

## 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., Percin 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., Percin 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., BİDEÇİ A., Camurdan O., CİNAZ P.

## Diğer Dergilerde Yayınlanan Makaleler

- I. **SMN1 gen delesyonu dışlanmış Spinal Musküler Atrofi ön tanılı çocuklarda etiolojinin 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**  
GÜNDOĞDU 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 Epidermolysis Bullosa**  
SAVAŞ A., SEZER A., KAYHAN G., ADIŞEN 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., Gücüyener 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 DERGISI, 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 WITH INTERSTITIAL 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 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)
- XIV. **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, 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 THE RAB3GAP1 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 çocuklarda 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. **Hereditör Spastik Parapleji: 6 olgu sunumu**  
Menderes D., Erçelebi H., Özbudak P., Sezer A., Perçin F. E., Demir E., Gücüyener K.  
3. Nöromusküler Hastalıklar Kongresi, İzmir, Türkiye, 1 - 03 Kasım 2019, ss.25
- III. **WES analizi ile otozomal resesif hereditör spastik parapleji tanısı alan altı yeni hasta**  
Sezer A., Kayhan G., Gücüyener 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 JENERALİZE LİPODİSTROFİ TİP 4 - CAVIN1 GENİNDEYENİ MUTASYON**  
DÖĞER E., SEZER A., KILINÇ UĞURLU A., DEMET AKBAŞ E., PERÇİN F. E., BİDECİ A., ÇAMURDAN M. O., CİNAZ 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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