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VICI SYNDROME IN A MID-EASTERN INFANT

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ABSTRACT:

We describe an infant with Vici syndrome born to healthy, Consanguineous parents, with Oculocutnaeous Hypopigmentation, Agenesis of Corpus Callosum, and cataracts. He had postnatal growth Deficiency and profound developmental retardation. Decreased ejection Fraction of the left ventricle was demonstrated on echocardiographic examination. He also had micrognathia and high-arched palate, and Developed postnatal hydrocephalus (Dandy Walker malformation). The infant died at the age of 4 ½ months. Death was presumably due to Cardiac arrhythmia. These findings strongly suggest the diagnosis of Vici syndrome.

Chiyonobu et al [2002] suggeted autosomal recessive inheritance on the basis of occurrence of the syndrome in three pairs of siblings of both sexes to unaffected parents. Positive consanguinity of unaffected parents in our case may support this hypothesis but still needs to be proved.

INTRODUCTION

In 1988 Vici et al. described 2 brothers with agenesis of corpus callosum (ACC), cutaneous hypopigmentation, cataracts, cleft lip and palate, Combined immunodeficiency, and severe psychomotor retardation. In 1999, Del Campo et al reported on 4 children, 2 sibs and 2 other Unrelated children, with similar findings confirming a unique syndrome Designated Vici syndrome. Chiyonobu et al [2002] described 2 sibs, Miyata et al [2007] described another 2 sibs, McClelland et al [2010] Described one case and Al-Owain et al[2010] added another case.

CLINICAL REPORT

A boy was born normally at term after an uneventful pregnancy to Consanguineous parents, a 27 year old mother and 32 year old father. They had 2 previous healthy children. Family history was unremarkable. There was premature rupture of membranes 3 days prior to delivery. Birth weight was 3 Kg. He had light-pigmented skin and moderately hypopigmented retinue. His parents have dark skin color and deep Brown retinue appropriate to their ethnic group. On day 2 of life he was Jaundiced and treatment with phototherapy began. On day 5, he Developed cardiac arrest and was resuscitated by mechanical ventilation And ionotropic drugs.

Magnetic resonance image showed agenesis of corpus callosum and Early changes of Ischemic Encephalopathy. No evidence of Hydrocephalus was seen at that time. (Fig.1a, b).

Echocardiography showed an open foramen ovale and patent ductus arteriosus with elevated pulmonary artery pressure. After 20 days the Infant was extubated. He was discharged from the neonatal unit at age of 40 days.

At 3 months of age the child presented with fever, respiratory distress, low oxygen tension, bilateral inspiratory crackles due to acute chest Infection, which was treated with antibiotics and oxygen therapy. He responded favorably. He was again noted to have Oculocutaneous Hypopigmentation. (Fig.2); the head was enlarged with OFC on the 95 th centile and with wide anterior fontanel, suggesting Hydrocephalus.

Growth was deficient with length < 3 rd centile for age; weight was on the 5 th centile. He had cataracts (Fig.3), roving nystagmus, coloboma of the optic discs and Hypopigmented retinue, Micrognathia and high-arched palate. (Fig. 4). He had an ejection murmur at left sternal edge; ECG Showed biventricular hypertrophy and chest radiogram showed Cardiomegaly. An echocardiogram showed decreased ejection fraction of the left ventricle (50%). The child did not show any signs of Development, he appeared disinterested in his surroundings with marked Head lag and no response to light or sound. Motor power was Decreased; he had truncal hypotonia with hypertonicity and brisk deep Tendon reflexes in upper and lower limbs. The child developed repeated Seizures that were controlled in the acute stage by diazepam and Phenobarbitone intravenously. He was then maintained on oral Barbiturates.

Computerized scan of the brain showed evidence of hydrocephalus. There was thinning of cerebral parenchyma, posterior fossa cyst and Dilatation of all ventricles (Dandy Walker malformation) (Fig. 5). A Ventriculo-peritoneal shunt was inserted when he was 4 months old. One Week later, he developed fever and respiratory symptoms.

Bronchopneumonia was diagnosed and treated with antibiotics and Oxygen. Immununological investigation showed normal T-cell; however, Serum immoglobulins were not tested because of the young age of the Patient. Urine testing showed no abnormalities and blood gas analysis showed no acidosis. No abnormalities were detected on metabolic screening. Full blood counts were within the expected values for age.

The child was taken home on oral barbiturates. At the age of 4 ½ Months, he arrived dead at the hospital. Cardiac arrhythmia was thought to be the direct cause of death. Autopsy was not authorized.

DISCUSSION

Prior to this report 12 cases have been described with features Suggesting the diagnosis of Vice syndrome. The common features found in all those cases include ACC, hypopigmentation of skin and retina or Oculocutaneous albinism, deficient growth and development, recurrent Infections, and cardiac abnormalities.

Hypotonia, (particularly truncal), Poor motor function, cataracts, and nystagmus were among the very Common features. ACC, hypo pigmentation, deficient growth and Deficient development probably constitute criteria of diagnosis.

Chiyonobu et al suggested Autosomal recessive inheritance on the Basis of occurrence of Vici syndrome in 3 pares of sibs, we support this Suggestion on the basis of occurrence of the syndrome on another pair of sibs reported by Miyata et al and consanguineous marriage in both our case and Al-Owain et al case.

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