Prof. Yasemin ALANAY

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Education Information

Post Doctorate of Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Turkey 2002 - 2011
Doctorate, Hacettepe University, Sağlık Bilimleri Enstitüsü, Pediyatrik Temel Bilimler A.B.D., Turkey 2004 - 2009
Post Doctorate, University of California, Los Angeles, Cedars-Sinai Medical Center, United States Of America 2005 - 2006
Expertise In Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Turkey 1997 - 2002
Undergraduate, Hacettepe University, Tıp Fakültesi (İngilizce), Turkey 1990 - 1996

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Child Health and Diseases, Pediatric Genetics and Teratology, Medical Genetics

Academic Titles / Tasks

Professor, Acibadem Mehmet Ali Aydinlar University, Graduate School Of Health Sciences, Department Of Genomic Studies, 2021 - Continues
Professor, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2020 - Continues
Professor, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2013 - Continues
Associate Professor, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2011 - 2012
Associate Professor, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2008 - 2010
Assistant Professor, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2005 - 2008
Expert, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2002 - 2005

Academic and Administrative Experience

Ethics Committee Member, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2021 - Continues
Rectorate Commissioner, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2020 - Continues
Rectorate Commissioner, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2020 - Continues
Uygulama ve Araştırma Merkezi Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2018 - Continues
Dean, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2015 - 2021

Courses

Developmental Genetics, Postgraduate, 2021 - 2022
Genetic Approach to Short Stature, Undergraduate, 2021 - 2022
Developmental Genetics, Undergraduate, 2021 - 2022
Clinical Cytogenetics, Undergraduate, 2021 - 2022
Pediatric Genetics, Undergraduate, 2021 - 2022
Treatment of Genetic Diseases and Future of Clinical Genomics , Undergraduate, 2021 - 2022
Nonmendelian Inheritance, Undergraduate, 2021 - 2022
From Genotype to Phenotype, Undergraduate, 2021 - 2022
Pediatric History Taking, Undergraduate, 2021 - 2022
Mendelian Inheritance Patterns, Undergraduate, 2021 - 2022

Jury Memberships

Associate Professor Exam, Associate Professor Exam, Acibadem Mehmet Ali Aydinlar University, March, 2022
Associate Professor Exam, Associate Professor Exam, Acibadem Mehmet Ali Aydinlar University, September, 2021

Published journal articles indexed by SCI, SSCI, and AHCI

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II. **The clinical phenotype of Koolen-de Vries syndrome in Turkish patients and literature review**
KARAMIK G., Tuysuz B., Isik E., Yilmaz A., ALANAY Y., Sunamak E. C., Durmusalioglu E. A., Ozkinay F., Cetin G. O., ÖZTÜRK N., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.191, no.7, pp.1814-1825, 2023 (SCI-Expanded)

III. **DNAJC21-related thrombocytopenia in a young adult female**
ASLAN D., AKGÜN DOĞAN Ö., Ay B., ÇAMURDAN M. O., Mancilar H., ALANAY Y.
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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
Polgreen L. E., Savarirayan R., Tofts L., Irving M., Wilcox W., Bacino C. A., Hoover-Fong J., Font R. U. F. U., Harmatz P., Rutsch F., et al.
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VI. **Obstacles and expectations of rare disease patients and their families in Turkiye: ISTisNA project survey results**
Hatirnaz 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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IX. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**
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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**
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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**
Bertoli-Avella A. M., Beetz C., Ameziane N., Rocha M. E., Guatibonza P., Pereira C., Calvo M., Herrera-Ordonez N., Segura-Castel M., Diego-Alvarez D., et al.
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XV. **Whole Exome Sequencing of consanguineous families of clinically diagnosed with Neurodevelopmental Disorders**
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XXI. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome**
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AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.7, pp.1157-1172, 2019 (SCI-Expanded)

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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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XXVII. **Biallelic loss of human CTNNA2, encoding alpha N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration**
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XXXI. **Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases**
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XXXII. **A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl Hydroxylation of Type I Procollagen**
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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.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.100, no.2, pp.216-227, 2017 (SCI-Expanded)

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.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.170, no.12, pp.3231-3236, 2016 (SCI-Expanded)

XXXV. **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., Simsek-Kiper P. O., Kroes H. Y., et al.
GENETICS IN MEDICINE, vol.18, no.9, pp.882-891, 2016 (SCI-Expanded)

XXXVI. **Is 1p36 Deletion Associated with Anterior Body Wall Defects?**
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XXXVII. **Late Sequel of Meningococcemia: Skeletal Dysplasia**
Karabulut G. S., Acar T., Yuksel A. B., ALANAY Y., Hatun S.
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XXXVIII. **Three cases of spondyloenchondrodysplasia (SPENCD) with systemic lupus erythematosus: a case series and review of the literature**
BİLGİNER Y., DÜZOVA A., TOPALOĞLU R., BATU AKAL E. D., BODUROĞLU O. K., Gucer S., Bodur I., ALANAY Y.
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XXXIX. **Nonsense Mutation in SPARC Gene Causing Autosomal Recessive Ostegenesis Imperfecta**
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XL. **Intrauterine Imaging Strategies for Bone Disease**
Alanay Y.
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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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Boegershausen N., Tsai I., Pohl E., Kiper P. O. S., Beleggia F., PERÇİN F. E., Keupp K., Matchan A., Milz E., ALANAY Y., et al.
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LI. **Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy**
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LV. **A case of fucosidosis type II: diagnosed with dysmorphological and radiological findings**
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Supported Projects

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Activities in Scientific Journals

Clinical Dysmorphology, Committee Member, 2010 - Continues
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Memberships / Tasks in Scientific Organizations

Çocuk Genetik Hastalıkları Derneği, Member, 2011 - Continues, Turkey
Türk Pediatri Kurumu, Member, 2009 - Continues, Turkey
International Skeletal Dysplasia Society, Member, 2005 - Continues, Switzerland
European Society of Human Genetics, Member, 2003 - Continues, Austria
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Tasks In Event Organizations

Alanay Y., European Society of Human Genetics Meeting, Scientific Congress, Berlin, Germany, Haziran 2020
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