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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 Tibbi 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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