LETTERS TO THE EDITOR

HYALINOSIS CUTIS ET MUCOSAE WITH RARE OCULAR INVOLVEMENT

To the Editor,

Hyalinosis cutis et mucosae (HCM) is a rare, generalised, recessively inherited disorder involving multiple organs besides skin, oral cavity and larynx.[1] Crops of papules, bullae, pustules and haemorrhagic crusts appear in first stage and scars of face, limbs with plaques/nodules over face, axillae, scrotum, extensors of elbows and knees are seen in second stage and both stages can overalp.[2] Usually hoarseness is present from infancy or early childhood and skin lesions can precede it, appear simultaneously or shortly after it.[2] The teeth tend to be lost early and pathognomonic intracranial calcifications were seen in 51/100 cases.[2] Hyaline deposits in the retina between membrane and pigment epithelium have been observed.[3] Presence of lipid differentiates HCM from other conditions associated with hyalinosis of subepidermal or submucosal connective tissue.[3] Probably hyaline deposits in HCM consist of glycoproteins associated with free or loosely bound lipid.

A 30-year-old unmarried Kashmiri Muslim male had deformity of nose due to extensive and progressive scarring following appearance of discrete erythematous, papules, vesicles, pustules, occasional haemorrhagic crusts, and similar though less severe lesions on the cheeks, face, ears, neck, limbs, buccal mucosa and palate for 20 years. 2.5-3 cm yellow, soft, plaques were seen on the extensors of elbows. A big pale yellow scar was seen on the left upper arm at the donor graft site. He had progressive hoarseness of voice for 1 year. He also had progressive diminution of vision for 2 years. Both corneas were hazy due to hyaline deposits and left eye also had hyaline deposits in retina. He had got keratoplasty done on the right eye. There was positive history of consanguinity in parents. X-ray skull revealed characteristic bean shaped calcified areas along sella turcica and calcified spots in anterior cranial fossa. Strongly PAS positive fibres with fibroblast nuclei running perpendicular to skin surface were seen in upper dermis along with prominent basement membrane zone and focal deposits in deep dermis. Similar fibers were seen around blood vessels and sweat glands also. Histopathology of epiglottis was similar.

Three major alterations observed in HCM are (i) massive deposition of amorphous material between collagen fibres and around blood vessels, (ii) marked reduction in number and size of collagen fibers, and (iii) considerable thickening of basal lamina due to more of collagen type IV and fibronectin. The fibroblasts secrete less of collagen type I and III and endothelial cells produced more of collagen type IV and V in HCM. Recent studies have established that HCM represents defect in collagen synthesis.

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References

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