

## Expert PhD Tuğba KALAYCI

### Personal Information

**Email:** tugba.kalayci@istanbul.edu.tr

**Web:** <https://avesis.istanbul.edu.tr/tugba.kalayci>

### International Researcher IDs

ORCID: 0000-0002-9963-5916

Yoksis Researcher ID: 194878

### Education Information

Doctorate, Istanbul University, Health Sciences Institute, Aziz Sancar Deneysel Tıp Araştırma Enstitüsü Bölümü, Turkey  
2019 - Continues

Expertise In Medicine, Istanbul University, Istanbul Medical Faculty, Tıbbi Genetik Anabilim Dalı, Turkey 2013 - 2017

Undergraduate, Selcuk University, School Of Medicine, Turkey 2006 - 2012

### Foreign Languages

English, C2 Mastery

### Dissertations

Expertise In Medicine, FETAL İSKELET DİSPLAZİLERİNİN KLİNİK, RADYOLOJİK BULGULAR ve MOLEKÜLER ETİYOPATOGENEZE GÖRE SINIFLANDIRILMASI, Istanbul University, Istanbul Medical Faculty, Tıbbi Genetik Anabilim Dalı, 2016

### Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Medical Genetics

### Academic Titles / Tasks

Student, Istanbul University, Health Sciences Institute, Aziz Sancar Deneysel Tıp Araştırma Enstitüsü Bölümü, 2019 - Continues

Expert PhD, Istanbul University, Istanbul Medical Faculty, Division of Medical Sciences , 2018 - Continues

Research Assistant PhD, Istanbul University, Istanbul Medical Faculty, Division of Medical Sciences , 2013 - 2017

### Courses

Prenatal ve Preimplantasyon Genetik Tanı ve Danışmanlığı, Postgraduate, 2021 - 2022

İskelet Sistemi Malformasyonları, Undergraduate, 2022 - 2023, 2021 - 2022, 2020 - 2021, 2019 - 2020, 2018 - 2019

Malformations of Human Skeletal System, Undergraduate, 2022 - 2023, 2021 - 2022, 2020 - 2021, 2019 - 2020

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

- I. **Fetal skeletal dysplasia cohort of a single tertiary referral center in Istanbul, Turkey.**  
Kalaycı T., Altunoglu U., Çorbacıoğlu Esmir A., Avcı Ş., Sarac Sivrikoz T., Karaman B., Kalelioğlu İ., Has R., Uyguner Z. O., Yüksel A., et al.  
American journal of medical genetics. Part A, vol.191, no.2, pp.498-509, 2023 (SCI-Expanded)
- II. **A new enrichment approach for candidate gene detection in unexplained recurrent pregnancy loss and implantation failure**  
Berkay E. G., Şoroğlu C. V., Kalaycı T., Uyguner Z. O., Akçapınar G. B., Başaran S.  
MOLECULAR GENETICS AND GENOMICS, vol.298, no.1, pp.253-272, 2023 (SCI-Expanded)
- III. **Clinical, Cytogenetic and Molecular Cytogenetic Outcomes of Cell-Free DNA Testing for Rare Chromosomal Anomalies**  
Başaran S., Has R., Kalelioğlu İ. H., Saraç Sivrikoz T., Karaman B., Kırgız M., Dehgan T., Kalaycı T., Selçuk B. Ş., Mıny P., et al.  
GENES, vol.13, no.12, pp.1-11, 2022 (SCI-Expanded)
- IV. **Functional characterization of a novel TP53RK mutation identified in a family with Galloway-Mowat syndrome**  
Treimer E., Kalaycı T., Schumann S., Suer I., Greco S., Schanze D., Schmeisser M. J., Kuhl S. J., Zenker M.  
HUMAN MUTATION, vol.43, no.12, pp.1866-1871, 2022 (SCI-Expanded)
- V. **Cytogenetic and molecular characterization of a patient having infertility and mild intellectual disability with a very rare unstable ring chromosome 13**  
Kaya M., Suer İ., Kalaycı T., Karaman B., Ozturk Ş., Palanduz S.  
SCOTTISH MEDICAL JOURNAL, vol.67, no.4, pp.173-177, 2022 (SCI-Expanded)
- VI. **Prenatal ultrasonographic features in Blomstrand osteochondrodysplasia: Antenatal case series confirmed by postmortem radiology and molecular diagnosis**  
Saraç Sivrikoz T., Kalaycı T., Senturk L., Karaman V., Kalelioğlu İ. H., Has R., Kayserili H., Uyguner Z. O., Nishimura G., Altunoglu U.  
PRENATAL DIAGNOSIS, vol.42, no.12, pp.1503-1510, 2022 (SCI-Expanded)
- VII. **Mutations in SCN11A cause orofaciodigital syndrome due to minor intron splicing defects affecting primary cilia**  
Iturrate A., Rivera-Barahona A., Flores C., Otaify G. A., Elhossini R., Perez-Sanz M. L., Nevado J., Tenorio-Castano J., Triviño J. C., Garcia-Gonzalo F. R., et al.  
American Journal of Human Genetics, vol.109, no.10, pp.1828-1849, 2022 (SCI-Expanded)
- VIII. **Clinical and molecular genetic findings of Crisponi/cold-induced sweating syndrome (CS/CISS) spectrum in patients from Turkey.**  
Yılmaz Gulec E., Turgut G. T., Gezdirici A., Karaman V., Ozturk F. N., Avcı S., Kalaycı T., Senturk L., Ayaz A., Kayserili H., et al.  
Clinical genetics, vol.102, no.3, pp.201-217, 2022 (SCI-Expanded)
- IX. **Distal renal tubular acidosis, autoimmune thyroiditis, enamel hypomaturation, and tooth agenesis caused by homozygosity of a novel double-nucleotide substitution in SLC4A4**  
Kantaputra P., Güven Y., Aksu B., Kalaycı T., Dogan C., Intachai W., Olsen B., Tongsimma S., Ngamphiw C., Noppakun K.  
Journal of the American Dental Association, vol.153, no.7, pp.668-676, 2022 (SCI-Expanded)
- X. **Expanding genotypic and phenotypic spectrums of LTBP3 variants in dental anomalies and short stature syndrome.**  
Kantaputra P., Guven Y., Kalaycı T., Özer P., Panyarak W., Intachai W., Olsen B., Carlson B. M., Praditsap O., Tongsimma S., et al.  
Clinical genetics, vol.102, no.1, pp.66-71, 2022 (SCI-Expanded)

- XI. **Functional loss of ubiquitin-specific protease 14 may lead to a novel distal arthrogyrosis phenotype.**  
Turgut G. T., Altunoglu U., Sivriköz T. S., Toksoy G., Kalayci T., Avci S., Karaman B., Gulec C., Basaran S., Sayin G., et al.  
Clinical genetics, vol.101, no.4, pp.421-428, 2022 (SCI-Expanded)
- XII. **A mysterious cause of recurrent acute liver dysfunction for over a decade**  
Dirim A. B., Kalayci T., Guzel Dirim M., Demir S., Cavus B., Cifcibasi Ormeci A., Akyuz F., Kaymakoglu S.  
GASTROENTEROLOGY REPORT, vol.10, 2022 (SCI-Expanded)
- XIII. **Clinical Characteristics and Mutation Spectrum of Neurofibromatosis Type 1 in 27 Turkish Families**  
Sharifi S., Kalayci T., Palanduz S., Ozturk S., Cefle K.  
BALKAN MEDICAL JOURNAL, vol.38, no.6, pp.365-373, 2021 (SCI-Expanded)
- XIV. **Re: Indication for Y Chromosome Microdeletion Analysis in Infertile Men: Is a New Sperm Concentration Threshold Needed?**  
Ortac M., Ergul R., Gurcan M., Kalayci T., Palanduz S., Aydin R., Kadioglu A.  
JOURNAL OF UROLOGY, vol.206, no.4, pp.1050, 2021 (SCI-Expanded)
- XV. **Skeletal and molecular findings in 51 Cleidocranial dysplasia patients from Turkey**  
Berkay E. G., Elkanova L., Kalayci T., ULUDAĞ ALKAYA D., Altunoglu U., Cefle K., Mihci E., NUR B., Tasdelen E., Bayramoglu Z., et al.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.8, pp.2488-2495, 2021 (SCI-Expanded)
- XVI. **Indication for Y Chromosome Microdeletion Analysis in Infertile Men: Is a New Sperm Concentration Threshold Needed?**  
Ortac M., Ergul R., Gurcan M., Kalayci T., Palanduz S., Aydin R., Kadioğlu A.  
UROLOGY, vol.146, pp.113-117, 2020 (SCI-Expanded)
- XVII. **The first case of Dyssegmental Dysplasia Rolland-Desbuquois type with a variant in HSPG2**  
KALAYCI T., Balanda N., Ferreira C. R., Altunoglu U.  
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, no.SUPPL 1, pp.237, 2020 (SCI-Expanded)
- XVIII. **Delineation of phenotypes and genotypes related to cohesin structural protein RAD21.**  
Krab L., Marcos-Alcalde I., Assaf M., Balasubramanian M., Andersen J., Bisgaard A., Fitzpatrick D., Gudmundsson S., Huisman S., Kalayci T., et al.  
Human genetics, vol.139, pp.575-592, 2020 (SCI-Expanded)
- XIX. **Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay.**  
Lee S., Chen D., Zaki M., Maroofian R., Houlden H., Di D., Abdin D., Morsy H., Mirzaa G., Dobyns W., et al.  
American journal of human genetics, vol.105, pp.844-853, 2019 (SCI-Expanded)
- XX. **Heterogeneous clinical phenotypes and cerebral malformations reflected by rotatin cellular dynamics.**  
Vandervore L., Schot R., Kasteleijn E., Oegema R., Stouffs K., Gheldof A., Grochowska M., van d., van U., Wilke M., et al.  
Brain : a journal of neurology, vol.142, pp.867-884, 2019 (SCI-Expanded)

## Articles Published in Other Journals

- I. **Fetal Hand Anomalies: 18 Cases Diagnosed Between 2020-2022 from a Single Tertiary Care Center**  
ASLANGER A. D., SARAÇ SİVRİKOZ T., KALAYCI T., BAŞARAN S., UYGUNER Z. O.  
Experimed, vol.12, no.3, pp.149-154, 2022 (Peer-Reviewed Journal)
- II. **CLINICAL AND MOLECULAR RESULTS OF SIX CASES WITH ROBERTS SYNDROME: REVIEW OF CASES FROM TURKIYE**  
ASLANGER A. D., KALAYCI T., KONUR E. N., GÜLEÇ Ç., AVCI Ş., ALTUNOĞLU U., KARAMAN V., TOKSOY G., KARAMAN B., BAŞARAN S., et al.  
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESİ DERGISI, vol.85, no.4, pp.501-510, 2022 (Scopus)

- III. **Association between HBA locus copy number gains and pathogenic HBB gene variants**  
Toksoy G., Akay N., Aghayev A., Karaman V., Avcı Ş., Kalaycı T., Altunoğlu U., Karakaş Z., Uyguner Z. O.  
INTERNATIONAL JOURNAL OF MEDICAL BIOCHEMISTRY, vol.4, no.2, pp.91-95, 2021 (Peer-Reviewed Journal)

## Refereed Congress / Symposium Publications in Proceedings

- I. **Frank-Ter Haar Sendromu Tanılı 3 Olgu ve Literatür Derlemesi**  
Konur E. N., Aslanger A. D., Kalaycı T., Altunoğlu U., Karaman V., Yeşil Sayın G., Kayserili Karabay H., Uyguner Z. O.  
15. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 09 November 2022, pp.91
- II. **Osteogenezis Imperfekta Tanılı 15 Olgunun Moleküler Sonuçları**  
Hacer Ö., Aslanger A. D., Kalaycı T., Güleç Ç., Demir K., Toksoy G., Karaman V., Öztürk A. P., Baş F., Yeşil Sayın G., et al.  
15. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 09 November 2022, pp.149
- III. **Epilepsinin Nadir, Nonkonvulzif Status Epileptikusun Nadir Olmayan Bir Sebebi: Ring 20 Kromozomu**  
Gezegen H., İlgezdi Kaya İ., Kalaycı T., Şirin İnanc N. G., Karaman B., Bebek N., Baykan B.  
58. ulusal Nöroloji Kongresi, İstanbul, Turkey, 19 - 24 November 2022, pp.650
- IV. **Prenatal Diagnosis Of 1P36 Deletion Syndrome Due To Pericentric Inversion On Chromosome 1: A Case Report**  
Yıldırım B. T., Kalaycı T., Bulut G., Saraç Sivriköz T., Selçuk B. Ş., Başaran S., Karaman B.  
15.ULUSLARARASI KATILIMLI, ULUSAL TIBBİ GENETİK KONGRESİ, Muğla, Turkey, 9 - 13 November 2022, pp.25
- V. **Fetal Dönemde Kontraktürler İle Seyreden Fenotiplerin Moleküler Tanısında Tüm Ekzom Dizileme Analizinin Katkısı**  
Turgut G. T., Altunoğlu U., Güleç Ç., Kalaycı T., Saraç Sivriköz T., Toksoy G., Karaman B., Yeşil Sayın G., Kayserili Karabay H., Uyguner Z. O.  
15.ULUSLARARASI KATILIMLI, ULUSAL TIBBİ GENETİK KONGRESİ, Muğla, Turkey, 9 - 13 November 2022, pp.85
- VI. **Trans Erkek; Gebelik Terminasyonu**  
Cenger C. D., Saraç Sivriköz T., Polat I., Kalaycı T., Şenbaş Z. A., Arıcan N.  
2.Uluslararası ve 18. Ulusal Adli Bilimler Kongresi, 14 - 17 October 2021, pp.414-416
- VII. **Sitokrom P450 oksidoredüktaz eksikliğine bağlı konjenital adrenal hiperplazi hastalarının klinik ve laboratuvar özellikleri: Olgu serisi**  
Bayrak Demirel Ö., Baş F., Kalaycı T., Yıldız M., Konur E. N., Poyrazoğlu Ş., Yeşil Sayın G., Darendeliler F. F.  
25. Ulusal Pediatrik Endokrinoloji Kongresi, Antalya, Turkey, 6 - 10 October 2021, pp.305-307
- VIII. **Three Nance Horan Syndrome Families from Turkey; Three Different Approaches for Molecular Diagnosis**  
Saraçoğlu H. P., Güven Y., Aksakal S. D., Kalaycı T., Altunoğlu U., Uyguner Z. O., Eraslan S., Börklü E., Kayserili Karabay H.  
European Human Genetics Conference, 28 - 31 August 2021, pp.1
- IX. **MECP2 Spektrumundan Etkilenmiş 27 Olgunun Klinik ve Moleküler Bulguları**  
Kalaycı T., Aslanger A. D., Altunoğlu U., Toksoy G., Konur E. N., Avcı Ş., Karaman V., Karaman B., Yeşil Sayın G., Kayserili Karabay H., et al.  
14. TIBBİ GENETİK KONGRESİ, 20 - 22 December 2020, vol.31, no.4, pp.53
- X. **Nadir Hastalıkların Tanı ve Takibinde Biyokimyasal Testlerin Tamamlayıcı Rolü: Olgu Sunumu**  
Gedikbaşı A., Toksoy G., Kalaycı T., Gelmez M. Y., Karaman B., Deniz G., Uyguner Z. O.  
Uluslararası Laboratuvar Tıbbı ve XX.Ulusal Klinik Biyokimya Kongresi, İstanbul, Turkey, 25 - 26 December 2020, pp.1
- XI. **Alport sendromlu 15 olgunun klinik ve moleküler bulguları**  
Aslanger A. D., Yürük Yıldırım Z. N., Toksoy G., Aksu B., Durmaz D., Göksu Çetinkaya A. P., Kalaycı T., Çam Delebe E. Ö., Karaman V., Yavuz S., et al.  
14. TIBBİ GENETİK KONGRESİ, İstanbul, Turkey, 20 - 22 December 2020, vol.31, no.4, pp.49
- XII. **Steroide duyarlı kronik anemisi ve osteosklerozu olan erişkin olguda moleküler tanının klinik izleme etkisi**

- Sharifi S., Kalaycı T., Kaya M., Suer İ., Öztürk Ş., Çefle K., Yenerel M. N., Palanduz Ş.  
14. Ulusal Tıbbi Genetik Kongresi (Uluslararası katılımlı), İstanbul, Turkey, 20 - 23 November 2020, pp.37
- XIII. **46,XX,t(8;9)(q12;q12) translokasyon taşıyıcısı tekrarlayan gebelik kayıp öykülü olgu sunumu**  
Suer İ., Kaya M., Sharifi S., Kalaycı T., Öztürk Ş., Çefle K., Palanduz Ş.  
14. Ulusal Tıbbi Genetik Kongresi (Uluslararası katılımlı), İstanbul, Turkey, 20 November - 22 December 2020, pp.89
- XIV. **Fetal skeletal dysplasia experience of a single tertiary referral center in Istanbul, Turkey**  
Kalaycı T., Altunoğlu U., Saraç Sivriköz T., Çorbacıoğlu A., Avcı Ş., Has R., Uyguner Z. O., Yüksel A., Başaran S., Kayserili H.  
14th International Skeletal Dysplasia Society Meeting, Oslo, Norway, 11 - 14 September 2019, pp.35
- XV. **LAMB2 Gen Mutasyonu Saptanan Dört Hasta Ve Dört Farklı Klinik Durum**  
Öner H. A., Toksoy G., Yürük Yıldırım Z. N., Yılmaz A., Çam Delebe E. Ö., Göksu Çetinkaya A. P., Tğrkkan Ö. N., Kalaycı T., Nayır A. N.  
Uluslararası Katılımlı 10. Çocuk Nefroloji Kongresi, Muğla, Turkey, 1 - 04 May 2019, pp.158
- XVI. **Clinical and Molecular Characterization of Stuve-Wiedmann Syndrome in Six Cases**  
Şentürk L., Güleç Ç., Kayserili Karabay H., Kalaycı T., Uyguner Z. O., Altunoğlu U.  
13. Uluslararası Katılımlı Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 7 - 11 November 2018, pp.123
- XVII. **Clinical and molecular findings of seven Turkish nonphotosensitive trichothiodystrophy patients with two novel mutations in MPLKIP**  
Kalaycı T., Altunoğlu U., Karaman B., Uyguner Z. O., Kayserili Karabay H.  
50th European Society of Human Genetics Conference, Kobenhavn, Denmark, 27 - 30 May 2017, pp.394
- XVIII. **Whole exome sequencing in fetal structural abnormalities: experience of 8 cases**  
Altunoglu U., Kalayci T., Kalelioglu I. H., Kayserili H.  
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.166-167
- XIX. **Clinical and molecular findings of seven Turkish non-photosensitive trichothiodystrophy patients with two novel mutations in MPLKIP**  
Kalayci T., Altunoglu U., Karaman B., Uyguner Z., Kayserili H.  
50th European-Society-of-Human-Genetics (ESHG) Conference, Copenhagen, Denmark, 27 - 30 May 2017, vol.26, pp.506
- XX. **idic(Y)(q11.2) ABNORMALITY IN CASES WITH MIXT GONADAL DYSGENESIS AND INFERTILITY**  
Kaya M., Suer İ., Kalaycı T., Karaman B., Dön B., Bağatır Ozan G., Uçur A., Öztan G., Bayrak A. G., Çefle K., et al.  
Erciyes Medical Genetics Days, Kayseri, Turkey, 7 - 10 March 2018, pp.16
- XXI. **New Fetal Case of Blomstrand Chondrodysplasia and Review of the Literature**  
Kalaycı T., Altunoğlu U., Şentürk L., Uyguner Z. O., Kayserili Karabay H.  
12th International Skeletal Dysplasia Society Meeting, İstanbul, Turkey, 11 - 13 September 2015

## Supported Projects

KALAYCI T., ÖZTÜRK Ş., PALANDUZ Ş., ÇEFLE K., SHARIFI S., Project Supported by Higher Education Institutions, FMR1 ve Karyotip Analizi Normal Sonuçlanan Prematür Over Yetmezliği POF Olgularında Moleküler Etyopatogenezin Araştırılması, 2019 - 2021

BAŞARAN S., BERKAY E. G., KARAMAN B., UYGUNER Z. O., KALAYCI T., Project Supported by Higher Education Institutions, Sebabi Açıklanamayan İnfertilite ile İlişkili Genlerin Araştırılması, 2018 - 2021

ALTUNOĞLU U., KALELİOĞLU İ. H., KALAYCI T., KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, Whole exome sequencing in fetal structural abnormalities: experience of 8 cases, 2017 - 2018

BAŞARAN S., KALAYCI T., Project Supported by Higher Education Institutions, FETAL İSKELET DİSPLAZİLERİNİN KLİNİK RADYOLOJİK BULGULAR ve MOLEKÜLER ETİYOPATOGENEZE GÖRE SINIFLANDIRILMASI, 2017 - 2017

## **Metrics**

Publication: 44

Citation (WoS): 53

Citation (Scopus): 52

H-Index (WoS): 4

H-Index (Scopus): 4

## **Non Academic Experience**

University, Erasmus Medical Center, Clinical Genetics