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Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Blended Phenotypes of Sexual Development Disorder and Coenzyme Q10 Deficiency, Together with a Sibling with Homozygous Variants in the <i>AHI1</i> Gene**
ATASAY R., YILMAZ L. N., Gulec A., CANPOLAT M., PER H., KARDAŞ F., SELÇUK B. Ş., KARAMAN B., KİRAZ A., DÜNDAR M.
MOLECULAR SYNDROMOLOGY, 2024 (SCI-Expanded)
- II. **Biochemical risk factors associated with refractory epilepsy: alpha synuclein and adenosine deaminase**
Sener N., Ketı D., GÜLEÇ A., CANPOLAT M., PER H., GÜMÜŞ H., Muhtaroglu S.
REVISTA ROMANA DE MEDICINA DE LABORATOR, vol.32, no.3, 2024 (SCI-Expanded)
- III. **Loss of TBC1D2B causes a progressive neurological disorder with gingival overgrowth**
Harms F. L., Rexach J. E., Efthymiou S., Aynekin B., PER H., Gulec A., Nampoothiri S., Sampaio H., Sachdev R., Stoeva R., et al.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.32, no.5, pp.558-566, 2024 (SCI-Expanded)
- IV. **Pediatric-Onset Chronic Inflammatory Demyelinating Polyneuropathy: A Multicenter Study**
Sarıkaya Uzan G., Vural A., Yüksel D., Aksoy E., Öztoprak Ü., CANPOLAT M., YILDIRIM S., YILDIRIM Ç., GÜLEÇ A., PER H., et al.
Pediatric Neurology, vol.145, pp.3-10, 2023 (SCI-Expanded)
- V. **Congenital Myasthenic Syndromes in Turkey: Clinical and Molecular Characterization of 16 Cases With Three Novel Mutations**
YILDIRIM S., GÜLEÇ A., Erdoğan M., Demir M., CANPOLAT M., GÜMÜŞ H., ÇAĞLAYAN A. O., DÜNDAR M., PER H.
Pediatric Neurology, vol.136, pp.43-49, 2022 (SCI-Expanded)

Refereed Congress / Symposium Publications in Proceedings

- I. **A novel pathogenic variant in VARS1 associated with a syndromic neurodevelopmental disorder with craniofacial, and neuroradiological abnormalities in a Turkish family**
Aynekin B., GÜLEÇ A., Karagoz I., YEŞİL SAYIN G., PER H., Houlden H., Efthymiou S.
56th Annual Conference of the European-Society-of-Human-Genetics (ESHG), Glasgow, England, 10 - 13 June 2023, pp.207-208
- II. **A novel homozygous variant inSUOXgene causes classic isolated sulfite oxidase deficiency: a case report**
USLU K., GÜLEÇ A., ARSLAN S., BAŞGÖZ N., KARDAŞ F., PER H., ÖZKUL Y., DÜNDAR M.
6.ULUSLARARASI ERCİYES TIP TIBBİ GENETİK KONGRESİ, Kayseri, Turkey, 16 September 2021, pp.30

Metrics

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