

Res. Asst. CAN KOŞUKCU

Personal Information

Office Phone: [+90 312 305 3038](tel:+903123053038)

Fax Phone: [+90 312 305 3039](tel:+903123053039)

Email: cankosukcu@hacettepe.edu.tr

Web: <https://avesis.hacettepe.edu.tr/cankosukcu>

Address: Hacettepe Üniversitesi Merkez Kampüsü Sağlık Bilimleri Enstitüsü Altındağ, 06100, ANKARA

Education Information

Doctorate, Hacettepe University, Çocuk Sağlığı Enstitüsü, Moleküler Metabolizma, Turkey 2016 - 2016

Postgraduate, Hacettepe University, Sağlık Bilimleri Enstitüsü, Biyoinformatik, Turkey 2013 - 2016

Undergraduate, Hacettepe University, Fen Fakültesi, Biyoloji, Turkey 2009 - 2013

Foreign Languages

English, C1 Advanced

Dissertations

Postgraduate, Otozomal dominant peters anomalisi tanısı alan bir ailede yeni gen araştırması, Hacettepe Üniversitesi, Sağlık Bilimleri Enstitüsü, Biyoinformatik (YI) (Tezli), 2015

Research Areas

Medicine, Health Sciences, Fundamental Medical Sciences, Biostatistics and Medical Informatics, Medical Biology

Academic Titles / Tasks

Research Assistant, Hacettepe University, Sağlık Bilimleri Enstitüsü, Biyoinformatik A.B.D., 2016 - Continues

Published journal articles indexed by SCI, SSCI, and AHCI

I. **One Disease with two Faces: Semidominant Inheritance of a Novel HTRA1 Mutation in a Consanguineous Family**

Bekircan-Kurt C. E. , ÇETİNKAYA A., GÖÇMEN R., KOŞUKCU C., Soylemezoglu F., ARSAVA E. M. , Tuncer A., ERDEM ÖZDAMAR S., Akarsu N. A. , TOPÇUOĞLU M. A.

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II. **Clinical and molecular characteristics of carnitine-acylcarnitine translocase deficiency with c.270delC and a novel c.408C>A variant**

BİLGİNER GÜRBÜZ B., YÜCEL YILMAZ D., ÖZGÜL R. K. , KOŞUKCU C., DURSUN A., SİVRİ H. S. , COŞKUN T., TOKATLI A.

TURKISH JOURNAL OF PEDIATRICS, vol.63, no.4, pp.691-696, 2021 (Journal Indexed in SCI)

- III. **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.
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- IV. **Frequency of HLA Class I and Class II Alleles in Patients with COVID from Turkey**
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- V. **Atypical Presentation of Sengers Syndrome: A Novel Mutation Revealed with Postmortem Genetic Testing.**
Guleray N., Kosukcu C., Taskiran Z., Simsek K., Utine G., Gucer S., Tokatli A., Boduroglu K., Alikasifoglu M.
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- VI. **Lymphocyte Subgroups and KREC Numbers in Common Variable Immunodeficiency: A Single Center Study**
Yaz I., Ozbek B., Ng Y. Y. , Cetinkaya P. G. , Halacli S., TAN Ç., KAŞIKCI M., KOŞUKCU C., Tezcan I., Cagdas D. N.
JOURNAL OF CLINICAL IMMUNOLOGY, vol.40, no.3, pp.494-502, 2020 (Journal Indexed in SCI)
- VII. **Detection of allele frequencies of common c. 511C > T and c.625G > A variants in the ACADS gene in the Turkish population**
Kilic M., Erguner B., KOŞUKCU C., ÖZGÜL R. K.
TURKISH JOURNAL OF PEDIATRICS, vol.62, no.1, pp.19-23, 2020 (Journal Indexed in SCI)
- VIII. **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 (Journal Indexed in SCI)
- IX. **Hyperphosphatasia with mental retardation syndrome type 4 In two siblings-expanding the phenotypic and mutational spectrum**
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- X. **A Novel Missense LIG4 Mutation in a Patient With a Phenotype Mimicking Behcet's Disease**
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- XI. **A novel NKX3-2 mutation associated with perinatal lethal phenotype of spondylo-megaepiphyseal-metaphyseal dysplasia in a neonate**
Simsek-Kiper P. O. , KOŞUKCU C., Akgun-Dogan O., GÖÇMEN R., ÜTİNE G. E. , SOYER T., Korkmaz-Toygar A., Nishimura G., ALİKAŞİFOĞLU M., Boduroglu K.
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- XII. **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., Alikasifoglu M., et al.
JOURNAL OF RHEUMATOLOGY, vol.45, no.12, pp.1671-1679, 2018 (Journal Indexed in SCI)
- XIII. **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.
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- XIV. **Hypomorphic RAG1 defect in a child presented with pulmonary hemorrhage and digital necrosis**
Taşkıran E., Sönmez H., Ayvaz D. N. , Koşukcu C., Batu E. D. , Esenboğa S., Topaloğlu R., Orhan D., Bilginer Y., Alikasifoglu M., et al.

CLINICAL IMMUNOLOGY, vol.187, pp.92-94, 2018 (Journal Indexed in SCI)

- XV. **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 (Journal Indexed in SCI)
- XVI. **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.
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- XVII. **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 (Journal Indexed in SCI)
- XVIII. **HERC1 mutations in idiopathic intellectual disability**
ÜTİNE G. E. , Taskiran E. Z. , KOŞUKCU C., KARAOŞMANOĞLU B., GÜLERAY N., Dogan O. A. , Kiper P. O. S. , Boduroglu K., ALİKAŞİFOĞLU M.
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- XIX. **Whole Exome Sequencing in Early Onset Systemic Lupus Erythematosus**
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- XX. **The effects of larval diet restriction on developmental time, preadult survival, and wing length in Drosophila melanogaster**
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TURKISH JOURNAL OF ZOOLOGY, vol.39, no.3, pp.395-403, 2015 (Journal Indexed in SCI)
- XXI. **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 (Journal Indexed in SCI)
- XXII. **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.
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Articles Published in Other Journals

- I. **Current Diagnostic Methods in Primary Ciliary Dyskinesia: Hacettepe University Experience**
EMİRALIOĞLU N., TAŞKIRAN Z. E. , KOŞUKCU C., BİLGİÇ E., ATILLA P., KAYA Z. B. , GÜNAYDIN R. Ö. , yüzbaşıoğlu a., Ademhan D., Eryılmaz Polat S., et al.
Turkish Thoracic Journal, vol.20, pp.17, 2019 (Refereed Journals of Other Institutions)

Refereed Congress / Symposium Publications in Proceedings

- I. **Sendromik olmayan zihinsel yetersizlikte tüm ekzom dizilemenin tanısal verimi**
TAŞKIRAN Z. E. , KARAOŞMANOĞLU 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

- II. **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
- III. **Severe Motor Mental Retardation with Microcephaly and Hypomyelination due to PYCR2 Gene variant in a Large Family**
BİLGİNER GÜRBÜZ B., EROĞLU ERTUĞRUL N. G. , KOŞUKCU C., YÜCEL YILMAZ D., ÖZGÜL R. K. , YALNIZOĞLU D.,
DURSUN A.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism 2019 (SSIEM-2019), Rotterdam,
Netherlands, 3 - 06 September 2019, vol.42, pp.1-479
- IV. **The Correlation Between Clinical Characteristics and Molecular Genetic Analysis Results of Primary Ciliary Dyskinesia Patients: Hacettepe University Experience**
EMİRALİOĞLU N., Taskiran E., KOŞUKCU C., BİLGİÇ E., ATİLLA P., Kaya B., Gunaydin O., YÜZBAŞIOĞLU A., Tugcu G. D.
, Ademhan D., et al.
European-Respiratory-Society (ERS) International Congress, Madrid, Spain, 28 September - 02 October 2019,
vol.54
- V. **TIP III HEREDİTER ANJIYOÖDEM ÖN TANILI HASTALARDA YENİ NESİL DİZİLEME İLE YENİ GENLERİN ARAŞTIRILMASI: ÖN VERİLER**
Koşukcu C., Uysal Soyer Ö., Karakaya G., Şahiner Ü. M. , Damadoğlu E., Ocak M., Kaya S. B. , Çakmak M., Kalyoncu A. F.
, Şekerel B. E. , et al.
XXV. Ulusal Allerji ve Klinik İmmünoloji Kongresi, Antalya, Turkey, 17 - 21 November 2018
- VI. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Costello Syndrome**
VURALI 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,
pp.351-352
- VII. **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
- VIII. **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
- IX. **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
- X. **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
- XI. **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

XII. 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, Kopenhag, Denmark, 27 - 30 May 2017

XIII. A novel mutation in the HTRA1 gene in a Turkish family with CARASIL

Bekircan-Kurt C. E. , KOŞUKCU C., KURNE A. T. , TOPÇUOĞLU M. A. , ERDEM-OZDAMAR S., GÖÇMEN R., AKARSU N.
1st Congress of the European-Academy-of-Neurology, Berlin, Germany, 20 - 23 June 2015, vol.22, pp.258

Supported Projects

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 Kraniyosinostozis, 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

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Citations

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