

## Original Article

# Neurofibroma and lipoma in association with giant congenital melanocytic nevus coexisting in one nodule: a case report

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**Abstract:** Giant congenital melanocytic nevi (GCMN) are rare conditions that defined as melanocytic lesion recognized at birth, which will reach a diameter larger than 20 cm, and they occur in about 1 per 500,000 newborns. Despite its rarity, they may associate with severe abnormalities like spina bifida occulta, meningocele, club foot and hypertrophy or atrophy of deeper structures of a limb, Carney complex, premature aging syndromes, neurofibroma, vitiligo, lipoma and dysplasia of bilateral hip impact on the patient. In this case, we report a 3-years-old male child presenting a GCMN with large, blackish, and thick nevus covering over the entire neck, back, and lower to the waist level. We highlight the importance of proper histopathological examination of the biopsy taken from the single huge nodule which revealed features of both neurofibroma and lipoma coexisting. The objective of this paper is to report a rare case with the clinical and pathologic findings.

**Keywords:** Giant congenital melanocytic nevi, neurofibroma, lipoma

## Introduction

The congenital melanocytic nevi (CMN) are pigmented cutaneous lesions occurred normally within nests in the epidermis, dermis or in other tissue. According to their projected adult surface diameter, CMN can be categorized into three different types [1]. The diameter of lesions larger than 20 cm is considered giant in approximately 1:20,000 of the incidence. Giant congenital melanocytic nevus (GCMN) is at greatest risk for complications such as malignant melanoma, neurofibroma, lipoma and other central nervous system [2]. Despite its rarity, GCMN are important because of association with other abnormalities. In this study, we are reporting the clinic manifestation, imaging findings and histopathology features in one case of GCMN associated with neurofibroma and lipoma coexisting in a single nodule.

## Case report

A 3-years-old male child was referred to our department, resident of village of Luanchuan

district in Luoyang, China, for follow up of his large, blackish skin lesions covered with thick, black and long hairs over the entire neck, back and lower to the waist level that had been present since birth and extended from the neck to the lower back posteriorly. Initially, the lesion was flat and smooth. Over time it became thicker and thicker, developed in symptom with black and long hairs gradually. At the age of two years, a nodule was emerged in size of walnut in the lesion at the junction of the neck and back, which gradually increased to attain the present size. There was no other significant history. Nobody in his family had similar complaints. His general physical examination and systemic examination was within normal limits. The physical examination revealed lesions, which the maximum diameter were measured more than 40 cm, on his entire neck, back, and the waist covered 15% of the skin surface area with a large, blackish, and thick plaque. Several satellite lesions were also distributed over the arms, lower trunk, thighs and legs. At the junction of the neck and back, a nodule of size, 9 × 7.5 cm



**Figure 1.** Posterior view of lesion in patient: GCMN spread over neck and back, an tumor measuring about 9 × 7.5 cm in size beneath the GCMN.

was present in symptoms of no erosion, drainage and ulceration. The nodule, covering with thick and terminal hairs, was soft, mobile, slipping under the palpating fingers and non-tender with a smooth surface (**Figure 1**). No abnormalities were observed in hemogram, biochemical parameters, urine routine examination, X-ray of the vertebral spines, whereas a 8.5 × 5.5 cm lump with T1- and T2-hyperintensity lesion of clear boundary and homogeneous signal was visible size in Magnetic Resonance Image (MRI), though low and nonuniform signal on fat saturation sequence, (**Figure 2**). The nodule biopsy specimens were done at the epidermal-dermal junction, dermis, and upper dermis. The specimens were sent for histopathological examination and characterized by densely packed epithelioid nevus cell nests and wavy spindled cells, which arranged in fibroblast and collagen like matrix, exhibiting light staining,

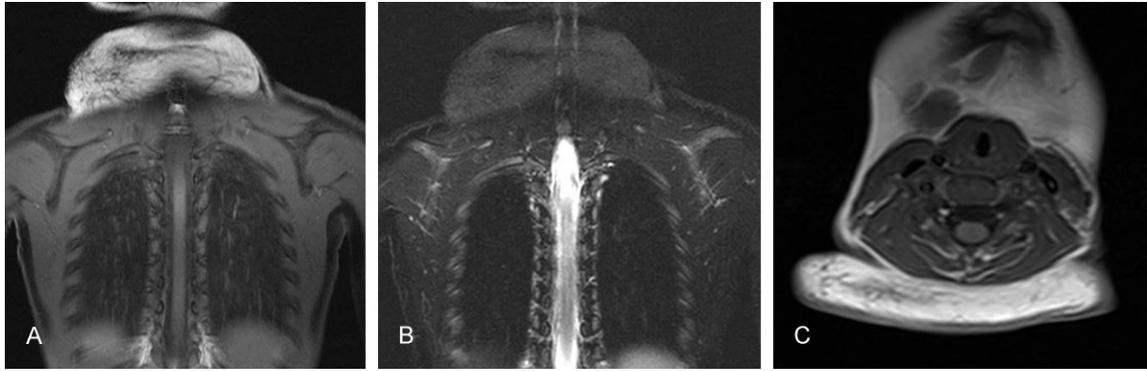
scant cytoplasm, slender nuclei. A large number of single vacuole mature fat cells were also visible in deep subcutaneous. Nuclei and cytoplasm for extruding mostly to one side and Slender fiber spacing separated the tumor into leaflike components with intervals in which there were thin-walled vessels, with no significant cell atypia and nuclear fission. All areas showed features of both neurofibroma and lipoma (**Figure 3**). The child received surgery for excision of the nodule and recovered well after operation. Two weeks after operation the child discharged from hospital.

### Discussion

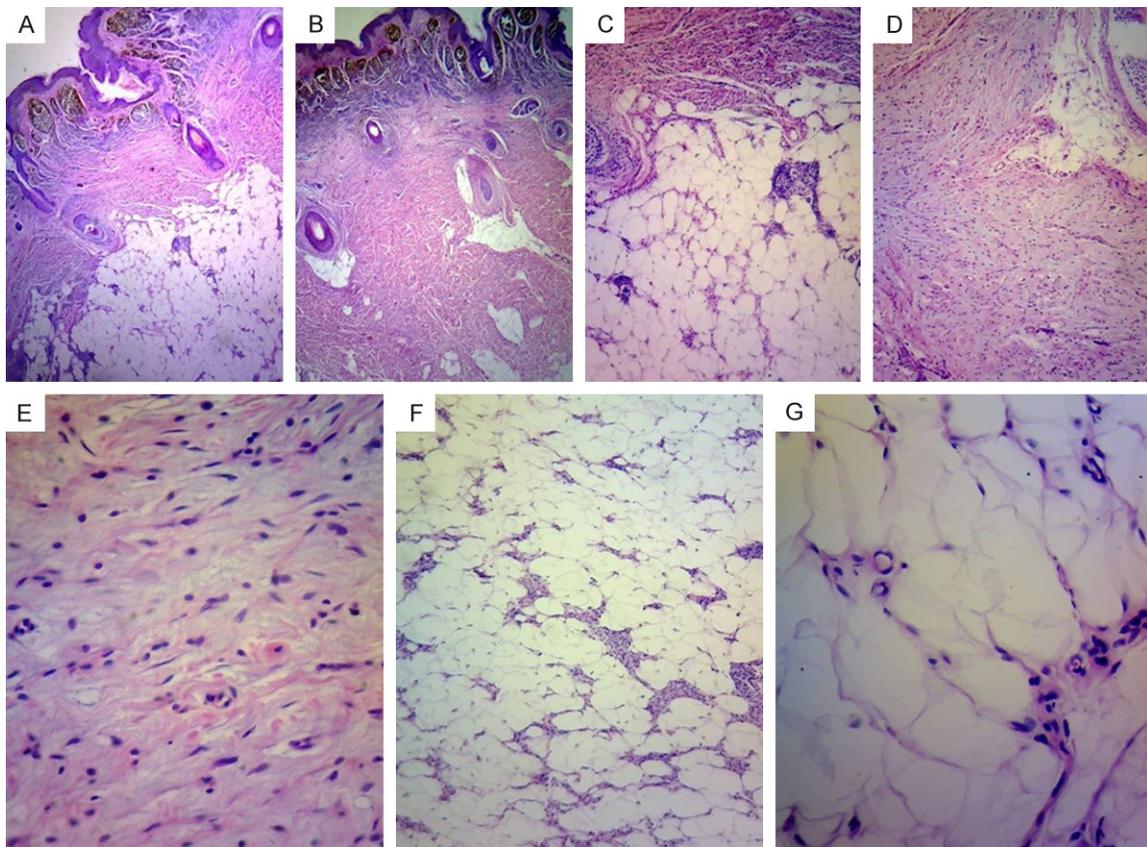
GCMN are the term to be synonymous to nevus measuring more than 20 cm in its greatest dimension or more than 2% of the body surface size, which often have a garment-like or bathing-trunk distribution [3]. The estimated lifetime risk of malignancy for GCMN is approximately 4.5-10% [4]. CMN have nevus cells in the dermis during the intrauterine life, which emigrated from the neural crest to the dermis, GCMN from a compound nevus are the presence of the back, buttocks, and posterior scalp that these areas are at higher risk because of the presence of more immature embryonic and neural cells [5].

There had been several reported that GCMN associated with other abnormalities including spina bifida occulta, meningocele, club foot and hypertrophy or atrophy of deeper structures of a limb, Carney complex, premature aging syndromes, neurofibroma, vitiligo, lipoma and dysplasia of bilateral hip [6-10]. Neurofibroma (NF) is a common benign tumor originated from neural crest, which can be categorized into two different types. NF-1 is one of the autosomal dominant inheritance and about 1/3000 of neonatal morbidity. The culture of NF-1 showed long arm of seventeenth chromosome abnormalities which involved the region 17q11.2 in Gene linkage studies [11], appearing single in the skin with single polypoid, nodular, and soft lesion. In addition to histology study, NF-1 lesion comprises scanty stained cytoplasm, slender nucleus and wavy spindled cells which arranged in the fibrillar, collagenous or mucoid matrix [12]. Lipoma is a common benign tumor of connective tissue, which occurs rarely in children. About 75% lipoma

## A case of neurofibroma and lipoma



**Figure 2.** MRI view showing about 8.5 × 5.5 cm lump with T1- and T2-hyperintensity, with clear boundary and homogeneous signal. Fat saturation sequence was in low and nonuniform signal.



**Figure 3.** HE staining ( $\times 40$ ,  $\times 100$ ,  $\times 400$ ) densely packed nevus cell nests in the epidermal-dermal junction and upper dermis, which arranged in fibroblast and collagen like matrix, exhibiting light staining, scant cytoplasm, slender nuclei. A large number of single vacuole mature fat cells were also visible in deep subcutaneous. Nuclei and cytoplasm for extruding mostly to one side and slender fiber spacing separated the tumor into leaflike components with intervals in which there were thin-walled vessels, with no significant cell atypia and nuclear fission. All areas showed features of both neurofibroma and lipoma.

cases exist gene rearrangement which are mostly located in 13~15 zone in long arm of the 12th chromosome [13]. Lipoma histologically show a lot of vacuolated mature fat cells, in

which nucleus are often on one side by the extrusion. The body of lipoma are often formed leafs separated by slender fiber intervals within thin-walled vessels [14].

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There have been reported that NF-1 or lipoma may occur in patients with GCMN in rare previous studies. It was first described by Bhagwat et al. reported that multiple nodules, which histopathological examination of the biopsy revealed features of both neurofibroma and lipoma, distributed over the forearms, face, upper chest and legs in the case of an eight-year-old girl [7]. However, in our case, a 3-years-old child with lesions distributed over entire neck, back, and the waist, whereas, a nodule from the lesion was also characterized by both neurofibroma and lipoma coexistence on histopathological examination. Although the exact mechanisms are not fully understood, it is generally acknowledged that their coexistence can be explained by the fact that both melanocytes and Schwann cells originate from the neural crest [10]. Due to the neural crest development dysplasia or defects, it may lead to abnormal development of its derivatives such as melanin cells, Schwann cells, fat, blood vessels, sensory neurons and muscles [15]. Because of the lack of more clinical observation data, the inherent relevance of GCMN, neurofibroma and lipoma occurring coincidentally in a patient has not been known. The advance research of GCMN may focus on gene inspections in the future.

### Disclosure of conflict of interest

None.

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