

Prof. Dr. AYŞE NURTEN AKARSU

Kişisel Bilgiler

İş Telefonu: [+90 3052559](tel:+903052559)

İş Telefonu: [+90 3051173](tel:+903051173)

Fax Telefonu: [+90 4268592](tel:+904268592)

E-posta: nakarsu@hacettepe.edu.tr

Web: <https://avesis.hacettepe.edu.tr/nakarsu>

Posta Adresi: Tıbbi Genetik Anabilim Dalı, D Blok (Konukevi 2) Kat 6 Sıhhiye, 06100 Ankara

Uluslararası Araştırmacı ID'leri

ORCID: 0000-0001-5432-0032

Publons / Web Of Science ResearcherID: E-9758-2013

Yoksis Araştırmacı ID: 9774

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

Akademik İdari Deneyim

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- I. **Defining mitochondrial protein functions through deep multiomic profiling**
Rensvold J. W., Shishkova E., Sverchikov Y., Miller I. J., ÇETİNKAYA A., Pyle A., Manicki M., Brademan D. R., ALANAY Y., Raiman J., et al.
NATURE, cilt.606, sa.7913, ss.382-388, 2022 (SCI-Expanded)
- II. **HEATR3 variants impair nuclear import of uL18 (RPL5) and drive Diamond-Blackfan anemia**
O'Donohue M., Da Costa L., Lezzerini M., Unal S., Joret C., Bartels M., Brilstra E., Scheijde-Vermeulen M., Wacheul L., De Keersmaecker K., et al.
Blood, cilt.139, sa.21, ss.3111-3126, 2022 (SCI-Expanded)
- III. **Long-Term Follow-Up Outcomes of 19 Patients with Osteogenesis Imperfecta Type XI and Bruck Syndrome Type I Caused by FKBP10 Variants**
YÜKSEL ÜLKER A., ULUDAĞ ALKAYA D., Elkanova L., ŞEKER A., Akpınar E., Akarsu N. A., Uyguner Z. O., TÜYSÜZ B.
CALCIFIED TISSUE INTERNATIONAL, cilt.109, sa.6, ss.633-644, 2021 (SCI-Expanded)
- IV. **Hematopoietic Cell Transplantation Cures Adenosine Deaminase 2 Deficiency: Report on 30 Patients**
Hashem H., Bucciol G., ÖZEN S., ÜNAL Ş., Bozkaya I. O., Akarsu N., Taskinen M., Koskenvuo M., Saarela J., Dimitrova D., et al.
JOURNAL OF CLINICAL IMMUNOLOGY, cilt.41, sa.7, ss.1633-1647, 2021 (SCI-Expanded)
- V. **One Disease with two Faces: Semidominant Inheritance of a Novel HTRA1 Mutation in a Consanguineous Family**
Bekircan-Kurt C. E., ÇETİNKAYA A., GÖÇMEN R., KOŞUKCU C., Soylemezoglu F., ARSAVA E. M., Tuncer A., ERDEM ÖZDAMAR S., Akarsu N. A., TOPÇUOĞLU M. A.
JOURNAL OF STROKE & CEREBROVASCULAR DISEASES, cilt.30, sa.9, 2021 (SCI-Expanded)
- VI. **Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy**
Wong H. H., Seet S. H., Maier M., GÜREL A., Traspas R. M., Lee C., Zhang S., TALİM B., Loh A. Y. T., Chia C. Y., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.108, sa.7, ss.1301-1317, 2021 (SCI-Expanded)
- VII. **Comprehensive clinical, biochemical, radiological and genetic analysis of 28 Turkish cases with suspected metachromatic leukodystrophy and their relatives**
Pekgul F., Eroglu-Ertugrul N. G., Bekircan-Kurt C. E., Erdem-Ozdamar S., Cetinkaya A., Tan E., Konuskan B., Karaagaoglu E., Topcu M., Akarsu N. A., et al.
MOLECULAR GENETICS AND METABOLISM REPORTS, cilt.25, 2020 (SCI-Expanded)
- VIII. **Laboratory diagnosis of metachromatic leukodystrophy requires more than arylsulfatase A assay**
Pekgul F., Bekircan-Kurt C. E., Konuskan B., Erdem-Ozdamar S., Tan E., Akarsu N., Topcu M., Anlar B., Ozkara H. A.
FEBS OPEN BIO, cilt.9, ss.199, 2019 (SCI-Expanded)
- IX. **Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene BRCA1**
Seo A., Steinberg-Shemer O., Unal Ş., Casadei S., Walsh T., Gumruk F., Shalev S., Shimamura A., Akarsu N., Tamary H., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.115, sa.20, ss.5241-5246, 2018 (SCI-Expanded)
- X. **Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamond-like features.**
Carapito R., Konantz M., Paillard C., Miao Z., Pichot A., Leduc M., Yang Y., Bergstrom K., Mahoney D., Shardy D., et al.
The Journal of clinical investigation, cilt.127, ss.4090-4103, 2017 (SCI-Expanded)
- XI. **Analysis of centrosome and DNA damage response in PLK4 associated Seckel syndrome.**
Dinçer T., Yorgancıoğlu-Budak G., Ölmez A., Er İ., Dodurga Y., Özdemir Ö., Toraman B., Yıldırım A., Sabir N., Akarsu N., et al.
European journal of human genetics : EJHG, cilt.25, ss.1118-1125, 2017 (SCI-Expanded)

- XII. **Identification of two novel PNPLA1 mutations in Turkish families with autosomal recessive congenital ichthyosis.**
Dökmeçi-Emre S, TAŞKIRAN Z. E., YÜZBAŞI OĞLU A., ÖNAL G., AKARSU A. N., KARADUMAN A., ÖZGÜÇ M.
The Turkish journal of pediatrics, cilt.59, ss.475-482, 2017 (SCI-Expanded)
- XIII. **Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling**
Cetinkaya A, Xiong J. R., Vargel İ., Kösemehmetoğlu K., Canter H. I., Gerdan O. F., Longo N., Alzahrani A., Camps M. P., Taskiran E. Z., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.99, sa.2, ss.299-317, 2016 (SCI-Expanded)
- XIV. **Recurrent viral infections associated with a homozygous CORO1A mutation that disrupts oligomerization and cytoskeletal association**
Yee C., Massaad M., Bainter W., Ohsumi T., Föger N., Chan A., Akarsu N., Aytakin C., Ayvaz D. N., Tezcan I., et al.
Journal of Allergy and Clinical Immunology, cilt.137, sa.3, ss.879, 2016 (SCI-Expanded)
- XV. **Nonsense Mutation in SPARC Gene Causing Autosomal Recessive Osteogenesis Imperfecta**
Abali S., ARMAN A., Atay Z., Bas S., Cam S., Gormez Z., Demirci H., ALANAY Y., Akarsu N., BEREKET A., et al.
HORMONE RESEARCH IN PAEDIATRICS, cilt.86, ss.167, 2016 (SCI-Expanded)
- XVI. **Frequency of Recessive Osteogenesis Imperfecta in a Turkish Cohort and Genetic Causes**
Abali S., ARMAN A., Atay Z., BEREKET A., Bas S., Haliloglu B., GÜRAN T., Gormez Z., Demirci H., Akarsu N., et al.
HORMONE RESEARCH IN PAEDIATRICS, cilt.86, ss.175-176, 2016 (SCI-Expanded)
- XVII. **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)
- XVIII. **Mutations Affecting the BHLHA9 DNA-Binding Domain Cause MSSD, Mesoaxial Synostotic Syndactyly with Phalangeal Reduction, Malik-Percin Type**
Malik S., Percin F. E., Bornholdt D., Albrecht B., Percesepe A., Koch M. C., Landi A., Fritz B., Khan R., Mumtaz S., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.95, sa.6, ss.649-659, 2014 (SCI-Expanded)
- XIX. **Novel splice-site and missense mutations in the ALDH1A3 gene underlying autosomal recessive anophthalmia/microphthalmia**
Semerci C. N., KALAY E., Yildirim C., DİNÇER T., Olmez A., TORAMAN B., Kocyigit A., Bulgu Y., Okur V., Satiroglu-Tufan L., et al.
BRITISH JOURNAL OF OPHTHALMOLOGY, cilt.98, sa.6, ss.832-840, 2014 (SCI-Expanded)
- XX. **Expanding the phenotypic spectrum of ECEL1-related congenital contracture syndromes**
Shaaban S., Duzcan F., Yildirim C., Chan W. -, Andrews C., Akarsu N. A., Engle E. C.
CLINICAL GENETICS, cilt.85, sa.6, ss.562-567, 2014 (SCI-Expanded)
- XXI. **TMCO1 Deficiency Causes Autosomal Recessive Cerebrofaciothoracic Dysplasia**
ALANAY Y., BEKİR E., ÜTİNE G. E., ORÇUN H., ŞİMŞEK KİPER P. Ö., TAŞKIRAN E. Z., PERÇİN F. E., UZ E., MAHMUT ŞAMİL S., BAYRAM Y., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.164, sa.2, ss.291-304, 2014 (SCI-Expanded)
- XXII. **Mutations in the interleukin receptor IL11RA cause autosomal recessive Crouzon-like craniosynostosis**
Keupp K., Li Y., VARGEL İ., Hoischen A., Richardson R., Neveling K., ALANAY Y., Uz E., Elcioglu N., Rachwalski M., et al.
MOLECULAR GENETICS & GENOMIC MEDICINE, cilt.1, sa.4, ss.223-237, 2013 (SCI-Expanded)
- XXIII. **Three Patients Resembling Teebi-Shaltout Syndrome**
Aldemir O., Ozen S., Erdem S., Kiraz A., Akarsu N., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.161, sa.10, ss.2570-2575, 2013 (SCI-Expanded)
- XXIV. **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)
- XXV. **Attenuated BMP1 Function Compromises Osteogenesis, Leading to Bone Fragility in Humans and**

Zebrafish

Asharani P. V., Keupp K., Semler O., Wang W., Li Y., Thiele H., Yigit G., Pohl E., Becker J., Frommolt P., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.90, sa.4, ss.661-674, 2012 (SCI-Expanded)

- XXVI. **Mutations in RIPK4 Cause the Autosomal-Recessive Form of Popliteal Pterygium Syndrome**
KALAY E., Sezgin O., Chellappa V., MUTLU M., Morsy H., Kayserili H., Kreiger E., CANSU A., TORAMAN B., Abdalla E. M., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.90, sa.1, ss.76-85, 2012 (SCI-Expanded)
- XXVII. **Disruption of the ptpro gene causes childhood onset nephrotic syndrome**
Ozaltin F., Ibsirlioglu T., Taskiran Z. E., Baydar D. E., Kaymaz F., Buyukcelik M., Iatropoulos P., Akarsu N. A., Schaefer F., Bakkaloglu A.
PEDIATRIC NEPHROLOGY, cilt.26, sa.9, ss.1578, 2011 (SCI-Expanded)
- XXVIII. **Disruption of PTPRO Causes Childhood-Onset Nephrotic Syndrome**
ÖZALTIN F., Ibsirlioglu T., Taskiran E. Z., Baydar D. E., Kaymaz F., Buyukcelik M., Kilic B. D., Balat A., Iatropoulos P., Asan E., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.89, sa.1, ss.139-147, 2011 (SCI-Expanded)
- XXIX. **KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes**
Putoux A., Thomas S., Coene K. L. M., Davis E. E., Alanay Y., Ogur G., Uz E., Buzas D., Gomes C., Patrier S., et al.
NATURE GENETICS, cilt.43, sa.6, ss.601-607, 2011 (SCI-Expanded)
- XXX. **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.
NEUROPSYCHOBIOLOGY, cilt.63, sa.1, ss.22-28, 2011 (SCI-Expanded)
- XXXI. **Mutation in exon 1f of PLEC, leading to disruption of plectin isoform 1f, causes autosomal-recessive limb-girdle muscular dystrophy.**
Gundesli H., TALİM B., KORKUSUZ P., Balci-Hayta B., Cirak S., Akarsu N. A., Topaloglu H., Dincer P. R.
American journal of human genetics, cilt.87, sa.6, ss.834-41, 2010 (SCI-Expanded)
- XXXII. **Congenital hypertelorism and osteopenia: A novel autosomal recessive disease**
Bonnard C., Merriman B., Lee H., Kayserili H., Akarsu N., Strobl A., Shboul M., Hamamy H., Reversade B.
DIFFERENTIATION, cilt.80, 2010 (SCI-Expanded)
- XXXIII. **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, sa.10, ss.2628-2633, 2010 (SCI-Expanded)
- XXXIV. **Intracranial and Extracranial Malformations in Patients With Craniofacial Anomalies**
TUNCBILEK G., Alanay Y., UZUN H., KAYIKCIOGLU A., AKARSU N. A., Benli K.
JOURNAL OF CRANIOFACIAL SURGERY, cilt.21, sa.5, ss.1460-1464, 2010 (SCI-Expanded)
- XXXV. **ASSOCIATION OF POLYNEUROPATHY, MENTAL RETARDATION, SENSORINEURAL HEARING LOSS, 6th NERVE PALSY, CONVULSIONS, AND ORAL DYSKINESIA; A PROPABLE NEW NEUROMETABOLIC DISORDER**
DURSUN A., YALNIZOĞLU D., DÜNDAR H., ERDEM S., AKARSU A. N., ÖZGÜL R. K.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- XXXVI. **Disruption of ALX1 Causes Extreme Microphthalmia and Severe Facial Clefing: Expanding the Spectrum of Autosomal-Recessive ALX-Related Frontonasal Dysplasia**
Uz E., Alanay Y., Aktas D., Vargel I., Gucer S., Tuncbilek G., von Eggeling F., Yilmaz E., DEREN Ö., Posorski N., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.86, sa.5, ss.789-796, 2010 (SCI-Expanded)
- XXXVII. **Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta**
Alanay Y., Avaygan H., Camacho N., ÜTİNE G. E., Boduroglu K., Aktas D., ALİKAŞİFOĞLU M., Tuncbilek E., ORHAN D., Bakar F. T., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.86, sa.4, ss.551-559, 2010 (SCI-Expanded)
- XXXVIII. **The effect of depression, BDNF gene val66met polymorphism and gender on serum BDNF levels**

- Ozan E., Okur H., Eker C., Eker O. D., GÖNÜL A. S., Akarsu N.
BRAIN RESEARCH BULLETIN, cilt.81, sa.1, ss.61-65, 2010 (SCI-Expanded)
- XXXIX. **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**
Wieczorek D., Pawlik B., Li Y., Akarsu N. A., Caliebe A., May K. J. W., Schweiger B., Vargas F. R., Balci S., Gillissen-Kaesbach G., et al.
HUMAN MUTATION, cilt.31, sa.1, ss.81-89, 2010 (SCI-Expanded)
- XL. **Derivative chromosome 1 and GLUT1 deficiency syndrome in a sibling pair**
Aktas D., Utine E. G., Mrasek K., Weise A., von Eggeling F., Yalaz K., Posorski N., Akarsu N., ALİKAŞIYOĞLU M., Liehr T., et al.
MOLECULAR CYTOGENETICS, cilt.3, 2010 (SCI-Expanded)
- XLI. **ALX4 dysfunction disrupts craniofacial and epidermal development**
Kayserili H., Uz E., Niessen C., VARGEL İ., Alanay Y., Tuncbilek G., Yigit G., Uyguner O., Candan S., Okur H., et al.
HUMAN MOLECULAR GENETICS, cilt.18, sa.22, ss.4357-4366, 2009 (SCI-Expanded)
- XLII. **Disease causing nature of homozygous missense, p.A523D, alteration in the perforin gene.**
Oner A., Okur H., Balta G., Unal Ş., Deger I., Akarsu N., Gurgey A.
Leukemia research, cilt.33, 2009 (SCI-Expanded)
- XLIII. **Homozygous feature of isolated triphalangeal thumb-preaxial polydactyly linked to 7q36: no phenotypic difference between homozygotes and heterozygotes**
Semerci C. N., Demirkan F., Ozdemir M., Biskin E., Akin B., Bagci H., Akarsu N. A.
CLINICAL GENETICS, cilt.76, sa.1, ss.85-90, 2009 (SCI-Expanded)
- XLIV. **A new autosomal dominant Peters' anomaly phenotype expanding the anterior segment dysgenesis spectrum**
Berker N., Alanay Y., Elgin U., Volkan-Salanci B., Simsek T., Akarsu N., ALİKAŞIYOĞLU M.
ACTA OPHTHALMOLOGICA, cilt.87, sa.1, ss.52-57, 2009 (SCI-Expanded)
- XLV. **Clinical and molecular aspects of Turkish familial hemophagocytic lymphohistiocytosis patients with perforin mutations**
Okur H., Balta G., Akarsu N., Oner A., Patiroglu T., Bay A., Sayli T., Unal Ş., Gurgey A.
LEUKEMIA RESEARCH, cilt.32, sa.6, ss.972-975, 2008 (SCI-Expanded)
- XLVI. **Mutations in the very low-density lipoprotein receptor VLDLR cause cerebellar hypoplasia and quadrupedal locomotion in humans**
Ozcelik T., Akarsu N., Uz E., Caglayan S., Gulsuner S., Onat O. E., Tan M., Tan U.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.105, sa.11, ss.4232-4236, 2008 (SCI-Expanded)
- XLVII. **Environmental effect and genetic influence: a regional cancer predisposition survey in the Zonguldak region of Northwest Turkey**
Kadir S., Onen-Hall A. P., Aydin S. N., Yakicier C., Akarsu N., Tuncer M.
ENVIRONMENTAL GEOLOGY, cilt.54, sa.2, ss.391-409, 2008 (SCI-Expanded)
- XLVIII. **Skewed X inactivation in an X linked nystagmus family resulted from a novel, p.R229G, missense mutation in the FRMD7 gene**
Kaplan Y., VARGEL İ., Kansu T., Akin B., Rohmann E., Kamaci S., Uz E., Ozcelik T., Wollnik B., Akarsu N. A.
BRITISH JOURNAL OF OPHTHALMOLOGY, cilt.92, sa.1, ss.135-141, 2008 (SCI-Expanded)
- XLIX. **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, sa.12, ss.1308-1312, 2007 (SCI-Expanded)
- L. **Molecular and clinical analysis of Turkish patients with HLH**
Balta G., Okur H., Akarsu N., Oner A., Sayli T., Gurgey A.
JOURNAL OF PEDIATRIC HEMATOLOGY ONCOLOGY, cilt.29, 2007 (SCI-Expanded)
- LI. **A fourth locus for hereditary hemorrhagic telangiectasia maps to chromosome 7.**
Bayrak-Toydemir P., McDonald J., Alkarsu N., Toydemir R. M., Calderon F., Tuncali T., Tang W., Miller F., Mao R.

American journal of medical genetics. Part A, cilt.140, sa.20, ss.2155-62, 2006 (SCI-Expanded)

- LII. **Evidence from autoimmune thyroiditis of skewed X-chromosome inactivation in female predisposition to autoimmunity**
Ozcelik T., Uz E., Akyerli C. B., Bagislar S., Mustafa C. A., Gursoy A., Akarsu N., Toruner G., Kamel N., Gullu S.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.14, sa.6, ss.791-797, 2006 (SCI-Expanded)
- LIII. **Autosomal recessive mesoaxial synostotic syndactyly with phalangeal reduction maps to chromosome 17p13.3**
Malik S., Percin F., Ahmad W., Percin S., Akarsu N., Koch M., Grzeschik K.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.4, ss.404-408, 2005 (SCI-Expanded)
- LIV. **Identification of KIF21A mutations as a rare cause of congenital fibrosis of the extraocular muscles type 3 (CFEOM3)**
Yamada K., Chan W., Andrews C., Bosley T., Sener E., Zwaan J., Mullaney P., Ozturk B., Akarsu A. N., Sabol L., et al.
INVESTIGATIVE OPHTHALMOLOGY & VISUAL SCIENCE, cilt.45, sa.7, ss.2218-2223, 2004 (SCI-Expanded)
- LV. **Is vitamin D hypothesis for schizophrenia valid? Independent segregation of psychosis in a family with vitamin-D-dependent rickets type IIA.**
Ozer S., Ulusahin A., Ulusoy S., Okur H., Coskun T., Tuncali T., Gogus A., Akarsu A. N.
Progress in neuro-psychopharmacology & biological psychiatry, cilt.28, sa.2, ss.255-66, 2004 (SCI-Expanded)
- LVI. **Heterozygous mutations of the kinesin KIF21A in congenital fibrosis of the extraocular muscles type 1 (CFEOM1)**
Yamada K., Andrews C., Chan W., McKeown C., Magli A., de Berardinis T., Loewenstein A., Lazar M., O'Keefe M., Letson R., et al.
NATURE GENETICS, cilt.35, sa.4, ss.318-321, 2003 (SCI-Expanded)
- LVII. **Molecular characterization of Turkish patients with pyrimidine 5 ' nucleotidase-I deficiency**
Balta G., GUMRUK F., AKARSU N., GURGEY A., ALTAY C.
BLOOD, cilt.102, sa.5, ss.1900-1903, 2003 (SCI-Expanded)
- LVIII. **Genome scan meta-analysis of schizophrenia and bipolar disorder, part III: Bipolar disorder**
Segurado R., Detera-Wadleigh S., Levinson D., Lewis C., Gill M., Nurnberg J., Craddock N., DePaulo J., Baron M., Gershon E., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.73, sa.1, ss.49-62, 2003 (SCI-Expanded)
- LIX. **Male cells in female recipients of hematopoietic-cell transplants**
Kaygusuz A., Akarsu A. N., Tuncer A.
NEW ENGLAND JOURNAL OF MEDICINE, cilt.347, sa.3, ss.219, 2002 (SCI-Expanded)
- LX. **Disruption of a long-range cis-acting regulator for Shh causes preaxial polydactyly**
Lettice L., Horikoshi T., Heaney S., van Baren M., van der Linde H., Breedveld G., Joosse M., Akarsu N., Oostra B., Endo N., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.99, sa.11, ss.7548-7553, 2002 (SCI-Expanded)
- LXI. **Hereditary intraosseous vascular malformation of the craniofacial region: An apparently novel disorder**
Vargel I., Cil B., Er N., Ruacan S., Akarsu A. N., Erk Y.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.109, sa.1, ss.22-35, 2002 (SCI-Expanded)
- LXII. **Van der Woude syndrome associated with ankyloblepharon filiforme adnatum is not linked to chromosome 1q32-q41 region.**
Okur H., Vargel I., Ozgur F., Erk Y., Akarsu A. N.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.69, sa.4, ss.289, 2001 (SCI-Expanded)
- LXIII. **DACH: Genomic characterization, evaluation as a candidate for postaxial polydactyly type A2, and developmental expression pattern of the mouse homologue**
Ayres J., Shum L., Akarsu A. N., Dashner R., Takahashi K., Ikura T., Slavkin H., Nuckolls G.
GENOMICS, cilt.77, ss.18-26, 2001 (SCI-Expanded)
- LXIV. **X-linked recessive inheritance of radial ray deficiencies in a family with four affected males**
Galjaard R., Kostakoglu N., Hoogeboom J., Breedveld G., van der Linde H., Hovius S., Oostra B., Sandkuijl L., Akarsu

- A. N., Heutink P.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.9, sa.9, ss.653-658, 2001 (SCI-Expanded)
- LXV. **An apparently dominant bipolar affective disorder (BPAD) locus on chromosome 20p11.2-q11.2 in a large Turkish pedigree**
Radhakrishna U., Senol S., Herken H., Gucuyener K., Gehrig C., Blouin J., Akarsu N., Antonarakis S.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.9, sa.1, ss.39-44, 2001 (SCI-Expanded)
- LXVI. **BIGH(3) gene analysis in the differential diagnosis of corneal dystrophies**
Kocak-Altintas A., Kocak-Midillioglu I., Akarsu A. N., Duman S.
CORNEA, cilt.20, sa.1, ss.64-68, 2001 (SCI-Expanded)
- LXVII. **Autosomal recessive severe intraosseous hemangioma in the skull. A new syndrome?**
Akarsu A. N., Vargel I., Erk Y.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.67, sa.4, ss.57, 2000 (SCI-Expanded)
- LXVIII. **Mutation of the gene encoding the ROR2 tyrosine kinase causes autosomal recessive Robinow syndrome**
van Bokhoven H., Celli J., Kayserili H., van Beusekom E., Balci S., Brussel W., Skovby F., Kerr B., Percin E., Akarsu N., et al.
NATURE GENETICS, cilt.25, sa.4, ss.423-426, 2000 (SCI-Expanded)
- LXIX. **A clinically variant fibrosis syndrome in a Turkish family maps to the CFEOM1 locus on chromosome 12**
Sener E., Lee B., Turgut B., Akarsu A. N., Engle E.
ARCHIVES OF OPHTHALMOLOGY, cilt.118, sa.8, ss.1090-1097, 2000 (SCI-Expanded)
- LXX. **Phenotypic variability of triphalangeal thumb-polysyndactyly syndrome linked to chromosome 7q36**
Balci S., Demirtas M., Civelek B., Piskin M., Sensoz O., Akarsu A. N.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.87, sa.5, ss.399-406, 1999 (SCI-Expanded)
- LXXI. **Multiple synostosis type 2 (SYNS2) maps to 20q11.2 and caused by a missense mutation in the growth/differentiation factor 5 (GDF5).**
Akarsu A. N., Rezaie T., Demirtas M., Farhud D., Sarfarazi M.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.65, sa.4, 1999 (SCI-Expanded)
- LXXII. **Clinical and genetic studies on 12 preaxial polydactyly families and refinement of the localisation of the gene responsible to a 1.9 cM region on chromosome 7q36**
Zguricas J., Heus H., Morales-Peralta E., Breedveld G., Kuyt B., Mumcu E., Bakker W., Akarsu N., Kay S., Hovius S., et al.
JOURNAL OF MEDICAL GENETICS, cilt.36, sa.1, ss.32-40, 1999 (SCI-Expanded)
- LXXIII. **Mesoaxial complete syndactyly and synostosis with hypoplastic thumbs: an unusual combination or homozygous expression of syndactyly type I?**
Percin E., Percin S., Egilmez H., Sezgin I., Ozbas F., Akarsu A. N.
JOURNAL OF MEDICAL GENETICS, cilt.35, sa.10, ss.868-874, 1998 (SCI-Expanded)
- LXXIV. **Clinical observations in autosomal recessive spastic paraplegia in childhood and further evidence for genetic heterogeneity**
Topaloglu H., Pinarli G., Erdem H., Gucuyener K., Karaduman A., Topcu M., Akarsu A. N., Ozguc M.
NEUROPEDIATRICS, cilt.29, sa.4, ss.189-194, 1998 (SCI-Expanded)
- LXXV. **A large family with type IV radial polydactyly**
Seyhan A., Akarsu N., Keskin F.
JOURNAL OF HAND SURGERY-BRITISH AND EUROPEAN VOLUME, sa.4, ss.530-533, 1998 (SCI-Expanded)
- LXXVI. **Sequence analysis and homology modeling suggest that primary congenital glaucoma on 2p21 results from mutations disrupting either the hinge region or the conserved core structures of cytochrome P4501B1**
Stoilov I., Akarsu A. N., Alozie I., Child A., Barsoum-Homsy M., Turacli M., Or M., Lewis R., Ozdemir N., Brice G., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.62, sa.3, ss.573-584, 1998 (SCI-Expanded)
- LXXVII. **Mapping of the second locus of postaxial polydactyly type A (PAP-A2) to chromosome 13q21-q32.**
Akarsu A. N., Ozbas F., Kostakoglu N.

- AMERICAN JOURNAL OF HUMAN GENETICS, cilt.61, sa.4, 1997 (SCI-Expanded)
- LXXVIII. **Identification of cytochrome P4501B1 as the gene mutated in primary congenital glaucoma families linked to the GLC3A locus on 2p21.**
Stoilov I., Akarsu A. N., Sarfarazi M.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.61, sa.4, 1997 (SCI-Expanded)
- LXXIX. **Genetic linkage study of familial Mediterranean fever (FMF) to 16p13.3 and evidence for genetic heterogeneity in the Turkish population**
Akarsu A. N., Saatci U., Ozen S., Bakkaloglu A., Besbas N., Sarfarazi M.
JOURNAL OF MEDICAL GENETICS, cilt.34, sa.7, ss.573-578, 1997 (SCI-Expanded)
- LXXX. **Identification of three different truncating mutations in cytochrome P4501B1 (CYP1B1) as the principal cause of primary congenital glaucoma (Buphthalmos) in families linked to the GLC3A locus on chromosome 2p21**
Stoilov I., Akarsu A. N., Sarfarazi M.
HUMAN MOLECULAR GENETICS, cilt.6, sa.4, ss.641-647, 1997 (SCI-Expanded)
- LXXXI. **A second locus (GLC3B) for primary congenital glaucoma (Buphthalmos) maps to the 1p36 region**
Akarsu A. N., Turacli M., Aktan S., BarsoumHomsy M., Chevrette L., Sayli B., Sarfarazi M.
HUMAN MOLECULAR GENETICS, cilt.5, sa.8, ss.1199-1203, 1996 (SCI-Expanded)
- LXXXII. **Genomic structure of HOXD13 gene: A nine polyalanine duplication causes synpolydactyly in two unrelated families**
Akarsu A. N., Stoilov I., Yilmaz E., Sayli B., Sarfarazi M.
HUMAN MOLECULAR GENETICS, cilt.5, sa.7, ss.945-952, 1996 (SCI-Expanded)
- LXXXIII. **Exclusion of primary congenital glaucoma (Buphthalmos) from two candidate regions of chromosome arm 6p and chromosome 11**
Akarsu A. N., Turacli M., Aktan S., Hossain A., BarsoumHomsy M., Chevrette L., Sayli B., Sarfarazi M.
AMERICAN JOURNAL OF MEDICAL GENETICS, cilt.61, sa.3, ss.290-292, 1996 (SCI-Expanded)
- LXXXIV. **ASSIGNMENT OF A LOCUS (GLC3A) FOR PRIMARY CONGENITAL GLAUCOMA (BUPHTHALMOS) TO 2P21 AND EVIDENCE FOR GENETIC-HETEROGENEITY**
SARFARAZI M., AKARSU A. N., HOSSAIN A., TURACLI M., AKTAN S., BARSOUMHOMSY M., CHEVRETTE L., SAYLI B.
GENOMICS, cilt.30, sa.2, ss.171-177, 1995 (SCI-Expanded)
- LXXXV. **LOCALIZATION OF THE SYNDACTYLY TYPE-II (SYNPOLYDACTYLY) LOCUS TO 2Q31 REGION AND IDENTIFICATION OF TIGHT LINKAGE TO HOXD8 INTRAGENIC MARKER**
SARFARAZI M., AKARSU A. N., SAYLI B.
HUMAN MOLECULAR GENETICS, cilt.4, sa.8, ss.1453-1458, 1995 (SCI-Expanded)
- LXXXVI. **Anophthalmos-syndactyly (Waardenburg) syndrome without oligodactyly of toes.**
Sayli B. S., Akarsu A. N., Altan S.
American journal of medical genetics, cilt.58, ss.18-20, 1995 (SCI-Expanded)
- LXXXVII. **A LARGE TURKISH KINDRED WITH SYNDACTYLY TYPE-II (SYNPOLYDACTYLY) .1. FIELD INVESTIGATION, CLINICAL AND PEDIGREE DATA**
SAYLI B., AKARSU A. N., SAYLI U., AKHAN O., CEYLANER S., SARFARAZI M.
JOURNAL OF MEDICAL GENETICS, cilt.32, sa.6, ss.421-434, 1995 (SCI-Expanded)
- LXXXVIII. **A LARGE TURKISH KINDRED WITH SYNDACTYLY TYPE-II (SYNPOLYDACTYLY) .2. HOMOZYGOUS PHENOTYPE**
AKARSU A. N., AKHAN O., SAYLI B., SAYLI U., BASKAYA G., SARFARAZI M.
JOURNAL OF MEDICAL GENETICS, cilt.32, sa.6, ss.435-441, 1995 (SCI-Expanded)
- LXXXIX. **GENETIC FEATURES OF RETINITIS-PIGMENTOSA IN TURKEY**
ATMACA L., SAYLI B., AKARSU N., GUNDUZ K.
DOCUMENTA OPHTHALMOLOGICA, cilt.89, sa.4, ss.387-392, 1995 (SCI-Expanded)
- XC. **THERAPEUTIC AND GENETIC-ASPECTS OF CONGENITAL GLAUCOMAS**
TURACLI M., AKTAN S., SAYLI B., AKARSU N.
INTERNATIONAL OPHTHALMOLOGY, cilt.16, ss.359-362, 1992 (SCI-Expanded)
- XCI. **Therapeutical and genetical aspects of congenital glaucomas.**

Turaçlı M. E., Aktan S. G., Saylı B. S., Akarsu N.

International ophthalmology, cilt.16, ss.359-62, 1992 (SCI-Expanded)

XCII. NASOPALPEBRAL LIPOMA-COLOBOMA SYNDROME

AKARSU A. N., SAYLI B.

CLINICAL GENETICS, cilt.40, sa.5, ss.342-344, 1991 (SCI-Expanded)

Kitap & Kitap Bölümleri

- I. **Nötropenin Nadir Bir Nedeni: Bileşik Heterozigot Mutasyonu Olan Shwachman-Diamond Sendromu**
AKSU T., AKARSU A. N., ÜNAL CANGÜL Ş.
Olgularla Kemik İliği Yetmezlikleri, Şule Ünal Cangül, Didem Atay, Turan Bayhan, Yusuf Ziya Aral, Editör, Galenos Yayın Evi, ss.171-173, 2023
- II. **YÜZ YARIKLARININ GENETİĞİ**
AKARSU A. N.
DUDAK ve DAMAK YARIKLARI HACETTEPE EKİP YAKLAŞIMI, Fatma FİGEN ÖZGÜR, Arda KÜÇÜKGÜVEN, Editör, HEKİM TIP KİTABEVİ, Ankara, ss.518-523, 2020
- III. **The ALX Homeobox Gene Family and Frontonasal Dysplasias**
Çetinkaya A., Akarsu A. N.
Epstein's Inborn Errors of Development: The Molecular Basis of Clinical Disorders of Morphogenesis (3 ed.), Robert P. Erickson, Anthony J. Wynshaw-Boris, Editör, Oxford University Press, London, London, ss.747-752, 2016

Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

- I. **In vitro Translasyon ile Ribozom İşlev Tayini**
KILIÇ H. B., ÇETİNKAYA A., AKARSU A. N., KOCAEFE Y. Ç.
26. Ulusal Tıbbi Biyoloji ve Genetik Kongresi 2021, Türkiye, 28 - 31 Ekim 2021, cilt.1, ss.277-278
- II. **Abstracts from the 53rd European Society of Human Genetics (ESHG) Conference: Interactive e-Posters**
GÜREL A., ÜNAL CANGÜL Ş., Yaralı N., ŞİMŞEK KİPER P. Ö., CEYLANER S., Bilir O. A., GÜMRÜK F., AKARSU A. N., ÇETİNKAYA A.
European Society of Human Genetics Virtual Conference, June 6-9, 2020., 6 - 09 Haziran 2020, cilt.28, ss.302
- III. **Plant-based milk alternative preference for children with cow milk protein allergy: Affecting factors and effect on nutritional status**
Parlak Z., ŞAHİNER Ü. M., KAHVECİ M., AKARSU A. N., Buyuktiryaki B., ŞEKEREL B. E., Soyer Ö.
European-Academy-of-Allergology-and-Clinical-Immunology Digital Congress (EAACI), London, Kanada, 6 - 08 Haziran 2020, cilt.75, ss.539-540
- IV. **Kalıtımsal nonsendromik yarık dudak/damak hastalığında genetik etiyolojinin araştırılması**
TORAMAN B., LİVAOĞLU M., DİNÇER T., BUDAK G., NALKIRAN İ., ÜNSAL S., YILDIZ G., AKARSU A. N., KALAY E.
XV. Ulusal Tıbbi Biyoloji ve Genetik Kongresi, Muğla, Türkiye, 26 - 29 Ekim 2017
- V. **Combination of UBR1 and UBR5 mutations in a severe form of Johanson-Blizzard Syndrome with total agenesis of lateral process and situs inversus**
ÇETİNKAYA A., GÜLSEN A. T., ERGÜNER B., TÜTÜN A., MUTLU M. B., KARADAĞ N., KARATEKİN G., KARAMAN A., AKARSU A. N.
American Society of Human Genetics 67. Annual Meeting, Orlando, Florşda, Amerika Birleşik Devletleri, 17 - 21 Ekim 2017
- VI. **Identification of Molecular Pathology of Peters' Anomaly Segregating in a Large Autosomal Dominant Family**
KOŞUKCU C., ASLI K., ALANAY Y., KAVAK P., BERKER N., TAŞKIRAN Z. E., ALİKAŞİFOĞLU M., SEZERMAN O. U., AKARSU A. N.

10th International Symposium on Health Informatics and Bioinformatics (HIBIT 2017), KALKANLI, GUZELYURT, Kıbrıs (Kktc), 29 - 30 Haziran 2017

VII. MULTICENTER RESULTS OF SCHWACHMAN-DIAMOND SYNDROME PATIENTS

ÜNAL S., Kalkan N., CELKAN T. T., Ozdemir G. N., Ozbek N., Yarali N., Cakmaklı H. F., Yenicesu I., Celik S. F., Kizilocak H., et al.

22nd Congress of the European-Hematology-Association, Madrid, İspanya, 22 - 25 Haziran 2017, cilt.102, ss.704-705

VIII. The clinical experiences with intraosseous vascular malformations related with ELMO2 mutations

VARGEL İ., ÇALIŞ M., CANTER H. İ., AKARSU A. N.

EURAPS 28.ANNUAL MEETING, 25 - 27 Mayıs 2017

IX. The clinical experiences with intraosseous vascular malformations related with ELMO2 mutations

VARGEL İ., ÇALIŞ M., CANTER H. İ., AKARSU A. N.

EURAPS 28. ANNUAL MEETING, İtalya, 25 - 27 Mayıs 2017

X. A Homozygous Germ Line Nonsense Mutation in BRCA1 Leading Fanconi Anemia and Neuroblastoma

Mehmet D., ÜNAL S., GÜMRÜK F., Akarsu N. A.

58th Annual Meeting and Exposition of the American-Society-of-Hematology (ASH), California, Amerika Birleşik Devletleri, 3 - 06 Aralık 2016, cilt.128

XI. A novel splicing site mutation of PLK4 that is required for centriole biogenesis and genomic stability causes Seckel syndrome

DİNÇER T., BUDAK G., SEMERCİ C. N., ÖLMEZ A., DODURGA Y., ÖZMERT M., TORAMAN B., YILDIRIM A., ERGİN H., ALVER A., et al.

European Society of Human Genetics Conference, 21 - 24 Mayıs 2016

XII. Loss of function mutations in ELMO2 impeding RSC1 signalling and cell migration cause intraosseous vascular malformation

ÇETİNKAYA A., Xiong J., VARGEL İ., KÖSEMEHMETOĞLU K., CANTER H. İ., GERDAN Ö., LONGO N., ALZHRANI A., TAŞKIRAN E. Z., BOTTO L., et al.

European Society of Human Genetics Conference, 21 - 24 Mayıs 2016

XIII. A Hypomorphic Coronin-1A Mutation that Abolishes Oligomerization and Cytoskeletal Association Is Associated with Impaired CD4+T Cell Survival

Yee C., Sanal O., AKARSU A. N., Ayetkin C., ÇAĞDAŞ AYVAZ D. N., Geha R. S., Chou J. S.

16th Biennial Meeting of the European-Society-for-Immunodeficiencies (ESID), Prague, Çek Cumhuriyeti, 29 Ekim - 01 Kasım 2014, cilt.34

XIV. Coronin-1A Oligomerization Is Critical For Host Defense Against Viral Pathogens

Yee C. S. K., Sanal O., Chou J. S., Geha R. S., Ayvaz D. N., Aytekin C., AKARSU A. N.

Annual Meeting of the American-Academy-of-Allergy-Asthma-and-Immunology (AAAAI), California, Amerika Birleşik Devletleri, 28 Şubat - 04 Mart 2014, cilt.133

XV. Congenital hypertelorism & osteopenia, a novel autosomal recessive disease of development

Bonnard C., Shboul M., Akarsu N., Kayserili H., Merriman B., Lee H., Hamamy H., Reversade B.

16th Annual Conference of the International-Society-of-Development-Biologists, Edinburgh, Saint Helena, 6 - 10 Eylül 2009, cilt.126

XVI. Linkage analysis of a four generation hereditary spherocytosis family with spectrin deficiency

Tasdemir P., Akarsu N., Demirel S.

7th European Cytogenetics Conference, Stockholm, İsveç, 4 - 07 Temmuz 2009, cilt.17, ss.79

XVII. The association of BDNF gene val66met polymorphism with the serum BDNF levels in drug-free depressed patients

Ozan E., Gonul A. S., Okur H., Akarsu N., EKER M. Ç., EKER M. Ç., Yulug B., Aydin N., Kirpinar I.

21st Congress of the European-College-of-Neuropsychopharmacology, Barcelona, İspanya, 30 Ağustos - 03 Eylül 2008, cilt.18

XVIII. Hunting the susceptibility gene for psychosis: A study of a family overloaded with different forms of psychotic disorders

Ozer S., Ulusoy S., Okur H., Gogus A., Akarsu A. N., Ulusahin A.

16th Congress of the European-College-of-Neuropsychopharmacology, PRAGUE, Çek Cumhuriyeti, 20 - 24 Eylül 2003, cilt.13

XIX. A family overloaded with psychosis and rickets-alopecia syndrome challenges vitamin-D hypothesis for schizophrenia

Ozer S., Ulusahin A., Ulusoy S., Okur H., Coskun T., Gogus A., Akarsu A. N.

16th Congress of the European-College-of-Neuropsychopharmacology, PRAGUE, Çek Cumhuriyeti, 20 - 24 Eylül 2003, cilt.13

XX. Molecular characterization of pyrimidine 5 prime nucleotidase (P5N-1) deficiency: Identification of 3 novel mutations.

Balta G., Gurgey A., Gumruk F., Akarsu N., Altay C.

44th Annual Meeting of the American-Society-of-Hematology, PHILADELPHIA, PENNSYLVANIA, 6 - 10 Aralık 2002, cilt.100

XXI. Phenotype description of Van der Woude Syndrome (VWS) unlinked to chromosome 1 containing IRF6

Okur H., Vargel I., Kondo S., Schutte B., Ozgur F., Balci S., Ozusta S., Cekirge I., Girgin B., Erk Y., et al.

52nd Annual Meeting of the American-Society-of-Human-Genetics, BALTIMORE, MARYLAND, 15 - 19 Ekim 2002, cilt.71, ss.285

XXII. Juvenile Hyalin Fibromatosis in three sibs from a consanguineous family: Clinical, histopathological and immunochemical findings

Balci S., Kulacoglu S., Senoz O., Vargel I., Erk Y., Onder S., Gokoz A., Akarsu A. N.

European-Society-of-Human-Genetics European Human Genetics Conference in Conjunction With European Meeting on Psychosocial Aspects of Genetics, Strasbourg, Fransa, 25 - 28 Mayıs 2002, cilt.10, ss.121-122

Desteklenen Projeler

AKARSU A. N., TÜBİTAK Projesi, Avrupa Diamond Blackfan Anemi Konsorsiyumu EuroDBA, 2016 - Devam Ediyor
ÖZKARA H. A., KURT C. E., ERDEM ÖZDAMAR S., TAN M. E., KONUŞKAN B., KARAAĞAOĞLU A. E., PEKGÜL F., TOPÇU M., ANLAR B., AKARSU A. N., Yükseköğretim Kurumları Destekli Proje, Metakromatik lökistrofi hastalığının alt tiplerinin tanımlanması, patojenik mutasyonların belirlenmesi, patogeneizde inflamasyonun incelenmesi, 2017 - 2019

TAŞKIRAN Z. E., ÜTİNE G. E., ŞİMŞEK KİPER P. Ö., BODUROĞLU O. K., AKARSU A. N., ALİKAŞİFOĞLU M., Yükseköğretim Kurumları Destekli Proje, Hacettepe Ekzom Projesi, 2015 - 2019

ALİKAŞİFOĞLU M., AKARSU A. N., BEKSAÇ M. S., GÜLERAY N., KABAÇAM S., Yükseköğretim Kurumları Destekli Proje, Maternal Kanda Hücre Dışı Serbest Dolaşan Fetal DNA İzolasyon Yöntemlerinin Karşılaştırılması, 2015 - 2016

YÜZBAŞIOĞLU A., DÖKMECİ S., AKARSU A. N., TAŞKIRAN Z. E., KARADUMAN A., ÖNAL G., Yükseköğretim Kurumları Destekli Proje, Otozomal Resesif Konjenital İktiyoz Hastalarında Genetik Analiz, 2015 - 2016

ÜNAL CANGÜL Ş., ÇETİN M., AKARSU A. N., GÜMRÜK F., TAŞKIRAN Z. E., BAYHAN T., Yükseköğretim Kurumları Destekli Proje, Diamond Blackfan anemili hastalarda yüksek çözünürlüklü dizileme yöntemi ile genom boyu analizi, 2015 - 2015

Metrikler

Yayın: 117

Atf (WoS): 4411

Atf (Scopus): 4859

H-İndeks (WoS): 32

H-İndeks (Scopus): 34

Akademi Dışı Deneyim

University of Geneva, Switzerland
University of Connecticut Health Center (UConn), USA
Ankara Üniversitesi Tıbbi Biyoloji ve Genetik B1
University of Connecticut Health Center (UConn), USA
Ankara-Akdere Ana Çocuk Sağlığı
Rize Güneysu Sağlık Ocağı