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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 (İngilizce), Dahili Tıp Bilimleri Bölümü, 2014 - Devam Ediyor

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayımlanan Makaleler

- I. Author's reply "Comment on 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.
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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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- 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**
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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**
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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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- XVI. **The effects of the covid-19 pandemic on puberty: a cross-sectional, multicenter study from Turkey**
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- XVII. **RZ358 in Congenital Hyperinsulinism: Results from a Multi-Center, Global, Phase 2b Study (RIZE)**

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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**
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- XX. **Clinical and genetic heterogeneity of HNF4A/HNF1A mutations in a multicentre paediatric cohort with hyperinsulinaemic hypoglycaemia**
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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.**
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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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- XXVIII. **CORRELATION OF STRENGTH, MOBILITY AND BONE MINERAL DENSITY IN DUCHENNE MUSCULAR DYSTROPHY**
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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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- 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**
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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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- XLI. **Congenital Hyperinsulinism and Evolution to Sulfonylurea-responsive Diabetes Later in Life due to a Novel Homozygous p.L171F ABCC8 Mutation**
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- XLII. The Genetic and Molecular Mechanisms of Congenital Hyperinsulinism**
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- XLIII. A Distinct Clinical Phenotype in Two Siblings with X-linked Adrenoleukodystrophy**
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- XLIV. Systemic Pseudohypoadosteronism Type 1 due to 3 Novel Mutations in SCNN1A and SCNN1B Genes**
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- XLV. TUBULOINTERSTITIAL NEPHRITIS AND IGA NEPHROPATHY ASSOCIATED WITH VALPROATE USE: A CASE REPORT**
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- XLVI. Evaluation of psychological characteristics of Turkish children with type 1 diabetes mellitus from two demographically and geographically distinct regions**
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- XLIX. 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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- L. Systemic Pseudohypoadosteronism 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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- 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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- 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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- 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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