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abstract

A Novel Protein C Mutation Presenting As Severe Purpura Fulminans: A Case Report

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abstract

A Novel Protein C Mutation Presenting As Severe Purpura Fulminans: A Case Report

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Introduction: Congenital Protein C deficiency is a rare cause of purpura fulminans which presents as rapidly progressive thrombotic disorder in neonatal period. Here in, we describe a case of severe congenital Protein C deficiency who presented with severe phenotype in early neonatal period and succumbed to illness due multiorgan thrombosis. A novel mutation was detected in protein C gene with an autosomal recessive inheritance.

Case Presentation: A newborn male child, born of non-consanguineous marriage, presented to Emergency department at 30 hours of life with multiple necrotic lesions on Buttocks and lower limbs. On examination, vitals were stable. The lesions progressed rapidly in irregular central haemorrhagic necrosis involving right foot. Baseline investigations revealed mild thrombocytopenia (Platelet count -130/microlitre), mildly increased PT and APTT with high D-Dimer suggestive of DIC. Further investigations revealed Protein C activity below <1%, while Protein S and ATIII were normal. Congenital Protein C diagnosis was made and child was started on FFP transfusions and anticoagulation. The causative molecular defect was homozygous novel mutation in PROC gene p.Gln91ter (C>T) transversion at codon 271 leading to stop codon and premature truncation. In view of unavailability of recombinant Protein C, child was continued on FFP transfusion and anticoagulation. However, despite best supportive care, child succumbed to multiorgan thrombosis.

Conclusion: Herein, we report a novel mutation in PROC gene presenting as severe phenotype in early neonatal period. Molecular diagnosis along prenatal counselling is vital in purpura fulminans considering the dismal outcome.

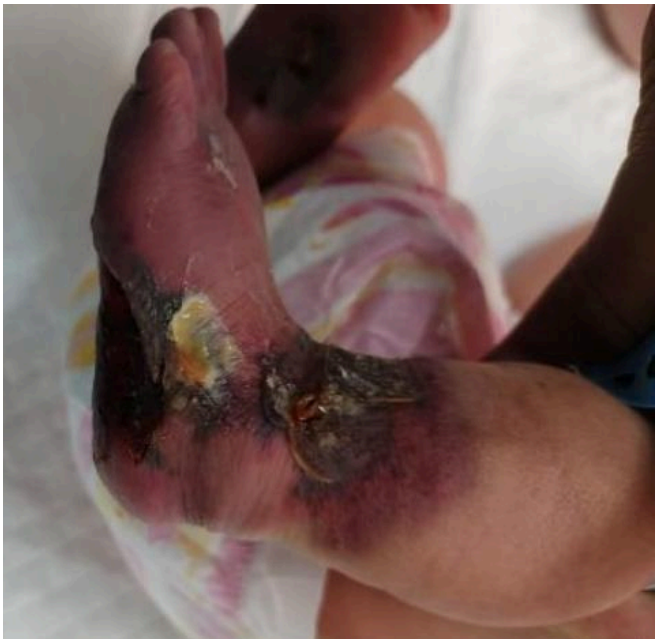

Figure 1