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Published journal articles indexed by SCI, SSCI, and AHCI

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, vol.194, no.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, vol.66, no.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, vol.191, no.8, pp.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, vol.191, no.1, pp.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, vol.188, no.6, pp.1890-1895, 2022 (SCI-Expanded)

VI. **An infant with two de novo variants causing recessive and dominant disorders: Diagnostic challenge**
Sezer A., Ogutlu O. B. G., TÜRKYILMAZ Z., GÜCÜYENER K., KAYHAN G., PERÇİN F. E.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, pp.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, vol.160, no.6, pp.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, vol.235, no.5, pp.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, vol.62, no.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., CİNAZ P.

HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.303-304, 2019 (SCI-Expanded)

Articles Published in Other Journals

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**
GÜNDOĞDU ÖĞÜTLÜ Ö. B., SEZER A., DEMİRBAŞ M. H., KAYHAN G., PERÇİN F. E.
Gazi Medical Journal, vol.31, no.2, 2020 (ESCI)

III. **Two patients with Epidemolisis Bullosa**
SAVAŞ A., SEZER A., KAYHAN G., ADIŞEN E., PERÇİN F. E.
Gazi Medical Journal, vol.31, no.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, vol.31, no.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, vol.30, no.1, pp.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, vol.30, no.1, pp.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, vol.40, no.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, vol.40, no.2, 2018 (Scopus)

IX. **Haploınsufficıency of ZNF462 gene ın a patıent wıth ınterstıtıal deletıon of chromosome 9q**
Sezer A., Kayhan G., Mermer S., Ergün M. A., Perçin F. E.
ERCIYES TIP DERGISI, vol.40, no.2, pp.51, 2018 (Scopus)

X. **A patıent wıth two syndromes due to paternal unıparental dısomy of chromosome 2 (pUPD2) related wıth homozygous novel mutatıons of the RAB3GAP1 and UNC80 genes**
Perçin F. E., Kayhan G., Sezer A., Koc A., Ergün M. A.
ERCIYES MEDICAL JOURNAL, vol.40, no.2, pp.69, 2018 (Peer-Reviewed Journal)

XI. **Lethal multiple pterygıum 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, vol.40, no.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.
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XIII. **Lethal Multıple Pterygıum Syndrome related wıth RYR1 gene gene mutatıon**
KAYHAN G., SEZER A., Ozdemır H., ERGÜN M. A., BAYRAM M., YİRMİBEŞ KARAOĞUZ M., PERÇİN F. E.
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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, vol.40, no.2, pp.70-73, 2018 (Peer-Reviewed Journal)

XV. **A PATIENT WITH TWO SYNDROMES DUE TO PATERNALUNIPARENTAL 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, vol.40, no.2, pp.35-79, 2018 (Peer-Reviewed Journal)

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, vol.39, pp.51, 2017 (Peer-Reviewed Journal)

Refereed Congress / Symposium Publications in Proceedings

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", Turkey, 20 - 22 November 2020

II. **Herediter Sapastik Parapleji: 6 olgu sunumu**
Menderes D., Erçelebi H., Özbudak P., Sezer A., Perçin F. E., Demir E., Gücüyener K.
3. Nöromusküler Hastalıklar Kongresi, İzmir, Turkey, 1 - 03 November 2019, pp.25

III. **WES analizi ile otozomal resesif herediter 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, Turkey, 1 - 03 November 2019, pp.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, Turkey, 1 - 03 November 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, Turkey, 7 - 09 March 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, Italy, 16 - 19 June 2018, vol.27, pp.384-385

VII. **IS HYPOPIGMENTED SKIN PATCH A NEW SYMPTOM OFROBERTS / SC PHOCOMELIA SYNDROME?**
SEZER A., KAYHAN G., SARI S., PERÇİN F. E.
Erciyes Medical Genetics Days 2017, Kayseri, Turkey, 11 - 13 May 2017, vol.39, pp.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, Denmark, 27 - 30 May 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, Turkey, 5 - 09 October 2016

Metrics

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