Setleis Syndrome - Bitemporal "Forceps Marks" Syndrome in a 6-day-old Taiwanese Infant

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Setleis syndrome, a variant of aplasia cutis congenita, is a rare genetic disease characterized by bitemporal scarring resembling forceps marks, abnormal eyebrows and eyelashes, low frontal hairline and 'pursed lips'. We report the first Taiwanese case in a 6-day-old full-term male infant who was born spontaneously to unrelated, normal-appearing parents with atrophic scar-like lesions on the bilateral temporal areas. In addition, upward slanting of the eyebrows and partial absence of the lateral brows were found. There were no other obvious development anomalies. Low frontal hair line and ‘pursed lips’ appearance became more apparent at 6-month of age. The baby was otherwise normal, alert and active. The clinical features are consistent with Setleis syndrome. (Dermatol Sinica 24: 266-268, 2006)

Key words: Bitemoral forceps marks, Atrophic scar, Setleis syndrome, Aplasia cutis congenita

Setleis氏徵候群，是種少見的先天性表皮發育不全：臨床上，兩側顱部有類似產錯夾傷的疤痕，合併異常傾斜的眉毛、部分睫毛缺損、前額髮線較低及綻起的嘴型。我們報告第一例台灣的Setleis氏徵候群，並整理相關的文獻：一個台灣出生，六天大的男嬰，雙親非近親結婚且外觀正常，發現兩側顱部有萎縮、類似疤痕的斑塊，傾斜的眉毛、部分睫毛缺損，沒有合併其他的發育異常；追蹤六個月後，前額髮線較低及綻起的嘴型變得益發明顯，但生長、發育都正常。（中華皮誌 24: 266-268, 2006）
INTRODUCTION
Setleis syndrome, a variant of aplasia cutis congenita, is a rare genetic disease characterized by bitemporal scarring resembling forceps marks, abnormal eyebrows and eyelashes, low frontal hairline and “pursed lips”. Most reported cases of Setleis syndrome were Puerto Ricans, but the disease also has been observed in Spanish, German, Arabic and Japanese patients. We report the first Taiwanese case in a 6-day-old infant who presented with atrophic scar-like lesions on the bilateral temporal areas.

CASE REPORT
A 6-day-old male infant presented with atrophic patches over the bilateral temporal areas (Fig. 1). He is the first child born to unrelated, normal-appearing parents. Close examination of the face of both parents revealed no scars. The pregnancy and delivery were normal. No forceps were used. Physical examination revealed several well-demarcated, erythematous, scar-like depressions with visible blood vessels on both temples without palpable bony defect. In addition, there were upward slanting of the eyebrows and partial absence of the lateral brows. The face seemed coarser than most newborns. Neurological examination, and sonography of the heart and kidneys revealed no abnormal findings. Re-examination of the patient at 6 months of age revealed that the scars darkened in color, low frontal hair line, and 3 short linear depressions on the left side of the chin (Fig. 2). The lips appeared pursed up and the scalp hair was somewhat coarse. The baby was otherwise

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**Fig. 1**
A 6-day-old baby presents with bitemporal atrophic scar-like lesions with upward slanting of eyebrows and absence of lateral brows.

**Fig. 2**
(A) At age of 6 months, the low frontal hair line and the appearance of “pursed lips” become more apparent. (B) Note that there are 3 short linear depressions on the left side of the chin (arrows).
normal with normal development.

**DISCUSSION**

Setleis syndrome is a rare disease first described by Setleis et al in 1963. It is characterized by “coarse” face, bitemporal scarring resembling forceps marks, and anomalies of eyebrows and eyelashes. Additional facial anomalies include low frontal hairline, flattening of the nasal bridge with a bulbous nasal tip and “pursed lips”. The hair may be unruly. Although most cases are said to have normal growth and development, development delay has been reported.

Setleis syndrome is a type of aplasia cutis congenita (ACC), which manifests a spectrum of cutaneous and subcutaneous tissue defects, and is supposed to be caused by an intrauterine disruption of skin development. Setleis syndrome also belongs to focal facial dermal dysplasia (FFDD), which is a heterogenous group of disorders characterized by congenital bilateral scar-like facial lesions, with or without associated anomalies. FFDD is classified into 3 types: type I, autosomal dominant FFDD; type II, autosomal recessive FFDD; and type III, FFDD with other facial features (Setleis syndrome). The cause of Setleis syndrome is unknown, but has been suggested to result from an insufficient migration of neural crest cells into the frontonasal process and the first branchial arch.

Setleis syndrome is initially reported as an autosomal recessive disease, but later studies reported observed milder expression in parents, suggestive of autosomal dominant inheritance with variable expression and penetrance. Diagnosis of Setleis syndrome is essentially clinical. Histopathology of atrophic lesions is characterized by atrophic epidermis with a thinned dermis devoid of adnexal structure and elastic fibers. In the present case, the inheritance appears to be autosomal recessive. The diagnosis was made as early as 6 days of age based on the typical bitemporal forceps mark-like atrophic scars and eyebrow anomalies. Additional features of low frontal hair line and pursed lips became more obvious at 6-month of age. No obvious neurological developmental abnormalities have been observed so far, but long term follow-up is necessary. Most reported cases of Setleis syndrome are Puerto Ricans, but the disease also has been observed in Spanish, German, Arabic and Japanese patients. To our knowledge, our patient represents the first reported Taiwanese case.

**REFERENCES**