Abstracts

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Graph 1

Results

1 North West & North Wales Paediatric Transport Service, Alder Hey Children’s hospital (RMCH) only has cardiology on site. (AHCH) is the cardiac surgical centre and Royal Manchester Children’s Hospital, Liverpool, Wales

Patients

In this study, this new method was examined on 3 non-compartment, 26 dilated cardiomyopathy, and 25 normal subjects.

Results

The mean percentages of myocardial hypertrabeculation/noncompaction were 3.59±2.27 in control group, 8.86±5.52 in dilated cardiomyopathic patients, and 38.1±30.8 in noncompaction patients. A value of 16% could distinguish left ventricular noncompaction from dilated cardiomyopathy with 92% specificity and 100% sensitivity and from normal subjects with 100% specificity and sensitivity. This percentage had a statistically significant association with noncompacted to compacted myocardial thickness ratio (P<0.001).

Key Conclusions

This method showed good correlations with the other echocardiographic and magnetic resonance criteria. However, it is not dependent on finding the area of maximal involvement.

Background

Only eleven paediatric intensive care units (PICUs) in UK have cardiac surgical services on-site. Small proportion of patients admitted to non-cardiac surgical PICUs require transport to surgical centres. North West and North Wales Paediatric Transport Service (NWTS) provides PIC transport service, including ability to conference call specialists. Alder Hey Children’s hospital (AHCH) is the cardiac surgical centre and Royal Manchester Children’s hospital (RMCH) only has cardiology on site.

Methods

Retrospective review suspected cardiac cases transferred in 12 months- assessing if time critical surgical patients underwent single transfer to surgical centre- interventions by referring hospital.

Results

Total 29 patients of suspected cardiac diagnosis.

INTERVENTIONS REFERRING HOSPITAL NWTS

Intubation

Central Venous Access

Dinoprostone (PGE2)

Inotropes

Inhaled Nitric oxide

22 4

6 7

20 2

7 10

0 3

Seven patients had time critical cardiac surgical lesions. One patient required a second immediate transfer to surgical centre. Retrospectively, this could have been predicted from clinical picture. 68% patients had dinoprostone started by referring team following advice from cardiologist or NWTS. Inotropes initiated on advice or by NWTS.

Conclusions

Regional cardiac network can work effectively with improved communication particularly in acute scenario. Potential surgical cases may be predicted from clinical picture, especially if not resolving with full medical treatment.

PREVALENCE OF SIDEROGENIC ANEMIA IN CHILDREN WITH CONGENITAL HEART DISEASE LESS THAN ONE YEAR IN COMPARISON WITH HEALTH CHILDREN

Introduction

Sideropenic anemia is prevalent in all children with congenital heart disease, especially in children with signs of heart failure.

Objectives

The purpose of this study is to determine the prevalence of sideropenic anemia in children with CHD and compares these results with data of healthy children less than one year in Kosovo population.

Methodology

Between 2000 and 2010 4236 children were evaluated retrospectively clinically and by echocardiography for congestive heart failure (CHF) caused by CHD. CHD was diagnosed in 832 patients (19.6%) while 78 patients (1.84%). Also from the study have been exclude children with complex CHD caused central cyanosis. In all patients were analyzed heart failure symptoms, hemoglobin levels, age of diagnosis, duration of hospitalization per year and death during medical attendance.

Results

Anemia was present in 78% of children with CHD, and in 85% children with CHF. Hospitalization days per year in anemic patients had a significantly higher than in non-anemic patients (mean 32.5±19.6 days per year versus 12.3±14.3 days per year (p < 0.05). There was not found significant relation onset of heart failure symptoms and the risk of developing anemia between anemic and non-anemic patients. The evaluation showed also needed for the two times longer period of the treatment of patients with CHD with antianemic medication in compare with healthy anemic children.

Conclusion

Anemia is prevalent in pediatric patients with CHD especially patients with heart failure. Study showed no higher mortality in anemic patients.

A NOVEL MYH7 VARIANT IDENTIFIED IN A CHILD WITH RESTRICTIVE CARDIOMYOPATHY

Restrictive cardiomyopathy (RCM) is very rare in children and usually associated with a poor prognosis. Identification and thorough understanding of the molecular basis for this condition is essential for selecting appropriate therapeutic treatments.

Background

A novel MYH7 variant was identiﬁed in a child with restrictive cardiomyopathy (RCM) and systemic disease, providing support for the hypothesis that RCM can be caused by mutations in the MYH7 gene.

Methods

Whole-exome sequencing revealed a novel heterozygous variant (c.1268T>A; p.L423F) in the MYH7 gene, which was absent from control DNA samples.

Results

This variant was predicted to be disease causing, with a pathogenic score of 97.2% and a 100% allele frequency. The patient presented with severe heart failure and dilated cardiomyopathy, with a mean ejection fraction of 8.5% at 0.4 months of age and 23.9% at 1.9 years of age. The patient also had a restrictive phenotype on echocardiography, with a mean left ventricular end-diastolic diameter of 54.2 mm and a mean fractional shortening of 12.3% at 0.4 months of age and 32.1 mm and 11.1% at 1.9 years of age, respectively. The patient also had a restrictive phenotype on echocardiography, with a mean left ventricular end-diastolic diameter of 54.2 mm and a mean fractional shortening of 12.3% at 0.4 months of age and 32.1 mm and 11.1% at 1.9 years of age, respectively.

Conclusion

This novel MYH7 variant is associated with severe heart failure and dilated cardiomyopathy, with a restrictive phenotype on echocardiography. This finding supports the hypothesis that RCM can be caused by mutations in the MYH7 gene.

Key Points

- A novel MYH7 variant was identified in a child with restrictive cardiomyopathy (RCM) and systemic disease.
- This variant was predicted to be disease causing, with a pathogenic score of 97.2% and a 100% allele frequency.
- The patient presented with severe heart failure and dilated cardiomyopathy, with a mean ejection fraction of 8.5% at 0.4 months of age and 23.9% at 1.9 years of age.
- The patient also had a restrictive phenotype on echocardiography, with a mean left ventricular end-diastolic diameter of 54.2 mm and a mean fractional shortening of 12.3% at 0.4 months of age and 32.1 mm and 11.1% at 1.9 years of age, respectively.