Nostril Bleeding with Severe Thrombocytopenia in A 6-Year-Old Female with Congenital Amegakaryocytic Thrombocytopenia: A Case Report

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Abstract — Congenital Amegakaryocytic Thrombocytopenia (CAMT) is a rare bone marrow failure disorder that offers with separated thrombocytopenia within the first year of life. Although CAMT is an unusual root cause of thrombocytopenia in the newborn, it needs to be taken into consideration for youngsters with a lingering, unexplained thrombocytopenia without anomalies particular for various other bone marrow failure disorders. In our case report we intended to show the platelets evaluation test before and after the treatment, however with the consideration that the CAMT is a rare disease, our patient came with platelet level of 0/μL which was very rare case with not physical abnormalities.

Index Terms— Thrombocytopenia, Amegakaryocyte, Congenital Disease

1 INTRODUCTION

Congenital amegakaryocytic thrombocytopenia (CAMT) is an unusual inherited autosomal recessive bone marrow failure disorder identified by a separated and severe reduction in the number of platelets and

also megakaryocytes which could develop right into aplastic anemia and leukemia throughout the initial years of life or develops into bone marrow failing with pancytopenia later in youth [1], [2]. CAMT is normally identified by lack of physical malformations. Recently, small malformations in the heart, eyes, and mind with or without physical abnormalities have been identified [3]. The normal platelet counts among healthy neonates are always in at the range close to 150×10^9/L. In neonatal patients with thrombocytopenia, newborns will show a pattern of low/low-normal platelet (PLT) counts ($100–200 \times 10^9$/L), with raging levels of falling to $50–100 \times 10^9$/L at the first week then returns too normal at 8^{th} to 10^{th} days. Late-onset neonatal thrombocytopenia shows typically after the initial 3 days of life in which platelet counts usually falls dramatically to ranks of 50×10^9/L [4].

Bone marrow analysis in infants with CAMT normally shows normal general cellularity with a separated decrease or absence of megakaryocytes, although in many cases of bone marrows studied early during the condition could have misleadingly serial marrows and also very little searching for could be needed to confirm the diagnosis [5], [6] The only curative treatment is bone marrow transplantation. It commonly results in fatality [the disease is not treated after development of pancytopenia [5], [6]

2 CASE REPORT:

We present a case of a 6-year-old Saudi female who presented with Right nostril bleeding for 3 hrs before coming arriving to the hospital. Bleeding stopped spontaneously. Medical history showed no history of trauma, no history of bleeding from other orifices. She is known case of congenital thrombocytopenia which was diagnosed at birth, delivery was spontaneous vaginal delivery (SVD) without any complications. She was admitted two times before, first one before 9 months, she presented with otitis media and fever, PLT was 2000/μL and after management, PLT increased to 41,000/μL, and discharged after one day. Second admission before 5 months with nostril bleeding, PLT was 4000/μL and after management, it was increased to 21000/μL, discharged after 3 days and referred to specialist hospital when is under follow up and platelet transfusion during emergency. In this admission, physical examination showed that the patient showed mild ecchomotic patches over both lower limb, other than that she was well during inspection and she was active, no neck swelling was found, normal heart sound, no murmur, no gallop, vesicular breathing, equal bilateral air entry, abdomen was lax, no distention, no hepatomegaly neither splenomegaly, tone and reflex was normal, on x-ray, radius was normal. PLT was 0/μL (**Fig. 1**) and she was managed by 4 units of platelet concentrate then 2 units of platelet

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concentrate. Then it was increased to 38.000/μL. vaccination is incomplete, Normal development, consanguineous marriage, her older brother 10 years with same condition and in good health, there's other family member from parental side with same condition. patient have stayed until her platelet count was elevated normal (**Fig. 2**), then she was discharged after 3 days of admission.

3 Discussion

We can now to differentiate severe CAMT includes thrombocytopenia from absent radii (TAR) and Wiskott-Aldrich syndrome (WAS). Furthermore, antiplatelet antibodies may be moved from the mother to the fetus in-uterus or at the time of delivery, triggering extreme thrombocytopenia. women who do not have common platelet antigens might produce antibodies against paternal antigens that are revealed in the establishing fetus [3], [4]. In our study the patients which was presented with CAMT, had platelet count level of 0/μL, which was very interesting because the patients did not have any physical symptoms as hepatomegaly or continues bleeding. in one case report study [7], similarly, to ours present three CAMT cases that presented with different clinical diagnoses, with various physical anomalies in two of those cases, showed no specific somatic abnormalities that accompany this deadly disease. In our patient treatment was initiated at the first day of admission throughout the 3 days long of patient staying at the hospital, with platelet transfusions, at the time of discharge, she was fully engrafted, independent of platelet transfusions. However, there has been only some rare cases reported a malignant transformation in a patient with CAMT [8]. Unlike our patients which was still stable with no sign of malignant transformation.

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4 Conclusion

CAMT is a genetically heterogeneous illness with the capacity for other genes being associated with the pathogenesis. The somatic abnormalities could be subordinate or they could be related to genetics that has actually not yet been determined. It ought to be born in mind that patients with CAMT may present outside the neonatal period. Although CAMT is an unusual root cause of thrombocytopenia in the newborn, it needs to be taken into consideration for youngsters with a lingering, unexplained thrombocytopenia without anomalies particular for various other bone marrow failure disorders. In our case report we

intended to show the platelets evaluation test before and after the treatment, however with the consideration that the CAMT is a rare disease, our patient came with platelet level of 0/μL which was very rare case with not physical abnormalities.

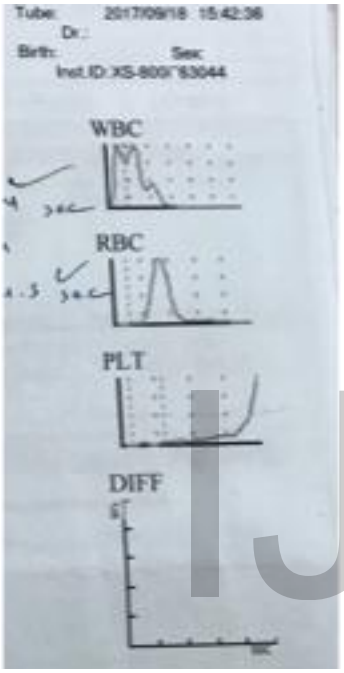

Fig. 1: Lab result at time of admission

Fig.2: Lab result after correction.

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