# Assoc. Prof. AYBERK TÜRKYILMAZ

## Personal Information

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**International Researcher IDs**
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## Education Information

Expertise In Medicine, Marmara University, School Of Medicine, Internal Medical Sciences, Turkey 2014 - 2019
Undergraduate, Gazi University, Tıp Fakültesi, Turkey 2005 - 2012

## Dissertations

Expertise In Medicine, Ankilozan spondilit'te mikroRNA ekspresyonu ve etklili epigenetik mekanizmaların incelenmesi, Marmara University, School Of Medicine, Internal Medical Sciences, 2018

## Research Areas

Medicine, Internal Medicine Sciences, Medical Genetics

## Academic Titles / Tasks

Associate Professor, Karadeniz Technical University, Tıp Fakültesi, Dahili Tıp, 2023 - Continues
Assistant Professor, Karadeniz Technical University, Tıp Fakültesi, Dahili Tıp, 2020 - 2023
Expert, University Of Health Sciences, Erzurum Regıon Health Research Center, Department Of Internal Medıcıne, 2019 - 2020
Research Assistant, Marmara University, School Of Medicine, Internal Medical Sciences, 2014 - 2019

## Academic and Administrative Experience

Head of Department, Karadeniz Technical University, Tıp Fakültesi, Dahili Tıp, 2023 - Continues

## Courses

AYAKTAN HASTA BAKIMI (POLİKLİNİK), Expertise In Medicine, 2024 - 2025, 2023 - 2024, 2021 - 2022
Doğumsal Metabolizma Hastalıkları ve Toplum Tarama Programları, Expertise In Medicine, 2024 - 2025, 2023 - 2024
BİYOİNFORMATİK, Expertise In Medicine, 2024 - 2025, 2023 - 2024
KALITIM ŞEKİLLERİ, Expertise In Medicine, 2024 - 2025, 2023 - 2024, 2021 - 2022
MİKRODELESYON SENDROMLARI, Expertise In Medicine, 2023 - 2024
TEZ DANIŞMANLIĞI, Expertise In Medicine, 2023 - 2024
PRENATAL TANI, Expertise In Medicine, 2023 - 2024
GEN TEDAVİSİ, Expertise In Medicine, 2023 - 2024
MOLEKÜLER GENETİK UYGULAMALAR, Expertise In Medicine, 2023 - 2024
GENETİK HASTALIKLARDAN KORUNMA, Expertise In Medicine, 2023 - 2024
KONJENİTAL MALFORMASYONLAR, DİSMORFOLOJİ, Expertise In Medicine, 2023 - 2024
SUNUM/SEMİNER/MAKALE TARTIŞMASI, Expertise In Medicine, 2021 - 2022
LABORATUVAR EĞİTİMİ, Expertise In Medicine, 2021 - 2022
HÜCRE VE GENOM ORGANİZASYONU, Expertise In Medicine, 2021 - 2022
İmmün sistem genetiği ve hastalıkları, Undergraduate, 2021 - 2022
Onkogenler ve Tümör Supressor genler, Undergraduate, 2021 - 2022
OLGU/DOSYA TARTIŞMASI, Expertise In Medicine, 2021 - 2022

## Advising Theses

Türkyılmaz A., Obstrüktif Olmayan Azospermi Hastalarında Genetik Etiyolojilerin Araştırılması, Expertise In Medicine, K.ADANUR(Student), Continues

## Jury Memberships

Post Graduate, Post Graduate, Marmara Üniversitesi, September, 2022
Doctoral Examination, Doctoral Examination, Biruni Üniversitesi, January, 2022

## Published journal articles indexed by SCI, SSCI, and AHCI

I. **Analysis of TSC1 and TSC2 genes and evaluation of phenotypic correlations with tuberous sclerosis.**
Eser M., Hekimoglu G., Kutlubay B., Sager S. G., Turkyilmaz A.
Molecular genetics and genomics : MGG, vol.300, no.1, pp.6, 2024 (SCI-Expanded)

II. **Unveiling New Clinical and Genetic Insights in Ultra-Rare Intellectual Disability Phenotypes: A Study of a Turkish Cohort.**
Turkyilmaz A., Sager S. G., Terali K., Kart P. O., Kamasak T., Ayaz A., Cebi A. H., Cansu A.
Clinical genetics, 2024 (SCI-Expanded)

III. **Novel compound heterozygous P4HTM variants in a girl with developmental and epileptic encephalopathy: First case report of P4HTM variant-associated epileptic encephalopathy.**
Alomarı O., Bebek O., Turkyilmaz A., Sager S. G.
Seizure, vol.124, pp.35-38, 2024 (SCI-Expanded)

IV. **Epilepsy genetics in the paediatric population of the Eastern Anatolia region of Turkey.**
Yarali O., Gündoğdu Öğütlü Ö. B., Saritaş S., Guler M. C., Keskin F., Türkyilmaz A.
Journal of neurogenetics, pp.1-10, 2024 (SCI-Expanded)

V. **Secondary findings in genes related to cancer phenotypes in Turkish exome sequencing data from 2020 individuals**

Demir O., Saglam K., Yilmaz M., Apuhan T., Cebi A. H., Turkyilmaz A.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.194, 2024 (SCI-Expanded)

VI. **Natural history of ENPP1 deficiency: Nationwide Turkish Cohort Study of autosomal-recessive hypophosphataemic rickets type 2.**
Dursun F., Turan İ., Bitkin E. Ç., Bayramoğlu E., Çayır A., Erdeve Ş. S., Çakır E. D. P., Çamtosun E., Dilek S. O., Kırmızıbekmez H., et al.
Clinical endocrinology, vol.101, no.5, pp.475-484, 2024 (SCI-Expanded)

VII. **Somatic STK11 mosaicism in a Turkish patient with Peutz-Jeghers syndrome.**
Yilmaz M., Bebek O., Colak Y., Türkyılmaz A.
Familial cancer, vol.23, no.4, pp.641-645, 2024 (SCI-Expanded)

VIII. **A Strong Candidate Gene for Nonsyndromic Intellectual Disability Phenotype: SGSM3.**
Turkyilmaz A., Saglam K., Yilmaz M., Cebi A. H.
Clinical genetics, 2024 (SCI-Expanded)

IX. **Genetic analyses in a cohort of pediatric patients with congenital hypothyroidism based on congenital hypothyroidism consensus guideline.**
Kurnaz E., Türkyılmaz A., Yaralı O., Dönmez A. S., Çayır A.
Hormone research in paediatrics, pp.1-16, 2024 (SCI-Expanded)

X. **A Rare Presentation of Homozygous Pathogenic Variant in MC2R Gene with Salt-Wasting Crisis in a Neonate**
Kardas Yildiz A., Bulbul A., Ozer Bekmez B., TÜRKYILMAZ A., Terali K., Dagdeviren Cakir A., Ucar A.
Molecular Syndromology, vol.15, no.1, pp.77-82, 2024 (SCI-Expanded)

XI. **Biallelic Deletion of PEX26 Exon 4 in a Boy with Phenotypic Features of both Zellweger Syndrome and Infantile Refsum Disease**
Yalçınkaya B., Sağlam K., Terali K., Tekin E., Taslak H., Türkyılmaz A.
Molecular Syndromology, vol.15, no.5, pp.380-388, 2024 (SCI-Expanded)

XII. **Discovery of a Novel CUL3 Variant: Unveiling Epilepsy and Newly Associated Dysmorphic Traits in a Turkish Patient**
Colak Y., YILMAZ M., Kart P. O., Terali K., TÜRKYILMAZ A., CANSU A.
MOLECULAR SYNDROMOLOGY, 2024 (SCI-Expanded)

XIII. **Classification of Brain Magnetic Resonance Imaging Abnormalities and Spectrum of Neurological Findings in a Cohort with Copy Number Variation-Related Disorders**
TÜRKYILMAZ A., Sager S. G., Caliskan E., Akcay M., DEMİR O., Baytar B., Akin Y.
MOLECULAR SYNDROMOLOGY, 2024 (SCI-Expanded)

XIV. **Smith-Lemli-Opitz Syndrome with Biallelic c.1295A>G (p.Tyr432Cys) Variant in the DHCR7 Gene in a 73-Year-Old Woman: Report of the Oldest Patient**
Yılmaz M., Bebek O., Türkyılmaz A.
MOLECULAR SYNDROMOLOGY, vol.15, no.4, pp.317-323, 2024 (SCI-Expanded)

XV. **A triple molecular diagnosis in a Turkish individual with hypotrichosis, deafness, and diabetes**
Türkyılmaz A., Cimbek E. A., Kardeş H., Çebi A. H., Acar Arslan E., Karagüzel G.
CLINICAL DYSMORPHOLOGY, vol.33, no.3, pp.118-120, 2024 (SCI-Expanded)

XVI. **The first Turkish family with a novel biallelic missense variant of the ALKBH8 gene: A study on the clinical and variant spectrum of ALKBH8-related intellectual developmental disorders**
Yılmaz M., Kamaşak T., Teralı K., Çebi A. H., Türkyılmaz A.
American Journal of Medical Genetics, Part A, vol.194, no.5, 2024 (SCI-Expanded)

XVII. **Experience with the Ketogenic Diet in a Boy with CLCN4 Related Neurodevelopmental Disorder**
Sağer G., Yükselmiş U., Güzel O., Türkyılmaz A., Akçay M.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.26, no.2, pp.77-82, 2024 (SCI-Expanded)

XVIII. **Prediction of molecular phenotypes for novel SCN1A variants from a Turkish genetic epilepsy syndromes cohort and report of two new patients with recessive Dravet syndrome**
Teralı K., Türkyılmaz A., Sağer S. G., Çebi A. H.
CTS-CLINICAL AND TRANSLATIONAL SCIENCE, vol.17, no.1, 2024 (SCI-Expanded)

XIX. **Severe Early-Onset Obesity and Diabetic Ketoacidosis due to a Novel Homozygous c.169C>T p.Arg57*Variant in CEP19 Gene**
Cayir A., Turkyilmaz A., Rabenstein H., Guven F., Karagoz Y. S., Vuralli D., Wabitsch M., DEMİRBİLEK H.
MOLECULAR SYNDROMOLOGY, vol.15, no.2, pp.104-113, 2023 (SCI-Expanded)

XX. **Identification of novel variants in Turkish families with non-syndromic congenital cataracts using whole-exome sequencing.**
Türkyılmaz A., Kaplan A. T., Öskan Yalçın S., Sağer S. G., Şimşek Ş.
International ophthalmology, vol.43, no.12, pp.4573-4583, 2023 (SCI-Expanded)

XXI. **Expanding the clinical and immunological phenotypes of PAX1-deficient SCID and CID patients**
YAKICI N., Kreins A. Y., Catak M. C., Babayeva R., ERMAN B., Kenney H., Gungor H. E., Cea P. A., Kawai T., Bosticardo M., et al.
Clinical immunology (Orlando, Fla.), vol.255, pp.109757, 2023 (SCI-Expanded)

XXII. **Pyrroline-5-carboxylate reductase 2 (PYCR2) deficiency causes hereditary spastic paraplaegia in late childhood**
Sager G., TÜRKYILMAZ A., Gunbey H. P., Tas I., Ozhelvaci F., Akin Y.
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, vol.44, pp.51-56, 2023 (SCI-Expanded)

XXIII. **Novel, homozygous RAB3GAP1 c.2606+1G>A, p.Glu830ValfsTer9 variant and chromosome 3q29 duplication in a Turkish individual with Warburg micro syndrome**
Geckinli B., TÜRKYILMAZ A., Alavanda C., Sager G., ARSLAN ATEŞ E., SÖYLEMEZ M. A., ARMAN A.
CLINICAL DYSMORPHOLOGY, vol.32, no.2, pp.55-61, 2023 (SCI-Expanded)

XXIV. **De novo Pure Partial Trisomy 6p Associated with Facial Dysmorphism, Developmental Delay, Brain Anomalies, and Primary Congenital Hypothyroidism**
Türkyılmaz A., Cimbek E. A., Çebi A. H., Acar Arslan E., Karagüzel G.
MOLECULAR SYNDROMOLOGY, vol.14, no.1, pp.35-43, 2023 (SCI-Expanded)

XXV. **Evaluation of long-term neurocognitive functions in patients with epileptic encephalopathy with continuous spike-and-wave during sleep (CSWS)/epileptic encephalopathy with spike-and-wave activation in sleep (EE-SWAS)**
Sager G., Takis G., Pinar Z. V., Duzkalir H., TÜRKYILMAZ A., Cag Y., Akin Y.
NEUROPHYSIOLOGIE CLINIQUE-CLINICAL NEUROPHYSIOLOGY, vol.53, no.1, 2023 (SCI-Expanded)

XXVI. **A novel de novo TET3 loss-of-function variant in a Turkish boy presenting with neurodevelopmental delay and electrical status epilepticus during slow-wave sleep**
Sager S. G., TÜRKYILMAZ A., Gunbey H. P., Karatoprak E. Y., Aslan E. S., Akın Y.
Brain and Development, vol.45, no.2, pp.140-145, 2023 (SCI-Expanded)

XXVII. **Novel guanosine diphosphate-mannose pyrophosphorylase A variant in an individual with achalasia, alacrima, and intellectual disability**
Sager G., TÜRKYILMAZ A., Hanedar Y. D., Günbey H. P., Akin Y.
Clinical dysmorphology, vol.32, no.1, pp.18-20, 2023 (SCI-Expanded)

XXVIII. **Evaluation of optical coherence tomography findings and visual evoked potentials in Charcot-Marie-Tooth disease**
Kaplan A. T., Oskan Yalcin S., Sager S. G., Türkyılmaz A., İnan R.
INTERNATIONAL OPHTHALMOLOGY, vol.43, no.1, pp.333-341, 2023 (SCI-Expanded)

XXIX. **Chromosomal microarray and exome sequencing in unexplained early infantile epileptic encephalopathies in a highly consanguineous population**
TÜRKDOĞAN D., Turkyilmaz A., Sager G., Ozturk G., ÜNVER O., Say M.
INTERNATIONAL JOURNAL OF NEUROSCIENCE, vol.133, no.7, pp.683-700, 2023 (SCI-Expanded)

XXX. **Microstructural evaluation of the brain with advanced magnetic resonance imaging techniques in cases of electrical status epilepticus during sleep (ESES)**
Düzkalir H. G., Genç B., Sağer S. G., TÜRKYILMAZ A., Günbey H. P.
Turkish Journal of Medical Sciences, vol.53, no.6, pp.1840-1851, 2023 (SCI-Expanded)

XXXI. **Autosomal Recessive Primary Microcephaly (MCPH) and Novel Pathogenic Variants in ASPM and WDR62 Genes**

Bolat H., Sağer S. G., TÜRKYILMAZ A., ÇEBİ A. H., Akln Y., Onay H., Özklnay F., Ünsel-Bolat G.
Molecular Syndromology, vol.13, no.5, pp.363-369, 2022 (SCI-Expanded)

XXXII. **Novel LNPK variant causes progressive cerebral atrophy: Expanding the clinical phenotype.**
Türkyılmaz A., Sağer S. G., Günbey H. P., Akın Y.
Clinical genetics, vol.102, no.3, pp.218-222, 2022 (SCI-Expanded)

XXXIII. **Differential Diagnosis of Acromegaly: Pachydermoperiostosis Two New Cases from Turkey**
Kartal Baykan E., Türkyılmaz A.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.14, no.3, pp.350-355, 2022 (SCI-Expanded)

XXXIV. **Genotype, phenotype characteristics and long-term follow-up of patients with Vitamin D Dependent Rickets Type IA (VDDR1a): A nationwide multicentre retrospective cross-sectional study**
Cayir A., DEMİRBİLEK H., TÜRKYILMAZ A., DEMİRCİOĞLU S., BEREKET A., Darendeliler F., Ozbek M. N., Unal E., Okdemir D., Esen I., et al.
HORMONE RESEARCH IN PAEDIATRICS, no.SUPPL 2, pp.100, 2022 (SCI-Expanded)

XXXV. **Genetic Landscape of SCN1A Variants in a Turkish Cohort with GEFS plus Spectrum and Dravet Syndrome**
TÜRKYILMAZ A., Tekin E., Yarali O., ÇEBİ A. H.
MOLECULAR SYNDROMOLOGY, vol.13, no.4, pp.270-281, 2022 (SCI-Expanded)

XXXVI. **Molecular characterization of Turkish patients with demyelinating Charcot-Marie-Tooth disease.**
Karakaya T., Turkyilmaz A., Sager G., Inan R., Yarali O., Cebi A. H., Akin Y.
Neurogenetics, vol.23, no.3, pp.213-221, 2022 (SCI-Expanded)

XXXVII. **HACE1, GLRX5, and ELP2 gene variant cause spastic paraplegies**
Sager G., Turkyilmaz A., Ates E. A., Kutlubay B.
ACTA NEUROLOGICA BELGICA, vol.122, no.2, pp.391-399, 2022 (SCI-Expanded)

XXXVIII. **Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis Due to CLDN16 Gene Mutations: Novel Findings in Two Cases with Diverse Clinical Features**
Eltan M., Abali Z. Y., TÜRKYILMAZ A., GÖKCE İ., Abali S., ALAVANDA C., ARMAN A., Kirkgoz T., GÜRAN T., Hatun S., et al.
CALCIFIED TISSUE INTERNATIONAL, vol.110, no.4, pp.441-450, 2022 (SCI-Expanded)

XXXIX. **Whole-exome sequencing reveals new potential genes and variants in patients with premature ovarian insufficiency**
TÜRKYILMAZ A., ALAVANDA C., Ates E. A., GEÇKİNLİ B. B., Polat H., GÖKCÜ M., Karakaya T., ÇEBİ A. H., SÖYLEMEZ M. A., GÜNEY A. İ., et al.
JOURNAL OF ASSISTED REPRODUCTION AND GENETICS, vol.39, no.3, pp.695-710, 2022 (SCI-Expanded)

XL. **Two New Cases of Primary Microcephaly with Neuronal Migration Defect Caused by Truncating Mutations in the ASPM Gene**
TÜRKYILMAZ A., Sager S. G.
MOLECULAR SYNDROMOLOGY, vol.13, no.1, pp.56-63, 2022 (SCI-Expanded)

XLI. **A Novel ATM Gene Mutation Affecting Splicing in an Ataxia-Telangiectasia Patient**
Ates E. A., TÜRKYILMAZ A., Eltan S. B., BARIŞ S., GÜNEY A. İ.
MOLECULAR SYNDROMOLOGY, vol.13, no.1, pp.80-84, 2022 (SCI-Expanded)

XLII. **Novel SH3PXD2B variant identified by whole-exome sequencing in a Turkish newborn with Frank-Ter Haar Syndrome.**
Türkyilmaz A., Sager S. G., Topcu B., Kaplan A. T., Günbey H. P., Akin Y.
Clinical dysmorphology, vol.31, pp.45-49, 2022 (SCI-Expanded)

XLIII. **Array-Based Comparative Genomic Hybridization Analysis in Children with Developmental Delay/Intellectual Disability**
TÜRKYILMAZ A., GEÇKİNLİ B. B., Tekin E., Ates E. A., Yarali O., ÇEBİ A. H., ARMAN A.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.24, no.2, pp.15-24, 2021 (SCI-Expanded)

XLIV. **Secondary findings in 622 Turkish clinical exome sequencing data**
Ates E. A., TÜRKYILMAZ A., Yildirim O., ALAVANDA C., Polat H., Demir S., ÇEBİ A. H., GEÇKİNLİ B. B., GÜNEY A. İ., ATA

P., et al.
JOURNAL OF HUMAN GENETICS, vol.66, no.11, pp.1113-1119, 2021 (SCI-Expanded)

XLV. **A novel frameshift variant in proximal exon 18 of KAT6B gene associated with an overlapping genitopatellar/say barber Biesecker-Young-Simpson syndrome phenotype.**
Turkyilmaz A., Ozden A.
Clinical dysmorphology, vol.30, pp.197-200, 2021 (SCI-Expanded)

XLVI. **The Spectrum of Low-Density Lipoprotein Receptor Mutations in a Large Turkish Cohort of Patients with Familial Hypercholesterolemia**
TÜRKYILMAZ A., Kurnaz E., ALAVANDA C., Yarali O., Kartal Baykan E., YAVUZ D., Cayir A., ATA P.
METABOLIC SYNDROME AND RELATED DISORDERS, vol.19, pp.340-346, 2021 (SCI-Expanded)

XLVII. **First Report of a de novo 10q23.31q23.33 Microdeletion: Obesity, Intellectual Disability and Microcephaly**
TÜRKYILMAZ A., Kurnaz E., Cayir A.
MOLECULAR SYNDROMOLOGY, vol.12, no.4, pp.258-262, 2021 (SCI-Expanded)

XLVIII. **A very rare skeletal dysplasia: spondyloepimetaphyseal dysplasia, sponastrime type.**
Kalaoglu E. E., Turkyilmaz A., Geckinli B. B., Arslan Ates E., Mentes A., Arman A.
Clinical dysmorphology, vol.30, pp.150-153, 2021 (SCI-Expanded)

XLIX. **Two novel CYP2R1 mutations in a family with vitamin D-dependent rickets type 1b**
Ozden A., DÖNERAY H., Turkyilmaz A.
ENDOCRINE, vol.72, no.3, pp.852-864, 2021 (SCI-Expanded)

L. **Meckel-Gruber Syndrome: Clinical and Molecular Genetic Profiles in Two Fetuses and Review of the Current Literature.**
Turkyilmaz A., Geckinli B. B., Alavanda C., Arslan Ates E., Buyukbayrak E. E., Eren S. F., Arman A.
Genetic testing and molecular biomarkers, vol.25, pp.445-451, 2021 (SCI-Expanded)

LI. **Biallelic Mutations in DNAJB11are Associated with Prenatal Polycystic Kidney Disease in a Turkish Family**
Ates E. A., TÜRKYILMAZ A., DELİL K., ALAVANDA C., SÖYLEMEZ M. A., GEÇKİNLİ B. B., ATA P., ARMAN A.
MOLECULAR SYNDROMOLOGY, vol.12, no.3, pp.179-185, 2021 (SCI-Expanded)

LII. **Clinical characteristics and molecular genetic analysis of a cohort with idiopathic congenital hypogonadism.**
Turkyilmaz A., Cayir A., Yarali O., Kurnaz E., Kartal Baykan E., Arslan Ates E., Demirbilek H.
Journal of pediatric endocrinology & metabolism : JPEM, vol.34, pp.771-780, 2021 (SCI-Expanded)

LIII. **Genotypic Sex and Severity of the Disease Determine the Time of Clinical Presentation in Steroid 17 alpha-Hydroxylase/17,20-Lyase Deficiency**
Kurnaz E., Kartal Baykan E., Turkyilmaz A., Yarali O., Yavas Abali Z., DEMİRCİOĞLU S., BEREKET A., Cayir A., GÜRAN T.
HORMONE RESEARCH IN PAEDIATRICS, vol.93, pp.558-566, 2021 (SCI-Expanded)

LIV. **Novel clinical features and pleiotropic effect in three unrelated patients with LMNA variant**
Turkyilmaz A., GEÇKİNLİ B. B., ALAVANDA C., Ates E. A., ARMAN A.
CLINICAL DYSMORPHOLOGY, vol.30, no.1, pp.10-16, 2021 (SCI-Expanded)

LV. **A Novel ELP2 Compound Heterozygous Mutation in a Boy with Severe Intellectual Disability, Spastic Diplegia, Stereotypic Behavior and Review of the Current Literature**
Turkyilmaz A., Sager G.
MOLECULAR SYNDROMOLOGY, vol.11, pp.315-319, 2020 (SCI-Expanded)

LVI. **FGF3-Related Phenotypes: A Study of LAMM Syndrome and Otodental Dysplasia Patients with Two Novel Mutations in FGF3 Gene**
Turkyilmaz A., GEÇKİNLİ B. B., ALAVANDA C., Zengin G., Ates E. A., ARMAN A.
INTERNATIONAL JOURNAL OF HUMAN GENETICS, vol.20, no.4, pp.179-190, 2020 (SCI-Expanded)

LVII. **Expansion of the phenotypic spectrum of SMC1A nonsense variants: a patient with cerebellar atrophy and review of the literature**
Tuerkyilmaz A., TÜRKDOĞAN D., Goermez Z., Ekinci G.

CLINICAL DYSMORPHOLOGY, vol.29, no.4, pp.217-223, 2020 (SCI-Expanded)

LVIII. **A large Turkish pedigree with multiple endocrine neoplasia type 1 syndrome carrying a rare mutation: c.1680_1683 del TGAG**
DEMİRTAŞ C. Ö., ATA P., Cetin A., Turkyilmaz A., Duman D. G.
TURKISH JOURNAL OF GASTROENTEROLOGY, vol.31, no.7, pp.508-514, 2020 (SCI-Expanded)

LIX. **THE EXPRESSION LEVELS OF microRNAs ASSOCIATED WITH T AND B CELL DIFFERENTIATION/STIMULATION IN ANKYLOSING SPONDYLITIS**
Turkyilmaz A., ATA P., Akbas F., YAĞCI İ.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.23, no.1, pp.25-31, 2020 (SCI-Expanded)

LX. **A VERY RARE PARTIAL TRISOMY SYNDROME: DE NOVO DUPLICATION OF 16q12.1q23.3 IN A TURKISH GIRL WITH DEVELOPMENTAL DELAY AND FACIAL DYSMORPHIC FEATURES**
Turkyilmaz A., Yarali O.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.23, no.1, pp.103-107, 2020 (SCI-Expanded)

LXI. **Low DHEAS Concentration in a Girl Presenting with Short Stature and Premature Pubarche: A Novel PAPSS2 Gene Mutation**
ELTAN M., Yavas Abali Z., Arslan Ates E., Kirkgoz T., KAYGUSUZ S. B., Turkyilmaz A., BEREKET A., DEMİRCİOĞLU S., GÜRAN T.
HORMONE RESEARCH IN PAEDIATRICS, vol.92, no.4, pp.262-268, 2020 (SCI-Expanded)

LXII. **A novel DCAF17 homozygous mutation in a girl with Woodhouse-Sakati syndrome and review of the current literature**
Kurnaz E., Turkyilmaz A., Yarali O., Demir B., Cayir A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.32, no.11, pp.1287-1293, 2019 (SCI-Expanded)

LXIII. **A NOVEL SPLICE-SITE MUTATION ON THE MLC1 GENE LEADING TO EXON 9 SKIPPING AND MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS IN A TURKISH PATIENT**
Turkyilmaz A., ÜNVER O., Ekinci G., TÜRKDOĞAN D.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.22, no.2, pp.89-91, 2019 (SCI-Expanded)

LXIV. **A novel truncating mutation of DOCK7 gene with an early-onset non-encephalopathic epilepsy**
TÜRKDOĞAN D., Turkyilmaz A., Gormez Z., Sager G., Ekinci G.
SEIZURE-EUROPEAN JOURNAL OF EPILEPSY, vol.66, pp.12-14, 2019 (SCI-Expanded)

LXV. **Chanarin-Dorfman syndrome**
Kalyon S., Gokden Y., Demirel N., Erden B., Turkyilmaz A.
TURKISH JOURNAL OF GASTROENTEROLOGY, vol.30, no.1, pp.105-108, 2019 (SCI-Expanded)

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## Memberships / Tasks in Scientific Organizations

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## Scientific Refereeing

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MOLECULAR BIOLOGY REPORTS, SCI Journal, May 2024
CLINICAL GENETICS, SCI Journal, March 2023
AMERICAN JOURNAL OF MEDICAL GENETICS, PART A, SCI Journal, February 2022
ANNALS OF LABORATORY MEDICINE, SCI Journal, January 2022
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## Congress and Symposium Activities

European Human Genetics Conference, Audience, Berlin, Germany, 2024
2. Hematoonkogenetik Kongresi, Audience, Girne, Cyprus (Kktc), 2023
European Human Genetics Conference , Attendee, Vienna, Austria, 2022
Session Moderator, Antalya, Turkey, 2021
XVII. Tıbbi Biyoloji ve Genetik Kongresi, Attendee, Ankara, Turkey, 2021
43. Pediatri Günleri ve 22.Pediatri Hemşireliği Günleri, Attendee, İstanbul, Turkey, 2021
INTERNATIONAL GEVHER NESIBE HEALTH SCIENCES CONFERENCE-VII, Attendee, Kayseri, Turkey, 2021
CERRAHPAŞA PEDİATRİ GÜNLERİ SEMPTOMDAN TANIYA, Attendee, İstanbul, Turkey, 2021
İSTANBUL MEDENİYET ÜNİVERSİTESİ ÇOCUK MEDENİYETİ ÇEVRİM İÇİ SEMPOZYUM, Attendee, İstanbul, Turkey, 2021
17. Ulusal Uludağ Pediatri Kış Kongresi, Attendee, Bursa, Turkey, 2021
4. Ege Endokrin Hastalıklar ve Genetik Sempozyumu, Attendee, İzmir, Turkey, 2021
American Society of Human Genetics Conference, Attendee, Texas, United States Of America, 2019
13th Balkan Congress of Human Genetics, Attendee, Edirne, Turkey, 2019
13. Ulusal Tıbbi Genetik Kongresi, Attendee, Antalya, Turkey, 2018
51st European Society of Human Genetics Conference, Attendee, Milan, Italy, 2018
50th European Society of Human Genetics Conference, Attendee, Kobenhavn, Denmark, 2017
12. Ulusal Tıbbi Genetik Kongresi, Attendee, İzmir, Turkey, 2016