DYSKERATOSIS CONGENITA WITH ADENOCARCINOMA OF STOMACH

R P Singh, V D Tiwari, A K Misuriya and S P Chattopadhyay

A 32-year-old male had dyskeratosis congenita with adenocarcinoma of stomach along with multiple cases in the family, rare mode of inheritance and occurrence of carcinoma of stomach in four affected family members. The patient presented with progressive reticulate hyperpigmentation of the face, neck, upper trunk and arms, hypopigmented macules, lacrimation and epigastric pain.

Key words : Dyskeratosis congenita, Carcinoma stomach.

Most cases of dyskeratosis congenita have an X-linked recessive inheritance, though a few cases with autosomal dominant inheritance have also been reported.[1,2] It is characterised by skin pigmentation, nail dystrophy and leukoplakia,[2-4] the serious features being bone marrow hypofunction and increased risk of malignant diseases. Burton and Rook[5] had noted that the onset is usually between 5 and 13 years of age with skin or nail changes as the first manifestation. Our patient had an unusual mode of inheritance (autosomal dominant) and carcinoma of stomach in four affected family members.

Case Report

A 32-year-old male, noticed dark discoloration of the skin of his neck and chest at the age of 10 years which progressively spread until 3 years back. Thereafter, the progression of pigmentation of face, arms and back was rapid. Dystrophy of all nails, photophobia and photosensitivity appeared during the past 3 years. Two years back, the patient developed pain in the epigastrium and excessive lacrimation. He also had an episode of malena 1½ years back. The family tree of the patient showed that 6 members of his family including the mother and sibs had similar skin changes. Four of them died due to carcinoma stomach.

From the Departments of Dermatology and Pathology, Command Hospital (SC) Pune and Armed Forces Medical College, Pune-411 040, India.

Address correspondence to : Major R P Singh, Military Hospital, Jabalpur-482 001 (MP).

Examination revealed reticulate hyperpigmentation of the face, neck, upper trunk and arms with hypopigmented macules at places. All the nails were dystrophic. Mucous membranes and palms and soles were normal. Epigastric tenderness was present. No abdominal lump was palpable. Routine hematological and biochemical investigations were normal. Barium meal study and endoscopy suggested a large malignant gastric ulcer which on operative biopsy proved to be a well differentiated adenocarcinoma.

Skin biopsy showed hyperkeratosis, basal cell degeneration at places, flattened rete ridges, absence of epidermal appendages and occasional melanophores in the upper dermis.

Comments

The family tree of the patient showed that the mother and five other members of his family were affected suggesting an autosomal dominant inheritance.[1] Complete syndrome occurring in the females has also been described earlier.[2] The usual mode of inheritance is X-linked recessive affecting only the males. Females can only be affected if they are homozygote. So if the mother of the proband was having homozygous genes for dyskeratosis congenita then the inheritance in this case can easily be explained on the basis of X-linked recessive inheritance.

Nearly 16% of the patients develop one or more neoplasms by the time they report.[6] Our

patient also had adenocarcinoma of the stomach when he was first examined. But occurrence of gastric carcinoma in 4 of his family members is unusual and not reported earlier.

References

1. Scoggins RB, Prescott KJ, Asher GH et al : Dyskeratosis congenita with Fanconi type anaemia : Investigations of immunologic and other defects, Clin Res, 1971; 19 : 409-410.
2. Bryan HG and Nixon RK : Dyskeratosis congenita and familial pancytopenia, JAMA, 1965; 192 : 203-208.
3. Connor JM and Teague RH : Dyskeratosis congenita : report of a large kindred, Brit J Dermatol, 1981; 105 : 321-325.
4. Milgrom H. Stoll HL and Crissey TJ : Dyskeratosis congenita, Arch Dermatol, 1964; 89 : 345-349.
5. Burton JL and Rook A : Genetics in Dermatology, Fourth ed, Editors : Rook A, Wilkinson DS and Ebling FJG et al : Blackwell Scientific Publications, Oxford, 1987; p 149.
6. Sirinavin C and Trowbridge AA : Dyskeratosis congenita : clinical features and genetic aspects : Report of a family and review of the literature, J Med Genet, 1975; 12 : 339-342.