Prof. OSMAN KORAY BODUROĞLU

Personal Information

Office Phone: +90 312 305 1173 Extension: 121
Email: kbodur@hacettepe.edu.tr
Web: https://avesis.hacettepe.edu.tr/kbodur

International Researcher IDs
ORCID: 0000-0001-6260-1942
ScopusID: 6701572368
Yoksis Researcher ID: 28175

Education Information

Post Doctorate of Medicine, Hacettepe University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, Turkey 2008 - 2013
Doctorate, Hacettepe University, Sağlık Bilimleri Enstitüsü, Genetik, Turkey 1998 - 2002
Expertise In Medicine, Hacettepe University, Tıp Fakültesi, Pediatri, Turkey 1988 - 1993
Undergraduate, Hacettepe University, Tıp Fakültesi, Turkey 1982 - 1988

Foreign Languages

English, C1 Advanced

Dissertations

Doctorate, Nöral tüp defektlerinin etiyolojisinde 5, 10 metilentetrahidrofolat redüktaz gen polimorfizmlerinin rolü, Hacettepe Üniversitesi, Sağlık Bilimleri Fakültesi, 2002

Research Areas

Medicine, Internal Medicine Sciences, Child Health and Diseases, Medical Genetics, Health Sciences

Academic Titles / Tasks

Professor, Hacettepe University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 1995 - Continues

Academic and Administrative Experience

Hacettepe Üniversitesi, Çocuk Sağlığı Enstitüsü, 2007 - Continues
Hacettepe Üniversitesi, Hacettepe Üniversitesi Hastaneleri Araştırma Ve Uygulama Merkezi, 2008 - 2010
Hacettepe Üniversitesi, Hacettepe Üniversitesi Hastaneleri Araştırma Ve Uygulama Merkezi, 2000 - 2008

Published journal articles indexed by SCI, SSCI, and AHCI

I. **A spectrum of TP63-related disorders with eight affected individuals in five unrelated families**
SOĞUKPINAR M., ÜTİNE G. E., Boduroğlu K., ŞİMŞEK KİPER P. Ö.
European Journal of Medical Genetics, vol.68, 2024 (SCI-Expanded)

II. **Further Expanding the Mutational Spectrum of Gorlin Syndrome in Three Unrelated Families**
KOLKIRAN A., ŞİMŞEK KİPER P. Ö., TOPALOĞLU YASAN G., KARAOSMANOĞLU B., Taşklran E., ÜTİNE G. E., TÜZ H. H., Boduroğlu K.
Molecular Syndromology, 2024 (SCI-Expanded)

III. **A Long-Term Follow-Up of a Patient with a Novel PORCN Variant and Additional Clinical Features**
Akalin A., Grzeschik K., ÜTİNE G. E., Boduroğlu K., ŞİMŞEK KİPER P. Ö.
Molecular Syndromology, 2024 (SCI-Expanded)

IV. **A Novel ZBTB20 Variant in a Patient with Primrose Syndrome: A Rare Clinical Entity with Distinctive Features**
Soğukplnar M., KARAOSMANOĞLU B., ÜTİNE G. E., Boduroğlu K., ŞİMŞEK KİPER P. Ö.
Molecular Syndromology, 2024 (SCI-Expanded)

V. **A novel biallelic CRIPT variant in a patient with short stature, microcephaly, and distinctive facial features**
AKALIN A., ŞİMŞEK KİPER P. Ö., Taşkıran E. Z., KARAOSMANOĞLU B., ÜTİNE G. E., BODUROĞLU O. K.
American Journal of Medical Genetics, Part A, vol.191, no.4, pp.1119-1127, 2023 (SCI-Expanded)

VI. **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., Özşin C., KOÇ N., Demir G. Ü., ALANAY Y., Utine E., BODUROĞLU O. K., Tekçiçek M., ŞİMŞEK KİPER P. Ö.
European Journal of Medical Genetics, vol.66, no.4, 2023 (SCI-Expanded)

VII. **Typical Face, Developmental Delay, and Hearing Loss in a Patient with 3M Syndrome: The Co-Occurrence of Two Rare Conditions**
AKALIN A., Simsek-Kiper P. O., Taskiran E., ÜTİNE G. E., BODUROĞLU O. K.
MOLECULAR SYNDROMOLOGY, vol.13, no.6, pp.537-542, 2023 (SCI-Expanded)

VIII. **A lethal and rare cause of arthrogryposis: Glyt1 encephalopathy**
Dasar T., ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., ÇAĞAN M., ÖZYÜNCÜ Ö., DEREN Ö., ÜTİNE G. E., GÜÇER K. Ş., BODUROĞLU O. K.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.65, no.12, 2022 (SCI-Expanded)

IX. **Biallelic loss-of-function variants in EXOC6B are associated with impaired primary ciliogenesis and cause spondylo-epi-metaphyseal dysplasia with joint laxity type 3**
ŞİMŞEK KİPER P. Ö., Jacob P., Upadhyai P., TAŞKIRAN Z. E., Guleria V. S., KARAOSMANOĞLU B., İMREN G., GÖÇMEN R., Bhavani G. S., Kausthubham N., et al.
HUMAN MUTATION, vol.43, no.12, pp.2116-2129, 2022 (SCI-Expanded)

X. **Diagnostic distribution and postnatal evaluation of prenatally detected short femur: A single center experience**
KAHRAMAN A. B., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.188, no.8, pp.2367-2375, 2022 (SCI-Expanded)

XI. **Al-Gazali skeletal dysplasia constitutes the lethal end of ADAMTSL2-related disorders**
Batkovskyte D., McKenzie F., Taylan F., ŞİMŞEK KİPER P. Ö., Nikkel S. M., Ohashi H., Miyahara H., Eriksson G., Ha T., ÜTİNE G. E., et al.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.SUPPL 1, pp.41-42, 2022 (SCI-Expanded)

XII. **Obstructive sleep apnea in children with Down syndrome: is it possible to predict severe apnea?**
Hizal M., ŞATIRER Ö., Polat S. E., Tural D. A., Ozsezen B., SUNMAN B., KARAHAN S., EMİRALİOĞLU N., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., et al.
EUROPEAN JOURNAL OF PEDIATRICS, vol.181, no.2, pp.735-743, 2022 (SCI-Expanded)

XIII. **Diagnostic yield of microarrays in individuals with non-syndromic developmental delay and intellectual disability**
Oguz S., Arslan U. E., Kiper P. O. S., Alikasifoglu M., BODUROĞLU O. K., Utine G. E.

JOURNAL OF INTELLECTUAL DISABILITY RESEARCH, vol.65, no.12, pp.1033-1048, 2021 (SSCI)

XIV. **Biallelic ITGB4 variants in familial pyloric atresia without epidermolysis bullosa: Report of two families with five siblings**
SOYER T., KARAOSMANOĞLU B., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., KARNAK İ., BODUROĞLU O. K., ÜTİNE G. E.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.11, pp.3427-3432, 2021 (SCI-Expanded)

XV. **Main Physical Features, Echocardiographic and Renal Ultrasonographic Findings of Turner Syndrome in 107 Pediatric Patients**
AKALIN A., ERTUĞRUL İ., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
MOLECULAR SYNDROMOLOGY, vol.12, no.6, pp.335-341, 2021 (SCI-Expanded)

XVI. **Spondyloepimetaphyseal dysplasia EXTL3-deficient type: Long-term follow-up and review of the literature**
AKALIN A., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., Utine E., ALANAY Y., Ozcelik U., BODUROĞLU O. K.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.10, pp.3104-3110, 2021 (SCI-Expanded)

XVII. **Sleep disordered breathing in patients with Prader willi syndrome: Impact of underlying genetic mechanism**
Ozsezen B., EMİRALİOĞLU N., Ozon A., Akin O., Tural D. A., SUNMAN B., Hejiyeva A., Hizal M., Alikasifoglu A., ŞİMŞEK KİPER P. Ö., et al.
RESPIRATORY MEDICINE, vol.187, 2021 (SCI-Expanded)

XVIII. **Kohlschutter-Tonz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity**
AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., Taskiran E., Schossig A., ÜTİNE G. E., Zschocke J., BODUROĞLU O. K.
JOURNAL OF CHILD NEUROLOGY, vol.36, no.10, pp.816-822, 2021 (SCI-Expanded)

XIX. **Diagnostic yield of whole-exome sequencing in non-syndromic intellectual disability**
Taşkıran Z. E., Karaosmanoglu B., Kosukcu C., Urel-Demir G., Akgun-Dogan O., Simsek-Kiper P. O., Alikasifoglu M., Boduroğlu O. K., Utine G. E.
JOURNAL OF INTELLECTUAL DISABILITY RESEARCH, vol.65, no.6, pp.577-588, 2021 (SSCI)

XX. **Further expanding the mutational spectrum of brain abnormalities, neurodegeneration, and dysosteosclerosis: A rare disorder with neurologic regression and skeletal features**
KINDIŞ 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)

XXI. **Natural history of TRPV4-Related disorders: From skeletal dysplasia to neuromuscular phenotype**
ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö., Oncel I., ÜTİNE G. E., Haliloglu G., BODUROĞLU O. K.
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, vol.32, pp.46-55, 2021 (SCI-Expanded)

XXII. **Two Siblings with Kaufman Oculocerebrofacial Syndrome Resembling Oculoauriculovertebral Spectrum**
ÜREL DEMİR G., Aydin B., KARAOSMANOĞLU B., AKGÜN DOĞAN Ö., Taskiran E. Z., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
MOLECULAR SYNDROMOLOGY, vol.12, no.2, pp.106-111, 2021 (SCI-Expanded)

XXIII. **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)

XXIV. **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., Alikaşifoğlu M., Boduroğlu O. K., Coşkun T., Ütine G. E.
Molecular Syndromology, vol.11, pp.302-308, 2020 (SCI-Expanded)

XXV. **Atypical Presentation of Sengers Syndrome: A Novel Mutation Revealed with Postmortem Genetic Testing.**
Guleray N., Kosukcu C., Taskiran Z., Simsek K., Utine G., Gucer S., Tokatli A., Boduroglu K., Alikasifoglu M.

Fetal and pediatric pathology, vol.39, no.2, pp.163-171, 2020 (SCI-Expanded)

XXVI. **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-Lafcl N., Şimşek-Kiper P. Ö., Eda Utine G., ALİKAŞİFOĞLU M., Boduroğlu K.
MOLECULAR SYNDROMOLOGY, vol.11, no.1, pp.38-42, 2020 (SCI-Expanded)

XXVII. **ADA2 deficiency in a patient with Noonan syndrome-like disorder with loose anagen hair: The co-occurrence of two rare syndromes**
Akgun-Dogan O., Simsek-Kiper P. O., Taskiran E., Lissewski C., Brinkmann J., Schanze D., Göçmen R., Cagdas D. N., Bilginer Y., Utine G. E., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.12, pp.2474-2480, 2019 (SCI-Expanded)

XXVIII. **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)

XXIX. **Chromosome Analysis in the Assessment for Gender Affirmation Process: A Retrospective Study**
Bagcaz A., BODUROĞLU O. K., BAŞAR K.
TURK PSIKIYATRI DERGISI, vol.30, no.3, pp.157-162, 2019 (SSCI)

XXX. **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)

XXXI. **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)

XXXII. **Hyperphosphatasia with mental retardation syndrome type 4 In two siblings-expanding the phenotypic and mutational spectrum**
Akgün D., Demir G., Kosukcu C., Taskiran E., Simsek-Kiper P., Utine G., Alikaşifoğlu M., Boduroğlu K.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.6, 2019 (SCI-Expanded)

XXXIII. **Syrian children in Turkey: A model of action for national pediatric societies**
ÖZMERT E. N., DERMAN O., Bideci A., Okumuş N., Boduroğlu K., Bakkaloğlu S., Hasanoğlu E., Alden E.
Pediatrics, vol.143, no.2, 2019 (SCI-Expanded)

XXXIV. **Effects of Vitamin D and estrogen receptor polymorphisms on bone mineral density in adolescents with anorexia nervosa**
I Nan-Erdoğan I., AKGÜL S., Işgln-Atlcl K., Tuğrul-Yücel T., Boduroğlu K., DERMAN O., KANBUR N.
Journal of Pediatric Endocrinology and Metabolism, 2019 (SCI-Expanded)

XXXV. **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)

XXXVI. **Non-immune hydrops fetalis: A retrospective analysis of 151 autopsies performed at a single center**
KAYKI G., Gucer S., AKÇÖREN Z., ORHAN D., TALİM B., YURDAKÖK M., YİĞİT Ş., BODUROĞLU O. K., ÜTİNE G. E., Orgul G., et al.
TURKISH JOURNAL OF PEDIATRICS, vol.60, no.5, pp.471-477, 2018 (SCI-Expanded)

XXXVII. **Further expansion of the mutational spectrum of spondylo-meta-epiphyseal dysplasia with abnormal calcification**
Urel-Demir G., Simsek-Kiper P. O., AKGÜN-DOĞAN O., GÖÇMEN R., WANG Z., MATSUMOTO N., MİYAKE N., Utine G. E., NİSHİMURA G., IKEGAWA S., et al.
Journal of Human Genetics, vol.63, no.9, pp.1003-1007, 2018 (SCI-Expanded)

XXXVIII. **Further delineation of spondyloepimetaphyseal dysplasia Faden-Alkuraya type: A RSPRY1-associated spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis**
Simsek-Kiper P., Taskiran E., Kosukcu C., Urel-Demir G., Akgun-Dogan O., Yilmaz G., Utine G., Nishimura G., Boduroglu K., Alikasifoglu M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.176, no.9, pp.2009-2016, 2018 (SCI-Expanded)

XXXIX. **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)

XL. **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)

XLI. **Clinical and molecular evaluation of 16 patients with rett syndrome**
Zengin-Akkuş P., Taşkıran E. Z., Kabaçam S., Şimşek-Kiper P. Ö., Haliloğlu G., Boduroğlu K., ÜTİNE G. E.
Turkish Journal of Pediatrics, vol.60, no.1, pp.1-9, 2018 (SCI-Expanded)

XLII. **Epigenotype and phenotype correlations in patients with Beckwith-Wiedemann syndrome**
Bilgin B., Kabaçam S., Taşkıran E., Şimşek-Kiper P. Ö., Alanay Y., Boduroğlu K., ÜTİNE G. E.
Turkish Journal of Pediatrics, vol.60, no.5, pp.506-513, 2018 (SCI-Expanded)

XLIII. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Costello Syndrome**
VURALLI 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)

XLIV. **Anauxetic dysplasia: A rare clinical entity**
Akgün-Doğan Ö., Şimsek-Kiper P. Ö., ÜTİNE G. E., Boduroğlu K.
Turkish Journal of Pediatrics, vol.60, no.1, pp.89-93, 2018 (SCI-Expanded)

XLV. **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)

XLVI. **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)

XLVII. **Dermal fibroblast transcriptome indicates contribution of WNT signaling pathways in the pathogenesis of Apert syndrome**
Çetinkaya A., Taşkıran E., Soyer T., Şimşek-Kiper P., Utine G., Tunçbilek G., Boduroğlu K., Alikaşifoğlu M.
TURKISH JOURNAL OF PEDIATRICS, vol.59, no.6, pp.619-624, 2017 (SCI-Expanded)

XLVIII. **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)

XLIX. **HERC1 mutations in idiopathic intellectual disability**
ÜTİNE G. E., Taskiran E. Z., KOŞUKCU C., KARAOSMANOĞLU 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)

L. **Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2**
Boegershausen N., Gatinois V., Riehmer V., Kayserili H., Becker J., Thoenes M., Simsek-Kiper P. O., Barat-Houari M., Elcioglu N. H., Wieczorek D., et al.
Human Mutation, vol.37, no.9, pp.847-864, 2016 (SCI-Expanded)

LI. **Bi-allelic Mutations in KLHL7 Cause a Crisponi/CISS1-like Phenotype Associated with Early-Onset**

Retinitis Pigmentosa
Angius A., Uva P., Buers I., Oppo M., Puddu A., Onano S., Persico I., Loi A., Marcia L., Höhne W., et al.
American Journal of Human Genetics, vol.99, no.1, pp.236-245, 2016 (SCI-Expanded)

LII. **Cortical-bone fragility - Insights from sFRP4 deficiency in Pyle's disease**
Kiper P. Ö., Saito H., Gori F., Unger S., Hesse E., Yamana K., Kiviranta R., SOLBAN N., LIU J., BROMMAGE R., et al.
New England Journal of Medicine, vol.374, no.26, pp.2553-2562, 2016 (SCI-Expanded)

LIII. **A novel de novo mutation involving the MLL2 gene in a Kabuki syndrome patient presenting with seizures**
Bekircan-Kurt C. E., Şimşek-Kiper P. Ö., Boduroğlu K., DERİCİOĞLU N.
Turkish Journal of Pediatrics, vol.58, no.1, pp.97-100, 2016 (SCI-Expanded)

LIV. **A Diagnosis to Consider in Intellectual Disability: Mowat-Wilson Syndrome**
Kılıç E., Cetinkaya A., Ütine G. E., Boduroğlu K.
Journal of Child Neurology, vol.31, no.7, pp.913-917, 2016 (SCI-Expanded)

LV. **Experience of a skeletal dysplasia registry in Turkey: A five-years retrospective analysis**
Kurt-Sukur E. D., Simsek-Kiper P. O., ÜTİNE G. E., Boduroglu K., Alanay Y.
American Journal of Medical Genetics, Part A, vol.167, no.9, pp.2065-2074, 2015 (SCI-Expanded)

LVI. **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)

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., Boduroglu K., Braunholz D., Caliebe A., Chrzanowska K. H., Czeschik J. C., et al.
Human Genetics, vol.134, no.6, pp.553-568, 2015 (SCI-Expanded)

LVIII. **A novel mutation in RNU4ATAC in a patient with microcephalic osteodysplastic primordial dwarfism type I**
Kılıç E., Yigit G., ÜTİNE G. E., Wollnik B., MIHÇI E., Nur B. G., Boduroglu K.
American Journal of Medical Genetics, Part A, vol.167, no.4, pp.919-921, 2015 (SCI-Expanded)

LIX. **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
Bögershausen N., Tsai I., Pohl E., Kiper P. O. S., Beleggia F., Ferda Percin E., Keupp K., Matchan A., Milz E., Alanay Y., et al.
Journal of Clinical Investigation, vol.125, no.9, pp.3585-3599, 2015 (SCI-Expanded)

LX. **A case of 22q11.2 deletion syndrome with right microphthalmia and left corneal staphyloma**
Tarlan B., Kiratli H., Kılıç E., Utine E., Boduroğlu K.
Ophthalmic genetics, vol.35, no.4, pp.248-251, 2014 (SCI-Expanded)

LXI. **Novel homozygous mutations in the osteoprotegerin gene TNFRSF11B in two unrelated patients with juvenile Paget's disease**
Naot D., Choi A., Musson D. S., Kiper P. O. S., ÜTİNE G. E., Boduroglu K., Peacock M., DiMeglio L. A., Cundy T.
BONE, vol.68, pp.6-10, 2014 (SCI-Expanded)

LXII. **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, no.4, pp.147-151, 2014 (SCI-Expanded)

LXIII. **Wildervanck syndrome: An uncommon cause of Duane syndrome**
Sekeroglu H. T., Simsek-Kiper P. O., ÜTİNE G. E., Boduroglu K., Sanac A. S., Sener E. C.
JOURNAL FRANCAIS D OPHTALMOLOGIE, vol.37, no.8, 2014 (SCI-Expanded)

LXIV. **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)

LXV. **Prenatal diagnosis in a fetus with de-novo 20q11.2q13.1 deletion and review of the literature**
Turgal M., ÖZYÜNCÜ Ö., ÜTİNE G. E., KILIÇ E., Boduroglu K.

CLINICAL DYSMORPHOLOGY, vol.23, no.3, pp.111-113, 2014 (SCI-Expanded)

LXVI. **Etiological yield of SNP microarrays in idiopathic intellectual disability**
Ütine G. E., Halıloglu G., Volkan-Salancı B., Cetinkaya A., Kıper 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)

LXVII. **Expanding the Mutational Spectrum of CRLF1 in Crisponi/CISS1 Syndrome**
Piras R., Chiappe F., La Torraca I., Buers I., Usala G., Angius A., Akin M. A., Basel-Vanagaite L., Benedicenti F., Chiodin E., et al.
HUMAN MUTATION, vol.35, no.4, pp.424-433, 2014 (SCI-Expanded)

LXVIII. **Cathepsin K analysis in a pycnodysostosis cohort: demographic, genotypic and phenotypic features**
ARMAN A., BEREKET A., ÇOKER A., ŞİMŞEK KİPER P. Ö., GÜRAN T., Ozkan B., Atay Z., Akcay T., Haliloglu B., BODUROĞLU O. K., et al.
ORPHANET JOURNAL OF RARE DISEASES, vol.9, 2014 (SCI-Expanded)

LXIX. **Celiac disease in Williams-Beuren syndrome**
Simsek-Kiper P. O., Sahin Y., Arslan U., Alanay Y., Boduroglu K., ORHAN D., ÖZEN H., ÜTİNE G. E.
TURKISH JOURNAL OF PEDIATRICS, vol.56, no.2, pp.154-159, 2014 (SCI-Expanded)

LXX. **TMCO1 Deficiency Causes Autosomal Recessive Cerebrofaciothoracic Dysplasia**
ALANAY Y., BEKİR E., ÜTİNE G. E., ORÇUN H., ŞİMŞEK KİPER P. Ö., TAŞKIRAN E. Z., PERÇİN F. E., UZ E., MAHMUT ŞAMİL S., BAYRAM Y., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.164, no.2, pp.291-304, 2014 (SCI-Expanded)

LXXI. **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)

LXXII. **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., Boduroglu K., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.164, no.1, pp.99-105, 2014 (SCI-Expanded)

LXXIII. **A de novo 11q23 deletion in a patient presenting with severe ophthalmologic findings, psychomotor retardation and facial dysmorphism**
Şimşek-Kiper P. Ö., Bayram Y., ÜTİNE G. E., Alanay Y., Boduroğlu K.
Turkish Journal of Pediatrics, vol.56, no.1, pp.80-84, 2014 (SCI-Expanded)

LXXIV. **Barraquer-Simons syndrome: A rare clinical entity**
Simsek-Kiper P. O., Roach E., ÜTİNE G. E., Boduroglu K.
American Journal of Medical Genetics, Part A, vol.164, no.7, pp.1756-1760, 2014 (SCI-Expanded)

LXXV. **Vesiculopustular eruption in neonatal transient myeloproliferative disorder**
Nar I., Surmeli-Onay O., Aytac Ş. S., TALİM B., Kiper P. O., Boduroglu K., YURDAKÖK M.
Indian Journal of Pediatrics, vol.81, no.4, pp.391-393, 2014 (SCI-Expanded)

LXXVI. **Positive effects of an angiotensin II type 1 receptor antagonist in Camurati-Engelmann disease: A single case observation**
Simsek-Kiper P. O., Dikoglu E., Campos-Xavier B., ÜTİNE G. E., Bonafe L., Unger S., Boduroglu K., Superti-Furga A.
American Journal of Medical Genetics, Part A, vol.164, no.10, pp.2667-2671, 2014 (SCI-Expanded)

LXXVII. **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, no.25, pp.5121-5135, 2013 (SCI-Expanded)

LXXVIII. **Homozygosity for a Novel Truncating Mutation Confirms TBX15 Deficiency as the Cause of Cousin Syndrome**
Dikoglu E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., Campos-Xavier B., BODUROĞLU O. K., Bonafe L., Superti-Furga A., Unger

S.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.161, no.12, pp.3161-3165, 2013 (SCI-Expanded)

LXXIX. **Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused by PAPSS2 Mutations**
Iida A., ŞİMŞEK KİPER P. Ö., Mizumoto S., Hoshino T., Elcioglu N., Horemuzova E., Geiberger S., Yesil G., Kayserili H., ÜTİNE G. E., et al.
HUMAN MUTATION, vol.34, no.10, pp.1381-1386, 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, vol.28, no.7, pp.926-932, 2013 (SCI-Expanded)

LXXXI. **A Case of 13q22. 2 q33. 3 Deletion**
KILIÇ E., ÜTİNE G. E., Alikagsifoglu M., BODUROĞLU O. K.
CHROMOSOME RESEARCH, vol.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, vol.98, pp.578, 2013 (SCI-Expanded)

LXXXIII. **Deletion of GNAS in a girl presenting with severe pre- and post-natal growth retardation, developmental delay and facial dysmorphism**
ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
CHROMOSOME RESEARCH, vol.21, 2013 (SCI-Expanded)

LXXXIV. **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, vol.21, 2013 (SCI-Expanded)

LXXXV. **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, vol.22, no.8, pp.1643-1653, 2013 (SCI-Expanded)

LXXXVI. **A case of Sotos syndrome with 5q35 microdeletion and novel clinical findings**
KILIÇ E., ÜTİNE G. E., Boduroglu K.
TURKISH JOURNAL OF PEDIATRICS, vol.55, no.2, pp.207-209, 2013 (SCI-Expanded)

LXXXVII. **MUCOLIPIDOSIS TYPE III IN AN ADOLESCENT PRESENTING WITH ATYPICAL FACIAL FEATURES AND SKELETAL DEFORMITIES**
Simsek-Kiper P. O., Topalglu R., Sahin Y., Utine G. E., BODUROĞLU O. K.
GENETIC COUNSELING, vol.24, no.1, pp.7-12, 2013 (SCI-Expanded)

LXXXVIII. **Congenital partial arhinia: a rare malformation of the nose coexisting with holoprosencephaly**
Takci S., Korkmaz A., Simsek-Kiper P. O., ÜTİNE G. E., BODUROĞLU O. K., YURDAKÖK M.
TURKISH JOURNAL OF PEDIATRICS, vol.54, no.4, pp.440-443, 2012 (SCI-Expanded)

LXXXIX. **PRRX1 is mutated in an otocephalic newborn infant conceived by consanguineous parents**
Celik T., Simsek P. O., Sozen T., Ozyuncu O., Utine G. E., Talim B., Yigil Ş., BODUROĞLU O. K., Kamnasaran D.
CLINICAL GENETICS, vol.81, no.3, pp.294-297, 2012 (SCI-Expanded)

XC. **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, vol.54, no.2, pp.198-202, 2012 (SCI-Expanded)

XCI. **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, vol.130, no.6, pp.715-724, 2011 (SCI-Expanded)

XCII. **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)

XCIII. **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, no.9, pp.2288-2292, 2011 (SCI-Expanded)

XCIV. **A patient with a duplication of 16q and a deletion of 3p presenting with coloboma and buphthalmos**
BODUROĞLU O. K., Simsek P. O., ÜTİNE G. E.
CHROMOSOME RESEARCH, vol.19, 2011 (SCI-Expanded)

XCV. **A case of deletion of 4q and duplication of 10q presenting with tetralogy of Fallot, developmental delay and facial dysmorphism**
Simsek P. O., ÜTİNE G. E., BODUROĞLU O. K.
CHROMOSOME RESEARCH, vol.19, 2011 (SCI-Expanded)

XCVI. **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)

XCVII. **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, vol.22, no.4, pp.401-409, 2011 (SCI-Expanded)

XCVIII. **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, vol.34, 2011 (SCI-Expanded)

XCIX. **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, no.4, pp.947-949, 2010 (SCI-Expanded)

C. **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, no.6, pp.1317-1318, 2009 (SCI-Expanded)

CI. **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, vol.51, no.3, pp.294-297, 2009 (SCI-Expanded)

CII. **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)

CIII. **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)

CIV. **KABUKI SYNDROME AND TRISOMY 10p**
Utine G. E., ALANAY Y., Aktas D., BODUROĞLU O. K., Alikasifoglu M., Tuncbilek E.
GENETIC COUNSELING, vol.19, no.3, pp.291-300, 2008 (SCI-Expanded)

CV. **Cerebro-facio-thoracic dysplasia: expanding the phenotype**
Cilliers D., ALANAY Y., BODUROĞLU O. K., Utine E., Tuncbilek E., Clayton-Smith J.
CLINICAL DYSMORPHOLOGY, vol.16, no.2, pp.121-125, 2007 (SCI-Expanded)

CVI. **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, vol.51, pp.151-161, 2007 (SCI-Expanded)

CVII. **Coexistent mosaic monosomy 21 and fragile X syndrome in a mentally retarded male patient**
Utine G. E., Aktas D., BODUROĞLU O. K., Alikasifoglu M., Tuncbilek E.
GENETIC COUNSELING, vol.18, no.2, pp.171-177, 2007 (SCI-Expanded)

CVIII. **Neocentric small supernumerary marker chromosomes (sSMC) - three more cases and review of the literature**
Liehr T., Utine G. E., Trautmann U., Rauch A., Kuechler A., Pietracz J., Bocian E., Kosyakova N., Mrasek K., BODUROĞLU O. K., et al.
CYTOGENETIC AND GENOME RESEARCH, vol.118, no.1, pp.31-37, 2007 (SCI-Expanded)

CIX. **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, vol.47, no.4, pp.327-333, 2005 (SCI-Expanded)

CX. **Celiac disease screening in 100 Turkish children with Down syndrome**
ALANAY Y., BODUROĞLU O. K., Tuncbilek E.
TURKISH JOURNAL OF PEDIATRICS, vol.47, no.2, pp.138-140, 2005 (SCI-Expanded)

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

CXII. **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, no.1, pp.92-95, 2004 (SCI-Expanded)

CXIII. **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, no.1, pp.5-10, 2004 (SCI-Expanded)

CXIV. **Cervical diastematomyelia in cervico-oculo-acoustic (Wildervanck) syndrome: MRI findings**
Balci S., Oguz K., Firat M., BODUROĞLU O. K.
CLINICAL DYSMORPHOLOGY, vol.11, no.2, pp.125-128, 2002 (SCI-Expanded)

Articles Published in Other Journals

I. **Clinical Evaluation of the Five Patients with Mosaic Trisomy 8 Syndrome: Case Series**
DAŞAR T. N., Soğukpınar M., Simsek-Kiper P. O., UTİNE G. E., BODUROĞLU O. K.
Türkiye Klinikleri Pediatri Dergisi, vol.32, no.2, pp.91-95, 2023 (Scopus)

II. **A Life-Threatening Complication in a Patient with Ehlers-Danlos Syndrome Musculocontractural Type**
Dasar T., Donkervoort S., ŞİMŞEK KİPER P. Ö., GÖÇMEN R., ÜTİNE G. E., BODUROĞLU O. K., Bonnemann C., Haliloglu G.
JOURNAL OF PEDIATRIC RESEARCH, vol.9, no.3, pp.297-301, 2022 (ESCI)

III. **Achondroplasiaand Down SyndromeIn An Infant: A RareCo-Occurrence**
ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö., GÖÇMEN R., ÜTİNE G. E., BODUROĞLU O. K.
Asia Pacific Journal of Pediatric and Child Health, 2020 (Scopus)

IV. **Akondroplazide baba yaşı: İleri baba yaşı kaçtır?**
ATAR S., ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
Çocuk Sağlığı ve Hastalıkları, vol.62, no.62, pp.7-9, 2019 (Scopus)

V. **Fragile X Syndrome: A Genetic Disorder to Consider in Patients with Speech Delay**
ÇELEN YOLDAŞ T., ÜTİNE G. E., ÖZMERT E. N., BODUROĞLU O. K.

Türkiye Çocuk Hastalıkları Dergisi, vol.12, no.4, pp.289-292, 2018 (Peer-Reviewed Journal)

VI. **3M Sendromu**
ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
Çocuk Sağlığı ve Hastalıkları Dergisi, 2017 (Scopus)

VII. **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)

VIII. **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)

IX. **3M syndrome 3M sendromu**
Kiper P. Ö. Ş., ÜTİNE G. E., Boduroğlu K.
Cocuk Sagligi ve Hastaliklari Dergisi, vol.60, no.2, pp.56-63, 2017 (Scopus)

X. **Noonan sendromunda göz bulguları: İki olgu ve literatürün gözden geçirilmesi**
TAYLAN ŞEKEROĞLU H., ERKAN TURAN K., ŞİMŞEK KİPER P. Ö., ÇOLAK D., Utine G. E., BODUROĞLU O. K.
Glokom-Katarakt, vol.12, pp.229-232, 2017 (Peer-Reviewed Journal)

XI. **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)

XII. **1p36 Microdeletion Syndrome: A Case Report**
ZENGİN AKKUŞ P., ŞAHİN Y., UTİNE G. E., BODUROĞLU O. K.
Acta Medica, vol.45, no.1, pp.26-28, 2014 (Peer-Reviewed Journal)

XIII. **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, vol.2, no.2, pp.64-71, 2011 (ESCI)

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. **Expanding the clinical and mutational spectrum of Roberts Syndrome with previously unreported endocrine findings**
Guleray N., Kiper P. O. S., Utine G. E., 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, Italy, 16 - 19 June 2018, vol.27, pp.127

V. **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, Italy, 16 - 19 June 2018, vol.27, pp.130

VI. **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, Italy, 16 - 19 June 2018, vol.27, pp.97

VII. **A novel homozygous ROGD1 mutation in two siblings with kohlschutter-tonz syndrome: a rare entity**
Kiper P. O. S., Utine G. E., Zschocke J., BODUROĞLU O. K.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.346

VIII. **17q24.3 Duplication In A Patient Presenting With SRY-Negative 46,XX Disorders of Sex Development**
Kiper P. O. S., Ozon A., Gonc N., Alikasifoglu A., Utine G. E., BODUROĞLU O. K., Alikasifoglu M.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.196

IX. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Costello Syndrome**
VURALLI 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

X. **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

XI. **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

XII. **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

XIII. **Effects of estrogen and vitamin D polymorphisms on bone density in adolescent anorexia nervosa patients**
Inan Erdogan I., AKGÜL S., IŞGIN K., BODUROĞLU O. K., DERMAN O., KANBUR N.
14th İnternational conferans on Clinical pediatrics., 14 - 16 June 2018

XIV. **Rett sendromlu 16 hastanın klinik ve moleküler açıdan değerlendirilmesi**
ZENGİN AKKUŞ P., TAŞKIRAN Z. E., KABAÇAM S., ŞİMŞEK KİPER P. Ö., HALİLOĞLU V. G., BODUROĞLU O. K., UTİNE G. E.
61. Türkiye Milli Pediatri Kongresi, Antalya, Turkey, 15 - 19 November 2017

XV. **Oftalmo-akromelik sendrom**
ÜREL DEMİR G., TAŞKIRAN Z. E., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

XVI. **Peters Plus Sendromu**
ÜREL DEMİR G., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

XVII. **Çok nadir iki sendromun birlikteliği: Noonan syndrome like with loose anagen hair ile ICF2 sendromu**
AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., Lissewski C., Erman B., ÇAĞDAŞ AYVAZ D. N., Boztuğ K., ÜTİNE G. E., Zenker M., TEZCAN F. İ., BODUROĞLU O. K.

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

XVIII. **KID Sendromu: Nadir bir klinik antite**
AKGÜN DOĞAN Ö., OĞUZ S., MENTEŞOĞLU D., ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., DOĞAN GÜNAYDIN S., ERSOY EVANS S., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

XIX. **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

XX. **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

XXI. **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

XXII. **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

XXIII. **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

XXIV. **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

XXV. **Tüberoskleroz hemihiperplazi birlikteliği**
AKGÜN DOĞAN Ö., ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

XXVI. **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

XXVII. **Teebi hipertelorizm sendromu**
ÜREL DEMİR G., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

XXVIII. **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

XXIX. **SOX9 gene dublication-related 46,XX ovotesticular disorder of sex development**
ÖZÖN Z. A., ALİKAŞİFOĞLU A., GÖNÇ E. N., VURALLI KARAOĞLAN D., BÜYÜKYILMAZ G., ŞİMŞEK KİPER P. Ö., UTİNE E., ORHAN D., SOYER T., BODUROĞLU O. K., et al.
10th International Meeting of Pediatric Endocrinolgy, 14 - 17 September 2017

XXX. **ICF SYNDROME: CLINICAL, IMMUNOLOGICAL AND CYTOGENETIC ANALYSIS OF SEVENCASES**
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

XXXI. **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

XXXII. **Duplication ina 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

XXXIII. **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, Kopenhag, Denmark, 27 - 30 May 2017

XXXIV. **A novel homozygous ROGD1 mutation in two siblings with Kohlschutter-Tönz syndrome: A rare entity**
ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., Zschocke J., BODUROĞLU O. K.
European Society of Human Genetics Conference 2017, Kopenhag, 27 - 30 May 2017

XXXV. **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

XXXVI. **Cinsiyetinden hoşnutsuzluk ve 46,XX gonadal disgenezi birlikteliği**
KAMIŞ G. Z., BODUROĞLU O. K., BAYRAKTAR M., BAŞAR K.
21. TPD Yıllık Toplantı ve Klinik Eğitim Sempozyumu, Turkey, 19 - 22 April 2017, vol.28, pp.72-73

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

XXXVIII. **Arthrogryposis multiplex congenita AMC Spectrum and classification at a tertiary referral center**
ÖNCEL İ., HALİLOĞLU V. G., ÜTİNE G. E., AKSOY M. C., BODUROĞLU O. K., TOPALOĞLU H. A.
21st International Congress of the World-Muscle-Society, Granada, Nicaragua, 4 - 08 October 2016, vol.26, pp.107

XXXIX. **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

XL. **Clinical and Molecular aspects and genotype phenotype correlation in Rett syndrome**
ZENGİN AKKUŞ P., ÜTİNE G. E., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., HALİLOĞLU V. G., BODUROĞLU O. K.
European Society of Human Genetics Conference 2016 Barcelona, 21 - 24 May 2016

XLI. **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

XLII. **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 May 2016

XLIII. **Clinical molecular aspects and genotype phenotype correlation in Rett syndrome**
ZENGİN AKKUŞ P., ÜTİNE G. E., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., HALİLOĞLU V. G., BODUROĞLU O. K.
European Society of Human Genetics 2016, Barcelona, Spain, 21 - 24 May 2016

XLIV. **Clinical aspects and genotype-phenotype correlation in Rett syndrome**
ZENGİN AKKUŞ P., ÜTİNE G. E., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., HALİLOĞLU V. G., BODUROĞLU O. K.
European Human Genetics Conference, Barcelona, Spain, 21 May 2016

XLV. **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., UTİNE G. E., BODUROĞLU O. K.
2. Klinik İmmünoloji Kongresi, Antalya, Turkey, 31 March - 03 April 2016

XLVI. **Bir vaka nedeniyle Goltz sendromu**
TAŞTEMEL T., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
2. Ulusal Çocuk Genetik Sempozyumu, 22-24 Ekim 2015, Samsun, Türkiye, Turkey, 22 - 24 October 2015

XLVII. **Nadir görülen bir iskelet displazisi Stüve Wiedemann sendromu**
AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
2. Ulusal Çocuk Genetik Sempozyumu, 22-24 Ekim 2015, Samsun, Türkiye, Sempozyum Kitabı Sayfa 72., Turkey, 22 - 24 October 2015

XLVIII. **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

XLIX. **WAGR Sendromu Aniriden daha fazlası**
PINAR Z. A., AKGÜN DOĞAN Ö., TAYLAN ŞEKEROĞLU H., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
2. Ulusal Çocuk Genetik Sempozyumu, 22-24 Ekim 2015, Samsun, Türkiye, Turkey, 22 - 24 October 2015

L. **Hennekam sendromu Otozomal resesif geçişli bir konjenital lenfödem**
GÜLERAY N., ÖZER M., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
2. Ulusal Çocuk Genetik Sempozyumu, 22-24 Ekim 2015, Samsun, Türkiye, Turkey, 22 - 24 October 2015

LI. **Mowat Wilson sendromu Klinik değerlendirme ve ZEB2 gen mutasyon delesyon analizi**
Kılıç E., Çetinkaya A., ÜTİNE G. E., BODUROĞLU O. K.
II. Ulusal Çocuk Genetik Sempozyumu, Turkey, 22 - 24 October 2015

LII. **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

LIII. **Frontometaphyseal dysplasia with a novel FLNA gene mutation**
Kılıç E., ÜTİNE G. E., Robertson S., BODUROĞLU O. K.
12th International Skeletal Dysplasia Meeting, 29 July - 01 August 2015

LIV. **Keutel Syndrome A Rare Clinical Entity**
DEMİREL M., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
International Skeletal Dysplasia Society 12th Biennial Meeting, July 29th-August 1, 2015-Istanbul, Turkey, Abstract Book p.109., İstanbul, Turkey, 29 July - 01 August 2015

LV. **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

LVI. **Chondrodysplasia Punctata Brachytelephalangic Type With A Novel ARSE Mutation A Clinical Report Poster sunumu Poster No 26 July 29th August 1 2015 Istanbul Turkey Abstract Book p 108**
CEYLAN A. C., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ANLAR F. B., BODUROĞLU O. K., SHİRO İ., GEN N.
International Skeletal Dysplasia Society 12th Biennial Meeting, 29 July - 01 August 2015

LVII. **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

LVIII. **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

LIX. **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

LX. **A Novel SMARCAL1 Mutation Associated With Schimke Immunoosseous Dysplasia A Clinical Report**
AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., MARTİN Z., BODUROĞLU O. K.
International Skeletal Dysplasia Society 12th Biennial Meeting, 29 July - 01 August 2015

LXI. **Skeletal Dysplasia With Intellectual Disability Dyggve Melchior Clausen Dysplasia**
ÜTİNE G. E., ŞİMŞEK KİPER P. Ö., BODUROĞLU O. K., SHİRO İ., GEN N.
International Skeletal Dysplasia Society 12th Biennial Meeting, 29 July - 01 August 2015

LXII. **Chondrodysplasia punctata brachytelephalangic type with a novel ARSE mutation A clinical report**
CEYLAN A. C., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ANLAR F. B., BODUROĞLU O. K., Ikegawa S., Nishimura G.
12th International Skeletal Dysplasia Meeting, 29 July - 01 August 2015

LXIII. **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

LXIV. **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

LXV. **Many faces of Rett syndrome: Is there still a diagnostic delay?**
UTİNE G. E., ZENGİN AKKUŞ P., BODUROĞLU O. K., HALİLOĞLU V. G.
11th European Paediatric Neurology Society (EPNS) Congress, Viyana, Austria, 27 - 30 May 2015, vol.19, pp.148

LXVI. **1p36 delesyonu: bir vaka takdimi**
ZENGİN AKKUŞ P., ŞAHİN Y., ÜTİNE G. E., BODUROĞLU O. K.
3. PUADER Kongresi, Turkey, 30 April 2014

LXVII. **Novel Mutations in the Osteoprotegerin Gene TNFRSF11B in Two Patients with Juvenile Paget's Disease.**
Naot D., Choi A., Musson D., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., Peacock M., Dimeglio L., Cundy T.
Annual Meeting of the American-Society-for-Bone-and-Mineral-Research, Texas, United States Of America, 12 - 15 September 2014, vol.29

LXVIII. **Case Presentation: Sjögren-Larsson Syndrome**
KILIÇ E., KILIÇ M., ÜTİNE G. E., COŞKUN T., NAKANO H., BODUROĞLU O. K.
12th International Congress of Inborn Errors of Metabolism. (ICIEM, 2013, Barcelona, Spain)., Barcelona, Spain, 03 September 2013

LXIX. **Hacettepe Üniversitesi Tıp Fakültesi İhsan Doğramacı Çocuk Hastanesi Çocuk Acil Ünitesi'nde Triaj Uygulama Tecrübesi ve Triaj Performans Analizi**
TEKŞAM Ö., TESTİK M. C., ÖZÖN Z. A., SÖNMEZ V., pehlivan C., KALE G., BODUROĞLU O. K.
II. Uluslararası Sağlıkta Performans ve Kalite Kongresi, Antalya, Turkey, 28 April 2010, pp.255-262

LXX. **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, Sweden, 4 - 07 July 2009, vol.17, pp.209

LXXI. **Prospective analysis of a second-trimester biochemical screening test for trisomy 21**
Portakal O., Deren O., BODUROĞLU O. K., Hascelik G.
60th Annual Meeting of the American-Association-for-Clinical-Chemistry, Washington, Kiribati, 27 - 31 July 2008, vol.54

LXXII. **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, Turkey, 7 - 10 July 2007, vol.15, pp.142-143

LXXIII. **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, Turkey, 7 - 10 July 2007, vol.15, pp.100

LXXIV. **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, Turkey, 7 - 10 July 2007, vol.15, pp.70

LXXV. **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, Turkey, 7 - 10 July 2007, vol.15, pp.96

LXXVI. **Partial monosomy of distal 6q**
Utine E., ALANAY Y., Aktas D., Boduroglu K., Alikasifoglu A., Tuncbilek E.
6th European Cytogenetics Conference, İstanbul, Turkey, 7 - 10 July 2007, vol.15, pp.70

Supported Projects

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 Spondiloepmetafizyeal displazi, Konobrakidaktili ve Kraniyosinostozis, 2017 - 2017

BODUROĞLU O. K., ÜTİNE G. E., ŞİMŞEK KİPER P. Ö., Project Supported by Higher Education Institutions, KohlschutterTonz sendromlu iki kız kardeşte yeni homozigot ROGD1 mutasyonu Nadir Bir Antite, 2017 - 2017

AKGÜL S., KANBUR N., BODUROĞLU O. K., DERMAN O., İNAN ERDOĞAN I., Project Supported by Higher Education Institutions, Adolesan Anoreksiya Nervoza Vakalarında Kemik Mineral Yoğunluğunun Belirlenmesinde Östrojen ve Vitamin D Reseptör Gen Polimorfizminin Rolü, 2015 - 2017

BODUROĞLU O. K., Project Supported by Higher Education Institutions, Silver-Russell Sendromu (SRS) bulunan hastalarda epigenotip ve fenotip ilişkisinin araştırılması, 2014 - 2017

ŞİMŞEK KİPER P. Ö., BODUROĞLU O. K., ÜTİNE G. E., Project Supported by Higher Education Institutions, 3M Sendromunda Ana Gen Defekti Olarak OBSL1 Mutasyonları: Türkiyeden Bir Çalışma, 2016 - 2016

ÜTİNE G. E., HALİLOĞLU V. G., BODUROĞLU O. K., Project Supported by Higher Education Institutions, Rett sendromunun klinik ve moleküler özellikleri ve genotip-fenotip korelasyonu, 2016 - 2016

BODUROĞLU O. K., ÜTİNE G. E., Project Supported by Higher Education Institutions, Yeni Tanı ve Tedavi Yöntemlerinin Klinik Genetik Uygulamalardaki Yeri, 2016 - 2016

ALİKAŞİFOĞLU M., BODUROĞLU O. K., ÜTİNE G. E., Project Supported by Higher Education Institutions, Genetikte Teknolojik İlerlemeler, 2016 - 2016

ÜTİNE G. E., BODUROĞLU O. K., Project Supported by Higher Education Institutions, Klinik Genetikte Tanı ve Tedavi Alanlarında İlerlemeler, 2016 - 2016

ŞİMŞEK KİPER P. Ö., TAŞKIRAN Z. E., BODUROĞLU O. K., ÜTİNE G. E., ALANAY Y., Project Supported by Higher Education Institutions, 3M Sendromlu Bir Grup Türk Hastada Klinik ve Moleküler Özelliklerin Analizi, 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

ALARCON MARTINEZ T., ÜTİNE G. E., BODUROĞLU O. K., Project Supported by Higher Education Institutions, 22q11.2 Delesyon Sendromlu Hastaların Klinik ve İleri Moleküler Değerlendirilmesi ve Genotip-Fenotip Korelasyonunun Araştırılması, 2015 - 2016

ÜTİNE G. E., BODUROĞLU O. K., ZENGİN AKKUŞ P., HALİLOĞLU V. G., Project Supported by Higher Education Institutions, Rett Sendromlu Hastaların Klinik ve Moleküler Değerlendirmesi, 2015 - 2015

Metrics

Publication: 204
Citation (WoS): 1163
Citation (Scopus): 1240
H-Index (WoS): 19
H-Index (Scopus): 19