



DOÇ. ÖZLEM ÖZ

Kişisel Bilgiler

Eposta: ozlemez@harran.edu.tr

Birimi : Tıbbi Genetik

Dahili : 4559

Makaleler (YOKSIS)

1 46,XX erkek sendromlu bir olgu

ONUR CURA DUYGU,ÇANKAYA TUFAN,ÖZ ÖZLEM,BORA ELÇİN,ÜLGENALP AYFER,ERÇAL MURAT DERYA

Pediatrici Uzmanlık Akademisi Dergisi,

2 A Novel Mutation in Neurofibromatosis Type 1 with Optic Glioma

ÖZ ÖZLEM

Oncologie,<http://dx.doi.org/10.32604/oncologie.2020.014087>

3 A Pilot Study for Investigation of Plasma Amino Acid Profile in Neurofibromatosis Type 1 Patients

ÖZ ÖZLEM,KOYUNCU İSMAİL,GÖNEL ATAMAN

Combinatorial Chemistry & High Throughput

Screening,<http://dx.doi.org/10.2174/1386207323666201204143206>

4 Ailevi Akdeniz Ateşi Hastalarında MEFV Geninin NGS ile Analizi: Tek Merkez Deneyimi

DÜZKALE NESLİHAN,ÖZ ÖZLEM

Harran Üniversitesi Tıp Fakültesi Dergisi,<https://dergipark.org.tr/tr/doi/10.35440/hutfd.826687>

5 Amino acid metabolism disorders and PAH gene mutations in Southeastern Anatolia Region

ÖZ ÖZLEM, Akbulut Emiş Deniz, ERCAN KARADAĞ MÜJGAN, GÖNEL ATAMAN, KOYUNCU İSMAİL

Turkish Journal of Biochemistry,<http://dx.doi.org/10.1515/tjb-2020-0338>

6 Analytical Interference with Contrast Agents in Genetic Test Result for Thrombophilia

ÖZ ÖZLEM, GÖNEL ATAMAN

Bentham Science Publishers Ltd.,<http://dx.doi.org/10.2174/1570180820666230105164237>

7 Are H1 and H3 haplotypes of endothelial protein C receptor (PROCR) an important factor in contracting COVIDu201019?

CEYLAN MEHMET REŞAT, KANKILIÇ NAZIM, ÖZ ÖZLEM

- 7 Journal of Medical Virology,<http://dx.doi.org/10.1002/jmv.27938>
- 8 **Are Monocyte, %M and WBC a Biomarker in the First Trimester Screening Test?**
ÖZ ÖZLEM, GÖNEL ATAMAN
New Emirates Medical Journal,<http://dx.doi.org/10.2174/03666221006083724>
- 9 **Beta-thalassemia mutation types and the relationship with the demographic factors in Sanliurfa, Turkey**
AKINCI AYŞE BURCU, DEMİR YENİGÜRBÜZ FATMA, TUNÇEZ EBRU, ÖZ ÖZLEM
FAMILY PRACTICE AND PALLIATIVE CARE,<http://www.fppc.com.tr/tr/pub/issue/64686/805164>
- 10 **BTD Gene Mutations in Biotinidase Deficiency: Genotype-Phenotype Correlation**
ÖZ ÖZLEM, KARACA MERYEM, ATAŞ NURGÜL, GÖNEL ATAMAN, ERCAN KARADAĞ MÜJGAN
JCPSP-JOURNAL OF THE COLLEGE OF PHYSICIANS AND SURGEONS
PAKISTAN,<http://dx.doi.org/10.29271/jcpsp.2021.07.780>
- 11 **Clinical and exome sequencing findings in seven children with Bardet-Biedl syndrome from Turkey**
GÜMÜŞ EVREN, TUNCEZ EBRU, ÖZ ÖZLEM, SAKA GÜVENÇ MERVE
Annals of Human Genetics,<http://dx.doi.org/10.1111/ahg.12401>
- 12 **Contribution of genotypes in Prothrombin and Factor V Leiden to COVID-19 and disease severity in patients at high risk for hereditary thrombophilia**
SEZER ÖZLEM, KIRAZ ASLIHAN, ALEMDAR ADEM, ERGÖREN MAHMUT ÇERKEZ, DÜNDAR MUNİS, GÜRKAN HAKAN, TEMEL ŞEHİME GÜLSÜN
Journal of Medical Virology,<http://dx.doi.org/10.1002/jmv.28457>
- 13 **Effects of a PPAR gamma receptor agonist and an angiotensin receptor antagonist on aortic contractile responses to alpha receptor agonists in diabetic and or hypertensive rats**
TUĞRUL İBRAHİM, DOST TURHAN, DEMİR ÖMER, GÖKALP FİLİZ, ÖZ ÖZLEM, GİRİT NECİP, BİRİNCİOĞLU MUSTAFA
Cardiovascular Journal of Africa,http://cvja.co.za/onlinejournal/vol27/vol27_issue3/#42/z
- 14 **Effects of APOE, ACE, PICALM, and CYP2D6 Gene Variants on Alzheimer's Disease**
ÖZ ÖZLEM, YENER GÖRSEV, BORA ELÇİN, ÇANKAYA TUFAN, ATAMAN ESRA, ERÇAL MURAT DERYA, ÜLGENALP AYFER
Current Psychiatry Research and Reviews,<http://dx.doi.org/10.2174/2666082217666210907104004>
- 15 **Evaluation of the efficiency of serum biotinidase activity as a newborn screening test in Turkey**
ERCAN KARADAĞ MÜJGAN, AKBULUT EMİŞ DENİZ, ÖZ ÖZLEM, ATAŞ NURGÜL, KARACA MERYEM, YILMAZ FATMA MERİÇ
Journal of Pediatric Endocrinology and Metabolism,<https://www.degruyter.com/view/journals/jpem/ahead-of-print/article-10.1515-jpem-2020-0382/article-10.1515-jpem-2020-0382.xml>
- 16 **Evaluation of Y chromosome microdeletions and chromosomal anomalies in infertile men**
ÖZ ÖZLEM
Hormone Molecular Biology and Clinical Investigation,<http://dx.doi.org/10.1515/hmbci-2021-0003>
- 17 **Genotype-Phenotype Correlation of Novel NF1 Gene Variants Detected by NGS in Patients with Neurofibromatosis Type 1**
ÖZ ÖZLEM

- 17 Neurochemical Journal,<http://dx.doi.org/10.1134/s1819712421040097>
- 18 **Identification of PSEN1 and PSEN2 Gene Variants and Clinical Findings with the Literature**
Nadide Cemre Randa, BORA ELÇİN, ATAMAN ESRA, ÖZ ÖZLEM, YENER GÖRSEV, ÜLGENALP AYFER
International Journal of Neurodegenerative Disorders,<https://www.clinmedjournals.org/articles/ijnd/international-journal-of-neurodegenerative-disorders-ijnd-2-007.php?jid=ijnd>
- 19 **Investigation of BRCA2 Gene K3326X Variant in Patients with Breast and Ovarian Cancer by Next-Generation Sequencing Technique**
DÜZKALE NESLİHAN, KARAÇİN CENGİZ, YÖRÜBULUT SERAP, ERDEM HAKTAN BAĞIŞ, ÖZ ÖZLEM, ALTINBAŞ MUSTAFA
Acta Oncologica Turcica,https://jag.journalagent.com/aot/pdfs/AOT-68815-ORIGINAL_ARTICLE-DUZKALE.pdf
- 20 **Investigation of Hereditary Cancer Predisposition Genes of Patients with Colorectal Cancer: Single-centre Experience**
DÜZKALE NESLİHAN, ÖZ ÖZLEM, TAŞKIN TÜRKMENOĞLU TUĞBA, ÇETİNKAYA KADİR, EREN TÜLAY, YALÇIN ŞUAYİP
Journal of the College of Physicians and Surgeons Pakistan,<http://dx.doi.org/10.29271/jcpsp.2021.07.811>
- 21 **Kronik Miyeloproliferatif Hastalık Olgularında JAK2 V617F Mutasyon Sıklığı**
ÖZ ÖZLEM
Harran Üniversitesi Tıp Fakültesi Dergisi,<http://dx.doi.org/10.35440/hutfd.645383>
- 22 **Nöromusküler Hastalık Genleri ve Moleküler Tanı Yaklaşımı: Duchenne Musküler Distrof**
ÖZ ÖZLEM, ÜLGENALP AYFER
Türkiye Klinikleri Pediatric Sciences,<https://www.turkiyeklinikleri.com/article/en-noromuskuler-hastalik-genleri-ve-molekuler-tani-yaklasimi-duchenne-muskuler-distrofi-67952.html>
- 23 **Rare Pericentrin (PCNT) gene mutation detected in a patient with microcephalic osteodysplastic primordial dwarfism in Turkey**
ÖZ ÖZLEM, GÖNEL ATAMAN
KUWAIT MEDICAL JOURNAL,<https://www.kmj.org.kw/current-issue>
- 24 **Response to Letter to the Editor: "Atrioventricular canal defect is an infrequent congenital heart disease that can be observed in Bardet-Biedl syndrome"**
GÜMÜŞ EVREN, TUNÇEZ EBRU, ÖZ ÖZLEM, SAKA GÜVENÇ MERVE
Annals of Human Genetics,<http://dx.doi.org/10.1111/ahg.12420>
- 25 **The Association Between Parameters of Erythrocytes Morphology and Thrombophilia-Related Mutations**
ÖZ ÖZLEM, GÖNEL ATAMAN
Reviews on Recent Clinical Trials,<http://dx.doi.org/10.2174/1574887116666211123092603>
- 26 **The link between miR-96 levels and the developmental dysplasia of the hip**
GÜMÜŞ EVREN, TEMİZ EBRU, DEMİRCAN TURAN, SARIKAYA BARAN, Yüksekdağ Özgür, BOZKURT CELAL, KOYUNCU İSMAİL, ÖZ ÖZLEM
Gazi Medical Journal,<https://medicaljournal.gazi.edu.tr/index.php/GMJ/article/view/3156/2580>

27 The Mutation of CD27 Deficiency Presented With Familial Hodgkin Lymphoma and a Review of the Literature

KÖSE DOĞAN, GÜZELÇİÇEK AHMET, ÖZ ÖZLEM, erdem arzu, Haliloğlu Yeşim, Witzel Maximilian, Klein Christoph, ÜNAL EKREM

Journal of Pediatric Hematology/Oncology, <http://dx.doi.org/10.1097/mp.0000000000002453>

28 Trizomi 13 ve 18 ile ilişkili prenatal sonografik bulgular; tek bir merkezde prenatal tanı konmuş olguların sunumu

EKMEKÇİ Emre, ÖZ ÖZLEM, TUNÇEZ Ebru, ERCAN Fedi, DEMİR EMİNE

Pamukkale Medical Journal, <http://dx.doi.org/10.31362/patd.757219>

Bildiriler (YOKSIS)

1 A NOVEL MUTATION IN PERICENTRIN (PCNT) GENE CAUSED MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM

ÖZ ÖZLEM, ÇATLI GÖNÜL, BORA ELÇİN, ANIK AHMET, ABACI AYHAN, BÖBER ECE, ERÇAL MURAT DERYA, GİRAY BOZKAYA ÖZLEM

27th COURSE IN MEDICAL GENETICS ,

2 ANTIPROLIFERATIVE AND APOPTOTIC EFFECTS OF HYPERICUM PERFORATUM ON PEDIATRIC SARCOMA CELLS

KILIÇ EREN MEHTAP, ÖZ ÖZLEM, KOZACI LEYLA DİDEM, BİRİNCİOĞLU MUSTAFA

International Symposium on Drug Research and Development "From Chemistry to Medicine" ,

3 DEVELOPMENTAL DELAY DUE TO DE NOVO MOSAIC CHROMOSOME 14Q PARTIAL DUPLICATION

BAŞDEMİRCİ MÜŞERREF, ÖZ ÖZLEM, ZAMANİ AYŞE GÜL

Erciyes Tıp Genetik Günleri 2018 , <https://erciyesmedj.com/jvi.aspx?un=EMJ-57984&volume=40&supp=2>

4 EDWARDS SENDROMU BENZERİ BULGULAR İLE PREZENTE OLAN 46,XX,DER(11)İNS(11;18) (Q21;Q12.2Q23) KARYOTİPE SAHİP BİR OLGU

BURHANLI TÜRKAN, DÜZ MEHMET BUĞRAHAN, AKIN DUMAN TUĞBA, ÖZYAVUZ ÇUBUK PELİN, ÖZ ÖZLEM, ERDEM FATİH MEHMET

Uluslararası Katılımlı 13. Ulusal Tıbbi Genetik Kongresi ,
<https://medicaljournal.gazi.edu.tr/index.php/GMJ/article/view/1978/1381>

5 ERKEN BAŞLANGIÇLI ALZHEİMER TANILI TÜRK HASTALARDA APP, PSEN1 VE PSEN2 MUTASYON SIKLIĞI VE HASTALARIN KARŞILAŞTIRILMASI

RANDA NADİDE CEMRE, BORA ELÇİN, ATAMAN ESRA, ÖZ ÖZLEM, YENER GÖRSEV, ÜLGENALP AYFER

Uluslararası Katılımlı 13. Ulusal Tıbbi Genetik Kongresi ,
<https://medicaljournal.gazi.edu.tr/index.php/GMJ/article/view/1978/1381>

6 Evaluation of the case with a new mutation in the PTPN11 gene in the light of the literature

ÖZ ÖZLEM

International Eurasian Conference on Biotechnology and Biochemistry ,
https://www.biotechbiochem.org/bildiri%20taslaklar%C4%B1/Proceeding_Book_BioTechBioChem_2020.pdf

7 Experimental Approach to Drug Interference in Biotidinase Assay with LC-MS / MS

ÖZ ÖZLEM, GÖNEL ATAMAN

3rd International Congress on Analytical and Bioanalytical Chemistry ,
http://icabc2021.firat.edu.tr/documentation/Abstract_Book3rdICABC.pdf

7

8 HBB GENİNDE NADİR GÖRÜLEN IVS-II-745 (C>G) VE 5'UTR +20 (C>T) MUTASYONLARININ BİRLİKTELİĞİ

AKINCI AYŞE BURCU, DEMİR YENİGÜRBÜZ FATMA, ÖZ ÖZLEM, TUNÇEZ EBRU

6. Ulusal Pediatrik Hematoloji Sempozyumu ,

9 Hypericum Perforatum'un HT115 Kolorektal Karsinoma Hücreleri Üzerinde Antiproliferatif ve Apoptotik Etkileri

KILIÇ EREN MEHTAP, ÖZ ÖZLEM, ural mürüvvet, BİRİNCİOĞLU MUSTAFA

XII. Ulusal Tıbbi Biyoloji ve Genetik Kongresi ,

10 Investigating consanguineous families from Turkey to identify autosomal recessive neurodevelopmental disorders

GÜMÜŞLÜ KUDRET ESEN, GÜMÜŞ EVREN, ÖZ ÖZLEM, ÖZKAN MELİS, KARAER KADRI, EKİCİ ARİF, YILDIZ EDİBE PEMBEGÜL, AYDINLI NUR, REİS ANDRE

European Society of Human Genetics 2021 ,

<https://www.abstractsonline.com/pp8/#!/10372/presentation/788>

11 Investigation of Chromosomal Anomalies and Copy Number Variations in Children Diagnosed with Autism Spectrum

KILIÇASLAN FETHİYE, ÖZ ÖZLEM

14th International Congress on Psychopharmacology & International Symposium on Child and Adolescent Psychopharmacology , <https://www.psychopharmacology2023.org/?p=proceedings-page>

12 Investigation of Chromosomal Anomalies and Copy Number Variations in Children Diagnosed with Autism Spectrum Disorder by Array Cgh Method

KILIÇASLAN FETHİYE, ÖZ ÖZLEM

14th International Congress on Psychopharmacology & International Symposium on Child and Adolescent Psychopharmacology , <https://www.psychopharmacology2023.org/program/sozel/>

13 Konjenital Nötropenide HAX1 Geninde Yeni Bir Mutasyon Tanımlanan Olgu

ÖZ ÖZLEM, DEMİR YENİGÜRBÜZ FATMA, AKINCI AYŞE BURCU

3. HEMATOLOJİK GENETİK SEMPOZYUMU ,

14 Kraniosinostozlu Olgu

ONUR CURA DUYGU, ÇANKAYA TUFAN, Uyanık Bülent, ÖZ ÖZLEM, BORA ELÇİN, GİRAY BOZKAYA ÖZLEM, ÜLGENALP AYFER, ERÇAL MURAT DERYA

2. Nörometabolik Dismorfoloji Sempozyumu ,

15 Mikrosefali primordial dwarfizm MOPD sendromunda perisentrin PCNT geninde yeni bir mutasyon Olgu sunumu

ÖZ ÖZLEM, ÇATLI GÖNÜL, BORA ELÇİN, ANIK AHMET, ABACI AYHAN, BÖBER ECE, ERÇAL MURAT DERYA, GİRAY BOZKAYA ÖZLEM

1. Ulusal Çocuk Genetik Sempozyumu ,

16 Multipl Konjenital Anomali Ve Motor Mental Retardasyonlu Olgu

ÖZ ÖZLEM, BORA ELÇİN, ÇANKAYA TUFAN, ONUR CURA DUYGU, Aksel Kılıçaslan Özge, GİRAY BOZKAYA ÖZLEM, ERÇAL MURAT DERYA

2. Nörometabolik Dismorfoloji Sempozyumu ,

17 Nörofibromatozis Tip 1 Hastalarında Tam Kan Sayımı Parametrelerinin Değerlendirilmesi

ÖZ ÖZLEM, ERCAN KARADAĞ MÜJGAN

- 17 4th International Eurasian Conference on Science, Engineering and Technology ,
[https://www.eurasiansciencetech.org/bildiri%20taslaklar
%C4%B1/Proceeding_Book_EurasianSciEnTech_2022.pdf](https://www.eurasiansciencetech.org/bildiri%20taslaklar%C4%B1/Proceeding_Book_EurasianSciEnTech_2022.pdf)
- 18 **Nörofibromatozis Tip 1 Sendromunda Yeni Bir Mutasyon Tanımlanan Olgu**
ÖZ ÖZLEM, TUNÇEZ EBRU, GÜNEY İLKER
12.Ulusal Tıbbi Genetik Kongresi ,
- 19 **Otozomal Resesif Konjenital İktiyozlu Yenidoğanda ALOX12B Geninde Nadir Bir Mutasyon**
ÖZ ÖZLEM
1. Bursa Uluslararası Katılımlı Genetik Günleri Dermatogenetik Sempozyumu ,
<https://medicaljournal.gazi.edu.tr/index.php/GMJ/article/view/2391/2110>
- 20 **PPAR-gama reseptör agonisti ve anjiotensin reseptör blokörlerinin izole sıçan aortasında alfa-1 ve alfa-2 adrenoseptör agonist cevapları üzerine olan etkileri**
TUĞRUL İBRAHİM,DEMİR ÖMER,Gökalp Filiz,ÖZ ÖZLEM,GİRİT NECİP,DOST TURHAN,BİRİNCİOĞLU MUSTAFA
21. Ulusal Farmakoloji Kongresi ,
- 21 **PRENATAL DIAGNOSIS OF DOUBLE TRISOMY OF EDWARDS SYNDROME AND KLINEFELTER SYNDROME**
ÖZ ÖZLEM, BAŞDEMİRCİ MÜŞERREF, EKMEKÇİ EMRE, ZAMANI AYŞE GÜL
Erciyes Tıp Genetik Günleri 2018 , <https://erciyesmedj.com/jvi.aspx?un=EMJ-57984&volume=40&supp=2>
- 22 **Prenatal Tanıda Tespit Edilmiş Cinsiyet Kromozom Anomalileri-Olgu Sunumu**
ÖZ ÖZLEM, ÇANKAYA TUFAN, BORA ELÇİN, ÜLGENALP AYFER, ERÇAL MURAT DERYA
10.Ulusal Tıbbi Genetik Kongresi ,
- 23 **The Effect of Plasminogen Activator Inhibitor-1 (PAI-1) (Serpine-1) 4G/5G Promoter Polymorphism on Predicting the COVID-19 Clinic.**
KANKILIÇ NAZIM, CEYLAN MEHMET REŞAT, ÖZ ÖZLEM
17th International Congress of Update in Cardiology and Cardiovascular Surgery. 5-7 November, 2021. , https://cms.galenos.com.tr/Uploads/Article_49659/ejcm-9-0-En.pdf
- 24 **The genetic landscape of neurodevelopmental disorders in a large cohort of multiplex consanguineous families from Turkey**
GÜMÜŞLÜ KUDRET ESEN, GÜMÜŞ EVREN, ÖZ ÖZLEM, ÖZKAN MELİS, KARAER KADRI, EKİCİ ARİF, YILDIZ EDİBE PEMBEGÜL, rauch anita, AYDINLI NUR, REİS ANDRE
ASHG 2022 ,
- 25 **The Role of Whole Exome Sequencing in Diagnosis of Autism and Developments in Genetic Therapy**
ÖZ ÖZLEM
14th International Congress on Psychopharmacology & International Symposium on Child and Adolescent Psychopharmacology , <https://www.psychopharmacology2023.org/>
- 26 **Two novel KMT2D variants in a series of 7 patients with Kabuki syndrome**
ÖZÇELİK FIRAT, DUMAN NİLGÜN, KIRAZ ASLIHAN, ÖZ ÖZLEM, GÖKÇE NURİYE, ÇİÇEK DİLEK, PER HÜSEYİN, ÖZKUL YUSUF, DÜNDAR MUNİS
6. Uluslararası Katılımlı Erciyes Tıp Tıbbi Genetik Kongresi ,
<https://medicaljournal.gazi.edu.tr/index.php/GMJ>

