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### Research Areas

Medicine

### Academic Titles / Tasks

Research Assistant PhD, Canakkale Onsekiz Mart University, Tıp Fakültesi, Dahili Tıp Bilimleri, 2019 - Continues

### Published journal articles indexed by SCI, SSCI, and AHCI

- I. **A case report of a patient with neurodevelopmental disorder with impaired speech and hyperkinetic movements: A biallelic variant in the ZNF142 gene**  
Kaya D., CEYLAN KÖSE C., AKCAN M. B., SILAN F.  
American Journal of Medical Genetics, Part A, vol.194, no.9, 2024 (SCI-Expanded)
- II. **Exploring genetic variants in congenital monosaccharide-disaccharide metabolism: Carrier ratios and phenotypic insights**  
AKCAN M. B., SILAN F.  
Journal of Pediatric Gastroenterology and Nutrition, vol.78, no.6, pp.1251-1260, 2024 (SCI-Expanded)
- III. **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)
- IV. **Evaluating of colchicine use patterns and attack frequency of familial Mediterranean fever patients in the COVID-19 pandemic**  
AKCAN M. B., Albuz B., Ozdemir O., SILAN F.  
International Journal of Rheumatic Diseases, vol.26, no.5, pp.988-991, 2023 (SCI-Expanded)
- V. **A New Case of Rare Microdeletion 10q22.3q23 along with Mosaic Klinefelter Syndrome Associated with Facial Dysmorphic Finding, Atrial Ventricular Septal Defect, and Motor Retardation**  
Dincsoy Bir F., SILAN F., Velickovic J., Berkay Akcan M. B., ÖZDEMİR Ö.  
MOLECULAR SYNDROMOLOGY, vol.13, no.3, pp.254-260, 2022 (SCI-Expanded)

### Articles Published in Other Journals

- I. **Familial intragenic X-linked OPHN1 gene deletion in a newborn male infant with low birth weight and distinctive facial appearance that diagnosed by advanced microarray-CGH method**

## Refereed Congress / Symposium Publications in Proceedings

- I. **VHL geninde patojenik/muhtemel patojenik varyant saptanan olguların genotip-fenotip ilişkisi**  
Kaya D., Akcan M. B., Sönmez V., Silan F.  
2.Ulusal HematoOnkoGenetik Kongresi, İskele, Cyprus (Kkctc), 4 - 07 May 2023, vol.1, no.3308, pp.105
- II. **A patient with Mayer-Rokitansky-Küster-Hauser(MRKH) syndrome due to 16p11.2 deletion: a case report(MRKH) syndrome due to 16p11.2 deletion: a case reportfrom Çanakkalefrom Çanakkale**  
Çelik K. M., Çavuş E., Akcan M. B., Silan F.  
EUROPEAN BIOTECHNOLOGY CONGRESS 2022, Praha, Czech Republic, 05 October 2022, pp.69
- III. **Mowat-Wilson Syndrome: A Case Report With Novel Splice Site Mutation In Zeb2 Gene**  
TEKİN K., AKCAN M. B., KAYA D., SILAN F.  
15. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 9 - 13 November 2022
- IV. **Isolated Colobomatous Microphthalmia Due To A Nonsense Mutation In The Abcb6 Gene: A Rare Case Report**  
Akcan M. B., Kaya D., Erdoğan H., Silan F.  
15.Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 9 - 13 November 2022, pp.269
- V. **Ailesel Kanser Sendromu Tanısında Göz Ardı Edilmemesi Gereken Bir Gen "Pms1": Çanakkale'Den Vaka Serisi**  
çelik k. m., KAYA D., AKCAN M. B., SILAN F.  
15. Ulusal Tıbbi Genetik Kongresi, Muğla, Turkey, 9 - 13 November 2022
- VI. **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 BIOTECHNOLOGY CONGRESS 2022, PRAG, Czech Republic, 5 - 07 October 2022
- VII. **The significance of the BRCA2 c.9934A>G variant is really unknown?A series of patients with BRCA2 c.9934A>G variant among Çanakkale patients**  
AKCAN M. B., ÇELİK K. M., SILAN F.  
EUROPEAN BIOTECHNOLOGY CONGRESS 2022, PRAG, Czech Republic, 5 - 07 October 2022
- VIII. **A patient with Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome due to 16p11.2 deletion: a case report from Çanakkale**  
ÇELİK K. M., ÇAVUŞ E., AKCAN M. B., SILAN F.  
EUROPEAN BIOTECHNOLOGY CONGRESS 2022, PRAG, Czech Republic, 5 - 07 October 2022
- IX. **A novel BRPF1 variant in a family with intellectual disability and dysmorphic face, from Çanakkale**  
Silan F., Ceylan Köse C., Kaya D., Akcan M. B.  
EUROPEAN BIOTECHNOLOGY CONGRESS 2022, Praha, Czech Republic, 5 - 07 October 2022, pp.3
- X. **The significance of the BRCA2 c.9934A>G variant is really unknown?A series of patients with BRCA2 c.9934A>G variant among Çanakkale patients**  
Akcan M. B., Çelik K. M., Silan F.  
European Biotechnology Congress, Praha, Czech Republic, 5 - 07 October 2022, pp.48
- XI. **22q11.2 duplication syndrome showing clinical variability and overlapping with the features of Digeorge/Velocardiofacial Syndrome: case series from Çanakkale**  
Akcan M. B., Çelik K. M., Silan F.  
7.Uluslararası Erciyes Tıp Tıbbi Genetik Kongresi, Kayseri, Turkey, 26 May 2022, pp.161-162
- XII. **Türkiye NRXN1 intragenik delesyonlarının fenotipik spektrumu: Çanakkale'den bir vaka serisi**  
ÇELİK K. M., AKCAN M. B., SILAN F.  
7. Uluslararası Erciyes Tıp Tıbbi Genetik Kongresi, Kayseri, Turkey, 26 - 28 May 2022
- XIII. **İzole Pankreas Kanserli Bir Olgunun Ailesinde Saptanan M.patojenik Novel Vhl Mutasyonu Bildirimi**  
Sönmez V., Kaya D., Akcan M. B., Kablan A., Emre Mutluer Y., Silan F.

1. Ulusal HematoOnkoGenetik Kongresi, Antalya, Turkey, 25 - 28 November 2021, pp.0-1
- XIV. **Palb2 Genotip Fenotip Korelasyonu: Vaka Serisi**  
Sönmez V., Akcan M. B., Ecmel Akbaş N., Kaya D., Muge Celik K., Silan F.  
1. Ulusal HematoOnkoGenetik Kongresi, Antalya, Turkey, 25 - 28 November 2021, pp.0-1
- XV. **Ailevi Kanser Sendromu Ön Tanılı Vakalarda Tp53 Ve Chek2 Genlerinin Analizi: Bir Vaka Serisi**  
Akcan M. B., Ceylan Köse C., Kaya D., Gündüz O. R., Sönmez V., Silan F.  
1. Ulusal HematoOnkoGenetik Kongresi, Antalya, Turkey, 25 - 28 November 2021, pp.0-1
- XVI. **De novo GABRB3 c.103G>A mutation detected in a patient with epilepsy and speech retardation**  
AKBAŞ N. E., SÖNMEZ V., AKCAN M. B., MUTLUER Y. E., CELİK K. M., SILAN F., ÖZDEMİR Ö.  
6. Uluslararası Katılımlı Erciyes Tıp Tıbbi Genetik Kongresi, Kayseri, Turkey, 16 September 2021
- XVII. **Multiple pterygia, joint contractures, hypoplastic lungs and hydrops: A rare case of lethal multiple pterygium syndrome**  
BAKIOĞLU KAYA D., AKCAN M. B., SÖNMEZ V., KABLAN A., ÖZDEMİR Ö., SILAN F.  
Ulusal tıbbi genetik kongresi 2020, Online, Turkey, 20 November 2020
- XVIII. **Oculocutaneous albinism caused by a compound heterozygous mutations in TYR gene: a case report**  
AKCAN M. B., BAKIOĞLU KAYA D., SÖNMEZ V., KABLAN A., ÖZDEMİR Ö., SILAN F.  
Ulusal tıbbi genetik kongresi 2020, Online, Turkey, 20 November 2020
- XIX. **Chromatin remodelling dysfunction and CHD2 related epilepsy: Reports of two unrelated child.**  
KABLAN A., ALBUZ B., AKBAŞ N. E., KARAKAYA T., SÖNMEZ V., AKCAN M. B., SILAN F., ÖZDEMİR Ö.  
European Biotechnology Congress 2020, Prag, Czech Republic, 24 September 2020
- XX. **A novel variant for SQSTM1 gene in a patient with frontotemporal dementia.**  
OCAK Ö., KARAKAYA T., ALBUZ B., AKBAŞ N. E., SÖNMEZ V., AKCAN M. B., SILAN F., ÖZDEMİR Ö.  
European Biotechnology Congress 2020, Prag, Czech Republic, 24 September 2020
- XXI. **A rare lethal multiple pterygium syndrome caused by a homozygous point mutation In CHRNG gene:a case report.**  
AKCAN M. B., ALBUZ B., KABLAN A., ÖZDEMİR Ö., SILAN F.  
European Biotechnology Congress 2020, Czech Republic, 24 September 2020
- XXII. **A Case Of Coffin-Siris Syndrome With Atypical Phenotype Caused By A Novel De Novo Mutation In ARID1B Gene.**  
ALBUZ B., AKBAŞ N. E., KARAKAYA T., SÖNMEZ V., AKCAN M. B., SILAN F., ÖZDEMİR Ö.  
European Biotechnology Congress 2020, Prag, Czech Republic, 24 September 2020

## Metrics

Publication: 28

Citation (WoS): 2

Citation (Scopus): 2

H-Index (WoS): 1

H-Index (Scopus): 1

## Awards

Doğan D., Akcan M. B., Silan F., Antikor Negatif Diyabet ve Monojenik Diyabette Genetik Analiz Sonuçlarının Değerlendirilmesi: Tek Merkez Deneyimi, Bursa Uludağ Üniversitesi, March 2024

Akcan M. B., Albuz B., Özdemir Ö., Silan F., COVID-19 pandemic in patients with familial mediterranean fever; The possible protective role of colchicine in COVID-19 symptoms, Erciyes Üniversitesi, September 2021

Akcan M. B., Kaya D., Sönmez V., Kablan A., Özdemir Ö., Silan F., Fellowship, European Cytogeneticists Association (E.C.A.), July 2021