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Audit Of A Structured Approach In Paediatric Patients With Dilated Cardiomyopathy.

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Introduction
Paediatric dilated cardiomyopathy (DCM) is the most common form of paediatric cardiomyopathy, with a diverse range of aetiologies. However, many patients are still labelled as idiopathic DCM without appropriate investigations. Recently, genetic testing has gained prominence in the diagnostic workup. This study tested the hypothesis that molecular genetic analysis provides incremental value in the identification of the aetiology of paediatric DCM.

Methods:
Clinical data of 75 children with DCM were retrospectively analysed, to determine whether their initial diagnosis had changed due to genetic testing or other diagnostic tests. Furthermore, aetiology groups were characterised and a clinical profile of DCM patients in a tertiary referral centre was created.

Results:
Idiopathic DCM was diagnosed in 28 (37.3%) cases, myocarditis and familial DCM were found in 18 (24%) cases each, inborn errors of metabolism (IEOM) in 10 (13.3%) and cardiomyopathy due to vitamin D deficiency in one patient (1.3%). 15 out of 39 patients who had genetic testing were genotype-positive, resulting in more patients with either familial DCM or IEOM. As a result, a paediatric DCM diagnostic guideline was created to develop a personalised workup strategy for the Royal Brompton Hospital.

Conclusion:
Genetic testing provided the clinical breakthrough needed to correctly identify the aetiology in paediatric DCM and thereby decreased the prevalence of idiopathic cases. Increasing the number of specific causes for paediatric DCM may lead to management changes and reduced morbidity and mortality.