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

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

Manager of Research and Application Center, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2023 - 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

Advising Theses

Alanay Y., Yenidoğan ve Çocuk Yoğun Bakım Ünitesinde (YYBÜ/ÇYBÜ) Hızla Kötüleşen Kritik Yenidoğan ve Süt Çocuklarında Hızlı Yeni Nesil Dizileme ile Genetik Tanı, Expertise In Medicine, A.Gör.(Student), 2024

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

I. **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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II. **A new line method; A direct test in spinal muscular atrophy screening for DBS**
Kubar A., Temel S. G., Beken S., Onder G., Hatirnaz Ö., Korkmaz A., Alanay Y., Ozbek U., Sag S. O., Ergoren M. C., et al.
Molecular Genetics and Genomic Medicine, vol.11, no.12, 2023 (SCI-Expanded)

III. **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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IV. **Treatment dilemma in a prepubertal patient with ACAN mutation but without advanced bone age**
Kozler S. H., GÜRPINAR G., Kilci F., KOÇYİĞİT E., ALANAY Y., JONES J. H., Jones F. M. C.
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V. **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.
HORMONE RESEARCH IN PAEDIATRICS, pp.163, 2023 (SCI-Expanded)

VI. **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.
HORMONE RESEARCH IN PAEDIATRICS, no.SUPPL 3, pp.149-151, 2023 (SCI-Expanded)

VII. **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)

VIII. **DNAJC21-related thrombocytopenia in a young adult female**
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AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS, vol.193, no.2, pp.193-197, 2023 (SCI-Expanded)

IX. **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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X. **Clinical Heterogeneity and Different Phenotypes in Patients with SETD2 Variants: 18 New Patients and Review of the Literature**
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XI. **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)

XII. **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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XIII. **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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XIV. **Undiagnosed diseases: Needs and opportunities in 20 countries participating in the Undiagnosed Diseases Network International**
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XV. **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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XVI. **Growth parameters in children with achondroplasia: A 7-year, prospective, multinational,**

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XVII. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**

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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.

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XX. **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, vol.66, pp.585-596, 2021 (SCI-Expanded)

XXI. **Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders**

Botto L. D., Meeths M., Campos-Xavier B., Bergamaschi R., Mazzanti L., Scarano E., Finocchi A., Cancrini C., Zirn B., Kuehnle I., et al.

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XXII. **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.

EUROPEAN JOURNAL OF HUMAN GENETICS, vol.29, no.1, pp.141-153, 2021 (SCI-Expanded)

XXIII. **Whole Exome Sequencing of consanguineous families of clinically diagnosed with Neurodevelopmental Disorders**

Turkgenc B., Yararbas K., Karakoyun H. K., Yesilyurt A., Gezdirici A., Tatli B., Akcakaya N. H., Hacifazlioglu N. E., Coskun O., Yesil G., et al.

EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, no.SUPPL 1, pp.886-887, 2020 (SCI-Expanded)

XXIV. **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.

MOLECULAR SYNDROMOLOGY, vol.11, no.4, pp.183-196, 2020 (SCI-Expanded)

XXV. **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., Nagabhyrava P., et al.

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XXVI. **Once-daily, subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial**

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XXVII. **Recurrent hydatidiform mole: When to stop ?**

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XXVIII. **A Randomized Controlled Trial of Vosoritide in Children with Achondroplasia**
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XXIX. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome**
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XXX. **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**
Simsek-Kiper P. O., TAŞKIRAN Z. E., KOŞUKCU C., ARSLAN U. E., Cormier-Daire V., Gonc N., ÖZÖN Z. A., Alikasifoglu A., Kandemir N., ÜTİNE G. E., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.7, pp.1157-1172, 2019 (SCI-Expanded)

XXXI. **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome**
van der Sluijs P. J., Jansen S., Vergano S. A., Adachi-Fukuda M., ALANAY Y., AlKindy A., Baban A., Bayat A., Beck-Woedl S., Berry K., et al.
GENETICS IN MEDICINE, vol.21, no.6, pp.1295-1307, 2019 (SCI-Expanded)

XXXII. **p.Ser348Cys mutation in FGFR3 gene leads to "Mild ACH /Severe HCH" phenotype.**
Bengur F., Ekmekci C., Karaarslan E., Gunoz H., Alanay Y.
European journal of medical genetics, pp.103659, 2019 (SCI-Expanded)

XXXIII. **A possible founder mutation in FZD6 gene in a Turkish family with autosomal recessive nail dysplasia.**
Saygi C., ALANAY Y., Sezerman U., Yenenler A., Ozoren N.
BMC medical genetics, vol.20, pp.15, 2019 (SCI-Expanded)

XXXIV. **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.
TURKISH JOURNAL OF PEDIATRICS, vol.60, no.5, pp.506-513, 2018 (SCI-Expanded)

XXXV. **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.
NATURE GENETICS, vol.50, no.8, pp.1093-1107, 2018 (SCI-Expanded)

XXXVI. **Fragile x-associated premature ovarian failure in a large Turkish cohort: Findings of Hacettepe Fragile X Registry**
ÜTİNE G. E., Simsek-Kiper P. O., Akgun-Dogan O., Urel-Demir G., ALANAY Y., Aktas D., BODUROĞLU O. K., Tuncbilek E., ALİKAŞİFOĞLU M.
EUROPEAN JOURNAL OF OBSTETRICS & GYNECOLOGY AND REPRODUCTIVE BIOLOGY, vol.221, pp.76-80, 2018 (SCI-Expanded)

XXXVII. **Parieto-occipital alopecia in early infancy mandates cranial imaging**
Tuc E., KARAARSLAN E., Celik I., ALANAY Y.
CLINICAL DYSMORPHOLOGY, vol.27, no.1, pp.15-17, 2018 (SCI-Expanded)

XXXVIII. **Hematopoietic Stem Cell Transplantation for Myelodysplastic Syndrome in a Child With Klinefelter Syndrome**
Serdaroglu E., KUŞKONMAZ B. B., ALANAY Y., AYTAÇ EYÜPOĞLU Ş. S., Cetin M., ÇETİNKAYA F. D.
JOURNAL OF PEDIATRIC HEMATOLOGY ONCOLOGY, vol.40, no.1, pp.81-82, 2018 (SCI-Expanded)

XXXIX. **Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases**
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XL. **A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl**

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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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XLI. **Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa**
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AMERICAN JOURNAL OF HUMAN GENETICS, vol.100, no.2, pp.216-227, 2017 (SCI-Expanded)

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XLIV. **Is 1p36 Deletion Associated with Anterior Body Wall Defects?**
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AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.170, no.7, pp.1889-1894, 2016 (SCI-Expanded)

XLV. **Late Sequel of Meningococcemia: Skeletal Dysplasia**
Karabulut G. S., Acar T., Yuksel A. B., ALANAY Y., Hatun S.
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XLVI. **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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XLVII. **Intrauterine Imaging Strategies for Bone Disease**
Alanay Y.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.13, 2016 (SCI-Expanded)

XLVIII. **Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate**
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L. **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.
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LI. **Hoyeraal-Hreidarsson Syndrome: An Extremely Rare Dyskeratosis Congenita Phenotype**
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LII. **Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome**
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LIII. **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
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LIV. **Experience of a Skeletal Dysplasia Registry in Turkey: A Five-Years Retrospective Analysis**
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LV. **Mutations in LONP1, a Mitochondrial Matrix Protease, Cause CODAS Syndrome**
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XIV. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome**
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XX. **The Skeletal Dysplasia Registry: Hacettepe Experience**
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XXI. **Possible effect of IGFR1 gene on macrocephaly in a patient with unbalanced 6;15 translocation with 6p25 deletion and 15q26 duplication**
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XXII. **Clinical, demographic and nosologic characterisation of the genetic disorders of the skeleton in Turkey: The skeletal dysplasia registry**
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XXIII. **Camptodactyly-arthropathy-coxa vara-pericarditis syndrome in a large family: A clinical condition with a diagnostic challange**
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XXIV. **Ischiospinal Dysostosis in a boy with a novel homozygous missense mutation in the BMPER gene**
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XXVI. **BİLEŞİK HETEROZİGOT GALNS MUTASYONU İLE TANI ALAN KLASİK OLMAYAN MUKOPOLİSAKKARİDOZ TİP IVA**
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XXX. **Normal enzim düzeyleri ve Japon Bayrağı göz dibi ile GM2 Gangliyosidoz tanısı alan iki kardeş: olgu sunumu**
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XXXIII. **Identification of Molecular Pathology of Peters' Anomaly Segregating in a Large Autosomal Dominant Family**
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XLII. **Parsiyel Trizomi 19p13 ile iliişkili bir immün yetmezlik**
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LVIII. **Two patients with distal partial trisomy 1q**
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LX. **The detection of subtelomeric chromosomal rearrangements in 100 patients with idiopathic mental retardation: Hacettepe University Experience**
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LXI. **Unilateral peters' anomaly type I in an infant with 22q11.2 deletion syndrome**
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Supported Projects

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

Activities in Scientific Journals

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

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
Milli Pediatri Derneği, Member, 2002 - Continues, Turkey

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

Metrics

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H-Index (Scopus): 42

Congress and Symposium Activities

Achondroplasia Masterclass, Invited Speaker, Frankfurt, Germany, 2024

Çocuk Nöroloji Aylık Toplantısı, Invited Speaker, İstanbul, Turkey, 2024
Büyüme Bozuklukları Sempozyumu, Invited Speaker, İstanbul, Turkey, 2024
10. Ulusal İSTAHED Aile Hekimliği Kongresi, Invited Speaker, Girne, Cyprus (Kktc), 2023
6. Ulusal Çocuk Genetik Kongresi, Invited Speaker, Aydın, Turkey, 2023
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12th UDNI Conference, Invited Speaker, Tbilisi, Georgia, 2023
19th Manchester Dysmorphology Conferences 2023, Attendee, Manchester, England, 2023
ESPE European Society of Pediatric Endocrinology, Invited Speaker, The Hague, Netherlands, 2023
Akondroplazide Yeni Bir Dönem, Invited Speaker, İstanbul, Turkey, 2023
UDNI Undiagnosed Disease Hackathon, Attendee, Stockholm, Sweden, 2023
XXVII. Ulusal Pediyatrik Endokrinoloji ve Diyabet Kongresi, Invited Speaker, Antalya, Turkey, 2023
7. Ulusal Çocuk Ortopedisi Kongresi, Invited Speaker, Ankara, Turkey, 2023