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 Hospital, Intercampus Genetics Program, Skeletal Dysplasia Registry, United States Of America 2005 - 2006
Expertise In Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Nursing, 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

Academic Titles / Tasks

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 - 2013
Associate Professor, Hacettepe University, Tıp Fakültesi (Türkçe), Nursing, 2008 - 2011
Assistant Professor, Hacettepe University, Tıp Fakültesi (Türkçe), Nursing, 2007 - 2008

Academic and Administrative Experience

Uygulama ve Araştırma Merkezi Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydinlar University, School of Medicine, 2017 - Continues
Rectorate Commissioner, Acibadem Mehmet Ali Aydinlar University, School of Medicine, Department of Medical Sciences, 2016 - Continues
Uygulama ve Araştırma Merkezi Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydinlar University, School of Medicine, 2016 - Continues
Head of Department, Acibadem Mehmet Ali Aydinlar University, School of Medicine, Department of Medical Sciences, 2015 - Continues
Rectorate Commissioner, Acibadem Mehmet Ali Aydinlar University, School of Medicine, 2015 - Continues
Akreditasyon, Sürekli Gelişim ve Yenilenme Kurulu Üyesi, Acibadem Mehmet Ali Aydinlar University, School of Medicine, Department of Medical Sciences, 2018 - 2021
Academic Performance D. Board Member, Acibadem Mehmet Ali Aydinlar University, School of Medicine, Department of

Medical Sciences, 2018 - 2021
University Executive Board Member, Acibadem Mehmet Ali Aydinlar University, School of Medicine, Department of Medical Sciences, 2015 - 2021
Dean, Acibadem Mehmet Ali Aydinlar University, School of Medicine, 2015 - 2021
Rectorate Commissioner, Acibadem Mehmet Ali Aydinlar University, School of Medicine, 2015 - 2018

Courses

Genetic Approach to Short stature, Undergraduate, 2020 - 2021
Pedigree Drawing Workshop, Undergraduate, 2020 - 2021
Common Chromosomal Disorders, Undergraduate, 2020 - 2021
Genetic tests for the Clinician, Undergraduate, 2020 - 2021
Pediatric History taking, Undergraduate, 2020 - 2021
PANEL: FROM GENOTYPE TO PHENOTYPE, Undergraduate, 2020 - 2021
Genetic Counselling, Undergraduate, 2020 - 2021
NONMENDELIAN INHERITANCE, Undergraduate, 2020 - 2021
Examination of The Extremities , Undergraduate, 2020 - 2021

Jury Memberships

Award, Hacettepe Üniversitesi Tıp Ve Sağlık Bilimleri Ödüller, Hacettepe Üniversitesi, June, 2020
Award, Hacettepe Üniversitesi Tıp ve Sağlık Bilimleri Ödülleri, Hacettepe Üniversitesi, May, 2019

Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

I. **Determining T and B Cell development by TREC/KREC analysis in primary immunodeficiency patients and healthy controls**
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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, vol.23, no.12, pp.2443-2447, 2021 (Journal Indexed in SCI)

III. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**
GÜLERAY LAFCI N., KOŞUKCU C., OĞUZ S., ÜREL DEMİR G., Taskiran E. Z. , ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E. , ALANAY Y., BODUROĞLU O. K. , ALİKAŞİFOĞLU M.
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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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V. **Spondyloepimetaphyseal dysplasia EXTL3-deficient type: Long-term follow-up and review of the literature**
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VI. **Further defining the clinical and molecular spectrum of acromesomelic dysplasia type maroteaux: a Turkish tertiary center experience.**

Simsek-Kiper P. O. , Urel-Demir G., Taskiran E. Z. , Arslan U. E. , Nur B., Mihci E., Haliloglu M., Alanay Y., Utine G. E. , Boduroglu K.

Journal of human genetics, vol.66, pp.585-596, 2021 (Journal Indexed in SCI)

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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IX. **Clinical and Molecular Characterization of Fanconi Anemia Patients in Turkey.**

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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**

Savarirayan R., Tofts L., Irving M., Wilcox W., Bacino C. A. , Hoover-Fong J., Ullot Font R., Harmatz P., Rutsch F., Bober M. B. , et al.

LANCET, vol.396, no.10252, pp.684-692, 2020 (Journal Indexed in SCI)

XII. **Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort**

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XV. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome.**

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XVI. **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**

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Bilginer Y., Duzova A., Topaloglu R., Batu E. D. , Boduroglu K., Gucer S., Bodur I., Alanay Y.
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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.**
Aka S., ALANAY Y., Boodhansingh K. E. , Stanley C. A. , Semiz S.
The Turkish journal of pediatrics, vol.58, no.5, pp.541-544, 2016 (Journal Indexed in SCI)

XXXVII. **Nonsense Mutation in SPARC Gene Causing Autosomal Recessive Ostegenesis Imperfecta**
Abali S., ARMAN A., Atay Z., Bas S., Cam S., Gormez Z., Demirci H., ALANAY Y., Akarsu N., BEREKET A., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.167, 2016 (Journal Indexed in SCI)

XXXVIII. **Hoyeraal-Hreidarsson Syndrome: An Extremely Rare Dyskeratosis Congenita Phenotype**
Bakar O., Isik U., Canpolat C., ALANAY Y.
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XL. **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
Boegershausen N., Tsai I., Pohl E., ŞİMŞEK KİPER P. Ö. , Beleggia F., Percin E. F. , Keupp K., Matchan A., Milz E., Alanay Y., et al.
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XLI. **Experience of a Skeletal Dysplasia Registry in Turkey: A Five-Years Retrospective Analysis**
Kurt-Sukur E. D. , ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E. , Boduroglu K., Alanay Y.
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XLII. **Mutations in LONP1, a Mitochondrial Matrix Protease, Cause CODAS Syndrome**
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Hatırnaz Ng Ö., Özbek U., Alanay Y., Project Supported by Higher Education Institutions, Albinizm Hastalıklarında gen varyantlarının ekzom analizi ile belirlenmesi, 2020 - 2021
Hatırnaz Ng Ö., Özbek U., Alanay Y., Turkey Institutes of Health Administration Project, Bireysel ve Dönüşümsel Tıp Alanındaki Uygulamalı Proje Çağrısı: Albinizm, 2019 - 2021
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Özbek U., Hatırnaz Ng Ö., Alanay Y., Project Supported by Higher Education Institutions, Genetic predisposition to leukemia and lymphoma, 2019 - 2020
Karaayvaz S., Alanay Y., Abalı S., Project Supported by Private Organizations in Other Countries, The intend and design of this study is to assess BMN 111 as a therapeutic option for the treatment of children with Achondroplasia, 2018 - 2020

Activities in Scientific Journals

AMERICAN JOURNAL OF MEDICAL GENETICS, Committee Member, 2010 - Continues
Clinical Dysmorphology, Committee Member, 2010 - Continues

Memberships / Tasks in Scientific Organizations

European Calcified Tissue Society Scientific Program Committee, Member of Science Committee, 2019 - Continues, Belgium
European Society of Human Genetics, Scientific Program Committee, Member of Science Committee, 2017 - Continues, Austria

European Society of Human Genetics, Board Member, Executive Board Member, 2013 - 2018

Scientific Refereeing

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, Journal Indexed in SCI-E, May 2020

Tasks In Event Organizations

Alanay Y., European Society of Human Genetics Meeting, Scientific Congress, Berlin, Germany, Haziran 2020
Alanay Y., ECTS 2020 European Calcified Tissue Society Meeting, Scientific Congress, Marseille, France, Mayıs 2020
Akyerli Boylu C., Özbek U., Hatırnaz Ng Ö., Alanay Y., Ağırbaşlı D., Öz Arslan D., Clinical Genomics and NGS Hybrid Course, Scientific Congress, İstanbul, Turkey, Mayıs 2019

Edit Congress and Symposium Activities

Çukurova Pediatri Günleri, Invited Speaker, Adana, Turkey, 2020
Nadir İskelet Hastalıkları Sempozyumu, Invited Speaker, İstanbul, Turkey, 2020
4. Ulusal Çocuk Genetik Kongresi, Invited Speaker, Ankara, Turkey, 2019
13th Balkan Congress of Human Genetics, Invited Speaker, Edirne, Turkey, 2019
3. Ege Endokrin Hastalıkları ve Genetik Sempozyumu, Invited Speaker, İzmir, Turkey, 2019

Citations

Total Citations (WOS):3271
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Invited Talks

Klinisyen için Genetikte Yenilikler, Seminar, Zeynep Kamil Hastanesi, Turkey, December 2019
Kalıtsal Kemik Kırılganlığında Genetik, Conference, Hacettepe Üniversitesi, Turkey, October 2019
A Pediatrician's Experience with WGS, Conference, University of Pennsylvania, United States Of America, August 2019
Akondroplazi, Conference, Doğu Grubu Pediatrik Endokrin Toplantısı, Turkey, March 2019