Prof. Dr. HURİYE NURSEL ELÇİOĞLU

Kişisel Bilgiler

E-posta: nelcioglu@marmara.edu.tr
Web: https://avesis.marmara.edu.tr/nelcioglu

Uluslararası Araştırmacı ID'leri
ORCID: 0000-0003-0051-6720
Yoksis Araştırmacı ID: 8771

Eğitim Bilgileri

Tıpta Uzmanlık, İstanbul Üniversitesi, Tıp Fakültesi, Türkiye 1984 - 1989

Yaptığı Tezler

Tıpta Uzmanlık, Asfiksili term yenidoğanlarda böbrek fonksiyonlarının incelenmesi, İstanbul Üniversitesi, İstanbul Tıp Fakültesi, 1989

Araştırma Alanları

Sağlık Bilimleri

Akademik Unvanlar / Görevler

Prof. Dr., Marmara Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2004 - Devam Ediyor

Verdiği Dersler

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

Yönetilen Tezler

Elçioğlu H. N., Menteş A. R., Nörofibromatozis tip1 grubu çocuk hastaların ağız bulgularının ve ağız sağlığıyla ilgili yaşam kalitesinin incelenmesi, Tıpta Uzmanlık, E.KABACIOĞLU(Öğrenci), 2024
Haznedaroğlu E., Elçioğlu H. N., Noonan Sendromlu Çocuk Hastaların Oral Bulgularının Ağız Sağlığı ile İlgili Yaşam Kalitesine Etkisinin İncelenmesi, Diş Hekimliğinde Uzmanlık, S.İNCE(Öğrenci), 2024
Elçioğlu H. N., Çocuk Genetik Hastalıkları Polikliniğinde 2000-2023 yılları arasında takip ettiğimiz akondroplazi tanılı hastaların klinik izlemlerinin değerlendirilmesi, komplikasyonların yönetimi ve genetik danışma verilmesi, Tıpta Uzmanlık, S.ALIYEVA(Öğrenci), 2024

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, Tıpta Uzmanlık, E.SÖNMEZ(Öğrenci), 2023

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, Tıpta Uzmanlık, A.FATİH(Öğrenci), 2023

Elçioğlu H. N., Çocuk Genetik Kliniğimizde 2010-2022 yılları arasında takip ettiğimiz Nörofibromatozis hastalarının klinik izlemlerinin değerlendirilmesi ve genetik danışma verilmesi, Tıpta Uzmanlık, M.CAN(Öğrenci), 2022

Menteş A. R., Elçioğlu H. N., Mukopolisakkaridoz grubu çocuk hastaların oral bulgularının ve yaşam kalitesi anketlerinin değerlendirilmesi, Diş Hekimliğinde Uzmanlık, V.ÖZALP(Öğrenci), 2022

ELÇİOĞLU H. N., Kliniğimizde 2000-2018 Yılları Arasında Takip Ettiğimiz Mukopolisakkaridoz Hastalarının Kardiyak Tutulumlarının Değerlendirilmesi, Tıpta Uzmanlık, M.KARIMOVA(Öğrenci), 2019

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

Elçioğlu H. N., Kliniğimizde 2000-2016yılları arasında takip ettiğimiz sanfilipposendromlu hastaların tanı, biyokimyasal parametreler, tedavi ve klinikizlemlerinin değerlendirilmesi ve genetik danışma verilmesi, Tıpta Uzmanlık, B.NOYAN(Öğrenci), 2016

Elçioğlu H. N., Çocuk genetik hastalıkları polikliniğimize 2000-2015 yıllarında başvuran osteogenezis imperfektalı hastaların retrospektif değerlendirilmesi ve genetik danışmanlık verilmesi, Tıpta Uzmanlık, Ö.KAMER(Öğrenci), 2016

Elçioğlu H. N., Kliniğimizde takip ettiğimiz bardet biedl sendromlu hastaların fenotipik dağılımına yönelik klinik veri bankası oluşturulması ve genetik danışma verilmesi, Tıpta Uzmanlık, D.ALİKILIÇ(Öğrenci), 2015

Elçioğlu H. N., Herediter primer mikrosifalisi olan çocuklarda klinik değerlendirmeler, Tıpta Uzmanlık, M.SALİM(Öğrenci), 2013

Elçioğlu H., Kraniofasial malformasyonlara klinik ve genetik yaklaşımlar, Tıpta Uzmanlık, F.Sarı(Öğrenci), 2010

Elçioğlu H. N., Çocuklarda eklem hipermobilitesi ile seyreden kalıtsal bağ dokusu hastalıklarının klinik özellikleri, Tıpta Uzmanlık, B.ÇOLAK(Öğrenci), 2008

ELÇİOĞLU H. N., Nonsendromik konjenital yarık dudak/damak hastalıklarında metinletetrahidrofolat redüktaz gen polimorfizmin yeri, Tıpta Uzmanlık, A.Şemiç(Öğrenci), 2004

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

I. **Comparison of the natural course of clinical and radiologic features in 13 patients with TRPV4-related skeletal dysplasias.**
Güneş N., Alkaya D. U., Kurugoğlu S., Özyalvaç N., Bursalı A., Elçioğlu N. H., Tüysüz B.
Pediatric radiology, 2025 (SCI-Expanded)

II. **Clinical and molecular spectrum along with genotype-phenotype correlation of 25 patients diagnosed with 3 M syndrome: a study from Turkey.**
Akalın A., Özalkak Ş., Yıldırım R., Karakaya A. A., Kolbaşı B., Durmuşalioğlu E. A., Kökali F., Ürel-Demir G., Öz V., Ünal E., et al.
European journal of pediatrics, cilt.184, sa.1, ss.68, 2024 (SCI-Expanded)

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.
American Journal of Medical Genetics, Part A, cilt.194, sa.10, 2024 (SCI-Expanded)

IV. **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.
Nucleic acids research, cilt.52, 2024 (SCI-Expanded)

V. **Clinical and Molecular Characterization of Mucopolysaccharidosis Type 3A and 3B in a Turkish Series**

Noyan B., Elcioglu N. H., Tebani A., Bekri S.
Molecular Syndromology, cilt.15, sa.3, ss.194-201, 2024 (SCI-Expanded)

VI. **Autosomal recessive otospondylo-mega-epiphyseal dysplasia: Comprehensive clinical review of a pediatric cohort**
MUTLU H., ELÇİOĞLU H. N., Kiliç E.
Clinical Dysmorphology, cilt.32, sa.4, ss.151-155, 2023 (SCI-Expanded)

VII. **Management of acute metabolic crisis in TANGO2 deficiency: A case report**
Yllmaz-Gümüş E., ELÇİOĞLU H. N., Genç E., Arlcl Ş., Öztürk G., Yaplcl Ö., Akalln F., ÖZTÜRK HİŞMİ B.
Journal of Pediatric Endocrinology and Metabolism, 2023 (SCI-Expanded)

VIII. **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, cilt.14, ss.475-480, 2022 (SCI-Expanded)

IX. **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.
MOLECULAR VISION, cilt.28, ss.57-69, 2022 (SCI-Expanded)

X. **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, cilt.101, sa.5-6, ss.559-564, 2022 (SCI-Expanded)

XI. **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, cilt.59, sa.4, ss.377-384, 2022 (SCI-Expanded)

XII. **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, cilt.108, sa.10, ss.1981-2005, 2021 (SCI-Expanded)

XIII. **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, cilt.64, sa.10, 2021 (SCI-Expanded)

XIV. **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, cilt.185, sa.8, ss.2325-2334, 2021 (SCI-Expanded)

XV. **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, cilt.140, sa.8, ss.1229-1239, 2021 (SCI-Expanded)

XVI. **Genotype-phenotype correlations in hereditary multiple exostoses**
Akalin I., Kurosawa K., Yeter B., Akbas E., Nishimura G., Wuyts W., Elcioglu N. H.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.28, sa.SUPPL 1, ss.834, 2020 (SCI-Expanded)

XVII. **Jeune Syndrome with a novel DYNC2H1 mutation**
Elcioglu N. H., Yeter B., Xue J. Y., Wang Z., Guo L., Nishimura G., Ikegawa S.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.28, sa.SUPPL 1, ss.835-836, 2020 (SCI-Expanded)

XVIII. **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.
Journal of intellectual disability research : JIDR, cilt.64, sa.12, ss.956-969, 2020 (SCI-Expanded)

XIX. **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.
CALCIFIED TISSUE INTERNATIONAL, cilt.107, ss.96-103, 2020 (SCI-Expanded)

XX. **The genomic and clinical landscape of fetal akinesia**
Pergande M., Motameny S., Oezdemir O., Kreutzer M., Wang H., Daimagueler H., Becker K., Karakaya M., Ehrhardt H., Elcioglu N., et al.
GENETICS IN MEDICINE, cilt.22, sa.3, ss.511-523, 2020 (SCI-Expanded)

XXI. **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, cilt.34, sa.10, ss.1873-1879, 2019 (SCI-Expanded)

XXII. **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., Helvacioglu D., Güran T., et al.
HORMONE RESEARCH IN PAEDIATRICS, cilt.91, ss.384, 2019 (SCI-Expanded)

XXIII. **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.
HUMAN MUTATION, cilt.40, sa.8, ss.1084-1100, 2019 (SCI-Expanded)

XXIV. **The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance**
Pehlivan D., Bayram Y., Gunes N., Akdemir Z. C., Shukla A., Bierhals T., Tabakci B., Sahin Y., Gezdirici A., Fatih J. M., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.105, sa.1, ss.132-150, 2019 (SCI-Expanded)

XXV. **Ptosis as a unique hallmark for autosomal recessive WNT1-associated osteogenesis imperfecta**
Nampoothiri S., Guillemyn B., Elcioglu N., Jagadeesh S., Yesodharan D., Suresh B., Turan S., Symoens S., Malfait F.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.179, sa.6, ss.908-914, 2019 (SCI-Expanded)

XXVI. **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.
GENETICS IN MEDICINE, cilt.21, sa.6, ss.1295-1307, 2019 (SCI-Expanded)

XXVII. **Does the clinical phenotype of mucolipidosis-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., Radhakrishnan N., Kappanayil M., et al.
CLINICAL DYSMORPHOLOGY, cilt.28, sa.1, ss.7-16, 2019 (SCI-Expanded)

XXVIII. **Progressive pseudorheumotoid dysplasia: A presentation of four cases with slow and rapid progression and effects of early rehabilitation program**
Giray E., YAĞCI İ., ELÇİOĞLU H. N.
TURKISH JOURNAL OF PHYSICAL MEDICINE AND REHABILITATION, cilt.65, sa.3, ss.290-297, 2019 (SCI-Expanded)

XXIX. **Correction: Loss of the BMP Antagonist, SMOC-1, Causes Ophthalmo-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., Morrison H., et al.
PLoS genetics, cilt.14, sa.12, 2018 (SCI-Expanded)

XXX. **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.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.176, sa.12, ss.2740-2750, 2018 (SCI-Expanded)

XXXI. **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, cilt.13, sa.11, 2018 (SCI-Expanded)

XXXII. **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.

HUMAN MUTATION, cilt.39, sa.9, ss.1226-1237, 2018 (SCI-Expanded)

XXXIII. **Mutations in TOP3A Cause a Bloom Syndrome-like Disorder**

Martin C., Sarlos K., Logan C. V., Thakur R. S., Parry D. A., Bizard A. H., Leitch A., Cleal L., Ali N. S., Al-Owain M. A., et al.

AMERICAN JOURNAL OF HUMAN GENETICS, cilt.103, sa.2, ss.221-231, 2018 (SCI-Expanded)

XXXIV. **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., Tayfun G. A., et al.

AMERICAN JOURNAL OF HUMAN GENETICS, cilt.103, sa.1, ss.115-124, 2018 (SCI-Expanded)

XXXV. **Dysosteosclerosis is also caused by TNFRSF11A mutation**

Guo L., Elcioglu N. H., Karalar O. K., Topkar M. O., Wang Z., Sakamoto Y., Matsumoto N., Miyake N., Nishimura G., Ikegawa S.

JOURNAL OF HUMAN GENETICS, cilt.63, sa.6, ss.769-774, 2018 (SCI-Expanded)

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

Colombo E. A., Elcioglu N. H., Graziano C., Farinelli P., Di Fede E., Neri I., Facchini E., Greco M., Gervasini C., Larizza L.

JOURNAL OF CLINICAL IMMUNOLOGY, cilt.38, sa.4, ss.494-502, 2018 (SCI-Expanded)

XXXVII. **Rothmund-Thomson Syndrome: Insights from New Patients on the Genetic Variability Underpinning Clinical Presentation and Cancer Outcome**

Colombo E. A., Locatelli A., Cubells Sanchez L., Romeo S., Elcioglu N. H., Maystadt I., Esteve Martinez A., Sironi A., Fontana L., Finelli P., et al.

INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES, cilt.19, sa.4, 2018 (SCI-Expanded)

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

Michaud V., Lasseaux E., Plaisant C., Verloes A., Perdomo-Trujillo Y., Hamel C., Elcioglu N. H., Leroy B., Kaplan J., Jouk P., et al.

PIGMENT CELL & MELANOMA RESEARCH, cilt.30, sa.6, ss.563-570, 2017 (SCI-Expanded)

XXXIX. **Identification of biallelic EXTL3 mutations in a novel type of spondylo-epi-metaphyseal dysplasia**

Guo L., Elcioglu N. H., Mizumoto S., Wang Z., Noyan B., Albayrak H. M., Yamada S., Matsumoto N., Miyake N., Nishimura G., et al.

JOURNAL OF HUMAN GENETICS, cilt.62, sa.8, ss.797-801, 2017 (SCI-Expanded)

XL. **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.

HUMAN GENETICS, cilt.136, sa.7, ss.821-834, 2017 (SCI-Expanded)

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

Klingbeil K. D., Greenland C. M., Arslan S., Paneque A. L., GÜRKAN H., Ulusal S. D., Maroofian R., Carrera-Gonzalez A., Montufar-Armendariz S., Paredes R., et al.

INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, cilt.98, ss.59-63, 2017 (SCI-Expanded)

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

Bayram Y., White J. J., Elcioglu N., Cho M. T., Zadeh N., Gedikbasi A., Palanduz S., Ozturk S., Cefle K., KASAPÇOPUR Ö., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.101, sa.1, ss.149-156, 2017 (SCI-Expanded)

XLIII. **Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases**
Ranza E., Huber C., Levin N., Baujat G., Bole-Feysot C., Nitschke P., Masson C., Alanay Y., Al-Gazali L., Bitoun P., et al.
CLINICAL GENETICS, cilt.91, sa.6, ss.868-880, 2017 (SCI-Expanded)

XLIV. **Novel and recurrent XYLT1 mutations in two Turkish families with Desbuquois dysplasia, type 2**
Guo L., Elcioglu N. H., Iida A., Demirkol Y. K., Aras S., Matsumoto N., Nishimura G., Miyake N., Ikegawa S.
JOURNAL OF HUMAN GENETICS, cilt.62, sa.3, ss.447-451, 2017 (SCI-Expanded)

XLV. **Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2**
Boegershausen N., Gatinois V., Riehmer V., Kayserili H., Becker J., Thoenes M., Simsek-Kiper P. O., Barat-Houari M., Elcioglu N. H., Wieczorek D., et al.
HUMAN MUTATION, cilt.37, sa.9, ss.847-864, 2016 (SCI-Expanded)

XLVI. **Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin**
Bayram Y., Karaca E., Akdemir Z. C., Yilmaz E. O., Tayfun G. A., AYDIN H., Torun D., Bozdogan S. T., Gezdirici A., Isikay S., et al.
JOURNAL OF CLINICAL INVESTIGATION, cilt.126, sa.2, ss.762-778, 2016 (SCI-Expanded)

XLVII. **A THANATOPHORIC DYSPLASIA TYPE I CASE WITH A FGFR3 P.R248C MUTATION AND SURVIVAL BEYOND THE NEONATAL PERIOD**
Sahin S., Ograg H., Aslan E. A., Akcan A. B., Turkmen M. K., Moosa S., Elcioglu N. H.
GENETIC COUNSELING, cilt.27, sa.4, ss.513-517, 2016 (SCI-Expanded)

XLVIII. **TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism**
Harley M. E., Murina O., Leitch A., Higgs M. R., Bicknell L. S., Yigit G., Blackford A. N., Zlatanou A., Mackenzie K. J., Reddy K., et al.
NATURE GENETICS, cilt.48, sa.1, ss.36-45, 2016 (SCI-Expanded)

XLIX. **A thanatophoric dysplasia type i case with a fgfr3 p.r248c mutation and survival ibeyond the neonatal period**
Sahin S., Ograg H., Atasaslan E., Akcan A., Kaynak Turkmen M., Moosa S., ELÇİOĞLU H. N.
Genetic Counseling, cilt.27, sa.4, ss.513-517, 2016 (SCI-Expanded)

L. **A role for repressive complexes and H3K9 di-methylation in PRDM5-associated brittle cornea syndrome**
Porter L. F., Galli G. G., Williamson S., Selley J., Knight D., Elcioglu N., Aydin A., Elcioglu M., Venselaar H., Lund A. H., et al.
HUMAN MOLECULAR GENETICS, cilt.24, sa.23, ss.6565-6579, 2015 (SCI-Expanded)

LI. **Insights into genotype-phenotype correlations from CREBBP point mutation screening in a cohort of 46 Rubinstein-Taybi syndrome patients**
Spena S., Milani D., Rusconi D., Negri G., Colapietro P., Elcioglu N., Bedeschi F., Pilotta A., Spaccini L., Ficcadenti A., et al.
CLINICAL GENETICS, cilt.88, sa.5, ss.431-440, 2015 (SCI-Expanded)

LII. **Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease**
Karaca E., Harel T., Pehlivan D., Jhangiani S. N., Gambin T., Akdemir Z. C., Gonzaga-Jauregui C., Erdin S., Bayram Y., Campbell I. M., et al.
NEURON, cilt.88, sa.3, ss.499-513, 2015 (SCI-Expanded)

LIII. **Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome**
Atik T., Koparir A., Bademci G., Foster J., Altunoglu U., Mutlu G. Y., Bowdin S., Elcioglu N., Tayfun G. A., Atik S. S., et al.
ORPHANET JOURNAL OF RARE DISEASES, cilt.10, 2015 (SCI-Expanded)

LIV. **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**
Boegershausen N., Tsai I., Pohl E., ŞİMŞEK KİPER P. Ö., Beleggia F., Percin E. F., Keupp K., Matchan A., Milz E., Alanay

Y., et al.
JOURNAL OF CLINICAL INVESTIGATION, cilt.125, sa.9, ss.3585-3599, 2015 (SCI-Expanded)

LV. **Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability**
Rosin N., Elcioglu N. H., Beleggia F., Isguven P., Altmueller J., Thiele H., Steindl K., Joset P., Rauch A., Nuernberg P., et al.
HUMAN MOLECULAR GENETICS, cilt.24, sa.13, ss.3708-3717, 2015 (SCI-Expanded)

LVI. **Exome sequencing reveals homozygous TRIM2 mutation in a patient with early onset CMT and bilateral vocal cord paralysis**
Pehlivan D., Akdemir Z. C., Karaca E., Bayram Y., Jhangiani S., Yildiz E. P., Muzny D., ULUÇ K., Gibbs R. A., Elcioglu N., et al.
HUMAN GENETICS, cilt.134, sa.6, ss.671-673, 2015 (SCI-Expanded)

LVII. **CRIM1 haploinsufficiency causes defects in eye development in human and mouse**
Beleggia F., Li Y., Fan J., Elcioglu N. H., Toker E., Wieland T., Maumenee I. H., Akarsu N. A., Meitinger T., Strom T. M., et al.
HUMAN MOLECULAR GENETICS, cilt.24, sa.8, ss.2267-2273, 2015 (SCI-Expanded)

LVIII. **The phenotypic and molecular genetic spectrum of Alstrom syndrome in 44 Turkish kindreds and a literature review of Alstrom syndrome in Turkey**
Ozanturk A., Marshall J. D., Collin G. B., DÜZENLİ S., Marshall R. P., Candan S., Tos T., Esen I., TAŞKESEN M., Cayir A., et al.
JOURNAL OF HUMAN GENETICS, cilt.60, sa.1, ss.1-9, 2015 (SCI-Expanded)

LIX. **Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway**
Acuna-Hidalgo R., Schanze D., Kariminejad A., Nordgren A., Kariminejad M. H., Conner P., Grigelioniene G., Nilsson D., Nordenskjold M., Wedell A., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.95, sa.3, ss.285-293, 2014 (SCI-Expanded)

LX. **Whole Exome Sequencing Identifies Three Novel Mutations in ANTXR1 in Families with GAPO Syndrome**
Bayram Y., Pehlivan D., Karaca E., Gambin T., Jhangiani S. N., Erdin S., Gonzaga-Jauregui C., Wiszniewski W., Muzny D., Elcioglu N. H., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.164, sa.9, ss.2328-2334, 2014 (SCI-Expanded)

LXI. **Authors' response: evaluation of choroidal thickness among patients with oculocutaneous albinism**
Karabas L., Esen F., ÇELİKER ATABERK H., Elcioglu N., ÇERMAN E., ERASLAN M., Kazokoglu H., ŞAHİN Ö.
BRITISH JOURNAL OF OPHTHALMOLOGY, cilt.98, sa.8, ss.1135-1136, 2014 (SCI-Expanded)

LXII. **Decreased subfoveal choroidal thickness and failure of emmetropisation in patients with oculocutaneous albinism**
Karabas L., Esen F., ÇELİKER ATABERK H., Elcioglu N., ÇERMAN E., ERASLAN M., Kazokoglu H., ŞAHİN Ö.
BRITISH JOURNAL OF OPHTHALMOLOGY, cilt.98, sa.8, ss.1087-1090, 2014 (SCI-Expanded)

LXIII. **CNS involvement in OFD1 syndrome: a clinical, molecular, and neuroimaging study**
Del Giudice E., Macca M., Imperati F., D'Amico A., Parent P., Pasquier L., Layet V., Lyonnet S., Stamboul-Darmency V., Thauvin-Robinet C., et al.
ORPHANET JOURNAL OF RARE DISEASES, cilt.9, 2014 (SCI-Expanded)

LXIV. **Mutations and polymorphisms in N-acetylgalactosamine-6-sulfate sulfatase gene in Turkish Morquio A patients**
Khedhiri S., Chkioua L., Elcioglu N., Laradi S., Miled A.
Pathologie Biologie, cilt.62, sa.1, ss.38-40, 2014 (SCI-Expanded)

LXV. **Molecular Analysis of Turkish Maroteaux-Lamy Patients and Identification of One Novel Mutation in the Arylsulfatase B (ARSB)Gene**
Zanetti A., Onenli-Mungan N., Elcioglu N., Ozbek M. N., KOR D., Lenzini E., Scarpa M., Tomanin R.
JIMD REPORTS, VOL 14, cilt.14, ss.1-9, 2014 (SCI-Expanded)

LXVI. **WERNER SYNDROME: CLINICAL EVALUATION OF TWO CASES AND A NOVEL MUTATION**
Mansur A. T., Elcioglu N. H., Demirci G. T.

GENETIC COUNSELING, cilt.25, sa.2, ss.119-127, 2014 (SCI-Expanded)

LXVII. **A comprehensive molecular study on coffin-siris and nicolaides-baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling**
Wieczorek D., Boegershausen N., Beleggia F., Steiner-Haldenstaett S., Pohl E., Li Y., Milz E., Martin M., Thiele H., Altmueller J., et al.
Human Molecular Genetics, cilt.22, sa.25, ss.5121-5135, 2013 (SCI-Expanded)

LXVIII. **Mutations in the gene encoding IFT dynein complex component WDR34 cause jeune asphyxiating thoracic dystrophy**
Schmidts M., Vodopiutz J., Christou-Savina S., Cortes C. R., McInerney-Leo A. M., Emes R. D., Arts H. H., TÜYSÜZ B., D'Silva J., Leo P. J., et al.
American Journal of Human Genetics, cilt.93, sa.5, ss.932-944, 2013 (SCI-Expanded)

LXIX. **Mutations in the interleukin receptor IL11RA cause autosomal recessive crouzon-like craniosynostosis**
Keupp K., Li Y., Vargel I., Hoischen A., Richardson R., Neveling K., Alanay Y., Uz E., Elcioğlu N., Rachwalski M., et al.
Molecular Genetics and Genomic Medicine, cilt.1, sa.4, ss.223-237, 2013 (SCI-Expanded)

LXX. **Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused by PAPSS2 Mutations**
Iida A., ŞİMŞEK KİPER P. Ö., Mizumoto S., Hoshino T., Elcioglu N., Horemuzova E., Geiberger S., Yesil G., Kayserili H., ÜTİNE G. E., et al.
HUMAN MUTATION, cilt.34, sa.10, ss.1381-1386, 2013 (SCI-Expanded)

LXXI. **Confirming genes influencing risk to cleft lip with/without cleft palate in a case-parent trio study**
Beaty T. H., Taub M. A., Scott A. F., Murray J. C., Marazita M. L., Schwender H., Parker M. M., Hetmanski J. B., Balakrishnan P., Mansilla M. A., et al.
HUMAN GENETICS, cilt.132, sa.7, ss.771-781, 2013 (SCI-Expanded)

LXXII. **Exome sequencing identifies DYNC2H1 mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement**
Schmidts M., Arts H. H., Bongers E. M. H. F., Yap Z., Oud M. M., Antony D., Duijkers L., Emes R. D., Stalker J., Yntema J. L., et al.
JOURNAL OF MEDICAL GENETICS, cilt.50, sa.5, ss.309-323, 2013 (SCI-Expanded)

LXXIII. **Mutations in Multidomain Protein MEGF8 Identify a Carpenter Syndrome Subtype Associated with Defective Lateralization**
Twigg S. R. F., Lloyd D., Jenkins D., Elcioglu N. E., Cooper C. D. O., Al-Sannaa N., Annagur A., Gillessen-Kaesbach G., Huening I., Knight S. J. L., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.91, sa.5, ss.897-905, 2012 (SCI-Expanded)

LXXIV. **Association between C677T and A1298C MTHFR gene polymorphism and nonsyndromic orofacial clefts in the Turkish population: a case-parent study**
Semic-Jusufagic A., Bircan R., Celebiler O., Erdim M., Akarsu N., Elcioglu N. H.
TURKISH JOURNAL OF PEDIATRICS, cilt.54, sa.6, ss.617-625, 2012 (SCI-Expanded)

LXXV. **Treacher Collins syndrome: clinical implications for the paediatrician-a new mutation in a severely affected newborn and comparison with three further patients with the same mutation, and review of the literature**
Schlump J., Stein A., Hehr U., Karen T., Moeller-Hartmann C., Elcioglu N. H., Bogdanova N., Woike H. F., Lohmann D. R., Felderhoff-Mueser U., et al.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.171, sa.11, ss.1611-1618, 2012 (SCI-Expanded)

LXXVI. **The diagnostic challenge of progressive pseudorheumatoid dysplasia (PPRD): A review of clinical features, radiographic features, and WISP3 mutations in 63 affected individuals**
Segarra N. G., Mittaz L., Campos-Xavier A. B., Bartels C. F., TÜYSÜZ B., ALANAY Y., Cimaz R., Cormier-Daire V., Di Rocco M., Duba H., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS, cilt.160C, sa.3, ss.217-229, 2012 (SCI-Expanded)

LXXVII. **PAPSS2 mutations cause autosomal recessive brachyolmia**

Miyake N., Elcioglu N. H., Iida A., Isguven P., Dai J., Murakami N., Takamura K., Cho T., Kim O., Hasegawa T., et al.
JOURNAL OF MEDICAL GENETICS, cilt.49, sa.8, ss.533-538, 2012 (SCI-Expanded)

LXXVIII. **Next generation sequencing identifies mutations in Atonal homolog 7 (ATOH7) in families with global eye developmental defects**
Khan K., Logan C. V., McKibbin M., Sheridan E., Elcioglu N. H., Yenice Ö., Parry D. A., Fernandez-Fuentes N., Abdelhamed Z. I. A., Al-Maskari A., et al.
HUMAN MOLECULAR GENETICS, cilt.21, sa.4, ss.776-783, 2012 (SCI-Expanded)

LXXIX. **A SHORT RIB POLYDACTYLY SYNDROME OVERLAPPING BOTH LETHAL AND NONLETHAL TYPES**
Yigiter A. B., Guducu N., Kavak Z. N., Isci H., Elcioglu N.
GENETIC COUNSELING, cilt.23, sa.2, ss.231-237, 2012 (SCI-Expanded)

LXXX. **Novel C16orf57 mutations in patients with Poikiloderma with Neutropenia: bioinformatic analysis of the protein and predicted effects of all reported mutations**
Colombo E. A., Elcioglu N. H., Yucelten D., Altunay I., Cetincelik U., Teti A., Del Fattore A., Luciani M., Sullivan S. K., Yan A. C., et al.
ORPHANET JOURNAL OF RARE DISEASES, cilt.7, 2012 (SCI-Expanded)

LXXXI. **Loss of the BMP Antagonist, SMOC-1, Causes Ophthalmo-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., Morrison H., et al.
PLOS GENETICS, cilt.7, sa.7, 2011 (SCI-Expanded)

LXXXII. **Carpenter Syndrome: Extended RAB23 Mutation Spectrum and Analysis of Nonsense-mediated mRNA Decay**
Jenkins D., Baynam G., De Catte L., Elcioglu N., Gabbett M. T., Hudgins L., Hurst J. A., Jehee F. S., Oley C., Wilkie A. O. M.
HUMAN MUTATION, cilt.32, sa.4, 2011 (SCI-Expanded)

LXXXIII. **Mutations in FKBP10 Cause Recessive Osteogenesis Imperfecta and Bruck Syndrome**
Kelley B. P., Malfait F., Bonafe L., Baldridge D., Homan E., Symoens S., Willaert A., Elcioglu N., Van Maldergem L., Verellen-Dumoulin C., et al.
JOURNAL OF BONE AND MINERAL RESEARCH, cilt.26, sa.3, ss.666-672, 2011 (SCI-Expanded)

LXXXIV. **Marie Unna Hereditary Hypotrichosis: A Turkish Family With Loss of Eyebrows and a U2HR Mutation**
Mansur A. T., Elcioglu N. H., Redler S., Serdar Z. A., ÇETİNEL Ş., Betz R. C., Akarsu N. A.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.152A, sa.10, ss.2628-2633, 2010 (SCI-Expanded)

LXXXV. **LRP4 Mutations Alter Wnt/beta-Catenin Signaling and Cause Limb and Kidney Malformations in Cenani-Lenz Syndrome**
Li Y., Pawlik B., Elcioglu N., Aglan M., Kayserili H., Yigit G., Percin F., Goodman F., Nuernberg G., Cenani A., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.86, sa.5, ss.696-706, 2010 (SCI-Expanded)

LXXXVI. **A report of a patient with duplication of 7p13 -> pter and deletion of 2p23 -> pter due to a maternal 2p;7p translocation**
Turkover B. B., Sayar C., Toksoy G., Elcioglu N.
TURKISH JOURNAL OF PEDIATRICS, cilt.51, sa.2, ss.174-179, 2009 (SCI-Expanded)

LXXXVII. **Mucopolysaccharidosis Type II: long-term follow-up after bone marrow transplantation**
Elcioglu N. H., Vellodi A., Hall C. M.
INTERNATIONAL JOURNAL OF CLINICAL PHARMACOLOGY AND THERAPEUTICS, cilt.47, 2009 (SCI-Expanded)

LXXXVIII. **A NOVEL LOSS-OF-FUNCTION MUTATION IN THE GNS GENE CAUSES SANFILIPPO SYNDROME TYPE D**
Elcioglu N. H., Pawlik B., Colak B., Beck M., Wollnik B.
GENETIC COUNSELING, cilt.20, sa.2, ss.133-139, 2009 (SCI-Expanded)

LXXXIX. **Molecular diagnosis of oculocutaneous albinism: new mutations in the OCA1-4 genes and practical aspects**
Rooryck C., Morice-Picard F., Elcioglu N. H., Lacombe D., Taieb A., Arveiler B.
PIGMENT CELL & MELANOMA RESEARCH, cilt.21, sa.5, ss.583-587, 2008 (SCI-Expanded)

XC. **Mutational Spectrum of the Oral-Facial-Digital Type I Syndrome: A Study on a Large Collection of**

Patients

Prattichizzo C., Macca M., Novelli V., Giorgio G., Barra A., Franco B.

HUMAN MUTATION, cilt.29, sa.10, ss.1237-1246, 2008 (SCI-Expanded)

XCI. **Adult height in Turkish patients with Turner syndrome without growth hormone treatment**

BEREKET A., Turan S., Elcioglu N., Hacihanefioglu S., Memioglu N., Bas F., Bundak R., Darendeliler F., Guenoez H., Saka N., et al.

TURKISH JOURNAL OF PEDIATRICS, cilt.50, sa.5, ss.415-417, 2008 (SCI-Expanded)

XCII. **Spondylocheiro dysplastic form of the Ehlers-Danlos syndrome - An autosomal-recessive entity caused by mutations in the zinc transporter gene SLC39A13**

Giunta C., Elcioglu N. H., Albrecht B., Eich G., Chambaz C., Janecke A. R., Yeowell H., Weis M., Eyre D. R., Kraenzlin M., et al.

AMERICAN JOURNAL OF HUMAN GENETICS, cilt.82, sa.6, ss.1290-1305, 2008 (SCI-Expanded)

XCIII. **Mucopolysaccharidosis disorders - Abstracts**

Elcioglu N., Colak B., Beck M.

ACTA PAEDIATRICA, cilt.97, ss.97, 2008 (SCI-Expanded)

XCIV. **Colobomatous macrophthalmia with microcornea syndrome maps to the 2p23-p16 region**

Elcioglu N. H., Akin B., Toker E., Elcioglu M., Kaya A., Tuncali T., Wollnik B., Hornby S., Akarsu N. A.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.143A, sa.12, ss.1308-1312, 2007 (SCI-Expanded)

XCV. **IFT80, which encodes a conserved intraflagellar transport protein, is mutated in Jeune asphyxiating thoracic dystrophy**

Beales P. L., Bland E., Tobin J. L., Bacchelli C., Tuysuz B., Hill J., Rix S., Pearson C. G., Kai M., Hartley J., et al.

NATURE GENETICS, cilt.39, sa.6, ss.727-729, 2007 (SCI-Expanded)

XCVI. **Novel and recurrent KIND1 mutations in two patients with Kindler syndrome and severe mucosal involvement**

Mansur A. T., Elcioglu N. H., Aydingoz I. E., Akkaya A. D., Serdar Z. A., Herz C., Bruckner-Tuderman L., Has C.

ACTA DERMATO-VENEREOLOGICA, cilt.87, sa.6, ss.563-565, 2007 (SCI-Expanded)

XCVII. **Molecular characterisation of six patients with prolidase deficiency: identification of the first small duplication in the prolidase gene and of a mutation generating symptomatic and asymptomatic outcomes within the same family**

Lupi A., Rossi A., Campari E., Pecora F., Lund A. M., Elcioglu N. H., Gultepe M., Di Rocco M., Cetta G., Forlino A.

JOURNAL OF MEDICAL GENETICS, cilt.43, sa.12, 2006 (SCI-Expanded)

XCVIII. **Tracheobronchial calcification associated with Keutel syndrome**

Oezdemir N., Ersu R., Akalin F., Karadag B. T., Kut A., Karakoc F., Elcioglu N., Dagli E.

TURKISH JOURNAL OF PEDIATRICS, cilt.48, sa.4, ss.357-361, 2006 (SCI-Expanded)

XCIX. **Prenatal diagnosis of Wolf-Hirschhorn syndrome (4p-) in association with congenital diaphragmatic hernia, cystic hygroma and IUGR**

Basgul A., Kavak Z., Akman I., Basgul A., Gokaslan H., ELÇİOĞLU H. N.

Clinical and Experimental Obstetrics and Gynecology, cilt.33, sa.2, ss.105-106, 2006 (SCI-Expanded)

C. **When Mucolipidosis III meets Mucolipidosis II: GNPTA gene mutations in 24 patients**

Bargal R., Zeigler M., Abu-Libdeh B., Zuri V., Mandel H., Ben Neriah Z., Stewart F., Elcioglu N., Hindi T., Le Merrer M., et al.

MOLECULAR GENETICS AND METABOLISM, cilt.88, sa.4, ss.359-363, 2006 (SCI-Expanded)

CI. **Prolidase deficiency associated with hemoglobin O trait and microcytic anemia**

Aytug A., Ergun T., Ratip S., Elcioglu N., Gultepe M., Mercan E., Gurbuz O.

INTERNATIONAL JOURNAL OF DERMATOLOGY, cilt.45, sa.7, ss.867-868, 2006 (SCI-Expanded)

CII. **BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus**

Stoetzel C., Laurier V., Davis E., Muller J., Rix S., Badano J., Leitch C., Salem N., Chouery E., Corbani S., et al.

NATURE GENETICS, cilt.38, sa.5, ss.521-524, 2006 (SCI-Expanded)

CIII. **Antenatal diagnosis of velocardiofacial syndrome by 3D ultrasonography**

Basgul A., Kavak Z., Kotiloglu E., Akman I., Akalin F., Elcioglu N., Sav A.

JOURNAL OF PERINATAL MEDICINE, cilt.34, sa.2, ss.177-178, 2006 (SCI-Expanded)

CIV. **Novel truncating and missense mutations of the KCC3 gene associated with Andermann syndrome**
Uyanik G., Elcioglu N., Penzien J., Gross C., Yilmaz Y., Olmez A., Demir E., Wahl D., Scheglmann K., Winner B., et al.
NEUROLOGY, cilt.66, sa.7, ss.1044-1048, 2006 (SCI-Expanded)

CV. **Muir-Torre syndrome**
Tuncel A., Çelebiler Ö., ELÇİOĞLU H. N.
European Journal of Plastic Surgery, cilt.27, sa.5, ss.241-243, 2004 (SCI-Expanded)

CVI. **Progeria: A new kind of Laminopathy Report of the First European Symposium on Progeria and creation of EURO-Progeria, a European Consortium on Progeria and related disorders**
Brune T., Bonne G., Denecke J., Elcioglu N., Hennekam R. C., Marquardt T., Ozgen H., Stamsnijder M., Steichen E., Steinmann B., et al.
Pediatric Endocrinology Reviews, cilt.2, sa.1, ss.39-45, 2004 (SCI-Expanded)

CVII. **Recessive omodysplasia: five new cases and review of the literature**
Elcioglu N., Gustavson K., Wilkie A., Yuksel-Apak M., Spranger J.
PEDIATRIC RADIOLOGY, cilt.34, sa.1, ss.75-82, 2004 (SCI-Expanded)

CVIII. **Neonatal Marfan syndrome caused by an exon 25 mutation of the Fibrillin-1 gene**
Elcioglu N., Akalin F., Elcioglu M., Comeglio P., Child A.
GENETIC COUNSELING, cilt.15, sa.2, ss.219-225, 2004 (SCI-Expanded)

CIX. **Metatropic dysplasia lethal variants**
Hall C., Elcioglu N.
PEDIATRIC RADIOLOGY, cilt.34, sa.1, ss.66-74, 2004 (SCI-Expanded)

CX. **Short rib-polydactyly syndrome: a case report**
Turkmen M., Temocin K., Acar C., Levi E., Karaman C., Inan G., Elcioglu N.
TURKISH JOURNAL OF PEDIATRICS, cilt.45, sa.4, ss.359-362, 2003 (SCI-Expanded)

CXI. **Colobomatous macrophthalmia with microcornea syndrome: Report of a new pedigree**
Toker E., Elcioglu N., Ozcan E., Yenice Ö., Ogut M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.121A, sa.1, ss.25-30, 2003 (SCI-Expanded)

CXII. **Alkaptonuria caused by compound heterozygote mutations**
Elcioglu N., Aytug A., Muller C., Gurbuz O., Ergun T., Kotiloglu E., Elcioglu M.
GENETIC COUNSELING, cilt.14, sa.2, ss.207-213, 2003 (SCI-Expanded)

CXIII. **Diagnostic dilemmas in the short rib - Polydactyly syndrome group**
Elcioglu N., Hall C.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.111, sa.4, ss.392-400, 2002 (SCI-Expanded)

CXIV. **Spondyloepimetaphyseal dysplasia with multiple dislocations (Hall type): three further cases and evidence of autosomal dominant inheritance**
Hall C., Elcioglu N., MacDermot K., Offiah A., Winter R.
JOURNAL OF MEDICAL GENETICS, cilt.39, sa.9, ss.666-670, 2002 (SCI-Expanded)

CXV. **Human Piebaldism: Six Novel Mutations of the Proto-oncogene KIT**
Syrris P., Heathcote K., Carrozzo R., Devriendt K., Elcioglu N., Garrett C., McEntagart M., Carter N. D.
HUMAN MUTATION, cilt.20, sa.3, 2002 (SCI-Expanded)

CXVI. **Dysosteosclerosis: a report of three new cases and evolution of the radiological findings**
Elcioglu N., Vellodi A., Hall C.
JOURNAL OF MEDICAL GENETICS, cilt.39, sa.8, ss.603-607, 2002 (SCI-Expanded)

CXVII. **Genetic and mutational analyses of a large multiethnic Bardet-Biedl cohort reveal a minor involvement of BBS6 and delineate the critical intervals of other loci**
Beales P., Katsanis N., Lewis R., Ansley S., Elcioglu N., Raza J., Woods M., Green J., Parfrey P., Davidson W., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.68, sa.3, ss.606-616, 2001 (SCI-Expanded)

CXVIII. **Pseudo-Torch syndrome: the autosomal recessive syndrome of microcephaly, intracranial calcification and dysmorphism, resembling intrauterine infection.. An additional observation**
Elcioglu N., Alper G., Yilmaz Y.
JOURNAL OF MEDICAL GENETICS, cilt.37, 2000 (SCI-Expanded)

CXIX. **Fraser syndrome: Diagnosed in a 50-year-old museum specimen**

Elcioglu H. N., Berry A.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.94, sa.3, ss.262-264, 2000 (SCI-Expanded)

CXX. **A variant of Cenani-Lenz type syndactyly**
SEVEN M., Yuksel A., Ozkilic A., Elcioglu N.
GENETIC COUNSELING, cilt.11, sa.1, ss.41-47, 2000 (SCI-Expanded)

CXXI. **Peroxisomal disorders: Clinical and biochemical studies in 15 children and prenatal diagnosis in 7 families**
Steinberg S., Elcioglu N., Slade C., Sankaralingam A., Dennis N., Mohammed S., Fensom A.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.85, sa.5, ss.502-510, 1999 (SCI-Expanded)

CXXII. **New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey**
Beales P., Elcioglu N., Woolf A., Parker D., Flinter F.
JOURNAL OF MEDICAL GENETICS, cilt.36, sa.6, ss.437-446, 1999 (SCI-Expanded)

CXXIII. **A Rapp-Hodgkin like syndrome in three sibs: Clinical, dental and dermatoglyphic study**
Atasu M., Akesi S., Elcioglu N., Yatmaz P., Ertas E.
CLINICAL DYSMORPHOLOGY, cilt.8, sa.2, ss.101-110, 1999 (SCI-Expanded)

CXXIV. **Persistent mullerian duct syndrome - A case report**
Erk A., Ozeren S., Ozbay O., Vural B., Elcioglu N.
JOURNAL OF REPRODUCTIVE MEDICINE, cilt.44, sa.2, ss.135-138, 1999 (SCI-Expanded)

CXXV. **Maternal systemic lupus erythematosus and chondrodysplasia punctata in two sibs: phenocopy or coincidence?**
Elcioglu N., Hall C.
JOURNAL OF MEDICAL GENETICS, cilt.35, sa.8, ss.690-694, 1998 (SCI-Expanded)

CXXVI. **A distinct form of spondyloepimetaphyseal dysplasia with multiple dislocations**
Hall C., Elcioglu N., Shaw D.
JOURNAL OF MEDICAL GENETICS, cilt.35, sa.7, ss.566-572, 1998 (SCI-Expanded)

CXXVII. **A lethal skeletal dysplasia with features of chondrodysplasia punctata and osteogenesis imperfecta: an example of Astley-Kendall dysplasia. Further delineation of a rare genetic disorder**
Elcioglu N., Hall C.
JOURNAL OF MEDICAL GENETICS, cilt.35, sa.6, ss.505-507, 1998 (SCI-Expanded)

CXXVIII. **Jeune's asphyxiating thoracic dystrophy of the newborn**
Sarimurat N., Elcioglu N., Tekant G., Elicevik M., Yeker D.
EUROPEAN JOURNAL OF PEDIATRIC SURGERY, cilt.8, sa.2, ss.100-101, 1998 (SCI-Expanded)

CXXIX. **Spondylometaphyseal dysplasia Sedaghatian type**
Elcioglu N., Hall C.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.76, sa.5, ss.410-414, 1998 (SCI-Expanded)

CXXX. **Temporal aspects in craniometaphyseal dysplasia: Autosomal recessive type**
Elcioglu N., Hall C.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.76, sa.3, ss.245-251, 1998 (SCI-Expanded)

CXXXI. **FISH analysis in patients with clinical diagnosis of Williams syndrome**
Elcioglu N., Mackie-Ogilvie C., Daker M., Berry A.
ACTA PAEDIATRICA, cilt.87, sa.1, ss.48-53, 1998 (SCI-Expanded)

CXXXII. **Tuberous sclerosis: Clinical evaluation in a family and implications for genetic counseling**
Elcioglu N., Karatekin G., Elcioglu M., Nuhoglu A., Cenani A.
GENETIC COUNSELING, cilt.9, sa.2, ss.131-138, 1998 (SCI-Expanded)

CXXXIII. **Partial trisomy of 15q due to inserted inverted duplication**
Elcioglu N., Fear C., Berry A.
CLINICAL GENETICS, cilt.52, sa.6, ss.442-445, 1997 (SCI-Expanded)

CXXXIV. **Dermatoglyphics in patients with Cenani-Lenz type syndactyly: Studies in a new case**
Elcioglu N., Atasu M., Cenani A.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.70, sa.4, ss.341-345, 1997 (SCI-Expanded)

CXXXV. **Monozygotic twins discordant for the Oculo-oto-radial syndrome (IVIC syndrome)**

Elcioglu N., Berry A.
GENETIC COUNSELING, cilt.8, sa.3, ss.201-206, 1997 (SCI-Expanded)

CXXXVI. **RENAL-FUNCTION DISORDERS IN NEWBORNS WITH PERINATAL ASPHYXIA**
ELCIOGLU N., SIRIN A., CAN G., EMRE S., NAYIR A., TANMAN F.
GEBURTSHILFE UND FRAUENHEILKUNDE, cilt.55, sa.3, ss.160-163, 1995 (SCI-Expanded)

CXXXVII. **THE FREQUENCY OF GENETIC EYE DISEASES IN A GENETIC-COUNSELING CENTER**
ELCIOGLU N., ELCIOGLU M., FUHRMANN W.
GENETIC COUNSELING, cilt.6, sa.4, ss.329-342, 1995 (SCI-Expanded)

CXXXVIII. **AN EPIDEMIOLOGIC APPROACH TO ACUTE-RENAL-FAILURE IN CHILDREN**
GÖKÇAY E. G., EMRE S., TANMAN F., SIRIN A., ELCIOGLU N., DOLUNAY G.
JOURNAL OF TROPICAL PEDIATRICS, cilt.37, sa.4, ss.191-193, 1991 (SCI-Expanded)

Diğer Dergilerde Yayınlanan Makaleler

I. **GestaltMatcher Database - A global reference for facial phenotypic variability in rare human diseases.**
Lesmann H., Hustinx A., Moosa S., Klinkhammer H., Marchi E., Caro P., Abdelrazek I. M., Pantel J. T., Ten Hagen M., Thong M., et al.
medRxiv : the preprint server for health sciences, 2024 (Hakemli Dergi)

II. **Developmental genomics of limb malformations: Allelic series in association with gene dosage effects contribute to the clinical variability**
Duan R., Hijazi H., Gulec E. Y., Eker H. K., Costa S. R., Sahin Y., Ocak Z., Isikay S., Ozalp O., Bozdogan S., et al.
HUMAN GENETICS AND GENOMICS ADVANCES, cilt.3, sa.4, 2022 (ESCI)

III. **Erken yaşlanma hastalığı Progeria: Hücresel mekanizma ve yeni Tedavi stratejileri**
Elçioğlu H. N.
Türkiye Klinikleri Pediatri Dergisi, cilt.1, sa.1, ss.157-164, 2021 (Scopus)

IV. **Novel and recurrent COL11A1 and COL2A1 mutations in the Marshall-Stickler syndrome spectrum**
Guo L., Elcioglu N. H., Wang Z., Demirkol Y. K., Isguven P., Matsumoto N., Nishimura G., Miyake N., Ikegawa S.
HUMAN GENOME VARIATION, cilt.4, 2017 (ESCI)

V. **Nijmegen-Breakage Syndrome; Two Siblings Presenting with Different Phenotypes**
Kiykim A., AYDINER E., ÖĞÜLÜR İ., BARIŞ S., Ozen A. O., Serifov K., Bademci G., Tekin M., Elcioglu N. H., Barlan I.
ASTIM ALLERJI IMMUNOLOJI, cilt.14, sa.2, ss.98-102, 2016 (ESCI)

VI. **Successful treatment of pityriasis lichenoides with narrow band ultraviolet B therapy in a patient with KID syndrome a case report**
salman A., SEÇKİN GENÇOSMANOĞLU D., YÜCELTEN A. D., elcioğlu n., Richard G., DEMİRKESEN C.
Dermatology Online Journal, cilt.22, sa.5, 2016 (Scopus)

VII. **Successful treatment of pityriasis lichenoides chronica with narrow-band ultraviolet B therapy in a patient with keratitis-ichthyosis-deafness syndrome: A case report**
SALMAN A., Gencosmanoglu D. S., YÜCELTEN A. D., Elcioglu N., Richard G., Demirkesen C.
Dermatology Online Journal, cilt.22, sa.5, 2016 (Scopus)

VIII. **Consanguineous Marriage in Turkey and its Meaning for Genetic Counselling Verwandtenehen in der Türkei und ihre Bedeutung für die genetische Praxis**
Elçioglu N., Cenani A.
Munchener Medizinische Wochenschrift, cilt.139, sa.48, ss.715-718, 1997 (Scopus)

IX. **A survey amongst German human genetic specialists about the situation of genetic counselling of Turkish patients in Germany EINE UMFRAGE BEI DEUTSCHEN HUMANGENETIKERN ZUR SITUATION DER GENETISCHEN BERATUNG VON TURKISCHEN RATSUCHENDEN IN DEUTSCHLAND**
Elcioglu N.
Medizinische Genetik, cilt.7, sa.4, ss.442-447, 1995 (Scopus)

X. **Renal function in full-term newborn following neonatal asphyxia**

Elcioglu N., Sirin A., Can G., Tanman F., Emre S., Nayir A.
Istanbul Tip Fakultesi Mecmuasi, cilt.55, sa.3, ss.437-443, 1992 (Scopus)

XI. **Our experience with the patients of complete and corrected transposition of great arteries**
Elcioglu N., Eker R., Sarioglu T., Aytac A., Barlas C., Cantez T.
Istanbul Tip Fakultesi Mecmuasi, cilt.53, sa.3, ss.15-20, 1990 (Scopus)

Kitaplar

I. **Artrogripozis ve Eklemkontraktürleri ile seyreden İskelet Displazileri**
Yeter B., Elçioğlu H. N.
Genetik İskelet Bızuklukları, Hatice Mutlu, Editör, Türkiye Klinikleri Yayınevi, Ankara, ss.132-139, 2024

II. **Letal İskelet Displazilerine Prenatal Dönemde Tanısal Yaklaşım**
Elçioğlu H. N.
Genetik İskelet Bozuklukları, Hatice Mutlu, Editör, Türkiye Klinikleri Yayınevi, Ankara, ss.127-131, 2024

III. **DNA tamir hastalıkları**
ELÇİOĞLU H. N., ARAS S.
İzmir Behçet Uz Çocuk Hastanesi Pediatri Kitabı, Editör Behzat Özkan, Derya Erçal, Editör, Nobel Kitapevi, Ankara, ss.222-225, 2022

IV. **Erken yaşlanma hastalığı progeria: Hücresel mekanizma ve yeni tedavi stratejileri**
ELÇİOĞLU H. N.
Çocuk Genetik Uygulamalarında Sık Görülen Hastalıkların Takip ve Tedavisi, Ercan Mıhçı, Editör, Türkiye Klinikleri Pediatri Dergisi, Ankara, ss.157-164, 2021

V. **Malformasyon Sendromlarına Yaklaşım**
Elçioğlu H. N.
NEONATOLOJİ 3. baskı, Türkan Dağoğlu,Fahri Ovalı, Editör, Nobel Tıp Kitapevi, İstanbul, ss.361-388, 2017

VI. **Bağ Doku Hastalıkları**
Elçioğlu H. N.
TİBBİ GENETİK ve KLİNİK UYGULAMALARI, Munis Dündar, Editör, Hipokrat, Kayseri, ss.817-834, 2016

Hakemli Bilimsel Toplantılarda Yayımlanmış Bildiriler

I. **Famililal Chilblain Lupus preading to two countries, affecting three Generations with variable phenotypic expressivity. Oral presentation [OP-45] 03.2022 Istanbul, Turkey**
SERDAR Z., ELÇİOĞLU H. N., ALTAN FERHATOĞLU Z., Serdar İ.
7.INDERCOS International Dermatology and Cosmetology Congress, İstanbul, Türkiye, 9 - 12 Mart 2022, (Tam Metin Bildiri)

II. **Çocuk Genetik Polikliniğinde Değerlendirilen Osteogenezis İmperfekta Olgularının Klinik veGenomik Özellikleri: Tek Merkez Deneyimi S110**
KARALAR PEKUZ Ö. K., ELÇİOĞLU H. N.
3.Uluslararası Behçet Uz Çocuk Kongresi, 23-25 Eylül 2021 İzmir, İzmir, Türkiye, 23 - 25 Eylül 2021, ss.182-183, (Tam Metin Bildiri)

III. **Katepsin C Gen Mutasyonuyla İlişkili Agresif Periodontitisli İki Kardeşin Ağız İçi Bulguları**
GÜNGÖRMEK H. S., HAZNEDAROĞLU E., MENTEŞ A. R., ÖZEMRİ SAĞ Ş., ZEYBEK S., TEMEL Ş. G., ELÇİOĞLU H. N.
1. Bursa Uluslararası Katılımlı Genetik Günleri: Dermatogenetik Sempozyumu, Bursa, Türkiye, 9 - 11 Ocak 2020, (Özet Bildiri)

IV. **DASS Sendromlu Çocuk Hastanın Oro-dental Bulguları**
Karaman G. E., AKYÜZ S. H., ELÇİOĞLU H. N., MENTEŞ A. R.
26. Uluslararası Türk Pedodonti Derneği Kongresi, 10 - 13 Ekim 2019, (Özet Bildiri)

V. **S-17 MULTİPLE EXOSTOSES SENDROMLARINDA GENOTİP-FENOTİP İLİŞKİSİ**

AKALIN İ., Tabakçı B., Kurosawa K., Nishimura G., Wuyts W., ELÇİOĞLU H. N.
4. Çocuk Genetik Kongresi, Türkiye, 25 - 27 Eylül 2019, (Tam Metin Bildiri)

VI. **Hipokondroplazi: FGFR3 mutasyonlu 4 Olgu Sunumu**
Aliyeva S., Tabakçı B., AKALIN İ., ELÇİOĞLU H. N.
4. Çocuk Genetik Kongresi, Türkiye, 25 - 27 Eylül 2019, (Tam Metin Bildiri)

VII. **A novel intermediate type osteopetrosis associated with a new homozygous CLCN7 mutation**
AKALIN İ., Xue J., Doğan B., Yılmaz S., Long G., ASLAN E., Wang Z., Albayrak K., Nishimura G., İkegawa S., et al.
14th International Skeletal Dysplasia Society Meeting, 11 - 14 Eylül 2019, cilt.1, ss.91, (Tam Metin Bildiri)

VIII. **A rare cause of hypophosphatemia: Raine Syndrome**
ELTAN M., ATA P., KIRKGÖZ T., ALAVANDA C., KAYGUSUZ S. B., SEVEN M. T., GURPINAR TOSUN B., YAVAŞ ABALI Z., GÜRAN T., ELÇİOĞLU H. N., et al.
European Society for Paediatric Endocrinology (ESPE)58th Annual Meeting, Vienna, VİYANA, Avusturya, 19 - 21 Eylül 2019, cilt.91, (Tam Metin Bildiri)

IX. **Exome sequencing reveals novel candidate genes and potential oligogenic inheritance in patients with the complex trait arthrogryposis**
Pehlivan D., Bayram Y., Akdemir Z. C., Fatih J., Gibbs R., Posey J., Elcioglu N., TÜYSÜZ B., Lupski J.
71st Annual Meeting of the American-Academy-of-Neurology (AAN), Pennsylvania, Amerika Birleşik Devletleri, 4 - 10 Mayıs 2019, cilt.92, (Özet Bildiri)

X. **Molecular characterization and haplotype analysis in a large group of Mucopolysaccharidosis type IIIC (MPS IIIC) patients reveal the evolutionary history of the disease**
Martins C., Frassinetti de Medeiros P., Leistner-Segal S., Elcioglu N., Behnam M., Lacerda L., Lefebvre J. F., Giugliani R., Pshezhetsky A. V.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Danimarka, 27 - 30 Mayıs 2017, cilt.26, ss.778, (Özet Bildiri)

XI. **Çocukluk Çağında Diyabetin Nadir bir Nedeni: Glukokinaz Mutasyonu (MODY 2)**
İşgüven Ş. P., Bingöl Aydın D., Dönmez Ersavaş D., Elçioğlu H. N.
62. Türkiye Milli Pediatri kongresi-2. Kosova -Türkiye Pediatri kongresi, Antalya, Türkiye, 15 - 19 Kasım 2017, (Özet Bildiri)

XII. **Whole exome sequencing reveals potetial oligenic inheritance and candidate novel genes in patients with arthrogryposis**
BAYRAM Y., PEHLİVAN D., TÜYSÜZ B., ELÇİOĞLU H. N.
ASHG 67th Annual meeting, 17 Ekim 2017 - 21 Ocak 2018, (Özet Bildiri)

XIII. **Whole exome sequencing reveals potential oligogenic inheritance andcandidate novel genes in patients with arthrogryposis**
BAYRAM Y., ULUDAĞ ALKAYA D., pehlivan d., Gezdirici A., SILAN F., ÖZDEMİR Ö., ELÇİOĞLU H. N., YILDIZ O., yavuz ş., TÜYSÜZ B.
American Society of Human Genetics 67thAnnual Meeting, 17 - 21 Ekim 2017, (Özet Bildiri)

XIV. **Molecular characterization and haplotype analysis in a large group of Mucoplysachrarisosis type IIC (MPS IIIC) patients reveal the evolutionary hystory of the disease**
Martins C., Segal S L., Pshezhetsky A., ELÇİOĞLU H. N.
ESHG 2017 Kopenhagen, 27 - 30 Mayıs 2017, (Özet Bildiri)

XV. **Mutation spectrum and haplotype study of mucopolysaccharidosis type IIIC patients reveal possible migration events and founder effects of HGSNAT mutations**
Martins C., de Medeiros P. F., Leistner-Segal S., Elcioglu N., Behnam M., Lacerda L., Lefebvre J., Giugliani R., Pshezhetsky A. V.
13th Annual Research Meeting on We're Organizing Research for Lysosomal Diseases (WORLD), California, Amerika Birleşik Devletleri, 13 - 17 Şubat 2017, cilt.120, (Özet Bildiri)

XVI. **A novel homozygous IFT122 p I460N c 1379T A mutation inSensenbrenner syndrome a rare disorder within two cousins**
AKALIN İ., ELÇİOĞLU H. N., CANDAN C., YILMAZ S., Yücetürk B.
ESHG kongresi, 21 - 24 Mayıs 2016, (Özet Bildiri)

XVII. **RASopaties: two case reports**
DEMİRKOL KENDİR Y., Alp Ünkar Z., ÖZDEMİR H., BİLGEN H. S., ELÇİOĞLU H. N.
1st Congress of the European The Young Paediatricians' Association, İstanbul, Türkiye, 4 - 06 Aralık 2015, (Özet Bildiri)

XVIII. **Double aneuploidy: Down – Klinefelter Syndrome**
ALP UNKAR Z., KENDİR DEMİRKOL Y., MEMİŞOĞLU A., ELÇİOĞLU H. N., ÖZEK E.
1st Congress Of The European Young Paediatricians' Association (EURYPA), 4 - 06 Aralık 2015, (Özet Bildiri)

XIX. **RASopaties two case reports**
Demirkol Y., Alp Z., BİLGEN H. S., ELÇİOĞLU H. N.
1. EURYPA congress, İstanbul, Türkiye, 4 - 06 Aralık 2015

XX. **Molecular etiology of arthrogryposis in a cohort of families of Turkish origin Y Bayram 1 E Karaca 1 Z Coban Akdemir 1 H Aydin 2 A Gezdirici 3 D Torun 4 S Tug Bozdogan 5 S Isikay 6 M M Atik 1 T Gambin 1 A Karaman 7 D Pehlivan 1 H Aslan 8 O Ozalp Yuregir 9 S N Jhangiani 10 E Boerwinkle 10 11 R A Gibbs 10 N Elcioglu 12 B Tuysuz 13 J R Lupski**
BAYRAM Y., Karaca E., Çoban A. Z., Aydın H., Gezdirici A., TORUN D., PEHLİVAN D., Aslan H., ELÇİOĞLU H. N., Lupski J.
Congress ASHG 2015, 6 - 10 Ekim 2015

XXI. **The Skeletal Changes in Hurler s Syndrome afterBone Marrow Transplantation**
ELÇİOĞLU H. N.
12thINTERNATIONAL SKELETAL DYSPLASIA SOCIETY MEETING, 29 Temmuz - 01 Ağustos 2015

XXII. **CLINICAL AND MOLECULAR STUDY OF A SERIES OF 31 PATIENTS WITH CHONDRODYSPLASIA WITH MULTIPLE DISLOCATIONS**
Ranza E., Huber C., Levin N., Baujat G., ALANAY Y., Al Gazali L., Bitoun P., Boute O., Coubes C., ELÇİOĞLU H. N.
12thINTERNATIONAL SKELETAL DYSPLASIA SOCIETY MEETING', 29 Temmuz - 01 Ağustos 2015, (Özet Bildiri)

XXIII. **Mucolipidosis III Gamma Patients**
ELÇİOĞLU H. N.
12thINTERNATIONAL SKELETAL DYSPLASIA SOCIETY MEETING, 29 Temmuz - 01 Ağustos 2015

XXIV. **A molecular network surrounding dysregulated H3K9 di methylation in PRDM5 associated disease**
Porter L., Gall G., Williamson S., Selley J., Knight D., ELÇİOĞLU H. N., Elçioğlu M., Lund A.
European Human GeneticsConference 2015, 6 - 09 Haziran 2015, (Özet Bildiri)

XXV. **Spotlight on the pathogenesis of Kabuki syndrome**
Bögershausen N., Tsai I., Pohl E., ŞİMŞEK KİPER P. Ö., Beleggia F., Percin F., Keupp K., ALANAY Y., KAYSERİLİ KARABEY H., ELÇİOĞLU H. N.
European Human GeneticsConference 2015, 6 - 09 Haziran 2015, (Özet Bildiri)

XXVI. **Brakio Okülo Fasiyel Sendromlu Bir Olguda Axenfeld Rieger Anomalisi**
AKAY TAYFUN G., ELÇİOĞLU H. N., ÇERMAN E., ÖZARSLAN ÖZCAN D., ERASLAN M.
1. MARMARA PEDİATRİ KONGRESİ, Türkiye, 17 - 19 Ocak 2014

XXVII. **Brankio Oküler Fasiyel Sendromlu Bir Olguda Göz Bulguları**
ÇERMAN E., ÖZARSLAN ÖZCAN D., ERASLAN M., AKKAYA TAYFUN G., ELÇİOĞLU H. N.
47. TÜRK OFTALMOLOJİ DERNEĞİ ULUSAL KONGRESİ, Türkiye, 6 - 10 Kasım 2013

XXVIII. **Succesful treatment of Pityriasis Lichenoides Chronica with Narrow-Band Ultraviolet B Therapy in a Patient with Keratitis-Ichthyosis-Deafness Syndrome: A Case Report**
SALMAN A., SEÇKİN GENÇOSMANOĞLU D., YÜCELTEN A. D., ELÇİOĞLU H. N., RICHARD G., DEMİRKESEN C.
11th European Society for Pediatric Dermatology Congress, 16 - 19 Mayıs 2012

XXIX. **Phenotype, Genetics and Natural History in 146 Patients with SEPN1-Related Myopathy: On the Way to Therapeutic Trials in a Rare Disorder**
Malfatti E., Martinez V. G., Von der Hagen M., Chabalier D., Quijano-Roy S., David O., Moghadaszadeh B., Boenne-Mann C., Bushby K., Castiglioni C., et al.
63rd AAN Annual Meeting, Hawaii, Amerika Birleşik Devletleri, 9 - 16 Nisan 2011, cilt.76, (Özet Bildiri)

XXX. **Werner Syndrome and Rothmund-Thomson syndrome: are they some cases with overlapping phenotypes resembling both disease?**

Elcioglu N. H., Mansur A. T., Aslan-Kayiran M.
British Human Genetics Conference, York, Sierra Leone, 15 - 17 Eylül 2008, cilt.45, (Özet Bildiri)

XXXI. **POMPE DISEASE AND EXPERIENCE WITH PRENATAL DIAGNOSIS**
Elcioglu N., Akalin F., van der Luijt R. B.
5th Symposium on Lysosomal Storage Disorders, Paris, Fransa, 10 - 12 Nisan 2008, cilt.30, (Özet Bildiri)

XXXII. **SEPN-related myopathy: an emerging entity phenotypical and molecular analysis of 80 cases**
Gonzalez V., Quijano-Roy S., Parain K., Bonnemann C., Bushby K., Castiglioni C., Ceuterick C., Chaigne D., Colomer J., Desguerre I., et al.
10th International Congress of the World-Muscle-Society, Iguassu Falls, Brezilya, 28 Eylül - 10 Ekim 2005, cilt.15, ss.715, (Özet Bildiri)

XXXIII. **Isolated fetal choroid plexus cysts and association with chromosome anomalies**
Sayar C., Sahinoglu Z., Uludogan M., Turkover B., Elcioglu N.
European-Society-of-Human-Genetics European Human Genetics Conference in Conjuction With European Meeting on Psychosocial Aspects of Genetics, Strasbourg, Fransa, 25 - 28 Mayıs 2002, cilt.10, ss.276-277, (Özet Bildiri)

XXXIV. **Spondylothoracic dysplasia (Jarcho-Levin syndrome) and Spondylocostal dysostosis, the confusing vertebral malsegmentation syndromes. Report of six cases**
Semic A., Elcioglu N., Yalcin S., Biren T.
European-Society-of-Human-Genetics European Human Genetics Conference in Conjuction With European Meeting on Psychosocial Aspects of Genetics, Strasbourg, Fransa, 25 - 28 Mayıs 2002, cilt.10, ss.113, (Özet Bildiri)

Desteklenen Projeler

Elçioğlu H. N., TÜBİTAK Uluslararası İkili İşbirliği Projesi, Genetik İskelet Sistemi Hastalıklarının Büyük Ölçekli Genom Çalışması (SBAG / 217S675), 2018 - 2020

Metrikler

Yayın: 191
Atıf (WoS): 4101
Atıf (Scopus): 5387
H-İndeks (WoS): 33
H-İndeks (Scopus): 39

Ödüller

Elçioğlu H. N., Kökali F., Üçüncülük KS Otozomal Resesif Primer Mikrosefali nedenş ASPM mutasyonu: Klinik değerlendirme, Çocuk Genetik Hastalıkları Derneği, Kasım 2023
Elçioğlu H. N., Dizi Işık A., Yeter Doğan B., Demir S., Üçünçülük ödülü/ Ulna Hipoplazisi ile Nörofibromatozis Tip 1, Çocuk Genetik Derneği, Ekim 2021
Yıldırım H. S., Haznedaroğlu E., Menteş A. R., Özemri Sağ Ş., Zeybek S., Temel Ş. G., Elçioğlu H. N., Poster bildiri üçüncülük ödülü, Dermatogenetik Sempozyumu Kurulu, Ocak 2020