

Prof. AYŞE NURTEN AKARSU

Personal Information

Office Phone: [+90 3052559](tel:+903052559)

Office Phone: [+90 3051173](tel:+903051173)

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

Email: nakarsu@hacettepe.edu.tr

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

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

International Researcher IDs

ORCID: 0000-0001-5432-0032

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

Yoksis Researcher ID: 9774

Education Information

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

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

Undergraduate, Hacettepe University, 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 University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 2008 - Continues

Associate Professor, Hacettepe University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 1999 - 2008

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

Academic and Administrative Experience

Published journal articles indexed by SCI, SSCI, and AHCI

- 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, vol.606, no.7913, pp.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, vol.139, no.21, pp.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, vol.109, no.6, pp.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, vol.41, no.7, pp.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, vol.30, no.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, vol.108, no.7, pp.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, vol.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, vol.9, pp.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, vol.115, no.20, pp.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, vol.127, pp.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, vol.25, pp.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, vol.59, pp.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, vol.99, no.2, pp.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, vol.137, no.3, pp.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, vol.86, pp.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, vol.86, pp.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, vol.24, no.8, pp.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, vol.95, no.6, pp.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, vol.98, no.6, pp.832-840, 2014 (SCI-Expanded)
- XX. **Expanding the phenotypic spectrum of ECEL1-related congenital contracture syndromes**
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CLINICAL GENETICS, vol.85, no.6, pp.562-567, 2014 (SCI-Expanded)
- XXI. **TMC01 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 (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, vol.1, no.4, pp.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, vol.161, no.10, pp.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, vol.54, no.6, pp.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, vol.90, no.4, pp.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, vol.90, no.1, pp.76-85, 2012 (SCI-Expanded)
- XXVII. **Disruption of the ptpro gene causes childhood onset nephrotic syndrome**
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- 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.
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- XXIX. **KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes**
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NATURE GENETICS, vol.43, no.6, pp.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, vol.63, no.1, pp.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.
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- 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.
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- 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, no.10, pp.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, vol.21, no.5, pp.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, vol.33, 2010 (SCI-Expanded)
- XXXVI. **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 (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, vol.86, no.4, pp.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, vol.81, no.1, pp.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, vol.31, no.1, pp.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, vol.3, 2010 (SCI-Expanded)
- XLI. **ALX4 dysfunction disrupts craniofacial and epidermal development**
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- XLII. **Disease causing nature of homozygous missense, p.A523D, alteration in the perforin gene.**
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- XLIII. **Homozygous feature of isolated triphalangeal thumb-preaxial polydactyly linked to 7q36: no phenotypic difference between homozygotes and heterozygotes**
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CLINICAL GENETICS, vol.76, no.1, pp.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, vol.87, no.1, pp.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.
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- XLVI. **Mutations in the very low-density lipoprotein receptor VLDLR cause cerebellar hypoplasia and quadrupedal locomotion in humans**
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- XLVII. **Environmental effect and genetic influence: a regional cancer predisposition survey in the Zonguldak region of Northwest Turkey**
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ENVIRONMENTAL GEOLOGY, vol.54, no.2, pp.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**
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- XLIX. **Colobomatous macrophthalmia with microcornea syndrome maps to the 2p23-p16 region**
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- L. **Molecular and clinical analysis of Turkish patients with HLH**
Balta G., Okur H., Akarsu N., Oner A., Sayli T., Gurgey A.
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- LI. **A fourth locus for hereditary hemorrhagic telangiectasia maps to chromosome 7.**
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- LII. **Evidence from autoimmune thyroiditis of skewed X-chromosome inactivation in female predisposition to autoimmunity**
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- LIII. **Autosomal recessive mesoaxial synostotic syndactyly with phalangeal reduction maps to chromosome 17p13.3**
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- LIV. **Identification of KIF21A mutations as a rare cause of congenital fibrosis of the extraocular muscles type 3 (CFEOM3)**
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- LV. **Is vitamin D hypothesis for schizophrenia valid? Independent segregation of psychosis in a family with vitamin-D-dependent rickets type IIA.**
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- LVII. **Molecular characterization of Turkish patients with pyrimidine 5' nucleotidase-I deficiency**
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- LVIII. **Genome scan meta-analysis of schizophrenia and bipolar disorder, part III: Bipolar disorder**
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- LIX. **Male cells in female recipients of hematopoietic-cell transplants**
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- LX. **Disruption of a long-range cis-acting regulator for Shh causes preaxial polydactyly**
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- LXI. **Hereditary intraosseous vascular malformation of the craniofacial region: An apparently novel disorder**
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- LXII. **Van der Woude syndrome associated with ankyloblepharon filiforme adnatum is not linked to chromosome 1q32-q41 region.**
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- LXIII. **DACH: Genomic characterization, evaluation as a candidate for postaxial polydactyly type A2, and developmental expression pattern of the mouse homologue**
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- LXIV. **X-linked recessive inheritance of radial ray deficiencies in a family with four affected males**
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Metrics

Publication: 117

Citation (WoS): 4411

Citation (Scopus): 4859

H-Index (WoS): 32

H-Index (Scopus): 34

Non Academic Experience

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