Case report

SCALP syndrome: What is it and its ophthalmic manifestations

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1. Introduction

SCALP syndrome is a rare neuro-cutaneous condition which is characterized by the presence of Sebaceous nevus, Central nervous system (CNS) malformations, Aplasia cutis congenita, Limbal dermoid and Pigmented (giant melanocytic) nevus. Whilst sharing several common features with sebaceous naevus syndrome (SNS), it represents a distinct clinical entity with a range of cutaneous and extra-cutaneous manifestations involving the eyes, brain and skeletal system. A genetic aetiology has been proposed however this has not been formally confirmed. To date, only 4 cases of SCALP syndrome have been reported, and they were all discussed in the dermatological literature. We present a case of a 3-month old girl with SCALP syndrome with multiple ocular anomalies which have not been previously described.

2. Case report

A 3 month old girl with infantile seizures and global developmental delay was referred for opinion regarding her eyes. This girl was the first child born to non-consanguineous parents. Perinatal history was complicated by prematurity at 35 weeks gestation and intrauterine growth retardation.

On examination, she was fixing and following with her right eye only. The left eye demonstrated no visual response. A large angle esotropia was present (Fig. 1A). She was not able to abduct either eye. There were bilateral large temporal limbal dermoids (Fig. 1B and C). The cornea adjacent to the dermoids showed evidence of exposure keratopathy secondary to poor tear film coverage. Anterior segment was normal. Fundus examination revealed bilateral optic nerve pallor with peri-papillary atrophy and a small optic disc on the left.

Systemic examination revealed numerous cutaneous abnormalities. Giant melanocytic naevi were seen across the left side of her scalp, face, neck, limbs and torso (Fig. 2). A linear sebaceous naevus was present across her left scalp was consistent with a region of aplasia cutis congenita (Fig. 3). Magnetic Resonance Imaging (MRI) showed the presence of a hypoplastic left optic nerve and mild hypoplasia of the optic chiasm (Fig. 4A). There was polymicrogyria affecting the posterior parietal lobe and tempo-occipital cortex (Fig. 4B). Extensive abnormality of bilateral hemispheric white matter and dilatation of the right lateral ventricle was noted with numerous prominent perivascular spaces secondary to parenchymal loss.

At the age of 12 months old, surgical debulking of the limbal lesions was performed under general anesthesia, with excision extending temporally into the superficial orbit. Histopathology demonstrated the presence of limbal dermoids with dense fibrous tissue lined by non-keratinising stratified squamous epithelium. In addition, subconjunctival lymphoid follicles, small amounts of mature adipose tissue, islands of lacrimal tissue, sebaceous glands, multiple thick walled vessels and occasional nerves were visualized. Molecular genetic studies performed on skin biopsy of the melanocytic naevi of the scalp detected the presence of somatic mosaic NRAS mutation.

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ABSTRACT

Purpose: To present the ophthalmic manifestations of a 3-month old female with SCALP syndrome.

Observations: The patient presented with multiple ocular anomalies including bilateral limbal dermoids, esotropia and left optic nerve hypoplasia.

Conclusions: We describe systemic and ocular anomalies in a rare case of SCALP syndrome. This report provides additional information on the ocular anomalies not previously described that may be associated with this clinical entity.
In this report, we describe a case of SCALP syndrome with numerous ocular anomalies. To our knowledge, it is the fifth pediatric case of SCALP syndrome reported in the literature and the first description of multiple ophthalmic anomalies associated with the syndrome.

From a dermatological perspective, our patient has multiple sebaceous naevi, a region of aplasia cutis congenita and pigmented giant melanocytic naevi distributed across the left scalp, neck and torso.
melanocytic) naevi.

Sebaceous naevi are hamartomatous naevi with aggregations of sebaceous glands and hair follicles most commonly distributed on the head and neck. These lesions have malignant potential, most often to basal cell carcinomas. Aplasia cutis congenita is a rare disorder characterized by discrete areas of absent skin and less commonly subcutaneous tissue with which the scalp is typically affected. Giant congenital melanocytic naevi like those described in our case are typically distributed over the back and thighs and like sebaceous naevi they possess malignant potential resulting in melanomas.

The detection of mosaic NRAS gene mutation explains the development of giant congenital melanocytic nevus in this syndrome. This mutation is localized to chromosome 1p13.2 and results in a GTPase proto-oncogene product implicated in other rasopathies and variants of epidermal nevus syndrome. This gene is likely to be implicated in other cases of SCALP syndrome, although further studies utilizing modern molecular diagnostic techniques will be required to confirm this finding.

CNS involvement including neurocutaneous melanosis and brain malformations resulting in seizures and developmental delay have also been described in SCALP syndrome. Our patient suffers from epilepsy and has developmental issues.

In addition to the previously described ophthalmic feature of limbal dermoid in SCALP syndrome, our report is the first to present numerous additional ophthalmic associations other than dermoids. These include strabismus, exposure keratopathy secondary to poor tear film coverage and optic nerve and chiasmal hypoplasia. Mimouni et al. gave brief mention to the surgical management of limbal dermoids in SCALP syndrome. The surgical debulking of the limbal dermoids in our patient posed a particular challenge. Due to the size and depth of the lesions, excision was extended temporally into the superficial orbit. Following debulking, both eyes were looking straight, thus eliminating the need for additional strabismus surgery. The corneas were also better covered by a healthier tear film.

4. Conclusion

SCALP syndrome is a rare clinical entity. This report describes the features of this condition, and serves as a reminder that this condition needs multidisciplinary management because of the various clinical problems and potential malignant changes. Malignant complications of SCALP syndrome can be life-threatening with poor outcomes resulting from missed or delayed diagnosis. Timely management of ophthalmic conditions such as strabismus and ocular surface issues is imperative to achieve the best possible long-term visual outcome. It is important that ophthalmologists are aware of this syndrome as they may be the first medical contact patients have.

Patient consent

The patient's legal guardian provided written consent to publish details and photographs of this case.

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