

## FACIAL PLEXIFORM NEUROFIBROMATOSIS IN A PATIENT WITH NEUROFIBROMATOSIS TYPE 1: A CASE REPORT

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The two major types of neurofibromatosis are neurofibromatosis type 1 (NF-1) and neurofibromatosis type 2 (NF-2) [1,2]. NF-1 is characterized clinically by multiple café-au-lait spots associated with discrete neurofibromas along peripheral nerves, while NF-2 is characterized clinically by acoustic neuromas associated with various tumors of the nervous systems, particularly meningiomas [1,2]. NF-1 is a synonym for von Recklinghausen's disease, named after its first publication by von Recklinghausen in 1882 [3]. The diagnosis of NF-1 is made according to the list already mentioned in the text of Hassan et al [4]. Major findings found in NF-1 other than Café-au-lait spots and discrete neurofibromas are intertriginous freckling, optic pathway tumors, Lisch nodules, plexiform neurofibromas, oral lesions, osseous lesions, skeletal abnormalities, vasculopathy, endocrine disturbances, urinary symptoms, neurological manifestations, sarcomatous changes of neurofibromas and other malignant diseases [2,5]. Among these, Café-au-lait spots and freckling should be the earliest manifestations of NF-1 [2,5]. However, clinicians often have difficulty in diagnosing NF-1 when patients show minimum numbers of Café-au-lait spots or freckling in childhood cases. In such cases in the early diagnosis of NF-1, a knowledge of plexiform neurofibromas may be extremely helpful, as the association of plexiform neurofibromas in NF-1 is less common than that of discrete, classic neurofibromas and as it is noticeable by the age of 2 years [2,4,5].

Plexiform neurofibroma is histologically similar to discrete neurofibroma, and is a diffuse elongated fibromatous tumor involving the trigeminal or upper cervical nerves [5,6]. As plexiform neurofibromas occur at an early age with cosmetically disfiguring miserable features, it affects the patient both physically and mentally [4]. Therefore, elucidation of the pathogenesis and the development of a treatment for plexiform neurofibromas are highly essential. Surgical resection of plexiform neurofibroma should be the first choice, but offers no favorable results in patients with huge extensive lesions like Hassan's case [4]. Currently, chemotherapeutic options for the management of

plexiform neurofibromas are is under way, including retinoic acid, angiogenesis inhibitors such as pegylated interferon  $\alpha$ -2b and thalidomide, farnesyl protein transferase inhibitors or cytokine modulators, as mentioned in Hassan's report [4]. In addition, pirfenidone, an antifibrotic agent that decreases proliferation of fibroblasts and collagen matrix synthesis, and conventional chemotherapy including a combination of methotrexate and vinblastine were also applied to this horrible disease of NF-1 [5]. Hassan et al. had reported a typical, but extremely huge lesion of plexiform neurofibromas with best quality figures.

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