CASE 8

Patient: A 9-year-old Thai girl from Chiang Rai

Chief Complaint: A solitary progressive erythematous rash on the left cheek since birth

Present Illness: The patient first presented with asymptomatic erythematous papules on her left cheek since she was born. Later, those papules gradually increased in size and number with age. Finally, all papules congregated into a solitary erythematous plaque. The lesion no longer changed in size when the girl was the age of 6. Apart from the cosmetic concern, the lesion is asymptomatic.

Past History: Healthy, normal development and intelligent child

Family History: No one in her family has the same lesion.

Dermatological Examination (Figure 8.1): A solitary, well-defined, erythematous, rough-surfaced bleachable, not tender plaque with irregular border, size 2x2.5 cm. No other skin manifestations.

Figure 8.1

Histopathology (S10-06915) (Figure 8.2): Proliferation of dilated capillaries and scattered stellate fibroblasts with coarse collagen bundles arranged haphazard array

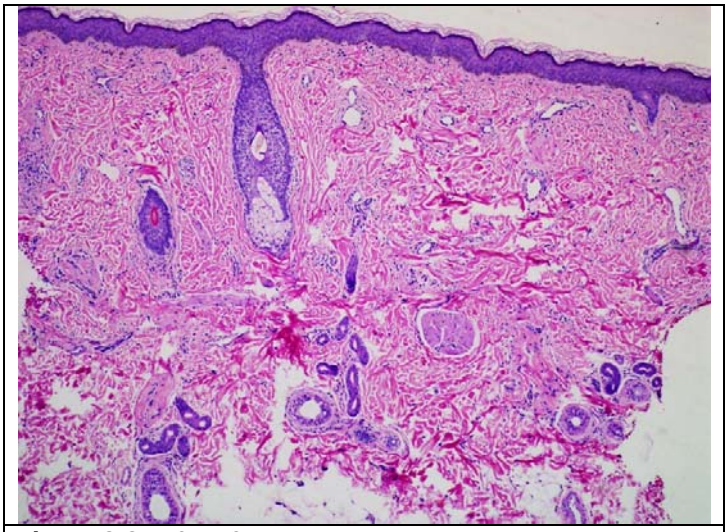

Figure 8.2, H&E 40x

Diagnosis: Solitary angiofibroma

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Discussion:

Angiofibromas are benign cutaneous tumor which can present sporadically or associate with inherited disease in multiple angiofibromas. Previously multiple facial angiofibromas were regarded as a pathognomonic sign for tuberous sclerosis; however, more than a decade ago researchers found that angiofibromas may also present in multiple endocrine hyperplasia type 1 (MEN1) .[1]

Histopathology demonstrates an increased of small blood vessels association with perivascular and periadnexal fibrosis arranged centrically. The fibrosis tissue usually contains stellate fibroblasts. Immunohistochemistry has shown that the large stellate fibroblastic cells express factor XIIIa but not S-100 protein. Factor XIIIa appears to be important in the promulgation of the fibroplasia. Ultrastructural studies have suggested that the stellate cells are fibroblastic or fibrohistiocytic.

A patient with multiple facial angiofibromas should be examined skin and other systemic manifestations carefully because the multiple lesions may be relevant to tuberous sclerosis or multiple endocrine hyperplasia type 1 (MEN1).

Tuberous sclerosis complex (TSC) is an autosomal dominant multisystem neuro-cutaneous syndrome characterized by the development of multiple harmatomas in various systems such as skin, brain, heart, kidney, liver and lungs.[2] Two-thirds of TSC patients represent sporadic mutations. Only 29% of patients have the full triad(seizure, mental retardation and cutaneous angiofibromas) and 6% of them lack all three of the triad.[2] Dermatologic manifestations of TSC are Ash leaf (polygonal hypomelanotic macules), Shagreen patch (a yellowish red or pink, flattened, slightly elevated surface with a rough texture), Angiofibromas (bilateral, symmetrical distribution of red to pink papules or nodules with a smooth, glistering surface with predilection for the butterfly area of the face and nasolabial grooves)[3], forehead fibrous plaques (yellowish-brown/skin-coloured plaques on scalp or forehead), ungual or periungual fibromas (skin-colored/reddish nodules usually arising from the nail bed of a finger or toe). Other manifestations are teeth enamel pitting, gingival fibromas, brain harmatomas (85% of patients have their first

epileptic episode in the first two years of life)[4], rhabdomyoma, renal, ocular and pulmonary involvement.

MEN1 is a dominantly inherited tumor syndrome consisting of tumor of parathyroid glands, entero-pancreatic endocrine tissue and posterior pituitary.[5] MEN1 patients may have a risk to develop cutaneous tumors such as multiple facial angiofibromas, collagenoma and lipoma.[1] In1998, Böni *et al* investigated the relationship of sporadic angiofibroma and MEN1 gene mutations by gene mapping. They suggested that alterations of MEN1 gene affect not only on MEN1 patients' clinical manifestations, but also on the initiation or/ and progression of a subset of sporadic angiofibromas.[6]

In this case, although her skin biopsy is angiofibroma, the girl has negative for family history and clinical findings for both TSC and MEN1. Therefore, the most likely diagnosis in this patient is sporadic solitary angiofibroma. Solitary giant facial angiofibroma can be seen in patients without any associated syndromes.[7] Cutaneous angiofibromas are benign lesions and can be removed by shave excision or dermabrasion.[8, 9] Excellent cosmetic outcomes have been revealed in facial angiofibromas of tuberous sclerosis treated with laser.[10-13]

Theoretically, the lesion with prominent vascular component like this case tends to respond well to vascular specific laser. Moreover, the erythematous plaque is on patient's face where is easily noticeable so the issue of cosmetic will be the major concern. By these two reasons, the treatment of choice in this case would be pulse dye laser. This girl has been treated with pulse dye laser (V-beam,Candela USA) by using parameters: spot size 7 mm., 6 msec and 8 J/cm^2 . She needs multiple treatment sessions which are 4-week apart. Her lesion responds fairly good after the first session.

References

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