Unequaled overgrowth inside and out—an exceptional example of hypertrichosis overlying plexiform neurofibroma

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INTRODUCTION

Plexiform neurofibroma is classically associated with neurofibromatosis type I (NF1). However, there are relatively few cases that involve isolated neurofibroma of the orbit of varying pathologies (myxoid, plexiform, diffuse) in the absence of clinical criteria of NF1.[1] Plexiform and diffuse neurofibromas usually present in childhood; however, isolated neurofibroma of the orbit with overlying hypertrichosis appears to be a rare finding.[2-5] Hypertrichosis is the excessive growth of hair on non—androgen-dependent areas of the body and is classified based on the time of onset and distribution.[6] We present a case of extensive hypertrichosis overlying a plexiform neurofibroma in a patient without stigmata of NF1.

Abbreviations used:

NF1: neurofibromatosis type I

CASE REPORT

A 10-year-old girl presented to the neurology clinic with an 8-year history of progressive hypertrichosis with associated discoloration and swelling in the right periorbital region (Fig 1). The hypertrichosis covered most of the right half of her face, excluding only the mandibular, malar, and nasolabial regions. The coarse terminal hair was dense with a uniform, thick distribution. The swelling was confined to the periorbital region and did not have associated proptosis of the eye.

Five years prior, ophthalmology had evaluated her, and magnetic resonance imaging of the brain showed an extensive periorbital lesion compressing the optic nerve and extending posteriorly into the right orbit, retromaxillary, and suprazygomatic spaces (Fig 2). Characteristic of plexiform neurofibroma, the magnetic resonance imaging showed the lesion to be infiltrative, ropelike, and hyperintense on T2-weighted images with a target-like appearance. In addition, possible sphenoid wing dysplasia was found.

Alcohol injections to the lesion in periorbital region garnered no improvement. Over the next 3 years, the swelling below her eye increased, as did the surface area of hair coverage. Surgical resection was performed, and unavailable pathology was consistent with plexiform neurofibroma.

Detailed examination by ophthalmology and neurology showed 1 hyperpigmented macule on the right upper chest, but no stigmata of NF1, including no axillary or inguinal freckling, bone abnormalities, or Lisch nodules. Her mother revealed a personal history of numerous small nodules, biopsied and diagnosed as benign unspecified nodules; however, the pathology report was unavailable. The result of subsequent blood genetic testing for NF1 was negative. The patient was diagnosed with isolated periorbital plexiform neurofibroma with associated hypertrichosis.

The patient has done well with her atypical hair pattern and plexiform neurofibroma. Currently, the family has opted for minimal management because she currently suffers no ophthalmologic consequences. Her hairstyle is adapted to match the hair growth pattern, and she anticipates exploring hair removal options in the future. She will follow-up

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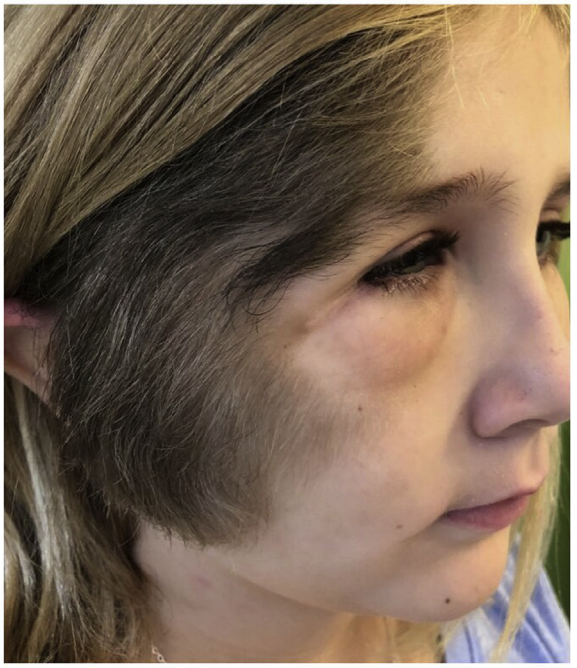

Fig 1. Hypertrichosis of the right side of the face and periorbital swelling.

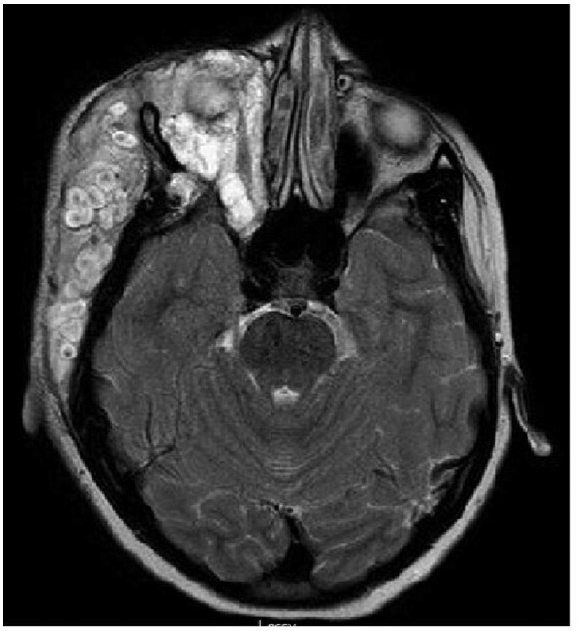

Fig 2. Plexiform neurofibroma extending into the right orbit.

with plastic surgery and ophthalmology, but has no current plans to pursue surgical resection.

DISCUSSION

Hypertrichosis overlying plexiform neurofibroma is a known finding; however, hypertrichosis to the extent seen in this patient is unusual.[2] Her lack of visual or oculomotor disturbances from the orbital lesion is impressive, and her prognosis for ocular function is uncertain. This highlights the importance of early recognition of abnormal hair patterns, because they have the potential to distract from more serious underlying pathology.

This patient's magnetic resonance imaging findings are consistent with NF1, but her genetic testing was negative. Several possibilities arise from this, including mosaic NF1, segmental NF1, and isolated plexiform neurofibroma. Another resection and further pathologic testing of the lesion is required to clarify the diagnosis, which the family does not desire.

The mechanism of hypertrichosis is not well defined,[5] and the genetic link of this overgrowth to isolated plexiform neurofibroma has yet to be elicited.[4] Multiple hypotheses for the etiology of hypertrichosis include abnormal signals enlarging the follicular papilla, including growth factors such as bone epidermal growth factor, and prolonged production of the normal anagen growth cycle.[4] However, a direct connection to plexiform neurofibroma does not exist.

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