SPITZ NEVUS IN A CASE OF NEUROFIBROMATOSIS TYPE1 – A CASE REPORT

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ABSTRACT

We present here an interesting case report of a patient presenting with Type1 Neurofibromatosis and a Spitz Nevus. A Spitz nevus is a benign nevus of spindle cells, usually pink, brown, or black in color. After an initial period of growth, the majority of Spitz nevi tend to stabilize their growth, reaching a final size smaller than 6 mm in about 95% of patients. The treatment indicated is complete excision to determine the lesion's depth and extension. Despite the fact that the Spitz nevus is a benign lesion, its transformation into a malignant lesion has been reported. Type 1 Neurofibromatosis is a rare genetic condition characterised by the development of multiple small nodules of a benign neural tumor which are neurofibromas, distributed all over the body posing a cosmetic problem to the patient.

Key words: *spitz nevus, neurofibromatosis type1*

CASE HISTORY

A 27 yr old female, presented to the outpatient wing of the hospital with complaints of a small, well circumscribed pink lesion, on her upper lip for 6 months and multiple nodules all over her body since birth. She had been previously diagnosed to suffer from Neurofibromatosis Type 1 . On detailed clinical examination, she was found to have multiple café au lait spots on her thorax and abdomen with multiple neurofibromas , the largest one in the nape of the neck, measuring 8x5cms and a lesion on her upper lip that was approximately 5mm in diameter, pink and soft. She was otherwise normal and had no complaints suggestive of any other diseases. All haematological investigations were found to be within normal limits. Excision biopsies were performed on the largest of the neurofibromas and the lesion on the upper lip. Grossly, multiple grey white soft tissue fragments were received and submitted to tissue processing.

Microscopically, Multiple sections from the material received from the lesion on the lip showed a hyperplastic epidermis with an admixture of spindle and epithelioid nevoid cells with abundant amphophilic cytoplasm and eosinophilic nuclei arranged in nests and alveoli extending into the mid-dermis. An occasional giant cell and few mitotic figures were also seen. The nevoid cells were uniform throughout the lesion, pigment was limited to the cells in the papillary dermis and a diagnosis of Spitz Nevus was offered and confirmed by positivity for S-100 and HMB-45 antibodies by immunohistochemistry. Sections from the mass on the nape of the neck showed a lesion composed of sweeping fascicles of benign cells with scant cytoplasm and elongated wavy nuclei having a serpentine configuration – Suggestive of a Neurofibroma.

DISCUSSION

The Spitz nevus, also known as benign juvenile melanoma or a nevus of epitheloid and spindle cells, was first described by Sophie Spitz in 1948 and is most common in children and adolescents. They can present in three different ways: solitary nodular, multiple grouped, and multiple disseminated. The great majority of lesions are solitary, nodular, and are present in children and adolescents (57%–70%), with a slight preference for the female gender. The regions where they are most frequently found are the lower extremities, the head and neck, and the trunk. Histologically, the spitz nevus is very similar to a melanoma due to the large size of the spindle cells with considerable nuclear and cellular pleomorphisms, presence of inflammatory infiltrate at the base of the lesion, and spread among collagen fibres. A classical spitz nevus consists of nests of large spindle or epithelioid melanocytes with a "raining down" pattern(Fig 1). The nests are fairly uniform, evenly spaced with no pagetoid spread. Epidermal changes such as acanthosis and hyperkeratosis maybe observed. Eosinophilic "Kamino Bodies"(Fig 2) are found intraepidermally, which are globular clusters of apoptotic melanocytes.The cells are often positive for S-100 (Fig 3) immunohistochemistry. The differential diagnosis includes malignant melanoma, mastocytoma, and pyogenic granuloma. Those lesions that are particularly hard to distinguish from malignant melanoma are called atypical or malignant Spitz nevi and Spitzoid cells. Immunohistochemical studies can help, but they are still not definitive. A Spitz Nevus expresses the S100 protein, but HMB-45 tends to be distributed more within the

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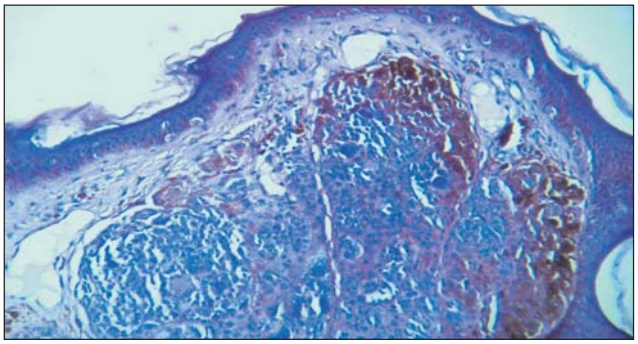
junctional and superficial components of the skin. HMB-45 correlates with melanosome production and thus, HMB-45-positive cells are of melanocytic origin. Therefore, HMB-45 may correlate best with factors that stimulate melanocytic proliferation and the production of melanosomes.The lesion is conservatively treated and surgical wide local excision is usually curative.

Neurofibromatosis Type 1 (NF1) formerly known as Von Recklinghausen's Disease is a congenital genetic disorder characterised by gene mutations in Chr 17 coding for a protein called neurofibrin which codes for RAS oncogene , leading to multiple lesions in the nervous system(neurofibromas) (Fig4), bones(scoliosis) , skin(café au lait spots) and eyes(lisch nodules). Treatment is usually only symptomatic.

CONCLUSION

To the best of our knowledge, this is the first time in literature that these two lesions have been reported in the same patient.Spitz nevus should always be clearly distinguished from malignant melanoma as they are two separate entities at ends of a spectrum and treatment differs. Neurofibromatosis 1 is a separate entity by itself that is congenital and seems to co-exist with spitz nevus in this case. The association of these two conditions occurring together may probably point to a common etiology which needs to be determined.



1.Fig 1 . Low power view (H&E x 10) Nests of large epitheloid melanocytes with a "raining down" pattern located subepidermally

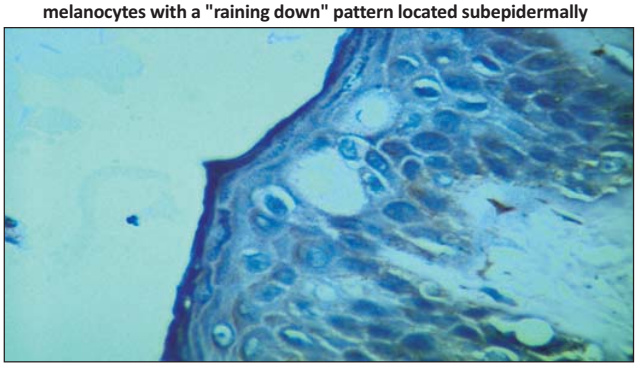


Fig2. High power view (H&E x 40) Kamino body

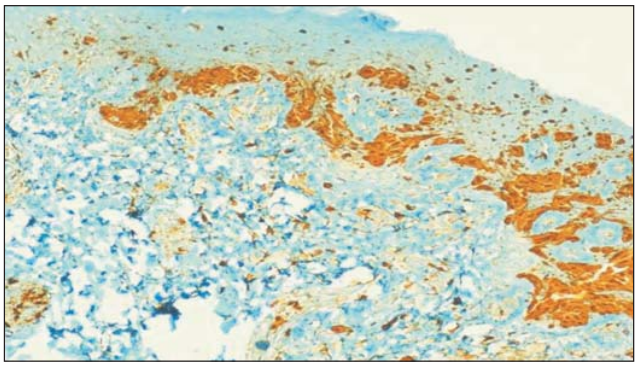


Fig3. Scanner view (H&E x4) IHC – intense nuclear S-100 positivity of the melanocytes

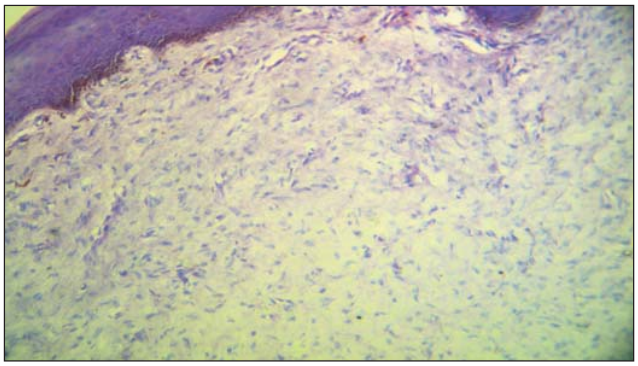


Fig4. Low power view (H&Ex10) Spindle cells with wavy nuclei - neurofibroma

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