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TÜRKYILMAZ



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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 etkili 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 Region Health Research Center, Department Of Internal Medicine, 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
BİYOBİYOİNFORMATİK, 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. **Epilepsy genetics in the paediatric population of the Eastern Anatolia region of Turkey.**
Yarali O., Gündoğdu Öğütlü Ö. B., Sarıtaş S., Guler M. C., Keskin F., Türkyılmaz A.
Journal of neurogenetics, pp.1-10, 2024 (SCI-Expanded)
- II. **Somatic STK11 mosaicism in a Turkish patient with Peutz-Jeghers syndrome.**
Yılmaz M., Bebek O., Colak Y., Türkyılmaz A.
Familial cancer, vol.23, no.4, pp.641-645, 2024 (SCI-Expanded)
- III. **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)
- IV. **A Strong Candidate Gene for Nonsyndromic Intellectual Disability Phenotype: SGSM3.**
Türkyılmaz A., Sağlam K., Yılmaz M., Cebi A. H.
Clinical genetics, 2024 (SCI-Expanded)
- V. **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)

- VI. **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, TÜRKİYILMAZ A, Terali K, Dagdeviren Cakir A, Ucar A.
Molecular Syndromology, vol.15, no.1, pp.77-82, 2024 (SCI-Expanded)
- VII. **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)
- VIII. **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., TÜRKİYILMAZ A., CANSU A.
MOLECULAR SYNDROMOLOGY, 2024 (SCI-Expanded)
- IX. **Classification of Brain Magnetic Resonance Imaging Abnormalities and Spectrum of Neurological Findings in a Cohort with Copy Number Variation-Related Disorders**
TÜRKİYILMAZ A., Sager S. G., Caliskan E., Akçay M., DEMİR O., Baytar B., Akin Y.
MOLECULAR SYNDROMOLOGY, 2024 (SCI-Expanded)
- X. **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)
- XI. **A triple molecular diagnosis in a Turkish individual with hypotrichosis, deafness, and diabetes**
Türkyılmaz A., Cimbeç E. A., Kardeş H., Çebi A. H., Acar Arslan E., Karagüzel G.
CLINICAL DYSMORPHOLOGY, vol.33, no.3, pp.118-120, 2024 (SCI-Expanded)
- XII. **Secondary findings in genes related to cancer phenotypes in Turkish exome sequencing data from 2020 individuals**
Demir O., Sağlam K., Yılmaz M., Apuhan T., Cebi A. H., Turkyılmaz A.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, 2024 (SCI-Expanded)
- XIII. **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., Terali K., Çebi A. H., Türkyılmaz A.
American Journal of Medical Genetics, Part A, vol.194, no.5, 2024 (SCI-Expanded)
- XIV. **Experience with the Ketogenic Diet in a Boy with CLCN4 Related Neurodevelopmental Disorder**
Sağır G., Yükselmiş U., Güzel O., Türkyılmaz A., Akçay M.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.26, no.2, pp.77-82, 2024 (SCI-Expanded)
- XV. **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**
Terali K., Türkyılmaz A., Sağır S. G., Çebi A. H.
CTS-CLINICAL AND TRANSLATIONAL SCIENCE, vol.17, no.1, 2024 (SCI-Expanded)
- XVI. **Severe Early-Onset Obesity and Diabetic Ketoacidosis due to a Novel Homozygous *c.169C>T p.Arg57** Variant in *CEP19* Gene**
Çayır A., Turkyılmaz A., Rabenstein H., Guven F., Karagoz Y. S., Vuralli D., Wabitsch M., DEMİR BİLİK H.
MOLECULAR SYNDROMOLOGY, vol.15, no.2, pp.104-113, 2023 (SCI-Expanded)
- XVII. **Identification of novel variants in Turkish families with non-syndromic congenital cataracts using whole-exome sequencing.**
Türkyılmaz A., Kaplan A. T., Öskan Yalçın S., Sağır S. G., Şimşek Ş.
International ophthalmology, vol.43, no.12, pp.4573-4583, 2023 (SCI-Expanded)
- XVIII. **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)
- XIX. **Pyrroline-5-carboxylate reductase 2 (PYCR2) deficiency causes hereditary spastic paraplaegia in late childhood**
Sager G., TÜRKYILMAZ A., Gunbey H. P., Tas I., Ozhelvacı F., Akin Y.
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, vol.44, pp.51-56, 2023 (SCI-Expanded)
- XX. **Novel, homozygous <i>RAB3GAP1</i> 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)
- XXI. **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., Akin Y.
Brain and Development, vol.45, no.2, pp.140-145, 2023 (SCI-Expanded)
- XXII. **De novo Pure Partial Trisomy 6p Associated with Facial Dysmorphism, Developmental Delay, Brain Anomalies, and Primary Congenital Hypothyroidism**
Türkyılmaz A., Cimbeke E. A., Çebi A. H., Acar Arslan E., Karagüzel G.
MOLECULAR SYNDROMOLOGY, vol.14, no.1, pp.35-43, 2023 (SCI-Expanded)
- XXIII. **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)
- XXIV. **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)
- XXV. **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)
- XXVI. **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., Sağır S. G., TÜRKYILMAZ A., Günbey H. P.
Turkish Journal of Medical Sciences, vol.53, no.6, pp.1840-1851, 2023 (SCI-Expanded)
- XXVII. **Chromosomal microarray and exome sequencing in unexplained early infantile epileptic encephalopathies in a highly consanguineous population**
TÜRKDOĞAN D., Türkyılmaz A., Sager G., Ozturk G., ÜNVER O., Say M.
INTERNATIONAL JOURNAL OF NEUROSCIENCE, vol.133, no.7, pp.683-700, 2023 (SCI-Expanded)
- XXVIII. **Autosomal Recessive Primary Microcephaly (MCPH) and Novel Pathogenic Variants in ASPM and WDR62 Genes**
Bolat H., Sağır S. G., TÜRKYILMAZ A., ÇEBİ A. H., Akin Y., Onay H., Özknay F., Ünşel-Bolat G.
Molecular Syndromology, vol.13, no.5, pp.363-369, 2022 (SCI-Expanded)
- XXIX. **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., DEMİRBILEK 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)
- XXX. **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)

- XXXI. **Novel LNPk variant causes progressive cerebral atrophy: Expanding the clinical phenotype.**
Türkyılmaz A., Sağır S. G., Günbey H. P., Akin Y.
Clinical genetics, vol.102, no.3, pp.218-222, 2022 (SCI-Expanded)
- XXXII. **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)
- XXXIII. **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)
- XXXIV. **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)
- XXXV. **HACE1, GLRX5, and ELP2 gene variant cause spastic paraplegias**
Sager G., Turkyilmaz A., Ates E. A., Kutlubay B.
ACTA NEUROLOGICA BELGICA, vol.122, no.2, pp.391-399, 2022 (SCI-Expanded)
- XXXVI. **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)
- XXXVII. **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)
- XXXVIII. **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)
- XXXIX. **Novel SH3PXD2B variant identified by whole-exome sequencing in a Turkish newborn with Frank-Ter Haar Syndrome.**
Türkyılmaz A., Sager S. G., Topcu B., Kaplan A. T., Günbey H. P., Akin Y.
Clinical dysmorphology, vol.31, pp.45-49, 2022 (SCI-Expanded)
- XL. **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)
- XLI. **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)
- XLII. **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)
- XLIII. **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)
- XLIV. **A very rare skeletal dysplasia: spondyloepimetaphyseal dysplasia, sponastrime type.**

- Kalaoglu E. E., Turkyilmaz A., Geckinli B. B., Arslan Ates E., Menten A., Arman A.
Clinical dysmorphology, vol.30, pp.150-153, 2021 (SCI-Expanded)
- XLV. **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)
- XLVI. **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)
- XLVII. **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)
- XLVIII. **Biallelic Mutations in DNAJB11 are 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)
- XLIX. **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)
- L. **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)
- LI. **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)
- LII. **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)
- LIII. **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)
- LIV. **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)
- LV. **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)
- LVI. **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)
- LVII. **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)
- LVIII. **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., Turkeyilmaz A., BEREKET A., DEMİRCİOĞLU S., GÜRAN T.
HORMONE RESEARCH IN PAEDIATRICS, vol.92, no.4, pp.262-268, 2020 (SCI-Expanded)
- LIX. **A novel DCAF17 homozygous mutation in a girl with Woodhouse-Sakati syndrome and review of the current literature**
Kurnaz E., Turkeyilmaz A., Yarali O., Demir B., Cayir A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.32, no.11, pp.1287-1293, 2019 (SCI-Expanded)
- LX. **A NOVEL SPLICE-SITE MUTATION ON THE MLC1 GENE LEADING TO EXON 9 SKIPPING AND MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS IN A TURKISH PATIENT**
Turkeyilmaz A., ÜNVER O., Ekinci G., TÜRKDOĞAN D.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.22, no.2, pp.89-91, 2019 (SCI-Expanded)
- LXI. **A novel truncating mutation of DOCK7 gene with an early-onset non-encephalopathic epilepsy**
TÜRKDOĞAN D., Turkeyilmaz A., Gormez Z., Sager G., Ekinci G.
SEIZURE-EUROPEAN JOURNAL OF EPILEPSY, vol.66, pp.12-14, 2019 (SCI-Expanded)
- LXII. **Chanarin-Dorfman syndrome**
Kalyon S., Gokden Y., Demirel N., Erden B., Turkeyilmaz 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**
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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 NESİBE HEALTH SCIENCES CONFERENCE-VII, Attendee, Kayseri, Turkey, 2021

CERRAHPAŞA PEDIATRİ GÜNLERİ SEMPTOMDAN TANIYA, Attendee, İstanbul, Turkey, 2021

İSTANBUL MEDENİYET ÜNİVERSİTESİ ÇOCUK MEDENİYETİ ÇEVİRİM 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