Congenital Naevus Sebaceous of Jadassohn in a Neonate: an Early Presentation of a Rare Lesion

Kathleen Moedts, Karlijn Van Damme

University of Antwerp, Antwerp University Hospital, Division of Neonatology, Edegem, Belgium

kathleenmoedts@hotmail.com

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Abstract

Naevus sebaceous of Jadassohn is a congenital hamartoma of the skin which, when associated with multisystemic complications, may manifest as linear naevus sebaceous syndrome or Schimmelpenning-Feuerstein-Mims syndrome. Naevus sebaceous is associated with postzygotic mutations, which may increase the risk of secondary tumour development within the lesion. Notably, secondary tumour transformation appears to be predominantly observed in adults, leading to controversy regarding the optimal timing for surgical excision. In this report, we describe a unique case of a neonate with a naevus sebaceous of Jadassohn, shedding light on the early onset of this rare lesion.

Case presentation

This case report describes the birth and clinical course of a male neonate born at term with a birth weight of 4200 grams (>90th percentile), a birth length of 55 centimetres (>90th percentile) and a head circumference of 35 centimetres (25-50th percentile).

He was born after an uneventful vaginal delivery with Apgar scores of 9/10/9 at 1, 5 and 10 minutes respectively. Pregnancy and antenatal ultrasound showed no abnormalities.

Initial clinical examination revealed a parietal erythematous, papillomatous and irregularly defined mass 3-4 cm in diameter and 2 cm thick. There was a small necrotic zone in the centre. The mass wasn't covered by skin (see Figures 1 and 2). An underlying skull defect could not be assessed.

There were no other clinical abnormalities, the patient was asymptomatic and well.

Because of this mass, he was transferred to a level 3 neonatal intensive care unit for further evaluation.

Magnetic resonance imaging (MRI) of the head revealed a soft tissue mass in the cutis and subcutis in the midline above the vertex. There was no evidence of intracranial extension or damage to the skull, nor were there any intracranial abnormalities. As computed tomography (CT) is considered to be a more sensitive examination for skull involvement, a head CT was performed, which showed a polylobular vascular tissue structure centred in the cutis/subcutis high parietal on the midline. There was no communication with the underlying skull.

Based on clinical examination and additional imaging, the differential diagnosis included fibroepithelial polyp, infantile myofibroma, or slow-flow vascular malformation.

Surgical removal, performed within the first 24 hours, was uneventful.

Histopathological examination of the parietal mass revealed a lesion consistent with naevus sebaceous (NS) of Jadassohn, which was completely excised. Sections through the various fragments showed skin with underlying adipose tissue. The epithelium, which was locally slightly verrucous, showed no signs of atypia. There were several hair follicles in the dermis and direct outflow of sebaceous glands onto the epithelium. Elsewhere in the fragment a proliferation of blood vessels was seen. The endothelial cells were not atypical. The blood vessels were dilated and congested with red blood cells. Extravasation of erythrocytes and areas of haemorrhage were seen at several sites. There were no atypical features. This benign haemangiomatous lesion was morphologically consistent with a cavernous haemangioma or vascular malformation, which appeared to have been incompletely removed.

Genetic studies could not be performed because the resection material was not preserved.

Further investigations ruled out nevus sebaceous syndrome, with ophthalmological examination, cardiac ultrasound and abdominal ultrasound showing no abnormalities. Head MRI showed no evidence of intracranial abnormalities and there was no clear evidence of skeletal abnormalities.

A medical genetics consultation was arranged for follow-up. The patient was also re-evaluated by a neurosurgeon approximately 5 months after surgery, which showed a favourable evolution, there was no recurrence of any lesions.

Follow-up was performed by the local paediatrician. No developmental problems or other associated symptoms were noted up to the present age of 2 years.

Discussion

NS of Jadassohn, first characterised by the dermatologist Josef Jadassohn in 1895, is a congenital hamartoma of the skin with hyperplasia of the epidermis, hair follicles and sebaceous and apocrine glands (1).

Clinical manifestations include linear or oval lesions with a smooth or verrucous texture, generally alopecic and in a range of colours. The scalp is the most commonly affected area, followed by the cephalic region. Involvement of the trunk and neck is less common. NS can vary in size from a few millimetres to 10cm, and giant nevi are extremely rare (2).

The association of multisystemic complications in NS leads to its classification as linear naevus sebaceous syndrome (NSS) or Schimmelpenning-Feuerstein-Mims syndrome (2, 3).

The estimated incidence of NS in newborns ranges from 0.1% to 0.3%, with no apparent gender or ethnic predilection. NSS is rare and the exact prevalence and incidence in the general population is unknown (2).

The differential diagnosis encompasses different syndromes, including cutaneous-skeletal hypophosphatemia syndrome, naevus comedonicus syndrome, Becker naevus syndrome, phakomatosis pigmentokeratotica, congenital hemidysplasia with ichthyosiform erythroderma and limb defects, and segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal naevus syndrome (4).

NS is characterised histologically by immature and abnormally formed pilosebaceous units. Epidermal changes may show some acanthosis and mild papillomatosis. With age, the lesion may increase in size, with a more prominent location of the sebaceous glands high in the dermis and an increase in the number of sebaceous lobules and malformed ducts.

Despite its prevalence as a common cutaneous lesion, the phenotype described in our case can be considered a rare and atypical case, with histological changes consistent with naevus sebaceous or naevus-sebaceous-like, in addition to a vascular malformation. Few case reports document phenotypes comparable to this case and are described as papillomatous pedunculated NS (5).

NS can be caused by various genetic factors, as demonstrated by Groesser et al. who studied 65 sebaceous naevi. Their study showed that 95% of these lesions had mutations in the Harvey rat sarcoma virus (*HRAS*) gene, while 5% had mutations in the Kirsten rat sarcoma virus (*KRAS*) gene. Nonlesional tissue from 18 individuals had a wild-type sequence, confirming genetic mosaicism. Their results suggest that NS and NSS are caused by postzygotic *HRAS* and *KRAS* mutations (6).

During puberty, hormonal fluctuations can cause proliferation and hyperplasia of the lesion, resulting in enlargement and a more verrucous appearance (7).

In addition to aesthetic concerns, NS carries the risk of secondary benign or malignant neoplasms (2). Various mutations may predispose individuals to the development of secondary tumours in NS (6). Specific *HRAS* mutations identified by Groesser et al. were also present in all associated secondary tumours studied, suggesting a common genetic basis. In particular, NS and basal cell carcinoma (BCC) also share deletions in the Patched Tumour Suppressor (*PTCH*) gene, which may account for the possibility of BCC arising within NS. Other possible risk factors for BCC in NS include Fitzpatrick phototypes I and II, family history, prolonged sun exposure, use of sunbeds or radiotherapy (8).

Studies suggest that secondary neoplasms occur in approximately 10% to 20% of cases of NS, the majority of which are benign. Only about 3% of cases show some degree of malignancy, which is a rare occurrence (2). Secondary tumour transformation appears to occur almost exclusively in adults, as a retrospective analysis found that 96% of all NS-derived malignancies occurred in patients over 18 years of age, with the remaining 4% in the 11-17 year age group. Common benign tumours include trichoblastoma and syringocystadenoma papilliferum, while the most common malignant tumour is BCC (9).

A conservative estimate suggests that the lifetime risk of malignant transformation in NS is less than 2%. However, determining the true lifetime risk is complicated by divergent numbers in children versus adults from studies on NS specimens, given that the majority are excised in childhood and adolescence (1).

As secondary tumour transformation appears to be almost exclusively seen in adults, the timing of surgical intervention remains controversial. Although smaller lesions may be technically easier to remove, it is important to consider the risks of general anaesthesia in younger patients.

Figure 1: Parietal erythematous, papillomatous and irregularly defined mass with a diameter of 3-4 centimetres and thickness of 2 centimetres.

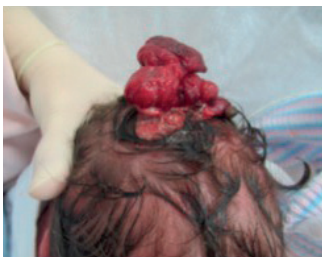
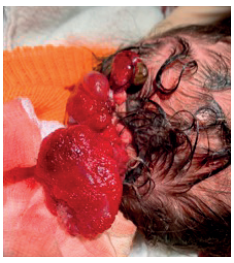


Figure 2: Close-up of the lesion, showing small necrotic zones.



Therefore, it has been suggested that excision should be performed prior to pubertal enlargement, provided that local and general anaesthesia are well tolerated, rather than waiting until malignant features develop (3).

Due to the size of the lesion in our patient, early excision appeared to be justified.

Other methods are frequently used to treat and improve Jadassohn lesions, including curettage, cauterization, cryotherapy, photodynamic therapy, topical salicylic acid, topical and systemic retinoid, topical application of vitamin D analogue, laser treatment, and dermabrasion (2).

The Schimmelpenning-Feuerstein-Mims syndrome was originally described as a triad of symptoms, including neurological impairment, seizures, and intellectual disability, associated with the presence of a NS. However, this syndrome has evolved to encompass multisystemic, extracutaneous complications involving a variety of organs including the nervous, ocular, cardiovascular, muscular, genitourinary and bone systems. The most frequently reported complications in the literature are hypophosphatemic rickets, intellectual disability, cognitive impairment, coloboma, and strabismus (2).

Due to the rarity of NSS and the extensive assessment that should follow, not all patients with NS require these examinations unless there are associated symptoms. However, if there are other associated symptoms, such as developmental delay, it is crucial to conduct assessments, including prenatal, developmental, and family histories, alongside neurological, ophthalmological, and cutaneous examinations. Any child with NS on the head or neck and developmental delay should undergo brain imaging. To evaluate for kyphoscoliosis, gait, and limb length, skeletal examinations should be conducted. Additionally, skin biopsies and relevant laboratory studies, such as serum/urine calcium and phosphate, liver and renal function tests, are critical (4).

In our patient, no resection material was retained for genetic testing. Given the lack of associated features and the low likelihood of detecting a genetic abnormality outside the lesion, it was decided not to pursue further genetic research at this time. Histopathological examination revealed findings consistent with a benign haemangiomatous lesion in addition to the sebaceous naevus. An extensive literature search did not reveal any association between haemangiomatous lesions and NS. There are few studies that have focused on assessing the prevalence of vascular malformations in NSS. Greene et al reviewed the medical records of 9 patients with NSS and concluded that 3 of these patients had various forms of vascular malformation including aortic aneurysm, carotid stenosis and lymphatic malformation (10). It remains unclear whether the vascular lesion described in our case should be considered as a separate entity.

Conclusion

This case report describes a rare and extensive phenotype of NS associated with an additional vascular malformation. The nature of this vascular malformation, whether it is associated with NS or a distinct entity, remains unknown.

NS is a common lesion in neonates. It is important to be aware of this pathology and its possible occurrence in Schimmelpenning-Feuerstein-Mims syndrome, which can affect multiple organ systems. Long-term follow-up is mandatory for the evaluation of associated symptoms and requires a comprehensive assessment.

While there is ongoing debate about the ideal management of NS, it is important to make decisions on a case-by-case basis and to recognise that there is a window of observation in affected children, given the gradual nature of secondary tumour transformation.

Conflict of interest

The authors have no conflicts of interest to declare with regard to the topic discussed in this manuscript.

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