

Doç.Dr. HÜSEYİN DEMİRBILEK

Kişisel Bilgiler

E-posta: huseyindr@hacettepe.edu.tr

Diğer E-posta: dr_huseyin@hotmail.com

Web: <https://avesis.hacettepe.edu.tr/huseyindr>

Eğitim Bilgileri

Tıpta Yandal Uzmanlık, Hacettepe Üniversitesi, Hacettepe Tıp Fakültesi, Türkiye 2006 - 2010

Tıpta Uzmanlık, Hacettepe Üniversitesi, Hacettepe Tıp Fakültesi, Türkiye 1999 - 2004

Lisans, Hacettepe Üniversitesi, Tıp Fakültesi, Türkiye 1991 - 1998

Araştırma Alanları

Sağlık Bilimleri, Temel Bilimler

Akademik Unvanlar / Görevler

Doç.Dr., Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 2014 - Devam Ediyor

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayımlanan Makaleler

- I. **Clinical Characteristics and Long-term Follow-up of Patients with Diabetes Due To PTF1A Enhancer Mutations.**
Demirbilek H., Cayir A., Flanagan S., Yıldırım R., Kor Y., Gurbuz F., Haliloğlu B., Yıldız M., Baran R., Akbas E., et al.
The Journal of clinical endocrinology and metabolism, cilt.105, 2020 (SCI Expanded İndekslerine Giren Dergi)
- II. **Identification of Three Novel and One Known Mutation in the *WFS1* Gene in Four Unrelated Turkish Families; the Role of Homozygosity Mapping in the Early Diagnosis.**
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- III. **Ectopic Posterior Pituitary, Polydactyly, Midfacial Hypoplasia and Multiple Pituitary Hormone Deficiency due to a Novel Heterozygous IVS11-2A > C(c.1957-2A > C) Mutation in the GLI2 Gene**
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- IV. **Evaluation of the Final Adult Height and Its Determinants in Patients with Growth Hormone Deficiency: A Single-centre Experience from the South-Eastern Region of Turkey**
Demiral M., Unal E., Baysal B., Baran R. T. , DEMİRBILEK H., Ozbek M. N.
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- V. **Validity of Six Month L-Thyroxine Dose for Differentiation of Transient or Permanent Congenital Hypothyroidism**
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- VI. **Frequency of Celiac Disease and Spontaneous Normalization Rate of Celiac Serology in Children and Adolescent Patients with Type 1 Diabetes.**
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- VII. **Neonatal diabetes due to homozygousINSgene promoter mutations: Highly variable phenotype, remission and early relapse during the first 3 years of life**
Demiral M., DEMİRBILEK H., Celik K., Okur N., Hussain K., Ozbek M. N.
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- VIII. **A novel diagnostic tool for the evaluation of hypothalamic-pituitary region and diagnosis of growth hormone deficiency: pons ratio**
Demiral M., Karaca M. S. , Unal E., Baysal B., Baran R. T. , DEMİRBILEK H., Ozbek M. N.
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- IX. **Ectopic posterior pituitary, polydactyly, midfacial hypoplasia and panhypopituitarism due to a novel heterozygous IVS 11-2AC(c.1957-2AC) mutation in GLI2 gene**
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- XI. **Clinical, biochemical and echocardiographic evaluation of patients with congenital rickets due to maternal vitamin D deficiency**
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- XII. **A novel approach for the evaluation of hypothalamic-pituitary region in patients with growth hormone deficiency: Pons ratio**
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- XIII. **Ion Transporters, Channelopathies, and Glucose Disorders**
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- XIV. **Evaluation of intraocular pressure and retinal nerve fiber layer, retinal ganglion cell, central macular thickness, and choroidal thickness using optical coherence tomography in obese children and healthy controls**
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- XV. **Congenital Hyperinsulinism and Evolution to Sulfonylurea-responsive Diabetes Later in Life due to a Novel Homozygous p.L171F ABCC8 Mutation**
Isik E., DEMİRBILEK H., Houghton J. A. , Ellard S., Flanagan S. E. , Hussain K.
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- XVI. **The Genetic and Molecular Mechanisms of Congenital Hyperinsulinism**
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- XVII. **Systemic Pseudohypoadosteronism Type 1 due to 3 Novel Mutations in SCNN1A and SCNN1B Genes**
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- XVIII. **A Distinct Clinical Phenotype in Two Siblings with X-linked Adrenoleukodystrophy**

- Ozdemir Kutbay N., Ozbek M. N. , Sarer Yurekli B., DEMİRBILEK H.
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- XX. **Evaluation of psychological characteristics of Turkish children with type 1 diabetes mellitus from two demographically and geographically distinct regions**
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- XXI. **Permanent neonatal diabetes mellitus and neurological abnormalities due to a novel homozygous missense mutation in NEUROD1**
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- XXII. **Comprehensive Genetic Testing Shows One in Five Children with Diabetes and Non-Autoimmune Extra-Pancreatic Features Have Monogenic Aetiology**
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- XXIII. **Growth Hormon Deficiency in Identical Twins with Gitelman Syndrome Due to Compound Heterozygous Mutation (p.R80fs*35/p.K957X) of the SLC12A3 Gene and the Evaluation of the Response to Growth Hormone Replacement Therapy**
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- XXIV. **Evaluation of Intraocular Pressure and Retinal Nerve Fiber Layer, Retinal Ganglion Cell, Central Macular Thickness and Choroidal Thickness Using Optical Coherence Tomography in Obese Children and Healthy Controls**
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- XXV. **Clinical Characterstics, Genotype-phenotype Correlations and Follow Up of Patients with Congenital Hyperinsulinaemic Hypoglycaemia; Single Center Experience from a Southeastern City of Turkey**
Ozbek M. N. , DEMİRBILEK H., Haliloglu B., Demiral M., Baran R. T. , Ellard S., Houghton J., Flanagan S. E. , Hussain K.
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- XXVI. **Systemic Pseudohypoaldosteronism Type 1 due to 3 Novel Mutations in SCNN1A and SCNN1B Genes; Report of 3 Cases**
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- XXVII. **Presenting Features, Clinical Characteristics and Follow Up of Familial Isolated Glucocorticoid Deficiency (FGD) Due to Mutations in MC2R and MRAP Genes**
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- XXIX. **Hyperinsulinaemic hypoglycaemia in children and adults**
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- XXX. **Evaluation of vitamin D levels in patients with acute rheumatic fever**
Onan S. H. , Demirbilek H., Aldudak B., Bilici M., Demir F., Yilmazer M. M.
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- XXXI. **Clinical and biochemical characteristics and bone mineral density of homozygous, compound**

- heterozygous and heterozygous carriers of three novel IGFALS mutations**
Isik E., Haliloglu B., van Doorn J., DEMİRBILEK H., Scheltinga S. A., Losekoot M., Wit J. M.
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- XXXII. **PSYCHOMETRIC ANALYSIS USING CHILD BEHAVIOR CHECKLIST (CBCL) IN TYPE 1 DIABETIC TURKISH CHILDREN FROM TWO DISTINCT DEMOGRAPHIC AND GEOGRAPHICAL REGIONS**
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- XXXIII. **Evaluation of therapeutics management patterns and glycemic control of pediatric type 1 diabetes mellitus patients in Turkey: A nationwide cross-sectional study**
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- XXXIV. **Bone Mineral Density in Adolescent Girls with Hypogonadotropic and Hypergonadotropic Hypogonadism**
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- XXXV. **A novel ALMS1 homozygous mutation in two Turkish brothers with Alstrom syndrome**
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- XXXVI. **Severe Hypercalcemia in a Child With Acute Lymphoblastic Leukemia Relapse: Successful Management With Combination of Calcitonin and Bisphosphonate**
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- XXXVII. **Anthropometric findings from birth to adulthood and their relation with karyotype distribution in Turkish girls with Turner syndrome**
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- XXXVIII. **Potential Role of Vitamin D in Pathogenesis of Acute Rheumatic Fever**
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- XXXIX. **A Syndrome of Permanent Neonatal Diabetes Mellitus and Neurological Abnormalities due to a Novel Homozygous Missense c.449T > A (p.I150N) Mutation in NEUROD1 Gene**
HATİPOĞLU N., DEMİRBILEK H., Gul U., Tati Z. U., Flanagan S., Ellard S., De Franco E., Kurtoglu S.
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- XL. **Kallmann Syndrome Due to a Homozygous Missense c.217C > T (p.R73C) Mutation Detected in the Exon-2 of the PROK2 Gene**
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- XLI. **Evaluation of the Epidemiological, Presenting and Follow-up Characteristics and their Impacts on the Glycemic Control in a Large Cohort of Pediatric Type 1 Diabetes Mellitus Patients from Southeastern Anatolian Region of Turkey**
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- XLII. **Severe Hyponatremia and Repeated Intestinal Resections for Intestinal Dysmotility Mimicking Congenital Aganglionic Megacolon due to Delay in the Diagnosis of Congenital Hypothyroidism**
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- XLIII. **Rare Causes of Primary Adrenal Insufficiency: Genetic and Clinical Characterization of a Large Nationwide Cohort**

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- XLIV. **Genotype and Phenotype Characteristics in 22 Patients with Vitamin D-Dependent Rickets Type I**
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- XLV. **Growth curves for Turkish Girls with Turner Syndrome: Results of the Turkish Turner Syndrome Study Group**
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- XLVI. **Clinical characteristics and molecular genetic analysis of 22 patients with neonatal diabetes from the South-Eastern region of Turkey: predominance of non-K-ATP channel mutations**
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- XLVII. **Turner Syndrome and Associated Problems in Turkish Children: A Multicenter Study**
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- XLVIII. **Normosmic idiopathic hypogonadotropic hypogonadism due to a novel homozygous nonsense c.C969A (p.Y323X) mutation in the KISS1R gene in three unrelated families**
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- XLIX. **Capillary Bedside Blood Glucose Measurement in Neonates: Missing a Diagnosis of Galactosemia**
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- L. **Prepubertal Unilateral Gynecomastia: Report of 2 Cases**
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- LI. **Familial Isolated Growth Hormone Deficiency Due to A Novel Homozygous Missense Mutation in the Growth Hormone Releasing Hormone Receptor Gene: Clinical Presentation With Hypoglycemia**
Demirbilek H., Tahir S., Baran R. T. , Sherif M., Shah P., Ozbek M. N. , Hatipoglu N., Baran A., Arya V. B. , Hussain K.
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- LII. **Phenotypic severity of homozygous GCK mutations causing neonatal or childhood-onset diabetes is primarily mediated through effects on protein stability**
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- LIII. **Long-Term Follow-Up of Children With Congenital Hyperinsulinism on Octreotide Therapy**
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- LVI. **Persistent hyperinsulinaemic hypoglycaemia in infancy**
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- LVII. **Oncologic manifestations in children with neurofibromatosis type 1 in Turkey**
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- LVIII. **Incidence of Type 1 Diabetes Mellitus in Turkish Children from the Southeastern Region of the Country: A Regional Report**
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- LIX. **Prevalence of type 1 diabetes mellitus in school children 6-18 years old in Diyarbakir, Southeastern Anatolian Region of Turkey**
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- LX. **Multiple Pituitary Hormone Deficiency Due to Gunshot Injury in a 6-Year-Old Girl**
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- LXI. **17 beta-Hydroxysteroid dehydrogenase type 3 deficiency as a result of a homozygous 7 base pair deletion in 17 beta HSD3 gene**
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- LXII. **Evaluation of serum kisspeptin levels in girls in the diagnosis of central precocious puberty and in the assessment of pubertal suppression**
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Atıflar

Toplam Atıf Sayısı (WOS):530

h-ineksi (WOS):14