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Education Information

Doctorate, Hacettepe University, Çocuk Sağlığı Enstitüsü, Genetik, Turkey 1990 - 1996

Undergraduate, Ankara University, Tıp Fakültesi, Turkey 1977 - 1984

Foreign Languages

English, B2 Upper Intermediate

Dissertations

Doctorate, Amnion hücre izolasyonu ve interfaz floresan in situ hibridizasyon tekniğinin uygulanması, Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 1996

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Medical Genetics, Life Sciences, Bioinformatics, Biological Information, Biotechnology, Industrial Biotechnology, Molecular Biology and Genetics, Genetic Disorders, Genomics, Cytogenetic, Natural Sciences

Academic Titles / Tasks

Professor, Hacettepe University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 2006 - Continues

Academic and Administrative Experience

Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 2007 - Continues

Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Ex vivo disease modelling of Rett syndrome: the transcriptomic and metabolomic implications of direct neuronal conversion**
KARAOSMANOĞLU B., İMREN G., Ozisin M. S., REÇBER T., ŞİMŞEK KİPER P. Ö., Haliloglu G., ALİKAŞİFOĞLU M., NEMUTLU E., TAŞKIRAN Z. E., ÜTİNE G. E.
MOLECULAR BIOLOGY REPORTS, no.1, 2024 (SCI-Expanded)
- II. **A novel variant in severe disease of DADA2: involving vasculitic and haematologic features**
AYAN G., Yagiz B., ÇINAR O. E., ÇAĞDAŞ AYVAZ D. N., ÖZBEK D. A., TUNCER A., Oğuz K. K., ÖZEN S., ALİKAŞİFOĞLU M., KARADAĞ Ö.
SCANDINAVIAN JOURNAL OF RHEUMATOLOGY, vol.52, no.1, pp.93-95, 2023 (SCI-Expanded)
- III. **FXR1-related congenital myopathy: expansion of the clinical and genetic spectrum**
Mroczek M., Longman C., Farrugia M. E., Garcia S. K., ARDIÇLI D., Topaloglu H., Hernandez-Lain A., ORHAN D., ALİKAŞİFOĞLU M., Duff J., et al.
JOURNAL OF MEDICAL GENETICS, vol.59, no.11, pp.1069-1074, 2022 (SCI-Expanded)
- IV. **Interaction between Dietary Fat Intake and Metabolic Genetic Risk Score on 25-Hydroxyvitamin D Concentrations in a Turkish Adult Population**
IŞGIN ATICI K., Alathari B. E., TURAN DEMİRCİ B., ŞENDUR S. N., LAY İ., Ellahi B., ALİKAŞİFOĞLU M., Erbas T., BÜYÜKTUNCER DEMİREL Z., Vimalaswaran K. S.
NUTRIENTS, vol.14, no.2, 2022 (SCI-Expanded)
- V. **Is conventional treatment still the first choice in pediatric patients with PHEX mutations in an era of monoclonal FGF-23 antibody?**
Alikasifoglu A., Unsal Y., Gonc N., Ozon A., Kandemir N., Alikasifoglu M.
HORMONE RESEARCH IN PAEDIATRICS, vol.94, no.SUPPL 1, pp.210-211, 2021 (SCI-Expanded)
- VI. **Long-term effect of conventional phosphate and calcitriol treatment on metabolic recovery and catch-up growth in children with PHEX mutation**
ALİKAŞİFOĞLU A., ÜNSAL Y., GÖNÇ E. N., ÖZÖN Z. A., KANDEMİR N., ALİKAŞİFOĞLU M.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.12, pp.1573-1584, 2021 (SCI-Expanded)
- VII. **Further expanding the mutational spectrum of brain abnormalities, neurodegeneration, and dysosteosclerosis: A rare disorder with neurologic regression and skeletal features**
KINDİŞ E., ŞİMŞEK KİPER P. Ö., KOŞUKCU C., TAŞKIRAN Z. E., GÖÇMEN R., Utine E., Haliloglu G., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, pp.1888-1896, 2021 (SCI-Expanded)
- VIII. **FTO gene-lifestyle interactions on serum adiponectin concentrations and central obesity in a Turkish population**
IŞGIN ATICI K., Alsulami S., TURAN DEMİRCİ B., Surendran S., ŞENDUR S. N., LAY İ., KARABULUT E., Ellahi B., Lovegrove J. A., ALİKAŞİFOĞLU M., et al.
INTERNATIONAL JOURNAL OF FOOD SCIENCES AND NUTRITION, vol.72, no.3, pp.375-385, 2021 (SCI-Expanded)
- IX. **Genetic disorders with symptoms mimicking rheumatologic diseases: A single-center retrospective study**
KAYA AKCA Ü., ŞİMŞEK KİPER P. Ö., ÜREL DEMİR G., SAĞ E., ATALAY E., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K., BİLGİNER Y., ÖZEN S.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.64, no.4, 2021 (SCI-Expanded)
- X. **A Revisited Diagnosis of Collagen VI Related Muscular Dystrophy in a Patient with a Novel COL6A2 Variant and 21q22.3 Deletion**
ŞİMŞEK KİPER P. Ö., OĞUZ S., ERGEN F. B., ÜTİNE G. E., ALİKAŞİFOĞLU M., Haliloglu G.
NEUROPEDIATRICS, vol.51, no.6, pp.445-449, 2020 (SCI-Expanded)
- XI. **Molecular etiology of isolated congenital cataract using next-generation sequencing: Single center exome sequencing data from Turkey**
Taylan Şekeroğlu H., Karaosmanoğlu B., Taşkıran E. Z., Şimşek Kiper P. Ö., Alikasifoglu M., Boduroğlu O. K., Coşkun T., Ütine G. E.

Molecular Syndromology, vol.11, pp.302-308, 2020 (SCI-Expanded)

- XII. **Poikiloderma with Neutropenia, Clericuzio-Type Accompanied by Loss of Digits Due to Severe Osteomyelitis**
AKDOĞAN N., KINDİŞ E., BOSTAN E., Utine E., ALİKAŞİFOĞLU M., ERSOY EVANS S.
JOURNAL OF CLINICAL IMMUNOLOGY, vol.40, no.6, pp.934-939, 2020 (SCI-Expanded)
- XIII. **Further Phenotypic Delineation of Partial Trisomy 17q and Partial Monosomy 20q due to Rare t(17;20)**
Ürel-Demir G., Akgün-Doğan Ö., OĞUZ S., Güleray-Lafci N., Şimşek-Kiper P. Ö., Eda Utine G., ALİKAŞİFOĞLU M., Boduroğlu K.
MOLECULAR SYNDROMOLOGY, vol.11, no.1, pp.38-42, 2020 (SCI-Expanded)
- XIV. **Intrafamilial variability of XYLT2-related spondyloocular syndrome**
Guleray N., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., Boduroglu K., ALİKAŞİFOĞLU M.
European Journal of Medical Genetics, vol.62, no.11, 2019 (SCI-Expanded)
- XV. **Metabolic Infrastructure of Pregnant Women With Trisomy 21 Fetuses; Metabolomic Analysis**
NEMUTLU E., ÖRGÜL G., REÇBER T., AYDIN E., ÖZKAN E., TURÇAL M., ALİKAŞİFOĞLU M., KIR S., BEKSAÇ M. S.
ZEITSCHRIFT FÜR GEBURTSCHILFE UND NEONATOLOGIE, vol.223, no.5, pp.297-303, 2019 (SCI-Expanded)
- XVI. **A case of Woodhouse-Sakati syndrome with pituitary iron deposition, cardiac and intestinal anomalies, with a novel mutation in DCAF17**
ŞENDUR S. N., OĞUZ S., ÜTİNE G. E., DAĞDELEN S., Oguz K. K., Erbas T., ALİKAŞİFOĞLU M.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.8, 2019 (SCI-Expanded)
- XVII. **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**
Simsek-Kiper P. O., Taskiran E., KOŞUKCU C., ARSLAN U. E., Cormier-Daire V., Gonc N., Ozon A., ALİKAŞİFOĞLU A., KANDEMİR N., ÜTİNE G. E., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.7, pp.1157-1172, 2019 (SCI-Expanded)
- XVIII. **An eight-case 1q21 region series: novel aberrations and clinical variability with new features**
CEYLAN A., ŞAHİN İ. F., Erdem H., Kayhan G., Simsek-Kiper P., ÜTİNE G. E., Percin F., Boduroglu K., ALİKAŞİFOĞLU M.
Journal of Intellectual Disability Research, vol.63, no.6, pp.548-557, 2019 (SSCI)
- XIX. **Evaluation of Pregnancies in 25 Families with Balanced/Unbalanced Chromosomal Translocations**
GÜLERAY N., Yucesoy H. M., FADİLOĞLU E., TANAÇAN A., ALİKAŞİFOĞLU M., BEKSAÇ M. S.
INTERNATIONAL JOURNAL OF HUMAN GENETICS, vol.19, no.1, pp.22-28, 2019 (SCI-Expanded)
- XX. **Diagnostic Pathway to Nonsense Mutation Dystrophinopathy: A Tertiary-Center, Retrospective Experience**
ARDIÇLI D., Haliloglu G., ALİKAŞİFOĞLU M., Topaloglu H.
NEUROPEDIATRICS, vol.50, no.1, pp.41-45, 2019 (SCI-Expanded)
- XXI. **A novel NKX3-2 mutation associated with perinatal lethal phenotype of spondylo-megaepiphyseal-metaphyseal dysplasia in a neonate**
Simsek-Kiper P. O., KOŞUKCU C., Akgun-Dogan O., GÖÇMEN R., ÜTİNE G. E., SOYER T., Korkmaz-Toygar A., Nishimura G., ALİKAŞİFOĞLU M., Boduroglu K.
European Journal of Medical Genetics, vol.62, no.1, pp.21-26, 2019 (SCI-Expanded)
- XXII. **Impact of Fat Mass and Obesity Associated (FTO) Gene Variants and Lifestyle Factors on Obesity Traits in A Turkish Population**
Alsulami S., Isgin-Atici K., Turan-Demirci B., Surendran S., ŞENDUR S. N., LAY İ., KARABULUT E., Ellahi B., Lovegrove L., ALİKAŞİFOĞLU M., et al.
PROCEEDINGS OF THE NUTRITION SOCIETY, vol.78, no.OCE2, 2019 (SCI-Expanded)
- XXIII. **Genetic testing for DADA2: How can we avoid missing patients?**
SÖNMEZ H. E., BATU AKAL E. D., Taskiran E. Z., ALİKAŞİFOĞLU M., BİLGİNER Y., ÖZEN S.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.26, no.11, pp.1563-1564, 2018 (SCI-Expanded)
- XXIV. **Prenatal and Postnatal Follow-up in Trisomies 13 and 18: A 20-Year Experience in a Tertiary Center**
Dogan O. A., Demir G. U., Arslan U. E., Simsek-Kiper P. O., ÜTİNE G. E., ALİKAŞİFOĞLU M., Boduroglu K.

- American Journal of Perinatology, vol.35, no.5, pp.427-433, 2018 (SCI-Expanded)
- XXV. **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., AKTAŞ D., Boduroglu K., Tuncbilek E., ALİKAŞİFOĞLU M.
European Journal of Obstetrics and Gynecology and Reproductive Biology, vol.221, pp.76-80, 2018 (SCI-Expanded)
- XXVI. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Costello Syndrome**
VURALI KARAOĞLAN D., KOŞUKCU C., Taskiran E., Simsek P. O., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU A., ALİKAŞİFOĞLU M.
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.363, 2018 (SCI-Expanded)
- XXVII. **Clinical, demographic and nosologic characterisation of the genetic disorders of the skeleton in Turkey: The Skeletal Dysplasia registry**
ŞİMŞEK KİPER P. Ö., Utine G. E., TAŞKIRAN Z. E., KOŞUKCU C., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
European Journal Of Human Genetics, 2018 (SCI-Expanded)
- XXVIII. **Prospective Turkish Cohort Study to Investigate the Frequency of Niemann-Pick Disease Type C Mutations in Consanguineous Families with at Least One Homozygous Family Member**
TOPÇU M., Aktas D., ÖZTOPRAK M., Mungan N. O., YÜCE A., ALİKAŞİFOĞLU M.
MOLECULAR DIAGNOSIS & THERAPY, vol.21, no.6, pp.643-651, 2017 (SCI-Expanded)
- XXIX. **Neonatal-Onset Recurrent Guillain-Barre Syndrome-Like Disease: Clues for Inherited CD59 Deficiency**
Ardicli D., TASKIRAN E. Z., KOŞUKCU C., Temucin Ç. M., OGUZ K. K., HALILOGLU G., ALİKAŞİFOĞLU M., TOPALOĞLU H.
NEUROPEDIATRICS, vol.48, no.6, pp.477-481, 2017 (SCI-Expanded)
- XXX. **Coexistence of Trisomy 13 and SRY (-) XX Ovotesticular Disorder of Sex Development**
Demir G. U., Dogan O. A., Kiper P. O. S., ÜTİNE G. E., Boduroglu K., Gucer S., ALİKAŞİFOĞLU M.
Fetal and Pediatric Pathology, vol.36, no.6, pp.445-451, 2017 (SCI-Expanded)
- XXXI. **Partial Trisomy 18 From a marker chromosome to a rare chromosomal disorder**
OĞUZ S., GÜLERAY N., DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
MOLECULAR CYTOGENETICS, vol.10, 2017 (SCI-Expanded)
- XXXII. **HERC1 mutations in idiopathic intellectual disability**
ÜTİNE G. E., Taskiran E. Z., KOŞUKCU C., KARAOĞLAN B., GÜLERAY N., Dogan O. A., Kiper P. O. S., Boduroglu K., ALİKAŞİFOĞLU M.
European Journal of Medical Genetics, vol.60, no.5, pp.279-283, 2017 (SCI-Expanded)
- XXXIII. **Unusual presentations of Carney Complex in patient with a novel PRKAR1A mutation**
AKİN S., NOYAN S., DAĞDELEN S., Pasaoglu I., KAYNAROĞLU V., MUT AŞKUN M., Bilen C. Y., Kiratli H., BAYDAR D. E., ÖNDER S., et al.
Neuroendocrinology Letters, vol.38, pp.248-254, 2017 (SCI-Expanded)
- XXXIV. **Whole Exome Sequencing in Early Onset Systemic Lupus Erythematosus**
BATU AKAL E. D., KOŞUKCU C., Taskiran E. Z., AKMAN S., Ozturk K., SÖZERİ B., Unsal E., Ekinci Z., BİLGİNER Y., ALİKAŞİFOĞLU M., et al.
ARTHRITIS & RHEUMATOLOGY, vol.68, 2016 (SCI-Expanded)
- XXXV. **A novel missense mutation of the GRK1 gene in Oguchi disease**
Teke M. Y., Citirik M., Kabacam S., Demircan S., ALİKAŞİFOĞLU M.
MOLECULAR MEDICINE REPORTS, vol.14, no.4, pp.3129-3133, 2016 (SCI-Expanded)
- XXXVI. **Genetic Variations in Attention Deficit Hyperactivity Disorder Subtypes and Treatment Resistant Cases**
Ünal D., Ünal M. F., Alikasıfoğlu M., Cetinkaya A.
PSYCHIATRY INVESTIGATION, vol.13, no.4, pp.427-433, 2016 (SCI-Expanded)
- XXXVII. **Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease**

Karaca E., Harel T., Pehlivan D., Jhangiani S. N., Gambin T., Akdemir Z. C., Gonzaga-Jauregui C., Erdin S., Bayram Y., Campbell I. M., et al
NEURON, vol.88, no.3, pp.499-513, 2015 (SCI-Expanded)

- XXXVIII. **A Case Series of Adenosine Deaminase 2-deficient Patients Emphasizing Treatment and Genotype-phenotype Correlations**
Batu E. D., KARADAĞ Ö., TASKIRAN E. Z., KALYONCU U., AKSENTIJEVICH I., ALİKAŞİFOĞLU M., ÖZEN S.
JOURNAL OF RHEUMATOLOGY, vol.42, no.8, pp.1532-1534, 2015 (SCI-Expanded)
- XXXIX. **Two patients with microdeletion and microduplication involving 1q21.1**
CEYLAN A. C., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
CHROMOSOME RESEARCH, vol.23, 2015 (SCI-Expanded)
- XL. **Systems-level analysis of genome wide association study results for a pilot juvenile idiopathic arthritis family study**
Aydin-Son Y., Batu E. D., DEMIRKAYA E., BİLGİNER Y., Kasapcopur O., Unsal E., ALİKAŞİFOĞLU M., ÖZEN S.
TURKISH JOURNAL OF PEDIATRICS, vol.57, no.4, pp.324-333, 2015 (SCI-Expanded)
- XLI. **Jervell and Lange-Nielsen syndrome with homozygous missense mutation of the KCNQ1 gene**
KILIÇ E., ERTUĞRUL İ., Ozer S., ALİKAŞİFOĞLU M., Aktas D., Boduroglu K., ÜTİNE G. E.
TURKISH JOURNAL OF PEDIATRICS, vol.56, no.5, pp.542-545, 2014 (SCI-Expanded)
- XLII. **Etiological yield of SNP microarrays in idiopathic intellectual disability**
Ütine G. E., Haliloglu G., Volkan-Salanci B., Cetinkaya A., Kiper P. O., Alanay Y., Aktas D., Anlar B., Topçu M., Boduroglu K., et al.
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, vol.18, no.3, pp.327-337, 2014 (SCI-Expanded)
- XLIII. **Methylene tetrahydrofolate reductase polymorphisms and homocysteine level in heart defects**
ŞAHİNER Ü. M., ALANAY Y., ALEHAN D., TUNÇBİLEK E., ALİKAŞİFOĞLU M.
PEDIATRICS INTERNATIONAL, vol.56, no.2, pp.167-172, 2014 (SCI-Expanded)
- XLIV. **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, no.1, pp.53-62, 2014 (SCI-Expanded)
- XLV. **Assessment of Whole-Brain White Matter by DTI in Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay**
Oguz K. K., Haliloglu G., TEMUÇİN Ç. M., GÖÇMEN R., Has A. C., Doerschner K., Dolgun A., ALİKAŞİFOĞLU M.
AMERICAN JOURNAL OF NEURORADIOLOGY, vol.34, no.10, pp.1952-1957, 2013 (SCI-Expanded)
- XLVI. **A Homozygous Deletion in GRID2 Causes a Human Phenotype With Cerebellar Ataxia and Atrophy**
Ütine G. E., Haliloglu G., Volkan Salanci B., Çetinkaya A., Şimşek Kiper P. Ö., Alanay Y., Aktas D., Boduroğlu O. K., Alikasıfoğlu M.
JOURNAL OF CHILD NEUROLOGY, vol.28, no.7, pp.926-932, 2013 (SCI-Expanded)
- XLVII. **Two Siblings With Similar Phenotypes: One of Them Had Ring 20 Chromosome**
Tezer F. I., Aktas D., ALİKAŞİFOĞLU M., SAYGI S.
CLINICAL EEG AND NEUROSCIENCE, vol.44, no.1, pp.58-61, 2013 (SCI-Expanded)
- XLVIII. **Association of LOXL1 gene polymorphisms with exfoliation syndrome/glaucoma and primary open angle glaucoma in a Turkish population**
Kasim B., Irkeç M., ALİKAŞİFOĞLU M., Orhan M., MOCAN M. C., Aktas D.
MOLECULAR VISION, vol.19, pp.114-120, 2013 (SCI-Expanded)
- XLIX. **The relationship between changes in functional cardiac parameters following anthracycline therapy and carbonyl reductase 3 and glutathione S transferase Pi polymorphisms**
VOLKAN-SALANCI B., AKSOY H., Kiratli P. O., TULUMEN E., GULER N., OKSUZOĞLU B., TOKGOZOĞLU L., Erbas B., ALİKAŞİFOĞLU M.
JOURNAL OF CHEMOTHERAPY, vol.24, no.5, pp.285-291, 2012 (SCI-Expanded)
- L. **Serum PON-1 Activity but not Q192R Polymorphism is Related to the Extent of Atherosclerosis**
Bayrak A., Bayrak T., TOKGÖZOĞLU S. L., VOLKAN SALANCI B., Deniz A., YAVUZ B., ALİKAŞİFOĞLU M., Demirpençe E.

JOURNAL OF ATHEROSCLEROSIS AND THROMBOSIS, vol.19, no.4, pp.376-384, 2012 (SCI-Expanded)

- LI. **Relationship of PON2 gene Ser311Cys polymorphism and serum paraoxonase activity with coronary artery disease in Turkish population**
Bayrak T., Bayrak A., VOLKAN SALANCI B., Deniz A., TOKGÖZOĞLU S. L., YAVUZ B., ALİKAŞİFOĞLU M., Demirpence E.
TURKISH JOURNAL OF BIOCHEMISTRY-TURK BIYOKIMYA DERGISI, vol.37, no.2, pp.150-155, 2012 (SCI-Expanded)
- LII. **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, vol.53, no.5, pp.558-560, 2011 (SCI-Expanded)
- LIII. **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, vol.19, 2011 (SCI-Expanded)
- LIV. **CYP1A1 polymorphism in adolescents with polycystic ovary syndrome**
AKGÜL S., DERMAN O., ALİKAŞİFOĞLU M., Aktas D.
INTERNATIONAL JOURNAL OF GYNECOLOGY & OBSTETRICS, vol.112, no.1, pp.8-10, 2011 (SCI-Expanded)
- LV. **Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta**
Alanay Y., Avaygan H., Camacho N., ÜTİNE G. E., Boduroglu K., Aktas D., ALİKAŞİFOĞLU M., Tuncbilek E., ORHAN D., Bakar F. T., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.86, no.4, pp.551-559, 2010 (SCI-Expanded)
- LVI. **THE RELATION BETWEEN FUNCTIONAL CARDIAC PARAMETERS AND SINGLE NUCLEOTIDE POLYMORPHISMS IN GLUTATHIONE S TRANSFERASE P1 AND CARBONYL REDUCTASE3 GENES**
VOLKAN SALANCI B., Tulumen E., Aksoy H., OKUTUCU S., Kiratli P. O., Oksuzoglu B., Guler N., Tokgozoglu L., Erbas B., ALİKAŞİFOĞLU M., et al.
INTERNATIONAL JOURNAL OF CARDIOLOGY, vol.140, 2010 (SCI-Expanded)
- LVII. **Derivative chromosome 1 and GLUT1 deficiency syndrome in a sibling pair**
Aktas D., Utine E. G., Mrasek K., Weise A., von Eggeling F., Yalaz K., Posorski N., Akarsu N., ALİKAŞİFOĞLU M., Liehr T., et al.
MOLECULAR CYTOGENETICS, vol.3, 2010 (SCI-Expanded)
- LVIII. **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, vol.53, no.2, pp.208-210, 2009 (SCI-Expanded)
- LIX. **Partial Distal Aphyalangia, 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, no.6, pp.1317-1318, 2009 (SCI-Expanded)
- LX. **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, vol.51, no.3, pp.199-206, 2009 (SCI-Expanded)
- LXI. **Impact of renin-angiotensin system polymorphisms on renal haemodynamic responsiveness to acute angiotensin-converting enzyme inhibition in type 2 diabetes mellitus**
VOLKAN-SALANCI B., DAĞDELEN S., ALİKAŞİFOĞLU M., ERBAS T., HAYRAN M., Erbas B.
JOURNAL OF THE RENIN-ANGIOTENSIN-ALDOSTERONE SYSTEM, vol.10, no.1, pp.41-50, 2009 (SCI-Expanded)
- LXII. **A new autosomal dominant Peters' anomaly phenotype expanding the anterior segment dysgenesis spectrum**
Berker N., Alanay Y., Elgin U., Volkan-Salanci B., Simsek T., Akarsu N., ALİKAŞİFOĞLU M.
ACTA OPHTHALMOLOGICA, vol.87, no.1, pp.52-57, 2009 (SCI-Expanded)
- LXIII. **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, vol.51, no.4, pp.343-350, 2008 (SCI-Expanded)

LXIV. Association of assisted reproductive technology with twinning and congenital anomalies

Balci S., Engiz O., ALİKAŞİFOĞLU M., ESİNLER İ., BEKSAÇ M. S.

INDIAN JOURNAL OF PEDIATRICS, vol.75, no.6, pp.638-640, 2008 (SCI-Expanded)

LXV. CYP1A1 gene polymorphism and polycystic ovary syndrome

ESİNLER İ., Aktas D., Otegen U., ALİKAŞİFOĞLU M., Yarali H., TUNÇBİLEK E.

REPRODUCTIVE BIOMEDICINE ONLINE, vol.16, no.3, pp.356-360, 2008 (SCI-Expanded)

LXVI. Factor V Leiden mutation and type 1 diabetes mellitus

Demirer A. N., ALİKAŞİFOĞLU M., TUNÇBİLEK E., Karakus S., Erbas T.

BLOOD COAGULATION & FIBRINOLYSIS, vol.19, no.1, pp.70-74, 2008 (SCI-Expanded)

Articles Published in Other Journals

I. Relationship between the Common Variants of the ADAM19, FAM13A, and IREB2 Genes and COPD Susceptibility and Severity

Senel M. Y., Kabacam S., KAŞIKCI ÇAVDAR M., Onder B. S., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M.

INDIAN JOURNAL OF RESPIRATORY CARE, no.2, pp.83-90, 2024 (ESCI)

II. Reduced irisin levels in patients with acromegaly

Şendur S. N., Baykal G., Firlatan B., Aydin B., Lay İ., Dağdelen S., Alikasıfoğlu M., Erbas T.

HORMONE MOLECULAR BIOLOGY AND CLINICAL INVESTIGATION, vol.43, no.3, pp.251-261, 2022 (ESCI)

III. Gorlin Syndrome in Eleven Patients

ÜTİNE G. E., ALANAY Y., Aktas D., BODUROĞLU O. K., ALİKAŞİFOĞLU M., TUNÇBİLEK E.

JOURNAL OF PEDIATRIC RESEARCH, vol.4, no.2, pp.63-67, 2017 (ESCI)

IV. A diagnosis to consider in an adult patient with facial features and intellectual disability: Williams syndrome

Dogan O. A., Kiper P. O. S., ÜTİNE G. E., ALİKAŞİFOĞLU M., Boduroglu K.

Korean Journal of Family Medicine, vol.38, no.2, pp.102-105, 2017 (Scopus)

V. A Baseline Algorithm for Molecular Diagnosis of Genetic Eye Diseases: Ophthalmologist's Perspective

TAYLAN ŞEKEROĞLU H., ÜTİNE G. E., ALİKAŞİFOĞLU M.

TURK OFTALMOLOJİ DERGİSİ-TURKISH JOURNAL OF OPHTHALMOLOGY, vol.46, no.6, pp.299-300, 2016 (ESCI)

VI. An Adult Patient with Monosomy 18p Growth Hormone Deficiency and Selective IgA Deficiency

ZENGİN AKKUŞ P., çetinkaya a., ÇAĞDAŞ AYVAZ D. N., ALİKAŞİFOĞLU M., ALİKAŞİFOĞLU A., KANDEMİR N., TEZCAN F. İ., utine G. E., BODUROĞLU O. K.

Journal of Genetic Syndromes & Gene Therapy, vol.7, no.2, 2016 (Peer-Reviewed Journal)

Books & Book Chapters

I. Tıbbi Genetik Bütünleşik Yaklaşım. (ed). Çeviri Editörleri:, 2019, Hipokrat Yayıncılık, Ankara.

ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M.

Hipokrat Yayıncılık, Ankara, 2019

Refereed Congress / Symposium Publications in Proceedings

I. Sendromik olmayan zihinsel yetersizlikte tüm ekzom dizilemenin tanısal verimi

TAŞKIRAN Z. E., KARAOSMANOĞLU B., KOŞUKCU C., ÜREL DEMİR G., akgün doğan ö., Kiper şimşek P. Ö., ALİKAŞİFOĞLU M., BODUROĞLU O. K., Utine G. E.

16. Tıbbi Biyoloji ve Genetik Kongresi, Turkey, 25 - 27 October 2019

- II. **Çocuklarda romatizmal bulgularla gelen genetik hastalıklar**
KAYA AKCA Ü., Bilginer Y., Özen S., Sag E., UTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M., ATALAY E., ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö.
20. ULUSAL ROMATOLOJİ KONGRESİ, Turkey, 16 - 20 October 2019
- III. **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, Norway, 11 - 14 September 2019
- IV. **SECONDARY VASCULITIS DUE TO SINGLE GENE DEFECTS**
ÖZEN S., BİLGİNER Y., Batu E., KARADAĞ Ö., ALİKAŞİFOĞLU M.
19th International Vasculitis and ANCA Workshop, Pennsylvania, United States Of America, 7 - 10 April 2019, vol.58
- V. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Costello Syndrome**
VURALI KARAOĞLAN D., KOŞUKCU C., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., UTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU A., ALİKAŞİFOĞLU M.
57th Annual ESPE European Society for Paediatric Endocrinology, Atina, Greece, 27 - 29 September 2018, vol.90, pp.351-352
- VI. **IGF1 Receptor Deletion is a Rare Cause of Prenatal Onset Short Stature.**
GÖNÇ E. N., ÖZÖN Z. A., Kabaçam S., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU A., KANDEMİR N., ALİKAŞİFOĞLU M.
International Congress of the Growth Hormone Research IGF Societies, Sep 14 - 17, 2018, Seattle, Washington, United States of America, 14 - 17 September 2018
- VII. **Expanding the clinical and mutational spectrum of Roberts syndrome with previously unreported endocrine findings.**
GÜLERAY N., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
European Human Genetics Conference, 16 - 19 June 2018
- VIII. **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 June 2018
- IX. **Clinical, demographic and nosologic characterization of the genetic disorders of the skeleton in Turkey: The skeletal dysplasia registry.**
ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., TAŞKIRAN Z. E., KOŞUKCU C., ARSLAN U. E., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
European Human Genetics Conference, Milan, Italy, June 16-19, 2018., Milan, Italy, 16 - 19 June 2018
- X. **Metabolic Infrastructure of Pregnant Women with Down Syndrome**
NEMUTLU E., ÖRGÜL G., Recber T., Aydın E., Özkan E., Turğal M., ALİKAŞİFOĞLU M., KIR S., BEKSAÇ M. S.
Seventh Congress of the South East European Society of Perinatal Medicine, 10 May 2018 - 12 January 2019
- XI. **Oküloaurikülovertebral spektrumda 5p delesyonu**
ÜREL DEMİR G., OĞUZ S., GÜLERAY N., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017
- XII. **Woodhouse-Sakati sendromunda iki yeni mutasyon**
OĞUZ S., AKGÜN DOĞAN Ö., ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
3.Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017
- XIII. **6p25.3 delesyonu**
AKGÜN DOĞAN Ö., ÜREL DEMİR G., GÜLERAY N., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., KUTLUK M. T., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
3.Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017
- XIV. **PGAP3 geninde yeni tanımlanmış mutasyona bağlı hiperfosfatazya mental retardasyon sendromu**
AKGÜN DOĞAN Ö., ÜREL DEMİR G., KOŞUKCU C., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU

M., BODUROĞLU O. K.

3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

- XV. **Taşıyıcı kız kardeşlerin oğullarında zihinsel yetersizlik**
ÜREL DEMİR G., OĞUZ S., GÜLERAY N., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017
- XVI. **Camurati-Engelmann hastalığı**
OĞUZ S., ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017
- XVII. **Juvenil Paget Hastalığı**
OĞUZ S., ÜREL DEMİR G., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017
- XVIII. **Further delineation of spondyloepimetaphyseal dysplasia Faden-Alkuraya type: a RSPRY1-associated spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis.**
ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., KOŞUKCU C., AKGÜN DOĞAN Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
International Skeletal Dysplasia Society Meeting, 20 - 23 September 2017
- XIX. **SOX9 gene duplication-related 46,XX ovotesticular disorder of sex development**
ÖZÖN Z. A., ALİKAŞİFOĞLU A., GÖNÇ E. N., VURALI KARAOĞLAN D., BÜYÜKYILMAZ G., ŞİMŞEK KİPER P. Ö., ÜTİNE E., ORHAN D., SOYER T., BODUROĞLU O. K., et al.
10th International Meeting of Pediatric Endocrinology, 14 - 17 September 2017
- XX. **ICF SYNDROME: CLINICAL, IMMUNOLOGICAL AND CYTOGENETIC ANALYSIS OF SEVEN CASES**
AKARSU A., ÇAĞDAŞ AYVAZ D. N., METİN A., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K., SANAL Ö., TEZCAN F. İ.
ESID 2017, 11 - 14 September 2017
- XXI. **Partial Trisomy 18 From a marker chromosome to a rare chromosomal disorder**
OĞUZ S., GÜLERAY N., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
European Cytogenetics Conference, 1 - 04 July 2017
- XXII. **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, Cyprus (Kktc), 29 - 30 June 2017
- XXIII. **Association between Klotho gene polymorphisms, circulating Klotho and FGF23 levels and echocardiographic parameters among patients with acromegaly.**
NAFİYE H., HEKİMSOY V., KABACAM S., LAY İ., ŞENER Y. Z., DAĞDELEN S., MUT AŞKUN M., EVRANOS B., YORGUN H., KABAKCI M. G., et al.
ENDO 2017, The Endocrine Society's 99th Annual Meeting and Expo., Orlando, United States Of America, 1 - 04 April 2017, vol.38
- XXIV. **Duplication in a patient presenting with SRY negative 46XX disorders of sex development**
ŞİMŞEK KİPER P. Ö., ÖZÖN Z. A., GÖNÇ E. N., ALİKAŞİFOĞLU A., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
50th The European Society of Human Genetics, 27 - 29 May 2017
- XXV. **Homozygous novel variant in MUT in a patient with intellectual disability without metabolic derangement**
ÜTİNE G. E., TAŞKIRAN Z. E., KOŞUKCU C., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
European Society of Human Genetics Conference 2017, Kopenhagen, Denmark, 27 - 30 May 2017
- XXVI. **RSPRY1 associated skeletal dysplasia: Spondylo-epi-metaphyseal dysplasia with conobrachydactyly and craniosynostosis**
ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., KOŞUKCU C., AKGÜN DOĞAN Ö., YILMAZ G., ÜTİNE G. E., NISHIMURA G.,

BODUROĞLU O. K., ALİKAŞİFOĞLU M.

European Society of Human Genetics Conference 2017, Kopenhag, Denmark, 27 - 30 May 2017

XXVII. **Whole Exoma Sequencing in early onset systemic lupus erythematosus**

BATU E. D., kosukcu c., TAŞKIRAN Z. E., AKMAN S., Unsal e., ekinci z., BİLGİNER Y., ALİKAŞİFOĞLU M., ÖZEN S.
2016 ACR/ARHP Annual Meeting, 6 - 11 November 2016

XXVIII. **Analysis of chromosome 22q11.2 copy number variations by multiplex ligation dependent probe amplification**

ALARCON MARTÍNEZ T., KABAÇAM S., CEYLAN A. C., ŞİMŞEK KİPER P. Ö., ALİKAŞİFOĞLU M., BODUROĞLU O. K., ÜTİNE G. E.

American Society of Human Genetics 66th Meeting, 18 - 22 October 2016

XXIX. **KLOTHO GENE POLYMORPHISM A NOVEL PREDICTIVE MARKER FOR CANCER IN ACROMEGALIC PATIENTS**

HELVACI N., kabaçam s., ŞENER Y. Z., DAĞDELEN S., MUT AŞKUN M., ALİKAŞİFOĞLU M., ERBAŞ A. T.

European Neuroendocrine Association Congress 2016, Milan, Italy, 19 - 22 October 2016

XXX. **Array CGH'te Saptanan Kopya Sayısı Değişikliklerinin Klinikle ve Kantitatif PCR ile Değerlendirilmesi**

CEYLAN A. C., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.

12. Ulusal Tıbbi Genetik Kongresi, İzmir, Turkey, 5 - 09 October 2016, pp.356

XXXI. **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 August 2016

XXXII. **A novel mutation of the menin gene in a family with multiple endocrine neoplasia type 1**

Şendur S. N., DOĞRUL A. B., Kabaçam S., ABBASOĞLU O., ÖNDER S. Ç., SÖKMENSÜER C., Erbaş B., DAĞDELEN S., ALİKAŞİFOĞLU M., ERBAŞ A. T.

18th European Congress of Endocrinology, 28 - 31 May 2016

XXXIII. **Does IGF2BP2 gene polymorphism have an effect on the development of gestational diabetes mellitus**

Çıkman D. İ., Çıkman M. S., ALİKAŞİFOĞLU M., DAĞDELEN S.

18th European Congress of Endocrinology, 28 - 31 May 2016

XXXIV. **Clinical and quantitative PCR confirmation of copy number variations detected by array CGH**

CEYLAN A. C., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.

European Society of Human Genetics Conference 2016, 21 - 24 May 2016

XXXV. **Dismorfik Bulguları ve Selektif IgA Eksikliği Tanısıyla İzlenen Monozomi 18p Olgusu**

ZENGİN AKKUŞ P., ÇETİNKAYA A., ÇAĞDAŞ AYVAZ D. N., ALİKAŞİFOĞLU M., ALİKAŞİFOĞLU A., KANDEMİR N., TEZCAN F. İ., ÜTİNE G. E., BODUROĞLU O. K.

2. Klinik İmmünoloji Kongresi, Antalya, Turkey, 31 March - 03 April 2016

XXXVI. **Erişkin dönemde tanı alan Williams sendromu vakası**

AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.

2. Ulusal Çocuk Genetik Sempozyumu, 22-24 Ekim 2015, Samsun, Türkiye, Turkey, 22 - 24 October 2015

XXXVII. **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, Turkey, 22 - 24 October 2015

XXXVIII. **A Novel NKX3.2 Mutation Associated With Spondylo Megaepiphyseal Metaphyseal Dysplasia In A Neonate A Rare Clinical Entity**

ŞİMŞEK KİPER P. Ö., YAVUZ Ş., ÜTİNE G. E., SOYER T., KORKMAZ TOYGAR A., ALİKAŞİFOĞLU M., GEN N., BODUROĞLU O. K.

International Skeletal Dysplasia Society 12th Biennial Meeting, 29 July - 01 August 2015

XXXIX. **Roberts SC Phocomelia Syndrome A Rare Clinical Entity**

GÜLERAY N., ŞİMŞEK KİPER P. Ö., DEMİREL M., ÇETİNKAYA A., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.

International Skeletal Dysplasia Society 12th Biennial Meeting, 29 July - 01 August 2015

- XL. Clinical and Molecular Analysis of 3M Syndrome In A Group of Turkish Patients**
ŞİMŞEK KİPER P. Ö., EKİM ZİHNİ T., ÜTİNE G. E., ALİKAŞİFOĞLU A., KANDEMİR N., VALERİE C. D., ALANAY Y., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
International Skeletal Dysplasia Society 12th Biennial Meeting, 29 July - 01 August 2015
- XLI. Experience of a Skeletal Dysplasia Registry In Turkey A Five Years Retrospective Analysis**
KURT ŞÜKÜR E. D., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ERGÜL T., ALİKAŞİFOĞLU M.
International Skeletal Dysplasia Society 12th Biennial Meeting, 29 July - 01 August 2015
- XLII. Two patients with microdeletion and microduplication involving 1q21.1**
CEYLAN A. C., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
10th European Cytogenetics Conference, 4 - 07 July 2015, vol.23, pp.49
- XLIII. Microdeletion and microduplication of 1q21.1 in two separate patients**
CEYLAN A. C., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
10th European Cytogenetics Conference, France, 4 - 07 July 2015
- XLIV. APPLICATION OF A CYTOGENETIC PROGRESSION SCORE (GPS), TUMOR LOCATION, AND GENETIC PROGRESSION IN MENINGIOMAS: HACETTEPE EXPERIENCE**
Mut M., Aktas D., ALİKAŞİFOĞLU M., Ozgen T.
3rd Quadrennial Meeting of the World-Federation-of-Neuro-Oncology/6th Meeting of the Asian-Society-for-Neuro-Oncology, Yokohama, Japan, 11 - 14 May 2009, vol.11, pp.964-965

Supported Projects

- ÜNAL D., ÜTİNE G. E., ÜNAL M. F., KOŞUKCU C., ŞİMŞEK KİPER P. Ö., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, İkiz Otistik Olgularında Genetik Farklılıklar, 2018 - 2021
- ALİKAŞİFOĞLU M., GÜLERAY N., Project Supported by Higher Education Institutions, Okuloaurikulovertebral Spektrum Etyolojisinde Genetik Nedenlerin Araştırılması, 2018 - 2019
- TAŞKIRAN Z. E., ÜTİNE G. E., ŞİMŞEK KİPER P. Ö., BODUROĞLU O. K., AKARSU A. N., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, Hacettepe Ekzom Projesi, 2015 - 2019
- ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., KOŞUKCU C., AKGÜN DOĞAN Ö., YILMAZ G., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, Spondiloepimetafizyeal displazi Faden Alkuraya Tipi İskelet Displazisinin Daha Geniş Detaylandırılması: RSPRY1-ilişkili Spondiloepimetafizyeal displazi, Konobrakidaktili ve Kraniyosinostozis, 2017 - 2017
- ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, RSPRY1 İlişkili İskelet Displazisi: Multipl Epifizyeal Displazi, Konobrakidaktil ve Kraniyosinostozis Birlikteliği, 2017 - 2017
- ALİKAŞİFOĞLU M., ÜTİNE G. E., ŞİMŞEK KİPER P. Ö., Project Supported by Higher Education Institutions, 46,XX SRY Negatif Cinsel Gelişim Bozukluğu Olan Bir Hastada 17q243 Duplikasyonu, 2017 - 2017
- PURALI N., ALİKAŞİFOĞLU M., BABAOĞLU M. Ö., Project Supported by Higher Education Institutions, Hacettepe Üniversitesi ArGe Strateji Belgesi Hazırlanması Moleküler Tıp Alanı Çalıştayı, 2016 - 2017
- ALİKAŞİFOĞLU M., BODUROĞLU O. K., ÜTİNE G. E., Project Supported by Higher Education Institutions, Genetikte Teknolojik İlerlemeler, 2016 - 2016
- ALİKAŞİFOĞLU M., AKARSU A. N., BEKSAÇ M. S., GÜLERAY N., KABAÇAM S., Project Supported by Higher Education Institutions, Maternal Kanda Hücre Dışı Serbest Dolaşan Fetal DNA İzolasyon Yöntemlerinin Karşılaştırılması, 2015 - 2016
- ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, 1q211 Mikrodelesyon Sendromu, 2015 - 2016
- BODUROĞLU O. K., ÜTİNE G. E., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, 1q211 Mikrodelesyon ve Mikroduplikasyon Sendromları, 2015 - 2016
- ALİKAŞİFOĞLU M., ÜTİNE G. E., BODUROĞLU O. K., Project Supported by Higher Education Institutions, Segmental Mikroduplikasyonlar ve Kopya Sayısı Değişiklikleri, 2015 - 2016
- ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, Array CGH'te Saptanan Kopya Sayısı Değişikliklerinin Klinikle ve Kantitatif PCR ile Değerlendirilmesi, 2014 - 2015

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