# DiGeorge syndrome: a case report

Kankanan arachchi I [1], Devasiri IV[1], Rathuwithana C[2], Wackwella WGHC[2],Atapattu N[3]

## Introduction

DiGeorge syndrome (DGS) is a genetic disorder caused by deletion of chromosome 22. The main features are congenital heart disease, absence or hypoplasia of thymus, hypoparathyroidism with consecutive hypocalcaemia. We report a case of an infant presented with hypocalcaemia-induced seizures with an associated thymic aplasia.

## Case Report

A 2 month old baby boy presented with recurrent seizures. They were brief in nature and occurring frequently throughout the day but there was no response to anti epileptics. He didn't have fever or other symptoms suggestive of central nervous system infection. His neurological examination was normal. Cardiovascular system reveals a systolic murmur at left upper sternal edge. The Laboratory investigations showed hypocalcaemia, hyperphosphatemia and low para thyroid hormone (PTH) levels. His serum sodium, potassium and magnesium were normal. Even though absolute lymphocyte count was normal his lymphocytes subsets testing revealed diminished activity in CD3 T cell lineage. A narrow superior mediastinum was seen on chest radiography. Ultrasonography showed an absent thymus in the anterior mediastinum.(Figure 1)

Echocardiography revealed an Osteum secondum atrial septal defect (ASD) Kidney and brain ultrasound were normal. Chromosome culture and karyotyping showed a karyotype of 46,XY,del(22)(p11.2).(Figure 2) Parental karyotypes were not carried out. Child was started on calcium and vitamin D supplementation and his serum calcium levels became normalize and seizures were subsided

## Discussion

Digeorge syndrome has a wide phenotypic spectrum and an estimated incidence of one in 4000 births. (1) In 90% of patients, the condition is caused by a deletion of chromosome 22q11.2. Neonatal hypocalcemia occurs in 60% of affected patients. Associated abnormalities of the 3rd and 4th pharyngeal pouches are common; these include conotruncal defects of the heart in 25%, velopharyngeal insufficiency in 32%, cleft palate in 9%, renal anomalies in 35%, and aplasia of the thymus with severe immunodeficiency in 1% (1). This child had the cardinal features of DGS (hypocalcaemia, absent thymus and congenital heart defects) but no characteristic facial features or cleft palate.

The diagnosis of DGS is based upon reduced numbers of CD3+ T cells, combined with either characteristic clinical findings (eg, congenital cardiac anomalies, hypocalcemia, SCID) or a demonstrated deletion in chromosome 22q11.2.(3) Complete DGS is diagnosed by the presence of profound immune deficiency (low or absent T cell numbers and lack of proliferation to mitogens) and athymia (4). Despite the radiological features being compatible with the diagnosis of thymic agenesis, there were no features suggestive of profound immunodeficiency in this child. Therefore this child would be precisely diagnosed as a case of an incomplete digeorge syndrome. Optimal management of patients with confirmed chromosome 22q11.2 deletion syndromes requires a multidisciplinary team. Ideally, this would include a cardiologist, endocrinologist, otolaryngologist, speech/language pathologist, developmental pediatric specialist, and an immunologist, although the need for these subspecialists depends upon the patient's phenotype. (5) In children with complete Digeorge, curative therapy requires thymic or hematopoietic cell transplantation. The prognosis for partial DGS patients and for complete DGS patients who survive transplantation is largely dependent upon the severity of the cardiac defect, degree of hypoparathyroidism, and intellectual development. The overall mortality rate was found to be 8 percent in a large series of 558 DGS patients (6). In most cases, death occurred in the first six months of life and was secondary to cardiac-related complications.

1. *Department of Paediatrics, Faculty of Medicine, University of Ruhuna*
2. *Professorial Paediatric unit, Teaching Hospital Karapitiya*
3. *Consultant Paediatric Endocrinologist, Lady Ridgeway Hospital, Borella*

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