

## Dr. Öğr. Üyesi FİLİZ BAŞAK ERGİN

### Kişisel Bilgiler

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### 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. Association between CLOCK gene polymorphisms with circadian rhythm, chrononutrition, dietary intake, and metabolic parameters in adolescents**  
Uyar G. O., YILDIRAN H., Teker-Duztas D., DALGIÇ B., KARAKAŞ N. M., ÇAMURDAN M. O., ERGİN F. B., EZGÜ F. S. FRONTIERS IN PUBLIC HEALTH, 2024 (SCI-Expanded)
- II. A very rare presentation of mitochondrial elongation factor Tu deficiency-TUFM mutation and literature review**  
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- III. A possibly new autoinflammatory disease due to compound heterozygous phosphomevalonate kinase gene mutation**  
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- IV. **Expected or unexpected clinical findings in liver glycogen storage disease type IX: distinct clinical and molecular variability**  
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- V. **The first case with FBXL4 mutation successfully treated with a parenteral ketogenic diet for lactic acidosis**  
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- VI. **Congenital defects of glycosylation: Novel presentations with mainly neurological involvement and variable dysmorphic features**  
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- VII. **Hypophosphatasia: is it an underdiagnosed disease even by expert physicians?**  
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- VIII. **Two patients from Turkey with a novel variant in the GM2A gene and review of the literature**  
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- IX. **Long-range cis-regulatory elements controlling GDF6 expression are essential for ear development**  
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- X. **Spectrum of Genetic Variants Associated with Anterior Segment Dysgenesis in South Florida**  
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- XI. **FOXF2 is required for cochlear development in humans and mice**  
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- XII. **Dysfunction of GRAP, encoding the GRB2-related adaptor protein, is linked to sensorineural hearing loss**  
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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. **Ripor2 is involved in auditory hair cell stereociliary bundle structure and orientation**  
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- XV. **Monosomy chromosome 21 compensated by 21q22.11q22.3 duplication in a case with small size and minor anomalies**  
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- XVI. **MPZL2 is a novel gene associated with autosomal recessive nonsyndromic moderate hearing loss**

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- XVII. **Novel pathogenic variants underlie SLC26A4-related hearing loss in a multiethnic cohort**  
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- XVIII. **Novel EYA1 variants causing Branchio-oto-renal syndrome**  
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- XIX. **Targeted Resequencing of Deafness Genes Reveals a Founder MYO15A Variant in Northeastern Brazil**  
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- XX. **Audiological findings in Noonan syndrome**  
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- XXI. **Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents**  
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- XXIII. **ROR1 is essential for proper innervation of auditory hair cells and hearing in humans and mice**  
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- XXV. **HPSE2 Mutations in Urofacial Syndrome, Non-Neurogenic Neurogenic Bladder and Lower Urinary Tract Dysfunction**  
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- XXVI. **Identification of Copy Number Variants Through Whole-Exome Sequencing in Autosomal Recessive Nonsyndromic Hearing Loss**  
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- XXVII. **FAM65B is a membrane-associated protein of hair cell stereocilia required for hearing**  
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- XXVIII. **Branchio-oculo-facial syndrome in a newborn caused by a novel TFAP2A mutation.**  
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- XXIX. **SLITRK6 mutations cause myopia and deafness in humans and mice**  
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- XXX. **Whole-Exome Sequencing Efficiently Detects Rare Mutations in Autosomal Recessive Nonsyndromic Hearing Loss**  
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- XXXI. **Screening of 38 Genes Identifies Mutations in 62% of Families with Nonsyndromic Deafness in Turkey**  
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- XXXII. **Recurrent and Private MYO15A Mutations Are Associated with Deafness in the Turkish Population**  
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- XXXIII. **A Truncating Mutation in SERPINB6 Is Associated with Autosomal-Recessive Nonsyndromic Sensorineural Hearing Loss**  
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- XXXV. **A FGF3 Mutation Associated With Differential Inner Ear Malformation, Microtia, and Microdontia**  
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- XXXVI. **Mutations in TMC1 contribute significantly to nonsyndromic autosomal recessive sensorineural hearing loss: A report of five novel mutations**  
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- XXXVIII. **The effect of p.Arg25Cys alteration in NKX2-5 on conotruncal heart anomalies: Mutation or polymorphism?**  
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- XXXIX. **Familial neonatal marfan syndrome due to parental mosaicism of a missense mutation in the FBN1 gene**  
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- XL. **SLC26A4 mutations are associated with a specific inner ear malformation**

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- XLI. **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.  
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- XLII. **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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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**  
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- V. **A New SOX9 Gene Mutation In A Case of Campomelic Dysplasia**  
Yuksel-Konuk E. B., CENGİZ F. B., FİTOZ Ö. S., Tekin M.  
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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: 53

Atıf (WoS): 1181

Atıf (Scopus): 1321

H-İndeks (WoS): 18

H-İndeks (Scopus): 20