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DOWN'S SYNDROME : A CASE REPORT

Gynaecology

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ABSTRACT

Down's Syndrome or Trisomy 21 is the commonest form of chromosomal abnormality which occurs in new borns/infants which leads to stunted growth and mental retardation. It is considered as a genetic disorder with a defect in all or a part of a third copy of chromosome 21. This case report highlights clinical presentation of 3 months old patient with Down's Syndrome.

KEYWORDS

Chromosome, Ultrasonography, Syndrome

INTRODUCTION :

Downs syndrome is a congenital disorder characterised by several delayed psychomotor dvelopment and malformed features with an increased risk of concomitant congenital disorders such as congenital heart and gastrointestinal defects, celiac disease and hypothyroidism. In humanbeings it affects 10 per 10,000 live births although in recent years prevalence has increased significantly.[1] Down syndrome is a chromosomal disorder caused by an error in cell division that results in the presence of an additional third chromosome 21 or " trisomy 21.2 Death certificates of patients with DS has reported most frequent medical disorder is congenital heart defects and respiratory infections[1].Down syndrome is named after John Langdon Down. The features usually range from mild to severe[3].

The most prominent physical characteristics of this syndrome are delayed physical and mental growth, small chin, slanted eye, poor muscle tone, a flat nasal bridge, a single crease of the palm and a large tongue along with other features which includes big toe, abnormal pattern of fingerprint and short fingers[4].

Diagnostic hypothesis of DS include chromosome analysis (karyotype examination) which can be performed in the pre and postnatal period .To determinate the recurrence risks of DS Cytogenetic investigation should be done, helping the genetic counseling process[5].

As present there is no cure for cognitive impairment in downs disease , the primary objective is to understanding and treating DS is linking individual genes to particular manifestations of disease[6].Respiratory tract infection is 2nd leading cause of death in children with Downs syndrome so use of prophylactic measures and respiratory syncytial virus (RSV) prophylaxis , additional immunizations are preventive interventions to be undertaken [7].

CASE REPORT :

A 03 months old female patient presented with complaints of Fever since 3 days, subsided on taking treatment not associated with excessive cry while micturation. Initially Patient had fever which is followed by cold and cough with nasal discharge, since past 2 days patient having noisy breathing and respiratory distress. Patient Birth weight was 1.5 kgs and did not cried immediately after birth. He also noticed weight loss prior to admission .

On physical examination patient her vital signs were
Heart rate- 100bpm, pulse rate-56/min
Respiratory system- BAE(+), Bilateral wheeze (+)
Epicanthal folds of eyes,
depressed nasal bridge,
protruded tongue
clinodactyly (+)
short neck.

Past Medical History -: Patient has history of NICU admissions for 5 days due to Neonatal Jaundice
Patent was immunized till date

Obstetric History of mother: A 31 years old female patient was admitted into hospital with history of pregnancy induced hypertension since 3rd trimester with average BP value (150/100 mg) and was on medication with antihypertensives for a period of 3 months, during delivery patient BP was raised and emergency LSCS done, patient delivered a female baby of weight 2.1 kg , did not cry immediately after birth.

Past medication History of mother: During gestation period patient was given T. nifedipine -10 mg/BD for last 3 months of 3rd trimister , before onset of labour inj labetolol -100mg and T.nifedipine-5 mg given for about 5 hours apart reduce sudden increase in BP. After postpartum period patient BP was not normalized and advised to continue antihypertensive drug such as T.labetolol-10 mg/day.

Lab investigations: Apart from the above mentioned physical examinations the patient underwent routine test as well as definitive tests.

Routine Test: complete blood picture revealed decrease Hb levels-8.5 gm/dl as well as decrease RBC count -3.1 mill/cumm, a total WBC count of 5,000/cumm with a differential count of 25% lymphocytes, and Neutrophil 70 %. Platelet count – 1.0 lakhs/cumm.- which revealed normocytic normochromic anaemia with relative neutrophilia and moderate thrombocytopenia.

Electrolytes revealed decrease Potassium and calcium levels of 3.2 mEq/L and 0.84 mmol/L respectively

Blood gas analysis was done which revealed decreased pO2 and SPO2 levels that is 2.5mm/Hg and 43.3% respectively and increased HCO3 and Base excess levels such as 19.6 mml/L and -5.1 mmol/L respectively

Patient was perfomed Chest X ray to rule out respiratory tract infections and was found to have broncholitis, with increased bronchovascular margins noted in bilateral lungs field more prominent on right side. Thyroid Profile was found to be normal.

Difinitive Tests: chorionic Villus Sampling (CVS)- revealed downs syndrome .

Lab investigations of mother :

Ultrasonography : Ultrasonography during 3rd trimester of mother detects fluid at the back of foetus neck which revealed Downs syndrome

Treatment: Patient was given therapy only to give symptomatic relief such as Syp.. PCM-5ml/TID, Neb Asthalin-3cc 6th hourly, Neb Adrenaline 0.5 cc+2 ccNS, suspension- cefuroxime-5ml

Discharge medication:- Patient was discharged after 5 days of appropriate treatment after ruling out the disease. Patient guardians were educated about disease and management mainly includes physical therapy.

DISCUSSION :

Downs syndrome is a chromosomal abnormality in new born infants associated of phenotypes such as including learning disability, heart problems, leukemia in childhood and as well as Alzhemers disease[8]. Age of mother conceiving pregnancy is significant risk factor because as age increases risk of chromosomal abnormality increases [9].

Clinical features of Downs syndrome include, flat nasal bridge upward slanting palpebral fissures, brushfield spots, , small ears, small nose, excessive skin at the nape of neck, common medical problems include- hearing problems, vision problems, cataracts, obstructive sleep apnea,otitis media, congenital heart disease, thyroid diseases, seizures, hematologic problems [10] .In the present case Clinical features of the patient were similar to the previous studies such as Epicanthal folds of eyes, depressed nasal bridge, protruded tongue, short neck.

Diagnosis of Downs syndrome primarily includes screening test which are non invasive test such as ultrasonography , definitive diagnostic test includes Karyotyping cultured fetal cells such as chorionic Villus Sampling (CVS), aminocentesis[11].During the 2nd and 3rd trimistersof pregnancy Percutanious umbilical blood sampling (PUBS) is a methodfor obtaining fetal blood along with ultrasnography[12] . In the present case the main risk factor that is the age of mother is present. Other diagnostic parameters include ultrasonography, Chorionic Villuys Sampling were done to confirm the diagnosis of the patient.

Individuals with DS have behavioural and psychiatric problems , at present current evidence based therapies established based on three major approaches such as1) previous experience on on other diseases such as AD 2) unspecific nutritional approaches 3)early interventions programmes.Therapies to improve CNS functions include specific(Acetylcholine, serotonin, Antiinflammatory) and nonspecific treatments such as Nootropic , harmone and, nutritional12 . As patient was suffering from bronchiolitis treatment was given to treat the same.

CONCLUSION:

Down's syndrome or Trisomy 21 is regarded as one of the most common chromosomal abnormality occurring in new borns/ infants.To improve our understanding of the disease numerous theories have been put forward. This case report highlights different aspects of this condition which might help in the diagnosis of the condition.

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