

## Assoc. Prof. ZİHNİ EKİM TAŞKIRAN

### Personal Information

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### International Researcher IDs

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### Education Information

Doctorate, Hacettepe University, Sağlık Bilimleri Enstitüsü, Tıbbi Biyoloji, Turkey 2008 - 2012

Postgraduate, Hacettepe University, Sağlık Bilimleri Enstitüsü, Tıbbi Biyoloji, Turkey 2006 - 2007

Undergraduate, Hacettepe University, Fen Fakültesi, Biyoloji Bölümü, Turkey 2001 - 2005

### Foreign Languages

English, C1 Advanced

### Research Areas

Medicine, Medical Biology, Internal Medicine Sciences, Medical Genetics, Health Sciences, Fundamental Medical Sciences

### Academic Titles / Tasks

Assistant Professor, Hacettepe University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2018 - Continues

Assistant Professor, Hacettepe University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2014 - 2018

Expert, Hacettepe University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2012 - 2014

### Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Neonatal ichthyosis-sclerosing cholangitis syndrome: report of a novel mutation and a review of the literature**  
Demir E., TUNA KIRSAÇLIOĞLU C., SALTİK TEMİZEL İ. N., ÜREL DEMİR G., KARAOSMANOĞLU B., Taşkıran E. Z., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., KULOĞLU Z., KANSU TANCA A.  
Clinical Dysmorphology, vol.32, no.2, pp.88-91, 2023 (SCI-Expanded)
- II. **A novel biallelic CRIPT variant in a patient with short stature, microcephaly, and distinctive facial features**  
AKALIN A., ŞİMŞEK KİPER P. Ö., Taşkıran E. Z., KARAOSMANOĞLU B., ÜTİNE G. E., BODUROĞLU O. K.  
American Journal of Medical Genetics, Part A, vol.191, no.4, pp.1119-1127, 2023 (SCI-Expanded)
- III. **Mutated Transcripts of ZEB2 Do Not Undergo Nonsense-Mediated Decay in Mowat-Wilson Syndrome**

Güleray Lafci N., KARAOSMANOĞLU B., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E.

Molecular Syndromology, 2023 (SCI-Expanded)

- IV. **Typical Face, Developmental Delay, and Hearing Loss in a Patient with 3M Syndrome: The Co-Occurrence of Two Rare Conditions**  
AKALIN A., Simsek-Kiper P. O., Taskiran E., ÜTİNE G. E., BODUROĞLU O. K.  
MOLECULAR SYNDROMOLOGY, vol.13, no.6, pp.537-542, 2023 (SCI-Expanded)
- V. **A lethal and rare cause of arthrogyriposis: Glyt1 encephalopathy**  
Dasar T., ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., ÇAĞAN M., ÖZYÜNCÜ Ö., DEREN Ö., ÜTİNE G. E., GÜÇER K. Ş., BODUROĞLU O. K.  
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.65, no.12, 2022 (SCI-Expanded)
- VI. **Biallelic loss-of-function variants in EXOC6B are associated with impaired primary ciliogenesis and cause spondylo-epi-metaphyseal dysplasia with joint laxity type 3**  
ŞİMŞEK KİPER P. Ö., Jacob P., Upadhyai P., TAŞKIRAN Z. E., Guleria V. S., KARAOSMANOĞLU B., İMREN G., GÖÇMEN R., Bhavani G. S., Kausthubham N., et al.  
HUMAN MUTATION, vol.43, no.12, pp.2116-2129, 2022 (SCI-Expanded)
- VII. **Effector Th1 cells under PD-1 and CTLA-4 checkpoint blockade abrogate the upregulation of multiple inhibitory receptors and by-pass exhaustion**  
Horzum U., Yanik H., Taskiran E. Z., Esendagli G.  
IMMUNOLOGY, vol.167, no.4, pp.640-650, 2022 (SCI-Expanded)
- VIII. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**  
Güleray N., Koşukcu C., Oğuz S., Ürel Demir G., Taşkıran E. Z., Kiper P. Ö. Ş., Utine G. E., Alanay Y., Boduroğlu K., Alikasıfoğlu M.  
CLEFT PALATE-CRANIOFACIAL JOURNAL, vol.59, no.9, pp.1114-1124, 2022 (SCI-Expanded)
- IX. **Identification of a shared genetic risk locus for Kawasaki disease and immunoglobulin A vasculitis by a cross-phenotype meta-analysis**  
Carmona E. G., Garcia-Gimenez J. A., Lopez-Mejias R., Khor C. C., Lee J., Taskiran E., ÖZEN S., Hocevar A., Liu L., Gorenjak M., et al.  
RHEUMATOLOGY, vol.61, no.3, pp.1204-1210, 2022 (SCI-Expanded)
- X. **De novo cloning and functional characterization of potassium channel genes and proteins in the crayfish *Astacus leptodactylus* (Eschscholtz, 1823) (Decapoda: Astacidea: Astacidae)**  
ERGİN B., SAĞLAM B., TAŞKIRAN Z. E., BAŞTUĞ T., PURALI N.  
JOURNAL OF CRUSTACEAN BIOLOGY, vol.42, no.1, 2022 (SCI-Expanded)
- XI. **Small cell lung cancer stem cells display mesenchymal properties and exploit immune checkpoint pathways in activated cytotoxic T lymphocytes**  
KURŞUNEL M. A., Taskiran E. Z., Tavukcuoglu E., Yanik H., Demirag F., KARAOSMANOĞLU B., Ozbay F. G., ÜNER A., Esendagli D., Kizilgoz D., et al.  
CANCER IMMUNOLOGY IMMUNOTHERAPY, vol.71, no.2, pp.445-459, 2022 (SCI-Expanded)
- XII. **Biallelic ITGB4 variants in familial pyloric atresia without epidermolysis bullosa: Report of two families with five siblings**  
SOYER T., KARAOSMANOĞLU B., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., KARNAK İ., BODUROĞLU O. K., ÜTİNE G. E.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.11, pp.3427-3432, 2021 (SCI-Expanded)
- XIII. **Spondyloepimetaphyseal dysplasia EXTL3-deficient type: Long-term follow-up and review of the literature**  
AKALIN A., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., Utine E., ALANAY Y., Ozcelik U., BODUROĞLU O. K.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.10, pp.3104-3110, 2021 (SCI-Expanded)
- XIV. **Kohlschütter-Tonz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity**  
AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., Taskiran E., Schossig A., ÜTİNE G. E., Zschocke J., BODUROĞLU O. K.  
JOURNAL OF CHILD NEUROLOGY, vol.36, no.10, pp.816-822, 2021 (SCI-Expanded)
- XV. **Diagnostic yield of whole-exome sequencing in non-syndromic intellectual disability**  
Taşkıran Z. E., Karaosmanoglu B., Kosukcu C., Urel-Demir G., Akgun-Dogan O., Simsek-Kiper P. O., Alikasifoglu M., Boduroğlu O. K., Utine G. E.

JOURNAL OF INTELLECTUAL DISABILITY RESEARCH, vol.65, no.6, pp.577-588, 2021 (SSCI)

- XVI. **Further expanding the mutational spectrum of brain abnormalities, neurodegeneration, and dysosteosclerosis: A rare disorder with neurologic regression and skeletal features**  
KINDIŞ E., ŞİMŞEK KİPER P. Ö., KOŞUKCU C., TAŞKIRAN Z. E., GÖÇMEN R., Utine E., Haliloglu G., BODUROĞLU O. K., ALİKAŞİFOĞLU M.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, pp.1888-1896, 2021 (SCI-Expanded)
- XVII. **FARS1-related disorders caused by bi-allelic mutations in cytosolic phenylalanyl-tRNA synthetase genes: Look beyond the lungs!**  
Schuch L. A., Forstner M., Rapp C. K., Li Y., Smith D. E. C., Mendes M., Delhommel F., Sattler M., EMİRALİOĞLU N., Taskiran E. Z., et al.  
CLINICAL GENETICS, vol.99, no.6, pp.789-801, 2021 (SCI-Expanded)
- XVIII. **Expanding the phenotypic spectrum of TNFRSF11A-associated dysosteosclerosis: a case with intracranial extramedullary hematopoiesis**  
Xue J., Simsek-Kiper P. O., ÜTİNE G. E., Yan L., Wang Z., Taskiran E. Z., KARAOSMANOĞLU B., İMREN G., GÖÇMEN R., Nishimura G., et al.  
JOURNAL OF HUMAN GENETICS, vol.66, no.6, pp.607-611, 2021 (SCI-Expanded)
- XIX. **Proerythroblast Cells of Diamond-Blackfan Anemia Patients With RPS19 and CECR1 Mutations Have Similar Transcriptomic Signature**  
KARAOSMANOĞLU B., KURŞUNEL M. A., ÇETİNKAYA F. D., GÜMRÜK F., ESENDAĞLI G., ÜNAL Ş., Taskiran E. Z.  
FRONTIERS IN PHYSIOLOGY, vol.12, 2021 (SCI-Expanded)
- XX. **Two Siblings with Kaufman Oculocerebrofacial Syndrome Resembling Oculoauriculovertebral Spectrum**  
ÜREL DEMİR G., Aydin B., KARAOSMANOĞLU B., AKGÜN DOĞAN Ö., Taskiran E. Z., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.  
MOLECULAR SYNDROMOLOGY, vol.12, no.2, pp.106-111, 2021 (SCI-Expanded)
- XXI. **Whole exome sequencing in unclassified autoinflammatory diseases: more monogenic diseases in the pipeline?**  
Kosukcu C., Taskiran E., Batu E., Sag E., Bilginer Y., Alikasifoglu M., Ozen S.  
RHEUMATOLOGY, vol.60, no.2, pp.607-616, 2021 (SCI-Expanded)
- XXII. **CD66b(+) monocytes represent a proinflammatory myeloid subpopulation in cancer**  
Horzum U., Yoyen-Ermis D., Taskiran E. Z., Yilmaz K. B., HAMALOĞLU E., KARAKOÇ D., ESENDAĞLI G.  
CANCER IMMUNOLOGY IMMUNOTHERAPY, vol.70, no.1, pp.75-87, 2021 (SCI-Expanded)
- XXIII. **Genetic IGF1R defects: new cases expand the spectrum of clinical features**  
Gonc E. N., Ozon Z., Oguz S., Kabacam S., Taskiran E., Kiper P., Utine G., Alikasifoglu A., Kandemir N., Boduroglu O., et al.  
JOURNAL OF ENDOCRINOLOGICAL INVESTIGATION, vol.43, no.12, pp.1739-1748, 2020 (SCI-Expanded)
- XXIV. **Novel insights into diabetes mellitus due toDNAJC3-defect: Evolution of neurological and endocrine phenotype in the pediatric age group**  
ÖZÖN Z. A., ALİKAŞİFOĞLU A., KANDEMİR N., Aydin B., GÖNÇ E. N., KARAOSMANOĞLU B., Celik N. B., Eroglu-Ertugrul N. G., Taskiran E. Z., Haliloglu G., et al.  
PEDIATRIC DIABETES, vol.21, no.7, pp.1176-1182, 2020 (SCI-Expanded)
- XXV. **A novel mutation of keratin 5 in epidermolysis bullosa simplex with migratory circinate erythema**  
YALICI ARMAĞAN B., Kabacam S., TAŞKIRAN Z. E., GÖKÖZ Ö., ÜTİNE G. E., ERSOY EVANS S.  
PEDIATRIC DERMATOLOGY, vol.37, no.2, pp.358-361, 2020 (SCI-Expanded)
- XXVI. **The Effect of Boron-Containing Nano-Hydroxyapatite on Bone Cells**  
Gizer M., Köse S., Karaosmanoglu B., Taskiran E., Berkkan A., Timuçin M., Korkusuz F., Korkusuz P.  
BIOLOGICAL TRACE ELEMENT RESEARCH, vol.193, no.2, pp.364-376, 2020 (SCI-Expanded)
- XXVII. **Genotype and phenotype evaluation of patients with primary ciliary dyskinesia: First results from Turkey**  
Emiralioglu N., Taşkıran E., Koşukcu C., Bilgiç E., Atilla P., Kaya B., Günaydın Ö., Yüzbaşıoğlu A., Tuğcu G., Ademhan D., et al.

- PEDIATRIC PULMONOLOGY, vol.55, no.2, pp.383-393, 2020 (SCI-Expanded)
- XXVIII. **A Monogenic Disease with a Variety of Phenotypes: Deficiency of Adenosine Deaminase 2**  
Ozen S., Bilginer Y., Batu E., Taşkıran E., Özkara H., Ünal Ş., Güleray N., Erden A., Karadağ Ö., Gümrük F., et al.  
JOURNAL OF RHEUMATOLOGY, vol.47, no.1, pp.117-125, 2020 (SCI-Expanded)
- XXIX. **ADA2 deficiency in a patient with Noonan syndrome-like disorder with loose anagen hair: The co-occurrence of two rare syndromes**  
Akgun-Dogan O., Simsek-Kiper P. O., Taskiran E., Lissewski C., Brinkmann J., Schanze D., Göçmen R., Cagdas D. N., Bilginer Y., Utine G. E., et al.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.12, pp.2474-2480, 2019 (SCI-Expanded)
- XXX. **Primary tumor resection for initially staged IV breast cancer An emphasis on programmed death-ligand 1 expression, promoter methylation status, and survival**  
Erol T., İmamoğlu N., Aydın B., Taşkıran Z. E., Esendağlı G., Kösemehmetoğlu K., Baykal A.  
MEDICINE, vol.98, no.33, 2019 (SCI-Expanded)
- XXXI. **Development, characterization and research of efficacy on in vitro cell culture of glucosamine carrying hyaluronic acid nanoparticles**  
Şahin Ş., BİLGİÇ E., Salimi K., Tuncel A., KARAOSMANOĞLU B., Taşkıran E. Z., KORKUSUZ P., KORKUSUZ F.  
JOURNAL OF DRUG DELIVERY SCIENCE AND TECHNOLOGY, vol.52, pp.393-402, 2019 (SCI-Expanded)
- XXXII. **Ophthalamo-acromelic syndrome in an infant**  
ÜREL DEMİR G., Taskiran E. Z., AKGÜN DOĞAN Ö., Simek-Kiper P. O., ÜTİNE G. E.  
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.7, 2019 (SCI-Expanded)
- XXXIII. **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**  
Simsek-Kiper P. O., Taskiran E., KOŞUKCU C., ARSLAN U. E., Cormier-Daire V., Gonc N., Ozon A., ALİKAŞİFOĞLU A., KANDEMİR N., ÜTİNE G. E., et al.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.7, pp.1157-1172, 2019 (SCI-Expanded)
- XXXIV. **Hyperphosphatasia with mental retardation syndrome type 4 In two siblings-expanding the phenotypic and mutational spectrum**  
Akgün D., Demir G., Kosukcu C., Taskiran E., Simsek-Kiper P., Utine G., Alikasıfoğlu M., Boduroğlu K.  
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.6, 2019 (SCI-Expanded)
- XXXV. **Transcriptome analysis reveals differentially expressed genes between human primary bone marrow mesenchymal stem cells and human primary dermal fibroblasts**  
TASKIRAN E. Z., KARAOSMANOĞLU B.  
TURKISH JOURNAL OF BIOLOGY, vol.43, no.1, pp.21-27, 2019 (SCI-Expanded)
- XXXVI. **A Novel Missense LIG4 Mutation in a Patient With a Phenotype Mimicking Behcet's Disease**  
Taskiran E., Sonmez H., Kosukcu C., Tavukcuoglu E., Yazici G., Esendagli G., Batu E., Kiper P., Bilginer Y., Alikasifoglu M., et al.  
JOURNAL OF CLINICAL IMMUNOLOGY, vol.39, no.1, pp.99-105, 2019 (SCI-Expanded)
- XXXVII. **Whole Exome Sequencing in Early-onset Systemic Lupus Erythematosus**  
Batu E. D., Koşukcu C., Taşkıran E., Sahin S., Akman S., Sözeri B., Ünsal E., Bilginer Y., Kasapcopur O., Alikasıfoğlu M., et al.  
JOURNAL OF RHEUMATOLOGY, vol.45, no.12, pp.1671-1679, 2018 (SCI-Expanded)
- XXXVIII. **Genetic testing for DADA2: How can we avoid missing patients?**  
SÖNMEZ H. E., BATU AKAL E. D., Taskiran E. Z., ALİKAŞİFOĞLU M., BİLGİNER Y., ÖZEN S.  
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.26, no.11, pp.1563-1564, 2018 (SCI-Expanded)
- XXXIX. **Further delineation of spondyloepimetaphyseal dysplasia Faden-Alkuraya type: A RSPRY1-associated spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis**  
Simsek-Kiper P., Taskiran E., Kosukcu C., Urel-Demir G., Akgun-Dogan O., Yilmaz G., Utine G., Nishimura G., Boduroglu K., Alikasifoglu M.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.176, no.9, pp.2009-2016, 2018 (SCI-Expanded)
- XL. **Clinical, demographic and nosologic characterisation of the genetic disorders of the skeleton in Turkey: The Skeletal Dysplasia registry**

ŞİMŞEK KİPER P. Ö., Utine G. E., TAŞKIRAN Z. E., KOŞUKCU C., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.  
European Journal Of Human Genetics, 2018 (SCI-Expanded)

- XLII. **Human bone marrow mesenchymal stem cells secrete endocannabinoids that stimulate in vitro hematopoietic stem cell migration effectively comparable to beta-adrenergic stimulation**  
KOSE S., AERTS-KAYA F. S. F., Kopru C. Z., NEMUTLU E., Koskonmaz B., KARAOSMANOĞLU B., TASKIRAN E. Z., ALTUN B., CETINKAYA D. U., KORKUSUZ P.  
EXPERIMENTAL HEMATOLOGY, vol.57, pp.30-41, 2018 (SCI-Expanded)
- XLIII. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Costello Syndrome**  
VURALLI KARAOĞLAN D., KOŞUKCU C., Taskiran E., Simsek P. O., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU A., ALİKAŞİFOĞLU M.  
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.363, 2018 (SCI-Expanded)
- XLIII. **Neonatal-Onset Recurrent Guillain-Barre Syndrome-Like Disease: Clues for Inherited CD59 Deficiency**  
Ardicli D., TASKIRAN E. Z., KOŞUKCU C., Temucin Ç. M., OGUZ K. K., HALILOGLU G., ALİKAŞİFOĞLU M., TOPALOGLU H.  
NEUROPEDIATRICS, vol.48, no.6, pp.477-481, 2017 (SCI-Expanded)
- XLIV. **HERC1 mutations in idiopathic intellectual disability**  
ÜTİNE G. E., Taskiran E. Z., KOŞUKCU C., KARAOSMANOĞLU B., GÜLERAY N., Dogan O. A., Kiper P. O. S., Boduroglu K., ALİKAŞİFOĞLU M.  
European Journal of Medical Genetics, vol.60, no.5, pp.279-283, 2017 (SCI-Expanded)
- XLV. **Identification of two novel PNPLA1 mutations in Turkish families with autosomal recessive congenital ichthyosis.**  
Dökmeçi-Emre S., TAŞKIRAN Z. E., YÜZBAŞIOĞLU A., ÖNAL G., AKARSU A. N., KARADUMAN A., ÖZGÜÇ M.  
The Turkish journal of pediatrics, vol.59, pp.475-482, 2017 (SCI-Expanded)
- XLVI. **Whole Exome Sequencing in Early Onset Systemic Lupus Erythematosus**  
BATU AKAL E. D., KOŞUKCU C., Taskiran E. Z., AKMAN S., Ozturk K., SÖZERİ B., Unsal E., Ekinci Z., BİLGİNER Y., ALİKAŞİFOĞLU M., et al.  
ARTHRITIS & RHEUMATOLOGY, vol.68, 2016 (SCI-Expanded)
- XLVII. **The Characteristic Features of the Patients with Deficiency of Adenosine Deaminase 2 (DADA2)**  
ERDEN A., BATU AKAL E. D., Taskiran E. Z., SÖNMEZ H. E., SARI A., ARMAĞAN B., KILIÇ L., ARICI Z. S., BİLGİNER Y., AKDOĞAN A., et al.  
ARTHRITIS & RHEUMATOLOGY, vol.68, 2016 (SCI-Expanded)
- XLVIII. **Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling**  
Cetinkaya A., Xiong J. R., Vargel İ., Kösemehmetoğlu K., Canter H. I., Gerdan O. F., Longo N., Alzahrani A., Camps M. P., Taskiran E. Z., et al.  
AMERICAN JOURNAL OF HUMAN GENETICS, vol.99, no.2, pp.299-317, 2016 (SCI-Expanded)
- XLIX. **A Case Series of Adenosine Deaminase 2-deficient Patients Emphasizing Treatment and Genotype-phenotype Correlations**  
Batu E. D., KARADAĞ Ö., TASKIRAN E. Z., KALYONCU U., AKSENTIJEVICH I., ALİKAŞİFOĞLU M., ÖZEN S.  
JOURNAL OF RHEUMATOLOGY, vol.42, no.8, pp.1532-1534, 2015 (SCI-Expanded)
- L. **Mutations In Anks6 Cause A Nephronophthisis-like Phenotype With End Stage Renal Disease**  
Taskiran E., Korkmaz E., Gucer S., KOŞUKCU C., Kaymaz F., Koyunlar C., Bryda E. C., Chaki M., Lu D. D., Vadnagara K., et al.  
PEDIATRIC NEPHROLOGY, vol.29, no.9, pp.1820, 2014 (SCI-Expanded)
- LI. **Mutations in ANKS6 Cause a Nephronophthisis-Like Phenotype with ESRD**  
Taskiran E. Z., Korkmaz E., Gucer S., KOŞUKCU C., Kaymaz F., Koyunlar C., Bryda E. C., Chaki M., Lu D., Vadnagara K., et al.  
JOURNAL OF THE AMERICAN SOCIETY OF NEPHROLOGY, vol.25, no.8, pp.1653-1661, 2014 (SCI-Expanded)
- LII. **TMCO1 Deficiency Causes Autosomal Recessive Cerebrofaciothoracic Dysplasia**  
ALANAY Y., BEKİR E., ÜTİNE G. E., ORÇUN H., ŞİMŞEK KİPER P. Ö., TAŞKIRAN E. Z., PERÇİN F. E., UZ E., MAHMUT

- ŞAMİL S., BAYRAM Y., et al.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.164, no.2, pp.291-304, 2014 (SCI-Expanded)
- LIII. **DGKE Variants Cause a Glomerular Microangiopathy That Mimics Membranoproliferative GN**  
ÖZALTIN F., Li B., Rauhauser A., An S., Soylemezoglu O., Gonul I. I., Taskiran E. Z., Ibsirlioglu T., Korkmaz E., BİLGİNER Y., et al.  
JOURNAL OF THE AMERICAN SOCIETY OF NEPHROLOGY, vol.24, no.3, pp.377-384, 2013 (SCI-Expanded)
- LIV. **The effect of colchicine on pyrin and pyrin interacting proteins.**  
Taskiran E. Z., Cetinkaya A., Balci-Peynircioglu B., Akkaya Y. Z., Yilmaz E.  
Journal of cellular biochemistry, vol.113, no.11, pp.3536-46, 2012 (SCI-Expanded)
- LV. **Frequency of mutations in PROP-1 gene in Turkish children with combined pituitary hormone deficiency**  
KANDEMİR N., VURALLI KARAOĞLAN D., Taskiran E., Gonc N., Ozon A., ALİKAŞİFOĞLU A., YILMAZ E.  
TURKISH JOURNAL OF PEDIATRICS, vol.54, no.6, pp.570-575, 2012 (SCI-Expanded)
- LVI. **Expression of ASC in post-mortem brain samples of Alzheimer's disease patients: A possible role for ASC in A beta amyloid formation**  
Taskiran E. Z., Balci-Peynircioglu B., Soylemezoglu F., YILMAZ E.  
TURKISH JOURNAL OF BIOCHEMISTRY-TURK BIYOKIMYA DERGISI, vol.36, no.4, pp.350-355, 2011 (SCI-Expanded)
- LVII. **MEFV mutations in systemic onset juvenile idiopathic arthritis**  
Ayaz N. A., ÖZEN S., BİLGİNER Y., Erguven M., Taskiran E., YILMAZ E., Besbas N., TOPALOĞLU R., Bakkaloglu A.  
RHEUMATOLOGY, vol.48, no.1, pp.23-25, 2009 (SCI-Expanded)
- LVIII. **Expression of ASC in renal tissues of familial mediterranean fever patients with amyloidosis: postulating a role for ASC in AA type amyloid deposition.**  
Balci-Peynircioglu B., Waite A. L., Schaner P., TAŞKIRAN Z. E., Richards N., ORHAN D., Gucer S., ÖZEN S., Gumucio D., Yilmaz E.  
Experimental biology and medicine (Maywood, N.J.), vol.233, no.11, pp.1324-33, 2008 (SCI-Expanded)

## Articles Published in Other Journals

- I. **Recurrent squamous cell carcinoma and a novel mutation in a patient with xeroderma pigmentosum: a case report**  
Sahin E. A., Taskiran E. Z., ŞİMŞEK KİPER P. Ö., Aydın B., Utine E.  
JOURNAL OF MEDICAL CASE REPORTS, vol.16, no.1, 2022 (ESCI)
- II. **Current Diagnostic Methods in Primary Ciliary Dyskinesia: Hacettepe University Experience**  
EMİRALİOĞLU N., TAŞKIRAN Z. E., KOŞUKCU C., BİLGİÇ E., ATİLLA P., KAYA Z. B., GÜNAYDIN R. Ö., yüzbaşıoğlu a., Ademhan D., Eryılmaz Polat S., et al.  
Turkish Thoracic Journal, vol.20, pp.17, 2019 (Scopus)
- III. **The Effect of Erythropoietin Treatment on Gene Expression Profile of Mesenchymal Stem Cells**  
Dalkiran D., KARAOSMANOĞLU B., TAŞKIRAN Z. E.  
Acta Medica, vol.50, no.2, pp.16-24, 2019 (Peer-Reviewed Journal)
- IV. **Neuronal Conversion of Dermal Fibroblasts as a Disease Model**  
TAŞKIRAN Z. E., KARAOSMANOĞLU B.  
Cumhuriyet Medical Journal, vol.40, no.4, pp.392-399, 2018 (Peer-Reviewed Journal)

## Refereed Congress / Symposium Publications in Proceedings

- I. **Büyük veriden hücre karakterizasyonuna: Transkripsiyon faktörü kataloglarının oluşturulması**  
İMREN G., KARAOSMANOĞLU B., ÇELEBİ SALTİK B., ESENDAĞLI G., TAŞKIRAN Z. E.  
Tıbbi Biyoloji ve Genetik Kongresi, Turkey, 27 - 30 October 2019
- II. **Sendromik olmayan zihinsel yetersizlikte tüm ekzom dizilemenin tanısal verimi**

TAŞKIRAN Z. E., KARAOSMANOĞLU B., KOŞUKCU C., ÜREL DEMİR G., akgün doğan ö., Kiper şimşek P. Ö., ALİKAŞİFOĞLU M., BODUROĞLU O. K., Utine G. E.

16. Tıbbi Biyoloji ve Genetik Kongresi, Turkey, 25 - 27 October 2019

- III. **Boron-Containing Nano-Hydroxyapatite Composites Alters Mesenchymal Stem Cell Proliferation and Osteogenic Differentiation**  
GİZER M., KÖSE S., KARAOSMANOĞLU B., TAŞKIRAN Z. E., BERKKAN A., TİMUÇİN M., KORKUSUZ F., KORKUSUZ P.  
European Orthopedic Research Society 27th Annual Anniversary Meeting, 2 - 05 October 2019
- IV. **The Skeletal Dysplasia Registry: Hacettepe Experience**  
ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., KOŞUKCU C., ARSLAN U. E., ÜTİNE G. E., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.  
The 14th biannual International Skeletal Dysplasia Society Meeting, Oslo, Norway, 11 - 14 September 2019
- V. **Development of a new antiproliferative arachydonoylcyclopropilamide (ACPA) releasing nanoparticle-based drug for endometrial cancers by targeting cannabinoid 1 receptors**  
BİLGİÇ E., BOYACIOĞLU Ö., VARAN C., BİLENSOY E., NEMUTLU E., KARAOSMANOĞLU B., TAŞKIRAN Z. E., KORKUSUZ P.  
EACR Conference Nanotechnology in Cancer: Engineering for Oncology, 12 - 14 September 2019
- VI. **Cannabinoid Receptor 1-Mediated Antiproliferative Effect of ACPA and ACPA-PCL Controlled Release System on Non-Small Cell Lung Cancer Lines**  
BOYACIOĞLU Ö., BİLGİÇ E., VARAN C., BİLENSOY E., NEMUTLU E., KARAOSMANOĞLU B., TAŞKIRAN Z. E., KORKUSUZ P.  
EACR Conference Nanotechnology in Cancer: Engineering for Oncology, Cambridge, Canada, 12 - 14 September 2019
- VII. **AB0574 A MONOGENIC DISEASE WITH WIDE RANGE OF SYMPTOMS: DEFICIENCY OF ADENOSINE DEAMINASE 2**  
BATU AKAL E. D., TAŞKIRAN Z. E., TEZCAN F. İ., ÖZKARA H. A., ÇAĞDAŞ AYVAZ D. N., ÖZEN S., ÜNAL CANGÜL Ş., BİLGİNER Y., KARADAĞ Ö., ÇETİN M., et al.  
Annual European Congress of Rheumatology (EULAR), Madrid, Spain, 12 - 15 June 2019, vol.78, pp.1748
- VIII. **Small cell lung cancer stem cells show adaptive resistance through effectively inducing t cell activation and modulating the expression of co-inhibitory receptor**  
KURŞUNEL M. A., TAŞKIRAN Z. E., ESENDAĞLI G.  
International Molecular Immunology Immunogenetics Congress IV (MIMIC IV), Bursa, Turkey, 27 - 29 April 2019, pp.29
- IX. **Kemik hücrelerine bor içeren nano-hidroksiapatit kompozitlerin moleküler etki mekanizması**  
Gizer M., KÖSE S., KARAOSMANOĞLU B., TAŞKIRAN Z. E., BERKKAN A., TİMUÇİN M., KORKUSUZ F., KORKUSUZ P.  
24. Ulusal Elektron Mikroskopi Kongresi, Edirne, Turkey, 24 - 26 April 2019
- X. **Endometriyum kanserinde araşidonilsiklopropilamid (ACPA)'in kannabinoid 1 reseptörü aracılı antiproliferatif etkisinin gösterilmesi**  
BİLGİÇ E., BOYACIOĞLU Ö., KARAOSMANOĞLU B., TAŞKIRAN Z. E., KORKUSUZ P.  
24. Ulusal Elektron Mikroskopi Kongresi, Edirne, Turkey, 24 - 26 April 2019, pp.112-113
- XI. **Primer Siliyer Diskinezi Tanısında Güncel Yöntemlerin Kullanımı: Hacettepe Deneyimi**  
EMİRALİOĞLU N., TAŞKIRAN Z. E., Kosukcu C., Bilgic E., Atilla P., Kaya B., GÜNAYDIN R. Ö., YÜZBAŞIOĞLU A., Ademhan D., Polat S. E., et al.  
Türk Toraks Derneği 22. Ulusal Kongresi, Antalya, Turkey, 10 - 14 April 2019
- XII. **Two siblings with primary ciliary dyskinesia and hepatic involvement**  
hizal m., BİLGİÇ E., TAŞKIRAN Z. E., ATİLLA P., AKÇÖREN Z., GÜNAYDIN R. Ö., ÖZEN H., esref s., yalcin e., DOĞRU ERSÖZ D., et al.  
3rd BEAT-PCD Conference and 4th PCD Training School, Lisbon, Portugal, 6 - 09 February 2018
- XIII. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Costello Syndrome**  
VURALLI KARAOĞLAN D., KOŞUKCU C., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU A., ALİKAŞİFOĞLU M.  
57th Annual ESPE European Society for Paediatric Endocrinology, Atina, Greece, 27 - 29 September 2018, vol.90,

- XIV. **Comparative Transcriptome Analysis of Distinct Stromal Cell Populations During Extracellular Matrix Remodeling in Skeletal Muscle**  
ÖZDEMİR SAKA C., AKÇAY D., YÖYEN ERMİŞ D., TAŞKIRAN Z. E., ESENDAĞLI G., KOCAEFE Y. Ç.  
13th Meeting of the Mediterranean Society of Myology, 27 - 29 June 2018, vol.37, pp.129-183
- XV. **Clinical, demographic and nosologic characterization of the genetic disorders of the skeleton in Turkey: The skeletal dysplasia registry.**  
ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., TAŞKIRAN Z. E., KOŞUKCU C., ARSLAN U. E., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.  
European Human Genetics Conference, Milan, Italy, June 16-19, 2018., Milan, Italy, 16 - 19 June 2018
- XVI. **Mezenkimal Kök Hücreler Endokannabinoidler Aracılığı ile Hematopoietik Kök Hücre Mobilizasyonunu Düzenlemektedir**  
KÖSE S., KÖPRÜ Ç. Z., KARAOSMANOĞLU B., TAŞKIRAN Z. E., AERTS KAYA F. S. F., NEMUTLU E., ÇETİNKAYA F. D., KORKUSUZ P.  
Elektron Mikroskopu Kongresi, Turkey, 24 - 26 April 2019
- XVII. **Rett sendromlu 16 hastanın klinik ve moleküler açıdan değerlendirilmesi**  
ZENGİN AKKUŞ P., TAŞKIRAN Z. E., KABAÇAM S., ŞİMŞEK KİPER P. Ö., HALİLOĞLU V. G., BODUROĞLU O. K., ÜTİNE G. E.  
61. Türkiye Milli Pediatri Kongresi, Antalya, Turkey, 15 - 19 November 2017
- XVIII. **Oftalmo-akromelik sendrom**  
ÜREL DEMİR G., TAŞKIRAN Z. E., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.  
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017
- XIX. **PGAP3 geninde yeni tanımlanmış mutasyona bağlı hiperfosfatazya mental retardasyon sendromu**  
AKGÜN DOĞAN Ö., ÜREL DEMİR G., KOŞUKCU C., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.  
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017
- XX. **Further delineation of spondyloepimetaphyseal dysplasia Faden-Alkuraya type: a RSPRY1-associated spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis.**  
ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., KOŞUKCU C., AKGÜN DOĞAN Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.  
International Skeletal Dysplasia Society Meeting, 20 - 23 September 2017
- XXI. **Identification of Molecular Pathology of Peters' Anomaly Segregating in a Large Autosomal Dominant Family**  
KOŞUKCU C., ASLI K., ALANAY Y., KAVAK P., BERKER N., TAŞKIRAN Z. E., ALİKAŞİFOĞLU M., SEZERMAN O. U., AKARSU A. N.  
10th International Symposium on Health Informatics and Bioinformatics (HIBIT 2017), KALKANLI, GUZELYURT, Cyprus (Kktc), 29 - 30 June 2017
- XXII. **A Section of Rare Diseases in Hacettepe Adult Clinics in Rheumatology: Deficiency of Adenosine Deaminase 2 (DADA2)**  
BÖLEK E. Ç., ERDEN A., ÖZEN S., KARADAĞ Ö., KALYONCU U., BATU E. D., TAŞKIRAN Z. E., arici z. s., SARI A., KILIÇ L., et al.  
5th Rare Diseases Summer School, 7 - 09 June 2017
- XXIII. **RSPRY1 associated skeletal dysplasia: Spondylo-epi-metaphyseal dysplasia with conobrachydactyly and craniosynostosis**  
ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., KOŞUKCU C., AKGÜN DOĞAN Ö., YILMAZ G., ÜTİNE G. E., Nishimura G., BODUROĞLU O. K., ALİKAŞİFOĞLU M.  
European Society of Human Genetics Conference 2017, Kopenhag, Denmark, 27 - 30 May 2017
- XXIV. **Homozygous novel variant in MUT in a patient with intellectual disability without metabolic derangement**  
ÜTİNE G. E., TAŞKIRAN Z. E., KOŞUKCU C., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., BODUROĞLU O. K., ALİKAŞİFOĞLU M.



European Society of Human Genetics Conference 2017, Kopenhagen, Denmark, 27 - 30 May 2017

- XXV. **Whole Exoma Sequencing in early onset systemic lupus erythematosus**  
BATU E. D., kosukcu c., TAŞKIRAN Z. E., AKMAN S., Unsal e., ikinci z., BİLGİNER Y., ALİKAŞİFOĞLU M., ÖZEN S.  
2016 ACR/ARHP Annual Meeting, 6 - 11 November 2016
- XXVI. **Clinical and Molecular Analysis of 3M Syndrome Patients A Study From Turkey**  
ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., ÜTİNE G. E., ALİKAŞİFOĞLU A., KANDEMİR N., Cormier Daire V., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.  
28th International Congress of Pediatrics, 17 - 22 August 2016
- XXVII. **Clinical and Molecular aspects and genotype phenotype correlation in Rett syndrome**  
ZENGİN AKKUŞ P., ÜTİNE G. E., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., HALİLOĞLU V. G., BODUROĞLU O. K.  
European Society of Human Genetics Conference 2016 Barcelona, 21 - 24 May 2016
- XXVIII. **Clinical and quantitative PCR confirmation of copy number variations detected by array CGH**  
CEYLAN A. C., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.  
European Society of Human Genetics Conference 2016, 21 - 24 May 2016
- XXIX. **Mendel Hastalıklarında Tüm Ekzom Dizileme**  
TAŞKIRAN Z. E.  
XIV. ULUSAL TIBBİ BİYOLOJİ VE GENETİK KONGRESİ, Turkey, 27 - 30 October 2015
- XXX. **Yağ Damlacıklarının Lipofaji Aracılı Regülasyonunda PNPLA1 Proteininin Rolü**  
ÖNAL G., ORAL Ö., TAŞKIRAN Z. E., YÜZBAŞIOĞLU A., KARADUMAN A., GÖZÜAÇIK D., DÖKMECİ S.  
XIV. Ulusal Tıbbi Biyoloji ve Genetik Kongresi, Muğla, Turkey, 27 - 30 October 2015

## Supported Projects

- ORHAN D., AKYÜZ C., KURUCU N., KARAOSMANOĞLU B., YET İ., TAŞKIRAN Z. E., GÜÇER K. Ş., Project Supported by Higher Education Institutions, Pediatrik Rabdomiyosarkom Alt Tiplerinde Moleküler Patolojik Belirteçlerin Analizi, 2017 - 2022
- ORHAN D., KARAOSMANOĞLU B., YET İ., YÜCE A., GÜMÜŞ E., TAŞKIRAN Z. E., AKÇÖREN Z., Project Supported by Higher Education Institutions, Neonatal Kolestatik Hastalıkların Moleküler Genetik Analizi, 2017 - 2022
- ESENDAĞLI G., TAŞKIRAN Z. E., Horzum U., Project Supported by Higher Education Institutions, Farklı aktivasyon basamaklarına ilerletilen yardımcı T hücrelerinde adezyon ve migrasyon mekanizmalarının incelenmesi, 2018 - 2021
- AKBAL VURAL Ö., TAŞKIRAN Z. E., BOLAT G., TUĞÇE YAMAN Y., Project Supported by Higher Education Institutions, mikroRNA Teşhisi İçin Tek Kullanımlık, Etiketsiz Nanobiyosensörlerin Geliştirilmesi ve Uygulaması, 2018 - 2020
- PURALI N., ERGİN B., TAŞKIRAN Z. E., SAĞLAM B., Project Supported by Higher Education Institutions, Bir primer reseptör nöronda mekanoelektriksel çevrimden sorumlu yapılara ait genlerin klonlanması ve kodlanan proteinlerin moleküler özelliklerinin belirlenmesi, 2017 - 2020
- KARAKOÇ D., YORGUN H., ÜNAL CANGÜL Ş., ER ÖZTAŞ Y., ESENDAĞLI G., KONAŞ E., ÇALIK BAŞARAN N., KILIÇASLAN B., EKİNCİ S., HELVACI E., et al., Project Supported by Higher Education Institutions, Hacettepe Üniversitesi Tıp Fakültesi Dergisi Acta Medica'nın Yapılandırılması, 2017 - 2019
- TAŞKIRAN Z. E., ÜTİNE G. E., ŞİMŞEK KİPER P. Ö., BODUROĞLU O. K., AKARSU A. N., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, Hacettepe Ekzom Projesi, 2015 - 2019
- ELÇİN G., ÇAKIR A., TAŞKIRAN Z. E., Project Supported by Higher Education Institutions, Herediter melanomda CDKN2A ve MC1R germline gen mutasyonlarının araştırılması, 2017 - 2018
- ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., KOŞUKCU C., AKGÜN DOĞAN Ö., YILMAZ G., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, Spondiloepimetafizyeal displazi Faden Alkuraya Tipi İskelet Displazisinin Daha Geniş Detaylandırılması: RSPRY1-ilişkili Spondiloepimetafizyeal displazi, Konobrakidaktili ve Kraniosinostozis, 2017 - 2017
- ÜTİNE G. E., AKGÜN DOĞAN Ö., KOŞUKCU C., ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., Project Supported by Higher Education Institutions, Zihinsel Yetersizliği Bulunan Ancak Metabolik Bozukluğu Olmayan Bir Hastada Tüm Ekzom Sekanslaması ile MUT Geninde Bulunan Yeni Homozigot Varyasyon, 2017 - 2017
- ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., BODUROĞLU O. K., ÜTİNE G. E., ALANAY Y., Project Supported by Higher Education

Institutions, 3M Sendromlu Bir Grup Türk Hastada Klinik ve Moleküler Özelliklerin Analizi, 2015 - 2016  
YÜZBAŞIOĞLU A., DÖKMECİ S., AKARSU A. N., TAŞKIRAN Z. E., KARADUMAN A., ÖNAL G., Project Supported by Higher  
Education Institutions, Otozomal Resesif Konjenital İktiyoz Hastalarında Genetik Analiz, 2015 - 2016  
ÜNAL CANGÜL Ş., ÇETİN M., AKARSU A. N., GÜMRÜK F., TAŞKIRAN Z. E., BAYHAN T., Project Supported by Higher  
Education Institutions, Diamond Blackfan anemili hastalarda yüksek çözünürlüklü dizileme yöntemi ile genom boyu  
analizi, 2015 - 2015

## **Metrics**

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H-Index (Scopus): 13