**Fuad Fazei N. Al Mutairi. MD**

Deputy of Genetics and Precision Medicine Department. Associate Professor of Pediatrics, Consultant of Medical Genetics, Riyadh, Saudi Arabia, +966557894444, Email: dr.fmutairi@gmail.com

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| Professional Summary |

To promote health care for children in community, especially about genetic disorders and advocate for their rights in having good health care systems in all aspects (prevention to advance health care) and work as a team to help all health care workers to gain needed knowledge and research skills in clinical genetics.

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| Experience |

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| King Abdulaziz Medical City **|** Riyadh**,** Saudi Arabia | September 2014 - Current |

* Deputy of Genetics and Precision Medicine Department (MARCH 2022- Current)
* Consultant of Pediatrics and Medical Genetics, Genetics and Precision Medicine Department at KASCH-KAMC.

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| Associate Professor of PediatricsKing Saud bin Abdulaziz University for Health Sciences **|** Riyadh**,** Saudi Arabia | September 2019 - Current |

* Consultant of Pediatrics and Medical Genetics, Genetics and Precision Medicine Department at KASCH-KAMC.

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| Assistant Professor of PediatricsKing Saud bin Abdulaziz University for Health Sciences **|** Riyadh**,** Saudi Arabia | May 2015 - September 2019 |

* Consultant of Pediatrics and Medical Genetics, Genetics and Precision Medicine Department at KASCH-KAMC.

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| Consultant of Pediatrics and Head **of Medical Genetics Division** Prince Sultan Military Medical City **|** Riyadh**,** Saudi Arabia | April 2012 - September 2014 |

* Consultant of Pediatrics, Head of Medical Genetics Division

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| Clinical FellowBC Children Hospital University of British Columbia **|** vancouver **,** Canada | March 2010 - March 2012 |

* Clinical Fellow

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| Senior RegistrarPrince Sultan Military Medical City **|** Riyadh**,** Saudi Arabia | January 2008 - February 2010 |

* Medical Genetics Division, Department of Pediatrics at PSMMC, Riyadh Saudi Arabia.

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| Pediatrics ResidentPrince Sultan Military Medical City **|** Riyadh**,** Saudi Arabia | October 2002 - January 2008 |

* Pediatrics Resident

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| Education |

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| Clinical Biochemical Genetics  The University of British Columbia, Vancouver, British Columbia, Canada | February 2012 |

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| Saudi Board in Pediatrics, (SSC-Ped)  Saudi Commission for Health Specialties, Riyadh, Saudi Arabia | January 2008 |

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| MBBS,  King Saud University, Riyadh, Saudi Arabia | April 2001 |

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| Memberships |

* Member of the Board of Directors of the Saudi Society of Medical Genetics (2012-Now).
* Member of the Board and Co-founder of Middle East and North Africa- Medical Genetic Association. MENA-MGA (2022-Now)
* Member of Institutional Review Board at King Abdulaziz Medical City (2021-Now)
* Member of KASCH Precision Medicine Executive Board Committee (2021-Now)
* Member of Middle East Hyperammonemia Expert Group (2013-Now).
* Member of Middle East Medical Metabolic Group (MEMG) (2009-2018).
* Member of Society of Inborn Error of Metabolism (SSIEM) (2012- Now).
* Member of Newborn Screening Advisory Board, MOH, Saudi Arabia 2014 -2016.
* Regional leader for the Rare Diseases Initiatives from Excellence in Pediatrics 2013-2014.

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| **ACADEMIC POSITIONS** |

* Chairman of SCHS Supervisory Board for Medical Genetic Fellowship Program (2021-Now)
* Member of SCHS Supervisory Board for Medical Genetic Fellowship Program (2016-2021)
* Program Director, Medical Genetics Fellowship Program at KAMC (2016-Dec 2022)
* Certified trainer in postgraduate medical training (Sep 2021-now)
* Member of Institutional Training Committee, KAMC (2016-2022)
* Pediatrics block Chief coordinator for medical Students, King Saud bin Abdulaziz University for Health Sciences (KSAU-HS) (2018-2020)
* Pediatrics block Co-coordinator for medical Students, King Saud bin Abdulaziz University for Health Sciences (KSAU-HS) (2015- 2018)
* Formal Clinical Tutor of Pediatrics Residency program in PSMMC (2012- 2014).

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| **MAIN GRANT RESEARCH PROJECTS** |
| * Certified GOOD CLINICAL PRACTICE (GCP) from NIDA Clinical Trials Network (Jan 2022) * NRC23R/145/03 - Implementation of new guidelines for dietary management of propionic acidemia and methylmalonic acidemia: A prospective cohort study (Co-Investigator) Ongoing * SCT22R/004/04 Sponsored multicenter clinical trial reLiver-1 and reLiver-2 studies (Principal Investigator) * RC20/453/R Genetic and Rare Disease Registry (Co-Investigator) Ongoing * RC20/621/R Outcomes of cases with elevated 3-hydroxyisovaleryl carnitine - Report from the Inborn Errors of Metabolism Information System (Principal Investigator) Ongoing * RC19/315/R Saudi Genome Database Proposal (Co-Investigator) Completed * RC19/120/R Genetic and Rare Diseases (GARD) Programme Saudi Arabia: A Step Towards Therapeutic Genomics (Co-Investigator) Ongoing * RC19/115/R Preventative Genome medicine for inherited genetic disorder in Saudi Arabia (Co-Investigator) Ongoing * RC18/313/R Phenotypic characterization of Dihydrolipoamide Dehydrogenase Deficiency (Principal Investigator) Completed * RC17/103/R Phenotypic and Molecular Spectrum of Aicardi-Goutières Syndrome: A Study of 24 Patients (Principal Investigator) Completed * RC16/211/R2 Identification of disease-causing mutations by WGS that are not detected by WES in families with hereditary disorders (Co-Investigator) Completed * RC13/116/R Randomized Multicentre Comparative Trial to Evaluate the Long Term Effectiveness of the use of Carbaglu® in Patients with Propionic Acidemia (PA) or Methylmalonic Acidemia (MMA) (Co-Investigator) Completed |  |

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| Publications |

1. Maroofian R, Kaiyrzhanov R, Cali E, Zamani M, Zaki MS, Ferla M, Tortora D, Sadeghian S, Saadi SM, Abdullah U, Ghayoor Karimiani E, Efthymiou S, Yeşil G, Alavi S, Al Shamsi AM, Tajsharghi H, Abdel-Hamid MS, Saadi NW, **Al Mutairi F,** ... Biallelic MED27 variants lead to variable ponto-cerebello-lental degeneration with movement disorders. Brain. 2023 Jul 30:awad257.
2. Langhammer F, Maroofian R, Badar R, Gregor A, .., **Al Mutairi F**, Tamim AM, Abdulwahab F, Alkuraya FS, Khouj EM, Alvi JR, Sultan T, Hashemi N, Karimiani EG, Ashrafzadeh F, Imannezhad S, Efthymiou S, Houlden H, Sticht H, Zweier C. Genotype-phenotype correlations in RHOBTB2-associated neurodevelopmental disorders. Genet Med. 2023 Aug;25(8):100885.
3. Alzahrani A, Alshalan M, Alfurayh M, Bin Akrish A, Alsubeeh NA, **Al Mutairi F.** Case Report: Clinical delineation of CACNA1D mutation: New cases and literature review. Front Neurol. 2023 Apr 14;14:1131490. doi: 10.3389/fneur.2023.
4. Balsam AlMaarik, Taghrid Aloraini, Roselyn Paclejan, Mohammed Balwi, Lamia Alsubaie, Abdulrahman Alswaid, Wafaa Eyiad, **Fuad Al Mutairi**, Faroug Ababneh, Majid Alfadhel, Ahmed A. Alfares. Supplementary testing after negative or inconclusive exome sequencing results. JBCGenetics. 2023; 6(1): 1-13
5. **Fuad Al Mutairi**, Meshal Alberreet, Lara Alkuhaimi, Khalid Aleisa, Rana Almana, Asma Awadalla. Coexistence of atopic dermatitis and thrombocytosis: diagnostic odyssey: a case reportJBCGenetics. 2023; 6(1): 80-84
6. Alfadhel M, Umair M, Al Tuwaijri A, **Al Mutairi F**. A Patient with Coarse Facial Features and Molecular Odyssey: Lessons Learned and Best Practice. Clin Chem. 2023 Jan 4;69(1):17-20.
7. Calì E, Lin SJ, Rocca C, Sahin Y, Al Shamsi A, **Al Mutairi F**, …. Gerard B, Zifarelli G, Beetz C, Fortuna S, Soler M, Valente EM, Varshney G, Maroofian R, Salpietro V, Houlden H. A homozygous MED11 C-terminal variant causes a lethal neurodegenerative disease. Genet Med. 2022 Oct;24(10):2194-2203.
8. Marafi D, Kozar N, Duan R, Bradley S, Yokochi K, **Al Mutairi F**, … Jhangiani SN, Gibbs RA, Miyatake S, Matsumoto N, Wagstaff LJ, Posey JE, Lupski JR, Meijer D, Wagner M. A reverse genetics and genomics approach to gene paralog function and disease: Myokymia and the juxtaparanode. Am J Hum Genet. 2022 Sep 1;109(9):1713-1723.
9. Moreno Traspas R, Teoh TS, Wong PM, Maier M, Chia CY, Lay K, Ali NA, Larson A, **Al Mutairi F,** Bertoli-Avella AM, Vincent M, Girisha KM, Reversade B. Loss of FOCAD, operating via the SKI messenger RNA surveillance pathway, causes a pediatric syndrome with liver cirrhosis. Nat Genet. 2022 Aug;54(8):1214-1226.
10. Alfadhel M, Abadel B, Almaghthawi H, Umair M, Rahbeeni Z, Faqeih E, Almannai M, Alasmari A, Saleh M, Eyaid W, Alfares A, **Al Mutairi F.** HMG-CoA Lyase Deficiency: A Retrospective Study of 62 Saudi Patients. Front Genet. 2022 May 13;13:880464.
11. Aloraini T, Aljouie A, Alniwaider R, Alharbi W, Alsubaie L, AlTuraif W, Qureshi W, Alswaid A, Eyiad W, **Al Mutairi F,** Ababneh F, Alfadhel M, Alfares A. The variant artificial intelligence easy scoring (VARIES) system. Comput Biol Med. 2022 Jun;145:105492.
12. Aloraini T, Alsubaie L, Alasker S, Al Muitiri A, Alswaid A, Eyiad W, **Al Mutairi F**, Ababneh F, Alfadhel M, Alfares A. The rate of secondary genomic findings in the Saudi population. Am J Med Genet A. 2022 Jan;188(1):83-88.
13. Aleissa M, Aloraini T, Alsubaie LF, Hassoun M, Abdulrahman G, Swaid A, Eyaid WA, **Mutairi FA,** Ababneh F, Alfadhel M, Alfares A. Common disease-associated gene variants in a Saudi Arabian population. Ann Saudi Med. 2022 Jan-Feb;42(1):29-35.
14. Alfarsi A, Alfadhel M, Alameer S, Alhashem A, Tabarki B, Ababneh F, Al Fares A, **Al Mutairi F.** The phenotypic spectrum of dihydrolipoamide dehydrogenase deficiency in Saudi Arabia. Mol Genet Metab Rep. 2021 Oct 23;29:100817. doi: 10.1016/j.ymgmr.2021.100817.
15. Alawbathani S, Westenberger A, Ordonez-Herrera N, Al-Hilali M, Al Hebby H, Alabbas F, Alhashem AM, Elyamany G, Megarbane A, Kose M, Alhashmi N, Al Sukaiti N, Al-Raqad M, Al-Tawalbeh S, Abu Adas Blanco O, Alkhattabi F, Sng D, Al-Ali R, Khan S, Tawamie H, Tripolszki K, Karageorgou V, Trunzo R, **Al Mutairi F,** Reversade B, Bauer P, Bertoli-Avella AM. Biallelic ZNFX1 variants are associated with a spectrum of immuno-hematological abnormalities. Clin Genet. 2021 Oct 27. doi: 10.1111/cge.14081.
16. Alfadhel M, Nashabat M, Saleh M, Elamin M, Alfares A, Al Othaim A, Umair M, Ahmed H, Ababneh F, **Al Mutairi F,** Eyaid W, Alswaid A, Alohali L, Faqeih E, Almannai M, Aljeraisy M, Albdah B, Hussein MA, Rahbeeni Z, Alasmari A. Long-term effectiveness of carglumic acid in patients with propionic acidemia (PA) and methylmalonic acidemia (MMA): a randomized clinical trial. Orphanet J Rare Dis. 2021 Oct 11;16(1):422. doi: 10.1186/s13023-021-02032-8.
17. Alfadhel M, Almuqbil M, **Al Mutairi F**, Umair M, Almannai M, Alghamdi M, Althiyab H, Albarakati R, Bashiri FA, Alshuaibi W, Ba-Armah D, Saleh MA, Al-Asmari A, Faqeih E, Altuwaijri W, Al-Rumayyan A, Balwi MA, Ababneh F, Alswaid AF, Eyaid WM, Almontashiri NAM, Alhashem A, Hundallah K, Bertoli-Avella A, Bauer P, Beetz C, Alrifai MT, Alfares A, Tabarki B. The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. Front Pediatr. 2021 May 13;9:633385. doi: 10.3389/fped.2021.633385. PMID: 34055681; PMCID: PMC8155587.
18. Le Voyer T, Neehus AL, Yang R, Ogishi M, Rosain J, Alroqi F, **Al Mutairi, F**,…et al. Inherited deficiency of stress granule ZNFX1 in patients with monocytosis and mycobacterial disease. Proc Natl Acad Sci U S A. 2021;118(15).
19. Almannai, M., Luo, S., Faqeih, E., **Almutairi, F**., Li, Q., &amp; Agrawal, P. B. (2021). Homozygous SPEG Mutation Is Associated With Isolated Dilated Cardiomyopathy. Circulation: Genomic and Precision Medicine, 14(2). https://doi.org/10.1161/circgen.120.003310
20. Althagafi, A., Alsubaie, L., Kathiresan, N., Mineta, K., Aloraini, T., **Almutairi, F**., … Hoehndorf, R. (2021). DeepSVP: Integration of genotype and phenotype for structural variant prioritization using deep learning. https://doi.org/10.1101/2021.01.28.428557
21. Alsubaie, L., Aloraini, T., Alfarsi, A., **Mutairi, F**. A., Alswaid, A., Alothaim, A., … Alfares, A. (2021). Amended Informative Negative Whole Exome Sequencing Results. <https://doi.org/10.21203/rs.3.rs-147916/v1>
22. Beetz C, Westenberger A, Al-Ali R, Ameziane N, Alhashmi N, **Al Mutairi, F**, ..Boustany RM, et al. LRRK2 Loss-of-Function Variants in Patients with Rare Diseases: No Evidence for a Phenotypic Impact. Mov Disord. 2021;36(4):1029-31.
23. Neuray C, Maroofian R, Scala M, Sultan T, Pai GS, **Al Mutairi, F**,..Mojarrad M, et al. Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain. 2020;143(8):2388-97.
24. **Al Mutairi F.** Hyperhomocysteinemia: Clinical Insights. J Cent Nerv Syst Dis. 2020;12:1179573520962230.
25. De Nittis P, Efthymiou S, Sarre A, Guex N, Chrast J, **Al Mutairi, F**,..Putoux A, et al. Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. J Med Genet. 2020.
26. Li C, Beauregard-Lacroix E, Kondratev C, **Al Mutairi, F,..** Rousseau J, Heo AJ, Neas K, et al. UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. Am J Hum Genet. 2021;108(1):134-47.
27. Maddirevula S, Shamseldin HE, Sirr A, AlAbdi L, Lo RS, **Al Mutairi, F**,..Ewida N, et al. Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. Front Genet. 2020;11:580484.
28. Bertoli-Avella, A. M., Kandaswamy, K. K., Khan, S., **Al Mutairi, F**, Ordonez-Herrera, N., Tripolszki, K., Beetz, C., … Bauer, P. (2021). Combining exome/genome sequencing with data repository analysis reveals novel gene–disease associations for a wide range of genetic disorders. Genetics in Medicine.
29. Bertoli-Avella, A. M., Beetz, C., Ameziane, N., Rocha, M. E., Guatibonza, **Al Mutairi, F**, P., Pereira, C., Bauer, P. (2020). Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort. European Journal of Human Genetics. doi:10.1038/s41431-020-00713-9
30. Neuray C, Maroofian R, Scala M, Sultan T, Pai GS, **Al Mutairi, F**, Mojarrad M, et al. Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain. 2020;143(8):2388-97.
31. Alfares A, Alsubaie L, Aloraini T, Alaskar A, Althagafi A, **Al Mutairi, F**, Alahmad A, et al. What is the right sequencing approach? Solo VS extended family analysis in consanguineous populations. BMC Med Genomics. 2020;13(1):103.
32. Alsubaie L, Aloraini T, Amoudi M, Swaid A, Eyiad W, **Al Mutairi, F**, et al. Genomic testing and counseling: The contribution of next-generation sequencing to epilepsy genetics. Ann Hum Genet. 2020.
33. Almutairi, M., Al‐Khenaizan, S., Sufiani, F. A., Balwi, M. A., &amp; **Al Mutairi, F.** (2020). Peeling of skin as presenting manifestation in congenital disorders of glycosylation. The Journal of Dermatology. doi:10.1111/1346-8138.15459
34. Almutairi, R., Alrashidi, S., Umair, M., Alshalan, M., Alsubaie, L., Aloraini, T., **Al Mutairi, F**. (2020). Novel mutation of the FHL1 gene associated with congenital myopathy and early respiratory muscles involvement: A case report. Journal of Biochemical and Clinical Genetics, 45-51. doi:10.24911/jbcgenetics/183-1585821994
35. Alsubaie, L., Alkhalaf, R., Aloraini, T., Amoudi, M., Swaid, A., **Al Mutairi, F.**, Alfares, A. (2020). MEFV c.2230GT p.(Ala744Ser) rs61732874 previously misclassified as pathogenic variant due to lack of a population specific database. Annals of Human Genetics. doi:10.1111/ahg.12385
36. **Al Mutairi, F.**, Alkhalaf, R., Alkhorayyef, A., Alroqi, F., Yusra, A., Umair, M., Alfadhel, M. (2020). Homozygous truncating NEK10 mutation, associated with primary ciliary dyskinesia: A case report. BMC Pulmonary Medicine, 20(1). doi:10.1186/s12890-020-1175-1
37. Schottlaender LV, Abeti R, Jaunmuktane Z, **Al Mutairi, F**,…. Aurrand-Lions M, Houlden HBi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. Am J Hum Genet. 2020 Mar 5;106(3):412-421.
38. Wagner, M., Skorobogatko, Y., Pode-Shakked, B., Powell, C. M., Alhaddad, B., **Al Mutairi, F**, … Distelmaier, F. (2020). Bi-allelic Variants in RALGAPA1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. *The American Journal of Human Genetics*, *106*(2), 246–255.
39. Nashabat, M., Obaid, A., **Al Mutairi, F**. A., Saleh, M., Elamin, M., Ahmed, H., … Alfadhel, M. (2019). Evaluation of long-term effectiveness of the use of carglumic acid in patients with propionic acidemia (PA) or methylmalonic acidemia (MMA): study protocol for a randomized controlled trial. *BMC Pediatrics*, *19*(1)
40. Monies D, Abouelhoda M, Assoum M, Moghrabi N, Rafiullah R, Almontashiri N, Alowain M, Alzaidan H, Alsayed M, Wali S, **Al Mutairi, F**, … Murad H, Meyer BF, Alkuraya FS. (2019). Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. *The American Journal of Human Genetics 105(4):879*
41. Salpietro V, Dixon CL, **Al Mutairi, F** … Houlden H(2019) AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nat Commun. 2019 Jul 12;10(1):3094. doi: 10.1038/s41467-019-10910-w
42. Alaqeel, B., Babakir, A., **Al Mutairi, F**., & Dubayee, M. (2019). Coexistence of genetic conditions: Exploring a possible relationship. *Sudanese Journal of Paediatrics*, 60–66.
43. Efthymiou S, Salpietro V, **Al Mutairi, F**, Alkuraya FS, Nolano M, Devaux J,… Houlden H (2019) Biallelic mutations in neurofascin cause neurodevelopmentalimpairment and peripheral demyelination. Brain. 2019 Oct 1;142(10):2948-2964
44. Alsahli, S., Alfares, A., Guzmán-Vega, F. J., Arold, S. T., Ba-Armah, D., & **Al Mutairi, F**. A. (2019). Truncating biallelic variant in DNAJA1, encoding the co-chaperone Hsp40, is associated with intellectual disability and seizures. *Neurogenetics*, *20*(2), 109–115
45. Chelban V, Wilson MP, Warman Chardon, **Al Mutairi, F**, Houlden H; Care4Rare Canada Consortium and the SYNaPS Study Group (2019) PDXK mutations cause polyneuropathy responsive to pyridoxal 5'-phosphate supplementation. Ann Neurol. 2019 Aug;86(2):225-240.
46. Al-Hamed, M. H., Imtiaz, F., Al-Hassnan, Z., Al-Owain, M., Al-Zaidan, H., Alamoudi, M. S., **Al Mutairi, F** … Alsayed, M. (2019). Spectrum of mutations underlying Propionic acidemia and further insight into a genotype-phenotype correlation for the common mutation in Saudi Arabia. *Molecular Genetics and Metabolism Reports*, *18*, 22–29.
47. Al-Harbi, M., Al-Amir, A., Al-Qahtani, A., & **Al-Mutairi, F**. (2019). Heterozygous mutation in SLC36A2 gene causing hyperglycinuria and nephrolithiasis. *Journal of Biochemical and Clinical Genetics*, 74–76.
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49. Ahmed, H., Al-Ghamdi, S., & **Al** **Mutairi, F**. (2018). Dilated cardiomyopathy in a child with truncating mutation in NRAP gene. *Journal of Biochemical and Clinical Genetics*, 77–80.
50. Alsahli, S,…  **Al Mutairi, F**. (2018). Severe Crohn's Disease Manifestations in a Child with Cystathionine β-Synthase Deficiency. ACG Case Reports Journal, 5. doi:10.14309/crj.2018.93
51. Mackie DI, **Al Mutairi F**, Davis RB, Kechele DO, Nielsen NR, Snyder JC, et al. hCALCRL mutation causes autosomal recessive nonimmune hydrops fetalis with lymphatic dysplasia. J Exp Med. 2018;215(9):2339-53.
52. AlGhamdi A, Alrifai MT, Al Hammad AI, **Al Mutairi F**, Alswaid A, Eyaid W, et al. Epilepsy in Propionic Acidemia: Case Series of 14 Saudi Patients. J Child Neurol. 2018;33(11):713-7.
53. Almannai M, Alasmari A, Alqasmi A, Faqeih E, **Al Mutairi F**, Alotaibi M, et al. Expanding the phenotype of SLC25A42-associated mitochondrial encephalomyopathy. Clin Genet. 2018;93(5):1097-102.
54. **Al Mutairi F**, Alzahrani F, Ababneh F, Kashgari AA, Alkuraya FS. A mendelian form of neural tube defect caused by a de novo null variant in SMARCC1 in an identical twin. Ann Neurol. 2018;83(2):433-6.
55. Alsahli S, Alrifai MT, Al Tala S, **Mutairi FA**, Alfadhel M. Further Delineation of the Clinical Phenotype of Cerebellar Ataxia, Mental Retardation, and Disequilibrium Syndrome Type 4. J Cent Nerv Syst Dis. 2018;10:1179573518759682.
56. Mohammed Al-Dubayee, Reem Fattouh , **Al Mutairi F**, Amir Babiker et al Novel homozygous mutation in CYP27B1 gene of vitamin D dependent rickets type 1A: a case report
57. Alfares A, Aloraini T, Subaie LA, **Al Mutairi F**, Alfadhel M et al. Whole-genome sequencing offers additional but limited clinical utility compared with reanalysis of whole-exome sequencing. Genet Med. 2018.
58. **Al Mutairi F**, Alfadhel M, Nashabat M, El-Hattab AW, Ben-Omran T, Hertecant J, et al. Phenotypic and Molecular Spectrum of Aicardi-Goutieres Syndrome: A Study of 24 Patients. Pediatr Neurol. 2018;78:35-40.
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60. Alfadhel M, Nashabat M, Alrifai MT, Alshaalan H, **Al Mutairi F**, Al-Shahrani SA, et al. Further delineation of the phenotypic spectrum of ISCA2 defect: A report of ten new cases. Eur J Paediatr Neurol. 2017.
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65. Alfares A, Alfadhel M, Wani T, Alsahli S, Alluhaydan I, **Al Mutairi F**, et al. A multicenter clinical exome study in unselected cohorts from a consanguineous population of Saudi Arabia demonstrated a high diagnostic yield. Mol Genet Metab. 2017;121(2):91-5.
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68. Alfadhel M, Benmeakel M, Hossain MA, **Al Mutairi F**, Al Othaim A, Alfares AA, et al. 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;11(1):126.
69. Alamri H, **Al Mutairi F**, Alothman J, Alothaim A, Alfadhel M, Alfares A. Diabetic ketoacidosis in vanishing white matter. Clin Case Rep. 2016;4(8):717-20.
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| **PRESENTATIONS:** |

* Prevention Genetic Program as an efficient model for rare diseases management. Saudi Rare Diseases Summit, Riyadh. 16th-17th March 2023.
* Pyruvate Complex related disorders, DLD phenotypic characterization, 3rd MEGMA Symposium, Abu Dhabi, UAE. 4th-6th November 2022.
* Hyperhomocysteinemia and related disorders, SSMG annual conference (SSMG 2022), Riyadh. 23rd-24th March 2022.
* Genetic disorders session, 22nd Intensive Pediatric Course, HMG-Takhassusi Hospital, Riyadh. 15th-19th Aug 2020.
* Update on Novel treatment for Aminoacidopathies, 1st International MEGMA symposium in collaboration with SSMG, Riyadh. 4th-6th November 2020.
* Pompe Disease-Lesson learned for newborn screening Program, 1st International MEGMA symposium in collaboration with SSMG, Riyadh. 4th-6th November 2020.
* Genetic disorders session, 21st Intensive Pediatric Course, HMG-Takhassusi Hospital, Riyadh. 30th Aug - 3rd Sep 2020.
* Non-Immune Hydrops Fetalis and Sub-fertility, Geneticist view. SSMG annual conference (SSMG 2019), KAUST Campus, Thuwal, Jeddah. 15th-16th April 2019.
* The role of transcription factors in embryogenesis: BAF complex and neural tube defect. SSMG annual conference (SSMG 2018), Riyadh. 14th-15th May 2018.
* Guidelines for Acute Management of Hyperammonemia in the Middle East Region, Pediatrics Egypt Congress, Cairo, Egypt. April 2018.
* Genomic advances in Neurodevelopment disorders: A clinician Guide, 1st National Developmental & Behavioral Pediatrics Conference, KASCH-KAMC, Riyadh. April 2018.
* Neurometabolic Disorders in Saudi Arabia, 8th Pediatrics Symposium, KAH-Al Ahsa. March 2018.
* Phenotypic and Molecular Spectrum of Aicardi-Goutieres Syndrome in Arab population: A Study of 24 Patients MEMG meeting, Athens, Greece. Feb 2018.
* Clinical Approach and recognition of IEM- The 7th Pediatrics Symposium, KAH-Al Ahsa. April 2017.
* Challenging cases with Hyperammonemia, Hyperammonemia and related Disorders (Tyrosinemia) at Doha Qatar. December 2016.
* Neurometabolic disorders in SA, Newborn Screening program in KSA, toward preventing disability, Dammam. Feb 2016.
* Lysosomal Storage diseases, clinical and biochemical approach. The 8th basic and advanced Medical Genetic Course KFMC-Riyadh. 28th May 2015.
* Neurometabolic Disorders in Saudi Arabia, 1ST International Pediatric Neurometabolic Conference, PSMMC, Riyadh. February 2015
* Quiz the Expert Session, Lysosomal Storage disease. Excellence in Pediatrics Conference Dubai, UAE. December 2014.
* Acute Liver Failure in Propionic Acidemia, Hyperammonemia and related Disorders (Homocystinuria) at Doha Qatar, December 2014.
* Metabolic Emergency, 7th Medical Genetics workshop at Khamis Mushait Military Hospital. December 2014.
* Fundamental concepts of Genetics. The 7th SSMG workshop at Collage of Medicine, King Faisal University (Al-Hassa Feb 2014.
* Genetic heterogeneity of Childhood Rhabdomyolysis, Pediatric Ground rounds, PSMMC January 2014.
* Aminoacidopathy, 5th Medical Genetics workshop at MCH Makkah, December 2013.
* Clinical and Molecular variability in Ethylmalonic Encephalopathy, MEMG meeting, Istanbul Turkey, October 2013.
* Congenital Disorders of Glycosylation, 6th Basic Genetic Course. KFMC, Riyadh. May 2013.
* Fatty Acid Oxidation Defect, hot topic in inherited metabolic disorders, 4th Genetic workshop for SSMG) at Prince Salman, North West Armed Force Hospital, Tabuk. April 2013.
* Metabolic Emergency (approach and early treatment), Pediatric Emergency Department, British Columbia Children Hospital UBC, Canada. January 2012.
* Congenital Disorder of Glycosylation -Update and suggested diagnostic approach, Biochemical Diseases Department, British Columbia Children Hospital, UBC, Canada. Dec 2011.
* Liver Transplantation as treatment option for Propionic Acidemia- evidence review, Biochemical Diseases Department, British Columbia Children Hospital UBC, Canada. Sep 2010.
* Cardiomyopathy in Propionic Acidemia, Biochemical Diseases Department, British Columbia Children Hospital UBC, Canada. June 2010.
* Case with Ketotic Hypoglycemia – What is our next step? Biochemical Diseases Department, British Columbia Children Hospital, UBC, Canada. May 2010.