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### Eğitim Bilgileri

Post Doktora, University Of Connecticut, Connecticut Health Center, Surgical Research Center, Amerika Birleşik Devletleri  
1994 - 1996

Doktora, Ankara Üniversitesi, Tıp Fakültesi, Tıbbi Biyoloji Ve Genetik, Türkiye 1988 - 1994

Lisans, Hacettepe Üniversitesi, Tıp Fakültesi, Tıp, Türkiye 1978 - 1984

### Yabancı Diller

İngilizce, B2 Orta Üstü

### Yaptığı Tezler

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

### Araştırma Alanları

Tıp, Sağlık Bilimleri, Dahili Tıp Bilimleri, Tıbbi Genetik

### Akademik Unvanlar / Görevler

Prof.Dr., Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 2008 - Devam Ediyor

Doç.Dr., Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 1999 - 2008

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

### Mesleki Deneyim

Anabilim/Bilim Dalı Başkanı, Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 2013 - 2014

### SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- Comprehensive clinical, biochemical, radiological and genetic analysis of 28 Turkish cases with

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- III. **Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene BRCA1**  
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- IV. **Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamond-like features.**  
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- V. **Analysis of centrosome and DNA damage response in PLK4 associated Seckel syndrome.**  
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- VI. **Identification of two novel PNPLA1 mutations in Turkish families with autosomal recessive congenital ichthyosis.**  
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- VII. **Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling.**  
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- X. **Frequency of Recessive Osteogenesis Imperfecta in a Turkish Cohort and Genetic Causes**  
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- XI. **CRIM1 haploinsufficiency causes defects in eye development in human and mouse**  
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- XII. **Mutations Affecting the BHLHA9 DNA-Binding Domain Cause MSSD, Mesoaxial Synostotic Syndactyly with Phalangeal Reduction, Malik-Percin Type**  
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- XIII. **Expanding the phenotypic spectrum of ECEL1-related congenital contracture syndromes**  
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- XIV. **Novel splice-site and missense mutations in the ALDH1A3 gene underlying autosomal recessive**

## **anophthalmia/micropthalmia**

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### **XV. TMC01 Deficiency Causes Autosomal Recessive Cerebrofaciothoracic Dysplasia**

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### **XVI. Three Patients Resembling Teebi-Shaltout Syndrome**

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### **XVII. Association between C677T and A1298C MTHFR gene polymorphism and nonsyndromic orofacial clefts in the Turkish population: a case-parent study**

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### **XIX. Mutations in RIPK4 Cause the Autosomal-Recessive Form of Popliteal Pterygium Syndrome**

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### **XXI. Disruption of PTPRO Causes Childhood-Onset Nephrotic Syndrome**

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### **XXII. KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes**

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### **XXIII. Smaller Hippocampus Volume Is Associated with Short Variant of 5-HTTLPR Polymorphism in Medication-Free Major Depressive Disorder Patients**

EKER M. Ç. , KİTİŞ Ö., OKUR H., Eker O. D. , Ozan E., Isikli S., Akarsu N., GÖNÜL A. S.

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### **XXIV. Mutation in Exon 1f of PLEC, Leading to Disruption of Plectin Isoform 1f, Causes Autosomal-Recessive Limb-Girdle Muscular Dystrophy**

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### **XXV. Congenital hypertelorism and osteopenia: A novel autosomal recessive disease**

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### **XXVI. Marie Unna Hereditary Hypotrichosis: A Turkish Family With Loss of Eyebrows and a U2HR Mutation**

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- XXVIII. **ASSOCIATION OF POLYNEUROPATHY, MENTAL RETARDATION, SENSORINEURAL HEARING LOSS, 6th NERVE PALSY, CONVULSIONS, AND ORAL DYSKINESIA; A PROPABLE NEW NEUROMETABOLIC DISORDER**  
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- XXXIII. **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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- XXXVI. **Homozygous feature of isolated triphalangeal thumb-preaxial polydactyly linked to 7q36: no phenotypic difference between homozygotes and heterozygotes**  
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- XLII. **Colobomatous macrophthalmia with microcornea syndrome maps to the 2p23-p16 region**  
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- XLV. **Evidence from autoimmune thyroiditis of skewed X-chromosome inactivation in female predisposition to autoimmunity**  
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- XLVI. **Autosomal recessive mesoaxial synostotic syndactyly with phalangeal reduction maps to chromosome 17p13.3**  
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- XLVIII. **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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