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CASE REPORT

KABUKI SYNDROME–A RARE CASE REPORT.

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Abstract

Kabuki syndrome is a rare congenital anomaly, characterized by five fundamental features, the "Pentad of Niikawa": dysmorphic facies, skeletal anomalies, dermatoglyphic abnormalities, mild to moderate mental retardation and postnatal growth deficiency.In addition,they may also manifest cardiac anomalies, urinary anomaly, feeding difficulties,hearing loss and hypotonia.

Here we present a case of 1year old female baby who had abnormal facies, global developmental delay and poor weight gain with delayed dentition and microcephaly. The ECHO cardiography showed large Atrial septal defect(ASD).Because of presence of these features a diagnosis of Kabuki syndrome was made.

There are few reported cases of kabuki syndrome from India. This is first case reported from Odisha, signifying its global prevalance but rare reporting.

Introduction:-

Kabuki syndrome (KS) (Kabuki make-up syndrome, Niikawa – Kuroki syndrome) is a rare congenital syndrome, which was initially described simultaneously by Niikawa *et al*.[1] and Kuroki *et al*.,[2] in 1981. It was described initially in Japan but is now known to occur in many other ethnic groups. The prevalence of Kabuki syndrome in the Japanese population has been estimated to be 1/32,000[3] .Sporadic cases of KS are found all over the world, and in India it is mainly found in the eastern regions.[4]..It is characterized by distinctive facial features (eversion of the lower lateral eyelid, arched eyebrows with the lateral one-third dispersed or sparse, depressed nasal tip, and prominent ears), skeletal anomalies, dermatoglyphic abnormalities, short stature, and mental retardation. The designation Kabuki make-up refers to the resemblance of the facial features with the characteristic make-up used by actors of Kabuki, a traditional Japanese theatrical form[5].The children may have seizure,microcephaly,hypotonia,nystagmus,strabismus,short stature,scoliosis,short fifth finger,problems with the hip and knee joints,cleft palate,high arched palate and dental problems.[6]

Case Report:-

A 1 year old first order female baby was admitted to the emergency department of MKCG paediatric department with complains of fever and fast breathing for 10 days.She was born out of a non consanguineous marriage by normal vaginal delivery after term gestation to a primigravida mother. There was history of failure to gain weight and developmental delay and history of seizure in the neonatal period but was not on any anti-epileptic drugs.There

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were feeding difficulties.The current weight was 4.5kg with a birth weight of the baby was 2.7kg .Her height was 60cm and head circumference was 38cm.(<3 SD) The baby had not attained developmental milestones like neck holding ,sitting and standing with or without support.She had generalized hypotonia and facial dysmorphism with high arched eyebrows sparse laterally, long palpebral fissures with everted lower eyelids, long eyelashes accompanying microcephaly. Her nose was broad with depressed tip, and ears were prominent and protruding.(Fig.1 and 2). On oral examination she had high arched palate and only 2 lower incisior tooth(Fig 3).There was a loud systolic murmur of grade 4/6 at the mitral area which was heard all over the chest.On doing ECHO cardiography ,a large 25mm ASD was found(Fig 4).There was tachypnoea and hepatomegaly. The other system findings were normal. She was diagnosed as Kabuki syndrome due to her clinical findings with large ASD with congestive cardiac failure. She improved clinically during her stay at the hospital with decongestive therapy and antibiotics and was discharged in 10 days with referal to higher center for appropriate intervention.

Fig 1 and 2:- showing dysmorphic facies with prominent protruding ears.

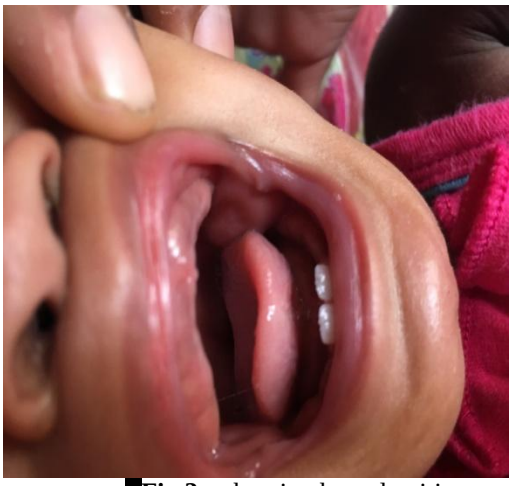

Fig 3:- showing hypodentition

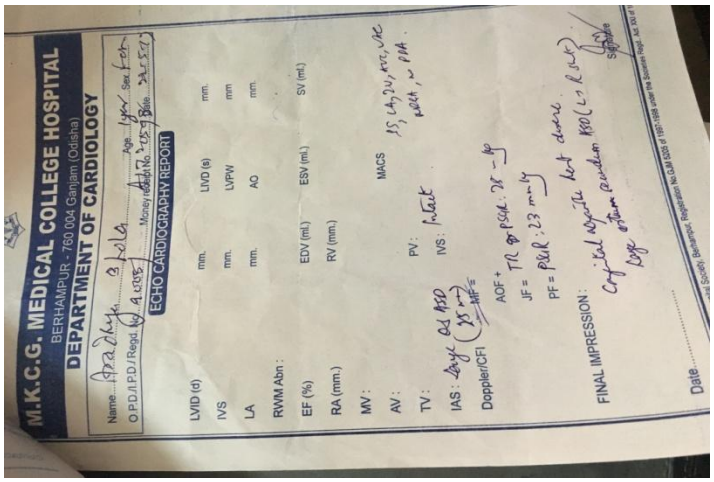

Fig 4:- showing ECHO cardiography report

Disscussion:-

Kabuki make-up syndrome was established with clinical findings. Currently, there is no consensus on the diagnostic criteria for Kabuki syndrome and there is no clinically available genetic test to confirm the diagnosis. In 1988, Niikawa et al. reported on the clinical findings in 62 patients diagnosed with Kabuki syndrome. Based on the findings in these patients, five cardinal manifestations were defined. These included a 'peculiar face' (eversion of

the lower lateral eyelid, arched eyebrows with the lateral one-third dispersed or sparse, depressed nasal tip, and prominent ears) in 100% of their patients, skeletal anomalies (deformed spinal column with or without sagittal cleft vertebrae, and brachydactyly V) in 92% of their patients, dermatoglyphic abnormalities (fingertip pads, absence of digital triradius, and increased digital ulnar loop and hypothenar loop patterns) in 93% of their patients, mild to moderate mental retardation in 92% of their patients, and postnatal growth deficiency in 83% of their patients. There have also been a number of less frequent findings reported in Kabuki syndrome, including visceral abnormalities, premature breast development in females, and susceptibility to frequent infections[7,10] In accordance with the literature, the female patient presented with developmental delay and seizures. On the other hand she also had microcephaly. Schrander-Stumpel et al. reported 29 Caucasian patients and reviewed 60 Japanese and 29 non-Japanese patients, noting that nonJapanese patients with this syndrome had more marked neurological symptoms. In over 80% of non-Japanese patients, neurological symptoms were a major clinical problem [11]. Precocious puberty is an occasional finding in the syndrome. In recent studies, it has been reported that congenital heart defect is present in 58% of patients with Kabuki syndrome[12]. The most common finding appears to be juxtaductal coarctation of the aorta, a relatively rare heart defect, followed by VSD and ASD[13].

Conclusion:-

Careful dysmorphological examination should be performed in all patients presenting with mental retardation and epilepsy to diagnose Kabuki syndrome. The patients diagnosed as Kabuki syndrome should be followed for premature thelarche and precocious puberty.Congenital heart defects and other malformations should be ruled out.Treatment requires co-ordinated efforts of the team of specialists like Paediatricians, surgeons, cardiologist, dentist, speech pathologst, audiologist and other health care professional.The prognosis depends on associated malformations specifically Congenital heart defects,but in general it has good prognosis and most children have normal life expectancy.Early intervention is important to ensure that children with kabaki syndrome reach their potential.

Consent Was Taken From Parents For Publishing The Case.

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