

Lama AlAbdi, Ph.D.

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Education

Biochemistry Department, King Saud University (KSU) | Riyadh, Saudi Arabia | B.S. | July 2009

Bioscience, King Abdullah University of Science and Technology (KAUST)| Thuwal, Saudi Arabia | M.S. | December 2011

Department of Biochemistry, Purdue University | West Lafayette, IN, USA | PhD | August 2019

Thesis Title: “Molecular Mechanisms That Govern Stem Cell Differentiation and Their Implications in Cancer” with Humaira Gowher, Ph.D.

Appointments

- Assistant professor, Zoology Department, King Saud University, College of Science (2019-Current).
- Adjunct Scientist in Developmental Genetics, Translational Genomics Department, Center of Genome Medicine, KFSHRC (2019-Current).
- University Lecturer, Zoology Department, King Saud University, College of Science (2011-2019).
- Research Assistant (Developmental Genetics), King Faisal Specialist Hospital and Research Center (2009-2010).
- Research Intern (DNA repair & Apoptosis), King Faisal Specialist Hospital and Research Center (2009).

Skills & Abilities

PUBLICATIONS

AlAbdi L., Maddirevula S., Shamseldin H.E., Khouj E., Helaby R., Hamid H., Almulhim A., Hashem M.O., Abdulwahab F., Abouyousef O., Alqahtani M., Altuwaijri N., Jaafar A., Alshidi T., Alzahrani F.; Mendeliome Group; Alkuraya F.S. (2023). Diagnostic implications of pitfalls in causal variant identification based on 4577 molecularly characterized families. *Nat Commun.* 14(1):5269. doi: 10.1038/s41467-023-40909-3.

Magliyah M.S., Almarek F., Nowilaty S.R., **Al-Abdi L.**, Alkuraya F.S., Alowain M., Schatz P., Alfaadhel T., Khan A.O., Alsulaiman S.M. (2023). LEPREL1-related giant retinal tear detachments mimic the phenotype of ocular stickler syndrome. *Retina.* 43(3):498-505. doi: 10.1097/IAE.0000000000003691.

Mahajan S., Ng B.G., **AlAbdi L.**, Earnest P.D.J., Sosicka P., Patel N., Helaby R., Abdulwahab F., He M., Alkuraya F.S., Freeze H.H. (2023). Homozygous truncating variant in MAN2A2 causes a novel congenital disorder of glycosylation with neurological involvement. *J Med Genet.* 60(7):627-635. doi: 10.1136/jmg-2022-108821. Epub 2022 Nov 10.

Maroofian R., Kaiyrzhanov R., Cali E., Zamani M., Zaki M.S., Ferla M., Tortora D., Sadeghian S., Saadi S.M., Abdullah U., Ghayoor Karimiani E., Efthymiou S., Yeşil G., Alavi S., Al Shamsi A.M., Tajsharghi H., Abdel-Hamid M.S., Saadi N.W., Al Mutairi F., **Alabdi L.**, Beetz C., Ali Z., Toosi M.B., Rudnik-Schöneborn S., Babaei M., Isohanni P., Muhammad J., Sheraz K., Al Shalan M., Hickey S.E., Marom D., Elhanan E., Kurian M.A., Marafi D., Saberi A., Hamid M., Spaull R., Meng L., Lalani S., Maqbool S., Rahman F., Seeger J., Palculict T.B., Lau T., Murphy D., Mencacci N.E., Steindl K., Begemann A., Rauch A., Akbas S., Dilruba A.A., Salpietro V., Yousaf H., Ben-Shachar S., Ejeskär K., Al Aqeel A.I., High F.A., Armstrong-Javors A.E., Zahraei S.M., Seifi T., Zeighami J., Shariati G., Sedaghat A., Asl S.N., Shahrooei M., Zifarelli G., Burglen L., Ravelli C., Zschocke J., Schatz U.A., Ghavideldarestani M.,

Kamel W.A., Van Esch H., Hackenberg A., Taylor J.C., Al-Gazali L., Bauer P., Gleeson J.J., Alkuraya F.S., Lupski J.R., Galehdari H., Azizimalamiri R., Chung W.K., Baig S.M., Houlden H., Severino M. (2023). Biallelic MED27 variants lead to variable ponto-cerebello-lental degeneration with movement disorders. *Brain*. awad257. doi: 10.1093/brain/awad257. Online ahead of print.

Badawi A., Magliyah M., Alabbasi O., **AlAbdi L.**, Alkuraya F.S., Schatz P., ALBalawi H.B., Mura M. (2023). Cone dystrophy associated with autoimmune polyglandular syndrome type 1. *Sci Rep*. 13(1):11223. doi: 10.1038/s41598-023-38419-9.

Almannai M., **AlAbdi L.**, Maddirevula S., Alotaibi M., Alsaeem B.M., Aljadhai Y.I., Alsaif H.S., Abukhalid M., Alkuraya F.S. (2023). KIF26A is mutated in the syndrome of congenital hydrocephalus with megacolon. *Hum Genet*. 142(3):399-405. doi: 10.1007/s00439-022-02513-1. Epub 2022 Dec 23

AlAbdi L., Alshammari M., Helaby R., Khan A.O., Alkuraya F.S. (2023). PMEL is mutated in oculocutaneous albinism. *Hum Genet*. 142(1):139-144. doi: 10.1007/s00439-022-02489-y. Epub 2022 Sep 27.

Saida K., Maroofian R., Sengoku T., Mitani T., Pagnamenta A.T., Marafi D., Zaki M.S., O'Brien T.J., Karimiani E.G., Kaiyrzhanov R., Takizawa M., Ohori S., Leong H.Y., Akay G., Galehdari H., Zamani M., Romy R., Carroll C.J., Toosi M.B., Ashrafzadeh F., Imannezhad S., Malek H., Ahangari N., Tomoum H., Gowda V.K., Srinivasan V.M., Murphy D., Dominik N., Elbendary H.M., Rafat K., Yilmaz S., Kanmaz S., Serin M., Krishnakumar D., Gardham A., Maw A., Rao T.S., Alsubhi S., Srour M., Buhas D., Jewett T., Goldberg R.E., Shamseldin H., Frengen E., Misceo D., Strømme P., Magliocco Ceroni J.R., Kim C.A., Yesil G., Sengenc E., Guler S., Hull M., Parnes M., Aktas D., Anlar B., Bayram Y., Pehlivan D., Posey J.E., Alavi S., Madani Manshadi S.A., Alzaidan H., Al-Owain M., **Alabdi L.**, Abdulwahab F., Sekiguchi F., Hamanaka K., Fujita A., Uchiyama Y., Mizuguchi T., Miyatake S., Miyake N., Elshafie R.M., Salayev K., Guliyeva U., Alkuraya F.S., Gleeson J.G., Monaghan K.G., Langley K.G., Yang H., Motavaf M., Safari S., Alipour M., Ogata K., Brown A.E.X., Lupski J.R., Houlden H., Matsumoto N. (2023). Brain monoamine vesicular transport disease caused by homozygous SLC18A2 variants: A study in 42 affected individuals. *Genet Med*. 25(1):90-102. doi: 10.1016/j.gim.2022.09.010. Epub 2022 Oct 31.

AlAbdi L., Desbois M., Rusnac D.V., Sulaiman R.A., Rosenfeld J.A., Lalani S., Murdock D.R., Burrage L.C.; Undiagnosed Diseases Network; Billie Au P.Y., Towner S., Wilson W.G., Wong L., Brunet T., Strobl-Wildemann G., Burton J.E., Hoganson G., McWalter K., Begtrup A., Zarate Y.A., Christensen E.L., Opperman K.J., Giles A.C., Helaby R., Kania A., Zheng N., Grill B., Alkuraya F.S. (2023). Loss-of-function variants in MYCBP2 cause neurobehavioural phenotypes and corpus callosum defects. *Brain*. 146(4):1373-1387. doi: 10.1093/brain/awac364.

Shankar SP, Grimsrud K, Lanoue L, Egense A, Willis B, Hörberg J, **AlAbdi L**, Mayer K, Ütkür K, Monaghan KG, Krier J. (2022). A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. *Genetics in Medicine*.

Mensah, I.K., Norvil, A.B, **AlAbdi, L.**, McGovern, S., Petell, C.J., He, M., Gowher, H. (2021). Misregulation of the expression and activity of DNA methyltransferases in cancer. *Nucleic Acids Res Cancer*, 3 (4).

Shamseldin, H.E., **AlAbdi, L.**, Maddirevula, S., Alsaif, H.S., Alzahrani, F., Ewida, N., Hashem, M., Abdulwahab, F., Abuyousef, O., Kuwahara, H. and Gao, X., Alkuraya, F.S. (2021). Lethal variants in humans: lessons learned from a large molecular autopsy cohort. *Genome medicine*, 13(1), pp.1-11.

AlAbdi, L., Alrashseed, S., Alsulaiman, A., Helaby, R., Imtiaz, F., Alhamed, M., Alkuraya, F.S. (2021). Residual risk for additional recessive diseases in consanguineous couples. *Genetics in Medicine*, 1-7.

Khan, A. O., **AlAbdi, L.**, Patel, N., Helaby, R., Hashem, M., Abdulwahab, F., F., AlBadr, F.B., Alkuraya, F.S (2021). Genetic testing results of children suspected to have Stickler syndrome type collagenopathy after ocular examination. *Molecular genetics & genomic medicine*, e1628.

Maddirevula, S., Shamseldin, H. E., Sirr, A., **AlAbdi, L.**, Lo, R. S., Ewida, N., Al-Qahtani, M., Hashem, M., Abdulwahab, F., Aboyousef, O., Kaya, N., Monies, D., Salem, M.H., Al Harbi, N., Aldhalaan, H.M., Alzaidan, H., Almanea, H.M., Alsalamah, A.K., Al Mutairi, F., Ismail, S., Abdel-Salam, G.M. H., Alhashem, A., Asery, A., Faqeih, E., AlQassmi, A., Al-Hamoudi, W., Algoufi, T., Shagrani, M., Dudley, A.M., Alkuraya, F.S. (2020). Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. *Front Genet*. <https://doi.org/10.3389/fgene.2020.580484>

Zha, C., Farah, C.A., Holt, R.J., Ceroni, F., **AlAbdi, L.**, Thuriot, F., Khan, A.O., Helaby, R., Levesque, S., Alkuraya, F.S., et al. (2020). Biallelic variants in the small optic lobe calpain CAPN15 are associated with congenital eye anomalies, deafness and other neurodevelopmental deficits. *Hum Mol Genet*.

Al-Abdi, L., Al Murshedi, F., Elmanzalawy, A., Al Habsi, A., Helaby, R., Ganesh, A., Ibrahim, N., Patel, N., Alkuraya, F. S. (2020) CNP deficiency causes severe hypomyelinating leukodystrophy in humans. *Hum Genet* <https://doi.org/10.1007/s00439-020-02144-4>

Norvil, A. B., **AlAbdi, L.**, Liu, B., Tu, Y. H., Forstoffer, N. E., Michie, A. R., Chen, T., Gowher, H. (2020) The acute myeloid leukemia variant DNMT3A Arg882His is a DNMT3B-like enzyme. *Nucleic Acids Res* [10.1093/nar/gkaa139](https://doi.org/10.1093/nar/gkaa139)

AlAbdi, L., Saha, D., He, M., Dar, M. S., Utturkar, S. M., Sudyanti, P. A., McCune, S., Spears, B. H., Breedlove, J. A., Lanman, N. A., Gowher, H. (2020) Oct4-Mediated Inhibition of Lsd1 Activity Promotes the Active and Primed State of Pluripotency Enhancers. *Cell Rep* [30](https://doi.org/10.1016/j.celrep.2020.107300), 1478-1490

Norvil, A. B., Petell, C. J., **Alabdi, L.**, Wu, L. C., Rossie, S., and Gowher, H. (2018) Dnmt3b Methylates DNA by a Noncooperative Mechanism, and Its Activity Is Unaffected by Manipulations at the Predicted Dinner Interface. *Biochemistry-US* [57](https://doi.org/10.1021/acs.biochem.7b01200), 4312-4324

AlAbdi, L., He, M., Yang, Q. Y., Norvil, A. B., and Gowher, H. (2018) The transcription factor Vezf1 represses the expression of the antiangiogenic factor Cited2 in endothelial cells. *J Biol Chem* [293](https://doi.org/10.1074/jbc.R117.74000), 11109-11118

Petell, C. J., **Alabdi, L.**, He, M., San Miguel, P., Rose, R., and Gowher, H. (2016) An epigenetic switch regulates de novo DNA methylation at a subset of pluripotency gene enhancers during embryonic stem cell differentiation. *Nucleic Acids Res* [44](https://doi.org/10.1093/nar/gkv370), 7605-7617

Abu-Safieh, L., Alrashed, M., Anazi, S., Alkuraya, H., Khan, A. O., Al-Owain, M., Al-Zahrani, J., **Al-Abdi, L.**, Hashem, M., Al-Tarimi, S., Sebai, M. A., Shamia, A., Ray-zack, M. D., Nassan, M., Al-Hassnan, Z. N., Rahbeeni, Z., Waheed, S., Alkharashi, A., Abboud, E., Al-Hazzaa, S. A. F., and Alkuraya, F. S. (2013) Autozygome-guided exome sequencing in retinal dystrophy patients reveals pathogenetic mutations and novel candidate disease genes. *Genome Res* [23](https://doi.org/10.1101/gr.133303), 236-247

Abu-Safieh, L., Al-Anazi, S., **Al-Abdi, L.**, Hashem, M., Alkuraya, H., Alamr, M., Sirelkhatim, M. O., Al-Hassnan, Z., Alkuraya, B., Mohamed, J. Y., Al-Salem, A., Alrashed, M., Faqeih, E., Softah, A., Al-Hashem, A., Wali, S., Rahbeeni, Z., Alsayed, M., Khan, A. O., Al-Gazali, L., Taschner, P. E. M., Al-Hazzaa, S., and Alkuraya, F. S. (2012) In search of triallelism in Bardet-Biedl syndrome. *Eur J Hum Genet* [20](https://doi.org/10.1038/ejhg.2012.10), 420-427

Khan, A. O., Aldahmesh, M. A., **Al-Abdi, L.**, Mohamed, J. Y., Hashem, M., Al-Ghamdi, I., and Alkuraya, F. S. (2011) Molecular Characterization of Newborn Glaucoma Including a Distinct Aniridic Phenotype. *Ophthalmic Genet* [32](https://doi.org/10.3109/14682378.2011.550000), 138-142

Khan, A. O., **Al-Abdi, L.**, Mohamed, J. Y., Aldahmesh, M. A., and Alkuraya, F. S. (2011) Familial juvenile glaucoma with underlying homozygous p.G61E CYP1B1 mutations. *J Aapos* [15](https://doi.org/10.1016/j.jaapos.2011.07.007), 198-199

Aldahmesh, M. A., Nowilaty, S. R., Alzahrani, F., **Al-Ebdi, L.**, Mohamed, J. Y., Rajab, M., Khan, A. O., and Alkuraya, F. S. (2011) Posterior Microphthalmos as a Genetically Heterogeneous Condition That Can Be Allelic to Nanophthalmos. *Arch Ophthalmol-Chic* [129](https://doi.org/10.1001/archophthalmol.2011.27), 805-807

Abu-Safieh, L., Abboud, E. B., Alkuraya, H., Shamseldin, H., Al-Enzi, S., **Al-Abdi, L.**, Hashem, M., Colak, D., Jarallah, A., Ahmad, H., Bobis, S., Nemer, G., Bitar, F., and Alkuraya, F. S. (2011) Mutation

of IGFBP7 Causes Upregulation of BRAF/MEK/ERK Pathway and Familial Retinal Arterial Macroaneurysms. *Am J Hum Genet* 89, 313-319

Al-Dosari, M. S., Almazyad, M., **Al-Ebdi, L.**, Mohamed, J. Y., Al-Dahmash, S., Al-Dhibi, H., Al-Kahtani, E., Al-Turkmani, S., Alkuraya, H., Hall, B. D., and Alkuraya, F. S. (2010) Ocular manifestations of branchio-oculo-facial syndrome: Report of a novel mutation and review of the literature. *Mol Vis* 16, 813-818

MENTORSHIP

- Graduate student supervision: Layali Alshammari (2019-2020), Yusra AlMarshadi (2020-2021), Halima Hamid and Aisha Almulhim (2020-Current), and Atheer Aljuhani (2022)
- Undergraduate research mentees: Andjela Djokovic (Summer 2016), Stephen McCune (Fall 2016-Spring 2017), Rmaya Modi (Fall 2017), Brice Hurling-Spears (Summer 2017), James Breedlove (fall 2017-Present), Sophia Tu (Summer 2018) and Nicole Adkins (Spring 2018-Spring 2020).
- Graduate students (rotations): Eliana Torres Zelada (Spring 2016), Sristi Chakravotry (Fall 2016), Smriti Hoda (Fall 2017), Debasmita Saha (Fall 2018), Isaiah Mensah (Fall 2018), and Sarah McGrevor (Spring 2019).

Synergistic Activities

- Requested as Full-time consultant by KFSH&RC (2022-Current)
- Initiated two seminar series at KSU, Collage of Science: Academic Reading and writing Seminar series and student-led Journal club (2020 and 2021)
- L'Oréal – UNESCO (Middle East) for Women in Science Award (2020)
- Purdue University Center for Cancer Research (PCCR) travel award (2018).
- BirdStair Fellowship (2017).
- Recipient of Saudi Arabian Cultural Mission (SACM) fellowship (2013-2019).
- Recipient of Interdisciplinary Life Science-PULSe Excellence Award (2015).
- Co-Chair of the Science in School outreach program (2016-2018).
- Ranked 3rd in College of Science. Granted the Dean's Award (2009).

Talks and Conferences

- Genomics of Rare Diseases Meeting (Virtual, 2023). Talk title: **Mendelian Hunt Gone Awry: The Diagnostic Implications of Pitfalls in Causal Variant Identification Based on 4,510 Molecularly Characterized Families**
- MEDLAB (Dubai 2023). Talk title: **Mendelian Hunt Gone Awry: The Diagnostic Implications of Pitfalls in Causal Variant Identification Based on 4,375 Molecularly Characterized Families**
- ICES2023 (KSU 2023). Talk title: **Mendelian Hunt Gone Awry: The Diagnostic Implications of Pitfalls in Causal Variant Identification Based on 4,375 Molecularly Characterized Families**
- Genomics of Rare Disease 2022 (March 28-29, 2022). Talk title: **Loss of function variants in MYCBP2 cause neurodevelopmental phenotypes and corpus callosum dysgenesis.**
- Dose of Science 2022 (February 15, 2022). Talk title: **Methylated DNA immunoprecipitation (MeDIP)**
- Graduate student Discussion panel (2021), KSU, Collage of Science, Department of Zoology
- Scholarship Outcomes (2020): Lama and Epigenetic Inheritance in America.

- 5th European Days of Albinism conference. Virtually (November 4th through 7th, 2020). Attendance.
- Saudi Ophthalmology 2020 Virtual (August 6-8, 2020). Talk title: **The Burden of Inherited Retinal Diseases in Saudi Arabia: A Genetic Epidemiology Overview**.
- Hitchhiker's Guide to the Biomolecular Galaxy. Purdue University, West Lafayette, IN (May 8-9, 2019). Talk title: **Oct4 inhibits Lsd1 activity and affects gain of DNA methylation at pluripotency enhancers**.
- FASEB's Biological Methylation: Fundamental Mechanisms in Human Health and Disease, Florence, Italy (June 17-22, 2018). Poster title: **Regulation of pluripotency by DNA methylation in F9 Embryonal carcinoma cells**.
- Academic leadership workshop (January 21st, 2018).
- Midwest Chromatin & Epigenetics Meeting. Purdue University, West Lafayette, IN (June 10-12, 2018). Talk title: **Regulation of pluripotency by DNA methylation in F9 Embryonal carcinoma cells**.
- Interdisciplinary Life Science-PULSe 5min thesis competition (2016 and 2017).
- Biochemical Horizons Symposium. Purdue University, West Lafayette, IN (November 17, 2017). Poster title: **Biological outcomes of the catalytic specialization of DNA methyltransferases**.
- American Society for Biochemistry and Molecular Biology- Experimental Biology. Chicago, IL (April 22-26, 2017).