CURRICULUM VITAE

PERSONAL STATEMENT

I am a computational biologist with extensive experience in genetics and analyses of omics data, and I am the PI of this application focused on developing methods/tools coupled with high-throughput functional assays for improved variant classification. I am a part of the NHGRI's Baylor College of Medicine Genomics Research to Elucidate the Genetics of Rare Diseases Program (BCM-GREGoR) as a PI of the UTHealth School of Public Health (UTSPH) subaward. My Ph.D. studies in the laboratory of Dr. Barton performed an epigenetic profiling to examine the function of a very well-studied tumor suppressor p53 in normal proliferating liver cells and a potent oncogene Myc in association with Gcn5 during somatic cell programming in mice. During my postdoctoral fellowship with Dr. Lupski, I worked extensively with the data we propose to analyze from the Baylor College of Medicine: Genomics Research to Elucidate the Genetics of Rare diseases (BCM-GREGoR), spearheading a wide variety of projects that improved understanding of the relationship between gene function and human phenotypes and fascinating yet understudied genetic mechanisms of disease in Mendelian disease biology through development of computational algorithms, tools and analytical frameworks. These include development of a nonsense-mediated decay (NMD) escape intolerance score metric to rank genes based on their depletion of protein truncating variants (PTVs) within the gene region predicted to escape NMD in control databases that would render them to cause potential dominant disease traits through potential dominant-negative or gain-of-function (GoF) effects. Collectively, I have been at the forefront of Mendelian genetics and genomics for the last 9 years and carried this role forward as an Assistant Professor of Epidemiology in the UTHealth School of Public Health (UTSPH).

My lab is currently developing novel predictive models and computational tools to examine the prevalence of transcripts present with NMD-escape alleles that are associated with a wide range of human phenotypes due to production of defective proteins. To achieve this, I spearheaded collaborations with investigators at the UTHSC and other institutes all over the world to identify NMD escape alleles associated with diverse human phenotypes including Mendelian and complex disease traits and congenital heart defects. The extension of this proposed approach to the dataset available from the NHLBI's Trans-Omics Precision Medicine (TOPMed) program and Simons Foundation Autism Reseach Initiative (SFARI) to characterize the impact of predicted NMD-escape alleles on quantitative hematological and hemostatic traits and neurodevelopmental disorders were awarded awarded. We are also working on benchmarking those NMD escape alleles' computational predictions with both molecular validations (Dr. Sujatha Jagannathan). I am highly committed to mentoring the next generation of scientists including mentoring my own graduate students and postdoctoral scholar and serving in the thesis committees of five graduate students from the Baylor College of Medicine, UTHealth McGovern Medical School and University of Pittsburg. Last three summers, I also lectured a course for graduate students introductory for statistical methods and software for analyzing measured genetic variation in human studies. I am also currently teaching an introductory course in epidemiology.

GENERAL BIOGRAPHICAL INFORMATION

A. Personal

1. Zeynep H Coban-Akdemir
2. PERMANENT RESIDENT (USA)

B. Education (include institution/location, degree, and dates of attendance)

Sabanci University, Faculty of Engineering and Natural Sciences, Istanbul, Turkey
B.S. in Biological Sciences and Bioengineering, 2002-2007 Harvard
University, Department of Biostatistics, Boston, MA, USA
A.M. in Biostatistics, 2007-2009
The University of Texas Health Science Center at Houston Graduate School of
Biomedical Sciences, Houston, TX, USA
Ph.D. in Biomedical Sciences, 2010-2014
Thesis: p53 Maintains Hepatic Cell Identity During Liver Regeneration Advisor:
Michelle C. Barton
Baylor College of Medicine, Molecular and Human Genetics, Houston, TX, USA
Postgraduate Training, 2014-2019
Advisor: James R. Lupski

C. Academic Appointments

The University of Texas Health Science Center at Houston School of Public Health,
Epidemiology, Human Genetics & Environmental Sciences, Houston, TX, USA
Assistant Professor, January 2020

D. Awards

Simons Foundation Pilot Award, 2024

TOPMed fellowship, 2023

ASHG/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research (Semifinalist), 2017
Harvard PQG conference - Whole Genome Sequencing Analysis: Comprehensive Capture of Genetic Variants (Stellar Abstract Award), 2016
ASHG/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research (Semifinalist), 2015
The University of Health Science Center at Houston: Genes & Development Research

Achievement Award (Post-candidacy category, second place), 2013
The University of Health Science Center at Houston: The Center for Cancer Epigenetics Fellowship, 2013
The University of Health Science Center at Houston: Genes & Development Off-site Course Award (First place), 2013
The University of Health Science Center at Houston: Genes & Development Retreat Poster Award (Pre-candidacy category), 2012
The University of Health Science Center at Houston: Hearst Foundation Student Research & Education Award (Pre-candidacy category), 2012
The University of Health Science Center at Houston: Genes & Development Research Achievement Award (Pre-candidacy category), 2012
Harvard University: Pfizer Fellowship, 2008
Sabanci University: ranked 2nd amongst Biological Sciences and Bioengineering 2007 graduate batch with CGPA 3.72/4.00, 2007
Sabanci University: Certificate of High Honor for 6 semesters, 2002-2007 Sabanci University: Certificate of Honor for 2 semesters, 2002-2007
Sabanci University: Honor Scholarship for ranking 95th out of approximately 1,700,000 test takers in the 2002 annual central nationwide university examination, 2002

II. RESEARCH INFORMATION

A. Research Support

1U01HG011758
Posey (PI), Role: Co-Investigator
07/15/2021 – 07/15/2026
“Baylor College of Medicine - Mendelian Genomics Research Center (BCM-MGRC)”
Development of new algorithms and methods and also integration of different kinds of genomic dataset to facilitate a better understanding of Mendelian disease biology.

NHLBI Trans-Omics for Precision Medicine (TOPMed) fellowship
Zeynep Coban Akdemir, Role: Principal Investigator
03/13/2023 – 03/12/2024
“Systematic profiling and characterization of transcripts that are associated with quantitative blood disease traits through putative gain-of-function variants”
The goal of this project is to establish enhanced predictive models and computational pipelines for comprehensive annotation of nonsense-mediated decay (NMD) outcomes of protein truncating variants (PTVs) (NMD-triggering vs. NMD-escape) using the whole genome sequencing (WGS) coupled with RNA-Sequencing (RNA-Seq) data available from the Genotype-Tissue Expression (GTEx) and TOPMed datasets and assess the impact of NMD-escape PTVs (putative GoF alleles) on quantitative hematological and hemostatic traits in the setting of the TOPMed Hematology and Hemostasis (H&H) Working Group

1X01HL148621-01
Brown (PI), Role: Co-Investigator
05/2020 –12/2023
“Transcriptome and Whole Genome analysis of HIPS samples: A prospectivecohort study of previously untreated patients with hemophilia A during their first 50 exposures to rFVIII”
The goal of this project is to characterize changes in immune cell gene expression during the first 50 exposure days, changes in gene expression in FVIII-specific lymphocytes during the first 50 FVIII

exposure days and the changes in gene expression during Immune Tolerance Induction (ITI) in patients with inhibitors.

The Simons Foundation Autism Research Initiative (SFARI) Pilot award
Zeynep Coban Akdemir, Role: Principal Investigator
06/01/2024 – 05/31/2026
"Systematic Identification and Characterization of Transcripts Associated with ASD through Potential Gain-of-Function Alleles"
The objective of this project is to develop advanced predictive models and computational pipelines for thorough annotation of nonsense-mediated decay (NMD) outcomes in protein truncating variants (PTVs) (differentiating between NMD-triggering and NMD-escape) using whole genome sequencing (WGS) combined with RNA-Sequencing (RNA-Seq) data from the Simons Simplex Collection dataset. Additionally, the project aims to evaluate the impact of NMD-escape PTVs (potential gain-of-function alleles) on autism spectrum disorder.

B. National Scientific Participation (include dates and titles)

Invited Lectures, Presentations, Seminars

Yale Genetics Walter J Burdette Trainee Symposium, New Haven, Connecticut (2019)
NYU Langone Health Center for Human Genetics and Genomics Talk, New York, NY (2019)
UTHealth Epidemiology, Human Genetics and Environmental Sciences Talk, Houston, TX (2019)
UTHealth SPH HGC Seminar Series Talk, Houston, TX (2021)
UTHealth SPH HGC Seminar Series Talk, Houston, TX (2022)
BCM Human Genome Sequencing Center Virtual Seminar Series Talk, Houston, TX (2022)
GREGoR Science Seminar Series Talk, Houston, TX (2023)
CPH Spring Seminar Series Talk, Houston, TX (2023)
TOPMed All Hands Workshop, Houston, TX (2023)
TOPMed Annual Meeting, Betheda, TX (2024)
UTHealth SPH HGC Seminar Series Talk, Houston, TX (2024)
GREGoR In-Person Meeting, Boston, MA (2024)
Horizons in Genomics Seminar, Virtual (2024)

C. Publications

Bozkurt-Yozgatli, T., Pehlivan, D., Gibbs, R.A., Sezerman, U., Posey, J.E., Lupski, J.R., and **Coban-Akdemir, Z.** (2024). Multilocus pathogenic variants contribute to intrafamilial clinical heterogeneity: a retrospective study of sibling pairs with neurodevelopmental disorders. BMC Med Genomics *17*, 85. 10.1186/s12920-024-01852-4.

Calame, D.G., Wong, J.H., Panda, P., Nguyen, D.T., Leong, N.C.P., Sangermano, R., Patankar, S.G., Abdel-Hamid, M.S., AlAbdi, L., Safwat, S., Flannery, K.P., Dardas, Z., Fatih, J.M., Murali, C., Kannan, V., Lotze, T.E., Herman, I., Ammouri, F., Rezich, B., Efthymiou, S., Alavi, S., Murphy, D., Firoozfar, Z., Nasab, M.E., Bahreini, A., Ghasemi, M., Haridy, N.A., Goldouzi, H.R., Eghbal, F., Karimiani, E.G., Begtrup, A., Elloumi, H., Srinivasan, V.M., Gowda, V.K., Du, H., Jhangiani, S.N., **Coban-Akdemir, Z.** …& Lupski, J.R., and Nguyen, L.N. (2024). Biallelic variation in the choline and ethanolamine transporter FLVCR1 underlies a severe developmental disorder spectrum. Genet Med, 101273. 10.1016/j.gim.2024.101273.

Chong, J.X., Berger, S.I., Baxter, S., Smith, E., Xiao, C., Calame, D.G., Hawley, M.H., Rivera-Munoz, E.A., DiTroia, S., Genomics Research to Elucidate the Genetics of Rare Diseases, C., Bamshad, M.J., and Rehm, H.L. (2024). Considerations for reporting variants in novel candidate genes identified during clinical genomic testing. Genet Med *26*, 101199. 10.1016/j.gim.2024.101199.

Coban-Akdemir, Z., Song, X., Ceballos, F. C., Pehlivan, D., Karaca, E., Bayram, Y., ... & Lupski, J. R. (2024). The impact of the Turkish population variome on the genomic architecture of rare disease traits. Genetics in Medicine Open, 101830.

Copeland, I., Wonkam-Tingang, E., Gupta-Malhotra, M., Hashmi, S.S., Han, Y., Jajoo, A., Hall, N.J., Hernandez, P.P., Lie, N., Liu, D., Xu, J., Rosenfeld, J., Haldipur, A., Desire, Z., **Coban-Akdemir, Z.H.**, Scott, D.A., Li, Q., Chao, H.T., Zaske, A.M., Lupski, J.R., Milewicz, D.M., Shete, S., Posey, J.E., and Hanchard, N.A. (2024). Exome sequencing implicates ancestry-related Mendelian variation at SYNE1 in childhood-onset essential hypertension. JCI Insight *9*. 10.1172/jci.insight.172152.

Dardas, Z., Fatih, J.M., Jolly, A., Dawood, M., Du, H., Grochowski, C.M., Jones, E.G., Jhangiani, S.N., Wehrens, X.H.T., Liu, P., Bi, W., Boerwinkle, E., Posey, J.E., Muzny, D.M., Gibbs, R.A., Lupski, J.R.,

Coban-Akdemir, Z.*, and Morris, S.A.* (2024). NODAL variants are associated with a continuum of laterality defects from simple D-transposition of the great arteries to heterotaxy. Genome Med *16*, 53. 10.1186/s13073-024-01312-9. (* Co-corresponding authors)/

Du, H., Dardas, Z., Jolly, A., Grochowski, C.M., Jhangiani, S.N., Li, H., Muzny, D., Fatih, J.M., Yesil, G., Elcioglu, N.H., Gezdirici, A., Marafi, D., Pehlivan, D., Calame, D.G., Carvalho, C.M.B., Posey, J.E., Gambin, T., **Coban-Akdemir, Z.**, and Lupski, J.R. (2024). HMZDupFinder: a robust computational approach for detecting intragenic homozygous duplications from exome sequencing data. Nucleic Acids Res *52*, e18. 10.1093/nar/gkad1223.

Duan, R., Marafi, D., Xia, Z.J., Ng, B.G., Maroofian, R., Sumya, F.T., Saad, A.K., Du, H., Fatih, J.M., Hunter, J.V., Elbendary, H.M., Baig, S.M., Abdullah, U., Ali, Z., Efthymiou, S., Murphy, D., Mitani, T., Withers, M.A., Jhangiani, S.N., **Coban-Akdemir, Z.**, Calame, D.G., Pehlivan, D., Gibbs, R.A., Posey, J.E., Houlden, H., Lupashin, V.V., Zaki, M.S., Freeze, H.H., and Lupski, J.R. (2023). Biallelic missense variants in COG3 cause a congenital disorder of glycosylation with impairment of retrograde vesicular trafficking. J Inherit Metab Dis *46*, 1195-1205. 10.1002/jimd.12679.

Petit, F., Longoni, M., Wells, J., Maser, R.S., Bogenschutz, E.L., Dysart, M.J., Contreras, H.T.M., Frenois, F., Pober, B.R., Clark, R.D., Giampietro, P.F., Ropers, H.H., Hu, H., Loscertales, M., Wagner, R., Ai, X., Brand, H., Jourdain, A.S., Delrue, M.A., Gilbert-Dussardier, B., Devisme, L., Keren, B., McCulley, D.J., Qiao, L., Hernan, R., Wynn, J., Scott, T.M., Calame, D.G., **Coban-Akdemir, Z.**, Hernandez, P., Hernandez-Garcia, A., Yonath, H., Lupski, J.R., Shen, Y., Chung, W.K., Scott, D.A., Bult, C.J., Donahoe, P.K., and High, F.A. (2023). PLS3 missense variants affecting the actin-binding domains cause X-linked congenital diaphragmatic hernia and body-wall defects. Am J Hum Genet *110*, 1787-1803. 10.1016/j.ajhg.2023.09.002.

Poli, M.C., Rebolledo-Jaramillo, B., Lagos, C., Orellana, J., Moreno, G., Martin, L.M., Encina, G., Bohme, D., Faundes, V., Zavala, M.J., Hasbun, T., Fischer, S., Brito, F., Araya, D., Lira, M., de la Cruz, J., Astudillo, C., Lay-Son, G., Cares, C., Aracena, M., Martin, E.S., **Coban-Akdemir, Z.**, Posey, J.E., Lupski, J.R., and Repetto, G.M. (2024). Decoding complex inherited phenotypes in rare disorders: the DECIPHERD initiative for rare undiagnosed diseases in Chile. Eur J Hum Genet. 10.1038/s41431-023-01523-

Holmstrom, L., Chaudhary, N.S., Nakamura, K., Chugh, H., Uy-Evanado, A., Norby, F., Metcalf, G.A., Menon, V.K., Yu, B., Boerwinkle, E., Chugh, S.S., **Akdemir, Z.**, and Kransdorf, E.P. (2023). Rare Genetic Variants Associated With Sudden Cardiac Arrest in the Young: A Prospective, Population-Based Study. Circ Genom Precis Med, e004105. 10.1161/CIRCGEN.123.004105.

Pan, X., Yun, J., **Coban Akdemir, Z.H.***, Jiang, X., Wu*, E., Huang, J.H.*, Sahni, N.*, and Yi, S.S.* (2023). AI-DrugNet: A network-based deep learning model for drug repurposing and combination therapy in neurological disorders. Comput Struct Biotechnol J *21*, 1533-1542. 10.1016/j.csbj.2023.02.004. (*Co-corresponding authors)

Klonowski, J., Liang, Q., **Akdemir, Z.C.**, Lo, C., and Kostka, D. (2023). aenmd: Annotating escape from nonsense-mediated decay for transcripts with protein-truncating variants. bioRxiv. 10.1101/2023.03.17.533185.

Li, M.M., Awasthi, S., Ghosh, S., Bisht, D., **Coban Akdemir, Z.H.**, Sheynkman, G.M., Sahni, N., and Yi, S.S. (2023). Gain-of-Function Variomics and Multi-omics Network Biology for Precision Medicine. Methods Mol Biol *2660*, 357-372. 10.1007/978-1-0716-3163-8_24.

Pan, X., **Coban Akdemir, Z.H.**, Gao, R., Jiang, X., Sheynkman, G.M., Wu, E., Huang, J.H., Sahni, N., and Yi, S.S. (2023). AD-Syn-Net: systematic identification of Alzheimer's disease-associated mutation and co-mutation vulnerabilities via deep learning. Brief Bioinform *24*. 10.1093/bib/bbad030.

Calame, D.G., Guo, T., Wang, C., Garrett, L., Jolly, A., Dawood, M., Kurolap, A., Henig, N.Z., Fatih, J.M., Herman, I., Du, H., Mitani, T., Becker, L., Rathkolb, B., Gerlini, R., Seisenberger, C., Marschall, S., Hunter, J.V., Gerard, A., Heidlebaugh, A., Challman, T., Spillmann, R.C., Jhangiani, S.N., **Coban-Akdemir, Z.**, Lalani, S., Liu, L., Revah-Politi, A., Iglesias, A., Guzman, E., Baugh, E., Boddaert, N., Rondeau, S., Ormieres, C., Barcia, G., Tan, Q.K.G., Thiffault, I., Pastinen, T., Sheikh, K., Biliciler, S., Mei, D., Melani, F., Shashi, V., Yaron, Y., Steele, M., Wakeling, E., Ostergaard, E., Nazaryan-Petersen, L., Undiagnosed Diseases, N., Millan, F., Santiago-Sim, T., Thevenon, J., Bruel, A.L., Thauvin-Robinet, C., Popp, D., Platzer, K., Gawlinski, P., Wiszniewski, W., Marafi, D., Pehlivan, D., Posey, J.E., Gibbs, R.A., Gailus-Durner, V., Guerrini, R., Fuchs, H., Hrabe de Angelis, M., Holter, S.M., Cheung, H.H., Gu, S., and Lupski, J.R. (2023). Monoallelic variation in DHX9, the gene encoding the DExH-box helicase DHX9, underlies neurodevelopment disorders and Charcot-Marie-Tooth disease. Am J Hum Genet *110*, 1394-1413. 10.1016/j.ajhg.2023.06.013.

Duan, R., Marafi, D., Xia, Z.J., Ng, B.G., Maroofian, R., Sumya, F.T., Saad, A.K., Du, H., Fatih, J.M., Hunter, J.V., Elbendary, H.M., Baig, S.M., Abdullah, U., Ali, Z., Efthymiou, S., Murphy, D., Mitani, T., Withers, M.A., Jhangiani, S.N., **Coban-Akdemir, Z.**, Calame, D.G., Pehlivan, D., Gibbs, R.A., Posey, J.E., Houlden, H., Lupashin, V.V., Zaki, M.S., Freeze, H.H., and Lupski, J.R. (2023). Biallelic missense

variants in COG3 cause a congenital disorder of glycosylation with impairment of retrograde vesicular trafficking. J Inherit Metab Dis. 10.1002/jimd.12679.

Petit, F., Longoni, M., Wells, J., Maser, R.S., Bogenschutz, E.L., Dysart, M.J., Contreras, H.T.M., Frenois, F., Pober, B.R., Clark, R.D., Giampietro, P.F., Ropers, H.H., Hu, H., Loscertales, M., Wagner, R., Ai, X., Brand, H., Jourdain, A.S., Delrue, M.A., Gilbert-Dussardier, B., Devisme, L., Keren, B., McCulley, D.J., Qiao, L., Hernan, R., Wynn, J., Scott, T.M., Calame, D.G., **Coban-Akdemir, Z.**, Hernandez, P., Hernandez-Garcia, A., Yonath, H., Lupski, J.R., Shen, Y., Chung, W.K., Scott, D.A., Bult, C.J., Donahoe, P.K., and High, F.A. (2023). PLS3 missense variants affecting the actin-binding domains cause X-linked congenital diaphragmatic hernia and body-wall defects. Am J Hum Genet *110*, 1787-1803. 10.1016/j.ajhg.2023.09.002.

Boschann, F., Cogulu, M.O., Pehlivan, D., Balachandran, S., Vallecillo-Garcia, P., Grochowski, C.M., Hansmeier, N.R., **Coban Akdemir, Z.H.**, Prada-Medina, C.A., Aykut, A., Fischer-Zirnsak, B., Badura, S., Durmaz, B., Ozkinay, F., Hagerling, R., Posey, J.E., Stricker, S., Gillessen-Kaesbach, G., Spielmann, M., Horn, D., Brockmann, K., Lupski, J.R., Kornak, U., and Schmidt, J. (2022). Biallelic variants in ADAMTS15 cause a novel form of distal arthrogryposis. Genet Med *24*, 2187-2193. 10.1016/j.gim.2022.07.012.

Dawood, M., Akay, G., Mitani, T., Marafi, D., Fatih, J.M., Gezdirici, A., Najmabadi, H., Kahrizi, K., Punetha, J., Grochowski, C.M., Du, H., Jolly, A., Li, H., **Coban-Akdemir, Z.**, Sedlazeck, F.J., Hunter, J.V., Jhangiani, S.N., Muzny, D., Pehlivan, D., Posey, J.E., Carvalho, C.M.B., Gibbs, R.A., and Lupski, J.R. (2023). A biallelic frameshift indel in PPP1R35 as a cause of primary microcephaly. Am J Med Genet A *191*, 794-804. 10.1002/ajmg.a.63080.

Du, H., Jolly, A., Grochowski, C.M., Yuan, B., Dawood, M., Jhangiani, S.N., Li, H., Muzny, D., Fatih, J.M., **Coban-Akdemir, Z.**, Carlin, M.E., Scheuerle, A.E., Witzl, K., Posey, J.E., Pendleton, M., Harrington, E., Juul, S., Hastings, P.J., Bi, W., Gibbs, R.A., Sedlazeck, F.J., Lupski, J.R., Carvalho, C.M.B., and Liu, P. (2022). The multiple de novo copy number variant (MdnCNV) phenomenon presents with peri-zygotic DNA mutational signatures and multilocus pathogenic variation. Genome Med *14*, 122. 10.1186/s13073-022-01123-w.

Faqeih, E.A., Alghamdi, M.A., Almahroos, M.A., Alharby, E., Almuntashri, M., Alshangiti, A.M., Clement, P., Calame, D.G., Qebibo, L., Burglen, L., Doco-Fenzy, M., Mastrangelo, M., Torella, A., Manti, F., Nigro, V., Alban, Z., Alharbi, G.S., Hashmi, J.A., Alraddadi, R., Alamri, R., Mitani, T., Magalie, B., **Coban-Akdemir, Z.**, Geckinli, B.B., Pehlivan, D., Romito, A., Karageorgou, V., Martini, J., Colin, E., Bonneau, D., Bertoli-Avella, A., Lupski, J.R., Pastore, A., Peake, R.W.A., Dallol, A., Alfadhel, M., and Almontashiri, N.A.M. (2023). Biallelic variants in HECT E3 paralogs, HECTD4 and UBE3C, encoding ubiquitin ligases cause neurodevelopmental disorders that overlap with Angelman syndrome. Genet Med *25*, 100323. 10.1016/j.gim.2022.10.006.

Hijazi, H., Reis, L.M., Pehlivan, D., Bernstein, J.A., Muriello, M., Syverson, E., Bonner, D., Estiar, M.A., Gan-Or, Z., Rouleau, G.A., Lyulcheva, E., Greenhalgh, L., Tessarech, M., Colin, E., Guichet, A., Bonneau, D., van Jaarsveld, R.H., Lachmeijer, A.M.A., Ruaud, L., Levy, J., Tabet, A.C., Ploski, R., Rydzanicz, M., Kepczynski, L., Polatynska, K., Li, Y., Fatih, J.M., Marafi, D., Rosenfeld, J.A., Coban-**Akdemir, Z.**, Bi, W., Gibbs, R.A., Hobson, G.M., Hunter, J.V., Carvalho, C.M.B., Posey, J.E., Semina, E.V., and Lupski, J.R. (2022). TCEAL1 loss-of-function results in an X-linked dominant neurodevelopmental syndrome and drives the neurological disease trait in Xq22.2 deletions. Am J Hum Genet *109*, 2270-2282. 10.1016/j.ajhg.2022.10.007.

Jolly, A., Du, H., Borel, C., Chen, N., Zhao, S., Grochowski, C.M., Duan, R., Fatih, J.M., Dawood, M., Salvi, S., Jhangiani, S.N., Muzny, D.M., Koch, A., Rouskas, K., Glentis, S., Deligeoroglou, E., Bacopoulou, F., Wise, C.A., Dietrich, J.E., Van den Veyver, I.B., Dimas, A.S., Brucker, S., Sutton, V.R., Gibbs, R.A., Antonarakis, S.E., Wu, N., **Coban-Akdemir, Z.H.**, Zhu, L., Posey, J.E., and Lupski, J.R. (2023). Rare variant enrichment analysis supports GREB1L as a contributory driver gene in the etiology of Mayer-Rokitansky-Kuster-Hauser syndrome. HGG Adv *4*, 100188. 10.1016/j.xhgg.2023.100188.

Khalaf-Nazzal, R., Fasham, J., Inskeep, K.A., Blizzard, L.E., Leslie, J.S., Wakeling, M.N., Ubeyratna, N., Mitani, T., Griffith, J.L., Baker, W., Al-Hijawi, F., Keough, K.C., Gezdirici, A., Pena, L., Spaeth, C.G., Turnpenny, P.D., Walsh, J.R., Ray, R., Neilson, A., Kouranova, E., Cui, X., Curiel, D.T., Pehlivan, D., **Akdemir, Z.C.**, Posey, J.E., Lupski, J.R., Dobyns, W.B., Stottmann, R.W., Crosby, A.H., and Baple, E.L. (2022). Bi-allelic CAMSAP1 variants cause a clinically recognizable neuronal migration disorder. Am J Hum Genet *109*, 2068-2079. 10.1016/j.ajhg.2022.09.012.

Serey-Gaut, M., Cortes, M., Makrythanasis, P., Suri, M., Taylor, A.M.R., Sullivan, J.A., Asleh, A.N., Mitra, J., Dar, M.A., McNamara, A., Shashi, V., Dugan, S., Song, X., Rosenfeld, J.A., Cabrol, C., Iwaszkiewicz, J., Zoete, V., Pehlivan, D., **Akdemir, Z.C.**, Roeder, E.R., Littlejohn, R.O., Dibra, H.K., Byrd, P.J., Stewart, G.S., Geckinli, B.B., Posey, J., Westman, R., Jungbluth, C., Eason, J., Sachdev, R., Evans, C.A., Lemire, G., VanNoy, G.E., O'Donnell-Luria, A., Mau-Them, F.T., Juven, A., Piard, J., Nixon, C.Y.,

Zhu, Y., Ha, T., Buckley, M.F., Thauvin, C., Essien Umanah, G.K., Van Maldergem, L., Lupski, J.R., Roscioli, T., Dawson, V.L., Dawson, T.M., and Antonarakis, S.E. (2023). Bi-allelic TTI1 variants cause an autosomal-recessive neurodevelopmental disorder with microcephaly. Am J Hum Genet *110*, 499-515. 10.1016/j.ajhg.2023.01.006.

Coban-Akdemir, Z., Song, X., Ceballos, F.C., Pehlivan, D., Karaca, E., Bayram, Y., Mitani, T., Gambin, T., Jhangiani, S.N., and Muzny, D.M. (2022). De novo mutation and identity-by-descent drive disease haplotypes, biallelic traits and multilocus pathogenic variation. bioRxiv, 2020.2004. 2027.064824.

Zhang, C., Jolly, A., Shayota, B.J., Mazzeu, J.F., Du, H., Dawood, M., Soper, P.C., Ramalho de Lima, A., Ferreira, B.M., **Coban-Akdemir, Z.**, White, J., Shears, D., Thomson, F.R., Douglas, S.L., Wainwright, A., Bailey, K., Wordsworth, P., Oldridge, M., Lester, T., Calder, A.D., Dumic, K., Banka, S., Donnai, D., Jhangiani, S.N., Potocki, L., Chung, W.K., Mora, S., Northrup, H., Ashfaq, M., Rosenfeld, J.A., Mason, K., Pollack, L.C., McConkie-Rosell, A., Kelly, W., McDonald, M., Hauser, N.S., Leahy, P., Powell, C.M., Boy, R., Honjo, R.S., Kok, F., Martelli, L.R., Filho, V.O., Genomics England Research, C., Muzny, D.M., Gibbs, R.A., Posey, J.E., Liu, P., Lupski, J.R., Sutton, V.R., and Carvalho, C.M.B. (2022). Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. HGG Adv 3, 100074.

Tasdelen, E., Calame, D.G., Akay, G., Mitani, T., Fatih, J.M., Herman, I., Du, H., **Coban-Akdemir, Z.**, Marafi, D., Jhangiani, S.N., Posey, J.E., Gibbs, R.A., Altiparmak, T., Kutlay, N.Y., Lupski, J.R., and Pehlivan, D. (2022). Novel RETREG1 (FAM134B) founder allele is linked to HSAN2B and renal disease in a Turkish family. Am J Med Genet A 188, 2153-2161.

Rodrigues, E.D.S., Griffith, S., Martin, R., Antonescu, C., Posey, J.E., **Coban-Akdemir, Z.**, Jhangiani, S.N., Doheny, K.F., Lupski, J.R., Valle, D., Bamshad, M.J., Hamosh, A., Sheffer, A., Chong, J.X., Einhorn, Y., Cupak, M., and Sobreira, N. (2022). Variant-level matching for diagnosis and discovery: Challenges and opportunities. Hum Mutat 43, 782-790.

Penon-Portmann, M., Eldomery, M.K., Potocki, L., Marafi, D., Posey, J.E., **Coban-Akdemir, Z.**, Harel, T., Grochowski, C.M., Loucks, H., Devine, W.P., Van Ziffle, J., Doherty, D., Lupski, J.R., and Shieh, J.T. (2022). De novo heterozygous variants in SLC30A7 are a candidate cause for Joubert syndrome. Am J Med Genet A 188, 2360-2366.

Munch, J., Engesser, M., Schonauer, R., Hamm, J.A., Hartig, C., Hantmann, E., Akay, G., Pehlivan, D., Mitani, T., **Coban Akdemir, Z**., Tuysuz, B., Shirakawa, T., Dateki, S., Claus, L.R., van Eerde, A.M., Genomics England Research, C., Smol, T., Devisme, L., Franquet, H., Attie-Bitach, T., Wagner, T., Bergmann, C., Hohn, A.K., Shril, S., Pollack, A., Wenger, T., Scott, A.A., Paolucci, S., Buchan, J., Gabriel, G.C., Posey, J.E., Lupski, J.R., Petit, F., McCarthy, A.A., Pazour, G.J., Lo, C.W., Popp, B., and Halbritter, J. (2022). Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. Kidney Int 101, 1039-1053.

Marafi, D., Kozar, N., Duan, R., Bradley, S., Yokochi, K., Al Mutairi, F., Saadi, N.W., Whalen, S., Brunet, T., Kotzaeridou, U., Choukair, D., Keren, B., Nava, C., Kato, M., Arai, H., Froukh, T., Faqeih, E.A., AlAsmari, A.M., Saleh, M.M., Pinto, E.V.F., Pichurin, P.N., Klee, E.W., Schmitz, C.T., Grochowski, C.M., Mitani, T., Herman, I., Calame, D.G., Fatih, J.M., Du, H., **Coban-Akdemir, Z.**, Pehlivan, D., Jhangiani, S.N., Gibbs, R.A., Miyatake, S., Matsumoto, N., Wagstaff, L.J., Posey, J.E., Lupski, J.R., Meijer, D., and Wagner, M. (2022). A reverse genetics and genomics approach to gene paralog function and disease: Myokymia and the juxtaparanode. Am J Hum Genet 109, 1713-1723.

Marafi, D., Fatih, J.M., Kaiyrzhanov, R., Ferla, M.P., Gijavanekar, C., Al-Maraghi, A., Liu, N., Sites, E., Alsaif, H.S., Al-Owain, M., Zakkariah, M., El-Anany, E., Guliyeva, U., Guliyeva, S., Gaba, C., Haseeb, A., Alhashem, A.M., Danish, E., Karageorgou, V., Beetz, C., Subhi, A.A., Mullegama, S.V., Torti, E., Sebastin, M., Breilyn, M.S., Duberstein, S., Abdel-Hamid, M.S., Mitani, T., Du, H., Rosenfeld, J.A., Jhangiani, S.N., **Coban Akdemir, Z.**, Gibbs, R.A., Taylor, J.C., Fakhro, K.A., Hunter, J.V., Pehlivan, D., Zaki, M.S., Gleeson, J.G., Maroofian, R., Houlden, H., Posey, J.E., Sutton, V.R., Alkuraya, F.S., Elsea, S.H., and Lupski, J.R. (2022). Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain 145, 909-924.

Ma, C., Chen, N., Jolly, A., Zhao, S., **Coban-Akdemir, Z.**, Tian, W., Kang, J., Ye, Y., Wang, Y., Koch, A. , Zhang, Y., Qin, C., Bonilla, X., Borel, C., Rall, K., Chen, Z., Jhangiani, S., Niu, Y., Li, X., Qiu, G., Zhang, S., Luo, G., Wu, Z., Bacopoulou, F., Deligeoroglou, E., Zhang, T.J., Rosenberg, C., Gibbs, R.A., Dietrich, J.E., Dimas, A.S., Liu, P., Antonarakis, S.E., Brucker, S.Y., Posey, J.E., Lupski, J.R., Wu, N., Zhu, L., Deciphering Disorders Involving, S., and Group, C.O.S. (2022). Functional characteristics of a broad spectrum of TBX6 variants in Mayer-Rokitansky-Kuster-Hauser syndrome. Genet Med.

Lima, A.R., Ferreira, B.M., Zhang, C., Jolly, A., Du, H., White, J.J., Dawood, M., Lins, T.C., Chiabai, M.A., van Beusekom, E., Cordoba, M.S., Caldas Rosa, E.C.C., Kayserili, H., Kimonis, V., Wu, E., Mellado, C., Aggarwal, V., Richieri-Costa, A., Brunoni, D., Cano, T.M., Jorge, A.A.L., Kim, C.A., Honjo, R., Bertola, D.R., Dandalo-Girardi, R.M., Bayram, Y., Gezdirici, A., Yilmaz-Gulec, E., Gumus, E., Yilmaz, G.C., Okamoto, N., Ohashi, H., **Coban-Akdemir, Z.**, Mitani, T., Jhangiani, S.N., Muzny, D.M., Regattieri, N.A.P., Pogue, R., Pereira, R.W., Otto, P.A., Gibbs, R.A., Ali, B.R., van Bokhoven, H., Brunner, H.G., Sutton, V.R., Lupski, J.R., Vianna-Morgante, A.M., Carvalho, C.M.B., and Mazzeu, J.F. (2022). Phenotypic and mutational spectrum of ROR2-related Robinow syndrome. Hum Mutat 43, 900-918.

Herman, I., Jolly, A., Du, H., Dawood, M., Abdel-Salam, G.M.H., Marafi, D., Mitani, T., Calame, D.G., **Coban-Akdemir, Z.**, Fatih, J.M., Hegazy, I., Jhangiani, S.N., Gibbs, R.A., Pehlivan, D., Posey, J.E., and Lupski, J.R. (2022). Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. Am J Med Genet A 188, 735-750.

Forbes, L.R., Eckstein, O.S., Gulati, N., Peckham-Gregory, E.C., Ozuah, N.W., Lubega, J., El-Mallawany, N.K., Agrusa, J.E., Poli, M.C., Vogel, T.P., Chaimowitz, N.S., Rider, N.L., Mace, E.M., Orange, J.S., Caldwell, J.W., Aldave-Becerra, J.C., Jolles, S., Saettini, F., Chong, H.J., Stray-Pedersen, A., Heslop, H.E., Kamdar, K.Y., Rouce, R.H., Muzny, D.M., Jhangiani, S.N., Gibbs, R.A., **Coban-Akdemir, Z.H.**, Lupski, J.R., McClain, K.L., Allen, C.E., and Chinn, I.K. (2022). Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. J Allergy Clin Immunol 149, 758-766.

Fasham, J., Lin, S., Ghosh, P., Radio, F.C., Farrow, E.G., Thiffault, I., Kussman, J., Zhou, D., Hemming, R., Zahka, K., Chioza, B.A., Rawlins, L.E., Wenger, O.K., Gunning, A.C., Pizzi, S., Onesimo, R., Zampino, G., Barker, E., Osawa, N., Rodriguez, M.C., Neuhann, T.M., Zackai, E.H., Keena, B., Capasso, J., Levin, A.V., Bhoj, E., Li, D., Hakonarson, H., Wentzensen, I.M., Jackson, A., Chandler, K.E., **Coban-Akdemir, Z.H.**, Posey, J.E., Banka, S., Lupski, J.R., Sheppard, S.E., Tartaglia, M., Triggs-Raine, B., Crosby, A.H., and Baple, E.L. (2022). Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genet Med 24, 631-644.

Duan, R., Hijazi, H., Gulec, E.Y., Eker, H.K., Costa, S.R., Sahin, Y., Ocak, Z., Isikay, S., Ozalp, O., Bozdogan, S., Aslan, H., Elcioglu, N., Bertola, D.R., Gezdirici, A., Du, H., Fatih, J.M., Grochowski, C.M., Akay, G., Baylor-Hopkins Center for Mendelian, G., Jhangiani, S.N., Karaca, E., Gu, S., **Coban-Akdemir, Z.**, Posey, J.E., Bayram, Y., Sutton, V.R., Carvalho, C.M.B., Pehlivan, D., Gibbs, R.A., and Lupski, J.R. (2022). Developmental genomics of limb malformations: Allelic series in association with gene dosage effects contribute to the clinical variability. HGG Adv 3, 100132.

Calame, D.G., Herman, I., Maroofian, R., Marshall, A.E., Donis, K.C., Fatih, J.M., Mitani, T., Du, H., Grochowski, C.M., Sousa, S.B., Gijavanekar, C., Bakhtiari, S., Ito, Y.A., Rocca, C., Hunter, J.V., Sutton, V.R., Emrick, L.T., Boycott, K.M., Lossos, A., Fellig, Y., Prus, E., Kalish, Y., Meiner, V., Suerink, M., Ruivenkamp, C., Muirhead, K., Saadi, N.W., Zaki, M.S., Bouman, A., Barakat, T.S., Skidmore, D.L., Osmond, M., Silva, T.O., Murphy, D., Karimiani, E.G., Jamshidi, Y., Jaddoa, A.G., Tajsharghi, H., Jin, S.C., Abbaszadegan, M.R., Ebrahimzadeh-Vesal, R., Hosseini, S., Alavi, S., Bahreini, A., Zarean, E., Salehi, M.M., Al-Sannaa, N.A., Zifarelli, G., Bauer, P., Robson, S.C., **Coban-Akdemir, Z.**, Travaglini, L., Nicita, F., Jhangiani, S.N., Gibbs, R.A., Posey, J.E., Kruer, M.C., Kernohan, K.D., Morales Saute, J.A., Houlden, H., Vanderver, A., Elsea, S.H., Pehlivan, D., Marafi, D., and Lupski, J.R. (2022). Biallelic Variants in the Ectonucleotidase ENTPD1 Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Ann Neurol 92, 304-321.

Boschann, F., Cogulu, M.O., Pehlivan, D., Balachandran, S., Vallecillo-Garcia, P., Grochowski, C.M., Hansmeier, N.R., **Coban Akdemir, Z.H.**, Prada-Medina, C.A., Aykut, A., Fischer-Zirnsak, B., Badura, S., Durmaz, B., Ozkinay, F., Hagerling, R., Posey, J.E., Stricker, S., Gillessen-Kaesbach, G., Spielmann, M., Horn, D., Brockmann, K., Lupski, J.R., Kornak, U., and Schmidt, J. (2022). Biallelic variants in ADAMTS15 cause a novel form of distal arthrogryposis. Genet Med 24, 2187-2193.

Baxter, S.M., Posey, J.E., Lake, N.J., Sobreira, N., Chong, J.X., Buyske, S., Blue, E.E., Chadwick, L.H., **Coban-Akdemir, Z.H.**, Doheny, K.F., Davis, C.P., Lek, M., Wellington, C., Jhangiani, S.N., Gerstein, M., Gibbs, R.A., Lifton, R.P., MacArthur, D.G., Matise, T.C., Lupski, J.R., Valle, D., Bamshad, M.J., Hamosh, A., Mane, S., Nickerson, D.A., Centers for Mendelian Genomics, C., Rehm, H.L., and O'Donnell-Luria, A. (2022). Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genet Med 24, 784-797.

Zhang, C., Mazzeu, J.F., Eisfeldt, J., Grochowski, C.M., White, J., **Akdemir, Z.C.**, Jhangiani, S.N., Muzny, D.M., Gibbs, R.A., Lindstrand, A., Lupski, J.R., Sutton, V.R., and Carvalho, C.M.B. (2021). Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. Am J Med Genet A 185, 3593-3600.

Saettini, F., Poli, C., Vengoechea, J., Bonanomi, S., Orellana, J.C., Fazio, G., Rodriguez, F.H., Noguera, L.P., Booth, C., Jarur-Chamy, V., Shams, M., Iascone, M., Vukic, M., Gasperini, S., Quadri, M., Barroeta Seijas, A., Rivers, E., Mauri, M., Badolato, R., Cazzaniga, G., Bugarin, C., Gaipa, G., Kroes, W.G.M., Moratto, D., van Oostaijen-Ten Dam, M.M., Baas, F., van der Maarel, S., Piazza, R., **Coban-Akdemir, Z.H.**, Lupski, J.R., Yuan, B., Chinn, I.K., Daxinger, L., and Biondi, A. (2021). Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood 137, 493-499.

Calame, D.G., Bakhtiari, S., Logan, R., **Coban-Akdemir, Z.**, Du, H., Mitani, T., Fatih, J.M., Hunter, J.V., Herman, I., Pehlivan, D., Jhangiani, S.N., Person, R., Schnur, R.E., Jin, S.C., Bilguvar, K., Posey, J.E., Koh, S., Firouzabadi, S.G., Alehabib, E., Tafakhori, A., Esmkhani, S., Gibbs, R.A., Noureldeen, M.M., Zaki, M.S., Marafi, D., Darvish, H., Kruer, M.C., and Lupski, J.R. (2021a). Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genet Med.

Calame, D.G., Fatih, J., Herman, I., **Akdemir, Z.C.**, Du, H., Jhangiani, S.N., Gibbs, R.A., Marafi, D., Pehlivan, D., Posey, J.E., Lotze, T., Mancias, P., Bhattacharjee, M.B., and Lupski, J.R. (2021b). Biallelic Pathogenic Variants in TNNT3 Associated With Congenital Myopathy. Neurol Genet *7*, e589.

Calame, D.G., Fatih, J.M., Herman, I., **Coban-Akdemir, Z.**, Du, H., Mitani, T., Jhangiani, S.N., Marafi, D., Gibbs, R.A., Posey, J.E., Mehta, V.P., Mohila, C.A., Abid, F., Lotze, T.E., Pehlivan, D., Adesina, A.M., and Lupski, J.R. (2021c). Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic EMD splice variant. Ann Clin Transl Neurol.

Calame, D.G., Herman, I., Fatih, J.M., Du, H., Akay, G., Jhangiani, S.N., **Coban-Akdemir, Z.**, Milewicz, D.M., Gibbs, R.A., Posey, J.E., Marafi, D., Hunter, J.V., Fan, Y., Lupski, J.R., and Miyake, C.Y. (2021d). Risk of sudden cardiac death in EXOSC5-related disease. Am J Med Genet A *185*, 2532-2540.

Chen, N., Zhao, S., Jolly, A., Wang, L., Pan, H., Yuan, J., Chen, S., Koch, A., Ma, C., Tian, W., Jia, Z., Kang, J., Zhao, L., Qin, C., Fan, X., Rall, K., **Coban-Akdemir, Z.**, Chen, Z., Jhangiani, S., Liang, Z., Niu, Y., Li, X., Yan, Z., Wu, Y., Dong, S., Song, C., Qiu, G., Zhang, S., Liu, P., Posey, J.E., Zhang, F., Luo, G., Wu, Z., Deciphering Disorders Involving, S., group, C.O.s., Su, J., Zhang, J., Chen, E.Y., Rouskas, K., Glentis, S., Bacopoulou, F., Deligeoroglou, E., Chrousos, G., Lyonnet, S., Polak, M., Rosenberg, C., Dingeldein, I., Bonilla, X., Borel, C., Gibbs, R.A., Dietrich, J.E., Dimas, A.S., Antonarakis, S.E., Brucker, S.Y., Lupski, J.R., Wu, N., and Zhu, L. (2021). Perturbations of genes essential for Mullerian duct and Wolffian duct development in Mayer-Rokitansky-Kuster-Hauser syndrome. Am J Hum Genet *108*, 337-345.

Contreras, J.L., Ladino, M.A., Aranguiz, K., Mendez, G.P., **Coban-Akdemir, Z.**, Yuan, B., Gibbs, R.A., Burrage, L.C., Lupski, J.R., Chinn, I.K., Vogel, T.P., Orange, J.S., and Poli, M.C. (2021). Immune Dysregulation Mimicking Systemic Lupus Erythematosus in a Patient With Lysinuric Protein Intolerance: Case Report and Review of the Literature. Front Pediatr *9*, 673957.

Duan, R., Saadi, N.W., Grochowski, C.M., Bhadila, G., Faridoun, A., Mitani, T., Du, H., Fatih, J.M., Jhangiani, S.N., **Akdemir, Z.C.**, Gibbs, R.A., Pehlivan, D., Posey, J.E., Marafi, D., and Lupski, J.R. (2021). A novel homozygous SLC13A5 whole-gene deletion generated by Alu/Alu-mediated rearrangement in an Iraqi family with epileptic encephalopathy. Am J Med Genet A *185*, 1972-1980.

Dworschak, G.C., Punetha, J., Kalanithy, J.C., Mingardo, E., Erdem, H.B., **Akdemir, Z.C.**, Karaca, E., Mitani, T., Marafi, D., Fatih, J.M., Jhangiani, S.N., Hunter, J.V., Dakal, T.C., Dhabhai, B., Dabbagh, O., Alsaif, H.S., Alkuraya, F.S., Maroofian, R., Houlden, H., Efthymiou, S., Dominik, N., Salpietro, V., Sultan, T., Haider, S., Bibi, F., Thiele, H., Hoefele, J., Riedhammer, K.M., Wagner, M., Guella, I., Demos, M., Keren, B., Buratti, J., Charles, P., Nava, C., Heron, D., Heide, S., Valkanas, E., Waddell, L.B., Jones, K.J., Oates, E.C., Cooper, S.T., MacArthur, D., Syrbe, S., Ziegler, A., Platzer, K., Okur, V., Chung, W.K., O'Shea, S.A., Alcalay, R., Fahn, S., Mark, P.R., Guerrini, R., Vetro, A., Hudson, B., Schnur, R.E., Hoganson, G.E., Burton, J.E., McEntagart, M., Lindenberg, T., Yilmaz, O., Odermatt, B., Pehlivan, D., Posey, J.E., Lupski, J.R., and Reutter, H. (2021). Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genet Med *23*, 1715-1725.

Dyment, D.A., O'Donnell-Luria, A., Agrawal, P.B., **Coban Akdemir, Z.**, Aleck, K.A., Antaki, D., Al Sharhan, H., Au, P.B., Aydin, H., Beggs, A.H., Bilguvar, K., Boerwinkle, E., Brand, H., Brownstein, C.A., Buyske, S., Chodirker, B., Choi, J., Chudley, A.E., Clericuzio, C.L., Cox, G.F., Curry, C., de Boer, E., de Vries, B.B.A., Dunn, K., Dutmer, C.M., England, E.M., Fahrner, J.A., Geckinli, B.B., Genetti, C.A., Gezdirici, A., Gibson, W.T., Gleeson, J.G., Greenberg, C.R., Hall, A., Hamosh, A., Hartley, T., Jhangiani, S.N., Karaca, E., Kernohan, K.,

Lauzon, J.L., Lewis, M.E.S., Lowry, R.B., Lopez-Giraldez, F., Matise, T.C., McEvoy-Venneri, J., McInnes, B., Mhanni, A., Garcia Minaur, S., Moilanen, J., Nguyen, A., Nowaczyk, M.J.M., Posey, J.E., Ounap, K., Pehlivan, D., Pajusalu, S., Penney, L.S., Poterba, T., Prontera, P., Doriqui, M.J.R., Sawyer, S.L., Sobreira, N., Stanley, V., Torun, D., Wargowski, D., Witmer, P.D., Wong, I., Xing, J., Zaki, M.S., Zhang, Y., Care4Rare, C., Centers for Mendelian, G., Boycott, K.M., Bamshad, M.J., Nickerson, D.A., Blue, E.E., and Innes, A.M. (2021). Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. Am J Med Genet A *185*, 119-133.

Gambin, T., Liu, Q., Karolak, J.A., Grochowski, C.M., Xie, N.G., Wu, L.R., Yan, Y.H., Cao, Y., **Coban Akdemir, Z.H.**, Wilson, T.A., Jhangiani, S.N., Chen, E., Eng, C.M., Muzny, D., Posey, J.E., Yang, Y., Zhang, D.Y., Shaw, C., Liu, P., Lupski, J.R., and Stankiewicz, P (2020). Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genet Med *22*, 1768-1776.

Gonzaga-Jauregui, C., Yesil, G., Nistala, H., Gezdirici, A., Bayram, Y., Nannuru, K.C., Pehlivan, D., Yuan, B., Jimenez, J., Sahin, Y., Paine, I.S., **Akdemir, Z.C.**, Rajamani, S., Staples, J., Dronzek, J., Howell, K., Fatih, J.M., Smaldone, S., Schlesinger, A.E., Ramirez, N., Cornier, A.S., Kelly, M.A., Haber, R., Chim, S.M., Nieman, K., Wu, N., Walls, J., Baylor-Hopkins Center for Mendelian Genetics, R.G.C., Poueymirou, W., Siao, C.J., Sutton, V.R., Williams, M.S., Posey, J.E., Gibbs, R.A., Carlo, S., Tegay, D.H., Economides, A.N., and Lupski, J.R. (2020). Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. Eur J Hum Genet *28*, 1243-1264.

Li, H., Dawood, M., Khayat, M.M., Farek, J.R., Jhangiani, S.N., Khan, Z.M., Mitani, T., **Coban-Akdemir, Z.,**Lupski, J.R., Venner, E., Posey, J.E., Sabo, A., and Gibbs, R.A. (2021). Exome variant discrepancies due to reference-genome differences. Am J Hum Genet *108*, 1239-1250.

Marafi, D., Fatih, J.M., Kaiyrzhanov, R., Ferla, M.P., Gijavanekar, C., Al-Maraghi, A., Liu, N., Sites, E., Alsaif, H.S., Al-Owain, M., Zakkariah, M., El-Anany, E., Guliyeva, U., Guliyeva, S., Gaba, C., Haseeb, A., Alhashem, A.M., Danish, E., Karageorgou, V., Beetz, C., Subhi, A.A., Mullegama, S.V., Torti, E., Sebastin, M., Breilyn, M.S., Duberstein, S., Abdel-Hamid, M.S., Mitani, T., Du, H., Rosenfeld, J.A., Jhangiani, S.N., **Coban Akdemir, Z.**, Gibbs, R.A., Taylor, J.C., Fakhro, K.A., Hunter, J.V., Pehlivan, D., Zaki, M.S., Gleeson, J.G., Maroofian, R., Houlden, H., Posey, J.E., Sutton, V.R., Alkuraya, F.S., Elsea, S.H., and Lupski, J.R. (2021). Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain.

Mitani, T., Isikay, S., Gezdirici, A., Gulec, E.Y., Punetha, J., Fatih, J.M., Herman, I., Akay, G., Du, H., Calame, D.G., Ayaz, A., Tos, T., Yesil, G., Aydin, H., Geckinli, B., Elcioglu, N., Candan, S., Sezer, O., Erdem, H.B., Gul, D., Demiral, E., Elmas, M., Yesilbas, O., Kilic, B., Gungor, S., Ceylan, A.C., Bozdogan, S., Ozalp, O., Cicek, S., Aslan, H., Yalcintepe, S., Topcu, V., Bayram, Y., Grochowski, C.M., Jolly, A., Dawood, M., Duan, R., Jhangiani, S.N., Doddapaneni, H., Hu, J., Muzny, D.M., Baylor-Hopkins Center for Mendelian, G., Marafi, D., **Akdemir, Z.C.**, Karaca, E., Carvalho, C.M.B., Gibbs, R.A., Posey, J.E., Lupski, J.R., and Pehlivan, D. (2021). High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. Am J Hum Genet *108*, 1981-2005.

Rickman, O.J., Salter, C.G., Gunning, A.C., Fasham, J., Voutsina, N., Leslie, J.S., McGavin, L., Cross, H.E., Posey, J.E., **Akdemir, Z.C.**, Jhangiani, S.N., Lupski, J.R., Baple, E.L., and Crosby, A.H. (2021). Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific C19orf12 isoform. Parkinsonism Relat Disord *82*, 84-86.

Robak, L.A., Du, R., Yuan, B., Gu, S., Alfradique-Dunham, I., Kondapalli, V., Hinojosa, E., Stillwell, A., Young, E., Zhang, C., Song, X., Du, H., Gambin, T., Jhangiani, S.N., **Coban Akdemir, Z.**, Muzny, D.M., Tejomurtula, A., Ross, O.A., Shaw, C., Jankovic, J., Bi, W., Posey, J.E., Lupski, J.R., and Shulman, J.M. (2020). Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurol Genet *6*, e498.

Saad, A.K., Marafi, D., Mitani, T., Du, H., Rafat, K., Fatih, J.M., Jhangiani, S.N., **Coban-Akdemir, Z.**, Baylor-Hopkins Center for Mendelian, G., Gibbs, R.A., Pehlivan, D., Hunter, J.V., Posey, J.E., Zaki, M.S., and Lupski, J.R. (2021). Neurodevelopmental disorder in an Egyptian family with a biallelic ALKBH8 variant. Am J Med Genet A *185*, 1288-1293.

Wohler, E., Martin, R., Griffith, S., Rodrigues, E.D.S., Antonescu, C., Posey, J.E., **Coban-Akdemir, Z.**, Jhangiani, S.N., Doheny, K.F., Lupski, J.R., Valle, D., Hamosh, A., and Sobreira, N. (2021). PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and

sharing of sequence data. Orphanet J Rare Dis *16*, 365.

Zhang, C., Mazzeu, J.F., Eisfeldt, J., Grochowski, C.M., White, J., **Akdemir, Z.C.**, Jhangiani, S.N., Muzny, D.M., Gibbs, R.A., Lindstrand, A., Lupski, J.R., Sutton, V.R., and Carvalho, C.M.B. (2020). Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. Am J Med Genet A.

Assia Batzir, N., Kishor Bhagwat, P., Larson, A., **Coban Akdemir, Z.**, Baglaj, M., Bofferding, L., Bosanko, K.aB., Bouassida, S., Callewaert, B., Cannon, A., Enchautegui Colon, Y., Garnica, A.D., Harr, M.H., Heck, S., Hurst, A.C.E., Jhangiani, S.N., Isidor, B., Littlejohn, R.O., Liu, P., Magoulas, P., Mar Fan, H., Marom, R., McLean, S., Nezarati, M.M., Nugent, K.M., Petersen, M.B., Rocha, M.L., Roeder, E., Smigiel, R., Tully, I., Weisfeld-Adams, J., Wells, K.O., Baylor-Hopkins Center for Mendelian, G., Posey, J.E., Lupski, J.R., Beaudet, A.L., and Wangler, M.F. (2020a). Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Hum Mutat *41*, 641-654.

Assia Batzir, N., Posey, J.E., Song, X., **Akdemir, Z.C.**, Rosenfeld, J.A., Brown, C.W., Chen, E., Holtrop, S.G., Mizerik, E., Nieto Moreno, M., Payne, K., Raas-Rothschild, A., Scott, R., Vernon, H.J., Zadeh, N., Baylor-Hopkins Center for Mendelian, G., Lupski, J.R., and Sutton, V.R. (2020b). Phenotypic expansion of POGZ-related intellectual disability syndrome (White-Sutton syndrome). Am J Med Genet A *182*, 38-52.

Burk, C.M., Coffey, K.E., Mace, E.M., Bostwick, B.L., Chinn, I.K., **Coban-Akdemir, Z.H.**, Jhangiani, S.N., Lupski, J.R., Ortiz, D., Barnum, J.L., Allen, S.W., Robertson, L.M., Orange, J.S., and Chong, H.J. (2020). Immunodeficiency, centromeric instability, and facial anomalies (ICF) syndrome with NK dysfunction and EBV-driven malignancy treated with stem cell transplantation. J Allergy Clin Immunol Pract *8*, 1103-1106 e1103.

Carvalho, C.M.B., **Coban-Akdemir, Z.**, Hijazi, H., Yuan, B., Pendleton, M., Harrington, E., Beaulaurier, J., Juul, S., Turner, D.J., Kanchi, R.S., Jhangiani, S.N., Muzny, D.M., Gibbs, R.A., Baylor-Hopkins Center for Mendelian, G., Stankiewicz, P., Belmont, J.W., Shaw, C.A., Cheung, S.W., Hanchard, N.A., Sutton, V.R., Bader, P.I., and Lupski, J.R. (2019). Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Med *11*, 25.

Chen, S., Jain, M., Jhangiani, S., **Akdemir, Z.C.**, Campeau, P.M., Klein, R.F., Nielson, C., Dai, H., Muzny, D.M., Boerwinkle, E., Gibbs, R.A., Orwoll, E.S., Lupski, J.R., Posey, J.E., and Lee, B. (2020a). Genetic Burden Contributing to Extremely Low or High Bone Mineral Density in a Senior Male Population From the Osteoporotic Fractures in Men Study (MrOS). JBMR Plus *4*, e10335.

Chen, W., Lin, J., Wang, L., Li, X., Zhao, S., Liu, J., **Akdemir, Z.C.**, Zhao, Y., Du, R., Ye, Y., Song, X., Zhang, Y., Yan, Z., Yang, X., Lin, M., Shen, J., Wang, S., Gao, N., Yang, Y., Liu, Y., Li, W., Liu, J., Zhang, N., Yang, X., Xu, Y., Zhang, J., Delgado, M.R., Posey, J.E., Qiu, G., Rios, J.J., Liu, P., Wise, C.A., Zhang, F., Wu, Z., Lupski, J.R., and Wu, N. (2020b). TBX6 missense variants expand the mutational spectrum in a non-Mendelian inheritance disease. Hum Mutat *41*, 182-195.

Coban-Akdemir, Z.H., Charng, W.L., Azamian, M., Paine, I.S., Punetha, J., Grochowski, C.M., Gambin, T., Valdes, S.O., Cannon, B., Zapata, G., Hernandez, P.P., Jhangiani, S., Doddapaneni, H., Hu, J., Boricha, F., Muzny, D.M., Boerwinkle, E., Yang, Y., Gibbs, R.A., Posey, J.E., Wehrens, X.H.T., Belmont, J.W., Kim, J.J., Miyake, C.Y., Lupski, J.R., and Lalani, S.R. (2020). Wolff-Parkinson-White syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. Am J Med Genet A *182*, 1387-1399.

Cook, S.A., Comrie, W.A., Poli, M.C., Similuk, M., Oler, A.J., Faruqi, A.J., Kuhns, D.B., Yang, S., Vargas-Hernandez, A., Carisey, A.F., Fournier, B., Anderson, D.E., Price, S., Smelkinson, M., Abou Chahla, W., Forbes, L.R., Mace, E.M., Cao, T.N., **Coban-Akdemir, Z.H.**, Jhangiani, S.N., Muzny, D.M., Gibbs, R.A., Lupski, J.R., Orange, J.S., Cuvelier, G.D.E., Al Hassani, M., Al Kaabi, N., Al Yafei, Z., Jyonouchi, S., Raje, N., Caldwell, J.W., Huang, Y., Burkhardt, J.K., Latour, S., Chen, B., ElGhazali, G., Rao, V.K., Chinn, I.K., and Lenardo, M.J. (2020). HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. Science *369*, 202-207.

Dias, C.M., Punetha, J., Zheng, C., Mazaheri, N., Rad, A., Efthymiou, S., Petersen, A., Dehghani, M., Pehlivan, D., Partlow, J.N., Posey, J.E., Salpietro, V., Gezdirici, A., Malamiri, R.A., Al Menabawy, N.M., Selim, L.A., Vahidi Mehrjardi, M.Y., Banu, S., Polla, D.L., Yang, E., Rezazadeh Varaghchi, J., Mitani, T., van Beusekom, E., Najafi, M., Sedaghat, A., Keller-Ramey, J., Durham, L., **Coban-Akdemir, Z.**, Karaca, E., Orlova, V., Schaeken, L.L.M., Sherafat, A., Jhangiani, S.N., Stanley, V., Shariati, G., Galehdari, H., Gleeson, J.G., Walsh,

C.A., Lupski, J.R., Seiradake, E., Houlden, H., van Bokhoven, H., and Maroofian, R. (2019). Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. Am J Hum Genet *105*, 1048-1056.

Farria, A.T., Mustachio, L.M., **Akdemir, Z.H.C.**, and Dent, S.Y.R. (2019). GCN5 HAT inhibition reduces human Burkitt lymphoma cell survival through reduction of MYC target gene expression and impeding BCR signaling pathways. Oncotarget *10*, 5847-5858.

Hansen, A.W., Murugan, M., Li, H., Khayat, M.M., Wang, L., Rosenfeld, J., Andrews, B.K., Jhangiani, S.N., **Coban Akdemir, Z.H.**, Sedlazeck, F.J., Ashley-Koch, A.E., Liu, P., Muzny, D.M., Task Force for Neonatal, G., Davis, E.E., Katsanis, N., Sabo, A., Posey, J.E., Yang, Y., Wangler, M.F., Eng, C.M., Sutton, V.R., Lupski, J.R., Boerwinkle, E., and Gibbs, R.A. (2019). A Genocentric Approach to Discovery of Mendelian Disorders. Am J Hum Genet *105*, 974-986.

Karaca, E., Posey, J.E., Bostwick, B., Liu, P., Gezdirici, A., Yesil, G., **Coban Akdemir, Z.**, Bayram, Y., Harms, F.L., Meinecke, P., Alawi, M., Bacino, C.A., Sutton, V.R., Kortum, F., and Lupski, J.R. (2019). Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycle-opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. Am J Med Genet A*179*, 2056-2066.

Karolak, J.A., Szafranski, P., Kilner, D., Patel, C., Scurry, B., Kinning, E., Chandler, K., Jhangiani, S.N., **Coban Akdemir, Z.H.**, Lupski, J.R., Popek, E., and Stankiewicz, P. (2019). Heterozygous CTNNB1 and TBX4 variants in a patient with abnormal lung growth, pulmonary hypertension, microcephaly, and spasticity. Clin Genet *96*, 366-370.

Kuhny, M., Forbes, L.R., Cakan, E., Vega-Loza, A., Kostiuk, V., Dinesh, R.K., Glauzy, S., Stray-Pedersen, A., Pezzi, A.E., Hanson, I.C., Vargas-Hernandez, A., Xu, M.L., **Coban-Akdemir, Z.H.**, Jhangiani, S.N., Muzny, D.M., Gibbs, R.A., Lupski, J.R., Chinn, I.K., Schatz, D.G., Orange, J.S., and Meffre, E. (2020). Disease-associated CTNNBL1 mutation impairs somatic hypermutation by decreasing nuclear AID. J Clin Invest *130*, 4411-4422.

Lam, M.T., Coppola, S., Krumbach, O.H.F., Prencipe, G., Insalaco, A., Cifaldi, C., Brigida, I., Zara, E., Scala, S., Di Cesare, S., Martinelli, S., Di Rocco, M., Pascarella, A., Niceta, M., Pantaleoni, F., Ciolfi, A., Netter, P., Carisey, A.F., Diehl, M., Akbarzadeh, M., Conti, F., Merli, P., Pastore, A., Levi Mortera, S., Camerini, S., Farina, L., Buchholzer, M., Pannone, L., Cao, T.N., **Coban-Akdemir, Z.H.**, Jhangiani, S.N., Muzny, D.M., Gibbs, R.A., Basso-Ricci, L., Chiriaco, M., Dvorsky, R., Putignani, L., Carsetti, R., Janning, P., Stray-Pedersen, A., Erichsen, H.C., Horne, A., Bryceson, Y.T., Torralba-Raga, L., Ramme, K., Rosti, V., Bracaglia, C., Messia, V., Palma, P., Finocchi, A., Locatelli, F., Chinn, I.K., Lupski, J.R., Mace, E.M., Cancrini, C., Aiuti, A., Ahmadian, M.R., Orange, J.S., De Benedetti, F., and Tartaglia, M. (2019). A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. J Exp Med *216*, 2778-2799.

Lin, M., Liu, Z., Liu, G., Zhao, S., Li, C., Chen, W., **Coban Akdemir, Z.**, Lin, J., Song, X., Wang, S., Xu, Q., Zhao, Y., Wang, L., Zhang, Y., Yan, Z., Liu, S., Liu, J., Chen, Y., Zuo, Y., Yang, X., Sun, T., Yang, X.Z., Niu, Y., Li, X., You, W., Qiu, B., Ding, C., Liu, P., Zhang, S., Carvalho, C.M.B., Posey, J.E., Qiu, G., Deciphering Disorders Involving, S., study, C.O., Lupski, J.R., Wu, Z., Zhang, J., and Wu, N. (2020). Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in FBN1. Mol Genet Genomic Med *8*, e1023.

Mace, E.M., Paust, S., Conte, M.I., Baxley, R.M., Schmit, M.M., Patil, S.L., Guilz, N.C., Mukherjee, M., Pezzi, A.E., Chmielowiec, J., Tatineni, S., Chinn, I.K., **Akdemir, Z.C.**, Jhangiani, S.N., Muzny, D.M., Stray-Pedersen, A., Bradley, R.E., Moody, M., Connor, P.P., Heaps, A.G., Steward, C., Banerjee, P.P., Gibbs, R.A., Borowiak, M., Lupski, J.R., Jolles, S., Bielinsky, A.K., and Orange, J.S. (2020). Human NK cell deficiency as a result of biallelic mutations in MCM10. J Clin Invest*130*, 5272-5286.

Marafi, D., Mitani, T., Isikay, S., Hertecant, J., Almannai, M., Manickam, K., Abou Jamra, R., El-Hattab, A.W., Rajah, J., Fatih, J.M., Du, H., Karaca, E., Bayram, Y., Punetha, J., Rosenfeld, J.A., Jhangiani, S.N., Boerwinkle, E., **Akdemir, Z.C.**, Erdin, S., Hunter, J.V., Gibbs, R.A., Pehlivan, D., Posey, J.E., and Lupski, J.R. (2020). Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Ann Clin Transl Neurol *7*, 610-627.

Mitani, T., Punetha, J., Akalin, I., Pehlivan, D., Dawidziuk, M., **Coban Akdemir, Z.**, Yilmaz, S., Aslan, E., Hunter, J.V., Hijazi, H., Grochowski, C.M., Jhangiani, S.N., Karaca, E., Fatih, J.M., Iwanowski, P., Gambin, T., Wlasienko, P., Goszczanska-Ciuchta, A., Bekiesinska-Figatowska, M., Hosseini, M., Arzhangi, S., Najmabadi, H., Rosenfeld, J.A., Du, H., Marafi, D., Blaser, S., Teitelbaum, R., Silver, R., Baylor-Hopkins Center for Mendelian, G., Posey,

J.E., Ropers, H.H., Gibbs, R.A., Wiszniewski, W., Lupski, J.R., Chitayat, D., Kahrizi, K., and Gawlinski, P. (2019). Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. Am J Hum Genet *105*, 1005-1015.

Ngo, K.J., Rexach, J.E., Lee, H., Petty, L.E., Perlman, S., Valera, J.M., Deignan, J.L., Mao, Y., Aker, M., Posey, J.E., Jhangiani, S.N., **Coban-Akdemir, Z.H.**, Boerwinkle, E., Muzny, D., Nelson, A.B., Hassin-Baer, S., Poke, G., Neas, K., Geschwind, M.D., Grody, W.W., Gibbs, R., Geschwind, D.H., Lupski, J.R., Below, J.E., Nelson, S.F., and Fogel, B.L. (2020). A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Hum Mutat *41*, 487-501.

Van Bergen, N.J., Guo, Y., Al-Deri, N., Lipatova, Z., Stanga, D., Zhao, S., Murtazina, R., Gyurkovska, V., Pehlivan, D., Mitani, T., Gezdirici, A., Antony, J., Collins, F., Willis, M.J.H., **Coban Akdemir, Z.H.**, Liu, P., Punetha, J., Hunter, J.V., Jhangiani, S.N., Fatih, J.M., Rosenfeld, J.A., Posey, J.E., Gibbs, R.A., Karaca, E., Massey, S., Ranasinghe, T.G., Sleiman, P., Troedson, C., Lupski, J.R., Sacher, M., Segev, N., Hakonarson, H., and Christodoulou, J. (2020). Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain *143*, 112-130.

Carvalho, C. M., **Coban-Akdemir, Z.**, Hijazi, H., Yuan, B., Pendleton, M., Harrington, E., ... & Jhangiani, S. N. (2019). Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. *Genome medicine*, *11*(1), 25.

Batzir, N. A., Posey, J. E., Song, X., **Akdemir, Z. C.**, Rosenfeld, J. A., Brown, C. W., ... & Payne, K. (2019). Phenotypic expansion of POGZ-related intellectual disability syndrome (White-Sutton syndrome). *American Journal of Medical Genetics Part A*.

Lin, M., Liu, Z., Liu, G., Zhao, S., Li, C., Chen, W., ... & Xu, Q. (2019). Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in FBN1. *Molecular genetics & genomic medicine*.

Eckstein, O. S., Gulati, N., Forbes, L., Peckham-Gregory, E., Ozuah, N. W., Poli, M. C., ... & Jolles, S. (2019). Genomic Characterization of a Pediatric Cohort with Non-Malignant Lymphoproliferative Disorders.

Dias, C. M., Punetha, J., Zheng, C., Mazaheri, N., Rad, A., Efthymiou, S., ... & Posey, J. E. (2019). Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. *The American Journal of Human Genetics*, *105*(5), 1048-1056.

Hansen, A. W., Murugan, M., Li, H., Khayat, M. M., Wang, L., Rosenfeld, J., ... & Ashley-Koch, A. E. (2019). A Genocentric Approach to Discovery of Mendelian Disorders. *The American Journal of Human Genetics*, *105*(5), 974-986.

Mitani, T., Punetha, J., Akalin, I., Pehlivan, D., Dawidziuk, M., **Akdemir, Z. C**., ... & Grochowski, C. M. (2019). Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. *The American Journal of Human Genetics*, *105*(5), 1005-1015.

Ngo, K. J., Rexach, J. E., Lee, H., Petty, L. E., Perlman, S., Valera, J. M., ... & Jhangiani, S. N. (2019). A Diagnostic Ceiling for Exome Sequencing in Cerebellar Ataxia and Related Neurological Disorders. *Human mutation*.

Lam, M. T., Coppola, S., Krumbach, O. H., Prencipe, G., Insalaco, A., Cifaldi, C., ... & Martinelli, S. (2019). A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. *Journal of Experimental Medicine*, *216*(12), 2778-2799.

Farria, A. T., Mustachio, L. M., **Akdemir, Z. H. C**., & Dent, S. Y. (2019). GCN5 HAT inhibition reduces human Burkitt lymphoma cell survival through reduction of MYC target gene expression and impeding BCR signaling pathways. *Oncotarget*, *10*(56), 5847.

Karaca, E., Posey, J. E., Bostwick, B., Liu, P., Gezdirici, A., Yesil, G., ... & Alawi, M. (2019). Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycle-opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. *American Journal of Medical Genetics Part A*.

Karolak, J. A., Szafranski, P., Kilner, D., Patel, C., Scurry, B., Kinning, E., ... & Popek, E. (2019). Heterozygous CTNNB1 and TBX4 variants in a patient with abnormal lung growth, pulmonary hypertension, microcephaly, and spasticity. *Clinical genetics*.

Burk, C. M., Coffey, K. E., Mace, E. M., Bostwick, B. L., Chinn, I. K., Coban-Akdemir, Z. H., ... & Allen, S. W. (2019). Immunodeficiency, centromeric instability, and facial anomalies (ICF) syndrome with NK dysfunction and EBV-driven malignancy treated with stem cell transplantation. *The Journal of Allergy and Clinical Immunology: In Practice*.

Chen, W., Lin, J., Wang, L., Li, X., Zhao, S., Liu, J., ... & Song, X. (2019). TBX6 missense

variants expand the mutational spectrum in a non-Mendelian inheritance disease. *Human mutation*.

Punetha, J., Karaca, E., Gezdirici, A., Lamont, R. E., Pehlivan, D., Marafi, D., ... & Jhangiani, S. N. (2019). Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. *Annals of clinical and translational neurology*.

Colombo, R., Jolly, A., Bayram, Y., Karaca, E., Di Simone, N., Scambia, G., ... & Lupski, J. R. (2019, July). Exome sequencing reveals novel IGSF10 variation in patients with hypergonadotropic hypogonadism. In *EUROPEAN JOURNAL OF HUMAN GENETICS* (Vol. 27, pp. 15-16). MACMILLAN BUILDING, 4 CRINAN ST, LONDON N1 9XW, ENGLAND: NATURE PUBLISHING GROUP.

Paine, I., Posey, J. E., Grochowski, C. M., Jhangiani, S. N., Rosenheck, S., Kleyner, R., ... & Cappuccio, G. (2019). Paralog Studies Augment Gene Discovery: DDX and DHX Genes. *The American Journal of Human Genetics*.

Pehlivan, D., Bayram, Y., Gunes, N**., Akdemir, Z. C.**, Shukla, A., Bierhals, T., ... & Gulec, E. Y. (2019). The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. *The American Journal of Human Genetics*.

Jolly, A., Bayram, Y., Turan, S., Aycan, Z., Tos, T., Abali, Z. Y., ... & Atay, Z. (2019). Exome sequencing of a primary ovarian insufficiency cohort reveals common molecular etiologies for a spectrum of disease. *The Journal of Clinical Endocrinology & Metabolism*, *104*(8), 3049-3067.

Jansen, S., van der Werf, I. M., Innes, A. M., Afenjar, A., Agrawal, P. B., Anderson, I. J., ... & **Coban-Akdemir, Z. H.** (2019). De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. *European Journal of Human Genetics*, *27*(5), 738.

Posey, J. E., O'Donnell-Luria, A. H., Chong, J. X., Harel, T., Jhangiani, S. N., **Akdemir, Z. H. C.**, ... & Sobreira, N. (2019). Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, *21*(4), 798-812.

Li, A. H., Hanchard, N. A., Azamian, M., D'Alessandro, L. C**., Coban-Akdemir, Z**., Lopez, K. N., ... & Boone, P. M. (2019). Genetic architecture of laterality defects revealed by whole exome sequencing. *European Journal of Human Genetics*, *27*(4), 563.

Cogné, B., Ehresmann, S., Beauregard-Lacroix, E., Rousseau, J., Besnard, T., Garcia, T., ... & Sanders, S. J. (2019). Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. *The American Journal of Human Genetics*, *104*(3), 530-541.

Beck, C. R., Carvalho, C. M., **Akdemir, Z. C.,** Sedlazeck, F. J., Song, X., Meng, Q., ... & Thornton, P. C. (2019). Megabase length hypermutation accompanies human structural variation at 17p11. 2. *Cell*, *176*(6), 1310-1324.

Karolak, J. A., Vincent, M., Deutsch, G., Gambin, T., Cogné, B., Pichon, O., ... & Dipple, K. (2019). Complex compound inheritance of lethal lung developmental disorders due to disruption of the TBX-FGF pathway. *The American Journal of Human Genetics*, *104*(2), 213-228.

Batzir, N. A., Bhagwat, P. K., Larson, A., **Akdemir, Z. C.**, Bagłaj, M., Bofferding, L., ... & Colon, Y. E. (2019). Recurrent Arginine Substitutions in the ACTG2 Gene are the Primary Driver of Disease Burden and Severity in Visceral Myopathy. *Human mutation*.

Aird, A., Lagos, M., Vargas-Hernández, A., Posey, J. E., **Coban-Akdemir, Z.**, Jhangiani, S., ... & Forbes, L. R. (2019). Novel heterozygous mutation in NFKB2 is associated with early onset CVID and a functional defect in NK cells complicated by disseminated CMV infection and severe nephrotic syndrome. *Frontiers in pediatrics*, *7*.

Karaca, E., Posey, J. E., **Akdemir, Z. C.**, Pehlivan, D., Harel, T., Jhangiani, S. N., ... & Bozdogan, S. (2018). Phenotypic expansion illuminates multilocus pathogenic variation. *Genetics in Medicine*, *20*(12), 1528.

Morimoto, M., Waller-Evans, H., Ammous, Z., Song, X., Strauss, K. A., Pehlivan, D., ... & Brigatti, K. W. (2018). Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. *The American Journal of Human Genetics*, *103*(5), 794-807.

Punetha, J., Mackay-Loder, L., Harel, T., **Coban-Akdemir, Z.**, Jhangiani, S. N., Gibbs, R. A., ... & Posey, J. E. (2018). Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. *Molecular genetics and metabolism*, *125*(3), 302-304.

Wang, X., Posey, J. E., Rosenfeld, J. A., Bacino, C. A., Scaglia, F., Immken, L., ... & Zhang, J. (2018). Phenotypic expansion in DDX 3X–a common cause of intellectual disability in

females. *Annals of Clinical and Translational Neurology*, *5*(10), 1277-1285.
Lumaka, A., Race, V., Peeters, H., Corveleyn, A., **Coban-Akdemir, Z.**, Jhangiani, S. N., ... & Vermeesch, J. R. (2018). A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. *American Journal of Medical Genetics Part A*, *176*(9), 1897-1909.
Du, R., Dinckan, N., Song, X., **Coban-Akdemir, Z.**, Jhangiani, S. N., Guven, Y., ... & Below, J. E. (2018). Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. *Human genetics*, *137*(9), 689-703.
Støve, S. I., Blenski, M., Stray-Pedersen, A., Wierenga, K. J., Jhangiani, S. N., Akdemir, Z. C., ... & McNall-Knapp, R. (2018). A novel NAA10 variant with impaired acetyltransferase activity causes developmental delay, intellectual disability, and hypertrophic cardiomyopathy. *European Journal of Human Genetics*, *26*(9), 1294.
Li, L., Jiao, X., D'Atri, I., Ono, F., Nelson, R., Chan, C. C., ... & Zhang, L. (2018). Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. *PLoS genetics*, *14*(8), e1007504.
Coban-Akdemir, Z.*, White, J. J.*, Song, X., Jhangiani, S. N., Fatih, J. M., Gambin, T., ... & Poli, C. (2018). Identifying genes whose mutant transcripts cause dominant disease traits by potential gain-of-function alleles. *The American Journal of Human Genetics*, *103*(2), 171-187. (*Co-first authors).
Song, X., Beck, C. R., Du, R., Campbell, I. M., **Coban-Akdemir, Z.**, Gu, S., ... & Lupski, J. R. (2018). Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. *Genome research*, *28*(8), 1228-1242.
Wiszniewski, W., Gawlinski, P., Gambin, T., Bekiesinska-Figatowska, M., Obersztyn, E., Antczak-Marach, D., ... & Sobecka, K. (2018). Comprehensive genomic analysis of patients with disorders of cerebral cortical development. *European journal of human genetics: EJHG*, *26*(8), 1121-1131.
Ruiz-García, R., Vargas-Hernández, A., Chinn, I. K., Angelo, L. S., Cao, T. N., **Coban-Akdemir, Z.**, ... & Allende, L. M. (2018). Mutations in PI3K110δ cause impaired natural killer cell function partially rescued by rapamycin treatment. *Journal of Allergy and Clinical Immunology*.
Chinn, I. K., Eckstein, O. S., Peckham-Gregory, E. C., Goldberg, B. R., Forbes, L. R., Nicholas, S. K., ... & Heslop, H. E. (2018). Genetic and mechanistic diversity in pediatric hemophagocytic lymphohistiocytosis. *Blood*, *132*(1), 89.
Liu, J., Zhou, Y., Liu, S., Song, X., Yang, X. Z., Fan, Y., ... & Du, R. (2018). The coexistence of copy number variations (CNVs) and single nucleotide polymorphisms (SNPs) at a locus can result in distorted calculations of the significance in associating SNPs to disease. *Human genetics*, *137*(6-7), 553-567.
Poli, M. C., Ebstein, F., Nicholas, S. K., de Guzman, M. M., Forbes, L. R., Chinn, I. K., ... & **Coban-Akdemir, Z. H.** (2018). Heterozygous truncating variants in POMP escape nonsense-mediated decay and cause a unique immune dysregulatory syndrome. *The American Journal of Human Genetics*, *102*(6), 1126-1142.
Cheng, H., Dharmadhikari, A. V., Varland, S., Ma, N., Domingo, D., Kleyner, R., ... & Crews, S. R. (2018). Truncating variants in NAA15 are associated with variable levels of intellectual disability, autism spectrum disorder, and congenital anomalies. *The American Journal of Human Genetics*, *102*(5), 985-994.
Dinckan, N., Du, R., **Akdemir, Z. C.**, Bayram, Y., Jhangiani, S. N., Doddapaneni, H., ... & Kayserili, H. (2018). A biallelic ANTXR1 variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. *American Journal of Medical Genetics Part A*, *176*(4), 1015-1022.
Gawliński, P., Pelc, M., Ciara, E., Jhangiani, S., Jurkiewicz, E., Gambin, T., ... & Muzny, D. (2018). Phenotype expansion and development in Kosaki overgrowth syndrome. *Clinical genetics*, *93*(4), 919-924.
Chen, A., Tiosano, D., Guran, T., Baris, H. N., Bayram, Y., Mory, A., ... & Jhangiani, S. N. (2018). Mutations in the mitochondrial ribosomal protein MRPS22 lead to primary ovarian insufficiency. *Human molecular genetics*, *27*(11), 1913-1926.
Makrythanasis, P., Maroofian, R., Stray-Pedersen, A., Musaev, D., Zaki, M. S., Mahmoud, I. G., ... & Gambin, T. (2018). Biallelic variants in KIF14 cause intellectual disability with microcephaly. *European Journal of Human Genetics*, *26*(3), 330.
Bostwick, B. L., McLean, S., Posey, J. E., Streff, H. E., Gripp, K. W., Blesson, A., ... & Hudgins, L. (2017). Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. *Genome medicine*, *9*(1), 73.

Pursley, A. N., Breman, A. M., Beck, A. E., Patel, A., Beaudet, A. L., Yuan, B., ... & Treadwell-Deering, D. (2017). Identification of novel candidate disease genes from de novo exonic copy number variants. *Genome medicine*, *9*(1), 83.

Lessel, D., Schob, C., Küry, S., Reinders, M. R., Harel, T., Eldomery, M. K., ... & Stegmann, A. P. (2017). De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. *The American Journal of Human Genetics*, *101*(5), 716-724.

Jehee, F. S., de Oliveira, V. T., Gurgel-Giannetti, J., Pietra, R. X., Rubatino, F. V., Carobin, N. V., ... & Brüggenwirth, H. T. (2017). Dual molecular diagnosis contributes to atypical Prader–Willi phenotype in monozygotic twins. *American Journal of Medical Genetics Part A*, *173*(9), 2451-2455.

Dinckan, N., Du, R., Petty, L. E., Coban-Akdemir, Z., Jhangiani, S. N., Paine, I., ... & Hu, J. (2018). Whole-exome sequencing identifies novel variants for tooth agenesis. *Journal of dental research*, *97*(1), 49-59.

White, J. J., Mazzeu, J. F., **Coban-Akdemir, Z.**, Bayram, Y., Bahrambeigi, V., Hoischen, A., ... & Touraine, R. (2018). WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. *The American Journal of Human Genetics*, *102*(1), 27-43.

Gambin, T., Yuan, B., Bi, W., Liu, P., Rosenfeld, J. A**., Coban-Akdemir, Z.**, ... & Dengle, L. (2017). Identification of novel candidate disease genes from de novo exonic copy number variants. *Genome medicine*, *9*(1), 83.

Eldomery, M. K.*, **Coban-Akdemir, Z.***, Harel, T., Rosenfeld, J. A., Gambin, T., Stray-Pedersen, A., ... & Wiszniewski, W. (2017). Lessons learned from additional research analyses of unsolved clinical exome cases. *Genome medicine*, *9*(1), 26. (*Co-first authors).

Luo, X., Rosenfeld, J. A., Yamamoto, S., Harel, T., Zuo, Z., Hall, M., ... & Rotenberg, J. (2017). Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. *PLoS genetics*, *13*(7), e1006905.

Bayram, Y., White, J. J., Elcioglu, N., Cho, M. T., Zadeh, N., Gedikbasi, A., ... **& Akdemir, Z. C.**, …(2017). REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. *The American Journal of Human Genetics*, *101*(1), 149-156.

Moyses-Oliveira, M., Fish, R., Giannuzzi, G., Karaca, E., **Akdemir, Z.**, Petit, F., ... & Zamariolli, M. (2017, June). Balanced X autosome translocation suggests association of AMMECR1 disruption with hearing loss short stature bone and heart alterations. In *MOLECULAR CYTOGENETICS* (Vol. 10). 236.

Halim, D., Brosens, E., Muller, F., Wangler, M. F., Beaudet, A. L., Lupski, J. R., ... & Brouwer, R. W. (2017). Loss-of-function variants in MYLK cause recessive megacystis microcolon intestinal hypoperistalsis syndrome. *The American Journal of Human Genetics*, *101*(1), 123-129.

Gambin, T. *****, **Akdemir, Z. C.*,** Yuan, B., Gu, S., Chiang, T., Carvalho, C., ... & Karaca, E. (2017). Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. *Nucleic acids research*, *45*(4), 1633-1648. (*Co-first authors).

Liu, P., Yuan, B., Carvalho, C. M., Wuster, A., Walter, K., Zhang, L., ... & Gelowani, V. (2017). An Organismal CNV Mutator Phenotype Restricted to Early Human Development. *Cell*, *168*(5), 830-842.

Schoch, K., Meng, L., Szelinger, S., Bearden, D. R., Stray-Pedersen, A., Busk, O. L., ... & Rosenfeld, J. A. (2017). A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. *The American Journal of Human Genetics*, *100*(2), 343-351.

Harms, F. L., Girisha, K. M., Hardigan, A. A., Kortüm, F., Shukla, A., Alawi, M., ... & Ceulemans, S. (2017). Mutations in EBF3 disturb transcriptional profiles and cause intellectual disability, ataxia, and facial dysmorphism. *The American Journal of Human Genetics*, *100*(1), 117-127.

Posey, J. E., Harel, T., Liu, P., Rosenfeld, J. A., James, R. A., **Coban Akdemir, Z. H.**, ... & Xia, F. (2017). Resolution of disease phenotypes resulting from multilocus genomic variation. *New England Journal of Medicine*, *376*(1), 21-31.

Stray-Pedersen, A., Sorte, H. S., Samarakoon, P., Gambin, T., Chinn, I. K., **Akdemir, Z. H. C.**, ... & Jhangiani, S. N. (2017). Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. *Journal of Allergy and Clinical Immunology*, *139*(1), 232-245.

Eldomery, M. K.*, **Akdemir, Z. C.***, Vögtle, F. N., Charng, W. L., Mulica, P., Rosenfeld, J. A., ... & Penney, S. (2016). MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. *Genome medicine*, *8*(1), 106. (*Co-first authors).

Mace, E. M., Bigley, V., Gunesch, J. T., Chinn, I. K., Angelo, L. S., Care, M. A., ... & LaRosa, D.

F. (2017). Biallelic mutations in IRF8 impair human NK cell maturation and function. *The Journal of clinical investigation*, *127*(1), 306.

Chinn, I. K., Sanders, R. P., Stray-Pedersen, A., **Coban-Akdemir, Z. H.**, Kim, V. H. D., Dadi, H., ... & Hanson, I. C. (2017). Novel Combined Immune Deficiency and Radiation Sensitivity Blended Phenotype in an Adult with Biallelic Variations in ZAP70 and RNF168. *Frontiers in Immunology*, *8*.

Pedroza, L. A., Guerrero, N., Stray-Pedersen, A., Tafur, C., Macias, R., Muñoz, G., ... & Lupski, J. R. (2017). First Case of CD40LG Deficiency in ecuador, Diagnosed after Whole exome sequencing in a patient with severe Cutaneous Histoplasmosis. *Frontiers in pediatrics*, *5*.

Loviglio, M. N., Beck, C. R., White, J. J., Leleu, M., Harel, T., Guex, N., ... & Yan, J. (2016). Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. *Genome medicine*, *8*(1), 105.

Charng, W. L., Karaca, E., **Akdemir, Z. C.**, Gambin, T., Atik, M. M., Gu, S., ... & Hu, J. (2016). Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. *BMC medical genomics*, *9*(1), 42.

Gu, S., Szafranski, P., **Akdemir, Z. C.**, Yuan, B., Cooper, M. L., Magriñá, M. A., ... & Patel, A. (2016). Mechanisms for complex chromosomal insertions. *PLoS genetics*, *12*(11), e1006446.

Sorte, H. S., Osnes, L. T., Fevang, B., Aukrust, P., Erichsen, H. C., Backe, P. H., ... & Muzny, D. M. (2016). A potential founder variant in CARMIL2/RLTPR in three Norwegian families with warts, molluscum contagiosum, and T-cell dysfunction. *Molecular genetics & genomic medicine*, *4*(6), 604-616.

Harel, T., Yoon, W. H., Garone, C., Gu, S., **Coban-Akdemir, Z.**, Eldomery, M. K., ... & Fox, S. (2016). Recurrent de novo and biallelic variation of ATAD3A, encoding a mitochondrial membrane protein, results in distinct neurological syndromes. *The American Journal of Human Genetics*, *99*(4), 831-845.

Vetrini, F., D'Alessandro, L. C., **Akdemir, Z. C.**, Braxton, A., Azamian, M. S., Eldomery, M. K., ... & Rijhsinghani,A. (2016). Bi-allelic mutations in PKD1L1 are associated with laterality defects in humans. *The American Journal of Human Genetics*, *99*(4), 886-893.

Bekheirnia, M. R., Bekheirnia, N., Bainbridge, M. N., Gu, S., **Akdemir, Z. H. C.**, Gambin, T., ... & Brewer, E. D. (2016). Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. *Genetics in Medicine*, *19*(4), 412-420.

Lodder, E. M., De Nittis, P., Koopman, C. D., Wiszniewski, W., de Souza, C. F. M., Lahrouchi, N., ... & Nannenberg, E. A. (2016). GNB5 mutations cause an autosomal-recessive multisystem syndrome with sinus bradycardia and cognitive disability. *The American Journal of Human Genetics*, *99*(3), 704-710.

Szafranski, P.*, **Coban-Akdemir, Z. H.***, Rupps, R., Grazioli, S., Wensley, D., Jhangiani, S. N., ... & Stankiewicz, P. (2016). Phenotypic expansion of TBX4 mutations to include acinar dysplasia of the lungs. *American Journal of Medical Genetics Part A*, *170*(9), 2440-2444. (*Co-first authors)

Prescott, T. E., Kulseth, M. A., Heimdal, K. R., Stadheim, B., Hopp, E., Gambin, T., ... & Lupski, J. R. (2016). Two male sibs with severe micrognathia and a missense variant in MED12. *European Journal of Medical Genetics*.

Harel, T., Yesil, G., Bayram, Y., **Coban-Akdemir, Z.**, Charng, W. L., Karaca, E., ... & Rosenfeld, J. A. (2016). Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. *The American Journal of Human Genetics*, *98*(3), 562-570.

Lalani, S. R., Liu, P., Rosenfeld, J. A., Watkin, L. B., Chiang, T., Leduc, M. S., ... & Miyake, C. Y. (2016). Recurrent muscle weakness with rhabdomyolysis, metabolic crises, and cardiac arrhythmia due to bi-allelic TANGO2 mutations. *The American Journal of Human Genetics*, *98*(2), 347-357.

Bayram, Y., Karaca, E., **Akdemir, Z. C.**, Yilmaz, E. O., Tayfun, G. A., Aydin, H., ... & Atik, M. M. (2016). Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. *The Journal of clinical investigation*, *126*(2), 762.

Farlow, J. L., Robak, L. A., Hetrick, K., Bowling, K., Boerwinkle, E., **Coban-Akdemir, Z. H**., ... & Jankovic, J. (2016). Whole-exome sequencing in familial Parkinson disease. *JAMA neurology*, *73*(1), 68-75.

Posey, J. E., Rosenfeld, J. A., James, R. A., Bainbridge, M., Niu, Z., Wang, X., ... & Xia, F. (2015). Molecular diagnostic experience of whole-exome sequencing in adult patients. *Genetics in Medicine*.

Burrage, L. C., Charng, W. L., Eldomery, M. K., Willer, J. R., Davis, E. E., Lugtenberg, D., ... &

Zapata, G. (2015). De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. *The American Journal of Human Genetics*, *97*(6), 904-913.

Karaca, E., Harel, T., Pehlivan, D., Jhangiani, S. N., Gambin, T., **Akdemir, Z. C.**, ... & Hunter, J. V. (2015). Genes that affect brain structure and function identified by rare variant analyses of mendelian neurologic disease. *Neuron*, *88*(3), 499-513.

Karaca, E., Yuregir, O. O., Bozdogan, S. T., Aslan, H., Pehlivan, D., Jhangiani, S. N., ... & Erdin, S. (2015). Rare variants in the notch signaling pathway describe a novel type of autosomal recessive Klippel–Feil syndrome. *American Journal of Medical Genetics Part A*, *167*(11), 2795-2799.

Pehlivan, D., Beck, C. R., Okamoto, Y., Harel, T., **Akdemir, Z. H.**, Jhangiani, S. N., ... & Gonzaga-Jauregui, C. (2015). The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. *Genetics in Medicine*, *18*(5), 443-451.

Bayram, Y., Aydin, H., Gambin, T., **Akdemir, Z. C.**, Atik, M. M., Karaca, E., ... & Lupski, J. R. (2015). Exome sequencing identifies a homozygous C5orf42 variant in a Turkish kindred with oral-facial-digital syndrome type VI. *American Journal of Medical Genetics Part A*, *167*(9), 2132-2137.

Ramasamy, R., Bakırcıoğlu, M. E., Cengiz, C., Karaca, E., Scovell, J., Jhangiani, S. N., ... & Gibbs, R. A. (2015). Whole-exome sequencing identifies novel homozygous mutation in NPAS2 in family with nonobstructive azoospermia. *Fertility and sterility*, *104*(2), 286-291.

Pehlivan, D., **Akdemir, Z. C.**, Karaca, E., Bayram, Y., Jhangiani, S., Yildiz, E. P., ... & Lupski, J. R. (2015). Exome sequencing reveals homozygous TRIM2 mutation in a patient with early onset CMT and bilateral vocal cord paralysis. *Human genetics*, *134*(6), 671-673.

Hirsch, C. L**.*, Akdemir, Z. C**.*, Wang, L., Jayakumaran, G., Trcka, D., Weiss, A., ... & Xia, Z. (2015). Myc and SAGA rewire an alternative splicing network during early somatic cell reprogramming. *Genes & development*, *29*(8), 803-816. (*Co-first authors).

Chong, J. X., Buckingham, K. J., Jhangiani, S. N., Boehm, C., Sobreira, N., Smith, J. D., ... & **Akdemir, Z. H. C., …** (2015). The genetic basis of Mendelian phenotypes: discoveries, challenges, and opportunities. *The American Journal of Human Genetics*, *97*(2), 199-215.

Jiang, S., Minter, L. C., Stratton, S. A., Yang, P., Abbas, H. A., **Akdemir, Z. C.**, ... & Lozano, G. (2015). TRIM24 suppresses development of spontaneous hepatic lipid accumulation and hepatocellular carcinoma in mice. *Journal of hepatology*, *62*(2), 371-379.

Kurinna, S., Stratton, S. A., **Coban, Z.**, Schumacher, J. M., Grompe, M., Duncan, A. W., & Barton, M. C. (2013). p53 regulates a mitotic transcription program and determines ploidy in normal mouse liver. *Hepatology*, *57*(5), 2004-2013.

Coban, Z., & Barton, M. C. Integration and Distillation: Global Comparisons of Liver Regeneration and HCC.

Coban, Z., & Barton, M. C. (2012). Integrative genomics: liver regeneration and hepatocellular carcinoma. *Journal of cellular biochemistry*, *113*(7), 2179-2184.

Bodenreider, O**., Coban, Z. H.**, Doganay, M. C., Erdem, E., & Kosucu, H. (2008). A preliminary report on answering complex queries related to drug discovery using answer set programming. *ALPSWS 2008: Applications of Logic Programming to the (Semantic) Web and Web Services*, 85.

D. Abstracts Presented Based on Posters or Oral Presentations

American Society of Human Genetics Conference 2022

Unraveling the genomic basis of congenital heart disease from Gabriella Miller Kids First Pediatric Research Program: A Preliminary data.

American Society of Human Genetics Conference 2022

Escape from Nonsense-Mediated Decay: Annotation of Transcripts with Protein Truncating Variants.

American Society of Human Genetics Conference 2022

Comparison of chromosomal inversions in three different datasets.

European Society of Human Genetics Conference 2021

Elucidating the non-canonical rules of NMD in human using genomic approaches.

American Society of Human Genetics Virtual Conference 2021

Organizer of the invited session entitled "Nonsense-mediated decay: A double-edged sword in cancer and genetic diseases"

International Society of Computational Biology 2021

Organizer of the invited session entitled "Emerging gain-of-function mutations and their characterization by multi-omics network biology"

European Human Genetics Virtual Conference 2021
De novo mutation in ancestral generations evolves haplotypes contributing to disease
American Society of Human Genetics Houston, TX (2019)
Long-sized AOH regions reveal IBD haplotypes that drive Mendelian disease through mutational burden at a locus
Genomics of Rare Disease, Wellcome Genome Campus, Hinxton, UK (2018)
A Systematic **Analysis of *De Novo* Mutations in 837 trios with clinical suspicion of Mendelian phenotypes**
American Society of Human Genetics San Diego, CA (2018)
A Systematic Analysis of De Novo Mutations in 837 Trios with Clinical Suspicion of Mendelian Phenotypes
The NHGRI Genome Sequencing Program (GSP) In-Person Meeting, Bethesda, MD (2017)
When are (LOF) mutations not LOF mutations?
American Society of Human Genetics Orlando, FL (2017)
When are predicted loss-of-function (LOF) mutations not LOF mutations?

III. TEACHING INFORMATION

A. Educational Leadership Roles (title, dates, responsibilities)

N/A

B. Didactic Coursework

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas USA
Instructor
Duties have included leading lectures and preparing homework and exams
• Epidemiology I, Spring 2024

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas USA
Instructor
Duties have included leading lectures and preparing homework and exams
• Epidemiology I, Spring 2023

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas USA
Instructor
Duties have included leading lectures, weekly labs and grading homework and exams
• Genetic Epidemiology, Summer 2024.

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas USA
Instructor
Duties have included leading lectures, weekly labs and grading homework and exams
• Genetic Epidemiology, Summer 2023.

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas USA
Instructor
Duties have included leading lectures, weekly labs and grading homework and exams
• Genetic Epidemiology, Summer 2022.

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas USA
Instructor
Duties have included leading lectures and preparing homework and exams
• Epidemiology I, Spring 2022

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas USA
Co-instructor
Duties have included leading lectures, weekly labs, grading homework and exams
• Applied Genetic Methods in Public Health, Summer 2021.

The University of Texas Health Science Center at Houston, Graduate School of Biomedical Sciences, Houston, Texas USA
Teaching Assistant

Duties have included leading discussion classes and grading exams

• Molecular Biology of Eukaryotic Cells, Spring 2013.
Harvard University, Boston, Massachusetts USA
Teaching Assistant
• Analysis of Rates&Proportions, Fall 2009
Duties have included office hours, leading weekly labs and grading homework and exams
Sabanci University, Istanbul, Turkey
Teaching Assistant
Duties have included leading weekly labs
• Statistical Modeling, Spring 2007

Teaching Assistant
Duties have included leading weekly labs

• Introduction to Probability & Statistics, Fall 2006.
Peer-Teaching Assistant, Academic Support Program (ASP)
Duties have included helping students in their studies by organizing workshops, seminars, and social events and providing guidance to student and providing assistance for the Science of Nature course

C. Curriculum Development Work (include institution where work was done and whether work was disseminated beyond home institution)

1. N/A
2. N/A
3. N/A

D. Non-didactic Teaching (include institution where work was done)

1. N/A
2. N/A
3. N/A
4. N/A
5. N/A

E. Faculty Development or Continuing Medical Education (list contributions as a course developer, presenter, etc.)

N/A

IV. SERVICE CONTRIBUTIONS

A. Administrative Assignments and Committees

Faculty advisor to the students:
Renee Acquaye
(MPH,Graduated)
Shoaib Aziz (MPH,
Graduated)
Barbara Bell (MPH)
Sasha Borisewitz (MPH)
Christopher Burris (MPH)
Amelia Collins (MPH)
Harsh Chauhan (MPH)
Sagar Mukhida (MPH)
Iman Egab (PhD)
Jiaoyang Xu (PhD)

Thesis committee member to the students:
Jonathan Klonowski (UPittsburg, Ph.D. Thesis Committee,
Graduated)
Andy Rivera Munoz (BCM, Ph.D. Thesis Committee)
Zain Dardas (BCM, Ph.D. Thesis Committee, Graduated)
Angad Jolly (BCM, Ph.D. Thesis Committee, Graduated)
Moez Dawood (BCM, Ph.D. Thesis Committee)
Nikhita Gogate (BCM, Ph.D. Thesis Committee)
Rachel Friedman (UTHealth SPH, Master's Thesis Committee)
Tugce Bozkurt (Acibadem University, Ph.D. Thesis Committee)

Alanna Cecchi (GSBS, Ph.D. Thesis Committee)

B. National, Regional or Local Participation in Professional or Voluntary Organizations

Member, American Society of Human Genetics Faculty
Member, GSBS, MD Anderson
Early Career Reviewer, NIH GCAT study section Member,
Epidemiology Curriculum Committee
Member, The Genomics Research to Elucidate the Genetics of Rare Diseases (GREGoR) program Data Standards and Analysis Committee Member

C. Other Pertinent Information (not given above, including community service)

N/A