Prof. AHMET OKAY ÇAĞLAYAN

Personal Information

Email: ahmetokay.caglayan@deu.edu.tr
Web: https://avesis.deu.edu.tr/ahmetokay.caglayan

International Researcher IDs
ORCID: 0000-0002-2332-322X
Publons / Web Of Science ResearcherID: D-1066-2012
ScopusID: 16641940600
Yoksis Researcher ID: 218863

Biography

I am a Professor of Medical Genetics and clinical geneticist with specific interest in neurodevelopmental disorders. I have extensive experience in next-generation and high-throughput molecular genetics, with special focus on the use of homozygosity mapping, linkage analysis and whole-exome sequencing in recessive forms of structural and functional abnormalities of the brain. I have successfully detected rare and somatic genetic variant identification in diverse range of diseases using high-throughput and next-generation genomics approaches at Yale. In the last ten years, I have published papers in Cell, Science, Nature, Nature Genetics and Neuron including one of the first successful applications of exome sequencing in the identification of genetic basis of recessive forms of structural and functional abnormalities of the brain.

I have served as a member of editorial boards, a reviewer in top-tier journals and conferences, and was invited to serve as a program committee member for international workshops and conferences as well. I established Medical Genetics Department and giving lectures to under and post graduate students, assisting in seminar groups or laboratories, and grading. Teaching activities are drawn from a diverse menu of lecture, laboratory, and seminar courses given at the undergraduate, graduate, and medical school level.

Education Information

Expertise In Medicine, Erciyes University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Turkey 2003 - 2007
Undergraduate, Eskisehir Osmangazi University, Tıp Fakültesi, Tıp Pr., Turkey 1997 - 2001

Certificates, Courses and Trainings

Vocational Training, Publons Academy Peer Reviewer, Publons Academy, 2020

Research Areas

Medicine, Childrens Mental Health and Disorders, Child Health and Diseases, Neurology, Medical Genetics, Obstetrics and Gynecology, Bioinformatics, Molecular Biology and Genetics, Neurosystems

Academic Titles / Tasks

Professor, Dokuz Eylul University, Sağlık Bilimleri Enstitüsü, Moleküler Tıp Anabilim Dalı (Disiplinlerarası), 2019 - Continues
Professor, Demiroğlu Bilim Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2018 - 2019
Associate Professor, Istanbul Bilim University, School Of Medıcıne, Department Of Internal Medıcıne, 2015 - 2017

Academic and Administrative Experience

Deputy Chief Physician, Dokuz Eylul University, Uygulama Ve Araştırma Hastanesi, Dahili Tıp Bilimleri, 2023 - Continues
Bölüm Akademik Teşvik Değerlendirme Komisyonu Üyesi, Dokuz Eylul University, Sağlık Bilimleri Enstitüsü, 2022 - Continues
Member of the Commission, Dokuz Eylul University, Sağlık Bilimleri Enstitüsü, 2022 - Continues
Ethics Committee Member, Dokuz Eylul University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2021 - Continues
Director of the Center, Dokuz Eylul University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2021 - Continues
Merkez Laboratuvar Başkan Yardımcısı, Dokuz Eylul University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2020 - Continues
Anabilim Dalı Akademik Kurul Üyesi, Dokuz Eylul University, Sağlık Bilimleri Enstitüsü, Sağlık Bilimleri Enstitüsü, 2019 - Continues
Anabilim Dalı Akademik Kurul Üyesi, Dokuz Eylul University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2019 - Continues
Head of Department, Dokuz Eylul University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2023 - 2023
Genetik Testler Muayene Komisyonu Asil Üyesi, Dokuz Eylul University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2023 - 2023
Assistant Director of the Institute, Dokuz Eylul University, Sağlık Bilimleri Enstitüsü, 2022 - 2023
Enstitü Yönetim Kurulu Üyesi, Dokuz Eylul University, Sağlık Bilimleri Enstitüsü, 2022 - 2023
Istanbul Bilim University, 2015 - 2019
Istanbul Bilim University, 2015 - 2017

Courses

Tıbbi Genetik, Undergraduate, 2022 - 2023, 2021 - 2022, 2016 - 2017
Tıbbi Biyoloji ve Genetik, Postgraduate, 2018 - 2019, 2017 - 2018, 2016 - 2017, 2015 - 2016
Tıbbi Biyoloji ve Genetik, Undergraduate, 2015 - 2016

Jury Memberships

Expertise In Medicine, Expertise In Medicine, Ege Üniversitesi, June, 2024
Doctorate, Doctorate, Ege Üniversitesi, January, 2024
Doctorate, Doctorate, İzmir Biyotıp ve Genom Merkezi, November, 2023
Doctorate, Doctorate, Ege Üniversitesi, October, 2023
Doctoral Examination, Doctoral Examination, Ege Üniversitesi, October, 2023
Expertise In Medicine, Expertise In Medicine, Ege Üniversitesi, September, 2023
Expertise In Medicine, Expertise In Medicine, Dokuz Eylül Üniversitesi, August, 2023
Appointment to Academic Staff - Associate Professorship, Appointment to Academic Staff - Associate Professorship, İstanbul Medipol Üniversitesi, July, 2023
Appointment to Academic Staff - Associate Professorship, Appointment to Academic Staff - Associate Professorship, İstanbul Medipol Üniversitesi, July, 2023
Appointment to Academic Staff - Associate Professorship, Appointment to Academic Staff - Associate Professorship, Balıkesir Üniversitesi, June, 2023
Appointment to Academic Staff-Assistant Professorship, Appointment to Academic Staff-Assistant Professorship,

Acıbadem Mehmet Ali Aydınlar Üniversitesi, June, 2023
Expertise In Medicine, Expertise In Medicine, Dokuz Eylül Üniversitesi, June, 2023
Associate Professor Exam, Associate Professor Exam, Sağlık Bilimleri Üniversitesi, May, 2023
PhD Thesis Monitoring Committee Member, PhD Thesis Monitoring Committee Member, Balıkesir Üniversitesi, March, 2023
Appointment to Academic Staff-Professorship, Appointment to Academic Staff-Professorship, İstanbul Üniversitesi, February, 2023
Appointment to Academic Staff - Associate Professorship, Appointment to Academic Staff - Associate Professorship, Sağlık Bilimleri Üniversitesi, February, 2023
Appointment to Academic Staff - Associate Professorship, Appointment to Academic Staff - Associate Professorship, Afyonkarahisar Sağlık Bilimleri Üniversitesi, February, 2023
Competition, TEKNOFEST Sağlık ve İlk Yardım Kategorisi-2023, Teknofest, January, 2023
Doctoral Examination, Doctoral Examination, Dokuz Eylül Üniversitesi, January, 2023
Appointment to Academic Staff-Assistant Professorship, Appointment to Academic Staff-Assistant Professorship, Bezmiâlem Vakıf Üniversitesi, December, 2022
Associate Professor Exam, Associate Professor Exam, Trakya Üniversitesi, December, 2022
Competition, TEKNOFEST Sağlıkta Yapay Zekâ Yarışmaları-2023, Teknofest, December, 2022
Associate Professor Exam, Associate Professor Exam, Atatürk Üniversitesi, October, 2022
Doctorate, Doctorate, Ege Üniversitesi, August, 2022
Appointment to Academic Staff-Assistant Professorship, Appointment to Academic Staff-Assistant Professorship, Van Yüzüncü Yıl Üniversitesi, August, 2022
Post Graduate, Post Graduate, Dokuz Eylül Üniversitesi, July, 2022
Competition, TEKNOFEST HAVACILIK, UZAY VE TEKNOLOJİ FESTİVALİ SAĞLIKTA YAPAY ZEKÂ YARIŞMASI, Dokuz Eylül Üniversitesi, June, 2022
Associate Professor Exam, Associate Professor Exam, Afyon Kocatepe Üniversitesi, June, 2022
Doctoral Examination, Doctoral Examination, Dokuz Eylül Üniversitesi, June, 2022
Competition, TEKNOFEST HAVACILIK, UZAY VE TEKNOLOJİ FESTİVALİ, Dokuz Eylul University, April, 2022
Associate Professor Exam, Associate Professor Exam, Dokuz Eylül Üniversitesi, December, 2021
Doctoral Examination, Doctoral Examination, Ege Üniversitesi, October, 2021
PhD Thesis Monitoring Committee Member, PhD Thesis Monitoring Committee Member, Dokuz Eylül Üniversitesi, September, 2021
Associate Professor Exam, Associate Professor Exam, Dokuz Eylül Üniversitesi, August, 2021
PhD Thesis Monitoring Committee Member, PhD Thesis Monitoring Committee Member, Ege Üniversitesi, August, 2021
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, Ege Üniversitesi, July, 2021
Appointment to Academic Staff-Professorship, Appointment Academic Staff, Lokman Hekim Üniversitesi, July, 2021
Doctoral Examination, Doctoral Examination, İstanbul Üniversitesi, May, 2021
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, Balıkesir Üniversitesi, February, 2021
Doctorate, Doctorate, İstanbul Üniversitesi, December, 2020
Doctoral Examination, Doctoral Examination, Ege Üniversitesi, October, 2020
Associate Professor Exam, Associate Professor Exam, Dokuz Eylül Üniversitesi, October, 2020
Doctoral Examination, Doctoral Examination, Ege Üniversitesi, October, 2020
Doctorate, Doctorate, İstanbul Üniversitesi, July, 2020
Associate Professor Exam, Associate Professor Exam, Dokuz Eylül Üniversitesi, March, 2020
Associate Professor Exam, Associate Professor Exam, Dokuz Eylül Üniversitesi, August, 2019
Associate Professor Exam, Associate Professor Exam, Dokuz Eylül Üniversitesi, September, 2018
Associate Professor Exam, Associate Professor Exam, Dokuz Eylül Üniversitesi, June, 2018

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Investigation of genotype-phenotype and familial features of Turkish dystrophinopathy patients.**
Ozkalayci H., Bora E., Cankaya T., Kocabey M., Zubari N. C., Yis U., Giray Bozkaya O., Turan S., Pekcanlar Akay A., Caglayan A. O., et al.
Neurogenetics, 2024 (SCI-Expanded)

II. **Phenotypic comparison of a novel variant (p.P164R) and A founder mutation (c.748+1G>A) in Warburg Micro syndrome**
Cinleti T., Sarıkaya Uzan G., Bürçe B., Küçümen Y., Yalçın H. Y., Gürsoy S., Yiş U., Çağlayan A. O., Giray Bozkaya Ö.
NEUROLOGY ASIA, vol.28, no.4, pp.1085-1094, 2023 (SCI-Expanded)

III. **Genetic, serological and clinical evaluation of childhood myasthenia syndromes- single center subgroup analysis experience in Turkey**
Özsoy Ö., Cinleti T., Günay Ç., Sarıkaya Uzan G., Giray Bozkaya Ö., Çağlayan A. O., Hız Kurul S., Yiş U.
Acta Neurologica Belgica, vol.123, no.6, pp.2325-2335, 2023 (SCI-Expanded)

IV. **Genetic evaluation of 50 Turkish patients with neurofibromatosis type 1: 2 years experience of a single center**
Kocabey M., Özkalaycı H., Çankaya T., Yılmaz Uzman C., Çağlayan A. O., Ülgenalp A., Erçal M. D.
International Journal of Developmental Neuroscience, vol.83, no.5, pp.456-465, 2023 (SCI-Expanded)

V. **MIRAGE syndrome in a 10-year-old girl with a novel Lys1024Glu missense variant in SAMD9**
Cinleti T., Gülen A., Sönmez B., Gürsoy S., Boyacioğlu Ö. K., Asilsoy S., Ulgenalp A., Bozkaya Ö., Çağlayan A. O.
CLINICAL DYSMORPHOLOGY, no.3, pp.133-138, 2023 (SCI-Expanded)

VI. **Clinical Heterogeneity in Patients with Long QT Syndrome and Segregation of Single Nucleotide Variants and Clinical Symptoms in 17 Affected Families**
Bora E., Bulut A. Y., Çankaya T., Cinleti T., Genç H. Z., Özcan E. E., Özpelit E., Ülgenalp A., Çağlayan A. O.
MOLECULAR SYNDROMOLOGY, 2023 (SCI-Expanded)

VII. **Investigation of different genomic variants in familial Mediterranean fever cases with monoallelic MEFV mutation.**
Kocabey M., Cankaya T., Bayram M. T., Ulgenalp A., Caglayan A. O., Giray Bozkaya Ö.
Clinical and experimental rheumatology, 2023 (SCI-Expanded)

VIII. **An investigation of the etiology and follow-up findings in 35 children with overgrowth syndromes, including biallelic SUZ12 variant.**
Yüksel Ülker A., Uludağ Alkaya D., Çağlayan A. O., Usluer E., Aykut A., Aslanger A., Vural M., Tüysüz B.
American journal of medical genetics. Part A, 2023 (SCI-Expanded)

IX. **Unexpected finding in kidney biopsy of a child with nephrotic proteinuria: Questions**
Bayram M., Yildiz G., Cağlayan A. O., Ulgenalp A., Unlu S. M., Soylu A., Kavukcu S.
PEDIATRIC NEPHROLOGY, no.1, pp.113-114, 2023 (SCI-Expanded)

X. **Unexpected finding in kidney biopsy of a child with nephrotic proteinuria: Answers**
Bayram M., Yildiz G., Cağlayan A. O., Ulgenalp A., Unlu S. M., Soylu A., Kavukcu S.
PEDIATRIC NEPHROLOGY, no.1, pp.115-117, 2023 (SCI-Expanded)

XI. **Hemoglobin A(1C) can differentiate subjects with GCK mutations among patients suspected to have MODY.**
Yılmaz Uzman C., Erbaş İ. M., Giray Bozkaya Ö., Paketçi A., Çağlayan A. O., Abacı A., Kulalı M. A., Böber E., Kekilli A., Çinleti T., et al.
Journal of pediatric endocrinology & metabolism : JPEM, vol.35, no.12, pp.1528-1536, 2022 (SCI-Expanded)

XII. **Biallelic BICD2 variant is a novel candidate for Cohen-like syndrome.**
Caglayan A. O., Tuysuz B., Gül E., Alkaya D. U., Yalcinkaya C., Gleeson J. G., Bilguvar K., Gunel M.
Journal of human genetics, vol.67, no.9, pp.553-556, 2022 (SCI-Expanded)

XIII. **Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes.**
Laugwitz L., Seibt A., Herebian D., Peralta S., Kienzle I., Buchert R., Falb R., Gauck D., Müller A., Grimmel M., et al.
Journal of medical genetics, vol.59, no.9, pp.878-887, 2022 (SCI-Expanded)

XIV. **Analysis of genotype-phenotype correlation in Walker-Warburg syndrome with a novel CRPPA mutation in different clinical manifestations**

Bayram N., Bayram A. K., Per H., Gümüş H., Ozsaygili C., Dogan M. S., Çağlayan A. O.
EUROPEAN JOURNAL OF OPHTHALMOLOGY, vol.32, no.5, 2022 (SCI-Expanded)

XV. **Congenital Myasthenic Syndromes in Turkey: Clinical and Molecular Characterization of 16 Cases With Three Novel Mutations.**
Öztürk S., Güleç A., Erdoğan M., Demir M., Canpolat M., Gümüş H., Çağlayan A. O., Dündar M., Per H.
Pediatric neurology, vol.136, pp.43-49, 2022 (SCI-Expanded)

XVI. **Clinical and molecular evaluation of MEFV gene variants in the Turkish population: a study by the National Genetics Consortium**
Dundar M., Fahrioglu U., Yildiz S. H., Bakir-Gungor B., Temel S. G., Akin H., Artan S., Cora T., Sahin F. I., Dursun A., et al.
FUNCTIONAL & INTEGRATIVE GENOMICS, vol.22, no.3, pp.291-315, 2022 (SCI-Expanded)

XVII. **Evaluation of hereditary/familial breast cancer patients with multigene targeted next generation sequencing panel and MLPA analysis in Turkey**
Bora E., Caglayan A. O., Koc A., Cankaya T., Ozkalayci H., Kocabey M., Kemer D., Aksoy S. Ö., Alicikus Z. A., Başara Akın I., et al.
Cancer Genetics, vol.262-263, pp.118-133, 2022 (SCI-Expanded)

XVIII. **Importance of multigene panel test in patients with consanguineous marriage and family history of breast cancer**
Ozmen V., Çağlayan A. O., Yararbas K., Ordu C., Aktepe F., Ozmen T., Ilgun A. S., Soybir G., Alco G., Tsaousis G. N., et al.
ONCOLOGY LETTERS, vol.23, no.4, 2022 (SCI-Expanded)

XIX. **Further delineation of familial polycystic ovary syndrome (PCOS) via whole-exome sequencing: PCOS-related rare FBN3 and FN1 gene variants are identified.**
Karakaya C., Çil A. P., Bilguvar K., Çakir T., Karalok M. H., Karabacak R. O., Caglayan A. O.
The journal of obstetrics and gynaecology research, 2022 (SCI-Expanded)

XX. **Clinical and genetic studies of thiamine metabolism dysfunction syndrome-4: case series and review of the literature.**
Samur B. M., Gümüş G., Canpolat M., Gümüş H., Per H., Cağlayan A. O.
Clinical dysmorphology, 2022 (SCI-Expanded)

XXI. **Familial clustering of nasopharyngeal carcinoma in the family of an adolescent with nasopharyngeal carcinoma**
Kara B., Ertan K., Düzova M., ÇAĞLAYAN A. O., Köksal Y.
Turkish Journal of Pediatrics, vol.64, no.6, pp.1130-1135, 2022 (SCI-Expanded)

XXII. **Cerebral developmental venous anomalies in children with mismatch repair deficiency.**
Kara B., Paksoy Y., Çağlayan A. O., Seher N., Akbaş H., Köksal Y.
The Turkish journal of pediatrics, vol.64, no.6, pp.1106-1116, 2022 (SCI-Expanded)

XXIII. **Clinical Features, Treatment and Outcome of Childhood Glial Tumors.**
Kara B., Ertan K., Karabagli P., Karabagli H., Yavas G., Caglayan A. O., Koksal Y.
Turkish neurosurgery, vol.32, pp.135-142, 2022 (SCI-Expanded)

XXIV. **A novel homozygous frameshift mutation in the TUSC3 gene identified in siblings with intellectual disability.**
Özmansur E. N., Pedük Y., Gümüş H., Çağlayan A. O., Per H.
Clinical dysmorphology, vol.31, pp.36-38, 2022 (SCI-Expanded)

XXV. **Peripheral Expression of MACROD2 Gene Is Reduced Among a Sample of Turkish Children with Autism Spectrum Disorder**
Alnak A., Ozucer I. K., ÇAĞLAYAN A. O., COŞKUN M.
PSYCHIATRY AND CLINICAL PSYCHOPHARMACOLOGY, vol.31, no.3, pp.261-269, 2021 (SCI-Expanded)

XXVI. **Biallelic ZNF335 mutations cause basal ganglia abnormality with progressive cerebral/cerebellar atrophy.**
ÇAĞLAYAN A. O., Yaghouti K., Kockaya T., Kemer D., ÇANKAYA T., Ameziane N., ÇOĞULU M. Ö., ÇOKER M., YALÇINKAYA C.
Journal of neurogenetics, vol.35, no.1, pp.23-28, 2021 (SCI-Expanded)

XXVII. **METAP1mutation is a novel candidate for autosomal recessive intellectual disability**
ÇAĞLAYAN A. O., Aktar F., Bilguvar K., Baranoski J. F., Akgumus G. T., Harmanci A. S., Erson-Omay E. Z., Yasuno K., ÇAKSEN H., Gunel M.
JOURNAL OF HUMAN GENETICS, vol.66, no.2, pp.215-218, 2021 (SCI-Expanded)

XXVIII. **Nasopharyngeal carcinoma in a child with Kartagener's syndrome**
Kara B., Seher N., Karanis M. I. E., KOPLAY M., ARTAÇ H., Koc M., ÇAĞLAYAN A. O., KÖKSAL Y.
TURKISH JOURNAL OF PEDIATRICS, vol.63, no.1, pp.155-160, 2021 (SCI-Expanded)

XXIX. **Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants**
Herkert J. C., Verhagen J. M. A., Yotti R., Haghighi A., Phelan D. G., James P. A., Brown N. J., Stutterd C., Macciocca I., Leong K., et al.
AMERICAN HEART JOURNAL, vol.225, pp.108-119, 2020 (SCI-Expanded)

XXX. **COQ4 Mutation Leads to Childhood-Onset Ataxia Improved by CoQ10 Administration**
ÇAĞLAYAN A. O., GÜMÜŞ H., Sandford E., Kubisiak T. L., Ma Q., Ozel A. B., PER H., Li J. Z., Shakkottai V. G., Burmeister M.
CEREBELLUM, vol.18, no.3, pp.665-669, 2019 (SCI-Expanded)

XXXI. **MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctive cerebellar, ocular, craniofacial and genital features (COFG syndrome)**
Rad A., Altunoglu U., Miller R., Maroofian R., James K. N., ÇAĞLAYAN A. O., Najafi M., Stanley V., Boustany R., YEŞİL SAYIN G., et al.
JOURNAL OF MEDICAL GENETICS, vol.56, no.5, pp.332-339, 2019 (SCI-Expanded)

XXXII. **Loss of Protocadherin-12 Leads to Diencephalic-Mesencephalic Junction Dysplasia Syndrome**
Guemez-Gamboa A., ÇAĞLAYAN A. O., Stanley V., Gregor A., Zaki M. S., Saleem S. N., Musaev D., McEvoy-Venneri J., Belandres D., Akizu N., et al.
ANNALS OF NEUROLOGY, vol.84, no.5, pp.638-647, 2018 (SCI-Expanded)

XXXIII. **Biallelic loss of human CTNNA2, encoding alpha N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration**
Schaffer A. E., Breuss M. W., ÇAĞLAYAN A. O., Al-Sanaa N., Al-Abdulwahed H. Y., Kaymakcalan H., Yilmaz C., Zaki M. S., Rosti R. O., Copeland B., et al.
NATURE GENETICS, vol.50, no.8, pp.1093-1107, 2018 (SCI-Expanded)

XXXIV. **Homozygous CAPN1 mutations causing a spastic-ataxia phenotype in 2 families**
Kocoglu C., Gundogdu A., Kocaman G., KAHRAMAN KOYTAK P., ULUÇ K., Kiziltan G., ÇAĞLAYAN A. O., Bilguv K., Vural A., Basak A. N.
NEUROLOGY-GENETICS, vol.4, no.1, 2018 (SCI-Expanded)

XXXV. **Biallelic mutations in the 3 ' exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing**
Lardelli R. M., Schaffer A. E., Eggens V. R. C., Zaki M. S., Grainger S., Sathe S., Van Nostrand E. L., Schlachetzki Z., Rosti B., Akizu N., et al.
NATURE GENETICS, vol.49, no.3, pp.457-464, 2017 (SCI-Expanded)

XXXVI. **Disruptions in asymmetric centrosome inheritance and WDR62-Aurora kinase B interactions in primary microcephaly**
Sgourdou P., Mishra-Gorur K., Saotome I., Henagariu O., Tuysuz B., Campos C., Ishigame K., Giannikou K., Quon J. L., Sestan N., et al.
SCIENTIFIC REPORTS, vol.7, 2017 (SCI-Expanded)

XXXVII. **Impaired Amino Acid Transport at the Blood Brain Barrier Is a Cause of Autism Spectrum Disorder**
Tarlungeanu D. C., Deliu E., Dotter C. P., Kara M., Janiesch P. C., Scalise M., Galluccio M., Tesulov M., Morelli E., Sonmez F. M., et al.
CELL, vol.167, no.6, pp.1481-1512, 2016 (SCI-Expanded)

XXXVIII. **Biallelic Mutations in TMTC3, Encoding a Transmembrane and TPR-Containing Protein, Lead to Cobblestone Lissencephaly**
Jerber J., Zaki M. S., Al-Aama J. Y., Rosti R. O., Ben-Omran T., Dikoglu E., Silhavy J. L., ÇAĞLAR C., Musaev D., Albrecht

B., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.99, no.5, pp.1181-1189, 2016 (SCI-Expanded)

XXXIX. **Mutations in MBOAT7, Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features**
Johansen A., Rosti R. O., Musaev D., Sticca E., Harripaul R., Zaki M., ÇAĞLAYAN A. O., Azam M., Sultan T., Froukh T., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.99, no.4, pp.912-916, 2016 (SCI-Expanded)

XL. **Biallelic Mutations in Citron Kinase Link Mitotic Cytokinesis to Human Primary Microcephaly**
Li H., Bielas S. L., Zaki M. S., Ismail S., Farfara D., Um K., Rosti R. O., Scott E. C., Tu S., Chi N. C., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.99, no.2, pp.501-510, 2016 (SCI-Expanded)

XLI. **A patient with a novel homozygous missense mutation in FTO and concomitant nonsense mutation in CETP**
ÇAĞLAYAN A. O., Tuysuz B., Coskun S., Quon J., Harmanci A. S., Baranoski J. F., Baran B., Erson-Omay E. Z., Henegariu O., Mane S. M., et al.
JOURNAL OF HUMAN GENETICS, vol.61, no.5, pp.395-403, 2016 (SCI-Expanded)

XLII. **Genome-Wide Association and Exome Sequencing Study of Language Disorder in an Isolated Population**
Kornilov S. A., Rakhlin N., Koposov R., Lee M., Yrigollen C., ÇAĞLAYAN A. O., Magnuson J. S., Mane S., Chang J. T., Grigorenko E. L.
PEDIATRICS, vol.137, no.4, 2016 (SCI-Expanded)

XLIII. **Clinical, Electrodiagnostic, and Genetic Features of Tangier Disease in an Adolescent Girl with Presentation of Peripheral Neuropathy**
PER H., CANPOLAT M., Bayram A. K., Ulgen E., Baran B., KARDAŞ F., GÜMÜŞ H., Kumandas S., Bilguvar K., ÇAĞLAYAN A. O.
NEUROPEDIATRICS, vol.46, no.6, pp.420-423, 2015 (SCI-Expanded)

XLIV. **A rare case of congenital fibrosis of extraocular muscle type IA due to KIF2IA mutation with Marcus Gunn jaw-winking phenomenon**
Bayram A. K., PER H., Quon J., CANPOLAT M., Uelgen E., Dogan H., GÜMÜŞ H., Kumandas S., Bayram N., Bilguvar K., et al.
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, vol.19, no.6, pp.743-746, 2015 (SCI-Expanded)

XLV. **The Effects of Ketogenic Diet on Seizures, Cognitive Functions, and Other Neurological Disorders in Classical Phenotype of Glucose Transporter 1 Deficiency Syndrome**
GÜMÜŞ H., Bayram A. K., KARDAŞ F., CANPOLAT M., ÇAĞLAYAN A. O., Kumandas S., KENDİRCİ M., PER H.
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Metrics

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Congress and Symposium Activities

Galen Days 2024, Invited Speaker, İzmir, Turkey, 2024
VII. Nöromusküler Hastalıklar Kongresi, Invited Speaker, Nevşehir, Turkey, 2024
Türkiye Genom Çalıştayı, Working Group, Ankara, Turkey, 2024
59. Ulusal Nöroloji Kongresi, Invited Speaker, Antalya, Turkey, 2023
Geleceğin Sağlık Teknolojileri Genomiks Kongresi, Working Group, İstanbul, Turkey, 2023
8. Erciyes Tıp Tıbbi Genetik Kongresi, Session Moderator, Kayseri, Turkey, 2023
ULUSAL NEONATOLOJİ KONGRESİ, Invited Speaker, Antalya, Turkey, 2023
Dokuz Eylül Bilimsel Araştırmalar Topluluğu Nadir Hastalıklar Sempozyumu, Invited Speaker, İzmir, Turkey, 2023
BİYOİNFORMATİK FORUMU - 2022, Working Group, Kocaeli, Turkey, 2022

15. Uluslararası Katılımlı Ulusal Tıbbi Genetik Kongresi, Session Moderator, Muğla, Turkey, 2022
15. Uluslararası Katılımlı Ulusal Tıbbi Genetik Kongresi, Attendee, Muğla, Turkey, 2022
TuPA Uluslarası Proteomik Kongresi, Session Moderator, İzmir, Turkey, 2022
TuPA Uluslarası Proteomik Kongresi, Invited Speaker, İzmir, Turkey, 2022
TuPA Uluslarası Proteomik Kongres, Attendee, İzmir, Turkey, 2022
Genç Meraklılar için Nörogenetik Semineri, Invited Speaker, İstanbul, Turkey, 2022
31. Ulusal Çocuk ve Ergen Ruh Sağlığı ve Hastalıkları Kongresi, Invited Speaker, İzmir, Turkey, 2022