VENTRICULAR DYSFUNCTION AND AORTIC DILATION IN PATIENTS WITH RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA

Poster Contributions
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Background: Epidermolysis bullosa (EB) is a group of diseases characterized by skin fragility and blistering with minimal stimulus and occurring in 8:1,000,000 children. Previously, recessive dystrophic EB (RDEB) has been associated with patient reported or clinically diagnosed heart failure. The current investigation reviewed structural and functional cardiac abnormalities in RDEB.

Methods: Retrospective analysis of patients followed at a single institution was performed. Diagnosis of RDEB type was based on one or more of the following: history, exam, skin biopsy, and genetic testing. Demographic, laboratory, and echocardiographic data were reviewed.

Results: 224 patients with EB were identified during the study period. Of these, 45 patients with RDEB had at least 1 echocardiogram (total 154 echocardiograms). In this group there was decreased shortening fraction in 7/45 (15.6%) and ejection fraction in 6/45 (13.3%), and increased left ventricular end-diastolic dimension in 7/45 (15.6%) patients. Normalized left ventricular mass was increased in 11/42 (26.2%). In addition to ventricular remodeling, 8/43 (18.6%) patients with RDEB had a dilated aortic root; of those, 4/8 (50%) had z-score >2.5.

Conclusion: Patients with RDEB demonstrated echocardiographic evidence of left ventricular dilation and systolic dysfunction at an incidence higher than previously reported for symptomatic heart failure alone, as well as aortic dilation not heretofore described. While ventricular dysfunction may be genetically or environmentally driven, aortic dilation suggests an inherent abnormality related to RDEB. Long-term follow up is necessary to further define the significance of these findings and clinically important outcomes.