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CASE STUDY

# Congenital Dyserythropoeitic Anaemia: A Case Report from Tanzania

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## ABSTRACT

**Background:** Congenital Dyserythropoietic Anaemia (CDA) is an inherited anaemia characterised by ineffective red cell production and erythroblast structural abnormalities.

**Case Report:** We present a case of a 9 month old boy with leg pain and microcytic hypochromic anaemia of 5.8g/dL with reticulocytopenia, which was preceded by an episode of acute gastrointestinal infection. Upon investigation of the bone marrow aspirate findings were internuclear bridges between incompletely separated polychromatic erythroblasts highly suggestive of CDA type I within proper clinical settings.

**Conclusion:** Careful analysis of bone marrow aspirate through light microscopy should be done in any child presented with microcytic hypochromic anaemia with reticulocytopenia.

## BACKGROUND

Congenital Dyserythropoietic Anaemia (CDA) is a group of hereditary anaemia distinguished by poor erythropoiesis, morphologic abnormalities of erythroblasts and hemolysis.[1]

Because of its rarity, it is difficult to determine the true incidence and prevalence of CDA. Frequency in Europe ranges from 0.08 cases/ million to 2.6 cases/ million for all types of CDA.[2]

Prevalence in Africa remains unknown, although several case reports have been published from North Africa. To our knowledge, no published case reports from Sub-Saharan Africa were identified in PubMed-indexed journals at the time of writing.[1]

There are 3 main types of CDA (I, II and III). Type I and II are autosomal recessive caused by bi-allelic mutations. Type I occurs as result of mutation in *CDAN1* and *C15 or f41* genes which have critical roles in Deoxyribonucleic acid (DNA) repair and/or chromatin reassembly following DNA replication. Type II is the commonest and results from mutation in *SEC23B* gene that codes for proteins involved in intracellular vesicle transport. Type III is autosomal dominant and rarest of the three, is as a result of mutation in *KIF23* gene which is responsible for coding mitotic kinesin-like protein 1, a key component of the central mitotic spindle, that results in separation of the cells during late mitosis.[3]

Clinical presentation of patients with CDA is variable not only among unrelated patients with different mutations, but also between siblings who share the same mutations. It ranges from asymptomatic individuals to those having transfusion dependent anaemia. Symptoms may be worse at the time of infection or after trauma. Some present only with prolonged neonatal jaundice.[1] Limb anomalies such as dysostosis and syndactyly occurs in 4-14% of affected population.[4]

We present a case of congenital dyserythropoietic anaemia from Sub Saharan Africa.

## CASE PRESENTATION

A 9-month-old African boy, appropriately immunised for age presented with 2-week history of persistent refusal to bear weight on the left leg noted by mother while bathing him, associated with crying while being touched on the left knee joint, and low-grade fever, not associated with swelling of the feet, knee or hands, rash, cough, irritability, or focal neurological deficit. No history of trauma was reported. In the preceding 3 weeks he was admitted twice for acute viral gastroenteritis. The refusal to bear weight and associated symptoms began during the second admission for acute viral gastroenteritis and persisted following discharge, prompting further evaluation.

His past medical history was unremarkable with no previous admissions, or surgical procedures and he had normal growth and development. He was delivered at term following an uncomplicated pregnancy to a non-consanguineous couple and had an

uneventful postnatal period. He is the second of the two siblings; the elder brother has cerebral palsy. No known family history of chronic anaemia, transfusion dependence, jaundice, or inherited haematological disorders.

On examination he was noted to be afebrile, moderately pale, not jaundiced, with no palpable lymph nodes. Vital signs assessment revealed normal blood pressure, absence of tachypnea, normal heart rate and oxygen saturation of >95% on room air. His left leg was in a normal posture, no swelling, bruising, crepitus or change in skin colour was noted; was not warm, it was equal in length and circumference to the right leg, however tenderness was noted upon touching the knee joint. The rest of the examination was normal.

Diagnosis of probable septic arthritis with anaemia without heart failure was made with differential diagnosis of osteomyelitis, sickle cell anaemia, acute lymphoblastic leukaemia and juvenile idiopathic arthritis.

A complete blood count (CBC) showed anaemia with haemoglobin of 5.8g/dl (range: 10.5-14.5g/dl) with microcytic hypochromic red cell indices. White blood cell count was normal at 9.88 x$10^9$/l (range: 4.5-13.0 x$10^9$/l), absolute neutrophils count 3.57x$10^9$/l (range: 1.78-5.38 x$10^9$/l) and lymphocytes – 4.9x$10^9$/l (range: 0.4 - 4.0x$10^9$/l) slight elevated likely related to the recent viral gastroenteritis. C reactive protein (CRP) was elevated at 113 mg/l (normal: 0.5 – 5.0mg/l) and Erythrocyte sedimentation rate (ESR) was elevated at 138.02 mm/hr (normal: 0-15). His hemoglobin electrophoresis was HbAA and had relative reticulocytopenia of 0.057% (normal: 0.3-3%), Absolute reticulocyte count was low at 1,898 cells/microlitre, with normal ferritin level of 56.75µg/l and peripheral blood smear showed microcytic hypochromic anaemia. X-ray of the leg which was done on the previous admission was normal.

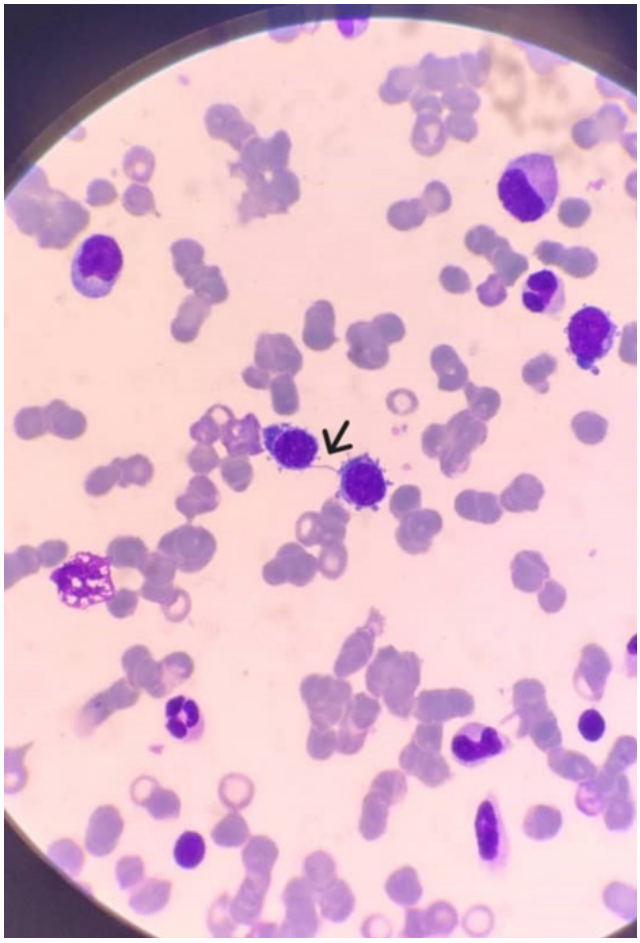
He was started on intravenous ceftriaxone at 75mg/kg, paracetamol at 15mg/kg, ibuprofen at 10mg/kg and ketoprofen gel, and was transfused with 78 ml of packed red blood cell (10ml/kg). Orthopaedic consult was sought, and Magnetic Resonance Imaging (MRI) of the leg was done to rule out infective process which was reported normal.

Other work up done included Antistreptolysin O Titer (ASOT) was normal at 3.41 IU/mL (normal: 0-150, Rheumatoid factor (RA Factor) was done to rule out autoimmune disorders like Rheumatoid Arthritis or juvenile idiopathic arthritis and was normal at 10.02 IU/mL (normal: 0-14.0). Flow cytometry ruled out acute lymphoblastic leukaemia.

On 3rd day post admission, a repeat CRP dropped to 58 mg/l and haemoglobin improved to 6.2g/dl.

Due to relative reticulocytopenia a bone marrow aspiration and biopsy was done on 4th day post admission to rule out congenital causes of anaemia, and reported: increased erythropoiesis, predominantly left-shifted erythroblasts occupying 39% of the marrow cells, with dysmorphic features- internuclear bridges, and incompletely separated polychromatic erythroblasts; features highly suggestive of Congenital Dyserythropoietic Anaemia (CDA), as seen in Figure 1 Genetic analysis to determine the type of CDA was not done due to financial constraints.

**FIGURE 1: Bone Marrow Aspirate Slide**



Bone Marrow aspirate slide, Hematoxylin and Eosin stain; showing internuclear bridges, and incompletely separated polychromatic erythroblasts (black arrow)

He was discharged on 5th day post admission and readmitted 11days after discharge for blood transfusion due persistent symptomatic anaemia with haemoglobin of 6.1g/dl.

He has since faired well and his repeat haemoglobin at 1 month after 2nd blood transfusion was stable at 9.6g/dl.

### Ethics and Consent

Written informed consent for publication of the patient's clinical details and images was obtained from the patient's parent/legal guardian.

## DISCUSSION

CDA is among the rare causes of anaemia, hence diagnosis is often made on high index of suspicion or coincidentally during anaemia work up.[1] This child presented with microcytic hypochromic anaemia of

unknown cause affecting only red cell lineage associated with relative reticulocytopenia. There are several causes of anemia associated with reticulocytopenia that specifically affect the red cell line, including Fanconi anemia, Diamond-Blackfan anemia, and CDA.[1] These conditions can be differentiated by bone marrow aspirate, with CDA displaying pathognomonic features of internuclear chromatin bridges between polychromatic and orthochromatic erythroblasts on light microscopy, along with marked erythroid hyperplasia.[5] This finding was noted in our patient's bone marrow aspirate, leading to the diagnosis.

Unlike most individuals with CDA who present with mild macrocytosis with anispoikilocytosis, and 30% who will have normal mean cell volume[1], our patient had microcytic hypochromic anaemia with normal serum ferritin levels of 56.75µg/l.

Affected individuals present with intense symptoms at the time of acute infection or trauma, like our patient, whose symptoms of leg pain and anaemia began after acute gastroenteritis. This could be as a result of exacerbation of underlying anaemia by an inflammatory state. Inflammation causes anaemia through increasing red cell clearance by increasing their phagocytosis, apoptosis and adherence to vascular endothelium. inflammatory cytokines produced during infective process also suppress erythropoiesis directly.[6]

While genetic analysis is important to determine the genetic mutations responsible for his

condition, type of disease, guide the genetic counselling and aid anticipatory guidance provided to the family; this has not been performed due to financial constraints and unavailability of tests locally. Diagnosis was based on characteristic bone marrow morphology seen under light microscopy, including erythroid hyperplasia and internuclear bridges, which are highly suggestive of CDA type I. Additional iron studies including serum iron, total iron-binding capacity, and transferrin saturation were also not available at the time of evaluation adding to this paper's limititation.

The expected prognosis and complications of congenital dyserythropoietic anaemia (CDA) varies widely by type and severity, ranging from mild to life-threatening and often requires long standing managements in cases like chronic anaemia, splenomegaly, gallstones, and progressive iron loads

## CONCLUSION

CDA should be considered in children with unexplained anaemia associated with reticulocytopenia and normal nutritional studies. High index of suspicion and careful analysis of bone marrow aspirate through light microscopy can lead to diagnosis of CDA.

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