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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. 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)
- II. 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., HALİLOĞLU B., DEMİRCİOĞLU S., HAKLAR G., BEREKET A., GÜRAN T.
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- III. 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., HALİLOĞLU B., KAYGUSUZ S. B., Yavas Abali Z., SEVEN MENEVŞE T., et al.
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- IV. Two new cases with novel pathogenic variants reflecting the clinical diversity of Schaaf-Yang syndrome**
ALAVANDA C., Arslan Ateş E., Yavaş Abali Z., GEÇKİNLİ B. B., DEMİRCİOĞLU S., ARMAN A.
Clinical Genetics, cilt.104, sa.1, ss.127-132, 2023 (SCI-Expanded)

- V. **Decline in the Age of Menarche in Istanbul Schoolgirls Over the Last 12 Years**
GÜRAN T., HELVACIOĞLU D., TOSUN B. G., ABALI Z. Y., Alır F., Arslan Y. t., Molla G., Şahin B., Sayar M. E., Atay Z., et al.
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- VI. **Molecular analysis of MKRN3 gene in Turkish girls with sporadic and familial idiopathic central**
KIRKGÖZ T., KAYGUSUZ S. B., ALAVANDA C., Helvacioğlu D., Abali Z. Y., GÜRPINAR TOSUN B., ELTAN M., SEVEN MENEVŞE T., GÜRAN T., ARMAN A., et al.
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- VII. **Osteopetrosis: Gene-based nosology and significance dysosteosclerosis.**
Turan S.
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- VIII. **Liraglutide for Weight Management in Children and Adolescents With Prader-Willi Syndrome and Obesity**
Diene G., Angulo M., Hale P. M., Jepsen C. H., Hofman P. L., Hokken-Koelega A., Ramesh C., DEMİRCİOĞLU S., Tauber M.
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- IX. **Basal cortisol measurements in the prediction of low-dose ACTH stimulation test outcomes**
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- X. **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., Menevse T. S., Ata P., Guran T., et al.
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- XI. **Change of menarcheal age in schoolgirls living in Istanbul over the last 12 years**
Güran T., Alır F., Arslan Y. T., Molla G., Şahin B., Sayar M. E., Atay Z., Helvacioğlu D., Gürpınar Tosun B., Haliloğlu B., et al.
HORMONE RESEARCH IN PAEDIATRICS, cilt.95, sa.SUPPL 2, ss.364, 2022 (SCI-Expanded)
- XII. **Breast ultrasonography: How useful in the diagnosis of precocious puberty?**
Helvacioğlu D., BIYIKLI E., BUĞDAYCI O., DEMİRCİOĞLU S., GÜRAN T., BEREKET A.
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- XIII. **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., Ispir T., GÖKDEMİR Y., ERDEM ERALP E., BEREKET A.
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- XIV. **Low-dose ACTH Stimulation Test: Comparison of Cortisol Response at 30, 40, and 60 Minutes**
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- XV. **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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- XVI. **A Case of Short Stature Presenting with Multiple Exocytosis**
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- XVII. **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.
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- XVIII. **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.,

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- XXIX. **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**
Abali Z. Y., Ates E. A., ELTAN M., GÜRPINAR TOSUN B., BEREKET A., GÜRAN T., DEMİRCİOĞLU S.
HORMONE RESEARCH IN PAEDIATRICS, cilt.95, sa.SUPPL 2, ss.377-378, 2022 (SCI-Expanded)
- XX. **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.
HORMONE RESEARCH IN PAEDIATRICS, cilt.95, sa.SUPPL 2, ss.407, 2022 (SCI-Expanded)
- XXI. **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., TÜRKİYILMAZ A., DEMİRCİOĞLU S., BEREKET A., Darendeliler F., Ozbek M. N., Unal E., Okdemir D., Esen I., et al.
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- XXII. **Wide phenotypical spectrum with the same karyotype: Mixed gonadal dysgenesis**
Seven Menevşe T., Gürpınar Tosun B., Helvacioğlu D., Abali Z. Y., Kirmizibekmez H., Dursun F., Demircioğlu S., Bereket A., Güran T.
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- XXIII. **Adrenal steroids reference ranges in infancy determined by LC-MS/MS**
Enver E. O., Vatansever P., Guran O., Bilgin L., Boran P., Demircioğlu S., Haklar G., Bereket A., Güran T.
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- XXIV. **Homozygosity for a novel INHA mutation in two male siblings with hypospadias, primary hypogonadism, and high normal testicular volume**
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- XXV. **Steroid hormone profiles and molecular diagnostic tools in pediatric patients with non-CAH primary adrenal insufficiency.**
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- XXVI. **A novel deletion involving the first GNAS exon encoding G α causes PHP1A without methylation changes at exon A/B**
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- XXVII. **Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis Due to CLDN16 Gene Mutations: Novel Findings in Two Cases with Diverse Clinical Features**
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- XXVIII. **Homozygosity for a novel INHA mutation in two male siblings with hypospadias, primary hypogonadism, and high-normal testicular volume.**
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- XXIX. **Lack of GNAS re-methylation during oogenesis may be a cause of sporadic pseudohypoparathyroidism type 1b (PHP1B).**
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- XXX. **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.

Kirkgoz T., Eltan M., Kaygusuz S. B., Yavas Abali Z., Helvacioğlu D., Seven Menevşe T., Gürpınar Tosun B., Guran T., Bereket A., Turan S.

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XXXI. Long-term efficacy of T3 analogue Triac in children and adults with MCT8 deficiency: a real-life retrospective cohort study.

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XXXII. Broad-spectrum XX and XY gonadal dysgenesis in patients with a homozygous L193S variant in PPP2R3C.

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XXXIII. Catch-up growth and discontinuation of fludrocortisone treatment in aldosterone synthase deficiency.

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XXXIV. Non-hormonal Clitoromegaly due to Clitoral Priapism Caused by Appendicitis/Appendectomy.

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XXXV. Dysgenesis and Dysfunction of the Pancreas and Pituitary Due to FOXA2 Gene Defects.

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XXXVI. Rahman Syndrome: Hypopituitarism might be responsible for changing height pattern

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XXXVII. Biallelic PPP2R3C mutations are associated with partial and complete gonadal dysgenesis in 46,XY and 46,XX individuals

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XXXVIII. Adrenocortical hormone profiles do not predict the molecular etiology in non-CAH primary adrenal insufficiency

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XXXIX. Endocrine disrupting chemicals and bone.

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XL. A rare cause of hypercalcemia: Congenital Lactase Deficiency

Eltan M., Alavanda C., Abalı S., Abalı Z. Y., Kaygusuz S. B., Gürpınar Tosun B., Seven Menevşe T., Helvacioğlu D., Guran T., Ata P., et al.

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XLI. Clinical and hormonal evolution of aldosterone synthase deficiency: Is complete remission possible?

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XLII. Clinical and Hormonal Profiles Correlate With Molecular Characteristics in Patients With 11 beta-Hydroxylase Deficiency

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- XLIII. **Severe Hypophosphatemic Rickets due to Tumor-Induced Osteomalasia**
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- XLIV. **Is quail egg a potential endocrine disruptor?**
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- XLV. **Acetazolamide treatment in a patient with pseudohypoparathyroidism with venous calcification**
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- XLVI. **46,XY DSD due to biallelic DHX37 gene mutations**
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- XLVIII. **Genotypic Sex and Severity of the Disease Determine the Time of Clinical Presentation in Steroid 17 alpha-Hydroxylase/17,20-Lyase Deficiency**
Kurnaz E., Baykan E. K., Turkyılmaz A., Yaralı O., Abali Z. Y., Demircioğlu S., Bereket A., Cayir A., Güran T.
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- XLIX. **Correlation of 11-oxygenated C19 androgens with the clinical and biochemical characteristics in premature adrenarcho**
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- L. **Hyperinsulinemic hypoglycemia due to biallelic mutations in the DNAJC3 gene**
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- LI. **Cranial MRI Abnormalities and Long-term Follow-up of the Lesions in 770 Girls With Central Precocious Puberty.**
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- LII. **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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- LIII. **Use of Insulin Degludec/Insulin Aspart in the Management of Diabetes Mellitus: Expert Panel Recommendations on Appropriate Practice Patterns**
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- LIV. **Cinacalcet as a First-Line Treatment in Neonatal Severe Hyperparathyroidism Secondary to Calcium Sensing Receptor (CaSR) Mutation**
Gulcan-Kersin S., Kirkgoz T., Eltan M., Rzayev T., Ata P., Bilgen H. S., Özek E., Bereket A., Turan S.
HORMONE RESEARCH IN PAEDIATRICS, cilt.93, sa.5, ss.313-321, 2020 (SCI-Expanded)
- LV. **Persistent Mullerian Duct Syndrome: A Rare But Important Etiology of Inguinal Hernia and Cryptorchidism**

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- LVI. **Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients**
Mantovani G., Bastepe M., Monk D., De Sanctis L., Thiele S., Ahmed S. F., Bufo R., Choplin T., De Filippo G., Devernois G., et al.
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- LVII. **A Rare Cause of Hypophosphatemia: Raine Syndrome Changing Clinical Features with Age**
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- LVIII. **Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study**
Groeneweg S., Van Geest F. S., Abacı A., Alcantud A., Ambegaonkar G. P., Armour C. M., Bakhtiani P., Barca D., Bertini E. S., Van Beynum I. M., et al.
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- LIX. **Management of Systemic Hypersensitivity Reactions to Gonadotropin-Releasing Hormone Analogues during Treatment of Central Precocious Puberty**
Kirkgöz T., Karakoc-Aydiner E., Bugrul F., Yavas Abali Z., Helvacioğlu D., Kiykim A., Bilgic Eltan S., Aruci Kasap N., Baris S., Ozen A. O., et al.
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- LX. **Clinical Significance of Hypophosphatasemia in Children**
Bayramli R., Cevlik T., Güran T., Atay Z., Bas S., Haklar G., Bereket A., Demircioğlu S.
CALCIFIED TISSUE INTERNATIONAL, cilt.106, sa.6, ss.608-615, 2020 (SCI-Expanded)
- LXI. **Nationwide Turkish Cohort Study of Hypophosphatemic Rickets**
Şıklar Z., Demircioğlu S., Bereket A., Bas F., Güran T., Akberzade A., Abacı A., Demir K., Böber E., Ozbek M. N., et al.
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- LXII. **Once-Weekly Somapacitan vs Daily GH in Children With GH Deficiency: Results From a Randomized Phase 2 Trial**
Sävendahl L., Battelino T., Brod M., Rasmussen M. H., Horikawa R., Juul R. V., Saenger P., Furthner D., Piringner B., Auer-Hackenberg L., et al.
JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, cilt.105, sa.4, 2020 (SCI-Expanded)
- LXIII. **Hereditary vitamin D-resistant rickets: a report of four cases with two novel variants in the VDR gene and successful use of intermittent intravenous calcium via a peripheral route**
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