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Education Information

Expertise In Medicine, Istanbul University, Tıp Fakültesi, Turkey 1984 - 1989

Dissertations

Expertise In Medicine, Asfiksili term yenidoğanlarda böbrek fonksiyonlarının incelenmesi, Istanbul University, Istanbul Medical Faculty, 1989

Research Areas

Health Sciences

Academic Titles / Tasks

Professor, Marmara University, School of Medicine, Internal Medical Sciences, 2004 - Continues

Courses

Tıp Fakültesi 5.sınıf dersleri Yaşam Boyu sağlık, Undergraduate, 2020 - 2021
Congenitale Anomalies in Childhood, Undergraduate, 2020 - 2021
Tıp Fakültesi 6.sınıf dersler ve pratikler, Undergraduate, 2020 - 2021

Advising Theses

Elçioğlu H. N., Çocuk genetik kliniğimizde 2000-2023 yılları arasında takip ettiğimiz marfan sendromu tanılı ve marfanoid hastaların klinik izlemlerinin değerlendirilmesi ve genetik danışma verilmesi, Expertise In Medicine, A.FATİH(Student), 2023
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Elçioğlu H. N., Çocuk Genetik Kliniğimizde 2010-2022 yılları arasında takip ettiğimiz Nörofibromatozis hastalarının klinik izlemlerinin değerlendirilmesi ve genetik danışma verilmesi, Expertise In Medicine, M.CAN(Student), 2022
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Elçioğlu H. N., Herediter primer mikrosifalisi olan çocuklarda klinik değerlendirmeler, Expertise In Medicine, M.SALİM(Student), 2013
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II. **Clinical and Molecular Characterization of Mucopolysaccharidosis Type 3A and 3B in a Turkish Series**
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III. **Further defining the molecular spectrum and long-term follow-up of 17 patients with Dyggve–Melchior–Clausen and Smith–McCort dysplasia type 2**
Akalın A., Ayaz E., Soğukpınar M., Avcı-Durmuşalioğlu E., Ürel-Demir G., YILDIZ A. E., Atik T., ELÇİOĞLU H. N., Eda Utine G., Şimşek-Kiper P. Ö.
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IV. **Autosomal recessive otospondylo-mega-epiphyseal dysplasia: Comprehensive clinical review of a pediatric cohort**
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VI. **A novel mutation in the TRIP11 gene: Diagnostic approach from relatively common skeletal dysplasias to an extremely rare Odontochondrodysplasia.**
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VII. **Novel SIX6 mutations cause recessively inherited congenital cataract, microcornea, and corneal opacification with or without coloboma and microphthalmia**
Panagiotou E. S., Fernandez-Fuentes N., Farraj L. A., McKibbin M., ELÇİOĞLU H. N., Jafri H., ÇERMAN E., Parry D. A., V. Logan C., Johnson C. A., et al.
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VIII. **Phenotypic spectrum of BLM- and RMI1-related Bloom syndrome**
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IX. **Disentangling molecular and clinical stratification patterns in beta-galactosidase deficiency**
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XI. **High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population**
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XVII. **A Rare Cause of Hypophosphatemia: Raine Syndrome Changing Clinical Features with Age**
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XVIII. **The genomic and clinical landscape of fetal akinesia**
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XIX. **TNFRSF11A-Associated Dysosteosclerosis: A Report of the Second Case and Characterization of the Phenotypic Spectrum**
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XXVII. **Correction: Loss of the BMP Antagonist, SMOC-1, Causes Ophthalmo-Acromelic (Waardenburg Anophthalmia) Syndrome in Humans and Mice.**
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XXVIII. **The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance**
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XXXVII. **Identification of biallelic EXTL3 mutations in a novel type of spondylo-epi-metaphyseal dysplasia**
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