Asst. Prof. NAZ GÜLERAY LAFCI

Personal Information

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International Researcher IDs
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Education Information

Expertise In Medicine, Hacettepe University, Tıbbi Genetik Anabilim Dalı, Turkey 2014 - 2019
Undergraduate, Gazi University, Tıp Fakültesi, Tıp, Turkey 2007 - 2014

Foreign Languages

Spanish, B2 Upper Intermediate
English, C1 Advanced

Dissertations

Expertise In Medicine, Investigation of genetic causes in oculoauriculovertebral spectrum etiology, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2019

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Medical Genetics

Academic Titles / Tasks

Lecturer PhD, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2021 - Continues
Research Assistant, Hacettepe University, Tıp Fakültesi (Türkçe), Tıp Fakültesi, 2014 - 2019

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Decreased calcium permeability caused by biallelic TRPV5 mutation leads to autosomal recessive renal calcium-wasting hypercalciuria**
GÜLERAY LAFCI N., van Goor M., Cetinkaya S., van der Wijst J., Acun M., Kurt Colak F., ÇETİNKAYA A., Hoenderop J.
European Journal of Human Genetics, 2024 (SCI-Expanded)
II. **Mutated Transcripts of ZEB2 Do Not Undergo Nonsense-Mediated Decay in Mowat-Wilson Syndrome**
Güleray Lafcl N., KARAOSMANOĞLU B., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E.

Molecular Syndromology, vol.14, no.3, pp.258-265, 2023 (SCI-Expanded)

III. **Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum**
Güleray N., Koşukcu C., Oğuz S., Ürel Demir G., Taşkıran E. Z., Kiper P. Ö. Ş., Utine G. E., Alanay Y., Boduroğlu K., Alikaşifoğlu M.
CLEFT PALATE-CRANIOFACIAL JOURNAL, vol.59, no.9, pp.1114-1124, 2022 (SCI-Expanded)

IV. **A Case of SHOX Deletion Due to Isodicentric Y Chromosome Anomaly with Multiple Endocrine Disorders**
Orman B., Kucukali G. K., GÜLERAY LAFCI N., Donmez B. O., Erdeve S. S., Cetinkaya S.
HORMONE RESEARCH IN PAEDIATRICS, vol.95, no.SUPPL 2, pp.533-534, 2022 (SCI-Expanded)

V. **Hypergonadotrophic hypogonadism in a patient with transaldolase deficiency: novel mutation in the pentose phosphate pathway**
Lafci N., Colak F. K., Sahin G., Sakar M., Cetinkaya S., Savas-Erdeve S.
HORMONES-INTERNATIONAL JOURNAL OF ENDOCRINOLOGY AND METABOLISM, vol.20, no.3, pp.581-585, 2021 (SCI-Expanded)

VI. **Retrospective evaluation of patients with X-linked adrenoleukodystrophy with a wide range of clinical presentations: a single center experience**
Olgac A., Kasaplcara C. S., Derinkuyu B., Yuksel D., Cetinkaya S., Aksoy A., Ceylaner S., Guleray N., Yesilipek A., Aydin H. I., et al.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.9, pp.1169-1179, 2021 (SCI-Expanded)

VII. **Clinical Management in Systemic Type Pseudohypoaldosteronism Due to SCNN1B Variant and Literature Review.**
Karacan Küçükali G., Çetinkaya S., Tunç G., Oğuz M. M., Çelik N., Akkaş K. Y., Şenel S., Güleray Lafcı N., Savaş Erdeve Ş.
Journal of clinical research in pediatric endocrinology, vol.13, pp.446-451, 2021 (SCI-Expanded)

VIII. **An intronic variant inBRAT1creates a cryptic splice site, causing epileptic encephalopathy without prominent rigidity**
Colak F. K., Guleray N., Azapagasi E., Yazici M. U., Aksoy E., Ceylan N.
ACTA NEUROLOGICA BELGICA, vol.120, no.6, pp.1425-1432, 2020 (SCI-Expanded)

IX. **Atypical Presentation of Sengers Syndrome: A Novel Mutation Revealed with Postmortem Genetic Testing.**
Guleray N., Kosukcu C., Taskiran Z., Simsek K., Utine G., Gucer S., Tokatli A., Boduroglu K., Alikasifoglu M.
Fetal and pediatric pathology, vol.39, no.2, pp.163-171, 2020 (SCI-Expanded)

X. **Café noir spots: a feature of familial progressive hyper- and hypopigmentation.**
Gulseren D., Guleray N., Akgun-Dogan O., Simsek-Kiper P. O., Utine E. G., Alikasifoglu M., Ersoy-Evans S.
Journal of the European Academy of Dermatology and Venereology : JEADV, vol.34, no.2, 2020 (SCI-Expanded)

XI. **Further Phenotypic Delineation of Partial Trisomy 17q and Partial Monosomy 20q due to Rare t(17;20)**
Ürel-Demir G., Akgün-Doğan Ö., OĞUZ S., Güleray-Lafcl N., Şimşek-Kiper P. Ö., Eda Utine G., ALİKAŞİFOĞLU M., Boduroğlu K.
MOLECULAR SYNDROMOLOGY, vol.11, no.1, pp.38-42, 2020 (SCI-Expanded)

XII. **A Monogenic Disease with a Variety of Phenotypes: Deficiency of Adenosine Deaminase 2**
Ozen S., Bilginer Y., Batu E., Taşkıran E., Özkara H. A., Ünal Ş., Güleray N., Erden A., Karadağ Ö., Gümrük F., et al.
JOURNAL OF RHEUMATOLOGY, vol.47, no.1, pp.117-125, 2020 (SCI-Expanded)

XIII. **Peters Plus syndrome: a recognizable clinical entity**
ÜREL DEMİR G., Lafci N., Dogan O. A., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E.
TURKISH JOURNAL OF PEDIATRICS, vol.62, no.1, pp.136-140, 2020 (SCI-Expanded)

XIV. **Intrafamilial variability of XYLT2-related spondyloocular syndrome**
Guleray N., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., Boduroglu K., ALİKAŞİFOĞLU M.
European Journal of Medical Genetics, vol.62, no.11, 2019 (SCI-Expanded)

XV. **Retrospective analysis of indications for termination of pregnancy**
ÖZYÜNCÜ Ö., ÖRGÜL G., TANAÇAN A., AKTOZ F., GÜLERAY N., FADILOĞLU E., BEKSAÇ M. S.
JOURNAL OF OBSTETRICS AND GYNAECOLOGY, vol.39, no.3, pp.355-358, 2019 (SCI-Expanded)

XVI. **Evaluation of Pregnancies in 25 Families with Balanced/Unbalanced Chromosomal Translocations**
GÜLERAY N., Yucesoy H. M., FADILOĞLU E., TANAÇAN A., ALİKAŞİFOĞLU M., BEKSAÇ M. S.
INTERNATIONAL JOURNAL OF HUMAN GENETICS, vol.19, no.1, pp.22-28, 2019 (SCI-Expanded)

XVII. **Growth Hormon Deficiency in Identical Twins with Gitelman Syndrome Due to Compound Heterozygous Mutation (p.R80fs*35/p.K957X) of the SLC12A3 Gene and the Evaluation of the Response to Growth Hormone Replacement Therapy**
Yaman B., Celegen K., Korkmaz E., Lafci N., Balik Z., DEMİRBİLEK H., DÜZOVA A.
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.461-462, 2018 (SCI-Expanded)

XVIII. **Partial Trisomy 18 From a marker chromosome to a rare chromosomal disorder**
OĞUZ S., GÜLERAY N., DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
MOLECULAR CYTOGENETICS, vol.10, 2017 (SCI-Expanded)

XIX. **HERC1 mutations in idiopathic intellectual disability**
ÜTİNE G. E., Taskiran E. Z., KOŞUKCU C., KARAOSMANOĞLU B., GÜLERAY N., Dogan O. A., Kiper P. O. S., Boduroglu K., ALİKAŞİFOĞLU M.
European Journal of Medical Genetics, vol.60, no.5, pp.279-283, 2017 (SCI-Expanded)

Refereed Congress / Symposium Publications in Proceedings

I. **Expanding the clinical and mutational spectrum of Roberts syndrome with previously unreported endocrine findings.**
GÜLERAY N., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
European Human Genetics Conference, 16 - 19 June 2018

II. **Okuloaurikülovertebral spektrumda 5p delesyonu**
ÜREL DEMİR G., OĞUZ S., GÜLERAY N., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

III. **6p25.3 delesyonu**
AKGÜN DOĞAN Ö., ÜREL DEMİR G., GÜLERAY N., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., KUTLUK M. T., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
3.Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

IV. **Taşıyıcı kız kardeşlerin oğullarında zihinsel yetersizlik**
ÜREL DEMİR G., OĞUZ S., GÜLERAY N., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
3. Ulusal Çocuk Genetik Sempozyumu, Antalya, Turkey, 11 - 13 October 2017

V. **Partial Trisomy 18 From a marker chromosome to a rare chromosomal disorder**
OĞUZ S., GÜLERAY N., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K., ALİKAŞİFOĞLU M.
European Cytogenetics Conference, 1 - 04 July 2017

VI. **Hennekam sendromu Otozomal resesif geçişli bir konjenital lenfödem**
GÜLERAY N., ÖZER M., AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
2. Ulusal Çocuk Genetik Sempozyumu, 22-24 Ekim 2015, Samsun, Türkiye, Turkey, 22 - 24 October 2015

VII. **Roberts SC Phocomelia Syndrome A Rare Clinical Entity**
GÜLERAY N., ŞİMŞEK KİPER P. Ö., DEMİREL M., ÇETİNKAYA A., ÜTİNE G. E., ALİKAŞİFOĞLU M., BODUROĞLU O. K.
International Skeletal Dysplasia Society 12th Biennial Meeting, 29 July - 01 August 2015

Supported Projects

ALİKAŞİFOĞLU M., GÜLERAY N., Project Supported by Higher Education Institutions, Okuloaurikulovertebral Spektrum Etyolojisinde Genetik Nedenlerin Araştırılması, 2018 - 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

Metrics

Publication: 28
Citation (WoS): 78
Citation (Scopus): 135
H-Index (WoS): 4
H-Index (Scopus): 5

Non Academic Experience

Public Hospital
Public Hospital
ANKARA DR.SAMİ ULUS KADIN DOĞUM ÇOCUK SAĞLIĞI VE HASTALIKLARI EĞİTİM VE ARAŞTIRMA HASTANESİ