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*Case Report*

# Congenital afibrinogenemia presenting with cellulitis in the lower back: A case report

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## ABSTRACT

Afibrinogenemia is a rare inherited bleeding disorder characterized by the complete absence of fibrinogen, a clotting protein essential for normal blood coagulation. We present a case of a 13-year-old girl diagnosed with afibrinogenemia at the age of 1 month, now admitted due to cellulitis in the lower back. Here, we have examined how to diagnose a case of these uncommon conditions and emphasized the management and treatment strategy for such a case.

**Keywords:** Afibrinogenemia, Cellulitis, Treatment, Counseling

## INTRODUCTION

Congenital afibrinogenemia is a very rare genetic illness with a deficiency or complete absence of fibrinogen or factor 1. It is an autosomal recessive disease discovered in 1920.[1] Fibrinogen plays a crucial role in the final step of the coagulation pathway by facilitating the formation of insoluble fibrin clots. It serves as a bridge between thrombocytes during the clotting process. In addition to its role in hemostasis, fibrinogen contributes to angiogenesis, tissue repair, and immune response. Recent research emphasizes the intricate interplay between fibrinogen, the vascular system, the immune system, and inflammation. Its functions extend beyond coagulation, encompassing direct cell signaling and host defense, and reflecting its ancestral roles.[2,3] Long-lasting bleeding from the umbilical cord stump occurs often in newborns with this disease following delivery.[4] It is estimated to have an incidence of 1–2 in a million in the world.[2]

## CASE STUDY

A 13-year-old girl born to 3rd-degree consanguineous parents who was a known case of afibrinogenemia diagnosed at the age of 1 month. She was admitted to the pediatric department of SS Institute of Medical Science and Research Center, Davangere, with complaints of fever for 5 days and lower back pain for 5 days, it was sudden in onset and progressive more in the back region aggravated on walking, sleeping, and even on sitting.

### Past history

There was a bloody discharge from the umbilical stump at the age of 1 month for which fibrinogen levels were checked and diagnosed the same. Since then, if any bleeding presents that the child

was given with fresh frozen plasma (FFP) transfusion and was on iron and folic acid tablets, for the past 4 years, no transfusion was done.

Physical examinations revealed temperature: 101* F, respiratory rate: 20 cpm, and pulse rate: 107 bpm. She was conscious and active with her abdomen soft and bowel sounds were heard.

Local examinations showed swelling present in the lower back (lower spine) with the size of 10*4 cm, tenderness +ve, local rise of temperature +ve, and transillumination absent.

**Relevant investigations were done as stated below**

Hemoglobin 11.1 g/dL, total count: 14490 cells/mm$^3$, neutrophils: 77%, lymphocytes: 18%, hematocrit: 33.3%, mean corpuscular volume: 80 fl, mean corpuscular hemoglobin: 26.4 pg, mean corpuscular hemoglobin concentration: 33%, red cell distribution width: 15.2, platelet count: 2.77 lakhs/mm$^3$, prothrombin time: >120 s, and activated partial thromboplastin clotting time: 190 s.

Ultrasound lower back suggests significant subcutaneous and perifascial edema noted along the right lower back over the swelling – features of inflammation.

The child received treatment with injection of ceftriaxone 750 mg 1-0-1, Tab. amoxiclav 625 mg (Amoxicillin + Clavulanic Acid) 1-0-1, Tab. Imol TD/SOS (Ibuprofen), and Tab. Serratio 10 mg (Serratiopeptidase) 1-0-1. On discharge, Tab. Imol (Ibuprofen) 1-0-1 and Tab. Bactoclav (Amoxicillin + Clavulanic Acid) 1-0-1 were advised to continue.

Counseling to the parents was done about the disease, treatment, and protection of the child.

## DISCUSSION

Afibrinogenemia is a relatively rare hereditary bleeding illness that can affect people of all racial and ethnic backgrounds, including both sexes.[1] About 1 in 1 million people are thought to have afibrinogenemia.[5] Bleeding can begin during the newborn era, and 85% of cases involve the umbilical cord, as well as 5% of cases involving the skin, gastrointestinal tract, genitourinary tract, or central nervous system. In 54% of patients, musculoskeletal hemorrhage, including hemarthroses, occurs.[1] In this case, the patient had umbilical cord bleeding at the age of 1 month. Patients with afibrinogenemia can get replacement treatment using fibrinogen concentrates. Its main advantages over conventional replacement treatments are virus inactivation, small-volume infusions, and reduced risk of adverse response. Khayat *et al*.[6] conducted a study on the efficacy and safety of fibrinogen concentrate for on-demand treatment of bleeding and surgical prophylaxis in pediatric patients with congenital fibrinogen deficiency in the year 2020 to find a new horizon of treatment for this disease so that the patient need not always get admitted to the hospital for cryotherapy. Only in cases of extreme emergency and in the absence of fibrinogen concentrates, cryoprecipitate, and FFP can be infused.[2] This patient gets an FFP transfusion whenever there is bleeding.

## CONCLUSION

Afibrinogenemia is an uncommon and challenging bleeding disorder that necessitates a comprehensive diagnostic approach and tailored management strategy. This case study of a 13-year-old girl with cellulitis in the lower back highlights the importance of early diagnosis and a multidisciplinary treatment approach involving replacement therapy and infection control measures. Advances in medical research and awareness will continue to improve the prognosis and quality of life for patients living with afibrinogenemia.

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**Authors' contributions**

LGS conceptualized the study, SM took the correspondence of the study, did the follow-up of the case, did a literature review, and wrote the original draft along with SG and SM.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent.

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**Conflicts of interest**

There are no conflicts of interest.

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