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Education Information

Doctorate, Baskent University, Institute Of Health Sciences, Tıbbi Genetik (Dr), Turkey 2018 - Continues

Undergraduate, Anadolu University, Faculty Of Business Administration, Department Of Business, Turkey 2009 - 2012

Associate Degree, Anadolu University, Open Education Faculty, Department Of Management And Organization, Turkey 2007 - 2009

Associate Degree, Anadolu University, Open Education Faculty, Department Of Management And Organization, Turkey 2007 - 2009

Doctorate, Bursa Uludağ University, TIP FAKÜLTESİ, TEMEL TIP BİLİMLERİ, Turkey 1996 - 2002

Foreign Languages

German, B1 Intermediate

Italian, B1 Intermediate

English, C1 Advanced

Research Areas

Health Sciences, Natural Sciences, Engineering and Technology

Academic Titles / Tasks

Professor, Bursa Uludağ University, TIP FAKÜLTESİ, DAHİLİ TIP BİLİMLERİ, 2022 - Continues

Associate Professor, Bursa Uludağ University, TIP FAKÜLTESİ, DAHİLİ TIP BİLİMLERİ, 2013 - 2022

Academic and Administrative Experience

Head of Department, Bursa Uludağ University, TIP FAKÜLTESİ, 2018 - 2021

Head of Department, Bursa Uludağ University, TIP FAKÜLTESİ, DAHİLİ TIP BİLİMLERİ, 2017 - 2019

Courses

Translasyonel Tıp, Doctorate, 2017 - 2018

Tıbbi Genetik, Undergraduate, 2017 - 2018

Published journal articles indexed by SCI, SSCI, and AHCI

- I. **SARS-CoV-2 brainstem encephalitis in human inherited DBR1 deficiency.**
Chan Y., Lundberg V., Le Pen J., Yuan J., Lee D., Pinci F., Volpi S., Nakajima K., Bondet V., Åkesson S., et al.
The Journal of experimental medicine, vol.221, no.9, 2024 (SCI-Expanded)
- II. **Genotype and Phenotype Correlation of Patients with Osteogenesis Imperfecta**
Aliyeva L., Denkboy Öngen Y., Eren E., Sarisözen M. B., Alemdar A., Temel Ş. G., Özemri Sağ Ş.
JOURNAL OF MOLECULAR DIAGNOSTICS, vol.26, no.10, pp.1-20, 2024 (SCI-Expanded)
- III. **Pyruvate kinase deficiency in 29 Turkish patients with two novel intronic variants.**
Gök V., Leblebisatan G., Gürlek Gökçebay D., Güler S., Doğan M. E., Tuğ Bozdoğan S., Koca Yozgat A., Özcan A., Pekpak Şahinoğlu E., Tokgöz H., et al.
British journal of haematology, 2024 (SCI-Expanded)
- IV. **Alzheimer Disease Associated Loci: APOE Single Nucleotide Polymorphisms in Marmara Region**
Badee A., Dündar M. S., Örnek Ergüzeloğlu C., Ergoren M. C., Alemdar A., Özemri Sağ Ş., Temel Ş. G.
BIOMEDICINES, vol.12, no.5, pp.1-11, 2024 (SCI-Expanded)
- V. **Homozygous missense variants in YKT6 result in loss of function and are associated with developmental delay, with or without severe infantile liver disease and risk for hepatocellular carcinoma.**
Ma M., Ganapathi M., Zheng Y., Tan K., Kanca O., Bove K. E., Quintanilla N., Sag S. O., Temel S. G., LeDuc C. A., et al.
Genetics in medicine : official journal of the American College of Medical Genetics, no.-, pp.101125, 2024 (SCI-Expanded)
- VI. **Higher COVID-19 pneumonia risk associated with anti-IFN- α than with anti-IFN- ω auto-Abs in children.**
Bastard P., Gervais A., Taniguchi M., Saare L., Särekannu K., Le Voyer T., Philippot Q., Rosain J., Bizien L., Asano T., et al.
The Journal of experimental medicine, vol.221, no.2, 2024 (SCI-Expanded)
- VII. **Investigation of infectious droplet dispersion in a hospital examination room cooled by split-type air conditioner**
Yüce B. E., Kalay O. C., KARPAT F., ALEMDAR A., TEMEL Ş. G., Dilektaşlı A. G., Başkan E. B., ÖZAKIN C., COŞKUN B.
Journal of Environmental Health Science and Engineering, 2024 (SCI-Expanded)
- VIII. **Autoantibodies against type I IFNs in humans with alternative NF- κ B pathway deficiency.**
Le Voyer T., Parent A. V., Liu X., Cederholm A., Gervais A., Rosain J., Nguyen T., Perez Lorenzo M., Rackaityte E., Rinchai D., et al.
Nature, vol.623, no.7988, pp.803-813, 2023 (SCI-Expanded)
- IX. **From Death to Life/Back to the Future: Detailed Premorbid Clinical and Family History Can Save Lives and Address the Final Diagnosis in Sudden Unexplained Deaths With Negative Autopsy**
Türkgenç B., Baydar Ç. L., Akçay A., Deniz I., Ergoren M. Ç., Özemri Sağ Ş., Yakıcıer C., Temel Ş. G.
APPLIED IMMUNOHISTOCHEMISTRY AND MOLECULAR MORPHOLOGY, vol.11, pp.1-8, 2023 (SCI-Expanded)
- X. **A new line method; A direct test in spinal muscular atrophy screening for DBS**
Kubar A., Temel Ş. G., Ergören M. C., Hatırnaz Ng Ö., Özemri Sağ Ş., Alanay Y., Özbek U.
MOLECULAR GENETICS AND GENOMICS, no.6, pp.1-8, 2023 (SCI-Expanded)
- XI. **Human inherited complete STAT2 deficiency underlies inflammatory viral diseases**
Bucciol G., Moens L., Ogishi M., Rinchai D., Matuoizzo D., Momenilandi M., Kerrouche N., Cale C. M., Treffeisen E. R., Al Salamah M., et al.
JOURNAL OF CLINICAL INVESTIGATION, vol.133, no.12, 2023 (SCI-Expanded)
- XII. **PGT for structural chromosomal rearrangements in 300 couples reveals specific risk factors but an interchromosomal effect is unlikely**

Ogur C., Kahraman S., Grif D. K., Yapan C. C., Tufekci M. A., Cetinkaya M., TEMEL Ş. G., YILMAZ A.
REPRODUCTIVE BIOMEDICINE ONLINE, vol.46, no.4, pp.713-727, 2023 (SCI-Expanded)

- XIII. **Interfering with Interferons: A Critical Mechanism for Critical COVID-19 Pneumonia**
Su H. C., Jing H., Zhang J., Human Genetic Effort M. O. T. C., Temel Ş. G., Casanova J.
ANNUAL REVIEW OF IMMUNOLOGY, no.41, pp.561-585, 2023 (SCI-Expanded)
- XIV. **Inherited and acquired errors of type I interferon immunity govern susceptibility to COVID-19 and multisystem inflammatory syndrome in children**
Buccio G., Abel L., Al-Muhsen S., Aiuti A., Al-Mulla F., Andreakos E., Antonio N., Arias A. A., Trouillet-Assant S., Belot A., et al.
JOURNAL OF ALLERGY AND CLINICAL IMMUNOLOGY: IN PRACTICE, vol.151, no.4, pp.832-840, 2023 (SCI-Expanded)
- XV. **Contribution of genotypes in Prothrombin and Factor V Leiden to COVID-19 and disease severity in patients at high risk for hereditary thrombophilia.**
Kiraz A., Sezer O., Alemdar A., Canbek S., Duman N., Bisgin A., Cora T., Ruhi H. I., Ergoren M. C., Geçkinli B. B., et al.
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- XVI. **An Endocrinological Perspective on 22q11.2 Deletion Syndrome: A Single-center Experience.**
Denkboy Ongen Y., Ozemri Sag S., Temel Ş. G., Eren E.
Journal of clinical research in pediatric endocrinology, vol.15, pp.285-292, 2023 (SCI-Expanded)
- XVII. **Autoantibodies against type I IFNs in patients with critical influenza pneumonia.**
Zhang Q., Pizzorno A., Miorin L., Bastard P., Gervais A., Le Voyer T., Bizien L., Manry J., Rosain J., Philippot Q., et al.
The Journal of experimental medicine, vol.219, no.11, 2022 (SCI-Expanded)
- XVIII. **"Analysis of ACE2 and TMPRSS2 coding variants as a risk factor for SARS-CoV-2 from 946 whole exome sequencing data in the Turkish Population.**
Duman N., Tuncel G., Bisgin A., Bozdogan S. T., Sag S. O., Gul S., Kiraz A., Balta B., Erdogan M., Uyanik B., et al.
Journal of medical virology, vol.94, no.11, pp.5225-5243, 2022 (SCI-Expanded)
- XIX. **Decoding the Human Genetic and Immunological Basis of COVID-19 mRNA Vaccine-Induced Myocarditis.**
Bolze A., Mogensen T. H., Zhang S., Abel L., Andreakos E., Arkin L. M., Borghesi A., Brodin P., Hagin D., Novelli G., et al.
Journal of clinical immunology, vol.42, pp.1354-1359, 2022 (SCI-Expanded)
- XX. **Characterization of a Novel Frameshift Mutation Within the TRPS1 Gene Causing Trichorhinophalangeal Syndrome Type 1 in a Kindred Cypriot Family.**
Ergoren M. C., Akcan N., Manara E., Paolacci S., Fahrioglu U., Betmezoglu M., Bundak R., Mocan G., Temel S. G., Bertelli M.
Applied immunohistochemistry & molecular morphology : AIMM, vol.30, no.9, pp.635-639, 2022 (SCI-Expanded)
- XXI. **The expression profile of WNT/beta-catenin signalling genes in human oocytes obtained from polycystic ovarian syndrome (PCOS) patients**
Ismail A. B., Naji M. ' S., Tuncel G., Ozbakir B., TEMEL Ş. G., Tulay P., Mocan G., Ergoren M. C., Nebih I.
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- XXII. **Increased radiosensitivity and impaired DNA repair in patients with STAT3-LOF and ZNF341 deficiency, potentially contributing to malignant transformations.**
Cekic Ş., Huriyet H., Hortoglu M., Kasap N., Ozen A., Karakoc-Aydiner E., Metin A., Ocakoglu G., Demiroz Abakay C., Temel Ş. G., et al.
Clinical and experimental immunology, vol.209, no.1, pp.83-89, 2022 (SCI-Expanded)
- XXIII. **Germline landscape of BRCA by 7-site collaborations as a BRCA consortium in Turkey.**
Bisgin A., Sag S. O., Dogan M. E., Yildirim M. S., Gumus A. A., Akkus N., Balasar O., Durmaz C. D., Ersoz R., Altiner S., et al.
Breast (Edinburgh, Scotland), vol.65, no.-, pp.15-22, 2022 (SCI-Expanded)
- XXIV. **Clinical and molecular evaluation of MEFV gene variants in the Turkish population: a study by the National Genetics Consortium.**
Dundar M., Fahrioglu U., Yildiz S. H., Bakir-Gungor B., Temel Ş. G., Akin H., Artan S., Cora T., Sahin F. I., Dursun A., et

al.

Functional & integrative genomics, vol.22, no.3, pp.291-315, 2022 (SCI-Expanded)

- XXV. **Novel homozygous missense mutation in NARS1 gene: A new neurodevelopmental disorder with microcephaly**
TEMEL Ş. G., ÖZEMRİ SAĞ Ş., EREN E., Deniz E.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.SUPPL 1, pp.352, 2022 (SCI-Expanded)
- XXVI. **Characterizing a de novo TRIO gene variant as a likely cause of autosomal dominant Intellectual developmental disorder type 63 with macrocephaly**
Tekguc D. C., Tuncel G., Karanlik S., Koreken N., TEMEL Ş. G., Ergoren M. C.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.SUPPL 1, pp.255, 2022 (SCI-Expanded)
- XXVII. **Structural analysis of M1AP variants associated with severely impaired spermatogenesis causing male infertility**
Gerlevik U., Ergoren M. C., SEZERMAN O. U., TEMEL Ş. G.
PEERJ, vol.10, 2022 (SCI-Expanded)
- XXVIII. **Consistency of variant interpretations among bioinformaticians and clinical geneticists in hereditary cancer panels.**
Agaoglu N. B., Unal B., Akgun Dogan O., Kanev M. O., Zolfagharian P., Ozemri Sag S., Temel Ş. G., Doganay L.
European journal of human genetics : EJHG, vol.30, pp.378-383, 2022 (SCI-Expanded)
- XXIX. **Determining T and B Cell development by TREC/KREC analysis in primary immunodeficiency patients and healthy controls**
Senturk G., Ng Y. Y., Eltan S. B., Baser D., Ogulur I., ALTINDİREK D., Firtina S., YILMAZ H., Kocamis B., KIYKIM A., et al.
SCANDINAVIAN JOURNAL OF IMMUNOLOGY, vol.95, no.3, 2022 (SCI-Expanded)
- XXX. **Birt-Hogg-Dube Syndrome: Diagnostic Journey of Three Cases from Skin to Gene**
Hasal E., Baskan E. B., Gul S., Dilektasli A. G., Sag S. O., Adird Ş., TEMEL Ş. G.
ANNALS OF DERMATOLOGY, vol.34, no.1, pp.66-71, 2022 (SCI-Expanded)
- XXXI. **Nanobubble Ozone Stored in Hyaluronic Acid Decorated Liposomes: Antibacterial, Anti-SARS-CoV-2 Effect and Biocompatibility Tests**
Sabanci A. U., Alkan P. E., Mujde C., Polat H. U., Erguzeloglu C. O., BİŞGİN A., ÖZAKIN C., TEMEL Ş. G.
INTERNATIONAL JOURNAL OF NANOMEDICINE, vol.17, pp.351-379, 2022 (SCI-Expanded)
- XXXII. **Psoriasis and 5HT-R2C Gene Polymorphism: Association between Clinical, Demographic and Therapeutic Parameters in the Turkish Population.**
Temel Ş. G., Yazici S., Yilmaz İ., Tosun Ö., Cerkez Ergoren M., Bulbul Baskan E., Oral B., Aydogan K.
Acta dermatovenerologica Croatica : ADC, vol.29, no.3, pp.121-126, 2021 (SCI-Expanded)
- XXXIII. **BRCA Variations Risk Assessment in Breast Cancers Using Different Artificial Intelligence Models**
Senturk N., Tuncel G., Dogan B., Aliyeva L., Dundar M. S., Ozemri Sag S., Mocan G., Temel S. G., Dundar M., Ergoren M. C.
GENES, vol.12, no.11, 2021 (SCI-Expanded)
- XXXIV. **Associations of the ITGB3 gene rs5918T > C and the APOA1 gene rs1799837C > T markers with serum lipid metabolism in coronary artery disease patients**
Conkbayir C., Ergoren M., Cobanogullari H., Balcioglu O., Abras I., Eminsel T., Oztas D., Ugurlucan M., TEMEL Ş. G.
EUROPEAN HEART JOURNAL, vol.42, pp.3192, 2021 (SCI-Expanded)
- XXXV. **Identification of a Novel De Novo COMP Gene Variant as a Likely Cause of Pseudoachondroplasia.**
Tuncel G., Akcan N., Gul S., Sag S. O., Bundak R., Mocan G., Temel Ş. G., Ergoren M. C.
Applied immunohistochemistry & molecular morphology : AIMM, vol.29, pp.546-550, 2021 (SCI-Expanded)
- XXXVI. **Mitochondrial estrogen receptors alter mitochondrial priming and response to endocrine therapy in breast cancer cells.**
Karakas B., Aka Y., Giray A., Temel Ş. G., Acikbas U., Basaga H., Gul O., Kutuk O.
Cell death discovery, vol.7, pp.189, 2021 (SCI-Expanded)
- XXXVII. **Natural selection at work? Vitamin D deficiency rates and rising health problems in young Turkish Cypriot professionals.**
Kandemiş E., Tuncel G., Fahrioğlu U., Temel Ş. G., Mocan G., Ergören M. Ç.

Central European journal of public health, vol.29, pp.130-133, 2021 (SCI-Expanded)

- XXXVIII. **MUTATION STATUS AND IMMUNOHISTOCHEMICAL CORRELATION OF EGFR MUTATIONS IN GASTROINTESTINAL STROMAL TUMORS**
Ozkayalar H., Ergoren M. C., Tuncel G., Kurt S., Cevik E., Sag O. S., Ozguven Y. B., Kabukcuoglu F., Mocan G., TEMEL Ş. G.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.24, no.1, pp.67-71, 2021 (SCI-Expanded)
- XXXIX. **A Homozygous Synonymous Variant Likely Cause of Severe Ciliopathy Phenotype**
Tuncel G., Kaymakamzade B., Engindereli Y., TEMEL Ş. G., Ergoren M. C.
GENES, vol.12, no.6, 2021 (SCI-Expanded)
- XL. **Evaluation of bioaccessibility and functional properties of kombucha beverages fortified with different medicinal plant extracts**
Tamer C. E., Temel Ş. G., Suna S., Karabacak A., Ozcan T., Ersan L. Y., Kaya B. T., Copur Ö. U.
TURKISH JOURNAL OF AGRICULTURE AND FORESTRY, vol.45, no.1, pp.13-32, 2021 (SCI-Expanded)
- XLI. **Targeted High-Throughput Sequencing Analysis Results of Osteogenesis Imperfecta Patients from Different Regions of Turkey.**
DEMİR S., YALÇINTEPE S., ATLI E. İ., Sanri A., Yildirim R., TÜTÜNCÜLER F., Celik M., ATLI E., ÖZEMİR SAĞ Ş., Eker D., et al.
Genetic testing and molecular biomarkers, vol.25, no.1, pp.59-67, 2021 (SCI-Expanded)
- XLII. **The importance of multiple gene analysis for diagnosis and differential diagnosis in charcot marie tooth disease**
YALÇINTEPE S., GÜRKAN H., DEMİR S., ÖZEMİR SAĞ Ş., ATLI E. İ., ATLI E., EKER D., TEMEL Ş. G.
Turkish Neurosurgery, vol.31, pp.888-895, 2021 (SCI-Expanded)
- XLIII. **Functional coding/non-coding variants in EGFR, ROS1 and ALK genes and their role in liquid biopsy as a personalized therapy.**
Ergoren M., Cobanogulları H., Temel Ş. G., Mocan G.
Critical reviews in oncology/hematology, vol.156, pp.103113, 2020 (SCI-Expanded)
- XLIV. **CC2D1A AS A NOVEL CILIOPATHY GENE**
Sakin I., Tuncel G., Sag S. O., KAPLAN O. İ., Khokha M. K., Ergoren M. C., Deniz E., TEMEL Ş. G.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, pp.454, 2020 (SCI-Expanded)
- XLV. **Biallelic mutations in M1AP are associated with meiotic arrest, severely impaired spermatogenesis and male infertility**
Friedrich C., TEMEL Ş. G., Nagirnaja L., Oud M. S., Lopes A. M., van der Heijden G. W., Heald J., Rotte N., Wistuba J., Woeste M., et al.
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- XLVI. **Identification of unsolved rare genetic cases of North Cyprus**
Ergoren M. C., Manara E., Paolacci S., Tuncel G., TEMEL Ş. G., Mocan G., DÜNDAR M., Bertelli M.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, pp.944, 2020 (SCI-Expanded)
- XLVII. **Unique combination and in silico modeling of biallelic POLR3A variants as a cause of Wiedemann-Rautenstrauch syndrome**
TEMEL Ş. G., Ergoren M. C., Manara E., Paolacci S., Tuncel G., Gul S., Bertelli M.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, no.12, pp.1675-1680, 2020 (SCI-Expanded)
- XLVIII. **Diagnostic Efficiency of Clinical Exome Solution Panel in patients with Hearing loss/Hereditary Deafness by using Next Generation Sequencing**
Temel Ş. G., Alemdar A., Yilmaz M., Aliyeva L., Sag S. O.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, pp.190-191, 2020 (SCI-Expanded)
- XLIX. **Characterization and in silico analyses of the BRCA1/2 variants identified in individuals with personal and/or family history of BRCA-related cancers**
Pirim D., Kaya N., Yildirim E., Sag S., Temel Ş. G.
INTERNATIONAL JOURNAL OF BIOLOGICAL MACROMOLECULES, vol.162, pp.1166-1177, 2020 (SCI-Expanded)
- L. **A rare case of fructose-1,6-bisphosphatase deficiency: a delayed diagnosis story**
Ergoren M. C., Tuncel G., ÖZEMİR SAĞ Ş., TEMEL Ş. G.

TURKISH JOURNAL OF BIOCHEMISTRY-TURK BIYOKIMYA DERGISI, vol.45, no.5, pp.613-616, 2020 (SCI-Expanded)

- LI. **Bi-allelic Mutations in M1AP Are a Frequent Cause of Meiotic Arrest and Severely Impaired Spermatogenesis Leading to Male Infertility**
Wyrwoll M. J., TEMEL Ş. G., Nagirnaja L., Oud M. S., Lopes A. M., van der Heijden G. W., Heald J. S., Rotte N., Wistuba J., Woeste M., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.107, no.2, pp.342-351, 2020 (SCI-Expanded)
- LII. **Strong Association between Serotonin Transporter 5-HTTVNTR Variant and Psychoactive Substance (Nicotine) Use in the Turkish Cypriot Population**
Kandemis E., Tuncel G., Asut O., TEMEL Ş. G., Ergoren M. C.
CURRENT DRUG METABOLISM, vol.21, no.6, pp.466-470, 2020 (SCI-Expanded)
- LIII. **European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death**
Fellmann F., van El C. G., Charron P., Michaud K., Howard H. C., Boers S. N., Clarke A. J., Duguet A., Forzano F., Kaufenstein S., et al.
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- LIV. **A novel homozygous nonsense mutation in CAST associated with PLACK syndrome**
TEMEL Ş. G., Karakas B., Seker U., Turkgenç B., Zorlu O., Saricaoğlu H., Oğur C., Kutuk O., Kellsell D. P., Yakicier M. C.
CELL AND TISSUE RESEARCH, vol.378, no.2, pp.267-277, 2019 (SCI-Expanded)
- LV. **Identification and characterization of a novel FBN1 gene variant in an extended family with variable clinical phenotype of Marfan syndrome**
Ergoren M. C., Turkgenç B., Terali K., Rodoplu O., Verstraeten A., Van Laer L., Mocan G., Loeys B., Tetik O., TEMEL Ş. G.
CONNECTIVE TISSUE RESEARCH, vol.60, no.2, pp.146-154, 2019 (SCI-Expanded)
- LVI. **Investigation of KCNQI polymorphisms as biomarkers for cardiovascular diseases in the Turkish Cypriots for establishing preventative medical measures**
Tulay P., Temel Ş. G., Ergoren M. C.
INTERNATIONAL JOURNAL OF BIOLOGICAL MACROMOLECULES, vol.124, pp.537-540, 2019 (SCI-Expanded)
- LVII. **The use of ACE INDEL polymorphism as a biomarker of coronary artery Chock disease (CAD) in humans with Mediterranean-style diet**
TEMEL Ş. G., Ergoren M. C., Yılmaz I., ORAL H. B.
INTERNATIONAL JOURNAL OF BIOLOGICAL MACROMOLECULES, vol.123, pp.576-580, 2019 (SCI-Expanded)
- LVIII. **The association between the chromosome 9p21 CDKN2B-AS1 gene variants and the lipid metabolism: A pre-diagnostic biomarker for coronary artery disease**
TEMEL Ş. G., Ergoren M. C.
ANATOLIAN JOURNAL OF CARDIOLOGY, vol.21, no.1, pp.31-38, 2019 (SCI-Expanded)
- LIX. **Arterial tortuosity syndrome: 40 new families and literature review.**
Beyens A., Albuissou J., Boel A., Al-Essa M., Al-Manea W., Bonnet D., Bostan Ö. M., Boute O., Busa T., Canham N., et al.
Genetics in medicine : official journal of the American College of Medical Genetics, vol.20, no.10, pp.1236-1245, 2018 (SCI-Expanded)
- LX. **STUB1 polyadenylation signal variant AACAAA does not affect polyadenylation but decreases STUB1 translation causing SCAR16**
Turkgenç B., Sanlidag B., Eker A., Giray A., Kutuk O., Yakicier C., TOLUN A., TEMEL Ş. G.
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- LXI. **Phenotype does not necessarily follow genotype: Identification of an incompletely penetrant novel POLR1D variant as a likely cause of Treacher Collins syndrome**
Sah H., Sanlidag B., Manara E., Terali K., Paolacci S., Mocan G., Temel Ş. G., Dirik E., Bertelli M., Ergoren M. C.
FEBS OPEN BIO, vol.8, pp.143, 2018 (SCI-Expanded)
- LXII. **Characterization and in silico modelling of bi-allelic POLR3A mutations as a cause of Wiedemann-Rautenstrauch syndrome**
Betmezoglu M., Terali K., Manara E., Mocan G., Temel Ş. G., Bertelli M., Ergoren M. C.
FEBS OPEN BIO, vol.8, pp.143-144, 2018 (SCI-Expanded)

- LXIII. **Homozygous, and compound heterozygous mutation in 3 Turkish family with Jervell and Lange-Nielsen syndrome: case reports**
Uysal F., Turkgenç B., Toksoy G., BOSTAN Ö. M., Evke E., Uyguner O., Yakicier C., Kayserili H., ÇİL E., Temel Ş. G.
BMC MEDICAL GENETICS, vol.18, 2017 (SCI-Expanded)
- LXIV. **Biphasic ROS production, p53 and BIK dictate the mode of cell death in response to DNA damage in colon cancer cells**
Kutuk O., Aytan N., Karakas B., Kurt A. G., Acikbas U., TEMEL Ş. G., Basaga H.
PLOS ONE, vol.12, no.8, 2017 (SCI-Expanded)
- LXV. **A Novel TBX19 Gene Mutation in a Case of Congenital Isolated Adrenocorticotrophic Hormone Deficiency Presenting with Recurrent Respiratory Tract Infections**
Akcan N., SERAKINCI N., Turkgenç B., Bundak R., Bahceciler N., Temel Ş. G.
FRONTIERS IN ENDOCRINOLOGY, vol.8, 2017 (SCI-Expanded)
- LXVI. **Letter to the editor regarding the article "A case of hypertrophic and dilated cardiomyopathic sudden cardiac death: de novo mutation in TTN and SGCD genes"**
Ergoren M. C., Temel S. G.
ANATOLIAN JOURNAL OF CARDIOLOGY, vol.17, no.1, pp.76-77, 2017 (SCI-Expanded)
- LXVII. **Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5**
McMillin M. J., Beck A. E., Chong J. X., Shively K. M., Buckingham K. J., Gildersleeve H. I. S., Aracena M. I., Aylsworth A. S., Bitoun P., Carey J. C., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.94, no.5, pp.734-744, 2014 (SCI-Expanded)
- LXVIII. **Duplication of SOX9 is not a common cause of 46,XX testicular or 46,XX ovotesticular DSD**
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Metrics

Publication: 195

Citation (WoS): 830

Citation (Scopus): 884

H-Index (WoS): 14

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