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### Personal Information

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### Education Information

Post Doctorate, University Of Connecticut, Connecticut Health Center, Surgical Research Center, United States Of America  
1994 - 1996

Doctorate, Ankara Üniversitesi, Tıp Fakültesi, Tıbbi Biyoloji Ve Genetik, Turkey 1988 - 1994

Under Graduate, Hacettepe Üniversitesi, Tıp Fakültesi, Tıp, Turkey 1978 - 1984

### Foreign Languages

English, B2 Upper Intermediate

### Dissertations

Doctorate, Sindaktili tip II (sin polidaktili): Genetik özellikleri, aile ve populasyon çalışması, Ankara Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1994

### Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Medical Genetics

### Academic Titles / Tasks

Professor, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 2008 - Continues

Associate Professor, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 1999 - 2008

Expert, Hacettepe Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1997 - 1999

### Professional Experience

Head of Department, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 2013 - 2014

### Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

- I. 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, vol.25, 2020 (Journal Indexed in SCI)

- II. **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, vol.9, pp.199, 2019 (Journal Indexed in SCI)
- III. **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, vol.115, no.20, pp.5241-5246, 2018 (Journal Indexed in SCI)
- IV. **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, vol.127, pp.4090-4103, 2017 (Journal Indexed in SCI Expanded)
- V. **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, vol.25, pp.1118-1125, 2017 (Journal Indexed in SCI)
- VI. **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, vol.59, pp.475-482, 2017 (Journal Indexed in SCI Expanded)
- VII. **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., ALZHRANI A., CAMPS M. P. , Taskiran E. Z. , et al.  
American journal of human genetics, vol.99, no.2, pp.299-317, 2016 (Journal Indexed in SCI Expanded)
- VIII. **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., Aytekin C., Ayvaz D. N. , Tezcan I., et al.  
Journal of Allergy and Clinical Immunology, vol.137, no.3, pp.879, 2016 (Journal Indexed in SCI Expanded)
- IX. **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, vol.86, pp.167, 2016 (Journal Indexed in SCI)
- X. **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, vol.86, pp.175-176, 2016 (Journal Indexed in SCI)
- XI. **CRIM1 haploinsufficiency causes defects in eye development in human and mouse**  
Beleggia F., Li Y., Fan J., Elcioglu N. H. , Tokar E., Wieland T., Maumenee I. H. , Akarsu N. A. , Meitinger T., Strom T. M. , et al.  
HUMAN MOLECULAR GENETICS, vol.24, no.8, pp.2267-2273, 2015 (Journal Indexed in SCI)
- XII. **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, vol.95, no.6, pp.649-659, 2014 (Journal Indexed in SCI)
- XIII. **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, vol.85, no.6, pp.562-567, 2014 (Journal Indexed in SCI)
- XIV. **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, vol.98, no.6, pp.832-840, 2014 (Journal Indexed in SCI)

- XV. **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, vol.164, no.2, pp.291-304, 2014 (Journal Indexed in SCI)
- XVI. **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, vol.161, no.10, pp.2570-2575, 2013 (Journal Indexed in SCI)
- XVII. **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, vol.54, no.6, pp.617-625, 2012 (Journal Indexed in SCI)
- XVIII. **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, vol.90, no.4, pp.661-674, 2012 (Journal Indexed in SCI)
- XIX. **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, vol.90, no.1, pp.76-85, 2012 (Journal Indexed in SCI)
- XX. **Disruption of the ptpo 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, vol.26, no.9, pp.1578, 2011 (Journal Indexed in SCI)
- XXI. **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, vol.89, no.1, pp.139-147, 2011 (Journal Indexed in SCI)
- XXII. **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, vol.43, no.6, pp.601-607, 2011 (Journal Indexed in SCI)
- XXIII. **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, vol.63, no.1, pp.22-28, 2011 (Journal Indexed in SCI)
- XXIV. **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.  
AMERICAN JOURNAL OF HUMAN GENETICS, vol.87, no.6, pp.834-841, 2010 (Journal Indexed in SCI)
- XXV. **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, vol.80, 2010 (Journal Indexed in SCI)
- XXVI. **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, no.10, pp.2628-2633, 2010 (Journal Indexed in SCI)
- XXVII. **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, vol.21, no.5, pp.1460-1464, 2010 (Journal Indexed in SCI)
- XXVIII. **ASSOCIATION OF POLYNEUROPATHY, MENTAL RETARDATION, SENSORINEURAL HEARING LOSS, 6th NERVE PALSY, CONVULSIONS, AND ORAL DYSKINESIA; A PROPABLE NEW NEUROMETABOLIC DISORDER**  
DURŞUN A., YALNIZOĐLU D., DÜNDAR H., ERDEM S., AKARSU A. N. , ÖZGÜL R. K.  
JOURNAL OF INHERITED METABOLIC DISEASE, vol.33, 2010 (Journal Indexed in SCI)
- XXIX. **Disruption of ALX1 Causes Extreme Microphthalmia and Severe Facial Clefting: 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, vol.86, no.5, pp.789-796, 2010 (Journal Indexed in SCI)
- XXX. **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., ALİKAŞİFOĐLU M., Tuncbilek E., ORHAN D., Bakar F. T. , et al.  
AMERICAN JOURNAL OF HUMAN GENETICS, vol.86, no.4, pp.551-559, 2010 (Journal Indexed in SCI)
- XXXI. **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, vol.81, no.1, pp.61-65, 2010 (Journal Indexed in SCI)
- XXXII. **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., ALİKAŞİFOĐLU M., Liehr T., et al.  
MOLECULAR CYTOGENETICS, vol.3, 2010 (Journal Indexed in SCI)
- XXXIII. **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., Gillessen-Kaesbach G., et al.  
HUMAN MUTATION, vol.31, no.1, pp.81-89, 2010 (Journal Indexed in SCI)
- XXXIV. **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, vol.18, no.22, pp.4357-4366, 2009 (Journal Indexed in SCI)
- XXXV. **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, vol.33, 2009 (Journal Indexed in SCI Expanded)
- XXXVI. **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, vol.76, no.1, pp.85-90, 2009 (Journal Indexed in SCI)
- XXXVII. **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., ALİKAŞİFOĐLU M.  
ACTA OPHTHALMOLOGICA, vol.87, no.1, pp.52-57, 2009 (Journal Indexed in SCI)
- XXXVIII. **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, vol.32, no.6, pp.972-975, 2008 (Journal Indexed in SCI)
- XXXIX. **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, vol.105, no.11, pp.4232-4236, 2008 (Journal Indexed in SCI)

- XL. 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, vol.54, no.2, pp.391-409, 2008 (Journal Indexed in SCI)
- XLI. 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, vol.92, no.1, pp.135-141, 2008 (Journal Indexed in SCI)
- XLII. 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, no.12, pp.1308-1312, 2007 (Journal Indexed in SCI)
- XLIII. 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, vol.29, 2007 (Journal Indexed in SCI)
- XLIV. A fourth locus for hereditary hemorrhagic telangiectasia maps to chromosome 7.**  
Bayrak-Toydemir P., McDonald J., Akarsu N., Toydemir R. M. , Calderon F., Tuncali T., Tang W., Miller F., Mao R.  
American journal of medical genetics. Part A, vol.140, no.20, pp.2155-62, 2006 (Journal Indexed in SCI Expanded)
- XLV. 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, vol.14, no.6, pp.791-797, 2006 (Journal Indexed in SCI)
- XLVI. 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, no.4, pp.404-408, 2005 (Journal Indexed in SCI)
- XLVII. 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, vol.45, no.7, pp.2218-2223, 2004 (Journal Indexed in SCI)
- XLVIII. 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, vol.28, no.2, pp.255-266, 2004  
(Journal Indexed in SCI)
- XLIX. 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, vol.35, no.4, pp.318-321, 2003 (Journal Indexed in SCI)
- L. Molecular characterization of Turkish patients with pyrimidine 5 ' nucleotidase-I deficiency**  
Balta G., GUMRUK F., AKARSU N., GURGEY A., ALTAY C.  
BLOOD, vol.102, no.5, pp.1900-1903, 2003 (Journal Indexed in SCI)
- LI. 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, vol.73, no.1, pp.49-62, 2003 (Journal Indexed in SCI)
- LII. Male cells in female recipients of hematopoietic-cell transplants**  
Kaygusuz A., Akarsu A. N. , Tuncer A.  
NEW ENGLAND JOURNAL OF MEDICINE, vol.347, no.3, pp.219, 2002 (Journal Indexed in SCI)
- LIII. 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, vol.99, no.11, pp.7548-7553, 2002 (Journal Indexed in SCI)

- LIV. **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, vol.109, no.1, pp.22-35, 2002 (Journal Indexed in SCI)
- LIV. **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, vol.69, no.4, pp.289, 2001 (Journal Indexed in SCI)
- LVI. **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, vol.9, no.9, pp.653-658, 2001 (Journal Indexed in SCI)
- LVII. **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, vol.77, pp.18-26, 2001 (Journal Indexed in SCI)
- LVIII. **BIGH(3) gene analysis in the differential diagnosis of corneal dystrophies**  
Kocak-Altintas A., Kocak-Midillioglu I., Akarsu A. N. , Duman S.  
CORNEA, vol.20, no.1, pp.64-68, 2001 (Journal Indexed in SCI)
- LIX. **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, vol.9, no.1, pp.39-44, 2001 (Journal Indexed in SCI)
- LX. **Autosomal recessive severe intraosseous hemangioma in the skull. A new syndrome?**  
Akarsu A. N. , Vargel I., Erk Y.  
AMERICAN JOURNAL OF HUMAN GENETICS, vol.67, no.4, pp.57, 2000 (Journal Indexed in SCI)
- LXI. **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, vol.118, no.8, pp.1090-1097, 2000 (Journal Indexed in SCI)
- LXII. **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, vol.25, no.4, pp.423-426, 2000 (Journal Indexed in SCI)
- LXIII. **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, vol.87, no.5, pp.399-406, 1999 (Journal Indexed in SCI)
- LXIV. **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, vol.65, no.4, 1999 (Journal Indexed in SCI)
- LXV. **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, vol.36, no.1, pp.32-40, 1999 (Journal Indexed in SCI)
- LXVI. **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, vol.35, no.10, pp.868-874, 1998 (Journal Indexed in SCI)

- LXVII. **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, vol.29, no.4, pp.189-194, 1998 (Journal Indexed in SCI)
- LXVIII. **A large family with type IV radial polydactyly**  
Seyhan A., Akarsu N., Keskin F.  
JOURNAL OF HAND SURGERY-BRITISH AND EUROPEAN VOLUME, no.4, pp.530-533, 1998 (Journal Indexed in SCI)
- LXIX. **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, vol.62, no.3, pp.573-584, 1998 (Journal Indexed in SCI)
- LXX. **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, vol.61, no.4, 1997 (Journal Indexed in SCI)
- LXXI. **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, vol.61, no.4, 1997 (Journal Indexed in SCI)
- LXXII. **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, vol.34, no.7, pp.573-578, 1997 (Journal Indexed in SCI)
- LXXIII. **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, vol.6, no.4, pp.641-647, 1997 (Journal Indexed in SCI)
- LXXIV. **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, vol.5, no.8, pp.1199-1203, 1996 (Journal Indexed in SCI)
- LXXV. **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, vol.5, no.7, pp.945-952, 1996 (Journal Indexed in SCI)
- LXXVI. **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, vol.61, no.3, pp.290-292, 1996 (Journal Indexed in SCI)
- LXXVII. **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, vol.30, no.2, pp.171-177, 1995 (Journal Indexed in SCI)
- LXXVIII. **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, vol.4, no.8, pp.1453-1458, 1995 (Journal Indexed in SCI)
- LXXIX. **Anophthalmos-syndactyly (Waardenburg) syndrome without oligodactyly of toes.**

Sayli B. S. , Akarsu A. N. , Altan S.

American journal of medical genetics, vol.58, pp.18-20, 1995 (Journal Indexed in SCI)

- LXXX. **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, vol.32, no.6, pp.421-434, 1995 (Journal Indexed in SCI)
- LXXXI. **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, vol.32, no.6, pp.435-441, 1995 (Journal Indexed in SCI)
- LXXXII. **GENETIC FEATURES OF RETINITIS-PIGMENTOSA IN TURKEY**  
ATMACA L., SAYLI B., AKARSU N., GUNDUZ K.  
DOCUMENTA OPHTHALMOLOGICA, vol.89, no.4, pp.387-392, 1995 (Journal Indexed in SCI)
- LXXXIII. **THERAPEUTIC AND GENETIC-ASPECTS OF CONGENITAL GLAUCOMAS**  
TURACLI M., AKTAN S., SAYLI B., AKARSU N.  
INTERNATIONAL OPHTHALMOLOGY, vol.16, pp.359-362, 1992 (Journal Indexed in SCI)
- LXXXIV. **Therapeutical and genetical aspects of congenital glaucomas.**  
Turaçlı M. E. , Aktan S. G. , Sayli B. S. , Akarsu N.  
International ophthalmology, vol.16, pp.359-62, 1992 (Journal Indexed in SCI Expanded)
- LXXXV. **NASOPALPEBRAL LIPOMA-COLOBOMA SYNDROME**  
AKARSU A. N. , SAYLI B.  
CLINICAL GENETICS, vol.40, no.5, pp.342-344, 1991 (Journal Indexed in SCI)

## Articles Published in Other Journals

- I. **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, vol.1, no.4, pp.223-237, 2013 (Journal Indexed in ESCI)

## Books & Book Chapters

- I. **YÜZ YARIKLARININ GENETİĞİ**  
AKARSU A. N.  
in: DUDAK ve DAMAK YARIKLARI HACETTEPE EKİP YAKLAŞIMI, Fatma FİGEN ÖZGÜR, Arda KÜÇÜKGÜVEN, Editor, HEKİM TIP KİTABEVİ, Ankara, pp.518-523, 2020
- II. **The ALX Homeobox Gene Family and Frontonasal Dysplasias**  
Çetinkaya A., Akarsu A. N.  
in: Epstein's Inborn Errors of Development: The Molecular Basis of Clinical Disorders of Morphogenesis (3 ed.), Robert P. Erickson, Anthony J. Wynshaw-Boris, Editor, Oxford University Press, London , London, pp.747-752, 2016

## Refereed Congress / Symposium Publications in Proceedings

- I. **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 June 2020, vol.28, pp.302



- II. **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, Canada, 6 - 08 June 2020, vol.75, pp.539-540
- III. **Kalıtısal 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, Turkey, 26 - 29 October 2017
- IV. **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, Florida, United States Of America, 17 - 21 October 2017
- V. **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, Cyprus (Kkct), 29 - 30 June 2017
- VI. **MULTICENTER RESULTS OF SCHWACHMAN-DIAMOND SYNDROME PATIENTS**  
ÜNAL S., Kalkan N., CELKAN T. T. , Ozdemir G. N. , Ozbek N., Yarali N., Cakmakli H. F. , Yenicesu I., Celik S. F. , Kizilocak H., et al.  
22nd Congress of the European-Hematology-Association, Madrid, Spain, 22 - 25 June 2017, vol.102, pp.704-705
- VII. **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 May 2017
- 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, Italy, 25 - 27 May 2017
- IX. **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, United States Of America, 3 - 06 December 2016, vol.128
- X. **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 May 2016
- XI. **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 May 2016
- XII. **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, Czech Republic, 29 October - 01 November 2014, vol.34
- XIII. **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. , Ayetkin C., AKARSU A. N.

Annual Meeting of the American-Academy-of-Allergy-Asthma-and-Immunology (AAAAI), California, United States Of America, 28 February - 04 March 2014, vol.133

- XIV. **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 September 2009, vol.126
- XV. **Linkage analysis of a four generation herediter spherocytosis family with spectrin deficiency**  
Tasdemir P., Akarsu N., Demirel S.  
7th European Cytogenetics Conference, Stockholm, Sweden, 4 - 07 July 2009, vol.17, pp.79
- XVI. **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, Spain, 30 August - 03 September 2008, vol.18
- XVII. **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, Czech Republic, 20 - 24 September 2003, vol.13
- XVIII. **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, Czech Republic, 20 - 24 September 2003, vol.13
- XIX. **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 December 2002, vol.100
- XX. **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 October 2002, vol.71, pp.285
- XXI. **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, France, 25 - 28 May 2002, vol.10, pp.121-122

## Supported Projects

AKARSU A. N. , TUBITAK Project, Avrupa Diamond Blackfan Anemi Konsorsiyumu EuroDBA, 2016 - Continues  
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. , Project Supported by Higher Education Institutions, Metakromatik lökodikrofi 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., Project Supported by Higher Education Institutions, Hacettepe Ekzom Projesi, 2015 - 2019  
ALİKAŞİFOĞLU M., AKARSU A. N. , BEKSAÇ M. S. , GÜLERAY N., KABAÇAM S., Project Supported by Higher Education Institutions, 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., Project Supported by Higher Education Institutions, Otozomal Resesif Konjenital İktiyoz Hastalarında Genetik Analiz, 2015 - 2016

ÜNAL CANGÜL Ş., ÇETİN M., AKARSU A. N. , GÜMRÜK F., TAŞKIRAN Z. E. , BAYHAN T., Project Supported by Higher Education Institutions, Diamond Blackfan anemili hastalarda yüksek çözünürlüklü dizileme yöntemi ile genom boyu analizi, 2015 - 2015

## Citations

Total Citations (WOS):4313

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