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Case Study
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A Case of Prolidase Deficiency Presenting as Upper Gastrointestinal Bleed

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Introduction

Prolidase deficiency is one the rare diseases known to the field of medicine. It is a rare autosomal recessive, inborn error of collagen metabolism manifesting as skin changes, recurrent infections, unusual facial features, organomegaly (hepatosplenomegaly) with elevated liver enzymes and varied intellectual dysfunction. There have been around 70 cases reported worldwide but our case is worth reporting as the patient has all features with portal hypertension and presented as massive upper gut bleed (variceal bleed). We present a 22 year old unmarried male with no underlying comorbidity. The patient was immediately stabilized by securing airway, breathing and circulation and given two units packed cell transfusion with hemodynamic monitoring and intravenous fluids. Simultaneously all base line investigations including were sent. Seeing the nature of bleed, Gastroenterologist was called and an urgent Upper GI endoscopy was done which revealed multiple varices (3 grade 3 and 2 grade 2) for which banding was done and patient starts on injection terlipressin 2 mg 8 hourly and pantoprazole infusion at a rate of 8 mg/h for 72 h (Figure 1). While the patient was being managed and after stabilization a complete general physical examination with relevant systemic examination revealed pallor, depressed nasal bridge, hepatosplenomegaly and multiple ulcers on bilateral lower limbs which according to the patient were chronic and present for more than 3 years. These ulcers were typically non-healing and the patient had never sought medical attention for them but was given some local treatment by a quack. Further he gave history that one of his siblings had a similar history of developing multiple ulcers on limbs which heal by atropic scarring. Keeping all these points in mind the possibility of prolidase deficiency was thought of. Thin layer chromatography of his overnight urine revealed marked increase in iminopeptide after gelatin loading (prolidase deficiency).

Discussion

Prolidase deficiency is a rare genetic disease with autosomal recessive inheritance first described by Goodman in 1968 in a male patient with intellectual disability and characteristic ulcers on legs [1]. Prolidase is an important enzyme involved in recycling of proline and hydroxyproline that is approximately one fourth of collagen [2]. The deficiency of this enzyme results in urinary loss of proline which is in the range of 3 gm per day [3]. These patients are intellectually subnormal, have short stature, saddle nose, hypertelorism, narrowed eyes and hypoplasia of jaw. The most characteristic feature remains skin fragility with leg ulceration and pitted scarring [4]. Other cutaneous

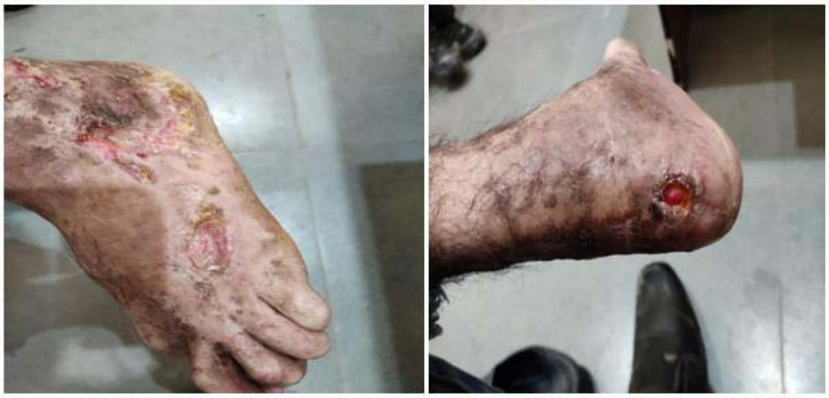

Figure 1: Upper gastrointestinal bleed.

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manifestations are photosensitivity, purpura, telangiectasias, and dry crusted lesions on face and fissured erythematous palms and soles [5]. They also present with recurrent infections such as otitis media, respiratory tract infections and sinusitis. In addition other features like simian crease, wasting of small muscles of hand, talipes equines, osteoporosis, hyperextensible joints, deafness, corneal opacities, amblyopia, optic atrophy, splenomegaly and protuberant abdomen [5]. The diagnosis is made by determining iminopeptiduria greater than 5 mmol/ 24 h with predominant peptide glycoproline [6]. A characteristic feature is resistance to all treatment including skin graft rejection. Treatment modalities of help are dapsone, diphenylhydantoin, ascorbic acid and manganese [6]. Topical preparation of 5 percent glycine and proline is effective [7]. Pulse corticosteroids show good response with inhibition of iminodipeptide primed neutrophil superoxide generation [8]. Plasma apheresis was also tried and successful in around 2 patients [9].

Conclusion

No specific treatment can be recommended; rather greater awareness and improved understanding of pathophysiology will improve recognition and treatment of this multisystem disease.

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