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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., Taşkıran 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**
Soğukpınar 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 arthrogyriposis: 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**
Batkovskytė 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?**
Hızal M., ŞATIRER Ö., Polat S. E., Tural D. A., Ozsezen B., SUNMAN B., KARAHAN S., EMİRALIOĞLU 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.

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- 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., EMİRALIOĞLU N., Ozon A., Akin O., Tural D. A., SUNMAN B., Hejjiyeva A., Hizal M., Alikasifoglu A., ŞİMŞEK KİPER P. Ö., et al.
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- XVIII. **Kohlschutter-Tonz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity**
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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.
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- XX. **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.
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- 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., Alikasıfoğlu 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.
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- 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.
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- 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., Alikasıfoğlu 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., Işğln-Atcl 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.
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- 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.
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- XLIV. **Anauxetic dysplasia: A rare clinical entity**
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- 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.
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- 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., Alikasifoglu M.
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- 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.
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- 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.
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- L. **Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2**
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- LI. **Bi-allelic Mutations in KLHL7 Cause a Crisponi/CISS1-like Phenotype Associated with Early-Onset**

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- LII. **Cortical-bone fragility - Insights from sFRP4 deficiency in Pyle's disease**
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- LIII. **A novel de novo mutation involving the MLL2 gene in a Kabuki syndrome patient presenting with seizures**
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- LIV. **A Diagnosis to Consider in Intellectual Disability: Mowat-Wilson Syndrome**
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- LV. **Experience of a skeletal dysplasia registry in Turkey: A five-years retrospective analysis**
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- LVI. **Two patients with microdeletion and microduplication involving 1q21.1**
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- LVII. **Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffin-Siris and Nicolaides-Baraitser syndromes**
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- LVIII. **A novel mutation in RNU4ATAC in a patient with microcephalic osteodysplastic primordial dwarfism type I**
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- LIX. **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
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- LX. **A case of 22q11.2 deletion syndrome with right microphthalmia and left corneal staphyloma**
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- LXI. **Novel homozygous mutations in the osteoprotegerin gene TNFRSF11B in two unrelated patients with juvenile Paget's disease**
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