical Genetics, Indianapolis, IN, USA; Medical Genetics; Post-doctoral fellow
1987-1989	Indiana University, Department of Medical Genetics, Indianapolis, IN, USA; Medical Genetics; Master of Science (M.Sc.) Genetics
1989-1990	University of Iowa Hospitals & Clinics, Iowa City, IA, USA; Pediatrics, Internship
1990-1992	University of Iowa Hospitals & Clinics, Iowa City, IA, USA; Pediatrics, Residency
1992-1993	University of Iowa Hospitals & Clinics, Iowa City, IA, USA; Medical Genetics, Fellow

Certification

1985	Jordan Medical Council Comprehensive Test
1989	ECFMG Certification
1992	American Board of Pediatrics; # 48716, *valid until 12/31/2018*
1993	American Board of Medical Genetics, Clinical Genetics; # 93078, *valid till 12/31/2019*
1993	American Board of Medical Genetics, Clinical Cytogenetics; # 93078, *valid till 12/31/2019*
1993	Jordanian Board of Pediatrics, *unlimited validity*
2001	Jordanian Board of Medical Genetics, *unlimited validity*
2012	Diplôme d'Habilitation à Diriger des Recherches (Diploma of ability to direct research); Universite Paris-Des Cartes

Licensure

1987	Jordan Licence #5825 *(Permanent, to practice medicine)*
1989-1992	Iowa Resident Physician License # MD-R-4213
1992	Iowa License # 28647 *(valid until 1/1/2018)*
2016	Jordan Licence #1159/10020 *(Permanent, to practice pediatrics)*

B. Professional and Academic Positions

1985-1987	Teaching Assistant of Anatomy & General Practitioner; School of Medicine, Jordan University of Science & Technology (JUST), Irbid, Jordan
1993-1998	Assistant Professor of Pediatrics; JUST, Irbid, Jordan
1994-2002	Director, Cytogenetics laboratory; JUST, Irbid, Jordan
1996-2002	Clinical Geneticist and dysmorphologist; The National Center for Diabetes, Endocrinology & Genetics, Amman, Jordan
1996-1998	Assistant Professor of Medical Laboratory Sciences; JUST, Irbid, Jordan

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1998-2002	Associate Professor of Pediatrics & Medical Laboratory Sciences; JUST, Irbid, Jordan
2000-2001	Interim Chairman of Pediatrics; JUST, Irbid, Jordan
2000-2001	Medical Director, Princess Rahma Children Hospital, Irbid, Jordan
2003-2006	Associate Professor of Pediatrics; University of Iowa, Carver College of Medicine and Children's Hospital of Iowa, Iowa City, IA, USA
2004-2010	Faculty mentor, the interdisciplinary PhD program in Genetics, Carver College of Medicine, Iowa City, IA, USA
2006-2010	Associate Professor, with tenure, of Pediatrics; University of Iowa, Carver College of Medicine and Children's Hospital of Iowa, Iowa City, IA, USA
2007-2014	Managing Director and Principal Research Scientist, Shafallah Medical Genetics Center; Shafallah Center Foundation, Doha, Qatar
2011-2015	Director, SMGC – Diagnostic Molecular Genetics laboratory, Shafallah Medical Genetics Center; Shafallah Center Foundation, Doha, Qatar
2010-2017	Adjunct Associate Professor of Pediatrics; University of Iowa, Carver College of Medicine and Children's Hospital of Iowa, Iowa City, IA, USA
2014-2016	Scientific Director and Principal Research Scientist, QBRI-Medical Genetics Center; Qatar Biomedical Research Institute, Qatar Foundation, Research and Development, Doha, Qatar
2015-2016	Professor (joint appointment), College of Science and Engineering, Hamad bin Khalifa University, Qatar Foundation, Doha, Qatar
2016-now	Professor of Pediatrics; The University of Jordan, School of Medicine, Amman, Jordan
2016-now	Consultant of Pediatrics and Medical Genetics; Jordan University Hospital, Amman, Jordan
2017-now	Adjunct Professor of Pediatrics; University of Iowa, Carver College of Medicine and Children's Hospital of Iowa, Iowa City, IA, USA

C. Honors, Awards, Recognitions, Outstanding Achievements

1978	Certificate of Appreciation for excellent performance in the Premedical Studies; School of Medicine, Cairo University, Cairo, Egypt
1987	Scholarship for Pediatrics & Medical Genetics Training; JUST, Irbid, Jordan
1992	PL-3 Teacher of the Year Award; Department of Pediatrics, University of Iowa Hospitals & Clinics, Iowa City, IA, USA
1996	"Human Genome" Fellowship; UNESCO/TWAS
1997	Biography entered in "Who's Who in the World"
1997	First prize for best research project; Sixth Conference of Jordan Pediatric Society, Amman, Jordan
1998	Biography entered in "Who's Who in Medicine & Health Care"
1998	Certificate of Praise by the committee for the Abdul-Hamid Shuman Prize for the young Arab investigators, Amman, Jordan
1999-2000	Chaire Internationale de Recherche "Blaise Pascal" de l'état et de la région d'ile-de France, Paris, France. Highly competitive award with 3-5 yearly awardees (Chair Blaise Pascal scholars) since 1996, includes all fields of Science and Liberal Arts and supports research with a budget €200,000 over one year (equivalent to ~ $250,000). http://www.chaires-blaise-pascal.ens.fr/laureats-2014/laureats-1999/?lang=fr
2000	Biography entered in "Who's Who in Finance & Industry"
2001	Abdul-Hamid Shuman Prize in Medical Sciences for the Young Arab Investigators, Amman, Jordan

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2013 Best Research Presentation in Biomedical Sciences, ARC'13 (Annual Research Conference, 2013), Doha, Qatar

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II. TEACHING

A. Teaching Assignments

Classroom teaching

1985-1987	Teaching Assistant of Anatomy (4 semesters), Anatomy for Medical, Dental and Nursing Students; JUST, Irbid, Jordan
1995-1996	Human Cytogenetics (Course director; Biol 442, 3 credit hours/ semester); Department of Biology, JUST, Irbid, Jordan
1996-1997	Basic Human Genetics (Course director; graduate, 3 credit hours); Department of Medical Laboratory Sciences, JUST, Irbid, Jordan
1996-1997	Population Genetics (Course director; graduate, 3 credit hours); Department of Medical Laboratory Sciences, JUST, Irbid, Jordan
1996-1997	Cytogenetics (Course director; graduate, 3 credit hours); Department of Medical Laboratory Sciences, JUST, Irbid, Jordan
1998-1999	Human Cytogenetics (Course director; Biol 442, 3 credit hours); Department of Biology, JUST, Irbid, Jordan
1999-2000	Medical Genetics for medical students, Hopital Necker-Enfants Malades de l'Université Paris V, Paris, France. *Gave 2 one and half hour lectures*
2000-2001	Basic Human Genetics (Course director; graduate, 3 credit hours); Department of Medical Laboratory Sciences, JUST, Irbid, Jordan
2000-2001	Advanced Cytogenetics (Course director; graduate, 3 credit hours); Department of Medical Laboratory Sciences, JUST, Irbid, Jordan
2000-2001	Advanced topics in Genetics (Course director; graduate, 3 credit hours); Department of Medical Laboratory Sciences, JUST, Irbid, Jordan
2003-2007	Small group discussion in the Medical Genetics for M1 Students (Facilitator) 12 hours / semester; fall); Carver College of Medicine, University of Iowa, Iowa City, IA, USA
2005	Human Molecular Genetics (127:191) Spring 2005, Genetics PhD program; Carver College of Medicine, University of Iowa, Iowa City, IA, USA. *One and half hour lecture*
2005	Genetic Analysis of Biological Systems (127:150) Fall 2005, Genetics PhD program; Carver College of Medicine, University of Iowa, Iowa City, IA, USA. *One and half hour lecture*
2006	Genetic Analysis of Biological Systems (127:150) Fall 2006, Genetics PhD program; Carver College of Medicine, University of Iowa, Iowa City, IA, USA. *One and half hour lecture*
2010	Epidemiology and Prevention of Genetic Diseases, PGY1 residents in community medicine, Primary Health Care, Ministry of Health, Doha, Qatar
2014	Pathophysiology (BIOM 510) Fall 2014, Biomedical graduate program (Department of Health Sciences); College of Arts and Sciences, Qatar University, Doha, Qatar. *Three-hour lecture*
2014	IBRO-MENA Neurogenetics School, December 10 – 15, International Brain Research Organization – Middle East and North Africa Chapter, Qatar Biomedical Research Institute, Doha, Qatar. *Three one hour lectures*
2016	Advanced Genetics (LS:504) Spring 2016, Biomedical and Biological Science graduate program; College of Science and Engineering, Hamad bin Khalifa University, Doha, Qatar. *Course Coordinator and teaching one third of the course*

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Clinical Teaching

1993-2002 Pediatrics (outpatient clinic & attending for inpatients, lectures, seminars) for 5th and 6th year Medical Students & for Pediatrics residents (Clinical teaching, 20 hours / week for 46 weeks / year); Jordan University of Science & Technology, Irbid, Jordan

1995 Introduction to clinical medicine for 3rd year Medical Students (Instructor; 30 hours / week for 6 weeks); JUST, Irbid, Jordan

1996-2002 Genetics (outpatient clinic, lectures, seminars) for 5th and 6th year Medical Students & for Pediatrics residents (Clinical teaching, 6 hours / week for 46 weeks / year); The National Center for Diabetes, Endocrinology & Genetics and Jordan University School of Medicine, Amman, Jordan

2003-2007 Pediatric residents and 4th year medical student, rotating through Genetics, Children's Hospital of Iowa, University of Iowa, Iowa City, Iowa, USA

2016-now Pediatrics (outpatient clinic & attending for inpatients, lectures, seminars) for 5th and 6th year Medical Students & for Pediatrics residents; School of Medicine, University of Jordan, Amman, Jordan

B. Undergraduate, Medical, Graduate Students and Postdoctoral Fellows Supervised

Thesis Committee Member and/or Examination Committee Member

1994 Nahla S. El-Bayyari, MSc, Department of Public Health. "The Pattern of Intra-Uterine Fetal Growth for Normal Singleton Pregnancies in Irbid" JUST, Irbid, Jordan (Examination Committee Member)

1997 Mahmoud A. Al-Bashtawi, MSc, Department of Public Health. "Characteristics of Epileptic Children Attending Princess Rahma Teaching Hospital" JUST, Irbid, Jordan (Examination Committee Member)

1997 Hamid Abu-Ebiela, MSc, Department of Public Health. "A Study of Mild & Moderate Handicap in Children Registered at the Special Education Centers in Northern Jordan" JUST, Irbid, Jordan (Examination Committee Member)

1998 Ahmad Y.H. Abu-Dalou, MSc, Department of Anthropology. "Head Shape of Adult Males as a Possible Indicator of Economic Change in Northern Jordan (1900-1978)" Yarmouk University, Irbid, Jordan (Examination Committee Member)

1999 Fatin Y. Atrooz, MSc, Department of Biology. "The Antimutagenic Effect of Coumarin on Sister Chromatid Exchange Induced by Paracetamol & Mitomycin C in Cultured Human Lymphocytes" Yarmouk University, Irbid, Jordan (Examination Committee Member)

2000 Ayman I. Al-Jaru, MSc, Department of Medical Laboratory Sciences. "Validation studies of Random Amplified Polymorphic DNA (RAPD) in human & the possibilities of its implementation in forensic casework" University of Jordan, Amman, Jordan (Examination Committee Member)

2001 Zaid A. Abu-Rubaiha, MSc, Department of Medical Laboratory Sciences. "Survey of FMF Gene Mutations in Jordan" University of Jordan, Amman, Jordan (Examination Committee Member)

2004 Jane Kimani, Genetics PhD program; Carver College of Medicine, University of Iowa, Iowa City, Iowa, USA (Thesis and Comprehensive Examination Committee Member); graduated May 2007

2005 Fedick Rahimov, Genetics PhD program; Carver College of Medicine, University of Iowa, Iowa City, Iowa, USA (Thesis and Comprehensive Examination Committee Member)

2006 Luciana Miranda Van Westen, DDS, Genetics PhD program; Carver College of Medicine, University of Iowa, Iowa City, Iowa, USA (Comprehensive Examination Committee Member)

2008 Mohamed Abdul-Rasool Al-Drees, MSc, Biotechnology program. "A Novel Deletion Down Regulating Expression of the Low Density Lipoprotein Receptor in a Kuwaiti Kindred" Arabian Gulf University, Manama, Bahrain (Examination Committee Member)

2009 Omar M. Al-Suliaman, MSc, Biotechnology program. "Phenotype-Genotype Correlation by Mutation Position in a Twin Brothers and Their Sister Affected by Familial Mediterranean Fever" Arabian Gulf University, Manama, Bahrain (Examination Committee Member)

2012 Noura N. Al-Sakran, MSc, Biotechnology program. "Expression of Macrophage Migration Inhibitory Factor and Caspase-1 in Serum and Peripheral Blood Lymphocytes of a Familial Mediterranean Fever Patient and Control." Arabian Gulf University, Manama, Bahrain (Examination Committee Member)

Advisor

1996 Mohannad B. Al-Lahham, MSc, Department of Public Health, School of Medicine. "Normative Standards of Clinical Anthropometric Measurements for Jordanian Newborns." JUST, Irbid, Jordan

1998 Nahla N. M. Abu-Dehies, MSc, Department of Public Health, School of Medicine. "Normative Standards of Craniofacial Measurements in Jordanian Infants." JUST, Irbid, Jordan

1999 Asem Alkhateeb, MSc, Department of Medical Laboratory Sciences, Faculty of Applied Medical Sciences. "Testing heterogeneity, Investigating a Common Haplotype & Fine Mapping for Progressive Pseudorheumatoid Dysplasia." JUST, Irbid, Jordan

1999 Rula Abu-Dalu, MSc, Department of Medical Laboratory Sciences, Faculty of Applied Medical Sciences. "Molecular Studies on Periodic Fever Syndromes." JUST, Irbid, Jordan

1999 Marwan Tayeh, MSc, Department of Medical Laboratory Sciences, Faculty of Applied Medical Sciences. "Clinical & Molecular Approaches to the Mapping of Congenital Pernicious Anemia in an Inbred Kindred From Jordan." JUST, Irbid, Jordan

1999 Suhad M. Hamad, MSc, Department of Medical Laboratory Sciences, Faculty of Applied Medical Sciences. "Neonatal Screening for Biotinidase Deficiency." JUST, Irbid, Jordan (co-supervisor)

2002 Asmaa Al-Jamali, MSc, Department of Medical Laboratory Sciences, Faculty of Applied Medical Sciences. "Folic acid status among Jordanian women." JUST, Irbid, Jordan

2002 Dana Najib, MSc, Department of Medical Laboratory Sciences, Faculty of Applied Medical Sciences. "Heterozygote identification of the FMF gene among Jordanians." JUST, Irbid, Jordan

2003 Noor Jarboo, MSc, Department of Medical Laboratory Sciences, Faculty of Applied Medical Sciences. "Allele frequency of the thermolabile variants of MTHFR gene." JUST, Irbid, Jordan

2006 Amy Buhr, Genetics PhD program; Carver College of Medicine, University of Iowa, Iowa City, Iowa, USA
Amy Buhr obtained a Pre-doctoral Research Training Fellowship for her thesis project entitled "Gene discovery in autosomal recessive epilepsy" for the calendar year 2007 from the Epilepsy Foundation

2015 Yasser Al-Sarraj, MSc, Department of Life Sciences, College of Science and Engineering. "The Identifcation of Genetic Factors in the Etiology of Autism Spectrum Disorder in Arab Families." Hamad Bin Khalifa University, Doha, Qatar

2017 Areen Al-Abdallat, MSc, Department of Medical Laboratory Science, College of Medicine. "Study of the genotype – phenotype correlation patterns in Jordanian Familial Mediterranean Fever (FMF) patients and examination of some inflammatory markers." University of Jordan, Amman, Jordan

Rotational Supervisor

2003 Shawn Sato, (undergraduate student); 10 week summer rotation, "Homozygosity mapping of isolated ectopia lentis" University of Iowa, Iowa City, Iowa, USA

2004 Shawn Sato, (undergraduate student); 10 week summer rotation, "Candidate gene examination in isolated ectopia lentis" University of Iowa, Iowa City, Iowa, USA

2004 Meredith Smith, (undergraduate student); 10 week summer rotation, "Identification of the parent of origin of Trisomy 21" University of Iowa, Iowa City, Iowa, USA

2004 Leah Franck, (Medical Student); 12 week summer rotation, "Identification of the parent of origin of Trisomy 21" University of Iowa, Iowa City, Iowa, USA

2004 Luis Ochoa, (Medical Student); 12 week summer rotation, "Examination of the Role of *LPIN2* in psoriasis" University of Iowa, Iowa City, Iowa, USA

2005 Meredith Smith, (undergraduate student); 10 week summer rotation, "Homozygosity mapping of autosomal recessive epilepsy utilizing the GeneChip SNP genotyping technology" University of Iowa, Iowa City, Iowa, USA

2005 Pryanka Rao, (undergraduate student); 10 week summer rotation, "Creation of a *Lpin2* knock-out mouse" University of Iowa, Iowa City, Iowa, USA

2005 Tim Helmes, (undergraduate student); 10 week summer rotation, "Candidate gene examination in autosomal recessive epilepsy" University of Iowa, Iowa City, Iowa, USA

2005 Maisam Abul-Haija, MD, (Pediatrics resident); 8 week rotation, "Mutation analysis of G-6-PD gene" University of Iowa, Iowa City, Iowa, USA

2006 Amy Buhr, (graduate student) Genetics PhD program; Carver College of Medicine, University of Iowa, Iowa City, Iowa, USA (Laboratory Rotation, 12 weeks)

2006 Andrew Buller, (undergraduate student); 10 week summer rotation, "Break point identification in familial 4;16 translocation" University of Iowa, Iowa City, Iowa, USA

2006 Annie Porter, (undergraduate student); 10 week summer rotation, "Homozygosity mapping of autosomal recessive epilepsy" University of Iowa, Iowa City, Iowa, USA

2006 Roya Ijadi-Maghsoodi, (Medical Student); 12 week summer rotation, "Examination of the role of *LPIN2* in psoriasis" University of Iowa, Iowa City, Iowa, USA
2006 Leah Moellers, (Medical Student); 12 week summer rotation, "Examination of the role of *LPIN2* in psoriasis" University of Iowa, Iowa City, Iowa, USA
2006 Sara Francis, (MSTP candidate, SUMR program); 10 week summer rotation, "Establishment of MLPA technique for *LPIN2*" University of Iowa, Iowa City, Iowa, USA
2006 Samer Bani Hani, MD, (Internal Medicine resident); 8 week rotation, "Application of MLPA for the detection of *LPIN2* deletions and duplications" University of Tennessee, Memphis, Tennessee, USA
2009 Diana Mena, (Medical Student, University of Iowa, Carver college of Medicine); 12 week summer rotation, "Linkage Mapping of an X-linked Blindness Syndrome" Shafallah Medical Genetics Center, Doha, Qatar
2009 Djouher Ait Idir, (Ph.D. candidate, University of Sciences and Technology, Houari Boumediene); 9 week rotation, "Mutation Analysis of Algerian FMF patients" Shafallah Medical Genetics Center, Doha, Qatar
2010 Hala Mint El-Moctar, (Medical Student, Weill-Cornel Medical college - Qatar); 9 month research period "Genetics of autosomal recessive childhood epilepsy" Shafallah Medical Genetics Center, Doha, Qatar

Postdoctoral Fellows Supervisor

2006-2007 Firas Rabi, (Pediatrics, Critical Care Fellow)
2011-2014 Fatma Baoumi Abdalla (PhD Molecular Biology)
2014-2015 Reem Al Olaby (PhD Pharmacology)
2015-2016 Dina Ahram (PhD Molecular Genetics)

C. Other Contributions to Institutional Programs

1988-1989 Cytogenetics journal club (weekly), Indiana University, Indianapolis, IN, USA.
1989-1992 Case Conference (biweekly), Morbidity & Mortality Conference (biweekly), Pediatrics, University of Iowa, Iowa City, IA, USA
1992-1993 Medical Genetics journal club (weekly), University of Iowa, Iowa City, IA, USA
1992-1993 Genetics chromosome and clinical conference (weekly), University of Iowa, Iowa City, IA, USA
1993-2002 Pediatrics journal club & clinical meetings (weekly), JUST, Irbid, Jordan.
1997-1999 Medical Technology seminar, JUST, Irbid, Jordan
2003-2007 Medical Genetics journal club (weekly), University of Iowa, Iowa City, IA, USA
2003-2007 Craniofacial journal club (weekly), University of Iowa, Iowa City, IA, USA
2003-2007 Pediatrics morning report (2 times/week), University of Iowa, Iowa City, IA, USA
2003-2007 Genetics chromosome and clinical conference (weekly), University of Iowa, Iowa City, IA, USA
2007-2009 Autism working group journal club (weekly), Shafallah Center, Doha, Qatar
2007-2014 SMGC journal club (weekly), Shafallah Center, Doha, Qatar
2014-2016 QBRI weekly seminar series, QBRI, Doha, Qatar
2014-2016 QBRI-MGC journal club (weekly), QBRI, Doha, Qatar

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III- SCHOLARSHIP

ORCID ID: 0000-0001-6230-8316
Google Scholar: http://scholar.google.com/citations?user=h20e87YAAAAJ&hl=en
Research Gate: https://www.researchgate.net/profile/Hatem_El-Shanti2

Indices	All	Last 5 years
All citations	**4305**	**2207**
h-index	**34**	**24**
i10-index	**58**	**39**

A. Publications and Creative Work

Peer-Reviewed Publications (Role is outlined in italics)

1) El-Shanti H, Bell WE., Waziri MH. Epidermal nevus syndrome: Subgroup with neuronal migration defects. J Child Neurol 1992; 7:29-34. *Clinical description, and manuscript writing.* Times Cited: **34**
2) Toth PP, El-Shanti H, Eivins S, Rhead WJ, Klein JM. Transient improvement of congenital lactic acidosis in a male with pyruvate decarboxylase deficiency treated with dichloroacetate. J Pediatr 1993; 123:427-430. *Clinical description, and manuscript writing.* Times Cited: **20**
3) Mears AJ, El-Shanti H, Murray JC, McDermid HE, Patil SR. Minute supernumerary ring chromosome 22 associated with cat eye syndrome: Further delineation of the critical region. Am J Hum Genet 1995; 57:667-673. *Clinical description, FISH karyotyping and manuscript writing.* Times Cited: **59**
4) Al-Sheyyab M, El-Shanti H, Ajlouni S, Sawalha D, Daoud AS. The clinical spectrum of Henoch-Schonlein purpura in infants and young children. Eur J Pediatr 1995;154:969-972. *Clinical description, and manuscript writing.* Times Cited: **42**
5) Daoud AS, Al-Kaysi F, El-Shanti H, Batieha A, Obeidat A, Al-Sheyyab M. Neural tube defects in Northern Jordan. Saudi Med J 1996; 17:78-81. *Collection of clinical data, and manuscript writing.* Times Cited: **12**
6) Al-Sheyyab M, El-Shanti H, Ajlouni S, Batieha A, Daoud AS. Henoch-Schonlein Purpura: Clinical experience and contemplation on a streptococcal association. J Trop Pediatr 1996; 42:200-203. *Study design, clinical description, and manuscript writing.* Times Cited: **57**
7) Daoud AS, Al-Sheyyab M, Abu Ekteish F, Obeidat A, Ali AA, El-Shanti H. Neonatal meningitis in northern Jordan. J Trop pediatr 1996; 42:267-270. *Clinical description, and manuscript writing.* Times Cited: **20**
8) Al-Sheyyab M, El-Shanti H, Todd D, Shurman A. Autosomal recessive lamellar ichthyosis and acute lymphoblastic leukemia. Eur J Hum Genet 1996; 4:105-107. *Clinical description, and manuscript writing.* Times Cited: **5**
9) Daoud AS, Al-Sheyyab M, Al-Qudah A, Khouri-Bulos N, El-Shanti H. An outbreak of Poliomyelitis in Jordan: Clinical observations. Indian Pediatr 1997; 34:51-54. *Collection of clinical data, and manuscript writing.* Times Cited: **0**
10) El-Shanti H, Khasawneh M, Hulsburg D, Major H, Patil S. A rare case of a liveborn with free, de novo and partial trisomy 12 and an unusual phenotype. Ann Genet 1997; 40:175-180. *Clinical description, karyotyping, FISH karyotype, and manuscript writing.* Times Cited: **8**
11) El-Shanti H, Omari HZ, Qubain HI. Progressive pseudorheumatoid dysplasia: Report of a family and review. J Med Genet 1997; 34:559-563. *Clinical description, pedigree data analysis, and manuscript writing.* Times Cited: **59**
12) El-Shanti H, Murray JC, Semina EV, Buetow KH, Scherpbier T, Al-Alami J. The assignment of the gene responsible for progressive pseudorheumatoid dysplasia to chromosome six and

examination of COL10A1 as a candidate gene. Eur J Hum Genet 1998; 6:251-256. *Clinical description, genotyping, linkage analysis and manuscript writing.* Times Cited: **22**

13) El-Shanti H, Al-Lahham MB, Batieha A. Normative standards of trunk and limb anthropometric measurements for Jordanian newborns. Saudi Med J 1998; 19:702-706. *Supervising the collection of data, generation of normative standard graphs, preparation of data for analysis, manuscript writing, and corresponding author.* Times Cited: **3**
14) Al-Sheyyab M, El-Shanti H, Daoud AS, Gharabieh N, Ektiesh F. Fanconi anemia and hypothyroidism. Ann Saudi Med 1998; 18:58-59. *Clinical description, and manuscript writing.* Times Cited: **1**
15) Alkhateeb A, Al-Alami J, Leal SM, El-Shanti H. Fine mapping of progressive pseudorheumatoid dysplasia: A tool for heterozygote identification. Genetic Testing 1999; 3:329-333. *Clinical description, pedigree analysis, genotyping, manuscript writing, and senior author.* Times Cited: **4**
16) Jarrah NS, El-Shanti H, Shennak MM, Ajlouni KM. Wolfram syndrome in triplets with newly recognized features. Ann Saudi Med 1999; 19:132-134. *Clinical description, pedigree analysis, and manuscript writing.* Times Cited: **5**
17) Al-Rimawi H, Al-Sheyyab M, Batieha A, El-Shanti H, Abukteish F. The effect of desferroxamine in acute hemolytic anemia of G-6-PD deficiency. Acta Hematologica 1999; 101:145-148. *Clinical description, and manuscript writing.* Times Cited: **8**
18) Majeed HA, Rawashdeh M, El-Shanti H, Qubain H, Khuri-Bulos N, Shahin HM. Familial Mediterranean Fever in children: the expanded clinical profile. Q J Med, 1999; 92:309-318. *Study design, clinical description, genetic analysis, and manuscript writing.* Times Cited: **108**
19) Al-Sheyyab M, Batieha A, El-Shanti H, Daoud A. Henoch-Schonlein purpura and streptococcal infection: a prospective case-control study. Ann Trop Paediatr 1999; 19:253-5. *Study design, clinical description, and manuscript writing.* Times Cited: **47**
20) Hurvitz JR, Suwairi WM, Van Hul W, El-Shanti H, Superti-Furga A, Roudier J, Holderbaum D, Pauli RM, Herd JK, Hul EV, Rezai-Delui H, Legius E, Le Merrer M, Al-Alami J, Bahabri SA, Warman ML. Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. Nat Genet 1999; 23:94-98. *Case contribution, clinical description, haplotype analysis and manuscript writing.* Times Cited: **249**
21) El-Shanti H, Daoud AS, Batieha A. A clinical study of a large inbred kindred with pure familial spastic paraplegia. Brain Dev 1999; 21:478-482. *Clinical description, pedigree analysis, and manuscript writing.* Times Cited: **5**
22) El-Shanti H, Al-Salem M, El-Najjar M, Ajlouni K, Beck J, Sheffield VC, Stone EM. A nonsense mutation in the retinal-specific guanylate cyclase gene is the cause of Leber congenital amaurosis in a large inbred-kindred from Jordan. J Med Genet 1999; 36:862-865. *Clinical description, pedigree analysis, linkage analysis, mutation detection and manuscript writing.* Times Cited: **17**
23) El-Shanti H, Al-Lahham MB, Batieha A. Craniofacial anthropometric measurements in a population of normal Jordanian newborns. J Med Liban 2000; 48:23-28. *Supervising the collection of data, generation of normative standard graphs, preparation of data for analysis, manuscript writing, and corresponding author*. Times Cited: **4**
24) Al-Sheyyab M, Daoud AS, El-Shanti H. Chediak-Higashi syndrome: A report of eight cases from three families. Indian Pediatr 2000; 37:69-75. *Clinical description, analysis of pedigree data, and manuscript writing*. Times Cited: **7**
25) El-Shanti H, Lidral A, Jarrah N, Druhan L, Ajlouni K. Homozygosity mapping identifies an additional locus for Wolfram syndrome on 4q. Am J Hum Genet 2000; 66:1229-1236. *Collecting families, analysis of pedigree data, clinical description, genotyping, haplotype analysis, and manuscript writing*. Times Cited: **93**
26) Sadiq MF, Khabour OF, El-Shanti HE, Samawi HM. The effect of trifluoperazine on the genotoxicity of Bleomycin in cultured human lymphocytes. Drug Chem Toxicol 2000; 23:361-

369. *Supervising the cytogenetic analysis and special cytogenetic techniques, preparation of data for analysis, manuscript review*. Times Cited: **12**

27) Sadiq MF, Zaghal MH, El-Shanti H. Induction of chromosomal aberrations by the rhodium(III) complex cis-[Rh(biq)(2)Cl(2)]Cl in cultured human lymphocytes. Mutagenesis; 2000; 15:375-378. *Supervising the cytogenetic analysis and special cytogenetic techniques, preparation of data for analysis, review of manuscript.* Times Cited: **13**

28) Majeed HA, El-Shanti H, Al-Rimawi H, Al- Masri N. On mice and men: An autosomal recessive syndrome of chronic recurrent multifocal osteomyelitis and congenital dyserythropoietic anemia. J Pediatr. 2000; 137:441-442. *Collecting and updating clinical and pedigree data, manuscript writing and review.* Times Cited: **38**

29) Jarrah N, El-Shanti H, Kheir A, Obeidat FN, Haddidi A, Ajlouni K. Familial disorder of sex determination in seven individuals from three related sibships. Eur J Ped 2000; 159:912-8. *Collecting clinical and pedigree data, analysis of the mode of inheritance, manuscript writing, and corresponding author*. Times Cited: **27**

30) Al-Sheyyab M, Jarrah N, Younis E, Shennak MM, Haddidi A, Awidi A, El-Shanti H, Ajlouni K. Bleeding tendency in Wolfram Syndrome: A newly identified feature with phenotype / genotype correlation. Eur J Ped 2001; 160:243-246. *Collecting clinical and pedigree data, analysis of genotype/phenotype correlation and manuscript review.* Times Cited: **23**

31) Wang K, Pugh EW, Griffin S, Doheny KF, Mostafa WZ, Al-aboosi MM, El-Shanti H, Gitschier J. Homozygosity mapping places the acrodermatitis enteropathica gene on chromosome 8q24.3. Am J Hum Genet 2001; 68:1055-1060. *Collecting clinical, pedigree and biologic data on the largest family in the analysis, analysis of the mode of inheritance, manuscript editing.* Times Cited: **106**

32) Mahafza T, El-Shanti H, Omari H. Raine syndrome: Report of a case with hand and foot anomalies. Clin Dysmorph 2001; 10:227-229. *Clinical description, analysis of pedigree data, manuscript writing and corresponding author*. Times Cited: **19**

33) Majeed HA, Al-Tarawna M, El-Shanti H, Kamel B, Al-Khalialeh F. The syndrome of chronic recurrent multifocal osteomyelitis and congenital dyserythropoietic anemia. Report of a new family and a review. Eur J Ped 2001; 160:705-710. *Clinical description, analysis of pedigree data and manuscript writing and review*. Times Cited: **96**

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Books

None

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Reviews

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*** The work was partially or totally performed at the University of Iowa**
^ The work was partially or totally performed at the Shafallah Medical Genetics Center / QBRI-Medical Genetics Center
 # The work was partially or totally performed at the University of Jordan

B. Areas of Research Interest and Current Projects

1) Gene mapping and positional cloning of human autosomal recessive disorders
2) Genetics of Autism Spectrum Disorder and Intellectual Developmental Disorder
3) Genetics of childhood epilepsy
4) Mutation analysis of autoinflammatory disorder genes, in particular bone and skin inflammation
5) Genetics of prematurity
6) Genetics of the process of nondisjunction

C. Grants Received

Completed

1995 – 1997

JUST / Deanship of Scientific Research
Molecular studies of G-6-PD deficiency
This study aimed at the identification of mutations of the G-6-PD gene in the Jordanian population
Role: PI

1995 – 1997

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JUST / Deanship of Scientific Research
Genetic epidemiology of multifactorial disorders & traits,
This study aimed at the identification of prevalence rates of selected birth defects in the Jordanian population and the establishment of a nucleus for a birth defect registry in a teaching hospital in Northern Jordan.
Role: PI

1995 – 1998

JUST / Deanship of Scientific Research
Normative standards of growth & clinical anthropometric measurements for Jordanian children
This study aimed at developing normative standard curves for some clinical craniofacial, limb and trunk anthropometric measurements in Jordan newborns and toddlers.
Role: PI

1996 – 1999

JUST / Deanship of Scientific Research
Gene mapping of autosomal recessive familial spastic paraplegia
This study aimed at the identification of the genetic locus for an autosomal recessive form of pure familial spastic paraplegia.
Role: PI

1997 – 1999

Higher council for Science & Technology, Amman, Jordan
Gene mapping of autosomal recessive disorders & traits
This study was a preliminary study for the identification and characterization of inbred families from Jordan segregating autosomal recessive disorders and predicting the feasibility of taking a homozygosity approach for mapping the responsible genes to genetic loci.
Role: PI

1 RO3 DE12533-01 Lidral (PI) 1998 – 2000
NIH / NIDCR
Homozygosity mapping of genes for craniofacial anomalies.
This study aimed at identifying families segregating craniofacial defects and applying homozygosity mapping strategies to identify the loci for the responsible genes.
Role: Co-Investigator

Chaire Internationale de Recherche “Blaise Pascal” 1999 – 2001
L'etat et de la region d'ile'de France € 200,000
Homozygosity mapping of autosomal recessive disorders
This study aimed at the identification of genetic loci responsible for the etiology of various autosomal recessive disorders in an inbred population (Jordan)
Role: PI

2001 – 2002

Higher Council for Science and Technology, Amman, Jordan
Population supplementation of folic acid in Jordan
This study aimed at the feasibility of countrywide supplementation of the diet of Jordanians with folic acid, by adding it to the milled wheat with the final goal of reducing birth defects.
Role: PI

2002 – 2002

JUST / Deanship of Scientific Research

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Folic acid status amongst Jordanian women
This study aimed at the estimation of the proportion of Jordanian women who are at risk for deficiency of folic acid.
Role: PI
2002 – 2002
JUST / Deanship of Scientific Research
Heterozygote identification of the FMF gene among Jordanians
This study aimed at the identification of the carrier frequency of the autosomal recessive condition Familial Mediterranean Fever in the Jordanian population with calculation of disease incidence and prevalence.
Role: 2002 – 2002
JUST / Deanship of Scientific Research
Allele frequency of the thermolabile variants of MTHFR gene
This study aimed at the identification of the genotypes of the MTHFR gene in the Jordanian population and the correlation between the folic acid stores and the genotypes with reflection on risk for birth defects.
Role: Principal Investigator

CMN # 1615 El-Shanti (PI) 2004 – 2005
Children Miracle Network $ 40,000
Role of *LPIN2* in CRMO.
This study examines the role of the gene LPIN2, which is responsible for the Majeed syndrome (bone inflammation is a component), in isolated chronic recurrent multifocal osteomyelitis.
Role: PI, no salary support.

El-Shanti (PI) 1/1/05 – 12/31/06
Carver Medical Research Initiative grant $ 30,000
Investigation of the Role of LPIN2 in the etiology of psoriasis
This study aims at the examination of *LPIN2* in the etiology of psoriasis
Role: PI, 10 % effort (no salary support)

El-Shanti (PI) 7/1/06 – 6/30/07
General Clinical Research Center grant $ 10,000
Recruitment of patients with idiopathic generalized epilepsy
This study aims at recruiting patients with specialized forms of epilepsy for genetic studies
Role: PI, 5 % effort (no salary support)

U50 CCU713238 Romitti (PI) 9/1/02 – 8/31/07
CDC/NCBDDD $ 774,998
Iowa Center of Excellence for Birth Defects Research and Prevention
A Center of Excellence to investigate genetic and environmental risk factors for major birth defects. There is no Funding overlap
Role: Co-Investigator, 5% effort (5% salary support)

U50/CCU725183-01 Romitti (PI) 9/30/05 – 9/29/07
CDC/NCBDDD $ 461,975 / year
Iowa Stillbirth Surveillance Project
Pilot project to evaluate the feasibility of expanding the Iowa Registry for Congenital and Inherited Disorders to conduct population-based surveillance on the occurrence of fetal deaths in the state of Iowa and to serve as a registry for etiologic studies of fetal deaths and as a resource for prevention programs that aim to reduce the occurrence of fetal deaths. There is no funding overlap
Role: Co-Investigator, 5% effort (5% salary support)

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1 RO3 AR051130-01A1 Ferguson (PI) 7/1/05 – 6/30/08
NIH / NIAMS $ 150,000
Genetic basis of chronic multifocal osteomyelitis
This study aims at the examination of the genetic causes of isolated chronic recurrent multifocal osteomyelitis. There is no funding overlap.
Role: Collaborator, 5 % effort (no salary support)

1 R21 AR053924-01 El-Shanti (PI) 7/15/06 – 5/31/08
NIH/NIAMS $ 169,000/ year, direct costs
Examination of the role of LPIN2 variations in skin and bone inflammation
This study aims at the examination of the role played by *LPIN2*, the gene responsible for the autoinflammatory disorder Majeed syndrome, in the etiology of psoriasis, pustulosis and CRMO. There is no funding overlap.
Role: PI, 10% effort (10% salary support)

NAF El-Shanti (PI) 7/1/2007 – 6/30/2008
National Ataxia Foundation $ 15,000
Mapping of a distinct autosomal dominant ataxia gene
This study aims at the localization of an autosomal dominant ataxia gene in a single family using genome wide SNP arrays
Role: PI, 5% effort (no salary support)

SF Intramural Funding El-Shanti (PI) 7/1/2007 – 3/31/2014
Shafallah Foundation, Shafallah Medical Genetics Center
Study of the genetic and environmental etiologic factors in Autism Spectrum Disorder
This study aims at the identification of variants in novel genes that play a role in the genetic predisposition to Autism Spectrum Disorder, as well as confirm the role of known genes.
Role: PI

NPRP09-374-3-092 El-Shanti (PI) 12/15/2010 – 2/1/2015
Qatar National Research Fund $ 1,037,040
Mutation analysis and genotype/phenotype correlation studies in Familial Mediterranean Fever
This study aims at the identification of the spectrum of *MEFV* mutations in Arabic FMF patients and the identification of the genotype/phenotype correlation pattern
Role: PI, 10% effort

NPRP09-215-3-049 Zalloua (PI) 12/15/2010 – 2/1/2015
Qatar National Research Fund $ 1,049,100
Genetic determinants of type 2 diabetes
This study aims at the identification of the genetic factors contributing to the etiology of adult onset diabetes mellitus
Role: Co-PI, 10% effort

NPRP09-367-3-087 Al-Alami (PI)(Kambouris) 12/15/2010 – 3/14/2015
Qatar National Research Fund $ 890,591
Genetic factors in autosomal recessive disorders among consanguineous Qatari families
This study aims at utilizing homozygosity mapping for the identification genes responsible for rare autosomal recessive disorders amongst Arabic populations
Role: Co-PI, 10% effort

NPRP09-374-3-091 El-Shanti (PI) 12/15/2010 – 5/1/2015

Qatar National Research Fund $ 1,021,260
Characterization of the LPIN2 gene and its protein and examination of its role in psoriasis
This study aims at characterization of *LPIN2* and the exploration of its etiologic role in psoriasis
Role: PI, 10% effort

NPRP09-943-3-246 Yunis (PI) 12/15/2010 – 6/15/2015
Qatar National Research Fund $ 998,700
The Consanguinity Factor in the Genetics of Prematurity
This study aims at the dissection of the etiologic role of consanguinity in premature onset of labor
Role: Co-PI, 10% effort

NPRP-6-359-3-095 El-Shanti (PI) 9/1/2014 – 3/15/2016
Qatar National Research Fund $ 1,005,000
Gene Identification in Autosomal Recessive Familial Epilepsy
This study aims at the identification of the autosomal recessive genetic factors contributing to the etiology of familial epilepsy
Role: LPI, 10% effort

QBRI Intramural Funding El-Shanti (PI) 9/1/2014 – 3/1/2016
Qatar Foundation, Qatar Biomedical Research Institute, Medical Genetics Center
Gene discovery in Autism Spectrum Disorder and associated comorbidities
This study aims at the identification of genetic variants that plays a role in the genetic predisposition to Autism Spectrum Disorder and its associated comorbidities, particularly epilepsy and intellectual developmental disorder.
Role: PI

Active

D. Invited Lectures

Lectures, Workshop Presentations and Media Presentation

1994 Chromosomal abnormalities, The Jordanian Medical Association, Amman, Jordan.
1994 Invited panelist, Jordanian Television panel discussion on “premarital testing & counseling”
1995 Introduction to genetic disorders, Workshop on Genetics & Genetic diseases, Business & Professional Women Club & UNICEF, Amman, Jordan.
1995 Dissection of multifactorial disorders, Fourth Jordanian French Medical Congress on Genetics & Genetic diseases, Jordanian French Medical Association, Amman, Jordan.
1995 Medical Genetics in Jordan, Service du Genetique Medicale, INSERM U393, Paris, France.
1996 Mapping of autosomal recessive disease genes utilizing inbred Jordanian families, Workshop on Gene mapping in isolated populations, American Society of Human Genetics Annual Meeting, San Francisco, California, USA.
1996 Mapping of autosomal recessive disorder genes utilizing inbred populations, Laboratory of Statistical Genetics, Rockefeller University, New York, NY, USA.
1997 Guest, Jordanian Radio question & answer on “genetic disorders” (2 episodes)

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1997 Chromosomal disorders & prenatal diagnosis, The Jordanian Society of Obstetricians & Gynecologists, Amman, Jordan.
1997 Invited panelist, Jordanian Television panel discussion on "biological cloning" (2 episodes)
1997 Chromosomal disorders, The Jordanian Medical Association, Zarqaa, Jordan.
1997 Invited panelist, Jordanian Radio panel discussion on "genetic diseases & birth defects"
1998 Molecular methods in genetic disorders & their diagnosis, workshop on the application of molecular biology in diagnostics & forensics, Al-Amal Center, Amman, Jordan.
1998 Neurogenetic disorders, The Jordanian Society for Neuroscience, Amman, Jordan.
1998 Introduction to inborn Errors of Metabolism, Neonatology seminars at Jordan University Hospital & Swiss Tropical Institute, Amman, Jordan.
1999 Molecular biology in forensic medicine, Workshop on Forensic Medicine, Amman, Jordan
1999 Genetics of Wolfram syndrome amongst Jordanians, National Center for Diabetes, Endocrinology & Genetics conference on Diabetes & Obesity, Amman, Jordan.
1999 Genetics for the clinician, Workshop. The National Center for Diabetes, Endocrinology & Genetics, Amman, Jordan.
2000 Mutation analysis of the MEFV gene in the Jordanian population, R.K.H. pediatric symposium "Molecular Genetics for the Clinician", Riyadh, Saudi Arabia.
2000 A family with the True hermaphroditism, seminar on Sex determination problems. Necker-Enfants Malades, Paris, France.
2000 Conférence de clôture, Chaire International de Recherche "Blaise Pascal", Ecole de Médecine, Paris, France. "Genetic disorders amongst the Jordanians; Strategies towards prevention.
2000 Progressive pseudorheumatoid dysplasia & chronic recurrent multifocal osteomyelitis; Genetic disorders mimicking rheumatic disorders, Laboratoire d'Immuno-Rheumatologie, Faculté de Médicine de Marseille, Marseille, France.
2000 Genetic disorders amongst the Jordanians: Strategies towards prevention, Second Pan Arab Congress of Diabetes & Endocrinology & the 10th International Clinical Genetics Seminar, Amman, Jordan.
2000 Temporary advisor for the World Health Organization. International collaborative research on craniofacial anomalies. Geneva, Switzerland.
2001 Guest speaker, Jordanian TV show, "consanguinity", Amman, Jordan.
2001 Temporary advisor for the World Health Organization. International collaborative research on craniofacial anomalies. WHO meeting on the prevention of craniofacial anomalies. Park City, Utah, USA.
2001 Rare syndromes, Jordan Pediatric Society, Amman, Jordan.
2001 Guest, Jordan T.V. program "Your Physician," issues in pediatrics, Amman, Jordan.
2002 Recombinant DNA technology, National Center for Diabetes, Endocrinology & Genetics, Amman, Jordan.
2002 The different approaches to understand the genetic component of Diabetes etiology, 3rd meeting on Genetics & Registry of Diabetes in Saudi Arabia, Riyadh, Saudi Arabia.
2002 The burden of genetic disorders on the Jordanians, Ministry of Health & WHO, Workshop on community control of genetic disorders, Amman, Jordan.

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2002 Folic acid status amongst Jordanian women of childbearing age: a preliminary study. National Center for Diabetes, Endocrinology & Genetics, Amman, Jordan.

2002 Genetics of FMF, FMF2002, Third international conference on familial mediterranean fever & hereditary inflammatory disorders, La Grande Motte, France.

2003 Les Tables Rondes des chaires internationales de recherche "Blaise Pascal", Continuité et Evolution, Paris, France, May 2003.

2005 The role of cytogenetics in infertility and recurrent fetal loss, Al-Amal Maternity Hospital, Amman, Jordan.

2005 Molecular and cytogenetic testing in preimplantation genetic diagnosis, Workshop and round table discussion (2 days) for assisted reproductive technology and genetics unit, Al-Amal Maternity Hospital, Amman, Jordan.

2005 Multiple approaches to the identification of the genetic factors involved in chronic recurrent multifocal osteomyelitis (CRMO), 12th European Pediatric Rheumatology congress, Versailles, France, September 15-18, 2005.

2005 The genetics of chronic recurrent multifocal osteomyelitis (CRMO), FMF and Beyond: The Fourth International Congress on the Systemic Autoinflammatory Diseases. Bethesda, MD, November 6-10, 2005

2005 Lecture "The health care of a child with Down syndrome", The 2nd Annual Pediatric Review Course, Amman, Jordan, November 17-18, 2005.

2005 Workshop "The work-up of child with multiple congenital anomalies and dysmorphism", The 2nd Annual Pediatric Review Course, Amman, Jordan, November 17-18, 2005

2006 Lecture "The Utilization of Mendelian disorders genes for the identification of genes responsible for multifactorial disorders", King Faisal Specialty Hospital and Research Center, Riyadh, Saudi Arabia, September 5, 2006.

2006 Lecture "Research interests, from Mendelian disorders to multifactorial disorders", King Fahad National Children's Cancer Center, Riyadh, Saudi Arabia, September 5, 2006

2006 commentator, Fellowship seminars, University of Iowa, Iowa City, IA.

2006 Panelist, Panel discussion on the "Research and Infrastructure Needs to Strengthen Newborn screening in the Region". Conference on Strengthening Newborn Screening in the Middle East and North Africa, Marrakech, Morocco, November 13-15

2007 Lecture "Ambiguous genitalia in the newborn," NICU enrichment day, Children's Hospital of Iowa, Iowa City, Iowa, USA, May 2007

2007 Lecture "Implications of Genetics in Prevention and Care of Children with Special Needs," The 2nd Annual International forum, Shafallah Center for Children with Special Needs, Doha, Qatar, April 2007

2007 Guest, Al-Jazeera International TV program "Everywoman," special issue on autism, Doha, Qatar, April 2007

2008 Lecture "Healthcare issues in Down syndrome," Shafallah Center for Children with Special needs weekly educational lecture, Doha, Qatar, February 2008

2008 Lecture "The genetics of Chronic Recurrent Multifocal Osteomyelitis." The Fifth International Congress on FMF and Systemic Autoinflammatory Diseases, Rome, Italy, April 2008

2008 Lecture "Laboratory Genetics for the Clinician," Tawam Hospital in Association with Johns Hopkins Medicine, The 2nd Annual International Pediatric Conference "Hot Topics in Pediatrics," Al-Ain, UAE, May 2008

2008 Presentation "Majeed syndrome," New therapies for CRMO, SAPHO syndrome and Behcet disease, Bethesda, MD, USA September 3-5

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2008 Presentation "Familial Mediterranean Fever and Other Autoinflammatory Disorders," The 2nd Al-Ain International Genetics Conference, Al-Ain, UAE, October 2008

2008 Presentation "Genes, Environment and Health," The biannual National Collaborative Perinatal Neonatal Network (NCPNN) meeting, American University in Beirut, Beirut, Lebanon, November 2008

2008 Presentation "Consanguinity and Health," The biannual National Collaborative Perinatal Neonatal Network (NCPNN) meeting, American University in Beirut, Beirut, Lebanon, November 2008

2009 Presentation "Gene Hunting in the Arab World," The third International Conference on Disability and Rehabilitation; "Scientific Research in the Field of Disability." Riyadh, Saudi Arabia, March 2009

2009 Speaker "Strategic Responses to Autism Challenge," Luncheon Briefing on Autism: Awareness, Implication, and Responses. United Nations Headquarters, New York, NY, USA, April 2nd, 2009 (World Autism Awareness Day)

2009 Speaker, Department of Public Information/NGO Briefing "Autism and Human Rights: Understanding and Safeguarding the Rights of People with Autism" in observance of World Autism Awareness Day 2009. United Nations Headquarters, New York, NY, USA, April 2, 2009

2009 Speaker, The 17th Conference of the Union of Arab Pediatricians & the 5th Meeting of the Pan Arab Neonatology Forum "The Burden of Birth Defects and Genetic Disorders and the Strategies for Prevention or Reduction of Their Toll" Amman, Jordan, May 5-8, 2009

2009 Speaker, The 17th Conference of the Union of Arab Pediatricians & the 5th Meeting of the Pan Arab Neonatology Forum "The Genetics of Preterm Labor" Amman, Jordan, May 5-8, 2009

2009 Lecturer, The 6th Annual Pediatric Review Course "Ambiguous Genitalia in the Newborn; Laboratory Genetics for the Pediatrician; Introduction to Metabolic Disorders; Burden of Genetic Disorders and Birth Defects and Strategies for Prevention" Amman, Jordan, November 5-6, 2009

2009 Speaker, the 2nd Qatar Genetics symposium: Advances in Medical Genetics "Gene Identification in Arabic Population" Doha, Qatar, October 31, 2009

2010 Speaker, the 3rd PanArab Human Genetics Conference: Post-Conference workshop "The Utilization of High Through-put SNP Genotyping for Gene Discovery in Genetic Disorders" Dubai, United Arab Emirates, March 13-15, 2010

2010 Speaker, Séminaire d'Immunologie, d'Hématologie et de Rhumatologie Pédiatriques - Pierre Royer "The Genetics of Bone and Skin Inflammation" Paris, France, March 26, 2010

2010 Speaker, Diplome Interuniversitaire de Rhumatologie Pediatrique "The Genetics of Bone and Skin Inflammation" Lyon, France, March 27, 2010

2010 Speaker, the 3rd MENA Newborn Screening conference: Prevention, Screening and Treatment of Developmental Disorders in the MENA region "Genetic and Metabolic Approaches to Screening for Developmental Disorders Beyond the Newborn Period" Doha, Qatar, April 26-29, 2010

2010 Speaker, Autism Awareness symposium "Study of the Genetics of Autism in Qatar" Doha, Qatar, May 1, 2010

2010 Lecturer, The 7th Annual Pediatric Review Conference "Autoinflammatory Disorders; Clinical Profile of Autism; Genetics of Autism" Amman, Jordan, October 28-29, 2010

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2011 The first Middle East Orphan Europe Neurometabolic Course "The molecular basis of childhood ataxia" Doha, Qatar, January 20-22, 2011

2011 Weil-Cornell Medical College – Qatar weekly research seminar, "Research program for gene discovery in neurodevelopmental disorders" Doha, Qatar

2011 Neurogenetics Workshop 2011, Microcephaly & Associated Brain Malformations "Genetic causes of Epilepsy in Arab populations" Doha, Qatar, April 16-17, 2011

2011 The First Middle East Lysosomal Storage Disease Forum "Molecular Aspects and Diagnosis of LSDs" Doha, Qatar, November 23-24, 2011

2012 Lecturer, the 8th Annual Pediatric Review Conference "Rare syndromes; Prenatal Diagnosis" Amman, Jordan, February 23-24, 2012

2012 Lecture "Prenatal Diagnosis," Tawam Hospital in Association with Johns Hopkins Medicine, The 6nd Annual International Pediatric Conference "Best Practice in Modern Pediatrics," Al-Ain, UAE, March 2012

2013 Lecture "Utilization of Advanced Molecular Genetic Technology in Pediatrics: Experience from Qatar," Hamad Medical Corporation in Partnership with Sick Kids International, The 3rd Annual Child Health Research Day, Doha, Qatar, March 2013

2014 Lecturer, the 9th Annual Pediatric Review Conference "Rare syndromes; Multiple congenital defects; chromosomal abnormalities" Amman, Jordan, February 27-28, 2014

2014 Seminar "Genetic and Genomic Approaches to the Elucidation of Autism Spectrum Disorder Etiology," Qatar University Seminar Series, Doha, Qatar, November 2014

2015 Presentation "The Genetic Etiology of Autism Spectrum Disorder: Research Experience from Qatar," Sidra Symposia Series – Autism Spectrum Disorder, Doha, Qatar, February 2015

2015 Lecturer, the 10th Annual Pediatric Review Conference "Pediatricians and ASD; The genetics of polycystic kidney disease; Rare syndromes" Amman, Jordan, February 26-27, 2015

2015 Presentation "The Genetic Etiology of Autism Spectrum Disorder" Carnegie Mellon University - Qatar, Doha, Qatar, March 2015

2015 Presentation "The Genetics of Childhood Epilepsy" The 20th Congress of the Union of Arab Pediatric Societies and The 15th International Conference of the Jordan Pediatric Society, Amman, April 2015

2015 Presentation "The Genetic Etiology of Autism Spectrum Disorder" The 20th Congress of the Union of Arab Pediatric Societies and The 15th International Conference of the Jordan Pediatric Society, Amman, April 2015

2015 Seminar "Genetic and Genomic Approaches to the Elucidation of Autism Spectrum Disorder Etiology," Institut National de la Santé et de la Recherche Médicale (INSERM); Université d'Evry Val d'Essonne, Evry, France

2016 Presentation "The Role of Next Generation Sequencing in Clinical Practice" Laboratory Medicine: Current Aspects and Future Prospects, Dammam, October 2016

2016 Presentation "Molecular Karyotyping in Clinical Practice" Laboratory Medicine: Current Aspects and Future Prospects, Dammam, October 2016

2017 Lecturer "Rare syndromes and genetic diseases" the 12th Annual Pediatric Review Conference, Amman, Jordan, March 9-10, 2017

2017 Presentation "Autoinflammation: The molecular pathophysiology of the innate immunity" The 4th International Congress of the Jordan Rheumatism Society, Amman, Jordan, April 4-6, 2017

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2017 Presentation "Spectrum of mutations and carrier frequency of Familial Mediterranean Fever gene among Arabs" The 6th International Jordanian Congress of Allergy and Immunology and the 2nd Pediatric Rheumatology Arab Group Meeting, Amman, Jordan, July 26-28, 2017

Local Conference presentations and organization

1994 Association of Infantile Glaucoma & Turner Syndrome: Clinical & Epidemiologic Observations, Eight Scientific day, Jordan University of Science & Technology, Irbid, Jordan.

1995 coordinator, Fourth Jordanian French Medical Congress on Genetics & Genetic diseases, Jordanian French Medical Association, Amman, Jordan

1996 Normative standards of anthropometric measurements in Jordan, The First International Medical Congress, Amman, Jordan.

1997 Mapping of the Autosomal Recessive Progressive Pseudorheumatoid Dysplasia & Examination of a Candidate Gene, Eleventh Scientific day, Jordan University of Science & Technology, Irbid, Jordan.

1997 Mapping of autosomal recessive disease causing genes utilizing inbred Jordanian families, Eighth Arab conference of clinical biology & first Jordanian conference of medical laboratory sciences. Amman, Jordan.

1997 Mapping of the autosomal recessive progressive pseudorheumatoid dysplasia: Evidence of homogeneity, Sixth conference of Jordan Pediatric Society. Amman, Jordan.

1998 Mapping & gene identification of rare autosomal recessive disorders utilizing inbred Jordanian families, Twelfth Scientific day, Jordan University of Science & Technology, Irbid, Jordan.

1998 Member, scientific committee, First International Conference of the Faculty of Medicine, University of Jordan & The Third International Conference of the German-Arab Medical Association, Amman, Jordan.

1999 Member, scientific committee, Seventh conference of the Jordanian Pediatric Society in association with the Cystic Fibrosis Association, Amman, Jordan

1999 Al-Alami J, El-Shanti H: Screening for biotinidase deficiency in Jordanian children suffering from seizures: results of a one-year pilot study, The Thirteenth Scientific day, Jordan University of Science & Technology, Irbid, Jordan.

1999 Local abstract, The Seventh conference of Jordanian Pediatric Society/Cystic Fibrosis Association, Amman, Jordan. "Tayeh M, Al-Alami J, Al-Sheyyab M, El-Shanti H: Linkage analysis of a large inbred family with autosomal recessive vitamin B12 deficiency"

1999 Local abstract, The Seventh conference of Jordanian Pediatric Society/Cystic Fibrosis Association, Amman, Jordan. "AlKhateeb A, Al-Alami J, El-Shanti H: DNA based testing for carrier identification for progressive pseudorheumatoid dysplasia to be used in premarital counseling.

1999 Local abstract, The Seventh conference of Jordanian Pediatric Society/Cystic Fibrosis Association, Amman, Jordan. "Abu-Dalo R, Abdul-Majeed H, Al-Alami J, El-Shanti H: Clinical, immunological & molecular studies on period fever syndromes.

2000 Member, preparatory & scientific committees, Second Pan Arab Congress of Diabetes & Endocrinology & the 10th International Clinical Genetics Seminar, Amman, Jordan

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2000 Mutation Analysis, Genotype - Phenotype Correlation & Carrier Frequency of FMF in the Jordanian Population, The Eighth conference of Jordanian Pediatric Society, Amman, Jordan.

2000 Local abstract, Second Pan Arab Congress of Diabetes & Endocrinology & the 10th International Clinical Genetics Seminar, Amman, Jordan. El-Shanti HE, Al-Khateeb M, Abu Rabihah Z, Tayeh M, Majeed HA: Mutation Analysis of FMF in the Jordanian Population.

2000 Homozygosity Mapping Identifies a New Locus for Wolfram Syndrome, Second Pan Arab Congress of Diabetes & Endocrinology & the 10th International Clinical Genetics Seminar, Amman, Jordan.

2001 Contributor, London Dysmorphology & Neurogenetics Database.

2001 Organizer, Workshop on Molecular Biology, National Center for Diabetes, Endocrinology & Genetics, Amman, Jordan.

2003 Pediatrics Grand Rounds, Multiple approaches to the identification of the genetic factors involved in chronic recurrent multifocal osteomyelitis (CRMO). Carver College of Medicine, University of Iowa, Iowa City, IA, USA.

2004 Pediatrics research day, *LPIN2* plays a role in bone autoinflammation, Carver College of Medicine, University of Iowa, Iowa City, IA, USA.

2004 The identification of the parent of origin of the extrachromosome in trisomy 21. Leah Franck. 36th Annual Medical Student Research Day, CCOM, Iowa City, Iowa, USA.

2004 Role of *LPIN2* in psoriasis. Luis Ochoa. 36th Annual Medical Student Research Day, CCOM, Iowa City, Iowa, USA.

2004 Search of the genetic basis of murine chronic multifocal osteomyelitis. Amar Mahgoub. 36th Annual Medical Student Research Day, CCOM, Iowa City, Iowa, USA.

2005 Research interests minitalk, Genetics PhD program research day and poster session. Carver College of Medicine, University of Iowa, Iowa City, Iowa, USA

2005 Pediatrics Research Presentation, Multiple approaches to the identification of the genetic factors involved in chronic recurrent multifocal osteomyelitis (CRMO). Carver College of Medicine, University of Iowa, Iowa City, Iowa, USA

2005 Pediatrics research day, A distinct autosomal recessive ataxia maps to chromosome 12 in an inbred family from Jordan, Carver College of Medicine, University of Iowa, Iowa City, IA, USA

2005 Member of the organizing committee, David W. Smith
26th Annual Workshop on Malformation and Morphogenesis, Iowa City, IA; August 2-6, 2005

2007 ICBDRP site visit, Molecular Genetics of Human Nondisjunction, Iowa City, IA; May 22, 2007

2007 Annual orthopedics resident research day, evaluation of two research reports, Department of Orthopedics and Rehabilitation, University of Iowa, Iowa City, IA; June 8, 2007

2008 Workshop on the Genetics of Autism, The third International Forum of the Shafallah Center, Doha, Qatar; April 20-22, 2008

2008 UN world focus on autism forum, New York, NY, USA; September 26, 2008

2009 World focus on autism forum, New York, NY, USA; September 22, 2009

2010 Vice-Chairman of the local organizing committee and local scientific committee and member of the international scientific and organizing committee, the 3rd MENA Newborn Screening conference: Prevention,

Screening and Treatment of Developmental Disorders in the MENA region, Doha, Qatar, April 26-29, 2010

2010 Qatar Foundation Annual Research Forum, Doha, Qatar; December 12-13, 2010

2011 Qatar Foundation Annual Research Forum, Doha, Qatar; November 20-22, 2011

2014 Advances in Neurogenetics Research Symposium, Doha, Qatar; March 22, 2014. Identifying Autism Spectrum Disorder Causing Mechanisms: Experience from Qatar.

2014 Panelist, Islamic Ethics in the Era of Genomics, Research Center for Islamic Legislation & Ethics (CILE), Doha, Qatar; October 2, 2014

2014 Organizing committee member, IBRO-MENA Neuroscience School and symposium, International Brain Research Organization – Middle East and North Africa Chapter, Qatar Biomedical Research Institute, Doha, Qatar

2014 IBRO-MENA Neuroscience symposium, Doha, Qatar; December 16-17, 2014. Genomic Approaches to the Elucidation of ASD etiology

2015 Organizing committee member, IBRO-MENA Neuroscience School and symposium, International Brain Research Organization – Middle East and North Africa Chapter, Qatar Biomedical Research Institute, Hamad Bin Khalifa University, Doha, Qatar

2015 Presentation “The Basics of DNA and Gene Identification in Human Disease” IBRO-MENA Neuroscience School and symposium, International Brain Research Organization – Middle East and North Africa Chapter, Qatar Biomedical Research Institute, Hamad Bin Khalifa University, Doha, Qatar

2015 Presentation “The Identification of De Novo and Rare Inherited DNA Variants in Autism Spectrum Disorder” Sidra Symposia Series – Functional Genomics Symposium, Doha, Qatar, December 2015

2016 Presentation “Identification of *de novo* and Rare Inherited DNA Variants in Autism Spectrum Disorder” 13th Middle East Metabolic Group (MEMG) meeting, Amman, Jordan, October 2016

2016 Presentation “*CLDN10* Mutations in Hypokalemia, Ichthyosis, Hypohidrosis, Alacrimia, and Xerostomia” 13th Middle East Metabolic Group (MEMG) meeting, Amman, Jordan, October 2016

E. Pending Decisions

IV. SERVICE

A. Offices held in professional organizations

Editorship

1999- 2009	Editorial Board Member, Journal of Biomedicine & Biotechnology
2001-2002	Editorial Board Member, Saudi Medical Journal
2005-now	Editor and curator, Majeed syndrome / *LPIN2* webpage in INFEVERS internet database. http://fmf.igh.cnrs.fr/ISSAID/infevers/search.php?n=7
2006-now	Advisory Board Member, Jordan Medical Journal
2008-2011	Editorial Board Member, Human Genomics and Proteomics
2011-now	Advisory Board Member, Journal of Royal Medical Services (Jordan)
2014-now	Senior Editor, Advances in Rare Diseases
2015-now	SOJ, Pediatrics and Child Care (Open Access)

Reviewer

1994	Jordan Medical Journal
1998	Saudi Medical Journal
1999	Journal of Medical Genetics
2000	Journal of Royal Medical Services (Jordan)
2002	National Research Council and Institute of Medicine, USA "Reducing Birth Defects: Meeting the Challenge in the Developing World (2003)"
2004	Journal of Royal Medical Services (Jordan) X2
2005	Saudi Medical Journal; Journal of Royal Medical Services (Jordan); Birth Defects Research, (Journal of the Teratology Society)
2005	David W. Smith 26th Annual Workshop on Malformation and Morphogenesis
2006	Saudi Medical Journal; Journal of Royal Medical Services (Jordan); Birth Defects Research, (Journal of the Teratology Society); American Journal of Medical Genetics; European Journal of Human Genetics
2006	General Clinical Research Center grant review
2007	American Journal of Medical Genetics; Clinical Orthopedics and Related Research; Journal of Pediatric Neurology; Human Mutation; European Journal of Clinical Investigation; Neurosciences; Medical Principles and Practice (Kuwait); Journal of Clinical Periodontology; PLoS Genetics
2007	Grant review, "Building Interdisciplinary Research Careers in Women's Health," Washington University School of Medicine in St. Louis, Missouri
2007	Abstract review, NBDPS, Centers for Disease Control and Prevention, Birth Defects Branch
2007	External grant reviewer, Italian Telethon Foundation, Milan, Italy
2008	PLoS Genetics; Scandinavian Journal of Rheumatology; American Journal of Medical Genetics; Psychiatry Research; Journal of Clinical Rheumatology; American Journal of Case Reports; Tissue Antigens
2008	Abstract review, NBDPS, Centers for Disease Control and Prevention, Birth Defects Branch
2008	External grant reviewer, Italian Telethon Foundation, Milan, Italy
2008	External grant reviewer, Agency for Science, Technology and Research (A*STAR) and Biomedical Research Council (BMRC), Singapore
2009	Arthritis Care & Research; Journal of Royal Medical Services (Jordan); American Journal of Human Genetics; Molecular Biology Reports; Human

	Genetics; American Journal of Medical Genetics; Rheumatology; Molecular Biology Reports; Pediatrics (AAP)
2009	External grant reviewer, Italian Telethon Foundation, Milan, Italy
2009	External Evaluator for Academic Promotion, Arab Gulf University, Bahrain
2009	Abstract review, NBDPS, Centers for Disease Control and Prevention, Birth Defects Branch
2010	Journal of Neurology; Molecular Biology Reports; DNA and Cell Biology; Nature Genetics; American Journal of Human Genetics; Saudi Medical Journal – Neurosciences
2010	2011 Pediatric Academic Societies workshop review
2010	Qatar Foundation Annual Research Forum 2010
2011	Internal research grants, College of Arts and Science, Qatar University, Doha
2011	Journal of Royal Medical Services (Jordan); British Journal of Dermatology
2011	Bentham Science Publishers, e-book series
2011	Qatar Foundation Annual Research Forum 2011
2012	British Journal of Dermatology, Australasian Journal of Dermatology, Arthritis care & Research
2013	Molecular Biology Reports X 3; Saudi Medical Journal; Medical Science Monitor; Expert Review of Clinical Immunology; Clinical Genetics; Cytotechnology X 2;
2013	External grant Reviewer, Jazan University (Deanship of Scientific Research), Saudi Arabia / Society of the Advancement of Science and Technology in the Arab World (SASTA)
2014	Clinical Case Reports; Molecular Diagnosis and Therapy; BMC Medical Genetics; European Journal of Human Genetics; Rheumatology; Pediatric Radiology; Clinical Rheumatology; American Journal of Nephrology
2015	Medical Science Monitor X 4; Human Molecular Genetics X 2; Rheumatology; Clinical Case Reports; Molecular Genetics and Metabolism; Journal of Cellular and Molecular Medicine
2016	External promotion reviewer, Al-Musol University, Al-Musol, Iraq
2016	European Journal of Medical Genetics X 2
2017	Asian Pacific Journal of Cancer prevention; Jordan Medical Journal; Cognitive and Behavioral Neurology; BMC Medical Genetics
2017	External grant Reviewer, Philadelphia University (Deanship of Scientific Research), Amman, Jordan
2017	External grant Reviewer, Scientific Research Support Fund, Amman, Jordan

Collegiate and University committees and service

1993-1997	Member, Committee for Medical School Library, Jordan University of Science & Technology, Irbid, Jordan
1993-1994	Member, Mortality & Morbidity Committee, School of Medicine, Jordan University of Science & Technology, Irbid, Jordan
1996-1997	chairman, Committee for Medical School Library, Jordan University of Science & Technology, Irbid, Jordan
1998-1999	Member, committee for Scientific Research, School of Medicine, Jordan University of Science & Technology, Irbid, Jordan
2000-2001	Member, committee for Medical School curriculum, Jordan University of Science & Technology, Irbid, Jordan
2000-2001	Member, committee of Medical Records, King Abdullah University Hospital, Jordan University of Science & Technology, Irbid, Jordan

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2000-2001	Member, committee of Medical Staff Professional Fees, King Abdullah University Hospital, Jordan University of Science & Technology, Irbid, Jordan
2004	Judge, University of Iowa Medical Student Research (platform presentation session), Iowa City, IA, USA
2005	Judge, University of Iowa Medical Student Research (platform presentation session), Iowa City, IA, USA
2006	Member, Search Committee for the Director of the Division of Pediatric Hematology/Oncology, University of Iowa, Iowa City, IA, USA
2009	Judge, Weil-Cornell Medical College – Qatar, Student Research (platform presentation session), Doha, Qatar
2010-2011	Member, Committee for the strategic planning (2011-2016) for the Shafallah Foundation and its centers, Doha, Qatar
2014-2015	Member of the Management Team, Qatar Biomedical Research Institute, Doha, Qatar
2016- now	Member, Committee for the blood bank and tissues, Jordan University Hospital, Amman, Jordan

National or international committees and service

1996-1998	Member *AD HOC* committee for the establishment of the National Center for Diabetes, Endocrinology & Genetics, Amman, Jordan
1997-1998	Member, advisory committee for the practice of genetics-related procedures & Technology. Amman, Jordan
2000-2002	Member, committee of The Jordanian Board of Pediatrics Examination, The Jordanian Medical Council, Amman, Jordan
2002-2002	Member, advisory committee for the practice of assisted reproduction. Amman, Jordan
2005-2006	Item writer for the genetics specialty examinations, American Board of Medical Genetics, Bethesda, MD, USA
2009-2013	International League Against Epilepsy (ILAE)-Autism Speaks (AS) collaborative Epilepsy-Autism task force
2010-2012	Qatar Cardiovascular Research Center (QCRC) Coordination and Implementation committee, Doha, Qatar
2015-2016	Qatar Genome Program committee (QGP), Doha Qatar
2016	Judge, Abdul-Hamid Shuman Prize in Medical Sciences for the Arab Investigators, Amman, Jordan

Professional Affiliations

1985-now	Jordan Medical Association
1988-now	American Society of Human Genetics
1992-2008	American Association for the Advancement of Science
1993-now	American College of Medical Genetics
1993-now	American Academy of Pediatrics
1994-now	The Jordan Pediatric Society
1995-now	European Society of Human Genetics
2005-now	The American Epilepsy Society
2006-now	The Society for Pediatric Research
2016_now	Jordan Society for Scientific Research

B. Clinical Assignments since last promotion

The University of Jordan School of Medicine

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Hospital previliges, Jordan University Hospital, Amman, Jordan
Two half day clinics per week, Jordan University Hospital, Amman, Jordan
Two half day clinics per week, National Center for Diabetes, Endocrinology and Genetics, Amman, Jordan