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

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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 sebacous 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 hamartoma.

Ocular abnormalities which occur in 30% of sebaceous nevus syndrome including involvement of the eyelid or conjunction 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 cynanosis, jaundice or lymphadenophathy. 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 scaring 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 Sebacous nevus syndrome (Schimmelpenning-Feuerstein-Mims syndrome). Laboratory Investigation were including Complete Blood Counts, Liver Function Tests, Urea & Creatinin, Electrolytes, 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 germs. The histopathological finding was consistant with nevus sebaceous. Patients was refereed to pediatric were diagnosed as infantile spasm, Her seizures were controlled by sodium valporate and vigobatrin. Infant was referred to ophthalmologist where diagnosed as left microphthalmia and cortial blindness.

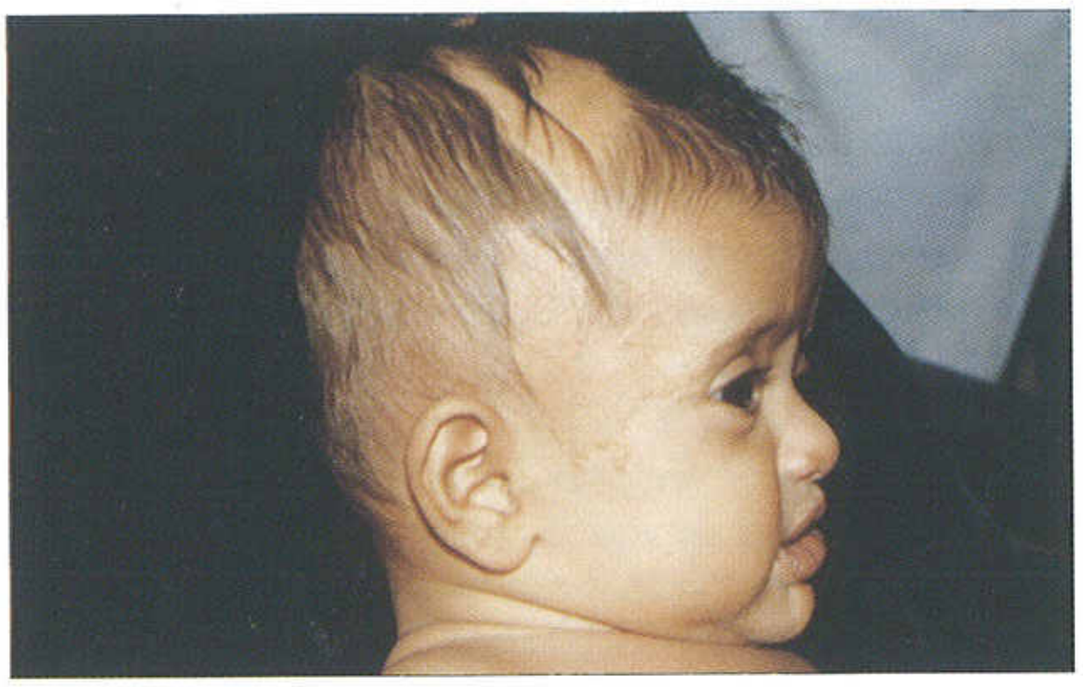

Fig. 1 Slightly elevated, yellowish, rough, circumscribed, plaques on the scalp and right temoral area, associated with scaring alopecia.

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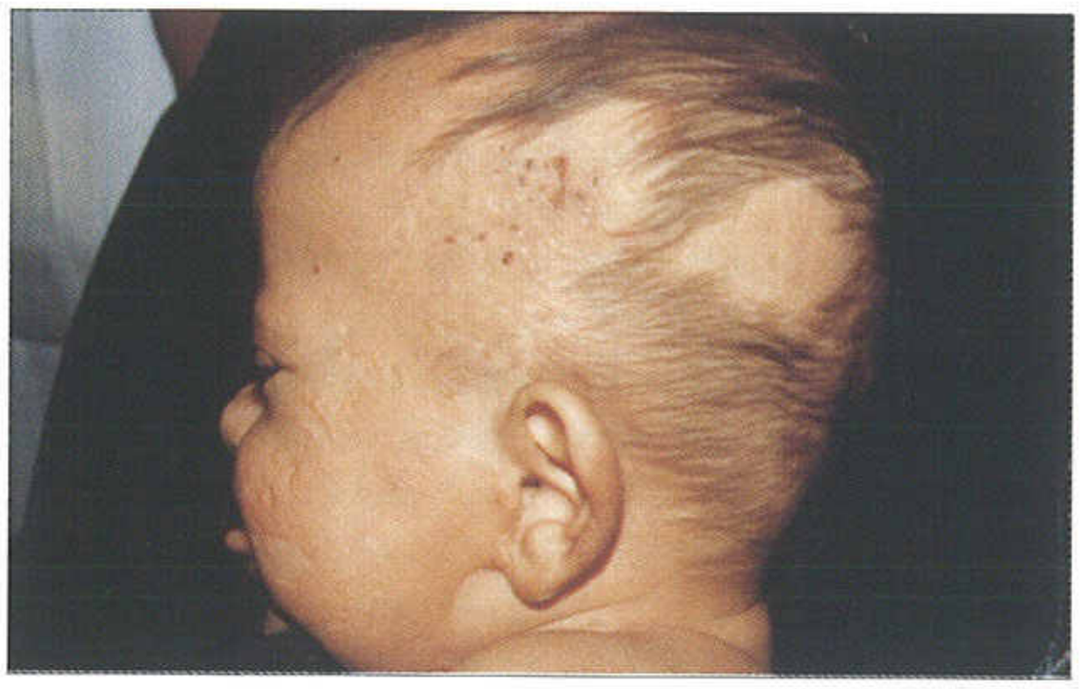

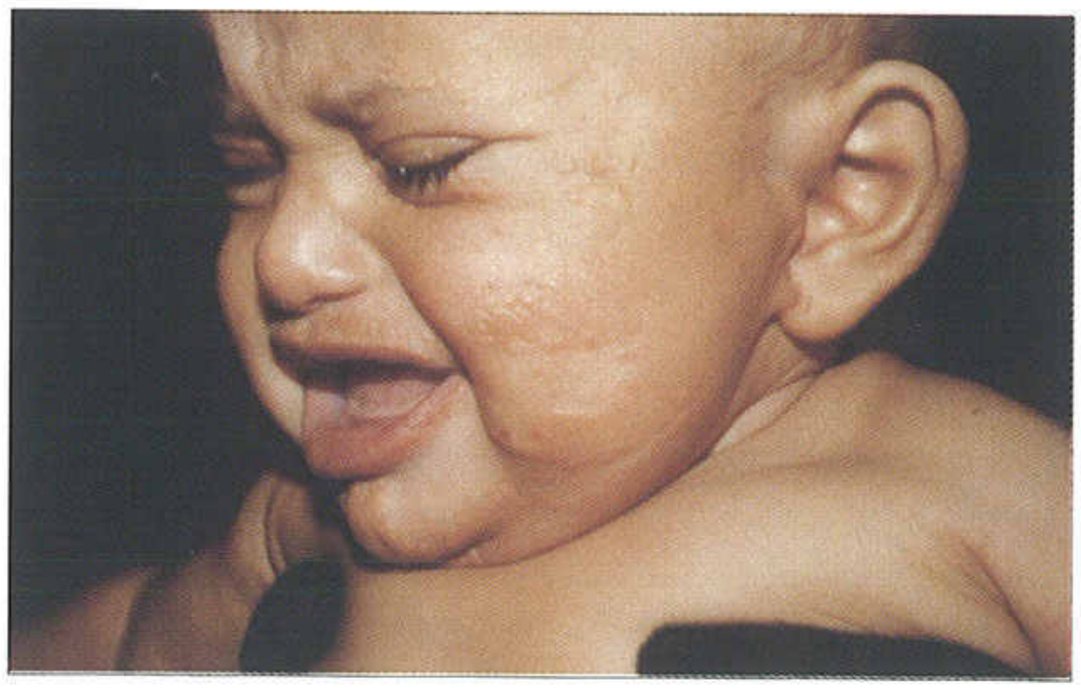

Fig. 2& 3 Slightly elevated, yellowish, rough, circumscribed, extensive plaques on the left temoral area and cheek. Also occipital area was involved.

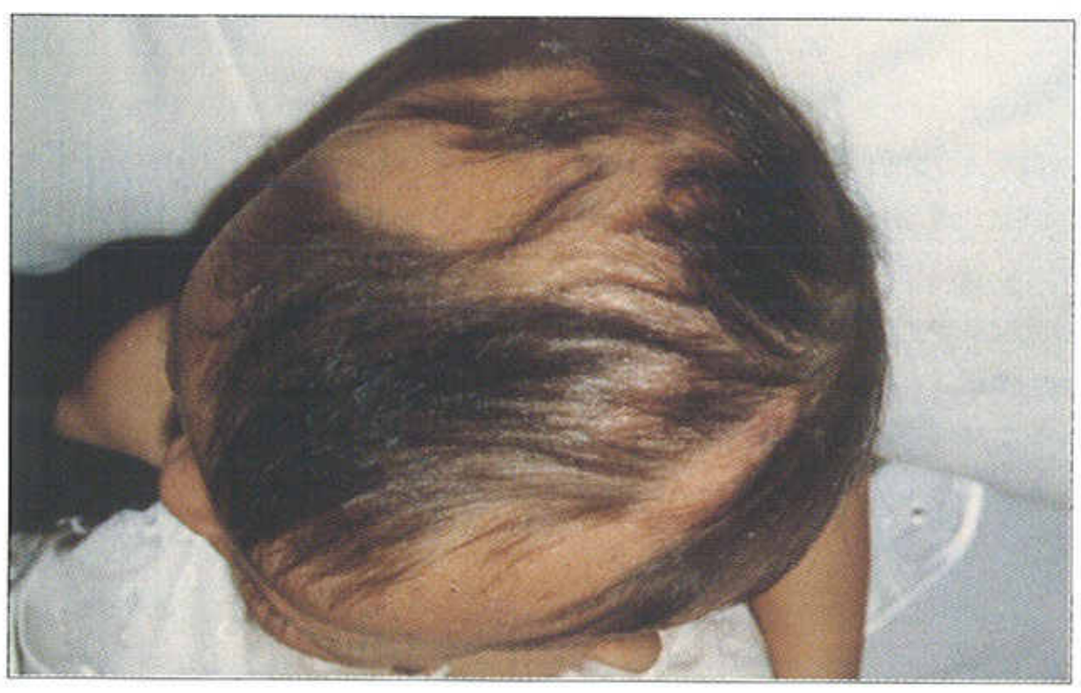

Fig. 4 Closed up view of skin lesions on the vertex. The scaring alopecia were cleared.

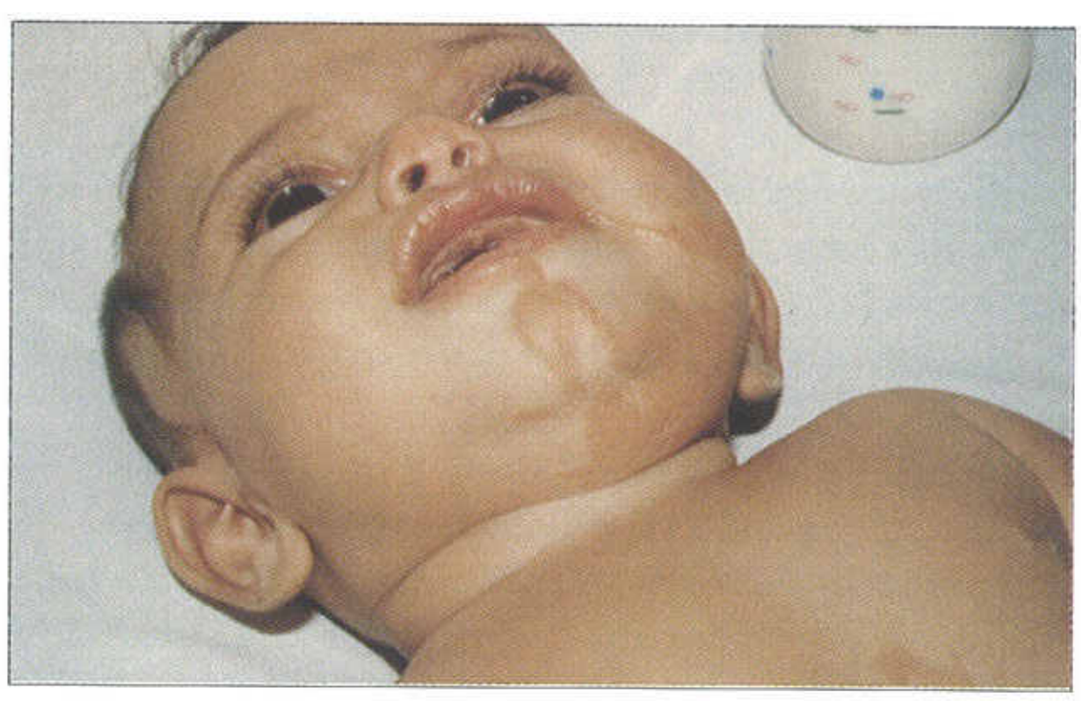

Fig. 5 Skin plaques on the chin and upper part of the chest.

The final diagnosis was Sebacous nevus syndrome with the following findings: Extensive nevus sebacous 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 Vigobatrin 125 mg BID. Patient discharged to be followed by Pediatrician, Ophthalmologist and Dermatologist

Discussion

Sebaceous nevi are present in approximately 0.3% of newborns 1 and appear as waxy to 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. [2] 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. [3,4,5]

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 microphalmia. In addition, colobomas and Lipodermoid tumors of the conjunctivae or sclerae are often present.[6] Examples of neurologic involvement include seizures, mental retardation, and intracranial hamrtomas. [7,8,9,10,11,12,13]

There is a trend for the sebaceous nevi to be more extensive in patients with this syndrome. [14] However, in the original description by Feurstein and Mims, there

was only a single midline plaque on the face in one patient. [6] 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

1. Alper J, Homes LB, Mihm MC Jr. Birthmarks with serious medical significance: Nevocellular veni, sebaceous nevi and multiple café au lait spots. J Pediatr 1979; 95: 696-700.

2. Mehregan AH, Pinkus H. Life history of organoid nevi: special reference to nevus sebaceus of Jadassohn. Arch Dermatol 1965; 91: 57-88.

3. Zaremba J, wislawski J, Bidzinski J et al. Jadassohn's naevus phakomatosis: 1. A report of two cases. J. Ment Defic Res 1978; 22: 91-102.

4. Hornstein OP, Knickenberg M. Zur Kenntnis des Schimmelpenning-Feurstein-Mims-Syndrome (organoide naevus-phakomatose). Arch Dermatol Forch 1974; 250: 33-50.

5. Lentz CL, Altman J, Mopper C. Nevus sebaceus of Jadassohn. Arch Dermatol 1968; 97: 294.

6. Diven DG, Solomon AR, MCNeely MC, et al. Nevus sebaceus associated with major ophthalmologic abnormalities. Arch Dermatol 1987; 123: 383-6.

7 Feurstein RC, Mims LC. Linear nevus sebaceus with convulsions and mental retardation. Am J Dis Child 1962; 104: 675-9.

8. Moskowitz R, Honig PJ. Nevus sebaceus in association with an intracranial mass. J. Am Acad Dermatol 1982; 6: 1078-80.

9. Prensky AL. Linear sebaceus nevus. In: Gomez MR, ed. Neurocutaneous diseases: a practical approach. Bosto: Butterworths, 1987: 335-44.

10. Marden PM, Venters HD. A new neurocutaneous syndrome. Am J Dis Child 1966; 112: 79.

11. Wausch Kuhn J, Rohde B. Systematisierte talydrusen-pigment – und epitheliale naevi mit neurologischer symptomatik: Feurstein-Mimassches neuroektodermales syndrome. Hautzt 1971; 22: 10.

12. Kuokkanen K, Koivikko M, Alavaikko M. Organoid nevus phakomatosis. Acta Derm Venereol (Stockh) 1980; 60-534.

13 Lantis S, Leyden J, Thew M et al. Nevus sebaceus of Judassohn: part of a new neurocutaneous syndrome. Arch Dermatol 1968; 98: 117.

14. Zaremba J. Judassohn's naevus phakomatosis: 2. A study based on a review of thirty-seven cases. J Ment Def Res 1978; 22: 103-23.

15. Sugarman GI, Reed WB. Two unusual neurocutaneous disorders with facial cutaneous signs. Arch Neurol 1969; 21: 242-7.

16. Caret DE, Drezner MK, Hamdan JA et al. Hypophosphatemic rickets/osteomalacia in linear sebaceus nevus syndrome: a variant of tumor – induced osteomalcia. J Pediatr 1986; 109: 994-1000

17. Skovby F, Svegaard E, Moller J. Hypophospjatemic rickets in linear sebaceus nevus sequence. J Pediatr 1987; 111: 855-7.

18. Solomon LM, Esterly NB. Epidermal and other congenital organoid nevi. Curr probl Pediatr 1975; 6: 1-56.

19. Solomon LM, Fretzin DF, Dewald RL. The epidermal nevus syndrome. Arch Dermatol 1968; 97: 273-85.

20. Domingo J, Helwing EB. Malignant neoplasms associated with nevus sebaceus of Judassohn. J AM ACAD Dermatol 1979; 1: 545-56.

21. Wilson Jones E, Heyl T, Naevus sebaceus. Br. J Dermatol 1970; 82-99

22. Bonvalet D, Barrandon V, Foix et. al. Tumeurs annexielles benignes de survenue tardive sur naevus verrucosebace (Jadassohn). Ann Dermatol Venerol 1983; 110-337.

23. Rahbair H, Mehregan AH. Development of proliferating triclemmal Cyst in organoid nevus. J Am Acad Dermatol 1986; 14: 123.

24. Brownstein MH, Shapiro L. The pilosebaceus tumors. Int J Dermatol 1977; 16: 340.

25. Morioka S. The natural history of nevus sebaceus. J Cutan Pathol 1985; 12: 200.

26. Allessi E, Wang SN, Advani HH et. al. Nevus sebaceus is associated with unusual neoplasms. Am J Dermatopathol 1988; 10: 116.

27. Tarkhan II, Domingo J. Metastasizing eccrine porocarcinoma developing in a sebaceus nevus of Jadassohn: Report of a case. Arch Dermatol 1985; 121: 413.