

## 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 (YI) (Tezli), Türkiye 2004 - 2006

Lisans, Ankara Üniversitesi, Fen Fakültesi, Biyoloji Bölümü, Türkiye 1999 - 2004

### Yaptığı Tezler

Doktora, Sendromik olmayan otozomal resesif işitme kaybı ve MYO15A genine bağlantı bulunan ailelerde mutasyon analizi, Ankara Üniversitesi, Biyoteknoloji Enstitüsü, Temel Biyoteknoloji (Dr), 2010

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### 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**

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II. **A possibly new autoinflammatory disease due to compound heterozygous phosphomevalonate kinase gene mutation**

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

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- IV. **The first case with FBXL4 mutation successfully treated with a parenteral ketogenic diet for lactic acidosis**  
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- V. **Congenital defects of glycosylation: Novel presentations with mainly neurological involvement and variable dysmorphic features**  
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- VI. **Hypophosphatasia: is it an underdiagnosed disease even by expert physicians?**  
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- VII. **Two patients from Turkey with a novel variant in the GM2A gene and review of the literature**  
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- VIII. **Long-range cis-regulatory elements controlling GDF6 expression are essential for ear development**  
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- IX. **Spectrum of Genetic Variants Associated with Anterior Segment Dysgenesis in South Florida**  
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- X. **FOXF2 is required for cochlear development in humans and mice**  
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- XII. **Ripor2 is involved in auditory hair cell stereociliary bundle structure and orientation**  
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- XIII. **Identification of candidate gene FAM183A and novel pathogenic variants in known genes: High genetic heterogeneity for autosomal recessive intellectual disability**  
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- XIV. **Monosomy chromosome 21 compensated by 21q22.11q22.3 duplication in a case with small size and minor anomalies**  
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- XV. **MPZL2 is a novel gene associated with autosomal recessive nonsyndromic moderate hearing loss**  
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- XVI. **Novel pathogenic variants underlie SLC26A4-related hearing loss in a multiethnic cohort**  
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- XVII. **Novel EYA1 variants causing Branchio-oto-renal syndrome**  
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- XVIII. **Targeted Resequencing of Deafness Genes Reveals a Founder MYO15A Variant in Northeastern Brazil**  
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- XIX. **Audiological findings in Noonan syndrome**  
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- XX. **Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents**  
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- XXI. **Variations in Multiple Syndromic Deafness Genes Mimic Non-syndromic Hearing Loss**  
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- XXV. **Identification of Copy Number Variants Through Whole-Exome Sequencing in Autosomal Recessive Nonsyndromic Hearing Loss**  
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- XXVI. **FAM65B is a membrane-associated protein of hair cell stereocilia required for hearing**  
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- XXIX. **Whole-Exome Sequencing Efficiently Detects Rare Mutations in Autosomal Recessive Nonsyndromic Hearing Loss**  
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- XXXI. **Recurrent and Private MYO15A Mutations Are Associated with Deafness in the Turkish Population**  
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- XXXII. **A Truncating Mutation in SERPINB6 Is Associated with Autosomal-Recessive Nonsyndromic Sensorineural Hearing Loss**  
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- XXXV. **Mutations in TMC1 contribute significantly to nonsyndromic autosomal recessive sensorineural hearing loss: A report of five novel mutations**  
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- XXXVII. **The effect of p.Arg25Cys alteration in NKX2-5 on conotruncal heart anomalies: Mutation or polymorphism?**  
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- XXXVIII. **Familial neonatal marfan syndrome due to parental mosaicism of a missense mutation in the FBN1 gene**  
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- XXXIX. **SLC26A4 mutations are associated with a specific inner ear malformation**  
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- 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**

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### **XLI. Analysis of NPHS2 mutations in Turkish steroid-resistant nephrotic syndrome patients**

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## **Diğer Dergilerde Yayınlanan Makaleler**

- I. **A Turkish case of incontinentia pigmenti with a deletion mutation at Inhibitor of kappa B kinase gamma gene**  
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- 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.  
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- 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.  
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- V. **A New SOX9 Gene Mutation In A Case of Campomelic Dysplasia**  
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## **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., ÇİFTÇİ B., TOPÇU B., TOKGÖZ D., YAZAR Ö. F., GÖKMENOĞLU H., RAJ Y., OKUR İ., et al.  
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- III. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases?**  
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## **Metrikler**

Yayın: 51

Atf (WoS): 1172

Atf (Scopus): 1318

H-İndeks (WoS): 18

H-İndeks (Scopus): 20