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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., Yldlz Y., SOĞUKPINAR M., ÜTİNE G. E., ÇELİK H. T.
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II. **TRAPPC6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions**
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III. **COVID-19 in inherited metabolic disorders: Clinical features and risk factors for disease severity**
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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**
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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**
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VI. **Challenging diagnosis and rare disease in children: Dermatitis artefacta**
ISIYEL 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 BTD Gene Mutation in A Case of Profound Biotinidase Deficiency**
DEVECİ K., AKAR H. T., YILDIZ Y., ÖZGÜL R. K.

Türkiye Çocuk Hastalıkları Dergisi, 2022 (Peer-Reviewed Journal)

Refereed Congress / Symposium Publications in Proceedings

I. **A Patient with Germline Heterozygous Missense IKZF1 Mutation**
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