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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

Doğumsal Metabolizma Hastalıkları ve Toplum Tarama Programları, Expertise In Medicine, 2023 - 2024
BİYOİNFORMATİK, Expertise In Medicine, 2023 - 2024
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
AYAKTAN HASTA BAKIMI (POLİKLİNİK), Expertise In Medicine, 2023 - 2024, 2021 - 2022
KONJENİTAL MALFORMASYONLAR, DİSMORFOLOJİ, Expertise In Medicine, 2023 - 2024
KALITIM ŞEKİLLERİ, Expertise In Medicine, 2023 - 2024, 2021 - 2022
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. **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)

II. **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)

III. **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, 2024 (SCI-Expanded)

IV. **Somatic STK11 mosaicism in a Turkish patient with Peutz-Jeghers syndrome.**
Yilmaz M., Bebek O., Colak Y., Türkyılmaz A.
Familial cancer, 2024 (SCI-Expanded)

V. **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)

VI. **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)

VII. **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, 2024 (SCI-Expanded)

VIII. **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, 2024 (SCI-Expanded)

IX. **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, 2024 (SCI-Expanded)

X. **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)

XI. **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)

XII. **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)

XIII. **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)

XIV. **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)

XV. **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)

XVI. **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)

XVII. **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)

XVIII. **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)

XIX. **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)

XX. **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)

XXI. **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)

XXII. **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)

XXIII. **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)

XXIV. **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)

XXV. **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)

XXVI. **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)

XXVII. **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)

XXVIII. **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)

XXIX. **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)

XXX. **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)

XXXI. **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)

XXXII. **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)

XXXIII. **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)

XXXIV. **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)

XXXV. **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)

XXXVI. **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)

XXXVII. **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)

XXXVIII. **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)

XXXIX. **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)

XL. **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)

XLI. **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)

XLII. **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)

XLIII. **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)

XLIV. **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)

XLV. **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)

XLVI. **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)

XLVII. **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)

XLVIII. **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)

XLIX. **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)

L. **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)

LI. **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)

LII. **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)

LIII. **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)

LIV. **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)

LV. **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)

LVI. **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)

LVII. **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)

Articles Published in Other Journals

I. **Frequency of Familial Mediterranean Fever Gene Mutation in Patients Presenting With Joint Pain and Diagnosed With Acute Rheumatic Fever**
Gullu U. U., Balaban I., Kara S. S., Yarali O., Tuerkyilmaz A., Ipek S., Guellue S. D., Caliskan O. F.
CUREUS JOURNAL OF MEDICAL SCIENCE, no.8, 2023 (ESCI)

II. **Lunapark deficiency leads to an autosomal recessive neurodevelopmental phenotype with a degenerative course, epilepsy and distinct brain anomalies**
Accogli A., Zaki M. S., Al-Owain M., Otaif M. Y., Jackson A., Argilli E., Chandler K. E., De Goede C. G. E. L., Cora T., Alvi J. R., et al.
BRAIN COMMUNICATIONS, vol.5, no.5, 2023 (ESCI)

III. **A Genetic Approach in the Evaluation of Short Stature**
TÜRKYILMAZ A., Donmez A. S., Cayir A.
Eurasian Journal of Medicine, vol.54, 2022 (ESCI)

IV. **Genetic Forms of Calciopenic Rickets**
Donmez A. S., Türkyılmaz A., Cayir A.
Eurasian Journal of Medicine, vol.54, 2022 (ESCI)

V. **Investigating CFTR gene variations in patient groups with positive newborn screening test results and preliminary clinical diagnosis of cystic fibrosis in the eastern anatolia region of Turkey**
TÜRKYILMAZ A., YARALI O.
Medicine Science | International Medical Journal, vol.10, no.2, pp.293-298, 2021 (Peer-Reviewed Journal)

VI. **Screening of MC4R, LEP, LEPR, POMC, SH2B1, and SIM1 genes in Turkish children with severe early-onset obesity**
TÜRKYILMAZ A., YARALI O., KURNAZ E., ÇAYIR A.
Medicine Science | International Medical Journal, vol.10, no.2, pp.328-333, 2021 (Peer-Reviewed Journal)

VII. **Can Uric Acid Be A Marker For Metabolically Unhealthy Obesity in Children and Adolescents?**
KURNAZ E., TÜRKYILMAZ A., ÇAYIR A.
Turkish Journal of Pediatric Disease, vol.15, pp.226-233, 2021 (Peer-Reviewed Journal)

Books & Book Chapters

I. **KALSİYOPENİK RAŞİTİZMİN GENETİK FORMLARI**
ÇAYIR A., TÜRKYILMAZ A., KURNAZ E.
in: Çocuk Endokrinolojisi ve Diyabet, DARENDELİLER FEYZA, AYCAN ZEHRA, KARA CENGİZ, ÖZEN SAMİM, EREN ERDAL, Editor, İSTANBUL TIP KİTAPEVLERİ, İstanbul, pp.1862-1874, 2021

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I. **2018-2022 Yılları Arasında AML Tanısı Almış Hastaların Genetik Test ve Tedavilerinin Değerlendirilmesi: Doğu Karadeniz Bölgesi Sonuçları**
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