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

- I. **Identification of novel MYH14 variants in families with autosomal dominant sensorineural hearing loss**  
DUMAN D., Ramzan M., Subasioglu A., MUTLU A., Peart L., Seyhan S., Guo S., Ila K., Balta B., KALCIOĞLU M. T., et al.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, 2024 (SCI-Expanded)
- II. **Genetic heterogeneity in hereditary hearing loss: Potential role of kinociliary protein TOGARAM2**  
Ramzan M., Zafeer M. F., Abad C., Guo S., Owrang D., Alper O., MUTLU A., Atik T., DUMAN D., Bademci G., et al.  
European Journal of Human Genetics, 2024 (SCI-Expanded)
- III. **Novel GPR156 variants confirm its role in moderate sensorineural hearing loss**  
Ramzan M., Bozan N., Seyhan S., Zafeer M. F., Ayral A., Duman D., Bademci G., Tekin M.  
SCIENTIFIC REPORTS, vol.13, no.1, 2023 (SCI-Expanded)
- IV. **Dispersed DNA variants underlie hearing loss in South Florida's minority population**  
Peart L., Gonzalez J., Morel Swols D., Duman D., Saridogan T., Ramzan M., Zafeer M. F., Liu X. Z., Eshraghi A. A., Hoffer M. E., et al.  
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- V. **Genome sequencing identifies coding and non-coding variants for non-syndromic hearing loss**  
Ramzan M., Duman D., Hendricks L. C. P., Guo S., Mutlu A., Kalcioğlu M. T., Seyhan S., Carranza C., Bonyadi M., Mahdieh N., et al.  
JOURNAL OF HUMAN GENETICS, vol.68, no.10, pp.657-669, 2023 (SCI-Expanded)
- VI. **Biallelic KITLG variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss**  
Vona B., Schwartzbaum D. A., Rodriguez A. A., Lewis S. S., Toosi M. B., Radhakrishnan P., Bozan N., Akın R., Doosti M., Manju R., et al.  
JOURNAL OF THE EUROPEAN ACADEMY OF DERMATOLOGY AND VENEREOLOGY, vol.36, no.9, pp.1606-1611, 2022 (SCI-Expanded)
- VII. **Mutations in MINAR2 encoding membrane integral NOTCH2-associated receptor 2 cause deafness in humans and mice**  
Bademci G., Lachgar-Ruiz M., Deokar M., Zafeer M. F., Abad C., Baylan M. Y., Ingham N. J., Chen J., Sineni C. J., Vadgama N., et al.  
Proceedings of the National Academy of Sciences of the United States of America, vol.119, no.26, 2022 (SCI-Expanded)
- VIII. **Whole Mitochondrial Genome Analysis in Turkish Patients with Mitochondrial Diseases**  
Gencer Öncül E. B., Duman D., Eminoğlu F. T., Aktuna S., Duman M. T.  
BALKAN MEDICAL JOURNAL, vol.39, no.2, pp.96-106, 2022 (SCI-Expanded)
- IX. **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, vol.130, no.8, pp.4213-4217, 2020 (SCI-Expanded)

- X. **Novel variant p.E269K confirms causative role of PLS1 mutations in autosomal dominant hearing loss**  
Diaz-Horta O., Bademci G., TOKGÖZ YILMAZ S., Guo S., Zafeer F., Sineni C. J., DUMAN D., Farooq A., Tekin M. CLINICAL GENETICS, vol.96, no.6, pp.575-578, 2019 (SCI-Expanded)
- XI. **Adams-Oliver syndrome caused by mutations of the EOGT gene**  
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- XII. **A truncating CLDN9 variant is associated with autosomal recessive nonsyndromic hearing loss**  
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- XIII. **Genetic Causes of Inner Ear Anomalies: a Review from the Turkish Study Group for Inner Ear Anomalies**  
OCAK E., DUMAN D., Tekin M. BALKAN MEDICAL JOURNAL, vol.36, no.4, pp.206-211, 2019 (SCI-Expanded)
- XIV. **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, vol.28, no.8, pp.1286-1297, 2019 (SCI-Expanded)
- XV. **Dysfunction of GRAP, encoding the GRB2-related adaptor protein, is linked to sensorineural hearing loss**  
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- XVI. **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., Elcioglu N. H., Celik P., Balci O., Cengiz F. B., Nunez D., Sineni C. J., Seyhan S., Kocaoglu D., et al. PLOS ONE, vol.13, no.11, 2018 (SCI-Expanded)
- XVII. **Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort**  
Meester J. A. N., Sukalo M., Schroeder K. C., Schanze D., Baynam G., Borck G., Bramswig N. C., DUMAN D., Gilbert-Dussardier B., Holder-Espinasse M., et al. HUMAN MUTATION, vol.39, no.9, pp.1246-1261, 2018 (SCI-Expanded)
- XVIII. **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, vol.137, no.6-7, pp.479-486, 2018 (SCI-Expanded)
- XIX. **Variants in CIB2 cause DFNB48 and not USH1J**  
Booth K. T., Kahrizi K., Babanejad M., Daghigh H., Bademci G., Arzhanghi S., Zareabdollahi D., Duman D., El-Amraoui A., Tekin M., et al. CLINICAL GENETICS, vol.93, no.4, pp.812-821, 2018 (SCI-Expanded)
- XX. **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., ATİK T., Huesca-Hernandez F., et al. INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, vol.101, pp.167-171, 2017 (SCI-Expanded)
- XXI. **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, vol.98, pp.59-63, 2017 (SCI-Expanded)
- XXII. **Variations in Multiple Syndromic Deafness Genes Mimic Non-syndromic Hearing Loss**  
Bademci G., Cengiz F. B., Foster J., Duman D., Sennaroglu L., Diaz-Horta O., Atik T., Kirazli T., Olgun L., Alper H., et al.

SCIENTIFIC REPORTS, vol.6, 2016 (SCI-Expanded)

- XXIII. **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.  
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- XXIV. **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.  
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- XXV. **Comprehensive analysis via exome sequencing uncovers genetic etiology in autosomal recessive nonsyndromic deafness in a large multiethnic cohort**  
Bademci G., Foster J., Mahdiah N., Bonyadi M., DUMAN D., Cengiz F. B., Menendez I., Diaz-Horta O., Shirkavand A., Zeinali S., et al.  
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- XXVI. **Novel mutations confirm that COL11A2 is responsible for autosomal recessive non-syndromic hearing loss DFNB53**  
Chakchouk I., Grati M., Bademci G., Bensaid M., Ma Q., Chakroun A., Foster J., Yan D., DUMAN D., Diaz-Horta O., et al.  
MOLECULAR GENETICS AND GENOMICS, vol.290, no.4, pp.1327-1334, 2015 (SCI-Expanded)
- XXVII. **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, vol.130, no.1, pp.54-58, 2015 (SCI-Expanded)
- XXVIII. **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.  
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- XXIX. **Analysis Of Hpse2 Gene Mutations In Children With Non-neurogenic Neurogenic Bladder And Urofacial (ochoa) Syndrome**  
Bulum B., ÖZÇAKAR Z. B., DUMAN D., Cengiz F. B., BURGU B., Baskin E., Cakar N., SOYGÜR Y. T., Ekim M., Tekin M., et al.  
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- XXX. **FAM65B is a membrane-associated protein of hair cell stereocilia required for hearing**  
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- XXXI. **Evidence for genotype-phenotype correlation for OTOF mutations**  
Yildirim-Baylan M., Bademci G., DUMAN D., Ozturkmen-Akay H., TOKGÖZ YILMAZ S., Tekin M.  
INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, vol.78, no.6, pp.950-953, 2014 (SCI-Expanded)
- XXXII. **BRANCHIO-OCULO-FACIAL SYNDROME IN A NEWBORN CAUSED BY A NOVEL TFAP2A MUTATION**  
Gunes N., Cengiz F. B., DUMAN D., Dervisoglu S., Tekin M., Tuysuz B.  
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- XXXIII. **High frequency of kidney and urinary tract anomalies in asymptomatic first-degree relatives of patients with CAKUT**  
BULUM AKBULUT B., ÖZÇAKAR Z. B., ÜSTÜNER E., DÜŞÜNCELİ ATMAN E., Kavaz A., DUMAN D., Walz K., FİTOZ Ö. S., Tekin M., Yalcinkaya F.  
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- XXXIV. **SLITRK6 mutations cause myopia and deafness in humans and mice**  
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K, Ota M., et al.

JOURNAL OF CLINICAL INVESTIGATION, vol.123, no.5, pp.2094-2102, 2013 (SCI-Expanded)

XXXV. **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, vol.7, no.11, 2012 (SCI-Expanded)

XXXVI. **MASP-1 is crucial for lectin pathway activation in human serum, while neither MASP-1 nor MASP-3 are required for alternative pathway function**

Degn S. E., Jensen L., Hansen A. G., DUMAN D., Tekin M., Jensenius J. C., Thiel S.

IMMUNOBIOLOGY, vol.217, no.11, pp.1218-1219, 2012 (SCI-Expanded)

XXXVII. **Mutations in OTOGL, Encoding the Inner Ear Protein Otogelin-like, Cause Moderate Sensorineural Hearing Loss**

Yariz K. O., DUMAN D., Seco C. Z., Dallman J., Huang M., Peters T. A., Sirmaci A., Lu N., Schraders M., Skromne I., et al.

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XXXVIII. **A truncating mutation in GPSM2 is associated with recessive non-syndromic hearing loss**

Yariz K. O., Walsh T., Akay H., Duman D., Akkaynak A. C., King M., Tekin M.

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XXXIX. **Cardiomyopathy with alopecia and palmoplantar keratoderma (CAPK) is caused by a JUP mutation**

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XL. **A founder TMIE mutation is a frequent cause of hearing loss in southeastern Anatolia**

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XLI. **Homozygous FGF3 mutations result in congenital deafness with inner ear agenesis, microtia, and microdontia**

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XLII. **Paternal X could relate to arithmetic function; study of cognitive function and parental origin of X chromosome in Turner syndrome**

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XLIII. **The c.IVS1+1G > A mutation in the GJB2 gene is prevalent and large deletions involving the GJB6 gene are not present in the Turkish population**

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XLIV. **A novel missense mutation in a C2 domain of OTOF results in autosomal recessive auditory neuropathy**

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XLV. **657del5 mutation of the Nijmegen breakage syndrome gene (NBS1) in the Turkish population**

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XLVI. **Screening the SLC26A4 gene in probands with deafness and goiter (Pendred syndrome) ascertained from a large group of students of the schools for the deaf in Turkey**

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## Articles Published in Other Journals

### I. Research of genetic bases of hereditary non-syndromic hearing loss

Subasioglu A., DUMAN D., Sirmaci A., Bademci G., Carkit F., SOMDAŞ M. A., Erkan M., Tekin M., DÜNDAR M.  
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## Refereed Congress / Symposium Publications in Proceedings

### I. Molecular diagnosis of lethal multiple pterygium syndrome in a fetus presenting with fetal akinesia deformation sequence

ÇETİN T., MUTLU H., DUMAN D., DUMAN M. T.

8. Uluslararası Erciyes Tıp Tıbbi Genetik Kongresi, Kayseri, Turkey, 21 September 2023

### II. Genome sequencing identifies coding and non-coding variants in hereditary deafness missed by exome sequencing

Ramzan M., Duman D., Hendricks L. C. P., Zafeer M. F., Bademci G., Tekin M.

American Society of Human Genetics 2022 Annual Meeting , California, United States Of America, 25 - 29 October 2022, pp.982

### III. Biallelic variants in KITLG cause Waardenburg syndrome type 2, albinism-deafness syndrome and oculocutaneous albinism

Girisha K., Vona B., Schwartzbaum D., Rodriguez A. A., Lewis S. S., Toosi M. B., Radhakrishnan P., Bozan N., Akın R., Doosti M., et al.

European Human Genetics Conference , Vienna, Austria, 11 - 14 June 2022, pp.369

### IV. Characterization of MINAR2 as a Novel Autosomal Recessive Deafness Gene

Bademci G., Lachgar-Ruiz ., Deokar M., Zafeer M. F., Abad ., Yildirim Baylan ., J. Ingham ., Chen ., J. Sinen C., Vadgama ., et al.

12th Molecular Biology of Hearing and Deafness Conference, Iowa, United States Of America, 24 - 27 May 2022, pp.18

### V. Expanding KITLG to Include Autosomal Recessive Inheritance, Oculocutaneous Albinism and Waardenburg/Albinism-Deafness Syndromes

Vona B., A. Schwartzbaum D., A. Rodriguez A., Lewis S. S., Toosi M. B., Radhakrishnan P., Bozan N., Akın R., Doosti M., Manju R., et al.

12th Molecular Biology of Hearing and Deafness Conference, Iowa, United States Of America, 24 - 27 May 2022, pp.97

### VI. Leigh Sendromu (LS) Ön Tanılı Bir Hastada Olası Genetik Etmenlerin Araştırılması

gencer öncül e. b., DUMAN D., EMİNOĞLU F. T., AKTUNA S., DUMAN M. T.

VI. Tıbbi Biyoloji ve Genetik Kongresi, Muğla, Turkey, 27 - 30 October 2019

### VII. işitme kaybının genetiği

DUMAN D.

V. Uluslararası Katılımlı Odyoloji Kongresi, Ankara, Turkey, 11 - 12 May 2019

### VIII. Whole Mitochondrial Genome Screening in Children with Suspected Mitochondrial Disease

GENCER ÖNCÜL E. B., DUMAN D., EMİNOĞLU F. T., AKTUNA S., DUMAN M. T.

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### IX. A founder mutation uncovers MPZL2 (DFNB11) as a novel autosomal recessive non-syndromic moderate hearing loss gene

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### X. Using Whole Genome Sequencing to Identify Novel Variants in Branchio-Oto-Renal Syndrome

Quynh P., hermidia a., amat d., gurkan h., DEMİR S., SEYHAN S., Sineni C. J., DUMAN D., Bademci G., Tekin M.  
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### XI. İŞİTME KAYBI GENETİĞİ

DUMAN D.

Ankara Tıp Kulak Burun Boğaz Kliniği 4. Alumni Toplantısı, Turkey, 14 - 15 December 2018

- XII. **Mitokondriyal kalıtım gösteren tanı konulamamış nadir nöroloji, psikiyatri, metabolizma hastalarında mitokondriyal DNA (mtDNA)'nın taranması**  
gencer oncül e. b., DUMAN D., EMİNOĞLU F. T., ERDOĞAN S., AKTUNA S., DUMAN B., DUMAN M. T.  
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- XIII. **Association of TOMM40 (Rs1160985 and Rs157581) Polymorphisms with Alzheimer Disease**  
RAJAB M. F., gencer oncül e. b., TAŞTAN H., DUMAN D., DİRİK E. B., DUMAN M. T.  
Uluslararası katılımlı 13. Ulusal Tıbbi Genetik Kongresi, Turkey, 7 - 11 November 2018
- XIV. **Three novel hearing loss genes reveal previously unrecognized roles of their protein products in the perception of sound.**  
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American Society of Human Genetics 2018 Annual Meeting, San Diego, 16 - 20 October 2018
- XV. **Sendromik olmayan işitme kaybılı Türk ailelerde sorumlu genlerin araştırılması**  
Duman D., Cengiz F. B., Bademci G., Subaşıoğlu A., Yılmaz S., Yıldırım Baylan M., Erbek H. S., Sennaroğlu L., Sennaroğlu G., Tekin M.  
Pediatrik Odyoloji Kongresi, Ankara, Turkey, 5 - 07 February 2018
- XVI. **A recessive variant in forkhead box domain of FOXF2 is associated with profound hearing loss and inner ear anomaly**  
Bademci G., Diaz-Horta O., İNCESULU Ş. A., Cengiz F. B., Abad C., Sineni C., Foster J., DUMAN D., FİTOZ Ö. S., Blanton S., et al.  
American Society of Human Genetics 2017 Annual Meeting, ORLANDO, United States Of America, 17 - 21 October 2017
- XVII. **Non Sendromik Ailesel İşitme Kayıplı Hastalarda Genetik Analiz**  
YILDIRIM H., YILDIRIM BAYLAN M., DUMAN D., Bademci G.  
5. Ulusal Otoloji Nörootoloji Kongresi, Antalya, Turkey, 4 - 07 May 2017
- XVIII. **Comprehensive Search for Deafness Genes Associated with Inner Ear Anomalies**  
Bademci G., Cengiz F. B., Diaz-Horta O., DUMAN D., Foster J., ATİK T., KİRAZLI T., ALPER H. H., Menendez I., PANDYA A., et al.  
American Society of Human Genetics 65th Annual Meeting, 6 - 10 October 2015
- XIX. **Tedavi Edilebilir Bir Otizm Nedeni Olarak BCKD-Kinaz Eksikliği: İki Olgu Sunumu**  
ÖZTÜRK HİŞMİ B., AKAY H., ÖZBEK M. N., BACANLI A., KUMRU B., DUMAN D., HİŞMİ A., Bademci G., Tekin M.  
XIII. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Adana, Turkey, 14 - 18 April 2015
- XX. **BCKD kinase deficiency Expanding the disease phenotypewith sensorineural hearing loss and transient acrodermatitisenteropathica like dermatitis**  
HİŞMİ B., ÖZTÜRKMEN AKAY H., BADEMCİ G., DUMAN D., BACANLI A., HİŞMİ A., KUMRU B., TEKİN M.  
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