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Published journal articles indexed by SCI, SSCI, and AHCI

I. **A novel RNPC3 gene variant expands the phenotype in patients with congenital hypopituitarism and neuropathy**
Abali Z. Y., Ili E., BAŞ F., Ozkan M. U., GÜLEÇ Ç., TOKSOY G., ÖZTÜRK A. P., KARAKILIÇ ÖZTURAN E., ASLANGER A. D., Caliskan M., et al.
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II. **Clinical and genetic spectrum from a prototype of ciliopathy: Joubert syndrome**
Uzunhan T. A., Erturk B., Aydin K., AYAZ A., Altunoglu U., Yarar M. H., Gezdirici A., Icagasioglu D. F., Ili E., UYANIK B., et al.
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III. **Expanding the genotypic and phenotypic landscapes of rhizomelic chondrodysplasia punctata type 3 (RCDP3) with two novel families, and a review of the literature**
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IV. **Phenotypic and molecular characterization of five patients with PIK3CA-related overgrowth spectrum (PROS)**
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V. **Whole-Exome Sequencing (WES) results of 50 patients with chronic kidney diseases: a perspective of Alport syndrome**
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REVISTA DA ASSOCIACAO MEDICA BRASILEIRA, vol.68, no.9, pp.1282-1287, 2022 (SCI-Expanded)

VI. **A Rare Cause of Syncope Naxos Disease Caused by Novel Homozygous Deletion in the JUP Gene**
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CIRCULATION-CARDIOVASCULAR IMAGING, no.10, pp.998-1001, 2021 (SCI-Expanded)

VII. **New Homozygous Missense MSMO1 Mutation in Two Siblings with SC4MOL Deficiency Presenting with Psoriasiform Dermatitis**
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VIII. **Promising effect of intravenous immunoglobulin therapy for epidermolysis bullosa pruriginosa**
Ertop P., Vural S., Ili E., Durmaz C. D., HEPER A., McGrath J. A., Ilgin R. H., BOYVAT A.
INTERNATIONAL JOURNAL OF DERMATOLOGY, no.7, pp.851-855, 2020 (SCI-Expanded)

IX. **Cytogenetic, Molecular, and Phenotypic Characterization of a Patient with de novo Derivative**

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Gokpinar E., Altiner Ş., KARABULUT H. G.
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X. **Association of pyrin mutations and autoinflammation with complex phenotype hidradenitis suppurativa: a case-control study**
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XI. **Optimizing the transport and storage conditions of current Good Manufacturing Practice - grade human umbilical cord mesenchymal stromal cells for transplantation (HUC-HEART Trial)**
TOPAL ÇELİKKAN F., Mungan C., Sucu M., Ulus A. T., ÇINAR Ö., Ili E., CAN A.
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Articles Published in Other Journals

I. **Genotype Phenotype Correlation of A Case Having Chromosome 3 Imbalance**
Taşdelen E., Gökpınar İli E., Altıner Ş., Ceylan A. C., Tuncalı T.
Ankara Üniversitesi Tıp Fakültesi Mecmuası, vol.74, no.3, pp.365-369, 2021 (Peer-Reviewed Journal)

II. **Hereditary Breast-Ovarian Cancer and BRCA1/BRCA2 Variants: A Single Center Experience**
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Refereed Congress / Symposium Publications in Proceedings

I. **Distrofik Epidermolizis Bülloza: Doku Mozaisizmine Giden Bir Aile Çalışması**
Sevim B., Öktem A., Gökpınar İli E., Gündüz K., Kaplan İ., Erdemli E., Kutlay N., Şanlı H.
XV. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 9 - 13 November 2022, pp.146

II. **Serebellar atrofide nadir bir genotip: GEMIN5 geninde compound heterozigot varyant saptanan bir olgu**
Gökpınar İli E., Uyur Yalçın E., Eldeş Hacıfazlıoğlu N.
XV. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 9 - 13 November 2022, pp.66

III. **A case of PTEN hamartoma tumor syndrome; a family study**
Ilgın Ruhi H., Gökpınar İli E., Güleray Lafcı N., Taşdelen E., Altıner Ş.
54th European Society of Human Genetics (ESHG) Conference, 28 - 31 August 2021, pp.424-425

IV. **Novel homozygous CEP41 mutation in a patient with Joubert syndrome**
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54th Conference of the European-Society-of-Human-Genetics (ESHG), ELECTR NETWORK, 28 - 31 August 2021, pp.270

V. **Saf kırmızı hücre aplazisinde genetik heterojenite**
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1. Ulusal Hematoonkogenetik Kongresi, Antalya, Turkey, 25 - 28 November 2021, pp.169

VI. **Novel Homozygous Missense MSMO1 Mutation in Two Patients With SC4MOL Deficiency**
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1. Uluslararası Katılımlı Genetik Günleri Dermatogenetik Sempozyumu, Bursa, Turkey, 9 - 11 January 2020

VII. **Assessing the limits of transport and storage conditions ofcGMP-grade humanumbilical cord mesenchymal stromal cells for clinical use**
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November 2019

VIII. **Sendromik Epidermolizis büllöza pruriginozanın genetik karakterizasyonu ve intravenöz immunglobulinin etkinliği**
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28.Ulusal Dermatoloji Kongresi, Antalya, Turkey, 24 - 28 September 2019

IX. **Intrafamilial phenotypic heterogeneity in dominant dystrophic epidermolysis bullosa associated with G2043R mutation in COL7A1**
Ili E., Vural S., Durmaz C. D., McGrath J. A., Ertop P., Heper A., Boyvat A., ILGIN RUHİ H.
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X. **Pyrin mutations in complex hidradenitis suppurativa**
Vural S., Gundogdu M., Ili E., Durmaz C. D., Vural A., Steinmueller-Magin L., Kleinhempel A., Holdt L. M., Ruzicka T., Giehl K. A., et al.
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XI. **A novel truncating mutation in LIG4 gene.**
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13. Balkan Congress of Human Genetics, 17 - 20 April 2019

XII. **ATN1 gene mutation in patients with Huntington disease-like phenotype**
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XIII. **Hidradenitis Suppurativa ile pyrin mutasyonları ve otoinflamasyon ilişkisi**
VURAL S., GÜNDOĞDU M., GÖKPINAR İLİ E., DURMAZ C. D., ILGIN RUHİ H., BOYVAT A.
28.Ulusal Dermatoloji Kongresi, Turkey, 24 - 28 September 2019

XIV. **Nonclassical adrenal hyperplasia and polycystic ovary syndrome: evaluation of four cases**
Ilgın Ruhi H., Taşdelen E., Gökpınar İli E., Gökoğlu M., Tuncalı T.
50th European Society of Human Genetics Conference, Kobenhavn, Denmark, 27 - 30 May 2017, pp.113-819

XV. **FRA16B: Cause or consequence? A case with Tourette syndrome and intellectual disability**
Kaba D., GÖKPINAR İLİ E., KUTLAY N., KILIÇ B. G.
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XVI. **NOVEL MUTATİONS İN THE ANDROGEN RECEPTOR GENE İN FOUR 46 xy FEMALES WİTH COMPLETE ANDROGEN İNSENSİTİVİTY SYNDROME**
ILGIN RUHİ H., KARABULUT H. G., ŞIKLAR Z., GÖKPINAR E., BERBEROĞLU M., TUKUN A.
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XVII. **MIXED GONADAL DYSGENESİS CYTOGENETİC AND PHENOTYPİC FİNDİNGS OF TWO CASES WİTH AMBİGOUS GENİTALİA**
GÖKPINAR E., DURMAZ C. D., ŞIKLAR Z., BERBEROĞLU M., ILGIN RUHİ H.
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XVIII. **Mixed gonadal dysgenesis cytogenetic and phenotypic findings of two cases with ambiguous genitalia**
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XIX. **Novel Mutations in the Androgen Receptor Gene in Four 46 XY Females with Complete Androgen Insensitivity Syndrome**
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XX. **Investigation of TBP gene mutations in patients with Huntington Disease phenotype**
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XXI. **22q11.2 delesyon sendromlu yetişkin dönem sunumlu bir olgu.**

GÖKPINAR E., ILGIN RUHİ H.
II. Hematolojik Genetik Sempozyumu, Kayseri, Turkey, 11 - 13 February 2016

Metrics

Publication: 36
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H-Index (Scopus): 3

Non Academic Experience

Akçakoca Devlet Hastanesi