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Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Coexistence of Two Rare Conditions Complicating the Other's Management: Propionic Acidemia and Apert Syndrome**
ENSERT CİHAN C. K., AKAR H. T., Yıldız Y., SOĞUKPINAR M., ÜTİNE G. E., ÇELİK H. T.
Molecular Syndromology, vol.15, no.1, pp.83-88, 2024 (SCI-Expanded)
- II. **TRAPPC6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions**
Almousa H., Lewis S. A., Bakhtiari S., Nordlie S. H., Pagnozzi A., Magee H., Efthymiou S., Heim J. A., Cornejo P., Zaki M. S., et al.
Brain : a journal of neurology, vol.147, no.1, pp.311-324, 2024 (SCI-Expanded)
- III. **COVID-19 in inherited metabolic disorders: Clinical features and risk factors for disease severity**
KAHRAMAN A. B., YILDIZ Y., ÇIKI K., Erdal I., AKAR H. T., DURSUN A., TOKATLI A., SİVRİ S.
Molecular Genetics and Metabolism, vol.139, no.2, 2023 (SCI-Expanded)
- IV. **Novel Cranial Imaging Findings and a Splice-Site Variant in a Patient with Tyrosinemia Type III, and a Summary of Published Cases**
KAHRAMAN A. B., AKAR H. T., Lafci N. G., YILDIZ Y., Tokatli A.
MOLECULAR SYNDROMOLOGY, vol.13, no.3, pp.193-199, 2022 (SCI-Expanded)
- V. **Biallelic mutations in ELFN1 gene associated with developmental and epileptic encephalopathy and joint laxity**
DURSUN A., YALNIZOĞLU D., Yılmaz D. Y., Oguz K. K., GÜLBAKAN B., KOŞUKCU C., AKAR H. T., Kahraman A. B., Acar N. V., GÜNBEY C., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.64, no.11, 2021 (SCI-Expanded)
- VI. **Challenging diagnosis and rare disease in children: Dermatitis artefacta**
ISİYEL E., ERSOY EVANS S., AKAR H. T., Gurbanov A., KARADUMAN A., FOTO ÖZDEMİR D., TEKŞAM Ö.
JOURNAL OF PAEDIATRICS AND CHILD HEALTH, vol.57, no.10, pp.1710-1712, 2021 (SCI-Expanded)
- VII. **Autism spectrum disorder in patients with inherited metabolic disorders-a large sample from a tertiary center**
ÇELEN YOLDAŞ T., BİLGİNER GÜRBÜZ B., AKAR H. T., ÖZMERT E. N., COŞKUN T.
TURKISH JOURNAL OF PEDIATRICS, vol.63, no.5, pp.767-779, 2021 (SCI-Expanded)
- VIII. **Complicated peripartum course in a patient with very long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency**
AKAR H. T., Çağan M., YILDIZ Y., SİVRİ H. S.
NEUROMUSCULAR DISORDERS, vol.31, no.6, pp.566-569, 2021 (SCI-Expanded)

Articles Published in Other Journals

- I. **A Novel Double Homozygous BTB Gene Mutation in A Case of Profound Biotinidase Deficiency**
DEVECİ K., AKAR H. T., YILDIZ Y., ÖZGÜL R. K.

Refereed Congress / Symposium Publications in Proceedings

I. A Patient with Germline Heterozygous Missense IKZF1 Mutation

YAKICI N., cetinkaya p., TAN Ç., AKAR H. T., ÖZBEK B., ÇAĞDAŞ AYVAZ D. N., TEZCAN F. İ.

ESID-2019, 18 - 21 September 2019

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