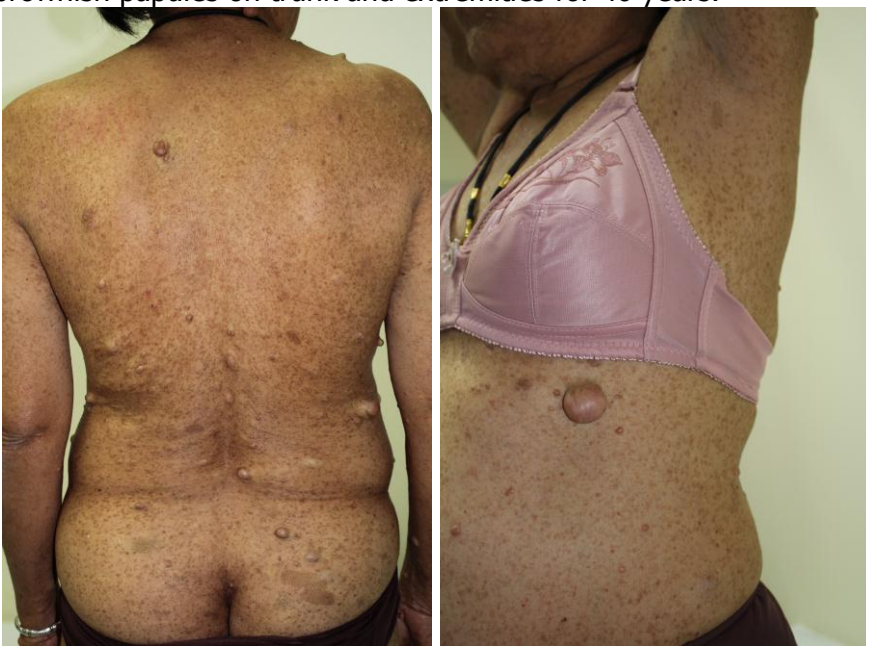
Case 10.1
A 64-year-old Thai woman from Saraburi.
Chief complaint: Multiple discrete asymptomatic skin colored to brownish papules on trunk and extremities for 40 years.



Present illness: The patient presented with 40 years history of multiple asymptomatic skin-colored to brownish papules on trunk and extremities. The lesions slowly increased in size and number.

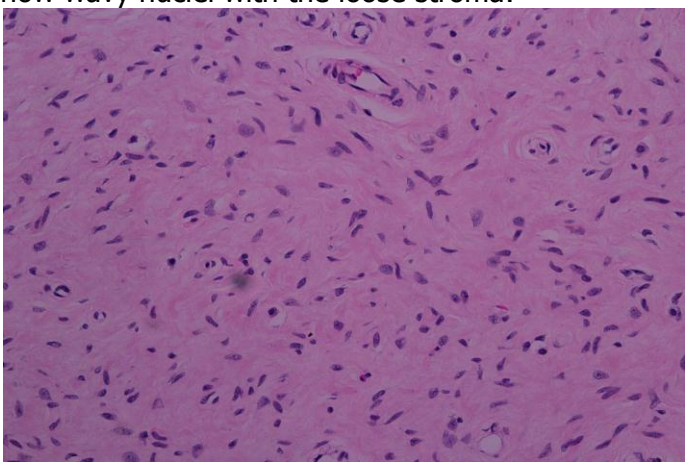
Past history
She has had brain tumor since 1978 and underwent surgery in Siriraj hospital. There was no evidence of recurrence. She also underwent TAH with BSO since 1987.

Family history: None of the patient's family has similar problems.

Physical examination:
Eye examination: Lisch nodules, both eyes.

Skin examination: Multiple discrete skin-colored to brownish papules(diameter 0.5-3 cm) on trunk and extremities. Multiple café-au-lait macules (diameter 1.5-5 cm) on trunk > 6 lesions, axillary freckling.

Histopathology
There is diffuse proliferation of oval and spindle cells, some of which show wavy nuclei with the loose stroma.



Investigation: NF 1 gene: pending

Diagnosis: Neurofibromatosis type 1

Treatment: Counseling and follow up for complication

Presenter: Siripan Lapsomboonsiri, M.D.

Consultant: Marinya Pongpudpunth, M.D.

Discussion

Neurofibromas are slowly growing,benign nerve sheath tumors that are composed of Schwann cells,perineural-like cells, and fibroblastic cells immersed in a collagenous or myxoid matrix. It is one of the most frequent tumours of neural origin and its presence is one of the clinical criteria for the diagnosis of type 1 neurofibromatosis (NF-1).[1]

Neurofibromatosis type 1 (NF1) is an autosomal dominant, multisystem disorder affecting approximately 1 in 3500 people.[2] NF1 causes by dysfunction of neurofibromin gene (NF1)which is located on chromosome 17q11.2 and encodes for neurofibromin,a tumor suppressor protein.[2] Characteristic clinical abnormalities for NF1 include pigmentation of the skin (café-au-lait spots), skinfold freckling, and cutaneous, sub-cutaneous or plexiform neurofibromas.[3] Other features such as macrocephaly,mental subnormalities with learning disorders, short stature, scoliosis, sphenoid bone dysplasia, congenital pseudarthrosis and cerebrovascular disorders. The clinical criteria for the diagnosis of NF1 are as follows (requiring ≥2 of the following features to be present): 1.Six or more café-au-lait spots: ≥5 mm in prepubertal individuals and ≥15 mm in postpubertal individuals, 2.≥2 neurofibromas of any type or 1 or more plexiform neurofibromas., 3.Freckling in the axilla or groin, 4.Optic glioma, 5.≥2 Lisch nodules (benign iris hamartomas), 6. A distinctive bony lesion (dysplasia of the sphenoid bone or dysplasia or thinning of the long bone cortex), 7.A first-degree relative with NF-1. Patients with NF1 also have an elevated risk for developing malignant peripheral nerve sheath tumor(MPNST), an aggressive cancer derived from cells associated with the nerve sheath mainly Schwann cells.[3]

NF1 is associated with vasculopathy, which is a significant but underrecognized complication that affects multiple sites, including thoracic, abdominal, renal, and intracranial vessels.[6] NF-1 vasculopathy can produce renal artery stenosis, coarctation of the

aorta, and other vascular lesions.[7] The incidence of hypertension in patients with NF1 is around 1% and is associated most commonly with renal artery stenosis in children and pheochromocytoma in adults.[8] Treatment of NF1: Neurofibromas are removed by surgery or CO_2 laser. Clinical trial with sirolimus, imatinib[9] is going for treatment plexiform neurofibroma.

References

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