

# A pale macule and a waxy pink lesion in a child with neurofibromatosis type 1

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## Case

A girl, three years of age, with neurofibromatosis type 1 (NF1) presented with two lesions for review. Both lesions had been present since birth and were not causing any concern. The lesion on her scalp (Figure 1) was absent of hair and the one on her left upper back (Figure 2) was more noticeable after a bath.

## Question 1

What is the lesion shown in Figure 1?

## Question 2

What are the possible complications of the lesion shown in Figure 1?

## Question 3

What is the management of the lesion shown in Figure 1?

## Question 4

What is the lesion shown in Figure 2?

## Question 5

How is the lesion shown in Figure 2 diagnosed and what is its pathophysiology?

## Question 6

Is there an association between the lesion shown in Figure 2 and NF1?

## Question 7

What is the management of the lesion shown in Figure 2?

## Answer 1

The lesion shown in Figure 1 is a sebaceous naevus or nevus sebaceous of Jadassohn.<sup>1,2</sup> It is an uncommon, benign, congenital hamartoma of the skin that comprises a combination of skin structures including hyperplastic epidermis, sebaceous glands, immature hair follicles and apocrine glands.<sup>1,2</sup> In infants and children, they present as pale, waxy, hairless plaques with a yellow–tan verrucous top and are usually diagnosed by clinical appearance alone.<sup>1,2</sup> In adolescence, under hormonal influences, they become more pronounced and verrucal in appearance.<sup>1,2</sup> Most lesions (95%) are located on the face or scalp,<sup>3</sup> and the size ranges from one to several centimetres.<sup>2</sup> Rarely, they form part of a syndrome called sebaceous naevus syndrome, which involves disorders of the eye, brain and skeleton.<sup>1,2</sup>

## Answer 2

Both benign and malignant skin lesions can arise in sebaceous naevi. The most common benign lesions include syringocystadenoma papilliferum and trichoblastoma.<sup>1</sup> Malignant growths are uncommon (5–22% of cases) and basal cell carcinoma (BCC) is by far the most common of these.<sup>2–4</sup> Squamous cell carcinoma (SCC), sebaceous carcinoma and adnexal carcinoma can also occur.<sup>1</sup> Most of these malignancies occur in adulthood, although rare cases of malignant growth in childhood have been reported.<sup>2,4,5</sup>

## Answer 3

Management of sebaceous naevi is controversial. Some studies suggest early excision and others advocate long-term monitoring with excision, as necessary, for malignant growth.<sup>1,2,5,6</sup> Long-term monitoring can be problematic as malignant lesions arising in sebaceous naevi may not present with their usual diagnostic features, making diagnosis difficult.<sup>3</sup> A sensible approach is to monitor



Figure 1. Waxy pink plaque on the scalp with absence of hair



Figure 2. Pale macules with surrounding erythema brought on by rubbing the area

**Box 1. Diagnostic criteria for neurofibromatosis type 1 (NF1)<sup>11,12</sup>**

A diagnosis of NF1 is established by the presence of at least two of the following criteria:

- six or more café au lait macules >5 mm (prepubertal) and >15 mm (postpubertal)
- two neurofibromas or one plexiform neurofibroma
- axillary or inguinal freckling
- Optic glioma
- two or more Lisch nodules (iris hamartomas)
- a characteristic bone lesion (pseudarthrosis or sphenoid dysplasia)
- a first-degree relative with NF1.

for any changes during childhood, when the risk of malignant transformation is low, and consider performing full thickness excision just prior to adolescence, when the risk increases.<sup>7</sup> The lesion may also be excised to reduce the nuisance value with grooming or to remove the hairless area for cosmetic reasons. At any stage, if any changes occur within a sebaceous naevus (eg rapid growth, irritation, ulceration), it should be reviewed by a dermatologist and biopsied or removed by full-thickness excision.<sup>1,3</sup>

**Answer 4**

The lesion shown in Figure 2 is a naevus anaemicus.

**Answer 5**

Naevus anaemicus is diagnosed clinically.<sup>8</sup> It presents as an asymptomatic pale macule and is roughly round, oval or linear in shape.<sup>8</sup> The main patch may be surrounded by satellite macules and is usually located on the trunk.<sup>8,9</sup> It is present from birth; however, it may not be noted until the individual is much older.<sup>8</sup>

Naevus anaemicus is caused by a localised vascular hypersensitivity to catecholamines that results in permanent vasoconstriction and pallor.<sup>8,9</sup> Putting a warm compress over the area or scratching it will not induce vasodilation

signified by erythema.<sup>8,9</sup> Diascopy, which involves putting a glass slide over the lesion and blanching the surrounding skin, reveals no border between the lesion and normal skin.<sup>8,9</sup> The diagnosis is normally made from history and examination alone. However, if the skin were biopsied, it would reveal normal skin and melanocytes.<sup>8</sup>

**Answer 6**

In one study, 8.8–51% of children with NF1 were found to have naevus anaemicus.<sup>10</sup> Before the age of two years, 50% of children with sporadic NF1 present with only one of the seven criteria for NF1 (Box 1), making the diagnosis of NF1 difficult during this time.<sup>11</sup> As naevus anaemicus is present since birth, it may be a possible indicator of NF1 in young infants with only one criteria of NF1 present,<sup>10,11</sup> but further research is required to substantiate this.

**Answer 7**

Management of naevus anaemicus involves reassurance and cosmetic measures as needed.<sup>1</sup>

**Key learning points**

- Malignant lesions may form in sebaceous naevi and close monitoring is recommended. Sebaceous naevi may be excised at the onset of puberty or they may be observed and full-thickness excision performed if any changes occur.
- Naevus anaemicus is a benign condition involving a localised abnormal vascular response to catecholamines that results in permanent vasoconstriction in the dermis. Diascopy results in loss of the border between the lesion and normal skin.

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