Introduction

Prolidase, the enzyme known to cleave the bond between proline and the other amino acids.

Prolidase deficiency is multisystem disease associated with massive imidodipeptiduria and lack of or reduced prolidase activity in erythrocytes, leukocytes, or cultured fibroblasts.

It is characterized by dysmorphic facial features, variable intellectual disability, chronic painful skin ulcers of the lower extremities and telangiectasia of the face and hands, recurrent skin and respiratory tract infections and hepatosplenomegaly with elevated liver enzymes.

Other common features include anemia, thrombocytopenia, hypergammaglobulinemia, and hypocomplementemia are common. Dermatologic features are the hallmark of the disease[1].
Case Report

We report here a 35-year-old lady with intellectual disability, dysmorphic features: depressed nasal bridge and high arch palate, recurrent bacterial infections. She had recurrent itchy lichenified patches and plaques over her extremities.

During her follow-up, she started to develop recurrent painful ulcerations over her legs with periods of complete healing. She underwent multiple skin grafts for non-healing ulcers with good result.

Parents are consanguineous and she had a sister, 16 years of age with recurrent bacterial infections and hearing disability post bacterial meningitis but no other manifestations.

The systemic examination revealed no abnormal findings except for a mild splenomegaly. Her ophthalmological examination was normal.

Skin biopsies were taken to rule out pyoderma gangrenosum, vasculitis and atypical mycobacterium and it showed only vasculopathy and her blood tests for autoimmune diseases, vasculitis and other were normal.

Whole exome sequencing revealed the known likely pathogenic variant in the PEPD gene c.202-2A>G in homozygous state. The result obtained is consistent with the genetic diagnosis of the autosomal recessive prolidase deficiency.

Her sister was tested for the same mutation and found positive.
Deep ulcer and atrophic scars on lower leg before and after treatment.
Discussion

Goodman in 1968 was the first to describe this condition in a male patient who had mental subnormality and characteristic recalcitrant ulcers on the lower legs.

The enzyme prolidase is widely distributed throughout the body and is important in recycling of proline and hydroxyproline which constitute about one quarter of the collagen[2,3].

The deficiency of this enzyme is responsible for massive loss of proline in the urine which is estimated to be as high as 3 gm per day.

The prolidase enzyme can be assayed in the erythrocytes, leucocytes and the fibroblasts and have been found to be undetectable in the patients with prolidase enzyme deficiency[1].

These patients are mentally subnormal and are of short stature. They have peculiar facies such as saddle nose, hypertelorism, narrowed eyes and hypoplasia of the jaws[4].

Among the clinical presentation, the most striking manifestation is the skin fragility with leg ulceration and characteristic pitted scarring. The other cutaneous changes seen include photosensitivity, purpura, telangiectasia, dry crusted lesions on the face and buttocks, dry fissured erythematous palms and soles[5].
Conclusion

Prolidase deficiency is multisystem disease with wide variable manifestations, skin legions characterize the disease but can be absent in the same affected family members

References: