International Journal of Pharmaceutical Research and Applications
Volume 8, Issue 1 Jan-Feb 2023, pp: 1109-1112 www.ijprajournal.com ISSN: 2249-7781

Down Syndrome: A Case Report

Kamali[1], Azhar[1], Anil[1], Rangaswamy[1], Vinod[2]
1. Pharm-D Intern
2. Assistant Professor
Department of Pharmacy Practice, TVM College of Pharmacy, Ballari, Karnataka, India.

Submitted: 12-01-2023 Accepted: 24-01-2023

ABSTRACT
Down syndrome is one of the leading causes of intellectual disability and millions of these patients face various health issues including learning and memory, Congenital heart disease (CHD), Alzheimer's disease (AD), leukemia, cancers and Hirschprungdisease(HD). This article highlights the varied clinical presentation of 4-month-old patients with Down's syndrome. The patient was diagnosed with Down's syndrome with Congenital Heart disease and Acute gastroenteritis with severe dehydration.

Keywords: Chromosome, Trisomy 21, Down syndrome, DS, Karyotype, CHD.

I. INTRODUCTION

Down‟s syndrome, also known as Trisomy 21. According to the National Institute of Child Health and Human Development, Down syndrome (DS) occurs in approximately 1 in 800 newborns, it is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21(47 chromosomes)[1]. It is the most frequently occurring chromosomal abnormality in humans and affects between 1 in 400-1500 babies born in different populations, depending on maternal age, and prenatal screening schedules. It is coupled with mental retardation, congenital heart defects, gastrointestinal anomalies, weak neuromuscular tone, dysmorphic features of the head, neck and airways, audiovestibular and visual impairment, characteristic facial and physical features, hematopoietic disorders and a higher incidence of other medical disorders[2]. Trisomicfetuses are at elevated risk of miscarriages and DS people have an increased incidence of developing several medical conditions[3]. Among the more common physical findings are small brachycephalic head,hypotonia, epicanthal folds, flat nasal bridge, upward-slanting palpebral fissures, small mouth, small ears, excessive skin at the nape of the neck, Brushfield spots,short fifth finger with clinodactyly,single transverse palmar crease, and wide spacing between the first and second toes, often with a deep plantar groove. The degree of cognitive impairment is variable and may be mild (IQ of 50–70), usually moderate (IQ of 35–50), or occasionally is severe (IQ of 20–35)[4]. The coincidence rate of both Klinefelter syndromes and Down in the same individual is estimated to lie in the range (of 0.27 to 0.7 × 10-5)[5]. Several cases of trisomy 21 and double aneuploidy of XXY have been published since the first report by Ford et al.In a pair of monozygotic twins,this abnormality has also been recently described. Further, both the sibs of the proband showing 48, XXY,+21 were found to exhibit trisomy 21 in yet another study[5]. The most prominent characteristics of the condition include physical growth delays, subnormal mentality and a severe degree of intellectual disability which is one of the most common feature[s7].

Advancing maternal age and aberrant recombination remain the only conclusive and well-documented risk factors for DS pregnancy[1]. About 85-88% of DS is associated with errors from the maternal egg about 5-9% originates from paternal sperm while the remaining 1-3% are attributed to mitotic cell division errors that happen after fertilisation[1]. The AMA risk factor in DS applies mainly to the trisomy 21 variant of DS although AMA is the primary risk factor for DS birth, due to higher birth rates in younger women about 80% of children with DS are born to women under 35 years of age[1].

The clinical features of a 4-month-old boy who exhibited the karyotype **48, XXY,+21** have been presented in this paper.

II. CASE REPORT

A 4-month-old baby has been admitted to the pediatrics department with complaints of loose stools 8-10 episodes since 1 day. The past history of a baby includes Trisomy. The baby cried at birth with a period of 8 months 5 days gestation with a birth weight of 2.5 Kgs. The baby was admitted

International Journal of Pharmaceutical Research and Applications
Volume 8, Issue 1 Jan-Feb 2023, pp: 1109-1112 www.ijprajournal.com ISSN: 2249-7781

and phototherapy was given for 2 days the baby was on formula feed since birth. Developmental history includes neck holding not attained recognized by mother since 4 months.

On objective examination, the pulse rate was 110bpm, Respiratory rate 30cpm and the Bilateral Dorsal pedal artery was positive.

On systemic examination, there is a shrunken abdomen soft non-tender, Bowel sounds positive, and the patient was drowsy with bilateral pupils reactive to light.

Parameters	Day-1	Day-2
Hemoglobin	12.4	
Total count	13100	
Neutrophils	52	
Lymphocytes	38	
Platelet	2.10	
Urea	182	50
Creatinine	2.5	0.6
Sodium	138	133
Potassium	5.9	4.3
Chloride	119	81
CRP	55.1	

Table 1.1 Laboratory Investigation of the patient

Other examination includes :

Investigations	**Results**
2D echo	• Sinus solitus : Levocardia • Complete AV canal defect • Large inlet ventricular septal defect bidirectional shunt • Large ostium primum and Secundum Atrial septal defect: left to right shunt • Bilateral branch pulmonary artery stenosis
Ultrasound Abdomen	No significant abnormality
neurosonography	Prominent right lateral Ventricle

Table 1.2 Various Investigations findings of a patient

International Journal of Pharmaceutical Research and Applications
Volume 8, Issue 1 Jan-Feb 2023, pp: 1109-1112 www.ijprajournal.com ISSN: 2249-7781

Fig 1.1 Showing the image of a child with Down Syndrome

Karyotype analysis :

Chromosome analysis revealed an abnormal female chromosome complement in all cells examined with three copies of chromosome 21, resulting in trisomy 21. These abnormalities are consistent with the diagnosis of Down syndrome.

The patient was diagnosed with Down's syndrome with Congenital Heart disease, and Acute gastroenteritis with severe dehydration.

The child was managed by IV fluids and Iv antibiotics, antidiarrhoeal measures were taken. The child gradually improved and is now hemodynamically stable and hence discharged.

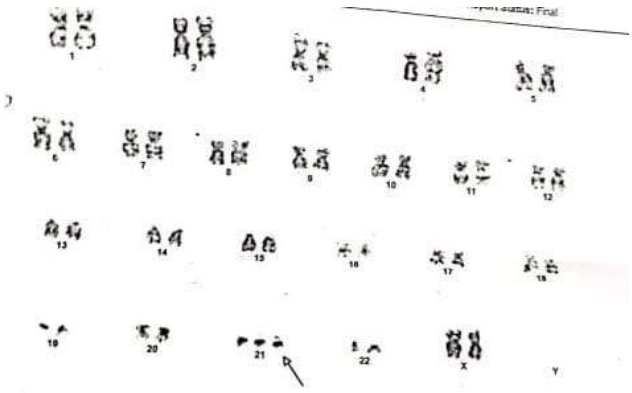

Fig 1.1 Karyotype Analysis showing Triple copies on Chromosome-21

III. DISCUSSION

The child is4 months old and exhibited features of typical Down Syndrome and has the karyotype **48, XXY+21**.Studies on the incidence of an XXY chromosome pattern among Down individuals have revealed that this double aneuploidy might be more frequent than predicted by multiplying the frequencies of individual aneuploidies. Recurrence risk as a factor expresses the possibility of a woman who had prior DS births having subsequent once[1]. For women who had prior trisomy childbirth, there is an approximately 1% recurrence risk of having another child with trisomy 21[1]. The carriers of a balanced

International Journal of Pharmaceutical Research and Applications
Volume 8, Issue 1 Jan-Feb 2023, pp: 1109-1112 www.ijprajournal.com ISSN: 2249-7781

translocation of chromosome 21 are also documented risk factors for DS pregnancy.

Hackshaw A K et al. in their study proposed new screening methods in which measurements obtained during the first and second trimesters are integrated to provide the risk status of having a pregnancy with DS[7].

Alzheimer's disease, heart defects, leukemia, hypertension, and gastrointestinal problems are the various clinical conditions associated with DS[3].

Development of Down syndrome brain is associated with a reduction in the neuronal number and abnormal neuronal differentiation[7]. It has been previously reported that Down syndrome neurons degenerate subsequently and undergo apoptosis[7].

Kernard et al. in their review stated that there are several ultrasound markers in Down syndrome which include nuchal fold thickness, cardiac abnormalities, duodenal atresia, femur length, and pyelectasis[7].In this case, the patient was noticed with cardiac abnormalities such as Sinus solitus: Levocardia, Complete AV canal defect, Large inlet ventricular septal defect bidirectional shunt and Large ostium primum and Secundum Atrial septal defect: left to right shunt. Seizure disorders are present in 5-10% of patients[7]. The most common seizures observed in infancy are Infantile spasms. However, this case has no history of seizures.

DS patients constitute approximately 5% of cases of HD. Duodenal stenosis(DST) and imperforate anus(IA) are 260 and 33 times more likely to occur in DS[3]. Early in the neonatal period infants with duodenal atresia or DST present with bilious vomiting. If left untreated it results in severe dehydration and electrolyte imbalance.In this, the patient was diagnosed with acute gastroenteritis with severe dehydration.

Development of Down's syndrome brain is associated with a reduction in the neuronal number and abnormal neuronal differentiation[7]. However, the Neurosonography of the patient reveals a Prominent Right lateral ventricle.

IV. CONCLUSION

Trisomy 21 or Down syndrome is a chromosomal abnormality that is most commonly detected in newborns and infants complications include cardiovascular diseases, Gastrointestinal diseases, Delayed developmental milestones, Mental retardation, Alzheimer's disease and other serious complications. Various theories have been proposed to understand the causal relationship between those complications with Down's Syndrome. This case indicates that various clinical conditions may be due to extrachromosomal abnormalities. Thus these various clinical aspects help in the diagnosis of the condition.

REFERENCES

[1]. Dr. Shilpa karkeraAssitant professor Department of Anatomy and neuroscience Trinity School of Medicine Rathomill, Kingston A current knowledge of “Down Syndrome: A Review” International Journal Dental and Medical Sciences Research Volume 3, Issue 1, Jan-Feb 2021 pp 805-810

[2]. Mohammed Kazemi, Mansoor Salehi, Majid Kheirolahi Down Syndrome: Current status, Challenges and Future Perspectives Int J Mol Cell Med Summer 2016; Vol 5 No 3 126

[3]. Ambreen Asim, Ashok Kumar, et al. "Down syndrome: an insight of the disease” Journal of Biomedical Science (2015) 22:14,

[4]. Marilyn J .Bull, MD, FAPP, Tracy Trotter, MD, FAPP, et al. Health Supervision for Children and Adolescence American Academy of Pediatrics Volume 149 No 5, May 2022

[5]. Cyrus Cyril, Chandra N, et al. Down Syndrome child with 48, XXY,+21 Karyotype Indian Journal of Human Genetics January-April 2005 VOL 11 Issue 1

[6]. Dr. Rahamathulla Khan M.D.S, Dr. Abdallah I, Abdallah PHD, Dr. Virginia Antony M.D.S Down Syndrome- A case report

[7]. Anoop Kurian Mathew, Antony ShijoyAmaladas, et al.Clinical presentation of Down's Syndrome: A Case report Journal of Medical Research 2017;3(3):107-109