Isolated plexiform neurofibroma in a patient with tuberous sclerosis: A rare association

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ABSTRACT
Plexiform neurofibroma is a rare tumor originating in peripheral nerve sheaths. It is mostly associated with neurofibromatosis type 1 and occasionally as an isolated entity. We report a case of isolated plexiform neurofibroma associated with tuberous sclerosis.

KEY WORDS: Isolated, plexiform neurofibroma, tuberous sclerosis

INTRODUCTION
Plexiform neurofibroma is a characteristic finding of neurofibromatosis one though isolated cases exist. Neurofibromatosis belongs to phakomatoses which are a group of neurocutaneous disorders characterized by involvement of structures that arise from the embryonic ectoderm including central nervous system, skin, and eyes. Every disorder of this syndrome has some characteristic diagnostic features, though overlap or associations have been reported [1,2]. We report a case of isolated plexiform neurofibroma associated with tuberous sclerosis.

CASE REPORT
A 7-year-old girl was referred to dermatology outdoor for pain and swelling over the right side of her neck for past 1 year. On examination, we found multiple nodular swellings over the right side of her neck. The nodules were present on an underlying hyper pigmented mass [Figure 1]. The nodules were firm and painful to touch. On puncture cheesy material came out. These were diagnosed as sebaceous cyst. The underlying mass was almost 5 cm × 7 cm in diameter and felt like a bag of worms on palpation. It was clinically diagnosed as plexiform neurofibroma. We also found multiple keratotic small papules over her centro-facial region [Figure 2]. These were diagnosed as angiofibromas by fine needle aspiration cytology. We looked for other associated cutaneous abnormalities in favor of neurofibromatosis and tuberous sclerosis, but nothing was found. On inquiry, her mother told that the facial lesions started developing for past 2 years and the neck mass was visible since past 3 years. There is no history of consanguity, no family history, no history of epilepsy. Development milestones were normal. Eye examination was normal. Computed tomography (CT) scan of the head showed multiple ependymomas that one of the major criteria of tuberous sclerosis [Figure 3]. Tuberous...
Tuberous sclerosis is a common genetically determined neurocutaneous syndrome characterized clinically by variable neuropsychiatric manifestations ranging from epilepsy to mental retardation and autism. Cutaneous characteristics are facial angiofibromas, subungual fibromas, shagreen patches, and hypomelanotic macules. Cerebral ependymomas, cardiac rhabdomyomas, renal angiomyolipomas, and pulmonary lymphangiomatosis are some of the visceral findings [5]. Plexiform neurofibroma is a rare tumor originating in peripheral nerve sheaths that histologically is comprised Schwann cell proliferation organized in myxoid stroma. It usually at 4-5 years of age, while deep lesions can manifest in adulthood [6].

They are hyperextensible plaques that feel like a bag of worms on palpation. They have been found commonly over head and neck region though some unusual locations have also been reported [7-9].

Plexiform neurofibroma is usually associated with neurofibromatosis type 1, though isolated cases without any association have also been seen [10]. However, association of plexiform neurofibroma with tuberous sclerosis has not been reported till date. As phakomatoses comprise a group of disorders with some common features, some overlap symptoms are possible. Hence, we report this case.

REFERENCES


DISCUSSION

Phakomatoses, also known as neurocutaneous syndrome, is derived from the Greek words phacos = lens, spot; phacos = light. They comprise a group of central nervous system disorders with concurrent lesions in the skin, eye, and sometimes other visceral organs [3]. They include neurofibromatosis, tuberous sclerosis, ataxia telangiectasia, Sturge–Weber syndrome Von Hippel–Lindau disease (retinocerebellar angiomatosis), incontinentia pigmenti, basal cell nevus syndrome, and Parry–Romberg syndrome [4].

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