

Cancer Prone Disease Section

Mini Review

Multiple self-healing squamous epithelioma

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Identity

Alias

Ferguson-Smith disease

Note

Multiple self-healing squamous epithelioma is characterized by the development of multiple invasive skin tumours with the histological appearances of well differentiated squamous cell carcinomas that resolve spontaneously if left untreated leaving deep pitted scars.

Inheritance

Autosomal dominant. Prevalence is unknown but over 100 cases have been reported worldwide with the majority of reported cases originating in Scotland.

Clinics

Phenotype and clinics

The age of onset of a first tumour is variable ranging from 8 to 62 years. Further tumours appear episodically throughout life with over a 100 tumours occurring in some more severely affected individuals. Tumours most commonly occur on areas of skin exposed to sunlight (the face, ears and limbs).

Lesions are typically painless. They first appear as red papules and progress to nodules, often with a central keratin plug.

Over time the lesions may ulcerate; when ulceration occurs, the edges are typically rolled and undermined.



Fig 1. Multiple lesions develop on sun-exposed areas of the body of patients, and all show spontaneous regression.

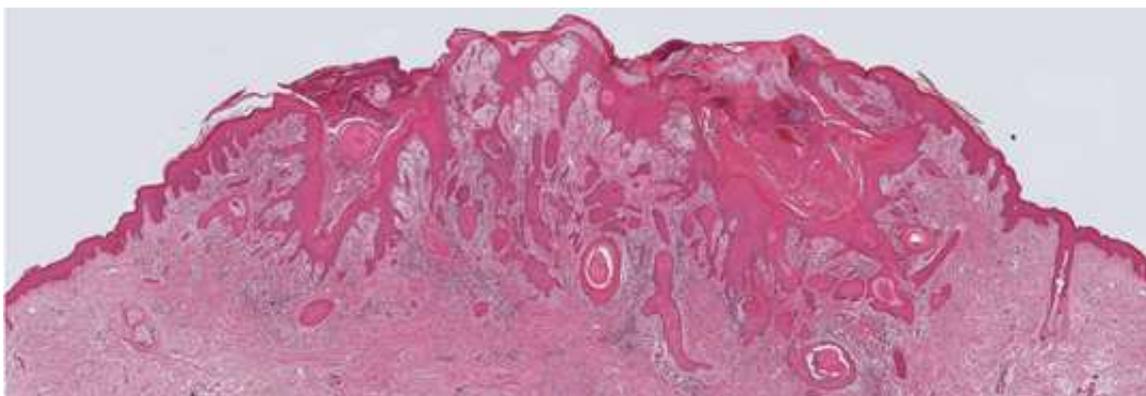


Fig 2. Histological section from a typical lesion. In this lesion, the collarette of epidermis on either edge and the eosinophilic cytoplasm, typical of a keratoacanthoma, are absent. (H&E; Scale bar: 100µm).

Characteristically these tumours undergo spontaneous regression within 4-6 months, resulting in scars that are typically deep and pitted on the face, scalp and ears. Scars occurring on the limbs tends to be smoother and shallower.

The tumours typically lack the collarette of epidermis on either edge and distinctive eosinophilic cytoplasm commonly observed in keratoacanthomas.

Neoplastic risk

An increased risk of neoplasia at other sites has not been reported. The skin tumours invade locally and may be destructive but do not appear to recur following excision. Aggressive local invasion after radiotherapy has been described.

Treatment

Surgical excision of tumours and cryotherapy of early lesions have been the most widely employed treatments. Etretinate was found to reduce the number of new lesions occurring in some patients.

Prognosis

Scarring following tumour resolution can be disfiguring. Although tumours can be locally invasive most resolve spontaneously and metastases are very rare.

Genes involved and proteins

Note

The locus has been mapped to a 4Mb region of chromosome 9q22.3 in studies of affected families but the genetic defect causing the condition has not been identified. Loss of heterozygosity for 9q22.3 markers in self-healing epitheliomas is consistent with a tumour suppressor function for the MSSE locus.

Location

9q22.3

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