Dr. Öğr. Üyesi FİLİZ BAŞAK ERGİN

Kişisel Bilgiler

E-posta: basakergin@gazi.edu.tr
Web: https://avesis.gazi.edu.tr/11361

Uluslararası Araştırmacı ID'leri
ORCID: 0000-0002-1374-5939
Yoksis Araştırmacı ID: 303242

Eğitim Bilgileri

Doktora, Ankara Üniversitesi, Biyoteknoloji Enstitüsü, Temel Biyoteknoloji (Dr), Türkiye 2007 - 2010
Yüksek Lisans, Ankara Üniversitesi, Biyoteknoloji Enstitüsü, Temel Biyoteknoloji (Yl) (Tezli), Türkiye 2004 - 2006
Lisans, Ankara Üniversitesi, Fen Fakültesi, Biyoloji Bölümü, Türkiye 1999 - 2004

Yaptığı Tezler

Doktora, Sendromik olmayan otozomal resesif işitme kayıplı ve MYO15A genine bağlantı bulunan ailelerde mutasyon analizi, Ankara Üniversitesi, Biyoteknoloji Enstitüsü, Temel Biyoteknoloji (Dr), 2010
Yüksek Lisans, PTPN11 gen mutasyonlarının noonan sendromlu hastalarda taranması, Ankara Üniversitesi, Biyoteknoloji Enstitüsü, Temel Biyoteknoloji (Yl) (Tezli), 2006

Araştırma Alanları

Sağlık Bilimleri

Akademik Unvanlar / Görevler

Öğretim Görevlisi, Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, 2019 - Devam Ediyor

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

I. **A very rare presentation of mitochondrial elongation factor Tu deficiency-TUFM mutation and literature review**
GÖKALP S., İNCİ A., KILIÇ A., Ozsaydi E., ALTUN A. N., DEMİR F., ERGİN F. B., Ozbek M. N., OKUR İ., EZGÜ F. S., et al.
Journal of Pediatric Endocrinology and Metabolism, cilt.37, sa.6, ss.571-574, 2024 (SCI-Expanded)

II. **A possibly new autoinflammatory disease due to compound heterozygous phosphomevalonate kinase gene mutation**
Yıldız Ç., Gezgin Yıldırım D., İnci A., Tümer L., Ergin F. B., Sunar Yayla E. N. S., Esmeray Şenol P., Karaçayır N., Eğritaş Gürkan Ö., Okur İ., et al.
Joint Bone Spine, cilt.90, sa.1, 2023 (SCI-Expanded)

III. **Expected or unexpected clinical findings in liver glycogen storage disease type IX: distinct clinical and molecular variability**

İnci A., Kılıç Yıldırım G., Cengiz Ergin F. B., Sarı S., Eğritaş Gürkan Ö., Okur İ., Biberoğlu G., Bükülmez A., Ezgü F. S., Dalgıç B., et al.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.35, sa.4, ss.451-462, 2022 (SCI-Expanded)

IV. **The first case with FBXL4 mutation successfully treated with a parenteral ketogenic diet for lactic acidosis**
İNCİ A., Aktas E., Cengiz Ergin F. B., OKUR İ., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
JOURNAL OF PARENTERAL AND ENTERAL NUTRITION, cilt.45, sa.8, ss.1788-1792, 2021 (SCI-Expanded)

V. **Congenital defects of glycosylation: Novel presentations with mainly neurological involvement and variable dysmorphic features**
İNCİ A., Cengiz B., BİBEROĞLU G., OKUR İ., ARHAN E., ÖNER A. Y., KASAPKARA Ç. S., Kucukcongar A., TÜMER L., Ezgu F. S.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.185, sa.9, ss.2739-2747, 2021 (SCI-Expanded)

VI. **Hypophosphatasia: is it an underdiagnosed disease even by expert physicians?**
İnci A., Ergin F. B., Yüce B. T., Çiftçi B., Demir E., Buyan N., Okur İ., Biberoğlu G., Öktem R. M., Tümer L., et al.
JOURNAL OF BONE AND MINERAL METABOLISM, cilt.39, sa.4, ss.598-605, 2021 (SCI-Expanded)

VII. **Two patients from Turkey with a novel variant in the GM2A gene and review of the literature**
İNCİ A., ERGİN F. B., BİBEROĞLU G., OKUR İ., EZGÜ F. S., TÜMER L.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.34, sa.6, ss.805-812, 2021 (SCI-Expanded)

VIII. **Long-range cis-regulatory elements controlling GDF6 expression are essential for ear development**
Bademci G., Abad C., Cengiz F. B., Seyhan S., İNCESULU Ş. A., Guo S., Fitoz S., ATLI E. İ., Gosstola N. C., DEMİR S., et al.
JOURNAL OF CLINICAL INVESTIGATION, cilt.130, sa.8, ss.4213-4217, 2020 (SCI-Expanded)

IX. **Spectrum of Genetic Variants Associated with Anterior Segment Dysgenesis in South Florida**
Thanikachalam S., Hodapp E., Chang T. C., Swols D. M., Cengiz F. B., Guo S., Zafeer M. F., Seyhan S., Bademci G., Scott W. K., et al.
GENES, cilt.11, sa.4, 2020 (SCI-Expanded)

X. **FOXF2 is required for cochlear development in humans and mice**
Bademci G., Abad C., İNCESULU Ş. A., Elian F., Reyahi A., Diaz-Horta O., Cengiz F. B., Sineni C. J., Seyhan S., ATLI E. İ., et al.
HUMAN MOLECULAR GENETICS, cilt.28, sa.8, ss.1286-1297, 2019 (SCI-Expanded)

XI. **Dysfunction of GRAP, encoding the GRB2-related adaptor protein, is linked to sensorineural hearing loss**
Li C., Bademci G., Subasioglu A., Diaz-Horta O., Zhu Y., Liu J., Mitchell T. G., Abad C., Seyhan S., DUMAN D., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.116, sa.4, ss.1347-1352, 2019 (SCI-Expanded)

XII. **Ripor2 is involved in auditory hair cell stereociliary bundle structure and orientation**
Diaz-Horta O., Abad C., Cengiz F. B., Bademci G., Blackwelder P., Walz K., Tekin M.
JOURNAL OF MOLECULAR MEDICINE-JMM, cilt.96, sa.11, ss.1227-1238, 2018 (SCI-Expanded)

XIII. **Identification of candidate gene FAM183A and novel pathogenic variants in known genes: High genetic heterogeneity for autosomal recessive intellectual disability**
McSherry M., Masih K. E., ELÇİOĞLU H. N., Celik P., Balci O., Cengiz F. B., Nunez D., Sineni C. J., Seyhan S., Kocaoglu D., et al.
PLOS ONE, cilt.13, sa.11, 2018 (SCI-Expanded)

XIV. **Monosomy chromosome 21 compensated by 21q22.11q22.3 duplication in a case with small size and minor anomalies**
Su M., Benke P. J., Bademci G., Cengiz F. B., Ouyang X., Peng J., Casas C. E., Tekin M., Fan Y.
MOLECULAR CYTOGENETICS, cilt.11, 2018 (SCI-Expanded)

XV. **MPZL2 is a novel gene associated with autosomal recessive nonsyndromic moderate hearing loss**
Bademci G., Abad C., İNCESULU Ş. A., Rad A., Alper O., Kolb S. M., Cengiz F. B., Diaz-Horta O., SILAN F., MIHÇI E., et al.
HUMAN GENETICS, cilt.137, sa.6-7, ss.479-486, 2018 (SCI-Expanded)

XVI. **Novel pathogenic variants underlie SLC26A4-related hearing loss in a multiethnic cohort**
Cengiz F. B., Yilmazer R., Olgun L., SENNAROĞLU L., Kirazli T., Alper H., Olgun Y., İNCESULU Ş. A., Atik T., Huesca-

Hernandez F., et al.
INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, cilt.101, ss.167-171, 2017 (SCI-Expanded)

XVII. **Novel EYA1 variants causing Branchio-oto-renal syndrome**
Klingbeil K. D., Greenland C. M., Arslan S., Paneque A. L., GÜRKAN H., Ulusal S. D., Maroofian R., Carrera-Gonzalez A., Montufar-Armendariz S., Paredes R., et al.
INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, cilt.98, ss.59-63, 2017 (SCI-Expanded)

XVIII. **Targeted Resequencing of Deafness Genes Reveals a Founder MYO15A Variant in Northeastern Brazil**
Manzoli G. N., Bademci G., Acosta A. X., Felix T. M., Cengiz F. B., Foster J., Da Silva D. S. D., Menendez I., Sanchez-Pena I., Tekin D., et al.
ANNALS OF HUMAN GENETICS, cilt.80, sa.6, ss.327-331, 2016 (SCI-Expanded)

XIX. **Audiological findings in Noonan syndrome**
Tokgoz-Yilmaz S., TÜRKYILMAZ M. D., Cengiz F. B., Sjostrand A. P., KÖSE S. K., Tekin M.
INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, cilt.89, ss.50-54, 2016 (SCI-Expanded)

XX. **Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents**
Yan D., Tekin D., Bademci G., Foster J., Cengiz F. B., Kannan-Sundhari A., Guo S., Mittal R., Zou B., Grati M., et al.
HUMAN GENETICS, cilt.135, sa.8, ss.953-961, 2016 (SCI-Expanded)

XXI. **Variations in Multiple Syndromic Deafness Genes Mimic Non-syndromic Hearing Loss**
Bademci G., Cengiz F. B., Foster J., DUMAN D., SENNAROĞLU L., Diaz-Horta O., Atik T., Kirazli T., Olgun L., Alper H., et al.
SCIENTIFIC REPORTS, cilt.6, 2016 (SCI-Expanded)

XXII. **ROR1 is essential for proper innervation of auditory hair cells and hearing in humans and mice**
Diaz-Horta O., Abad C., SENNAROĞLU L., Foster J., DeSmidt A., Bademci G., Tokgoz-Yilmaz S., DUMAN D., Cengiz F. B., Grati M., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.113, sa.21, ss.5993-5998, 2016 (SCI-Expanded)

XXIII. **Comprehensive analysis via exome sequencing uncovers genetic etiology in autosomal recessive nonsyndromic deafness in a large multiethnic cohort**
Bademci G., Foster J., Mahdieh N., Bonyadi M., DUMAN D., Cengiz F. B., Menendez I., Diaz-Horta O., Shirkavand A., Zeinali S., et al.
GENETICS IN MEDICINE, cilt.18, sa.4, ss.364-371, 2016 (SCI-Expanded)

XXIV. **HPSE2 Mutations in Urofacial Syndrome, Non-Neurogenic Neurogenic Bladder and Lower Urinary Tract Dysfunction**
BULUM AKBULUT B., ÖZÇAKAR Z. B., DUMAN D., Cengiz F. B., Kavaz A., BURGU B., Baskin E., Cakar N., SOYGÜR Y. T., Ekim M., et al.
NEPHRON, cilt.130, sa.1, ss.54-58, 2015 (SCI-Expanded)

XXV. **Identification of Copy Number Variants Through Whole-Exome Sequencing in Autosomal Recessive Nonsyndromic Hearing Loss**
Bademci G., Diaz-Horta O., Guo S., DUMAN D., Van Booven D., Foster J., Cengiz F. B., Blanton S., Tekin M.
GENETIC TESTING AND MOLECULAR BIOMARKERS, cilt.18, sa.9, ss.658-661, 2014 (SCI-Expanded)

XXVI. **FAM65B is a membrane-associated protein of hair cell stereocilia required for hearing**
Diaz-Horta O., Subasioglu-Uzak A., Grati M., DeSmidt A., Foster J., Cao L., Bademci G., TOKGÖZ YILMAZ S., DUMAN D., Cengiz F. B., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.111, sa.27, ss.9864-9868, 2014 (SCI-Expanded)

XXVII. **Branchio-oculo-facial syndrome in a newborn caused by a novel TFAP2A mutation.**
Günes N., Cengiz F. B., Duman D., Dervişoğlu S., Tekin M., Tüysüz B.
Genetic counseling (Geneva, Switzerland), cilt.25, sa.1, ss.41-7, 2014 (SCI-Expanded)

XXVIII. **SLITRK6 mutations cause myopia and deafness in humans and mice**
Tekin M., Chioza B. A., Matsumoto Y., Diaz-Horta O., Cross H. E., DUMAN D., Kokotas H., Moore-Barton H. L., Sakoori K., Ota M., et al.

JOURNAL OF CLINICAL INVESTIGATION, cilt.123, sa.5, ss.2094-2102, 2013 (SCI-Expanded)

XXIX. **Whole-Exome Sequencing Efficiently Detects Rare Mutations in Autosomal Recessive Nonsyndromic Hearing Loss**
Diaz-Horta O., DUMAN D., Foster J., Sirmaci A., Gonzalez M., Mahdieh N., Fotouhi N., Bonyadi M., Cengiz F. B., Menendez I., et al.
PLOS ONE, cilt.7, sa.11, 2012 (SCI-Expanded)

XXX. **Screening of 38 Genes Identifies Mutations in 62% of Families with Nonsyndromic Deafness in Turkey**
DUMAN D., Sirmaci A., Cengiz F. B., ÖZDAĞ SEVGİLİ H., Tekin M.
GENETIC TESTING AND MOLECULAR BIOMARKERS, cilt.15, sa.1-2, ss.29-33, 2011 (SCI-Expanded)

XXXI. **Recurrent and Private MYO15A Mutations Are Associated with Deafness in the Turkish Population**
Cengiz F. B., DUMAN D., Sirmaci A., TOKGÖZ YILMAZ S., Erbek S., Oztukmen-Akay H., İNCESULU Ş. A., Edwards Y. J. K., ÖZDAĞ SEVGİLİ H., Liu X. Z., et al.
GENETIC TESTING AND MOLECULAR BIOMARKERS, cilt.14, sa.4, ss.543-550, 2010 (SCI-Expanded)

XXXII. **A Truncating Mutation in SERPINB6 Is Associated with Autosomal-Recessive Nonsyndromic Sensorineural Hearing Loss**
Sirmaci A., Erbek S., Price J., Huang M., DUMAN D., Cengiz F. B., Bademci G., TOKGÖZ YILMAZ S., ÖZTÜRK HİŞMİ B., ÖZDAĞ SEVGİLİ H., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.86, sa.5, ss.797-804, 2010 (SCI-Expanded)

XXXIII. **GJB2 Mutations in Mongolia: Complex Alleles, Low Frequency, and Reduced Fitness of the Deaf**
Tekin M., Xia X., Erdenetungalag R., Cengiz F. B., White T. W., Radnaabazar J., Dangaasuren B., Tastan H., Nance W. E., Pandya A.
ANNALS OF HUMAN GENETICS, cilt.74, ss.155-164, 2010 (SCI-Expanded)

XXXIV. **A FGF3 Mutation Associated With Differential Inner Ear Malformation, Microtia, and Microdontia**
Ramsebner R., Ludwig M., Parzefall T., Lucas T., Baumgartner W., Bodamer O., Cengiz F. B., Schoefer C., Tekin M., Frei K.
LARYNGOSCOPE, cilt.120, sa.2, ss.359-364, 2010 (SCI-Expanded)

XXXV. **Mutations in TMC1 contribute significantly to nonsyndromic autosomal recessive sensorineural hearing loss: A report of five novel mutations**
Sirmaci A., DUMAN D., Ozturkmen-Akay H., Erbek S., İNCESULU Ş. A., ÖZTÜRK HİŞMİ B., Arici Z. S., Yuksel-Konuk E. B., Tasir-Yilmaz S., Tokgoz-Yilmaz S., et al.
INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, cilt.73, sa.5, ss.699-705, 2009 (SCI-Expanded)

XXXVI. **Homozygous FGF3 mutations result in congenital deafness with inner ear agenesis, microtia, and microdontia**
TEKİN M. N., Akay H. O., Fitoz S., Birnbaum S., Cengiz F. B., SENNAROĞLU L., İNCESULU Ş. A., Konuk E. B. Y., Bayrak A. H., Senturk S., et al.
CLINICAL GENETICS, cilt.73, sa.6, ss.554-565, 2008 (SCI-Expanded)

XXXVII. **The effect of p.Arg25Cys alteration in NKX2-5 on conotruncal heart anomalies: Mutation or polymorphism?**
Akcaboy M. I., Cengiz F. B., Inceoglu B., UÇAR T., Atalay S., Tutar E., TEKİN M. N.
PEDIATRIC CARDIOLOGY, cilt.29, sa.1, ss.126-129, 2008 (SCI-Expanded)

XXXVIII. **Familial neonatal marfan syndrome due to parental mosaicism of a missense mutation in the FBN1 gene**
Tekin M., Cengiz F. B., Ayberkin E., Kendirli T., FİTOZ Ö. S., Tutar E., ÇİFTCİ E., Conba A.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.143A, sa.8, ss.875-880, 2007 (SCI-Expanded)

XXXIX. **SLC26A4 mutations are associated with a specific inner ear malformation**
FİTOZ Ö. S., Sennaroglu L., Incesulu A., Cengiz F. B., Koc Y., Tekin M.
INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, cilt.71, sa.3, ss.479-486, 2007 (SCI-Expanded)

XL. **Homozygous mutations in fibroblast growth factor 3 are associated with a new form of syndromic**

deafness characterized by inner ear agenesis, microtia, and microdontia
Tekin M., Hismi B. O., FİTOZ Ö. S., ÖZDAĞ SEVGİLİ H., Cengiz F. B., Sirmaci A., Aslan I., Inceoglu B., Yuksel-Konuk E. B., Yilmaz S. T., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.80, sa.2, ss.338-344, 2007 (SCI-Expanded)

XLI. **Analysis of NPHS2 mutations in Turkish steroid-resistant nephrotic syndrome patients**
ÖZÇAKAR Z. B., Cengiz F. B., Cakar N., Uncu N., Kara N., Acar B., Yuksel S., Ekim M., Tekin M., Yalcinkaya F.
PEDIATRIC NEPHROLOGY, cilt.21, sa.8, ss.1093-1096, 2006 (SCI-Expanded)

Diğer Dergilerde Yayınlanan Makaleler

I. **A Turkish case of incontinentia pigmenti with a deletion mutation at Inhibitor of kappa B kinase gamma gene**
Ergin F. B., Tekin M., Gunes M., Gunes B., Baysun S., Akar N.
EGYPTIAN JOURNAL OF MEDICAL HUMAN GENETICS, cilt.23, sa.1, 2022 (ESCI)

II. **m.3010G>A Değişikliğinin Türk Populasyonunda Siklik Kusma Sendromuna Etkisi**
ERGİN F. B., İNCİ A., OKUR İ., BİBEROĞLU G., TÜMER L., EZGÜ F. S.
Celal Bayar Üniversitesi Sağlık Bilimleri Enstitüsü Dergisi, 2022 (Hakemli Dergi)

III. **Mitochondrial Variants in Leber's Hereditary Optic Neuropathy in Turkish Patients.**
CENGİZ F. B., Yüce B., Çiftçi B.
Turkish Journal of Molecular Biology Biotechnology, cilt.3, ss.6-9, 2018 (Hakemli Dergi)

IV. **Dominant deafness-onychodystrophy syndrome caused by an ATP6V1B2 mutation**
Menendez I., Carranza C., Herrera M., Marroquin N., Foster J., Cengiz F. B., Bademci G., Tekin M.
CLINICAL CASE REPORTS, cilt.5, sa.4, ss.376-379, 2017 (ESCI)

V. **A New SOX9 Gene Mutation In A Case of Campomelic Dysplasia**
Yuksel-Konuk E. B., CENGİZ F. B., FİTOZ Ö. S., Tekin M.
TURKIYE KLINIKLERI PEDIATRI, cilt.18, ss.253-256, 2009 (Hakemli Dergi)

Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

I. **Ultra-Nadir Bir Hastalık ve Yeni Bir Mutasyon; Smg9 Eksikliği, Bir Aile 4Etkilenmiş Birey**
Ergin F. B., İnci A., Ezgü F. S.
6. Ulusal Çocuk Genetik Kongresini 09-12 Kasım 2023, Aydın, Türkiye, 9 - 11 Kasım 2023

II. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal Storage Disease?**
CENGİZ F. B., İNCİ A., BİBEROĞLU G., Çiftçi B., Topçu B., Tokgöz D., Yazar Ö. F., Gökmenoğlu H., Raj Y., OKUR İ., et al.
International Inborn Errors of Metabolism and Nutrition Congress, 10 - 14 Nisan 2019

III. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases?**
ERGİN F. B., İNCİ A., BİBEROĞLU G., ÇİFTÇİ B., TOPÇU YÜCE A. B., TOKGÖZ D., YAZAR Ö. F., GÖKMENOĞLU H., RAJ Y., OKUR İ., et al.
INTERNATIONAL INBORN ERRORS OFMETABOLISM AND NUTRITION CONGRESS 10 - 14 April 2019 Istanbul-Turkey, Türkiye, 10 - 14 Nisan 2019

Metrikler

Yayın: 51
Atıf (WoS): 1172
Atıf (Scopus): 1318

H-İndeks (WoS): 18
H-İndeks (Scopus): 20