

# Prof. GÜLEN EDA ÜTİNE

## Personal Information

**Email:** geutine@hacettepe.edu.tr

**Web:** <https://avesis.hacettepe.edu.tr/geutine>

## Education Information

Doctorate, Hacettepe University, Çocuk Sağlığı Enstitüsü, Genetik Doktora Programı, Turkey 2005 - 2011

Post Doctorate of Medicine, Hacettepe University, Tıp Fakültesi, Çocuk Genetik Hastalıkları, Turkey 2004 - 2011

Expertise In Medicine, Hacettepe University, Tıp Fakültesi, Pediatri, Turkey 1998 - 2004

Undergraduate, Hacettepe University, Tıp Fakültesi, Turkey 1992 - 1998

## Foreign Languages

English, C1 Advanced

## Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Child Health and Diseases, Pediatric Genetics and Teratology

## Academic and Administrative Experience

Hacettepe Üniversitesi, Çocuk Sağlığı Enstitüsü, 2012 - 2016

## Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Apparent mineralocorticoid excess: A diagnosis beyond classical causes of severe hypertension in a child**  
GÜLHAN B., ÜNSAL Y., BALTU D., ÇELİK ERTAŞ N. B. , Ozdemir G., Utine E., ÖZCAN H. N. , DÜZOVA A., Gonc N.  
BLOOD PRESSURE MONITORING, vol.27, no.3, pp.208-211, 2022 (Journal Indexed in SCI)
- II. **Efficacy of flecainide in bidirectional ventricular tachycardia and tachycardia-induced cardiomyopathy with Andersen-Tawil syndrome**  
ÜNAL YÜKSEKGÖNÜL A., Azak E., AKALIN A., ERTUĞRUL İ., Kilic E., ÜTİNE G. E. , KARAGÖZ T.  
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.65, no.6, 2022 (Journal Indexed in SCI)
- III. **Typical Face, Developmental Delay, and Hearing Loss in a Patient with 3M Syndrome: The Co-Occurrence of Two Rare Conditions**  
AKALIN A., Simsek-Kiper P. O. , Taskiran E., ÜTİNE G. E. , BODUROĞLU O. K.  
MOLECULAR SYNDROMOLOGY, 2022 (Journal Indexed in SCI)
- IV. **Diagnostic distribution and postnatal evaluation of prenatally detected short femur: A single center experience**  
KAHRAMAN A. B. , ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E. , BODUROĞLU O. K.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, 2022 (Journal Indexed in SCI)
- V. **More than meets the eye: expanding and reviewing the clinical and mutational spectrum of brittle**

## **cornea syndrome**

Dhooge T., Van Damme T., Syx D., Mosquera L. M. , Nampoothiri S., Radhakrishnan A., ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E. , Bonduelle M., Migeotte I., et al.

EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.SUPPL 1, pp.156-157, 2022 (Journal Indexed in SCI)

- VI. **Al-Gazali skeletal dysplasia constitutes the lethal end of ADAMTSL2-related disorders**  
Batkovskytte D., McKenzie F., Taylan F., ŞİMŞEK KİPER P. Ö. , Nikkel S. M. , Ohashi H., Miyahara H., Eriksson G., Ha T., ÜTİNE G. E. , et al.  
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.SUPPL 1, pp.41-42, 2022 (Journal Indexed in SCI)
- VII. **MENSTRUATION RELATED QUALITY OF LIFE IN ADOLESCENTS WITH GENETIC SYNDROMES ACCOMPANYING AN INTELLECTUAL DISABILITY**  
ÜÇLER ÇINAR H., PEHLİVANTÜRK KIZILKAN M., KANBUR N., DERMAN O., TÜZÜN GÜN Z., ÜTİNE G. E. , ŞİMŞEK KİPER P. Ö. , AKALIN A., AKGÜL S.  
JOURNAL OF ADOLESCENT HEALTH, vol.70, no.4, 2022 (Journal Indexed in SCI)
- VIII. **A very rare case of a newborn with tetrasomy 9p and literature review**  
SÜLEYMAN M., OĞUZ S., KAYKI G., ÇELİK H. T. , ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E. , YİĞİT Ş.  
TURKISH JOURNAL OF PEDIATRICS, vol.64, no.1, pp.171-178, 2022 (Journal Indexed in SCI)
- IX. **SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype**  
Motta M., Fasano G., Gredy S., Brinkmann J., Bonnard A. A. , ŞİMŞEK KİPER P. Ö. , Gulec E. Y. , Essaddam L., ÜTİNE G. E. , Prandi I. G. , et al.  
AMERICAN JOURNAL OF HUMAN GENETICS, vol.108, no.11, pp.2112-2129, 2021 (Journal Indexed in SCI)
- X. **Sleep disordered breathing in patients with Prader willi syndrome: Impact of underlying genetic mechanism**  
Ozsezen B., EMİRALİOĞLU N., Ozon A., Akin O., Tural D. A. , SUNMAN B., Hejiyeva A., Hizal M., Alikasifoglu A., ŞİMŞEK KİPER P. Ö. , et al.  
RESPIRATORY MEDICINE, vol.187, 2021 (Journal Indexed in SCI)
- XI. **Obstructive sleep apnea in children with Down syndrome: is it possible to predict severe apnea?**  
Hizal M., ŞATIRER Ö., Polat S. E. , Tural D. A. , Ozsezen B., SUNMAN B., KARAHAN S., EMİRALİOĞLU N., ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E. , et al.  
EUROPEAN JOURNAL OF PEDIATRICS, 2021 (Journal Indexed in SCI)
- XII. **Main Physical Features, Echocardiographic and Renal Ultrasonographic Findings of Turner Syndrome in 107 Pediatric Patients**  
AKALIN A., ERTUĞRUL İ., ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E. , BODUROĞLU O. K.  
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- XIII. **Biallelic ITGB4 variants in familial pyloric atresia without epidermolysis bullosa: Report of two families with five siblings**  
SOYER T., KARAOSMANOĞLU B., TAŞKIRAN Z. E. , ŞİMŞEK KİPER P. Ö. , KARNAK İ., BODUROĞLU O. K. , ÜTİNE G. E.  
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- XIV. **Three new cases of Crisponi /cold induced sweating syndrome (CS/CISS1) in Turkish families**  
Kolkiran A., ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E.  
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- XV. **Refractory temporal lobe epilepsy in patients with mosaic turner syndrome: two case reports and literature review**  
ARSLAN D., Utine E., SAYGI S.  
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- XVI. **Spondyloepimetaphyseal dysplasia EXTL3-deficient type: Long-term follow-up and review of the literature**  
AKALIN A., TAŞKIRAN Z. E. , ŞİMŞEK KİPER P. Ö. , Utine E., ALANAY Y., Ozcelik U., BODUROĞLU O. K.  
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- XVII. **Further expanding the mutational spectrum of brain abnormalities, neurodegeneration, and dysosteosclerosis: A rare disorder with neurologic regression and skeletal features**  
KINDİŞ E., ŞİMŞEK KİPER P. Ö. , KOŞUKCU C., TAŞKIRAN Z. E. , GÖÇMEN R., Utine E., Haliloglu G., BODUROĞLU O. K. ,

- ALİKAŞİFOĞLU M.  
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- XVIII. **Natural history of TRPV4-Related disorders: From skeletal dysplasia to neuromuscular phenotype**  
ÜREL DEMİR G., ŞİMŞEK KİPER P. Ö. , Oncel I., ÜTİNE G. E. , Haliloglu G., BODUROĞLU O. K.  
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, vol.32, pp.46-55, 2021 (Journal Indexed in SCI)
- XIX. **Kohlschutter-Tonz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity**  
AKGÜN DOĞAN Ö., ŞİMŞEK KİPER P. Ö. , Taskiran E., Schossig A., ÜTİNE G. E. , Zschocke J., BODUROĞLU O. K.  
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- XX. **Genetic disorders with symptoms mimicking rheumatologic diseases: A single-center retrospective study**  
KAYA AKCA Ü., ŞİMŞEK KİPER P. Ö. , ÜREL DEMİR G., SAĞ E., ATALAY E., ÜTİNE G. E. , ALİKAŞİFOĞLU M.,  
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- XXI. **More than meets the eye: Expanding and reviewing the clinical and mutational spectrum of brittle cornea syndrome**  
Dhooge T., Van Damme T., Syx D., Mosquera L. M. , Nampoothiri S., Radhakrishnan A., Simsek-Kiper P. O. , ÜTİNE G. E. , Bonduelle M., Migeotte I., et al.  
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- XXII. **Two Siblings with Kaufman Oculocerebrofacial Syndrome Resembling Oculoauriculovertebral Spectrum**  
ÜREL DEMİR G., Aydin B., KARAOSMANOĞLU B., AKGÜN DOĞAN Ö., Taskiran E. Z. , ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E. ,  
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Xue J., Simsek-Kiper P. O. , ÜTİNE G. E. , Yan L., Wang Z., Taskiran E. Z. , KARAOSMANOĞLU B., İMREN G., GÖÇMEN R.,  
Nishimura G., et al.  
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- XXIV. **A Revisited Diagnosis of Collagen VI Related Muscular Dystrophy in a Patient with a Novel COL6A2 Variant and 21q22.3 Deletion**  
ŞİMŞEK KİPER P. Ö. , OĞUZ S., ERGEN F. B. , ÜTİNE G. E. , ALİKAŞİFOĞLU M., Haliloglu G.  
NEUROPEDIATRICS, vol.51, no.6, pp.445-449, 2020 (Journal Indexed in SCI)
- XXV. **Molecular etiology of isolated congenital cataract using next-generation sequencing: Single center exome sequencing data from Turkey**  
Taylan Şekeroğlu H., Karaosmanoğlu B., Taşkıran E. Z. , Şimşek Kiper P. Ö. , Alikasıfoğlu M., Boduroğlu O. K. , Coşkun T., Ütine G. E.  
Molecular Syndromology, vol.11, pp.302-308, 2020 (Journal Indexed in SCI)
- XXVI. **Clinical and Molecular Spectrum of Four Patients Diagnosed with Mowat-Wilson Syndrome**  
AYYILDIZ EMECEN D., IŞIK E., ÜTİNE G. E. , Simsek-Kiper P. O. , ATİK T., Ozkinay F.  
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- XXVII. **A rare cause of syndromic short stature: 3M syndrome in three families**  
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- XXVIII. **The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered**  
Brinkmann J., Lissewski C., Pinna V., Vial Y., Pantaleoni F., Lepri F., Daniele P., Burnyete B., Cuturilo G., Fauth C., et al.  
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- XXIX. **Poikiloderma with Neutropenia, Clericuzio-Type Accompanied by Loss of Digits Due to Severe Osteomyelitis**  
AKDOĞAN N., KINDİŞ E., BOSTAN E., Utine E., ALİKAŞİFOĞLU M., ERSOY EVANS S.  
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- XXX. **A novel mutation of keratin 5 in epidermolysis bullosa simplex with migratory circinate erythema**

- YALICI ARMAĞAN B., Kabacam S., TAŞKIRAN Z. E. , GÖKÖZ Ö. , ÜTİNE G. E. , ERSOY EVANS S.  
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- XXXI. **Crisponi/cold-induced sweating syndrome: Differential diagnosis, pathogenesis and treatment concepts**  
Buers I, Persico I, Schoening L, Nitschke Y, Di Rocco M, Loi A, Sahi P. K. , ÜTİNE G. E. , Bayraktar-Tanyeri B, Zampino G., et al.  
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- XXXII. **Peters Plus syndrome: a recognizable clinical entity**  
ÜREL DEMİR G., Lafci N., Dogan O. A. , ŞİMŞEK KİPER P. Ö. , ÜTİNE G. E.  
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- XXXIII. **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 (Journal Indexed in SCI)
- XXXIV. **Spondyloocular syndrome: Presentation of two siblings diagnosed with the rare disease and the results of Pamidronate Therapy**  
VURALI KARAOĞLAN D., ŞİMŞEK KİPER P. Ö. , Utine E., ÜNSAL Y., ALİKAŞİFOĞLU A., KANDEMİR N.  
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.387, 2019 (Journal Indexed in SCI)
- XXXV. **A case of Woodhouse-Sakati syndrome with pituitary iron deposition, cardiac and intestinal anomalies, with a novel mutation in DCAF17**  
ŞENDUR S. N. , OĞUZ S., ÜTİNE G. E. , DAĞDELEN S., Oguz K. K. , Erbas T., ALİKAŞİFOĞLU M.  
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.62, no.8, 2019 (Journal Indexed in SCI)
- XXXVI. **Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome**  
Simsek-Kiper P. O. , Taskiran E., KOŞUKCU C., ARSLAN U. E. , Cormier-Daire V., Gonc N., Ozon A., ALİKAŞİFOĞLU A., KANDEMİR N., ÜTİNE G. E. , et al.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.179, no.7, pp.1157-1172, 2019 (Journal Indexed in SCI)
- XXXVII. **Ophtho-acromelic syndrome in an infant**  
ÜREL DEMİR G., Taskiran E. Z. , AKGÜN DOĞAN Ö., Simek-Kiper P. O. , ÜTİNE G. E.  
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- XXXVIII. **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome**  
van der Sluijs P. J. , Jansen S., Vergano S. A. , Adachi-Fukuda M., ALANAY Y., AlKindy A., Baban A., Bayat A., Beck-Woedl S., Berry K., et al.  
GENETICS IN MEDICINE, vol.21, no.6, pp.1295-1307, 2019 (Journal Indexed in SCI)
- XXXIX. **An eight-case 1q21 region series: novel aberrations and clinical variability with new features**  
CEYLAN A., ŞAHİN İ. F. , Erdem H., Kayhan G., Simsek-Kiper P., ÜTİNE G. E. , Percin F., Boduroglu K., ALİKAŞİFOĞLU M.  
Journal of Intellectual Disability Research, vol.63, no.6, pp.548-557, 2019 (Journal Indexed in SSCI)
- XL. **A novel NKX3-2 mutation associated with perinatal lethal phenotype of spondylo-megaepiphyseal-metaphyseal dysplasia in a neonate**  
Simsek-Kiper P. O. , KOŞUKCU C., Akgun-Dogan O., GÖÇMEN R., ÜTİNE G. E. , SOYER T., Korkmaz-Toygar A., Nishimura G., ALİKAŞİFOĞLU M., Boduroglu K.  
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- XLI. **Non-immune hydrops fetalis: A retrospective analysis of 151 autopsies performed at a single center**  
KAYKI G., Gucer S., AKÇÖREN Z., ORHAN D., TALİM B., YURDAKÖK M., YİĞİT Ş., BODUROĞLU O. K. , ÜTİNE G. E. , Orgul G., et al.  
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- XLII. **Prenatal and Postnatal Follow-up in Trisomies 13 and 18: A 20-Year Experience in a Tertiary Center**  
Dogan O. A. , Demir G. U. , Arslan U. E. , Simsek-Kiper P. O. , ÜTİNE G. E. , ALİKAŞİFOĞLU M., Boduroglu K.  
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- XLIII. **Fragile x-associated premature ovarian failure in a large Turkish cohort: Findings of Hacettepe**

## **Fragile X Registry**

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- XLIV. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Costello Syndrome**  
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- XLV. **Anauxetic dysplasia: A rare clinical entity**  
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- XLVI. **Clinical and molecular evaluation of 16 patients with rett syndrome**  
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- XLVII. **Epigenotype and phenotype correlations in patients with Beckwith-Wiedemann syndrome**  
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- XLVIII. **Coexistence of Trisomy 13 and SRY (–) XX Ovotesticular Disorder of Sex Development**  
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- XLIX. **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.  
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- L. **HERC1 mutations in idiopathic intellectual disability**  
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- LI. **A novel TRAPPC11 mutation in two Turkish families associated with cerebral atrophy, global retardation, scoliosis, achalasia and alacrima**  
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- LII. **Polyposis deserves a perfect physical examination for final diagnosis: Bannayan-Riley-Ruvalcaba syndrome**  
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- LIII. **A Turkish BCS1L mutation causes GRACILE-like disorder**  
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- LIV. **Congenital multisegmental lymphatic dysplasia with systemic involvement: a case report**  
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- LV. **Congenital Mirror Movements in Gorlin Syndrome: A Case Report With DTI and Functional MRI Features**  
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- LVI. **A Diagnosis to Consider in Intellectual Disability: Mowat-Wilson Syndrome**  
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- LVII. **Experience of a skeletal dysplasia registry in Turkey: A five-years retrospective analysis**

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- LVIII. **Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffin–Siris and Nicolaides–Baraitser syndromes**  
Bramswig N. C. , Luedecke H., Alanay Y., Albrecht B., Barthelmie A., Boduroglu K., Braunholz D., Caliebe A., Chrzanowska K. H. , Czeschik J. C. , et al.  
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- LIX. **Two patients with microdeletion and microduplication involving 1q21.1**  
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- LX. **A novel mutation in RNU4ATAC in a patient with microcephalic osteodysplastic primordial dwarfism type I**  
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- LXI. **Partial distal aphyalangia, duplication of metatarsal IV, microcephaly, and borderline intelligence: A fourth patient with parental consanguinity and additional feature of massive cerebral thrombosis**  
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- LXII. **Novel homozygous mutations in the osteoprotegerin gene TNFRSF11B in two unrelated patients with juvenile Paget's disease**  
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