Prolidase deficiency

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Creation Date: August 2001
Update: January 2004

Scientific Editor: Professor Jean-Marie Saudubray

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Abstract
Prolidase deficiency is a very rare autosomal recessive disease. Prevalence remains unknown, some 40 cases have been reported. It is characterized by mild to severe skin lesions particularly on the face, palms, lower legs and soles, besides variable other features. Patients excrete massive amounts of iminodipeptides in urine due to a deficiency of the exopeptidase prolidase. The activity of this enzyme can be measured in hemolysates, leukocytes or fibroblasts. Mutations in the PEPD gene on chromosome 19p13.2 have been observed. The skin ulcers respond partially to local treatment.

Keywords
prolidase, iminodipeptiduria

Disease name
Prolidase deficiency

Excluded diseases
Lathyrism

Diagnostic criterium
Prolidase deficiency

Differential diagnosis
Systemic lupus erythematosus

Prevalence
Unknown; some 40 individuals have been reported.

Clinical description
First symptoms start between birth and young adult age. Most characteristic are the skin lesions ranging from mild to severe, including recalcitrant ulcerations. They are mostly located on the face, the palms, the lower legs and the soles. Other features are facial dysmorphy, psychomotor retardation and recurrent infections.

Management
This is limited to treatment of the skin ulcers with oral ascorbate, manganese (cofactor of prolidase), inhibitors of collagenase, and local

applications of L-proline-and glycine-containing ointments. Response to treatment is variable.

Etiology
Mutations in the *PEPD* gene on chromosome 19p13.2.

Diagnostic methods
- Partition and elution chromatography,
- direct chemical ionisation mass spectrometry,
- enzymatic test.

Genetic counseling
Autosomal recessive inheritance.

Antenatal diagnosis
It is possible on amniocytes and chorionic villi.

Unresolved questions
An efficient treatment is not available.

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