Prof. HURİYE NURSEL ELÇİOĞLU

Personal Information

Email: nelcioglu@marmara.edu.tr
Web: https://avesis.marmara.edu.tr/nelcioglu

International Researcher IDs
ORCID: 0000-0003-0051-6720
Yoksis Researcher ID: 8771

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., Çocuk genetik hastalıkları polikliniğimize 2000-2015 yıllarında başvuran osteogenezis imperfektalı hastaların retrospektif değerlendirilmesi ve genetik danışmanlık verilmesi, Expertise In Medicine, Ö.KAMER(Student), 2016
Elçioğlu H. N., Kliniğimizde takip ettiğimiz bardet biedl sendromlu hastaların fenotipik dağılımına yönelik klinik veri bankası oluşturulması ve genetik danışma verilmesi, Expertise In Medicine, D.ALİKILIÇ(Student), 2015
Elçioğlu H. N., Herediter primer mikrosifalisi olan çocuklarda klinik değerlendirmeler, Expertise In Medicine, M.SALİM(Student), 2013
Elçioğlu H., Kraniofasial malformasyonlara klinik ve genetik yaklaşımlar, Expertise In Medicine, F.Sarı(Student), 2010
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ELÇİOĞLU H. N., Nonsendromik konjenital yarık dudak/damak hastalıklarında metinletetrahidrofolat redüktaz gen polimorfizmin yeri, Expertise In Medicine, A.Şemiç(Student), 2004

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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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II. **HMZDupFinder: a robust computational approach for detecting intragenic homozygous duplications from exome sequencing data.**
Du H., Dardas Z., Jolly A., Grochowski C. M., Jhangiani S. N., Li H., Muzny D., Fatih J. M., Yesil G., Elçioglu N. H., et al.
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III. **Clinical and Molecular Characterization of Mucopolysaccharidosis Type 3A and 3B in a Turkish Series**
Noyan B., Elcioglu N. H., Tebani A., Bekri S.
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IV. **Autosomal recessive otospondylo-mega-epiphyseal dysplasia: Comprehensive clinical review of a pediatric cohort**
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V. **Management of acute metabolic crisis in TANGO2 deficiency: A case report**
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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. **Phenotypic spectrum of BLM- and RMI1-related Bloom syndrome**
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CLINICAL GENETICS, vol.101, no.5-6, pp.559-564, 2022 (SCI-Expanded)

VIII. **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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IX. **Disentangling molecular and clinical stratification patterns in beta-galactosidase deficiency**
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X. **MFSD2A-associated primary microcephaly-Expanding the clinical and mutational spectrum of this ultra-rare disease**
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XI. **High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population**
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XII. **From cataract to syndrome diagnosis: Revaluation of Warburg-Micro syndrome Type 1 patients**
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XIII. **Genome sequencing in families with congenital limb malformations**
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Eltan M., Alavanda C., Yavas Abali Z., Ergenekon P., Yalindag Ozturk N., Sakar M., Dağçınar A., Kırkgöz T., Kaygusuz S. B., Gökdemir Y., et al.
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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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XXIII. **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome**
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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**
Lehalle D., Altunoglu U., Bruel A., Assoum M., Duffourd Y., Masurel A., Baujat G., Bessieres B., Captier G., Edery P., et al.
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XXX. **Further delineation of Malan syndrome**
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AMERICAN JOURNAL OF HUMAN GENETICS, vol.103, no.1, pp.115-124, 2018 (SCI-Expanded)

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