Assoc. Prof. ÖZLEM AKGÜN DOĞAN

Personal Information

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Research Areas

Health Sciences

Academic Positions

Associate Professor, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2023 - Continues
Assistant Professor, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2021 - 2022

Academic and Administrative Experience

Fakülte Yönetim Kurulu Üyesi, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2023 - Continues
Program Koordinatörü, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2022 - Continues
ACURARE Vice President, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, 2022 - Continues
Toplumsal Katkı Komisyonu Üyeliği, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2022 - 2023

Courses

Postgraduate
Mendelian Inheritance Patterns, Postgraduate, 2022 - 2023
Applications of genetic testing from clinic point of view, Postgraduate, 2022 - 2023
Developmental Genetics, Postgraduate, 2022 - 2023
Undergraduate
Pediatric Genetics, Undergraduate, 2023 - 2024, 2022 - 2023
From Genotype to Phenotype, Undergraduate, 2023 - 2024
Mendelian Inheritance, Undergraduate, 2023 - 2024, 2022 - 2023
Translational Medicine, Undergraduate, 2022 - 2023
Developmental Genetics, Undergraduate, 2023 - 2024, 2022 - 2023

Non Mendelian Inheritance, Undergraduate, 2023 - 2024, 2022 - 2023
Introduction to Pediatrics, Undergraduate, 2023 - 2024, 2022 - 2023
Clinical Cytogenetics, Undergraduate, 2023 - 2024, 2022 - 2023

Jury Memberships

PhD Thesis Monitoring Committee Member, PhD Thesis Monitoring Committee Member, Acıbadem Mehmet Ali Aydınlar Üniversitesi, April, 2023

Taught Courses and Trainings

Tahir Turanlı E., Uğurlu S., AKGÜN DOĞAN Ö., Genetik ve İmmünoloji Kampı, 2023 - 2023
Akgün Doğan Ö., Hatırnaz Ng Ö., Özdemir Ö., İSTİSNA Nadir ve Tanısız Hastalıkların Çözümünde Kullanılacak Stratejiler, Genomik Yaklaşımlar ve Veri Analizi Eğitimi, 2023 - 2023
Akgün Doğan Ö., Serebral Palsi Sempozyumu, 2022 - 2023
Akgün Doğan Ö., Tıbbi Genomikte Varyant Yorumlama ve Önceliklendirme, 2022 - 2022

Designed Courses and Trainings

Akgün Doğan Ö., Tıbbi Genomikte Varyant Yorumlama ve Önceliklendirme, November 2022

Research Infrastructure Information

Hatırnaz Ng Ö., Alanay Y., Özdemir Ö., Akgün Doğan Ö., Bilgüvar K., İStanbul Tanısız ve Nadir Hastalıklara Çözüm Platformu, February 2023

Journal articles indexed in SCI, SSCI, and AHCI

I. **Exocrine pancreatic insufficiency as an unusual extrarenal manifestation of proximal renal tubular acidosis associated with a novel SLC4A4 mutation**
Hasturk B., Agbas A., AKGÜN DOĞAN Ö., Yilmaz E. K., Saygili S., Beser O. F., CANPOLAT N.
PEDIATRIC NEPHROLOGY, 2025 (SCI-Expanded)
II. **Statistical analysis of genomic in-silico pathogenicity predictors for the characterization of VUS in rare and undiagnosed disorders**
AYDIN E., Ergun B., AKGÜN DOĞAN Ö., ALANAY Y., HATIRNAZ NG Ö., ÖZDEMİR Ö.
EUROPEAN JOURNAL OF HUMAN GENETICS, pp.1172, 2024 (SCI-Expanded)
III. **Statistical analysis of genomic in-silico pathogenicity predictors for the characterization of VUS in rare and undiagnosed disorders**
AYDIN E., Ergun B., AKGÜN DOĞAN Ö., ALANAY Y., HATIRNAZ NG Ö., ÖZDEMİR Ö.
EUROPEAN JOURNAL OF HUMAN GENETICS, pp.1172, 2024 (SCI-Expanded)
IV. **CFAP99 deficiency leads to heterotaxy and scoliosis**
Ay B., Viviano S., AKGÜN DOĞAN Ö., ALANAY Y., Deniz E.
EUROPEAN JOURNAL OF HUMAN GENETICS, pp.1747-1748, 2024 (SCI-Expanded)
V. **Very Early-Onset Schizophrenia with Accompanying Obsessive-Compulsive Symptoms: A Case Report of a Female with 16p13.11 Duplication**
Kiziltan K., Kiziltan E. O., Oral E. Y., AKGÜN DOĞAN Ö., Ersoy M., Karacetin G.
PSYCHIATRY AND CLINICAL PSYCHOPHARMACOLOGY, vol.34, no.4, pp.356-357, 2024 (SCI-Expanded)

VI. **BCL11B-related disease: a single phenotypic entity?**
Vedovato-dos-Santos J. H., Tooze R., Mccann E., Sithambaram S., ALANAY Y., Johnson D., AKGÜN DOĞAN Ö., KILERCIK M., BİNGÖL A., ÖZEK M., et al.
EUROPEAN JOURNAL OF HUMAN GENETICS, pp.1559-1560, 2024 (SCI-Expanded)

VII. **Impact of deep phenotyping: High diagnostic yield in a diverse pediatric population of 172 patients through clinical whole-genome sequencing at a single center**
AKGÜN DOĞAN Ö., Bengur E. T., Ay B., Ozkose G. S., Kar E., Bengur F. B., Bulut A., Yigit A., Aydin E., Esen F. N., et al.
EUROPEAN JOURNAL OF HUMAN GENETICS, pp.1560, 2024 (SCI-Expanded)

VIII. **CFAP99 deficiency leads to heterotaxy and scoliosis**
Ay B., Viviano S., AKGÜN DOĞAN Ö., ALANAY Y., Deniz E.
EUROPEAN JOURNAL OF HUMAN GENETICS, pp.1747-1748, 2024 (SCI-Expanded)

IX. **Pushing the boundaries of rare disease diagnostics with the help of the first Undiagnosed Hackathon.**
Delgado-Vega A. M., Cederroth H., Taylan F., Ekholm K., Ek M., Thonberg H., Jemt A., Nilsson D., Eisfeldt J., Bilgrav Saether K., et al.
Nature genetics, vol.56, no.11, pp.2287-2294, 2024 (SCI-Expanded)

X. **Real-world experience with vosoritide treatment in achondroplasia: A single-center report from Turkey**
ABALI S., Ozkose G. S., AKGÜN DOĞAN Ö., DEMİRCİOĞLU S., ALANAY Y.
HORMONE RESEARCH IN PAEDIATRICS, pp.348-349, 2024 (SCI-Expanded)

XI. **Two new patients with acromesomelic dysplasia, PRKG2 type-identification and characterization of the first missense variant**
AKGÜN DOĞAN Ö., Diaz-Gonzalez F., de Lima Jorge A. A., Onenli-Mungan N., Menezes Andrade N. L., de Polli Cellin L., CEYLANER S., Barcellos Rosa Modkovski M., ALANAY Y., Heath K. E.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.32, no.10, pp.1250-1256, 2024 (SCI-Expanded)

XII. **Rapid genome sequencing for critically ill infants: an inaugural pilot study from Turkey.**
Guner Yilmaz B., Akgun-Dogan Ö., Ozdemir Ö., Yuksel B., Hatirnaz Ng Ö., Bilguvar K., Ay B., Ozkose G. S., Aydin E., Yigit A., et al.
Frontiers in pediatrics, vol.12, pp.1412880, 2024 (SCI-Expanded)

XIII. **Metabolic and other morbid complications in congenital generalized lipodystrophy type 4**
Akinci G., Alyaarubi S., Patni N., Alhashmi N., Al-Shidhani A., Prodam F., Gagne N., Babalola F., Al Senani A., Muniraj K., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.194, no.6, 2024 (SCI-Expanded)

XIV. **Implication of transcription factor FOXD2 dysfunction in syndromic congenital anomalies of the kidney and urinary tract (CAKUT)**
Riedhammer K. M., Nguyen T. T., Koşukcu C., Calzada-Wack J., Li Y., Assia Batzir N., Saygılı S., Wimmers V., Kim G., Chrysanthou M., et al.
Kidney International, vol.105, no.4, pp.844-864, 2024 (SCI-Expanded)

XV. **Schwartz-Jampel Syndrome Type-1: Compound Heterozygosity of Two Novel Variants.**
Atmaca F. G., Akgün Doğan Ö., Kutlubay B., Kırmızıbekmez H.
Journal of clinical research in pediatric endocrinology, 2024 (SCI-Expanded)

XVI. **Impact of deep phenotyping: high diagnostic yield in a diverse pediatric population of 172 patients through clinical whole-genome sequencing at a single center**
Akgün Doğan Ö., Bengur E. T., Ay B., Ozkose G. S., Kar E., Bengur F. B., Bulut A., Yigit A., Aydin E., Esen F. N., et al.
FRONTIERS IN GENETICS, vol.15, 2024 (SCI-Expanded)

XVII. **Magnetic resonance imaging based kidney volume assessment for risk stratification in pediatric autosomal dominant polycystic kidney disease**
Yilmaz K., SAYGILI S. K., CANPOLAT N., AKGÜN DOĞAN Ö., YÜRÜK YILDIRIM Z. N., Cicek-Oksuz R. Y., Oner H. A., AKSU B., Akyel N. G., Oguzhan-Hamis O., et al.
FRONTIERS IN PEDIATRICS, vol.12, 2024 (SCI-Expanded)

XVIII. **A novel homozygous missense variant in TBC1D31 in a consanguineous family with**

congenital anomalies of the kidney and urinary tract (CAKUT)
SAYGILI S. K., Kosukcu C., BAŞTUĞ T., AKGÜN DOĞAN Ö., Yilmaz E. K., Kalyoncu A. U., AĞBAŞ A., CANPOLAT N., Caliskan S., ÖZALTIN F.
CLINICAL GENETICS, vol.104, no.6, pp.679-685, 2023 (SCI-Expanded)

XIX. **DNAJC21-related thrombocytopenia in a young adult female**
ASLAN D., AKGÜN DOĞAN Ö., Ay B., ÇAMURDAN M. O., Mancilar H., ALANAY Y.
AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS, vol.193, no.2, pp.193-197, 2023 (SCI-Expanded)

XX. **Clinical Heterogeneity and Different Phenotypes in Patients with SETD2 Variants: 18 New Patients and Review of the Literature**
Parra A., Rabin R., Pappas J., Pascual P., Cazalla M., Arias P., Gallego-Zazo N., Santana A., Arroyo I., Artigas M., et al.
GENES, vol.14, no.6, 2023 (SCI-Expanded)

XXI. **Clinical features of generalized lipodystrophy in Turkey: A cohort analysis.**
Yildirim Simsir I., Tuysuz B., Ozbek M. N., Tanrikulu S., Celik Guler M., Karhan A. N., Denkboy Ongen Y., Gunes N., Soyaltin U. E., Altay C., et al.
Diabetes, obesity & metabolism, 2023 (SCI-Expanded)

XXII. **Diagnostic Pitfalls of a Newborn with Congenital Nephrogenic Diabetes Insipidus.**
Güran Ö., Güven S., Kırmızıbekmez H., Akgün Doğan Ö., Karadeniz Bilgin L.
Journal of clinical research in pediatric endocrinology, 2023 (SCI-Expanded)

XXIII. **Obstacles and expectations of rare disease patients and their families in Turkiye: ISTisNA project survey results**
Hatirnaz Ng Ö., Sahin I., Erbilgin Y., Ozdemir Ö., Yucesan E., Erturk N., Yemenici M., Akgun Dogan Ö., Ugur Iseri S. A., Satman I., et al.
FRONTIERS IN PUBLIC HEALTH, vol.10, 2023 (SCI-Expanded)

XXIV. **Concurrent Pathogenic Variants of BRCA1, MUTYH and CHEK2 in a Hereditary Cancer Family**
Agaoglu N. B., HATIRNAZ NG Ö., Unal B., AKGÜN DOĞAN Ö., Amanvermez U., Yildiz J., Doganay L., Ghazani A. A., Rana H. Q.
CANCER GENETICS, vol.268-269, pp.128-136, 2022 (SCI-Expanded)

XXV. **Familial early-onset obesity in Turkish children: variants and polymorphisms in the melanocortin-4 receptor (MC4R) gene**
Kirmizibekmez H., Demirkol Y. K., Dogan Ö., Seymen G., Balci E. I., Atla P., Dursun F.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.5, pp.657-662, 2022 (SCI-Expanded)

XXVI. **Consistency of variant interpretations among bioinformaticians and clinical geneticists in hereditary cancer panels**
Agaoglu N. B., Unal B., AKGÜN DOĞAN Ö., Kanev M. O., Zolfagharian P., Sag S. O., TEMEL Ş. G., Doganay L.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.3, pp.378-383, 2022 (SCI-Expanded)

XXVII. **COVID-19 PCR test performance on samples stored at ambient temperature**
Agaoglu N. B., Yildiz J., AKGÜN DOĞAN Ö., Kose B., Alkurt G., Demirkol Y. K., Irvem A., Doganay L., Dinler Doğanay G.
JOURNAL OF VIROLOGICAL METHODS, vol.301, 2022 (SCI-Expanded)

XXVIII. **Mutational spectrum of congenital long QT syndrome in Turkey; identification of 12 novel mutations across KCNQ1, KCNH2, SCN5A, KCNJ2, CACNA1C, and CALM1**
AKGÜN DOĞAN Ö., Agaoglu N. B., K. Demirkol Y., Doganay L., Ergul Y., Karacan M.
JOURNAL OF CARDIOVASCULAR ELECTROPHYSIOLOGY, vol.33, no.2, pp.262-273, 2022 (SCI-Expanded)

XXIX. **Determining the accuracy of next generation sequencing based copy number variation analysis in Hereditary Breast and Ovarian Cancer**
Agaoglu N. B., Unal B., Dogan Ö., Zolfagharian P., Sharifli P., Karakurt A., Senay B. C., Kizilboga T., Yildiz J., Dinler Doğanay G., et al.
EXPERT REVIEW OF MOLECULAR DIAGNOSTICS, vol.22, no.2, pp.239-246, 2022 (SCI-Expanded)

XXX. **Kohlschutter-Tonz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity**
Akgun-Dogan Ö., Simsek-Kiper P. O., Taskiran E., Schossig A., ÜTİNE G. E., Zschocke J., BODUROĞLU O. K.
JOURNAL OF CHILD NEUROLOGY, vol.36, no.10, pp.816-822, 2021 (SCI-Expanded)

XXXI. **Comparison of the clinical diagnostic criteria and the results of the next-generation sequence gene panel in patients with monogenic systemic autoinflammatory diseases**
Sozeri B., Demir F., SÖNMEZ H. E., Karadag S. G., Demirkol Y. K., AKGÜN DOĞAN Ö., Doganay H. L., Ayaz N. A.
CLINICAL RHEUMATOLOGY, vol.40, no.6, pp.2327-2337, 2021 (SCI-Expanded)

XXXII. **Does sampling saliva increase detection of SARS-CoV-2 by RT-PCR? Comparing saliva with oro-nasopharyngeal swabs**
Dogan Ö., Kose B., Agaoglu N. B., Yildiz J., Alkurt G., Demirkol Y. K., Irvem A., Doganay G. D., Doganay L.
JOURNAL OF VIROLOGICAL METHODS, vol.290, 2021 (SCI-Expanded)

XXXIII. **The same mutation in a family with adenosine deaminase 2 deficiency**
Sozeri B., Ercan G., Dogan Ö., Yildiz J., Demir F., Doganay L.
RHEUMATOLOGY INTERNATIONAL, vol.41, no.1, pp.227-233, 2021 (SCI-Expanded)

XXXIV. **Genetic panel screening in patients with clinically unclassified systemic autoinflammatory diseases**
Demir F., AKGÜN DOĞAN Ö., Demirkol Y. K., Tekkus K. E., Canbek S., Karadag S. G., SÖNMEZ H. E., Ayaz N. A., Doganay H. L., Sozeri B.
CLINICAL RHEUMATOLOGY, vol.39, no.12, pp.3733-3745, 2020 (SCI-Expanded)

XXXV. **H syndrome with a novel homozygousSLC29A3mutation in two sisters**
Demir D., Karabay E. A., Sozeri B., Gursoy F., Dogan Ö., Topaktas E., Zindanci I.
PEDIATRIC DERMATOLOGY, vol.37, no.6, pp.1135-1138, 2020 (SCI-Expanded)

XXXVI. **Peters Plus syndrome: a recognizable clinical entity**
Demir G. U., GÜLERAY LAFCI N., AKGÜN DOĞAN Ö., Kiper P. O. S., ÜTİNE G. E.
TURKISH JOURNAL OF PEDIATRICS, vol.62, no.1, pp.136-140, 2020 (SCI-Expanded)

XXXVII. **ADA2 deficiency in a patient with Noonan syndrome-like disorder with loose anagen hair: The co-occurrence of two rare syndromes**
Akgun-Dogan Ö., Simsek-Kiper P. O., TAŞKIRAN Z. E., Lissewski C., Brinkmann J., Schanze D., GÖÇMEN R., ÇAĞDAŞ AYVAZ D. N., BİLGİNER Y., ÜTİNE G. E., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.12, pp.2474-2480, 2019 (SCI-Expanded)

XXXVIII. **A Case of Leukoencephalopathy and Small Vessels Disease Caused by a Novel HTRA1 Homozygous Mutation**
GÜNDÜZ T., Demirkol Y., AKGÜN DOĞAN Ö., Demir S., Akcakaya N. H.
JOURNAL OF STROKE & CEREBROVASCULAR DISEASES, vol.28, no.11, 2019 (SCI-Expanded)

XXXIX. **Ophthalmo-acromelic syndrome in an infant**
Urel-Demir G., TAŞKIRAN Z. E., Akgun-Dogan Ö., Simek-Kiper P. O., ÜTİNE G. E.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.7, 2019 (SCI-Expanded)

XL. **Hyperphosphatasia with mental retardation syndrome type 4 In two siblings-expanding the phenotypic and mutational spectrum**
Dogan Ö., Demir G. U., KOŞUKCU C., TAŞKIRAN Z. E., Simsek-Kiper P. O., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.6, 2019 (SCI-Expanded)

XLI. **A novel NKX3-2 mutation associated with perinatal lethal phenotype of spondylo-megaepiphyseal-metaphyseal dysplasia in a neonate.**
ŞİMŞEK KİPER P. Ö., KOŞUKCU C., Akgun-Dogan Ö., GÖÇMEN R., ÜTİNE G. E., SOYER T., Korkmaz-Toygar A., Nishimura G., ALİKAŞİFOĞLU M., Boduroglu K.
European journal of medical genetics, vol.62, no.1, pp.21-26, 2019 (SCI-Expanded)

XLII. **Further expansion of the mutational spectrum of spondylo-meta-epiphyseal dysplasia with abnormal calcification**
Urel-Demir G., Simsek-Kiper P. O., Akgun-Dogan Ö., GÖÇMEN R., Wang Z., Matsumoto N., Miyake N., Utine G. E., Nishimura G., Ikegawa S., et al.
JOURNAL OF HUMAN GENETICS, vol.63, no.9, pp.1003-1007, 2018 (SCI-Expanded)

XLIII. **Further delineation of spondyloepimetaphyseal dysplasia Faden-Alkuraya type: A RSPRY1-associated spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis**
Simsek-Kiper P. O., Taskiran E. Z., KOŞUKCU C., Urel-Demir G., Akgun-Dogan Ö., YILMAZ G., ÜTİNE G. E., Nishimura G.,

BODUROĞLU O. K., ALİKAŞİFOĞLU M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.176, no.9, pp.2009-2016, 2018 (SCI-Expanded)

XLIV. **Prenatal and Postnatal Follow-up in Trisomies 13 and 18: A 20-Year Experience in a Tertiary Center**
Dogan Ö., Demir G. U., Arslan U., Simsek-Kiper P. O., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
AMERICAN JOURNAL OF PERINATOLOGY, vol.35, no.5, pp.427-433, 2018 (SCI-Expanded)

XLV. **Anauxetic dysplasia: A rare clinical entity**
Akgun-Dogan Ö., Simsek-Kiper P. O., ÜTİNE G. E., BODUROĞLU O. K.
TURKISH JOURNAL OF PEDIATRICS, vol.60, no.1, pp.89-93, 2018 (SCI-Expanded)

XLVI. **Coexistence of Trisomy.. and SRY (-) XX Ovotesticular Disorder of Sex Development**
Demir G. U., Dogan Ö., Kiper P. O. S., ÜTİNE G. E., BODUROĞLU O. K., Gucer S., ALİKAŞİFOĞLU M.
FETAL AND PEDIATRIC PATHOLOGY, vol.36, no.6, pp.445-451, 2017 (SCI-Expanded)

Articles Published in Other Journals

I. **A rare disease: ZAP70 deficiency**
Erdogan S., Cakmak S. C., Atay G., Akkus C. H., Karakayali B., Dogan Ö., Sozeri B.
NORTHERN CLINICS OF ISTANBUL, vol.11, no.2, pp.167-170, 2024 (ESCI)

II. **A Case with Hypotrichosis-Lymphedema-Telangiectasia Syndrome with Hair Shaft Fragility**
Atis G., Sari A. S., Soylu E., AKGÜN DOĞAN Ö.
SKIN APPENDAGE DISORDERS, vol.8, no.6, pp.511-514, 2022 (ESCI)

III. **Evaluation and Long-term Monitoring of Patients with MODY, and Description of Novel Mutations**
SAĞSAK E., ÖNDER A., KENDİRCİ H. N. P., Yildiz M., DOĞAN Ö. A.
JAREM, vol.12, no.2, pp.99-107, 2022 (Peer-Reviewed Journal)

IV. **Clinical Utility of Molecular Autopsy in Fetal and Pediatric Patients with Suspected Genetic Disorders**
DOĞAN Ö. A., AĞAOĞLU N. B., ALANAY Y.
Gümüşhane Üniversitesi Sağlık Bilimleri Dergisi, vol.11, no.1, pp.82-89, 2022 (Peer-Reviewed Journal)

V. **Clinical findings and molecular diagnosis in children with Bardet-Biedl Syndrome in Turkey: Identification of novel variants**
Doğan Ö., Ağaoğlu N. B.
Gulhane Medical Journal, vol.64, no.2, pp.144-151, 2022 (Scopus)

VI. **Evaluation of Clinical and Molecular Findings in a Group of Turkish Individuals with Marfan Syndrome**
AĞAOĞLU N. B., AKGÜN DOĞAN Ö.
İstanbul Kanuni Sultan Süleyman Tıp Dergisi, vol.14, no.1, pp.8-17, 2022 (Peer-Reviewed Journal)

VII. **Investigation of association between ABO blood groups and COVID-19 clinical severity**
İRVEM A., Sarmış A., AKGÜN DOĞAN Ö., YILDIZ J., HABİP Z., AĞAOĞLU N. B., Köse B., Alkurt G., Altunal L. N., Özel A. S., et al.
Medical Science and Discovery, vol.8, no.12, pp.671-674, 2021 (Peer-Reviewed Journal)

VIII. **Psychometric and Psychosocial Evaluation of Adolescents with Turner Syndrome in a Multidisiplinary Approach: A Preliminary Study**
Karakok B., AKDEMİR D., YALÇIN S. S., ÖZUSTA H. Ş., ÜTİNE G. E., AKGÜN DOĞAN Ö., Kiper P. O. S., Demir G. U.
GUNCEL PEDIATRI-JOURNAL OF CURRENT PEDIATRICS, vol.19, no.3, pp.363-372, 2021 (ESCI)

IX. **The origin of SARS-CoV-2 in Istanbul: Sequencing findings from the epicenter of the pandemic in Turkey**
Karacan I., Akgun T. K., Agaoglu N. B., Irvem A., Alkurt G., Yildiz J., Kose B., Ozel A. S., Altunal L. N., Can N. D., et al.
NORTHERN CLINICS OF ISTANBUL, vol.7, no.3, pp.203-209, 2020 (ESCI)

X. **The musculoskeletal system manifestations in children with familial Mediterranean fever**
Demir F., Bolac L. G., Merter T., Canbek S., Dogan Ö., Demirkol Y. K., Yildiz J., Doganay H. L., Sozeri B.
NORTHERN CLINICS OF ISTANBUL, vol.7, no.5, pp.438-442, 2020 (Peer-Reviewed Journal)

XI. **Responding to COVID-19 in Istanbul: Perspective from genomic laboratory**

Doganay L., Agaoglu N. B., Irvem A., Alkurt G., Yildiz J., Kose B., Demirkol Y. K., AKGÜN DOĞAN Ö., Dinler Doğanay G.
NORTHERN CLINICS OF ISTANBUL, vol.7, no.3, pp.311-312, 2020 (ESCI)

XII. **Varicella-Related Hospitalizations Among Immunocompetent and Immunocompromised Children in Pre-Vaccine Era: A Tertiary Care Center Experience in Turkey**
Dogan Ö., Topcu S., Tanir N. G.
JOURNAL OF PEDIATRIC RESEARCH, vol.5, no.1, pp.11-16, 2018 (Peer-Reviewed Journal)

XIII. **Monogenic diabetes Monogenik diyabet**
Doğan Ö., Utine G. E., Alikaşifoğlu A.
Cocuk Sagligi ve Hastaliklari Dergisi, vol.59, no.3, pp.140-151, 2016 (Scopus)

Books

I. **Marfan Sendromu**
AKGÜN DOĞAN Ö.
in: Çocuk Genetik Hastalıkları, Mıhçı Ercan, Editor, Türkiye Klinikleri Yayınevi, Ankara, pp.79-85, 2021

II. **Akondroplazi**
AKGÜN DOĞAN Ö., ALANAY Y.
in: Çocuk Genetik Hastalıkları, Mıhçı, Ercan, Editor, Türkiye Klinikleri Yayınevi, Ankara, pp.66-71, 2021

III. **Genetik Danışma**
AKGÜN DOĞAN Ö., ALANAY Y.
in: Cerebral Palsy ve Genetik, Özbek Uğur, Akçakaya Nihan Hande, Editor, Boyut Yayınevi, İstanbul, pp.57-65, 2019

IV. **Nörokütanöz Hastalıklar**
AKGÜN DOĞAN Ö., BODUROĞLU O. K.
in: Yurdakök Pediatri, Yurdakök, Murat, Editor, Güneş Tıp Kitabevleri, Ankara, pp.2098-2112, 2017

Papers Presented at Peer-Reviewed Scientific Conferences

I. **Unveiling the Protein Landscape in Cerebral Palsy Through AI-Based Structural Analysis**
Özkeserli Z., Yiğit A., Akgün Doğan Ö., Bilgüvar K., Özbek U., Bayram Akçapinar G.
17th The International Symposium on Health Informatics and Bioinformatics, İstanbul, Turkey, 18 - 20 December 2024, pp.161, (Summary Text)

II. **EARLY REAL-WORLD EXPERIENCE WITH VOSORITIDE TREATMENT IN ACHONDROPLASIA: A SINGLE-CENTER REPORT FROM TURKEY**
Abalı S., Özköse S. G., Akgün Doğan Ö., Semiz S., Alanay Y.
16. International Skeletal Dysplasia Society Meeting, Madrid, Spain, 18 - 21 September 2024, (Unpublished)

III. **Yoğun Bakımdaki Kritik Yenidoğan ve Süt Çocuklarında Hızlı Yeni Nesil Dizileme ile Genetik Tanı-İlk Türkiye Deneyimi**
Güner Yılmaz B., Akgün Doğan Ö., Özdemir Ö., Bilgüvar K., Hatırnaz Ng Ö., Özköse G. Ş., Demirel A., Aktaş S., Arcagok B. C., Kazancı E., et al.
Ulusal Neonatoloji Kongresi, Antalya, Turkey, 24 - 28 April 2024, (Unpublished)

IV. **Functional and structural landscape of Cerebral Palsy (CP) related proteins**
Özkeserli Z., Yiğit A., Akgün Doğan Ö., Bilgüvar K., Özbek U., Bayram Akçapınar G.
Applied Bioinformatics in Life Sciences (5th edition), Leuven, Belgium, 7 - 08 March 2024, pp.1, (Summary Text)

V. **First Report of Heterozygous Intragenic Deletion in NPR2 Presenting with Familial Short Stature**
Akgün Doğan Ö., Alanay Y.
19th Manchester Dysmorphology Conference 2023, Manchester, England, 16 - 18 November 2023, pp.10-12, (Full Text)

VI. **Camptodactyly, Tall Stature, And Hearing Loss Syndrome with a Novel Homozygous FGFR3 Variant**
Akgün Doğan Ö., Abalı S., Alanay Y.

19th Manchester Dysmorphology Conference 2023, Manchester, England, 16 - 18 November 2023, pp.13-14, (Full Text)

VII. **Türkiye'De Yoğun Bakımdaki Kritik Pediatrik Hastalarda Hızlı Tüm Genom Dizileme: Bir Ön Çalışma**
Akgün Doğan Ö., Alanay Y., Özdemir Ö., Hatırnaz Ng Ö., Beken S.
6. Çocuk Genetik Ulusal Kongresi, İzmir, Turkey, 9 - 12 November 2023, pp.87-88, (Full Text)

VIII. **Obstacles and Expectations of Rare Disease Patients and Their Families in Türkiye: ISTisNA Project Survey Results**
Hatırnaz Ng Ö., Özdemir Ö., Alanay Y., Akgün Doğan Ö.
12th International Conference on Rare and Undiagnosed Diseases, Tbilisi, Georgia, 22 - 23 October 2023, pp.1, (Summary Text)

IX. **From patient to function: modeling CRIM1 in xenopus tropicalis**
AKGÜN DOĞAN Ö., Viviano S., HATIRNAZ NG Ö., Agaoglu N. B., Ji W., Jeffries L., Ozbek U., Lakhani S., Khokha M., Deniz E., et al.
55th European-Society-of-Human-Genetics (ESHG) Conference, Vienna, Austria, 11 - 14 June 2022, pp.656-657, (Summary Text)

X. **Biallelic copy number variations in both upstream & downstream enhancers of SHOX gene causes mesomelia and clubfoot without short stature**
Yilmaz B. G., ABALI S., Akberzade A., Ay B., Tumer S., AKGÜN DOĞAN Ö., Nishimura G., ALANAY Y.
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UDNI Conference, Vienna, Austria, 7 - 08 November 2022, (Unpublished)

XII. **Severely Affected Newborn Female With De Novo Likely Pathogenic Variant In BCAP31**
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Undiagnosed diseases network international (UDNI), Vienna, Austria, 7 - 08 November 2022, (Unpublished)

XIII. **VERİ ANALİZİ VE YORUMLAMA**
AKGÜN DOĞAN Ö.
5.Ulusal Çocuk Genetik Kongresi, Turkey, 07 October 2021, (Full Text)

XIV. **Gilbert Sendromu: Genetik Test Yapılması Gerekli mıdır?**
GERENLİ N., KENDİR DEMİRKOL Y., AKGÜN DOĞAN Ö.
63. TÜRKİYE MİLLİ PEDİATRİ KONGRESİ, Gazi Mağosa, Cyprus (Kktc), 30 October 2019, (Summary Text)

XV. **Vascular ehlers-danlos syndrome**
Agaoglu N., Demirkol Y. K., Dogan Ö., Say M., Akgun T. K., Doganay H.
52nd Conference of the European-Society-of-Human-Genetics (ESHG), Gothenburg, Sweden, 15 - 18 June 2019, vol.27, pp.1279, (Summary Text)

XVI. **Stuve-Wiedemann syndrome: a rare clinical entity**
Dogan Ö., Demirkol Y. K., Say M., Akgun T. K., Doganay L.
52nd Conference of the European-Society-of-Human-Genetics (ESHG), Gothenburg, Sweden, 15 - 18 June 2019, vol.27, pp.1300-1301, (Summary Text)

XVII. **A novel mutation in COL2A1 leading to spondyloepiphyseal dysplasia congenita**
Demirkol Y. K., Dogan Ö., Say M., Akgun T. K., Doganay L.
52nd Conference of the European-Society-of-Human-Genetics (ESHG), Gothenburg, Sweden, 15 - 18 June 2019, vol.27, pp.1273-1274, (Summary Text)

XVIII. **GENETIC SCREENING IN PATIENTS WITH UNDIFFERENTIATED PERIODIC FEVER SYNDROME**
Demir F., Dogan Ö., Kendirci Y., Tekkus K. E., Canbek S., AKTAY AYAZ N., Doganay L., Sozeri B.
Annual European Congress of Rheumatology (EULAR), Madrid, Spain, 12 - 15 June 2019, vol.78, pp.974, (Summary Text)

XIX. **Ophthalmo-acromelic syndrome in an infant**
Demir G. U., TAŞKIRAN Z. E., DOĞAN Ö., Kiper P. O. S., ÜTİNE G. E.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26,

pp.959, (Summary Text)

XX. **RSPRY1-associated skeletal dysplasia: Spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis**
Simsek-Kiper P. O., TAŞKIRAN Z. E., KOŞUKCU C., Akgun-Dogan Ö., YILMAZ G., ÜTİNE G. E., Nishimura G., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.249-250, (Summary Text)

XXI. **Absence of major eye malformations further expands the phenotype of SOX2 deletions**
Guleray N., Demir G. U., DOĞAN Ö., Kiper P. O. S., ÜTİNE G. E., ALİKAŞİFOĞLU M.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.500-501, (Summary Text)

XXII. **Homozygous novel variant in MUT in a patient with intellectual disability without metabolic derangement**
Utine E. G., TAŞKIRAN Z. E., KOŞUKCU C., DOĞAN Ö., Kiper O. P. S., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.932, (Summary Text)

Funded Projects

Akgün Doğan Ö., Alanay Y., Abalı S., Özdemir Ö., TÜBİTAK International Bilateral Joint Cooperation Program Project, Combining Multidisciplinary Re-phenotyping with Re-analysis of WES/WGS Data in Patients with Undiagnosed Skeletal Dysplasias, 2024 - 2026

BAYRAM AKÇAPINAR G., DOĞAN T., Bilgüvar K., HATIRNAZ NG Ö., ÖZBEK U., AKGÜN DOĞAN Ö., TUBITAK Project, Deep-CP: Development of a Deep Learning Variant Pathogenicity Prediction Tool using AlphaFold for Cerebral Palsy , 2022 - 2025

Akgün Doğan Ö., Ağbaş A., Saygılı S. K., Canpolat N., Özaltın F., Project Supported by Higher Education Institutions, Kalıtsal nadir böbrek hastalıklarında yeni genlerin araştırılması, 2021 - 2025

Akgün Doğan Ö., Özbek U., Hatırnaz Ng Ö., Alanay Y., Development Agency, İStanbul Tanısız ve NAdir Hastalıklara Çözüm Platformu-İSTisNA, 2022 - 2024

Alanay Y., Özbek U., Hatırnaz Ng Ö., Akgün Doğan Ö., Özdemir Ö., Çıtak A., Korkmaz Toygar A., Beken S., Kazancı E., Demirel A., et al., Project Supported by Higher Education Institutions, Yenidoğan ve Çocuk Yoğun Bakım Ünitesinde (YYBÜ/ÇYBÜ) Hızla Kötüleşen Kritik Yenidoğan ve Süt Çocuklarında Hızlı Yeni Nesil Dizileme ile Genetik Tanı, 2022 - 2024

Akgün Doğan Ö., Alanay Y., Hatırnaz Ng Ö., Açıkel Elmas M., Project Supported by Higher Education Institutions, "Herediter Spastik Paraparezili Bir Ailede Klinik İnceleme ve ERLIN1 Geninin Hastalık Patofizyolojisindeki Yerinin Aydınlatılması", 2022 - 2023

Akgün Doğan Ö., Other International Funding Programs, Investigation of the Functions of Candidate Genes Determined by WholeExome Sequence Analysis in a Group of Congenital Malformations Using CRISPR /Cas9 Gene Regulation Technology in Xenopus Frog Model, 2021 - 2022

Activities in Scientific Journals

EUROPEAN JOURNAL OF MEDICAL GENETICS, Assistant Editor/Section Editor, 2023 - Continues

Memberships and Roles in Scientific Organizations

European Board of Clinical Genetics, Member, 2022 - Continues, Austria

Undiagnosed Disease Network International, Country Representative, 2022 - Continues, Turkey

Milli Pediatri Derneği, Member, 2019 - Continues, Turkey

European Society of Human Genetics, Member, 2019 - Continues, Austria
Çocuk Genetik Hastalıkları Derneği, Member, 2012 - Continues, Turkey

Scientific Consultations

Trispera İlaç, Scientific Consultancy, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, Turkey, 2024 - Continues

Roles in Event Organizations

Akgün Doğan Ö., 6. Çocuk Genetik Ulusal Kongresi, Scientific Congress, Turkey, November 2023

Scientific Research / Working Group Memberships

Udnı Functional Study Group, Baylor College of Medicine, United States Of America,
https://www.udninternational.org/schede-14-members, 2022 - Continues
Udnı Diagnostic Working Group, Karolinska Institutet (Karolinska Institute), Sweden,
https://www.udninternational.org/schede-10-groups_with_udn_programs_activities_ongoing, 2022 - Continues

Mobility Activity

Post Doc, Post Doc, Yale University, United States Of America, 2021 - 2022

Congress and Symposium Activities

3. Cerrahpaşa Pediatri Günleri, Invited Speaker, İstanbul, Turkey, 2023
12th Undiagnosed Diseases Network International Conference , Invited Speaker, Tbilisi, Georgia, 2023
Prof. Selçuk Apak İstanbul Çocuk Nöroloji Toplantıları , Attendee, İstanbul, Turkey, 2023
19th Manchester Dysmorphology Conference 2023, Attendee, Manchester, England, 2023
6. Ulusal Çocuk Genetik Kongresi, Invited Speaker, İzmir, Turkey, 2023
6. Çocuk Genetik Hastalıkları Kongresi, Session Moderator, İzmir, Turkey, 2023
II.Genetik ve İmmünoloji Kampı, Invited Speaker, İstanbul, Turkey, 2023
Undiagnosed Hackathon, Working Group, Stockholm, Sweden, 2023
5. Ege Endokrin Hastalıklar ve Genetik Sempozyumu, Session Moderator, İzmir, Turkey, 2023
11th Undiagnosed Disease Network International Conference, Attendee, Vienna, Austria, 2022
Cerrahpaşa Genetik ve İmmünoloji Kampı, Invited Speaker, İstanbul, Turkey, 2022
European Society of Human Genetics Conference, Attendee, Vienna, Austria, 2022
Çukurova Pediatri Kongresi , Invited Speaker, Adana, Turkey, 2022
5. Ulusal Çocuk Genetik Kongresi, Invited Speaker, İzmir, Turkey, 2021

Invited Talks

Use Cases from ACURAREACIBADEM University: Unsolved cases, Workshop, EJP RD Training on "Strategies to foster solutions of undiagnosed RD cases, Italy, March 2024
3. Cerrahpaşa Pediatri Günleri/Genetik testleri isterken nelere dikkat edelim? Nasıl yorumlayalım?, Conference, İstanbul Üniversitesi-Cerrahpaşa, Turkey, December 2023

İSTİSNA Nadir ve Tanısız Hastalıkların Çözümünde Kullanılacak Stratejiler, Genomik Yaklaşımlar ve Veri Analizi Eğitim, Workshop, İstanbul Üniversitesi-Cerrahpaşa, Turkey, November 2023
Zihinsel Yetersizlikte RNA ve Epigenetik/ 6. Çocuk Genetik Ulusal Kongresi, Conference, Çocuk Genetik Hastalıkları Derneği, Turkey, November 2023
II.Genetik ve İmmünoloji Kampı, Seminar, Romatoloji Derneği, Turkey, November 2023
NGS Veri Analizi/6. Çocuk Genetik Ulusal Kongresi, Conference, Çocuk Genetik Hastalıkları Derneği, Turkey, November 2023
Current Clinical Genetic Approaches for Rare and Undignosed Diseases/12th Undiagnosed Diseases Network International Conference, Conference, Undiagnosed Diseases Network International , Georgia, November 2023
Pediatride Genetik Testlerin Kullanımı, Conference, Çukurova Üniversitesi, Turkey, June 2022
Genetik Testlerin Yorumlanması, Workshop, Cerrahpaşa Romatoloji Derneği, Turkey, June 2022
Veri Analizi ve Yorumlama, Conference, Turkey, October 2021

Scholarships

Visiting Scholar, Fulbright Program, 2021 - 2022

Representation and Promotion Activities

Institutional Representation, 12th Undiagnosed Diseases Network International Conference , Georgia, Tbilisi, 2023 - 2023
Institutional Representation, Undiagnosed Disease Network International, Austria, Vienna, 2022 - 2022
Institutional Representation, Cerrahpaşa Romatoloji Derneği , Turkey, İstanbul, 2022 - 2022
Institutional Representation, European Society of Human Genetics, Austria, Vienna, 2022 - 2022
Institutional Representation, Pediatride Genetik Testlerin Kullanımı, Turkey, Adana, 2022 - 2022