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CONGENITAL LOCALIZED ABSENCE OF SKIN : A REPORT OF ONE CASE

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Abstract

Congenital skin defect is a rare entity even among the integument anomalies. The author had the opportunity of carefully observing a newborn with this anomaly at birth and covering the wound with free skin graft early in life. As the results of treatment proved to be better than originally expected, the author would like to report on the characteristics of this entity and review some of the literature.

CASE REPORT

Mother : 26 years old, primara, office clerk

Family history : There was no consanguinity and no history of any skin abnormality in parents.

Course of pregnancy and delivery : Mother underwent wedge resection of the right overy for corpus luteum cyst during 13th week of pregnancy. Delivery of a female infant was premature, 36 weeks and 6 days, birth weight 2600 g and height 45 cm with Apgar's scores of 6 and 9. Complications noted at time of delivery were dark green amniotic fluid and skin defects on both feet. No other anomalies were noted.

Local findings : As shown in Fig. 1, a roughly symmetrical skin defect was noted on the dorsal aspect of both feet near the ankles. The skin defect extended completely around the right ankle and almost completely around the left. The surface of the affected area was lower than the healthy skin area and the boundary was well demarcated. The area was moist and dorsal veins of the foot could be seen easily. In addition to the above, the physiological transverse arch of the foot disappeared and protruded markedly forming a state of pes planus, the toes were spread out in a fan-like shape and the big toe was in a state of overtoe.

Treatment and subsequent course : As the patient had icterus physiologicus neonatorum and marked weight loss, surgery was delayed until the 19th day

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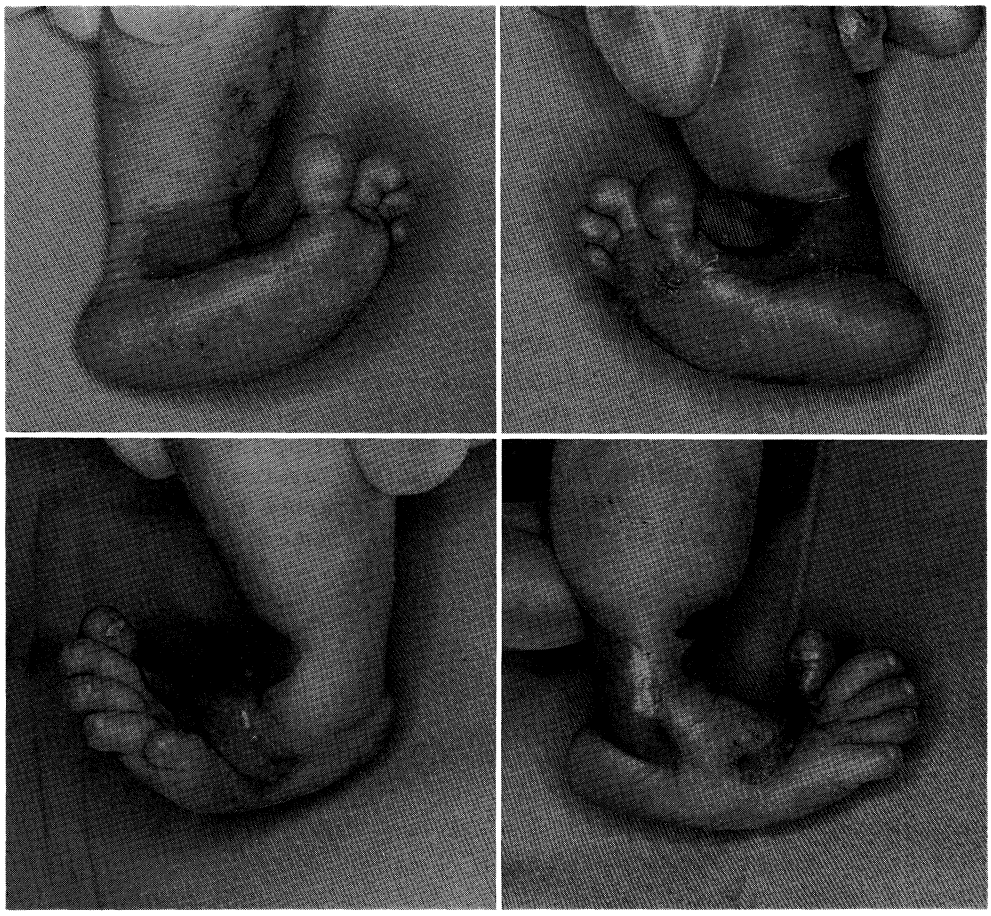

Fig. 1 Appearance of both feet on the next day after birth.

of birth (weight 2340 g). In approximately 50% of the area, there was epithelial formation, but as the regenerated epithelium was unstable and in a state of readily sloughing off, the regenerated epithelium was completely removed and returned to the state immediately after birth. Thiersch graft using skin cut from the buttock was performed. Due to hematoma, part of the graft of failed to take (approximately 1.5×2.0 cm). This area required about 1 month the left foot for healing by re-epithelization. A soft corrective cast was applied for 3 months to correct the foot deformity improved rapidly. At about 1 year after birth, the appearance and function were both almost normal. Fig. 2 shows the state 1 year 8 months after birth. There were deformities of the both great toenail (Fig. 3).

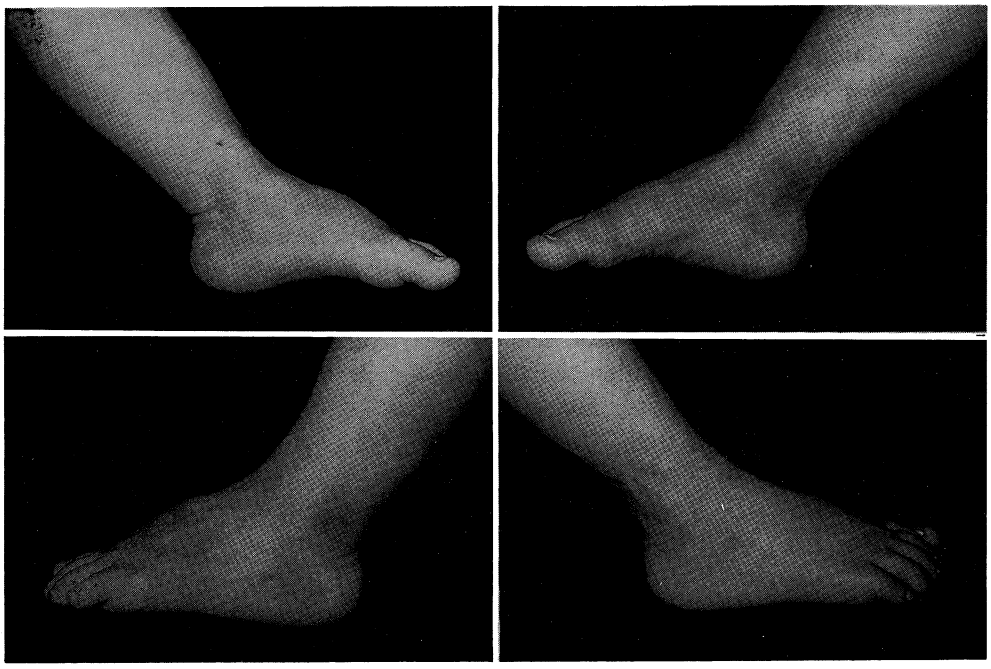

Fig. 2 Appearance of both feet at 1 year 8 months of age.

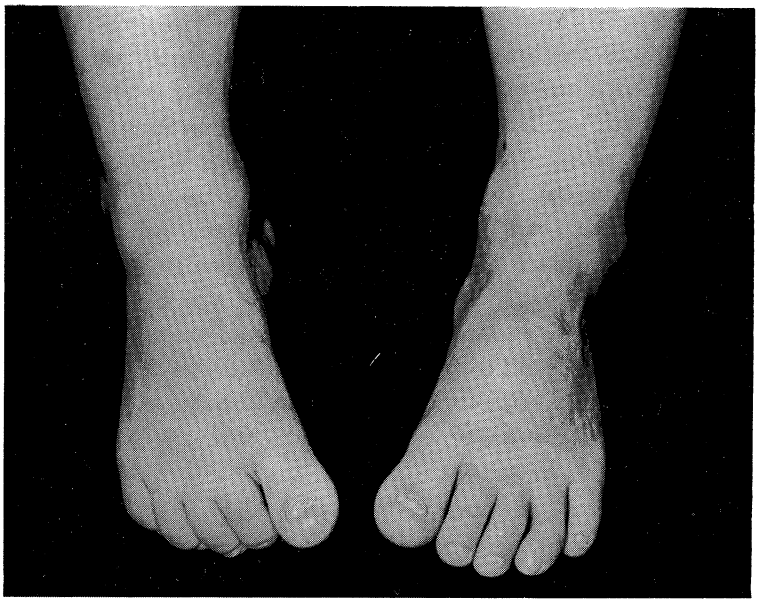

Fig. 3 Thickened and short big toenail seen at 1 year 8 months of age.

DISCUSSION

Campbell[1] is said to be the first to report this entity, and as of 1965 there have been 246 cases[2] presented. In Japan, Namba[3] reported the first case in 1931 and as 1971, 23 cases[4] have been introduced.

This anomaly develops over any surface of the body and is of varying sizes, but those involving the scalp are predominant, accounting for about 80% of the cases. For some unknown reason, symmetrical skin defects such as in this case are also quite numerous. There have also been reports on this anomaly in complication with other deformities such as deformities of the extremities and the heart, cleft lip and palate, hemangioma, hydrocephalus and meningocele. However, no special correlation with this condition and the complicating deformities has been introduced nor is the rate of such complication high.

Many theories have been advanced as to its etiology, but two main ones prevail. First, is heredity. There have been reports on multiple incidents in one family pedigree[5,6]. In Japan, cases among siblings have been reported by 3 investigators. The genetic trait points to an autosomal dominant gene. Cases in whom congenital skin defect occurred symmetrically near the distal portion of the extremities, are considered by some workers[7,8,9] to be a type of congenital epidermolysis bullosa. It is felt further study is required because the classification and categories of congenital epidermolysis bullosa are still fluid and concensus of opinion has not been reached. My case has not yet developed bullosa. She has a younger sister who is free of anomalies.

The second is the amnionic adhesion theory[10,11] in which it is said that a partial skin defect develops as a result of adhesion between the amnionic membrane and fetal epidermis. Following the publication of this theory, careful observation revealed that there are many cases without complication of adhesion. This finding has led to the thinking that amnionic adhesion occurred secondary to the skin defect. If amnionic adhesion occurs primarily, theoretically it would have to take place during the early embryologic stage, and if it does occur during such an early stage (within 3 weeks), the deformity would most likely not be limited to skin defect but also assume more severe deformities. Further, it would also be difficult to explain the reason for the development of the many cases with defects of the oral mucosa and the asymmetrical pattern. Thus, although it is a well known historical theory, at present it has been discredited.

There have been comparatively few cases in which surgery has been positively performed, and in many the long term follow-up findings are unknown. When review is restricted to re-epithelization alone, many are of the opinion that healing is good. However, it is said that re-epithelization occurred

only along the wound margin. In cases in whom skin graft was not performed, it took 3 weeks to 2 months to heal by scarring. The mortality rate because of skin defect is high, and about 50% of the cases reported in Japan have died. There have been reports[4] that severe contracture had occurred and adequate improvement could not be achieved because of the delay in performing graft. Thus, in view of the concept of wound healing, in cases with extensive wounds should be positively treated by free skin graft including homograft.

CONCLUSION

A newborn with skin defects on both feet and severe deformity was treated early by split thickness skin graft and application of corrective apparatus, and good results were achieved. The patient did not develop hypertrophic scars and contracture, and the results as compared to graft treatment for thermal burns were surprisingly good. In view of these finding my case is suspected to be a special type of epidermolysis bullosa[7,8,9], but as it is not yet general today, it was introduced as a case of congenital localized absence of skin.

Acknowledgment

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