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ÖNLÜKLÜ
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EDUCATION AND TRAINING ACTIVITIES

M.D., Hacettepe University Medical School, Ankara, 2001 (graduation date)

PROFESSIONAL EXPERIENCE

- **Attending Physician,**
Ankara City Hospital, Pediatric Genetic Unit, Ankara, Turkey,
- **Fellowship,** Hacettepe University , Dept. of Pediatrics, Pediatric Genetic Unit
Ankara, Turkey
- **Residency,** Hacettepe University , Dept. of Pediatrics, Ankara, Turkey

TEACHING EXPERIENCE

- **Supervised Graduate Thesis:** 2008, Ventricular Tachycardias in childhood

2013, Mowat-Wilson Syndrome, Clinical and Molecular Analysis

LANGUAGES: English

PUBLICATIONS

1. Kılıç E, Çetinkaya A, Utine GE, Boduroğlu OK (2016). **A Diagnosis to Consider in Intellectual Disability: Mowat-Wilson Syndrome.** Journal of Child Neurology, 31(7), 913-7. Doi: 10.1177/0883073815627884.
2. Kılıç E, Çeliker A, Karagöz T, Alehan D, Özkutlu S, Özer S (2012). **Analysis of idiopathic ventricular tachycardia in childhood.** The Turkish Journal of Pediatrics, 54(3), 269-272.



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3. Nuria GS, Mittaz L, Xavier C, Bartels C, Tüysüz B, Alanay Y, Cimaz R, Cormier Daire V, Rocco MD, Duba HC, Forzano F, Hospach T, **Kılıç E**, Kuemmerledeschner J, Mortier G, Mrusek S, Nampoothiri S, Obersztyn E, Pauli RM, Selicorni A, Tenconi R, Unger S, Utine GE, Wright M, Zabel B, Warman ML, Supertifurga A, Bonafe L (2012). **The Diagnostic Challenge of Progressive Pseudorheumatoid Dysplasia (PPRD): A Review of Clinical Features, Radiographic Features, and *WISP3* Mutations in 63 Affected Individuals.** American Journal of Medical Genetics Part C, 160(3), 217-229. Doi: 10.1002/ajmg.c.31333.
4. Ünal Ş, Alanay Y, Çetin M, Boduroğlu K, Utine GE, Cormier Daire V, Huber C, Özsürekeçi Y, **Kılıç E**, Şimsek Kiper PÖ (2014). **Striking Hematological Abnormalities in Patients With Microcephalic Osteodysplastic Primordial Dwarfism Type II (MOPD II): A Potential Role of Pericentrin in Hematopoiesis.** Pediatr Blood Cancer, 61(2), 301-305. Doi: 10.1002/pbc.24783.
5. Wieczorek D, Bogershausen N, Beleggia F, Steiner Haldensta S, Pohl E, Li Y, Milz E, Martin M, Thiele H, Altmüller J, Alanay Y, Klein Hitpass L, Bohringer S, Wollstein Andreas, Albrecht Beate, Boduroğlu K, Caliebe A, Chrzanowska K, Cristofoli F, Czeschik J, Devriendt K, Dotti MT, Elçioglu N, Gener B, Goecke TO, Krajewska WM, Guillen NE, Hayek J, Houge G, **Kılıç E**, Lopez Gonzalez V, Kuechler A, Lyonnet S, Mari F, Marozza A, Dramard MM (2013). **A comprehensive molecular study on Coffin–Siris and Nicolaides–Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling.** Human Molecular Genetics, 22(25), 5121-5135. Doi: 10.1093/hmg/ddt366.
6. Piras R, Chiappe F, Torraca IL, Buers I, Usala G, Angius A, Akin MA, Basel VL, Benedicenti F, Chiodin E, El Assy O, Feingold-Zadok M, Guibert J, Kamien B, Kasapkara ÇS, **Kılıç E**, Boduroğlu K, Manzur AY, Onal Eray E, Paderi E, Herrero Roche C, Ünal S, Utine G E, Zanda G,



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- Zankl A, Zampino G, Crisponi G, Crisponi L (2014). **Expanding the Mutational Spectrum of *CRLF1* in Crisponi/CISS1 Syndrome.** Human Mutation, 35(4), 424-433. Doi: 10.1002/humu.22522.
7. Kara A, Tezer H, Devrim İ, Kılıç E, Karagöz T, Özer S, Cengiz AB, Seçmeer G (2006). **Kawasaki Disease A Case Report in Extreme of Pediatrics.** Infectious Diseases in Clinical Practice, 14(5), 333-334. Doi:10.1097/01.idc.0000219055.28051.fd.
 8. Kılıç E, Ertugrul İ, Özer S, Alikasifoglu M, Aktas D, Boduroğlu OK, Utine GE (2014). **Jervell and Lange-Nielsen syndrome with homozygous missense mutation of the *KCNQ1* gene.** The Turkish Journal of Pediatrics, 56(5), 542-545.
 9. Kılıç E, Sahin M, Sahin S, Özer S (2009). **Isotretinoin (13-cis-retinoic acid)-associated premature ventricular contractions.** The Turkish Journal of Pediatrics, 51(4), 387-388.
 10. Kılıç E, Utine GE, Boduroğlu K (2013). **A case of Sotos syndrome with 5q35 microdeletion and novel clinical findings.** The Turkish Journal of Pediatrics, 55(2), 207-209.
 11. Tarlan B, Kıratlı H, Kılıç E, Utine GE, Boduroğlu OK (2013). **A Case of 22q11.2 Deletion Syndrome with Right Microphthalmia and Left Corneal Staphyloma.** Ophthalmic Genetics, 35(4), 248-251. Doi: 10.3109/13816810.2013.811269.
 12. Kılıç E, Yigit G, Utine GE, Wollnik B, Mihçı E, Nur BG, Boduroğlu K (2015). **A Novel Mutation in *RNU4ATAC* in a Patient with Microcephalic Osteodysplastic Primordial Dwarfism Type I.** American Journal of Medical Genetics Part A, 167(4), 919-921. Doi: 10.1002/ajmg.a.36955.
 13. Kılıç E, Kılıç M, Utine GE, Sivri S, Coskun T, Alanay Y (2014). **A case of fucosidosis type II: diagnosed with dysmorphological and radiological findings.** The Turkish Journal of Pediatrics, 56(4), 430-433.



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14. Kılıç E, Utine GE, Ünal S, Haliloglu G, Karlı Oguz K, Çetin M, Boduroğlu K, Alanay Y (2012). **Medical management of moyamoya disease and recurrent stroke in an infant with Majewski osteodysplastic primordial dwarfism type II (MOPD II).** European Journal of Pediatrics, 171, 1567-1571. Doi: 10.1007/s00431-012-1732-6.
15. Kılıç E, Alanay Y, Utine GE, Özgen-Mocan B, Robinson Peter N (2012). **Arterial tortuosity and aneurysm in a case of Loeys-Dietz syndrome type IB with a mutation p.R537P in the TGFBR2 gene.** The Turkish Journal of Pediatrics, 54(2), 198-202.
16. Turgal M, Özyüncü Ö, Utine GE, Kılıç E, Boduroğlu K (2014). **Prenatal diagnosis in a fetus with de-novo 20q11.2q13.1 deletion and review of the literature.** Clinical Dysmorphology, 23(3), 111-113. Doi:10.1097/MCD.0000000000000037._
17. Alan Y, HersHKovitz D, Indelman M, Galloway P, Whiteford M, Sprecher E, Kılıç E (2011). **Novel mutations in GALNT3 causing hyperphosphatemic familial tumoral calcinosis.** J Bone Miner Metab, 29(5), 621-625. Doi: 10.1007/s00774-011-0260-1.
18. Angius A, Uva P, Buers I, Oppo M, Puddu A, Onano S, Persico I, Loi A, Marcia L, Höhne W, Cuccuru G, Fotia G, Deiana M, Marongiu M, Atalay HT, Inan S, El Assy O, Smit LM, Okur I, Boduroğlu K, Utine GE, Kılıç E, Zampino G, Crisponi G, Crisponi L, Rutsch F (2016). **Bi-allelic Mutations in KLHL7 Cause a Crisponi/CISS1-like Phenotype Associated with Early-Onset Retinitis Pigmentosa.** Am J Hum Genet, 99(1), 236-45. Doi: 10.1016/j.ajhg.2016.05.026._
19. Hizarcioglu-Gulsen H, Kilic E, Dominguez-Garrido E, Aydemir Y, Utine GE, Saltik-Temizel IN (2017). **Polyposis deserves a perfect physical examination for final diagnosis: Bannayan-Riley-Ruvalcaba syndrome.** The Turkish Journal of Pediatrics 2017; 59: 80-83. DOI. 10.24953/turkjp.2017.01.014._



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20. Ehmke N, Graul-Neumann L, Smorag L, Koenig R, Segebrecht L, Magoulas P, Scaglia F, **Kilic E**, Hennig AF, Adolphs N, Saha N, Fauler B, Kalscheuer VM, Hennig F, Altmüller J, Netzer C, Thiele H, Nürnberg P, Yigit G, Jäger M, Hecht J, Krüger U, Mielke T, Krawitz PM, Horn D, Schuelke M, Mundlos S, Bacino CA, Bonnen PE, Wollnik B, Fischer-Zirnsak B, Kornak U (2017). **De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction.** Am J Hum Genet. 2017 Nov 2;101(5):833-843. doi: 10.1016/j. ajhg.2017.09.016.
21. Daphné Lehalle, Umut Altunoglu, Ange-Line Bruel, Patricia Blanchet, Jong-Woo Choi, Julie Désir, Yannis Duffourd, Hulya Kayserili, **Esra Kiliç**, Damien Lederer, Lucile Pinson, Christel Thauvin-Robinet, Julien Thevenon, Patrick Callier, Laurence Faivre (2017). **Clinical delineation of a subtype of frontonasal dysplasia with creased nasal dorsum and occasional limb anomalies: report of six novel cases and review of the literature.** Am J Med Genet. 2017;1-7. DOI: 10.1002/ajmg.a.38490.
22. **Kılıç E**, Özer S (2008). **Konjenital Kalp Hastalarında Ventriküler Aritmiler ve Ani Kardiyak Ölüm.** Türkiye Klinikleri Journal of Cardiology Special Topics, 1(6), 87-99.
23. **Kılıç E**, Tezer H, Kılıç M, Devrim İ, Haliloglu M, Cengiz AB, Yüce A, Kara A (2009). **Fasciola hepatica, bir parazitik enfestasyonda radyolojik tanı: Bir vaka takdimi.** Çocuk Sağlığı ve Hastalıkları Dergisi, 52(4), 216-218.
24. **Kılıç E**, Alanay Y, Korkmaz A, Utine GE, Karagöz T, Boduroğlu K (2014). **Tam ektopia kordisli bir Cantrell pentalojisi vakası.** Çocuk Sağlığı ve Hastalıkları Dergisi, 54(2), 83-86.
25. Kılıç M, Yıldız D, Özdemir O, Koçak M, Günbey S, Yasar D, **Kılıç E** (2013). **Tip II sialidoz: Bir vaka takdimi.** Çocuk Sağlığı ve Hastalıkları Dergisi, 56(4), 181-183.



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26. Kılıç M, Kılıç E, Utine GE, Nakano H, Boduroğlu K, Coskun T (2014). **Sjögren-Larsson sendromu: Yeni bir mutasyon ve Türk vakaların gözden geçirilmesi.** Çocuk Sağlığı ve Hastalıkları Dergisi, 57(3), 191-194.
27. Kılıç E (2017). **Progresif Psödoromatoid Kondrodisplazi, eklem şişliği ve hareket kısıtlılığının sıra dışı bir nedeni - Progressive pseudorheumatoid condrodysplasia, an unusual cause of joint swelling and stiffness.** The Journal of Pediatric Research. JPR-19970.
28. Gürkaş E, Maraş-Genç H, Kılıç E (2017). **Triple X Syndrome with a Rare Finding: Cleft Palate -Triple X Sendromunda Nadir Bir Bulgu: Yarık Damak.** The Journal of Pediatric Research. JPR-32154.
29. Kılıç M, Oğuz KK, Kılıç E, Yüksel D, Demirci H, Sağiroğlu MŞ, Yücel-Yılmaz D, Özgül RK (2017). **A patient with mitochondrial disorder due to a novel mutation in MRPS22.** Metab Brain Dis. 2017 Oct;32(5):1389-1393. doi: 10.1007/s11011-017-0074-5. Epub 2017 Jul 27.
30. Kılıç M, Ceylan AC, Örün UA, Kılıç E. Metab Brain Dis. 2018 Apr. **First cardiac manifestation of hypotonia-cystinuria syndrome.** doi: 10.1007/s11011-018-0226-2.
31. Kılıç M, Kılıç E, Yılmaz DY, Özgül RK. **Reply to 'contribution of the MRPS22 variant and a down mosaic to the phenotype'.** Metab Brain Dis. 2018 Dec;33(6):1779-1780. doi: 10.1007/s11011-018-0300-9.
32. Van der Sluijs EPJ, Jansen S, Vergano SA, Adachi-Fukuda M, Alanay Y, AlKindy A, Baban A, Bayat A, Beck-Wödl S, Berry K, Bijlsma EK, Bok LA, Brouwer AFJ, van der Burgt I, Campeau PM, Canham N, Chrzanowska K, Chu YWY, Chung BHY, Dahan K, DeRademaeker M, Destree A, Dudding-Byth T, Earl R, Elcioglu N, Elias ER, Fagerberg C, Gardham A, Gener B, Gerkes EH, Grasshoff U, van Haeringen A, Heitink KR, Herkert JC, den Hollander NS, Horn D, Hunt D,



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Kant SG, Kato M, Kayserili H, Kersseboom R, Kilic E, Krajewska-Walasek M, Lammers K, Laulund LW, Lederer D, Lees M, López-González V, Maas S, Mancini GMS, Marcelis C, Martinez F, Maystadt I, McGuire M, McKee S, Mehta S, Metcalfe K, Milunsky J, Mizuno S, Moeschler JB, Netzer C, Ockeloen CW, Oehl-Jaschkowitz B, Okamoto N, Olminkhof SNM, Orellana C, Pasquier L, Pottinger C, Riehmer V, Robertson SP, Roifman M, Rooryck C, Ropers FG, Rosello M, Ruivenkamp CAL, Sagioglu MS, Sallevelt SCEH, Sanchis Calvo A, Simsek-Kiper PO, Soares G, Solaeche L, Mujgan Sonmez F, Splitt M, Steenbeek D, Stegmann APA, Stumpel CTRM, Tanabe S, Uctepe E, Utine GE, Veenstra-Knol HE, Venkateswaran S, Vilain C, Vincent-Delorme C, Vulto-van Silfhout AT, Wheeler P, Wilson GN, Wilson LC, Wollnik B, Kosho T, Wieczorek D, Eichler E, Pfundt R, de Vries BBA, Clayton-Smith J, Santen GWE. **The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome.** Genet Med. 2018 Nov 8. doi: 10.1038/s41436-018-0330-z.

MEMBERSHIPS AND ASSOCIATIONS

Pediatric Genetic

SKILLS AND INTERESTS

Malformations, Intellectual Disability, Skeletal Dysplasias