Prof. Dr. YASEMİN ALANAY

Kişisel Bilgiler

İş Telefonu: +90 212 304 4528
Diğer E-posta: yaseminalanay@acualanaylab.org
Web: https://avesis.acibadem.edu.tr/527
Posta Adresi: Büyükdere Cad. No:40 Sarıyer İstanbul

Uluslararası Araştırmacı ID'leri
ScholarID: EBP2898AAAAJ
ORCID: 0000-0003-0683-9731
Publons / Web Of Science ResearcherID: A-1516-2016
ScopusID: 56247793800
Yoksis Araştırmacı ID: 125850

Eğitim Bilgileri

Tıpta Yandal Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Türkiye 2002 - 2011
Doktora, Hacettepe Üniversitesi, Sağlık Bilimleri Enstitüsü, Pediyatrik Temel Bilimler A.B.D., Türkiye 2004 - 2009
Post Doktora, University of California, Los Angeles, Cedars-Sinai Medical Center, Amerika Birleşik Devletleri 2005 - 2006
Tıpta Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Türkiye 1997 - 2002
Lisans, Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Türkiye 1990 - 1996

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
Pediatric Genetics, Lisans, 2021 - 2022
Treatment of Genetic Diseases and Future of Clinical Genomics , Lisans, 2021 - 2022
Nonmendelian Inheritance, Lisans, 2021 - 2022
From Genotype to Phenotype, Lisans, 2021 - 2022
Pediatric History Taking, Lisans, 2021 - 2022
Mendelian Inheritance Patterns, Lisans, 2021 - 2022

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

Doçentlik Sınavı, Doçentlik Sınavı, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Mart, 2022
Doçentlik Sınavı, Doçentlik Sınavı, Acıbadem Mehmet Ali Aydınlar Üniversitesi, Eylül, 2021

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

I. **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.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.32, sa.10, ss.1250-1256, 2024 (SCI-Expanded)
II. **Rapid genome sequencing for critically ill infants: an inaugural pilot study from Turkey.**
Guner Yilmaz B., Akgun-Dogan O., Ozdemir Ö., Yuksel B., Hatirnaz Ng Ö., Bilguvar K., Ay B., Ozkose G. S., Aydin E., Yigit A., et al.
Frontiers in pediatrics, cilt.12, ss.1412880, 2024 (SCI-Expanded)
III. **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.
FRONTIERS IN GENETICS, cilt.15, 2024 (SCI-Expanded)
IV. **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, cilt.11, sa.12, 2023 (SCI-Expanded)

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, ss.163, 2023 (SCI-Expanded)

VI. **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.
HORMONE RESEARCH IN PAEDIATRICS, ss.174-175, 2023 (SCI-Expanded)

VII. **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, sa.SUPPL 3, ss.149-151, 2023 (SCI-Expanded)

VIII. **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, cilt.191, sa.7, ss.1814-1825, 2023 (SCI-Expanded)

IX. **DNAJC21-related thrombocytopenia in a young adult female**
ASLAN D., AKGÜN DOĞAN Ö., Ay B., ÇAMURDAN M. O., Mancilar H., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS, cilt.193, sa.2, ss.193-197, 2023 (SCI-Expanded)

X. **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.
ORPHANET JOURNAL OF RARE DISEASES, sa.1, 2023 (SCI-Expanded)

XI. **Clinical Heterogeneity and Different Phenotypes in Patients with SETD2 Variants: 18 New Patients and Review of the Literature**
Parra A., Rabin R., Pappas J., Pascual P., Cazalla M., Arias P., Gallego-Zazo N., Santana A., Arroyo I., Artigas M., et al.
GENES, cilt.14, sa.6, 2023 (SCI-Expanded)

XII. **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, cilt.66, sa.4, 2023 (SCI-Expanded)

XIII. **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.
HORMONE RESEARCH IN PAEDIATRICS, sa.SUPPL 2, ss.28-29, 2023 (SCI-Expanded)

XIV. **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.
FRONTIERS IN PUBLIC HEALTH, cilt.10, 2023 (SCI-Expanded)

XV. **Unmet needs in countries participating in the undiagnosed diseases network international: an international survey considering national health care and economic indicators**
Sciascia S., Roccatello D., Salvatore M., Carta C., Cellai L. L., Ferrari G., Lumaka A., Groft S., ALANAY Y., Azam M., et al.
FRONTIERS IN PUBLIC HEALTH, cilt.11, 2023 (SCI-Expanded)

XVI. **Undiagnosed diseases: Needs and opportunities in 20 countries participating in the Undiagnosed Diseases Network International**
Taruscio D., Salvatore M., Lumaka A., Carta C., Cellai L. L., Ferrari G., Sciascia S., Groft S., ALANAY Y., Azam M., et al.
FRONTIERS IN PUBLIC HEALTH, cilt.11, 2023 (SCI-Expanded)

XVII. **Growth parameters in children with achondroplasia: A 7-year, prospective, multinational, observational study**
Savarirayan R., Irving M., Harmatz P., Delgado B., Wilcox W. R., Philips J., Owen N., Bacino C. A., Tofts L., Charrow J., et al.
GENETICS IN MEDICINE, cilt.24, sa.12, ss.2444-2452, 2022 (SCI-Expanded)

XVIII. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**
GÜLERAY LAFCI N., KOŞUKCU C., Oguz S., Demir G. U., Taskiran E. Z., Kiper P. O. S., ÜTİNE G. E., ALANAY Y., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
CLEFT PALATE-CRANIOFACIAL JOURNAL, cilt.59, sa.9, ss.1114-1124, 2022 (SCI-Expanded)

XIX. **Defining mitochondrial protein functions through deep multiomic profiling**
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.
NATURE, cilt.606, ss.382-388, 2022 (SCI-Expanded)

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**
Bozkurt T., ALANAY Y., ISIK U., SEZERMAN O. U.
BMC MEDICAL GENOMICS, cilt.14, sa.1, 2021 (SCI-Expanded)

XXI. **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, cilt.66, ss.585-596, 2021 (SCI-Expanded)

XXII. **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.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.185, sa.2, ss.517-527, 2021 (SCI-Expanded)

XXIII. **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, cilt.29, sa.1, ss.141-153, 2021 (SCI-Expanded)

XXIV. **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, cilt.28, sa.SUPPL 1, ss.886-887, 2020 (SCI-Expanded)

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.
MOLECULAR SYNDROMOLOGY, cilt.11, sa.4, ss.183-196, 2020 (SCI-Expanded)

XXVI. **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.
Brain : a journal of neurology, cilt.143, ss.2929-2944, 2020 (SCI-Expanded)

XXVII. **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, cilt.396, sa.10252, ss.684-692, 2020 (SCI-Expanded)

XXVIII. **Recurrent hydatidiform mole: When to stop ?**
ÖZBAŞLI E., TAKMAZ Ö., Gurkan H., ALANAY Y., GÜNGÖR M., DEDE F. S.
Clinical and Experimental Obstetrics and Gynecology, cilt.47, sa.3, ss.424-426, 2020 (SCI-Expanded)

XXIX. **A Randomized Controlled Trial of Vosoritide in Children with Achondroplasia**
Pogreen L., Savarirayan R., Tofts L., Rehab K., Irving M., Wilcox W., Bacino C., Hoover-Fong J., Ullot Font R., Harmatz P., et al.
HORMONE RESEARCH IN PAEDIATRICS, cilt.93, ss.169-170, 2020 (SCI-Expanded)

XXX. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome**
Tuc E., Bengur F. B., Aykut A., ŞAHİN Ö., ALANAY Y.
CLINICAL GENETICS, cilt.97, sa.5, ss.795-796, 2020 (SCI-Expanded)

XXXI. **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, cilt.179, sa.7, ss.1157-1172, 2019 (SCI-Expanded)

XXXII. **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, cilt.21, sa.6, ss.1295-1307, 2019 (SCI-Expanded)

XXXIII. **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, ss.103659, 2019 (SCI-Expanded)

XXXIV. **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, cilt.20, ss.15, 2019 (SCI-Expanded)

XXXV. **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, cilt.60, sa.5, ss.506-513, 2018 (SCI-Expanded)

XXXVI. **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, cilt.50, sa.8, ss.1093-1107, 2018 (SCI-Expanded)

XXXVII. **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, cilt.221, ss.76-80, 2018 (SCI-Expanded)

XXXVIII. **Parieto-occipital alopecia in early infancy mandates cranial imaging**
Tuc E., KARAARSLAN E., Celik I., ALANAY Y.
CLINICAL DYSMORPHOLOGY, cilt.27, sa.1, ss.15-17, 2018 (SCI-Expanded)

XXXIX. **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, cilt.40, sa.1, ss.81-82, 2018 (SCI-Expanded)

XL. **Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases**
Ranza E., Huber C., Levin N., Baujat G., Bole-Feysot C., Nitschke P., Masson C., ALANAY Y., Al-Gazali L., Bitoun P., et al.
CLINICAL GENETICS, cilt.91, sa.6, ss.868-880, 2017 (SCI-Expanded)

XLI. **A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl Hydroxylation of Type I Procollagen**
Duran I., Martin J. H., Weis M. A., Krejci P., Konik P., Li B., ALANAY Y., Lietman C., Lee B., Eyre D., et al.
JOURNAL OF BONE AND MINERAL RESEARCH, cilt.32, sa.6, ss.1309-1319, 2017 (SCI-Expanded)

XLII. **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, cilt.100, sa.2, ss.216-227, 2017 (SCI-Expanded)

XLIII. **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, cilt.170, sa.12, ss.3231-3236, 2016 (SCI-Expanded)

XLIV. **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, cilt.18, sa.9, ss.882-891, 2016 (SCI-Expanded)

XLV. **Is 1p36 Deletion Associated with Anterior Body Wall Defects?**
Collu M., Yuksel Ş., Sirin B. K., Abbasoglu L., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.170, sa.7, ss.1889-1894, 2016 (SCI-Expanded)

XLVI. **Late Sequel of Meningococcemia: Skeletal Dysplasia**
Karabulut G. S., Acar T., Yuksel A. B., ALANAY Y., Hatun S.
JOURNAL OF PEDIATRICS, cilt.173, ss.264-266, 2016 (SCI-Expanded)

XLVII. **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.
LUPUS, cilt.25, sa.7, ss.760-765, 2016 (SCI-Expanded)

XLVIII. **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, cilt.58, sa.5, ss.541-544, 2016 (SCI-Expanded)

XLIX. **Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate**
Breckpot J., Anderlid B., ALANAY Y., Blyth M., Brahimi A., Duban-Bedu B., Goze O., Firth H., YAKICIER M. C., Hens G., et al.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.24, sa.1, ss.51-58, 2016 (SCI-Expanded)

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.
HORMONE RESEARCH IN PAEDIATRICS, cilt.86, ss.167, 2016 (SCI-Expanded)

LI. **Intrauterine Imaging Strategies for Bone Disease**
Alanay Y.
HORMONE RESEARCH IN PAEDIATRICS, cilt.86, ss.13, 2016 (SCI-Expanded)

LII. **Hoyeraal-Hreidarsson Syndrome: An Extremely Rare Dyskeratosis Congenita Phenotype**
Bakar O., Isik U., Canpolat C., ALANAY Y.
PEDIATRIC DERMATOLOGY, cilt.32, sa.6, 2015 (SCI-Expanded)

LIII. **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
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.
JOURNAL OF CLINICAL INVESTIGATION, cilt.125, sa.9, ss.3585-3599, 2015 (SCI-Expanded)

LIV. **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.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.167, sa.9, ss.2065-2074, 2015 (SCI-Expanded)

LV. **Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome**
Atik T., Koparir A., Bademci G., Foster J., Altunoglu U., Mutlu G. Y., Bowdin S., ELÇİOĞLU H. N., Tayfun G. A., Atik S. S., et al.
ORPHANET JOURNAL OF RARE DISEASES, cilt.10, 2015 (SCI-Expanded)

LVI. **Mutations in LONP1, a Mitochondrial Matrix Protease, Cause CODAS Syndrome**
Dikoglu E., Alfaiz A., Gorna M., Bertola D., Chae J. H., Cho T., Derbent M., ALANAY Y., GÜRAN T., Kim O., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.167, sa.7, ss.1501-1509, 2015 (SCI-Expanded)

LVII. **Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffin-Siris and Nicolaides-Baraitser syndromes**
Bramswig N. C., Luedecke H., ALANAY Y., Albrecht B., Barthelmie A., BODUROĞLU O. K., Braunholz D., Caliebe A., Chrzanowska K. H., Czeschik J. C., et al.
HUMAN GENETICS, cilt.134, sa.6, ss.553-568, 2015 (SCI-Expanded)

LVIII. **Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3**
Chong J. X., Burrage L. C., Beck A. E., Marvin C. T., McMillin M. J., Shively K. M., Harrell T. M., Buckingham K. J., Bacino C. A., Jain M., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.96, sa.5, ss.841-849, 2015 (SCI-Expanded)

LIX. **A Study of the Clinical and Radiological Features in a Cohort of 93 Patients with a COL2A1 Mutation Causing Spondyloepiphyseal Dysplasia Congenita or a Related Phenotype**
Terhal P. A., Nievelstein R. J. A. J., Verver E. J. J., Topsakal V., van Dommelen P., Hoornaert K., Le Merrer M., Zankl A., Simon M. E. H., Smithson S. F., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.167, sa.3, ss.461-475, 2015 (SCI-Expanded)

LX. **Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy**
Simons C., Rash L. D., Crawford J., Ma L., Cristofori-Armstrong B., Miller D., Ru K., Baillie G. J., ALANAY Y., Jacquinet A., et al.
NATURE GENETICS, cilt.47, sa.1, ss.73-78, 2015 (SCI-Expanded)

LXI. **A VARIANT CASE OF 6p24 DELETION SYNDROME (OMIM #612582)**
Ergin R. N., Cigerciogullari E., Alanay Y., Yayla M.
GENETIC COUNSELING, cilt.26, sa.2, ss.237-241, 2015 (SCI-Expanded)

LXII. **Prepubertal Unilateral Gynecomastia: Report of 2 Cases**
Demirbilek H., Bacak G., Baran R. T., Avci Y., Baran A., Keles A., Ozbek M. N., ALANAY Y., Hussain K.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, cilt.6, sa.4, ss.250-253, 2014 (SCI-Expanded)

LXIII. **Partial monosomy 3q26.33-3q27.3 presenting with intellectual disability, facial dysmorphism, and diaphragm eventration: a case report**
Sahin Y., Kiper P. O., ALANAY Y., Liehr T., ÜTİNE G. E., BODUROĞLU O. K.
CLINICAL DYSMORPHOLOGY, cilt.23, sa.4, ss.147-151, 2014 (SCI-Expanded)

LXIV. **A case of fucosidosis type II: diagnosed with dysmorphological and radiological findings**
KILIÇ E., KILIÇ M., ÜTİNE G. E., Sivri S., COŞKUN T., ALANAY Y.
TURKISH JOURNAL OF PEDIATRICS, cilt.56, sa.4, ss.430-433, 2014 (SCI-Expanded)

LXV. **Etiological yield of SNP microarrays in idiopathic intellectual disability**
ÜTİNE G. E., Haliloglu G., Volkan-Salanci B., ÇETİNKAYA A., Kiper P. O., ALANAY Y., Aktas D., Anlar B., Topcu M., BODUROĞLU O. K., et al.
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, cilt.18, sa.3, ss.327-337, 2014 (SCI-Expanded)

LXVI. **Cathepsin K analysis in a pycnodysostosis cohort: demographic, genotypic and phenotypic features**
ARMAN A., BEREKET A., Coker A., Kiper P. O. S., GÜRAN T., Ozkan B., Atay Z., Akcay T., HALİLOĞLU B., BODUROĞLU O. K., et al.
ORPHANET JOURNAL OF RARE DISEASES, cilt.9, 2014 (SCI-Expanded)

LXVII. **Methylene tetrahydrofolate reductase polymorphisms and homocysteine level in heart defects**
ŞAHİNER Ü. M., ALANAY Y., ALEHAN D., TUNÇBİLEK E., ALİKAŞİFOĞLU M.
PEDIATRICS INTERNATIONAL, cilt.56, sa.2, ss.167-172, 2014 (SCI-Expanded)

LXVIII. **XYLT1 Mutations in Desbuquois Dysplasia Type 2**

BUI C., HUBER C., Tuysuz B., ALANAY Y., Bole-Feysot C., LEROY J. G., MORTIER G., Nitschke P., MUNNICH A., CORMIER-DAIRE V.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.94, sa.3, ss.405-414, 2014 (SCI-Expanded)

LXIX. **Report of a Patient With Temple-Baraitser Syndrome**
YEŞİL G., Guler S., Yuksel A., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.164, sa.3, ss.848-851, 2014 (SCI-Expanded)

LXX. **Celiac disease in Williams-Beuren syndrome**
Simsek-Kiper P. O., Sahin Y., Arslan U., ALANAY Y., BODUROĞLU O. K., ORHAN D., ÖZEN H., ÜTİNE G. E.
TURKISH JOURNAL OF PEDIATRICS, cilt.56, sa.2, ss.154-159, 2014 (SCI-Expanded)

LXXI. **TMCO1 Deficiency Causes Autosomal Recessive Cerebrofaciothoracic Dysplasia**
ALANAY Y., Erguner B., Utine E., Hacariz O., ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., Percin F., UZ YILDIRIM E., Sagiroglu M. S., Yuksel B., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.164, sa.2, ss.291-304, 2014 (SCI-Expanded)

LXXII. **Striking Hematological Abnormalities in Patients With Microcephalic Osteodysplastic Primordial Dwarfism Type II (MOPD II): A Potential Role of Pericentrin in Hematopoiesis**
ÜNAL S., ALANAY Y., Cetin M., BODUROĞLU O. K., Utine E., Cormier-Daire V., Huber C., ÖZSÜREKCİ Y., KILIÇ E., Kiper O. P. S., et al.
PEDIATRIC BLOOD & CANCER, cilt.61, sa.2, ss.302-305, 2014 (SCI-Expanded)

LXXIII. **A de novo 11q23 deletion in a patient presenting with severe ophthalmologic findings, psychomotor retardation and facial dysmorphism**
Simsek-Kiper P. O., Bayram Y., ÜTİNE G. E., ALANAY Y., BODUROĞLU O. K.
TURKISH JOURNAL OF PEDIATRICS, cilt.56, sa.1, ss.80-84, 2014 (SCI-Expanded)

LXXIV. **Neurochemical Evaluation of Brain Function With H-1 Magnetic Resonance Spectroscopy in Patients With Fragile X Syndrome**
ÜTİNE G. E., Akpinar B., ARSLAN U. E., Kiper P. O. S., Volkan-Salanci B., ALANAY Y., Aktas D., Haliloglu G., Oguz K. K., BODUROĞLU O. K., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.164, sa.1, ss.99-105, 2014 (SCI-Expanded)

LXXV. **PARENTAL FACTORS IN PRENATAL DECISION MAKING AND THE IMPACT OF PRENATAL GENETIC COUNSELING: A STUDY ON TURKISH FAMILIES**
Simsek-Kiper P. O., ÜTİNE G. E., Volkan-Salanci B., ALANAY Y., Aktas D., ALİKAŞİFOĞLU M., BODUROĞLU O. K., Tuncbilek E.
GENETIC COUNSELING, cilt.25, sa.1, ss.53-62, 2014 (SCI-Expanded)

LXXVI. **A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling**
Wieczorek D., Boegershausen N., Beleggia F., Steiner-Haldenstaett S., Pohl E., Li Y., Milz E., Martin M., Thiele H., Altmueller J., et al.
HUMAN MOLECULAR GENETICS, cilt.22, sa.25, ss.5121-5135, 2013 (SCI-Expanded)

LXXVII. **Three Patients Resembling Teebi-Shaltout Syndrome**
Aldemir O., Ozen S., Erdem S., Kiraz A., Akarsu N., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.161, sa.10, ss.2570-2575, 2013 (SCI-Expanded)

LXXVIII. **Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused by PAPSS2 Mutations**
Iida A., Simsek-Kiper P. O., Mizumoto S., Hoshino T., ELÇİOĞLU H. N., Horemuzova E., Geiberger S., YEŞİL SAYIN G., Kayserili H., ÜTİNE G. E., et al.
HUMAN MUTATION, cilt.34, sa.10, ss.1381-1386, 2013 (SCI-Expanded)

LXXIX. **Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome**
Czeschik J. C., Voigt C., Alanay Y., Albrecht B., Avci S., FitzPatrick D., Goudie D. R., Hehr U., Hoogeboom A. J., Kayserili H., et al.
HUMAN GENETICS, cilt.132, sa.8, ss.885-898, 2013 (SCI-Expanded)

LXXX. **A Homozygous Deletion in GRID2 Causes a Human Phenotype With Cerebellar Ataxia and Atrophy**
Ütine G. E., Haliloglu G., Volkan Salancı B., Çetinkaya A., Şimşek Kiper P. Ö., Alanay Y., Aktas D., Boduroğlu O. K.,

Alikaşifoğlu M.
JOURNAL OF CHILD NEUROLOGY, cilt.28, sa.7, ss.926-932, 2013 (SCI-Expanded)

LXXXI. **Microdeletions at 1q21.1 and 2q24.2 in a Patient with Developmental Delay and Dysmorphic Features**
ÜTİNE G. E., ŞİMŞEK KİPER P. Ö., ALANAY Y., BODUROĞLU O. K.
CHROMOSOME RESEARCH, cilt.21, 2013 (SCI-Expanded)

LXXXII. **STRIKING HEMATOLOGICAL ABNORMALITIES IN PATIENTS WITH MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II MAY INDICATE A POTENTIAL ROLE OF PERICENTRIN GENE IN HEMATOPOIESIS**
ÜNAL S., Cetin M., ALANAY Y., BODUROĞLU O. K., Utine E., Kilic E., Ozsurekci Y., Gumruk F.
HAEMATOLOGICA, cilt.98, ss.578, 2013 (SCI-Expanded)

LXXXIII. **Functional analysis of a duplication (p.E63_D69dup) in the switch II region of HRAS: new aspects of the molecular pathogenesis underlying Costello syndrome**
Lorenz S., Lissewski C., Simsek-Kiper P. O., ALANAY Y., Boduroglu K., Zenker M., Rosenberger G.
HUMAN MOLECULAR GENETICS, cilt.22, sa.8, ss.1643-1653, 2013 (SCI-Expanded)

LXXXIV. **Clinical and molecular analysis of RASopathies in a group of Turkish patients**
Simsek-Kiper P. O., ALANAY Y., GÜLHAN B., Lissewski C., Turkyilmaz D., ALEHAN D., ÇETİN M., ÜTİNE G. E., Zenker M., BODUROĞLU O. K.
CLINICAL GENETICS, cilt.83, sa.2, ss.181-186, 2013 (SCI-Expanded)

LXXXV. **Laboratory Genetic Testing in Clinical Practice**
ÇOĞULU M. Ö., ALANAY Y., Toruner G. A.
BIOMED RESEARCH INTERNATIONAL, cilt.2013, 2013 (SCI-Expanded)

LXXXVI. **A severity affected case with Schimke immuno-osseous dysplasia**
Candan C., Yilmaz Y., ALANAY Y., Turhan P., Candan F., Erguven M.
TURK PEDIATRI ARSIVI-TURKISH ARCHIVES OF PEDIATRICS, cilt.47, sa.4, ss.319-321, 2012 (SCI-Expanded)

LXXXVII. **Further characterization of ATP6V0A2-related autosomal recessive cutis laxa**
Fischer B., Dimopoulou A., Egerer J., Gardeitchik T., Kidd A., Jost D., Kayserili H., ALANAY Y., Tantcheva-Poor I., Mangold E., et al.
HUMAN GENETICS, cilt.131, sa.11, ss.1761-1773, 2012 (SCI-Expanded)

LXXXVIII. **MRI findings of intracranial malformations in a case with Fraser syndrome**
Yesilkaya Y., HIZAL M., Oguz K. K., ALANAY Y.
CLINICAL DYSMORPHOLOGY, cilt.21, sa.4, ss.234-236, 2012 (SCI-Expanded)

LXXXIX. **Medical management of moyamoya disease and recurrent stroke in an infant with Majewski osteodysplastic primordial dwarfism type II (MOPD II)**
KILIÇ E., Utine E., ÜNAL S., Haliloglu G., Oguz K. K., Cetin M., BODUROĞLU O. K., ALANAY Y.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.171, sa.10, ss.1567-1571, 2012 (SCI-Expanded)

XC. **IMPAD1 Mutations in Two Catel-Manzke Like Patients**
Nizon M., ALANAY Y., Tuysuz B., Kiper P. O. S., Genevieve D., Sillence D., Huber C., Munnich A., Cormier-Daire V.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.158A, sa.9, ss.2183-2187, 2012 (SCI-Expanded)

XCI. **Severe neurologic manifestations from cervical spine instability in spondylo-megaepiphyseal-metaphyseal dysplasia**
SIMON M., Campos-Xavier A. B., Mittaz-Crettol L., VALADARES E. R., CARVALHO D., SPECK-MARTINS C. E., NAMPOOTHIRI S., ALANAY Y., MIHÇI E., van Bever Y., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS, cilt.160C, sa.3, ss.230-237, 2012 (SCI-Expanded)

XCII. **Further Delineation of CANT1 Phenotypic Spectrum and Demonstration of Its Role in Proteoglycan Synthesis**
Nizon M., Huber C., De Leonardis F., Merrina R., Forlino A., Fradin M., Tuysuz B., Abu-Libdeh B. Y., ALANAY Y., Albrecht B., et al.
HUMAN MUTATION, cilt.33, sa.8, ss.1261-1266, 2012 (SCI-Expanded)

XCIII. **The diagnostic challenge of progressive pseudorheumatoid dysplasia (PPRD): A review of clinical**

features, radiographic features, and WISP3 mutations in 63 affected individuals
Segarra N. G., Mittaz L., Campos-Xavier A. B., Bartels C. F., Tuysuz B., ALANAY Y., Cimaz R., Cormier-Daire V., Di Rocco M., Duba H., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS, cilt.160C, sa.3, ss.217-229, 2012 (SCI-Expanded)

XCIV. **Wiedemann-Rautenstrauch syndrome: Report of a variant case**
Kiraz A., Ozen S., TUBAŞ F., Usta Y., Aldemir O., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.158A, sa.6, ss.1434-1436, 2012 (SCI-Expanded)

XCV. **Disease-associated mutations in the actin-binding domain of filamin B cause cytoplasmic focal accumulations correlating with disease severity**
Daniel P. B., Morgan T., ALANAY Y., Bijlsma E., Cho T., Cole T., Collins F., David A., Devriendt K., Faivre L., et al.
HUMAN MUTATION, cilt.33, sa.4, ss.665-673, 2012 (SCI-Expanded)

XCVI. **Arterial tortuosity and aneurysm in a case of Loeys-Dietz syndrome type IB with a mutation p.R537P in the TGFBR2 gene**
KILIÇ E., ALANAY Y., Utine E., ÖZGEN MOCAN B., Robinson P. N., BODUROĞLU O. K.
TURKISH JOURNAL OF PEDIATRICS, cilt.54, sa.2, ss.198-202, 2012 (SCI-Expanded)

XCVII. **Haploinsufficiency of a Spliceosomal GTPase Encoded by EFTUD2 Causes Mandibulofacial Dysostosis with Microcephaly**
Lines M. A., Huang L., Schwartzentruber J., Douglas S. L., Lynch D. C., Beaulieu C., Guion-Almeida M. L., Zechi-Ceide R. M., Gener B., Gillessen-Kaesbach G., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.90, sa.2, ss.369-377, 2012 (SCI-Expanded)

XCVIII. **Hypophosphatasia Presenting with Pyridoxine-Responsive Seizures, Hypercalcemia, and Pseudotumor Cerebri: Case Report**
DEMİRBİLEK H., ALANAY Y., Alikasifoglu A., Topcu M., Mornet E., Gonc N., Ozon A., Kandemir N.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, cilt.4, sa.1, ss.34-38, 2012 (SCI-Expanded)

XCIX. **A review of the principles of radiological assessment of skeletal dysplasias**
ALANAY Y., Lachman R. S.
JCRPE Journal of Clinical Research in Pediatric Endocrinology, cilt.3, sa.4, ss.163-178, 2011 (SCI-Expanded)

C. **A mutation screen in patients with Kabuki syndrome**
Li Y., Boegershausen N., ALANAY Y., ŞİMŞEK KİPER P. Ö., Plume N., Keupp K., Pohl E., Pawlik B., Rachwalski M., Milz E., et al.
HUMAN GENETICS, cilt.130, sa.6, ss.715-724, 2011 (SCI-Expanded)

CI. **Craniosynostosis and Multiple Skeletal Anomalies in Humans and Zebrafish Result from a Defect in the Localized Degradation of Retinoic Acid**
Laue K., Pogoda H., Daniel P. B., van Haeringen A., ALANAY Y., von Ameln S., Rachwalski M., Morgan T., Gray M. J., Breuning M. H., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.89, sa.5, ss.595-606, 2011 (SCI-Expanded)

CII. **Catel-Manzke Syndrome: A Clinical Report Suggesting Autosomal Recessive Inheritance**
ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.9, ss.2288-2292, 2011 (SCI-Expanded)

CIII. **A rare case of 2q37 microdeletion with Albright hereditary osteodystrophy-like phenotype**
ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALANAY Y., Aktas D., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
TURKISH JOURNAL OF PEDIATRICS, cilt.53, sa.5, ss.558-560, 2011 (SCI-Expanded)

CIV. **Beals Hecht Syndrome-Arthrogryposis Multiplex Congenita-Congenital Arachnodactyly: Case Report**
Gokkaya N. K. O., Ucan H., Uckun A. C., ALANAY Y.
TURKIYE FIZIKSEL TIP VE REHABILITASYON DERGISI-TURKISH JOURNAL OF PHYSICAL MEDICINE AND REHABILITATION, cilt.57, sa.3, ss.178-181, 2011 (SCI-Expanded)

CV. **Spondyloenchondrodysplasia: a rare cause of short stature**
Yesiltepe-Mutlu G., ÖZSU CAVGA E., ÇİZMECİOĞLU JONES F. M., ALANAY Y., Hatun S.
TURKISH JOURNAL OF PEDIATRICS, cilt.53, sa.4, ss.464-466, 2011 (SCI-Expanded)

CVI. **Mutations in the TGF beta Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromicric**

and Geleophysic Dysplasias
Le Goff C., Mahaut C., Wang L. W., Allali S., Abhyankar A., Jensen S., Zylberberg L., Collod-Beroud G., Bonnet D., ALANAY Y., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.89, sa.1, ss.7-14, 2011 (SCI-Expanded)

CVII. **Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia**
Allali S., Le Goff C., Pressac-Diebold I., Pfennig G., Mahaut C., Dagoneau N., Alanay Y., Brady A. F., Crow Y. J., Devriendt K., et al.
JOURNAL OF MEDICAL GENETICS, cilt.48, sa.6, ss.417-421, 2011 (SCI-Expanded)

CVIII. **KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes**
Putoux A., Thomas S., Coene K. L. M., Davis E. E., ALANAY Y., Ogur G., Uz E., Buzas D., Gomes C., Patrier S., et al.
NATURE GENETICS, cilt.43, sa.6, ss.601-607, 2011 (SCI-Expanded)

CIX. **Rapid prenatal diagnosis of common aneuploidies by QF-PCR in the Turkish population**
Aktas D., Kutukcu B., Bayram Y., ÜTİNE G. E., ALANAY Y., ÖZYÜNCÜ Ö., DEREN Ö., Beksac S., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
CHROMOSOME RESEARCH, cilt.19, 2011 (SCI-Expanded)

CX. **Spondyloenchondrodysplasia with Systemic Lupus Erythematosus: a report of three cases**
BİLGİNER Y., ALANAY Y., DÜZOVA A., TOPALOĞLU R., Superti-Furga A., ÖZEN S., ÖZALTIN F., BEŞBAŞ N.
CLINICAL AND EXPERIMENTAL RHEUMATOLOGY, cilt.29, sa.2, ss.430, 2011 (SCI-Expanded)

CXI. **Genetic deficiency of tartrate-resistant acid phosphatase associated with skeletal dysplasia, cerebral calcifications and autoimmunity**
Lausch E., Janecke A., Bros M., Trojandt S., ALANAY Y., De Laet C., Huebner C. A., Meinecke P., Nishimura G., Matsuo M., et al.
NATURE GENETICS, cilt.43, sa.2, ss.132-139, 2011 (SCI-Expanded)

CXII. **PHENOTYPICAL PROPERTIES AND RESPONSE TO CHOLESTEROL THERAPY OF SMITH-LEMLI-OPITZ SYNDROME CASES**
Kilic M., Tokatli A., ALANAY Y., Kilic E., Kalkanoglu-Sivri H. S., DURSUN A., Onol S., Haliloglu G., Utine G. E., BODUROĞLU O. K., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

CXIII. **OPINIONS OF TURKISH PHYSICIANS TOWARDS TERMINATION OF PREGNANCY FOR FETAL DISORDERS**
Utine G. E., Kiper P. O., Salanci B. V., ALANAY Y., Aktas D., Alikasifoglu M., BODUROĞLU O. K., Tuncbilek E.
GENETIC COUNSELING, cilt.22, sa.4, ss.401-409, 2011 (SCI-Expanded)

CXIV. **Cervical Spine Dysmorphism: Report of Two Unusual Cases of Craniovertebral Junction**
Daglioglu E., ALANAY Y., Ozdol C., Okay O., Dalgic A., Belen D.
TURKIYE KLINIKLERI TIP BILIMLERI DERGISI, cilt.30, sa.6, ss.2052-2056, 2010 (SCI-Expanded)

CXV. **Intracranial and Extracranial Malformations in Patients With Craniofacial Anomalies**
Tuncbilek G., ALANAY Y., UZUN H., Kayikcioglu A., AKARSU A. N., Benli K.
JOURNAL OF CRANIOFACIAL SURGERY, cilt.21, sa.5, ss.1460-1464, 2010 (SCI-Expanded)

CXVI. **FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification? Response**
ALANAY Y., Krakow D.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.87, sa.2, ss.308, 2010 (SCI-Expanded)

CXVII. **Cowden syndrome with bronchial asthma**
ÖZSÜREKCİ Y., Yavuz S. T., ALANAY Y., ÜTİNE G. E., Kalayci O.
TURKISH JOURNAL OF PEDIATRICS, cilt.52, sa.3, ss.330-332, 2010 (SCI-Expanded)

CXVIII. **Disruption of ALX1 Causes Extreme Microphthalmia and Severe Facial Clefting: Expanding the Spectrum of Autosomal-Recessive ALX-Related Frontonasal Dysplasia**
Uz E., ALANAY Y., Aktas D., Vargel I., Gucer S., Tuncbilek G., von Eggeling F., Yilmaz E., DEREN Ö., Posorski N., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.86, sa.5, ss.789-796, 2010 (SCI-Expanded)

CXIX. **A Second Patient With Tsukahara Syndrome: Type A1 Brachydactyly, Short Stature, Hearing Loss, Microcephaly, Mental Retardation, and Ptosis**

ÜTİNE G. E., Breckpot J., Thienpont B., ALANAY Y., Aksoy C., BODUROĞLU O. K., Devriendt K.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.4, ss.947-949, 2010 (SCI-Expanded)

CXX. **A Variant of Desbuquois Dysplasia Characterized by Advanced Carpal Bone Age, Short Metacarpals, and Elongated Phalanges: Report of Seven Cases**
Kim O., Nishimura G., Song H., Matsui Y., Sakazume S., Yamada M., Narumi Y., ALANAY Y., Unger S., Cho T., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.4, ss.875-885, 2010 (SCI-Expanded)

CXXI. **Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta**
ALANAY Y., Avaygan H., Camacho N., ÜTİNE G. E., BODUROĞLU O. K., Aktas D., ALİKAŞİFOĞLU M., Tuncbilek E., ORHAN D., Bakar F. T., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.86, sa.4, ss.551-559, 2010 (SCI-Expanded)

CXXII. **Clinical and radiographic findings in two brothers affected with a novel mutation in matrix metalloproteinase 2 gene**
Gok F., Crettol L. M., ALANAY Y., Hacihamdioglu B., Kocaoglu M., Bonafe L., ÖZEN S.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.169, sa.3, ss.363-367, 2010 (SCI-Expanded)

CXXIII. **Disruption of the Podosome Adaptor Protein TKS4 (SH3PXD2B) Causes the Skeletal Dysplasia, Eye, and Cardiac Abnormalities of Frank-Ter Haar Syndrome**
Iqbal Z., Cejudo-Martin P., de Brouwer A., van der Zwaag B., Ruiz-Lozano P., Scimia M. C., Lindsey J. D., Weinreb R., Albrecht B., Megarbane A., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.86, sa.2, ss.254-261, 2010 (SCI-Expanded)

CXXIV. **Hemihyperplasia-multiple lipomatosis syndrome: an underdiagnosed entity in children with asymmetric overgrowth**
Boybeyi Ö., ALANAY Y., Kayikcioglu A., KARNAK İ.
JOURNAL OF PEDIATRIC SURGERY, cilt.45, sa.1, 2010 (SCI-Expanded)

CXXV. **OBSL1 mutations in 3-M syndrome are associated with a modulation of IGFBP2 and IGFBP5 expression levels**
Huber C., Fradin M., Edouard T., Le Merrer M., Alanay Y., Da Silva D. B., David A., Hamamy H., Van Hest L., Lund A. M., et al.
Human Mutation, cilt.31, sa.1, ss.20-26, 2010 (SCI-Expanded)

CXXVI. **Homozygous Inactivating Mutations in the NKX3-2 Gene Result in Spondylo-Megaepiphyseal-Metaphyseal Dysplasia**
HELLEMANS J., SIMON M., DHEEDENE A., ALANAY Y., MIHÇI E., RIFAI L., SEFIANI A., VAN BEVER Y., MERADJI M., SUPERTI-FURGA A., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.85, sa.6, ss.916-922, 2009 (SCI-Expanded)

CXXVII. **Identification of CANT1 Mutations in Desbuquois Dysplasia**
Huber C., Oules B., Bertoli M., Chami M., Fradin M., ALANAY Y., Al-Gazali L. I., Ausems M. G. E. M., Bitoun P., Cavalcanti D. P., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.85, sa.5, ss.706-710, 2009 (SCI-Expanded)

CXXVIII. **ALX4 dysfunction disrupts craniofacial and epidermal development**
Kayserili H., Uz E., Niessen C., VARGEL İ., ALANAY Y., Tuncbilek G., Yigit G., Uyguner O., Candan S., Okur H., et al.
HUMAN MOLECULAR GENETICS, cilt.18, sa.22, ss.4357-4366, 2009 (SCI-Expanded)

CXXIX. **Infantile Systemic Hyalinosis: A Case Report**
Gunduz O., ERSOY EVANS S., BODUROĞLU O. K., ALANAY Y., Ozkaya O.
TURKDERM-TURKISH ARCHIVES OF DERMATOLOGY AND VENEROLOGY, cilt.43, sa.3, ss.112-115, 2009 (SCI-Expanded)

CXXX. **Mild clinical phenotype and subtle radiographic findings in an infant with cartilage-hair hypoplasia**
Turkkani-Asal G., ALANAY Y., Turul-Ozgur T., Zenker M., Thiel C., Rauch A., ÜNAL S., Gurgey A., TEZCAN F. İ.
TURKISH JOURNAL OF PEDIATRICS, cilt.51, sa.5, ss.493-496, 2009 (SCI-Expanded)

CXXXI. **Wilms Tumor, AML and Medulloblastoma in a Child With Cancer Prone Syndrome of Total Premature Chromatid Separation and Fanconi Anemia**
Sari N., AKYÜZ C., Aktas D., GÜMRÜK F., ORHAN D., ALİKAŞİFOĞLU M., AYDIN G. B., ALANAY Y., BÜYÜKPAMUKÇU M.

PEDIATRIC BLOOD & CANCER, cilt.53, sa.2, ss.208-210, 2009 (SCI-Expanded)

CXXXII. **Identification of loss-of-function mutations of SLC35D1 in patients with Schneckenbecken dysplasia, but not with other severe spondylodysplastic dysplasias group diseases**
Furuichi T., Kayserili H., Hiraoka S., Nishimura G., Ohashi H., ALANAY Y., Lerena J. C., ASLANGER A. D., Koseki H., Cohn D. H., et al.
JOURNAL OF MEDICAL GENETICS, cilt.46, sa.8, ss.562-568, 2009 (SCI-Expanded)

CXXXIII. **Le Fort III Bipartition Osteotomy to Treat a Rare Craniofacial Anomaly: Frontofacionasal Dysostosis**
Tuncbilek G., ALANAY Y., Kayikcioglu A.
JOURNAL OF CRANIOFACIAL SURGERY, cilt.20, sa.4, ss.1056-1058, 2009 (SCI-Expanded)

CXXXIV. **Partial Distal Aphalangia, Duplication of Metatarsal IV, Microcephaly and Borderline Intelligence: A Third Patient Suggesting Autosomal Recessive Inheritance**
ÜTİNE G. E., ALANAY Y., Aktas D., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.6, ss.1317-1318, 2009 (SCI-Expanded)

CXXXV. **Rare sex chromosome aneuploidies: 49,XXXXY and 48,XXXY syndromes**
Simsek P. O., ÜTİNE G. E., ALİKAŞİFOĞLU A., ALANAY Y., BODUROĞLU O. K., Kandemir N.
TURKISH JOURNAL OF PEDIATRICS, cilt.51, sa.3, ss.294-297, 2009 (SCI-Expanded)

CXXXVI. **PORCN Mutations in Focal Dermal Hypoplasia: Coping with Lethality**
Bornholdt D., Oeffner F., Koenig A., Happle R., ALANAY Y., Ascherman J., Benke P. J., del Carmen Boente M., van der Burgt I., Chassaing N., et al.
HUMAN MUTATION, cilt.30, sa.5, 2009 (SCI-Expanded)

CXXXVII. **Subtelomeric rearrangements in mental retardation: Hacettepe University experience in 130 patients**
ÜTİNE G. E., Celik T., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K., Tuncbilek E., Aktas D.
TURKISH JOURNAL OF PEDIATRICS, cilt.51, sa.3, ss.199-206, 2009 (SCI-Expanded)

CXXXVIII. **TBX15 mutations cause craniofacial dysmorphism, hypoplasia of scapula and pelvis, and short stature in cousin syndrome**
Lausch E., Hermanns P., Farin H., ALANAY Y., Unger S., Nikkel S., Steinwender C., Scherer G., Spranger J., Zabel B., et al.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.168, sa.3, ss.379, 2009 (SCI-Expanded)

CXXXIX. **A new autosomal dominant Peters' anomaly phenotype expanding the anterior segment dysgenesis spectrum**
Berker N., ALANAY Y., Elgin U., Volkan-Salanci B., ŞİMŞEK T., AKARSU A. N., ALİKAŞİFOĞLU M.
ACTA OPHTHALMOLOGICA, cilt.87, sa.1, ss.52-57, 2009 (SCI-Expanded)

CXL. **Cyclic pamidronate treatment in Bruck syndrome: Proposal of a new modality of treatment**
Andiran N., ALİKAŞİFOĞLU A., ALANAY Y., YORDAM N.
PEDIATRICS INTERNATIONAL, cilt.50, sa.6, ss.836-838, 2008 (SCI-Expanded)

CXLI. **TBX15 Mutations Cause Craniofacial Dysmorphism, Hypoplasia of Scapula and Pelvis, and Short Stature in Cousin Syndrome**
Lausch E., Hermanns P., Farin H. F., ALANAY Y., Unger S., Nikkel S., Steinwender C., Scherer G., Spranger J., Zabel B., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.83, sa.5, ss.649-655, 2008 (SCI-Expanded)

CXLII. **Unilateral Peters' anomaly in an infant with 22q11.2 deletion syndrome**
Erdogan M. K., ÜTİNE G. E., ALANAY Y., Aktas D.
CLINICAL DYSMORPHOLOGY, cilt.17, sa.4, ss.289-290, 2008 (SCI-Expanded)

CXLIII. **Evaluation of prenatal-onset osteochondrodysplasias by ultrasonography: A retrospective and prospective analysis**
Krakow D., ALANAY Y., Rimoin L. P., Lin V., Wilcox W. R., Lachman R. S., Rimoin D. L.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.15, ss.1917-1924, 2008 (SCI-Expanded)

CXLIV. **Cryptic trisomy 5q35.2qter and deletion 1p36.3 characterised using FISH and array-based CGH**
Utine E. G., ALANAY Y., Aktas D., ALİKAŞİFOĞLU M., BODUROĞLU O. K., Vermeesch J., TUNÇBİLEK E., Fryns J.
EUROPEAN JOURNAL OF MEDICAL GENETICS, cilt.51, sa.4, ss.343-350, 2008 (SCI-Expanded)

CXLV. **Pseudo-trisomy 13 in a fetus: further support for autosomal recessive inheritance**

ÜTİNE G. E., ALANAY Y., Aktas D., TALİM B., Kale G., Tuncbilek E.
TURKISH JOURNAL OF PEDIATRICS, cilt.50, sa.3, ss.287-290, 2008 (SCI-Expanded)

CXLVI. **Clinical and radiographic delineation of odontochondrodysplasia**
Unger S., Antoniazzi F., Brugnara M., ALANAY Y., ÇAĞLAYAN A. O., Lachlan K., Ikegawa S., Nishimura G., Zabel B., Spranger J., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.146A, sa.6, ss.770-778, 2008 (SCI-Expanded)

CXLVII. **KABUKI SYNDROME AND TRISOMY 10p**
Utine G. E., ALANAY Y., Aktas D., BODUROĞLU O. K., Alikasifoglu M., Tuncbilek E.
GENETIC COUNSELING, cilt.19, sa.3, ss.291-300, 2008 (SCI-Expanded)

CXLVIII. **Dynamic cervicomedullary cord compression and alterations in cerebrospinal fluid dynamics in children with achondroplasia: Report of four cases**
Danielpour M., Wilcox W. R., Alanay Y., Pressman B. D., Rimoin D. L.
Journal of Neurosurgery, cilt.107, ss.504-507, 2007 (SCI-Expanded)

CXLIX. **Distal partial trisomy 1q: report of two cases and a review of the literature**
Utine G. E., Aktas D., ALANAY Y., Guecer S., Tuncbilek E., Mrasek K., Liehr T.
PRENATAL DIAGNOSIS, cilt.27, sa.9, ss.865-871, 2007 (SCI-Expanded)

CL. **Infantile systemic hyalinosis, a case report**
Gunduz O., Evans S. E., Boduroglu K., ALANAY Y., Ozkaya O.
VIRCHOWS ARCHIV, cilt.451, sa.2, ss.535, 2007 (SCI-Expanded)

CLI. **Cavernous malformation with Poland-Mijbius syndrome - Case illustration**
MUT AŞKUN M., Palaoglu S., ALANAY Y., Ismailoglu O., Tuncbilek E.
JOURNAL OF NEUROSURGERY, cilt.107, sa.1, ss.79, 2007 (SCI-Expanded)

CLII. **Angulated femurs and the skeletal dysplasias: Experience of the International Skeletal Dysplasia Registry (1988-2006)**
ALANAY Y., Krakow D., Rimoin D. L., Lachman R. S.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.11, ss.1159-1168, 2007 (SCI-Expanded)

CLIII. **Cerebro-facio-thoracic dysplasia: expanding the phenotype**
Cilliers D., ALANAY Y., BODUROĞLU O. K., Utine E., Tuncbilek E., Clayton-Smith J.
CLINICAL DYSMORPHOLOGY, cilt.16, sa.2, ss.121-125, 2007 (SCI-Expanded)

CLIV. **A molecular and clinical study of Larsen syndrome caused by mutations in FLNB**
Bicknell L. S., Farrington-Rock C., Shafeghati Y., Rump P., ALANAY Y., Alembik Y., Al-Madani N., Firth H., Karimi-Nejad M. H., Kim C. A., et al.
JOURNAL OF MEDICAL GENETICS, cilt.44, sa.2, ss.89-98, 2007 (SCI-Expanded)

CLV. **A multidisciplinary approach to the management of individuals with fragile X syndrome**
ALANAY Y., Unal F., Turanli G., Alikasifoglu M., Alehan D., Akyol U., Belgin E., Sener C., Aktas D., BODUROĞLU O. K., et al.
JOURNAL OF INTELLECTUAL DISABILITY RESEARCH, cilt.51, ss.151-161, 2007 (SCI-Expanded)

CLVI. **The skeletal dysplasias: Clinical-molecular correlations**
Rimoin D. L., Cohn D., Krakow D., Wilcox W., Lachman R. S., ALANAY Y.
SKELETAL BIOLOGY AND MEDICINE, PT B, cilt.1117, ss.302-309, 2007 (SCI-Expanded)

CLVII. **Terminal phalangeal accessory ossification center of the thumb: an additional radiographic finding in Larsen syndrome**
ALANAY Y., Utine G. E., Lachman R. S., Krakow D., Tuncbilek E.
PEDIATRIC RADIOLOGY, cilt.36, sa.9, ss.970-973, 2006 (SCI-Expanded)

CLVIII. **Congenital contractural arachnodactyly (Beals syndrome)**
Tuncbilek E., Alanay Y.
ORPHANET JOURNAL OF RARE DISEASES, cilt.1, 2006 (SCI-Expanded)

CLIX. **Spondylo-ocular syndrome: A new entity involving the eye and spine**
ALANAY Y., Superti-Furga A., Karel F., Tuncbilek E.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.140A, sa.6, ss.652-656, 2006 (SCI-Expanded)

CLX. **Spondyloenchondrodysplasia with spasticity, cerebral calcifications, and immune dysregulation:**

Clinical and radiographic delineation of a pleiotropic disorder
Renella R., Schaefer E., LeMerrer M., ALANAY Y., Kandemir N., Eich G., Costa T., Ballhausen D., Boltshauser E., Bonafe L., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.6, ss.541-550, 2006 (SCI-Expanded)

CLXI. **Analysis of MTHFR 1298A > C in addition to MTHFR 677C > T polymorphism as a risk factor for neural tube defects in the Turkish population**
BODUROĞLU O. K., ALANAY Y., Alikasifoglu M., Aktas D., Tuncbilek E.
TURKISH JOURNAL OF PEDIATRICS, cilt.47, sa.4, ss.327-333, 2005 (SCI-Expanded)

CLXII. **Is Dandy-Walker malformation associated with "distal 13q deletion syndrome"? Findings in a fetus supporting previous observations**
ALANAY Y., Aktas D., Utine E., Talim B., Onderoglu L., Caglar M., Tuncbilek E.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.3, ss.265-268, 2005 (SCI-Expanded)

CLXIII. **Response to the letter to "MTHTR 677C-T polymorphism is not excluded as maternal risk for Down syndrome among Turkish women"**
Boduroglu K., Alanay Y., Tuncbilek E., Koldan B.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.4, ss.462, 2005 (SCI-Expanded)

CLXIV. **MTHFR 677C-T polymorphism is not excluded as maternal risk for Down syndrome among Turkish women [4] (multiple letters)**
Martínez-Frías M. L., Bermejo E., Rodríguez-Pinilla E., Prieto D., Prieto L., Boduroğlu K., Alanay Y., Tunçbilek E., Koldan B.
American Journal of Medical Genetics, sa.4, ss.461-462, 2005 (SCI-Expanded)

CLXV. **Celiac disease screening in 100 Turkish children with Down syndrome**
ALANAY Y., BODUROĞLU O. K., Tuncbilek E.
TURKISH JOURNAL OF PEDIATRICS, cilt.47, sa.2, ss.138-140, 2005 (SCI-Expanded)

CLXVI. **Clinical heterogeneity of hereditary optic atrophy in a Turkish family**
Acaroglu G., ALANAY Y., Reynier P., Amati-Bonneau P., Men G.
NEURO-OPHTHALMOLOGY, cilt.29, sa.1, ss.9-15, 2005 (SCI-Expanded)

CLXVII. **The evolution of sagittal segmental alignment of the spine during childhood**
Cil A., Yazici M., Uzumcugil A., Kandemir U., Alanay A., ALANAY Y., Acaroglu R., Surat A.
SPINE, cilt.30, sa.1, ss.93-100, 2005 (SCI-Expanded)

CLXVIII. **A case of ring chromosome 18 with mild phenotypic features**
BODUROĞLU O. K., ALANAY Y., Tuncbillek E.
CHROMOSOME RESEARCH, cilt.13, ss.66, 2005 (SCI-Expanded)

CLXIX. **Phenotypic and molecular characterization of Bruck syndrome (osteogenesis imperfecta with contractures of the large joints) caused by a recessive mutation in PLOD2**
Ha-Vinh R., ALANAY Y., Bank R., Campos-Xavier A., Zankl A., Superti-Furga A., Bonafe L.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.2, ss.115-120, 2004 (SCI-Expanded)

CLXX. **Oculo-palato-cerebral syndrome: A third case supporting autosomal recessive inheritance**
ALANAY Y., BODUROĞLU O. K., Sonmez B., Orhan M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.1, ss.92-95, 2004 (SCI-Expanded)

CLXXI. **Noonan syndrome and systemic lupus erythematosus: presentation in childhood**
ALANAY Y., Balci S., Ozen S.
CLINICAL DYSMORPHOLOGY, cilt.13, sa.3, ss.161-163, 2004 (SCI-Expanded)

CLXXII. **Methylenetetrahydrofolate reductase enzyme polymorphisms as maternal risk for Down syndrome among Turkish women**
BODUROĞLU O. K., ALANAY Y., Koldan B., Tuncbilek E.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.1, ss.5-10, 2004 (SCI-Expanded)

CLXXIII. **Perinatal mortality rate - Hospital based study during 1998-2001 at Hacettepe University**
Durukan T., Onderoglu L., Deren O., Saygan-Karamursel B., Erdem G., Oran O., Yurdakok M., Tekinalp G., Yigit S., Korkmaz A., et al.
JOURNAL OF PERINATAL MEDICINE, cilt.31, sa.5, ss.435-440, 2003 (SCI-Expanded)

CLXXIV. **Localized acquired hypertrichosis following cast application**
Kara A., Kanra G., Alanay Y.
PEDIATRIC DERMATOLOGY, cilt.18, sa.1, ss.57-59, 2001 (SCI-Expanded)

Diğer Dergilerde Yayınlanan Makaleler

I. **Variants in ACTC1 underlie distal arthrogryposis accompanied by congenital heart defects**
Chong J. X., Childers M. C., Marvin C. T., Marcello A. J., Gonorazky H., Hazrati L., Dowling J. J., Al Amrani F., ALANAY Y., Nieto Y., et al.
HUMAN GENETICS AND GENOMICS ADVANCES, cilt.4, sa.3, 2023 (ESCI)

II. **Genetik Hastalık Şüphesi Olan Fetal ve Pediatrik Hastalarda Moleküler Otopsinin Klinik Faydası**
AKGÜN DOĞAN Ö., AĞAOĞLU N. B., ALANAY Y.
Gümüşhane Üniversitesi Sağlık Bilimleri Dergisi, cilt.11, sa.1, ss.82-89, 2022 (Hakemli Dergi)

III. **Intrauterine Cataract Diagnosis and Follow-up**
Aksay S., BİLDİRİCİ İ., COŞAR C. B., ALANAY Y., Cigerciogullari E.
TURK OFTALMOLOJI DERGISI-TURKISH JOURNAL OF OPHTHALMOLOGY, cilt.50, sa.4, ss.245-247, 2020 (Hakemli Dergi)

IV. **The process of how mothers with down's syndrome children learn about the diagnosis: A qualitative research**
Basgul S. S., Mutlu D., ALANAY Y., Toker M.
ANNALS OF CLINICAL AND ANALYTICAL MEDICINE, cilt.11, sa.1, ss.54-58, 2020 (Hakemli Dergi)

V. **Dental Findings and Mutational Analysis of a Case withEhlers-Danlos Syndrome**
HAKKI S., Aktaş D., ALANAY Y., AVUNDUK M. C., HAKKI E. E.
Journal of Dentistry and Oral Biology, cilt.2, ss.1-5, 2017 (Hakemli Dergi)

VI. **Gorlin Syndrome in Eleven Patients**
ÜTİNE G. E., ALANAY Y., Aktas D., BODUROĞLU O. K., ALİKAŞİFOĞLU M., TUNÇBİLEK E.
JOURNAL OF PEDIATRIC RESEARCH, cilt.4, sa.2, ss.63-67, 2017 (Hakemli Dergi)

VII. **Clinical genetics and classification of craniosynostosis**
ŞAHİN O. N., Alanay Y.
Türk Nöroşirürji Dergisi, cilt.27, sa.3, ss.255-262, 2017 (Hakemli Dergi)

VIII. **FKBP10 (FKBP65 Protein), Osteogenesis Imperfecta and Bruck Syndrome**
Krakow D., ALANAY Y.
OSTEOGENESIS IMPERFECTA: A TRANSLATIONAL APPROACH TO BRITTLE BONE DISEASE, ss.151-157, 2014 (Hakemli Dergi)

IX. **Mutations in the interleukin receptor IL11RA cause autosomal recessive Crouzon-like craniosynostosis**
Keupp K., Li Y., VARGEL İ., Hoischen A., Richardson R., Neveling K., ALANAY Y., Uz E., ELÇİOĞLU H. N., Rachwalski M., et al.
MOLECULAR GENETICS & GENOMIC MEDICINE, cilt.1, sa.4, ss.223-237, 2013 (ESCI)

X. **Early physiotherapy interventions in a case with the Say/Barber/Biesecker/Young-Simpson type of Ohdo syndrome and arthrogryposis multiplex congenita**
KINIKLI G. İ., GÜR G., MUTLU A., KEREM GÜNEL M., ALANAY Y.
Yeni Tıp Dergisi, cilt.29, sa.2, ss.108-111, 2012 (Hakemli Dergi)

XI. **Pentalogy of Cantrell: A case report Tam ektopia kordisli bir Cantrell pentalojisi vakasi**
KILIÇ E., Alanay Y., Korkmaz A., Ütine E., KARAGÖZ T., Boduroğlu K.
Cocuk Sagligi ve Hastaliklari Dergisi, cilt.54, sa.2, ss.83-86, 2011 (Scopus)

XII. **Searching f(o)r Copy Number Changes in Nonsyndromic X-Linked Intellectual Disability**
Utine G. E., Kiper P. O., ALANAY Y., Haliloglu G., Aktas D., BODUROĞLU O. K., Tuncilek E., Alikasifoglu M.
MOLECULAR SYNDROMOLOGY, cilt.2, sa.2, ss.64-71, 2011 (Hakemli Dergi)

XIII. **Perinatal mortality analysis from 2001-2006 at Hacettepe University Hospital Hacettepe Üniversitesi**

Hastanesi 2001-2006 dönemi perinatal mortalite analizi
Korkmaz A., AKÇÖREN Z., Alanay Y., ÖZYÜNCÜ Ö., YİĞİT Ş., DEREN Ö., TALİM B., ORHAN D., Güçer Ş., YURDAKÖK M., et al.
Cocuk Sagligi ve Hastaliklari Dergisi, cilt.53, sa.3, ss.175-188, 2010 (Scopus)

XIV. **Femoral-facial syndrome: A case-report Femoral hipoplazi ve değişik yüz görünü mü sendromu: Bir vaka takdimi**
Korkmaz A., Çinar A., TEKŞAM Ö., YURDAKÖK M., Alanay Y., Balci S.
Cocuk Sagligi ve Hastaliklari Dergisi, cilt.47, sa.3, ss.205-208, 2004 (Scopus)

Kitap & Kitap Bölümleri

I. **Akondroplazi**
AKGÜN DOĞAN Ö., ALANAY Y.
Çocuk Genetik Hastalıkları, Mıhçı, Ercan, Editör, Türkiye Klinikleri Yayınevi, Ankara, ss.66-71, 2021

II. **Genetik Danışma**
AKGÜN DOĞAN Ö., ALANAY Y.
Cerebral Palsy ve Genetik, Özbek Uğur, Akçakaya Nihan Hande, Editör, Boyut Yayınevi, İstanbul, ss.57-65, 2019

III. **Kısım 10 Klinik Genetik ve Dismorfoloji, İskeletin Genetik Hastalıkları**
ŞİMŞEK KİPER P. Ö., ALANAY Y.
Yurdakök Pediatri, Murat Yurdakök, Editör, Güneş Tıp Kitabevi, Ankara, ss.2024-2047, 2017

Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

I. **Yoğun Bakımdaki Kritik Yenidoğan ve Süt Çocuklarında Hızlı Yeni Nesil Dizileme ile Genetik Tanı-İlk Türkiye Deneyimi**
Güner Yılmaz B., Akgün Doğan Ö., Özdemir Ö., Bilgüvar K., Hatırnaz Ng Ö., Özköse G. Ş., Demirel A., Aktaş S., Arcagok B. C., Kazancı E., et al.
Ulusal Neonatoloji Kongresi, Antalya, Türkiye, 24 - 28 Nisan 2024

II. **Neuroimaging features of MOPDII in ten patients with PCNT mutation: A Tertiary Centre Experience**
Akalin A., GÖÇMEN R., Simsek-Kiper P., TAŞKIRAN Z. E., ALANAY Y., HALİLOĞLU V. G., ÜTİNE G. E., Boduroglu K.
56th Annual Conference of the European-Society-of-Human-Genetics (ESHG), Glasgow, İngiltere, 10 - 13 Haziran 2023, ss.408-409

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.
55th European-Society-of-Human-Genetics (ESHG) Conference, Vienna, Avusturya, 11 - 14 Haziran 2022, ss.656-657

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

Yilmaz B. G., ABALI S., Akberzade A., Ay B., Tumer S., AKGÜN DOĞAN Ö., Nishimura G., ALANAY Y.
55th European-Society-of-Human-Genetics (ESHG) Conference, Vienna, Avusturya, 11 - 14 Haziran 2022, ss.127

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., KARAARSLAN E., Bildirici I., ALANAY Y.
55th European-Society-of-Human-Genetics (ESHG) Conference, Vienna, Avusturya, 11 - 14 Haziran 2022, ss.105

IX. **Multiple epiphyseal dysplasia: A diagnostic challenge with genetic heterogeneity**
Dasar T. N., TAŞKIRAN Z. E., Urel-Demir G., KARAOSMANOĞLU B., İMREN G., YILMAZ G., Alanay Y., ÜTİNE G. E., Boduroglu K., Simsek-Kiper P.
55th European-Society-of-Human-Genetics (ESHG) Conference, Vienna, Avusturya, 11 - 14 Haziran 2022, ss.397

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.
Ulusal Neonatoloji Kongresi, Antalya, Türkiye, 26 - 30 Nisan 2023

XI. **Severely Affected Newborn Female With De Novo Likely Pathogenic Variant In BCAP31**
Güner Yılmaz B., Aktas S., Kazancı E., Zeybel G., Özköse G. Ş., Akgün Doğan Ö., Korkmaz Toygar A., Alanay Y.
Undiagnosed diseases network international (UDNI), Vienna, Avusturya, 7 - 08 Kasım 2022

XII. **Evaluation of the etiology of epilepsy and/or developmental delay in children with next generation sequencing:A single center experience**
Kava H., Alanay Y., Yeşilyurt A., Isık U.
17. International Child Neurology Congress, Antalya, Türkiye, 3 - 07 Ekim 2022, ss.2

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.
54th Conference of the European-Society-of-Human-Genetics (ESHG), ELECTR NETWORK, 28 - 31 Ağustos 2021, ss.118

XIV. **The third family with TAF6-related phenotype: Alazami-Yuan syndrome**
Tuc E., Bengur F. B., Aykut A., ŞAHİN Ö., ALANAY Y.
53rd Conference of the European-Society-of-Human-Genetics (ESHG), ELECTR NETWORK, 6 - 09 Haziran 2020, ss.904

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.
Annual Meeting of the American-Society-for-Bone-and Mineral Research (ASBMR), ELECTR NETWORK, 11 - 15 Eylül 2020, cilt.35, ss.18

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.
52nd Conference of the European-Society-of-Human-Genetics (ESHG), Gothenburg, İsveç, 15 - 18 Haziran 2019, cilt.27, ss.1506-1507

XVII. **Screening TYR gene variations in Turkish oculocutaneuse albinism patients**
Ng O. H., YILMAZ E., Parlakgunes Z., YARARBAŞ K., Ziylan S., ALANAY Y., ÖZBEK U.
52nd Conference of the European-Society-of-Human-Genetics (ESHG), Gothenburg, İsveç, 15 - 18 Haziran 2019, cilt.27, ss.1233-1234

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**
ZEYBEK S., TEMEL Ş. G., NUR B., ÖZEMRİ SAĞ Ş., ALANAY Y., MIHÇI E.
4. Ulusal Çocuk Genetik Kongresi, Türkiye, 25 - 27 Eylül 2019

XX. **The Skeletal Dysplasia Registry: Hacettepe Experience**
ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., KOŞUKCU C., ARSLAN U. E., ÜTİNE G. E., ALANAY Y., ALİKAŞİFOĞLU M.,

BODUROĞLU O. K.
The 14th biannual International Skeletal Dysplasia Society Meeting, Oslo, Norveç, 11 - 14 Eylül 2019

XXI. **Possible effect of IGFR1 gene on macrocephaly in a patient with unbalanced 6;15 translocation with 6p25 deletion and 15q26 duplication**
YARARBAŞ K., Sayar C., Tumer S., Ozciger D., Tavukcu C., Oncu N., ALANAY Y.
51st Conference of the European-Society-of-Human-Genetics (ESHG) in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), Milan, İtalya, 16 - 19 Haziran 2018, cilt.27, ss.992

XXII. **Ischiospinal Dysostosis in a boy with a novel homozygous missense mutation in the BMPER gene**
Kablan A., Mat B., Temel Ş. G., ALANAY Y.
51st Conference of the European-Society-of-Human-Genetics (ESHG) in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), Milan, İtalya, 16 - 19 Haziran 2018, cilt.27, ss.916-917

XXIII. **Clinical, demographic and nosologic characterisation of the genetic disorders of the skeleton in Turkey: The skeletal dysplasia registry**
Simsek-Kiper P. O., Utine G. E., Taskiran E. Z., Kosukcu C., Arslan U., ALANAY Y., Alikasifoglu M., BODUROĞLU O. K.
51st Conference of the European-Society-of-Human-Genetics (ESHG) in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), Milan, İtalya, 16 - 19 Haziran 2018, cilt.27, ss.130

XXIV. **Camptodactyly-arthropathy-coxa vara-pericarditis syndrome in a large family: A clinical condition with a diagnostic challange**
Oguz S., Kiper P. O. S., Utine G. E., ALANAY Y., Ozen S., BODUROĞLU O. K., Alikasifoglu M.
51st Conference of the European-Society-of-Human-Genetics (ESHG) in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), Milan, İtalya, 16 - 19 Haziran 2018, cilt.27, ss.97

XXV. **Next Generation Sequencing (NGS) panel revealed new candidate genes and variants in 25 Hypertrophic Cardiomyopathy patients**
Turkgenc B., TEMEL Ş. G., UYSAL F., Atik S. U., Oztunc F., Sulu A., EKİCİ F., Ayabakan C., Odemis E., Saygili A., et al.
51st Conference of the European-Society-of-Human-Genetics (ESHG) in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), Milan, İtalya, 16 - 19 Haziran 2018, cilt.27, ss.150

XXVI. **BİLEŞİK HETEROZİGOT GALNS MUTASYONU İLE TANI ALAN KLASİK OLMAYAN MUKOPOLİSAKKARİDOZ TİP IVA**
ABALI S., YARARBAŞ K., Ersoy Olbak M., Karabıyık N., İçten İ., ÇELEBİ A. R. C., ARSLAN Ş., ALANAY Y.
3. EGE ENDOKRİN HASTALIKLAR VE GENETİK SEMPOZYUMU, Türkiye, 8 - 09 Mart 2019

XXVII. **Baraitser-Winter Syndrome in a boy with heterozygous missense mutation in the ACTB gene**
Temel Ş. G., Ekmekci G., Yenmis G., Kiper P. O. S., ALANAY Y.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Danimarka, 27 - 30 Mayıs 2017, cilt.26, ss.956

XXVIII. **Camptodactyly-arthropathy-coxa vara-pericarditis syndrome in a large family: A clinical condition with a diagnostic challenge.**
OĞUZ S., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALANAY Y., ÖZEN S., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
European Human Genetics Conference, 16 - 19 Haziran 2018

XXIX. **Otozomal resesif infantil bilateral striatal nekroz,nup 62 geninde mutasyon: olgu sunumu**
ÜNVER O., SAĞER S. G., ÖZTÜRK THOMAS G., EKİNCİ G., ALANAY Y., TÜRKDOĞAN D.
20. Çocuk Ulusal Nörolojisi Kongresi, Kıbrıs (Kktc), 2 - 06 Mayıs 2018

XXX. **Normal enzim düzeyleri ve Japon Bayrağı göz dibi ile GM2 Gangliyosidoz tanısı alan iki kardeş: olgu sunumu**
ÜNVER O., ÖZTÜRK THOMAS G., SAĞER S. G., EKİNCİ G., ALANAY Y., TÜRKDOĞAN D.
6. uluslararası katılımlı lizozomal hastalıklar kongresi, Türkiye, 11 - 15 Nisan 2018

XXXI. **Targeted custom gene panel sequencing for cardiac ion channelopathies: Efficiently detects candidate pathogenic mutations in Long QT syndrome**
TEMEL Ş. G., Turkgenc B., KARADAĞ Ö., AYKAN H. H., UYSAL F., Bastuhan I. Y., Sulu A., Atik S. U., Cinar B., Dedeoglu R., et al.
European Biotechnology Congress, Dubrovnik, Hırvatistan, 25 - 27 Mayıs 2017, cilt.256

XXXII. **A NEW MUTATION IN THE WISP3 GENE (c.935_936insTp. C314Lfs7) IN A PATIENT WITH**

PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA
ABUR Ü., OĞUR M. G., AYKUT A., ALANAY Y.
Erciyes Medical Journal, 11 - 13 Mayıs 2017, cilt.39

XXXIII. **Identification of Molecular Pathology of Peters' Anomaly Segregating in a Large Autosomal Dominant Family**
KOŞUKCU C., ASLI K., ALANAY Y., KAVAK P., BERKER N., TAŞKIRAN Z. E., ALİKAŞİFOĞLU M., SEZERMAN O. U., AKARSU A. N.
10th International Symposium on Health Informatics and Bioinformatics (HIBIT 2017), KALKANLI, GUZELYURT, Kıbrıs (Kktc), 29 - 30 Haziran 2017

XXXIV. **A report of mosaic Turner syndrome with a mild Kabuki like phenotype**
Yararbas K., Sayar C., Tumer S., Tavukcu C., Oncu N., Ozciger D., Ekmekci C., KUŞKUCU A., ALANAY Y.
cilt.10

XXXV. **Baraitser-Winter Syndromein a boy with heterozygote missense mutation in the ACTB gene.**
TEMEL Ş. G., ALANAY Y., Cumhur E., Pelin Özlem K. Ş.
ESHG Conference 2017, 27 - 30 Mayıs 2017

XXXVI. **Mitokondriyal hastalıklarda genetik testlerin rolü-iki olgu sunumu**
ÖZTÜRK THOMAS G., ÜNVER O., SAĞER S. G., KUTLUBAY B., ALANAY Y., Yüksel F., EKİNCİ G., TÜRKDOĞAN D.
19. Ulusal çocuk nörolojisi kongresi, Türkiye, 19 - 23 Nisan 2017

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.
28th International Congress of Pediatrics, 17 - 22 Ağustos 2016

XXXVIII. **Detection of 15q (Prader Willi/Angelman) deletion in maternal cell-free fetal dna test, a case report**
BATUKAN A. C., TAKMAZ Ö., YAKICIER M. C., ALANAY Y., ÖZBAŞLI E., Tosun O.
25th European Congress of Perinatal Medicine, Maastricht, Hollanda, 15 - 18 Haziran 2016, cilt.29, ss.21-22

XXXIX. **Meier Gorlin ear patella short stature syndrome A rare clinical entity**
AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ALANAY Y., ÜTİNE G. E., BODUROĞLU O. K.
European Society of Human Genetics Conference 2016, 21 - 24 Mayıs 2016

XL. **OBSL1 Mutations Represent The Major Gene Defect In A Group Of 3M Syndrome Patients A Study From Turkey**
ŞİMŞEK KİPER P. Ö., Taşkıran E. Z., ARSLAN U., Koşukçu C., AKGÜN DOĞAN Ö., ÜTİNE G. E., ALİKAŞİFOĞLU A., KANDEMİR N., Cormier Daire V., ALANAY Y., et al.
European Society of Human Genetics Conference 2016, 21 - 24 Mayıs 2016

XLI. **Detection of 15q (Prader Willi/ Angelman syndrome) deletion in maternal cell-free fetal DNA test; A case report**
BATUKAN C., TAKMAZ Ö., YAKICIER C., ALANAY Y., ÖZBAŞLI E.
XI. Türk Alman Jinekoloji Kongresi, Antalya, Türkiye, 11 - 15 Mayıs 2016

XLII. **Parsiyel Trizomi 19 9 ile ilşkili bir İmmün Yetmezlik**
BOZDOĞAN G., VARDAR G., ÇITAK A., ERTEKİN V., ALANAY Y.
2. Klinik İmmonoloji Kongresi, Türkiye, 31 Mart - 03 Nisan 2016

XLIII. **Parsiyel Trizomi 19p13 ile iliişkili bir immün yetmezlik**
BOZDOĞAN G., VARDAR G., ÇITAK A., ERTEKİN V., ALANAY Y.
2. Klinik İmmunoloji Kongresi, Türkiye, 31 Mart - 03 Nisan 2016

XLIV. **3M Sendromlu Bir Grup Hastada Klinik Ve Moleküler Bulguların Analizi**
ŞİMŞEK KİPER P. Ö., EKİM ZİHNİ T., ÜTİNE G. E., ALİKAŞİFOĞLU A., KANDEMİR N., VALERİE C. D., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
2. Ulusal Çocuk Genetik Sempozyumu, 22-24 Ekim 2015, Samsun, Türkiye, Türkiye, 22 - 24 Ekim 2015

XLV. **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**
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.
12thINTERNATIONAL SKELETAL DYSPLASIA SOCIETY MEETING', 29 Temmuz - 01 Ağustos 2015

XLVIII. **Spotlight on the pathogenesis of Kabuki syndrome**
Bögershausen N., Tsai I. C., Pohl E., ŞİMŞEK KİPER P. Ö., Beleggia F., PERÇİN F. E., Keupp K., Angela M., ALANAY Y., KAYSERİLİ KARABEY H., et al.
European Human Genetics Conference - ESHG 2015, Glasgow, Scotland, UK, 6 - 09 Haziran 2015

XLIX. **Spotlight on the pathogenesis of Kabuki syndrome**
NİNA B., TSAİ I., POHL E., ŞİMŞEK KİPER P. Ö., BELEGGIA F., PERÇİN F. E., KEUPP K., MATRCHAN A., MİLZ E., ALANAY Y., et al.
ESHG 2015, 6-9 June 2015, Glasgow, Scotland, United Kingdom., 6 - 09 Haziran 2015

L. **Spotlight on the pathogenesis of Kabuki syndrome**
Bögershausen N., Tsai I., Pohl E., ŞİMŞEK KİPER P. Ö., Beleggia F., Percin F., Keupp K., ALANAY Y., KAYSERİLİ KARABEY H., ELÇİOĞLU H. N.
European Human GeneticsConference 2015, 6 - 09 Haziran 2015

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.
16.Ulusal Pediatrik Endokrin ve Diyabet Kongresi, Samsun, Türkiye, 6 - 10 Kasım 2012

LII. **Cabezas Sendromu**
ÖZSU E., RAHİME GÜL Y. M., ÇİZMECİOĞLU F. M., ALANAY Y., HATUN Ş.
15. Ulusal Pediatrik Endokrinoloji ve Diyabet Kongresi, İzmir, Türkiye, 23 - 26 Kasım 2011

LIII. **Metabolik Sendrom Carnevale Sendrom Birlikteliği Vaka Sunumu**
RAHİME GÜL Y. M., ALANAY Y., ÖZSU E., ÇİZMECİOĞLU F. M., HATUN Ş.
15.Ulusal Pediatrik Endokrin ve Diyabet Kongresi, İzmir, Türkiye, 23 - 26 Kasım 2011

LIV. **BİR SPONDİLO ENDONDRO DİSPLAZİ VAKASI**
RAHİME GÜL Y. M., ÖZSU E., ÇİZMECİOĞLU F. M., ALANAY Y., HATUN Ş.
14. Ulusal Pediatrik Endokrin ve Diyabet Kongresi, Muğla, Türkiye, 4 - 10 Ekim 2010

LV. **Two siblings with severe deforming osteogenesis imperfecta with mutations in the newly idendified 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.
6th European Cytogenetics Conference, İstanbul, Türkiye, 7 - 10 Temmuz 2007, cilt.15, ss.70

Desteklenen Projeler

Akgün Doğan Ö., Sütçübaşı B., Alanay Y., Diğer Uluslararası Fon Programları, Arid1b Ile İlişkili Bozuklukta Gelişimsel İlerleme Alanları - Çok Yöntemli 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 (Kktc), 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