CASE 4

A 36-year-old Thai woman from Bangkok

Chief complaint:

Multiple brownish asymptomatic macules and skin color papules on trunk since she was born.

Present illness:

She has multiple brownish macules arranged in group on her chin, neck and trunk. She also noticed about soft skin color papules on her trunk since she was young.

Past history:

Her general heath is good.

Family history:

Her grandfather, mother, sister and brother also have brownish macules on their body.

Physical examination:

Skin: Multiple skin-colored, soft, dome-shaped papules of various sizes (ranging from 3 to 7 mm in greatest diameter) scattered on her back and multiple brownish macules arrange in segment followed Blaschko's lines on her back and neck.

Slit lamp examination: no Lisch nodules

Other systems were entirely normal.

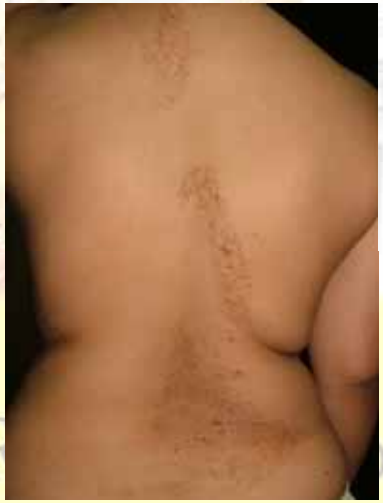

Fig. 4.1

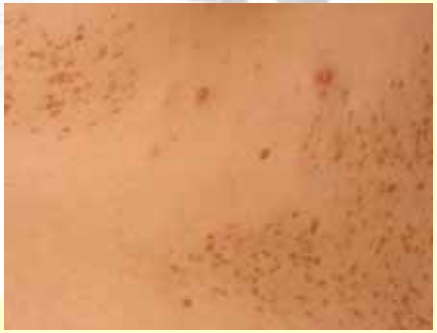

Fig. 4.2

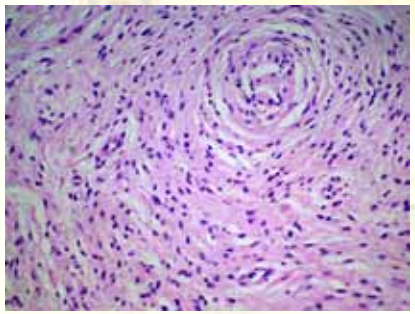

Fig. 4.3

Histopathology: (S05-05548)

Well-circumscribe, exophytic tumor composes of oval and spindle cell.

Some tumor cells show s or comma-shaped nuclei in the loose stroma with abundant mucin and thin wavy collagen strands.

Diagnosis: Segmental neurofibromatosis
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Consultant: Siripen Puavilai

Discussion:

Segmental neurofibromatosis (NF) is a rare disorder characterized by neurofibromas, with or without pigmentary changes that are localized most often to one region of the body. Its prevalence is estimated between 0.0014 and 0.002 percent. This form of NF was first described by Gammel in 1931. In 1956, Crowe *et al*. described additional patients with neurofibromas and *café-au-lait* macules in a dermatomal distribution and suggested the term sectorial NF. Two decades later, Miller and Sparkes proposed the term, segmental NF. In 1982, Riccardi classified NF into eight categories (Table 1). Type V correspond to segmental neurofibromatosis and was defined by the presence of neurofibromas and/or café-au-lait spots with segmental distribution and without systemic involvement or family history. Because of the heterogeneity of NF, in 1987, Roth *et al* proposed four possible subgroups of the segmental type, according to unilateral or bilateral disposition, family history and systemic involvement (Table 2). Our case can be classified as type III segmental neurofibrosis because

of the familial involvement. The proposed causative mechanism is a mosaicism,caused by a postzygotic somatic mutation in primitive neural crest cells, which affect the NF-1 gene.The more premature the mutations arise in the developing embryo, the more generalized would be the phenotype.The somatic mutation can explain the bilateral form, when coincidentally occurring in two contiguous regions of the body (NF type IV in Roth's classification). Transmission to the next generation is possible if a gonadal mosaicism coincides with a somatic mosaicism (NF type III in Roth's classification).

The management of segmental NF is fairly uncomplicated. Individual neurofibromas may be surgically removed if they cause the patient discomfort, compromise adjacent structures, cause cosmetic disfigurement, or show signs of malignant transformation. Segmental NF may rarely be associated with extracutaneous manifestations, all patients with segmental NF should undergo a thorough skin evaluation and ophthalmologic evaluation. Lisch nodules are usually not present during early childhood and ophthalmologic examination should be conducted after 6 years of age. With regard to genetic counseling, Sloan *et al*. recommend that patients without bilateral Lisch nodules should be informed that there is an extremely small risk of transmitting segmental or even generalized NF to their offspring, while patients with bilateral Lisch nodules should be informed that the chance of genetic transmission approaches that of type I NF.

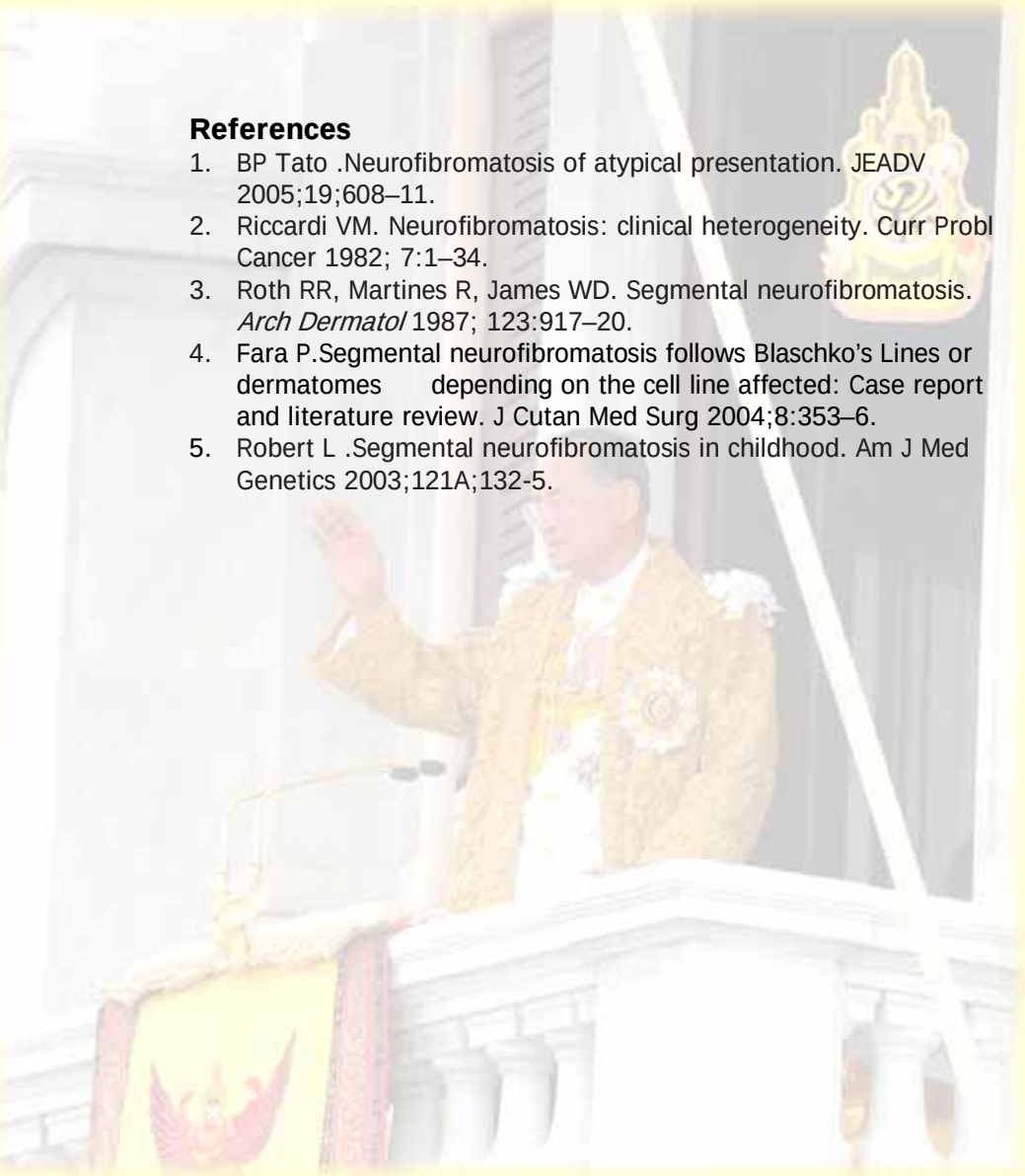
We report a case of segmental NF. Presentation of patients with cutaneous neurofibromas should prompt the physician to perform a thorough physical examination and to question the patient regarding family history. It is important for the physician to determine the type of NF the patient has so that, if indicated, an appropriate search for systemic disease and genetic counseling can be undertaken.

Table 1 Classification of neurofibromatosis (Riccardi, 1982)

Category	Description	Features
Type I	Von Recklinghausen's disease	Multiple café-au-lait spots; Lisch nodules; neurofibromas
Type II	Acoustic	Bilateral acoustic tumours; few café-au-lait spots and neurofibromas
Type III	Mixed	Intermediate between first two types
Type IV	Variant	Café-au-lait spots and neurofibromas; variable family history
Type V	Segmental	Café-au-lait spots or neurofibromas in one unilateral segment
Type VI	Café-au-lait spots	Café-au-lait spots only
Type VII	Late onset	Onset of disease after age 30
Type VIII	Not otherwise specified	Cases not characteristic of any other category

Table 2 Classification of segmental neurofibromatosis (Roth et al., 1987)

Category	Description	Features
Type I	True segmental	Segmental café-au-lait spots and /or neurofibromas, no systemic involvement, non-familial
Type II	Localized with deep involvement	Segmental with deep systemic involvement, non-familial
Type III	Hereditary segmental	Segmental, no deep involvement, familial
Type IV	Bilateral segmental	Bilateral segmental café-au-lait spots and /or neurofibromas, no deep involvement, non-familial



References

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3. Roth RR, Martines R, James WD. Segmental neurofibromatosis. *Arch Dermatol* 1987; 123:917–20.
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