Asst. Prof. ÖZLEM AKGÜN DOĞAN

Personal Information

Email: Ozlem.Dogan@acibadem.edu.tr
Web: https://avesis.acibadem.edu.tr/Ozlem.Dogan

Education Information

Doctorate, Hacettepe University, Sağlık Bilimleri Enstitüsü, Turkey 2015 - Continues
Post Doctorate of Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Nursing, Turkey 2012 - 2018
Expertise In Medicine, Diğer (Kurumlar,hastaneler Vb.), Ankara Dr.Sami Ulus Çocuk Sağlığı ve Hastalıkları Eğitim Araştırma Hastanesi, Çocuk Sağlığı ve Hastalıkları, Turkey 2007 - 2012
Undergraduate, Hacettepe University, Tıp Fakültesi (Türkçe), Turkey 2000 - 2006

Research Areas

Health Sciences

Academic Titles / Tasks

Assistant Professor, Acibadem Mehmet Ali Aydinlar University, School of Medicine, Department of Medical Sciences, 2021 - Continues

Courses

Mendelian Inheritance, Undergraduate, 2020 - 2021
Clinical Cytogenetics, Undergraduate, 2020 - 2021
Non-mendelian Inheritance, Undergraduate, 2020 - 2021
PANEL: FROM GENOTYPE TO PHENOTYPE, Undergraduate, 2020 - 2021

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

I. **Diagnostic yield of whole-exome sequencing in non-syndromic intellectual disability**
TAŞKIRAN Z. E. , KARAOSMANOĞLU B., KOŞUKCU C., Urel-Demir G., Akgun-Dogan Ö., Simsek-Kiper P. o. , ALİKAŞİFOĞLU M., BODUROĞLU O. K. , ÜTİNE G. E.
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III. **Kohlschutter-Tonz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity**
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IV. **Two Siblings with Kaufman Oculocerebrofacial Syndrome Resembling Oculoauriculovertebral**

Spectrum

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V. **Does sampling saliva increase detection of SARS-CoV-2 by RT-PCR? Comparing saliva with oro-nasopharyngeal swabs**

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VI. **The same mutation in a family with adenosine deaminase 2 deficiency**

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VII. **Genetic panel screening in patients with clinically unclassified systemic autoinflammatory diseases**

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VIII. **H syndrome with a novel homozygousSLC29A3mutation in two sisters**

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IX. **Cafe noir spots: a feature of familial progressive hyper- and hypopigmentation**

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X. **Further Phenotypic Delineation of Partial Trisomy 17q and Partial Monosomy 20q due to Rare t(17;20)**

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XI. **Peters Plus syndrome: a recognizable clinical entity**

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TURKISH JOURNAL OF PEDIATRICS, vol.62, no.1, pp.136-140, 2020 (Journal Indexed in SCI)

XII. **ADA2 deficiency in a patient with Noonan syndrome-like disorder with loose anagen hair: The co-occurrence of two rare syndromes**

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AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.12, pp.2474-2480, 2019 (Journal Indexed in SCI)

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XIV. **Ophthalmo-acromelic syndrome in an infant**

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XV. **Hyperphosphatasia with mental retardation syndrome type 4 In two siblings-expanding the phenotypic and mutational spectrum**

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XVI. **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 (Journal Indexed in SCI Expanded)

XVII. **Further expansion of the mutational spectrum of spondylo-meta-epiphyseal dysplasia with abnormal calcification**
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JOURNAL OF HUMAN GENETICS, vol.63, no.9, pp.1003-1007, 2018 (Journal Indexed in SCI)

XVIII. **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.
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XIX. **Prenatal and Postnatal Follow-up in Trisomies 13 and 18: A 20-Year Experience in a Tertiary Center**
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XX. **Fragile x-associated premature ovarian failure in a large Turkish cohort: Findings of Hacettepe Fragile X Registry.**
ÜTİNE G. E. , ŞİMŞEK KİPER P. Ö. , Akgun-Dogan Ö., ÜREL DEMİR G., Alanay Y., Aktas D., Boduroglu K., Tuncbilek E., ALİKAŞİFOĞLU M.
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XXII. **HERC1 mutations in idiopathic intellectual disability**
ÜTİNE G. E. , TAŞKIRAN Z. E. , KOŞUKCU C., KARAOSMANOĞLU B., Guleray N., Dogan Ö., Kiper P. O. S. , BODUROĞLU O. K. , ALİKAŞİFOĞLU M.
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XXIII. **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 (Journal Indexed in SCI)

Articles Published in Other Journals

I. **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. , Dogan Ö., Dinler Doğanay G.
NORTHERN CLINICS OF ISTANBUL, vol.7, no.3, pp.311-312, 2020 (Journal Indexed in ESCI)

II. **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 (Journal Indexed in ESCI)

III. **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 (Journal Indexed in ESCI)

IV. **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 (Journal Indexed in ESCI)

V. **A Diagnosis to Consider in an Adult Patient with Facial Features and Intellectual Disability: Williams Syndrome**
Dogan Ö., Kiper P. O. S. , ÜTİNE G. E. , ALİKAŞİFOĞLU M., BODUROĞLU O. K.

KOREAN JOURNAL OF FAMILY MEDICINE, vol.38, no.2, pp.102-105, 2017 (Journal Indexed in ESCI)

VI. **Monogenic diabetes Monogenik diyabet**
Doğan Ö., Utine G. E. , Alikaşifoğlu A.
Cocuk Sagligi ve Hastaliklari Dergisi, vol.59, no.3, pp.140-151, 2016 (Refereed Journals of Other Institutions)

Refereed Congress / Symposium Publications in Proceedings

I. **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

II. **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

III. **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

IV. **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

V. **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

VI. **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

VII. **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

VIII. **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

IX. **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

Memberships / Tasks in Scientific Organizations

Çocuk Genetik hastalıkları Derneği, Member, 2016 - Continues, Turkey

Citations

Total Citations (WOS):51
h-index (WOS):4

Scholarships

Visiting Scholar Programme, Fulbright Program, 2021 - Continues