Poliosis and Neurofibromatosis Type 1: Two Familiar Cases and Review of the Literature

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Established Facts

- Poliosis on the scalp in neurofibromatosis type 1 is rare.

Novel Insights

- We report two familiar cases of poliosis overlying plexiform neurofibromas in the same location in patients with neurofibromatosis type 1.

Keywords

Poliosis · Neurofibromatosis type 1 · Plexiform neurofibromas · Scalp

Abstract

Two brothers were referred to our clinic for reevaluation of neurofibromatosis type 1 (NF1). Both brothers presented a peculiarity that is not so common in NF1: poliosis overlying plexiform neurofibromas on the scalp. Poliosis overlying plexiform neurofibromas is rarely reported in the literature. The peculiarity of our cases is the familiarity and the presence of poliosis in the same location.

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Brief Report

Two brothers presented at our clinic for reevaluation of neurofibromatosis type 1 (NF1), which had been diagnosed, respectively, at 1 and 6 years. The oldest was 40 years old (case 1) and the other 34 years old (case 2). Family history revealed that their father, their paternal uncle, and their paternal grandmother had NF1, but there was no history of poliosis or achromic macules. We report the clinical cutaneous findings of the two brothers and the findings of other cases described in the literature in Table 1. Systemic symptoms of our cases are reported in Table 2.

Both brothers presented a peculiarity that is not so common in NF1: poliosis overlying plexiform neurofibromas on the scalp (Fig. 1). Neurofibromas are benign proliferations of the neural element of the peripheral nerve; they can develop from the Schwann cells or fibroblasts. Single or multiple skin neurofibromas are observed in more than 95% of cases of NF1. Plexiform neurofibromas, occurring in 30% of cases, are multiple neurofibromas with diffuse neural expansion along the peripheral nerve [1].

Table 1. Features of the patients with poliosis and neurofibromas

	Case 1	Case 2	Case 3	Case 4	Case 5
Sex	Man	Man	Man	Woman	Man
Age of the patient at dermatological visit, years	40	34	19	32	38
Site of poliosis	Right parieto-occipital region	Right temporo-parietal region	Left temporo-parietal region	Left temporal region	Right parieto-occipital region
Site of scalp neurofibroma	Right parieto-occipital region	Right temporo-parietal region	Left temporo-parietal region	Left temporal region	Right parieto-occipital region
Plexiform neurofibroma of the scalp	Yes	Yes	No	No	Yes
Clinical features of neurofibromatosis	Multiple nodular neurofibromas on the trunk, arms, and legs, plexiform neurofibromas in the pubic region and plantar region, blue-red pseudo-atrophic macules on the trunk, freckles in the axillary regions, café au lait spots, piezogenic papules on the feet	Multiple nodular neurofibromas, multiple plexiform neurofibromas, one of 2 cm in diameter on the right parietal region, another on the dorsal region	Isolated neurofibroma; no familiar history of neurofibromatosis type 1	Isolated neurofibroma; no familiar history of neurofibromatosis	Axillary freckling, multiple café au lait macules plus dome-shaped, soft, multiple superficial neurofibromas, trunk and extremities; no eye abnormality; normal physical and mental development; maternal aunt had piebaldism; a son with neurofibromatosis type 1
Age of onset of neurofibroma	First years of life	First years of life	4 years	22 years	Not indicated
Age of onset of poliosis	First years of life	First years of life	12 years	Six months after the neurofibroma began to grow	23 years
References	Patient we visited	Patient we visited	Patient described by Koplon and Shapiro [5]	Patient described by Kwon et al. [3]	Patient described by Sandoval-Tress and Nava-Jiménez [4]

Table 2. Systemic symptoms of the two brothers

Case 1	Case 2
Hypophosphatemic rickets Empty sella syndrome Hyperlipidemia Fatty liver disease Depression Panic attacks Bilateral sensorineural hearing loss Osteochondritis dissecans of the medial femoral condyle of the left knee Hypertension Prostatic hypertrophy Irises with Lisch nodules	Aortic coarctation surgically corrected at the age of 12 Incomplete right bundle branch block Three neurofibromas in the heart No ophthalmological abnormality

Fig. 1. Case 1 and case 2.

Poliosis circumscripta is a localized patch of white hair in a group of hair follicles, known as “white forelock,” secondary to the involvement of the central frontal scalp, but it can involve any hairy area of the body, including the eyebrows, eyelashes, and

beard [2]. Microscopically, poliosis demonstrates either decreased or absent melanin and/or melanocytes in the hair bulbs of the affected hair follicles [2]. The epidermal melanocytes are usually not affected, unless there is associated vitiligo [2].

In children, poliosis may be the presenting sign of several hereditary syndromes that should alert the physician to look for other cutaneous manifestations [2]. In fact it may occur in several genetic syndromes including piebaldism, Waardenburg syndrome, and tuberous sclerosis [2].

Poliosis overlying plexiform neurofibromas is rarely reported in the literature. To our knowledge, only three cases have been described, in two of which the neurofibromas are isolated, while in the third case, the patient was affected by NF1 [2–5]. In our two familial cases, the patients had NF1 onset in the first years of life.

In our cases and in all the cases described in the literature, the neurofibromas were localized on the scalp, in the lateral-temporoparietal region; they were acquired in all cases, including our own, except for one case, in which it is not indicated. It has been hypothesized that in such cases, poliosis is probably due to an autoimmune response in which the T cells cytotoxic to neurofibroma might cross-react with surrounding hair bulb melanocytes, resulting in their destruction with subsequent whitish discoloration of the hair surrounding the neurofibroma [2].

The peculiarity of our cases is the familiarity and the presence of poliosis in the same location. More studies are needed to explain the exact relation between poliosis and neurofibromas.

Statement of Ethics

All authors confirm that all the research meets the ethical guidelines, including adherence to the legal requirements of the study country. Patient consent has been obtained.

Disclosure Statement

All authors have not conflict of interest.

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