# DIGEORGE SYNDROME PRESENTING WITH IMPERFORATED ANUS AND RECURRENT INFECTIONS IN NEONATAL PERIOD: A CASE REPORT

PPC02

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## BACKGROUND

DiGeorge syndrome or 22q11.2 deletion syndrome (22q11.2 DS) is the most common chromosomal microdeletion disorder. Signs and symptoms are associated with abnormal development of 3rd and 4th pharyngeal pouches. Early diagnosis in primary care through prenatal ultrasound and combined management with multidisciplinary team improve the quality of life of a patient with this rare syndrome.

## CLINICAL PRESENTATION

A 3-month old baby boy was born term at 37 weeks of gestation via spontaneous vertex delivery with birth weight of 2.4kg. Prenatally, his mother has Gestational Diabetes Mellitus requiring total insulin of 26 unit per day and it was complicated with polyhydramnios. Detailed scan has been done which showed Atrial Septal Defect (ASD) and mild bilateral kidney enlargement. His birth event was uneventful however he was referred to paediatric team for further assessment and evaluation.

On examination, noted he had down slanting palpebral fissure, low set ears, bilateral preauricular skin tag, skin tag over left lower cheek, long tapering fingers, retrognathia, micrognathia, imperforated anus and sacral dimple 3cm away from imperforated anal. Other systemic examination was normal. He was referred to the surgical team for imperforated anus and left transverse colostomy was created for anorectal malformation at day 3 of life. In view of dysmorphism, chromosomal study and fluorescent in situ hybridization (FISH) for DiGeorge syndrome was also sent. He has normal chromosomal karyotyping but was turned out to be positive of DiGeorge Syndrome. Echocardiography done by Paediatric Consultant Cardiologist showed Truncus Arteriosus (Type A3 Van Pragh Classification) with major aortopulmonary collateral arteries (MAPCAs). The child was also tachycardic and slightly tachypneic under ventilation support. He was started on anti-failure medications and treated as Truncus Arteriosus in failure.

At two weeks of life, he was noted to have persistent hypocalcemia. His calcium level was low with high serum phosphate level. Serum Alkaline Phosphatase (ALP) was normal. Serum parathyroid hormone (PTH) was 1.6pg/mL. The child was started on regular calcium gluconate infusion and calcium supplements for his neonatal hypocalcemia after discussion with Paediatrics Endocrine Team.

His NICU stay was stormy with recurrent episodes of infections, cardiac events and dermatological issue. He finally succumbed to death at the age of 3-month old due to septicemic shock secondary to *Pseudomonas Aeruginosa* bacteremia with underlying Di George Syndrome with Primary Immune Deficiency and Infantile erythroderma.

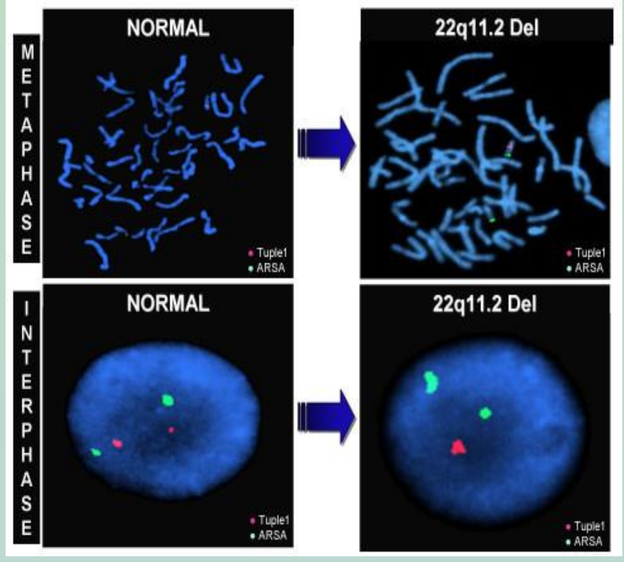


Figure 1: Fluorescent in situ hybridization (FISH) for DiGeorge Syndrome

## DISCUSSION

DiGeorge syndrome has a wide phenotypic spectrum and an estimated incidence is 1 in 4000 live births (1). 90% of cases, occur due to sporadic de-novo deletions of 22q11.2 and only 10% inherit as autosomal dominant trait. Male and female has been affected equally. Familial cases have also been found. In our case, it is most likely sporadic case as there was no similar family history of the illness. As a hemizygous deletion of chromosome 22q11.2 occurs, various clinical phenotypes are shown with a broad spectrum.

The classic triad of this syndrome includes conotruncal cardiac anomalies, hypoplastic thymus, and hypocalcemia (2). Most of these patients show mildly or moderately diminished circulating T cells (2). Additionally, neonatal hypocalcemia occurs in 60% of affected patients with this syndrome as presented in the case (3). Typical dysmorphic facies is another important clinical finding. Our patient had down slanting palpebral fissure, low set ears, retrognathia, micrognathia, and low set ear without any palatal involvement. Usually 17% patients with DiGeorge syndrome have palatal involvement (3).

Pregnancies at risk for the 22q11.2 deletion can be evaluated as two distinct groups: the first group is pregnancy with a family history of 22q11.2 deletion and the other group is pregnancy with abnormal fetal ultrasound findings (1). Usually, anomalies such as cleft palate, polyhydramnios and renal or skeletal anomalies, in addition to cardiac defects, may raise the suspicion (4). In the case presented, the patient had a negative family history and some ultrasound features. Although 22q11.2 deletion syndrome is usually characterized by multiple anomalies, the congenital heart disease being the most common (4). The improvements in routine ultrasound techniques are expected to increase significantly the number of cases with prenatally detected cardiac defects. Although there is no definite opinion for the prenatal diagnosis of 22q11.2 deletion, given the progress in ultrasound techniques and increasing awareness of physicians regarding 22q11.2 deletion syndrome appearance, there will probably be a dramatic increase in the demand for prenatal diagnosis.

Patients with DiGeorge Syndrome have a variable degree of immunodeficiency due to an underdeveloped or absent thymus. T cell immunodeficiency is of major significance in patients with “complete” DiGeorge syndrome and aplasia of the thymus, where affected patients need immune reconstitution, usually with bone marrow transplantation or thymic transplants. This is fortunately rare, occurring in less than 1% of reported cases (2). Infections are often problematic across the lifespan of an individual with complete DiGeorge syndrome. Unfortunately our case was one of those having recurrent bouts of infections and succumbed to death due to sepsis. For the majority of patients with DiGeorge syndrome who are severely immunodeficient, they are at risk of opportunistic infections such as *Pneumocystis jiroveci*, Cytomegalovirus and sometimes fungus (4). Thus, they should be treated with prophylactic broad spectrum antibiotics.

## CONCLUSION

Early diagnosis and combined management through multidisciplinary and coordinated care plan improves the quality of life of a patient with this rare syndrome. The case presented hereby emphasizes that a detailed ultrasound examination has remarkable importance during early pregnancy to choose the specific genetic test as FISH analysis in addition to the routine cytogenetic tests for an early prenatal diagnosis. Furthermore, prenatal genetic counselling should be offered to the couples regarding future pregnancy.

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