Osler-Weber-Rendu syndrome associated with vitiligo

Sir,

A 60-year-old male patient presented to our outpatient department with a complaint of asymptomatic, depigmented lesions over his hands, feet, and lips of more than 10 years’ duration. The patient was a known diabetic on insulin. He had been evaluated and treated for the past 10 years for recurrent epistaxis. No apparent cause was found for the epistaxis. The patient also had a persistently low hemoglobin level, for which he had been on vitamin and iron supplements. Findings from all relevant investigations, including bone marrow biopsy, were normal. He gave no history of other bleeding tendencies. There was no history of hematemesis, hemoptysis, bleeding from the gums, or melena.

On examination he was found to have acral vitiligo. Interestingly we found multiple telangiectasias over the hands, lips, tongue, and ear lobes [Figures 1-3]. The patient had noticed these red lesions increasing in number over the last few years but had attached no significance to the same. Apparently all his brothers too had similar lesions over the extremities, but none had a history of epistaxis or other significant bleeding tendency. A skin biopsy confirmed the lesions to be telangiectasia. Based on the history and clinical findings, the possibility of Osler-Weber-Rendu syndrome was considered. The patient was extensively investigated including abdominal, chest, and neurological imaging, to rule out other systemic involvement. All findings were within normal limits.

Osler-Weber-Rendu syndrome, or hereditary hemorrhagic telangiectasia (HHT), is easily recognized in individuals displaying the classical triad of epistaxis, telangiectasia, and a suitable family history, but the disease is more difficult to diagnose in many patients. In our case, we assume that the diagnosis was missed earlier probably because the lesions were few and not very evident initially. Serious consequences may have to be faced if visceral arteriovenous malformations (AVM), particularly in the pulmonary circulation, are unrecognized and left untreated. The diagnosis of Osler-Weber-Rendu syndrome is essentially clinical. The 4 criteria (epistaxis, telangiectasia, visceral lesions, and an appropriate family history) are carefully delineated. The HHT diagnosis is definite if 3 criteria are present (which was so in our case). A diagnosis of HHT cannot be established in patients with only 2 criteria, but should be recorded as ‘possible’ or
suspected’ to maintain a high index of clinical suspicion. If fewer than 2 criteria are present, HHT is unlikely, although children of affected individuals should be considered at risk in view of age-related penetration in this disorder.[1] Mutations in 2 genes, endoglin (ENG) and activin receptor-like kinase 1 (ACVRL1 or ALK1), have been associated with HHT.[2] HHT1 is caused by mutations in endoglin gene mapping on chromosome 9q, and HHT2 is caused by mutations in ALK1 located on chromosome 12q.[3] In view of the high prevalence of pulmonary and cerebral AVMs, all patients with HHT should be screened for their presence, and relatives of patients with HHT should be investigated for presence of the disease.[4] This case is interesting because of the association between Osler-Weber-Rendu syndrome and vitiligo. To the best of our knowledge, there has been only one previous report of a similar association.[5] We assume this to be a coincidental association as there seems to be no other definite basis to associate HHT with vitiligo, though recently independent studies have suggested a common chromosomal link (chromosome 7) for both vitiligo and HHT.[6,7] This case also highlights the importance of thinking of, and looking for, the possibility of Osler-Weber-Rendu syndrome in patients presenting with recurrent epistaxis without any other apparent cause.

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Sir,

Localized depigmented patches in a dermatomal distribution that do not cross the midline are called segmental vitiligo. The course of the segmental type tends to be earlier in onset and more stable than generalized vitiligo and is not familial.[1]

Twenty-nail dystrophy (TND) presents as rough surfaced nail plates and involving up to 20 nails. Two types of nail changes have been described. In the first type, the entire nail appears to have been sandpapered (sandpaper nails) in a longitudinal direction and shows excessive ridging and roughness. In the second type, the nail plate is shiny (shiny nails).[2] Here we report a case of an unusual association of segmental vitiligo and twenty-nail dystrophy.

A male patient aged 20 years presented with depigmented skin lesions since 5 years and also with nail changes of 2 years' duration.

On cutaneous examination, localized, multiple, round-to-oval–shaped, achromic macules were seen in a segmental distribution over the abdomen (corresponding to T9,10 dermatome). Examination of the nails revealed longitudinal ridging and roughness over the nail plates (sandpaper nails) in all the nails. Histopathology of an achromic macule showed marked absence of melanin granules, and Wood's lamp examination revealed amelanotic macules. Nail biopsy showed spongiosis in the nail matrix with mononuclear inflammatory infiltrate.

Association of cutaneous and systemic autoimmune diseases with vitiligo occurs more significantly in the non-segmental type than in the segmental type.[3] Though rarely, segmental vitiligo has been reported in association with a few skin disorders like poliosis,[1] halo nevus, [1,3] nevoid basal cell carcinoma syndrome,[4] Parry-Romberg syndrome[5] and linear scleroderma.[6]

The association of vitiligo and TND is very rare and has been sparsely reported,[7-10] and vitiligo in most of such associations was of non-segmental type, like generalized vitiligo,[7] scalp vitiligo (localized),[8] and acrofacial vitiligo.[9] However, association of segmental vitiligo with TND has been described in 2 patients.[10] The association of segmental vitiligo with TND in the present case can be explained by the autoimmune origin of these disorders. Although the etiology of segmental vitiligo is based primarily on the neurogenic theory of melanocyte destruction, an immune mechanism cannot be completely ruled out, because autoimmune disorders have been described in approximately 3.4% to 9.5% of cases of segmental vitiligo.[1,11] Further, systemic and topical steroids and psoralen with ultraviolet A (PUVA) therapy have shown encouraging responses in early lesions.[11] The strong association of TND with dermatoses which have autoimmune etiopathogenesis has led some to speculate that the nail changes are primarily due to an autoimmune process.[12,13] Thus our case emphasizes a common autoimmune insult to the melanocytes and nail matrix as the logical explanation for this rare association.

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