

# Prof. ŐEHİME GÜLSÜN TEMEL

## Personal Information

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## International Researcher IDs

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## Education Information

Doctorate, Baskent University, Institute Of Health Sciences, Tıbbi Genetik (Dr), Turkey 2018 - 2024

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, Tıp Fakültesi, Temel Tıp Bilimleri, 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. **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, vol.22, no.2, pp.471-482, 2024 (SCI-Expanded)
- II. **Identifying Hub Genes and Metabolic Pathways in Collagen VI-Related Dystrophies: A Roadmap to Therapeutic Intervention**  
Ceyhan A. B., Kaynar A., Altay O., Zang C., Temel Ş. G., Türkez H., Mardinoglu A.  
BIOMOLECULES, no.14, pp.1-20, 2024 (SCI-Expanded)
- III. **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)
- IV. **Investigation of the Release of Growth Factors from Apheresis Platelet Concentrate (APC) Loaded Three Layered Composite Nanofiber Surface**  
Yılmaz H., Aras Gül C., Karaçay M., Altuntuğ Cesur M. İ., Karaca E., Temel Ş. G., Bülbül Başkan E., Oral H. B., Kaya E.  
TEKSTİL VE KONFEKSİYON, vol.34, no.3, pp.244-252, 2024 (SCI-Expanded)
- V. **The impact and future of artificial intelligence in medical genetics and molecular medicine: an ongoing revolution.**  
Ozcelik F., Dundar M. S., Yildirim A. B., Henehan G., Vicente O., Sánchez-Alcázar J. A., Gokce N., Yildirim D. T., Bingol N. N., Karanfilska D. P., et al.  
Functional & integrative genomics, vol.24, no.4, pp.138, 2024 (SCI-Expanded)
- VI. **CC2D1A causes ciliopathy, intellectual disability, heterotaxy, renal dysplasia, and abnormal CSF flow**  
Kim A. H., Sakin I., Vivano S., Tuncel G., Aguilera S. M., Goles G., Jeffries L., Ji W., Lakhani S. A., Ceylan Köse C., et al.  
LIFE SCIENCE ALLIANCE, vol.7, no.10, pp.1-10, 2024 (SCI-Expanded)
- VII. **Genotype and Phenotype Correlation of Patients with Osteogenesis Imperfecta**  
Aliyeva L., Denkboy Öngen Y., Eren E., Sarısözen M. B., Alemdar A., Temel Ş. G., Özemri Sağ Ş.  
JOURNAL OF MOLECULAR DIAGNOSTICS, vol.26, no.10, pp.1-20, 2024 (SCI-Expanded)
- VIII. **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)
- IX. **Alzheimer Disease Associated Loci: APOE Single Nucleotide Polymorphisms in Marmara Region**  
Badee A., Dünder 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)
- X. **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)
- XI. **Higher COVID-19 pneumonia risk associated with anti-IFN- $\alpha$  than with anti-IFN- $\omega$  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.  
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- XII. **Autoantibodies against type I IFNs in humans with alternative NF- $\kappa$ 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)

- XIII. **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**  
Türkgenc 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)
- XIV. **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, no.3, pp.285-292, 2023 (SCI-Expanded)
- XV. **A new line method; A direct test in spinal muscular atrophy screening for DBS**  
Kubar A., Temel Ş. G., Ergören M. C., Hatrnaz Ng Ö., Özemri Sağ Ş., Alanay Y., Özbek U.  
MOLECULAR GENETICS AND GENOMICS, no.6, pp.1-8, 2023 (SCI-Expanded)
- XVI. **Human inherited complete STAT2 deficiency underlies inflammatory viral diseases**  
Bucciol G., Moens L., Ogishi M., Rinchai D., Matuozzo 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)
- XVII. **Interfering with Interferons: A Critical Mechanism for Critical COVID-19 Pneumonia**  
Su H. C., Jing H., Zhang ., Human Genetic Effort M. O. T. C., Temel Ş. G., Casanova J.  
ANNUAL REVIEW OF IMMUNOLOGY, no.41, pp.561-585, 2023 (SCI-Expanded)
- XVIII. **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)
- XIX. **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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- XX. **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.  
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- XXI. **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.  
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- XXII. **"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.  
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- XXIII. **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.  
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- XXIV. **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)

- XXV. **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.  
ZYGOTE, vol.30, no.4, pp.536-542, 2022 (SCI-Expanded)
- XXVI. **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)
- XXVII. **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)
- XXVIII. **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)
- XXIX. **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)
- XXX. **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)
- XXXI. **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)
- XXXII. **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)
- XXXIII. **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)
- XXXIV. **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)
- XXXV. **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)
- XXXVI. **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)
- XXXVII. **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)
- XXXVIII. **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)
- XXXIX. **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)
- XL. **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)
- XLI. **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)
- XLII. **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)
- XLIII. **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)
- XLIV. **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.  
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- XLV. **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)
- XLVI. **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)
- XLVII. **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)
- XLVIII. **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.  
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, pp.1002-1003, 2020 (SCI-Expanded)
- XLIX. **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)

- L. **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)
- LI. **Unique combination and in silico modeling of biallelic POLR3A variants as a cause of Wiedemann-Rautenstrauch syndrome**  
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EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, no.12, pp.1675-1680, 2020 (SCI-Expanded)
- LII. **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.  
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- LIII. **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., Yıldırım E., Sag S., Temel Ş. G.  
INTERNATIONAL JOURNAL OF BIOLOGICAL MACROMOLECULES, vol.162, pp.1166-1177, 2020 (SCI-Expanded)
- LIV. **A rare case of fructose-1,6-bisphosphatase deficiency: a delayed diagnosis story**  
Ergoren M. C., Tuncel G., ÖZEMRİ SAĞ Ş., TEMEL Ş. G.  
TURKISH JOURNAL OF BIOCHEMISTRY-TURK BIYOKIMYA DERGISI, vol.45, no.5, pp.613-616, 2020 (SCI-Expanded)
- LV. **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)
- LVI. **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)
- LVII. **European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death**  
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EUROPEAN JOURNAL OF HUMAN GENETICS, vol.27, no.12, pp.1763-1773, 2019 (SCI-Expanded)
- LVIII. **A novel homozygous nonsense mutation in *CAST* associated with PLACK syndrome**  
TEMEL Ş. G., Karakas B., Seker Ü., Turkgenc B., Zorlu O., Saricaoğlu H., Ogur C., Kutuk O., Kelsell D. P., Yakicier M. C.  
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- LIX. **Identification and characterization of a novel FBN1 gene variant in an extended family with variable clinical phenotype of Marfan syndrome**  
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- LX. **Investigation of KCNQ1 polymorphisms as biomarkers for cardiovascular diseases in the Turkish Cypriots for establishing preventative medical measures**  
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INTERNATIONAL JOURNAL OF BIOLOGICAL MACROMOLECULES, vol.124, pp.537-540, 2019 (SCI-Expanded)
- LXI. **The use of ACE INDEL polymorphism as a biomarker of coronary artery Chock disease (CAD) in humans with Mediterranean-style diet**  
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INTERNATIONAL JOURNAL OF BIOLOGICAL MACROMOLECULES, vol.123, pp.576-580, 2019 (SCI-Expanded)
- LXII. **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)

- LXIII. **Arterial tortuosity syndrome: 40 new families and literature review.**  
Beyens A., Albuisson 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,  
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- LXIV. **STUB1 polyadenylation signal variant AACAAA does not affect polyadenylation but decreases STUB1 translation causing SCAR16**  
Turkgenc B., Sanlidag B., Eker A., Giray A., Kutuk O., Yakicier C., TOLUN A., TEMEL Ş. G.  
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- LXV. **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.  
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- LXI. **GENETICS OF HEREDITARY ARRHYTHMIAS**  
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## Patent

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## Tasks In Event Organizations

Dündar M., Özkul Y., Temel Ş. G., Kiraz A., Akalın H., 8. Erciyes Tıp Tıbbi Genetik Kongresi, Scientific Congress, Kayseri, Turkey, Eylül 2023

## Metrics

Publication: 201

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## Congress and Symposium Activities

EGE ENDOKRİN HASTALIKLARI VE GENETİK SEMPOZYUMU, Session Moderator, İzmir, Turkey, 2021

## Awards

Yassı E., Temel Ş. G., PullExo Klinik ve Günlük Kullanım Amaçlı Mobil El Dış İskeleti, Teknofest, September 2023

Doğan B., Özemri Sağ Ş., Temel Ş. G., En İyi 2. Poster Ödülü , 1. Bursa Uluslararası Katılımlı Genetik Günleri

Dermatogenetik Sempozyumu, January 2020