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Tıbbi ilgi ve uzmanlık alanları:

Çocuk Sağlığı ve Hastalıkları

Çocuk Genetik

Eğitim:

2009-2013 Hacettepe Üniversitesi–Çocuk Sağlığı Hast.-Çocuk Genetik Ünitesi

2001-2008 Hacettepe Üniversitesi–Çocuk Sağlığı Hastalıkları AD.

1994-2001 Hacettepe Üniversitesi Tıp Fakültesi

Çalışılan Kurumlar:

2002/2008 Hacettepe Üniversitesi Çocuk Hastanesi Çocuk Sağlığı ve Hastalıkları

2009/2013 Hacettepe Üniversitesi Çocuk Hastanesi Çocuk Genetik Bölümü

2013/- Ankara Çocuk Sağlığı Hematoloji Onkoloji Hastanesi Çocuk Genetik

Yayınlar:

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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4. Ünal Ş, Alanay Y, Çetin M, Boduroğlu K, Utine GE, Cormier Daire V, Huber C, Özsürekçi 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.
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6. Piras R, Chiappe F, Torracca IL, Buers I, Usala G, Angius A, Akın 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, 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.



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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.
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.



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- 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._
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