Encephalocraniocutaneous Lipomatosis without Neurologic Anomalies

Dae-Hun Kim, M.D., Seung-Bae Park, M.D., Young Lee, M.D., Myung Im, M.D., Young-Joon Seo, M.D., Si-Hwan Choi, M.D.¹, Jeung-Hoon Lee, M.D.

Departments of Dermatology and ¹Ophthalmology, School of Medicine, Chungnam National University, Daejeon, Korea

Dear Editor:

Encephalocraniocutaneous lipomatosis (ECCL) is a rare, sporadically occurring congenital neurocutaneous syndrome that characteristically involves ectomesodermal tissues, such as the skin, eyes, and central nervous system (CNS). Since it was first described by Haberland and Perou¹ in 1970, the majority of new cases of ectomesodermal dysgenesis have been reported using the term ECCL.

A 1-month-old Asian male was referred for dermatologic evaluation of patchy alopecia located in the frontotem-

poral region. The hairless patch had been noted at birth. Physical examination revealed a soft, somewhat elevated, area of patchy hair loss in the frontotemporal region (Fig. 1a). A biopsy of the hairless scalp revealed diffuse loss of hair follicles and thickening of the subcutaneous fatty tissue with lobular proliferation (Fig. 2A). The clinical and histologic findings of the scalp lesion were consistent with nevus psiloliparus, which is considered a hallmark of ECCL. No sign of scalp inflammation or scarring was found. Additionally, some papular lesions had been present on the forehead and periorcular area since birth

Fig. 1. (a) Patchy hair loss in the frontotemporal area. (b) Erythematous papular lesions were seen on the forehead and periorcular area. (c) Epibulbar choristoma on the left eyeball. (d) Eyelid coloboma.

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Corresponding author: Jeung-Hoon Lee, M.D., Department of Dermatology, Chungnam National University Hospital, Munhwaro 282, Jung-gu, Daejeon 301-721, Korea. Tel: 82-42-280-7706, Fax: 82-42-280-8459, E-mail: jhoon@cnu.ac.kr

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Fig. 2. (A) Biopsy specimen from the scalp showed no significant dermal, perifollicular, or subcutaneous fibrosis. The thickness of the subcutaneous adipose tissue was increased with lobular proliferation of mature subcutaneous fatty tissue and fewer hair follicles (H&E $\times 40$). (B) Magnetic resonance imaging reveals an oval image suggestive of a lipoma, about 2 cm sized, in left parietal paramedian area of the scalp (arrow). No abnormal signal intensity lesion in the brain parenchyme.

(Fig. 1b) and eyelid coloboma and epibulbar choristoma on left eye were observed during an ophthalmologic investigation (Fig. 1c, d). The constellation of findings met the criteria for ECCL². A neurological examination including brain magnetic resonance imaging performed to identify any neurologic anomalies was normal (Fig. 2B). The electrocardiogram and echocardiography showed no evidence of congenital heart defects.

ECCL is a sporadically occurring neurocutaneous disorder of unknown etiology. The hypothesized genetic mechanism involves lethal autosomal dominant genes that survive by mosaicism, and the pathogenesis most likely involves dysgenesis of the cephalic neural crest and anterior neural tube³.

Clinically, ECCL is characterized by unilateral cutaneous and ophthalmologic lesions with CNS anomalies and some authors have proposed diagnostic criteria². The skin anomalies associated with ECCL include small nodular skin tags in the periocular area or on the eyelid, subcutaneous lipomas of the scalp, and patchy or streaky nonscarring alopecia with or without underlying fatty tissue. The hairless fatty tissue nevus of the scalp is called a nevus psiloliparus, and was once regarded as pathognomic of ECCL. However, nevus psiloliparus with no other features associated with ECCL has also been reported as an independent non-syndromic skin disorder⁴.

Although a few reports have showed ECCL without CNS anomalies⁵, various CNS anomalies have been detected in ECCL, including hemiatrophy, dilated ventricles, porencephalic cysts, abnormal calcifications, intracranial lipomas, and cranial asymmetry². The degree of the intracranial anomalies with ECCL does not correspond with the extent of associated skin and eye findings. There may be accompanying mental retardation or epilepsy. The neurodevelopmental spectrum is broad, from normal development to severe retardation². Because mild retardation or severe retardation are seen in roughly two-thirds of ECCL patients in developmental data², long term follow-up is needed to detect further abnormal neurodevelopment.

When confronted with congenital hairless lesions of the scalp, clinicians must consider the diagnosis of nevus psiloliparus and keep in mind its association with ECCL. They need to investigate other possible anomalies associated with ECCL using a multidisciplinary evaluation and follow-up by a neurosurgeon, pediatrician, ophthalmologist, and plastic surgeon.

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Multiple Bowen's Disease: In a Veteran Who Participated in Vietnam War

Na Hyun Kwon, M.D., Bo In Lee, M.D., Hei Sung Kim, M.D., Young Min Park, M.D.,
Hyung Ok Kim, M.D., Jun Young Lee, M.D.

Department of Dermatology, Seoul St. Mary's Hospital, College of Medicine, The Catholic University of Korea, Seoul, Korea

Dear Editor:

Many etiological factors have been suggested for Bowen's Disease (BD). However, the relationship between tetrachlorodibenzo-p-dioxin (TCDD) and BD has not yet been clarified. We report here on a case detailing TCDD as a possible cause of multiple BD.

A 63-year-old-man presented with multiple erythematous and scaly plaques over the whole body, which had first appeared over 10 years prior (Fig. 1). He had a history of contact with defoliants while he participated in the Vietnam War (2 years), 40 years ago and was treated for peripheral neuropathies that were possibly related to Agent Orange. The patient was otherwise in good health. The physical examination revealed six scaly erythematous papules and plaques on his back, arm, leg, and finger. According to the patient the lesions occurred spontaneously. Skin biopsies from the back and finger were performed. Histologic examination showed full thickness epidermal atypia with adnexal involvement (Fig. 2). The clinical appearance of the skin lesion and the microscopic descriptions were consistent with a diagnosis of multiple

BD.

TCDD is a kind of dioxin isomer, and TCDD is contained in Agent Orange, which was used as a defoliant during the Vietnam War from 1961 to 1970. TCDD has a half-life of approximately 8 years in humans and it accumulates primarily in fatty tissues over time, so even small exposures may eventually reach dangerous levels¹. Both the National Toxicology Programs and the International Agency for Research on Cancer have classified TCDD as a human carcinogen.

Over the past several years a number of studies have described the cutaneous effects of TCDD toxicity. The common TCDD-induced skin lesions are chloracne, porphyria cutanea tarda, hyperpigmentation, and malignant fibrous histiocytomas of the skin. In Korea, 2 cases of BCC, 1 case of SCC, and 1 case of merkel cell carcinoma in Vietnam Veterans exposed to TCDD have been reported^{2,3}. Ikuta et al.⁴ have reported that in primary cultured keratinocytes, TCDD increases post-confluent proliferation and late differentiation. TCDD also inhibits culture-induced senescence, which is a form of permanent

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Corresponding author: Jun Young Lee, M.D., Department of Dermatology, Seoul St. Mary's Hospital, College of Medicine, The Catholic University of Korea, 222 Banpo-daero, Seocho-gu, Seoul 137-701, Korea. Tel: 82-2-2258-1380, Fax: 82-2-594-3255, E-mail: jylee@catholic.ac.kr

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