Res. Asst. PhD CANAN CEYLAN KÖSE

Personal Information

Email: cananceylanköse@comu.edu.tr
Web: https://avesis.comu.edu.tr/cananceylank%C3%B6se

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Anemia and thrombocytopenia due to a novel BRPF1 variant in a family from Çanakkale with intellectual disability and dysmorphic facies: Case report and review of the literature**
Kose C. C., Kaya D., Akcan M. B., Silan F.
AMERICAN JOURNAL OF MEDICAL GENETICS, PART A, vol.191, no.6, pp.2209-2214, 2023 (SCI-Expanded)

Refereed Congress / Symposium Publications in Proceedings

I. **Importance of genetic diagnosis in demyelinating diseases; adult-onset Alexander Disease case with novel GFAP mutation**
CEYLAN KÖSE C., OCAK Ö., GÜNDÜZ O. R., SILAN F.
15. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 9 - 13 November 2022

II. **A novel BRPF1 variant in a family with intellectual disability and dysmorphic face, from Çanakkale**
CEYLAN KÖSE C., KAYA D., AKCAN M. B., SILAN F.
EUROPEAN BİOTECHNOLOGY CONGRESS 2022, PRAG, Czech Republic, 5 - 07 October 2022

III. **A rare Malpuech, Mignarelli, Michels, Carnevale Syndrome (3MC Syndrome); Novel variant in the MASP1 gene**
GÜNDÜZ O. R., CEYLAN KÖSE C., SILAN F.
EUROPEAN BİOTECHNOLOGY CONGRESS 2022, PRAG, Czech Republic, 5 - 07 October 2022

IV. **A novel homozygous mutation in CC2D1A gene: two case from a Turkish family in Canakkale**
KAYA D., CEYLAN KÖSE C., SILAN F.
EUROPEAN BİOTECHNOLOGY CONGRESS 2022, PRAG, Czech Republic, 5 - 07 October 2022

V. **A rare syndrome from Canakkale; Kindler Syndrome with loss of heterozygosity of FERMT1 gene region**
GÜNDÜZ O. R., IŞIK MERMUTLU S., CEYLAN KÖSE C., SILAN F.
7. Uluslararası Erciyes Tıp Tıbbi Genetik Kongresi, Kayseri, Turkey, 26 - 28 May 2022

Metrics

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