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Yoksis Araştırmacı ID: 200423

Eğitim Bilgileri

- I. Lisans Yandal, Marmara Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Türkiye 2001 - 2004
- II. Lisans, Marmara Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Türkiye 1995 - 2000
- III. Ön Lisans, Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Türkiye 1988 - 1995

Yabancı Diller

- I. İngilizce, C1 İleri

Araştırma Alanları

Sağlık Bilimleri

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayımlanan Makaleler

- I. Expanding the Clinical Features of Schimke Immuno-Osseous Dysplasia: A New Patient with a Novel Variant and Novel Clinical Findings.**
Alavanda C., Demir Ş., Güven S., Eltan M., Bilgiç Eltan S., Sefer A. P., Pul S., Güran T., Alpaz H., Arman A., et al.
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- II. A global survey on the use of the international classification of diseases codes for metabolic dysfunction-associated fatty liver disease**
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Hepatology International, cilt.18, sa.4, ss.1178-1201, 2024 (SCI-Expanded)
- III. Evaluating breast ultrasonography as a complementary diagnostic method in girls with central precocious puberty**
BIYIKLI E., Helvacioğlu D., BUĞDAYCI O., Tosun B. G., DEMİRCİOĞLU S., GÜRAN T., BEREKET A.
Pediatric Radiology, cilt.54, sa.7, ss.1156-1167, 2024 (SCI-Expanded)
- IV. Development of external genitalia during mini-puberty: is it related to somatic growth or reproductive hormones?**
Gacemer H. A., Tosun B. G., Helvacioğlu D., Yaman A., Abali Z. Y., HALILOĞLU B., DEMİRCİOĞLU S., HAKLAR G., BEREKET A., GÜRAN T.
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- V. **Venous Thrombosis in a Pseudohypoparathyroidism Patient with a Novel GNAS Frameshift Mutation and Complete Resolution of Vascular Calcifications with Acetazolamide Treatment.**
Menevse T. S., Iwasaki Y., Abali Z. Y., Tosun B. G., Helvacioğlu D., DOĞRU Ö., BUĞDAYCI O., Cyr S. M., GÜRAN T., BEREKET A., et al.
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- VI. **Challenges in the management of a 7 years old child with thyrotropin-secreting pituitary adenoma and the review of the literature**
KIRKGÖZ T., Abali S., Seker A., GÜRPINAR TOSUN B., ELTAN M., Helvacioğlu D., HALILOĞLU B., KAYGUSUZ S. B., Yavas Abali Z., SEVEN MENEVŞE T., et al.
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- VII. **Two new cases with novel pathogenic variants reflecting the clinical diversity of Schaaf-Yang syndrome**
ALAVANDA C., Arslan Ateş E., Yavaş Abalı Z., GEÇKİNLİ B. B., DEMİRCİOĞLU S., ARMAN A.
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- VIII. **Decline in the Age of Menarche in Istanbul Schoolgirls Over the Last 12 Years**
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- IX. **Molecular analysis of MKRN3 gene in Turkish girls with sporadic and familial idiopathic central**
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- X. **Osteopetrosis: Gene-based nosology and significance dysosteosclerosis.**
Turan S.
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- XI. **Liraglutide for Weight Management in Children and Adolescents With Prader-Willi Syndrome and Obesity**
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- XII. **Basal cortisol measurements in the prediction of low-dose ACTH stimulation test outcomes**
Gacemer H., Gürpınar Tosun B., Abali Z. Y., Helvacioğlu D., Haliloğlu B., Demircioğlu S., Bereket A., Güran T.
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- XIII. **A Case of Short Stature Presenting with Multiple Exocytosis**
Kaygusuz S. B., Gokoglu M., DEMİRCİOĞLU S.
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- XIV. **Low-dose ACTH Stimulation Test: Comparison of Cortisol Response at 30, 40, and 60 Minutes**
Gürpınar Tosun B., Arıkan H., Demircioğlu S., Bereket A., Güran T.
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- XV. **A rare cause of monogenic obesity: Schaaf-Yang syndrome due to a novel MAGEL2 gene variant**
Abali Z. Y., Ates E. A., GÜRAN T., BEREKET A., DEMİRCİOĞLU S.
HORMONE RESEARCH IN PAEDIATRICS, cilt.95, sa.SUPPL 2, ss.230, 2022 (SCI-Expanded)
- XVI. **Single Nucleotide Polymorphisms (SNPs) Profile as Predictive Markers of Lifestyle Modification Outcomes in Pediatric Obesity Treatment**
Gawlik A., Sobalska-Kwapis M., Antosz A., Strapagiel D., Seweryn M., Shmoish M., BEREKET A., Wasniewska M., KIRKGÖZ T., DEMİRCİOĞLU S., et al.
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- XVII. **Circulating mRNA and miRNA Signatures as Predictive Markers of Lifestyle Modification Outcomes in Pediatric Obesity Treatment**
Gawlik A., Shmoish M., BEREKET A., Wasniewska M., Antosz A., KIRKGÖZ T., DEMİRCİOĞLU S., GÜRAN T., Aversa T., Corica D., et al.
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- XVIII. **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., DEMİRBİLEK H., TÜRKYILMAZ A., DEMİRCİOĞLU S., BEREKET A., Darendeliler F., Ozbek M. N., Unal E., Okdemir D., Esen I., et al.
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- XIX. **Diagnostic Features and Risk Factors for Childhood Thyroid Cancers**
ŞAHİN P., GÜRPINAR TOSUN B., YUMUŞAKHUYLU A. C., GÜRAN T., Helvacioğlu D., Abali Z. Y., HALİLOĞLU B., OYSU Ç., BEREKET A., DEMİRCİOĞLU S.
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- XX. **Glucagon response to hypoglycemia during extended oral glucose tolerance test in children with cystic fibrosis and comparing with healthy peers**
HALİLOĞLU B., SEVEN MENEVŞE T., GÜRPINAR TOSUN B., GÜRAN T., DEMİRCİOĞLU S., İspir T., GÖKDEMİR Y., ERDEM ERALP E., BEREKET A.
HORMONE RESEARCH IN PAEDIATRICS, cilt.95, sa.SUPPL 2, ss.199-200, 2022 (SCI-Expanded)
- XXI. **Etiological analysis of hypophosphatemia: A single-center experience**
Eltan M., Alavanda C., Abali Z. Y., Bayramoğlu E., Kaygusuz S. B., Helvacioğlu D., Tosun B. G., Menevşe T. S., Ata P., Guran T., et al.
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- XXII. **Wide phenotypical spectrum with the same karyotype: Mixed gonadal dysgenesis**
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- XXIII. **Change of menarcheal age in schoolgirls living in Istanbul over the last 12 years**
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- XXIV. **Differences due to the variant type in the inheritance pattern of BMP15 gene-related primary ovarian insufficiency: a girl with a homozygous null BMP15 gene variant**
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- XXV. **Breast ultrasonography: How useful in the diagnosis of precocious puberty?**
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- XXVI. **Adrenal steroids reference ranges in infancy determined by LC-MS/MS**
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- XXVII. **Homozygosity for a novel INHA mutation in two male siblings with hypospadias, primary hypogonadism, and high normal testicular volume**
Guran T., Ates E. A., Eltan M., Sahin B., Tosun B. G., Seven Menevşe T., Geckinli B. B., Greenfield A., Turan S., Bereket A.
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- XXVIII. **Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis Due to CLDN16 Gene Mutations: Novel Findings in Two Cases with Diverse Clinical Features**
Eltan M., Abali Z. Y., Turkyilmaz A., Gökçe İ., Abali S., Alavanda C., Arman A., Kirkgöz T., Güran T., Hatun S., et al.
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- XXIX. **Steroid hormone profiles and molecular diagnostic tools in pediatric patients with non-CAH primary adrenal insufficiency.**
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- XXX. **A novel deletion involving the first GNAS exon encoding G α causes PHP1A without methylation**

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- XXXI. **Homozygosity for a novel INHA mutation in two male siblings with hypospadias, primary hypogonadism, and high-normal testicular volume.**
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- XXXII. **Efficacy of the Novel Degludec/Aspart Insulin Co-formulation in Children and Adolescents with Type 1 Diabetes: A Real-life Experience with 1-year IDeg/Asp Therapy in Poorly Controlled and Non-compliant Patients.**
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- XXXIII. **Lack of GNAS re-methylation during oogenesis may be a cause of sporadic pseudohypoparathyroidism type Ib (PHP1B).**
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- XXXIV. **Long-term efficacy of T3 analogue Triac in children and adults with MCT8 deficiency: a real-life retrospective cohort study.**
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- XXXV. **Catch-up growth and discontinuation of fludrocortisone treatment in aldosterone synthase deficiency.**
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- XXXVI. **Broad-spectrum XX and XY gonadal dysgenesis in patients with a homozygous L193S variant in PPP2R3C.**
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- XXXVII. **Non-hormonal Clitoromegaly due to Clitoral Priapism Caused by Appendicitis/Appendectomy.**
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- XXXVIII. **Dysgenesis and Dysfunction of the Pancreas and Pituitary Due to FOXA2 Gene Defects.**
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- XXXIX. **Severe Hypophosphatemic Rickets due to Tumor-Induced Osteomalasia**
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- XL. **Genotypic Sex and Severity of the Disease Determine the Time of Clinical Presentation in Steroid 17 alpha-Hydroxylase/17,20-Lyase Deficiency**
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- XLI. **Is quail egg a potential endocrine disruptor?**
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- XLII. **Correlation of 11-oxygenated C19 androgens with the clinical and biochemical characteristics in**

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- XLIII. **46,XY DSD due to biallelic DHX37 gene mutations**
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- XLIV. **Clinical and Hormonal Profiles Correlate With Molecular Characteristics in Patients With 11 beta-Hydroxylase Deficiency**
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- XLV. **Biallelic PPP2R3C mutations are associated with partial and complete gonadal dysgenesis in 46,XY and 46,XX individuals**
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- XLVI. **Acetazolamide treatment in a patient with pseudohypoparathyroidism with venous calcification**
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- XLVII. **Machine Learning Quest for Predictive Markers of Lifestyle Modification Outcomes in Pediatric Obesity Treatment**
Gawlik A., Shmoish M., BEREKET A., Wasniewska M., Antosz A., Kirkgoz T., DEMİRCİOĞLU S., GÜRAN T., Aversa T., Corica D., et al.
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- XLVIII. **Adrenocortical hormone profiles do not predict the molecular etiology in non-CAH primary adrenal insufficiency**
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- XLIX. **A rare cause of hypercalcemia: Congenital Lactase Deficiency**
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- L. **Endocrine disrupting chemicals and bone.**
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- LI. **Hyperinsulinemic hypoglycemia due to biallelic mutations in the DNAJC3 gene**
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- LII. **Clinical and hormonal evolution of aldosterone synthase deficiency: Is complete remission possible?**
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- LIII. **Rahman Syndrome: Hypopituitarism might be responsible for changing height pattern**
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- LIV. **Cranial MRI Abnormalities and Long-term Follow-up of the Lesions in 770 Girls With Central Precocious Puberty.**
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- LV. **Does Genotype–Phenotype Correlation Exist in Vitamin D-Dependent Rickets Type IA: Report of 13**

New Cases and Review of the Literature

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- LVI. **Use of Insulin Degludec/Insulin Aspart in the Management of Diabetes Mellitus: Expert Panel Recommendations on Appropriate Practice Patterns**
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- LVII. **Cinacalcet as a First-Line Treatment in Neonatal Severe Hyperparathyroidism Secondary to Calcium Sensing Receptor (CaSR) Mutation**
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- LVIII. **Persistent Mullerian Duct Syndrome: A Rare But Important Etiology of Inguinal Hernia and Cryptorchidism**
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SEXUAL DEVELOPMENT, cilt.13, ss.264-270, 2020 (SCI-Expanded)
- LIX. **Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients**
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- LX. **A Rare Cause of Hypophosphatemia: Raine Syndrome Changing Clinical Features with Age**
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- LXI. **Management of Systemic Hypersensitivity Reactions to Gonadotropin-Releasing Hormone Analogues during Treatment of Central Precocious Puberty**
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- LXII. **Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study**
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- LXIII. **Clinical Significance of Hypophosphatasemia in Children**
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