CASE REPORT

Schimmelpenning Syndrome: A Case Report and Literature Review

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We report a case of a male newborn with Schimmelpenning syndrome, which presented as diffuse sebaceous nevi covering the left side of the body, from the lower chin midface to the lower leg; cardiac–ocular comorbidities were also present. We present photographs of this patient’s sebaceous nevus, which may assist physicians in the early diagnosis of this condition and prevent unnecessary examinations and inadequate therapies.

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1. Introduction

Schimmelpenning syndrome is a congenital neurocutaneous disorder encompassing the cutaneous nevi (nevus sebaceous of Jadassohn) in conjunction with extracutaneous (neurologic, skeletal, cardiovascular, ophthalmic, and urologic) anomalies. Nevus sebaceous is a hallmark of Schimmelpenning syndrome, and if physicians are aware of this distinct skin lesion, an early diagnosis can be made. In a review of sebaceous nevus patients, the scalp was found to be the most common location (59.3%), followed by the face (32.6%). In contrast with reported cases, our patient presented with diffuse sebaceous nevi covering the left side of the body from the lower chin midface to the lower leg. We therefore report distinct skin lesions, which may assist physicians in the early diagnosis of this condition and prevent unnecessary examinations and inadequate therapies.

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Because of a great risk of multiorgan involvement, failure to recognize Schimmelpenning syndrome can result in failure to diagnose a potentially serious or life-threatening medical condition; hence, management of the syndrome using a multidisciplinary approach is mandatory.

2. Case Report

A male newborn was born to a G1P1 healthy mother via a normal spontaneous vaginal delivery at a gestational age of 40 weeks, with a birth weight of 3270 g. At birth, diffuse verrucous, granulated plaques were observed over the lower chin midface extending to the left side of the anterior chest, the upper limb, the thigh, and the lower leg (Figure 1). A hairless yellowy-red flat plaque was found over the patient’s scalp (Figure 2), and corneal opacity and microphthalmos were also noted. The findings of a general physical examination on admission were normal. Skin biopsies were taken from the left posterior auricle and left lower leg, and histopathological analysis showed epidermis hyperkeratosis, acanthosis, and papillomatosis (Figure 3). There was no evidence of malignancy. Ocular examination revealed microphthalmos, corneal vascularization, and epibulbar tumors. Cranial, abdominal, and renal ultrasonographs revealed no abnormalities, but cardiac ultrasonography revealed coarctation of the aorta and a septal secundum defect. A chromosome study revealed a normal male karyotype. After the necessary examinations were completed, the family requested transfer to another hospital. The patient was then discharged from that hospital at 4 days of age.

After discharge, he was lost to follow-up. He had visited a well-baby clinic for vaccination. According to his mother’s statements, he had visual impairment due to retinoblastoma and had received chemotherapy for retinoblastoma. There was no seizure history. He had developmental delay and was enrolled in an early intervention program.

3. Discussion

Epidermal nevus syndromes are a group of distinct disorders. There are nine epidermal nevus syndromes well defined by clinical, histopathologic, and genetic criteria.2 Schimmelpenning syndrome is the best-known epidermal nevus syndrome. Several names are interchangeable for this condition, such as linear sebaceous nevus syndrome, Solomon syndrome, Jadassohn nevus phacomatosis, organoid nevus phacomatosis, organoid nevus syndrome, sebaceous nevus syndrome, and Jadassohn sebaceous nevus syndrome.

In 1957, Gustav Schimmelpenning first detailed epidermal nevi with neurologic anomalies.3 Thereafter, Feuerstein and Mims4 reported a case of linear nevus, epilepsy, and mental retardation in 1962. Since then, the classic triad of linear nevus sebaceous syndrome (sebaceous nevus, seizures, and mental retardation) has been widely used. Since the original identification, several cases have been reported. A number of studies have revealed that the associated anomalies of the syndrome extend widely beyond the initial triad; hence, the diagnostic criteria of the triad have been abandoned.5

Van de Warrenburg et al5 reviewed the literature in 1998, and neurologic findings were noted in 66% of cases. The major neurologic abnormalities include mental retardation, seizures, and hemiparesis. Lovejoy and Boyle6 found that epilepsy occurs in up to 67% of cases, with seizures typically beginning during the 1st year of life. Structural abnormality of the cerebrum or cranium is present in 72% of cases.6 The most common abnormalities are hemimegalencephaly and ipsilateral gyral malformations.7 A correlation between epidermal nevi lesions and the incidence of central nervous system abnormalities has been reported. Menascu and Donner8 reported two cases of
linear nevus sebaceous syndrome with intractable seizure as the initial presentation at 3 and 4 months of life.

Skin lesions were noted upon admission. In our present case, nevus sebaceous was present from birth. Wu et al also reported a rare case of aplasia cutis congenita with refractory seizures. The ulcerative-like wounds identified after birth measured 2 and 3 cm in diameter, and were located on the scalp vertex just lateral to the midline. Two weeks later, these lesions became asymptomatic dome-shaped indurated plaques. Cranial magnetic resonance imaging, however, revealed two pieces that lacked ossification over the occipital bone. After consultation with a dermatologist, this was diagnosed clinically as aplasia cutis congenita.9

Ophthalmological abnormalities are present in 59% of cases. The major ocular abnormalities are colobomas and choristomas.10 Microphthalmos, corneal vascularization, and epibulbar tumors were found in our case.

Hypophosphatemic rickets has been reported.11 Cardiovascular abnormalities include patent ductus arteriosus, patent foramen ovale, ventricular septal defect, coarctation of the aorta, hypoplasia of the aorta, arterial flutter/fibrillation, and arterial premature systoles.12

Urinary system issues include horseshoe kidney and a duplicated urinary collection system, and undescended testes and an enlarged clitoris have also been reported.13

Nevus sebaceous is a hallmark of Schimmelpenning syndrome; it is a hamartoma with epidermal, follicular, sebaceous, and apocrine elements, and occurs in approximately 0.3% of neonates.1 In two reviews of sebaceous nevus patients, of those with parietal lesions (42%), the scalp was the most common location (59.3%), followed by the face (32.6%); lesions have also been identified in the preauricular area (3.8%), on the neck (3.2%), and in locations away from the head and neck (1.3%).1 In contrast with reported cases, our patient presented with diffuse sebaceous nevi covering the left side of the body from the lower chin midface to the lower leg.

Upon development of secondary malignant neoplasms, surgical excision is the treatment of choice and has traditionally been recommended, but according to present knowledge, there is no need to remove the sebaceous nevus as a prophylactic measure, because the rate of such malignancies is now known to be lower than previously estimated. Given the low risk of malignant transformation, especially in children, clinical follow-up is considered to be a safe alternative to prophylactic excision in children, and if one or more true basal cell carcinomas should develop later, these tumors can easily be removed.13

Whether the brain should be examined by imaging techniques in all children with large sebaceous nevi is a controversial issue. Happle4 suggested that if there are no clinical signs of central nervous system involvement, a “wait and see” attitude is preferable.

The cause of Schimmelpenning syndrome is still unknown, but it has been theorized to result from genomic mosaicism. In a recent study, Carlson et al14 found human papillomavirus (HPV) DNA integration in skin taken from the sebaceous nevi of Jadassohn patients, and the results of that study suggested that maternal transmission of HPV DNA to fetal ectodermal stem cells could result in epigenomic mosaicism and altered skin development. Whether the maternal HPV DNA plays an epigenetic mosaic role and then alters fetal skin development is currently unknown, and further research is necessary.

The clinical outcome of Schimmelpenning syndrome is usually not fulminant; however, Mollica et al15 presented a case of a newborn baby who died 36 hours postpartum. Schimmelpenning syndrome is a multisystem neurocutaneous syndrome in which epidermal nevi may be associated with neurological, skeletal, ocular, and other cutaneous anomalies. Nevus sebaceous is a diagnostic marker of Schimmelpenning syndrome. Awareness of this distinct skin lesion may help physicians to make an early diagnosis and prevent unnecessary examinations. In addition, Schimmelpenning syndrome is associated with a wide range of abnormalities, and careful evaluation of systems with the potential to be involved is necessary.

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**References**