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Variables Associated With a Known Etiology of Cardiomyopathy in Children : The Pediatric Cardiomyopathy Registry (PCMR) From 1990-1995

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Background: Most cardiomyopathy in children is idiopathic. We hypothesized that certain patient, disease, and clinical practice variables are more likely to be associated with known etiologies. Identification of these variables may lead to better strategies for diagnosis, and ultimately, etiology-specific treatments.

Methods: The NHLBI-sponsored Pediatric Cardiomyopathy Registry collected retrospective data on U.S. and Canadian individuals 18 years or younger who were diagnosed with cardiomyopathy between 1990 and 1995 and met study criteria.

Results: The sample consisted of 916 patients, 31% of whom had a known etiology for cardiomyopathy. Diagnostic categories included 27.5% myocarditis, 23.2% neuromuscular, 22.5% familial isolated cardiomyopathy, 13.9% inborn errors of metabolism, 8.9% malformation syndromes, and 3.9% other/unspecified. Patient characteristics associated with known etiology included older age, positive family histories of cardiomyopathy, sudden death, and genetic syndromes, and New England region ($P<.001$); disease characteristics included hypertrophy ($P<.05$), abnormal serum CPK ($P<.001$) and carnitine levels ($P<.05$); and clinical practice characteristics included increase amount of testing, specific testing for urine ketones and genetic disorders, cardiac catheterization, biopsy, and treatment with calcium channel antagonists ($P<.05$). After excluding patients with familial isolated cardiomyopathy, neuromuscular disorders, malformation syndromes, and other unspecified non-idiopathic disease who are diagnosed by other means, only 58% of patients underwent specified diagnostic testing, and most individual tests were performed in less than 20% of patients. Patients who underwent endocardial or skeletal muscle biopsies were more likely to have a known etiology (odds ratio 5.4, $P<.0001$) even after adjustment for patient differences and urine/blood testing. In patients who did not undergo biopsy, urine and blood testing was associated with known etiology, but the overall diagnosis rate was low (11.8%).

Conclusions: Several variables have been identified that are associated with known etiology of cardiomyopathy. Increased diagnostic testing may reduce the rate of idiopathic cardiomyopathy.