

Assoc. Prof. HÜSEYİN DEMİRBILEK

Personal Information

Email: huseyindr@hacettepe.edu.tr

Other Email: dr_huseyin@hotmail.com

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

Education Information

Post Doctorate of Medicine, Hacettepe Üniversitesi, Hacettepe Tıp Fakültesi, Turkey 2006 - 2010

Expertise In Medicine, Hacettepe Üniversitesi, Hacettepe Tıp Fakültesi, Turkey 1999 - 2004

Under Graduate, Hacettepe Üniversitesi, Tıp Fakültesi, Turkey 1991 - 1998

Research Areas

Health Sciences, Natural Sciences

Academic Titles / Tasks

Associate Professor, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 2014 - Continues

Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

- 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.
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- 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**
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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**
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- VIII. **A novel diagnostic tool for the evaluation of hypothalamic-pituitary region and diagnosis of growth hormone deficiency: pons ratio**
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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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- X. **Clinical characteristics and long term follow up of 17 patients with permanent neonatal diabetes due to PTF1A distal enhancer mutations**
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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**
DEMİRBILEK H., Galcheva S., VURALLI KARAOĞLAN D., Al-Khawaga S., Hussain K.
INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES, vol.20, 2019 (Journal Indexed in SCI)
- 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**
Baran R. T. , Baran S. O. , Toraman N. F. , Filiz S., DEMİRBILEK H.
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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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- 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.

NEUROENDOCRINOLOGY LETTERS, vol.40, pp.36-40, 2019 (Journal Indexed in SCI)

- XIX. TUBULOINTERSTITIAL NEPHRITIS AND IGA NEPHROPATHY ASSOCIATED WITH VALPROATE USE: A CASE REPORT**
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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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- XXV. Clinical Characterstics, Genotype-phenotype Correlations and Follow Up of Patients with Congenital Hyperinsulinaemic Hypoglycaemia; Single Center Experience from a Southeastern City of Turkey**
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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**
Ozbek M. N. , Karasin N. D. , DEMİR BİLEK H., Demiral M., Baran R. T. , GÜRAN T.
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- XXIX. Hyperinsulinaemic hypoglycaemia in children and adults**
Shah P., Rahman S. A. , DEMİR BİLEK H., Guemes M., Hussain K.
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- XXX. Evaluation of vitamin D levels in patients with acute rheumatic fever**
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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**
Onan S. H. , Demirbilek H., Aldudak B., Bilici M., Demir F., Yilmazer M. M.
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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., Tatli 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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- XLIII. **Rare Causes of Primary Adrenal Insufficiency: Genetic and Clinical Characterization of a Large Nationwide Cohort**
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- XLVII. **Turner Syndrome and Associated Problems in Turkish Children: A Multicenter Study**
Yesilkaya E., BEREKET A., Darendeliler F., BAŞ F., POYRAZOĞLU Ş., Aydin B. K. , DARCAN Ş., Dundar B., Buyukinan M., Kara C., et al.
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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**
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- LIII. **Long-Term Follow-Up of Children With Congenital Hyperinsulinism on Octreotide Therapy**
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- LV. **Pancreatic Endocrine and Exocrine Function in Children following Near-Total Pancreatectomy for Diffuse Congenital Hyperinsulinism**
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- LVII. **Oncologic manifestations in children with neurofibromatosis type 1 in Turkey**
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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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- LXIII. **Vitamin D-deficient rickets mimicking ankylosing spondylitis in an adolescent girl**
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- LXV. **Hypophosphatasia Presenting with Pyridoxine-Responsive Seizures, Hypercalcemia, and Pseudotumor Cerebri: Case Report**
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