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Genetic basis and hematologic manifestations of Sitosterolemia in a group of Turkish patients

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Background: Sitosterolemia is a rare disease characterized by premature atherosclerosis, xanthoma, and hematologic abnormalities that are caused by mutations in the ABCG5/ABCG8 genes. To evaluate the prevalence, risk factors, and genetic basis of sitosterolemia in a group of Turkish patients.

Methods: The 110 suspected patients for sitosterolemia were enrolled in this study. All participants underwent sterol level measurement by gas chromatography. Any individual with sterol level ≥15 µg/mL were genetically tested by Sanger sequencing analysis for ABCG5/ABCG8 defects. When an index case was identified, their relatives were invited to undergo cascade screening for sitosterolemia. All patients and 157 age matched healthy controls were tested for three common polymorphisms in the ABCG8 gene (c.1895T>C and c.161A>G) and in the ABCG5 gene (c.1810C>G). Results: The seven index cases (6.4%) were diagnosed sitosterolemia. They had elevated sterol levels (≥15 µg/mL) in association with four novel and one known mutations in the ABCG5/ABCG8 genes. The 13 family members (11.8%) had these same mutations. Seventy of the 90 patients with suspected sitosterolemia (63.6% of total) had elevated sterol levels, whereas the remaining 20 (18.2% of total) did not. The frequencies of polymorphisms were not significantly different between patients and controls (p>0.05 for all). Male sex (OR: 4.7 [95%CI: 1.1-19.3]), young age (≤18 years) (OR: 3.1 [95%CI: 1.2-7.2]), presence of macrothrombocytopenia (OR: 5.7 [95%CI: 1.9-7.1]) were significant risk factors for sitosterolemia in ABCG5/8 mutation-positive 20 patients/relatives. Conclusion: The findings suggest that sterol testing for sitosterolemia could be beneficial for Turkish children with unexplained macrothrombocytopenia.

Biography

Zühre Kaya is a Professor of Paediatric Haematology in the Department of Paediatrics at the Medical School of Gazi University in Ankara, Turkey. Dr Kaya earned her medical degree at the Medical School of Gazi University, where she also completed her residency in paediatrics and a fellowship in paediatric haematology. She currently serves as full professor of Pediatric Hematology at the University of Gazi and director of Haemostasis laboratory of the University of Gazi (Turkey). Dr Kaya's credit are some 100 articles published in such journals as Haemophilia, Blood Coagulation and Fibrinolysis, Pediatric Blood & Cancer, among others.