

CURRICULUM VITAE

Dr. Ayman W. El-Hattab, MD, FAAP, FACMG

**American Board of Clinical Genetics
American Board of Biochemical Genetics (Metabolic)
American Board of Pediatrics
Fellow of the American Academy of Pediatrics
Fellow of the American College of Medical Genetics**

Associate Professor (joint)

College of Medicine, University of Sharjah, Sharjah, United Arab Emirates

Consulting Editor for *GeneReviews*

<https://www.ncbi.nlm.nih.gov/books/NBK138604/>



Consultant Clinical Genetics and Metabolic

American Hospital Dubai, Dubai, United Arab Emirates

<https://www.ahdubai.com/doctors-profile/ayman-el-hattab>

Mediclinic City Hospital, Dubai, United Arab Emirates

Mediclinic Welcare Hospital, Dubai, United Arab Emirates

Mediclinic Al Noor Hospital, Abu Dhabi, United Arab Emirates

Mediclinic Al Ain Hospital, Al-Ain, United Arab Emirates

<https://www.mediclinic.ae/en/corporate/doctors/1/ayman-el-hattab.html>

King's College Hospital London, Dubai, United Arab Emirates

<https://kingscollegehospitaldubai.com/dr/ayman-el-hattab/>

University Hospital Sharjah, Sharjah, United Arab Emirates

<https://www.uhs.ae/doctor-profile/dr-ayman-el-hattab-209/>

Kanad Hospital, Al-Ain, United Arab Emirates

<https://kanadhospital.org/portfolio/dr-ayman-el-hattab/>

KidsHeart Medical Center, Abu Dhabi, United Arab Emirates

KidsHeart Medical Center, Dubai, United Arab Emirates

The Heart Medical Center, Al-Ain, United Arab Emirates

<https://www.kidsheart.ae/en/doctors/dr-ayman-el-hattab.html>

Neuropedia Children's Neuroscience Center, Dubai, United Arab Emirates

<http://neuropedia.ae/about-us/>

Hope Abilitation Medical Center, Dubai, United Arab Emirates

<https://hope-amc.com/doctor/dr-ayman-el-hattab/>

Genesis Perinatal Care Clinic, Dubai, United Arab Emirates

Genesis Healthcare Center, Dubai, United Arab Emirates

https://www.genesis-dubai.com/our_doctors/dr-ayman-el-hattab/

Personal data

Name: Ayman Warrad Mahmoud El-Hattab
 Date of Birth: 1 July 1977
 Citizenship: Jordanian
 Family status: Married with 4 children
 Address: Al Oyouun Village
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 Al-Ain
 United Arab Emirates
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Education

10/1996 – 06/2002: Bachelor Degree of Science in Medicine and Surgery, Jordan University of Science and Technology, Irbid, Jordan.
 07/2002 – 06/2003: Internship in medicine, surgery, obstetric, and pediatrics, the Ministry of Health hospitals, Irbid, Jordan.
 07/2003 – 06/2005: Pediatric residency, Jordan University of Science and Technology / King Abdullah University Hospital, Irbid, Jordan.
 07/2005 – 06/2008: Pediatric residency, SUNY Downstate Medical Center, Brooklyn, New York, USA.
 07/2008 – 06/2010: Clinical Genetics Fellowship, Baylor College of Medicine, Houston, Texas, USA.
 07/2010 – 06/2011: Medical Biochemical Genetics Fellowship, Baylor College of Medicine, Houston, Texas, USA.

Examinations

08/2004: Passed the USMLE Step 1 examination.
 10/2004: Passed the USMLE Step 2 CK examination.
 12/2004: Passed the USMLE Step 2 CS examination.
 06/2005: Passed the USMLE Step 3 examination.
 10/2008: Passed the American Board of Pediatrics examination.
 09/2011: Passed the American Board of Medical Genetics (ABMG) General examination.
 09/2011: Passed the American Board of Medical Genetics (ABMG) Clinical Genetics examination.
 09/2011: Passed the American Board of Medical Genetics (ABMG) Medical Biochemical Genetics examination.

Certificates

06/2002: Bachelor Degree of Science in Medicine and Surgery, Jordan University of Science and Technology, Irbid, Jordan.
 10/2008: The American Board of Pediatrics, USA.
 09/2011: The American Board of Medical Genetics, Clinical Genetics, USA.
 09/2011: The American Board of Medical Genetics, Medical Biochemical Genetics, USA.

Appointments

- 07/2011 – 06/2012: Assistant Professor of Child Health, University of Missouri Health Care, Columbia, Missouri, USA.
- 07/2011 – 06/2012: Assistant Professor of Pathology & Anatomical Sciences, University of Missouri Health Care, Columbia, Missouri, USA.
- 07/2011 – 06/2012: Director of Biochemical Genetics Laboratory, University of Missouri Health Care, Columbia, Missouri, USA.
- 07/2012 – 06/2014: Consultant, Metabolic and Clinical Genetics, Medical Genetics Section, Department of Pediatrics, The Children's Hospital, King Fahad Medical City, Riyadh, Saudi Arabia.
- 03/2013 – 06/2014: Assistant Professor of Pediatrics, Faculty of Medicine, King Saud bin Abdulaziz University for Health Sciences, Riyadh, Saudi Arabia.
- 01/2013 – 06/2014: Program Director, Clinical Genetics and Metabolic Disorders Fellowship, King Fahad Medical City, Riyadh, Saudi Arabia.
- 06/2014 – 11/2018: Consultant, Clinical Genetics and Metabolic, Division of Clinical Genetics and Metabolic Disorders, Pediatric Department, Tawam Hospital, Al-Ain, United Arab Emirates.
- 01/2019 – Present: Clinical Associate Professor (joint), The Department of Clinical Sciences, College of Medicine, University of Sharjah, Sharjah, United Arab Emirates
- 01/2019 – Present: Consultant Clinical Genetics and Metabolic, Genetics Clinics, KidsHeart Medical Center, Abu Dhabi, United Arab Emirates
- 01/2019 – Present: Consultant Clinical Genetics and Metabolic, Genetics Clinics, KidsHeart Medical Center, Dubai, and Al-Ain, United Arab Emirates
- 01/2019 – Present: Consultant Clinical Genetics and Metabolic, Genetics Clinics, The Heart Medical Center, Al-Ain, United Arab Emirates
- 04/2019 – Present: Consultant Clinical Genetics and Metabolic, University Hospital Sharjah, Sharjah, United Arab Emirates
- 04/2019 – Present: Consultant Clinical Genetics and Metabolic, Neuropedia Children's Neuroscience Center, Dubai, United Arab Emirates
- 06/2019 – Present: Consultant Clinical Genetics and Metabolic, Mediclinic City Hospital, Dubai, United Arab Emirates
- 07/2019 – Present: Consultant Clinical Genetics and Metabolic, American Hospital Dubai, Dubai, United Arab Emirates
- 08/2019 – Present: Consultant Clinical Genetics and Metabolic, Mediclinic Welcare Hospital, Dubai, United Arab Emirates
- 08/2019 – Present: Consultant Clinical Genetics and Metabolic, Hope Abilitation Medical Center, Dubai, United Arab Emirates
- 08/2019 – Present: Consultant Clinical Genetics and Metabolic, Mediclinic Al Noor Hospital, Abu Dhabi, United Arab Emirates
- 08/2019 – Present: Consultant Clinical Genetics and Metabolic, Oasis Hospital, Al-Ain, United Arab Emirates
- 09/2019 – Present: Consultant Clinical Genetics and Metabolic, Genesis Perinatal Care Clinic, Dubai, United Arab Emirates
- 11/2019 – Present: Consultant Clinical Genetics and Metabolic, Mediclinic Al Ain Hospital, Al-Ain, United Arab Emirates
- 04/2019 – Present: Consultant Clinical Genetics and Metabolic, King's College Hospital London, Dubai, United Arab Emirates

04/2019 – Present: Consultant Clinical Genetics and Metabolic, Genesis Healthcare Center, Dubai, United Arab Emirates

Licenses

07/2003: Licensed by the Ministry of Health, Jordan.
 08/2008: Licensed by Texas Medical Board, USA.
 08/2010: Licensed by Missouri State Board of Registration for the Healing Arts, USA.
 07/2102: Registered at the Saudi Commission for Health Specialties, Saudi Arabia.
 05/2014: Licensed by Department of Health - Abu Dhabi, United Arab Emirates.
 01/2019: Licensed by Dubai Healthcare City Authority, Dubai, United Arab Emirates.
 04/2019: Licensed by Dubai Health Authority, Dubai, United Arab Emirates.
 04/2019: Licensed by Ministry of Health, United Arab Emirates.

Membership

Membership in scientific societies

2006 – Present Member of the American Society of Human Genetics (ASHG), USA.
 2008 – Present Member of the United Mitochondrial Disease Foundation (UMDF), USA.
 2009 – Present Fellow of the American Academy of Pediatrics (AAP), USA.
 2012 – Present Fellow of the American College of Medical Genetics (ACMG), USA.
 2013 – Present Member of the European Society of Human Genetics (ESHG), Vienna, Austria.
 2013 – Present Member of the Society for the Study of Inborn Errors of Metabolism (SSIEM), London, UK.
 2013 – Present Member of the Saudi Society of Medical Genetics (SSMG), Saudi Arabia.
 2017 – Present Member of the Society for Inherited Metabolic Disorders (SIMD), USA

Membership in committees

09/2011 – 06/2012: Member at The Faculty Leadership and Development Committee, University of Missouri Health Care, Columbia, Missouri, USA.
 11/2011 – 06/2012: Member at The Genetic Advisory Committee, Department of Health and Senior Services, Missouri, USA.
 11/2011 – 06/2012: Member at The Newborn Screening Standing Committee, Department of Health and Senior Services, Missouri, USA.
 07/2012 – 05/2013: The Chairman of the organizing committee for The Basic and Advanced Medical Genetics 6th Annual Review Course, King Fahad Medical City, Riyadh, Saudi Arabia.
 09/2012 – 06/2014: Member at The Children's Hospital Undergraduates Committee (planning and organizing teaching pediatrics to medical students), King Fahad Medical City, Riyadh, Saudi Arabia.
 11/2012 – 06/2014: Member at The Children's Hospital Research Committee (creation of research culture at Children's Hospital), King Fahad Medical City, Riyadh, Saudi Arabia.
 11/2012 – 06/2014: Member at The Children's Hospital Regulatory Documentation Committee (review all internal policies and procedures of Children's Hospital), King Fahad Medical City, Riyadh, Saudi Arabia.
 01/2013 – 06/2014: The Children's Hospital representative for the Joint Commission International Accreditation (JCIA) Care of Patient (COP) Chapter, King Fahad Medical City, Riyadh, Saudi Arabia.
 02/2013 – 04/2013 The Organizer for The Applications of Advanced Genetic Testing in Medical Diagnosis Symposium, King Fahad Medical City, Riyadh, Saudi Arabia.
 05/2013 – 06/2014: Member of the Scientific Committee of the Saudi Society of Medical Genetics, Riyadh, Saudi Arabia.
 05/2013 – 04/2014: Member of the Organizing Committee for the First International Meeting for the Saudi Society of Medical Genetics, Riyadh, Saudi Arabia.

- 06/2013 – 06/2014: Member of the Children’s Hospital International Collaboration Task Force, King Fahad Medical City, Riyadh, Saudi Arabia.
- 02/2014 – 06/2014: Member of the Scientific Committee of the Children’s Hospital Annual Research Day, King Fahad Medical City, Riyadh, Saudi Arabia.
- 06/2015 – 11/2018: Member of Genetics Advisory Panel, Health Authority of Abu Dhabi (HAAD), Abu Dhabi, United Arab Emirates.
- 10/2017 – 11/2018: Member of Newborn Screening National Committee, Health Authority of Abu Dhabi (HAAD), Abu Dhabi, United Arab Emirates.

Awards

- 07/1997: Distinguished performance at Jordan University of Science and Technology Medical School. The name appeared on the Faculty Honor List.
- 06/2002: Rank of seventh among the graduates at Jordan University of Science and Technology Medical School.
- 03/2008: Received “Certificate of Appreciation” from King’s County Hospital Center for outstanding resident, New York, USA.
- 02/2009: Received the Society for Inherited Metabolic Disorders (SIMD)/Hyperion Fellowship in Inborn Errors of Metabolism Award.
- 02/2010: Received the Genzyme/ American College of Medical Genetics Foundation (ACMGF) Clinical Genetics Fellowship in Biochemical Genetics Award.
- 08/2010: Received the Trainee Research Award, the 60th annual meeting of American Society of Human Genetics (ASHG).
- 11/2010: Received the Research Subject Advocate (RSA) Award from the General Clinical Research Center (GCRC) at Texas Children’s Hospital, Texas, USA.
- 06/2011: Received the Best Abstract Award, first prize, United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine 2011 Meeting.
- 10/2011: Received the Semifinalist Trainee Research Award, the 12th International Congress of Human Genetics (ICHG) / 61st Annual Meeting of The American Society of Human Genetics (ASHG).
- 07/2013: Received the Publication Award, Research and Scientific Publication Center, King Fahad Medical City, Riyadh, Saudi Arabia.
- 09/2013: Received the 2012 Society for Inherited Metabolic Disorders (SIMD) Emmanuel Shapira Award. The award was announced at the 12th International Congress of Inborn Errors of Metabolism (ICIEM 2013). Barcelona, Spain, September 3-6, 2013.
- 03/2016: Recognized as “Top Poster” at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Tampa, Florida, USA, March 8-12, 2016.
- 03/2017: Recognized as “Top Poster” at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Phoenix, Arizona, USA, March 21-25, 2017.
- 09/2017: Received the 2016 Society for Inherited Metabolic Disorders (SIMD) Emmanuel Shapira Award (for the second time). The award was announced at the 13th International Congress of Inborn Errors of Metabolism (ICIEM 2017). Rio de Janeiro, Brazil, September 5-8, 2017.

Publications**Peer-reviewed articles**

1. **El-Hattab AW**, Smolarek TA, Walker ME, Schorry EK, Immken LL, Patel G, Abbott MA, Lanpher BC, Ou Z, Kang SH, Patel A, Scaglia F, Lupski JR, Cheung SW, Stankiewicz P. **Redefined genomic architecture in 15q24 directed by patient deletion/duplication breakpoint mapping.** Hum Genet. 2009;126(4):589-602.
<http://www.ncbi.nlm.nih.gov/pubmed/19557438>
2. **El-Hattab AW**, Li FY, Shen J, Powell BR, Bawle EV, Adams DJ, Wahl E, Kobori JA, Graham B, Scaglia F, Wong LJ. **Maternal systemic primary carnitine deficiency uncovered by newborn screening: clinical, biochemical, and molecular aspects.** Genet Med. 2010;12(1):19-24.
<http://www.ncbi.nlm.nih.gov/pubmed/20027113>
3. **El-Hattab AW**, Li FY, Schmitt E, Zhang S, Craigen WJ, Wong LJ. **MPV17-associated hepatocerebral mitochondrial DNA depletion syndrome: new patients and novel mutations.** Mol Genet Metab. 2010;99(3):300-308.
<http://www.ncbi.nlm.nih.gov/pubmed/20074988>
4. **El-Hattab AW**, Skorupski JC, Hsieh MH, Breman AM, Patel A, Cheung SW, Craigen WJ. **OEIS complex associated with chromosome 1p36 deletion: a case report and review.** Am J Med Genet A. 2010;152A(2):504-11.
<http://www.ncbi.nlm.nih.gov/pubmed/20101692>
5. Li FY, **El-Hattab AW**, Bawle E, Boles RG, Schmitt E, Scaglia F, Wong LJ. **Molecular spectrum of SLC22A5 (OCTN2) gene mutations detected in 143 subjects evaluated for systemic carnitine deficiency.** Hum Mutat. 2010;31(8):E1632-51.
<http://www.ncbi.nlm.nih.gov/pubmed/20574985>
6. **El-Hattab AW**, Eng PA, Wu JBS, Walker BA, Stankiewicz P, Cheung SW, Brown CW. **Microduplication of Xp11.23p11.3 with effects on cognition, behavior, and craniofacial development.** Clin Genet. 2011;79(6):531-538.
<http://www.ncbi.nlm.nih.gov/pubmed/20662849>
7. **El-Hattab AW**, Zhang F, Maxim R, Christensen KM, Ward JC, Hines-Dowell S, Scaglia F, Lupski JR, Cheung SW. **Deletion and duplication of 15q24: molecular mechanisms and potential modification by additional copy number variants.** Genet Med. 2010;12(9):573-586.
<http://www.ncbi.nlm.nih.gov/pubmed/20860070>
8. Sadikovic B, Wang J, **El-Hattab A**, Landsverk M, Douglas G, Brundage EK, Craigen WJ, Schmitt ES, Wong LJ. **Sequence homology at the breakpoint and clinical phenotype of mitochondrial DNA deletion syndromes.** PLoS One. 2010;5(12):e15687.
<http://www.ncbi.nlm.nih.gov/pubmed/21187929>
9. Fruhman G, **El-Hattab AW**, Belmont JW, Patel A, Cheung SW, Sutton VR. **Suspected trisomy 22: Modification, clarification, or confirmation of the diagnosis by aCGH.** Am J Med Genet A. 2011;155(2):434-438.
<http://www.ncbi.nlm.nih.gov/pubmed/21271668>
10. **El-Hattab AW**, Fang P, Jin W, Hughes JR, Gibson J, Patel GS, Grange DK, Manwaring L, Patel A, Stankiewicz P, Cheung SW. **Int22h-1/int22h-2-mediated Xq28 rearrangements: intellectual disability associated with duplications and in utero male lethality with deletions.** J Med Genet. 2011;48(12):840-850.
<http://www.ncbi.nlm.nih.gov/pubmed/21984752>
11. Magoulas PL, **El-Hattab AW**. **Chromosome 15q24 microdeletion syndrome.** Orphanet J Rare Dis. 2012;7(1):2.
<http://www.ncbi.nlm.nih.gov/pubmed/22216833>
12. Magoulas PL, **El-Hattab AW**, Roy A, Bali DS, Finegold MJ, Craigen WJ. **Diffuse reticuloendothelial system involvement in type IV glycogen storage disease with a novel**

- GBE1 mutation: A case report and review.** Hum Pathol. 2012;43(6):943-951.
<http://www.ncbi.nlm.nih.gov/pubmed/22305237>
13. **El-Hattab AW**, Hsu JW, Emrick LT, Wong LJ, Craigen WJ, Jahoor F, Scaglia F. **Restoration of impaired nitric oxide production in MELAS syndrome with citrulline and arginine supplementation.** Mol Genet Metab. 2012;105(4):607-614.
<http://www.ncbi.nlm.nih.gov/pubmed/22325939>
 14. **El-Hattab AW. Systemic Primary Carnitine Deficiency.** In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. 2012 Mar 15. Available from: <http://www.ncbi.nlm.nih.gov/books/NBK84551/>.
<http://www.ncbi.nlm.nih.gov/pubmed/22420015>
 15. Lee IC*, **El-Hattab AW***, Wang J, Li FY, Weng SW, Craigen WJ, Wong LJ. **SURF1-associated Leigh syndrome: A case-series and novel mutations.** Hum Mutat. 2012;33(8):1192-200.
(*These two authors contributed equally to this work)
<http://www.ncbi.nlm.nih.gov/pubmed/22488715>
 16. **El-Hattab AW**, Scaglia F, Craigen WJ, Wong L-JC . **MPV17-Related Hepatocerebral Mitochondrial DNA Depletion Syndrome.** In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. 2012 May 17.
<http://www.ncbi.nlm.nih.gov/pubmed/22593919>
 17. **El-Hattab AW**, Emrick LT, Craigen WJ, Scaglia F. **Citrulline and arginine utility in treating nitric oxide deficiency in mitochondrial disorders.** Mol Genet Metab. 2012;107(3):247-252.
<http://www.ncbi.nlm.nih.gov/pubmed/22819233>
 18. Magoulas PL, **El-Hattab AW. Systemic primary carnitine deficiency: An overview of clinical manifestations, diagnosis, and management.** Orphanet J Rare Dis. 2012;7(1):68.
<http://www.ncbi.nlm.nih.gov/pubmed/22989098>
 19. Magoulas PL, **El-Hattab AW. Glycogen Storage Disease Type IV.** In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. 2013 Jan 3. Available from: <http://www.ncbi.nlm.nih.gov/books/NBK115333/>
<http://www.ncbi.nlm.nih.gov/pubmed/23285490>
 20. Segal MM, Williams MS, Gropman AL, Torres AR, Forsyth R, Connolly AM, **El-Hattab AW**, Perlman SJ, Samanta D, Parikh S, Pavlakis SG, Feldman LK, Betensky RA, Gospe SM Jr. **Evidence-Based Decision Support for Neurological Diagnosis Reduces Errors and Unnecessary Workup.** J Child Neurol. 2014;29(4):487-92
<http://www.ncbi.nlm.nih.gov/pubmed/23576414>
 21. **El-Hattab AW**, Scaglia F. **Mitochondrial DNA depletion syndromes: review and updates of genetic basis, manifestations, and therapeutic options.** Neurotherapeutics. 2013;10(2):186-198.
<http://www.ncbi.nlm.nih.gov/pubmed/23385875>
 22. **El-Hattab AW**, Emrick L, K. Williamson K, Craigen W Scaglia F. **The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome.** Meta Gene. 2013;1:8-14.
<http://www.ncbi.nlm.nih.gov/pubmed/25411654>
 23. Al-Hussaini A, Faqeih E, **El-Hattab AW**, Alfadhel M, Asery A, Alsaleem B, Bakhsh E, Ali A, Alasmari A, Lone K, Nahari A, Eyaid W, Al Balwi M, Craig K, Butterworth A, He L, Taylor RW. **Clinical and molecular characteristics of mitochondrial DNA depletion syndrome associated with neonatal cholestasis and liver failure.** J Pediatr. 2014;164(3):553-559.
<http://www.ncbi.nlm.nih.gov/pubmed/24321534>
 24. **El-Hattab AW**, Emrick LT, Chanprasert S, Craigen WJ, Scaglia F. **Mitochondria: role of citrulline and arginine supplementation in MELAS syndrome.** Int J Biochem Cell Biol. 2014;48:85-91.
<http://www.ncbi.nlm.nih.gov/pubmed/24412347>

25. Meilleur KG, Zukosky K, Medne L, Fequiére P, Powell-Hamilton N, Winder TL, Alsaman A, **El-Hattab AW**, Dastgir J, Hu Y, Donkervoort S, Golden JA, Eagle R, Finkel R, Scavina M, Hood IC, Rorke-Adams LB, Bönnemann CG. **Clinical, pathologic, and mutational spectrum of dystroglycanopathy caused by LARGE mutations.** J Neuropathol Exp Neurol. 2014;73(5):425-41.
<http://www.ncbi.nlm.nih.gov/pubmed/24709677>
26. Segal MM, Abdellateef M, **El-Hattab AW**, Hilbush BS, De La Vega FM, Tromp G, Williams MS, Betensky RA, Gleeson J. **Clinical pertinence metric enables hypothesis-independent genome-phenome analysis for neurologic diagnosis.** J Child Neurol. 2015;30(7):881-888.
<http://www.ncbi.nlm.nih.gov/pubmed/25156663>
27. **El-Hattab AW**, Emrick LT, Chanprasert S, Hsu JW, Jahoor F, Scaglia F, Craigen WJ. **Glucose metabolism derangements in adults with the MELAS m.3243A>G mutation.** Mitochondrion. 2014;18:63-69.
<http://www.ncbi.nlm.nih.gov/pubmed/25086207>
28. **El-Hattab AW**, Schaaf CP, Fang P, Roeder E, Kimonis VE, Church JA, Patel A, Cheung SW. **Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review.** BMC Med Genet. 2015;16:12.
<http://www.ncbi.nlm.nih.gov/pubmed/25927380>
29. **El-Hattab AW.** **Inborn Errors of Metabolism.** Clin Perinatol. 2015;42(2):413-439.
<http://www.ncbi.nlm.nih.gov/pubmed/26042912>
30. **El-Hattab AW**, Adesina AM, Jones J, Scaglia F. **MELAS syndrome: Clinical manifestations, pathogenesis, and treatment options.** Mol Genet Metab. 2015;116(1-2):4-12.
<http://www.ncbi.nlm.nih.gov/pubmed/26095523>
31. **El-Hattab AW**, Scaglia F. **Disorders of carnitine biosynthesis and transport.** Mol Genet Metab 2015;116(3):107-112.
<http://www.ncbi.nlm.nih.gov/pubmed/26385306>
32. Bayram Y, Karaca E, Coban Akdemir Z, Yilmaz EO, Tayfun GA, Aydin H, Torun D, Bozdogan ST, Gezdirici A, Isikay S, Atik MM, Gambin T, Harel T, **El-Hattab AW**, Charng WL, Pehlivan D, Jhangiani SN, Muzny DM, Karaman A, Celik T, Yuregir OO, Yildirim T, Bayhan IA, Boerwinkle E, Gibbs RA, Elcioglu N, Tuysuz B, Lupski JR. **Molecular etiology of arthrogyposis in multiple families of mostly Turkish origin.** J Clin Invest. 2016;126(2):762-78.
<http://www.ncbi.nlm.nih.gov/pubmed/26752647>
33. **El-Hattab AW**, Saleh MA, Hashem A, Al-Owain M, Asmari AA, Rabei H, Abdelraouf H, Hashem M, Alazami AM, Patel N, Shaheen R, Faqeih EA, Alkuraya FS. **ADAT3-related intellectual disability: Further delineation of the phenotype.** Am J Med Genet A. 2016;170(5):1142-1147.
<http://www.ncbi.nlm.nih.gov/pubmed/26842963>
34. **El-Hattab AW**, Emrick LT, Hsu JW, Chanprasert S, Almannai M, Craigen WJ, Jahoor F, Scaglia F. **Impaired nitric oxide production in children with MELAS syndrome and the effect of arginine and citrulline supplementation.** Mol Genet Metab. 2016;117(4):407-412.
<http://www.ncbi.nlm.nih.gov/pubmed/26851065>
35. Harel T, Yesil G, Bayram Y, Coban-Akdemir Z, Charng WL, Karaca E, Al Asmari A, Eldomery MK, Hunter JV, Jhangiani SN, Rosenfeld JA, Pehlivan D, **El-Hattab AW**, Saleh MA, LeDuc CA, Muzny D, Boerwinkle E; Baylor-Hopkins Center for Mendelian Genomics, Gibbs RA, Chung WK, Yang Y, Belmont JW, Lupski JR. **Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy.** Am J Hum Genet. 2016;98(3):562-570.
<http://www.ncbi.nlm.nih.gov/pubmed/26942288>
36. **El-Hattab AW**, Shaheen R, Hertecant J, Galadari HI, Albaqawi BS, Nabil A, Alkuraya FS. **On the phenotypic spectrum of serine biosynthesis defects.** J Inherit Metab Dis. 2016;39(3):373-381.
<https://www.ncbi.nlm.nih.gov/pubmed/26960553>

37. **El-Hattab AW**, Schaaf CP, Cheung SW. **Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated**. 2016 Mar 10. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016.
<https://www.ncbi.nlm.nih.gov/pubmed/26962617>
38. **El-Hattab AW**, Scaglia F. **Mitochondrial cytopathies**. Cell Calcium. 2016;60(3):199-206.
<https://www.ncbi.nlm.nih.gov/pubmed/26996063>
39. **El-Hattab AW**. **Serine biosynthesis and transport defects**. Mol Genet Metab. 2016;118(3):153-159.
<https://www.ncbi.nlm.nih.gov/pubmed/27161889>
40. **El-Hattab AW**, Scaglia F. **SUCLA2-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria**. 2016 Jun 30. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016.
<http://www.ncbi.nlm.nih.gov/pubmed/20301762>
41. Charng WL, Karaca E, Coban Akdemir Z, Gambin T, Atik MM, Gu S, Posey JE, Jhangiani SN, Muzny DM, Doddapaneni H, Hu J, Boerwinkle E, Gibbs RA, Rosenfeld JA, Cui H, Xia F, Manickam K, Yang Y, Faqeih EA, Al Asmari A, Saleh MA, **El-Hattab AW**, Lupski JR. **Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate**. BMC Med Genomics. 2016;9(1):42.
<http://www.ncbi.nlm.nih.gov/pubmed/27435318>
42. Shaheen R, Al-Salam Z, **El-Hattab AW**, Alkuraya FS. **The syndrome dysmorphic facies, renal agenesis, ambiguous genitalia, microcephaly, polydactyly and lissencephaly (DREAM-PL): Report of two additional patients**. Am J Med Genet A. 2016;170(12):3222-3226.
<http://www.ncbi.nlm.nih.gov/pubmed/27480277>
43. **El-Hattab AW**, Scaglia F. **Mitochondrial Cardiomyopathies**. Front Cardiovasc Med. 2016;3:25.
<http://www.ncbi.nlm.nih.gov/pubmed/27504452>
44. Al Kaabi EH, **El-Hattab AW**. **N-acetylglutamate synthase deficiency: Novel mutation associated with neonatal presentation and literature review of molecular and phenotypic spectra**. Mol Genet Metab Rep. 2016;8:94-8.
<http://www.ncbi.nlm.nih.gov/pubmed/27570737>
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Book chapters

1. **El-Hattab AW & Reid VS. Inborn Errors of Metabolism.** In Cloherty JP, editor. Manual of Neonatal Care. 7th ed. Philadelphia, PA, USA: Lippincott Williams and Wilkins; 2011. p. 767-789.
2. **El-Hattab AW. MPV17-associated hepatocerebral mitochondrial DNA depletion syndrome.** In Wong LJ, editor. Mitochondrial Disorders Caused by Nuclear Genes. 1st ed. New York, NY, USA: Springer Science+Business Media LLC; 2013. p. 103-112.
3. **El-Hattab AW & Scaglia F. Mitochondrial disorders.** In Lee B, editor. Inborn Errors of Metabolism. 1st ed. Philadelphia, PA, USA: Elsevier Inc; 2014. p. 180-202.
4. **El-Hattab AW & Reid VS. Inborn Errors of Metabolism.** In Hansen AR, Eichenwald EC, Stark AR, Martin CR, editors. Cloherty and Stark's Manual of Neonatal Care, 8th ed. Philadelphia, PA, USA: Lippincott Williams and Wilkins; 2017. Chapter 60; p 858-891.
5. **El-Hattab AW & Scaglia F. Disorders of purine and pyrimidine metabolism.** In Kline MW, editor. Rudolph's Pediatrics, 23rd Edition. New York, USA: McGraw-Hill Education; 2018; chapter 163, p759-765.

Abstracts:

1. **El-Hattab AW, Craigen W, Wong LJ, Jahoor F, Scaglia F. Arginine flux and nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation: study design** (Abstract). Mol Genet Metab. 2010;99(3):195.
2. **El-Hattab AW, Li FY, Schmitt E, Zhang S, Craigen W, Wong LJ. MPV17-associated hepatocerebral mitochondrial DNA depletion syndrome: New patients and novel mutations.** Mitochondrion, 2010;10(2):222.
3. **El-Hattab AW, Hsu JWC, Jahoor J, Craigen W, Scaglia F. Nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation: Interim results** (Abstract). Mol Genet Metab. 2011;102(3):252.
4. **El-Hattab AW, Craigen W, Wong LJ, Jahoor F, Scaglia F. Nitric oxide production in subjects**

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6. Peck D, Fang P, **El-Hattab AW**. **ASS1 novel mutation in an asymptomatic infant with citrullinemia type 1** (Abstract). *Mol Genet Metab.* 2012;105(3):347.
7. **El-Hattab AW**, Emrick L, Hsu JWC, Jahoor F, Scaglia F, Craigen W. **Glucose kinetics in subjects with MELAS syndrome: Interim results.** *Mitochondrion* 2012;12(5):554.
8. **El-Hattab AW**, Emrick LT, Williamson KC, Craigen WJ, Scaglia F. **The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome.** *Mitochondrion* 2013; 13(6): 898.
9. **El-Hattab AW**, Emrick LT, Chanprasert S, Hsu JW, Almannai M, Craigen WJ, Jahoor F, Scaglia F. **Utility of citrulline in treating hypoargininemia in children with MELAS.** *Mitochondrion* 2015;24: S18-S19

Online databases:

1. **El-Hattab AW. 15q24 microdeletion syndrome.** Orphanet Encyclopedia of Rare Diseases. 2012. Available from: http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=94065
2. **El-Hattab AW. Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency.** Orphanet Encyclopedia of Rare Diseases. 2019. Available from: https://www.orpha.net/consor/www/cgi-bin/OC_Exp.php?lng=EN&Expert=279934

Editorial Responsibilities

- 04/2013 – 06/2014 Editor, KFMC e-Knowledge Exchange Newsletter, King Fahad Medical City, Riyadh, Saudi Arabia
- 10/2016 – present GeneReviews, Consulting Editor
(<https://www.ncbi.nlm.nih.gov/books/NBK138604/>)

Reviewer's Activities

Review articles for the following journals:

1. Acute Medicine & Surgery
2. Advances in Clinical Chemistry
3. American Association of Clinical Endocrinologists Journals
4. American Journal of Medical Genetics Part A
5. American Journal of Medical Genetics Part B
6. BMC Medical Genetics
7. BMC Pediatrics
8. BMJ Case Reports
9. Brazilian Journal of Medical and Biological Research
10. Child Neurology Open
11. Clinical Genetics
12. Current Drug Metabolism
13. Developmental Medicine & Child Neurology
14. Early Human Development

15. eNeurologicalSci
16. Epileptic Disorders
17. European Journal of Medical Genetics
18. Expert Opinion on Orphan Drugs
19. Expert Review of Molecular Diagnostics
20. Frontiers in Genetics
21. Future Microbiology
22. Gene
23. GeneReviews
24. Genomics
25. Human Mutation
26. International Invention Journal of Medicine and Medical Sciences
27. Journal of Cardiology and Cardiovascular Sciences
28. Journal of Human Genetics
29. Journal of Inherited Metabolic Disease
30. Journal of Medical Genetics
31. Journal of Nephrology Advances
32. Journal of Pediatric Genetics
33. Journal of Pediatric Neurology
34. Journal of the Neurological Sciences
35. Medical Science Monitor
36. Mitochondrion
37. Molecular Genetics and Metabolism
38. Molecular Genetics and Metabolism Reports
39. Multiple Sclerosis and Related Disorders
40. Neurodegenerative Disease Management
41. Neuropsychiatric Genetics
42. Orphanet Journal of Rare Diseases
43. Pediatric Dermatology
44. PeerJ
45. PLOS ONE
46. Pharmacological Research
47. Scientific Reports
48. Translational Science of Rare Diseases

Teaching activities (details in Appendix 1)

King Abdullah University Hospital (07/2003 – 06/2005)	4 lectures/presentations
SUNY Downstate Medical Center (07/2005 – 06/2008)	18 lectures/presentations
Baylor College of Medicine (07/2008 – 06/2011)	14 lectures/presentations
University of Missouri (07/2011 – 06/2012)	7 lectures/presentations
King Fahad Medical City (07/2012 – 06/2014)	26 lectures/presentations
Tawam Hospital (07/2014 – 11/2018)	36 lectures/presentations
University of Sharjah (01/2019 – Present)	14 lecture/presentations

Presentation at scientific meetings (details in Appendix 2)

Total presentations 77

Poster presentations 37

Oral presentations 40 (including 32 invited presentations)

The American Society of Human Genetics (ASHG) Annual Meetings: **11** presentations
 The American College of Medical Genetics (ACMG) Clinical Genetics Meetings: **11** presentations
 The United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meetings: **8** presentations.
 The International Congress of Inborn Errors of Metabolism (ICIM): **5** presentations
 The Society for Inherited Metabolic Disorders (SIMD) Annual Meetings: **5** presentations
 The European Society of Human Genetics (ESHG) European Human Genetics Conference: **3** presentation
 The Society for the Study of Inborn Errors of Metabolism (SSIEM) Annual Symposia: **3** presentations
 The College of American Pathologists (CAP) Pathologists Meeting: **1** presentation
 The Child Neurology Society Annual Meeting: **1** presentation
 The Basic and Advanced Medical Genetics Annual Review Courses: **2** presentations
 The SEHA International Pediatric Conferences: **2** presentations
 The Emirates Gynecology Oncology Group (EGOG)/ Emirates Oncology and Obstetric Society meeting: **1**
 The OBS-GYNE Exhibit and Congress: **1** presentation
 The Emirates Down Syndrome International Conference: **1** presentation
 Middle East Metabolic Group (MEMG) meeting: **1** presentation
 Arab International Pediatric Medical Congress: **2** presentations
 Abu Dhabi International Neonatal Multispecialty Conference: **1** presentation
 The International Pediatric Conference of Columbia University Medical Center/ NYP Hospital & KidsHeart: **5**
 United Arab Emirates Fetal Medicine Meeting: **1**
 Annual Dubai International Pediatric Neurology Congress: **1**
 Emirati Pediatric Neurology Network Meeting: **2**
 World Pediatric Cardiology and Cardiovascular Surgery Congress: **1**
 Emirates Orthopedics Society meeting: **1**
 Emirates Pediatric and Neonatal Intensive Care Conference (EPNIC): **1**
 Annual International Pediatric Medical Congress: **1**
 Gulf Rare Disease Academy: **1**
 NEUROPEDICON: **1**
 Neurosciences Update, Cleveland Clinic Abu Dhabi: **1**
 Pediatric & Neonatal International Conference of University Hospital Sharjah (UHS): **1**

Workshop and courses

1. Writing a Research Proposal Workshop for health care providers. King Fahad Medical City, Riyadh, Saudi Arabia, September 9, 2012.
2. Lysosomal Storage Diseases (LSD) Masterclass. Dubai, United Arab Emirates, September 27 – 28, 2012.
3. The Applications of Advanced Genetic Testing in Medical Diagnosis Symposium, King Fahad Medical City, Riyadh, Saudi Arabia, April 7, 2013.
4. The 25th Riyadh Autism Journal Club (Genetic Evaluation for Autism and A Neuronal Carnitine Deficiency Hypothesis for Prevention of Non-Dysmorphic Autism), Prince Nasser Bin Abdulaziz Center for Autism, Saudi Arabia, April 7, 2013.
5. The Basic and Advanced Medical Genetics 6th Annual Review Course. King Fahad Medical City, Riyadh, Saudi Arabia, May 21-22, 2013.
6. King Abdullah International Medical Research Center (KAIMRC) 4th Annual Scientific Forum- Research Day, King Saud bin Abdulaziz University for Health Sciences, Riyadh, Saudi Arabia, November 12-13, 2013.
7. The 10th Middle East Metabolic Group (MEMG) meeting, Istanbul, Turkey, December 5-7, 2013.
8. The First International Meeting for the Saudi Society of Medical Genetics, Riyadh, Saudi Arabia,

April 30th-May 1st, 2014. Moderator for the Mitochondrial Disorders Session.

9. The Basic and Advanced Medical Genetics 7th Annual Review Course. King Fahad Medical City, Riyadh, Saudi Arabia, May 21, 2014. Moderator for the Basics in Cytogenetics and Molecular Genetics Session.
10. How to publish your medical research in international scientific journals workshop. Tawam Hospital, Al-Ain, United Arab Emirate, October 14, 2014.
11. The 2nd rare disease Middle East leaders' forum. Dubai, United Arab Emirates, December 6, 2014.
12. KidsHeart Medical Center Lecture series, Abu Dhabi, United Arab Emirates, 7 January 2019
13. KidsHeart Medical Center Lecture series, Abu Dhabi, United Arab Emirates, 4 February 2019
14. The Pediatric Neuroscience Series, the fourth Chapter, Dubai, United Arab Emirates, 28 March 2019.
15. The 2nd MPS I masterclass, Dubai, United Arab Emirates, 26-27 April 2019

Research support

Title: The diagnostic yield of chromosomal microarray in pediatrics

Goals: Chromosomal microarray is a common test that has been utilized frequently to test for chromosomal abnormalities in children with developmental delay, congenital malformation, and growth failure. In this study, we aim to review children who received this test to determine the indications and the outcome of this test. Determining the fraction of cases that are diagnosed through chromosomal microarray can help in determining the role of this test in investigating children with developmental delay, congenital malformation, and growth failure.

Principle Investigator: Ayman W El-Hattab, MD.

Period: 1/2015 – 12/2019.

Role: Principle investigator

Title: The utility of whole exome sequencing (WES) in diagnosing neurogenetic diseases

Goals: Whole exome sequencing (WES) has been increasing used to diagnose genetic and neurological diseases. The test involves the sequencing of the coding region of all known genes and is utilized when the clinical picture is not consistent with a specific disease. This test have been used and helped in diagnosing several rare genetic, metabolic, and neurological diseases. This study aims to review the outcome of WES in diagnosing neurogenetic diseases and the clinical features of diseases diagnosed by WES through a retrospective chart review.

Principle Investigator: Ayman W El-Hattab, MD.

Period: 1/2017 – 12/2019.

Role: Principle investigator

Title: The effect of arginine and citrulline supplementation on endothelial dysfunction in mitochondrial diseases.

Goals: We hypothesize in this study that patients with mitochondrial diseases have endothelial dysfunction that will improve after arginine or citrulline supplementation. We will assess endothelial function using peripheral arterial tonometry before and after arginine supplementation, and before and after citrulline supplementation in children with mitochondrial diseases. We expect to have improvement in endothelial function after arginine and citrulline supplementation. This would add more evidence to the beneficial therapeutic effect of arginine and citrulline and more support to their use in patients with mitochondrial diseases.

Principle Investigator: Ayman W El-Hattab, MD.

Period: 12/2014 – 12/2018.

Annual direct cost: 175,000 AED (\$47,650).

Funding source: National Research Foundation, Ministry of Higher Education and Scientific

Research, United Arab Emirate.

Role: Principle investigator

Title: Whole exome sequencing to identify causes of Mendelian genetic diseases.

Goals: Performing whole exome sequencing on patients suspected to have Mendelian genetic diseases but have no molecular diagnosis aiming to 1) identify the genetic diagnosis in those patients which can help in treatment, prognosis, recurrent risk anticipation, and prevention via prenatal testing, 2) expand the knowledge about the phenotype associated with mutations in known gene, and 3) discover new genes in which mutations can be associated with Mendelian genetic diseases.

Principle Investigator: Ayman W El-Hattab, MD.

Period: 04/2013 – 12/2016.

Role: Principle investigator

Title: Arginine flux and nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation

Goals: To test whether nitric oxide production is lower in subjects with MELAS syndrome, whether arginine and citrulline supplementation will increase the nitric oxide production, and whether citrulline supplementation will increase nitric oxide production more than arginine supplementation.

Principle Investigator: Fernando Scaglia, MD.

Period: 7/2009-6/2011.

Annual direct cost: \$50,000 per year for 2 years.

Funding source: SIMD/Hyperion Fellowship in Inborn Errors of Metabolism.

Role: Recipient of the SIMD/Hyperion Fellowship in Inborn Errors of Metabolism for 2009.

Title: Glucose kinetics in subjects with MELAS syndrome

Goals: To assess glucose metabolism in individuals with MELAS syndrome who do or do not have diabetes mellitus (DM) and in healthy control individuals to investigate the pathophysiologic etiology of DM in MELAS syndrome and determine whether individuals with MELAS syndromes who do not yet have DM have incipient alterations in glucose metabolism.

Principle Investigator: William Craigen, MD, PhD.

Period: 7/2010-6/2011.

Annual direct cost: \$75,000 per year for 1 year.

Funding source: Genzyme/ACMGF Clinical Genetics Fellowship in Biochemical Genetics

Role: Recipient of the Genzyme/ACMGF Clinical Genetics Fellowship in Biochemical Genetics for 2010

Title: Long-term extension of a phase 2, open-label dose-finding study to evaluate the safety, efficacy, and tolerability of multiple subcutaneous doses of rAvPAL-PEG in subjects with PKU.

Goals: To evaluate the effect of long-term administration of multiple doses of SC injections of rAvPAL-PEG on blood phenylalanine concentrations in subjects with PKU.

Principle Investigator: Richard Hillman, MD, PhD.

Period: 07/2011 - 06/2012.

Annual direct cost: \$250,000 annually.

Funding source: BioMarin Pharmaceutical, Inc.

Role: Co-investigator (5% effort).

Title: PKUDOS – Phenylketonuria (PKU) Demographics, Outcomes, and Safety Registry.

Goals: Observational registry for patients with PKU receiving Kuvan. The purpose of this registry is to increase knowledge about the course of disease in Kuvan treated patients by evaluating changes in dietary phenylalanine prescriptions, blood phenylalanine levels, concomitant medications, neurocognitive, and behavioral status over time. Additionally, PKUDOS will collect additional data on the long-term safety and efficacy of Kuvan.

Principle Investigator: Richard Hillman, MD, PhD.

Period: 07/2011 – 06/2012.

Funding source: BioMarin Pharmaceutical, Inc.

Role: Co-investigator.

Title: Lysosomal storage disease (LSD) registry program.

Goals: This registry is a multi-center, observational program for patients diagnosed with a LSD including Gaucher, Fabry, MPS I, and Pompe disease. The objectives of the LSD Registry Program are: a) to enhance the understanding of the variability, progression, and natural history of these LSD diseases with the ultimate goal of better guiding and assessing therapeutic intervention, b) to provide the medical community treating patients with LSD diseases with recommendations for monitoring patients and reports on patient outcomes to optimize patient care, and c) to evaluate the long-term effectiveness of enzyme replacement therapy.

Principle Investigator: Thomas W Loew, MD.

Period: 07/2011 – 06/2012.

Funding source: Genzyme Corporation.

Role: Co-investigator.

Title: Inborn Errors of Metabolism Information System (IBEM-IS).

Goals: This is a long term outcome registry for patients with inborn errors of metabolism who are cared for in the seven state Region 4, and eight state Heartland area delineated by HRSA. (http://region4genetics.org/region4/ibem_is_registry.aspx). This cohort of states encompasses >1 million births per year, all of whom are screened using expanded newborn screening. This represents an opportunity to accumulate meaningful sample sizes for mining of data intrinsic to the database, as well as for recruitment for future interventional studies. It is anticipated that the IBEM-IS will result in the development of evidence-based treatment protocols, potentially improving the care, health and quality of life for children diagnosed with these rare conditions. The registry will also serve as an infrastructure for eventual access to intervention trials.

Principle Investigator: Richard Hillman, MD, PhD.

Period: 07/2011 – 06/2012.

Funding source: Heartland Regional Genetics Collaborative.

Role: Co-investigator.

Title: A phase 4, open-label, prospective study in patients with Pompe disease to evaluate the efficacy and safety of alglucosidase alfa produced at the 4000 L scale.

Goals: To evaluate the efficacy and safety of alglucosidase alfa produced at the 4000 L scale (Lumizyme®) in patients with Pompe disease 1-8 years old.

Principle Investigator: Ayman W El-Hattab, MD.

Period: 03/2012 – 06/2012.

Annual direct cost: \$80,000 annually.

Funding source: Genzyme Corporation.

Role: Principle investigator (6% effort).

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Appendix 1: Teaching activities**King Abdullah University Hospital (07/2003 – 06/2005)****Department of Pediatrics Case Conference:**

- 1 09/2003: Maple syrup urine disease
- 2 11/2003: Erythema nodosum
- 3 01/2005: Neonatal thrombosis

Department of Pediatrics Journal Club:

- 4 05/2004: Fluid resuscitation

SUNY Downstate Medical Center (07/2005 – 06/2008)**Department of Pediatrics Case Conference:**

- 1 11/2005: Congenital syphilis
- 2 01/2006: Dermatomyositis
- 3 02/2006: Clinical evaluation of substance abuse
- 4 04/2006: Infant of diabetic mother
- 5 05/2006: Complex chromosomal abnormality in a newborn with multiple congenital defects
- 6 05/2006: Cornelia de Lange syndrome
- 7 08/2006: Ambiguous genitalia
- 8 09/2006: Teratology
- 9 01/2007: Imprinting: Angelman syndrome and Prader-Willi syndrome
- 10 03/2007: Herpes Simplex Virus
- 11 03/2007: Depression
- 12 04/2007: Fanconi anemia
- 13 05/2007: Alport syndrome
- 14 10/2007: Cerebral palsy
- 15 01/2008: Amaurosis fugax
- 16 01/2008: Systemic lupus erythematosus
- 17 04/2008: Maternal hyperthyroidism
- 18 06/2008: Fetal alcohol syndrome

Baylor College of Medicine (07/2008 – 06/2011)**Human and Molecular Genetics Department Grand Rounds**

- 1 08/2008: Chromosome 15q24 deletion
- 2 10/2008: OEIS complex and 1p36 deletion
- 3 12/2008: Glycogen storage disease type IV
- 4 01/2009: Congenital disorders of glycosylation
- 5 03/2009: Systemic primary carnitine deficiency
- 6 05/2009: Norrie disease: case presentation
- 7 08/2009: Juvenile Huntington disease
- 8 10/2009: Gorlin syndrome
- 9 01/2010: Epidermolysis Bullosa
- 10 02/2010: Complex IV deficiency

Human and Molecular Genetics Department Clinical Conferences (CME)

- 11 10/2009: MELAS syndrome: Pathogenesis and potential therapies
- 12 11/2010: MELAS syndrome: Overview and ongoing research

Medical students teaching

- 13 09/2009: Facilitate medical student groups in discussing the medical and social aspects of genetic disorders
- 14 12/2010: Facilitate medical student groups in discussing the ethical aspects in genetic cases

University of Missouri (07/2011 – 06/2012)**Child Health Department Grand Rounds (CME)**

- 1 09/2011: Mitochondrial disorders: mechanisms, manifestations, and diagnostic approach

First year medical students:

- 2 09/2011: Problem based learning tutorial: OTC female carrier

Second year medical students:

- 3 04/2012: Problem based learning tutorial: Metabolic disorders

Third year medical students

- 4 11/2011- Metabolic disorders and newborn screening. A lecture during the 3rd year medical student clerkship rotation in the Department of Child Health (once every 8 weeks).
06/2012: clerkship rotation in the Department of Child Health (once every 8 weeks).
- 5 11/2011- Mitochondrial diseases. A lecture during the 3rd year medical student clerkship rotation in the Department of Child Health (once every 8 weeks).
06/2012: clerkship rotation in the Department of Child Health (once every 8 weeks).

Pediatric residents

- 6 09/2011: Lecture: How to write a manuscript
7 05/2012: Lecture: Metabolic disorders

King Fahad Medical City (07/2012 – 06/2014)**Pediatric Department Grand Rounds:**

- 1 11 Sep 2012 Lecture: Congenital disorders of glycosylation
2 15 Apr 2014 Lecture: Mitochondrial diseases: mechanisms and clinical manifestations

Pediatric residents teaching:

- 3 15 May 2013 Participation as an examiner at the Junior End Year Clinical Exam
4 05 May 2014 Participation as an examiner at the Junior End Year Clinical Exam
5 18 May 2012 Tutorial: Review of common metabolic disorder

Third year medical students:

- 6 03 Nov 2012 Lecture: Strategies for prevention and treatment of genetic disorders
7 24 Feb 2013 Lecture: Cystic fibrosis as a genetic disorder
8 25 Sep 2013 Lecture: Genetic information, its expression and variation
9 26 Sep 2013 Lecture: Chromosome and cell genetics
10 26 Sep 2013 Lecture: Principles of molecular genetics
11 10 Nov 2013 Lecture: Prevention and treatment of genetic disorders
12 24 Feb 2014 Lecture: Cystic fibrosis as a genetic disorder

Fifth year medical students:

- 13 03 Sep 2012 Lecture: Inborn errors of metabolism
14 21 Nov 2012 Participation in OSCE examination as an examiner
15 06 March 2013 Participation in OSCE examination as an organizer and examiner
16 03 April 2013 Lecture: Inborn errors of metabolism
17 21 Nov 2013 Lecture: Dymorphology and Chromosomal Disorder
18 30 Jan 2014 Participation in OSCE examination as an organizer and examiner
19 11 Jun 2014 Lecture: Dymorphology and Chromosomal Disorder

Clinical Genetics fellows

- 20 19 March 2014 Lecture: Malformation terminology
21 23 April 2014 Lecture: Prevention and treatment of genetic diseases

Other teaching activities

- 22 15 Sep 2012 Lecture: Fabry disease for Nephrology Department
23 16 Dec 2012 Examiner at the 4th Clinical Course in Pediatrics, Department of Pediatrics, King Khalid University Hospital; College of Medicine, King Saud University, Riyadh, Saudi Arabia.
24 06 Nov 2013 Lecture: Hunter syndrome. Security Forces Hospital, Riyadh, Saudi Arabia.
25 18 Nov 2013 Examiner at the 5th Clinical Course in Pediatrics, Department of Pediatrics, King Khalid University Hospital; College of Medicine, King Saud University, Riyadh, Saudi Arabia.
26 05 Feb 2014 Lecture: Genetic counseling for nursing students

Tawam Hospital (07/2014 – 11/2018)**Genetics and Metabolic Grand Round**

- 1 28 Sep 2014 Dymorphology and malformation terminology
2 26 Oct 2014 Introduction to metabolic disorders
3 02 Nov 2014 Treatment options of inborn errors of metabolism
4 07 Dec 2014 Cardiomyopathy: genetic and metabolic evaluation
5 21 Dec 2014 Prevention of genetic diseases
6 28 Dec 2014 Genomic disorders
7 25 Jan 2015 Principles of molecular genetics
8 05 Apr 2015 Glycogen storage diseases
9 19 Apr 2015 Overgrowth syndrome

Pediatrics Department Grand Round

- 10 24 Dec 2014 Carnitine deficiency
- 11 25 Jan 2016 How can we define “inborn errors of metabolism”?
- 12 03 Oct 2016 Storage diseases.
- 13 13 Dec 2017 Glycogen storage diseases
- Pediatric Resident Lectures**
- 14 19 Apr 2015 Introduction to clinical genetics and patterns of inheritance
- 15 20 Apr 2015 Dysmorphology and malformation terminology
- 16 21 Apr 2015 Principles of molecular genetics
- 17 22 Apr 2015 Cytogenetics and chromosomal abnormalities
- 18 26 Apr 2015 Introduction to inborn errors of metabolism
- 19 28 Apr 2015 Organic acidemias and approach for metabolic acidosis
- 20 30 Apr 2015 Urea cycle defects and approach for hyperammonemia
- 21 03 May 2015 Hypoglycemia due to inborn errors of metabolism
- 22 04 May 2015 Lysosomal and peroxisomal storage diseases
- 23 05 May 2015 Mitochondrial disorders
- 24 06 May 2015 Treatment and prevention of genetic diseases
- 25 31 July 2016 Hypoglycemia: case scenarios, approach, and common metabolic disorders
- 26 03 Aug 2016 Hyperammonemia: case scenarios, approach, and common metabolic disorders
- 27 07 Aug 2016 Metabolic acidosis: case scenarios, approach, and common metabolic disorders
- 28 09 Aug 2016 Dysmorphology and malformation terminology
- 29 15 Mar 2018 Mitochondrial diseases
- 30 29 Mar 2018 Genomic disorders
- Other teaching activities**
- 31 16-17 Sep 2014 Participated as an examiner in the assessment of the IVF clinical scientists, Health Authority-Abu Dhabi (HAAD).
- 32 17 Sep 2014 Al Ain Pediatric Club, lecture: Genomic disorders.
- 33 22 Dec 2014 Breast Cancer Master Class at Tawam Hospital, Lecture: *BRCA1* and *BRCA2*-related hereditary breast and ovarian cancer.
- 34 11 June 2015 Oasis Hospital, Al-Ain, lecture: Storage diseases.
- 35 17 Dec 2015 King Abdulla Specialized Children’s Hospital, Riyadh, Saudi Arabia. Lecture: How can we define “inborn errors of metabolism”?
- 36 31 Oct 2016 Jordan University, Amman, Jordan. Lecture: Mitochondrial diseases: mechanisms, manifestations, diagnostic approach, & therapeutic options
- University of Sharjah (01/2019 – Present)**
- 1 17 Feb 2019 American Hospital Dubai, Lecture: The scope of clinical genetics
- 2 24 Apr 2019 The Heart Medical Center Pediatric Lecture Series, Lecture: The scope of clinical genetics
- 3 30 Jun 2019 American Hospital Dubai, Lecture: The scope of clinical genetics
- 4 15 July 2019 University Hospital Sharjah, Lecture: The scope of clinical genetics
- 5 09 Sep 2019 University of Sharjah, College of Medicine, first year medical students: Briefing: A child with abnormal facies (Down syndrome).
- 6 16 Sep 2019 University of Sharjah, College of Medicine, first year medical students: Lecture: Chromosomal abnormalities.
- 7 17 Sep 2019 Mediclinic City Hospital, Lecture: The scope of clinical genetics
- 8 17 Oct 2019 University of Sharjah, College of Medicine, fourth year medical students: Lecture: Introduction to inborn errors of metabolism.
- 9 29 Oct 2019 Corniche Hospital, Neonatology fellows: Lecture: MCQs: Genetics and Neonatology
- 10 12 Dec 2019 University of Sharjah, College of Medicine, fourth year medical students: Lecture: Introduction to inborn errors of metabolism.
- 11 21 Dec 2019 Mediclinic Al Ain Hospital, Lecture: The scope of clinical genetics
- 12 06 Feb 2020 University of Sharjah, College of Medicine, fourth year medical students: Lecture: Introduction to inborn errors of metabolism.
- 13 04 Mar 2019 Mediclinic Al Noor Hospital, Lecture: The scope of clinical genetics
- 14 12 April 2020 University of Sharjah, College of Medicine, fourth year medical students: Lecture: Introduction to inborn errors of metabolism.

Appendix 2: Presentations at scientific meetings

1. **El-Hattab AW**, Velinov M, Valencia G, Perenyi A. Very complex, *de novo* chromosomal abnormality in a newborn with multiple congenital defects- a counseling challenging. Poster at the American Society of Human Genetics (ASHG) 56th Annual Meeting. New Orleans, Louisiana, USA, October 9-13, 2006.
2. Li FY, Bawle E, Boles RG, **El-Hattab AW**, Schmitt E, Wong LJ. Mutations in the *OCTN2 (SLC22A5)* gene detected in clinical testing of patients with carnitine deficiency. Poster at the American College of Medical Genetics (ACMG) Clinical Genetics Meeting. Tampa, Florida, USA, March 25-29, 2009.
3. **El-Hattab AW**, Li FY, Schmitt E, Zhang S, Craigen WJ, Wong LJ. *MPV17*-associated hepatocerebral mitochondrial DNA depletion syndrome: New patients and novel mutations. Poster at the United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meeting. Vienna, Virginia, USA, July 24-27, 2009.
4. Roy A, **El-Hattab AW**, Hicks MJ, Kearney DL, Bali DS, Finegold MJ. Novel mutation in glycogen branching enzyme in glycogen storage disease type IV. Poster at the College of American Pathologists (CAP) Pathologists Meeting. Washington, DC, USA, October 11-14, 2009.
5. **El-Hattab AW**, Li FY, Schmitt E, Zhang S, Craigen WJ, Wong LJ. *MPV17*-associated hepatocerebral mitochondrial DNA depletion syndrome: New patients and novel mutations. Poster at the American Society of Human Genetics (ASHG) 59th Annual Meeting. Honolulu, Hawaii, USA, October 20-24, 2009.
6. **El-Hattab AW**, Enciso V, Roeder ER, Grange DK, Sutton VR, Cheung SW, Patel A. Xq28 microduplications mediated by the homologous *int22h-1* and *int22h-2* in four cognitively impaired males. Poster at the American Society of Human Genetics (ASHG) 59th annual meeting. Honolulu, Hawaii, USA, October 20-24, 2009.
7. Sadikovic B, Wang J, **El-Hattab AW**, Douglas G, Landsverk M, Brundage EK, Schmitt ES, Wong LJ. Genetic mapping and clinical presentation of 65 patients with the mitochondrial deletion syndrome. Poster at the American College of Medical Genetics (ACMG) Clinical Genetics Meeting. Albuquerque, New Mexico, USA, March 24-28, 2010.
8. **El-Hattab AW**, Craigen W, Wong LJ, Jahoor F, Scaglia F. Arginine flux and nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation. **Invited oral presentation** at the Society for Inherited Metabolic Disorders (SIMD) 33rd annual meeting. Albuquerque, New Mexico, USA, March 28-31, 2010.
9. **El-Hattab AW**, Hsu JW, Craigen W, Wong LJ, Jahoor F, Scaglia F. Nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation: interim results. Poster at the United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meeting. Scottsdale, Arizona, USA, June 16-19, 2010.
10. **El-Hattab AW**, Hsu JW, Craigen W, Wong LJ, Jahoor F, Scaglia F. Nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation: interim results. Poster at the Society for the Study of Inborn Errors of Metabolism (SSIEM) Annual Symposium. Istanbul, Turkey, August 31 - September 3, 2010.
11. **El-Hattab AW**, Hsu JWC, Craigen W, Wong LJ, Jahoor F, Scaglia F. Nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation: interim results. **Platform oral presentation** at the American Society of Human Genetics (ASHG) 60th Annual Meeting. Washington, DC, USA, November 2-6, 2010.
12. **El-Hattab AW**, Hsu JWC, Jahoor J, Craigen W, Scaglia F. Nitric oxide production in MELAS syndrome and the effect of arginine and citrulline. **Invited oral presentation** at the Society for Inherited Metabolic Disorders (SIMD) 34th Annual Meeting. Pacific Grove, California, USA, February 27 - March 2, 2011.
13. **El-Hattab AW**, Hsu JWC, Jahoor F, Scaglia F, Craigen W. Glucose kinetics in subjects with MELAS syndrome: interim results. Poster at the American College of Medical Genetics (ACMG) Clinical Genetics Meeting. Vancouver, British Columbia, Canada, March 16-20, 2011.
14. **El-Hattab AW**, Hsu JW, Wong LJ, Craigen W, Jahoor F, Scaglia F. Nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation: interim results. **Platform oral presentation** at the United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meeting. Chicago, Illinois, USA, June 15-17, 2011.
15. **El-Hattab AW**, Hsu JW, Wong LJ, Jahoor F, Scaglia F, Craigen W. Glucose kinetics in subjects with MELAS syndrome: interim results. Poster at the United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meeting. Chicago, Illinois, USA, June 15-17, 2011.
16. **El-Hattab AW**, Hsu JW, Wong LJ, Craigen W, Jahoor F, Scaglia F. Restoration of impaired nitric oxide production in MELAS syndrome with citrulline and arginine supplementation. **Platform oral presentation** at the Society for the Study of Inborn Errors of Metabolism (SSIEM) Annual Symposium. Geneva, Switzerland.

August 30 - September 2, 2011.

17. **El-Hattab AW**, Hsu JW, Wong LJ, Craigen W, Jahoor F, Scaglia F. Restoration of impaired nitric oxide production in MELAS syndrome with citrulline and arginine supplementation. Poster at the 12th International Congress of Human Genetics (ICHG) / 61st Annual Meeting of the American Society of Human Genetics (ASHG). Montreal, Canada, October 11-15, 2011.
18. **El-Hattab AW**, Emrick L, Hsu JW, Wong LJ, Jahoor F, Scaglia F, Craigen W. Glucose kinetics in subjects with MELAS syndrome: interim results. Poster at the Society for Inherited Metabolic Disorders (SIMD) Annual Meeting. Charlotte, North Carolina, USA, March 31 – April 3, 2012.
19. Peck D, Fang P, **El-Hattab AW**. ASS1 novel mutation in an asymptomatic infant with citrullinemia type 1. Poster at the Society for Inherited Metabolic Disorders (SIMD) Annual Meeting. Charlotte, North Carolina, USA, March 31 – April 3, 2012.
20. Peck D, Fang P, **El-Hattab AW**. ASS1 novel mutation in an asymptomatic infant with citrullinemia type 1. Poster at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Charlotte, North Carolina, USA, March 28-30, 2012.
21. **El-Hattab AW**, Emrick L, Hsu JW, Wong LJ, Jahoor F, Scaglia F, Craigen W. Glucose kinetics in subjects with MELAS syndrome: interim results. Platform oral presentation at the United Mitochondrial Disease Foundation Mitochondrial (UMDF) Medicine Meeting. Bethesda, Maryland, USA, June 13-16, 2012.
22. Lee IC, **El-Hattab AW**, Wang J, Li FY, Weng SW, Wong LJ. SURF1-associated Leigh syndrome: A case-series and novel mutations. Poster at the United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meeting. Bethesda, Maryland, USA, June 13-16, 2012.
23. **El-Hattab AW**, Emrick L, Hsu JW, Wong LJ, Jahoor F, Scaglia F, Craigen W. Glucose kinetics in subjects with MELAS syndrome: interim results. Poster at the Society for the Study of Inborn Errors of Metabolism (SSIEM) Annual Symposium. Birmingham, UK, September 4-7, 2012.
24. Segal MM, Williams MS, Gropman AL, Torres AR, Forsyth R, Connolly AM, **El-Hattab AW**, Perlman SJ, Samanta D, Parikh S, Pavlakis SG, Betensky RA, Gospe SM Jr. An Evidence-Based, Open-Database Approach to Pediatric Neurologic Diagnostic Decision Support Reduces Errors in Diagnosis and Testing. Poster at the Child Neurology Society 41st Annual Meeting, Huntington Beach, California, USA, Oct 31 - Nov 3, 2012.
25. **El-Hattab AW**, Emrick L, Hsu JW, Wong LJ, Jahoor F, Scaglia F, Craigen W. Glucose kinetics in subjects with MELAS syndrome: interim results. Platform oral presentation at the American Society of Human Genetics (ASHG) 62nd Annual Meeting. San Francisco, California, USA, November 6-10, 2012.
26. **El-Hattab AW**, Emrick L, K. Williamson K, Craigen W, Scaglia F. The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome. Poster at the European Society of Human Genetics (ESHG) European Human Genetics Conference 2013. Paris, France, June 8-11, 2013.
27. **El-Hattab AW**, Emrick L, K. Williamson K, Craigen W, Scaglia F. The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome. Poster at the United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meeting. Newport Beach, California, USA, June 12-15, 2013.
28. **El-Hattab AW**, Emrick L, K. Williamson K, Craigen W, Scaglia F. The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome. Poster at the 12th International Congress of Inborn Errors of Metabolism (ICIEM 2013). Barcelona, Spain, September 3-6, 2013.
29. **El-Hattab AW**, Hsu JW, Emrick LT, Wong LJ, Craigen WJ, Jahoor F, Scaglia F. Restoration of impaired nitric oxide production in MELAS syndrome with citrulline and arginine supplementation. Invited oral presentation at the 12th International Congress of Inborn Errors of Metabolism (ICIEM 2013). Barcelona, Spain, September 3-6, 2013.
30. **El-Hattab AW**, Emrick L, K. Williamson K, Craigen W, Scaglia F. The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome. Poster at the American Society of Human Genetics (ASHG) 63rd Annual Meeting. Boston, Massachusetts, USA, October 22-26, 2013.
31. **El-Hattab AW**, Emrick LT, Chanprasert S, Hsu JW, Jahoor F, Scaglia F, Craigen WJ. Glucose metabolism derangements in adults with the MELAS mutation m.3243A>G. Poster at the Society for Inherited Metabolic Disorders (SIMD) Annual Meeting. Pacific Grove, California, USA, March 9-12, 2014.
32. **El-Hattab AW**, Emrick LT, Chanprasert S, Hsu JW, Jahoor F, Scaglia F, Craigen WJ. Glucose metabolism derangements in adults with the MELAS mutation m.3243A>G. Poster at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Nashville, Tennessee, USA, March 26-28, 2014.
33. **El-Hattab AW**. Genomic rearrangements. Invited oral presentation at the Basic and Advanced Medical Genetics 6th Annual Review Course. King Fahad Medical City, Riyadh, Saudi Arabia, May 21, 2013.
34. **El-Hattab AW**. Treatment options for inborn errors of metabolism. Invited oral presentation at the Basic and Advanced Medical Genetics 7th Annual Review Course. King Fahad Medical City, Riyadh, Saudi

Arabia, May 21, 2014.

35. **El-Hattab AW.** *BRCA1* and *BRCA2*-related hereditary breast and ovarian cancer. **Invited oral presentation** at the 4th Emirates Gynecology Oncology Group (EGOG)/ Emirates Oncology and Obstetric Society meeting (Gynecological Oncology Updates). Dubai, United Arab Emirates, December 5, 2014.
36. **El-Hattab AW.** Genomic disorders. **Invited oral presentation** at the SEHA International Pediatric Conference. Al-Ain, United Arab Emirates, March 19-21, 2015.
37. **El-Hattab AW.** *BRCA1* and *BRCA2*-related hereditary breast and ovarian cancer. **Invited oral presentation** at the OBS-GYNE Exhibit and Congress, Dubai, United Arab Emirates, 27-29 March 2015.
38. **El-Hattab AW.** Chromosomal abnormalities in Down syndrome. **Invited oral presentation** at the Emirates Down Syndrome International Conference. Dubai, United Arab Emirates, 30 April – 2 May 2015.
39. **El-Hattab AW,** Emrick LT, Chanprasert S, Hsu JW, Almannai M, Craigen WJ, Jahoor F, Scaglia F. Utility of citrulline in treating hypoargininemia in children with MELAS. **Poster** at the United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meeting. Herndon, Virginia, USA, June 17-20, 2015.
40. Charng WL, Karaca E, Akdemir ZB, Gambin T, Atik MM, Jhangiani SN, The Baylor-Hopkins Center for Mendelian Genomics, Gibbs RA, Faqeih EA, Al Asmari A, Saleh MAM, **El-Hattab AW,** James R. Lupski. Rare Variant Identification in Saudi Children with Mendelian Neurologic Diseases. **Poster** at the American Society of Human Genetics (ASHG) Annual Meeting. Baltimore, Maryland, USA, October 6-10, 2015.
41. **El-Hattab AW,** Emrick LT, Chanprasert S, Hsu JW, Almannai M, Craigen WJ, Jahoor F, Scaglia F. Utility of citrulline supplementation in treating hypoargininemia in children with MELAS syndrome. **Poster** at the American Society of Human Genetics (ASHG) Annual Meeting. Baltimore, Maryland, USA, October 6-10, 2015.
42. **El-Hattab AW.** Modalities of genetic testing. **Invited oral presentation** at the SEHA 10th International Pediatric Conference. Abu Dhabi, United Arab Emirates, February 11-13, 2016.
43. **El-Hattab AW,** Emrick LT, Hsu JW, Chanprasert S, Almannai M, Craigen WJ, Jahoor F, Scaglia F. Impaired nitric oxide production in children with MELAS syndrome and the effect of arginine and citrulline supplementation. **Poster** at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Tampa, Florida, USA, March 8-12, 2016.
44. Bayram Y, Harel T, Yesil G, Coban-Akdemir Z, Charng WL, Karaca E, Al Asmari A, Eldomery MK, Hunter JV, Jhangiani SN, Rosenfeld JA, Pehlivan D, **El-Hattab AW,** Saleh MA, LeDuc CA, Muzny D, Boerwinkle E, Baylor-Hopkins Center for Mendelian Genomics, Gibbs RA, WK Chung, Yang Y, Belmont JW, Lupski JR. Mono-allelic and bi-allelic variants in *EMC1*, implicated in ER-mitochondria communication, are associated with neurodegeneration. **Poster** at the European Society of Human Genetics (ESHG) European Human Genetics Conference 2016. Barcelona, Spain, May 21-24, 2016.
45. **El-Hattab AW,** Emrick LT, Hsu JW, Chanprasert S, Almannai M, Craigen WJ, Jahoor F, Scaglia F. Impaired nitric oxide production in children with MELAS syndrome and the effect of arginine and citrulline supplementation. **Platform oral presentation** at the United Mitochondrial Disease Foundation (UMDF) Mitochondrial Medicine Meeting. Seattle, Washington, USA, June 15-18, 2016.
46. Sampath S, **El-Hattab AW.** Autozygosity mapping using SNP based chromosomal microarray: opportunity to increase the clinical diagnostic yield in populations with high consanguinity. **Poster** at the American Society of Human Genetics (ASHG) Annual Meeting. Vancouver, Canada, October 18-22, 2016.
47. **El-Hattab AW.** N-acetylglutamate synthase (NAGS) deficiency: case presentation. **Invited oral presentation** at the 13th Middle East Metabolic Group (MEMG) meeting, Amman, Jordan, October 27-30, 2016.
48. **El-Hattab AW.** Mitochondrial diseases: mechanisms, manifestations, diagnostic approach, and therapeutic options. **Invited oral presentation** at the 4th Arab Pediatric Medical Congress, Dubai, United Arab Emirates, 2-4 March 2017.
49. Abdullatif MA, Al Dhaibani MA, Khassawneh MY, **El-Hattab AW.** Chromosomal microarray in a highly consanguineous population: diagnostic yield, utility of regions of homozygosity, and novel mutations. **Poster** at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Phoenix, Arizona, USA, March 21-25, 2017.
50. Al Zaabi N, Hertecant J, Al Jasmi F, **El-Hattab AW.** Endothelial dysfunction and the effect of arginine and citrulline supplementation in children with mitochondrial diseases: interim results. **Poster** at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Phoenix, Arizona, USA, March 21-25, 2017.
51. **El-Hattab AW,** Emrick LT, Hsu JW, Chanprasert S, Almannai M, Craigen WJ, Jahoor F, Scaglia F. Impaired nitric oxide production in children with MELAS syndrome and the effect of arginine and citrulline supplementation. **Invited oral presentation** at the 13th International Congress of Inborn Errors of Metabolism (ICIEM 2017). Rio de Janeiro, Brazil, September 5-8, 2017.

52. Al Zaabi N, Hertecant J, Al Jasmi F, **El-Hattab AW**. Endothelial dysfunction and the effect of arginine and citrulline supplementation in children with mitochondrial diseases: interim results. **Platform oral presentation** at the 13th International Congress of Inborn Errors of Metabolism (ICIEM 2017). Rio de Janeiro, Brazil, September 5-8, 2017.
53. **El-Hattab AW**, Dai H, Almannai M, Wang J, Wong LJ. Molecular and clinical spectra of FBXL4 deficiency. Poster at the 13th International Congress of Inborn Errors of Metabolism (ICIEM 2017). Rio de Janeiro, Brazil, September 5-8, 2017.
54. **El-Hattab AW**. Recognizable syndromes in the newborn period. **Invited oral presentation** at the 3rd Abu Dhabi International Neonatal Multispecialty Conference, Abu Dhabi, United Arab Emirates, 16-18 November 2017.
55. **El-Hattab AW**. Mitochondrial diseases. **Invited oral presentation** at the 5th Annual Arab International Pediatric Medical Congress, Dubai, United Arab Emirates, 1-4 February 2018.
56. **El-Hattab AW**. Case scenarios in inborn errors of metabolism presenting with hypoglycemia. **Invited oral presentation** at the 5th International Pediatric Conference of Columbia University Medical Center/ NYP Hospital & KidsHeart Medical Center, Al-Ain, United Arab Emirates, 14-16 March 2018.
57. **El-Hattab AW**. Dismorphology terminology and genetic syndromes. **Invited oral presentation** at the 5th International Pediatric Conference of Columbia University Medical Center/ NYP Hospital & KidsHeart Medical Center, Al-Ain, United Arab Emirates, 14-16 March 2018.
58. Hertecant J, Al Jasmi F, **El-Hattab AW**. Endothelial dysfunction and the effect of arginine and citrulline supplementation in children with mitochondrial diseases: interim results. Poster at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Charlotte, North Carolina, 10-14 April 2018.
59. Glinton KE, Benke PJ, Lines MA, Geraghty MT, Chakraborty P, Al-Dirbashi OY, Jiang Y, Kennedy AD, Grotewiel MS, Sutton VR, Elsea SH, **El-Hattab AW**. Disturbed phospholipid metabolism in serine biosynthesis defects revealed by metabolomic profiling. Poster at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Charlotte, North Carolina, 10-14 April 2018.
60. Sirr A, Scott A, Cromie G, Heyesus M, **El-Hattab AW**, Alkuraya F, Dudley A. Yeast surrogate genetic approaches for functional predictions of genetic variants related to inborn errors of metabolism. **Platform oral presentation** at the American Society of Human Genetics (ASHG) Annual Meeting. San Diego, California, USA, October 16-20, 2018.
61. Beetz C, Kdissa A, Karageorgou V, Ameziane N, Bauer P, Suleiman J, Sutton VR, **El-Hattab AW**. A homozygous VPS26C nonsense variant is associated with a novel syndromic phenotype. Poster at the European Society of Human Genetics (ESHG) European Human Genetics Conference 2019. Gothenburg, Sweden, June 15–18, 2019.
62. **El-Hattab AW**. The utility of whole exome sequencing in prenatal settings. **Invited oral presentation** at the United Arab Emirates Fetal Medicine Meeting, Raffles, Dubai, United Arab Emirates, 12 February 2019.
63. Al-Sweel N, Openshaw A, Lamb A, Hong B, **El-Hattab AW**, Andersen E. Typical and Atypical Duplications within the Recurrent Xq28 int22h1/int22h2-Flanked Region to Expand Genotype-Phenotype Correlation. Poster at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting. Seattle, Washington, USA, 2-6 April 2019.
64. **El-Hattab AW**. Dismorphology and genetic syndromes. **Invited oral presentation** at the 7th International Pediatric Conference of Columbia University Medical Center/NYP Hospital & KidsHeart Medical Center, Dubai, United Arab Emirates, 20 - 22 March 2019.
65. **El-Hattab AW**. Case scenarios in Clinical Genetics: Common complaints as presenting manifestations. **Invited oral presentation** at the 7th International Pediatric Conference of Columbia University Medical Center/NYP Hospital & KidsHeart Medical Center, Dubai, United Arab Emirates, 20 - 22 March 2019.
66. **El-Hattab AW**. Metabolic diseases presenting with seizures. **Invited oral presentation** at the 3rd Annual Dubai International Pediatric Neurology Congress, Dubai, United Arab Emirates, 04 – 06 April 2019.
67. **El-Hattab AW**. Modalities of genetic testing. **Invited oral presentation** at the Emirati Pediatric Neurology Network Meeting, Dubai, United Arab Emirates, 19 April 2019.
68. **El-Hattab AW**. Cardiovascular Genetics. **Keynote presentation** at the World Pediatric Cardiology and Cardiovascular Surgery Congress, Dubai, United Arab Emirates, 9-10 September 2019
69. **El-Hattab AW**. Genetic syndromes associated with bone tumors. **Invited oral presentation** at the Emirates Orthopedics Society meeting, Dubai, 25 September 2019
70. **El-Hattab AW**. Overgrowth syndrome. **Invited oral presentation** at the Emirati Pediatric Neurology Network Meeting, Dubai, United Arab Emirates, 4 October 2019.
71. **El-Hattab AW**. Utilities and limitations of various genetic testing modalities. **Invited oral presentation** at the 8th International Pediatric Conference of Columbia University Medical Center/NYP Hospital & KidsHeart

Medical Center, Al-Ain, United Arab Emirates, 9 – 11 October 2019.

72. **El-Hattab AW.** Metabolic emergencies of inborn errors of metabolism. **Invited oral presentation** at the Emirates Pediatric and Neonatal Intensive Care Conference (EPNIC), Al Jalila Children's Specialty Hospital, Dubai, 6-8 November 2019.
73. **El-Hattab AW.** Overgrowth syndrome. **Invited oral presentation** at the 5th Annual International Pediatric Medical Congress, Dubai, United Arab Emirates, 14-16 November 2019.
74. **El-Hattab AW.** From rare disease to LSD. **Invited oral presentation** at the 2nd Gulf Rare Disease Academy, Dubai, United Arab Emirates, 15-16 November 2019.
75. **El-Hattab AW.** Pediatric neuromuscular disorders: genetic basis and management updates. **Invited oral presentation** at 1st Neuropedia Conference on Neuromuscular Diseases in Children NEUROPEDICON 2020, Dubai, United Arab Emirates, 10 January 2020.
76. **El-Hattab AW.** Modalities of genetic testing and the utility of whole exome sequencing. **Invited oral presentation** at the Neurosciences Update, Cleveland Clinic Abu Dhabi, Abu Dhabi, United Arab Emirates, 29 January 2020.
77. **El-Hattab AW.** Modalities of genetic testing: Clinical implications. **Invited oral presentation** at the 8th Pediatric & Neonatal International Conference of University Hospital Sharjah (UHS), Dubai, 13-14 February 2020.