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Proceeding

Advances in Cardiology and Cardiovascular Disorders

Digeorge syndrome [22q11.2 Microdeletion] in a critically sick neonate with duct dependant systemic circulation and refractory hypocalcemic seizures

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Received: November 10, 2017 | **Published:** November 14, 2017

DiGeorge syndrome(DGS) is the most common microdeletion syndrome with an estimated incidence of approximately 1/4000 per live birth. More than 180 malformations are associated with 22q11.2 microdeletion; the most common are cardiac defects, a characteristic facial appearance, thymic hypoplasia, cleft palate/velopharyngeal insufficiency (VPI), hypoparathyroidism with hypocalcaemia, speech and language impairment and developmental delay. Here we report a case of DiGeorge syndrome with interrupted aortic arch with refractory hypocalcemic seizures.

A 28-days-old female child, 2nd by birth order was born of non consanguineous marriage by LSCS delivery. Baby had birth weight of 3 kg and did not have any significant NICU course. On day 7 of life she was found to have heart murmur. On evaluation, child had coarctation of aorta with ventricular septal defect (VSD) and was referred to the Department of Paediatric Cardiology. Child had refractory hypocalcemic seizures on day 15 of life. There was no family history of congenital heart disease. On Examination, a syndromic feature were present like, flat facies, small eyes, small chin, and low set ears and was referred for genetic testing when DGS was suspected clinically. FISH was performed using LSI TUPLE1/ ARSA probe from Vysis Inc., USA. Probe hybridization showed only one signal (red) on one of the chromosomes 22 at 22q11.2 region in all the metaphases analysed and the results turned out to be positive for DGS.

There was significant difference of around 30mmHg between right upper limb and the other 3 limbs. On cardiovascular examination, S1 normal, S2 normal and there was Grade 3/6 ESM in left upper sternal area. Her echocardiogram showed large doubly committed VSD with left to right shunt, severe juxtaductalcoarctation of aorta, good biventricular systolic function. The baby underwent repair of coarctation of aorta (resection and extended end to end anastomosis) and transpulmonarygoretex patch closure of VSD. She was discharged after 10 days of surgery with uneventful post-operative period. The baby is currently aged 2 years with good weight and has made a good cardiac recovery. Her developmental milestones are quite well and able to talk a few words.

DGS is rarely diagnosed before operation or necropsy. This may be because hypocalcaemia is not often the presenting feature and abnormal susceptibility to infections only becomes obvious in the first months of life in the few patients who have a normal heart or survive with a cardiovascular malformation. Clinical recognition is important if cardiac surgery is considered, because the transfusion of irradiated blood may avoid a serious graft versus host reaction. The clinician should suspect DGS in infants with interruption of the aortic arch or truncus arteriosus, particularly if they have facial stigmata and hypocalcaemia and the degree of suspicion should be even higher if the interruption of the aortic arch is associated with a right-sided descending aorta.

Citation: Wadile SR, Sivakumar K, Kar B. DiGeorge syndrome [22q11.2 Microdeletion] in a critically sick neonate with duct dependant systemic circulation and refractory hypocalcemic seizures. *Adv Cardiol Cardiovasc Disorders.* (2018);1(1):60.