

Personal Information

Email: akcahanakalin@hacettepe.edu.tr

Web: <https://avesis.hacettepe.edu.tr/akcahanakalin>

Published journal articles indexed by SCI, SSCI, and AHCI

- I. **A Long-Term Follow-Up of a Patient with a Novel PORCN Variant and Additional Clinical Features**
Akalin A., Grzeschik K., ÜTİNE G. E., Boduroğlu K., ŞİMŞEK KİPER P. Ö.
Molecular Syndromology, 2024 (SCI-Expanded)
- II. **Assessing the Menstrual Cycle and Related Problems in Adolescents with a Genetic Syndrome Associated with Intellectual Disability**
Çınar H. Ü., Kızılkın M. P., AKALIN A., Kiper P. Ö. Ş., ÜTİNE G. E., DERMAN O., KANBUR N., AKGÜL S.
Journal of Pediatric and Adolescent Gynecology, vol.36, no.4, pp.363-371, 2023 (SCI-Expanded)
- III. **A novel biallelic CRIPT variant in a patient with short stature, microcephaly, and distinctive facial features**
AKALIN A., ŞİMŞEK KİPER P. Ö., Taşkıran E. Z., KARAOSMANOĞLU B., ÜTİNE G. E., BODUROĞLU O. K.
American Journal of Medical Genetics, Part A, vol.191, no.4, pp.1119-1127, 2023 (SCI-Expanded)
- IV. **Spondylo-meta-epiphyseal dysplasia (SMED), short limb-hand abnormal calcification type: Further expanding the mutational spectrum and dental findings of three new patients**
AKALIN A., Özşin C., KOÇ N., Demir G. Ü., ALANAY Y., Utine E., BODUROĞLU O. K., Tekçiçek M., ŞİMŞEK KİPER P. Ö.
European Journal of Medical Genetics, vol.66, no.4, 2023 (SCI-Expanded)
- V. **Typical Face, Developmental Delay, and Hearing Loss in a Patient with 3M Syndrome: The Co-Occurrence of Two Rare Conditions**
AKALIN A., Simsek-Kiper P. O., Taskiran E., ÜTİNE G. E., BODUROĞLU O. K.
MOLECULAR SYNDROMOLOGY, vol.13, no.6, pp.537-542, 2023 (SCI-Expanded)
- VI. **Efficacy of flecainide in bidirectional ventricular tachycardia and tachycardia-induced cardiomyopathy with Andersen-Tawil syndrome**
ÜNAL YÜKSEKGÖNÜL A., Azak E., AKALIN A., ERTUĞRUL İ., Kilic E., ÜTİNE G. E., KARAGÖZ T.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.65, no.6, 2022 (SCI-Expanded)
- VII. **MENSTRUATION RELATED QUALITY OF LIFE IN ADOLESCENTS WITH GENETIC SYNDROMES ACCOMPANYING AN INTELLECTUAL DISABILITY**
ÜÇLER ÇINAR H., PEHLİVANTÜRK KIZILKAN M., KANBUR N., DERMAN O., TÜZÜN GÜN Z., ÜTİNE G. E., ŞİMŞEK KİPER P. Ö., AKALIN A., AKGÜL S.
JOURNAL OF ADOLESCENT HEALTH, vol.70, no.4, 2022 (SCI-Expanded)
- VIII. **Spondyloepimetaphyseal dysplasia EXTL3-deficient type: Long-term follow-up and review of the literature**
AKALIN A., TAŞKIRAN Z. E., ŞİMŞEK KİPER P. Ö., Utine E., ALANAY Y., Ozcelik U., BODUROĞLU O. K.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.10, pp.3104-3110, 2021 (SCI-Expanded)
- IX. **Main Physical Features, Echocardiographic and Renal Ultrasonographic Findings of Turner Syndrome in 107 Pediatric Patients**
AKALIN A., ERTUĞRUL İ., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., BODUROĞLU O. K.
MOLECULAR SYNDROMOLOGY, vol.12, no.6, pp.335-341, 2021 (SCI-Expanded)

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