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Education Information

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

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

Undergraduate, Hacettepe University, Tıp Fakültesi, Turkey 1991 - 1998

Research Areas

Health Sciences, Natural Sciences

Academic Titles / Tasks

Associate Professor, Hacettepe University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 2014 - Continues

Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Author's reply "Comment on diagnostic utility of the average peak LH levels measured during GnRH stimulation test"**
Koca S. B., DEMİRBILEK H.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, 2024 (SCI-Expanded)
- II. **Diagnostic utility of the average peak LH levels measured during GnRH stimulation test**
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- III. **Severe Early-Onset Obesity and Diabetic Ketoacidosis due to a Novel Homozygous c.169C>T p.Arg57*Variant in <i>CEP19</i> Gene**
Cayir A., TÜRKYILMAZ A., Rabenstein H., Guven F., Karagoz Y. S., VURALI KARAOĞLAN D., Wabitsch M., DEMİRBILEK H.
MOLECULAR SYNDROMOLOGY, no.2, pp.104-113, 2024 (SCI-Expanded)
- IV. **Type A insulin resistance syndrome due to a novel heterozygous c.3486_3503del (p.Arg1163_Ala1168del) INSR gene mutation in an adolescent girl and her mother**
Koca S. B., Kulali M. A., Göğüş B., DEMİRBILEK H.
Archives of Endocrinology and Metabolism, vol.68, 2024 (SCI-Expanded)
- V. **Evaluation Of Long-term Height and Pubertal Outcome Of Boys Presented With Delayed Puberty Due To Constitutional Delay In Growth And Puberty And Isolated Hypogonadotropic Hypogonadism**

DOĞAN Ö., VURALI KARAOĞLAN D., DEMİRBILEK H.

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- VI. **Clinical characteristics, molecular genetics analysis results and long-term follow-up of a large cohort of congenital hyperinsulinism from Turkey: A nationwide cross-sectional study**
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- VII. **Metreleptin Treatment in a Boy with Congenital Generalized Lipodystrophy due to Homozygous c.465_468delGACT (p.T156Rfs*8) Mutation in the BSCL2 Gene: Results From the First-year**
Özalkak Ş., Demiral M., Ünal E., Taş F. F., Onay H., DEMİRBILEK H., Özbek M. N.
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- VIII. **Serum kisspeptin, neurokinin B and inhibin B levels can be used as alternative parameters to distinguish idiopathic CPP from premature thelarche in the early stages of puberty**
Vuralli D., Ciftci N., DEMİRBILEK H.
Clinical Endocrinology, vol.98, no.6, pp.788-795, 2023 (SCI-Expanded)
- IX. **Incidence and Risk Factors of Hyperglycemia in Severe Multisystem Inflammatory Syndrome in Children: A Retrospective Case-Control Study**
SARITAŞ NAKİP Ö., KESİCİ S., BOZKURT B. S., ÖZSÜREKÇİ Y., DEMİRBILEK H., BAYRAKÇI B.
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- X. **Managing Severe Hypoglycaemia in Patients with Diabetes: Current Challenges and Emerging Therapies**
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- XI. **Results from a Global, Multi-Center, Phase 2b Study (RIZE) in Congenital Hyperinsulinism: Characterization of a High Unmet Treatment Need and Glycemic Response to RZ358**
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- XII. **Demographic, Clinical, Hormonal And Genetic Characteristics Of Children And Adolescents With Congenital Adrenal Hyperplasia Due To 11-Beta Hydroxylase Deficiency**
Kendirici H. N. P., BAYRAMOĞLU E., Aycan Z., HATİPOĞLU N., Ucakturk S. A., Ozalkak S., ÖZSU E., Akbas E. D., Aydin M., Dundar I., et al.
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- XIII. **Serum kisspeptin, neurokinin B and inhibin B levels can be used as an auxiliary parameter to distinguish idiopathic CPP from premature thelarche in the early stages of puberty**
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- XIV. **Severe early-onset obesity and diabetic ketoacidosis due to a novel homozygous c.169C > T p.Arg57*mutation in CEP19 gene**
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- XV. **Genotype, phenotype characteristics and long-term follow-up of patients with Vitamin D Dependent Rickets Type IA (VDDR1a): A nationwide multicentre retrospective cross-sectional study**
Cayir A., DEMİRBILEK H., TÜRKYILMAZ A., DEMİRCİOĞLU S., BEREKET A., Darendeliler F., Ozbek M. N., Unal E., Okdemir D., Esen I., et al.
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- XVI. **The effects of the covid-19 pandemic on puberty: a cross-sectional, multicenter study from Turkey**
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ITALIAN JOURNAL OF PEDIATRICS, vol.48, no.1, 2022 (SCI-Expanded)
- XVII. **RZ358 in Congenital Hyperinsulinism: Results from a Multi-Center, Global, Phase 2b Study (RIZE)**

- Thornton P, DEMİRBILEK H, Christesen H. T., Melikian M, Galcheva S, Dastamani A, Roberts B, Hood D., O'Boyle E., Raskin J., et al.
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- XVIII. Revisiting the Annual Incidence of Type 1 Diabetes Mellitus in Children from the Southeastern Anatolian Region of Turkey: A Regional Report**
Ozalkak S, Yildirim R., Tunc S., Unal E., Tas F. F., DEMİRBILEK H., Ozbek M. N.
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- XIX. Long-term follow-up of transient neonatal diabetes mellitus due to a novel homozygous c.7734C>T (p.R228C) mutation in ZFP57 gene: relapse at prepubertal age**
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- XX. Clinical and genetic heterogeneity of HNF4A/HNF1A mutations in a multicentre paediatric cohort with hyperinsulinaemic hypoglycaemia**
McGlacken-Byrne S. M., Mohammad J. K., Conlon N., Gubaeva D., Siersbaek J., Schou A. J., DEMİRBILEK H., Dastamani A., Houghton J. A. L., Brusgaard K., et al.
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- XXI. Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics**
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- XXII. Clinical, biochemical, and echocardiographic evaluation of neonates with vitamin D deficiency due to maternal vitamin D deficiency**
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- XXIII. A rare and preventable aetiology of neurodevelopmental delay and epilepsy: familial glucocorticoid deficiency**
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- XXIV. Clinical and Hormonal Profiles Correlate With Molecular Characteristics in Patients With 11 beta-Hydroxylase Deficiency**
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- XXV. 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.**
Sherif M., Demirbilek H., Cayir A., Tahir S., Cavdarli B., Demiral M., Cebeci A., Vuralli D., Rahman S., Unal E., et al.
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- XXVI. Frequency of Celiac Disease and Spontaneous Normalization Rate of Celiac Serology in Children and Adolescent Patients with Type 1 Diabetes.**
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- XXVII. Clinical characteristics and molecular genetic analysis of a cohort with idiopathic congenital hypogonadism**
Turkyilmaz A., Cayir A., Yarali O., Kurnaz E., Baykan E. K., Ates E. A., DEMİRBILEK H.
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- XXVIII. CORRELATION OF STRENGTH, MOBILITY AND BONE MINERAL DENSITY IN DUCHENNE MUSCULAR DYSTROPHY**
Baran R. T., Kutluk M. G., Toraman N. F., Filiz M. B., Filiz S., DEMİRBILEK H.
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- XXX. **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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- XXXI. **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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- XXXII. **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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- XXXIII. **Validity of Six Month L-Thyroxine Dose for Differentiation of Transient or Permanent Congenital Hypothyroidism**
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- XXXIV. **A novel diagnostic tool for the evaluation of hypothalamic-pituitary region and diagnosis of growth hormone deficiency: pons ratio**
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- XXXV. **Clinical, biochemical and echocardiographic evaluation of patients with congenital rickets due to maternal vitamin D deficiency**
Cayir A., Akyigit A., Gullu U. U., Kahveci H., Yildiz D., Kurnaz E., Vuralli D., Kaya A., DEMİRBILEK H.
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.163, 2019 (SCI-Expanded)
- XXXVI. **A novel approach for the evaluation of hypothalamic-pituitary region in patients with 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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- XXXVII. **Clinical characteristics and long term follow up of 17 patients with permanent neonatal diabetes due to PTF1A distal enhancer mutations**
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- XXXIX. **Ion Transporters, Channelopathies, and Glucose Disorders**
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- XL. **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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- XLII. Congenital Hyperinsulinism and Evolution to Sulfonylurea-responsive Diabetes Later in Life due to a Novel Homozygous p.L171F ABCC8 Mutation**
Isik E, DEMİR BİLEK H, Houghton J. A., Ellard S., Flanagan S. E., Hussain K.
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- XLIV. A Distinct Clinical Phenotype in Two Siblings with X-linked Adrenoleukodystrophy**
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- XLV. Systemic Pseudohypoaldosteronism Type 1 due to 3 Novel Mutations in SCNN1A and SCNN1B Genes**
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- XLVI. TUBULOINTERSTITIAL NEPHRITIS AND IGA NEPHROPATHY ASSOCIATED WITH VALPROATE USE: A CASE REPORT**
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- XLVII. Evaluation of psychological characteristics of Turkish children with type 1 diabetes mellitus from two demographically and geographically distinct regions**
Baran R. T., Surer-Adanir A., Karakurt M. N., Dundar M., Aydin M., Ozbek M. N., Demirebilek H.
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- L. Clinical Characteristics, 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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- L. Systemic Pseudohypoaldosteronism Type 1 due to 3 Novel Mutations in SCNN1A and SCNN1B Genes; Report of 3 Cases**
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- LI. 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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- LII. Comprehensive Genetic Testing Shows One in Five Children with Diabetes and Non-Autoimmune Extra-Pancreatic Features Have Monogenic Aetiology**
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- LIV. **Congenital Hyperinsulinism: Diagnosis and Treatment Update**
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- LV. **Hyperinsulinaemic hypoglycaemia in children and adults**
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- LVI. **Evaluation of vitamin D levels in patients with acute rheumatic fever**
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- LVII. **Clinical and biochemical characteristics and bone mineral density of homozygous, compound heterozygous and heterozygous carriers of three novel IGFALS mutations**
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- LIX. **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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- LX. **Bone Mineral Density in Adolescent Girls with Hypogonadotropic and Hypergonadotropic Hypogonadism**
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- LXI. **A novel ALMS1 homozygous mutation in two Turkish brothers with Alstrom syndrome**
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- LXII. **Anthropometric findings from birth to adulthood and their relation with karyotype distribution in Turkish girls with Turner syndrome**
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- LXIII. **Severe Hypercalcemia in a Child With Acute Lymphoblastic Leukemia Relapse: Successful Management With Combination of Calcitonin and Bisphosphonate**
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- LXIV. **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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- LXV. **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**
Ozbek M. N., Demirbilek H., Baysal B., Baran R. T., Haliloglu B., Ocal M.
HORMONE RESEARCH IN PAEDIATRICALS, vol.86, pp.242, 2016 (SCI-Expanded)
- LXVI. **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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Non Academic Experience

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