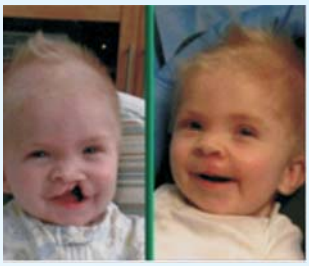
DIGEORGE SYNDROME (CATCH.22) CASE PRESENTATION

Prof. Ani Grace Kalaimathi M.Sc (N).,PGDNA.,DQA.,Ph.D., Principal, HOD, Child Health Nursing, MIOT College of Nursing
Mrs. Vijayakumari M.Sc.,(N) Lecturer, Child Health Nursing, MIOT College of Nursing
Ms.Gunavathi, Ms. Maheswari, Ms. Sudha, Ms. Sumithra, Ms. Yamunavathy,
II year M.sc (Nursing) students, Child Health Nursing, MIOT college of Nursing.

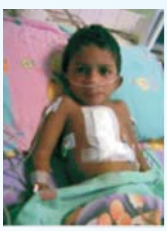
INTRODUCTION

Every mother's dream is to bring forth a healthy newborn and every health care professional dreams and works with one ultimate goal- healthy mother and healthy baby at the end of the each pregnancy. DGS is a common congenital disorder characterized by neural-crest-related developmental defects. The genetic pathways regulating cardiac neural crest development are reviewed and the evidence implicating TBX1 gene on chromosome 22q11 in the pathogenesis of DiGeorge syndrome is summarized. DiGeorge Syndrome (DGS) was first documented by Angelo DiGeorge 1965.



Patient history:-

Master Balakumaran 3.5 years old male child born on 2007 to non consanguineous parents, from Tamil Nadu (pudukottai) .He was born by lower segment cessarion section due to prolonged labour, with the birth weight of 2.6 kg. He cried immediately after birth, APGAR score was 7 /10. At birth the child was identified with Cleft lip and palate. After one week the child was identified with systolic murmur and tachypnea; diagnosed as pink TOF after the Echo screening test. Also the child had feeding difficulties and frequent upper respiratory infection for which the child was treated symptomatically till the month of April 2009 .After that the child underwent cheiloplasty on April 2009 and palatoplasty on August 2009. Then the child was referred to MIOT Hospitals Children's Care Centre for further management.



At the time of admission

The child was admitted on 03.03.2011. During admission, he was conscious, vital parameters showed Temperature 37˚C, HR-122 beats /mt, Respiration-28 breaths/mt and blood pressure-90/55mmHg. SpO2 level 92% pulse oximetry showed 92% oxygen saturation level. His weight was9.6 kg ,height-84 cm Physical findings revealed that dysmorphic facies like operated scar for cleft lip and palate ,bat ears, bulbar nostrils, growth retardation and mild developmental delay and mild mental retardation. Echocardiogram revealed velo-cardio facial syndrome, pink TOF. Electro cardio gram showed that SR,QUQ ,QRS axis RAH with early transition . Chest x-ray showed mildly lung vascularity. His hemoglobin value was 13.7 gm/dl ,pcv-40%,and elevated WBC count 13,000 cells/mm. Baby was admitted in pediatric ward and planned for surgery. Pre operative preparations like reservation of 2 units platelets and 2 units FFP was done.

Prognosis

Surgery (Intra cardiac repair) was performed on 4/3/11. Baby was kept on mechanical assisted ventilation (SIMV mode) with the support of ionotropics and prophylactic antibiotics therapy. Medications given to the includes, Inj. Cefuroxime 300 mg tds, Inj. Amikacin 75 mg /bd, Syp. Sucralfate 2.5 ml 6 th hourly , Syp . Calpol 140 mg 6th hourly Inj . Lasix ,Tab. Aldactone 6.25mg bd, Syp . Ostocalcium 5ml od. Urine output improved, it ranged from 620 to 700ml /24hrs for two days .Post operative investigation was taken , shows that elevated WBC count18,010cells/mm,Neutrophil-79%.On 1st post operative day child was identified with Right pleural effusion, good ventricular function ,tiny residual VSD mild tricuspid regurgitation for which Oxygen was delivered 4 liters/mt by mask. The chest physiotherapy was given for the following post operative days. Medications were continued as same as previously used along with repeated Echo done, the child had minimal right pleural effusion .Respiration was comfortable, with the saturation level 98% .He was awake, response only to sounds not by words. Then the child was shifted to pediatric ward. The same medications were continued. Suture removal was done on 7th post operative day, and got discharged. Echo done on the day before discharge and called for after 1 month review.

Follow Up Care

CBC, Total count, differential count, parathyroid level, FISH analysis, USG abdomen for renal system to be done later for further analysis and early identification and treatment of any problem.

Mast . Balakumaran, image of post operative status.

Age -3.5 years

REVIEW OF LITERATURE

Definition

DiGeorge Syndrone is a syndrome caused by the deletion of a small piece of chromosome 22. The deletion occurs near the middle of the chromosome at a location designated q11.2 i.e., on the long arm of one of the pair of chromosomes 22. the features described are as follows,

Cardiac Abnormality (especially Tetralogy of Fallot)

Abnormal facies

Thymic aplasia

Cleft palate

Hypocalcemia.

Incidence - 22q11.2 deletion syndrome affects an estimated 1 in 4000 live births.

Nomenclature - Velo-Cardio-facial syndrome, Sprintzen syndrome, DiGeorge syndrome, Sedlackova syndrome, conotruncal anomaly face syndrome.

Causes - Genetic deletions (loss of a small part of the genetic material), Other mechanical causes are unknown.

Pathophysiology

Deletion of large region on chromosome 22, Thymic hypoplasia leads to failure of formation of 3rd & 4th pharyngeal pouches early during embryogenesis. And cellular immune deficit and other structure forming at the same time are also affected. anomalies of great vessels (right sided aortic arch), esophageal atresia, congenital heart disease, dysmorphic facial features.

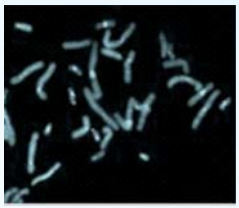
Clinical features

- Congenital heart disease (40% of individuals), particularly conotruncal malformations (tetralogy of Fallot, interrupted aortic arch, ventricular septal defect, and persistent truncus arteriosus)

- Palatal abnormalities (50%), particularly velopharyngeal incompetence (VPI), submucosal cleft palate, and cleft palate; characteristic facial features (present in the majority of Caucasian individuals) including hypertelorism.
- Learning difficulties (90%) but broad range, Hpocalcemia (50%)(due to hypoparathyroidism)
- Significant feeding problems (30%), Renal anomalies (37%), Hearing loss (both conductive and sensorineural) (Hearing loss with craniofacial syndromes), Laryngotracheoesophageal anomalies.
- Growth hormone deficiency, Autoimmune disorders, Seizures (without hypocalcemia), Skeletal abnormalities

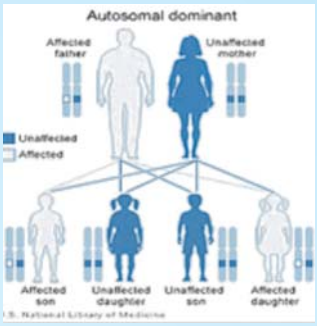
Diagnosis and testing

- Clinical observation
- Antenatal ultra sound to identify the palatine abnormalities and CHD
- Amniocentesis
- The deletion syndrome is diagnosed in individuals with a submicroscopic deletion of chromosome 22 detected by fluorescence in situ hybridization (FISH).



- Cytogenetic studies and negative FISH testing shows normal in 5 % individuals.
- Reduced calcium level
- Radiological study done to know the cardiomegaly.
- ECHO- to identify the cardiac anomalies.
- Genetics screening.

22q11.2 deletion syndrome is inherited in an autosomal dominant pattern.(22q11.2 deletion syndrome are missing about 3 million base on one copy of chromosome 22 in every cell of their body).



Treatment

There is no cure for deletion syndrome. Certain individual features are treatable using standard treatments.

- Blood transfusion for thrombocytopenia
- Immunization with live vaccines for immune problem
- Thymus transplantation performed for thymus aphasia.
- Bacterial infections are treated with antibiotics.
- Cardiac surgery is often required for congenital heart abnormalities.
- Palatal abnormalities (velopharyngeal inadequacy(VPI) are treated with prosthesis and surgery.
- Hypoparathyroidism causing hypocalcaemia often requires lifelong vitamin D and calcium supplements.

Nursing Diagnosis

- Cardiac output decreased related to structural defect of the heart.
- Tissue perfusion ineffective related to inadequate blood supply to the tissues
- Breathing pattern ineffective related to pulmonary congestion & aspiration of food

- Feeding interrupted related to cleft palate
- Nutritional imbalance less than body requirement related to less intake of food.
- Growth and development delay/impaired related to genetic defect.
- Communication interrupted related to cleft palate
- Parental coping ineffective related to chromosomal defect of the child.
- Aspiration risk for related to Cleft palate
- Infection risk for related to auto immune disorders.
- Therapeutic management ineffective related to lack of knowledge on chromosomal disorders.

Complication

Mental retardation. Cognitive impairments, Speech and language delay, Psychiatric illness (eg. Schizophrenia), Autoimmune disorders (hypothyroidism and hypoparathyroidism), Infection (problem with the immune system's T-cell mediated response due to absent / hypoplastic thymus, Convulsion due to hypocalcemia.

Pognosis- There is no cure for 22q11.2 deletion syndromes. Corrective surgeries can lengthen the child's lives.

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