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Education

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

Academic Positions

Professor 2013 - Present	Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences
Associate Professor 2011 - 2012	Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences

Associate Professor 2008 - 2010	Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü
Assistant Professor 2005 - 2008	Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü
Expert 2002 - 2005	Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü

Funded Projects

1. Akgün Doğan Ö., Sütçübaşı B., Alanay Y., Other International Funding Programs, Arid1b Ile İlişkili Bozuklukta Gelişimsel İlerleme Alanları - Çok Yöntemli Çok Merkezli Prospektif Doğal Seyir Çalışması, 2024 - 2027
2. Akgün Doğan Ö., Alanay Y., Abalı S., Özdemir Ö., TÜBİTAK International Bilateral Joint Cooperation Program Project, Combining Multidisciplinary Re-phenotyping with Re-analysis of WES/WGS Data in Patients with Undiagnosed Skeletal Dysplasias, 2024 - 2026
3. Akgün Doğan Ö., Alanay Y., Özek M., Project Supported by Other Official Institutions, ARID1B İle İlişkili Bozuklukta Gelişimsel İlerleme Alanları - Çok Yöntemli Çok Merkezli Prospektif Doğal Seyir Çalışması, 2024 - 2025
4. Akgün Doğan Ö., Alanay Y., Özek M., Project Supported by Other Official Institutions, Acıbadem Üniversitesi Tıp Fakültesi'nde Kraniosinostoz Kayıt Sisteminin Oluşturulması: Ulusal Kayıt Sistemi için Temel Oluşturma Çalışması (Establishing a Craniosynostosis Registry in Acibadem Universty School of Medicine, Turkey: A Nucleus for a National Registry), 2024 - 2025
5. Güven G., Tahir Turanlı E., Alanay Y., Project Supported by Other Official Institutions, Progresif Psödoromatoid Displazi Hastalığı Geni Olan CCN6'de in vitro Oluşturulan Patojenik Varyantların Fonksiyonel Analizleri , 2024 - 2025
6. Güven G., Alanay Y., Tahir Turanlı E., TUBITAK Project, Progresif Psödoromatoid Displazi Geni olan CCN6'daki Patojenik Varyantların İşlevsel Analizleri İçin Kondrosit Hücre Hatlarında Modellenmesi , 2022 - 2024
7. Bilgüvar K., Hatırnaz Ng Ö., Özdemir Ö., Akgün Doğan Ö., Alanay Y., Özbek U., Development Agency, İStanbul Tanısız ve NAdir Hastalıklara Çözüm Platformu-İSTisNA, 2022 - 2024
8. Alanay Y., Özbek U., Hatırnaz Ng Ö., Akgün Doğan Ö., Özdemir Ö., Çıtak A., Korkmaz Toygar A., Beken S., Kazancı E., Demirel A., et al., Project Supported by Higher Education Institutions, Yenidoğan ve Çocuk Yoğun Bakım Ünitesinde (YYBÜ/ÇYBÜ) Hızla Kötüleşen Kritik Yenidoğan ve Süt Çocuklarında Hızlı Yeni Nesil Dizileme ile Genetik Tanı, 2022 - 2024
9. Akgün Doğan Ö., Alanay Y., Hatırnaz Ng Ö., Açıkel Elmas M., Project Supported by Higher Education Institutions, "Herediter Spastik Paraparezili Bir Ailede Klinik İnceleme ve ERLIN1 Geninin Hastalık Patofizyolojisindeki Yerinin Aydınlatılması", 2022 - 2023
10. Özbek U., Alanay Y., Sezerman O. U., Julkowska D., EU Framework Program Project, European Research Program on Rare Diseases, 2019 - 2023
11. Hatırnaz Ng Ö., Özbek U., Alanay Y., Project Supported by Higher Education Institutions, Albinizm Hastalıklarında gen varyantlarının ekzom analizi ile belirlenmesi, 2020 - 2021
12. Hatırnaz Ng Ö., Özbek U., Alanay Y., Research Project of the Presidency of Turkey Health Institutes (TÜSEB), Bireysel ve Dönüşümsel Tıp Alanındaki Uygulamalı Proje Çağrısı: Albinizm, 2019 - 2021
13. Özbek U., Hatırnaz Ng Ö., Alanay Y., Development Agency, İSTisNA – İStanbul Tanısız ve NAdir Hastalıklara Çözüm Platformu Fizibilite Projesi, 2019 - 2021
14. Karaayvaz S., Alanay Y., Abalı S., Project Supported by Private Organizations in Other Countries, Akondroplazisi olan pediatrik hastalar için çok merkezli, çok uluslu klinik değerlendirme , 2016 - 2021

15. Özbek U., Hatırnaz Ng Ö., Alanay Y., Project Supported by Higher Education Institutions, Genetic predisposition to leukemia and lymphoma, 2019 - 2020
16. 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

Journal articles indexed in SCI, SSCI, and AHCI

1. **BCL11B-related disease: a single phenotypic entity?**
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   European journal of human genetics : EJHG, vol.33, no.4, pp.451-460, 2025 (SCI-Expanded)
2. **Evaluation of the etiology of epilepsy and/or developmental delay in children via next-generation sequencing: a single-center experience**
   Kava H., AKGÜN DOĞAN Ö., Yesilyurt A., ALANAY Y., Isik U.
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3. **A global survey about undiagnosed rare diseases: perspectives, challenges, and solutions**
   Baldovino S., Sciascia S., Carta C., Salvatore M., Cellai L. L., Ferrari G., Lumaka A., Groft S., Alanay Y., Azam M., et al.
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4. **HPDL Variant Type Correlates With Clinical Disease Onset and Severity**
   Lee E. H., Kim-Mcmanus O., Yang J. H., Haas R., Zaki M. S., Abdel-Salam G. M. H., Nakamura Y., Abdel-Hamind M. S., Ebrahimi-Fakhari D., Alecu J. E., et al.
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5. **CFAP99 deficiency leads to heterotaxy and scoliosis**
   Ay B., Viviano S., AKGÜN DOĞAN Ö., ALANAY Y., Deniz E.
   EUROPEAN JOURNAL OF HUMAN GENETICS, pp.1747-1748, 2024 (SCI-Expanded)
6. **Impact of deep phenotyping: High diagnostic yield in a diverse pediatric population of 172 patients through clinical whole-genome sequencing at a single center**
   AKGÜN DOĞAN Ö., Bengur E. T., Ay B., Ozkose G. S., Kar E., Bengur F. B., Bulut A., Yigit A., Aydin E., Esen F. N., et al.
   EUROPEAN JOURNAL OF HUMAN GENETICS, pp.1560, 2024 (SCI-Expanded)
7. **Statistical analysis of genomic in-silico pathogenicity predictors for the characterization of VUS in rare and undiagnosed disorders**
   AYDIN E., Ergun B., AKGÜN DOĞAN Ö., ALANAY Y., HATIRNAZ NG Ö., ÖZDEMİR Ö.
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10. **ITGB4-Related pyloric atresia without epidermolysis in two siblings.**
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13. **Real-world experience with vosoritide treatment in achondroplasia: A single-center report from Turkey**
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14. **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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15. **GROWTH-PROMOTING EFFECTS OF VOSORITIDE IN CHILDREN WITH ACHONDROPLASIA AGED=10 YEARS AT TREATMENT INITIATION: RESULTS FROM A PHASE 3 EXTENSION STUDY**
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16. **PERSISTENT GROWTH-PROMOTING EFFECTS OF VOSORITIDE IN CHILDREN WITH ACHONDROPLASIA FOR UP TO 4 YEARS: UPDATE FROM PHASE 3 EXTENSION STUDY**
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17. **PERSISTENT GROWTH-PROMOTING EFFECTS OF VOSORITIDE IN CHILDREN WITH ACHONDROPLASIA IS ACCOMPANIED BY IMPROVEMENT IN PHYSICAL ASPECTS OF QUALITY OF LIFE**
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18. **Rapid genome sequencing for critically ill infants: an inaugural pilot study from Turkey.**
Guner Yilmaz B., Akgun-Dogan Ö., Ozdemir Ö., Yuksel B., Hatirnaz Ng Ö., Bilguvar K., Ay B., Ozkose G. S., Aydin E., Yigit A., et al.
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19. **A Potentially Treatable Genetic Disorder Which Presented with Neuropsychiatric Involvement and Drug-Resistant Focal Epilepsy: Niemann-Pick Disease Type C**
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20. **Impact of deep phenotyping: high diagnostic yield in a diverse pediatric population of 172 patients through clinical whole-genome sequencing at a single center**
Akgün Doğan Ö., Bengur E. T., Ay B., Ozkose G. S., Kar E., Bengur F. B., Bulut A., Yigit A., Aydin E., Esen F. N., et al.
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21. **A new line method; A direct test in spinal muscular atrophy screening for DBS**
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22. **Treatment dilemma in a prepubertal patient with ACAN mutation but without advanced bone age**
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23. **An adolescent boy with PLS3 mutation causing severe thoracic kypho-scoliosis**
Kilci F., JONES J. H., GÜRPINAR G., KOÇYİĞİT E., ÇAKIR Ö., CEYLANER S., ALANAY Y., ÇİZMECİOĞLU JONES F. M.
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24. **The clinical phenotype of Koolen-de Vries syndrome in Turkish patients and literature review**
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25. **Persistent growth-promoting effects of vosoritide in children with achondroplasia for up to 3.5 years: update from Phase 3 extension study**
Polgreen L. E., Savarirayan R., Tofts L., Irving M., Wilcox W. W., Bacino C. A., Hoover-Fong J., Ullot Font R., Harmatz P., Rutsch F., et al.
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26. **Real-world evidence in achondroplasia: considerations for a standardized data set**
ALANAY Y., Mohnike K., Nilsson O., Alves I., AlSayed M., Appelman-Dijkstra N. M., Baujat G., Ben-Omran T., Breyer S., Cormier-Daire V., et al.
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27. **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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28. **Clinical Heterogeneity and Different Phenotypes in Patients with SETD2 Variants: 18 New Patients and Review of the Literature**
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29. **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.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.66, no.4, 2023 (SCI-Expanded)
30. **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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31. **Obstacles and expectations of rare disease patients and their families in Turkiye: ISTisNA project survey results**
Hatirnaz Ng Ö., Sahin I., Erbilgin Y., Ozdemir Ö., Yucesan E., Erturk N., Yemenici M., Akgun Dogan Ö., Ugur Iseri S. A., Satman I., et al.
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32. **Unmet needs in countries participating in the undiagnosed diseases network international: an international survey considering national health care and economic indicators**
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33. **Undiagnosed diseases: Needs and opportunities in 20 countries participating in the Undiagnosed Diseases Network International**
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34. **Growth parameters in children with achondroplasia: A 7-year, prospective, multinational, observational study**
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35. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**
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36. **Defining mitochondrial protein functions through deep multiomic profiling**
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37. **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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39. **Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders**
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41. **Whole Exome Sequencing of consanguineous families of clinically diagnosed with Neurodevelopmental Disorders**
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58. **A custom Amplisec arrhytmia panel comprimising 68 cardiac chanelopathy genes is a gold standard for the rapid and sensitive detection of genetic variation in long Qt syndrome**
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64. **Bir Olgu Nedeni ile PTH Direnci Pseudohipoparatiroidi tip1 A Albright Herediter Osteodistrofisi mi Akrodisostoz mu**
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RAHİME GÜL Y. M., ALANAY Y., ÖZSU E., ÇİZMECİOĞLU F. M., HATUN Ş.
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RAHİME GÜL Y. M., ÖZSU E., ÇİZMECİOĞLU F. M., ALANAY Y., HATUN Ş.
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69. **Rapid prenatal diagnosis of common aneuploidies by QF-PCR, four years experience of Hacettepe University**
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7th European Cytogenetics Conference, Stockholm, Sweden, 4 - 07 July 2009, vol.17, pp.209, (Summary Text)
70. **Two patients with distal partial trisomy 1q**
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6th European Cytogenetics Conference, İstanbul, Turkey, 7 - 10 July 2007, vol.15, pp.70-71, (Summary Text)
71. **Bloom syndrome in a child with severe short stature and wilms tumor**
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72. **Unilateral peters' anomaly type I in an infant with 22q11.2 deletion syndrome**
Erdogan K. M., Utine G. E., ALANAY Y., Volkan-Salanci B., Boduroglu K., Aktas D., Alikasifoglu M., Tuncbilek E.
6th European Cytogenetics Conference, İstanbul, Turkey, 7 - 10 July 2007, vol.15, pp.100, (Summary Text)
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6th European Cytogenetics Conference, İstanbul, Turkey, 7 - 10 July 2007, vol.15, pp.70, (Summary Text)
74. **Clinical evaluation of Prader-Willi and Angelman syndrome patients with 15q11-13 deletion**
Kurtul K., Boduroglu K., ALANAY Y., Utine E., Salanci B. V., Aktas D., Alikasifoglu M., Tuncbilek E.
6th European Cytogenetics Conference, İstanbul, Turkey, 7 - 10 July 2007, vol.15, pp.96, (Summary Text)
75. **The detection of subtelomeric chromosomal rearrangements in 100 patients with idiopathic mental retardation: Hacettepe University Experience**
Celik T., Utine E., ALANAY Y., Aktas D., Boduroglu K., Alikasifoglu M., Tuncbilek E.
6th European Cytogenetics Conference, İstanbul, Turkey, 7 - 10 July 2007, vol.15, pp.70, (Summary Text)
76. **Chromosomal abnormalities presented with seizures**
Volkan-Salanci V., Utine G. E., Alanay Y., Aktas D., Boduroglu K., Alikasifoglu M., Tuncbilek E.
6th European Cytogenetics Conference, İstanbul, Turkey, 7 - 10 July 2007, vol.15, pp.84, (Summary Text)

Academic and Administrative Experience

2023 - Present	**Manager of Research and Application Center**	Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences
2020 - Continues	**Member of Commissions Affiliated to the Rectorate**	Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences
2020 - Continues	**Member of Commissions Affiliated to the Rectorate**	Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences
2018 - Continues	**Applied Research Center 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, Department Of Medical Sciences

Activities in Scientific Journals

EUROPEAN JOURNAL OF MEDICAL GENETICS, Committee Member, 2023 - Continues
American Journal Of Medical Genetics Part A, Committee Member, 2010 - Continues
Clinical Dysmorphology, Committee Member, 2010 - Continues

Memberships and Roles 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
Milli Pediatri Derneği, Member, 2002 - Continues, Turkey

Metrics

Publication: 294
Citation (WoS): 4594
Citation (Scopus): 5636
H-Index (WoS): 40
H-Index (Scopus): 45

Research Areas

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