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Case Report

Pacinian neurofibroma: A rare case report

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Abstract A 25-year-old, Javanese, Indonesian, female presented with large bumps on the buttocks, hip, and lower abdomen for last 15 years. Local examination revealed multiple, soft, sagging tumor masses that seemed hanging, fused, hyperpigmented, with varying sizes, from the smallest diameter about 1 cm to the largest of 40 cm x 30 cm on the right buttock, 35 cm x 30 cm on the left buttock where both spread up to the hips and lower abdomen, with sharp demarcation, wrinkling and loosening of the surface of the skin. Routine investigations were normal. Histopathology revealed proliferation of fibrous connective tissue accompanied with whorled structures *Pacinian bodies* that showed no sign of malignancy. Immunohistochemistry examination with antibodies against protein S-100 showed tumor cells with positive immunoreactivity. A diagnosis of Pacinian neurofibroma was made.

Key words
Pacinian neurofibroma, Pacinian bodies, S-100 antibodies.

Introduction

Pacinian neurofibroma (PN) is a very rare benign dermal tumor that is characterized by the presence of Pacinian corpuscles-like structures in the dermal myxoid stroma.[1] The term pacinian neurofibroma was initially proposed by Thoma in 1894, then by Prichard and Custer in 1952, also by Prose *et al*. in 1957.[2]

Various other terms also had been used for the dermal neoplasms, such as neurothekeoma, nerve sheath myxoma, benign myxoid tumor of nerve sheath, perineural myxoma, bizarre cutaneous neurofibroma.[1,3] However, some authors consider that the term pacinian perineural cell fibroma may describe it better.[2]

In PN, there is Pacinian corpuscle-like differentiation in the myxoid stroma. While the ordinary neurofibroma would suggest the presence of mature Pacinian corpuscle, but there is no typical characteristic of Pacinian neurofibroma.[2]

Until now there has been no reported incidence about this case. Only a few cases have been reported before that involved fingers and toes, and scalp.[1,3,4,7]

Histopathologic features of PN usually show typical round or ovoid corpuscle, with pale concentric pacinian corpuscle-like lamellar structure attached to myxoid matrix. There is no complete Vater-Pacini structure.[1]

Clinically, there are dome-shaped papules or nodules with a diameter less than 1 cm. We report a case of PN with very large sized lesions that occured on the buttocks.

Case Report

A 25-year-old, Javanese, Indonesian, female presented with large swellings on the buttocks, hip, and lower abdomen for last 15 years. The swellings were initially of the size of a thumb in the lower abdominal area, then slowly expanded

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to left and right side of hip and buttocks, where they looked like hanging on patient's body. The patient never experienced pain or itching, and the lesions never bled. Patient also had brownish spots on skin since birth. The patient went to several physicians and hospitals but did not receive any treatment.

There was no history of seizures, impaired vision, hearing loss, learning disability, impaired defecation, abdominal pain or nausea, high blood pressure, joint pain, or spontaneous bone fracture.

The patient was the elder of two siblings. Her parents were not cousins and had no history of similar disease. Patient's younger brother, a 20-year-old male, also had brownish spots on the skin since birth but never developed bumps.

On physical examination, consciousness was compos mentis, blood pressure 100/70 mmHg and other vital signs within normal range. Examination of the head and neck, ENT, oral cavity and teeth, hair, eyes, neck, chest, back, extremities found no abnormalities. There was no lymphadenopathy and kyphoscoliosis.

Dermatologic status revealed soft, multiple, sagging tumor mass, that seemed hanging, fused, hyperpigmented, with varying size, from the smallest diameter about 1 cm to the largest of 40 x 30 cm on the right buttock, 35 x 30 cm on the left buttock where both spread up to the hips and lower abdomen. It was sharply demarcated with wrinkled and loose skin surface (**Figure 1** and **2**). On palpation it was soft, nontender and no 'bag of worms' feeling. There were 2 large café-au-lait macules that fused from the back to the right lateral abdominal wall, also to the left medial chest and abdomen wall. The second café-au-lait macule surrounded the tumor mass in the buttocks and groin area.

Histopathologic examination showed proliferation of fibrous connective tissue accompanied with whorled structure Pacinian bodies that showed no sign of malignancy (**Figure 3** and **4**). Immunohistochemistry showed antibodies against protein S-100.

Consultation with ophthalmologist found no Lisch nodule (iris hamartoma). Evaluation by neurologist found no sign of focal neurological deficit. Assessment by ENT department was in normal range without any sign of acoustic neuroma. IQ test by psychologist was within normal range.

X-ray thorax, posteroanterior/lateral view, showed no scoliosis, cardiomegaly, or metastases in lungs and bones. X-ray pelvis and cruris sinistra also showed no metastases. Blood examination was within normal range.

According to anamnesis, clinical examination, and histopathologic examination, diagnosis of this patient was pacinian neurofibroma.

Management plan for the patient included serial excision surgery after obtaining consent from the patient's family.

Discussion

Pacinian neurofibroma (PN) is a very rare dermal tumor, which usually presents as dome-shaped papules or nodules.[1,2,3,4] In this patient, PN showed very different clinical findings, hitherto unreported. Usually PN occurs on hand and feet, but can be found on the buttocks or any other body part.

Histopathology plays a very critical role in confirming diagnosis, because clinical manifestations are less typical in most cases. In this patient, histopathologic findings of proliferation of fibrous connective tissue

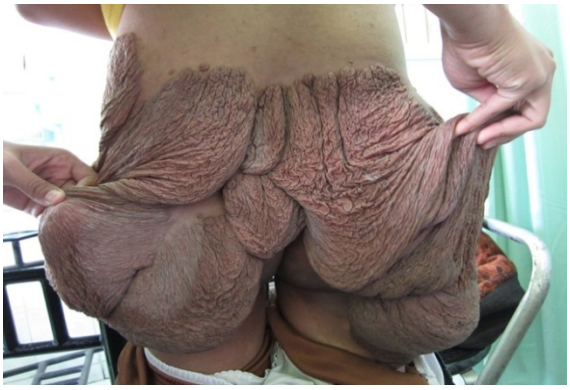

Figure 1 An extensive tumorous mass with wrinkled surface, thrown into folds and hanging involving both buttocks extending to upper thighs.

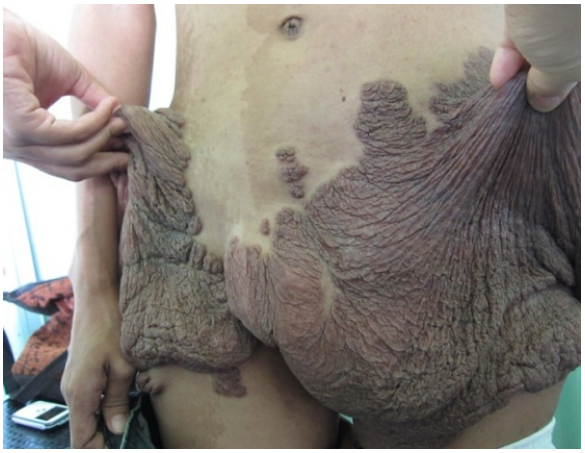

Figure 2 A massive pendulous mass, extending from lower abdomen to upper thigh

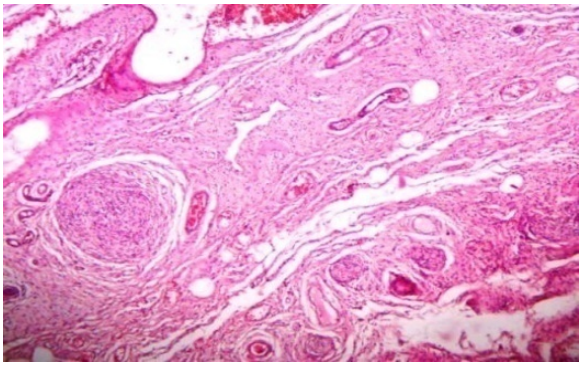

Figure 3 Pacinian corpuscle with different maturation stages.

accompanied with whorled structures (Pacinian bodies). Immunohistochemistry showed tumor cells with positive immunoreactivity against antibodies S-100.

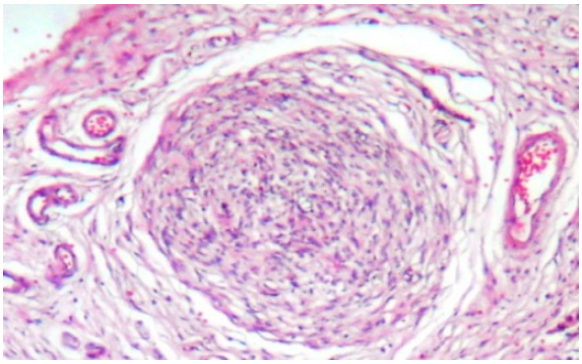

Figure 4 Pacinian corpuscle magnification 400x.

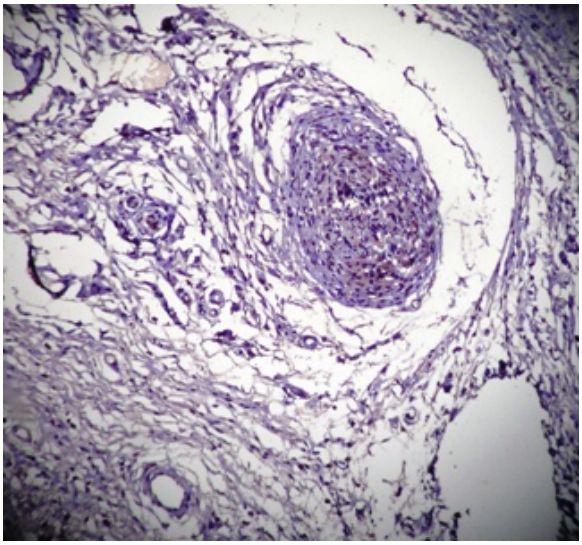

Figure 5 Pacinian corpuscle with nucleus and cytoplasm.

This is consistent with the literature where PN showed typical histopathologic image of round or ovoid lobules that contain Pacinian corpuscles embedded in myxoid stroma. In a well-differentiated Pacinian corpuscle, there is homogenous, acellular, eosinophilic central core with capillaries, while at a more immature stage it contains more cellular elements with spindle-shaped nuclei. The amount of variation in size and morphology of Pacinian corpuscle is caused by different maturation stages.[1]

The possible differential diagnosis in this case was plexiform neurofibroma. Plexiform neurofibroma is a variant of type 1 neurofibromatosis (Von Recklinghausen), where the diagnosis can be established when there are

at least 2 of 7 criteria of National Institutes of Health (NIH), namely.[3,8]

1. 6 or more macula café-au-lait with diameter > 5 mm in pre-puberty patients or > 15 mm on adult patients
2. 2 or more neurofibromas of any type or one plexiform neurofibroma
3. Freckles on axilla or inguinal folds (Crowe's sign)
4. Optic glioma
5. 2 or more Lisch nodules (iris hamartoma)
6. Distinct bone lesions such as sphenoid dysplasia or thinning of long bone cortex with or without pseudoarthrosis
7. History of first-degree family (parents, siblings, or offspring) who meet NF1 criteria.

This patient had only 1 of 7 criteria mentioned above. Thus the differential diagnosis of plexiform neurofibroma could be ruled out.

Until now the relationship between pacinian neurofibroma and neurofibromatosis has not been fully understood, and the literature did not show any malignant variant of this tumor.[1,2,9,10] The suggested treatment of choice is wide excision of tumor mass, after assessment of the factors e.g. size of tumor mass and the involvement of vascular and nerve structures.

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