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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 Medical Center, Amerika Birleşik Devletleri 2005 - 2006

Tıpta Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), 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, Tibbi Genetik

Akademik Unvanlar / Görevler

Prof.Dr., Acibadem Mehmet Ali Aydınlar Üniversitesi, Sağlık Bilimleri Enstitüsü, Genom Çalışmaları Ana Bilim Dalı, 2021 - Devam Ediyor

Prof.Dr., Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2020 - Devam Ediyor

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 - 2012

Doç.Dr., Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2008 - 2010

Dr.Öğr.Üyesi, Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2005 - 2008

Uzman, Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2002 - 2005

Akademik İdari Deneyim

Etik Kurul Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2021 - Devam Ediyor

Rektörlüğe Bağlı Komisyon Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü,

2020 - Devam Ediyor

Rektörlüğe Bağlı Komisyon Üyesi, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü,
2020 - Devam Ediyor

Uygulama ve Araştırma Merkezi Yönetim Kurulu Üyesi, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili
Tıp Bilimleri Bölümü, 2018 - Devam Ediyor

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

Verdiği Dersler

Developmental Genetics, Yüksek Lisans, 2021 - 2022

Genetic Approach to Short Stature, Lisans, 2021 - 2022

Developmental Genetics, Lisans, 2021 - 2022

Clinical Cytogenetics, Lisans, 2021 - 2022

Pediatric Genetics, Lisans, 2021 - 2022

Treatment of Genetic Diseases and Future of Clinical Genomics , Lisans, 2021 - 2022

Nonmendelian Inheritance, Lisans, 2021 - 2022

From Genotype to Phenotype, Lisans, 2021 - 2022

Pediatric History Taking, Lisans, 2021 - 2022

Mendelian Inheritance Patterns, Lisans, 2021 - 2022

Jüri Üyelikleri

Doçentlik Sınavı, Doçentlik Sınavı, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Mart, 2022

Doçentlik Sınavı, Doçentlik Sınavı, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Eylül, 2021

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- I. **Two new patients with acromesomelic dysplasia, PRKG2 type-identification and characterization of the first missense variant**
AKGÜN DOĞAN Ö., Diaz-Gonzalez F., de Lima Jorge A. A., Onenli-Mungan N., Menezes Andrade N. L., de Polli Cellin L., CEYLANER S., Barcellos Rosa Modkovski M., ALANAY Y., Heath K. E.
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- II. **The clinical phenotype of Koolen-de Vries syndrome in Turkish patients and literature review**
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AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.191, sa.7, ss.1814-1825, 2023 (SCI-Expanded)
- III. **< i >DNAJC21< /i >-related thrombocytopenia in a young adult female**
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AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS, cilt.193, sa.2, ss.193-197, 2023 (SCI-Expanded)
- IV. **Spondylo-meta-epiphyseal dysplasia (SMED), short limb-hand abnormal calcification type: Further expanding the mutational spectrum and dental findings of three new patients**
Akalin A., Ozsin C., KOÇ N., Demir G. U., Alanay Y., ÜTİNE G. E., BODUROĞLU O. K., Tekcicek M., Simsek-Kiper P. O.
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- V. **Persistent and stable growth promoting effects of vosoritide in children with achondroplasia for up to 3.5 years: results from an ongoing Phase 3 extension study**
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- HORMONE RESEARCH IN PAEDIATRICS, sa.SUPPL 2, ss.28-29, 2023 (SCI-Expanded)
- VI. **Obstacles and expectations of rare disease patients and their families in Turkiye: ISTisNA project survey results**
Hatinnaz Ng Ö., Sahin I., Erbilgin Y., Ozdemir O., Yucesan E., Erturk N., Yemenici M., Akgun Dogan Ö., Ugur Iseri S. A., Satman I., et al.
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- VII. **Undiagnosed diseases: Needs and opportunities in 20 countries participating in the Undiagnosed Diseases Network International**
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- VIII. **Growth parameters in children with achondroplasia: A 7-year, prospective, multinational, observational study**
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- IX. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**
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CLEFT PALATE-CRANIOFACIAL JOURNAL, cilt.59, sa.9, ss.1114-1124, 2022 (SCI-Expanded)
- X. **Defining mitochondrial protein functions through deep multiomic profiling**
Rensvold J. W., Shishkova E., Sverchkov Y., Miller I. J., ÇETİNKAYA A., Pyle A., Manicki M., Brademan D. R., Alanay Y., Raiman J., et al.
NATURE, cilt.606, ss.382-388, 2022 (SCI-Expanded)
- XI. **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**
Bozkurt T., ALANAY Y., ISIK U., SEZERMAN O. U.
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- XII. **Further defining the clinical and molecular spectrum of acromesomelic dysplasia type maroteaux: a Turkish tertiary center experience.**
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- XIII. **Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders**
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- XIV. **Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort**
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- XV. **Whole Exome Sequencing of consanguineous families of clinically diagnosed with Neurodevelopmental Disorders**
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- XVI. **Clinical and Molecular Characterization of Fanconi Anemia Patients in Turkey**
TOKSOY G., ULUDAĞ ALKAYA D., Bagirova G., Avci S., Aghayev A., Gunes N., Altunoglu U., ALANAY Y., BAŞARAN S., Berkay E. G., et al.
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- XVII. **Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia.**
 Ebrahimi-Fakhari D., Teinert J., Behne R., Wimmer M., D'Amore A., Eberhardt K., Brechmann B., Ziegler M., Jensen D., Nagabhyryava P., et al.
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- XVIII. **Once-daily, subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial**
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- XIX. **Recurrent hydatidiform mole: When to stop ?**
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- XX. **A Randomized Controlled Trial of Vosoritide in Children with Achondroplasia**
 Pogreen L., Savarirayan R., Tofts L., Rehab K., Irving M., Wilcox W., Bacino C., Hoover-Fong J., Ullot Font R., Harmatz P., et al.
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- XXI. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome**
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- XXII. **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**
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- XXIII. **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome**
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- XXIV. **p.Ser348Cys mutation in FGFR3 gene leads to "Mild ACH /Severe HCH" phenotype.**
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- XXV. **A possible founder mutation in FZD6 gene in a Turkish family with autosomal recessive nail dysplasia.**
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- XXVI. **Epigenotype and phenotype correlations in patients with Beckwith-Wiedemann syndrome**
 Bilgin B., Kabacam S., TAŞKIRAN Z. E., Simsek-Kiper P. O., ALANAY Y., BODUROĞLU O. K., ÜTİNE G. E.
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- XXVII. **Biallelic loss of human CTNNA2, encoding alpha N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration**
 Schaffer A. E., Breuss M. W., Caglayan A. O., Al-Sanaa N., Al-Abdulwahed H. Y., Kaymakcalan H., Yilmaz C., Zaki M. S., Rosti R. O., Copeland B., et al.
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- XXVIII. **Fragile x-associated premature ovarian failure in a large Turkish cohort: Findings of Hacettepe Fragile X Registry**
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- XXIX. Hematopoietic Stem Cell Transplantation for Myelodysplastic Syndrome in a Child With Klinefelter Syndrome**
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- XXX. Parieto-occipital alopecia in early infancy mandates cranial imaging**
Tuc E., KARAARSLAN E., Celik I., ALANAY Y.
CLINICAL DYSMORPHOLOGY, cilt.27, sa.1, ss.15-17, 2018 (SCI-Expanded)
- XXXI. A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl Hydroxylation of Type I Procollagen**
Duran I., Martin J. H., Weis M. A., Krejci P., Konik P., Li B., ALANAY Y., Lietman C., Lee B., Eyre D., et al.
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- XXXII. Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases**
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- XXXIII. Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa**
Van Damme T., Gardeitchik T., Mohamed M., Guerrero-Castillo S., Freisinger P., Guillemyn B., Kariminejad A., Dalloyaux D., Van Kraaij S., Lefeber D. J., et al.
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- XXXIV. AUTS2 Syndrome in a 68-Year-Old Female: Natural History and Further Delineation of the Phenotype**
Sengun E., Yararbas K., Kasakyan S., ALANAY Y.
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- XXXV. Expanding the clinical and mutational spectrum of the Ehlers-Danlos syndrome, dermatosparaxis type**
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- XXXVI. Is 1p36 Deletion Associated with Anterior Body Wall Defects?**
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- XXXVII. Three cases of spondyloenchondrodysplasia (SPENCD) with systemic lupus erythematosus: a case series and review of the literature**
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- XXXVIII. Late Sequel of Meningococcemia: Skeletal Dysplasia**
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- XXXIX. Intrauterine Imaging Strategies for Bone Disease**
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- XL. Seizures and diagnostic difficulties in hyperinsulinism-hyperammonemia syndrome.**
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- XLI. Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate**
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- XLII. Nonsense Mutation in SPARC Gene Causing Autosomal Recessive Osteogenesis Imperfecta**
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- XLIII. Hoyeraal-Hreidarsson Syndrome: An Extremely Rare Dyskeratosis Congenita Phenotype**
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- XLIV. Experience of a Skeletal Dysplasia Registry in Turkey: A Five-Years Retrospective Analysis**
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- XLV. RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
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- XLVI. Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome**
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- XLVII. Mutations in LONP1, a Mitochondrial Matrix Protease, Cause CODAS Syndrome**
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- XLVIII. Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffin-Siris and Nicolaides-Baraitser syndromes**
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- XLIX. Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3**
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- L. 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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- LI. A VARIANT CASE OF 6p24 DELETION SYNDROME (OMIM #612582)**
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- LII. Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy**
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- LIII. Prepubertal Unilateral Gynecomastia: Report of 2 Cases**
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- LIV. Partial monosomy 3q26.33-3q27.3 presenting with intellectual disability, facial dysmorphism, and diaphragm eventration: a case report**
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- LV. A case of fucosidosis type II: diagnosed with dysmorphological and radiological findings**
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