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Education Information

Doctorate, Hacettepe University, Sağlık Bilimleri Enstitüsü, Turkey 2015 - Continues

Post Doctorate of Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Turkey 2012 - 2018

Expertise In Medicine, Diğer (Kurumlar,hastaneler Vb.), Ankara Dr.Sami Ulus Çocuk Sağlığı ve Hastalıkları Eğitim Araştırma Hastanesi, Çocuk Sağlığı ve Hastalıkları, Turkey 2007 - 2012

Under Graduate, Hacettepe University, Tıp Fakültesi (Türkçe), Turkey 2000 - 2006

Research Areas

Health Sciences

Academic Titles / Tasks

Assistant Professor, Acibadem Mehmet Ali Aydınlar University, School Of Medicine, Department Of Medical Sciences, 2021 - Continues

Courses

Mendelian Inheritance, Under Graduate, 2020 - 2021

Clinical Cytogenetics, Under Graduate, 2020 - 2021

Non-mendelian Inheritance, Under Graduate, 2020 - 2021

PANEL: FROM GENOTYPE TO PHENOTYPE, Under Graduate, 2020 - 2021

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

- I. **Diagnostic yield of whole-exome sequencing in non-syndromic intellectual disability**
TAŞKIRAN Z. E. , KARAOSMANOĞLU B., KOŞUKCU C., Urel-Demir G., Akgun-Dogan Ö., Simsek-Kiper P. o. , ALİKAŞİFOĞLU M., BODUROĞLU O. K. , ÜTİNE G. E.
JOURNAL OF INTELLECTUAL DISABILITY RESEARCH, vol.65, no.6, pp.577-588, 2021 (Journal Indexed in SSCI)
- II. **Comparison of the clinical diagnostic criteria and the results of the next-generation sequence gene panel in patients with monogenic systemic autoinflammatory diseases**
Sozeri B., Demir F., SÖNMEZ H. E. , Karadag S. G. , Demirkol Y. K. , Dogan Ö., Doganay H. L. , Ayaz N. A.
CLINICAL RHEUMATOLOGY, vol.40, no.6, pp.2327-2337, 2021 (Journal Indexed in SCI)
- III. **Does sampling saliva increase detection of SARS-CoV-2 by RT-PCR? Comparing saliva with oronasopharyngeal swabs**
Dogan Ö., Kose B., Agaoglu N. B. , Yildiz J., Alkurt G., Demirkol Y. K. , Irvem A., Doganay G. D. , Doganay L.

JOURNAL OF VIROLOGICAL METHODS, vol.290, 2021 (Journal Indexed in SCI)

- IV. **Two Siblings with Kaufman Oculocerebrofacial Syndrome Resembling Oculoauriculovertebral Spectrum**
Urel-Demir G., Aydin B., KARAOSMANOĞLU B., Akgun-Dogan Ö., TAŞKIRAN Z. E. , Simsek-Kiper P. O. , ÜTİNE G. E. , BODUROĞLU O. K.
MOLECULAR SYNDROMOLOGY, vol.12, no.2, pp.106-111, 2021 (Journal Indexed in SCI)
- V. **Kohlschutter-Tonz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity**
Akgun-Dogan Ö., Simsek-Kiper P. O. , Taskiran E., Schossig A., ÜTİNE G. E. , Zschocke J., BODUROĞLU O. K.
JOURNAL OF CHILD NEUROLOGY, 2021 (Journal Indexed in SCI)
- VI. **The same mutation in a family with adenosine deaminase 2 deficiency**
Sozeri B., Ercan G., Dogan Ö., Yildiz J., Demir F., Doganay L.
RHEUMATOLOGY INTERNATIONAL, vol.41, no.1, pp.227-233, 2021 (Journal Indexed in SCI)
- VII. **Genetic panel screening in patients with clinically unclassified systemic autoinflammatory diseases**
Demir F., Dogan Ö., Demirkol Y. K. , Tekkus K. E. , Canbek S., Karadag S. G. , SÖNMEZ H. E. , Ayaz N. A. , Doganay H. L. , Sozeri B.
CLINICAL RHEUMATOLOGY, vol.39, no.12, pp.3733-3745, 2020 (Journal Indexed in SCI)
- VIII. **H syndrome with a novel homozygousSLC29A3mutation in two sisters**
Demir D., Karabay E. A. , Sozeri B., Gursoy F., Dogan Ö., Topaktas E., Zindanci I.
PEDIATRIC DERMATOLOGY, vol.37, no.6, pp.1135-1138, 2020 (Journal Indexed in SCI)
- IX. **Cafe noir spots: a feature of familial progressive hyper- and hypopigmentation**
Gulseren D., Guleray N., Akgun-Dogan Ö., Simsek-Kiper P. O. , Utine E. G. , ALİKAŞİFOĞLU M., Ersoy-Evans S.
JOURNAL OF THE EUROPEAN ACADEMY OF DERMATOLOGY AND VENEREOLOGY, vol.34, no.2, 2020 (Journal Indexed in SCI)
- X. **Further Phenotypic Delineation of Partial Trisomy 17q and Partial Monosomy 20q due to Rare t(17;20)**
Urel-Demir G., Akgun-Dogan Ö., OĞUZ S., Guleray-Lafci N., Simsek-Kiper P. O. , Utine G. E. , ALİKAŞİFOĞLU M., BODUROĞLU O. K.
MOLECULAR SYNDROMOLOGY, vol.11, no.1, pp.38-42, 2020 (Journal Indexed in SCI)
- XI. **Peters Plus syndrome: a recognizable clinical entity**
Demir G. U. , Lafci N. G. , Dogan Ö., Kiper P. O. S. , ÜTİNE G. E.
TURKISH JOURNAL OF PEDIATRICS, vol.62, no.1, pp.136-140, 2020 (Journal Indexed in SCI)
- XII. **ADA2 deficiency in a patient with Noonan syndrome-like disorder with loose anagen hair: The co-occurrence of two rare syndromes**
Akgun-Dogan Ö., Simsek-Kiper P. O. , TAŞKIRAN Z. E. , Lissewski C., Brinkmann J., Schanze D., GÖÇMEN R., ÇAĞDAŞ AYVAZ D. N. , BİLGİNER Y., ÜTİNE G. E. , et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.12, pp.2474-2480, 2019 (Journal Indexed in SCI)
- XIII. **A Case of Leukoencephalopathy and Small Vessels Disease Caused by a Novel HTRA1 Homozygous Mutation**
GÜNDÜZ T., Demirkol Y., Dogan Ö., Demir S., Akcakaya N. H.
JOURNAL OF STROKE & CEREBROVASCULAR DISEASES, vol.28, no.11, 2019 (Journal Indexed in SCI)
- XIV. **Ophthlmo-acromelic syndrome in an infant**
Urel-Demir G., TAŞKIRAN Z. E. , Akgun-Dogan Ö., Simek-Kiper P. O. , ÜTİNE G. E.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.7, 2019 (Journal Indexed in SCI)
- XV. **Hyperphosphatasia with mental retardation syndrome type 4 In two siblings-expanding the phenotypic and mutational spectrum**
Dogan Ö., Demir G. U. , KOŞUKCU C., TAŞKIRAN Z. E. , Simsek-Kiper P. O. , ÜTİNE G. E. , ALİKAŞİFOĞLU M., BODUROĞLU O. K.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.6, 2019 (Journal Indexed in SCI)
- XVI. **A novel NKX3-2 mutation associated with perinatal lethal phenotype of spondylo-megaepiphyseal-metaphyseal dysplasia in a neonate.**
ŞİMŞEK KİPER P. Ö. , KOŞUKCU C., Akgun-Dogan Ö., 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 (Journal Indexed in SCI Expanded)
- XVII. **Further expansion of the mutational spectrum of spondylo-meta-epiphyseal dysplasia with abnormal calcification**
Urel-Demir G., Simsek-Kiper P. O. , Akgun-Dogan Ö., GÖÇMEN R., Wang Z., Matsumoto N., Miyake N., Utine G. E. , Nishimura G., Ikegawa S., et al.
JOURNAL OF HUMAN GENETICS, vol.63, no.9, pp.1003-1007, 2018 (Journal Indexed in SCI)
- XVIII. **Further delineation of spondyloepimetaphyseal dysplasia Faden-Alkuraya type: A RSPRY1-associated spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis**
Simsek-Kiper P. O. , Taskiran E. Z. , KOŞUKCU C., Urel-Demir G., Akgun-Dogan Ö., YILMAZ G., ÜTİNE G. E. , Nishimura G., BODUROĞLU O. K. , ALİKAŞİFOĞLU M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.176, no.9, pp.2009-2016, 2018 (Journal Indexed in SCI)
- XIX. **Prenatal and Postnatal Follow-up in Trisomies 13 and 18: A 20-Year Experience in a Tertiary Center**
Dogan Ö., Demir G. U. , Arslan U., Simsek-Kiper P. O. , ÜTİNE G. E. , ALİKAŞİFOĞLU M., BODUROĞLU O. K.
AMERICAN JOURNAL OF PERINATOLOGY, vol.35, no.5, pp.427-433, 2018 (Journal Indexed in SCI)
- XX. **Fragile x-associated premature ovarian failure in a large Turkish cohort: Findings of Hacettepe Fragile X Registry.**
ÜTİNE G. E. , ŞİMŞEK KİPER P. Ö. , Akgun-Dogan Ö., ÜREL DEMİR G., Alanay Y., Aktas D., Boduroglu K., Tuncbilek E., ALİKAŞİFOĞLU M.
European journal of obstetrics, gynecology, and reproductive biology, vol.221, pp.76-80, 2018 (Journal Indexed in SCI Expanded)
- XXI. **Anauxetic dysplasia: A rare clinical entity**
Akgun-Dogan Ö., Simsek-Kiper P. O. , ÜTİNE G. E. , BODUROĞLU O. K.
TURKISH JOURNAL OF PEDIATRICS, vol.60, no.1, pp.89-93, 2018 (Journal Indexed in SCI)
- XXII. **HERC1 mutations in idiopathic intellectual disability**
ÜTİNE G. E. , TAŞKIRAN Z. E. , KOŞUKCU C., KARAOSMANOĞLU B., Guleray N., Dogan Ö., Kiper P. O. S. , BODUROĞLU O. K. , ALİKAŞİFOĞLU M.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.60, no.5, pp.279-283, 2017 (Journal Indexed in SCI)
- XXIII. **Coexistence of Trisomy.. and SRY (-) XX Ovotesticular Disorder of Sex Development**
Demir G. U. , Dogan Ö., Kiper P. O. S. , ÜTİNE G. E. , BODUROĞLU O. K. , Gucer S., ALİKAŞİFOĞLU M.
FETAL AND PEDIATRIC PATHOLOGY, vol.36, no.6, pp.445-451, 2017 (Journal Indexed in SCI)

Articles Published in Other Journals

- I. **The musculoskeletal system manifestations in children with familial Mediterranean fever**
Demir F., Bolac L. G. , Merter T., Canbek S., Dogan Ö., Demirkol Y. K. , Yildiz J., Doganay H. L. , Sozeri B.
NORTHERN CLINICS OF ISTANBUL, vol.7, no.5, pp.438-442, 2020 (Journal Indexed in ESCI)
- II. **The origin of SARS-CoV-2 in Istanbul: Sequencing findings from the epicenter of the pandemic in Turkey**
Karacan I., Akgun T. K. , Agaoglu N. B. , Irvem A., Alkurt G., Yildiz J., Kose B., Ozel A. S. , Altunal L. N. , Can N. D. , et al.
NORTHERN CLINICS OF ISTANBUL, vol.7, no.3, pp.203-209, 2020 (Journal Indexed in ESCI)
- III. **Responding to COVID-19 in Istanbul: Perspective from genomic laboratory**
Doganay L., Agaoglu N. B. , Irvem A., Alkurt G., Yildiz J., Kose B., Demirkol Y. K. , Dogan Ö., Dinler Doğanay G.
NORTHERN CLINICS OF ISTANBUL, vol.7, no.3, pp.311-312, 2020 (Journal Indexed in ESCI)
- IV. **Varicella-Related Hospitalizations Among Immunocompetent and Immunocompromised Children in Pre-Vaccine Era: A Tertiary Care Center Experience in Turkey**
Dogan Ö., Topcu S., Tanir N. G.
JOURNAL OF PEDIATRIC RESEARCH, vol.5, no.1, pp.11-16, 2018 (Journal Indexed in ESCI)
- V. **A Diagnosis to Consider in an Adult Patient with Facial Features and Intellectual Disability: Williams Syndrome**

Dogan Ö., Kiper P. O. S. , ÜTİNE G. E. , ALİKAŞİFOĞLU M., BODUROĞLU O. K.

KOREAN JOURNAL OF FAMILY MEDICINE, vol.38, no.2, pp.102-105, 2017 (Journal Indexed in ESCI)

VI. **Monogenic diabetes Monogenik diyabet**

Doğan Ö., Utine G. E. , Alikasıfoğlu A.

Cocuk Sagligi ve Hastaliklari Dergisi, vol.59, no.3, pp.140-151, 2016 (Refereed Journals of Other Institutions)

Refereed Congress / Symposium Publications in Proceedings

- I. **Gilbert Sendromu: Genetik Test Yapılması Gerekli midir?**
GERENLİ N., KENDİR DEMİRKOL Y., AKGÜN DOĞAN Ö.
63. TÜRKİYE MİLLİ PEDİATRİ KONGRESİ, Gazi Mağosa, Cyprus (Kktc), 30 October 2019
- II. **A novel mutation in COL2A1 leading to spondyloepiphyseal dysplasia congenita**
Demirkol Y. K. , Dogan Ö. , Say M. , Akgun T. K. , Doganay L.
52nd Conference of the European-Society-of-Human-Genetics (ESHG), Gothenburg, Sweden, 15 - 18 June 2019, vol.27, pp.1273-1274
- III. **Vascular ehlers-danlos syndrome**
Agaoglu N., Demirkol Y. K. , Dogan Ö. , Say M. , Akgun T. K. , Doganay H.
52nd Conference of the European-Society-of-Human-Genetics (ESHG), Gothenburg, Sweden, 15 - 18 June 2019, vol.27, pp.1279
- IV. **Stuve-Wiedemann syndrome: a rare clinical entity**
Dogan Ö. , Demirkol Y. K. , Say M. , Akgun T. K. , Doganay L.
52nd Conference of the European-Society-of-Human-Genetics (ESHG), Gothenburg, Sweden, 15 - 18 June 2019, vol.27, pp.1300-1301
- V. **GENETIC SCREENING IN PATIENTS WITH UNDIFFERENTIATED PERIODIC FEVER SYNDROME**
Demir F., Dogan Ö. , Kendirci Y. , Tekkus K. E. , Canbek S., AKTAY AYAZ N., Doganay L., Sozeri B.
Annual European Congress of Rheumatology (EULAR), Madrid, Spain, 12 - 15 June 2019, vol.78, pp.974
- VI. **Ophthalamo-acromelic syndrome in an infant**
Demir G. U. , TAŞKIRAN Z. E. , DOĞAN Ö. , Kiper P. O. S. , ÜTİNE G. E.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.959
- VII. **Homozygous novel variant in MUT in a patient with intellectual disability without metabolic derangement**
Utine E. G. , TAŞKIRAN Z. E. , KOŞUKCU C., DOĞAN Ö. , Kiper O. P. S. , BODUROĞLU O. K. , ALİKAŞİFOĞLU M.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.932
- VIII. **RSPRY1-associated skeletal dysplasia: Spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis**
Simsek-Kiper P. O. , TAŞKIRAN Z. E. , KOŞUKCU C., Akgun-Dogan Ö. , YILMAZ G., ÜTİNE G. E. , Nishimura G., BODUROĞLU O. K. , ALİKAŞİFOĞLU M.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.249-250
- IX. **Absence of major eye malformations further expands the phenotype of SOX2 deletions**
Guleray N., Demir G. U. , DOĞAN Ö. , Kiper P. O. S. , ÜTİNE G. E. , ALİKAŞİFOĞLU M.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.500-501

Memberships / Tasks in Scientific Organizations

Çocuk Genetik hastalıkları Derneği, Member, 2016 - Continues, Turkey

Citations

Total Citations (WOS):51

h-index (WOS):4

Scholarships

Visiting Scholar Programme, Fulbright Program, 2021 - Continues