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Education Information

Post Doctorate of Medicine, Cukurova University, Tıp Fakültesi, Çocuk Endokrinolojisi, Turkey 2010 - 2013
Expertise In Medicine, Dışkapı Çocuk Eğitim Ve Araştırma Hastanesi , Çocuk Sağlığı Ve Hastalıkları, Çocuk Sağlığı Ve Hastalıkları , Turkey 2004 - 2009

Foreign Languages

English, B2 Upper Intermediate

Dissertations

Expertise In Medicine, Dikkat eksikliği ve hiperaktivite tanılı hastalarda metilfenidat kullanımının büyüme ve iştah üzerine etkisi, Cukurova University, Çocuk Endokrinolojisi, Çocuk Endokrinolojisi, 2013
Expertise In Medicine, AKUT GASTROENTERİT NEDENİYLE HASTANEYE YATAN HASTALARDA ETKENLER VE KLİNİK BULGULAR: EPİDEMİYOLOJİK ÇALIŞMA, Dışkapı Çocuk Eğitim Ve Araştırma Hastanesi, Çocuk Sağlığı Ve Hastalıkları, Çocuk Sağlığı Ve Hastalıkları , 2009

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Child Health and Diseases, Pediatric Endocrinology and Metabolism

Academic Titles / Tasks

Assistant Professor, Cukurova University, Tıp Fakültesi, Çocuk Sağlığı Ve Hastalıkları , 2018 - Continues
Assistant Professor, Cukurova University, Tıp Fakültesi, Çocuk Sağlığı Ve Hastalıkları , 2016 - 2018

Professional Experience

Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

- **21-Hydroxylase deficiency: Mutational spectrum and Genotype-Phenotype relations analyses by next-generation sequencing and multiplex ligation-dependent probe amplification**
  Turan I., Tastan M., Boga D. D. , GÜRBÜZ F., KOTAN L. D. , TULİ A., YÜKSEL B.
  EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.63, 2020 (Journal Indexed in SCI)
- **BMP4 mutations as a novel cause of normosmic hypogonadotropic hypogonadism**
  Topaloglu A. K. , Yildirim R., KOTAN L. D. , Akkus G., Unal E., Turan I., Dilek S., Tastan M., GÜRBÜZ F., YÜKSEL B.
  HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.113-114, 2019 (Journal Indexed in SCI)
- **Gender decision in disorders of sex development (DSD) patients: 20 years' experience**

GÜRBÜZ F., ALKAN M., GÜL ÇELİK G., BİŞGİN A., ÇEKİN N., TOPALOĞLU A. K. , ZORLUDEMİR Ü., AVCI A., YÜKSEL B.
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.333-334, 2019 (Journal Indexed in SCI)

- **Unusual and early onset IPEX syndrome: a case report**
  DOĞRUEL D., GÜRBÜZ F., TURAN İ., ALTINTAŞ D. U. , YILMAZ M., YÜKSEL B.
  TURKISH JOURNAL OF PEDIATRICS, vol.61, pp.580-584, 2019 (Journal Indexed in SCI)
- **Efficiency of Single Dose of Tolvaptan Treatment During the Triphasic Episode After Surgery for Craniopharyngioma**
  GÜRBÜZ F., Tastan M., TURAN İ., YÜKSEL B.
  JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.11, pp.202-206, 2019 (Journal Indexed in SCI)
- **Neonatal Screening for Congenital Adrenal Hyperplasia in Turkey: A Pilot Study with 38,935 Infants**
  GÜRAN T., Tezel B., GÜRBÜZ F., EKLİOĞLU B. S. , HATİPOĞLU N., Kara C., ŞİMŞEK E., ÇİZMECİOĞLU JONES F. M. , Ozon A., Bas F., et al.
  JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.11, pp.13-23, 2019 (Journal Indexed in SCI)
- **Hyperphosphatemic Familial Tumoral Calcinosis in Two Siblings with a Novel Mutation in <i>GALNT3</i> Gene: Experience from Southern Turkey**
  Kışla E., Gürbüz F., BALCI S., Bişgin A., TAŞTAN M., Yüksel B., Yılmaz M.
  Journal of clinical research in pediatric endocrinology, vol.11, pp.94-99, 2019 (Journal Indexed in SCI)
- **Prevalence and associated phenotypes of PLXNA1 variants in normosmic and anosmic idiopathic hypogonadotropic hypogonadism**
  KOTAN L. D. , Isik E., TURAN İ., Mengen E., AKKUŞ G., Tastan M., GÜRBÜZ F., YÜKSEL B., TOPALOĞLU A. K.
  CLINICAL GENETICS, vol.95, pp.320-324, 2019 (Journal Indexed in SCI)
- **Molecular genetic studies in a case series of isolated hypoaldosteronism due to biosynthesis defects or aldosterone resistance.**
  TURAN İ., Kotan L. D. , TASTAN M., GURBUZ F., TOPALOĞLU A. K. , YÜKSEL B.
  Clinical endocrinology, vol.88, pp.799-805, 2018 (Journal Indexed in SCI)
- **Novel inactivating mutations of the DCAF17 gene in American and Turkish families cause male infertility and female subfertility in the mouse model.**
  GURBUZ F., DESAI S., DIAO F., TURKKAHRAMAN D., WRANITZ F., WOOD-TRAGESER M., SHIN Y., Kotan L. D. , JIANG H., WITCHEL S., et al.
  Clinical genetics, vol.93, pp.853-859, 2018 (Journal Indexed in SCI)
- **Novel Inactivating Mutations of the DCAF17 gene in American and Turkish families cause male infertility and female subfertility in the mouse model**
  KOTAN L. D. , TOPALOĞLU A. K. , YÜKSEL B., GÜRBÜZ F., WOOD-TRAGESER M.
  CLINICAL GENETICS, vol.4, pp.853-859, 2018 (Journal Indexed in SCI)
- **A Turkish Family with 46,XY Disorder of Sex Development Due to 17b-Hydroxysteroid Dehydrogenase Type 3 Deficiency**
  GÜRBÜZ F., TURAN İ., Tastan M., YÜKSEL B.
  HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.582, 2018 (Journal Indexed in SCI)
- **CCDC141 Mutations in Idiopathic Hypogonadotropic Hypogonadism**
  TURAN İ., Hutchins B. I. , Hacihamdioglu B., KOTAN L. D. , GÜRBÜZ F., Ulubay A., Mengen E., YÜKSEL B., Wray S., TOPALOĞLU A. K.
  JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, vol.102, pp.1816-1825, 2017 (Journal Indexed in SCI)
- **Hypogonadotropic Hypogonadism due to Novel FGFR1 Mutations**
  AKKUŞ G., KOTAN L. D. , Durmaz E., Mengen E., TURAN İ., Ulubay A., GÜRBÜZ F., YÜKSEL B., Tetiker T., TOPALOĞLU A. K.
  JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.9, pp.95-100, 2017 (Journal Indexed in SCI)
- **THE EFFECT OF RECOMBINANT GROWTH HORMONE TREATMENT ON INTRAOCULAR PRESSURE IN CHILDREN WITH GROWTH HORMONE DEFICIENCY**

GÜRBÜZ F., ERDEM E., Mengen E., Simdivar G., YAĞMUR M., Ersoz R., TOPALOĞLU A. K. , YÜKSEL B.
HORMONE RESEARCH IN PAEDIATRICS, vol.88, pp.293, 2017 (Journal Indexed in SCI)

- **A CASE OF CONGENITAL GENERALIZED LIPODYSTROPHY TYPE 2 WITH NOVEL BSCL2 GENE MUTATION**
  GÜRBÜZ F., TURAN İ., Tastan M., TOPALOĞLU A. K. , YÜKSEL B.
  HORMONE RESEARCH IN PAEDIATRICS, vol.88, pp.141-142, 2017 (Journal Indexed in SCI)
- **A RARE CAUSE OF CONGENITAL ADRENAL HYPERPLASIA: CONGENITAL LIPOID ADRENAL HYPERPLASIA**
  GÜRBÜZ F., TURAN İ., TOPALOĞLU A. K. , YÜKSEL B.
  HORMONE RESEARCH IN PAEDIATRICS, vol.88, pp.566, 2017 (Journal Indexed in SCI)
- **A Novel Homozygous Mutation in the KCNJ11 Gene of a Neonate with Congenital Hyperinsulinism and Successful Management with Sirolimus.**
  ÜNAL S., GÖNÜLAL D., UÇAKTÜRK A., Siyah B., FLANAGAN S., Gürbüz F., TAYFUN M., ELMAOĞULLARı S., ARASLı A., DEMIREL F., et al.
  Journal of clinical research in pediatric endocrinology, vol.8, pp.478-481, 2016 (Journal Indexed in SCI)
- **Wolcott-Rallison Syndrome with Novel EIF2AK3 Gene Mutation.**
  Gürbüz F., Yüksel B., Topaloğlu A. K.
  Journal of clinical research in pediatric endocrinology, vol.8, pp.496-497, 2016 (Journal Indexed in SCI)
- **Crouzonodermoskeletal Syndrome with Hypoplasia of Corpus Callosum and Inferior Vermis.**
  Gürbüz F., CEYLANER S., Topaloğlu A. K. , Yüksel B.
  Journal of clinical research in pediatric endocrinology, vol.8, pp.373-4, 2016 (Journal Indexed in SCI)
- **Sertoli cell only syndrome with ambiguous genitalia.**
  Gurbuz F., CEYLANER S., Erdogan S., Topaloglu A. K. , Yuksel B.
  Journal of pediatric endocrinology & metabolism : JPEM, vol.29, pp.849-52, 2016 (Journal Indexed in SCI)
- **Idiopathic Hypogonadotropic Hypogonadism Caused by Inactivating Mutations in SRA1.**
  Kotan L. D. , COOPER C., DARCAN Ş., CARR I., ÖZEN S., YAN Y., HAMEDANI M., Gürbüz F., MENGEN E., TURAN İ., et al.
  Journal of clinical research in pediatric endocrinology, vol.8, pp.125-34, 2016 (Journal Indexed in SCI)
- **CCDC141 Mutation Identified in Anosmic Hypogonadotropic Hypogonadism (Kallmann Syndrome) Alters GnRH Neuronal Migration.**
  HUTCHINS B., Kotan L. D. , TAYLOR-BURDS C., OZKAN Y., CHENG P., Gurbuz F., TIONG J., MENGEN E., Yuksel B., Topaloglu A. K. , et al.
  Endocrinology, vol.157, pp.1956-66, 2016 (Journal Indexed in SCI)
- **Anthropometric findings from birth to adulthood and their relation with karyotpye distribution in Turkish girls with Turner syndrome.**
  SARI E., BEREKET A., YEŞILKAYA E., BAŞ F., BUNDAK R., AYDıN B., DARCAN Ş., DÜNDAR B., BÜYÜKINAN M., KARA C., et al.
  American journal of medical genetics. Part A, pp.942-8, 2016 (Journal Indexed in SCI)
- **Coexistence of Kabuki Syndrome and Autoimmune Thyroiditis.**
  Gürbüz F., Özalp Y., CEYLANER S., Topaloğlu A. K. , Yüksel B.
  Journal of clinical research in pediatric endocrinology, vol.8, pp.105-6, 2016 (Journal Indexed in SCI)
- **Complete idiopathic hypogonadotropic hypogonadism due to homozygous GNRH1 mutations in the mutational hot spots in the region encoding the decapeptide**
  MENGEN E., TUNC S., KOTAN L. D. , NALBANTOGLU O., Demir K., GÜRBÜZ F., TURAN İ., ŞEKER G., YÜKSEL B., TOPALOĞLU A. K.
  Hormone Research in Paediatrics, vol.85, pp.107-111, 2016 (Journal Indexed in SCI)
- **Effects of methylphenidate on appetite and growth in children diagnosed with attention deficit and hyperactivity disorder**
  GÜRBÜZ F., GURBUZ B. B. , CELIK G. G. , Yildirim V., UCAKTURK S. A. , SEYDAOĞLU G., UCAKTURK E. M. , TOPALOĞLU A. K. , YÜKSEL B.
  JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.29, pp.85-92, 2016 (Journal Indexed in SCI)
- **Idiopathic Hypogonadotrophic Hypogonadism Caused by Inactivating Mutations in SRA1**
  KOTAN L. D. , Cooper C., DARCAN Ş., Carr I., ÖZEN S., Yan Y., Hamedani M. K. , GÜRBÜZ F., Mengen E.,

TURAN İ., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.58-59, 2016 (Journal Indexed in SCI)

- **Inactivating Mutations in CCDC141 Causing Idiopathic Hypogonadotrophic Hypogonadism/Kallmann Syndrome**
  TURAN İ., Hutchins B. I. , Hacihamdioglu B., Ozbek M. N. , KOTAN L. D. , ÖZKAN Y., Stoner H., Cheng P. J. , GÜRBÜZ F., Mengen E., et al.
  HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.58, 2016 (Journal Indexed in SCI)
- **Expanding the Clinical Spectrum Associated With GLIS3 Mutations.**
  DIMITRI P., HABEB A., Gurbuz F., MILLWARD A., WALLIS S., MOUSSA K., AKCAY T., TAHA D., HOGUE J., SLAVOTINEK A., et al.
  The Journal of clinical endocrinology and metabolism, vol.100, 2015 (Journal Indexed in SCI)
- **Growth curves for Turkish Girls with Turner Syndrome: Results of the Turkish Turner Syndrome Study Group**
  Darendeliler F., Yesilkaya E., BEREKET A., Bas 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, pp.183-191, 2015 (Journal Indexed in SCI)
- **EARLY DIABETIC NEPHROPATHY AND ENDOTHELIAL DYSFUNCTION IN CHILDREN WITH TYPE 1 DIABETES MELLITUS**
  Aynaci S., KARABAY BAYAZIT A., MELEK E., GÜRBÜZ F., ATMIŞ B., Anarat R., YÜKSEL B., Anarat A.
  PEDIATRIC NEPHROLOGY, vol.30, pp.1632, 2015 (Journal Indexed in SCI)
- **The effect of lifestyle change and metformin therapy on serum arylesterase and paraoxonase activity in obese children**
  Cayir A., Turan M. I. , GÜRBÜZ F., Kurt N., Yildirim A.
  JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.28, pp.551-556, 2015 (Journal Indexed in SCI)
- **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, pp.429-438, 2015 (Journal Indexed in SCI)
- **Turner Syndrome and Associated Problems in Turkish Children: A Multicenter Study**
  Yesilkaya E., BEREKET A., Darendeliler F., Bas F., Poyrazoglu S., Aydin B. K. , DARCAN Ş., Dundar B., Buyukinan M., Kara C., et al.
  JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.7, pp.27-36, 2015 (Journal Indexed in SCI)
- **MCM9 Mutations Are Associated with Ovarian Failure, Short Stature, and Chromosomal Instability**
  Wood-Trageser M. A. , GÜRBÜZ F., Yatsenko S. A. , Jeffries E. P. , Kotan L. D. , Surti U., Ketterer D. M. , Matic J., Chipkin J., Jiang H., et al.
  AMERICAN JOURNAL OF HUMAN GENETICS, vol.95, pp.754-762, 2014 (Journal Indexed in SCI)
- **Loss-of-Function Mutations in PNPLA6 Encoding Neuropathy Target Esterase Underlie Pubertal Failure and Neurological Deficits in Gordon Holmes Syndrome**
  Topaloğlu A. K. , Lomniczi A., Kretzschmar D., Dissen G. A. , Kotan L. D. , Mcardle C. A. , Koç A. F. , Hamel B. C. , Guclu M., Papatya E. D. , et al.
  JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, vol.99, 2014 (Journal Indexed in SCI)
- **Mutations in FEZF1 Cause Kallmann Syndrome**
  La Kotan L. D. , Hutchins B. I. , Ozkan Y., Demire F., Stoner H., Cheng P. J. , Esen I., GÜRBÜZ F., BIÇAKCI Y. K. , Mengen E., et al.
  AMERICAN JOURNAL OF HUMAN GENETICS, vol.95, pp.326-331, 2014 (Journal Indexed in SCI)
- **Etiological Evaluation of Patients Presenting with Isolated Micropenis to an Academic Health Care Center**
  Aslan T. B. , GÜRBÜZ F., Temiz F., YÜKSEL B., TOPALOĞLU A. K.
  INDIAN JOURNAL OF PEDIATRICS, vol.81, pp.775-779, 2014 (Journal Indexed in SCI)
- **Loss of Function Mutations in PNPLA6 Encoding Neuropathy Target Esterase Cause Pubertal Failure**

and Cerebellar Ataxia (Gordon Holmes Syndrome)
KOTAN L. D. , LOMNICZI A., Kretzschmar D., Dissen G. A. , McArdle C. A. , Koc F., Hamel B. C. , Guclu M., Papatya E. D. , Eren E., et al.
ENDOCRINE REVIEWS, vol.35, 2014 (Journal Indexed in SCI)

- **Epstein-barr virus encephalitis in infancy.**
Gurbuz F., GURBUZ B., ÇAYıR A., TEZER H.
The West Indian medical journal, vol.63, pp.206-7, 2014 (Journal Indexed in SCI)
- **The Novel Mutation p.Trp147Arg of the Steroidogenic Acute Regulatory Protein Causes Classic Lipoid Congenital Adrenal Hyperplasia with Adrenal Insufficiency and 46,XY Disorder of Sex Development**
YÜKSEL B., Kulle A. E. , GÜRBÜZ F., Welzel M., Kotan D., Mengen E., Holterhus P., TOPALOĞLU A. K. , Groetzinger J., Riepe F. G.
HORMONE RESEARCH IN PAEDIATRICS, vol.80, pp.163-169, 2013 (Journal Indexed in SCI)
- **Relationship between metabolic control and neurocognitive functions in children diagnosed with type I diabetes mellitus before and after 5 years of age**
TOLU-KENDIR O., KİRİŞ N., TEMIZ F., GÜRBÜZ F., Onenli-Mungan N., TOPALOĞLU A. K. , YÜKSEL B.
TURKISH JOURNAL OF PEDIATRICS, vol.54, pp.352-361, 2012 (Journal Indexed in SCI)
- **Unilateral exudative retinal detachment as the sole presentation of relapsing acute lymphoblastic leukemia.**
AZıK F., AKıNCı A. C. , SAYLı T., CULHA V., TEBERIK K., TEKE M., Gürbüz F.
Turkish journal of haematology : official journal of Turkish Society of Haematology, vol.29, pp.181-4, 2012 (Journal Indexed in SCI)
- **Inactivating KISS1 Mutation and Hypogonadotropic Hypogonadism**
TOPALOĞLU A. K. , Tello J. A. , KOTAN L. D. , OZBEK M. N. , YILMAZ M., ERDOĞAN Ş., GÜRBÜZ F., TEMIZ F., Millar R. P. , YÜKSEL B.
NEW ENGLAND JOURNAL OF MEDICINE, vol.366, pp.629-635, 2012 (Journal Indexed in SCI)
- **Distribution of Gene Mutations Associated with Familial Normosmic Idiopathic Hypogonadotropic Hypogonadism**
GÜRBÜZ F., KOTAN L. D. , Mengen E., ŞIKLAR Z., BERBEROĞLU M., Dokmetas S., Kilicli M. F. , Guven A., KİREL B., Saka N., et al.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.4, pp.121-126, 2012 (Journal Indexed in SCI)

Articles Published in Other Journals

- **Coexistence of type 1 diabetes mellitus and periventricular heterotopia in a child: A case report**
İNCECİK F., GÜRBÜZ F.
JOURNAL OF PEDIATRIC NEUROSCIENCES, vol.15, pp.67-68, 2020 (Journal Indexed in ESCI)
- **Predicted Benign and Synonymous Variants in CYP11A1 Cause Primary Adrenal Insufficiency Through Missplicing**
Maharaj A., Buonocore F., Meimaridou E., Ruiz-Babot G., Guasti L., Peng H., Capper C. P. , Burgos-Tirado N., Prasad R., Hughes C. R. , et al.
JOURNAL OF THE ENDOCRINE SOCIETY, vol.3, pp.201-221, 2019 (Journal Indexed in ESCI)
- **Vitamin D levels of children with chronic liver disease**
GÜRBÜZ F., AĞIN M., MENGEN E., ELCI H., ÜNAL İ., TÜMGÖR G., YÜKSEL B.
CUKUROVA MEDICAL JOURNAL, vol.43, pp.450-456, 2018 (Journal Indexed in ESCI)
- **Evaluation of Two Different Pamidronate Treatment Protocols in Children with Osteogenesis Imperfecta**
ÖNENLİ MUNGAN H., gürbüz F., mengen e., özgür ö., topaloğlu a. k. , yüksel b.
CUKUROVA MEDICAL JOURNAL, vol.39, pp.532-539, 2014 (National Refreed University Journal)

Refereed Congress / Symposium Publications in Proceedings

- **Kronik Karaciğer Hastalığı Olan Çocukların D Vitamini Düzeylerinin Değerlendirilmesi**

GÜRBÜZ F., AĞIN M., mengen e., elçi h., ÜNAL İ., TÜMGÖR G., YÜKSEL B.
XXII. Ulusal Pediatrik Endokrinoloji ve Diyabet Kongresi, Turkey, 18 - 22 April 2018

Citations

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