

# Nevus Sebaceous of Jadassohn - A Rare Case Report

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## 1. Introduction

Nevus sebaceous, also called nevus sebaceous of Jadassohn or organoid nevus, is a benign hamartoma of the skin, characterized by hyperplasia of the epidermis, immature hair follicles, and sebaceous and apocrine glands. Lesions are usually present at birth and appear as waxy, yellow-orange or tan, hairless plaques. They have a tendency to thicken and become more verrucous over time, especially around the time of puberty [1].

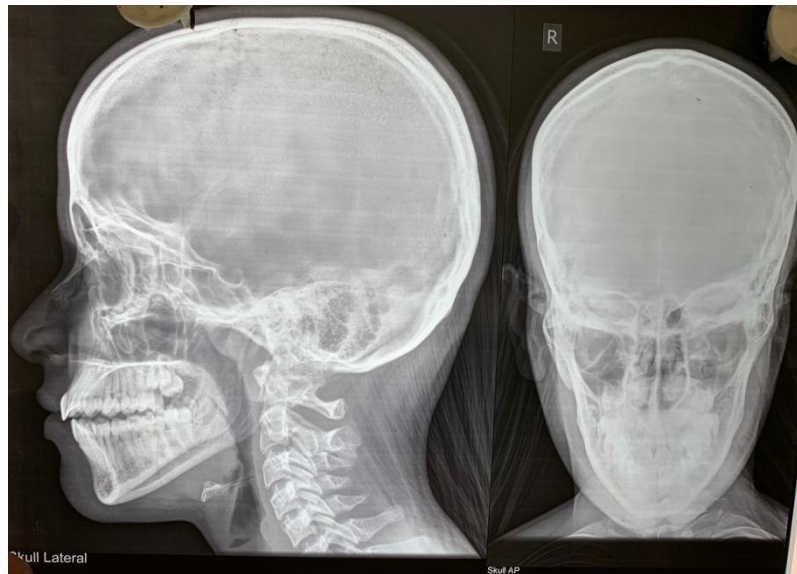
## 2. Case Report

An 14 year old girl presented to paediatric outpatient department with complaints of raised pigmented lesion over the right side of the scalp for the past 12 years. The lesion gradually increased in size to reach the present size. Parents gave history of hairless yellowish plaque present over the

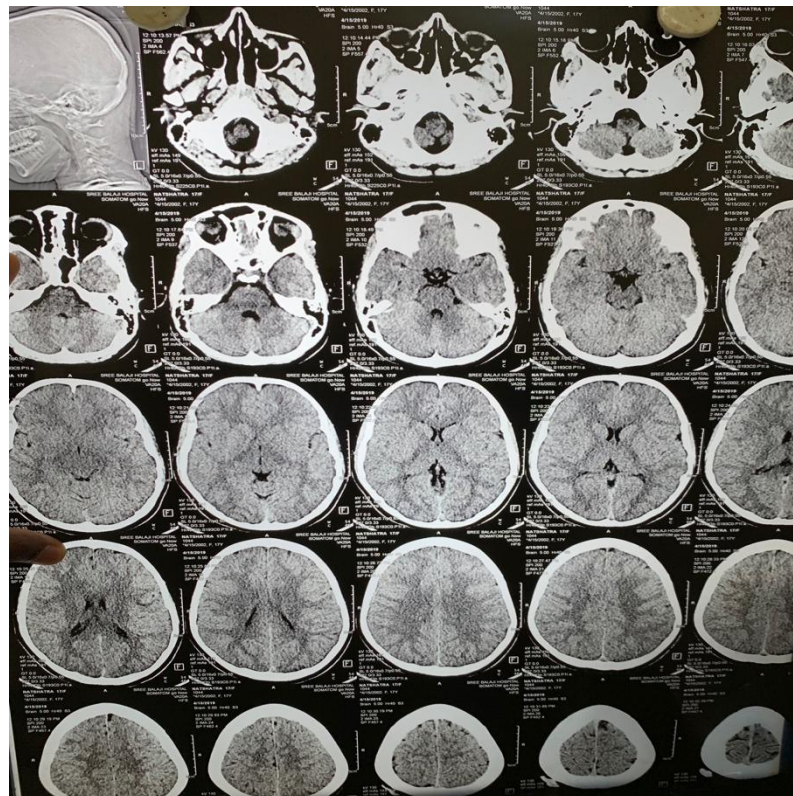
scalp at birth. There was no history of trauma. Her systemic investigation was normal and no developmental defect was noted. On cutaneous examination, a single 8\*7 cm blackish, soft, cerebriform and well demarcated nodular plaques was present over the scalp with multiple folds. (Figure 1). Baseline haematological investigations done were normal. X-ray skull done was normal with no bony extension (Figure 2), CT- Brain done showed an enhancing exophytic hyperdense skin lesion noted in the right parieto- occipital region with associated fat stranding, no adjacent bony erosion and no intracranial communication suggestive of Nevus Sebaceous. (Figure. 3). Histopathological examination showed papillomatous epidermal hyperplasia with hyperkeratosis and large number of mature sebaceous glands in the dermis along with follicular plugging with malformed hair follicles. The diagnosis of cerebriform type of nevus sebaceous was made.



**Figure 1:** A single cerebriform lesion of size 8\*7cm in the right parieto-occipital region



**Figure 2:** X-ray scalp with no bony extension of the lesion



**Figure 3:** CT-Brain with no intracranial extension

### 3. Discussion

Nevus sebaceous occurs in approximately 0.3 percent of new-born's, without sex predilection. It is usually sporadic, but familial cases have been reported [2,3]. Nevus sebaceous and nevus sebaceous syndrome (Schimmelpenning syndrome) are thought to be caused by postzygotic mosaic mutations in the *HRAS* or *KRAS* genes [4], although isolated cases due to mosaic mutations in *NRAS* and *FGFR2* have also been reported [5,6]. *RAS* promotes cell growth through activation of multiple pathways, including the mitogen-activated protein kinase (MAPK) signal-transduction pathway. Activating germline mutations in this gene family are involved in the pathogenesis of several inherited

malformation syndromes (example, Costello syndrome, Noonan syndrome, neurofibromatosis 1, Legius syndrome), some of which are associated with an increased risk of cancer.

In nevus sebaceous, the epidermis shows varying degrees of acanthosis and papillomatosis. In early lesions, sebaceous glands may be underdeveloped and decreased in number. The presence of immature hair follicles is characteristic and diagnostic. After puberty, the epidermis shows prominent papillomatous hyperplasia. Sebaceous glands are abundant and sometimes situated at an abnormally high level in the dermis. Ectopic apocrine glands can be seen in most lesions, sometimes deep in the dermis, beneath the sebaceous glands.

In most cases, nevus sebaceous is apparent at birth and presents as a well-defined, thin, yellow-orange or tan, oval, round, or linear plaque. Sometimes lesions are not noticed until later in childhood or after puberty. Most commonly, nevus sebaceous occurs on the scalp and is associated with a well-defined, localized area of alopecia. In older children and adults, lesions tend to be more elevated, verrucous, or nodular [7]. The size ranges from one to several centimetres.

In infancy and childhood, nevus sebaceous remains mostly unchanged, due to the quiescence of sebaceous glands. At puberty, under hormonal influences, lesions thicken and become verrucous or nodular. Rapid, circumscribed enlargement or ulceration should arouse suspicion for malignant transformation.

A variety of benign and malignant epithelial neoplasms may arise within sebaceous nevi. Benign tumors include viral warts, trichoblastoma, syringocystadenomacystoma, tricholemmoma, sebaceoma, nodular hidradenoma, hydrocystoma, and eccrine poroma [1,7-9]. Malignant neoplasms include basal cell carcinoma, apocrine carcinoma, trichilemmal carcinoma, sebaceous carcinoma, microcystic adnexal carcinoma, porocarcinoma, and squamous cell carcinoma. There is an isolated report of melanoma arising in a sebaceous nevus [10]. The exact incidence and the lifetime risk of malignancy arising in nevus sebaceous are unknown. In a review of nearly 5000 cases, benign and malignant tumors (mainly basal cell carcinomas) developed in 16 and 8 percent of patients, respectively [11]. However, the incidence of basal cell carcinoma may have been overestimated, due to misinterpretation of trichoblastoma. Large case series published after 1990 report rates of basal cell carcinoma of <1 percent [12,13]. The risk of malignancy increases with age, but basal cell carcinomas have also been reported in children. There are several reports of multiple different tumors arising simultaneously in the same sebaceous nevus [15, 16].

#### 4. Diagnosis

The diagnosis of nevus sebaceous is usually made clinically, based upon the finding of a circumscribed, slightly raised, yellow-orange plaque located on the scalp or face. If the diagnosis is in question, a tissue biopsy should be performed for histologic confirmation. Typical histopathologic findings include immature hair follicles; hyperplastic, immature sebaceous glands; dilated apocrine glands; and epidermal hyperplasia [8].

No additional evaluation is needed for children presenting with a small, solitary nevus sebaceous. However, in patients with large or extensive lesions and suspected nevus sebaceous syndrome, a thorough neurologic and ophthalmologic examination should be performed. Additional evaluation is based upon clinical findings and may include electroencephalography, neuroimaging studies with magnetic resonance imaging (MRI), skeletal radiography, and analysis of liver and renal function, including urine calcium and phosphate levels.

#### 5. Management

The definitive treatment of nevus sebaceous is full-thickness excision. However, the necessity and timing of excision to prevent possible future malignancy are still debated [11, 12, 13]. The decision to excise the lesion should be made in individual patients, based upon age, extension and location of the lesion, and the patient's or parents' concern about the cosmetic appearance and/or risk of malignancy. Since the risk of malignant transformation appears to be lower than previously believed, observation may be reasonable for lesions that do not cause cosmetic concern. Alternatives to surgical excision include photodynamic therapy, carbon dioxide laser resurfacing, and dermabrasion. However, since these treatment modalities do not completely remove the lesion, the risk of recurrence and potential for neoplasm development remains [1,8]. For children with Schimmelpenning syndrome and phacomatosis pigmentokeratocica, a multidisciplinary approach is needed, depending upon the type and extent of organ involvement, and may require the coordination of different specialists such as a dermatologist, pediatric neurologist, ophthalmologist, orthopedic surgeon, plastic surgeon, and psychologist [17].

#### 6. Conclusion

- Nevus sebaceous is a rare, benign hamartoma of the skin, characterized by hyperplasia of the epidermis, immature hair follicles, and sebaceous and apocrine glands.
- In most cases, nevus sebaceous is apparent at birth and presents as a well-defined, thin, yellow-orange or tan, linear plaque usually located on the scalp. In older children and adults, lesions tend to be more elevated, verrucous, or nodular. Nevus sebaceous syndrome is defined by the association of a nevus sebaceous with cerebral, ocular, or skeletal defects.
- A variety of benign and malignant epithelial neoplasms may arise within sebaceous nevi, including trichoblastoma, syringocystadenomacystoma, and basal cell carcinoma.
- The diagnosis of nevus sebaceous is usually based upon the clinical presentation. A biopsy for histopathologic confirmation may be warranted if the diagnosis is uncertain. For patients with suspected nevus sebaceous syndrome, a thorough neurologic and ophthalmologic examination should be performed. Additional evaluation includes electroencephalography, neuroimaging studies, and skeletal radiography.
- The definitive treatment of nevus sebaceous is full-thickness excision. The decision to excise should be made in individual patients, based upon age, extension and location of the lesion, and the patient's or parents' concern about the cosmetic appearance and/or risk of malignancy. Since the risk of malignant transformation appears to be lower than previously believed, observation may be reasonable for lesions that do not cause cosmetic concern.

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