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Bringing Cardiogenetics to the Clinic: Challenges and Opportunities

Background: Advances in genetics have improved understanding of health conditions, yet integration into routine practice remains limited. This study examines the benefits of office-based genetic testing and its impact on patient care and decision-making.
Methods: We conducted a cross-sectional study with 54 pediatric cardiology outpatients who underwent genetic testing through GeneDx, a genetic testing company that provides in-office testing kits for streamlined diagnostics. Samples were collected using oral cheek swabs, and clinical data were extracted from medical records for analysis.
Results: The most common indications for genetic testing were familial dyslipidemia, Left ventricular hypertrophy with a family history of cardiomyopathy, and early cardiac arrest in relatives. Other reasons included syndromic features (marfanoid habitus), recurrent syncope, and abnormal EKG/ECHO. Among 54 patients (28 males, 26 females), 16 (30%) had pathogenic or likely pathogenic mutations, 30 (55.5%) had variants of unknown significance (VUS), and 14 (26%) were mutation-negative. Familial hyperlipidemia was most common (10 patients), followed by Brugada syndrome (2), cardiomyopathies (2), BRAF-related RASopathy (1), and arrhythmogenic right ventricular dysplasia (1). Recommendations included ICD placement, sports clearance decisions, electrophysiological studies, lipid center referral, genetic counseling and testing for family members at an earlier time than would have happened with routine referral to Geneticist. High-risk VUS cases, based on clinical phenotype and family clustering, were referred for further evaluation.
Conclusion: Our study emphasizes that office-based genetic testing enables early diagnosis, timely treatment, and preventive care. As genomic databases grow, the rise of VUS is inevitable, making their interpretation a key challenge. Sharing emerging gene data and advancing molecular biology knowledge are essential for reclassifying VUS as benign or pathogenic.