

Abstract

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Inherited human collagen lysyl hydroxylase deficiency: ascorbic acid response.

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BACKGROUND: A patient is described with congenital hypotonia, lax joints, friable skin, hemorrhagic scars, high-arched palate, and borderline microcornea.

DISCUSSION OF CASE: Acid hydrolyzed whole skin collagen had a reduced hydroxylysine content of 0.5 residues per 1,000 as compared to 5.1 \pm 0.7 in control skin. Collagen lysyl hydroxylase in dialyzed subcellular fractions of cultured skin fibroblasts required L-ascorbate as a principal cofactor. Activity of this enzyme in cultured skin fibroblasts derived from this patient, his father, and mother were 17%, 66%, and 39% of control values, respectively. Collagen prolyl hydroxylase activity was normal.

RESULTS: Pharmacologic amounts of oral vitamin C (4 gm/day) produced an increase and withdrawal resulted in abrupt diminution of urinary excretion of hydroxylysine. Over a two-year period the patient's wound healing and muscle strength improved and corneal diameter increased. Hydroxylysine content of the skin did not increase.

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