Research and Publications

1. **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.

2. **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.

3. 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.

4. **Ababneh FA**, balwim, **Swaid A**, Yousseft, azzawim. Hereditary deletion of the entire FAM20C gene in a patient with Raine syndrome. AJMG accepted.

5. D.Sparrow, Efaqeih, Bsallout, **Swaid A, Ababneh FA**, Alsayed-moeen, **Rukban H**, Eyaid W, Rkageyam, Sian.Ellard, Peter.Turnpenny, S.Dunwoodie. Mutation of HES7 in a Large Extended Family with Spondylocostal Dysostosis and Dextrocardia with Situs Inversus. AJMG accepted

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

7. 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.

8. **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.

9. **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.

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

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

12. **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.

13. **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.

14. **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.

15. **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.

16. **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.

17. Aldahmesh MA, Khan AO, Mohamed JY, Hijazi H, Al-Owain M, **Alswaid A**, Alkuraya FS. Genomic analysis of pediatric cataract in Saudi Arabia reveals novel candidate disease genes. Genet Med. 2012 Dec;14(12):955-62..

18. Bachmann-Gagescu R, Ishak GE, Dempsey JC, Adkins J, O'Day D, Phelps IG, Gunay-Aygun M, Kline AD, Szczaluba K, Martorell L, **Alswaid A**, Alrasheed S, Pai S, Izatt L, Ronan A, Parisi MA, Mefford H, Glass I, Doherty D. Genotype-phenotype correlation in CC2D2A-related Joubert syndrome reveals an association with ventriculomegaly and seizures.

19. Christiansen HE, Schwarze U, Pyott SM, **AlSwaid A**, Al Balwi M, Alrasheed S, Pepin MG, Weis MA, Eyre DR, Byers PH. Homozygosity for a missense mutation in SERPINH1, which encodes the collagen chaperone protein HSP47, results in severe recessive osteogenesis imperfecta.

20. Gorden NT, Arts HH, Parisi MA, Coene KL, Letteboer SJ, van Beersum SE, Mans DA, Hikida A, Eckert M, Knutzen D, **Alswaid AF**., et al. CC2D2A is mutated in Joubert syndrome and interacts with the ciliopathy-associated basal body protein CEP290. Am J Hum Genet. 2008 Nov;83(5):559-71.

21. Faiyaz-Ul-Haque M, Zaidi SH, Al-Sanna N, **Alswaid A**, Momenah T, Kaya N, Al-Dayel F, Bouhoaigah I, Saliem M, Tsui LC, Teebi AS. A novel missense and a recurrent mutation in SLC2A10 gene of patients affected with arterial tortuosity syndrome. Atherosclerosis. 2009 Apr;203(2):466-71.

22. Dagoneau N, Benoist-Lasselin C, Huber C, Faivre L, Mégarbané A, **Alswaid A**, Dollfus H, Alembik Y, Munnich A, Legeai-Mallet L, Cormier-Daire V. ADAMTS10 mutations in autosomal recessive Weill-Marchesani syndrome. Am J Hum Genet. 2004 Nov;75(5):801-6.

23. Faivre L, Dollfus H, Lyonnet S, Alembik Y, Mégarbané A, Samples J, Gorlin RJ, **Alswaid A**, Feingold J, Le Merrer M, Munnich A, Cormier-Daire V. Clinical homogeneity and genetic heterogeneity in Weill-Marchesani syndrome. Am J Med Genet A. 2003 Dec 1;123A(2):204-7. Review.
24. Bond J, Scott S, Hampshire DJ, Springell K, Corry P, Abramowicz MJ, Mochida GH, Hennekam RC, Maher ER, Fryns JP, **Alswaid A**, Jafri H, Rashid Y, Mubaidin A, Walsh CA, Roberts E, Woods CG. Protein-truncating mutations in ASPM cause variable reduction in brain size. Am J Hum Genet. 2003 Nov;73(5):1170-7.
25. Astuto LM, Kelley PM, Askew JW, Weston MD, Smith RJ, **Alswaid AF**, Al-Rakaf M, Kimberling WJ. Searching for evidence of DFNB2. Am J Med Genet. 2002 May 15;109(4):291-7.
26. Faivre L, Mégarbané A, **Alswaid A**, Zylberberg L, Aldohayan N, Campos-Xavier B, Bacq D, Legeai-Mallet L, Bonaventure J, Munnich A, Cormier-Daire V. Homozygosity mapping of a Weill-Marchesani syndrome locus to chromosome 19p13.3-p13.2. Hum Genet. 2002 Apr;110(4):366-70.
27. Shaheen R, Al-Owain M, Khan AO, Aglan M, Hossni HA, Al-Tassan R, **Eyaid W,** Alkuraya FS. Identification of Three Novel ECEL1 Mutations in Three Families with Distal Arthrogryposis Type 5D. Clin Genet. 2013 Jul 6.
28. Uusimaa J, Evans J, Smith C, Butterworth A, Craig K, Ashley N, Liao C, Carver J, Diot A, Macleod L, Hargreaves I, Al-Hussaini A, Faqeih E, Asery A, Al Balwi M, **Eyaid W**., et al. Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. Eur J Hum Genet. 2013 May 29.
29. Shaheen R, Ansari S, Alshammari MJ, Alkhalidi H, Alrukban H, **Eyaid W,** Alkuraya FS. A novel syndrome of hypohidrosis and intellectual disability is linked to COG6 deficiency. J Med Genet. 2013 Jul;50(7):431-6.
30. Manzini MC, Tambunan DE, Hill RS, Yu TW, Maynard TM, Heinzen EL, Shianna KV, Stevens CR, Partlow JN, Barry BJ, Rodriguez J, Gupta VA, Al-Qudah AK, **Eyaid WM**,

Friedman JM, Salih MA, Clark R, Moroni I, Mora M, Beggs AH, Gabriel SB, Walsh CA. Exome sequencing and functional validation in zebrafish identify GTDC2 mutations as a cause of Walker-Warburg syndrome. Am J Hum Genet. 2012 Sep 7;91(3):541-7.

31. **Eyaid W**, Al-Tassan RS, Al-Nouri DM. Intracranial calcifications, microcephaly, and seizure. If not congenital infection, what could it be? Neurosciences (Riyadh). 2012 Jul;17(3):248-52.
32. **Eyaid W**, Al-Qattan MM, Al Abdulkareem I, Fetaini N, Al Balwi M. A novel homozygous missense mutation (c.610G>A, p.Gly204Ser) in the WNT7A gene causes tetra-amelia in two Saudi families. Am J Med Genet A. 2011 Mar;155A(3):599-604.
33. Li Y, Pawlik B, Elcioglu N, Aglan M, Kayserili H, Yigit G, Percin F, Goodman F, Nürnberg G, Cenani A, Urquhart J, Chung BD, Ismail S, Amr K, Aslanger AD, Becker C, Netzer C, Scambler P, **Eyaid W.**, et al. LRP4 mutations alter Wnt/beta-catenin signaling and cause limb and kidney malformations in Cenani-Lenz syndrome. Am J Hum Genet. 2010 May 14;86(5):696-706.
34. Shen J, Gilmore EC, Marshall CA, Haddadin M, Reynolds JJ, **Eyaid W**, Bodell A, Barry B, Gleason D, Allen K, Ganesh VS, Chang BS, Grix A, Hill RS, Topcu M, Caldecott KW, Barkovich AJ, Walsh CA. Mutations in PNKP cause microcephaly, seizures and defects in DNA repair. Nat Genet. 2010 Mar;42(3):245-9
35. Al-Qattan MM, Al-Balwi M, **Eyaid W**, Al-Abdulkarim I, Al-Turki S. Congenital duplication of the palm syndrome: gene analysis and the molecular basis of its clinical features. J Hand Surg Eur Vol. 2009 Apr;34(2):247-51.
36. Fromme JC, Ravazzola M, Hamamoto S, Al-Balwi M, **Eyaid W**, Boyadjiev SA, Cosson P, Schekman R, Orci L. The genetic basis of a craniofacial disease provides insight into COPII coat assembly. Dev Cell. 2007 Nov;13(5):623-34
37. Al-Qattan MM, **Eyaid W**, Al-Balwi M. Congenital duplication of the palm syndrome. Ann Plast Surg. 2007 Sep;59(3):341-3.

38. Boyadjiev SA, Fromme JC, Ben J, Chong SS, Nauta C, Hur DJ, Zhang G, Hamamoto S, Schekman R, Ravazzola M, Orci L, **Eyaid W**. Cranio-lenticulo-sutural dysplasia is caused by a SEC23A mutation leading to abnormal endoplasmic-reticulum-to-Golgi trafficking. Nat Genet. 2006 Oct;38(10):1192-7.
39. Shen J, **Eyaid W**, Mochida GH, Al-Moayyad F, Bodell A, Woods CG, Walsh CA. ASPM mutations identified in patients with primary microcephaly and seizures. J Med Genet. 2005 Sep;42(9):725-9.
40. Al Tawil K, Shataiwi A, Mutair A**, Eyaid W**, Saif SA. Hypoparathyroidism-retardation-dysmorphism (HRD) syndrome in triplets. Am J Med Genet A. 2005 Jun 1;135(2):200-1.
41. **Eyaid WM**, Al-Nouri DM, Rashed MS, Al-Rifai MT, Al-Wakeel AS. An inborn error of metabolism presenting as hypoxic-ischemic insult. Pediatr Neurol. 2005 Feb;32(2):134-6.
42. Currier SC, Lee CK, Chang BS, Bodell AL, Pai GS, Job L, Lagae LG, Al-Gazali LI, **Eyaid WM**, Enns G, Dobyns WB, Walsh CA. Mutations in POMT1 are found in a minority of patients with Walker-Warburg syndrome. Am J Med Genet A. 2005 Feb 15;133A(1):53-7
43. Rashed MS, Saadallah AA, Rahbeeni Z, **Eyaid W,** Seidahmed MZ, Al-Shahwan S, Salih MA, Osman ME, Al-Amoudi M, Al-Ahaidib L, Jacob M. Determination of urinary S-sulphocysteine, xanthine and hypoxanthine by liquid chromatography-electrospray tandem mass spectrometry. Biomed Chromatogr. 2005 Apr;19(3):223-30
44. Ferland RJ, **Eyaid W**, Collura RV, Tully LD, Hill RS, Al-Nouri D, Al-Rumayyan A, Topcu M, Gascon G, Bodell A, Shugart YY, Ruvolo M, Walsh CA. Abnormal cerebellar development and axonal decussation due to mutations in AHI1 in Joubert syndrome. Nat Genet. 2004 Sep;36(9):1008-13.
45. Mochida GH, Rajab A, **Eyaid W**, Lu A, Al-Nouri D, Kosaki K, Noruzinia M, Sarda P, Ishihara J, Bodell A, Apse K, Walsh CA. Broader geographical spectrum of Cohen syndrome due to COH1 mutations. J Med Genet. 2004 Jun;41(6):e87

46. Frank CG, Grubenmann CE, **Eyaid W**, Berger EG, Aebi M, Hennet T. Identification and functional analysis of a defect in the human ALG9 gene: definition of congenital disorder of glycosylation type IL. Am J Hum Genet. 2004 Jul;75(1):146-50

47. Hämäläinen RH, Avela K, Lambert JA, Kallijärvi J, **Eyaid W**, Gronau J, Ignaszewski AP, McFadden D, Sorge G, Lipsanen-Nyman M, Lehesjoki AE. Novel mutations in the TRIM37 gene in Mulibrey Nanism. Hum Mutat. 2004 May;23(5):522.

48. Yaghmai R, Kimyai-Asadi A, Rostamiani K, Heiss NS, Poustka A, **Eyaid W**, Bodurtha J, Nousari HC, Hamosh A, Metzenberg A. Overlap of dyskeratosis congenita with the Hoyeraal-Hreidarsson syndrome. J Pediatr. 2000 Mar;136(3):390-3

49. **Eyaid WM,** Clough MV, Root H, Scott KM, McCormick MK, Zhang X, Lisitsyn NA, Kearns WG, Francomano CA, Richards JE, McIntosh I. Physical mapping of the nail patella syndrome interval at 9q34: ordering of STSs and ESTs. Hum Genet. 1998 Oct;103(4):525-6.

50. McIntosh I, Dreyer SD, Clough MV, Dunston JA, **Eyaid W**, Roig CM, Montgomery T, Ala-Mello S, Kaitila I, Winterpacht A, Zabel B, Frydman M, Cole WG, Francomano CA, Lee B. Mutation analysis of LMX1B gene in nail-patella syndrome patients. Am J Hum Genet. 1998 Dec;63(6):1651-8.