A rare Endocrine association of Dilated Cardiomyopathy with Congenital Adrenal Hyperplasia due to 11 β hydroxylase deficiency

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Background:

- Cardiomyopathy – a heterogeneous group of disorder of myocardium associated with mechanical and/or electrical dysfunction that usually exhibit inappropriate ventricular hypertrophy or dilatation leading to heart failure.
- Congenital Adrenal Hyperplasia (CAH) due to 11β-hydroxylase deficiency is rare inherited disorder of cortisol biosynthesis due to genetic defects of CYP11B1 gene presenting as hypertension, features of androgen excess, and peripheral precocious puberty in children.
- We report rare association in a child with CAH secondary to 11 β hydroxylase deficiency with dilated cardiomyopathy.

Case :

- 2 year boy born to non-consanguineous parents diagnosed with CAH at 18 months of age along with hypertension & virilisation.
- Presented at 2.6 years with respiratory distress, tachycardia with gallop rhythm, and cardiogenic shock.

Investigations:

- Chest X-ray and 2D echocardiography suggested massive cardiomegaly
- Doppler studies revealed dilated cardiomyopathy with Ejection Fraction of 15-20%.

Management:

- Treated with furosemide, spironolactone, hydrocortisone and enalapril following which the blood pressure normalized
- Repeat echocardiography suggested improvement in cardiac function. [Ejection Fraction 36%]

Conclusion:

- Cardiac function improved following hydrocortisone therapy suggesting the possible direct effect of corticosteroids on cardiac function.
- Animal studies have shown reduced cardiac muscle contractility following adrenalectomy which is reversible with the use of dexamethasone.
- Significant decrease in calcium uptake from sarcoplasmic reticulum could also be potentially responsible for myocardial dysfunction.

Conflict of Interest: – none stated

Reference:

Minette, Mary S. et al Cardiac Function in Congenital Adrenal Hyperplasia: A Pattern of Reversible Cardiomyopathy. J Pediatr jun2013 162:6;1193-1198.

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