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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

Undergraduate, 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. **Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics**
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- II. **Clinical and Hormonal Profiles Correlate With Molecular Characteristics in Patients With 11 beta-Hydroxylase Deficiency**
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- III. **Frequency of Celiac Disease and Spontaneous Normalization Rate of Celiac Serology in Children and Adolescent Patients with Type 1 Diabetes.**
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- IV. **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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- V. **Clinical characteristics and molecular genetic analysis of a cohort with idiopathic congenital hypogonadism**
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- VII. **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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- VIII. **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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- IX. **Validity of Six Month L-Thyroxine Dose for Differentiation of Transient or Permanent Congenital Hypothyroidism**
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- X. **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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- XV. **Clinical, biochemical and echocardiographic evaluation of patients with congenital rickets due to maternal vitamin D deficiency**
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- XVII. **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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- NIGERIAN JOURNAL OF CLINICAL PRACTICE, vol.22, no.4, pp.539-545, 2019 (Journal Indexed in SCI)
- XVIII. Congenital Hyperinsulinism and Evolution to Sulfonylurea-responsive Diabetes Later in Life due to a Novel Homozygous p.L171F ABCC8 Mutation**
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- XXII. TUBULOINTERSTITIAL NEPHRITIS AND IGA NEPHROPATHY ASSOCIATED WITH VALPROATE USE: A CASE REPORT**
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- XXIII. Evaluation of psychological characteristics of Turkish children with type 1 diabetes mellitus from two demographically and geographically distinct regions**
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- XXVIII. 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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- XXXII. **Hyperinsulinaemic hypoglycaemia in children and adults**
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- XXXIV. **Clinical and biochemical characteristics and bone mineral density of homozygous, compound heterozygous and heterozygous carriers of three novel IGFALS mutations**
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- XXXVII. **Bone Mineral Density in Adolescent Girls with Hypogonadotropic and Hypergonadotropic Hypogonadism**
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- XXXVIII. **A novel ALMS1 homozygous mutation in two Turkish brothers with Alstrom syndrome**
Laxer C., Rahman S. A. , Sherif M., Tahir S., Cayir A., Demirbilek H., Hussain K.
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- XXXIX. **Anthropometric findings from birth to adulthood and their relation with karyotype distribution in Turkish girls with Turner syndrome**
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- XL. **Severe Hypercalcemia in a Child With Acute Lymphoblastic Leukemia Relapse: Successful Management With Combination of Calcitonin and Bisphosphonate**
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- XLI. **Rare Causes of Primary Adrenal Insufficiency: Genetic and Clinical Characterization of a Large Nationwide Cohort**
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- XLII. **Potential Role of Vitamin D in Pathogenesis of Acute Rheumatic Fever**
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- XLIII. **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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- XLV. **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**
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- L. **Normosmic idiopathic hypogonadotropic hypogonadism due to a novel homozygous nonsense c.C969A (p.Y323X) mutation in the KISS1R gene in three unrelated families**
Demirbilek H., Ozbek M. N. , Demir K., KOTAN L. D. , Cesur Y., Dogan M., Temiz F., Mengen E., GÜRBÜZ F., YÜKSEL B., et al.
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- LI. **Turner Syndrome and Associated Problems in Turkish Children: A Multicenter Study**
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- LII. **Capillary Bedside Blood Glucose Measurement in Neonates: Missing a Diagnosis of Galactosemia**
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- LIV. **Phenotypic severity of homozygous GCK mutations causing neonatal or childhood-onset diabetes is primarily mediated through effects on protein stability**
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- LV. Prepubertal Unilateral Gynecomastia: Report of 2 Cases**
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- LXI. Prevalence of type 1 diabetes mellitus in school children 6-18 years old in Diyarbakir, Southeastern Anatolian Region of Turkey**
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- LXII. Multiple Pituitary Hormone Deficiency Due to Gunshot Injury in a 6-Year-Old Girl**
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- LXIV. 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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- LXV. 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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