Asst. Prof. EMRAH KAYGUSUZ

Personal Information

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International Researcher IDs
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Education Information

Post Doctorate, Georg-August-Universitaet Göttingen, Germany 2018 - 2020
Doctorate, Universitaet zu Köln, Institute For Biochemistry, Medical Faculty And Cologne Center For Genomics, Germany 2014 - 2018
Postgraduate, Universität Zu Köln, Biyoloji Bilimi (Genetik), Germany 2011 - 2013

Foreign Languages

German, B2 Upper Intermediate
English, C1 Advanced

Dissertations

Doctorate, Role of CSNK2B encoding casein kinase II subunit beta in Filippi Syndrome, Universitaet zu Köln, Institute For Biochemistry, Medical Faculty And Cologne Center For Genomics, 2018
Postgraduate, Identification of RBBP8 mutation in Pakistani families affected with Jawad Syndrome, Universität Zu Köln, Biyoloji Bilimi (Genetik), 2013

Research Areas

Bioinformatics, Molecular Biology and Genetics, Genetic Disorders, Genomics, Animal Molecular Genetics

Academic Titles / Tasks

Assistant Professor, Bilecik Seyh Edebali University, Fen Fakültesi, Moleküler Biyoloji Ve Genetik, 2020 - Continues

Courses

Undergraduate
Biyoinformatiğe Giriş, Undergraduate, 2020 - 2021

Moleküler Biyolojide Uygulamalar-2, Undergraduate, 2020 - 2021
Moleküler Biyolojide Uygulamalar, Undergraduate, 2020 - 2021
Biyoinformatik-2, Undergraduate, 2020 - 2021
Moleküler Terminoloji, Undergraduate, 2020 - 2021
Kromozomal Hastalıklar, Undergraduate, 2020 - 2021
Moleküler Teknikler, Undergraduate, 2020 - 2021
Model Organizmalar, Undergraduate, 2020 - 2021
Moleküler Teknoloji, Undergraduate, 2020 - 2021

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Loss-of-function variants in DNM1 cause a specific form of developmental and epileptic encephalopathy only in biallelic state.**
Yigit G., Sheffer R., Daana M., Li Y., Kaygusuz E., Mor-Shakad H., Altmüller J., Nürnberg P., Douiev L., Kaulfuss S., et al.
Journal of medical genetics, vol.59, no.6, pp.549-553, 2022 (SCI-Expanded)

II. **A 24-generation-old founder mutation impairs splicing of RBBP8 in Pakistani families affected with Jawad syndrome.**
Kaygusuz E., Khayyat A. I. A., Abdullah U., Budde B. S., Asif M., Ahmed I., Makhdoom E. U. H., Sur-Erdem I., Baig J. M., Khan M. M. A., et al.
Clinical genetics, vol.100, no.4, pp.486-488, 2021 (SCI-Expanded)

III. **Modifier Genes in Microcephaly: A Report on WDR62, CEP63, RAD50 and PCNT Variants Exacerbating Disease Caused by Biallelic Mutations of ASPM and CENPJ.**
Makhdoom E. U. H., Waseem S. S., Iqbal M., Abdullah U., Hussain G., Asif M., Budde B., Höhne W., Tinschert S., Saadi S. M., et al.
Genes, vol.12, no.5, 2021 (SCI-Expanded)

IV. **An update of pathogenic variants in ASPM, WDR62, CDK5RAP2, STIL, CENPJ, and CEP135 underlying autosomal recessive primary microcephaly in 32 consanguineous families from Pakistan.**
Rasool S., Baig J. M., Moawia A., Ahmad I., Iqbal M., Waseem S. S., Asif M., Abdullah U., Makhdoom E. U. H., Kaygusuz E., et al.
Molecular genetics & genomic medicine, vol.8, no.9, 2020 (SCI-Expanded)

V. **Mutations in CKAP2L, the human homolog of the mouse radmis gene, cause filippi syndrome**
Hussain M. S., Battaglia A., Szczepanski S., Kaygusuz E., Toliat M. R., Sakakibara S., Altmüller J., Thiele H., Nürnberg G., Moosa S., et al.
American Journal of Human Genetics, vol.95, no.5, pp.622-632, 2014 (SCI-Expanded)

Articles Published in Other Journals

I. **De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway**
Asif M., Kaygusuz E., Shinawi M., Nickelsen A., Hsieh T., Wagle P., Budde B. S., Hochscherf J., Abdullah U., Höning S., et al.
Human Genetics and Genomics Advances, vol.3, no.3, 2022 (Scopus)

Books

I. **Describing genetic basis and disease pathogenesis of Filippi syndrome**
KAYGUSUZ E.
Cinius, İstanbul, 2022

II. **Identification of RBBP8 mutation in Pakistani families affected with Jawad Syndrome**
KAYGUSUZ E.
Cinius Yayınları, 2021

Papers Published in Refereed Scientific Meetings

I. **Role of encoding casein kinase II subunit beta in a new intellectual disability-craniodigital syndrome (Davetli Konuşmacı)**
KAYGUSUZ E.
Workshop on Rare Diseases, İstanbul, Turkey, 03 December 2022

II. **De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway (Davetli Konuşmacı)**
KAYGUSUZ E.
8th International Congress of the Molecular Biology Association of Turkey, İstanbul, Turkey, 9 - 12 June 2022

III. **Identification of RBBP8 mutation in Pakistani families affected with Jawad Syndrome**
KAYGUSUZ E.
5th International Congress of the Molecular Biology Association of Turkey, İstanbul, Turkey, 08 September 2017

IV. **De novo pathogenic variants in CSNK2B cause a new intellectual disability-craniodigital syndrome distinguished from Poirier-Bienvenu neurodevelopmental syndrome**
KAYGUSUZ E., Asif M.
European Society of Human Genetics (ESHG) Conference, Glasgow, England, 06 June 2020

V. **De novo mutation of CSNK2B encoding beta subunit of casein kinase 2 causes Filippi syndrome**
KAYGUSUZ E.
52nd European Society of Human Genetics (ESHG) Conference, Gothenburg, Sweden, 15 June 2019

VI. **Pathogenic consequences of impaired Wnt signaling and DNA damage response delineates Filippi syndrome**
KAYGUSUZ E., Asif M., Brancati F., Nickelsen A., Nienberg C., Jose J., Niefind K., Noegel A., Hussain M. S., Nürnberg P.
35th Ernst Klenk Symposium in Molecular Medicine, Cologne, Germany, 15 - 17 September 2019

VII. **Identification of a founder mutation in Pakistani families affected with Jawad syndrome**
KAYGUSUZ E., Nürnberg P., Noegel A., Hussain M. S.
Arbeitsgemeinschaft Für Gen-Diagnostik e.V., Berlin, Germany, 6 - 08 October 2017

VIII. **Identification of Founder Mutation in Pakistani Families Affected With Jawad Syndrome**
KAYGUSUZ E., Hussain M. S., Nürnberg P., Noegel A., Altmüller J., Budde B., Asif M., Baig S. M., Tariq M., Ahmed I.
5th International Congress of the Molecular Biology Association of Turkey, İstanbul, Turkey, 8 - 10 September 2017

IX. **Filippi syndrome is aheterogenic disorder with a high proportion of CKAP2L mutations**
KAYGUSUZ E., Hussain M. S., Szczepanski S., Nürnberg P., Noegel A.
31st Ernst Klenk Symposium in Molecular Medicine, Cologne, Germany, 27 - 29 September 2015

X. **Mutations in CKAP2L, the human homolog of the mouse Radmis gene, cause Filippi syndrome**
KAYGUSUZ E., Hussain M. S., Altmüller J., Wollnik B., Szczepanski S., Noegel A., Thiele H., Newman W., Sakakibara S., Gillessen-Kaesbach G., et al.
International Meeting of the German Society for Cell Biology, Cologne, Germany, 24 - 27 March 2015

Supported Projects

Aptullahoğlu E., Kaygusuz E., TUBITAK Project, Investigating the Combined Targeting of MDM2 and STAT3 as a New Therapeutic Approach in Acute Lymphoblastic Leukemia, 2021 - 2024

Kaygusuz E., Hussain M. S., Khayyat A. I., Iqbal M., Asif M., Noegel A., Nürnberg P., Project Supported by Public Organizations in Other Countries, Role of Wnt signaling in the etiology of Filippi syndrome and ectrodactyly ectodermal

dysplasia, 2017 - 2019

Peer Reviews in Scientific Publications

CLINICAL GENETICS, SCI Journal, May 2024
Bilecik Şeyh Edebali Üniversitesi Fen Bilimleri Dergisi, National Scientific Refreed Journal, June 2023

Scientific Project Refereeing

Project Supported by Higher Education Institutions, BAP MSc, May 2024
Project Supported by Higher Education Institutions, BAP Research Project, December 2021

Metrics

Publication: 18
Citation (Scopus): 94
H-Index (Scopus): 5

Congress and Symposium Activities

Workshop on Rare Diseases, Invited Speaker, İstanbul, Turkey, 2022
8th International Congress of the Molecular Biology Association of Turkey, Invited Speaker, İstanbul, Turkey, 2022