

## Down Syndrome with Congenital Heart Disease (Endocardial Cushion Defect) Presenting as Central Nervous System Stroke

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### Abstract:

**F**loppy baby is the characteristic feature of down syndrome (DS). But there may be new onset weakness which may be due to various reasons like stroke from moyamoya disease, stroke from vaso-occlusive disease, stroke from venous sinus thrombosis, traumatic subdural hematoma. Brain abscess, spinal cord injury and brachial plexus injury.<sup>[1]</sup> Here we report a case of 6 month old female baby diagnosed as down syndrome with endocardial cushion defect (ECD), presenting as CNS stroke to the paediatric emergency of MKCG Medical college Berhampur, Ganjam, Odisha.

**Keywords:** Floppy baby, ECD, CNS stroke

### Introduction:

Down syndrome occurs 1 in 800 newborns. In India about 1 million babies are born every year. It has characteristic facial appearance and hypotonia. Trisomy is the most common type of DS and accounts 95% of total DS population<sup>[2]</sup>. Translocation (3%) and mosaic (2%) are the less prevalent types. DS is predisposed to many abnormalities, 75% of DS suffer from hearing loss, 50-75% suffer from sleep disorders, 60% of DS have eye diseases, 22% of DS have psychiatric diseases and around 50% of DS have CHD, of these ECD is the most common type with 43%, followed by VSD (32%), secundum ASD (10%), TOF (6%) and isolated PDA (4%). About 70% of ECD are associated with DS<sup>[3]</sup>. The ECD may result in atrio-ventricular septal defect with or without AV canal defects. It may be partial or complete. A complete AV defect indicates a both atrial and ventricular septal defect with a common AV valve. A partial defect indicates atrial septal involvement with separate mitral and tricuspid valve orifices. There is predominantly left to right shunt but in patients with long standing pulmonary overload there may be reversal of shunting of blood, this may cause cyanosis and is a poor prognostic indicator.

### Case report:

A 6 month old female baby presented to the emergency paediatric department with complains of left sided weakness of upper and lower limb (fig 1). On examination, the baby was found to be floppy, having flat facies with a small nose, small abnormally shaped ears (fig 2). There was extra space between the big toe and second toe (sandal gap) and there was deep plantar crease (fig 4). These features helped making a diagnosis of DS. The anterior frontanelle was bulged and the vitals were stable but cyanosis was present around lips. There was a grade 1 systolic murmur heard at the apex. On doing an ECHO, a diagnosis of ECD was confirmed (fig 3). The diagnosis of meningitis was made but on doing lumbar puncture, CSF study came out to be normal and TFUSG was also normal. Neuroimaging (CT/MRI) was not done due to financial constraints. Because the TLC was high and CRP

positive, a course of antibiotics was given and the child improved. The child was discharged with an advice to consult cardiologist. Hence, a clinical diagnosis of CNS stroke due to thrombosis was made.



Fig 1 Floppy baby with paucity of movement on left side



Fig 2 Showing features of DS



Fig 3 Showing ECHO report showing ECD



Fig 4 Showing sandal gap and deep Plantar crease

### Discussion:

In >2years, the most common cause of stroke in a CHD is brain abscess and in <2 years age group it is mostly due to thrombosis. Thrombo-embolic complications in children are seen at a rate of 5.3 per 10,000 hospitalized children, but there is an increased incidence of thrombosis in DS. Children diagnosed with DS and CHD have 95% chances of thrombosis compared to normal children<sup>[5]</sup>.

### Conclusion:

DS is a risk factor for many diseases like CHD, leukemia, thyroid diseases, autoimmune disorders, epilepsy, intestinal obstruction and increased susceptibility to infections hence a complete investigation should be done periodically.

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