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Education Information

Expertise In Medicine, Ankara University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Turkey 2014 - 2018
Undergraduate, Hacettepe University, Tıp Fakültesi (Türkçe), Tıp Fakültesi, Turkey 2006 - 2013

Dissertations

Expertise In Medicine, Şizofreni hastalarında RASD1 geni mutasyonlarının araştırılması, Ankara University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2018

Research Areas

Health Sciences

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Novel nonsense CAST mutation in two siblings with PLACK syndrome**
DURMAZ ÖZDİNÇ C. D., Tekmenuray-Unal A.
International Journal of Dermatology, vol.62, no.10, pp.1295-1299, 2023 (SCI-Expanded)

II. **Phenotypic and molecular characterization of five patients with PIK3CA-related overgrowth spectrum (PROS)**
Ili E. G., Tasdelen E., Durmaz C. D., ALTINER Ş., TUNCALI T., Martinez-Glez V., KARABULUT H. G., Vural S., Ceylaner S., ACAR M. O., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.188, no.6, pp.1792-1800, 2022 (SCI-Expanded)

III. **Undifferentiated Melanoma Resembling Undifferentiated Round Cell Sarcoma: The Diagnostic Power of Molecular Melanoma Signature**
KAVUNCUOĞLU A., DURMAZ ÖZDİNÇ C. D., GÖKÖZ Ö., ÜNER A., KÖSEMEHMETOĞLU K.
INTERNATIONAL JOURNAL OF SURGICAL PATHOLOGY, vol.30, no.3, pp.346-349, 2022 (SCI-Expanded)

IV. **Extending Phenotypic Spectrum of 17q22 Microdeletion: Growth Hormone Deficiency**
Durmaz C. D., ALTINER Ş., Tasdelen E., KARABULUT H. G., ILGIN RUHİ H.
FETAL AND PEDIATRIC PATHOLOGY, vol.40, no.5, pp.486-492, 2021 (SCI-Expanded)

V. **MASP1-related 3MC syndrome in a patient from Turkey**

Durmaz C. D., Altiner S.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.7, pp.2267-2270, 2021 (SCI-Expanded)

VI. **Ectopic Posterior Pituitary, Polydactyly, Midfacial Hypoplasia and Multiple Pituitary Hormone Deficiency due to a Novel Heterozygous IVS11-2A > C(c.1957-2A > C) Mutation in the GLI2 Gene**
Demiral M., DEMİRBİLEK H., Unal E., Durmaz C. D., Ceylaner S., Ozbek M. N.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.12, no.3, pp.319-328, 2020 (SCI-Expanded)

VII. **Promising effect of intravenous immunoglobulin therapy for epidermolysis bullosa pruriginosa**
Ertop P., Vural S., Ili E. G., Durmaz C. D., HEPER A., McGrath J. A., Ilgin R. H., BOYVAT A.
INTERNATIONAL JOURNAL OF DERMATOLOGY, vol.59, no.7, pp.851-855, 2020 (SCI-Expanded)

VIII. **H syndrome: a genodermatosis characterised by hyperpigmented, and hypertrichotic skin**
An I., Durmaz C. D., ILGIN RUHİ H., Ertop P., Ozturk M., Sula B., Ecer N.
HONG KONG JOURNAL OF DERMATOLOGY & VENEREOLOGY, vol.27, no.3, pp.137-140, 2019 (SCI-Expanded)

IX. **Association of pyrin mutations and autoinflammation with complex phenotype hidradenitis suppurativa: a case-control study**
VURAL S., Gundogdu M., Illi E. G., Durmaz C. D., Vural A., Steinmuller-Magin L., Kleinhempel A., Holdt L. M., Ruzicka T., Giehl K. A., et al.
BRITISH JOURNAL OF DERMATOLOGY, vol.180, no.6, pp.1459-1467, 2019 (SCI-Expanded)

X. **Primary Hypertrophic Osteoarthropathy Mimicking Juvenile Idiopathic Arthritis: A Novel SLCO2A1 Mutation and Imaging Findings**
USLU YURTERİ E., Durmaz C. D., KARABULUT H. G., Seifert W., Horn D., AKKAYA Z., Turgay M.
CYTOGENETIC AND GENOME RESEARCH, vol.158, no.3, pp.126-132, 2019 (SCI-Expanded)

XI. **WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome**
White J. J., Mazzeu J. F., Coban-Akdemir Z., Bayram Y., Bahrambeigi V., Hoischen A., van Bon B. W. M., Gezdirici A., Gulec E. Y., Ramond F., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.102, no.1, pp.27-43, 2018 (SCI-Expanded)

XII. **A Novel PTCH1 Frameshift Mutation Leading to Nevoid Basal Cell Carcinoma Syndrome**
Durmaz C. D., Evans G., Smith M. J., Ertop P., AKAY B., TUNCALI T.
CYTOGENETIC AND GENOME RESEARCH, vol.154, no.2, pp.57-61, 2018 (SCI-Expanded)

XIII. **A Novel PORCN Frameshift Mutation Leading to Focal Dermal Hypoplasia: A Case Report**
Durmaz C. D., McGrath J., Liu L., KARABULUT H. G.
CYTOGENETIC AND GENOME RESEARCH, vol.154, no.3, pp.119-121, 2018 (SCI-Expanded)

XIV. **Autosomal Recessive Oculodentodigital Dysplasia: A Case Report and Review of the Literature**
Tasdelen E., Durmaz C. D., KARABULUT H. G.
CYTOGENETIC AND GENOME RESEARCH, vol.154, no.4, pp.181-186, 2018 (SCI-Expanded)

XV. **Skin-Dominant Phenotype in a Patient with H Syndrome: Identification of a Novel Mutation in the SLC29A3 Gene**
Vural S., Ertop P., Durmaz C. D., ŞANLI H., HEPER A., KUNDAKCI N., KARABULUT H. G., ILGIN RUHİ H.
CYTOGENETIC AND GENOME RESEARCH, vol.151, no.4, pp.186-190, 2017 (SCI-Expanded)

XVI. **Bilateral choanal atresia in an adult woman with pycnodysostosis**
Durmaz C. D., Tas V., Kocaay P., FİTOZ Ö. S., Onay H., BETON S., Ozkinay F., ILGIN RUHİ H.
CONGENITAL ANOMALIES, vol.57, no.3, pp.91-92, 2017 (SCI-Expanded)

XVII. **Unusual Chromosomal Rearrangement Resulted in Interstitial Monosomy 9p: Case Report**
Durmaz C. D., Yararbas K., KUTLAY N., Turedi O., Akin I., Gurbuz C., Karatas G., Tukun A.
CYTOGENETIC AND GENOME RESEARCH, vol.148, no.1, pp.19-24, 2016 (SCI-Expanded)

Refereed Congress / Symposium Publications in Proceedings

I. **Pyrin mutations in complex hidradenitis suppurativa**
Vural S., Gundogdu M., Ili E. G., Durmaz C. D., Vural A., Steinmueller-Magin L., Kleinhempel A., Holdt L. M., Ruzicka T.,

Giehl K. A., et al.
Annual Meeting of the British-Society-for-Investigative-Dermatology, Bradford, England, 1 - 03 April 2019, vol.180

Supported Projects

ÇETİNKAYA A., AKARSU A. N., DURMAZ ÖZDİNÇ C. D., GÜLERAY LAFCI N., ALİKAŞİFOĞLU M., Project Supported by Higher Education Institutions, Hacettepe Optik Genom Haritalama Merkezinin Oluşturulması, 2022 - Continues
ÖZGÜL N., ATEŞ ÖZDEMİR D., USUBÜTÜN A., DURMAZ ÖZDİNÇ C. D., ÇELİK Ö., Project Supported by Higher Education Institutions, Mikrosatelit İnstabilitesi Olan Endometrium Kanserli Olguların Klinik Patolojik ve Genetik Verilerinin Değerlendirilmesi, 2022 - Continues

Metrics

Publication: 18
Citation (WoS): 119
Citation (Scopus): 118
H-Index (WoS): 5
H-Index (Scopus): 5

Non Academic Experience

Wellcome Trust Sanger Institute