SEBACEOUS NEVUS SYNDROME (SCHIMMELPENNING-FEUERSTEIN-MIMS SYNDROME) ASSOCIATED WITH INFANTIL SPASMS, INTRACRANINAL HAMARTOMA, CORTICAL BLINDNESS AND MICROPHTHALMIA

Mohammed Mazen Mourad, MD PhD; Talal Tallab, MD; Khalid Bahamdan, MD

Introduction

Sebaceous nevus syndrome describe any combination of a sebaceous nevus with any single significant of skeletal abnormality, neurological abnormality and ocular abnormality. This syndrome is more common in-patient with multiple or extensive skin lesions. No genetic contribution in most cases. A wide range of skeletal abnormalities were found in sebaceous nevus syndrome including Kyphosis, Scoliosis, Hypertrophy, atrophy and short limbs, Syndactyly, Vitamin D resistant rickets and Cystic changes.

Also many neurological abnormalities which occur in 50% of cases including mental retardation in 40%, epilepsy in 30%, infantile spasms, spastic hemiparesis 20%, cerebral angiomas, cortical atrophy and intracranial hemartoma.

Ocular abnormalities which occur in 30% of sebaceous nevus syndrome including involvement of the eyelid or conjenction by the new sebaceous, cortical blindness, microphthalmia, macrophthalmia & anophthalmia, corneal opacities cataracts, and colobomas of the eyelids, iris and retina.

Case Report

Six month old, Saudi infant girl, from Jizan was admitted to A.C.H. under the derma care for investigation, with history of extensive skin lesions since birth, occurs on the head, neck and upper part of chest, and scalp.

The infant is the second child in the family. First child was normal. There is history of consanguinity. Infant was a product of FT uncomplicated pregnancy, has normal development and breast feeding. History of repeated attacks of convulsions since the age of 3 months. Physical Examination, generally the infant looks well. Vital signs were stable, no cyanosis, jaundice or lymphadenopathy. Chest, CVS, CNS and abdomen were normal. Left eye looks smaller compare to the right eye. Patient is not following bright object but pupils reacting normally to the light. Cutaneous Examination showed slightly elevated, yellowish, rough, circumscribed, extensive plaques on the scalp, and temporal area bilaterally, with extension to both cheeks, chin, and upper lip associated with scarving alopecia on the vertex and occipital area of the scalp. Small skin plaque of similar morphology present on the upper part of chest (Figs. 1.2.3.4.5). Mucous membrane is normal.

Clinical Impression was Sebaceous nevus syndrome (Schimmelpenning-Feuersstein-Mims syndrome). Laboratory Investigation were including Complete Blood Counts, Liver Function Tests, Urea & Creatinin, Electrolyates, Chest X-ray, Electro Cardio Gram, Abdominal ultrasound, all were within normal limits. Other investigation has been done including skeletal survey were normal. CT scan normal, MRI: Normal brain with superficial occipital nodular lesion (Hamartoma), EEG - Sleep - Abnormal finding, Skin biopsy from the scalp for histopathology showed incomplete differentiated hair structures, foci of undifferentiated cells representing malformation of hair germ. The histopathological finding was consistent with nevus sebaceous. Patients was refereed to pediatric were diagnosed as infantile spasm, Her seizures were controlled by sodium valporate and vigabatrin. Infant was referred to ophthalmologist where diagnosed as left microphthalmia and cortical blindness.

Correspondence should be addressed to:
Dr. Khalid Bahamdan
Division of Dermatology, Internal Medicine Department
College of Medicine & Medical Sciences, King Khalid University
P.O. Box 641, Abha, Kingdom of Saudi Arabia
Tel.: 966 7 2247800, Telefax: 966 7 2264659

Fig. 1 Slightly elevated, yellowish, rough, circumscribed, plaques on the scalp and right temporal area, associated with scarving alopecia.
The final diagnosis was Sebaceous nevus syndrome with the following findings: Extensive nevus sebaceous of Jadassohn on the head & neck, neurological abnormalities including: Infantile spasms and intracranial hamartoma. Ocular abnormalities including: cortical blindness and microphthalmia.

Treatment: Patient improved from her seizures by Sodium valporate 75 mg BID and Vigobartin 125 mg BID. Patient discharged to be followed by Pediatrician, Ophthalmologist and Dermatologist.

Discussion
Sebaceous nevi are present in approximately 0.3% of newborns and appear as waxy, verrucous plaque. Typically, there is yellow to orange hue that reflects hyperplasia of sebaceous glands.

Although the most common location is the head and neck, they can occur on the extremities as well as the trunk. The distribution of sebaceous nevi is along the lines of Blaschko, but this may be different to appreciate on the scalp, face or neck. For example, a forehead plaque that runs longitudinally along the midline is following Blaschko’s lines, as is a semicircular lesion that begins on the lateral cheek and ends the corner of the mouth.

In the sebaceous nevus syndrome (Schimmelpenning-Feuerstein-Mims Syndrome), patients have ocular, vascular, musculoskeletal, and central nervous system abnormalities in addition to sebaceous nevi.

The ocular manifestations can vary from corneal vascularization to microphthalmia. In addition, colobomas and Lipodermoid tumors of the conjunctivae or sclerae are often present. Examples of neurologic involvement include seizures, mental retardation, and intracranial hamartomas.

There is a trend for the sebaceous nevi to be more extensive in patients with this syndrome. However, in the original description by Feurstein and Mims, there...
was only a single midline plaque on the face in one patient. In 1969, Sugarman and Reed 15 described the association of sebaceous nevi with hypophosphatemic osteomalacia/rickets, this association has been confirmed more recently. 16-17 Although the histologic diagnosis of sebaceous nevus rests on the presence of an increased number of sebaceous glands in the dermis, additional clues include dermal apocrine sweat glands, absent or hypoplastic hair follicles, and papillomatosis of the epidermis. 2 However, if the patient is prepubescent or the nevus is located in areas such as the digits, there may not be hyperplasia of mature sebaceous glands. 18 For these reasons as well as the overlap in appearance with verrucous epidermal nevi, some authors prefer the term organoid nevus to describe both types of lesions. 4 The term organoid nevus phakomatosis has been introduced to reflect the similar systemic finding observed in the sebaceous nevus syndrome and the epidermal nevus syndrome as described by Solom et al. 19 When sebaceous nevi are limited in size, they can be excised; otherwise, longitudinal observation is indicated, given the possibility for development of basal cell carcinomas and rarely adnexal or squamous cell carcinoma. 20,21,22,23,24,25, 26,27

**Conclusion**

We report a rare case of sebaceous nevus syndrome in a Saudi infant patient, presented as extensive sebaceous nevus in the head, neck and chest associated with infantile spasms, intracranial hamartoma, cortical blindness and microphthalmia.

---

**References**