BİRUNİ ÜNİVERSİTESİ
"Bilimin Geleceği"

ÖZGEÇMİŞ VE ESERLER LİSTESİ



ÖZGEÇMİŞ

Adı ve Soyadı: Adnan YÜKSEL

Akademik Unvanı: Prof. Dr.

İş Adresi:

E-postası: ayuksel@biruni.edu.tr

Uzmanlık Alanı: Çocuk Sağlığı ve Hastalıkları
Sağlık Bilimleri

Derece	Bölüm/Program	Üniversite	Yıl
Doktora	GENETİK ANABİLİM DALI	İstanbul Üniversitesi	2002
Tıpta Yandal Uzmanlık	DAHİLİ TIP BİLİMLERİ BÖLÜMÜ	İstanbul Üniversitesi	1994
Tıpta Uzmanlık	DAHİLİ TIP BİLİMLERİ BÖLÜMÜ	İstanbul Üniversitesi	1991
Lisans		İstanbul Üniversitesi	1987

Görev Unvanı	Görev Yeri	Yıl
Prof. Dr.	Biruni Üniversitesi	2014-Devam Ediyor
Prof. Dr.	İstanbul Üniversitesi	2007-2010
Prof. Dr.	İstanbul Üniversitesi	1987-1991

Yönetilen Doktora Tezleri/Sanatta Yeterlik Çalışmaları:

1. Epilepsi hastalarında ilaç direncinin MDR1 genindeki C3435T VE G2677T/A polimorfizmleri ile ilişkisi (2009)
2. Vigabatrin uygulanan gebe wistar-albino ratların fetüslerinde teratojenik etkilerin morfolojik olarak araştırılması (2002)
3. Karbamazepin kullanan gebelerde ve çocuklarında antioksidan sistemin incelenmesi (2002)
4. Down sendromlu süt çocuklarında görsel ve işitsel fonksiyonların prospektif takibi (2001)
5. Down Sendromlu Türk Çocuklarının Büyüme ve Gelişme Çizelgeleri ile Nörolojik Gelişimlerinin Prospektif Tayini (1998)

İdari Görevler:

1. Rektör - Biruni Üniversitesi (2019 - Devam Ediyor)
2. Rektör - Biruni Üniversitesi (2015 - 2019)
3. Kurucu Rektör - Biruni Üniversitesi (2014 - 2015)
4. Rektör Danışmanı - İstanbul Üniversitesi (2013 - 2014)
5. Rektör - Bezmiâlem Vakıf Üniversitesi (2010 - 2012)
6. Anabilim/Bilim Dalı Başkanı - İstanbul Üniversitesi (2007 - 2010)

Bilimsel Kuruluşlara Üyelikler:

1. Mesleki Dernek (Üye) (2014 - Devam Ediyor)
2. Mesleki Dernek (Üye) (2009 - Devam Ediyor)

3. Mesleki Dernek (Üye) (2025 - Devam Ediyor)
4. Mesleki Dernek (Üye) (2025 - Devam Ediyor)

Ödüller:

1. Türkiye Bilim İnsanı Ödülü Birinciliği (2017)
2. Bilim Günleri Ödülü (2006)
3. İstanbul Üniversitesi Araştırma Fonu, Bilimsel Araştırma Ödülü (2002)

ESERLER

A. Uluslararası hakemli dergilerde yayımlanan makaleler:

A1. Bozlak S., Yavaş C., Yalcin E., Şeflekçi Y., Dogan T., Ece A., Akyel N. G., Yüksel A., "Neurovascular Involvement in Arterial Tortuosity Syndrome Associated with a Homozygous SLC2A10 p.(Trp162Ter) Variant: Clinical, Molecular, and In Silico Characterization", *International Journal of Molecular Sciences*, vol. 27, no. 15, 2026.

A2. Ömerustaoğlu A., Kırtaş A. T., Baran Karalar E., Şahin D., Satoğlu R., Satıcı S. A., Yüksel A., "Sustainable Digital Learning in Higher Education: Development of the Moodle-Based BirDeHa Usability Scale and Its Associations with Academic Locus of Control and Achievement Motivation", *Sustainability (Switzerland)*, vol. 18, no. 12, 2026.

A3. Manav Yiğit Z., Özyavuz Çubuk P., Bayrak K. U., Düz M. B., Yüksel A., "CHAMP1-Related Neurodevelopmental Disorder: Two Turkish Cases with Novel Truncating Variants and Literature Review", *Molecular Syndromology*, 2026.

A4. Yaman A., Sert A., Yüksel A., "Multi-organ failure secondary to eltrombopag overexposure: a novel utilization of hemodiafiltration in pediatric critical care management Multiorganversagen infolge einer Überexposition mit Eltrombopag: Eine neuartige Anwendung der Hämodiafiltration in der pädiatrischen Intensivmedizin", *Klinische Padiatrie*, 2026.

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A7. Alagoz M., Kherad N., Gunger E., Kaymaz S., Yüksel A., "The New CIC Mutation Associates with Mental Retardation and Severity of Seizure in Turkish Child with a Rare Class I Glucose-6-Phosphate Dehydrogenase Deficiency", *Journal of Molecular Neuroscience*, vol. 70, no. 12, pp. 2077-2084, 2020.

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A9. Alagoz M., Kherad N., Bozkurt S., Yüksel A., "New mutations in KCNT2 gene causing early infantile epileptic encephalopathy type 57: Case study and literature review", *Acta Biochimica Polonica*, vol. 67, no. 3, pp. 431-434, 2020.

A10. Alagoz M., Kherad N., Gavaz M., Yüksel A., "New Genetic Approaches for Early Diagnosis and Treatment of Autism Spectrum Disorders", *Review Journal of Autism and Developmental Disorders*, vol. 6, no. 4, pp. 367-380, 2019.

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A45. Brancati F., Iannicelli M., Travaglini L., Mazzotta A., Bertini E., Boltshauser E., D'arrigo S., Emma F., Fazzi E., Gallizzi R. vd., "MKS3/TMEM67 mutations are a major cause of COACH syndrome, a joubert syndrome related disorder with liver involvement", *Human Mutation*, vol. 30, no. 2, 2009.

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D. Ulusal hakemli dergilerde yayımlanan makaleler:

D1. Yıldız Karaahmet A., Bilgiç F. Ş., Yüksel A., "The impact of educational and behavioral self-management interventions for children diagnosed with epilepsy and their families on disease management, quality of life, and psychosocial outcomes: a systematic review and meta-analysis", *Epilepsy and Behavior*, vol. 12, no. 6, 2026.

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D6. Yüksel A., Kayserili H., Yeşil G., Apak M., "Evaluation of mental retardation - Part 2: The factors that elucidate the etiologic diagnosis of the patients with mental retardation or multiple congenital abnormality and mental retardation", *Journal of Pediatric*

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D10. Emiroğlu H. H., Yüksel A., Serdaroğlu P., Topaloğlu H., Erginel-ünaltuna N., Ataoğlu E., Elevli M., "Clinicohistopathologic features of sarcoglycanopathy in four siblings Sarkoglikanopatili dört kardeşin klinikohistopatolojik özellikleri", *Cocuk Sagligi ve Hastaliklari Dergisi*, vol. 47, no. 1, 2004.

D11. Seven M., Özkiliç A., Yüksel A., "Dysmorphic face in two siblings with infantile neuroaxonal dystrophy", *Genetic Counseling*, vol. 13, no. 4, 2002.

D12. Ozkilic A., Seven M., Yüksel A., "A case of acampomelic campomelic dysplasia", *Genetic Counseling*, vol. 13, no. 1, 2002.

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