

İbrahim Şahin

MD, EBMGG

Associate Professor of Medical Genetics · Consultant Clinical Geneticist

Department of Medical Genetics, Ankara Etlik City Hospital,
University of Health Sciences, Ankara, Türkiye

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PROFESSIONAL SUMMARY

European Board-certified clinical genetics consultant, associate professor and laboratory director with more than thirteen years of integrated clinical, academic and molecular diagnostic experience. Established and managed four genetic diagnostic laboratories, including Bahrain's first molecular genetics next-generation sequencing laboratory (CAP accredited). Experienced in cancer genetics (germline and somatic), human genetics and rare diseases, pharmacogenomics, and next-generation sequencing and bioinformatics.

Author of more than forty publications, including work in *The American Journal of Human Genetics*, *PLOS ONE* and *American Journal of Medical Genetics*; supervisor of three or more postgraduate students; and designer of clinical genetics and genomics curricula at MD, MSc, PhD and residency levels.

RESEARCH INTERESTS

Hereditary cancer · Cancer genetics · Human genetics · Rare diseases · Pharmacogenomics · Precision medicine · Systems biology

CORE COMPETENCIES

Clinical genetics

Variant classification (ACMG), genotype–phenotype analysis, reporting, metabolic and rare disease evaluation, prenatal genetics and cancer genetics.

Molecular and biochemical genetics laboratory leadership

NGS workflows, WES/WGS/panel/CGP analysis including CNV and mtDNA, biochemical metabolic assays, SOP creation, quality-control management, and accreditation readiness (CAP, ISO, national).

Academic and educational leadership

Curriculum design, course leadership, student supervision, assessment development and accreditation documentation.

EDUCATION

2012–2017 Medical Genetics Residency Programme
Department of Medical Genetics, Atatürk University, School of Medicine

2004–2011 Doctor of Medicine (MD)
Dokuz Eylül University, School of Medicine

ACADEMIC AND PROFESSIONAL APPOINTMENTS

2026–present Associate Professor, Department of Medical Genetics
Ankara Etlik City Hospital, University of Health Sciences, Ankara, Türkiye

2021–2026 Head of Molecular and Biochemical Genetics Laboratories; Assistant Professor; Consultant Clinical Geneticist
Princess Al-Jawhara Center for Molecular Medicine, Genetics and Inherited Disorders, College of Medicine & Medical Science, Arabian Gulf University, Bahrain

2023–2024	Head of Bahrain Oncology Centre Laboratory; Consultant Clinical Geneticist King Hamad University Hospital, Bahrain
2019–2021	Associate Director, Tissue Typing (HLA) Laboratory University of Health Sciences, Dışkapı Yıldırım Beyazıt Training and Research Hospital, Türkiye
2017–2022	Medical Geneticist University of Health Sciences, Dışkapı Yıldırım Beyazıt Training and Research Hospital, Department of Medical Genetics, Diagnostic Center for Genetic Diseases, Türkiye
2017–2019	Visiting Scientist, Cancer Systems Biology Bilkent University, Department of Molecular Biology and Genetics, Türkiye
2012–2017	Medical Geneticist Department of Medical Genetics, Diagnostic Center for Genetic Diseases, Atatürk University, School of Medicine

LICENSURE AND CERTIFICATION

06/2023	European Board of Medical Genetics (EBMGG)
11/2022	Associate Professor in Medical Genetics, Ministry of Health, Türkiye
03/2017	Medical Geneticist Diploma, Atatürk University Medical School
07/2011	Medical School Diploma, Dokuz Eylül University Medical School

PROFESSIONAL SOCIETY MEMBERSHIPS

2012–present	Turkish Society of Medical Genetics
2021–present	European Society for Medical Oncology (ESMO)
2021–present	International Society of Liquid Biopsy (ISLB)
2015–present	European Society of Human Genetics (ESHG)
2014–2020	American Association for Cancer Research (AACR)
2014–2020	European Association for Cancer Research (EACR)

LANGUAGES

Turkish (native) · English (fluent) · Arabic (intermediate) · German (elementary) · French (elementary)

TECHNICAL SKILLS

Bioinformatics; programming in Python and R; machine learning.

BOOK CHAPTERS

- Şahin İ. Hereditary cancer research and technological advances: contributions of CRISPR, artificial intelligence and advanced laboratory methods to understanding and treating hereditary cancers. In: *Hereditary Cancers*, 2026. ISBN 978-625-92736-0-0.
- Şahin İ. Liquid biopsy (non-invasive cancer genotyping, plasma genotyping). In: *Cancer in All Aspects*, 2020. ISBN 978-605-7902-34-4.

PUBLICATIONS

1. Al-Jamea LH, Woodman A, Al-Ansari RY, Alayidh FS, Alharbi KK, Al Ahmadi NS, Al-Jawair R, Sahin I, AlGhamdi SS, Al Johar EJ, Quiambao JV, Deifalla AH, Hawsawi YM. Association between leptin gene polymorphisms and hematological metabolic parameters in obese Saudi adults. *Frontiers in Medicine*, 2026.
2. Aygün H, Şahin İ, Taşkın A, Ceylan AC, Karaali K, Yayıcı Köken Ö. Expanding the phenotypic spectrum of PUS3 deficiency: a p.Tyr71Cys case demonstrating a stable, nonprogressive leukoencephalopathy pattern. *Molecular Syndromology*, 2026.
3. Aydın Z, Çaycı FŞ, İnözü M, Şahin İ, Konrad M, Bayrakçı US. Evaluation of children with familial hypomagnesemia with hypercalciuria and nephrocalcinosis. *Turkish Journal of Pediatric Disease*, 2025.
4. Alabbasi L, Ben Turkia H, Nass M, Sahin I. Diagnostic challenges in the myopathic variant of carnitine palmitoyltransferase II deficiency: a case report. *Cureus*, 2024.
5. Sahin I, Erdem HB, Bahsi T, Saat H. Expanding the genotype–phenotype correlations and mutational spectrum in inherited retinal diseases: novel and recurrent mutations. *Cureus*, 2024. doi:10.7759/cureus.53742
6. Erdem HB, Alay MT, Özdemir Z, Çevik E, Ateş Ö, Karaçin C, Şahin İ, Doğan M, Bahsi T. Higher TP53 somatic mutation prevalence from liquid biopsy analysis in ever smoker non-small-cell lung cancer patients. *Mutation Research*, 2023. doi:10.1016/j.mrfmmm.2023.111847
7. Sahin I, Kandemir N, Saat H. Expanding the genotype–phenotype correlations in Alport syndrome: novel mutations, digenic inheritance and genetic modifiers. *Egyptian Journal of Medical Human Genetics*, 2023. doi:10.1186/s43042-023-00441-x
8. Al-Kafaji G, Jassim G, AlHajeri A, Alawadhi AMT, Fida M, Sahin I, Alali F, Fadel E. Investigation of germline variants in Bahraini women with breast cancer using next-generation sequencing based multigene panel. *PLOS ONE*, 2023. doi:10.1371/journal.pone.0291015
9. Aydın Z, İnözü M, Şahin İ, Bayrakçı US. A case series of four patients with congenital nephrotic syndrome. *Journal of Child*, 2023.
10. Saat H, Sahin I, Erdem HB, Özgür S, Terlemez Tokgöz S, Bahsi T. Molecular approach of hereditary arrhythmias, long QT syndrome and arrhythmogenic right ventricular cardiomyopathy. *The Anatolian Journal of Cardiology*, 2022. doi:10.5152/anatoljcardiol.2022.1324
11. Saat H, Sahin I, Düzkale N, Gönül M, Bahsi T. Genetic etiology of ichthyosis in Turkish patients: next-generation sequencing identified seven novel mutations. *Medeniyet Medical Journal*, 2022.
12. Sahin I, Saat H, Erdem HB, Bahsi T. Non-invasive cancer genotyping: a single-centre experience on the challenges, limitations and methods. *Gazi Medical Journal*, 2022.
13. Sahin I, Saat H. New perspectives on the recurrent monoallelic germline mutations of DNA repair and checkpoint genes and clinical variability. *Genetic Testing and Molecular Biomarkers*, 2022.
14. Sahin I, Saat H, Aksoy S. New perspectives on the recurrent BRCA mutations and clinical variability. *International Journal of Hematology and Oncology*, 2022.
15. Sahin I, Erdem HB. Molecular characterization reveals the importance and diversity of germline and somatic RET mutations in cancer. *Acta Oncologica Turcica*, 2021.
16. Sahin I, Saat H, Aksoy S, Dizdar Ö, Erdem HB, Bahsi T. Liquid biopsy: novel perspectives on the importance and spectrum of PIK3CA, PTEN and RET mutations in solid tumors. *Molecular and Clinical Oncology*, 2021. doi:10.3892/mco.2021.2434
17. Sahin I, Saat H. Hereditary spastic paraplegia: new insights into clinical variability and spasticity–ataxia phenotype, and novel mutations. *Acta Neurologica Belgica*, 2021. doi:10.1007/s13760-021-01779-y
18. Sahin I, Gündoğdu B, Ceylan AC, Erdem HB, Tatar A. High expression of stem cell-related genes in polyps with villous features and high-grade dysplasia support malignant phenotype and colorectal carcinogenesis. *Asian Pacific Journal of Cancer Prevention*, 2021.
19. Sahin I, Saat H. A novel BRCA1 duplication and new insights on the spectrum and frequency of germline large genomic rearrangements in BRCA1/BRCA2. *Molecular Biology Reports*, 2021. doi:10.1007/s11033-021-06499-3

20. Saat H, Sahin I. Mutation spectrum of hereditary myopathies in Turkish patients and novel variants. *Annals of Human Genetics*, 2021. doi:10.1111/ahg.12429
21. Ceylan AC, Erdem HB, Şahin İ, Agarwal M. SMN1 gene copy number analysis for spinal muscular atrophy (SMA) in a Turkish cohort by CODE-SEQ technology, an integrated solution for detection of SMN1 and SMN2 copy numbers and the "2+0" genotype. *Neurological Sciences*, 2020.
22. Pehlivan D, Bayram Y, Gunes N, Coban Akdemir Z, Shukla A, Bierhals T, Tabakci B, Sahin Y, Gezdirici A, Fatih JM, Gulec EY, Yesil G, Punetha J, Ocak Z, Grochowski CM, Karaca E, Albayrak HM, Radhakrishnan P, Erdem HB, Sahin I, et al., Lupski JR. The genomics of arthrogryposis, a complex trait: candidate genes and further evidence for oligogenic inheritance. *The American Journal of Human Genetics*, 2019. doi:10.1016/j.ajhg.2019.05.015
23. Ceylan AC, Sahin I, Erdem HB, Kayhan G, Simsek-Kiper PO, Utine GE, Perçin F, Boduroglu K, Alikışifoğlu M. An 8-case 1q21 region series: novel aberrations and clinical variability with new features. *Journal of Intellectual Disability Research*, 2019.
24. Sahin I. Algorithms, guidelines and genetic consultation in non-invasive cancer genotyping (liquid biopsy, plasma genotyping). *Gazi Medical Journal*, 2019. Abstract, 13th National Congress of Medical Genetics with International Participation.
25. Ceylan AC, Citli S, Erdem HB, Sahin I, Acar Arslan E, Erdogan M. Importance and usage of chromosomal microarray analysis in diagnosing intellectual disability, global developmental delay and autism, and discovering new loci for these disorders. *Molecular Cytogenetics*, 2018. doi:10.1186/s13039-018-0402-4
26. Keles M, Sahin I, Kurt A, Bozoglu C, Simsek G, Kabalar E, Tatar A. Mitochondrial DNA deletions in patients with esophagitis, Barrett's esophagus, esophageal adenocarcinoma and squamous cell carcinoma. *African Health Sciences*, 2018.
27. Erdem HB, Ceylan AC, Sahin I, Sever-Erdem Z, Citli S, Tatar A. Mitochondrial DNA copy number alterations in familial Mediterranean fever patients. *Bratislava Medical Journal*, 2018.
28. Erdem HB, Ceylan AC, Kaya A, Bahsi T, Sever Erdem Z, Sahin I, Tatar A. Positive correlation between platelet parameters and genetic markers of thrombophilia panel in recurrent pregnancy loss. *Eurasian Journal of Family Medicine*, 2018.
29. Sahin I, Erdem HB, Tan H, Tatar A. Becker's myotonia: novel mutations and clinical variability in patients born to consanguineous parents. *Acta Neurologica Belgica*, 2018.
30. Erdem HB, Sahin I, Tatar A. Myhre syndrome with novel findings: bilateral congenital cortical cataract, bilateral papilledema, accessory nipple and adenoid hypertrophy. *Clinical Dysmorphology*, 2017.
31. Erdem HB, Sahin I, Tatar A. Genetic basis and approaches in skin disorders. *Türkiye Klinikleri Journal of Dermatology – Special Topics*, 2017.
32. Erdem HB, Sahin I, Tasdemir S, Tatar A. Partial trisomy 5p12-q11.2 resulting from a marker chromosome: a new case report with attention deficit hyperactivity disorder. *Genetic Counseling*, 2016.
33. Yuce Kahraman C, Tasdemir S, Sahin I, Marzioglu Ozdemir E, Yaralı O, Ziypak T, Adanur S, Kahraman M, Tatar A. The relationship between endothelial nitric oxide synthase gene (NOS3) polymorphisms, NOS3 expression and varicocele. *Genetic Testing and Molecular Biomarkers*, 2016.
34. Tasdemir S, Erozu R, Dogan H, Erdem HB, Sahin I, Kara M, Engin RI, Turkez H. Association between human hair loss and the expression levels of nucleolin, nucleophosmin and UBTF genes. *Genetic Testing and Molecular Biomarkers*, 2016.
35. Tasdemir S, Erdem HB, Sahin I, Ozel L, Ozdemir G, Erozu R, Tatar A. Correlation with platelet parameters and genetic markers of thrombophilia panel (factor II g.20210G>A, factor V Leiden, MTHFR C677T/A1298C, PAI-1, β -fibrinogen, factor XIII A V34L, glycoprotein IIIa L33P) in ischemic strokes. *NeuroMolecular Medicine*, 2016.
36. Tasdemir S, Sahin I, Çayır A, Yuce I, Ceylaner S, Tatar A. Vici syndrome in siblings born to consanguineous parents. *American Journal of Medical Genetics Part A*, 2016. doi:10.1002/ajmg.a.37398
37. Tasdemir S, Erdem HB, Sahin I, Kara M, Tatar A. Waardenburg syndrome type 1 and a rare finding of anal atresia. *Genetic Counseling*, 2015.

38. Tasdemir S, Sahin I, Morris-Rosendahl DJ, Marzioglu E, Çayır A, Yuce I, Tatar A. Recurrent RAB3GAP1 mutations in the Turkish population. *Genetic Counseling*, 2015.
39. Tatar A, Tasdemir S, Sahin I, Bozoglu C, Erdem HB, Yoruk O, Tatar A. Mitochondrial DNA deletions in patients with chronic suppurative otitis media. *European Archives of Oto-Rhino-Laryngology*, 2015.
40. Bayramoğlu A, Saritemur M, Tasdemir S, Omeroglu M, Erdem HB, Sahin I. A rare cause of dyspnea in emergency medicine: Keutel syndrome. *The American Journal of Emergency Medicine*, 2015.
41. Tasdemir S, Sahin I, Çayır A, Doneray H, Solomon B, Muenke M, Yuce I, Tatar A. Holoprosencephaly: ZIC2 mutation in a case with panhypopituitarism. *Journal of Pediatric Endocrinology and Metabolism*, 2014.
42. Taşdemir Ş, Yılmaz M, Şahin İ, Erdem HB, Al RA, İngeç M, Tatar A. Retrospective analysis of 1429 cases who underwent amniocentesis and cordocentesis. *Perinatal Journal*, 2014.
43. Aydın Z, Çaycı FŞ, İnözü M, Şahin İ, Konrad M, Bayrakçı US. Seven pediatric cases with familial hypomagnesemia with hypercalciuria and nephrocalcinosis. *Research Square*, 2022. Preprint.