Schimmelpenning Syndrome: A Neuro-Oculo-Cutaneous Disorder

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A 1.5-month-old baby girl with patchy hair loss over the scalp and yellow-to-black raised lesions over the face was brought in by the mother. The lesions were asymptomatic and constant in size since birth without any history of aggravation or regression. The baby was born following an uneventful full-term pregnancy with no perinatal insults. Cutaneous examination revealed six pale-to-hyperpigmented plaques over the left side of the scalp and face in a Blaschko-linear pattern. Ocular examination revealed a pedunculated mass and symblepharon at the left lateral canthus of the left eye and reacting pupils [Figure 1 and 2]. She had no abnormal neurological, cardiac, or per-abdomen clinical findings. She then presented 1.5 years later with delayed milestones, a developmental quotient of 27% and profound hearing loss in the left ear till 90 dB that was subsequently surfaced on conducting brainstem-evoked response audiometry (BERA). Schimmelpenning-Feuerstein-Mims syndrome is an epidermal nevus syndrome that characteristically presents with nevus sebaceous and cerebral, ocular, and skeletal defects, though other genitourinary, cardiovascular, endocrine, and dental abnormalities have been documented as well.[1] Clinically, most epidermal nevi are present at birth.[2] However, neurological symptoms may only develop over a period of time. There is currently no curative treatment

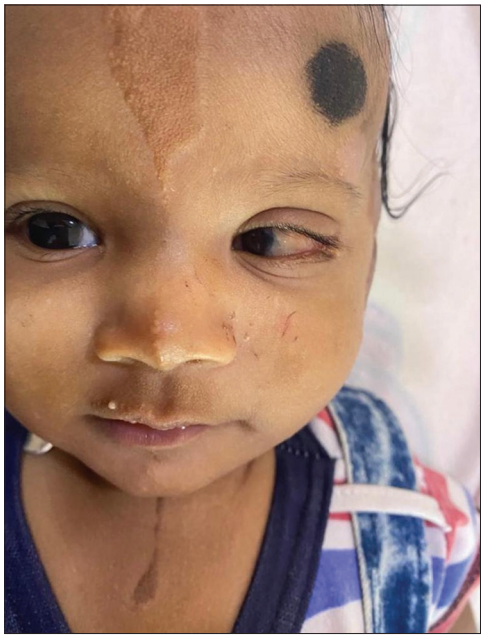

Figure 1: A linear, sharply demarcated, yellowish plaque with multiple papillomatous and verrucous areas over the forehead, tip of the nose, philtrum, and the chest. Left bulbar conjunctiva shows a well-defined, circular, pedunculated mass of size 2 × 1 cm and symblepharon at the lateral canthus

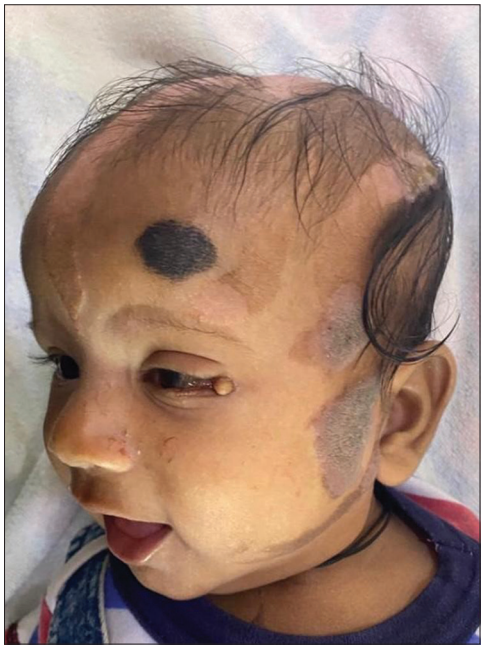

Figure 2: Multiple sharply demarcated, pale-to-hyperpigmented papillomatous plaques present in a Blaschko-linear pattern on the left side of the face and scalp and periocular nodule on the left lateral canthus

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for Schimmelpenning syndrome, but an inter-disciplinary approach coupled with careful evaluation of systems that have the potential to be involved is necessary. The skin lesion can be treated with emollients, topical salicylic acid, retinoids, vitamin D analog, laser, photodynamic therapy, shave dermabrasion, cryotherapy, or surgery for cosmetic reasons.[2] Similarly, the treatment of ocular lesion can range from simple excision, amniotic membrane transplantation, lamellar keratoplasty, and penetrating keratoplasty to autologous limbal stem cell allograft along with eyelid reconstruction, tailored according to the residual eyelid defect.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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