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Personal Information

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Education Information

Post Doctorate of Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Turkey 2002 - 2011

Doctorate, Hacettepe University, Sağlık Bilimleri Enstitüsü, Pediatrik Temel Bilimler A.B.D., Turkey 2004 - 2009

Post Doctorate, University of California, Los Angeles, Cedars-Sinai Hospital, Intercampus Genetics Program, Skeletal Dysplasia Registry, United States Of America 2005 - 2006

Expertise In Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Turkey 1997 - 2002

Under Graduate, Hacettepe University, Tıp Fakültesi (İngilizce), Turkey 1990 - 1996

Research Areas

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

Academic Titles / Tasks

Professor, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2013 - Continues

Associate Professor, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2011 - 2013

Associate Professor, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2008 - 2011

Assistant Professor, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2007 - 2008

Professional Experience

Academic Performance D. Board Member, Acibadem Mehmet Ali Aydınlar University, School Of Medicine, Department Of Medical Sciences, 2018 - Continues

Akreditasyon, Sürekli Gelişim ve Yenilenme Kurulu Üyesi, Acibadem Mehmet Ali Aydınlar University, School Of Medicine, Department Of Medical Sciences, 2018 - Continues

Uygulama ve Araştırma Merkezi Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, 2017 - Continues

Uygulama ve Araştırma Merkezi Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, 2016 - Continues

Rectorate Commissioner, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2016 - Continues

University Executive Board Member, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2015 - Continues

Dean, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, 2015 - Continues

Head of Department, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2015 - Continues

Rectorate Commissioner, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, 2015 - Continues

Rectorate Commissioner, Acibadem Mehmet Ali Aydınlar University, Tıp Fakültesi, 2015 - 2018

Courses

PANEL: FROM GENOTYPE TO PHENOTYPE, Under Graduate, 2019 - 2020

NONMENDELIAN INHERITANCE, Under Graduate, 2019 - 2020

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

- **The third family with TAF6-related phenotype: Alazami-Yuan syndrome.**
Tuc E., Bengur F., Aykut A., Sahin O., Alanay Y.
Clinical genetics, 2020 (Journal Indexed in SCI)
- **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**
Şimşek Kiper P. Ö. , Taskiran E., Koşukcu C., Arslan U. E. , Cormier-Daire V., Gonc N., Ozon A., Alikasıfoğlu A., Kandemir N., Ütine G. E. , et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, pp.1157-1172, 2019 (Journal Indexed in SCI)
- **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome**
van der Sluijs P. J. , Jansen S., Vergano S. A. , Adachi-Fukuda M., ALANAY Y., AlKindy A., Baban A., Bayat A., Beck-Woedl S., Berry K., et al.
GENETICS IN MEDICINE, vol.21, pp.1295-1307, 2019 (Journal Indexed in SCI)
- **p.Ser348Cys mutation in FGFR3 gene leads to "Mild ACH /Severe HCH" phenotype.**
Bengur F., Ekmekci C., Karaarslan E., Gunoz H., Alanay Y.
European journal of medical genetics, pp.103659, 2019 (Journal Indexed in SCI)
- **A possible founder mutation in FZD6 gene in a Turkish family with autosomal recessive nail dysplasia.**
Saygi C., ALANAY Y., Sezerman U., Yenenler A., Ozoren N.
BMC medical genetics, vol.20, pp.15, 2019 (Journal Indexed in SCI)
- **Epigenotype and phenotype correlations in patients with Beckwith-Wiedemann syndrome**
Bilgin B., Kabacam S., Taskiran E., Simsek-Kiper P. O. , Alanay Y., Boduroglu K., ÜTİNE G. E.
TURKISH JOURNAL OF PEDIATRICS, vol.60, pp.506-513, 2018 (Journal Indexed in SCI)
- **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., Yılmaz C., Zaki M. S. , Rosti R. O. , Copeland B., et al.
NATURE GENETICS, vol.50, pp.1093-1107, 2018 (Journal Indexed in SCI)
- **Fragile x-associated premature ovarian failure in a large Turkish cohort: Findings of Hacettepe Fragile X Registry**
ÜTİNE G. E. , ŞİMŞEK KİPER P. Ö. , Akgun-Dogan O., ÜREL DEMİR G., Alanay Y., Aktas D., Boduroglu K., Tuncbilek E., ALİKAŞİFOĞLU M.
EUROPEAN JOURNAL OF OBSTETRICS & GYNECOLOGY AND REPRODUCTIVE BIOLOGY, vol.221, pp.76-80, 2018 (Journal Indexed in SCI)
- **Hematopoietic Stem Cell Transplantation for Myelodysplastic Syndrome in a Child With Klinefelter Syndrome**
Serdaroglu E., Kuskonmaz B., ALANAY Y., Aytac S., Cetin M., Cetinkaya D. U.
JOURNAL OF PEDIATRIC HEMATOLOGY ONCOLOGY, vol.40, pp.81-82, 2018 (Journal Indexed in SCI)
- **Parieto-occipital alopecia in early infancy mandates cranial imaging**
Tuc E., KARAARSLAN E., Celik I., ALANAY Y.
CLINICAL DYSMORPHOLOGY, vol.27, pp.15-17, 2018 (Journal Indexed in SCI)
- **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.
MOLECULAR CYTOGENETICS, vol.10, 2017 (Journal Indexed in SCI)
- **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, vol.32, pp.1309-1319, 2017 (Journal Indexed in SCI)

- **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, vol.91, pp.868-880, 2017 (Journal Indexed in SCI)
- **Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa**
Van Damme T., Gardeitchik T., Mohamed M., Guerrero-Castillo S., Freisinger P., Guillemy B., Kariminejad A., Dalloyaux D., Van Kraaij S., Lefeber D. J. , et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.100, pp.216-227, 2017 (Journal Indexed in SCI)
- **AUTS2 Syndrome in a 68-Year-Old Female: Natural History and Further Delineation of the Phenotype**
Sengun E., Yazarbas K., Kasakyan S., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.170, pp.3231-3236, 2016 (Journal Indexed in SCI)
- **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., ŞİMŞEK KİPER P. Ö. , Kroes H. Y. , et al.
GENETICS IN MEDICINE, vol.18, pp.882-891, 2016 (Journal Indexed in SCI)
- **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, vol.170, pp.1889-1894, 2016 (Journal Indexed in SCI)
- **Three cases of spondyloenchondrodysplasia (SPENCD) with systemic lupus erythematosus: a case series and review of the literature**
Bilginer Y., Duzova A., Topaloglu R., Batu E. D. , Boduroglu K., Gucer S., Bodur I., Alanay Y.
LUPUS, vol.25, pp.760-765, 2016 (Journal Indexed in SCI)
- **Late Sequel of Meningococemia: Skeletal Dysplasia**
Karabulut G. S. , Acar T., Yuksel A. B. , ALANAY Y., Hatun S.
JOURNAL OF PEDIATRICS, vol.173, pp.264-266, 2016 (Journal Indexed in SCI)
- **Nonsense Mutation in SPARC Gene Causing Autosomal Recessive Osteogenesis Imperfecta**
Abali S., ARMAN A., Atay Z., Bas S., Cam S., Gormez Z., Demirci H., ALANAY Y., Akarsu N., BEREKET A., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.167, 2016 (Journal Indexed in SCI)
- **Seizures and diagnostic difficulties in hyperinsulinism-hyperammonemia syndrome.**
Aka S., ALANAY Y., Boodhansingh K. E. , Stanley C. A. , Semiz S.
The Turkish journal of pediatrics, vol.58, pp.541-544, 2016 (Journal Indexed in SCI)
- **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, vol.24, pp.51-58, 2016 (Journal Indexed in SCI)
- **Intrauterine Imaging Strategies for Bone Disease**
Alanay Y.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.13, 2016 (Journal Indexed in SCI)
- **Hoyeraal-Hreidarsson Syndrome: An Extremely Rare Dyskeratosis Congenita Phenotype**
Bakar O., Isik U., Canpolat C., ALANAY Y.
PEDIATRIC DERMATOLOGY, vol.32, 2015 (Journal Indexed in SCI)
- **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., Elcioglu N., Tayfun G. A. , Atik S. S. , et al.
ORPHANET JOURNAL OF RARE DISEASES, vol.10, 2015 (Journal Indexed in SCI)
- **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, vol.167, pp.2065-2074, 2015 (Journal Indexed in SCI)
- **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
Boegershausen N., Tsai I., Pohl E., ŞİMŞEK KİPER P. Ö. , Beleggia F., Percin E. F. , Keupp K., Matchan A., Milz E., Alanay Y., et al.
JOURNAL OF CLINICAL INVESTIGATION, vol.125, pp.3585-3599, 2015 (Journal Indexed in SCI)
- **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, vol.167, pp.1501-1509, 2015 (Journal Indexed in SCI)

- **Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffin-Siris and Nicolaiides-Baraitser syndromes**

Bramswig N. C. , Luedecke H., Alanay Y., Albrecht B., Barthelmie A., Boduroglu K., Braunholz D., Caliebe A., Chrzanowska K. H. , Czeschik J. C. , et al.

HUMAN GENETICS, vol.134, pp.553-568, 2015 (Journal Indexed in SCI)

- **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, vol.96, pp.841-849, 2015 (Journal Indexed in SCI)

- **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, vol.167, pp.461-475, 2015 (Journal Indexed in SCI)

- **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, vol.47, pp.73-78, 2015 (Journal Indexed in SCI)

- **A VARIANT CASE OF 6p24 DELETION SYNDROME (OMIM #612582)**

Ergin R. N. , Cigerciogullari E., Alanay Y., Yayla M.

GENETIC COUNSELING, vol.26, pp.237-241, 2015 (Journal Indexed in SCI)

- **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, vol.6, pp.250-253, 2014 (Journal Indexed in SCI)

- **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. , Boduroglu K.

CLINICAL DYSMORPHOLOGY, vol.23, pp.147-151, 2014 (Journal Indexed in SCI)

- **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, vol.56, pp.430-433, 2014 (Journal Indexed in SCI)

- **Etiological yield of SNP microarrays in idiopathic intellectual disability**

ÜTİNE G. E. , Haliloglu G., Volkan-Salanci B., Cetinkaya A., Kiper P. O. , Alanay Y., Aktas D., Anlar B., Topcu M., Boduroglu K., et al.

EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, vol.18, pp.327-337, 2014 (Journal Indexed in SCI)

- **Cathepsin K analysis in a pycnodysostosis cohort: demographic, genotypic and phenotypic features**

ARMAN A., BEREKET A., ÇOKER A., ŞİMŞEK KİPER P. Ö. , GÜRAN T., ÖZKAN B., Atay Z., Akcay T., Haliloglu B., Boduroglu K., et al.

ORPHANET JOURNAL OF RARE DISEASES, vol.9, 2014 (Journal Indexed in SCI)

- **Methylene tetrahydrofolate reductase polymorphisms and homocysteine level in heart defects**

ŞAHİNER Ü. M. , Alanay Y., ALEHAN D., Tuncbilek E., ALİKAŞİFOĞLU M.

PEDIATRICS INTERNATIONAL, vol.56, pp.167-172, 2014 (Journal Indexed in SCI)

- **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, vol.94, pp.405-414, 2014 (Journal Indexed in SCI)

- **Report of a Patient With Temple-Baraitser Syndrome**

YEŞİL G., Guler S., Yuksel A., ALANAY Y.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.164, pp.848-851, 2014 (Journal Indexed in SCI)

- **Celiac disease in Williams-Beuren syndrome**

ŞİMŞEK KİPER P. Ö. , Sahin Y., Arslan U., Alanay Y., Boduroglu K., ORHAN D., ÖZEN H., ÜTİNE G. E.

TURKISH JOURNAL OF PEDIATRICS, vol.56, pp.154-159, 2014 (Journal Indexed in SCI)

- **TMCO1 Deficiency Causes Autosomal Recessive Cerebrofaciothoracic Dysplasia**

Alanay Y., Erguner B., Utine E., Hacariz O., ŞİMŞEK KİPER P. Ö. , Taskiran E. Z. , Percin F., Uz E., Sagiroglu M. S. , Yuksel B., et al.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.164, pp.291-304, 2014 (Journal Indexed in SCI)

- **Striking Hematological Abnormalities in Patients With Microcephalic Osteodysplastic Primordial Dwarfism Type II (MOPD II): A Potential Role of Pericentrin in Hematopoiesis**

ÜNAL Ş., Alanay Y., Cetin M., Boduroglu K., Utine E., Cormier-Daire V., Huber C., ÖZSÜREKÇİ Y., KILIÇ E., Kiper O. P. S. , et al.

PEDIATRIC BLOOD & CANCER, vol.61, pp.302-305, 2014 (Journal Indexed in SCI)

- **Neurochemical Evaluation of Brain Function With H-1 Magnetic Resonance Spectroscopy in Patients With Fragile X Syndrome**

ÜTİNE G. E. , Akpınar B., ARSLAN U. E. , Kiper P. O. S. , Volkan-Salanci B., Alanay Y., Aktas D., Haliloglu G., Oguz K. K. , Boduroglu K., et al.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.164, pp.99-105, 2014 (Journal Indexed in SCI)

- **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., Boduroglu K., Tuncbilek E.

GENETIC COUNSELING, vol.25, pp.53-62, 2014 (Journal Indexed in SCI)

- **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., Boduroglu K.

TURKISH JOURNAL OF PEDIATRICS, vol.56, pp.80-84, 2014 (Journal Indexed in SCI)

- **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, vol.22, pp.5121-5135, 2013 (Journal Indexed in SCI)

- **Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused by PAPSS2 Mutations**

İlida A., ŞİMŞEK KİPER P. Ö. , Mizumoto S., Hoshino T., Elcioglu N., Horemuzova E., Geiberger S., YEŞİL G., Kayserili H., ÜTİNE G. E. , et al.

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- **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, vol.161, pp.2570-2575, 2013 (Journal Indexed in SCI)

- **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.

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- **A Homozygous Deletion in GRID2 Causes a Human Phenotype With Cerebellar Ataxia and Atrophy**

ÜTİNE G. E. , Haliloglu G., Salanci B., Cetinkaya A., Kiper P. O. , Alanay Y., Aktas D., Boduroglu K., ALİKAŞİFOĞLU M.

JOURNAL OF CHILD NEUROLOGY, vol.28, pp.926-932, 2013 (Journal Indexed in SCI)

- **STRIKING HEMATOLOGICAL ABNORMALITIES IN PATIENTS WITH MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II MAY INDICATE A POTENTIAL ROLE OF PERICENTRIN GENE IN HEMATOPOIESIS**

ÜNAL S., ÇETİN M., Alanay Y., Boduroglu K., Utine E., KILIÇ E., ÖZSÜREKÇİ Y., GÜMRÜK F.

HAEMATOLOGICA, vol.98, pp.578, 2013 (Journal Indexed in SCI)

- **Microdeletions at 1q21.1 and 2q24.2 in a Patient with Developmental Delay and Dysmorphic Features**
 ÜTİNE G. E. , Kiper P. O. , Alanay Y., Boduroglu K.
 CHROMOSOME RESEARCH, vol.21, 2013 (Journal Indexed in SCI)
- **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.
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- **Clinical and molecular analysis of RASopathies in a group of Turkish patients**
 Simsek-Kiper P. O. , Alanay Y., Gulhan B., Lissewski C., Turkyilmaz D., Alehan D., Cetin M., Utine G. E. , Zenker M., Boduroglu K.
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- **Laboratory Genetic Testing in Clinical Practice**
 Cogulu O., ALANAY Y., Toruner G. A.
 BIOMED RESEARCH INTERNATIONAL, 2013 (Journal Indexed in SCI)
- **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, vol.47, pp.319-321, 2012 (Journal Indexed in SCI)
- **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, vol.131, pp.1761-1773, 2012 (Journal Indexed in SCI)
- **MRI findings of intracranial malformations in a case with Fraser syndrome**
 Yesilkaya Y., Hizal M., Oguz K. K. , Alanay Y.
 CLINICAL DYSMORPHOLOGY, vol.21, pp.234-236, 2012 (Journal Indexed in SCI)
- **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 Ş., Haliloglu G., Oguz K. K. , Cetin M., Boduroglu K., ALANAY Y.
 EUROPEAN JOURNAL OF PEDIATRICS, vol.171, pp.1567-1571, 2012 (Journal Indexed in SCI)
- **IMPAD1 Mutations in Two Catel-Manzke Like Patients**
 Nizon M., Alanay Y., Tuysuz B., ŞİMŞEK KİPER P. Ö. , Genevieve D., Sillence D., Huber C., Munnich A., Cormier-Daire V.
 AMERICAN JOURNAL OF MEDICAL GENETICS PART A, pp.2183-2187, 2012 (Journal Indexed in SCI)
- **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.
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- **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.
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19. Ulusal çocuk nörolojisi kongresi, Turkey, 19 - 23 April 2017
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Activities in Scientific Journals

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

Clinical Dysmorphology, Committee Member, 2010 - Continues

Memberships / Tasks in Scientific Organizations

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

Tasks In Event Organizations

Alanay Y., European Society of Human Genetics Meeting, Scientific Congress, Berlin, Germany, Haziran 2020

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Invited Talks

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