# CURRICULUM VITAE AND PUBLICATIONS LIST

## CURRICULUM VITAE


Name and Surname: Adnan YÜKSEL

Academic Title: Professor Dr.

Work Address:

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Area of Expertise: Child Health and Diseases
Health Sciences

| Degree | Department/Program | University | Year |
|---|---|---|---|
| Doctorate | GENETİK ANABİLİM DALI | İstanbul University | 2002 |
| Sub-Specialty in Medicine | DAHİLİ TIP BİLİMLERİ BÖLÜMÜ | İstanbul University | 1994 |
| Medical Specialty | DAHİLİ TIP BİLİMLERİ BÖLÜMÜ | İstanbul University | 1991 |
| Bachelor's Degree | | İstanbul University | 1987 |

| Position Title | Workplace | Year |
|---|---|---|
| Professor Dr. | Biruni University | 2014-Continues |
| Professor Dr. | İstanbul University | 2007-2010 |
| Professor Dr. | İstanbul University | 1987-1991 |

Supervised Doctoral Theses/Proficiency Studies in Arts:

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)

Administrative Duties:

Rector - Biruni University (2019 - Continues)
Rector - Biruni University (2015 - 2019)
Founding Rector - Biruni University (2014 - 2015)
Rector Advisor - İstanbul University (2013 - 2014)
Rector - Bezmiâlem Vakıf University (2010 - 2012)
Head of Main Science/Science Department - İstanbul University (2007 - 2010)

Memberships in Scientific Organizations:

Professional Association (Member) (2014 - Continues)
Professional Association (Member) (2009 - Continues)
Professional Association (Member) (2025 - Continues)
Professional Association (Member) (2025 - Continues)

Awards:

First Prize in Turkey Scientist Award (2017)
Science Days Award (2006)
Istanbul University Research Fund, Scientific Research Award (2002)

# PUBLICATIONS

**A. Articles published in international peer-reviewed journals:**

A1. "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, 2020.

A2. "ASC-1 Is a Cell Cycle Regulator Associated with Severe and Mild Forms of Myopathy", Annals of Neurology, 2020.

A3. "New mutations in KCNT2 gene causing early infantile epileptic encephalopathy type 57: Case study and literature review", Acta Biochimica Polonica, 2020.

A4. "New Genetic Approaches for Early Diagnosis and Treatment of Autism Spectrum Disorders", Review Journal of Autism and Developmental Disorders, 2019.

A5. "Incidence of Type 1 Diabetes in Children Aged Below 18 Years during 2013-2015 in Northwest Turkey", JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018.

A6. "Characterization of greater middle eastern genetic variation for enhanced disease gene discovery", Nature Genetics, 2016.

A7. "Revealing the function of a novel splice-site mutation of CHD7 in CHARGE syndrome", Gene, 2016.

A8. "Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease", Neuron, 2015.

A9. "Novel POC1A mutation in primordial dwarfism reveals new insights for centriole biogenesis", Human Molecular Genetics, 2015.

A10. "Whole-exome sequencing revealed two novel mutations in Usher syndrome", Gene, 2015.

A11. "Erratum: Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy (Nat. Genet. (2015) 47: (73-77))", Nature Genetics, 2015.

A12. "Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy", Nature Genetics, 2015.

A13. "Poikiloderma with neutropenia: Genotype-ethnic origin correlation, expanding phenotype and literature review", American Journal of Medical Genetics, Part A, 2014.

A14. "Human CLP1 mutations alter tRNA biogenesis, Affecting both peripheral and central nervous system function", Cell, 2014.

A15. "Melatonin attenuates phenytoin sodium-induced DNA damage", Drug and Chemical Toxicology, 2014.

A16. "The drug-transporter gene MDR1 C3435T and G2677T/A polymorphisms and the risk of multidrug-resistant epilepsy in Turkish children", Molecular Biology Reports, 2014.

A17. "Report of a patient with Temple-Baraitser syndrome", American Journal of Medical Genetics, Part A, 2014.

A18. "Keutel syndrome: Report of two novel MGP mutations and discussion of clinical overlap with arylsulfatase E deficiency and relapsing polychondritis", American Journal of Medical Genetics, Part A, 2014.

A19. "Therapeutic plasma exchange for malignant refractory status epilepticus: A case report", Pediatric Neurology, 2014.

A20. "The effect of genetic polymorphisms of cytochrome P450 CYP2C9, CYP2C19, and CYP2D6 on drug-resistant epilepsy in Turkish children", Molecular Diagnosis and Therapy, 2014.

A21. "Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders", European Journal of Human Genetics, 2013.

A22. "A novel EFNB1 mutation in a patient with craniofrontonasal syndrome and right hallux duplication", Gene, 2013.

A23. "MicroRNA profiling in lymphocytes and serum of tyrosinemia type-I patients", Molecular Biology Reports, 2013.

A24. "Effects of memantine and melatonin on signal transduction pathways vascular leakage and brain injury after focal cerebral ischemia in mice", Neuroscience, 2013.

A25. "Circumferential skin folds and multiple anomalies: Confirmation of a distinct autosomal recessive Michelin tire baby syndrome", Clinical Dysmorphology, 2013.

A26. "Involvement of the corpus callosum splenium in a case with SSPE: Magnetic resonance spectroscopy findings", Archives of Medical Science, 2013.

A27. "Deficiency of selenium and zinc as a causative factor for idiopathic intractable epilepsy", Epilepsy Research, 2013.

A28. "Identification of a novel mutation in ZAP70 and prenatal diagnosis in a Turkish family with severe combined immunodeficiency disorder", Gene, 2013.

A29. "A rare case of split hand/foot malformation with sensorineural hearing loss and Mondini dysplasia", Clinical Dysmorphology, 2013.

A30. "Evidence that membrane-bound G protein-coupled melatonin receptors MT1 and MT2 are not involved in the neuroprotective effects of melatonin in focal cerebral ischemia", Journal of Pineal Research, 2012.

A31. "Glutathione S-transferase M1, GSTT1 and GSTP1 genetic polymorphisms and the risk of age-related macular degeneration", Ophthalmic Research, 2011.

A32. "Polymorphisms of the DNA repair genes XPD and XRCC1 and the risk of age-related macular degeneration", Investigative Ophthalmology and Visual Science, 2010.

A33. "Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies", Human Mutation, 2010.

A34. "Marked Improvement in Segawa Syndrome After l-Dopa and Selegiline Treatment", Pediatric Neurology, 2010.

A35. "Expanding CEP290 mutational spectrumin ciliopathies", American Journal of Medical Genetics, Part A, 2009.

A36. "Molecular genetic screening of MBS1 locus on chromosome 13 for microdeletions and exclusion of FGF9, GSH1 and CDX2 as causative genes in patients with Moebius syndrome", European Journal of Medical Genetics, 2009.

A37. "Mutational screening of BASP1 and transcribed processed pseudogene TPΨg-BASP1 in patients with Möbius syndrome", Journal of Genetics and Genomics, 2009.

A38. "Magnetic resonance imaging, magnetic resonance spectroscopy, and facial dysmorphism in a case of lowe syndrome with novel OCRL1 gene mutation", Journal of Child Neurology, 2009.

A39. "MKS3/TMEM67 mutations are a major cause of COACH syndrome, a joubert syndrome related disorder with liver involvement", Human Mutation, 2009.

A40. "Spontaneous intracranial hypotension syndrome in a patient with Marfan syndrome and autosomal dominant polycystic kidney disease", Headache, 2008.

A41. "CEP290 mutations are frequently identified in the oculo-renal form of Joubert syndrome-related disorders", American Journal of Human Genetics, 2007.

A42. "Facial Dysmorphism in Leigh Syndrome With SURF-1 Mutation and COX Deficiency", Pediatric Neurology, 2006.

A43. "Epilepsy in vacuolating megalencephalic leukoencephalopathy with subcortical cysts", Seizure, 2003.

A44. "Neuroblastoma in a dysmorphic girl with a partial duplication of 2p caused by an unbalanced translocation", Clinical Dysmorphology, 2002.

A45. "Changes in the antioxidant system in epileptic children receiving antiepileptic drugs: Two-year prospective studies", Journal of Child Neurology, 2001.

A46. "Infantile-onset megalencephalic leucoencephalopathy in two siblings", Journal of Paediatrics and Child Health, 2000.

A47. "Siblings with cystic leukoencephalopathy and megalencephaly", Journal of Child Neurology, 2000.

A48. "The effects of carbamazepine and valproic acid on the erythrocyte glutathione, glutathione peroxidase, superoxide dismutase and serum lipid peroxidation in epileptic children", Pharmacological Research, 2000.

A49. "Neuroimaging findings of four patients with Sandhoff disease", Pediatric Neurology, 1999.

A50. "N-acetyl-β-glucosaminidase and β-galactosidase activity in children receiving antiepileptic drugs", Pediatric Neurology, 1999.

A51. "Effects of carbamazepine and valproate on brainstem auditory evoked potentials in epileptic children", Child's Nervous System, 1995.

### D. Articles published in national peer-reviewed journals:

D1. "Demographic and clinical findings of cerebral palsy patients in Istanbul: A multicenter study Istanbul'daki serebral palsi olgularinin klinik ve demografik özellikleri: Çok merkezli çalişma", Turkiye Fiziksel Tip ve Rehabilitasyon Dergisi, 2014.

D2. "Vanishing white matter leukodystrophy, A rare case report Kaybolan beyaz madde hastalığı; nadir birolgu", Turk Pediatri Arsivi, 2013.

D3. "A new syndrome presenting with dysmorphic facies, oculocutaneous albinism, glaucoma, cryptorchidism and mental retardation", Genetic Counseling, 2011.

D4. "Keutel syndrome in a patient presenting with hearing loss", B-ENT, 2010.

D5. "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 Neurosciences, 2007.

D6. "Evaluation of mental retardation - Part 1: Etiologic classification of 4659 patients with mental retardation or multiple congenital abnormality and mental retardation", Journal of Pediatric Neurosciences, 2007.

D7. "Warburg Micro syndrome in a Turkish boy", Clinical Dysmorphology, 2007.

D8. "The effects of vigabatrin on rat liver antioxidant status", Drug Metabolism and Drug Interactions, 2005.

D9. "Clinicohistopathologic features of sarcoglycanopathy in four siblings Sarkoglikanopatili dört kardeşin klinikohistopatolojik özellikleri", Cocuk Sagligi ve Hastaliklari Dergisi, 2004.

D10. "Dysmorphic face in two siblings with infantile neuroaxonal dystrophy", Genetic Counseling, 2002.

D11. "A case of acampomelic campomelic dysplasia", Genetic Counseling, 2002.

D12. "A case of symbrachydactyly with oligodactyly", Genetic Counseling, 2001.

D13. "A variant of Cenani-Lenz type syndactyly", Genetic Counseling, 2000.

D14. "Vitamin and mineral status in Down syndrome", Trace Elements and Electrocytes, 2000.

D15. "Erythrocyte Glutathione, Glutathione Peroxidase, Superoxide Dismutase And Serum Lipid Peroxidation In Epileptic Children With Valproate And Carbamazepine Monotherapy", Journal of Basic and Clinical Physiology and Pharmacology, 2000.

D16. "Two female siblings with a previously unreported MCA/MR syndrome: Pre and postnatal growth retardation, iris colobomata, spasticity, facial dysmorphism and dilated ventricles", Genetic Counseling, 1999.

D17. "A family presenting Goltz syndrome (focal dermal hypoplasia) in three generations", Turkish Journal of Pediatrics, 1998.

D18. "Effect of valproate and carbamazepine on visual evoked potentials in epileptic children", Pediatrics International, 1995.

D19. "Tetra-amelia, lung hypo-/aplasia, cleft lip-palate, and heart defect: A new syndrome?", American Journal of Medical Genetics, 1994.

D20. "The effects of antiepileptic drugs on brainstem auditory evoked potentials", Medical Bulletin of Istanbul Medical Faculty, 1993.

D21. "Brainstem audiometer in the preterm babies given aminoglycosides", Cerrahpasa Tip Fakultesi Dergisi, 1993.

D22. "The evaluation of gonadal function in women with hepatocellular failure secondary to posthepatitic cirrhosis", Istanbul Tip Fakultesi Mecmuasi, 1993.

D23. "Apnea", SENDROM, 1993.

D24. "Influence of long-term carbamazepine treatment on thyroid function", Pediatrics International, 1993.

D25. "Serum thyroid hormones and pituitary response to thyrotropin-releasing hormone in epileptic children receiving anti-epileptic medication", Pediatrics International, 1993.