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Research Areas

Medical Genetics

Academic Titles / Tasks

Research Assistant PhD, Marmara University, School Of Medicine, Internal Medical Sciences, 2018 - Continues

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

- I. **KIDNEY DIMENSION IS THE MOST IMPORTANT PARAMETER ASSOCIATED WITH DETERIORATION IN KIDNEY FUNCTION IN CHILDREN WITH AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE**
Cicek N., GÖKCE İ., ALAVANDA C., Guven S., SAK M., Turkkan O. N. , Bodur E. D. , Polat S., ATA P., YILDIZ N., et al.
PEDIATRIC NEPHROLOGY, vol.36, no.10, pp.3421, 2021 (Journal Indexed in SCI)
- II. **PHENOTYPIC AND GENOTYPIC CHARACTERISTICS OF CHILDREN WITH BARTTER SYNDROME**
Guyen S., GÖKCE İ., ALAVANDA C., Bodur E. D. , Cicek N., SAK M., Pul S., Turkkan O. N. , ATA P., YILDIZ N., et al.
PEDIATRIC NEPHROLOGY, vol.36, no.10, pp.3430, 2021 (Journal Indexed in SCI)
- III. **CLINICAL-GENETIC CHARACTERISTICS AND PREDICTORS OF DISEASE PROGRESSION IN PATIENTS WITH AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE**
ALPAY H., Cicek N., ALAVANDA C., Guven S., SAK M., Turkkan O. N. , Bodur E. D. , Polat S., ATA P., YILDIZ N., et al.
PEDIATRIC NEPHROLOGY, vol.36, no.10, pp.3422, 2021 (Journal Indexed in SCI)
- IV. **THE FIRST CASE OF AMYLOIDOSIS DUE TO HOMOZYGOUS P.V377I MUTATION IN A PATIENT WITH HYPERIMMUNOGLOBULIN D SYNDROME**
Bodur E. D. , GÖKCE İ., Sozeri B., Alavanda C., Farmanli O., Ata P., KAYA H., ALPAY H.
PEDIATRIC NEPHROLOGY, vol.36, no.10, pp.3448, 2021 (Journal Indexed in SCI)
- V. **A rare cause of hypercalcemia: Congenital Lactase Deficiency**
ELTAN M., ALAVANDA C., ABALI S., Abali Z. Y. , KAYGUSUZ S. B. , GÜRPINAR TOSUN B., SEVEN MENEVŞE T., Helvacioğlu D., GÜRAN T., ATA P., et al.
HORMONE RESEARCH IN PAEDIATRICALS, vol.94, no.SUPPL 1, pp.75, 2021 (Journal Indexed in SCI)
- VI. **Meckel-Gruber Syndrome: Clinical and Molecular Genetic Profiles in Two Fetuses and Review of the Current Literature**
TÜRKYILMAZ A., GEÇKİNLİ B. B. , ALAVANDA C., Ates E. A. , Buyukbayrak E. E. , EREN Ş. F. , ARMAN A.
GENETIC TESTING AND MOLECULAR BIOMARKERS, 2021 (Journal Indexed in SCI)
- VII. **Secondary findings in 622 Turkish clinical exome sequencing data**
Ates E. A. , TÜRKYILMAZ A., Yildirim O., ALAVANDA C., Polat H., Demir S., ÇEBİ A. H. , GEÇKİNLİ B. B. , GÜNEY A. İ. , ATA P., et al.
JOURNAL OF HUMAN GENETICS, 2021 (Journal Indexed in SCI)
- VIII. **The Spectrum of Low-Density Lipoprotein Receptor Mutations in a Large Turkish Cohort of Patients with Familial Hypercholesterolemia**

TÜRKYILMAZ A., Kurnaz E., ALAVANDA C., Yarali O., Kartal Baykan E., YAVUZ D., Cayir A., ATA P.
METABOLIC SYNDROME AND RELATED DISORDERS, 2021 (Journal Indexed in SCI)

- IX. **Biallelic Mutations in DNAJB11 are Associated with Prenatal Polycystic Kidney Disease in a Turkish Family**
Ates E. A. , TÜRKYILMAZ A., DELİL K., ALAVANDA C., SÖYLEMEZ M. A. , GEÇKİNLİ B. B. , ATA P., ARMAN A.
MOLECULAR SYNDROMOLOGY, 2021 (Journal Indexed in SCI)
- X. **Novel clinical features and pleiotropic effect in three unrelated patients with LMNA variant**
Turkyilmaz A., GEÇKİNLİ B. B. , ALAVANDA C., Ates E. A. , ARMAN A.
CLINICAL DYSMORPHOLOGY, vol.30, no.1, pp.10-16, 2021 (Journal Indexed in SCI)
- XI. **Does Genotype–Phenotype Correlation Exist in Vitamin D-Dependent Rickets Type IA: Report of 13 New Cases and Review of the Literature**
Kaygusuz S. B. , Alavanda C., Kırkgöz T., Eltan M., Yavas Abali Z., Helvacioğlu D., Güran T., Ata P., Bereket A., Demircioğlu S.
Calcified Tissue International, 2021 (Journal Indexed in SCI Expanded)
- XII. **A Rare Cause of Hypophosphatemia: Raine Syndrome Changing Clinical Features with Age**
Eltan M., Alavanda C., Yavas Abali Z., Ergenekon P., Yalindag Ozturk N., Sakar M., Dağçınar A., Kırkgöz T., Kaygusuz S. B. , Gökdemir Y., et al.
CALCIFIED TISSUE INTERNATIONAL, vol.107, pp.96-103, 2020 (Journal Indexed in SCI)
- XIII. **A rare cause of hypophosphatemia: Raine Syndrome**
Eltan M., Ata P., Kırkgöz T., Alavanda C., Kaygusuz S. B. , Menevse T. S. , Tosun B. G. , Abali Z. Y. , Helvacioğlu D., Güran T., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.384, 2019 (Journal Indexed in SCI)

Refereed Congress / Symposium Publications in Proceedings

- I. **Nadir bir iskelet displazisi: Pknodizostoz tanısı alan iki kız kardeş**
ALAVANDA C., GEÇKİNLİ B. B. , ARSLAN ATEŞ E., POLAT H., SÖYLEMEZ M. A. , GÜNEY A. İ. , ATA P., ARMAN A.
4. Ulusal Çocuk genetik kongresi, Turkey, 25 - 27 September 2019
- II. **Schaaf Yang sendromu**
Alavanda C., Arslan Ateş E., Polat H., Geçkinli B. B. , Söylemez M. A. , Güney A. İ. , Ata P., Arman A.
4. Ulusal Çocuk Genetik kongresi, İstanbul, Turkey, 25 - 27 September 2019
- III. **A rare cause of hypophosphatemia: Raine Syndrome**
ELTAN M., ATA P., KIRKGÖZ T., ALAVANDA C., KAYGUSUZ S. B. , SEVEN M. T. , GURPINAR TOSUN B., YAVAŞ ABALI Z., GÜRAN T., ELÇİOĞLU H. N. , et al.
European Society for Paediatric Endocrinology (ESPE)58th Annual Meeting, Vienna, VIYANA, Austria, 19 - 21 September 2019, vol.91
- IV. **A cohort of patients with hypertrophic and dilated cardiomyopathy**
POLAT H., TÜRKYILMAZ A., ALAVANDA C., ARSLAN ATEŞ E., SÖYLEMEZ M. A. , GEÇKİNLİ B. B. , YILDIRIM Ö., ARMAN A.
13 Balkan Congress of Human Genetics, 17 - 20 April 2019
- V. **MARFAN SYNDROME: GENOTYPE-PHENOTYPE CORRELATIONS**
GEÇKİNLİ B. B. , ARSLAN ATEŞ E., TÜRKYILMAZ A., ALAVANDA C., YILDIRIM Ö., SÖYLEMEZ M. A. , ARMAN A.
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- VI. **Von Hippel Lindau Patients**
ALAVANDA C., TÜRKYILMAZ A., POLAT H., GEÇKİNLİ B. B. , GÜNEY A. İ. , ATA P., SÖYLEMEZ M. A. , ARMAN A.
13. BALKAN GENETİK KONGRESİ, Edirne, Turkey, 16 - 20 April 2019
- VII. **LAMM syndrome: Two new patients , one novel mutation and one new mechanism**
ALAVANDA C., TÜRKYILMAZ A., ARSLAN ATEŞ E., GEÇKİNLİ B. B. , SÖYLEMEZ M. A. , ARMAN A.
13 Balkan Congress of Human Genetics, 17 - 20 April 2019
- VIII. **TWO NOVEL MUTATIONS IN THREE DIFFERENT GENES ASSOCIATED WITH RETINITIS**

PIGMENTOSA/LEBER CONGENITAL AMAROSIS IN ONE PATIENT

ALAVANDA C., ATA P., YILMAZ Ö., POLAT H., GEÇKİNLİ B. B. , ARMAN A.

13. BALKAN GENETİK KONGRESİ, Edirne, Turkey, 16 - 20 April 2019

- IX. **NOVEL RAB3GAP1INTRONIC MUTATION CAUSING WARBURGG MICRO SYNDROME IN TWO PATIENTS**
GEÇKİNLİ B. B. , TÜRKİYILMAZ A., ARSLAN ATEŞ E., ALAVANDA C., TAŞLIDERE H., SÖYLEMEZ M. A. , ARMAN A.
13 ULUSAL TIBBİ GENETİK KONGRESİ, Turkey, 7 - 11 November 2018
- X. **Genetic analysis results of the patient cohort diagnosed with cardiomyopathy**
TÜRKİYILMAZ A., ALAVANDA C., ARSLAN ATEŞ E., SÖYLEMEZ M. A. , GEÇKİNLİ B. B. , YILDIRIM Ö., ARMAN A.
13 ULUSAL TIBBİ GENETİK KONGRESİ, Turkey, 7 - 11 November 2018
- XI. **Three genotypes causing three distinct phenotypes in a hereditary cancer family**
ARSLAN ATEŞ E., TÜRKİYILMAZ A., ALAVANDA C., YILDIRIM Ö., SÖYLEMEZ M. A. , GEÇKİNLİ B. B. , GÜNEY A. İ.
13 ULUSAL TIBBİ GENETİK KONGRESİ, Turkey, 7 - 11 November 2018
- XII. **Chromosomal array-CGH analysis in patients having neurodevelopmental delay and dysmorphic features**
ALAVANDA C., ARSLAN ATEŞ E., TÜRKİYILMAZ A., GEÇKİNLİ B. B. , ATA P., GÜNEY A. İ. , SÖYLEMEZ M. A. , ÖZYAVUZ ÇABUK P., ARMAN A.
13 ULUSAL TIBBİ GENETİK KONGRESİ, Turkey, 7 - 11 November 2018
- XIII. **Genetic analysis results of a patient cohort diagnosed with arrhythmia**
TÜRKİYILMAZ A., ALAVANDA C., ARSLAN ATEŞ E., SÖYLEMEZ M. A. , GEÇKİNLİ B. B. , YILDIRIM Ö., ARMAN A.
13 ULUSAL TIBBİ GENETİK KONGRESİ, Turkey, 7 - 11 November 2018
- XIV. **GENETİK KLİNİĞİNE PSODOBARTTER SENDROMU BULGULARIYLA BAŞVURAN KİSTİK FİBROZLU İKİ KIZ KARDEŞ**
ALAVANDA C., ATA P., TÜRKİYILMAZ A., Arslan E., YILDIZ N., ALPAY H.
Uluslararası Katılımlı 13. Ulusal Tıbbi Genetik Kongresi, Turkey, 7 - 11 November 2018
- XV. **A further case of autosomal recessive brachyolmia having a novel mutation in PAPSS2 gene**
ARSLAN ATEŞ E., ELTAN M., TÜRKİYILMAZ A., ALAVANDA C., SÖYLEMEZ M. A. , GEÇKİNLİ B. B. , GÜNEY A. İ. , GÜRAN T., ARMAN A.
13 ULUSAL TIBBİ GENETİK KONGRESİ, Turkey, 7 - 11 November 2018
- XVI. **PEDIATRİK AKUT LENFBLASTİK LÖSEMİDE TEŞHİSEL TESTLERİN GEÇERLİLİĞİ**
Yılmaz İ., ATA P., ALAVANDA C., Arslan E., Eren R., Yılmaz B., timur ç., pala S., Acıyiyen Y., sökmen b., et al.
13. Uluslararası Katılımlı Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 7 - 11 November 2018
- XVII. **REVERSE GENETİK İLE TANI KOYULAN NADİR BİR SPASTİK PARAPLEJİ AİLESİ**
ALAVANDA C., ates e., TÜRKİYILMAZ A., GÜNEY A. İ. , GEÇKİNLİ B. B. , ATA P., ARMAN A.
13.ULUSLARARASI KATILIMLI ULUSAL TIBBİ GENETİK ANABİLİM DALI, Turkey, 7 - 11 November 2018
- XVIII. **FARKLI GENLER,FARKLI MUTASYONLAR,FENOTİPTE FARKLILIK YARATIYOR MU ? : ALPORT SENDROMU**
ATA P., ALAVANDA C., TÜRKİYILMAZ A., YILDIZ N., ALPAY H.
Uluslararası Katılımlı 13. Ulusal Tıbbi Genetik Kongresi, Turkey, 7 - 11 November 2018
- XIX. **NÖROMOTOR GELİŞME GERİLİĞİ VE DISMORFİK BULGULARI OLAN HASTALARDA ARRAY-CGH ANALİZİ**
ATA P., ALAVANDA C., ATEŞ E., GEÇKİNLİ B. B. , GÜNEY A. İ. , SÖYLEMEZ M. A. , ÇABUK P., ARMAN A.
Uluslararası Katılımlı 13. Ulusal Tıbbi Genetik Kongresi, Antalya, Turkey, 7 - 11 November 2018

Citations

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