A familial syndromic association between cutaneous malignant melanoma and neural system tumours: reply from authors


Published in: British Journal of Dermatology

DOI: 10.1111/j.1365-2133.2004.06291.x

2004

Link to publication

Citation for published version (APA):

Total number of authors: 9

General rights
Unless other specific re-use rights are stated the following general rights apply:
Copyright and moral rights for the publications made accessible in the public portal are retained by the authors and/or other copyright owners and it is a condition of accessing publications that users recognise and abide by the legal requirements associated with these rights.
• Users may download and print one copy of any publication from the public portal for the purpose of private study or research.
• You may not further distribute the material or use it for any profit-making activity or commercial gain
• You may freely distribute the URL identifying the publication in the public portal

Read more about Creative commons licenses: https://creativecommons.org/licenses/

Take down policy
If you believe that this document breaches copyright please contact us providing details, and we will remove access to the work immediately and investigate your claim.
mechanism of immune-privilege collapse to that in alopecia areata may play some role in our case, and cause subsequent lymphocytic infiltration into the follicular epithelium, causing the follicular mucinosis. Various adverse effects of imatinib have been reported in the literature, but to our knowledge follicular mucinosis has never been reported previously.

Department of Dermatology, Hokkaido University Graduate School of Medicine, N15W7, Kita-ku, Sapporo 060-8638, Japan
Correspondence: Daisuke Sawamura.
E-mail: smartdai@med.hokudai.ac.jp

References

A familial syndromic association between cutaneous malignant melanoma and neural system tumours

DOI: 10.1111/j.1365-2133.2004.06292.x

Sirs, Nielsen et al. recently published the results of an analysis of Swedish patients with four primary tumours, at least one of which was a cutaneous malignant melanoma (MM). In this group there was a significantly higher than expected incidence of neural system tumours (NST) including meningioma, nonacoustic neurinoma and astrocytoma. The authors concluded that the association between multiple MM and NST might form a previously undescribed new syndrome.

In fact, a syndromic association between MM and NST has been described previously. In 1993, Kaufman et al. described a three-generation family where MM, cerebral astrocytoma or both developed in eight family members. Subsequently, our group surveyed 904 consecutive MM patients for the occurrence of NST within their first- and second-degree relatives. In that study, 15 Jewish families consisting of MM patients and at least one NST-affected relative were identified. The NST consisted of astrocytoma, medulloblastoma, glioblastoma multiforme, ependymoma, glioma, meningioma and acoustic neurinoma. Additionally, 10 patients with two primary tumours, MM and NST, meningioma in nine and acoustic neurinoma in one, were described. Further analyses from France and Finland confirmed the notion of familial cosegregation of MM–NST. Large-scale epidemiological studies in Scandinavia have confirmed an increased risk of NST in relatives of MM patients and an increased risk of MM as a second primary tumour among NST patients. Thus, familial–syndromic association of MM and NST has been recognized as a rare autosomal dominant familial cancer syndrome since the early 1990s and was designated as the Melanoma and Neural System Tumour syndrome (MM–NST, OMIM 155755).

The underlying genetic defect was sought only in a handful of MM–NST families. Analysis of two families with the MM–NST syndrome showed hemizygous germline deletion at the 9p21 region that ablated both CDKN2A (p16) and p14ARF. Analysis of 11 families with two or more cases of glioma revealed a hemizygous germline deletion in CDKN2A in one family with both glioma and melanoma. In another family with MM and NST (mainly astrocytoma), deletion was found in the p14ARF specific exon 1B. The deletion, leading to loss of p14ARF function, did not affect p16.

However, in other MM–NST families analysed, including the pedigree described by Nielsen et al., no germline mutations were identified in CDKN2A and other known familial MM candidate genes. The inherited predisposition to the MM–NST syndrome in these families probably lies at another, yet unknown genes.

In conclusion, the association of MM and NST is a well-established rare autosomal dominant trait whose underlying genetic defect is yet unknown in the majority of cases.

Department of Dermatology and Oncogenetics
Unit, Sheba Medical Centre, Tel-Hashomer, and the Sackler School of Medicine, Tel-Aviv University, Israel
E-mail: a_scope@netvision.net.il; astrauma@hotmail.com

References

© 2004 British Association of Dermatologists, British Journal of Dermatology, 151, 1272–1288
A familial syndromic association between cutaneous malignant melanoma and neural system tumours: reply from authors

DOI: 10.1111/j.1365-2133.2004.06291.x

Sir, We are grateful for the comments made by Scope et al. describing the possibility of an association between neural system tumours and melanoma. The Swedish data by Hemminki et al. were not published when we submitted our manuscript and we therefore had no chance of referring to their findings.

Our study did not address heredity as such, only the presence of associated tumours among individuals with four or more tumours of which at least one was a melanoma. Pedigree data are under evaluation and it is therefore premature to conclude that the association described by us follows a dominant inheritance. A recessive mechanism may be as pertinent in this extremely predisposed group of patients. Further pedigree analysis will help to solve this matter.

References


Espinosi cellulitis associated with molluscum contagiosum

DOI: 10.1111/j.1365-2133.2004.06285.x

Sir, Eosinophilic cellulitis is a rare dermatosis that has a clinical picture resembling acute cellulitis and a characteristic histopathology with dermal oedema and dense eosinophilic infiltration, which is called the flame figure. Although the aetiology and pathogenesis of this condition are still unknown, various associated disorders, such as several viral infectious diseases, have been documented. Recently, it has been reported that cryosurgery for molluscum contagiosum (MC) may be one of the triggering factors of eosinophilic cellulitis. Furthermore, a case of hypereosinophilic syndrome (HES), in which the initial diagnosis of eosinophilic cellulitis was made, has been reported to be concomitant with MC. Interestingly, some have reported overlapping clinical and histopathological findings in eosinophilic cellulitis and HES, which are also characterized by peripheral eosinophilia and eosinophilic infiltration of tissues. In this communication, we present an additional rare case of eosinophilic cellulitis closely associated with untreated MC.

A 10-year-old Japanese boy was referred to us for further investigations of a suspected acute cellulitis with a 7-day history of an asymptomatic erythematous rash that had developed over the left lower abdomen and rapidly enlarged. He had experienced a similar episode 7 months previously on almost the same region, but the condition had resolved spontaneously. His birth history was unremarkable. There was no history of a scratch or bite by either animals or arthropods. He had no history of atopic diseases and had not taken any drugs.

On physical examination, a rosy, well-defined, oedematous, erythematous plaque with a diameter of approximately 20 cm on the left lower abdomen extending to the left inguinal region was observed. The plaque was not painful, but there was a burning sensation. A superficial inguinal lymph node was palpable, but the patient was afebrile. A whitish papule covered with crust was visible near the centre of the affected skin.

Histology of a biopsy taken from this papular lesion revealed typical features of MC in the epidermis (Fig. 1A). The dermis was patchily infiltrated mainly by histiocytes and lymphocytes and a dense perivascular and diffuse infiltrate of