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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. **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)

II. **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)

III. **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)

IV. **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)

V. **A Strong Candidate Gene for Nonsyndromic Intellectual Disability Phenotype: SGSM3.**

Turkyilmaz A., Saglam K., Yilmaz M., Cebi A. H.
Clinical genetics, 2024 (SCI-Expanded)

VI. **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)

VII. **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)

VIII. **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)

IX. **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)

X. **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)

XI. **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)

XII. **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)

XIII. **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)

XIV. **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)

XV. **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)

XVI. **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)

XVII. **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)

XVIII. **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)

XIX. **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)

XX. **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)

XXI. **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)

XXII. **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)

XXIII. **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)

XXIV. **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)

XXV. **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)

XXVI. **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)

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. **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)

XXIX. **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)

XXX. **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)

XXXI. **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)

XXXII. **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)

XXXIII. **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)

XXXIV. **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)

XXXV. **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)

XXXVI. **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)

XXXVII. **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)

XXXVIII. **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)

XXXIX. **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)

XL. **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)

XLI. **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)

XLII. **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)

XLIII. **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)

XLIV. **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)

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. **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)

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 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)

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. **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)

LI. **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)

LII. **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)

LIII. **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)

LIV. **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)

LV. **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)

LVI. **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)

LVII. **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)

LVIII. **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)

LIX. **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)

LX. **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)

LXI. **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)

LXII. **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)

LXIII. **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**
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Türkyılmaz A., Sağer S. G.
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IX. **Tüm Ekzom Dizileme Yapılan Bilişsel Gerilik Fenotipi Ile Takipli 72 Olgunun Klinik ve Genetik Bulgularının Değerlendirilmesi**
Türkyılmaz A., Sağer S. G.
15. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 9 - 13 November 2022, pp.64

X. **Boy Kısalığının Nadir Bir Sebebi; Acan Mutasyonu ve Büyüme Hormonu Tedavisi**
Karagüzel G., Çebi A. H., Yıldız Boyraz A., Cimbek E. A., Gökcü M., Türkyılmaz A.
15. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 9 - 13 November 2022, pp.289

XI. **Kenny-Caffey Syndrome Type 2: Clinical And Molecular Features In Four Unrelated Patients And Report Of A Novel Fam111A Variant**
Kaya M., Eviz E., Koçak Eker H., Tekmenüray Ünal A., Türkyılmaz A., Çepni E., Çayır A., Özbek M. N., Büyükinan M., Altunoğlu U.
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XII. **Konjenital Kalp Hastalığının Nkx2-5 Gen Varyantları Ile Ilişkisi**
Geçkinli B. B., Demir Ş., Girgin G., Türkyılmaz A., Akalın F., Arman A.
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XIII. **Evaluation Of The Mutation Spectrum Of 20 Bardet-Biedl Syndrome Cases In Turkish Population**
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XIV. **A RARE CASE of IMMUNODEFICIENCY-ICF 1 SYNDROME DIAGNOSED FOLLOWING COVID-19 INFECTION**
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XV. **OTOİMMUN SKLEROZAN KOLANJİT VE HEM OKSİJENAZ EKSİKLİĞİ**
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XVII. **BOY KISALIĞI İLE GELEN FLOATİNG-HARBOR SENDROMLU BİR OLGU**
Yıldız Poyraz A., Türkyılmaz A., Cimbek E. A., Karagüzel G.
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XVIII. **CİNSİYET GELİŞİM BOZUKLUĞUNUN NADİR BİR NEDENİ: 1q4 DELESYON SENDROMU**
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XIX. **Cinsel gelişim bozukluğunun nadir bir nedeni; 1q4 delesyon sendromu**
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XX. **Genişletilmiş Gen Panelleri Moleküler Tanıya Beklenenin Ötesinde Katkı Sunabilir Mi?**
TÜRKYILMAZ A., ÇEBİ A. H.
1. Ulusal HematoOnkoGenetik Kongresi, Antalya, Turkey, 25 November 2021, pp.106

XXI. **Primer Mikrosefali Hasta Kohortunda Genetik Etyolojinin Araştırılması**
TÜRKYILMAZ A., SAĞER S. G., ÇEBİ A. H.
XVII. Tıbbi Biyoloji ve Genetik Kongresi, ONLİNE KONGRE, Turkey, 28 October 2021, vol.52, pp.257-258

XXII. **Sendromik Olmayan Konjenital Katarakt Olgularında Tek Gen Defektlerinin Araştırılması**
TÜRKYILMAZ A., KAPLAN A. T., Özkan Yalçın S., SAĞER S. G.
5. Ulusal Çocuk Genetik Kongresi, İzmir, Turkey, 07 October 2021, pp.118

XXIII. **ATOPİK DERMATİT VE İNEK SÜTÜ PROTEİN ALERJİSİ KLİNİĞİ İLE BAŞVURAN STAT 3 EKSİKLİĞİ OLGUSU**
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15. Uluslararası Katılımlı ÇOCUK ALERJİ VE ASTIM KONGRESİ, Muğla, Turkey, 30 October - 02 November 2021, pp.124

XXIV. **Herediter Ataksi Ön Tanılı Hasta Kohortunda Genetik Etyolojinin Araştırılması**
TÜRKYILMAZ A., SAĞER S. G.
43. Pediatri Günleri ve 22.Pediatri Hemşireliği Günleri, İstanbul, Turkey, 30 May 2021

XXV. **Serebellar Malformasyonlu Hasta Kohortunda Genetik Etyolojinin Araştırılması**
TÜRKYILMAZ A., SAĞER S. G.
CERRAHPAŞA PEDİATRİ GÜNLERİ SEMPTOMDAN TANIYA, İstanbul, Turkey, 15 April 2021

XXVI. **METABOLİK HASTALIK TANISIYLA TAKİPLİ HASTALARIMIZIN KLİNİK, GENETİK VE EEG BULGULARININ DEĞERLENDİRİLMESİ EVALUATION OF CLINICAL, GENETIC AND EEG FINDINGS OF THE PATIENTS FOLLOWED WITH THE DIAGNOSIS OF METABOLIC DISEASES**
SAĞER S. G., TÜRKYILMAZ A.
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XXVII. **GEFS+ SPEKTRUM Olgularında ARRAY-CGH Değerlendirmesi**
SAĞER S. G., TÜRKYILMAZ A.
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XXVIII. **Distoni Kliniğiyle Takipli 40 Olgunun Klinik ve Genetik Bulgularının Değerlendirilmesi**
SAĞER S. G., TÜRKYILMAZ A.
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XXIX. **KONJENİTAL HİPOTİROİDİ OLGULARIMIZIN KLİNİK VE GENETİK SONUÇLARININ DEĞERLENDİRİLMESİ: TEK MERKEZ DENEYİMİ**
KURNAZ E., TÜRKYILMAZ A., ÇAYIR A.
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XXX. **MODY Ön Tanılı 188 Olgunun Klinik ve Genetik Bulgularının Değerlendirilmesi**
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XXXI. **A COHORT OF PATIENTS WITH HYPERTROPHIC AND DILATED CARDIOMYOPATHY**
POLAT H., TÜRKYILMAZ A., ALAVANDA C., ARSLAN ATEŞ E., SÖYLEMEZ M. A., GEÇKİNLİ B. B., YILDIRIM Ö., ARMAN A.
13th Balkan Congress of Human Genetics, Edirne, Turkey, 17 April 2019, pp.86

XXXII. **LAMM SYNDROME; TWO NEW PATIENTS, ONE NOVEL MUTATION AND ONE NEW MECHANISM**
ALAVANDA C., TÜRKYILMAZ A., ARSLAN ATEŞ E., GEÇKİNLİ B. B., SÖYLEMEZ M. A., ARMAN A.
13th Balkan Congress of Human Genetics, Edirne, Turkey, 17 April 2019, pp.48

XXXIII. **Von Hippel Lindau Patients**
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13 Balkan Congress of Human Genetics, Edirne, Turkey, 17 April 2019, pp.150

XXXIV. **Marfan Syndrome: Genotype-Phenotype Correlations**
GEÇKİNLİ B. B., ARSLAN ATEŞ E., TÜRKYILMAZ A., ALAVANDA C., YILDIRIM Ö., SÖYLEMEZ M. A., ARMAN A.
13th Balkan Congress of Human Genetics, Edirne, Turkey, 17 April 2019, pp.151

XXXV. **A novel intronic ATM gene mutation affecting splicing in a patient with Ataxia-Telangiectasia**
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51st European Society of Human Genetics Conference, Milan, Italy, 16 June 2018, vol.27, pp.255

XXXVI. **FGF3 gene mutations related to two syndromic Congenital deafness cases: Congenital deafness with inner ear agenesis (Michel aplasia), microtia, and microdontia and Otodental dysplasia**
TÜRKYILMAZ A., GEÇKİNLİ B. B., ARSLAN ATEŞ E., SÖYLEMEZ M. A., GÜNEY A. İ., ATA P., ARMAN A.
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XXXVII. **BİR KALITIMSAL KANSER AİLESİNDE ÜÇ FARKLI FENOTİPE YOL AÇAN ÜÇ GENOTİP**
ARSLAN ATEŞ E., TÜRKYILMAZ A., ALAVANDA C., YILDIRIM Ö., SÖYLEMEZ M. A., GEÇKİNLİ B. B., GÜNEY A. İ.
13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 07 November 2018, vol.30, pp.80

XXXVIII. **FARKLI GENLER,FARKLI MUTASYONLAR,FENOTİPTE FARKLILIK YARATIYOR MU ?: ALPORT SENDROMU**
ATA P., ALAVANDA C., TÜRKYILMAZ A., YILDIZ N., ALPAY H.
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XXXIX. **REVERSE GENETİK İLE TANI KOYULAN NADİR BİR SPASTİK PARAPLEJİ AİLESİ**
ALAVANDA C., ARSLAN ATEŞ E., GEÇKİNLİ B. B., TÜRKYILMAZ A., YILDIRIM Ö., SÖYLEMEZ M. A., SAĞER S. G., GÜNEY A. İ., ATA P., ARMAN A.
13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 07 November 2018, vol.30, pp.127

XL. **İKİ LAMİNOPATİ OLGUSU: LMNA GENİ MUTASYONLARI İLE İLİŞKİLİ FENOTİPLER**
GEÇKİNLİ B. B., TÜRKYILMAZ A., ARSLAN ATEŞ E., YILDIRIM Ö., SÖYLEMEZ M. A., ATA P., ARMAN A.
13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 07 November 2018, vol.30, pp.308

XLI. **MARMARA ÜNİVERSİTESİ TIP FAKÜLTESİ TIBBİ GENETİK KLİNİĞİNE BAŞVURAN ARİTMİ TANILI HASTA KOHORTU VE GENETİK ANALİZ SONUÇLARI**

TÜRKYILMAZ A., ALAVANDA C., ARSLAN ATEŞ E., SÖYLEMEZ M. A., GEÇKİNLİ B. B., YILDIRIM Ö., ARMAN A.
13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 07 November 2018, vol.30, pp.165

XLII. **PAPSS2 GENİNDE NOVEL BİR MUTASYON SAPTANAN NADİR BİR OTOZOMAL RESESİF BRAKİYOLMİ OLGUSU**
ARSLAN ATEŞ E., ELTAN M., TÜRKYILMAZ A., ALAVANDA C., SÖYLEMEZ M. A., GEÇKİNLİ B. B., GÜNEY A. İ., GÜRAN T., ARMAN A.
13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 07 November 2018, vol.30, pp.333

XLIII. **GENETİK KLİNİĞİNE PSÖDOBARTTER SENDROMU BULGULARIYLA BAŞVURAN KİSTİK FİBROZLU İKİ KIZ KARDEŞ**
ALAVANDA C., ATA P., TÜRKYILMAZ A., ARSLAN E., YILDIZ N., ALPAY H.
13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 07 November 2018, vol.30, pp.299

XLIV. **NÖROMOTOR GELİŞME GERİLİĞİ VE DİSMORFİK BULGULARI OLAN HASTALARDA ARRAY-CGH ANALİZİ**
ALAVANDA C., ARSLAN ATEŞ E., TÜRKYILMAZ A., GEÇKİNLİ B. B., ATA P., GÜNEY A. İ., SÖYLEMEZ M. A., ÖZYAVUZ ÇABUK P., ARMAN A.
13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 07 November 2018, vol.30, pp.258

XLV. **2 KARDEŞTE WARBURG MİKRO SENDROMU' NA NEDEN OLAN RAB3GAP1 GENİ NOVEL İNTRONİK MUTASYONU**
GEÇKİNLİ B. B., TÜRKYILMAZ A., ARSLAN ATEŞ E., ALAVANDA C., TAŞLIDERE H., SÖYLEMEZ M. A., ARMAN A.
13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 07 November 2018, vol.30, pp.271

XLVI. **Nöromotor gelişme geriliği ve distonili Xq28 duplikasyon sendromu**
GEÇKİNLİ B. B., Şimşek H., DELİL K., TÜRKDOĞAN D., SAĞER S. G., SÖYLEMEZ M. A., ARSLAN ATEŞ E., TÜRKYILMAZ A., KARAKAYA T., GÜNEY A. İ., et al.
12. Ulusal Tıbbi Genetik Kongresi, İzmir, Turkey, 05 October 2016, pp.214

XLVII. **Inversion Y Having Different Phenotypic Expressions at Three Brothers**
TÜRKYILMAZ A., ATA P., DELİL K., SOYSAL S., ARSLAN ATEŞ E., GÜNEY A. İ.
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XLVIII. **Clinical Evaluation of a MDC1A Case Carrying LAMA2 Mutation**
SÖYLEMEZ M. A., TÜRKYILMAZ A., ARSLAN ATEŞ E., Şimşek H., KARAKAYA T., DELİL K., GEÇKİNLİ B. B., ATA P., GÜNEY A. İ., ARMAN A.
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XLIX. **Beckwith Wiedemann Sendromlu Olgunun Klinik Bulguları Ve Moleküler Tanısı**
KARAKAYA T., SÖYLEMEZ M. A., ARSLAN ATEŞ E., TÜRKYILMAZ A., Şimşek H., DELİL K., GEÇKİNLİ B. B., ATA P., GÜNEY A. İ., ARMAN A.
12. Ulusal Tıbbi Genetik Kongresi, İzmir, Turkey, 05 October 2016, pp.254

L. **Von Hippel-Lindau Sendromu Ailesindeki Asemptomatik 3 Çocuğun Genetik Test Sonuçlarının Değerlendirilmesi**
DELİL K., GEÇKİNLİ B. B., Şimşek H., TÜRKYILMAZ A., ARSLAN ATEŞ E., SÖYLEMEZ M. A., GÜNEY A. İ., ATA P., ARMAN A.
12. Ulusal Tıbbi Genetik Kongresi, İzmir, Turkey, 05 October 2016, pp.183

LI. **Nadir Görülen Bir Genetik Sendrom: Meacham Sendromu**
DELİL K., TÜRKYILMAZ A., Şimşek H., KARAKAYA T., ARSLAN ATEŞ E., SÖYLEMEZ M. A., GEÇKİNLİ B. B., GÜNEY A. İ., ATA P., ARMAN A.
12. Ulusal Tıbbi Genetik Kongresi, İzmir, Turkey, 05 October 2016, pp.301

LII. **Silver-Russell Sendromlu bir olgu**
GEÇKİNLİ B. B., DELİL K., Şimşek H., TÜRKYILMAZ A., ARSLAN ATEŞ E., KARAKAYA T., GÜNEY A. İ., ATA P., ARMAN A.
12. Ulusal Tıbbi Genetik Kongresi, İzmir, Turkey, 05 October 2016, pp.213

LIII. **Chromosome 17p11.2 deletion in a Turkish girl with Smith-Magenis Syndrome**
GEÇKİNLİ B. B., DELİL K., TÜRKYILMAZ A., SÖYLEMEZ M. A., Şimşek H., KARAKAYA T., ARSLAN ATEŞ E., GÜNEY A. İ., ATA P., ARMAN A.

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LIV. **Geç Tanı Almış Wıllıams Sendromlu Üç Olgunun Klinik Değerlendirmesi**
ARSLAN ATEŞ E., GEÇKİNLİ B. B., KARAKAYA T., TÜRKYILMAZ A., Şimşek H., SÖYLEMEZ M. A., DELİL K., ATA P., GÜNEY A. İ., ARMAN A.
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LV. **RAB3GAP1 Geninde Bilinen Bir Splice-Site Mutasyonunun Fonksiyonel Değerlendirmesi**
TÜRKYILMAZ A., SÖYLEMEZ M. A., ARSLAN ATEŞ E., Ergüner B., Şimşek H., KARAKAYA T., DELİL K., GEÇKİNLİ B. B., ATA P., GÜNEY A. İ., et al.
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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