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Kişisel Bilgiler

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SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- I. Letter to the Editor regarding “New cases of recently described Thauvin-Robinet-Faivre syndrome with a novel homozygous FIBP gene variant” by Kılıç and Koşukçu, “An investigation of the etiology and follow-up findings in 35 children with overgrowth syndromes, including biallelic SUZ12 variant” by Yüksel Ülker et al. and “Expanding the phenotype and genotype in Thauvin-Robinet-Faivre syndrome: A new patient with a novel variant and additional clinical findings” by Duzenli et al.
DÜZENLİ T., SEZER A., PERÇİN F. E.
American Journal of Medical Genetics, Part A, cilt.194, sa.4, 2024 (SCI-Expanded)
- II. Long-term follow-up and novel variant in Suleiman-El-Hattab syndrome: Expanding the genotypic and clinical spectrum of a rare neurodevelopmental disorder
SEZER A., KAYHAN G., Percin F. E.
European Journal of Medical Genetics, cilt.66, sa.9, 2023 (SCI-Expanded)
- III. Expanding the phenotype and genotype in Thauvin-Robinet-Faivre syndrome: A new patient with a novel variant and additional clinical findings
DÜZENLİ T., Sezer A., KAYHAN G., ARSLAN A., Percin F. E.
American Journal of Medical Genetics, Part A, cilt.191, sa.8, ss.2232-2239, 2023 (SCI-Expanded)
- IV. A homozygous missense variant in the WRN gene segregating in a family with progressive pulmonary failure with recurrent spontaneous pneumothorax and interstitial lung disease
Sezer A., KAYHAN G., RAMASLI GÜRSOY T., ŞİŞMANLAR EYÜBOĞLU T., PERÇİN F. E.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.191, sa.1, ss.220-227, 2023 (SCI-Expanded)
- V. A de novo heterozygous HOXA11 variant in a patient with mesomelic dysplasia with urogenital abnormalities
Sezer A., Perçin F. E., Kazan H. H., Kayhan G., Akturk M. Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.188, sa.6, ss.1890-1895, 2022 (SCI-Expanded)
- VI. An infant with two de novo variants causing recessive and dominant disorders: Diagnostic challenge
Sezer A., Oğutlu O. B. G., TÜRKYILMAZ Z., GÜCÜYENER K., KAYHAN G., PERÇİN F. E.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.28, ss.895, 2020 (SCI-Expanded)
- VII. Warburg Micro Syndrome 1 due to Segmental Paternal Uniparental Isodisomy of Chromosome 2 Detected by Whole-Exome Sequencing and Homozygosity Mapping
Sezer A., Kayhan G., Koc A., Ergün M. A., Perçin F. E.
CYTOGENETIC AND GENOME RESEARCH, cilt.160, sa.6, ss.309-315, 2020 (SCI-Expanded)
- VIII. 15-LOX-1 has diverse roles in the resensitization of resistant cancer cell lines to doxorubicin
Kazan H. H., Urfali-Mamatoglu C., Yalcin G. D., Bulut O., SEZER A., BANERJEE S., GÜNDÜZ U.
JOURNAL OF CELLULAR PHYSIOLOGY, cilt.235, sa.5, ss.4965-4978, 2020 (SCI-Expanded)
- IX. Hypopigmented patches in Roberts/SC phocomelia syndrome occur via aneuploidy susceptibility
SEZER A., KAYHAN G., Zenker M., Percin E. F.
EUROPEAN JOURNAL OF MEDICAL GENETICS, cilt.62, sa.12, 2019 (SCI-Expanded)
- X. Congenital generalized lipodystrophy type 4-New mutation in the CAVIN1 gene
DÖĞER E., SEZER A., Ugurlu A. K., Akbas E. D., Percin F. E., BİDECİ A., Camurdan O., CİNNAZ P.

Diğer Dergilerde Yayınlanan Makaleler

- I. **SMN1 gen delesyonu dışlanmış Spinal Musküler Atrofi ön tanılı çocuklarda etiyolojinin tüm ekzom dizi analizi verilerine dayanarak retrospektif olarak araştırılması**
SEZER A., DEMİR E., KAYHAN G., ERGÜN M. A., TUĞ E.
Gazi Medical Journal, 2020 (ESCI)
- II. **A report of two siblings diagnosed with Cutis Laxa**
GÜNDÖĞDU ÖĞÜTLÜ Ö. B., SEZER A., DEMİRBAŞ M. H., KAYHAN G., PERÇİN F. E.
Gazi Medical Journal, cilt.31, sa.2, 2020 (ESCI)
- III. **Two patients with Epidemolysis Bullosa**
SAVAŞ A., SEZER A., KAYHAN G., ADİŞEN E., PERÇİN F. E.
Gazi Medical Journal, cilt.31, sa.2, 2020 (ESCI)
- IV. **Two new patients diagnosed with Trichothiodystrophy type 1**
Sezer A., Kayhan G., Güçüyener K., Bideci A., Perçin F. E.
Gazi Medical Journal, cilt.31, sa.2, 2020 (ESCI)
- V. **A rare form of interstitial deletion of chromosome 9q21.33q22.31: A case report**
SEZER A., KAYHAN G., ERGÜN M. A., PERÇİN F. E.
Gazi Medical Journal, cilt.30, sa.1, ss.1-101, 2019 (Scopus)
- VI. **ADNP Gene in the Etiology of Syndromic Autism: A case report**
SEZER A., KAYHAN G., ERGÜN M. A., PERÇİN F. E.
Gazi Medical Journal, cilt.30, sa.1, ss.1-101, 2019 (Scopus)
- VII. **Background of a carrier family with along inversion of chromosome 2 detected via karyotyping and aCGH analysis**
YİRMİBEŞ KARAOĞUZ M., SEZER A., KAYHAN G.
ERCIYES MEDICAL JOURNAL, cilt.40, sa.2, 2018 (Scopus)
- VIII. **Clinical findings of the two fetuses with the pericentric inversion of chromosome Y relevant or coincidental**
SAVAŞ A., SEZER A., KAYHAN G., YİRMİBEŞ KARAOĞUZ M.
ERCIYES MEDICAL JOURNAL, cilt.40, sa.2, 2018 (Scopus)
- IX. **Haploinsufficiency of ZNF462 gene in a patient with interstitial deletion of chromosome 9q**
Sezer A., Kayhan G., Mermer S., Ergün M. A., Perçin F. E.
ERCIYES TIP DERGİSİ, cilt.40, sa.2, ss.51, 2018 (Scopus)
- X. **A patient with two syndromes due to paternal uniparental disomy of chromosome 2 (pUPD2) related with homozygous novel mutations of the RAB3GAP1 and UNC80 genes**
Perçin F. E., Kayhan G., Sezer A., Koc A., Ergün M. A.
ERCIYES MEDICAL JOURNAL, cilt.40, sa.2, ss.69, 2018 (Hakemli Dergi)
- XI. **Lethal multiple pterygium syndrome related with RYR1 gene mutation**
KAYHAN G., SEZER A., ÖZDEMİR H., ERGÜN M. A., BAYRAM M., YİRMİBEŞ KARAOĞUZ M., PERÇİN F. E.
ERCIYES MEDICAL JOURNAL, cilt.40, sa.2, 2018 (Scopus)
- XII. **HAPLOINSUFFICIENCY OF ZNF462 GENE IN A PATIENT WITHINTERSTITIAL DELETION OF CHROMOSOME 9q**
SEZER A., KAYHAN G., MERMER S., ERGÜN M. A., PERÇİN F. E.
Erciyes Medical Journal, cilt.40, sa.2, ss.35-79, 2018 (Hakemli Dergi)
- XIII. **Lethal Multiple Pterygium Syndrome related with RYR1 gene gene mutation**
KAYHAN G., SEZER A., Ozdemir H., ERGÜN M. A., BAYRAM M., YİRMİBEŞ KARAOĞUZ M., PERÇİN F. E.
ERCIYES MEDICAL JOURNAL, cilt.40, sa.2, 2018 (Scopus)
- XIV. **LETHAL MULTIPLE PTERYGIUM SYNDROME RELATED WITHRYR1 GENE MUTATION**
KAYHAN G., SEZER A., ÖZDEMİR H., ERGÜN M. A., BAYRAM M., YİRMİBEŞ KARAOĞUZ M., PERÇİN F. E.

- Erciyes Medical Journal, cilt.40, sa.2, ss.70-73, 2018 (Hakemli Dergi)
- XV. A PATIENT WITH TWO SYNDROMES DUE TO PATERNAL UNIPARENTAL DISOMY OF CHROMOSOME 2 (pUPD2) RELATED WITH HOMOZYGOUS NOVEL MUTATIONS OF THERAB3GAP1 AND UNC80 GENES
PERÇİN F. E., KAYHAN G., SEZER A., KOÇ A., ERGÜN M. A.
ERCIYES MEDICAL JOURNAL, cilt.40, sa.2, ss.35-79, 2018 (Hakemli Dergi)
- XVI. A report of two infertile patients with isodicentric short arm of chromosome Y
KAYHAN G., ALTAN M., SEZER A., ERGÜN M. A., YİRMİBEŞ KARAOĞUZ M.
ERCIYES MEDICAL JOURNAL, cilt.39, ss.51, 2017 (Hakemli Dergi)

Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

- I. SMN1 gen delesyonu dışlanmış Spinal Musküler Atrofi ön tanılı çocukların etiyolojinin tüm ekzom dizi analizi verilerine dayanarak retrospektif olarak araştırılması.
SEZER A., DEMİR E., KAYHAN G., ERGÜN M. A., TUĞ E.
14. Ulusal Tıbbi Genetik Kongresi "Uluslararası Katılımlı", Türkiye, 20 - 22 Kasım 2020
- II. Herediter Sapastik Parapleji: 6 olgu sunumu
Menderes D., Erçelebi H., Özbudak P., Sezer A., Perçin F. E., Demir E., Güçüyener K.
3. Nöromusküler Hastalıklar Kongresi, İzmir, Türkiye, 1 - 03 Kasım 2019, ss.25
- III. WES analizi ile otozomal resesif herediter spastik parapleji tanısı alan altı yeni hasta
Sezer A., Kayhan G., Güçüyener K., Erçelebi H., Cengiz B., Ergün M. A., Perçin F. E.
3. Nöromusküler Hastalıklar Kongresi, İzmir, Türkiye, 1 - 03 Kasım 2019, ss.122-123
- IV. SLC25A4 ilişkili bir aksiyel miyopati olgusu: Yeni bir fenotip
ERÇELEBİ H., SEZER A., ERDEM ÖZDAMAR S., ERGÜN M. A., DEMİR E.
3. Nöromusküler Hastalıklar Kongresi, 1-3 Kasım 2019, Çeşme, İzmir, Türkiye, 1 - 03 Kasım 2019
- V. KONJENİTAL JENERALİZİ LİPODISTROFİ TİP 4 - CAVIN1 GENİNDEYENİ MUTASYON
DÖĞER E., SEZER A., KILINÇ UĞURLU A., DEMET AKBAŞ E., PERÇİN F. E., BİDECİ A., ÇAMURDAN M. O., CİNNAZ P.
3. Ege Endokrin Hastalıklar ve Genetik Sempozyumu, İzmir, Türkiye, 7 - 09 Mart 2019
- VI. Dual overlapping phenotype recessively inherited due to paternal uniparental disomy of chromosome 2(pUPD2) in a patient
Perçin F. E., Kayhan G., Sezer A., Koç A., Ergün M. A.
51st Conference of Theocharis European-Society-of-Human-Genetics (ESHG) in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), Milan, İtalya, 16 - 19 Haziran 2018, cilt.27, ss.384-385
- VII. IS HYPOPIGMENTED SKIN PATCH A NEW SYMPTOM OF ROBERTS / SC PHOCOMELIA SYNDROME?
SEZER A., KAYHAN G., SARI S., PERÇİN F. E.
Erciyes Medical Genetics Days 2017, Kayseri, Türkiye, 11 - 13 Mayıs 2017, cilt.39, ss.48
- VIII. Is there any relationship between NRG1 gene duplication and cardiac findings in two prenatal cases with invdupdel(8p) syndrome?
SEZER A., BAYRAM M., KAYHAN G., ÜNAL A., ÖZDEMİR H., KARÇAALTINCABA D., YİRMİBEŞ KARAOĞUZ M.
European Human Genetics Conference, Kopenhag, Danimarka, Kopenhag, Danimarka, 27 - 30 Mayıs 2017
- IX. Disentrik ve Neosentrik Kromozomal Yeniden Düzenlenimle Sonuçlanan 4 9 Resiprokal Translokasyonlu İnfertil Olgu
TUĞ E., SEZER A., YİRMİBEŞ KARAOĞUZ M.
XII. Ulusal Tıbbi Genetik Kongresi, Türkiye, 5 - 09 Ekim 2016

Metrikler

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