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Araştırma Alanları

Tıp, Dahili Tıp Bilimleri, Çocuk Sağlığı ve Hastalıkları, Pediatrik Genetik ve Teratoloji, Tıbbi Genetik, Sağlık Bilimleri

Akademik Unvanlar / Görevler

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

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

Prof. Dr., Acıbadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2013 - Devam Ediyor

Doç. Dr., Acıbadem 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

Uygulama ve Araştırma Merkezi Müdürü, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri

Bölümü, 2023 - Devam Ediyor

Rektörlüğe Bağlı Komisyon Üyesi, Acıbadem 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

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

Nonmendelian Inheritance, Lisans, 2021 - 2022

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Yönetilen Tezler

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

Jüri Üyelikleri

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- V. **An adolescent boy with PLS3 mutation causing severe thoracic kypho-scoliosis**
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- VI. **Treatment dilemma in a prepubertal patient with ACAN mutation but without advanced bone age**
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- VIII. **The clinical phenotype of Koolen-de Vries syndrome in Turkish patients and literature review**
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- IX. **<i>DNAJC21</i>-related thrombocytopenia in a young adult female**
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- X. **Real-world evidence in achondroplasia: considerations for a standardized data set**
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- XI. **Clinical Heterogeneity and Different Phenotypes in Patients with <i>SETD2</i> Variants: 18 New Patients and Review of the Literature**
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- XII. **Spondylo-meta-epiphyseal dysplasia (SMED), short limb-hand abnormal calcification type: Further expanding the mutational spectrum and dental findings of three new patients**
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- XIV. **Obstacles and expectations of rare disease patients and their families in Türkiye: ISTisNA project survey results**
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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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- XIX. **Defining mitochondrial protein functions through deep multiomic profiling**
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- XX. **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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- XXI. **Further defining the clinical and molecular spectrum of acromesomelic dysplasia type maroteaux: a Turkish tertiary center experience.**
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- XXII. **Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders**
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- XXIII. **Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort**
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- XXIV. **Whole Exome Sequencing of consanguineous families of clinically diagnosed with Neurodevelopmental Disorders**
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- XXV. **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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- XXVII. **Once-daily, subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial**
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- XXVIII. **Recurrent hydatidiform mole: When to stop ?**
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- XXIX. **A Randomized Controlled Trial of Vosoritide in Children with Achondroplasia**
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- XXX. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome**
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- XXXI. **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**
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- XXXII. **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome**
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- XXXV. **Epigenotype and phenotype correlations in patients with Beckwith-Wiedemann syndrome**
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- XXXVII. **Fragile x-associated premature ovarian failure in a large Turkish cohort: Findings of Hacettepe Fragile X Registry**
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- XLV. Expanding the clinical and mutational spectrum of the Ehlers-Danlos syndrome, dermatosparaxis type**
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III. Camptodactyly, Tall Stature, And Hearing Loss Syndrome with a Novel Homozygous FGFR3 Variant

Akgün Doğan Ö., Abalı S., Alanay Y.

19th Manchester Dysmorphology Conference 2023, Manchester, İngiltere, 16 - 18 Kasım 2023, ss.13-14

IV. First Report of Heterozygous Intragenic Deletion in NPR2 Presenting with Familial Short Stature

Akgün Doğan Ö., Alanay Y.

19th Manchester Dysmorphology Conference 2023, Manchester, İngiltere, 16 - 18 Kasım 2023, ss.10-12

V. Obstacles and Expectations of Rare Disease Patients and Their Families in Türkiye: ISTisNA Project Survey Results

Hatırnaz Ng Ö., Özdemir Ö., Alanay Y., Akgün Doğan Ö.

12th International Conference on Rare and Undiagnosed Diseases, Tbilisi, Gürcistan, 22 - 23 Ekim 2023, ss.1

VI. From patient to function: modeling CRIM1 in xenopus tropicalis

AKGÜN DOĞAN Ö., Viviano S., HATIRNAZ NG Ö., Agaoglu N. B., Ji W., Jeffries L., Ozbek U., Lakhani S., Khokha M.,

Deniz E., et al.

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VII. Biallelic copy number variations in both upstream & downstream enhancers of SHOX gene causes mesomelia and clubfoot without short stature

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- VIII. **Retrospective analysis of fetal magnetic resonance imaging (feMRI) examinations in the last 10 years at a tertiary center: experience of a single radiologist and a single perinatologist**
Ay B., Sari E., ALİS D. C., KARARSLAN E., Bildirici I., ALANAY Y.
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- IX. **Multiple epiphyseal dysplasia: A diagnostic challenge with genetic heterogeneity**
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- X. **Geçici neonatal myasteni gravis mi? Fetal asetilkolin reseptör inaktivasyon sendromu mu?8**
Aktas S., Kazancı E., Güner Yılmaz B., Korkmaz Toygar A., Alanay Y., Çomu S.
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- XI. **Severely Affected Newborn Female With De Novo Likely Pathogenic Variant In BCAP31**
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- XII. **Evaluation of the etiology of epilepsy and/or developmental delay in children with next generation sequencing:A single center experience**
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- XIII. **Molecular Diagnosis of TYR Negative Albinism Patients by Clinical Exome Sequencing**
Akyoney S., Sahin I., Unal B., Agaoglu N. B., Mudun A., Parlakgunes Z., Yilmaz E., ALANAY Y., ÖZBEK U., HATIRNAZ NG Ö.
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- XIV. **The third family with <i>TAF6</i>-related phenotype: Alazami-Yuan syndrome**
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- XV. **A Randomized Controlled Trial of Vosoritide in Children with Achondroplasia**
Savarirayan R., Tofts L., Irving M., Wilcox W., Bacino C., Hoover-Fong J., Font R. U., Harmatz P., Rutsch F., Bober M., et al.
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- XVI. **Does my expertise still make a difference? A single-clinician's experience of genomic sequencing in 120 pediatric patients**
Bengur F. B., KAR E., YARARBAŞ K., ALANAY Y.
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- XVII. **Screening TYR gene variations in Turkish oculocutaneuse albinism patients**
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- XVIII. **WAC geninde tanımlanan yeni bir varyant ve DeSanto-Shinawi Sendromu**
Tunç E., TEMEL Ş. G., ALANAY Y.
4.Çocuk Genetik Kongresi, Türkiye, 25 - 27 Eylül 2019
- XIX. **Meester-Loeys sendromu: Marfan benzeri sendromlara yeni bir üye**
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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. **Ischiospinal Dysostosis in a boy with a novel homozygous missense mutation in the BMPER gene**
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- XXIII. **Clinical, demographic and nosologic characterisation of the genetic disorders of the skeleton in Turkey: The skeletal dysplasia registry**
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- XXIV. **Camptodactyly-arthropathy-coxa vara-pericarditis syndrome in a large family: A clinical condition with a diagnostic challenge**
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- XXV. **Next Generation Sequencing (NGS) panel revealed new candidate genes and variants in 25 Hypertrophic Cardiomyopathy patients**
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- XXVI. **BİLEŞİK HETEROZİGOT GALNS MUTASYONU İLE TANI ALAN KLASİK OLMAYAN MUKOPOLİSAKKARİDOZ TİP IVA**
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- XXVII. **Baraitser-Winter Syndrome in a boy with heterozygous missense mutation in the ACTB gene**
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- XXIX. **Otozomal resesif infantil bilateral striatal nekroz,nup 62 geninde mutasyon: olgu sunumu**
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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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- XXXI. **Targeted custom gene panel sequencing for cardiac ion channelopathies: Efficiently detects candidate pathogenic mutations in Long QT syndrome**
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- XXXII. **A NEW MUTATION IN THE WISP3 GENE (c.935_936insTp. C314Lfs7) IN A PATIENT WITH**

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ABUR Ü., OĞUR M. G., AYKUT A., ALANAY Y.

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- XXXIII. **Identification of Molecular Pathology of Peters' Anomaly Segregating in a Large Autosomal Dominant Family**
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- XXXIV. **A report of mosaic Turner syndrome with a mild Kabuki like phenotype**
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- XXXVI. **Mitokondriyal hastalıklarda genetik testlerin rolü-iki olgu sunumu**
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- XXXVII. **Clinical and Molecular Analysis of 3M Syndrome Patients A Study From Turkey**
ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., ÜTİNE G. E., ALİKAŞİFOĞLU A., KANDEMİR N., Cormier Daire V., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
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- XXXVIII. **Detection of 15q (Prader Willi/Angelman) deletion in maternal cell-free fetal dna test, a case report**
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- XXXIX. **Meier Gorlin ear patella short stature syndrome A rare clinical entity**
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- XL. **OBSL1 Mutations Represent The Major Gene Defect In A Group Of 3M Syndrome Patients A Study From Turkey**
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- XLI. **Detection of 15q (Prader Willi/ Angelman syndrome) deletion in maternal cell-free fetal DNA test; A case report**
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- XLII. **Parsiyel Trizomi 19 9 ile ilişkili bir İmmün Yetmezlik**
BOZDOĞAN G., VARDAR G., ÇITAK A., ERTEKİN V., ALANAY Y.
2. Klinik İmmunoloji Kongresi, Türkiye, 31 Mart - 03 Nisan 2016
- XLIII. **Parsiyel Trizomi 19p13 ile ilişkili bir immün yetmezlik**
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- XLIV. **3M Sendromlu Bir Grup Hastada Klinik Ve Moleküler Bulguların Analizi**
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- XLV. **A custom Ampliseq arrhythmia panel comprising 68 cardiac channelopathy genes is a gold standard for the rapid and sensitive detection of genetic variation in long Qt syndrome**
Burcu T., TEMEL Ş. G., AYKAN H. H., SÜLÜ A., UYSAL F., BAŞPINAR O., RAMOĞLU M. G., KARAGÖZ T., AKALIN F., ALANAY Y., et al.

American Society of Human Genetics Congress, 6 - 10 Ekim 2015

- XLVI. **3D Image analysis of facial skeletal and soft tissue changes after monobloc distraction**
CANTER H. İ., ÖZEK M., ÇAKAN D., ALANAY Y., YILDIZ M. E., YILDIZ K.
The 16th Congress of International Society of Craniofacial Surgery (ISCFs), 14 - 18 Eylül 2015
- XLVII. **CLINICAL AND MOLECULAR STUDY OF A SERIES OF 31 PATIENTS WITH CHONDRODYSPLASIA WITH MULTIPLE DISLOCATIONS**
Ranza E., Huber C., Levin N., Baujat G., ALANAY Y., Al Gazali L., Bitoun P., Boute O., Coubes C., ELÇİOĞLU H. N.
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- XLVIII. **Spotlight on the pathogenesis of Kabuki syndrome**
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- L. **Spotlight on the pathogenesis of Kabuki syndrome**
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- LI. **Bir Olgu Nedeni ile PTH Direnci Pseudohipoparatiroidi tip1 A Albright Herediter Osteodistrofisi mi Akrodisostoz mu**
RAHİME GÜL Y. M., ALANAY Y., ÖZSU E., YÜKSEL A., ÇİZMECİOĞLU F. M., HATUN Ş.
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- LII. **Cabezas Sendromu**
ÖZSU E., RAHİME GÜL Y. M., ÇİZMECİOĞLU F. M., ALANAY Y., HATUN Ş.
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- LIII. **Metabolik Sendrom Carnevale Sendrom Birlikteliği Vaka Sunumu**
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- LIV. **BİR SPONDİLO ENDONDRO DİSPLAZİ VAKASI**
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- LV. **Two siblings with severe deforming osteogenesis imperfecta with mutations in the newly identified recessive OI gene FKBP 65**
RAHİME GÜL Y. M., ALANAY Y., AKARSU A. N., ÖZSU E., ÇİZMECİOĞLU F. M., HATUN Ş.
ESPE 49. Annual Meeting, PRAGUE, 22 - 25 Eylül 2009
- LVI. **Rapid prenatal diagnosis of common aneuploidies by QF-PCR, four years experience of Hacettepe University**
Aktas D., Kutukcu B., Utine G., ALANAY Y., Deren O., BODUROĞLU O. K., Beksac S., Alikasifoglu M.
7th European Cytogenetics Conference, Stockholm, İsveç, 4 - 07 Temmuz 2009, cilt.17, ss.209
- LVII. **Two patients with distal partial trisomy 1q**
Aktas D., Utine E., ALANAY Y., Gucer S., Tuncbilek E., Mrasek K., Liehr T.
6th European Cytogenetics Conference, İstanbul, Türkiye, 7 - 10 Temmuz 2007, cilt.15, ss.70-71
- LVIII. **Bloom syndrome in a child with severe short stature and wilms tumor**
Boduroglu K., ALANAY Y., Alikasifoglu M., Aktas D., Utine G. E., Tuncbilek E.
6th European Cytogenetics Conference, İstanbul, Türkiye, 7 - 10 Temmuz 2007, cilt.15, ss.142-143
- LIX. **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, Türkiye, 7 - 10 Temmuz 2007, cilt.15, ss.84
- LX. **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, Türkiye, 7 - 10 Temmuz 2007, cilt.15, ss.96

LXI. Partial monosomy of distal 6q

Utine E., ALANAY Y., Aktas D., Boduroglu K., Alikasifoglu A., Tuncbilek E.

6th European Cytogenetics Conference, İstanbul, Türkiye, 7 - 10 Temmuz 2007, cilt.15, ss.70

LXII. 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, Türkiye, 7 - 10 Temmuz 2007, cilt.15, ss.100

LXIII. 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.

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Desteklenen Projeler

Akgün Doğan Ö., Sütçübaşı B., Alanay Y., Diğer Uluslararası Fon Programları, Arid1b ile İlişkili Bozuklukta Gelişimsel İlerleme Alanları - Çok Yönlü Çok Merkezli Prospektif Doğal Seyir Çalışması, 2024 - 2027

Bilimsel Dergilerdeki Faaliyetler

EUROPEAN JOURNAL OF MEDICAL GENETICS, Editörler Kurulu Üyesi, 2023 - Devam Ediyor

American Journal Of Medical Genetics Part A, Editörler Kurulu Üyesi, 2010 - Devam Ediyor

Clinical Dysmorphology, Editörler Kurulu Üyesi, 2010 - Devam Ediyor

Bilimsel Kuruluşlardaki Üyelikler / Görevler

Çocuk Genetik Hastalıkları Derneği, Üye, 2011 - Devam Ediyor , Türkiye

Türk Pediatri Kurumu, Üye, 2009 - Devam Ediyor , Türkiye

International Skeletal Dysplasia Society, Üye, 2005 - Devam Ediyor , İsviçre

European Society of Human Genetics, Üye, 2003 - Devam Ediyor , Avusturya

Milli Pediatri Derneği, Üye, 2002 - Devam Ediyor , Türkiye

Etkinlik Organizasyonlarındaki Görevler

Alanay Y., European Society of Human Genetics Meeting, Bilimsel Kongre / Sempozyum Organizasyonu, Berlin, Almanya, Haziran 2020

Alanay Y., ECTS 2020 European Calcified Tissue Society Meeting, Bilimsel Kongre / Sempozyum Organizasyonu, Marseille, Fransa, Mayıs 2020

Metrikler

Yayın: 270

Atıf (WoS): 4567

Atıf (Scopus): 5063

H-İndeks (WoS): 40

H-İndeks (Scopus): 42

Kongre ve Sempozyum Katılımı Faaliyetleri

- Achondroplasia Masterclass, Davetli Konuşmacı, Frankfurt, Almanya, 2024
Çocuk Nöroloji Aylık Toplantısı, Davetli Konuşmacı, İstanbul, Türkiye, 2024
Büyüme Bozuklukları Sempozyumu, Davetli Konuşmacı, İstanbul, Türkiye, 2024
10. Ulusal İSTAHED Aile Hekimliği Kongresi, Davetli Konuşmacı, Girne, Kıbrıs (Kkct), 2023
6. Ulusal Çocuk Genetik Kongresi, Davetli Konuşmacı, Aydın, Türkiye, 2023
6. Ulusal Çocuk Genetik Kongresi, Davetli Konuşmacı, Aydın, Türkiye, 2023
12th UDNI Conference, Davetli Konuşmacı, Tbilisi, Gürcistan, 2023
19th Manchester Dysmorphology Conferences 2023, Katılımcı, Manchester, İngiltere, 2023
ESPE European Society of Pediatric Endocrinology, Davetli Konuşmacı, The Hague, Hollanda, 2023
Akondroplazide Yeni Bir Dönem, Davetli Konuşmacı, İstanbul, Türkiye, 2023
UDNI Undiagnosed Disease Hackathon, Katılımcı, Stockholm, İsveç, 2023
XXVII. Ulusal Pediyatrik Endokrinoloji ve Diyabet Kongresi, Davetli Konuşmacı, Antalya, Türkiye, 2023
7. Ulusal Çocuk Ortopedisi Kongresi, Davetli Konuşmacı, Ankara, Türkiye, 2023