Facial Angiofibroma as an Initial Manifestation in Multiple Endocrine Neoplasia Type 1

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A 38-year-old woman presented to Shinshu University Hospital because her mother had recently been diagnosed as having MEN1. Although no abnormal findings were noted by biochemical screening and imaging studies, physical examination revealed a small angiofibroma on her face (Fig. 1A). Subsequent genetic testing revealed that she carries a heterozygous in-frame deletion (c.511_519del) of the MEN1 gene, which had been previously identified in her mother.

Despite its high frequency (1, 2), facial angiofibroma in MEN1 is not fully recognized. There might be ethnic differences in its frequency and clinical features. The majority of Caucasian patients have more than three facial angiofibromas, while most Japanese patients with angiofibroma have a single lesion or two lesions (Fig. 1B-D). The frequency of angiofibroma among Japanese patients is also lower than that of Caucasian patients (1-3). For all patients who developed MEN1-related tumors, careful examination for facial angiofibromas can be a clue to an early diagnosis of MEN1.

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