Treatable Cause of Ventricular Dysfunction in DiGeorge Syndrome

Hypocalcemia is a known cause of reversible cardiomyopathy and DiGeorge syndrome is an important cause of hypocalcemia. A 17 days old 2.3 kg female child, first in birth order, born to a primigravida mother by full term LSCS started having tonic-clonic jerks on day 8 of life and was also noticed to have bluishness of lips and nails, when she was referred to us. Child had intermittent episodes of tonic clonic seizures on admission. On examination, her hemodynamic parameters were stable; saturation was 74% in room air and there was cyanosis. Precordial examination revealed no cardiomegaly. S1was normal, S2 was single. There was a continuous murmur at infraclavicular area. Per abdomen – liver 2cm below right costal margin. Rest of the systemic examination was unremarkable. Echocardiography revealed pulmonary atresia with ventricular septal defect (VSD) with biventricular dysfunction (LVEF = 35%). Blood investigations revealed hypocalcemia (calcium = 4 mg/dL) for which correction was given intravenously. Qtc of the patient was 540 milliseconds. She underwent CT pulmonary angiography which revealed atresia of main pulmonary artery and hypoplastic confluent pulmonary arteries, VSD, two moderate size aorto-pulmonary collateral and aberrant right subclavian artery; thymus was not seen. FISH test showed Di-George Syndrome. Mother sample for calcium and alkaline phosphatase was within normal limits (calcium=9.5mg/dL, alkaline phosphatase = 144u/L). At discharge on oral calcium, there was marked improvement in ventricular function (LVEF = 50%) with no recurrence of seizure activity. As the baby was maintaining a saturation of about 86% it was advised to follow up for saturation monitoring and surgery at a later date.

22q11.2 deletion syndrome has several presentations including DiGeorge syndrome (DGS). Hypocalcemia is relatively common in children with DiGeorge syndrome with incidence rates varying from 17 to 60%. Especially young infants have a high incidence of hypocalcemia. Myocardial dysfunction in patients with hypocalcemia is well described in the literature [1-4] but their association has been reported scantily [1,2].

Considering such an impact of hypocalcemia on myocardial dysfunction, it is prudent to consider that a case of DiGeorge syndrome associated with hypocalcemia is likely to cause ventricular dysfunction. Hypocalcemic cardiomyopathy shows excellent results with treatment. In any child of DiGeorge syndrome presenting with myocardial dysfunction hypocalcemia should be included in the differential diagnosis and must be investigated as this is a reversible cause of cardiomyopathy.

NEERAJ AWASTHY AND SANJAY KHATRI
Department of Pediatric Cardiology,
Fortis Escorts Heart Institute,
Delhi, India.
n_awasthy@yahoo.com

REFERENCES

1. Uysal S, Kalayci AG, Bysal K. Cardiac function in children with vitamin D deficiency rickets. Pediatr Cardiol.1999;20:283–6.
2. Tomar M , Radhakrishnan S, Shrivastava S. Myocardial dysfunction due to hypocalcaemia. Indian Pediatr. 2009 Oct 14. pii: S097475590800421-2.
3. Goulet M, Rio M, Jacquette A, Ladouceur M, Bonnet D. Neonatal hypocalcaemic dilated myocardiopathy due to a 22q11 microdeletion. Arch Mal Coeur Vaiss. 2006;99:520-2.
4. Maiya S, Sullivan I, Allgrove J, Yates R, Malone M, Brain C, *et al*. Hypocalcemia and vitamin D deficiency: an important but preventable cause of life threatening infant heart failure. Heart. 2008;94:581-4.