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뇌두개피부지방종증 치료에 대한 미용적 고찰

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Aesthetic Consideration of Management in Encephalocutaneous Lipomatosis: How we do it

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Encephalocraniocutaneous lipomatosis (ECCL) is a rare congenital neurocutaneous syndrome that was characterized by unilateral, smooth, hairless fatty tissue nevi of the scalp, termed nevus psiloliparus, facial lesions, multiple anomalies involving the eye, and ipsilateral porencephalic cysts with cortical atrophy, cranial asymmetry, marked developmental delays, and mental retardation. A 12-month-old boy was referred to our clinic for evaluation of non-scarring alopecia on the left side with an underlying fatty mass in the left parietal scalp and left-sided multiple periocular masses. It showed a large lipomatous mass on the scalp with overlying alopecia. Multiple skin tags and defects in the left periocular area were also observed. Additional ocular anomalies included epibulbar lipodermoid, iris coloboma, and localized peripapillary hypopigmentation lesions. After complete excision, the wound was covered with a local flap. The histologic examination revealed a mass surrounded by a well-developed capsule within the dermal layer, adipose tissue and connective tissue septa extending into the reticular dermis. No recurrence was observed at follow-up. It is essential to differentiate suspected ECCL from other syndromes which present with epibulbar chorisotomas. Neuroimaging, and pathological studies may be helpful for correct diagnosis. We will correct multiple periocular lesions in preschool age and follow up developmental problems like developmental delay and mental retardation constantly.

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Key Words: Encephalocraniocutaneous lipomatosis, Neurocutaneous syndrome

I. INTRODUCTION

Encephalocraniocutaneous lipomatosis (ECCL) is rare neurocutaneous syndrome which involves the development of lipomatosis on the unilateral face or scalp. ECCL was first described by Haberland and Perou in 1970.[1] When Fishman et al.[2] reported a case of ECCL in 1978, the syndrome was called "Fishman syndrome" or "Haberland syndrome". Due to the lack of reported cases, ECCL is thought to be too rare to estimate the frequency of occurrence. Happle described ECCL as a mosaicism caused by mutant autosomal lethal genes, while Legius et al. suggested an association with mutations in neurofibromatosis type 1 (NF1) genes. Important diagnostic criteria include hairless fatty tissue nevus psiloliparus (NP) of the scalp and eyeball choristoma. Major symptoms include non-scarring lipomatosis accompanying alopecia, ocular choristomas, and multiple anomalies in the eye causing impaired vision.[3] In addition, leptomeningeal angiomatosis or porencephalic cysts may develop in the central nervous system (CNS) resulting in

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mental retardation, epilepsy, and accordingly, developmental delays. Differentiating ECCL from neurocutaneous syndromes, such as oculocerebrocutaneous syndrome (OCC), Goldenhar syndrome, and sebaceous nevus syndrome, which all include symptoms of CNS dysfunction, skin polyps, and masses appearing as lipomatoses, is required. For diagnosis, imaging and pathological examination on the skin tissues will be helpful. This article presents clinical manifestations and discussion about encephalocraniocutaneous lipomatosis.

II. CASE

A 12-month-old boy was referred to our clinic with the chief complaint of multiple periocular masses on the left side and an underlying fatty mass in the left parietal scalp (Fig. 1). The patient is a first child who was born through normal delivery after 41 weeks of gestation, weighing 3,300 g at birth. The child's mother had no definitive medical history. No developmental disorders were observed, but multiple periocular masses around the left eyebrow area and an approximately 3.5 × 2.5 cm-sized hairless mass on the left parietal scalp were observed (Fig. 2). Blepharocoloboma at the left lid margin and medial canthus (Fig. 3) and a cloudy epibulbar lesion invading into the upper corneal limbus were observed. Through funduscopic examination, peripapillary hypopigmentation was observed around the optic disc area. The cup/disc ratio in the left eye was 0.8, which is higher than normal. No definitive findings of pediatric neurological symptoms with ultrasonographic and brain MRI were observed.

Polyps (skin tags) of the left lid margin and medial canthal area were excised, and complete resection and histological examination for the mass in the left parietal scalp were performed (Fig. 4).

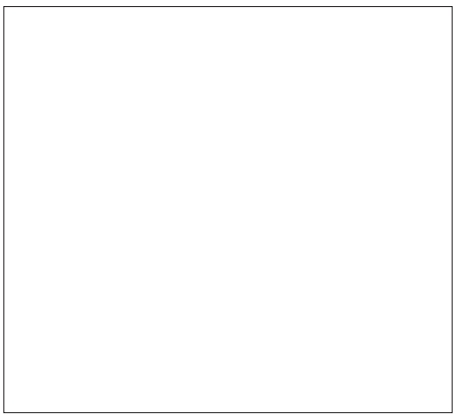

Fig. 1. Preoperative finding: Large lipomatous lesion with overlying alopecia on the left parietal scalp.

According to the pathological findings, masses of the medial canthus were the same as lipomatosis (fibrolipoma), which was a thick dermal layer of fatty tissues surrounded by fibrous tissues, and the scalp mass was also diagnosed as lipomatosis (lipoma; Fig. 5). The patient was discharged 12 days after surgery without definitive complications, and pediatric and ophthalmological follow-up care has continued for 12 months using MRI, neurological examinations, and funduscopic examinations. No definitive finding or recurrence has been observed.

III. DISCUSSION

Encephalocraniocutaneous lipomatosis was first described in 1970 with the results of a post-mortem autopsy by pathologists Haberland and Peoru, who reported a case of a child with alopecia and multiple masses in the unilateral head and neck and facial area with accompanying symptoms of mental retardation and epilepsy. Upon conducting the autopsy, lipomatosis was found in the face, scalp, heart, and central nervous system,

Fig. 2. There are multiple cutaneous masses located at the left eyebrow and medial canthal area.

Fig. 3. Blepharocoloboma of the left upper eyelid. Epibulbar dermoid on the superior limbal border.

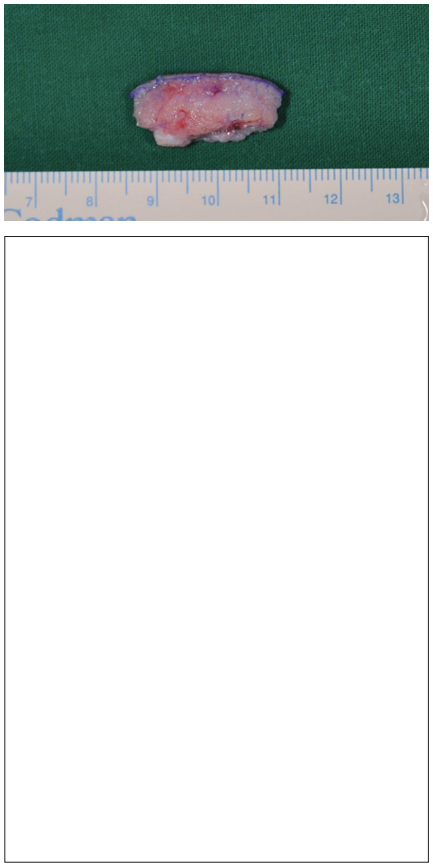

Fig. 4. Intraoperative excision & postoperative finding: Excised mass from the scalp contains a 2.5×1.5 cm sized lipoma. It had a well demarcated margin on the cutting section.

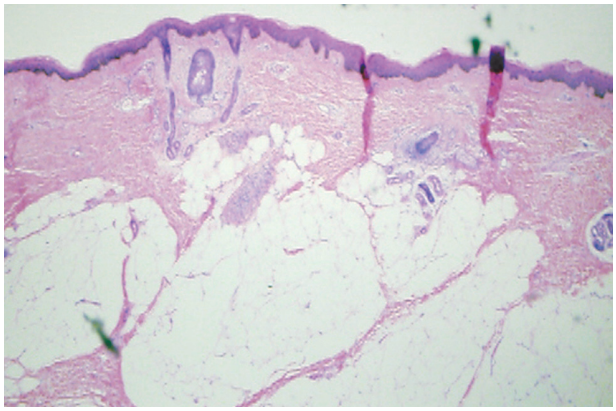

Fig. 5. Adipose tissue and connective tissue septa extending into the reticular dermis obscures the normal architecture of the reticular dermis and subcutaneous junction (H & E, ×40).

and cranial hyperostosis, excessive fatty tissues located between cortical bones, cerebral anomalies and calcification, and leptomeningeal angiomatosis were also observed. In the eyeball, lateral lipodermoid invaded into the cornea like a pterygium and included cartilage and glandular tissue. In addition, skin polyps, which are hamartomas composed of fatty and fibrous tissues, were found on the eyelid and surrounding area.

Non-scarring alopecia is the most typical symptom of ECCL. A mass composed of fatty tissues or subcutaneous fat can be the pathology of ECCL. A nevus that contains soft and hairless fatty tissues developing on the scalp is called nevus psiloliparus (NP) and is observed in almost all patients as the most typical skin symptom. Most NP cases develop unilaterally without crossing over the midline, but cases of crossing over have been reported.[3] According to these reported cases, lipomatosis can develop not only on the skin of the head and neck but in the heart (Haberland),[1] legs and torso (Al-Mefty),[4] lumbosacral area (Gokhale),[5] and suprapubic area (Chittenden). The skin covering the mass is always hairless.

In addition, findings in the CNS, such as leptomeningeal angiomatosis, porencephalic cysts, arachnoid cysts, and microencephalia, were reported through postmortem histological and radiological exams. These tumors or cysts may cause hypofunction of the CNS or atrophy of surrounding nerve tissues, which can result in severe or mild mental retardation and developmental delays in addition to dysphasia, spastic tetraplegia, and epilepsy.[3]

In the cranial bone, excessive fatty tissues proliferated among the bony trabeculae, and the cortical bone surrounding the fatty tissues became thin. Hyperosteosis may also develop in some other areas. Since the head and neck pathology of lipomatosis develops unilaterally, cranial asymmetry is observed.

Ocular anomalies are very diverse and found in all ECCL patients. The most common ocular anomalies include ipsilateral choristomas and skin tags developing on the eyelid and surrounding area on the same side as the scalp pathology. Choristomas are mostly dermoid or lipodermoid tumors developing on the surface of the eyeball and the accessory organs of the eye. Each skin polyp is about 5 mm in diameter, is unilaterally present on the eyelid and surrounding skin, and is histologically determined to be lipomatosis, lipomatosis, or angiofibroma.

The etiological mechanism of ECCL is not clearly understood. Happle suggested a hypothesis of mosaicism caused by mutations in autosomal lethal genes.[7] Mosaicism refers to the coexistence of normal cells and those with mutated lethal genes which should have been eliminated in the fetus during the gestation period but are still alive. The mutated genes are not transferred to the next generation because the genes are present on autosomal chromosomes. Legius et al. found mutated neurofibromatosis type 1 (NF1) genes in a 2-year-old male patient but could not clarify the relationship between the mutated genes and ECCL.[8]

Differential diseases include oculocerebrocutaneous syndrome (OCC), which shows epibulbar choristoma and sys-

temic symptoms, Goldenhar syndrome, and sebaceous nevus syndrome.

OCC displays neurocutaneous syndrome accompanying ocular, neural, and skin anomalies, which are similar to ECCL findings in terms of ocular anomalies, skin polyps, and cranial anomalies. However, OCC is different from ECCL because orbital cysts and agenesis of the corpus callosum are found only in the case of OCC with a definitive pathology of local dermal hypoplasia. By comparison, cerebral calcification is found only in the case of ECCL with a definitive pathology of lipomatosis in the head, neck and CNS; accordingly, CT and MRI are very useful methods for differential diagnosis.

Since first described in 1952, Goldenhar syndrome has been known to include ocular pathologies such as epibulbar choristoma and ocular motor dysfunction, in addition to the frequent development of systemic symptoms, such as preauricular skin tags, pretragal fissures, microtia, cleft lip, spinal anomalies, digital anomalies, urogenital organ anomalies, congenital heart anomalies, and hemifacial microsomia.

Sebaceous nevus syndrome, also called linear sebaceous nevus syndrome, shows frequent development of choristomas invading into the corneal limbus of both eyes. Epilepsy, mental retardation, and linear sebaceous nevi are the 3 major symptoms, and CNS anomalies, such as ventricular enlargement, spinal lipomatosis, cerebral cortex calcification, and bilateral choristomas, are common findings of both sebaceous nevus syndrome and ECCL. Skin pathology is important for differential diagnosis. In cases of ECCL, scalp symptoms never cross over the midline except the case of bilateral ECCL, in which symptoms developing on both sides of the scalp meet at the midline. In bilateral ECCL, linear sebaceous nevus without hair loss is a definitive manifestation. Since encephalocraniocutaneous lipomatosis rarely has bilateral head and neck skin manifestations, cases of bilateral linear sebaceous nevus without hair loss can be regarded as sebaceous nevus syndrome. We think that surgical correction for blepharocoloboma of the left upper eyelid and epibulbar dermoid on the superior limbal border should be done in appropriate moment.

In pediatric patients, attention should be given to neurological symptoms which may commonly develop due to eyeball anomalies, including amblyopia, and to effects from the CNS. The patient of the present case did not show definitive findings in the radiological and neurological examinations other than the ocular pathology, but appropriate ophthalmological, pediatric, and neurological treatments and follow-up care are necessary.

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