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CLINICAL REPORT

Congenital Spindle Cell Naevus with Unusual Transformation: Proliferative Nodule or Melanoma?

Katharina FLUX and Wolfgang HARTSCHUH
Department of Dermatology, University of Heidelberg, Heidelberg, Germany

Congenital melanocytic naevi can give rise to secondary melanocytic tumours, such as proliferative nodules and malignant melanoma. The clinical and histological features of both lesions may be nearly identical, which makes an unequivocal diagnosis impossible. In particular, it is difficult to differentiate clearly between benign and malignant proliferation in infants with secondary melanocytic proliferation. Reports on melanocytic proliferation and malignant melanoma within the paediatric age-group are very rare. There is limited expert knowledge on this subject and little is known about prognosis and outcome. We report here a case of an infant with an unusual transformation of a congenital spindle cell naevus of the umbilical region, and discuss clinical, histological and genomic criteria. ***Key words: proliferative nodule; congenital naevus; melanoma; spindle cell melanocytes.***

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Katharina Flux, Department of Dermatology, University of Heidelberg, Voss-Str. 2, DE-69115 Heidelberg, Germany. E-mail: katharina.flux@med.uni-heidelberg.de

Congenital melanocytic naevi occur in approximately 1% of newborns (1). These naevi bear a lifetime risk of developing secondary melanocytic proliferations, of which malignant melanoma is the most feared (2, 3). Diagnosis of malignant melanoma in the paediatric age group is rare and difficult (4, 5) since benign secondary proliferations may mimic the clinical features of malignant melanoma. Fortunately, the majority of these proliferations are benign (6–9); however, this makes identification of the exceptional case even more challenging. Conventional histology may not lead to an unequivocal decision between benign and malignant, because the criteria of malignancy are also partly met by benign proliferative nodules (1, 10–12).

CASE REPORT

Following a normal delivery, a first-born baby girl was found to have a congenital pigmented naevus in the umbilical region. There were no evident maternal lesions, and the placenta was of normal appearance. The naevus was asymmetrical, well circumscribed, consisted of both light and dark brown colour, with no satellites (Fig. 1A). No biopsy was taken of the naevus immediately after birth. The parents were advised to refer their daughter to the dermatologist regularly (every 6 months). Due to health problems in the father of the child, who developed a tumour of the testicles and was treated with chemotherapy, the baby girl's naevus was neglected and she was not referred to regular dermatological follow-up. At 1.5 years of age the girl was referred to our department because the naevus had developed a pink, fleshy nodule at the centre over a period of 6–8 weeks. The naevus had increased in size and had become papillary and partly verrucous, and the colour had changed to dark brown and black.

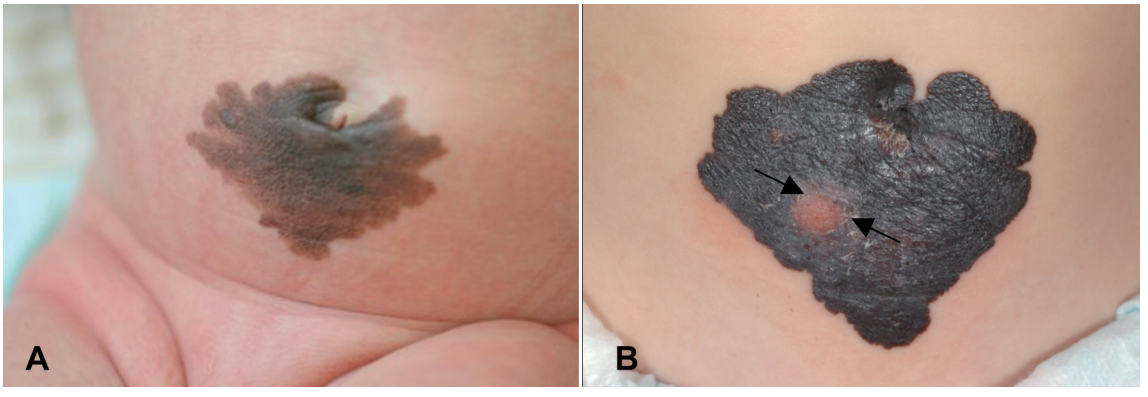

Fig. 1. (A) Congenital melanocytic naevus after birth. (B) The same lesion 1.5 years later. Note the disproportional growth, the increased thickness, the verrucous surface and the depigmentated ulcerated nodule (*arrows*).

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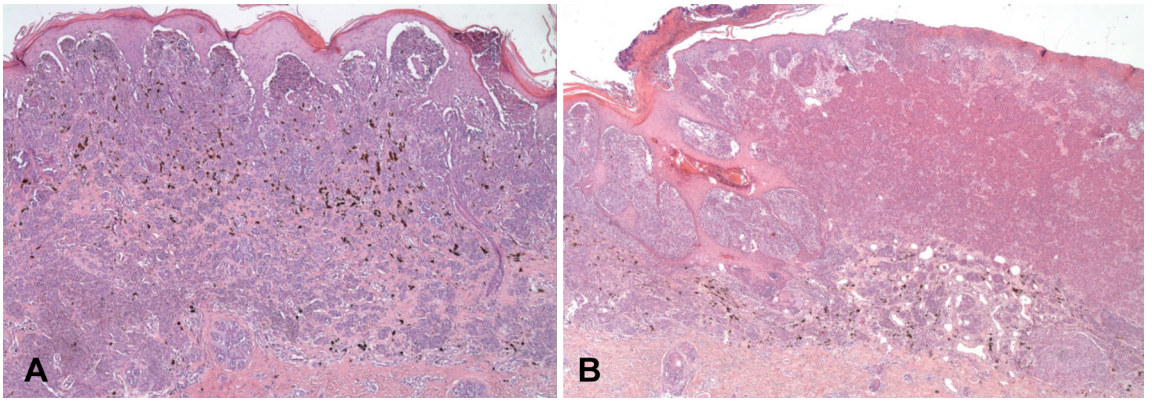

Fig. 2. Histological specimen of the surrounding naevus (A) and the ulcerated nodule (B) show two populations of cells (haematoxylin-eosin, low-power view ×2.5). (A) Pigmented, large spindle cell melanocytes with pleomorphic nuclei aggregated in vertically-oriented nests and fascicles distributed irregularly along the dermal–epidermal junction and reaching the deep corium resembling spindle cell naevus. (B) Population of melanocytes composed of discrete aggregates of small, round cells with monomorphous nuclei and loss of pigmentation.

The maximum diameter of the lesion was 7 cm (Fig. 1B). There were no other pigmented lesions elsewhere on the integument. The girl showed no evidence of lymphadenopathy or organomegaly.

The pink nodule in the centre of the naevus was excised including a margin of the surrounding dark tissue. Histology of the excised nodule revealed a pigmented congenital spindle cell naevus (Fig. 2A) with an ulcerated nodule at the centre composed of single and nested monomorphous melanocytes without definite maturation with descent into the dermis (Fig. 2B, 3A). The melanocytes were more densely packed than the surrounding naevus and had no pigmentation. There were multiple mitotic figures within the superficial parts of this nodule, some of them showing atypical forms (Fig. 3). The maximum vertical tumour thickness of the ulcerated nodule was 2 mm.

The diagnoses of malignant melanoma vs. a proliferative nodule in the background of a congenital spindle cell naevus were considered (Table I).

Immunohistochemical studies were performed using antibodies against Ki-67, S-100, HMB45 and Melan A. The superficial parts of the ulcerated nodule stained positively for Ki-67 and S-100, but did not express HMB45 or Melan A. In contrast to the central nodule, the surrounding congenital naevus did not express Ki-67, but stained positively for all melanocytic markers. A comparative genomic hybridization (CGH) was performed on the ulcerated area and the surrounding spitzoid area. No chromosomal aberrations were detected, either in the nodule itself or in the surrounding naevus. Thus, CGH supported the diagnosis of a benign secondary proliferation.

The clinical aspects and the histological material of this case were discussed within an expert forum of histopathologists. The majority opinion favoured a melanoma arising in a congenital naevus with features of a pigmented spindle cell naevus.

The remainder of the lesion was excised under general anaesthetic, with a small safety margin. There were no additional histological abnormalities in the remaining parts of the lesion. A sentinel biopsy was not performed. Magnetic resonance imaging of the cranium, lungs and abdominal organs, and ultrasound of the lymph-nodes did not reveal any metastases. Follow-up has been uneventful for 4 years.

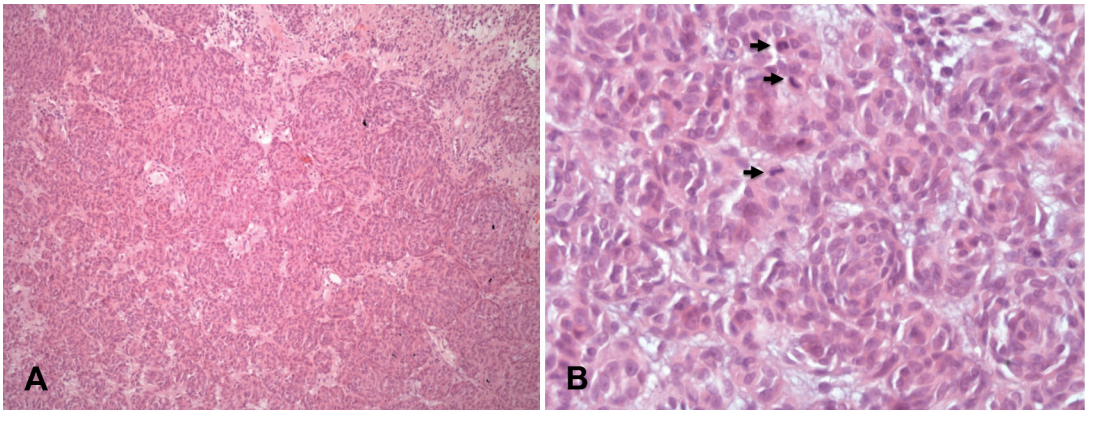

Fig. 3. (A) High-power view (×20) of the ulcerated nodule (haematoxylin-eosin) reveals monomorphous melanocytes that show a confluence of nests. (B) Multiple mitotic figures (*arrows*) in the superficial parts of the ulcerated nodule (high-power view ×40).

Table I. *Criteria for proliferative nodules within congenital naevi*

	Criteria
Clinical features	Naevus is usually large or giant Age: very young (within the first 2 years of life) Consistency: solid at first, progressive reduction in consistency. Loss of pigmentation over time Regression over time
Cytopathology	High cellular density – in contrast with congenital naevus in the background Monomorphism of cells Cells gradually merge with the surrounding melanocytes, intermediate forms of cells found at the border to the naevus Maturation of the melanocytes with descent
Other	Ulceration possible Necroses in exceptional cases only Slow growth (rapid growth possible in exceptional cases)

DISCUSSION

The case reported here is remarkable in two ways: first, the congenital naevus is a spindle cell naevus, which makes this case very rare compared with other cases in the literature; secondly, the proliferating tumour within this naevus was considered as melanoma by a majority of experts, although the CGH showed no aberrations. The confluence of melanocytic nests in the ulcerated focus of the lesion was one of the crucial histological criteria in favour of melanoma, since confluence of nests would be unusual for a diagnosis of benign proliferative nodule.

The transformational potential of congenital and non-congenital spindle cell naevi is unknown. Is has been discussed in the literature that Spitz's naevi may represent incomplete melanomas, since they show increased copy numbers of chromosome 11p. Therefore, it is generally accepted practice to excise these naevi. However, the congenital spindle cell naevus in our case is not synonymous with a Spitz's naevus. The question remains as to whether the naevus had always been a spindle cell naevus, or whether the spindle cell differentiation was due to an earlier transformation that developed over time. The CGH points to a true congenital spindle cell naevus in this case. Congenital spindle cell naevi have rarely been described in the literature (6, 13). According to Ackermann (13), congenital Spitz's naevi (referred to as one subgroup of spindle cell naevi) present as fawn-coloured patches, topped with red, brown or black papules, and congenital Spitz's naevi may present as segmental (systematized) lesions. They tend to grow more rapidly than other types of naevi, especially in childhood, and may become prominent within weeks (13). The risk of malignant transformation of the subgroup "congenital Spitz' naevi" cannot be assessed. In theory, melanocytes of a Spitz's naevus have no more or less capability of transforming into those of melanoma than do melanocytes of common congenital naevi (14). Cases of proliferative nodules within congenital Spitz' naevi have been reported (6).

Proliferative nodules usually present with a solid consistency at the beginning of their development, which reduces progressively. In addition, the pigmentation may be lost over time, and finally, the nodules should regress without medical interference (7, 13). In our case the nodular tumour within the naevus had been observed for 4 months prior to excision. No signs of regression were noted during that period, and the nodule did not show rapid growth. However, the total size of the congenital lesion had grown disproportionally to the size of the child. Congenital naevi usually grow in proportion with the rest of the skin. A disproportion of 15% is tolerated, and is seen in several congenital naevi during early childhood (15).

The following factors favour a benign lesion: the age of the girl (< 2 years); the lack of necroses within the nodule; the lack of mitotic figures in the *deep* parts of the lesion; and the lack of genomic aberrations (only 4% of all melanomas studied by Bastian et al. had normal CGH results) (15, 16). In addition, the nodule had developed in the superficial parts of the congenital naevus. This fact points more to a benign proliferation than to melanoma, since the latter is usually observed to have its origin in the dermal parts of congenital naevi. Immunohistochemical analyses are of limited use to distinguish melanoma from a proliferative nodule (13). In our case, staining with Ki-67 simply underlined the high mitotic rate in the superficial parts of the lesion. S-100, HMB45 and Melan A may be expressed by both melanoma and proliferative nodules (14). In this case HMB45 and Melan A were not expressed within the ulcerated lesion, matching the loss of pigmentation (the pink nodule).

CONCLUSION

Currently, the only reliable criterion of malignant melanoma is metastatic spread. Retrospective studies on young children with congenital naevi with secondary melanocytic proliferations are needed to provide long-term follow-up and an exact correlation with the histological findings. In the case reported here the authors favour a diagnosis of a proliferative nodule within a congenital spindle cell naevus.

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