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1. CURRENT POSITION:

- Consultant Pediatrics and Genetics
- Head of Genetics division, Department of Paediatrics,, King Abdulaziz Medical City, Riyadh, Saudi Arabia.
- Associate professor, King Saud bin Abdulaziz University for Health Sciences (KSAU-HS).

2. QUALIFICATIONS:

1. Bachelor of Medicine and Surgery (MBBS), 8/2000
2. SAUDI BOARD in Pediatrics(SSC-Ped) , 2/2006
3. ARAB BOARD in Pediatrics ABHS(CH), 3/2006
4. Master of Health Science(MHSc) in the field of epidemiology and research, 5/2010
5. Canadian Board in Biochemical Genetics (FCCMG), 10/2010
6. Neurometabolic fellowship certificate, 4/ 2011

3. EDUCATION:

Graduated from King Saud University in Riyadh, 2000.

Internship:

From June 17, 2000 – June 6, 2001 in the following hospitals:

- Pediatric Rotation, King Khalid University Hospital (KKUH)
- Surgery Rotation, King Khalid University Hospital (KKUH)
- Obstetrics & Gynecology Rotation, King Khalid University Hospital (KKUH)
- Medicine Rotation, King Khalid University Hospital (KKUH) & Riyadh Medical Complex Hospital.
- Elective Rotation (Radiology & Anesthesia), King Fahad National Guard Hospital (KFNGH) & King Khalid University Hospital.

Residency:

From 2001 – 2005 at King Fahad National Guard Hospital (KFNGH) in the following departments:

- General Pediatrics, 6 months
- Neonatal Intensive Care Unit (NICU), 7 months
- Pediatric Intensive Care Unit (PICU), 4 months

- Pediatric Emergency, 4 months
- Cardiology, 2 months
- Neurology, 3 months
- Endocrinology, 2 months
- Genetic & Metabolic (Elective), 2 months
- Immunology & Allergy, 1 month
- Ambulatory Care, 3 months
- Gastroenterology, 2 months
- Hematology & Oncology, 2 months
- Nephrology, 2 months
- Infectious Diseases, 2 months
- Pulmonology, 1 month

Post Doctoral Experience:

March 2006 – August 2006	Assistant Consultant in general pediatrics at King Fahad National Guard Hospital, Riyadh, Saudi Arabia.
September 2006 – June 2007	Assistant Consultant in Metabolic & Genetic services at King Fahad National Guard Hospital, Riyadh, Saudi Arabia.
July 2007 – October 2010	Clinical Fellow in the Division of Biochemical Diseases at the BC Children's Hospital, Vancouver, BC Canada
November 2010- May 2011	Clinical Fellow in Neurometabolic Diseases at the BC Children's Hospital, Vancouver, BC Canada
June 2011- April 2013	Consultant Paediatrics and Biochemical Genetics,Department of Paediatrics, Genetics division, King Fahad National Guard Hospital, King Abdulaziz Medical City, Riyadh, Saudi Arabia. Assisstant professor, King Saud bin Abdulaziz University for Health Sciences (KSAU-HS).
April 2013- Present	Head of Genetics division, Department of Pediatrics, King Abdulaziz Medical City, Riyadh, Saudi Arabia.

4. ADMINISTRATIVE & ACADEMIC APPOINTMENTS:

- Head of Genetics division, Department of Pediatrics, King Abdulaziz Medical City, Riyadh, Saudi Arabia.
- Chair of Middle East Hyperammonemia and UCDs Scientific Group (MHUSG)
- Vice president of Saudi Society of Medical Genetics
- Head of Newborn Screening program Committee, Department of Pediatrics, King Abdulaziz Medical City, Riyadh, Saudi Arabia.
- Member of advisory committee for National Newborn Screening Program.

- Members of examination committee for Saudi Genetics fellowship program, Saudi Council of Health Specialties , Riyadh, Saudi Arabia.
- I am involved in teaching activities lectures on biochemical genetics and general pediatrics for pediatrics residents and medical students including the following:
    - Resident teaching in preparation for Pediatric examinations (MCQ) and clinical sessions.
    - Bedside teachings for residents.
    - Rotating residents with our specialty.
    - Medical students theoretical lectures.

- **Supervisor for many medical students for research projects including:**
    - Prevalence of Inborn Errors of Metabolism at National Guard Health affairs between 2001-2013.
    - Asparagine Synthetase Deficiency: New Inborn Errors of Metabolism

- Evaluator for several research projects conducted by medical students at King Saud bin Abdulaziz University for Heath Sciences.

5. KEY SYMPOSIUMS AND COURSES ATTENDED RELATED TO MEDICAL EDUCATION:

- The subject matter expert workshops**,** Saudi Commission for health Specialties, Riyadh, November, 2011.
- Problem based learning workshops, Riyadh, January, 2012.

6. KEY SYMPOSIUMS AND COURSES ATTENDED(RELATED TO RESEARCH:

- Introduction to Clinical Research, Feb 2004.
- 1st Pediatric Research Day, June 2004.
- Evidence Based Medicine Workshop, Essential and Advanced modules, March 2004.
- Evidence Based Medicine International Conference, March 2004.
- 2nd Pediatric Research Day, Sep 2005.
- Subspecialty Oriented Evidence Based Medicine, 2005.
- Statistics for Health Research
- Epidemiological Methods
- Systematic review and Meta-analysis
- Leadership in Public Health
- Scientific Basis for Epidemiological Thinking
- Socioeconomic Determinants of Global Health
- Design & Analysis of Clinical Trials
- Health Survey Methods: A Practical Introduction
- Social determinants of global health: Neglected approach to Neglected disease.

7. KEY SYMPOSIUM AND COURSES ATTENDED RELATED TO GENETICS INCLUDING INBORN ERRORS OF METABOLISM:

- 10th International Symposium on Mucopolysaccharidosis , Vancouver, 2008
- Seattle Exchange, Vancouver, 2009 – Medical genetics all day seminar

- ACMG Genetics Review Course, 2009- Dallas, USA
- 11th International Congress of Inborn Error of Metabolism ICIEM 2009, San Diego, USA (present abstract there).
- Medical Genetics 530 course (January 2010-April 2010) - Master level course.
- Pyridoxine dependent epilepsy seminar, Vancouver, 27th July 2010 (I did presentation during that seminar with title: Pyridoxine dependant epilepsy: General background and 2 newly diagnosed adult siblings.
- 2011 ACMG Annual Clinical Genetics Meeting (March 16-20), Vancouver, Canada.
- .International Congress on Prevention of Congential Diseases: Screening Newborns – Current State and Future Challenges. May 2011.Vienna, Austria.
- 3rd Europian Gaucher Leadership Forum. 23-24 September, 2011.
- Intercontinental Merck Serono Symposium on PKU Advanced Control and Treatment Dubai, UAE, May 2012
- Annual symposium, society for study of inborn errors of metabolism. 4-7 September, 2012, Birmingham, UK.
- Lysosomal Storage diseases Middle East Meeting, 3-4 October 2012, Dubai, UAE.
- Annual Symposium, Middle East Metabolic Group. 12-14 December, 2012, Dubai, UAE.
- Annual symposium, society for study of inborn errors of metabolism. 4-7 September, 2013, Barcelona, Spain
- 12th Asian Oceanian Congress of Child Neurology, 14-18 September, 2013, Riyadh, Saudi Arabia
- 10th Middle East Metabolic Group, 12-14 December, 2013, Istanbul, Turkey
- Annual symposium, society for study of inborn errors of metabolism. 2-5 September, 2014, Innsbruck, Austria
- 11th Middle East Metabolic Group, 22-25 October, 2014, Marrakesh, Morocco
- 10th Hot Topics in Neonatal Medicine Conference on 3-5 February, 2015, Jeddah, Saudi Arabia.
- 1st International Neurometabolic Conference, 22-24 February, 2015, Riyadh, Saudi Arabia.
- 1st International workshop on rare diseases, 28th-2nd March, 2015, Riyadh, Saudi Arabia.

8. NATIONAL LECTURES RELATED TO BIOCHEMICAL GENETICS:

- Overview of an inborn errors of metabolism, Om Al Hamam Clinics, Riyadh, November, 2011.
- Energy and Health "The many faces of mitochondrial disease in children, KFSH, Riyadh, February, 2012
- Fabry disease, Al Qassim, March, 2012.
- Hunter disease, Al Qassim, March, 2012.
- Mitochondrial disorders, The 5th Review & Update of Basic Medical Genetic Course , King Fahad Medical City, May 2012.
- Mucopolysaccharidosis, ENT department, King Abdulaziz Medical City, Riyadh, November 2012.
- Chromosomes and more, 3rd Medical Genetics course: review and update, King Fahad Central Hospital, Jazan, January 2013.
- Mitochondrial disorders,4th Medical Genetics course: review and update, Tabouk, April 2013

- Clinical Approach to Metabolic Emergencies, Maternity and Children Hospital, 25-26 December 2013, Makkah, Saudi Arabia
- Web application for DRUG treatment of IEM, 10th Hot Topics in Neonatal Medicine Conference on 3-5 February, 2015, Jeddah, Saudi Arabia.
- Drug treatment of IEM, Systematic Review, 1st International Neurometabolic Conference, 22-24 February, 2015, Riyadh, Saudi Arabia.

9. Contribution to Novel Gene Discovery

Contribute to discovery of 35 novel genes or disease as follow:

Genes	Clinical Significance
PDL1, TUBA3E, INO80, NID1, TSEN15, DMBX1, CLHC1, C12orf4, WDR93, ST7, MATN4, SEC24D, PCDHB4, PTPN23, TAF6, TBCK, FAM177A1, KIAA1109, MTSS1L, XIRP1, KCTD3, CHAF1B, ARV1, ISCA2, PTRH2, GEMIN4, MYOCD, PDPR, DPH1, NUP107, TMEM92, EPB41L4A, and FAM120AOS)	Intellectual disability
GOLGA2	Neuromuscular disorder
ISCA2	Mitochondrial disorder
AGTPBP1	Spinal Muscular Atrophy

10. PUBLICATIONS:

1. **Alfadhel M,** AlAmir A. Senior – Loken Syndrome in a Saudi child. Saudi Journal of Kidney Diseases and Transplantation. 2008; 19(3):443-445.

2. Leitch CC, Zaghloul NA, Davis EE, Stoetzel C, Diaz-Font A, Rix S, **Alfadhel M**, Lewis RA, Eyaid W, Banin E, Dollfus H, Beales PL, Badano JL, Katsanis N. Hypomorphic mutations in syndromic encephalocele genes are associated with Bardet-Biedl syndrome, Nature Genetics. 2008;40(4):443-448.

3. **Alfadhel M,** Pugash D, Robinson AJ, Murphy JJ, Senger C, Afshar K, Armstrong L. Pre- and Postnatal Findings in a Boy with Duplication of the Bladder and Intestine. Report and Review. American Journal of Medical Genetics Part A. 2009; 149A (12):2795-2802. Review.

4. **Alfadhel M**, LillquistYP, Waters PJ, Sinclair G, Struys E, McFadden D, Hendson G, Hyams L, Shoffner J, D Vallance HD. Infantile cardioencephalopathy due to a COX 15 gene defect: Report and review.. Am J Med Genet A. 2011(4):840-844. (Abstract presented in 11th International Congress of Inborn Error of Metabolism). Review.

5. **Alfadhe M,** Sirrs S. Enzyme replacement therapy for Fabry disease: some answers but more questions Ther Clin Risk Manag. 2011;7:69-82. Review.

6. Mattman A, Sirrs S, Michelle MM, , Salvarinova R, **Alfadhel M**, Lillquist YP. Mitochondrial Disease Clinical Manifestations: An overview BCMJ, 2011; 53 (4): 183-187.

7. **Alfadhel M,** Yong SL, Lillquist YP, Langlois S. Precocious Puberty in Two Girls with PEHO Syndrome: a Clinical Feature Not Previously Described. Child Neurol. 2011 Jul;26(7):851-857. Review.

8. **Alfadhel M**, Lillquist YP, Davis S, Junker AK, Stockler-Ipsiroglu S. Eighteen Year Follow-up of a Patient with Cobalamin F disease (cblF) and Review of Case. Am J Med Genet A. 2011 Oct; 155(10):2571-2577.

9. **Alfadhel M,** Sirrs S, Waters PJ, Szeitz A, Struys E, Coulter-Mackie M, Stockler-Ipsiroglu. Variability of phenotype in two sisters with pyridoxine dependent epilepsy. Can J Neurol Sci. 2012 Jul;39(4):516-519.

10. **Alfadhel M,** Alhasan KA, Alotaibi M, Al Fakeeh K. Extreme intrafamilial variability of Saudi brothers with primary hyperoxaluria type 1. Ther Clin Risk Manag. 2012;8:373-376.

11. Eyaid W, Al Harbi T, Anazi S, Wamelink MM, Jakobs C, Al Salammah M, Al Balwi M, **Alfadhel M**, Alkuraya FS. Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. J Inherit Metab Dis. 2013 Jan 12.

12. **Alfadhel M**, Almuntashri M, Jadah RH, Bashiri FA, Al Rifai MT, Al Shalaan H, Al Balwi M, Al Rumayan A, Eyaid W, Al-Twaijri W. Biotin-responsive basal ganglia disease should be renamed biotin-thiamine-responsive basal ganglia disease: a retrospective review of the clinical, radiological and molecular findings of 18 new cases. Orphanet J Rare Dis. 2013 Jun 6;8:83. doi: 10.1186/1750-1172-8-83. (Highly Accessed)

13. **Alfadhel M**, Al-Thihli K, Moubayed H, Eyaid W, Al-Jeraisy M. Drug treatment of inborn errors of metabolism: a systematic review. Arch Dis Child. 2013 Jun;98(6):454-61.

14. Jassim N, Alghaihab M, Al Saleh S, **Alfadhel M**, Wamelink MM, Eyaid W. Pulmonary Manifestations of in Patient with Transaldolase Deficiency. JIMD Rep. 2013 Jul 12.

15. **Alfadhel M**, AlShehhi W, Alshaalan H, Al Balwi M, Eyaida W. Mucolipidosis II: first report from Saudi Arabia. Ann Saudi Med. 2013 Jul-Aug;33(4):382-6.

16. Tabarki B, Al-Hashem A, **Alfadhel M.** Biotin-Thiamine-Responsive Basal Ganglia Disease. In: Pagon RA, Adam MP, Bird TD, Dolan CR, Fong CT, Smith RJH, Stephens K, editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2013

17. 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 Is Associated with Neonatal Cholestasis and Liver Failure. J Pediatr. 2013 Dec 7. pii: S0022-3476(13)01378-4

18. **Alfadhel M**, Kattan R. Aromatic amino Acid decarboxylase deficiency not responding to pyridoxine and bromocriptine therapy: case report and review of response to treatment. J Cent Nerv Syst Dis. 2014 Jan 7;6:1-5.

19. Sarkhy AA, Al-Sunaid A, Abdullah A, **AlFadhel M**, Eiyad W. A novel MPV17 gene mutation in a Saudi infant causing fatal progressive liver failure. Ann Saudi Med. 2014;34(2):175-8

20. Alrifai MT, AlShaya MA, Abulaban A, **Alfadhel M**. Hereditary neurometabolic causes of infantile spasms in 80 children presenting to a tertiary care center. Pediatr Neurol. 2014 Sep;51(3):390-7

21. **Alfadhel M**, Saleh N, Alenazi H, Baffoe-Bonnie H. Acute intermittent porphyria caused by novel mutation in HMBS gene, misdiagnosed as cholecystitis. Neuropsychiatr Dis Treat. 2014 Nov 12;10:2135-7.
22. Al-Hassnan ZN, Al-Dosary M, **Alfadhel M**, Faqeih EA, Alsagob M, Kenana R, Almass R, Al-Harazi OS, Al-Hindi H, Malibari OI, Almutari FB, Tulbah S, Alhadeq F, Al-Sheddi T, Alamro R, AlAsmari A, Almuntashri M, Alshaalan H, Al-Mohanna FA, Colak D, Kaya N.
23. Alazami AM, Patel N, Shamseldin HE, Anazi S, Al-Dosari MS, Alzahrani F, Hijazi H, Alshammari M, Aldahmesh MA, Salih MA, Faqeih E, Alhashem A, Bashiri FA, Al-Owain M, Kentab AY, Sogaty S, Al Tala S, Temsah MH, Tulbah M, Aljelaify RF, Alshahwan SA, Seidahmed MZ, Alhadid AA, Aldhalaan H, AlQallaf F, Kurdi W, **Alfadhel M,** Babay Z, Alsogheer M, Kaya N, Al-Hassnan ZN, Abdel-Salam GM, Al-Sannaa N, Al Mutairi F, El Khashab HY, Bohlega S, Jia X, Nguyen HC, Hammami R, Adly N, Mohamed JY, Abdulwahab F, Ibrahim N, Naim EA, Al-Younes B, Meyer BF, Hashem M, Shaheen R, Xiong Y, Abouelhoda M, Aldeeri AA, Monies DM, Alkuraya FS. Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. Cell Rep. 2015 Jan 13;10(2):148-61.
24. **Alfadhel M**, Alrifai MT, Trujillano D, Alshaalan H, Al Othaim A, Al Rasheed S, Assiri H, Alqahtani AA, Alaamery M, Rolfs A, Eyaid W. Asparagine Synthetase Deficiency: New Inborn Errors of Metabolism. JIMD Rep. 2015 Feb 8.
25. Tabarki B, **Alfadhel M**, AlShahwan S, Hundallah K, AlShafi S, AlHashem A. Treatment of biotin-responsive basal ganglia disease: Open comparative study between the combination of biotin plus thiamine versus thiamine alone. Eur J Paediatr Neurol. 2015 Jun 12. pii: S1090-3798(15)00105-1. doi: 10.1016/j.ejpn.2015.05.008.

26. Hollink IH, **Alfadhel M**, Al-Wakeel AS, Ababneh F, Pfundt R, de Man SA, Jamra RA, Rolfs A, Bertoli-Avella AM, van de Laar IM. Broadening the phenotypic spectrum of pathogenic LARP7 variants: two cases with intellectual disability, variable growth retardation and distinct facial features. J Hum Genet. 2015 Nov 26.

27. Imtiaz F, Al-Mubarak BM, Al-Mostafa A, Al-Hamed M, Allam R, Al-Hassnan Z, Al-Owain M, Al-Zaidan H, Rahbeeni Z, Qari A, Faqeih EA, Alasmari A, Al-Mutairi F, **Alfadhel M**, Eyaid WM, Rashed MS, Al-Sayed M. Spectrum of Mutations in 60 Saudi Patients with Mut Methylmalonic Acidemia. JIMD Rep. 2015 Nov 29

28. Shamseldin HE, Bennett AH, **Alfadhel M**, Gupta V, Alkuraya FS. GOLGA2, encoding a master regulator of golgi apparatus, is mutated in a patient with a neuromuscular disorder. Hum Genet. 2016 Feb;135(2):245-51. doi: 10.1007/s00439-015-1632-8.

29. Alazami AM, Al-Qattan SM, Faqeih E, Alhashem A, Alshammari M, Alzahrani F, Al-Dosari MS, Patel N, Alsagheir A, Binabbas B, Alzaidan H, Alsiddiky A, Alharbi N, **Alfadhel M**, Kentab A, Daza RM, Kircher M, Shendure J, Hashem M, Alshahrani S, Rahbeeni Z, Khalifa O, Shaheen R, Alkuraya FS. Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Hum Genet. 2016 May;135(5):525-40.

30. **Alfadhel M.,** Mutairi FA, Makhseed N, Jasmi FA, Al-Thihli K, Al-Jishi E, AlSayed M, Al-Hassnan ZN, Al-Murshedi F, Häberle J, Ben-Omran T. Guidelines for acute management of hyperammonemia in the Middle East region. Ther Clin Risk Manag. 2016 Mar 31;12:479-87.

31. Alrifai MT, **Alfadhel M**. Worsening of Seizures after Asparagine Supplementation in a Child with Asparagine Synthetase Deficiency. Pediatr Neurol. 2016 May;58:98-100.

32. Tarailo-Graovac M, Shyr C, Ross CJ, Horvath GA, Salvarinova R, Ye XC, Zhang LH, Bhavsar AP, Lee JJ, Drögemöller BI, Abdelsayed M, **Alfadhel M,** Armstrong L, Baumgartner MR, Burda P, Connolly MB, Cameron J, Demos M, Dewan T, Dionne J, Evans AM, Friedman JM, Garber I, Lewis S, Ling J, Mandal R, Mattman A, McKinnon M,

Michoulas A, Metzger D, Ogunbayo OA, Rakic B, Rozmus J, Ruben P, Sayson B, Santra S, Schultz KR, Selby K, Shekel P, Sirrs S, Skrypnyk C, Superti-Furga A, Turvey SE, Van Allen MI, Wishart D, Wu J, Wu J, Zafeiriou D, Kluijtmans L, Wevers RA, Eydoux P, Lehman AM, Vallance H, Stockler-Ipsiroglu S, Sinclair G, Wasserman WW, van Karnebeek CD. Exome Sequencing and the Management of Neurometabolic Disorders. N Engl J Med. 2016 Jun 9;374(23):2246-55.

33. Sass JO, Gemperle-Britschgi C, Tarailo-Graovac M, Patel N, Walter M, Jordanova A, **Alfadhel M**, Barić I, Çoker M, Damli-Huber A, Faqeih EA, García Segarra N, Geraghty MT, Jåtun BM, Kalkan Uçar S, Kriewitz M, Rauchenzauner M, Bilić K, Tournev I, Till C, Sayson B, Beumer D, Ye CX, Zhang LH, Vallance H, Alkuraya FS, van Karnebeek CD. Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic OPLAH mutations: 20 new mutations in 14 families. Mol Genet Metab. 2016 Sep;119(1-2):44-9.

34. **Alfadhel M**, Nashabat M, Qahtani HA, Alfares A, Mutairi FA, Shaalan HA, Douglas GV, Wierenga K, Juusola J, Alrifai MT, Arold ST, Alkuraya F, Ali QA. Mutation in SLC6A9 encoding a glycine transporter causes a novel form of non-ketotic hyperglycinemia in humans. Hum Genet. 2016 Aug 1

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36. Alwadei AH, Benini R, Mahmoud A, Alasmari A, Kamsteeg EJ, **Alfadhel M**. Loss-of-function mutation in RUSC2 causes intellectual disability and secondary microcephaly. Dev Med Child Neurol. 2016 Sep 9

37. **Alfadhel M**, Benmeakel M, Hossain MA, Al Mutairi F, Al Othaim A, Alfares AA, Al Balwi M, Alzaben A, Eyaid W. Thirteen year retrospective review of the spectrum of inborn errors of metabolism presenting in a tertiary center in Saudi Arabia. Orphanet J Rare Dis. 2016 Sep 15;11(1):126.

38. Trujillano D, Bertoli-Avella AM, Kumar Kandaswamy K, Weiss ME, Köster J, Marais A, Paknia O, Schröder R, Garcia-Aznar JM, Werber M, Brandau O, Calvo Del Castillo M,

Baldi C, Wessel K, Kishore S, Nahavandi N, Eyaid W, Al Rifai MT, Al-Rumayyan A, Al-Twaijri W, Alothaim A, Alhashem A, Al-Sannaa N, Al-Balwi M, **Alfadhel M**, Rolfs A, Abou Jamra R. Clinical exome sequencing: results from 2819 samples reflecting 1000 families. Eur J Hum Genet. 2017 Feb;25(2):176-182.

39. **Alfadhel M,** Nashabat M, Abu Ali Q, Hundallah K. Mitochondrial iron-sulfur cluster biogenesis from molecular understanding to clinical disease. Neurosciences (Riyadh). 2017 Jan;22(1):4-13.
40. Obaid A, Nashabat M, Al Fakeeh K, Al Qahtani AT, **Alfadhel M** . Delineation of cystinuria in Saudi Arabia: A case series. BMC Nephrol. 2017 Feb 6;18(1):50.
41. Alfakeeh K, Azar M, **Alfadhel M**, Abdullah AM, Aloudah N, Alsaad KO. Rare genetic variant in the CFB gene presenting as atypical hemolytic uremic syndrome and immune complex diffuse membranoproliferative glomerulonephritis, with crescents, successfully treated with eculizumab. Pediatr Nephrol. 2017 Feb 16.
42. **Alfadhel M**, Al Othaim A, Al Saif S, El Mutairi F, Alsayed M, Rahbeeni Z, Alzaidan H, Alowain M, Al-Hassnan Z, Saeedi M, Aljohery S, Alasmari A, Faqeih E, Alwakeel M, AlMashary M, Almohameed S, Alzahrani M, Migdad A, Al-Dirbashi OY, Rashed M, Alamoudi M, Jacob M, Alahaidib L, El-Badaoui F, Saadallah A, Alsulaiman A, Eyaid W, Al-Odaib A. Expanded Newborn Screening Program in Saudi Arabia: Incidence of screened disorders. J Paediatr Child Health. 2017 Mar 24
43. Alfares A, **Alfadhel M**, Wani T, Alsahli S, Alluhaydan I, Al Mutairi F, Alothaim A, Albalwi M, Al Subaie L, Alturki S, Al-Twaijri W, Alrifai M, Al-Rumayya A, Alameer S, Faqeeh E, Alasmari A, Alsamman A, Tashkandia S, Alghamdi A, Alhashem A, Tabarki B, AlShahwan S, Hundallah K, Wali S, Al-Hebbi H, Babiker A, Mohamed S, Eyaid W, Zada AAP. A multicenter clinical exome study in unselected cohorts from a consanguineous population of Saudi Arabia demonstrated a high diagnostic yield. Mol Genet Metab. 2017 Jun;121(2):91-95.

44. Al Mutairi F, Shamseldin HE, **Alfadhel M**, Rodenburg RJ, Alkuraya FS. A lethal neonatal phenotype of mitochondrial short-chain enoyl-CoA hydratase-1 deficiency. Clin Genet. 2017 Apr;91(4):629-633

45. Alshenqiti A, Nashabat M, AlGhoraibi H, Tamimi O, **Alfadhel M**. Pulmonary hypertension and vasculopathy in incontinentia pigmenti: a case report. Ther Clin Risk Manag. 2017 May 9;13:629-634

46. Nashabat M, Maegawa G, Nissen PH, Nexo E, Al-Shamrani H, Al-Owain M, **Alfadhel M**. Long-term Outcome of 4 Patients With Transcobalamin Deficiency Caused by 2 Novel TCN2 Mutations. J Pediatr Hematol Oncol. 2017 May 22.

47. Hossain MA, Obaid A, Rifai M, Alem H, Hazwani T, Al Shehri A, **Alfadhel M**, Eto Y, Eyaid W. Early onset of Fazio-Londe syndrome: the first case report from the Arabian Peninsula. Hum Genome Var. 2017 May 25.

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landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. Hum Genet. 2017 Aug;136(8):921-939.

49. Alsubhi S, Alhashem A, Faqeih E, **Alfadhel M**, Alfaifi A, Altuwaijri W, Alsahli S, Aldhalaan H, Alkuraya FS, Hundallah K, Mahmoud A, Alasmari A, Mutairi FA, Abduraouf H, AlRasheed L, Alshahwan S, Tabarki B. Congenital disorders of glycosylation: The Saudi experience. Am J Med Genet A. 2017 Jul 25.
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51. Ortigoza-Escobar JD, **Alfadhel M**, Molero-Luis M, Darin N, Spiegel R, de Coo IF, Gerards M, Taylor RW, Artuch R, Nashabat M, Rodríguez-Pombo P, Tabarki B, Pérez-Dueñas B; Thiamine Deficiency Study Group. Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Ann Neurol. 2017 Jul 18.
52. Windpassinger C, Piard J, Bonnard C, **Alfadhel M**, Lim S, Bisteau X, Blouin S, Ali NB, Ng AYJ, Lu H, Tohari S, Talib SZA, van Hul N, Caldez MJ, Van Maldergem L, Yigit G, Kayserili H, Youssef SA, Coppola V, de Bruin A, Tessarollo L, Choi H, Rupp V, Roetzer K, Roschger P, Klaushofer K, Altmüller J, Roy S, Venkatesh B, Ganger R, Grill F, Ben Chehida F, Wollnik B, Altunoglu U, Al Kaissi A, Reversade B, Kaldis P. CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. Am J Hum Genet. 2017 Sep 7;101(3):391-403.

11. PROPOSALS:

- Biotin Thiamine Responsive Basal Ganglia Disease- Pilot study toward newborn screening program for treatable neurometabolic disorder.

12. TUTOR AT NATIONAL COURSES:

- Introduction to clinical research course Dammam 10-11 October 2011.
- Introduction to clinical research course Riyadh November and March 2012

13. INTERNATIONAL ORAL PRESENTATION:

- Filling the gaps, benefit minus harm of an extended newborn screening. Fabry disease model. International Congress on Prevention of Congenital Diseases: Screening Newborns – Current State and Future Challenges. May 2011.Vienna, Austria.
- Mitochondrial diseases and disorders, 12th Asian Oceanian Congress of Child Neurology, 14-18 September, 2013, Riyadh, Saudi Arabia
- Drug Treatment of Inborn Errors of Metabolism, 10th Middle East Metabolic Group symposium, 12-14 December, 2013, Istanbul, Turkey
- Web app application for drugs used in treatment of Inborn Errors of Metabolism, 1st International Saudi Society of Medical Genetics Symposium, 30th -1st May, 2014, Riyadh, Saudi Arabia
- Asparagine Synthetase Deficiency, Annual symposium, society for study of inborn errors of metabolism. 2-5 September, 2014, Innsbruck, Austria

14. POSTER PRESENTATION:

- **Alfadhel M**, LillquistYP, Waters PJ, Sinclair G, Struys E, McFadden D, Hendson G, Hyams L, Shoffner J, D Vallance HD. Infantile cardioencephalopathy due to a COX 15 gene defect: Report and review. 11th International Congress of Inborn Error of Metabolism ICIEM 2009, San Diego, USA
- Croft J, Clark S, Hind H, L. Hikin, **Alfadhel M,** Al Balwi MA, Manning NJ, etal**.** Diagnostic Difficulties in Glutaryl-CoA Dehydrogenase Deficiency. Annual symposium, society for study of inborn errors of metabolism. 4-7 September**,** 2013, Barcelona, Spain
- **Alfadhel M**, Al-Thihli K, Moubayed H, Eyaid W, Al-Jeraisy M. Drug treatment of inborn errors of metabolism: a systematic review. Annual symposium, society for study of inborn errors of metabolism. 4-7 September, 2013, Barcelona, Spain
- **Alfadhel M**, Almuntashri M, Jadah RH, Bashiri FA, Al Rifai MT, Al Shalaan H, Al Balwi M, Al Rumayan A, Eyaid W, Al-Twaijri W. Biotin-responsive basal ganglia disease should be renamed biotin-thiamine-responsive basal ganglia disease: a retrospective review of the clinical, radiological and molecular findings of 18 new cases.Annual symposium, society for study of inborn errors of metabolism. 4-7 September**,** 2013, Barcelona, Spain

15. CHAPTER IN THE BOOK:

- Human genetics and dysmorphology chapter and metabolic disorders chapter in H and M MCQ'S in Paediatrics book published by Huda Thabet Al Hussamy and Mansur Naser Al Howasi, 2013
- Tabarki B, Al-Hashem A, Alfadhel M. Biotin-Thiamine-Responsive Basal Ganglia Disease. In: Pagon RA, Adam MP, Bird TD, Dolan CR, Fong CT, Smith RJH, Stephens K, editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2013
- Nutrition Protocol for Mangement of Inborn Errors of Metabolism. **Majid Alfadhel**, Fuad Almutairi, Lina Alohali
- Manual of Establishing a Newborn Screening Program. Diagnosis and Management of Screened Disorders. **Majid Alfadhel**, Saif Alsaif , Abdullah Alzaben
- Protocol for Lysosomal Diseases Patients Started on Enzyme Replacement Therapy. **Majid Alfadhel**, Hiba Moubayed

16. MODERATOR AT NATIONAL SYMPOSIUM:

- Workshop on prevention of genetic diseases, KFSH, Riyadh, Saudi Arabia, 4th April 2012.
- 12th Asian Oceanian Congress of Child Neurology, 14-18 September, 2013, Riyadh, Saudi Arabia
- 5th Medical Genetics Workshop review and update, 25-26 December 2013, Maternity and Children Hospital, Makkah, Saudi Arabia
- 12th Medical Genetics Workshop 5th Medical Genetics Workshop review and update, 28-29December 2016, Maternity and Children Hospital, Al Baha, Saudi Arabia

17. LECTURES FOR MEDICAL STUDENTS AT KING SAUD BIN ABDULAZIZ UNIVERSITY FOR HEALTH SCIENCES (KSAU-HS) :

- Genes and cardiovascular diseases. June 2012 (for female section).
- Genes and cardiovascular diseases. June 2012 (for male section).
- Continuous teaching Round, November 2012 (for male section).

18. LECTURES FOR PEDIATRICS RESIDENTS:

- Case scenarios for patients with inborn error of metabolism. Yearly
- Weekly sessions about Inborn Errors of Metabolism.

19. COMMITTEES:

- Newborn screening committee at King Fahad National Guard Hospital, King Abdulaziz Medical City, Riyadh, Saudi Arabia since June 2011 – present.
- Pediatrics mortality committee at King Fahad National Guard Hospital, King Abdulaziz Medical City, Riyadh, Saudi Arabia since February 2012 – present

20. INVENTION & CREATION:

- Web app for Medication used in treatment of inborn errors of metabolism called IEM Drugs

21. AWARDS & PRIZES:

- The best resident in 2005 while I was R4.
- The best case report at Pediatrics Clinical Research Day, King Fahad National Guard Hospital (KFNGH) in Riyadh, Saudi Arabia in 2005.
- Certificate of appreciation in the Pediatrics Emergency Medicine Course in 2005.
- Academic excellence award from Saudi Cultural Bureau, 2008 and 2010
- Best Poster at 9th Middle East Metabolic Group (MEMG) conference, 12-14 December, 2012, Dubai, UAE.
- Best Poster at 5th Pediatric Research day, King Abdulaziz Medical City, Riyadh, Saudi Arabia.31 December, 2012, Riyadh, KSA

- Most Active researcher prize at 5th Pediatric Research day, King Abdulaziz Medical City, Riyadh, Saudi Arabia. 31 December, 2012, Riyadh, KSA

20. **LANGUAGES:** Arabic and English.