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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

Elçioğlu H. N., Çocuk genetik kliniğimizde 2000-2023 yılları arasında takip ettiğimiz noonan sendromu ve ilişkili rasopati sendromu tanılı hastaların klinik izlemlerinin değerlendirilmesi ve genetik danışma verilmesi, Expertise In Medicine, E.SÖNMEZ(Student), 2023

Elçioğlu H. N., Mukopolisakkaridoz grubu çocuk hastaların oral bulgularının ve yaşam kalitesi anketlerinin değerlendirilmesi, Dental Specialty, V.ÖZALP(Student), 2022

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

ELÇİOĞLU H. N., Kliniğimizde 2000-2018 Yılları Arasında Takip Ettiğimiz Mukopolisakkaridoz Hastalarının Kardiyak Tutulumlarının Değerlendirilmesi, Expertise In Medicine, M.KARIMOVA(Student), 2019

Elçioğlu H. N., Kliniğimizde 2000-2016 yılları arasında takip ettiğimiz Sanfilippo sendromlu hastaların tanı, biyokimyasal parametreler, tedavi ve klinik izlemlerinin değerlendirilmesi ve Genetik Danışma verilmesi, Expertise In Medicine, B.Noyan(Student), 2016

Elçioğlu H. N., Kliniğimizde takip ettiğimiz ehlers-danlos sendromlu hastaların fenotipik dağılımına yönelik klinik veri bankası oluşturulması ve genetik danışma verilmesi, Expertise In Medicine, S.ARAS(Student), 2016

Elçioğlu H. N., Çocuk genetik hastalıkları polikliniğimize 2000-2015 yıllarında başvuran osteogenezis imperfekta 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., Kraniofasial malformasyonlara klinik ve genetik yaklaşımlar, Expertise In Medicine, F.Sarı(Student), 2010

Elçioğlu H. N., Çocuklarda eklem hipermobilitesi ile seyreden kalıtsal bağ dokusu hastalıklarının klinik özellikleri, Expertise In Medicine, B.ÇOLAK(Student), 2008

ELÇİOĞLU H. N., Nonsendromik konjenital yarı dudak/damak hastalıklarında metilentetrahidrofolat redüktaz gen polimorfizmin yeri, Expertise In Medicine, A.Şemiç(Student), 2004

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Published journal articles indexed by SCI, SSCI, and AHCI

- I. **HMZDupFinder: a robust computational approach for detecting intragenic homozygous duplications from exome sequencing data.**
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Nucleic acids research, vol.52, 2024 (SCI-Expanded)
- II. **Autosomal recessive otospondylo-mega-epiphyseal dysplasia: Comprehensive clinical review of a pediatric cohort**
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Clinical Dysmorphology, vol.32, no.4, pp.151-155, 2023 (SCI-Expanded)
- III. **Management of acute metabolic crisis in TANGO2 deficiency: A case report**
Yılmaz-Gümüş E., ELÇİOĞLU H. N., Genç E., Arıç Ş., Öztürk G., Yapıcı Ö., Akallı F., ÖZTÜRK HİŞMİ B.
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- IV. **A novel mutation in the *TRIP11* gene: Diagnostic approach from relatively common skeletal dysplasias to an extremely rare Odontochondrodysplasia.**
Yeter B., Dilruba Aslanger A., Yesil G., Elcioglu N. H.
Journal of clinical research in pediatric endocrinology, vol.14, pp.475-480, 2022 (SCI-Expanded)
- V. **Novel SIX6 mutations cause recessively inherited congenital cataract, microcornea, and corneal opacification with or without coloboma and microphthalmia**
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MOLECULAR VISION, vol.28, pp.57-69, 2022 (SCI-Expanded)
- VI. **Phenotypic spectrum of BLM- and RMI1-related Bloom syndrome**
Goenenc I. I., ELÇİOĞLU H. N., Grijalva C. M., Aras S., Grossmann N., Praulich I., Altmueller J., Kaulfuss S., Li Y., Nuernberg P., et al.
CLINICAL GENETICS, vol.101, no.5-6, pp.559-564, 2022 (SCI-Expanded)
- VII. **Disentangling molecular and clinical stratification patterns in beta-galactosidase deficiency**
Tebani A., Sudrie-Arnaud B., Dabaj I., Torre S., Domitille L., Snanoudj S., Heron B., Levade T., Caillaud C., Vergnaud

S., et al.

JOURNAL OF MEDICAL GENETICS, vol.59, no.4, pp.377-384, 2022 (SCI-Expanded)

- VIII. **MFSD2A-associated primary microcephaly-Expanding the clinical and mutational spectrum of this ultra-rare disease**
Khuller K., Yigit G., Grijalva C. M., Altmueller J., Thiele H., Nurnberg P., Elcioglu N. H., YETER DOĞAN B., Hehr U., Stein A., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.64, no.10, 2021 (SCI-Expanded)
- IX. **High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population**
Mitani T., Isikay S., Gezdirici A., Gulec E. Y., Punetha J., Fatih J. M., Herman I., Akay G., Du H., Calame D. G., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.108, no.10, pp.1981-2005, 2021 (SCI-Expanded)
- X. **Genome sequencing in families with congenital limb malformations**
Elsner J., Mensah M. A., Holtgrewe M., Hertzberg J., Bigoni S., Busche A., Coutelier M., de Silva D. C., Elcioglu N., Filges I., et al.
HUMAN GENETICS, vol.140, no.8, pp.1229-1239, 2021 (SCI-Expanded)
- XI. **From cataract to syndrome diagnosis: Revaluation of Warburg-Micro syndrome Type 1 patients**
Albayrak H. M., Elcioglu N. H., YETER DOĞAN B., Karaer K.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.8, pp.2325-2334, 2021 (SCI-Expanded)
- XII. **Genotype-phenotype correlations in hereditary multiple exostoses**
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EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, no.SUPPL 1, pp.834, 2020 (SCI-Expanded)
- XIII. **Jeune Syndrome with a novel DYNC2H1 mutation**
Elcioglu N. H., Yeter B., Xue J. Y., Wang Z., Guo L., Nishimura G., Ikegawa S.
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- XIV. **Development, behaviour and sensory processing in Marshall-Smith syndrome and Malan syndrome: phenotype comparison in two related syndromes.**
Mulder P. A., van Balkom I. D. C., Landlust A. M., Priolo M., Menke L. A., Acero I. H., Alkuraya F. S., Arias P., Bernardini L., Bijlsma E. K., et al.
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- XV. **A Rare Cause of Hypophosphatemia: Raine Syndrome Changing Clinical Features with Age**
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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- XVI. **The genomic and clinical landscape of fetal akinesia**
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GENETICS IN MEDICINE, vol.22, no.3, pp.511-523, 2020 (SCI-Expanded)
- XVII. **TNFRSF11A-Associated Dysosteosclerosis: A Report of the Second Case and Characterization of the Phenotypic Spectrum**
Xue J., Wang Z., Shinagawa S., Ohashi H., Otomo N., Elcioglu N. H., Nakashima T., Nishimura G., Ikegawa S., Guo L.
JOURNAL OF BONE AND MINERAL RESEARCH, vol.34, no.10, pp.1873-1879, 2019 (SCI-Expanded)
- XVIII. **A rare cause of hypophosphatemia: Raine Syndrome**
Eltan M., Ata P., Kırkgöz T., Alavanda C., Kaygusuz S. B., Menevse T. S., Tosun B. G., Abali Z. Y., Helvacioğlu D., Güran T., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.384, 2019 (SCI-Expanded)
- XIX. **Molecular characterization of a large group of Mucopolysaccharidosis type IIIC patients reveals the evolutionary history of the disease**
Martins C., de Medeiros P. F., Leistner-Segal S., Dridi L., Elcioglu N., Wood J., Behnam M., Noyan B., Lacerda L., Geraghty M. T., et al.
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- XX. **The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for**

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Pehlivan D., Bayram Y., Gunes N., Akdemir Z. C., Shukla A., Bierhals T., Tabakci B., Sahin Y., Gezdirici A., Fatih J. M., et al.

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- XXI. **Ptois as a unique hallmark for autosomal recessive WNT1-associated osteogenesis imperfecta**
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- XXII. **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome**
van der Sluijs P. J., Jansen S., Vergano S. A., Adachi-Fukuda M., ALANAY Y., AlKindy A., Baban A., Bayat A., Beck-
Woedl S., Berry K., et al.
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- XXIII. **Does the clinical phenotype of mucopolidosis-III gamma differ from its alpha beta counterpart?: supporting facts in a cohort of 18 patients**
Nampoothiri S., Elcioglu N. H., KOCA S. S., Yesodharan D., Chandrababu K. K., Vinod K. V., Bhat M., Mohandas N. K.,
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- XXIV. **Progressive pseudorheumatoid dysplasia: A presentation of four cases with slow and rapid progression and effects of early rehabilitation program**
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- XXV. **Correction: Loss of the BMP Antagonist, SMOC-1, Causes Ophthlmo-Acromelic (Waardenburg Anophthalmia) Syndrome in Humans and Mice.**
Rainger J., van Beusekom E., Ramsay J. K., McKie L., Al-Gazali L., Pallotta R., Saponari A., Branney P., Fisher M.,
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- XXVI. **The oculaauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance**
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AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.176, no.12, pp.2740-2750, 2018 (SCI-Expanded)
- XXVII. **Identification of candidate gene FAM183A and novel pathogenic variants in known genes: High genetic heterogeneity for autosomal recessive intellectual disability**
McSherry M., Masih K. E., Elcioglu N. H., Celik P., Balci O., Cengiz F. B., Nunez D., Sineni C. J., Seyhan S., Kocaoglu D., et al.
PLOS ONE, vol.13, no.11, 2018 (SCI-Expanded)
- XXVIII. **Further delineation of Malan syndrome**
Priolo M., Schanze D., Tatton-Brown K., Mulder P. A., Tenorio J., Kooblall K., Hernandez Acero I., Alkuraya F. S., Arias P., Bernardini L., et al.
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- XXIX. **Mutations in TOP3A Cause a Bloom Syndrome-like Disorder**
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- XXX. **Recessive MYF5 Mutations Cause External Ophthalmoplegia, Rib, and Vertebral Anomalies**
Di Gioia S. A., Shaaban S., TÜYSÜZ B., Elcioglu N. H., Chan W., Robson C. D., Ecklund K., Gilette N. M., Hamzaoglu A.,
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AMERICAN JOURNAL OF HUMAN GENETICS, vol.103, no.1, pp.115-124, 2018 (SCI-Expanded)
- XXXI. **Dysosteosclerosis is also caused by TNFRSF11A mutation**
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Ikegawa S.

JOURNAL OF HUMAN GENETICS, vol.63, no.6, pp.769-774, 2018 (SCI-Expanded)

- XXXII. **Insights into Mutation Effect in Three Poikiloderma with Neutropenia Patients by Transcript Analysis and Disease Evolution of Reported Patients with the Same Pathogenic Variants**

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- XXXIV. **Clinico-molecular analysis of eleven patients with Hermansky-Pudlak type 5 syndrome, a mild form of HPS**

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- XXXVI. **Heterozygous HNRNPU variants cause early onset epilepsy and severe intellectual disability**

Bramswig N. C., Luedecke H., Hamdan F. F., Altmueller J., Beleggia F., Elcioglu N. H., Freyer C., Gerkes E. H., Demirkol Y. K., Knupp K. G., et al.

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- XXXVII. **REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis**

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- XXXVIII. **Novel EYA1 variants causing Branchio-oto-renal syndrome**

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- XXXIX. **Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases**

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- XL. **Novel and recurrent XYLT1 mutations in two Turkish families with Desbuquois dysplasia, type 2**

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- XLI. **Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2**

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- XLII. **Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin**

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- XLIII. **TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism**

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- XLIV. **A THANATOPHORIC DYSPLASIA TYPE I CASE WITH A FGFR3 P.R248C MUTATION AND SURVIVAL BEYOND THE NEONATAL PERIOD**
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- XLV. **A thanatophoric dysplasia type i case with a fgfr3 p.r248c mutation and survival ibeyond the neonatal period**
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- XLVI. **A role for repressive complexes and H3K9 di-methylation in PRDM5-associated brittle cornea syndrome**
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- XLVII. **Insights into genotype-phenotype correlations from CREBBP point mutation screening in a cohort of 46 Rubinstein-Taybi syndrome patients**
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- XLVIII. **Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease**
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- XLIX. **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
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- L. **Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome**
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- LI. **Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability**
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- LII. **Exome sequencing reveals homozygous TRIM2 mutation in a patient with early onset CMT and bilateral vocal cord paralysis**
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- LIII. **CRIM1 haploinsufficiency causes defects in eye development in human and mouse**
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- LIV. **The phenotypic and molecular genetic spectrum of Alstrom syndrome in 44 Turkish kindreds and a literature review of Alstrom syndrome in Turkey**
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- LV. **Whole Exome Sequencing Identifies Three Novel Mutations in ANTXR1 in Families with GAPO Syndrome**
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D., Elcioglu N. H., et al.

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