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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. **Three Birds With One Stone: Successful Management Of Peritonitis Induced Pediatric Septic Shock With Peritoneal Washing**
DEMİR O. O., Erdemir G. N., KESİCİ S., BOYBEYİ TÜNER Ö., ÖZSÜREKÇİ Y., DEMİRBİLEK H.
KLINISCHE PADIATRIE, 2024 (SCI-Expanded)
- II. **Author's reply "Comment on diagnostic utility of the average peak LH levels measured during GnRH stimulation test"**
Koca S. B., DEMİRBİLEK H.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, 2024 (SCI-Expanded)
- III. **Diagnostic utility of the average peak LH levels measured during GnRH stimulation test**
Koca S. B., DEMİRBİLEK H.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, no.9, pp.773-778, 2024 (SCI-Expanded)
- IV. **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., VURALLI KARAOĞLAN D., Wabitsch M., DEMİRBİLEK H.
MOLECULAR SYNDROMOLOGY, no.2, pp.104-113, 2024 (SCI-Expanded)
- V. **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)

- 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**
DEMİRBILEK H., Ozbek M. N., Yildiz M., Houghton J. L. A., Onal H., GÜRBÜZ F., Cetinkaya S., Cayir A., DENKBOY ÖNGEN Y., PARLAK M., et al.
HORMONE RESEARCH IN PAEDIATRICS, pp.589, 2023 (SCI-Expanded)
- VII. **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.
HORMONE RESEARCH IN PAEDIATRICS, pp.387, 2023 (SCI-Expanded)
- VIII. **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.
Journal of clinical research in pediatric endocrinology, vol.15, no.3, pp.329-333, 2023 (SCI-Expanded)
- IX. **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)
- X. **Managing Severe Hypoglycaemia in Patients with Diabetes: Current Challenges and Emerging Therapies**
DEMİRBILEK H., Vuralli D., Haris B., Hussain K.
Diabetes, Metabolic Syndrome and Obesity: Targets and Therapy, vol.16, pp.259-273, 2023 (SCI-Expanded)
- XI. **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.
Journal of Pediatric Infectious Diseases, vol.18, no.1, pp.31-37, 2023 (SCI-Expanded)
- XII. **Demographic, Clinical, Hormonal And Genetic Characteristics Of Children And Adolescents With Congenital Adrenal Hyperplasia Due To 11-Beta Hydroxylase Deficiency**
Kendirci 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.
HORMONE RESEARCH IN PAEDIATRICS, vol.95, no.SUPPL 2, pp.119-120, 2022 (SCI-Expanded)
- XIII. **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.
HORMONE RESEARCH IN PAEDIATRICS, vol.95, no.SUPPL 2, pp.100, 2022 (SCI-Expanded)
- XIV. **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**
DEMİRBILEK H., Melikyan M., Galcheva S., Dastamani A., Thornton P., De Leon D., Raskin J., Roberts B., Hood D., O'Boyle E., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.95, no.SUPPL 2, pp.32-33, 2022 (SCI-Expanded)
- XV. **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**
VURALI KARAOĞLAN D., Ciftci N., DEMİRBILEK H.
HORMONE RESEARCH IN PAEDIATRICS, vol.95, no.SUPPL 2, pp.62-63, 2022 (SCI-Expanded)
- XVI. **Severe early-onset obesity and diabetic ketoacidosis due to a novel homozygous c.169C > T p.Arg57*mutation in CEP19 gene**
Cayir A., Turkyilmaz A., Rabenstein H., GÜVEN F., Karagoz Y. S., Wabitsch M., DEMİRBILEK H.
HORMONE RESEARCH IN PAEDIATRICS, vol.95, no.SUPPL 2, pp.216, 2022 (SCI-Expanded)
- XVII. **The effects of the covid-19 pandemic on puberty: a cross-sectional, multicenter study from Turkey**

- Mutlu G. Y., Eviz E., Haliloglu B., Kirmizibekmez H., Dursun F., Ozalkak S., Cayir A., Sacli B. Y., Ozbek M. N., DEMİRBILEK H., et al.
ITALIAN JOURNAL OF PEDIATRICS, vol.48, no.1, 2022 (SCI-Expanded)
- XVIII. **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.
HORMONE RESEARCH IN PAEDIATRICS, vol.95, no.SUPPL 1, pp.265-266, 2022 (SCI-Expanded)
- XIX. **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.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.14, no.2, pp.172-178, 2022 (SCI-Expanded)
- XX. **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**
Kontbay T., Atar M., DEMİRBILEK H.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, pp.695-698, 2022 (SCI-Expanded)
- XXI. **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.
EUROPEAN JOURNAL OF ENDOCRINOLOGY, vol.186, no.4, pp.417-427, 2022 (SCI-Expanded)
- XXII. **Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics**
Patel K. A., Ozbek M. N., Yildiz M., GÜRAN T., Kocyigit C., ACAR S., ŞIKLAR Z., Atar M., Colclough K., Houghton J., et al.
DIABETOLOGIA, vol.65, no.2, pp.336-342, 2022 (SCI-Expanded)
- XXIII. **Clinical, biochemical, and echocardiographic evaluation of neonates with vitamin D deficiency due to maternal vitamin D deficiency**
Cayir A., Akyigit A., Gullu U. U., Kahveci H., Yildiz D., Kurnaz E., VURALLI KARAOĞLAN D., Kaya A., Buyukyilmaz G., DEMİRBILEK H.
CARDIOLOGY IN THE YOUNG, vol.32, no.1, pp.88-93, 2022 (SCI-Expanded)
- XXIV. **A rare and preventable aetiology of neurodevelopmental delay and epilepsy: familial glucocorticoid deficiency**
Ozbek M. N., Demiral M., Unal E., Karasin N. D., Baran R. T., DEMİRBILEK H.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.11, pp.1463-1468, 2021 (SCI-Expanded)
- XXV. **Clinical and Hormonal Profiles Correlate With Molecular Characteristics in Patients With 11 beta-Hydroxylase Deficiency**
YILDIZ M., Isik E., Abali Z. Y., KESKİN M., Ozbek M. N., Bas F., Ucakturk S. A., Buyukinan M., Onal H., Kara C., et al.
JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, vol.106, no.9, 2021 (SCI-Expanded)
- XXVI. **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.
Journal of clinical research in pediatric endocrinology, vol.13, pp.34-43, 2021 (SCI-Expanded)
- XXVII. **Frequency of Celiac Disease and Spontaneous Normalization Rate of Celiac Serology in Children and Adolescent Patients with Type 1 Diabetes.**
Unal E., Demiral M., Baysal B., Agın M., Devocioğlu E., Demirbilek H., Özbek M.
Journal of clinical research in pediatric endocrinology, vol.13, pp.72-79, 2021 (SCI-Expanded)
- 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.
ACTA MEDICA MEDITERRANEA, vol.37, no.6, pp.3647-3654, 2021 (SCI-Expanded)
- XXIX. **Clinical characteristics and molecular genetic analysis of a cohort with idiopathic congenital**

hypogonadism

Turkylmaz A, Cayir A, Yarali O, Kurnaz E, Baykan E. K, Ates E. A., DEMİRBILEK H.

JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.6, pp.771-780, 2021 (SCI-Expanded)

- XXX. **Clinical Characteristics and Long-term Follow-up of Patients with Diabetes Due To PTF1A Enhancer Mutations.**
Demirbilek H., Cayir A., Flanagan S., Yıldırım R., Kor Y., Gurbuz F., Haliloğlu B., Yıldız M., Baran R., Akbas E., et al.
The Journal of clinical endocrinology and metabolism, vol.105, 2020 (SCI-Expanded)
- XXXI. **Neonatal diabetes due to homozygousINSgene promoter mutations: Highly variable phenotype, remission and early relapse during the first 3 years of life**
Demiral M., DEMİRBILEK H., Celik K., Okur N., Hussain K., Ozbek M. N.
PEDIATRIC DIABETES, vol.21, no.7, pp.1169-1175, 2020 (SCI-Expanded)
- XXXII. **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**
Demiral M., Unal E., Baysal B., Baran R. T., DEMİRBILEK H., Ozbek M. N.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.12, no.3, pp.295-302, 2020 (SCI-Expanded)
- XXXIII. **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**
Demiral M., DEMİRBILEK H., Unal E., Durmaz C. D., Ceylaner S., Ozbek M. N.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.12, no.3, pp.319-328, 2020 (SCI-Expanded)
- XXXIV. **Validity of Six Month L-Thyroxine Dose for Differentiation of Transient or Permanent Congenital Hypothyroidism**
Asena M., Demiral M., Unal E., Ocal M., DEMİRBILEK H., Ozbek M. N.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.12, no.3, pp.275-280, 2020 (SCI-Expanded)
- XXXV. **A novel diagnostic tool for the evaluation of hypothalamic-pituitary region and diagnosis of growth hormone deficiency: pons ratio**
Demiral M., Karaca M. S., Unal E., Baysal B., Baran R. T., DEMİRBILEK H., Ozbek M. N.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.33, no.6, pp.735-742, 2020 (SCI-Expanded)
- XXXVI. **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 PAEDIATRICALS, vol.91, pp.163, 2019 (SCI-Expanded)
- XXXVII. **Clinical characteristics and long term follow up of 17 patients with permanent neonatal diabetes due to PTF1A distal enhancer mutations**
DEMİRBILEK H., Cayir A., DeFranco E., Kor Y., Yildiz M., Yildirim R., Baran R. T., Demiral M., Haliloglu B., Flanagan S. E., et al.
HORMONE RESEARCH IN PAEDIATRICALS, vol.91, pp.230, 2019 (SCI-Expanded)
- XXXVIII. **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.
HORMONE RESEARCH IN PAEDIATRICALS, vol.91, pp.253-254, 2019 (SCI-Expanded)
- XXXIX. **Ectopic posterior pituitary, polydactyly, midfacial hypoplasia and panhypopituitarism due to a novel heterozygous IVS 11-2AC(c.1957-2AC) mutation in GLI2 gene**
Demiral M., Unal E., Kardas B., DEMİRBILEK H., Ozbek M. N.
HORMONE RESEARCH IN PAEDIATRICALS, vol.91, pp.173, 2019 (SCI-Expanded)
- XL. **Ion Transporters, Channelopathies, and Glucose Disorders**
DEMİRBILEK H., Galcheva S., VURALLI KARAOĞLAN D., Al-Khawaga S., Hussain K.
INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES, vol.20, no.10, 2019 (SCI-Expanded)
- XLI. **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

Baran R. T., Baran S. O., Toraman N. F., Filiz S., DEMİRBILEK H.

NIGERIAN JOURNAL OF CLINICAL PRACTICE, vol.22, no.4, pp.539-545, 2019 (SCI-Expanded)

- XLII. **Congenital Hyperinsulinism and Evolution to Sulfonylurea-responsive Diabetes Later in Life due to a Novel Homozygous p.L171F ABCC8 Mutation**
Isik E., DEMİRBILEK H., Houghton J. A., Ellard S., Flanagan S. E., Hussain K.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.11, no.1, pp.82-87, 2019 (SCI-Expanded)
- XLIII. **The Genetic and Molecular Mechanisms of Congenital Hyperinsulinism**
Galcheva S., DEMİRBILEK H., Al-Khawaga S., Hussain K.
FRONTIERS IN ENDOCRINOLOGY, vol.10, 2019 (SCI-Expanded)
- XLIV. **Systemic Pseudohypoadosteronism Type 1 due to 3 Novel Mutations in SCNN1A and SCNN1B Genes**
Cayir A., Demirelli Y., Yildiz D., Kahveci H., Yarali O., Kurnaz E., VURALLI KARAOĞLAN D., DEMİRBILEK H.
HORMONE RESEARCH IN PAEDIATRICALS, vol.91, no.3, pp.175-185, 2019 (SCI-Expanded)
- XLV. **A Distinct Clinical Phenotype in Two Siblings with X-linked Adrenoleukodystrophy**
Ozdemir Kutbay N., Ozbek M. N., Sarer Yurekli B., DEMİRBILEK H.
NEUROENDOCRINOLOGY LETTERS, vol.40, no.1, pp.36-40, 2019 (SCI-Expanded)
- XLVI. **TUBULOINTERSTITIAL NEPHRITIS AND IGA NEPHROPATHY ASSOCIATED WITH VALPROATE USE: A CASE REPORT**
Celegen K., Saglam A., Koca S. B., DEMİRBILEK H., DÜZOVA A.
PEDIATRIC NEPHROLOGY, vol.33, no.10, pp.1974, 2018 (SCI-Expanded)
- 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., Demirbilek H.
TURKISH JOURNAL OF PEDIATRICALS, vol.60, no.5, pp.554-561, 2018 (SCI-Expanded)
- XLVIII. **Permanent neonatal diabetes mellitus and neurological abnormalities due to a novel homozygous missense mutation in NEUROD1**
DEMİRBILEK H., HATİPOĞLU N., Gul U., Tati Z. U., Ellard S., Flanagan S. E., De Franco E., Kurtoglu S.
PEDIATRIC DIABETES, vol.19, no.5, pp.898-904, 2018 (SCI-Expanded)
- XLIX. **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**
Baran R. T., Baran S., Toraman N. F., Filiz M. B., Filiz S., DEMİRBILEK H.
HORMONE RESEARCH IN PAEDIATRICALS, vol.90, pp.305, 2018 (SCI-Expanded)
- L. **Clinical Characteristics, Genotype-phenotype Correlations and Follow Up of Patients with Congenital Hyperinsulinaemic Hypoglycaemia; Single Center Experience from a Southeastern City of Turkey**
Ozbek M. N., DEMİRBILEK H., Haliloglu B., Demiral M., Baran R. T., Ellard S., Houghton J., Flanagan S. E., Hussain K.
HORMONE RESEARCH IN PAEDIATRICALS, vol.90, pp.358, 2018 (SCI-Expanded)
- LI. **Systemic Pseudohypoadosteronism Type 1 due to 3 Novel Mutations in SCNN1A and SCNN1B Genes; Report of 3 Cases**
Cayir A., Demirelli Y., Yildiz D., Kahveci H., Yarali O., Karaoglan D. V., Kurnaz E., DEMİRBILEK H.
HORMONE RESEARCH IN PAEDIATRICALS, vol.90, pp.366-367, 2018 (SCI-Expanded)
- LII. **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**
Yaman B., Celegen K., Korkmaz E., Lafci N., Balik Z., DEMİRBILEK H., DÜZOVA A.
HORMONE RESEARCH IN PAEDIATRICALS, vol.90, pp.461-462, 2018 (SCI-Expanded)
- LIII. **Comprehensive Genetic Testing Shows One in Five Children with Diabetes and Non-Autoimmune Extra-Pancreatic Features Have Monogenic Aetiology**
Patel K. A., Colclough K., Ozbek M. N., Yildiz M., GÜRAN T., Kocyigit C., Acar S., Siklar Z., Atar M., Johnson M. B., et al.
HORMONE RESEARCH IN PAEDIATRICALS, vol.90, pp.213, 2018 (SCI-Expanded)

- LIV. **Presenting Features, Clinical Characteristics and Follow Up of Familial Isolated Glucocorticoid Deficiency (FGD) Due to Mutations in MC2R and MRAP Genes**
Ozbek M. N., Karasin N. D., DEMİRBILEK H., Demiral M., Baran R. T., GÜRAN T.
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.161-162, 2018 (SCI-Expanded)
- LV. **Congenital Hyperinsulinism: Diagnosis and Treatment Update**
DEMİRBILEK H., Hussain K.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.9, pp.69-87, 2017 (SCI-Expanded)
- LVI. **Hyperinsulinaemic hypoglycaemia in children and adults**
Shah P., Rahman S. A., DEMİRBILEK H., Guemes M., Hussain K.
LANCET DIABETES & ENDOCRINOLOGY, vol.5, no.9, pp.729-742, 2017 (SCI-Expanded)
- LVII. **Evaluation of vitamin D levels in patients with acute rheumatic fever**
Onan S. H., Demirbilek H., Aldudak B., Bilici M., Demir F., Yilmazer M. M.
ANATOLIAN JOURNAL OF CARDIOLOGY, vol.18, no.1, pp.75-76, 2017 (SCI-Expanded)
- LVIII. **Clinical and biochemical characteristics and bone mineral density of homozygous, compound heterozygous and heterozygous carriers of three novel IGFALS mutations**
Isik E., Haliloglu B., van Doorn J., DEMİRBILEK H., Scheltinga S. A., Losekoot M., Wit J. M.
EUROPEAN JOURNAL OF ENDOCRINOLOGY, vol.176, no.6, pp.657-667, 2017 (SCI-Expanded)
- LIX. **PSYCHOMETRIC ANALYSIS USING CHILD BEHAVIOR CHECKLIST (CBCL) IN TYPE 1 DIABETIC TURKISH CHILDREN FROM TWO DISTINCT DEMOGRAPHIC AND GEOGRAPHICAL REGIONS**
Baran R. T., Adanir A. S., Karakurt M. N., Dundar M., Aydin M., Ozbek M. N., DEMİRBILEK H.
HORMONE RESEARCH IN PAEDIATRICS, vol.88, pp.603-604, 2017 (SCI-Expanded)
- LX. **Evaluation of therapeutics management patterns and glycemic control of pediatric type 1 diabetes mellitus patients in Turkey: A nationwide cross-sectional study**
Hatun S., Demirbilek H., DARCAN Ş., Yuksel A., Binay C., Simsek D. G., Kara C., Cetinkaya E., Unuvar T., Ucakurk A., et al.
DIABETES RESEARCH AND CLINICAL PRACTICE, vol.119, pp.32-40, 2016 (SCI-Expanded)
- LXI. **Bone Mineral Density in Adolescent Girls with Hypogonadotropic and Hypergonadotropic Hypogonadism**
Ozbek M. N., Demirbilek H., Baran R. T., Baran A.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.8, no.2, pp.163-169, 2016 (SCI-Expanded)
- LXII. **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.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.29, no.5, pp.585-589, 2016 (SCI-Expanded)
- LXIII. **Anthropometric findings from birth to adulthood and their relation with karyotype distribution in Turkish girls with Turner syndrome**
Sari E., BEREKET A., Yesilkaya E., BAŞ F., Bundak R., Aydin B. K., DARCAN Ş., Dundar B., Buyukinan M., Kara C., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.170, no.4, pp.942-948, 2016 (SCI-Expanded)
- LXIV. **Severe Hypercalcemia in a Child With Acute Lymphoblastic Leukemia Relapse: Successful Management With Combination of Calcitonin and Bisphosphonate**
Tagiyev A., DEMİRBILEK H., Tavit B., Buyukyilmaz G., Gumruk F., Cetin M.
JOURNAL OF PEDIATRIC HEMATOLOGY ONCOLOGY, vol.38, no.3, pp.232-234, 2016 (SCI-Expanded)
- LXV. **Severe Hyponatremia and Repeated Intestinal Resections for Intestinal Dysmotility Mimicking Congenital Aganglionic Megacolon due to Delay in the Diagnosis of Congenital Hypothyroidism**
Buyukyilmaz G., Baltu D., SOYER T., Tanyildiz M., DEMİRBILEK H.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.519, 2016 (SCI-Expanded)
- LXVI. **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 PAEDIATRICS, vol.86, pp.242, 2016 (SCI-Expanded)
- LXVII. **Kallmann Syndrome Due to a Homozygous Missense c.217C > T (p.R73C) Mutation Detected in the**

Exon-2 of the PROK2 Gene

Ozturk M. N., Demirbilek H., KOTAN L. D., Baysal B., Ocal M., TOPALOĞLU A. K.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.431-432, 2016 (SCI-Expanded)

LXVIII. Potential Role of Vitamin D in Pathogenesis of Acute Rheumatic Fever

Onan S. H., Demirbilek H., Aldudak B., Bilici M., Demir F., Yilmazer M. M.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.191-192, 2016 (SCI-Expanded)

LXIX. Genotype and Phenotype Characteristics in 22 Patients with Vitamin D-Dependent Rickets Type I

Tahir S., Demirbilek H., Ozbek M. N., Baran R. T., Tanriverdi S., Hussain K.
HORMONE RESEARCH IN PAEDIATRICS, vol.85, no.5, pp.309-317, 2016 (SCI-Expanded)

LXX. 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

HATİPOĞLU N., DEMİRBILEK H., Gul U., Tati Z. U., Flanagan S., Ellard S., De Franco E., Kurtoglu S.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.204-205, 2016 (SCI-Expanded)

LXXI. Rare Causes of Primary Adrenal Insufficiency: Genetic and Clinical Characterization of a Large Nationwide Cohort

GÜRAN T., Buonocore F., Saka N., Ozbek M. N., Aycan Z., BEREKET A., BAŞ F., DARCAN Ş., Bideci A., Guven A., et al.
JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, vol.101, no.1, pp.283-291, 2016 (SCI-Expanded)

LXXII. Growth curves for Turkish Girls with Turner Syndrome: Results of the Turkish Turner Syndrome Study Group

Darendeliler F., Yesilkaya E., BEREKET A., BAŞ F., Bundak R., Sari E., Aydin B. K., DARCAN Ş., Dundar B., Buyukinan M., et al.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.7, no.3, pp.183-191, 2015 (SCI-Expanded)

LXXIII. Clinical characteristics and molecular genetic analysis of 22 patients with neonatal diabetes from the South-Eastern region of Turkey: predominance of non-K-ATP channel mutations

Demirbilek H., Arya V. B., Nuri M., Houghton J. A. L., Baran R. T., Akar M., Tekes S., Tuzun H., Mackay D. J., Flanagan S. E., et al.
EUROPEAN JOURNAL OF ENDOCRINOLOGY, vol.172, no.6, pp.697-705, 2015 (SCI-Expanded)

LXXIV. Turner Syndrome and Associated Problems in Turkish Children: A Multicenter Study

Yesilkaya E., BEREKET A., Darendeliler F., BAŞ F., POYRAZOĞLU Ş., Aydin B. K., DARCAN Ş., Dundar B., Buyukinan M., Kara C., et al.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.7, no.1, pp.27-36, 2015 (SCI-Expanded)

LXXV. Capillary Bedside Blood Glucose Measurement in Neonates: Missing a Diagnosis of Galactosemia

Ozbek M. N., Ocal M., Tanriverdi S., Baysal B., Deniz A., Oncel K., DEMİRBILEK H.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.7, no.1, pp.83-85, 2015 (SCI-Expanded)

LXXVI. 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.
CLINICAL ENDOCRINOLOGY, vol.82, no.3, pp.429-438, 2015 (SCI-Expanded)

LXXVII. Phenotypic severity of homozygous GCK mutations causing neonatal or childhood-onset diabetes is primarily mediated through effects on protein stability

Raimondo A., Chakera A. J., Thomsen S. K., Colclough K., Barrett A., De Franco E., Chatelas A., Demirbilek H., Akcay T., Alawneh H., et al.
HUMAN MOLECULAR GENETICS, vol.23, no.24, pp.6432-6440, 2014 (SCI-Expanded)

LXXVIII. Familial Isolated Growth Hormone Deficiency Due to A Novel Homozygous Missense Mutation in the Growth Hormone Releasing Hormone Receptor Gene: Clinical Presentation With Hypoglycemia

Demirbilek H., Tahir S., Baran R. T., Sherif M., Shah P., Ozbek M. N., Hatipoglu N., Baran A., Arya V. B., Hussain K.
JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, vol.99, no.12, 2014 (SCI-Expanded)

LXXIX. Prepubertal Unilateral Gynecomastia: Report of 2 Cases

Demirbilek H., Bacak G., Baran R. T., Avci Y., Baran A., Keles A., Ozbek M. N., ALANAY Y., Hussain K.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.6, no.4, pp.250-253, 2014 (SCI-Expanded)

- LXXX. **Long-Term Follow-Up of Children With Congenital Hyperinsulinism on Octreotide Therapy**
Demirbilek H., Shah P., Arya V. B., Hinchey L., Flanagan S. E., Ellard S., Hussain K.
JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, vol.99, no.10, pp.3660-3667, 2014 (SCI-Expanded)
- LXXXI. **Clinical characteristics and phenotype-genotype analysis in Turkish patients with congenital hyperinsulinism; predominance of recessive K-ATP channel mutations**
Demirbilek H., Arya V. B., Ozbek M. N., AKINCI A., Dogan M., Demirel F., Houghton J., Kaba S., Guzel F., Baran R. T., et al.
EUROPEAN JOURNAL OF ENDOCRINOLOGY, vol.170, no.6, pp.885-892, 2014 (SCI-Expanded)
- LXXXII. **Pancreatic Endocrine and Exocrine Function in Children following Near-Total Pancreatectomy for Diffuse Congenital Hyperinsulinism**
Arya V. B., Senniappan S., Demirbilek H., Alam S., Flanagan S. E., Ellard S., Hussain K.
PLOS ONE, vol.9, no.5, 2014 (SCI-Expanded)
- LXXXIII. **Persistent hyperinsulinaemic hypoglycaemia in infancy**
Shah P., Demirbilek H., Hussain K.
SEMINARS IN PEDIATRIC SURGERY, vol.23, no.2, pp.76-82, 2014 (SCI-Expanded)
- LXXXIV. **Oncologic manifestations in children with neurofibromatosis type 1 in Turkey**
İNCECİK F., ALTUNBAŞAK Ş., HERGÜNER M., BAYRAM İ., KÜPELİ S., Demirbilek H.
TURKISH JOURNAL OF PEDIATRICS, vol.55, no.3, pp.266-270, 2013 (SCI-Expanded)
- LXXXV. **Multiple Pituitary Hormone Deficiency Due to Gunshot Injury in a 6-Year-Old Girl**
Demirbilek H., Ozbek M. N., Baran R. T., Baran A.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.5, no.3, pp.209-211, 2013 (SCI-Expanded)
- LXXXVI. **Incidence of Type 1 Diabetes Mellitus in Turkish Children from the Southeastern Region of the Country: A Regional Report**
Demirbilek H., Ozbek M. N., Baran R. T.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.5, no.2, pp.98-103, 2013 (SCI-Expanded)
- LXXXVII. **Prevalence of type 1 diabetes mellitus in school children 6-18 years old in Diyarbakir, Southeastern Anatolian Region of Turkey**
Demirbilek H., Ozbek M. N.
TURKISH JOURNAL OF MEDICAL SCIENCES, vol.43, no.5, pp.768-774, 2013 (SCI-Expanded)
- LXXXVIII. **17 beta-Hydroxysteroid dehydrogenase type 3 deficiency as a result of a homozygous 7 base pair deletion in 17 beta HSD3 gene**
ALİKAŞİFOĞLU A., Hiort O., Gonc N., DEMİRBILEK H., Isik E., KANDEMİR N.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.25, pp.561-563, 2012 (SCI-Expanded)
- LXXXIX. **Evaluation of serum kisspeptin levels in girls in the diagnosis of central precocious puberty and in the assessment of pubertal suppression**
DEMİRBILEK H., GÖNÇ E. N., Ozon A., ALİKAŞİFOĞLU A., KANDEMİR N.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.25, pp.313-316, 2012 (SCI-Expanded)
- XC. **Vitamin D-deficient rickets mimicking ankylosing spondylitis in an adolescent girl**
DEMİRBILEK H., Aydogdu D., Ozon A.
TURKISH JOURNAL OF PEDIATRICS, vol.54, no.2, pp.177-179, 2012 (SCI-Expanded)
- XCI. **Hypophosphatasia Presenting with Pyridoxine-Responsive Seizures, Hypercalcemia, and Pseudotumor Cerebri: Case Report**
DEMİRBILEK H., Alanay Y., ALİKAŞİFOĞLU A., TOPÇU M., Mornet E., Gonc N., Ozon A., KANDEMİR N.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.4, no.1, pp.34-38, 2012 (SCI-Expanded)
- XCII. **Distribution of Gene Mutations Associated with Familial Normosmic Idiopathic Hypogonadotropic Hypogonadism**
GÜRBÜZ F., KOTAN L. D., Mengen E., Siklar Z., Berberoglu M., Dokmetas S., Kilicli M. F., Guven A., Kirel B., Saka N., et al.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.4, no.3, pp.121-126, 2012 (SCI-Expanded)
- XCIII. **The role of the resistive index in Hashimoto's thyroiditis: a Sonographic pilot study in children**
Sarıkaya B., DEMİRBILEK H., AKATA D., KANDEMİR N.

CLINICS, vol.67, no.11, pp.1253-1257, 2012 (SCI-Expanded)

- XCIV. **Assessment of gonadotrophin suppression in girls treated with GnRH analogue for central precocious puberty; validity of single luteinizing hormone measurement after leuprolide acetate injection**
DEMİRBILEK H., ALİKAŞİFOĞLU A., Gonc N. E., Ozon A., KANDEMİR N.
CLINICAL ENDOCRINOLOGY, vol.76, no.1, pp.126-130, 2012 (SCI-Expanded)
- XCV. **GnRH stimulation Test in precocious puberty: Single sample is adequate for diagnosis and dose adjustment**
KANDEMİR N., DEMİRBILEK H., ÖZÖN Z. A., Gönç N., ALİKAŞİFOĞLU A.
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.3, no.1, pp.12-17, 2011 (SCI-Expanded)
- XCVI. **Two pediatric patients with Von Hippel-Lindau disease type 2b: from patient to screening, from screening to patient**
Gonc N., Engiz O., Neumann H. P. H., DEMİRBILEK H., Ozon A., ALİKAŞİFOĞLU A., KANDEMİR N.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.24, pp.109-112, 2011 (SCI-Expanded)
- XCVII. **Anemia and Neutropenic Fever with High Dose Diazoxide Treatment in a Case with Hyperinsulinism Due to Munchausen by Proxy**
Ozon A., DEMİRBILEK H., ERTUĞRUL A., ÜNAL S., GÜMRÜK F., KANDEMİR N.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.23, no.7, pp.719-723, 2010 (SCI-Expanded)
- XCVIII. **Assessment of thyroid function during the long course of Hashimoto's thyroiditis in children and adolescents**
DEMİRBILEK H., KANDEMİR N., GÖNÇ E. N., Ozon A., ALİKAŞİFOĞLU A.
CLINICAL ENDOCRINOLOGY, vol.71, no.3, pp.451-454, 2009 (SCI-Expanded)
- XCIX. **Single sample during gonadotropin stimulation test is adequate for the diagnosis of precocious puberty**
KANDEMİR N., DEMİRBILEK H., Ozon A., Gonc N., ALİKAŞİFOĞLU A.
HORMONE RESEARCH, vol.72, pp.336, 2009 (SCI-Expanded)
- C. **A clinical review of patients with 45 XO/46 XY genotype**
ALİKAŞİFOĞLU A., DEMİRBILEK H., Ozon A., Gonc N., Kandemir N.
HORMONE RESEARCH, vol.70, pp.156, 2008 (SCI-Expanded)
- CI. **Hashimoto's thyroiditis in children and adolescents: A retrospective study on clinical, epidemiological and laboratory properties of the disease**
DEMİRBILEK H., KANDEMİR N., GÖNÇ E. N., Ozon A., ALİKAŞİFOĞLU A., Yordam N.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.20, no.11, pp.1199-1205, 2007 (SCI-Expanded)
- CII. **Hypoglycaemia and hypothermia due to nimesulide overdose**
Yapakci E., Uysal O., Demirbilek H., Olgar S., Nacar N., Ozen H.
ARCHIVES OF DISEASE IN CHILDHOOD, vol.85, no.6, pp.510, 2001 (SCI-Expanded)

Articles Published in Other Journals

- I. **Continuous Hepatogonadal and Splenogonogal Fusion: A Rare Cause of Bilateral Infra-Abdominal Testis in an 18-Month-Old Boy**
Durmus G., BOYBEYİ TÜNER Ö., ÖZCAN H. N., GÖZMEN O., DEMİRBILEK H., SOYER T.
EUROPEAN JOURNAL OF PEDIATRIC SURGERY REPORTS, vol.10, no.01, 2022 (ESCI)
- II. **INTENSIVE PHYSICAL EXERCISE AND KETOSIS IN TYPE 1 DIABETES: LITERATURE REVIEW ON A CASE AFTER COVID-19 QUARANTINE**
Civil T., ÖZEN G., DEMİRBILEK H.
INTERNATIONAL JOURNAL OF LIFE SCIENCE AND PHARMA RESEARCH, pp.276-280, 2021 (ESCI)
- III. **Cystoscopy-Guided Laparoscopic Excision of Prostatic Utricle: Report of a Case**
Boybeyi-Turer O., DEMİRBILEK H., SOYER T.
EUROPEAN JOURNAL OF PEDIATRIC SURGERY REPORTS, vol.8, no.1, pp.35-38, 2020 (ESCI)

- IV. **Reversible Dilated Cardiomyopathy Due to Combination of Vitamin D-Deficient Rickets and Primary Hypomagnesemia in an 11-Month-Old Infant**
Yazici M. U., KESİCİ S., Demirbilek H., Tanyildiz M., Gumustas M., BAYRAKCI B.
JOURNAL OF PEDIATRIC INTENSIVE CARE, vol.7, no.1, pp.46-48, 2018 (ESCI)
- V. **A rare clinical condition in childhood comorbidity of idiopathic hypoparathyroidism and basal ganglia calcification: Fahr disease**
Baran R. T., DEMİRBILEK H., Baran S., Parlak M., Parlak E.
CUKUROVA MEDICAL JOURNAL, vol.43, no.1, pp.231-234, 2018 (ESCI)
- VI. **Diagnosis and treatment of hyperinsulinaemic hypoglycaemia and its implications for paediatric endocrinology**
DEMİRBILEK H., Rahman S. A., Buyukyilmaz G. G., Hussain K.
INTERNATIONAL JOURNAL OF PEDIATRIC ENDOCRINOLOGY, 2017 (Peer-Reviewed Journal)
- VII. **Severe hyponatremia and repeated intestinal resections for intestinal dysmotility mimicking congenital aganglionic megacolon due to delay in the diagnosis of congenital hypothyroidism.**
Buyukyilmaz G., Baltu D., Soyer T., Tanyıldız M., Demirbilek H.
Annals of pediatric endocrinology & metabolism, vol.21, pp.230-234, 2016 (Scopus)
- VIII. **Evaluation of Echocardiography and Holter Electrocardiography Findings in Patients with Mucopolysaccharidosis**
Aldudak B., Ozbek M. N., Demirbilek H., Saygi S., Celik M., Kangin M.
GUNCEL PEDIATRI-JOURNAL OF CURRENT PEDIATRICS, vol.13, no.3, pp.165-170, 2015 (ESCI)
- IX. **Combined nutritional anemia coexisting with microcytic anemia**
Yildirim A. T., Demirbilek H., Saygi S., Aliosmanoglu C., Soker M.
IZMIR DR BEHCET UZ COCUK HASTANESI DERGISI, vol.3, no.1, pp.44-48, 2013 (ESCI)
- X. **Neurofibromatosis Type 1 and Autoimmune Hyperthyroidism in a 10.5 Years-Old Girl**
Demirbilek H., Kupeli S., Ozbek M. N., Saygi S., Yildirim A. T.
CUKUROVA MEDICAL JOURNAL, vol.38, no.4, pp.805-808, 2013 (ESCI)
- XI. **A case of iatrogenic thyrotoxicosis presenting with malnutrition and fever of unknown origin**
Malnütrisyon ve tekrarlayan nedeni bilinmeyen ateş nedeni ile getirilen iatrojenik trikotoksikoz vakası
Arikan Z. T., Güder E., Aykan H., DEMİRBILEK H., Özön A., YALÇIN S. S.
Cocuk Sagligi ve Hastaliklari Dergisi, vol.51, no.2, pp.91-94, 2008 (Scopus)

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H-Index (Scopus): 14

Non Academic Experience

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