

## Prof.Dr. Yasemin ALANAY

### Kişisel Bilgiler

İş Telefonu: [+90 216 500 4024](tel:+902165004024)

E-posta: [yasemin.alanay@acibadem.edu.tr](mailto:yasemin.alanay@acibadem.edu.tr)

Web: <https://avesis.acibadem.edu.tr/yasemin.alanay>

Posta Adresi: Büyükdere Cad. No:40 Sarıyer İstanbul

### Eğitim Bilgileri

Tıpta Yandal Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Türkiye 2002 - 2011

Doktora, Hacettepe Üniversitesi, Sağlık Bilimleri Enstitüsü, Pediatrik Temel Bilimler A.B.D., Türkiye 2004 - 2009

Post Doktora, University of California, Los Angeles, Cedars-Sinai Hospital, Intercampus Genetics Program, Skeletal Dysplasia Registry, Amerika Birleşik Devletleri 2005 - 2006

Tıpta Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Hemşirelik Bölümü, Türkiye 1997 - 2002

Lisans, Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Türkiye 1990 - 1996

### Araştırma Alanları

Tıp, Sağlık Bilimleri, Dahili Tıp Bilimleri, Çocuk Sağlığı ve Hastalıkları, Pediatrik Genetik ve Teratoloji

### Akademik Unvanlar / Görevler

Prof.Dr., Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2013 - Devam Ediyor

Doç.Dr., Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2011 - 2013

Doç.Dr., Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Hemşirelik Bölümü, 2008 - 2011

Yrd.Doç.Dr., Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Hemşirelik Bölümü, 2007 - 2008

### Akademik İdari Deneyim

Uygulama ve Araştırma Merkezi Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, 2017 - Devam Ediyor

Rektörlüğe Bağlı Komisyon Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2016 - Devam Ediyor

Uygulama ve Araştırma Merkezi Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, 2016 - Devam Ediyor

Anabilim/Bilim Dalı Başkanı, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2015 - Devam Ediyor

Rektörlüğe Bağlı Komisyon Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, 2015 - Devam Ediyor

Akreditasyon, Sürekli Gelişim ve Yenilenme Kurulu Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2018 - 2021

Akademik Performans D. Kurulu Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2018 - 2021

Üniversite Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2015 - 2021

Dekan, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, 2015 - 2021

Rektörlüğe Bağlı Komisyon Üyesi, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, 2015 - 2018

## Verdiği Dersler

Genetic Approach to Short stature, Lisans, 2020 - 2021

Pedigree Drawing Workshop, Lisans, 2020 - 2021

Common Chromosomal Disorders, Lisans, 2020 - 2021

Genetic tests for the Clinician, Lisans, 2020 - 2021

Pediatric History taking, Lisans, 2020 - 2021

PANEL: FROM GENOTYPE TO PHENOTYPE, Lisans, 2020 - 2021

Genetic Counselling, Lisans, 2020 - 2021

NONMENDELIAN INHERITANCE, Lisans, 2020 - 2021

Examination of The Extremities , Lisans, 2020 - 2021

## Jüri Üyelikleri

Ödül, Hacettepe Üniversitesi Tıp Ve Sağlık Bilimleri Ödüller, Hacettepe Üniversitesi, Haziran, 2020

Ödül, Hacettepe Üniversitesi Tıp ve Sağlık Bilimleri Ödülleri, Hacettepe Üniversitesi, Mayıs, 2019

## SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- I. **Determining T and B Cell development by TREC/KREC analysis in primary immunodeficiency patients and healthy controls**  
Şentürk G., Ng Y. Y. , Eltan S. B. , Başer D., Ogulur I., Altındirek D., Fırtına S., Yılmaz H., Kocamış B., Kıyıkım A., et al.  
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- II. **Safe and persistent growth-promoting effects of vosoritide in children with achondroplasia: 2-year results from an open-label, phase 3 extension study**  
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GENETICS IN MEDICINE, cilt.23, sa.12, ss.2443-2447, 2021 (SCI İndekslerine Giren Dergi)
- III. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**  
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CLEFT PALATE-CRANIOFACIAL JOURNAL, 2021 (SCI İndekslerine Giren Dergi)
- IV. **Re-analysis of whole-exome sequencing data reveals a novel splicing variant in the SLC2A1 in a patient with GLUT1 Deficiency Syndrome 1 accompanied by hemangioma: a case report**  
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BMC MEDICAL GENOMICS, cilt.14, sa.1, 2021 (SCI İndekslerine Giren Dergi)
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- VI. **Further defining the clinical and molecular spectrum of acromesomelic dysplasia type maroteaux: a Turkish tertiary center experience.**  
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Journal of human genetics, cilt.66, ss.585-596, 2021 (SCI İndekslerine Giren Dergi)

- VII. **Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders**  
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- VIII. **Whole Exome Sequencing of consanguineous families of clinically diagnosed with Neurodevelopmental Disorders**  
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EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.28, sa.SUPPL 1, ss.886-887, 2020 (SCI İndekslerine Giren Dergi)
- IX. **Clinical and Molecular Characterization of Fanconi Anemia Patients in Turkey.**  
Toksoy G., Uludağ A., Bagirova G., Avci Ş., Aghayev A., Günes N., Altunoğlu U., Alanay Y., Başaran S., Berkay E., et al.  
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- X. **Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia.**  
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- XI. **Once-daily, subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial**  
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- XIV. **Recurrent hydatidiform mole: When to stop ?**  
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Clinical and Experimental Obstetrics and Gynecology, cilt.47, sa.3, ss.424-426, 2020 (SCI İndekslerine Giren Dergi)
- XV. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome.**  
Tuc E., Bengur F., Aykut A., Sahin O., Alanay Y.  
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- XVI. **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**  
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- XVII. **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome**  
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- XVIII. **p.Ser348Cys mutation in FGFR3 gene leads to "Mild ACH /Severe HCH" phenotype.**  
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- XXIX. **A possible founder mutation in FZD6 gene in a Turkish family with autosomal recessive nail dysplasia.**  
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- XX. **Epigenotype and phenotype correlations in patients with Beckwith-Wiedemann syndrome**  
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- XXIII. **Parieto-occipital alopecia in early infancy mandates cranial imaging**  
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- XXIV. **Hematopoietic Stem Cell Transplantation for Myelodysplastic Syndrome in a Child With Klinefelter Syndrome**  
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- XXV. **A report of mosaic Turner syndrome with a mild Kabuki like phenotype**  
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- XXVI. **A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl Hydroxylation of Type I Procollagen**  
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- XXVIII. **Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa**  
Van Damme T., Gardeitchik T., Mohamed M., Guerrero-Castillo S., Freisinger P., Guillemin B., Kariminejad A., Dalloyaux D., Van Kraaij S., Lefeber D. J. , et al.  
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- XXIX. **AUTS2 Syndrome in a 68-Year-Old Female: Natural History and Further Delineation of the Phenotype**  
Sengun E., Yararbas K., Kasakyan S., ALANAY Y.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.170, sa.12, ss.3231-3236, 2016 (SCI İndekslerine Giren Dergi)
- XXX. **Expanding the clinical and mutational spectrum of the Ehlers-Danlos syndrome, dermatosparaxis type**  
Van Damme T., Colige A., Syx D., Giunta C., Lindert U., Rohrbach M., Aryani O., ALANAY Y., ŞİMŞEK KİPER P. Ö. , Kroes H. Y. , et al.  
GENETICS IN MEDICINE, cilt.18, sa.9, ss.882-891, 2016 (SCI İndekslerine Giren Dergi)
- XXXI. **Is 1p36 Deletion Associated with Anterior Body Wall Defects?**

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**XXXII. Late Sequel of Meningococemia: Skeletal Dysplasia**

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**XXXIII. Three cases of spondyloenchondrodysplasia (SPENCD) with systemic lupus erythematosus: a case series and review of the literature**

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**XXXIV. Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate**

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**XXXV. Intrauterine Imaging Strategies for Bone Disease**

Alanay Y.

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**XXXVI. Seizures and diagnostic difficulties in hyperinsulinism-hyperammonemia syndrome.**

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**XXXVII. Nonsense Mutation in SPARC Gene Causing Autosomal Recessive Osteogenesis Imperfecta**

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**XXXVIII. Hoyeraal-Hreidarsson Syndrome: An Extremely Rare Dyskeratosis Congenita Phenotype**

Bakar O., Isik U., Canpolat C., ALANAY Y.

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**XXXIX. Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome**

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**XLII. Mutations in LONP1, a Mitochondrial Matrix Protease, Cause CODAS Syndrome**

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- XLV. **A Study of the Clinical and Radiological Features in a Cohort of 93 Patients with a COL2A1 Mutation Causing Spondyloepiphyseal Dysplasia Congenita or a Related Phenotype**  
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- XLVI. **A VARIANT CASE OF 6p24 DELETION SYNDROME (OMIM #612582)**  
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- XLVII. **Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy**  
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- XLVIII. **Prepubertal Unilateral Gynecomastia: Report of 2 Cases**  
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- XLIX. **Partial monosomy 3q26.33-3q27.3 presenting with intellectual disability, facial dysmorphism, and diaphragm eventration: a case report**  
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- L. **A case of fucosidosis type II: diagnosed with dysmorphological and radiological findings**  
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- LI. **Etiological yield of SNP microarrays in idiopathic intellectual disability**  
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- LII. **Cathepsin K analysis in a pycnodysostosis cohort: demographic, genotypic and phenotypic features**  
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## Bilimsel Dergilerdeki Faaliyetler

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## Bilimsel Kuruluşlardaki Üyelikler / Görevler

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## Bilimsel Hakemlikler

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## Etkinlik Organizasyonlarındaki Görevler

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## Kongre ve Sempozyum Katılımı Faaliyetleri

Çukurova Pediatri Günleri, Davetli Konuşmacı, Adana, Türkiye, 2020  
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## Atıflar

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