Vici Syndrome: A Case Report of Non-Identical Twins with Oculo- Cutaneous Albinism, Recurrent Infections and Absent Corpus Collasum

Abstract

Vici syndrome is a rapidly progressive, neurological disease of childhood. It is associated with complete agenesis of the corpus collasum, occulocutaneous albinism, immunodeficiency, cardiomyopathy and neuromuscular defects. This a case report of non identical twins who presented with failure to thrive, seizures, developmental delay associated with albinism, absent corpus collasum, and recurrent infections.

Keywords: Vici syndrome; Albinism; Immunodeficiency; Absent corpus collasum; Seizures; Developmental delay; Infections

Case Report

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Abbreviations: CBC: Complete Blood Count; SCID: Severe Combined Immunodeficiency; EEG: Electroencephalogram; MRI: Magnetic Resonance Imaging; PBP: Peripheral Blood Picture; IgG2: Immunoglobulin Gamma-2; Mg: Milligram; kg: Kilogram

Introduction

Vici Syndrome is an autosomal recessive multisystem disorder that was described by vici et al. [1], characterized by global developmental delay, seizures, cataracts, absent corpus collasum, cleft lip and palate and cardiomyopathy [1]. Several case reports have described this condition over the past 3 decades. Here we describe a case of non identical twins who presented with this condition early in infancy.

Case Presentation

We present two cases of 3 months old non identical twins, an outcome of a consanguineous marriage. They were delivered to an uneventful pregnancy via Caesarean Section due to twin pregnancy. They presented with, failure to thrive, feeding reflux regurgitation, developmental delay, left sided focal tonic clonic seizures since they were 7 days old. They had a past history of a chest infection, one week before their presentation to which they sought medical attention and were treated for by oral medications. Their family history was significant for Albinism, with normal development and outcome.

On examination: They had oculocutaneous albinism dehydrated, very floppy, with a weak cry, with a positive moro reflex. Their weight and height were below the 5th centiles for their age. They had silvery white hair with fair skin and blue reddish eyes. They had no palpable lymphnodes. Anterior fontanelle was normal with 3*3 centimeters in diameter; they had a high arched palate with oral mucocutaneouscandidaisis. Cardiovascular examination showed a systolic murmur in the mitral area, with normal 1st and 2nd heart sounds. Chest examination was clear. Abdominal examination was unremarkable. Central Nervous System examination revealed they had horizontal nystagmus, poor suckling reflex with generalized hypotonia and hyporeflexia.

The patients were admitted with the pediatric neurology unit; for seizure control, nutritional support and rehydration. One week into their admission they developed urinary tract infection, and were prescribed parentral antibiotics for 5 days. Their complete blood count (CBC), in twin 1 and twin 2 respectively: Haemoglobin: 9.2 g/dl and 9.5 g/dl WBCs: 10.700 cells and 7.500 cells /mm^3, Platelets: 453,000 and 444,000. Lymphocytes were: 65% and 51%, neutrophils: 28% and 38% with mixed cells: 7.4% and 11% respectively. Lymphocytosis in both twins was normal and corresponding with their age. Their peripheral blood picture showed features of mild hypochromic anaemia with normal Red and white blood cells in morphology and count, with large platelets. Five days later their mother complained of cough that was especially worse during the night, associated with diarrhoea and high grade fever that was not responding to oral paracetmol and cold sponging. They had coarse crackles all over the chest, chest X-rays revealed upper lobes consolidation and features of aspiration pneumonia. Blood cultures were taken for both twins and revealed MRSA in twin one and Enterococci in the 2nd twin. They were treated with intravenous antibiotics namely Ceftazidime (50mg/kg/ dose every 8 hours) and Vancomycin (15mg/kg/dose every 8 hours) for 14 days. The twins conditions improved gradually and they were discharged two weeks later after excessive nutritional counseling, as well as assuring they were infection-free. During the 2nd week of their admission, a consultation was done to the clinical immunology unit, and their immunological tests revealed: twin 1 having a relatively normal lymphocyte subsets count. Whilst twin 2 showed a global decrease in lymphocyte

subsets numbers suggestive of possible severe combined immunodeficiency (SCID) despite normal immunoglobulin levels.

Also, other special investigations done included Electroencephalogram (EEG): which revealed a suppression burst pattern indicating encephalopathy. MRI Brain scan showed: absent corpus collasum. Hair microscopy: silver whitish hair with clumping of melanin granules. Echocardiography was requested to check for signs of cardiomyopathy. Muscle biopsy not done.

Differential diagnosis: Vici Syndrome, Chediak Higashi Syndrome (CHS) and Hermansky Pudlak Syndrome. The patients were discharged to be followed up closely at the pediatric neurology and clinical immunology and allergy clinic monthly. The twins were considered as cases of Vici Syndrome. Chediak Higashi Syndrome was ruled out as the patients' platelets were large but did not contain any cytoplasmic granules. And their reflux and neurological features including the absent corpus collasum, and the EEG suppression burst pattern of encephalopathy with the absence of hepatosplenomegaly did not favor a diagnosis of CHS or Hermansky Pudlak Syndrome. These twins are the first two cases of Vici Syndrome to be reported in Sudan.

Discussion

Vici Syndrome, is a rare congenital multi system disorder that is inherited in an autosomal recessive pattern, it is characterized by failure to thrive, hypodigmentation, immunological defects evident by recurrent infections, neurological abnormalities of developmental delay, seizures, cataracts, cardiomyopathy and neuromuscular abnormalities; with the later only being described in a few recent case reports [2]. These non identical twins certainly express most of the features of Vici syndrome. During the first three weeks of their admission extensive efforts were done to reach a diagnosis of Chediak Higashi syndrome especially since they both had extensive oculocutaneous albinism, with silvery white hair, supported with their history of recurrent infections and developmental delay. However, they showed no hepatosplenomegaly and a few days later their MRI brain scans and EEG findings notably the absent corpus collasum guided the diagnosis towards a different direction. Also, their peripheral blood picture showed normal white cell morphology with relatively large, but non granular platelets. Hair light microscopy showed a few but almost absent melanin granules that are sparsely distributed in the hair shaft. All of which does not comply with the clinical picture of CHS decribed by of Shashikant CUP et al. [3] in their case report [3].

Furthermore, these twins did not exhibit any signs of Hermansky Pudlak Syndrome clinical phenotypes which were described by Laura D et al. [1], especially with the absence of bleeding diathesis, and normal neutrophil count and morphology in relation to their age, as described above.

In 1988, Vici et al. [4] described two siblings, brothers; who presented with a congenital malformation that featured cutaneous hypopigmentation, convulsions, neuropsychological developmental delay, ceflt lip and palate, bilateral cataracts with recurrent respiratory infections, both patients described in this study died early at the ages of 2 and 3 years old. Their investigations revealed absent corpus collasum, hypoplasia of cerebellar vermis, thymus and peripheral lymphoid tissue. Their immunological tests done at the time revealing CD4 T cells depletion with low serum levels of IgG2; therefore, pointing towards a pathologic mechanism characterizing this malformation which was later given his name to be known as Vici Syndrome [4].

Among the reported cases of Vici syndrome most of them described cardiomyopathy as a main feature of this malformation. Our two twin patients only had a systolic murmur in the mitral area which is later to be confirmed by Echocardiography. They had an intact but high arched palate but no cleft lip. Most reported cases of this syndrome described recurrent infections, with variable immunological phenotypes ranging from severe immunodeficiency to reported patients having a normal immune system, thus almost all cases of Vici syndrome old and new should be extensively investigated with their immunological systems largely studied as done and recommended by Finocchi A et al. [5] study done in 2011. Finocchi A et al. [5] patient's needed and was given immunoglobulin replacement despite a mildly affected cellular immunity, because he had low immunoglobulins levels [5].

Thereafter, a recent study described a phenotype of Vici syndrome with some neuromuscular abnormalities evident on muscle biopsy, hence increasing and widening the clinical picture and phenotype of Vici syndrome. This recent report study by McClelland V et al. [6] reviewed a Vici syndrome patient with sensorineural deafness, whose muscle biopsy showed several changes including increased muscle fiber size, increased internal nuclei with abnormalities on Gomori trichrome and oxidative stains. Thus, their report had lead to the inclusion of Vici syndrome to a wider clinical picture and differential diagnoses such as congenital agenesis of corpus Collasum, as well as neurgenic atrophy and Centro nuclear myopathy (CNM) or a metabolic (Mitochondrial) cytopathy [6].

Although our patients' clinical pictures are somewhat characteristic of Vici syndrome, they still need to be followed up and with more investigations to further confirm this diagnosis.

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