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Education Information

Post Doctorate, University Of Connecticut, Connecticut Health Center, Surgical Research Center, United States Of America
1994 - 1996

Doctorate, Ankara Üniversitesi, Tıp Fakültesi, Tıbbi Biyoloji Ve Genetik, Turkey 1988 - 1994

Undergraduate, Hacettepe Üniversitesi, Tıp Fakültesi, Tıp, Turkey 1978 - 1984

Foreign Languages

English, B2 Upper Intermediate

Dissertations

Doctorate, Sindaktili tip II (sin polidaktili): Genetik özellikleri, aile ve populasyon çalışması, Ankara Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1994

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Medical Genetics

Academic Titles / Tasks

Professor, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 2008 - Continues

Associate Professor, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 1999 - 2008

Expert, Hacettepe Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1997 - 1999

Academic and Administrative Experience

Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 2013 - 2014

Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

I. One Disease with two Faces: Semidominant Inheritance of a Novel HTRA1 Mutation in a

Consanguineous Family

Bekircan-Kurt C. E. , ÇETİNKAYA A., GÖÇMEN R., KOŞUKCU C., Soylemezoglu F., ARSAVA E. M. , Tuncer A., ERDEM ÖZDAMAR S., Akarsu N. A. , TOPÇUOĞLU M. A.

JOURNAL OF STROKE & CEREBROVASCULAR DISEASES, vol.30, no.9, 2021 (Journal Indexed in SCI)

- II. **Hematopoietic Cell Transplantation Cures Adenosine Deaminase 2 Deficiency: Report on 30 Patients**
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- III. **Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy**
Wong H. H. , Seet S. H. , Maier M., GÜREL A., Traspas R. M. , Lee C., Zhang S., TALİM B., Loh A. Y. T. , Chia C. Y. , et al.
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- IV. **Long-Term Follow-Up Outcomes of 19 Patients with Osteogenesis Imperfecta Type XI and Bruck Syndrome Type I Caused by FKBP10 Variants**
YÜKSEL ÜLKER A., ULUDAĞ ALKAYA D., Elkanova L., ŞEKER A., Akpınar E., Akarsu N. A. , Uyguner Z. O. , TÜYSÜZ B.
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- V. **Comprehensive clinical, biochemical, radiological and genetic analysis of 28 Turkish cases with suspected metachromatic leukodystrophy and their relatives**
Pekgul F., Eroglu-Ertugrul N. G. , Bekircan-Kurt C. E. , Erdem-Ozdamar S., Cetinkaya A., Tan E., Konuskan B., Karaagaoglu E., Topcu M., Akarsu N. A. , et al.
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- VI. **Laboratory diagnosis of metachromatic leukodystrophy requires more than arylsulfatase A assay**
Pekgul F., Bekircan-Kurt C. E. , Konuskan B., Erdem-Ozdamar S., Tan E., Akarsu N., Topcu M., Anlar B., Ozkara H. A.
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- VII. **Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene BRCA1**
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- VIII. **Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamond-like features.**
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- IX. **Analysis of centrosome and DNA damage response in PLK4 associated Seckel syndrome.**
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- X. **Identification of two novel PNPLA1 mutations in Turkish families with autosomal recessive congenital ichthyosis.**
Dökmeçi-Emre S., TAŞKIRAN Z. E. , YÜZBAŞIOĞLU A., ÖNAL G., AKARSU A. N. , KARADUMAN A., ÖZGÜÇ M.
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- XI. **Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling.**
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American journal of human genetics, vol.99, no.2, pp.299-317, 2016 (Journal Indexed in SCI Expanded)
- XII. **Recurrent viral infections associated with a homozygous CORO1A mutation that disrupts oligomerization and cytoskeletal association**
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- XIII. **Nonsense Mutation in SPARC Gene Causing Autosomal Recessive Osteogenesis Imperfecta**

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- XV. **CRIM1 haploinsufficiency causes defects in eye development in human and mouse**
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- XVI. **Mutations Affecting the BHLHA9 DNA-Binding Domain Cause MSSD, Mesoaxial Synostotic Syndactyly with Phalangeal Reduction, Malik-Percin Type**
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- XVII. **Novel splice-site and missense mutations in the ALDH1A3 gene underlying autosomal recessive anophthalmia/microphthalmia**
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- XIX. **TMCO1 Deficiency Causes Autosomal Recessive Cerebrofaciothoracic Dysplasia**
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- XXI. **Association between C677T and A1298C MTHFR gene polymorphism and nonsyndromic orofacial clefts in the Turkish population: a case-parent study**
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- XXIII. **Mutations in RIPK4 Cause the Autosomal-Recessive Form of Popliteal Pterygium Syndrome**
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- XXIV. **Disruption of the ptpro gene causes childhood onset nephrotic syndrome**
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- XXV. **Disruption of PTPRO Causes Childhood-Onset Nephrotic Syndrome**
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- XXVII. **Smaller Hippocampus Volume Is Associated with Short Variant of 5-HTTLPR Polymorphism in Medication-Free Major Depressive Disorder Patients**
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- XXX. **Marie Unna Hereditary Hypotrichosis: A Turkish Family With Loss of Eyebrows and a U2HR Mutation**
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- XXXI. **Intracranial and Extracranial Malformations in Patients With Craniofacial Anomalies**
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Uz E., Alanay Y., Aktas D., Vargel I., Gucer S., Tuncbilek G., von Eggeling F., Yilmaz E., DEREN Ö., Posorski N., et al.
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- XXXIV. **Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta**
Alanay Y., Avaygan H., Camacho N., ÜTİNE G. E. , Boduroglu K., Aktas D., ALİKAŞİFOĞLU M., Tuncbilek E., ORHAN D., Bakar F. T. , et al.
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- XXXV. **The effect of depression, BDNF gene val66met polymorphism and gender on serum BDNF levels**
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- XXXVI. **A Specific Mutation in the Distant Sonic Hedgehog (SHH) Cis-Regulator (ZRS) Causes Werner Mesomelic Syndrome (WMS) While Complete ZRS Duplications Underlie Haas Type Polysyndactyly and Preaxial Polydactyly (PPD) With or Without Triphalangeal Thumb**
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- XXXVII. **Derivative chromosome 1 and GLUT1 deficiency syndrome in a sibling pair**
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- XXXVIII. **ALX4 dysfunction disrupts craniofacial and epidermal development**
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- XXXIX. **Disease causing nature of homozygous missense, p.A523D, alteration in the perforin gene.**
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- XL. **Homozygous feature of isolated triphalangeal thumb-preaxial polydactyly linked to 7q36: no phenotypic difference between homozygotes and heterozygotes**
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- XLI. **A new autosomal dominant Peters' anomaly phenotype expanding the anterior segment dysgenesis spectrum**
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- XLIII. **Mutations in the very low-density lipoprotein receptor VLDLR cause cerebellar hypoplasia and quadrupedal locomotion in humans**
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- XLIV. **Environmental effect and genetic influence: a regional cancer predisposition survey in the Zonguldak region of Northwest Turkey**
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- XLV. **Skewed X inactivation in an X linked nystagmus family resulted from a novel, p.R229G, missense mutation in the FRMD7 gene**
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- XLVII. **Molecular and clinical analysis of Turkish patients with HLH**
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- L. **Autosomal recessive mesoaxial synostotic syndactyly with phalangeal reduction maps to chromosome 17p13.3**
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- LI. **Identification of KIF21A mutations as a rare cause of congenital fibrosis of the extraocular muscles type 3 (CFEOM3)**
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- LII. **Is vitamin D hypothesis for schizophrenia valid? Independent segregation of psychosis in a family with vitamin-D-dependent rickets type IIA.**
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- LIV. **Molecular characterization of Turkish patients with pyrimidine 5 ' nucleotidase-I deficiency**
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- LV. **Genome scan meta-analysis of schizophrenia and bipolar disorder, part III: Bipolar disorder**
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- LVII. **Disruption of a long-range cis-acting regulator for Shh causes preaxial polydactyly**
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- LXIII. **An apparently dominant bipolar affective disorder (BPAD) locus on chromosome 20p11.2-q11.2 in a large Turkish pedigree**
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- LXIV. **Autosomal recessive severe intraosseous hemangioma in the skull. A new syndrome?**
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