Congenital Absence of the Scalp and Skull *

HERBERT CONWAY, M.D., GEORGE JOHNSON, JR., M.D.
New York, N. Y.

From the Department of Surgery (Plastic) of The New York Hospital-Cornell Medical Center

DEFECTS of the vertex of the head of the new-born infant are very rare but very interesting congenital defects. As with other rare anomalies, no doubt many more have occurred than have been reported in the medical literature. In 1826, Campbell made the first report of a case of congenital absence of the scalp. In 1954, Weinberg reported the data in 134 collected cases. Combined osteocutaneous defects of the vertex, on the other hand, are quite rare. The first report of a case of partial agenesis of the skull and scalp was made by Billard in 1828. This case exhibited a defect of scalp and skull through an area one and one-half by one-third inches, the area replaced by smooth, depressed cicatricial tissue. The bony defect was oblong, about one inch in greatest measurement. The infant died at one month of age.

Review of the world literature has yielded 22 well documented cases of congenital defect of the scalp and skull (Table 1). In this report the authors provide the summarized records of two additional cases encountered at The New York Hospital, bringing the total number of reported cases of congenital defect of the skull and scalp to 24. In addition, two cases of congenital defect of scalp alone are reported from The New York Hospital. These congenitally defective infants were encountered in the course of 418,983 admissions to The New York Hospital, 68,947 of which were newborn infants, during the years 1932 to 1955. Most of the defects were single lesions presenting in the parietal region near the midsagittal line. Commonly, the description was that of a depressed, shiny hairless area, covered by a thin, almost transparent and vascular cicatrix, the adjacent scalp being devoid of hair through a circumferential radius of several centimeters. Associated congenital anomalies occurred frequently. Often the defect was noted in still-borns. In the survivors, hemorrhage or meningitis occurred frequently in post-natal development.

Discussion of the etiology of the defect can be no more revealing than our current knowledge of the causes of any or all congenital anomalies. Review of the cases reported in the literature as well as our own has thrown no light on the subject. Details regarding the possibility of maternal vitamin deficiency are not available. Syphilis, looked upon as a probable causative factor because of the high number of still-borns in which the defect has been described, and because of the serpiginous outline of the ulcerative and granulomatous lesion in many cases, has not been found to have direct causal relationship. The defect is both ectodermal and mesodermal, both in the cases of deficiency of scalp only and in those of combined osteocutaneous defect.

The importance of this congenital anomaly is brought out by the fact that the death rate in infants with congenital defect of the scalp alone has been established at 20 per cent. Of the 22 cases of combined defect (skull and scalp) collected from the literature, 13 died, a mortality rate of 59 per cent.

Therapy may be separated into initial treatment and definitive treatment. That spontaneous epithelization of open granulomatous areas will occur is attested to

* Submitted for publication March 20, 1956.

TABLE 1. *Congenital Absence of Skull and Scalp*

Author	Year	Location	Size of Defect		Comments
			Scalp	Skull	
1. Billard	1828	left parietal	3.3 cm. ×7 mm.	2.2 cm. ×?	Patient died at 1 month. Also had a "hernie du cerveau."
2. Schrader	1893	just to the left of the midline	*	7×2 cm.	Died at 3 months.
3. Matthes	1894		*	*	Lived.
4. Mace	1908	frontal-parietal region	*	*	Lived. Dura absent. Encephalocele also present.
5. Kehrer	1910	between parietal and sagittal fontanelle	*	*	Lived. Defect down to Dura.
6. Lequeux	1910	parietal	from large to small fontanelle	from large to small fontanelle	Died in 3 days. Defect down to Dura. Multiple other anomalies.
7. Lequeux		parietal near small fontanelle	3×4 mm. and 5×6 mm.	3×4 mm. and 5×6 mm.	Lived. Also had syndactylism of both feet. (Two defects.)
8. Bonnaire	1913	left temporal	5×3 cm.	4×3 cm.	Died at birth. No other anomalies noted.
9. Bonnaire		bregmatic region	*	*	Observed by Barr of Cologne Museum.
10. Möller	1923	midline	10×7 cm.	5×8 cm.	Died at 14 days.
11. Möller	1923	midline	*	2 cm.	Died in 17 hours. Defect joined amniotic membrane around edge of defect.
12. Heidler	1924	parietal	5×2.5 cm.	5×2.5 cm.	Died at 2 months of meningitis.
13. Gare	1924	midline	7×8 cm.	less than 7×8 cm.	Died at 20 days after bleeding from longitudinal sinus for 4 hours.
14. Heinrichsbauer	1926	in midline near small fontanelle	3×2.5 cm.	3×2.5 cm.	Died—was a premature infant.
15. Terruhn	1930	vertex	Two were 3 cm. in diameter, the other smaller	Two were 3 cm. in diameter, the other smaller	Born dead.
16. Scheyer	1934	in midline near small fontanelle	3×2.5 cm.	3×2.5 cm.	Lived. Healed spontaneously.
17. Carreño	1934	left posterior parietal	4×5 cm. diam.	4×5 cm. diam.	Lived. No other anomalies.
18. Franke	1937	in midline 1 cm. from small fontanelle	2×1 cm.	2×1 cm.	Died. Cleft palate, anomalies of hand and mongolism also present.
19. Robyn	1939	large defect from bregmatic fontanelle to inferior ⅓ of parietal fontanelle to lamboid fontanelle	*	* (see location)	Lived. No other anomalies.
20. Vignocchi	1940	in midline between parietal bones	5×4 cm.	5×4 cm.	Died in 4 days. Mongoloid face, intraventricular septal defect and supernumerary digits.
21. Turrettini	1949	between sagittal suture and left branch of lamboid suture	2 cm.	2 cm.	Lived. No other anomalies.
22. Ingram	1954	vertex	*	*	Lived.
23. The New York Hospital	1955	vertex	13.5×9 cm.**	13.5×9 cm.**	Lived. Defect down to pia mater.
24. The New York Hospital	1955	both parietal and posterior portion temporal	13×6 cm.	13×9 cm.	Lived.

* Not specified.
** At 8 years of age.

by the smooth scar-epithelial surface exhibited on examination of subjects who have survived. In two of our cases the defects presented as areas of thin granulation at birth. In one of these (Case 1) spontaneous epithelization was effected and skin grafting was not required in the initial therapy (Fig. 1). In the other infant (Case 4) accurate dressing care was rewarded by diminution of the size of the defect. Operation was refused and follow-up has not been obtainable. It is probable that many

of the deaths in the early weeks after birth have been due to hemorrhage from the granulomatous area. Therefore, closure by skin grafting probably is the early treatment of choice.

Definitive treatment may be delayed until the child is older and in good condition to stand operation. Cicatrix or skin graft over the area should be excised, the scalp and galea mobilized and sutured. Mobilization of these tissues, if the defect is a large one, may require the use of multiple relaxation incisions in the galea, parallel to the long axis of the wound (Fig. 2). If the size of the defect is such that linear closure does not apply, a rotation flap of scalp is applicable, using a thick-split skin graft to cover the area from which the flap has been rotated. Coverage of the brain with soft tissue flap from the scalp offers reasonable protection. In selected cases it may be advisable to offer further protection by insertion under the flap of a patterned bone graft or a fashioned prosthesis of inert material.

FIG. 1. Case 1. Congenital absence of the skin of the scalp present at birth as an open granulating area with serpiginous outline. Some epithelization took place in the six weeks after birth, at which time the above photograph was taken. There was no abnormal bony defect.

CASE REPORTS

Case 1. T. L. (New York Hospital Case #24 65 31). This male patient was born in The New York Hospital on September 14, 1939. The mother and father, both age 32, had been in good health. Blood serologic tests for syphilis were negative in parents and infant. Family history revealed no history of congenital anomalies. The first pregnancy had resulted in a miscarriage. The pregnancy leading to the birth of this patient was full term and uneventful. At the time of delivery, as the head presented on the perineum, a granulating defect, serpiginous in outline, was noted in the scalp of the infant. Following episiotomy an uncomplicated delivery was performed.

Initial examination by the pediatrician revealed a healthy infant with no anomalies, other than the lesion of the scalp. The birth weight was 3470 Gm. Blood Wassermann test was negative. On the vertex of the scalp there was a 6 by 6 cm. ulceration with a granulating base. The surrounding scalp had several smaller defects. No bony defect was palpated. Local dressing care was effected, and in 20 days the ulcer had healed. At one year of age the resulting scar (Fig. 1) was excised, and the surrounding scalp undermined and approximated. Residual scars in this area were excised on 4 occasions. Successive operations were required, because often the scar would stretch in spite of wide undermining. Finally, the technic of galeal relaxation incisions (Fig. 2) was successful. The latest photograph taken at the age of 11 reveals the final result (Fig. 3).

This case is instructive in demonstrating that as long as the defect involves the scalp alone and does not overlie one of the venous sinuses, primary treatment may safely be directed toward preventing infection until the wound has healed. Excision of the scarred scalp may be performed at a later date, as an elective procedure.

Case 2. S. S. (New York Hospital Case #53 75 98). This male infant was born at another hospital on April 17, 1949, one month prior to the expected date of confinement. At 4 months of the pregnancy the mother had a single episode of a clear vaginal discharge. At 7 months the mother had rather rapid enlargement of the abdomen, and was thought to have twins. Three days prior to the delivery the membranes ruptured and a copious amount of blood-tinged amniotic fluid escaped. The delivery was otherwise uneventful. The birth weight was 4 lbs., 8 ozs. (2041 Gm.). Three days after birth the patient was transferred to The New York Hospital.

Physical examination revealed an absence of the skin over the right side of the face (Fig. 4)

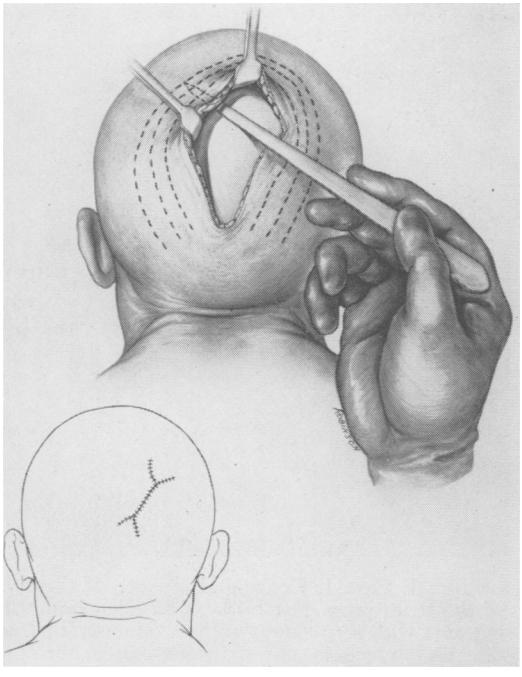

FIG. 2. Case 1. At one year of age the scarred area was excised. Margins were approximated after wide undercutting with the aid of multiple subcutaneous relaxation incisions as indicated by the dotted lines. These incisions extend through the galea and subcutaneous scalp but not through the skin.

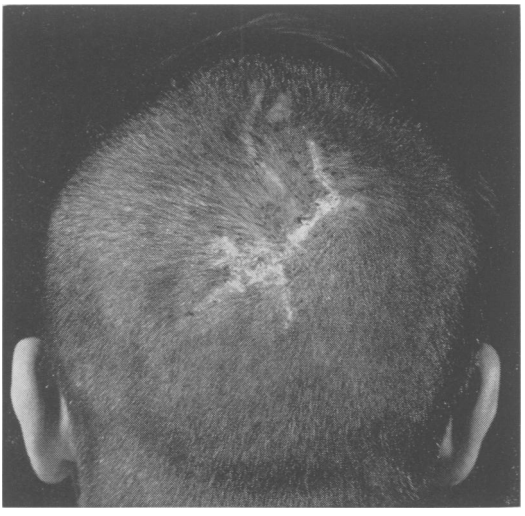

FIG. 3. Case 1. As the child's head grew, the residual scar of excision widened. Excisions of scar were required at intervals of two years on four different occasions to bring the child's scalp to the above acceptable condition without noticeable bald spots.

extending from a vertical line through the lateral canthus of the eye cephalad over the brow, caudad to the base of the neck, posteriorly to the posterior portion of the neck and over the ear and the parietal region of the scalp. The area was vascular, smooth, and glistening.

The finger nails were absent from the right second and third fingers. Over the right index finger an area denuded of skin measured 1 by 2 cm. Other cutaneous defects were as follows: an area .5 cm. in diameter over the lateral thigh; an area 1 cm. in diameter over the thigh; and an area 2 by 3 cm. over the left cheek. Lesions with the appearance of ruptured blebs were found above the medial malleolus, at the ankle, on the lateral aspect of the calf, and on the dorsum of the fifth toe and foot. Intact blebs were found on the lateral aspect of the thigh and on the surface of the tongue. There was congenital absence of the right ear.

The infant was placed in an incubator and given antibiotic therapy. Three days following admission (6 days after birth) the patient died. The final diagnoses after autopsy were prematurity, subarachnoid and intraventricular hemorrhage, bronchopneumonia, congenital ectodermal defects, and partial agenesis of the cerebellum.

A case such as this makes one wonder if the 20 per cent mortality quoted by Ingalls is really due to the congenital absence of the scalp or whether it may not be caused by the frequent association of other developmental anomalies.

Case 3. P. G. (New York Hospital Case #70 68 57). This male infant was born August 20, 1947, of a precipitous delivery while entering another hospital. The pregnancy, which was full term, had been uncomplicated. The mother age 30 and the father age 31 were in good health and had negative Wassermann tests. There was no family history of congenital anomalies, and two female siblings were free from anomalies. Birth weight was 5 lbs., 13 ozs. (2589 Gm.). The scalp and skull over the vertex were absent, the defect being covered by a thin, membranous tissue. No description as to size was obtainable. Congenital deformities of the right hand and foot also were diagnosed.

Five days after birth the infant was transferred to another hospital where it was noted that the scalp area was infected. Although accurate records are not available, the child was hospitalized for a total of 9 months, during which time, on 4 separate occasions, thick split skin grafts were applied to the defect. Healing of the wound by this technic proved to be life-saving. During the child's first 7 years many physicians were consulted. Varying opinions were offered, and the

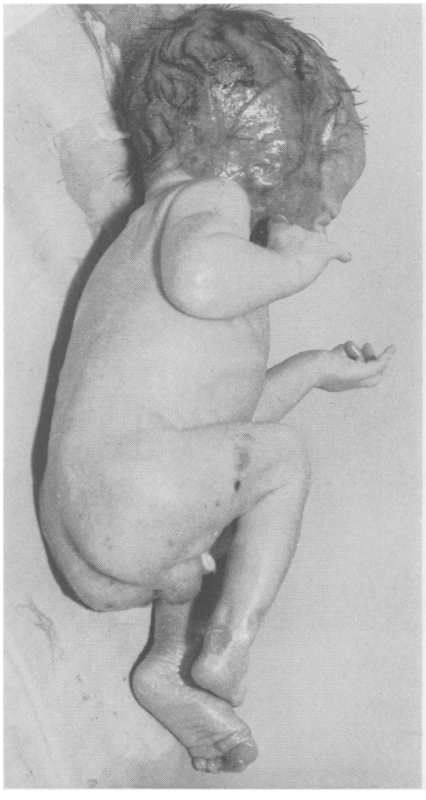

FIG. 4. Case 2. Congenital absence of the scalp of the right side of the head. The thin, shiny membrane is the only covering of the brain. The ear is absent.

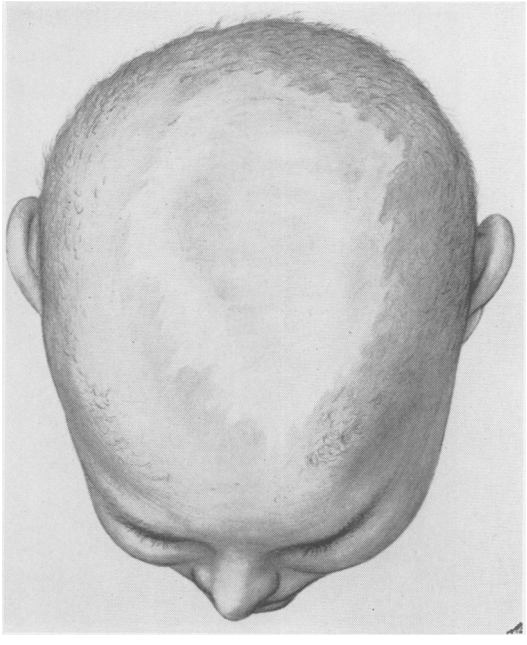

FIG. 5. Case 3. Artist's sketch of scalp when this boy was seven years old. The irregular bony defect could be palpated at the margins of the scarred area. Healing during infancy had been effected by the use of thin grafts of skin.

parents became confused. Because the brain was covered only by the thin skin graft and adjacent scar, the parents were afraid to send the child to school, fearing slight injury might result in serious complications.

The child was admitted for the first time to The New York Hospital in April, 1955. His development, both mentally and physically, had been in accordance with accepted standards. He had had the usual childhood diseases with no significant sequelae. His tonsils had been removed at the age of 5.

Examination on admission revealed a somewhat thin boy who was in good general health. On the vertex of the scalp there was a shiny, smooth, white area measuring 13.5 by 9.0 cm., which had the appearance of a scar with an irregular, slightly raised margin. The adjacent several centimeters of scalp were devoid of hair (Fig. 5). On palpation the above described area was very thin and an irregular defect of the skull could be made out which measured 13 cm. at its greatest length and 9 cm. at its greatest width. The osseous defect involved portions of both parietal bones and the anterior portion of the occipital bone. Exophoria of the right eye was noted. There were multiple healed scars on the back and abdomen. There was an absence of all but the proximal phalanges in each finger of the right hand. The left hand was normal. The first, second, and third toes of the right foot were absent with only one phalanx present in the fourth and fifth toes. There was syndactylism between the second, third, fourth and fifth toes of the left foot. The boy's gait was described as short, quick steps, thought to be due to the pedal anomalies. No neurologic defects were found. Intelligence was average, and there was no evidence of emotional disturbance.

Complete blood count and urinalysis were within normal limits. Skull x-ray studies (Fig. 6) revealed absence of a portion of the parietal bones along the vertex with smaller defects in the occipital and frontal bones. The outline of the defect of the skull was serpiginous. The remainder of the skull was normal. Sagittal sinus venograms (Fig. 7) revealed no opacification of the posterior portion of the superior sagittal sinus. Pneumoencephalogram (Fig. 8) showed a normal ventricular system with no evidence of cerebral atrophy.

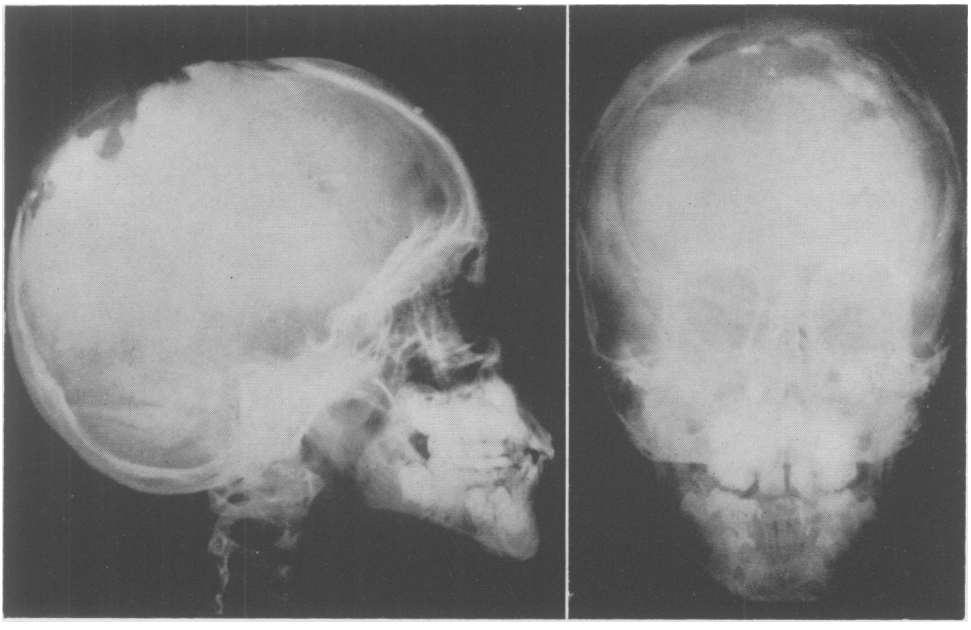

FIG. 6. Case 3. X-ray of the skull, showing bony defects.

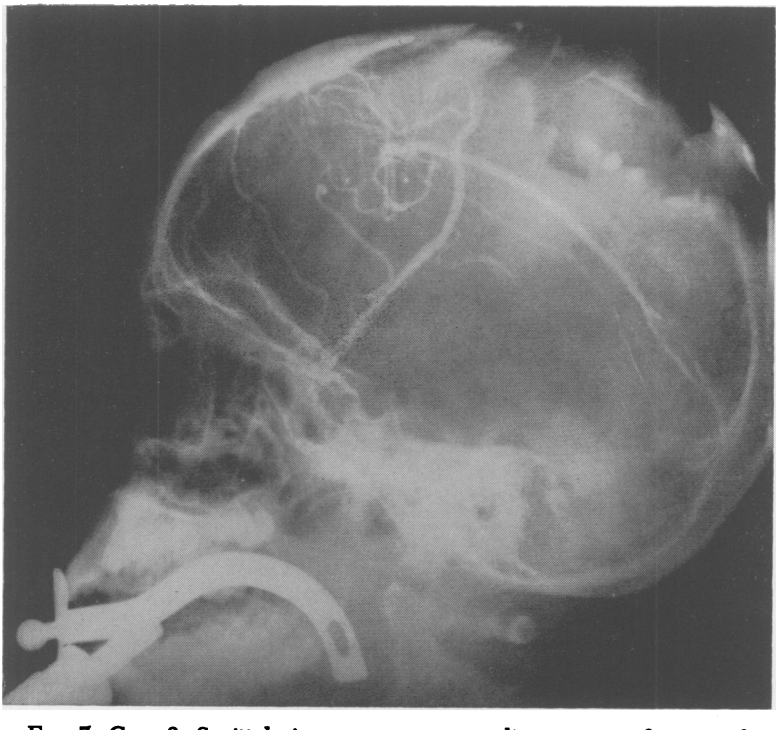

FIG. 7. Case 3. Sagittal sinus venogram revealing no opacification of posterior portion of superior sagittal sinus.

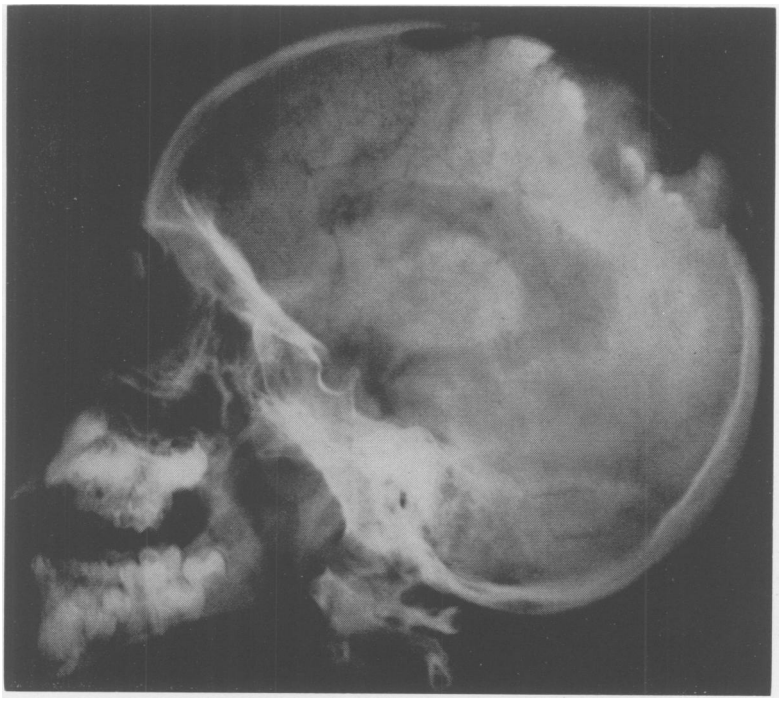

FIG. 8. Case 3. A pneumoencepahlogram demonstrates the normal ventricular system.

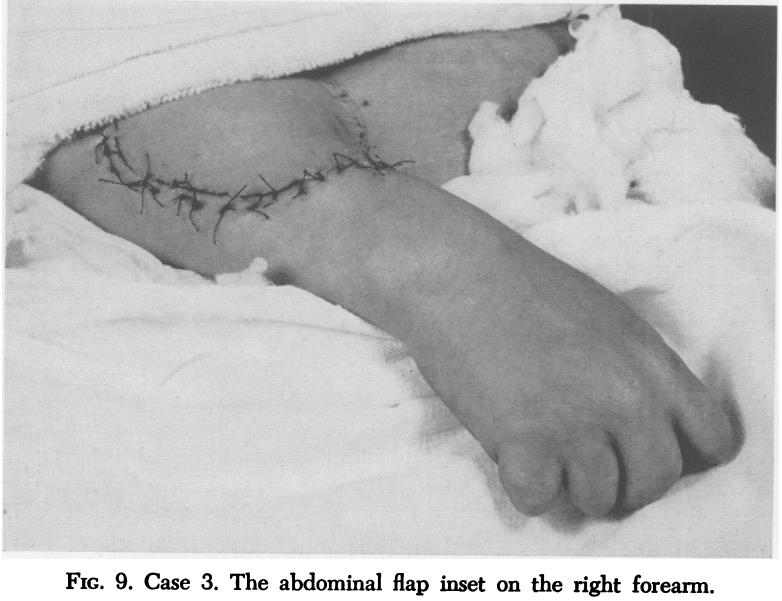

FIG. 9. Case 3. The abdominal flap inset on the right forearm.

In order to afford protection to the brain, the decision was made to replace the thin covering with pedicled tissue. On June 3, 1955, an abdominal flap was partially elevated and inset into the right arm (Fig. 9). Eleven days later partial elevation of the abdominal flap was performed. Minor infection developed in the site of partial elevation, which delayed the next procedure for several weeks.

On July 22, 1955, the split thickness graft was

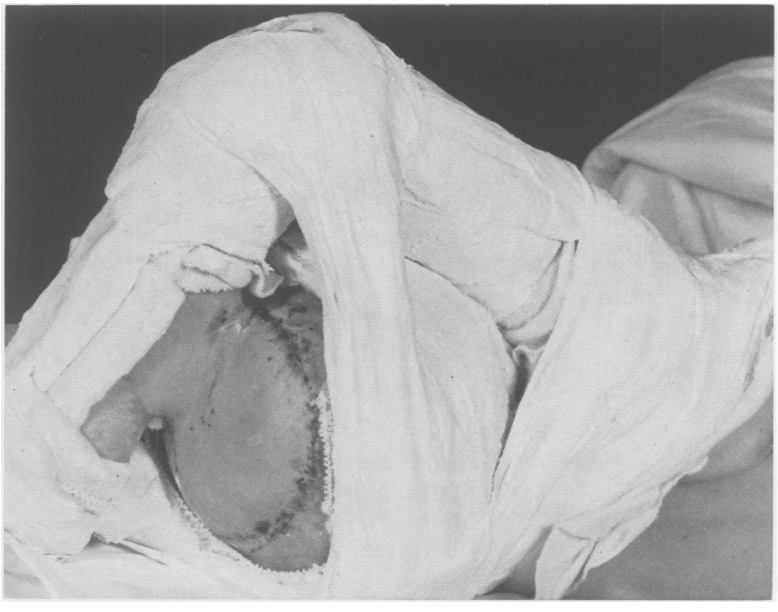

FIG. 10. Case 3. The abdominal flap migrated to the scalp via the forearm, with the position being maintained by plaster fixation.

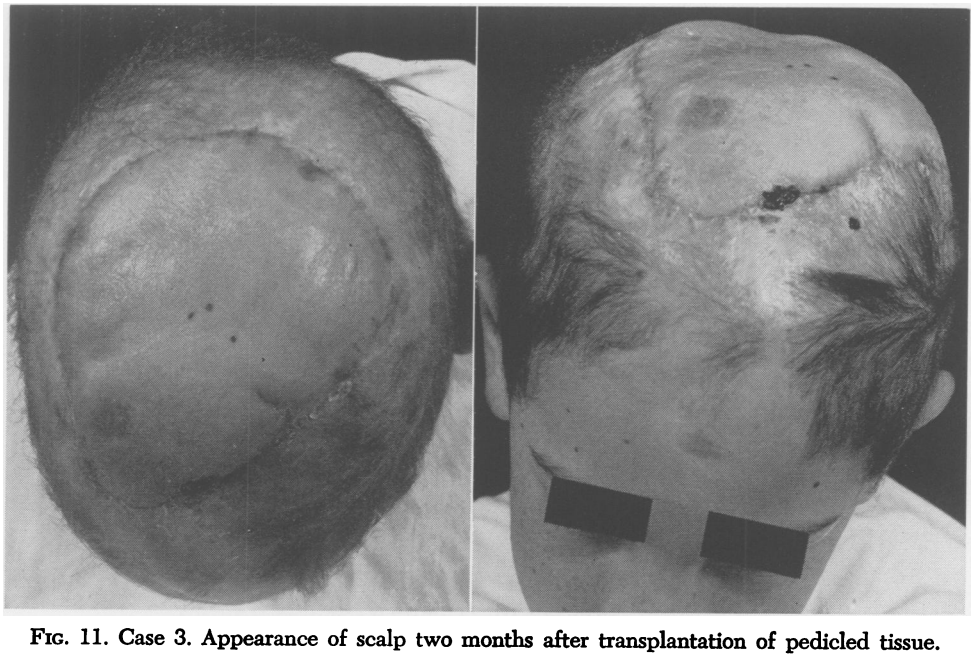

FIG. 11. Case 3. Appearance of scalp two months after transplantation of pedicled tissue.

removed from the defect of the scalp. This dissection was most tedious because the graft had been placed directly on the pia mater. As the dissection progressed, a small amount of cerebrospinal fluid drained from a minute hole in the floor of the wound. A cotton pledget was placed over the hole and at the end of the procedure no further leak was evident. The abdominal pedicle on the right arm was then freed from the abdomen and migrated via the forearm to the defect of the scalp

(Fig. 10). Immobilization was effected with plaster. In the ensuing few days clear fluid developed under the flap and leaked from the sutured edges of the wound. The child was kept in a sitting position and antibiotics were administered. The leakage subsided. On August 24, 1955, the abdominal pedicle flap was divided from the arm and further inset into the scalp. The postoperative course was uncomplicated, and the boy was discharged from the hospital to return in 6 months for insertion of a prosthesis. Fig. 11 was taken 2 months after the migration of pedicled tissue.

This case is one of unusual interest not only because it represents an example of the rare osteocutaneous defects, but because the defect in this case was larger than that of any case previously reported. The application of the skin graft in the patient's infancy undoubtedly was a life-saving maneuver. The dissection was most difficult when it had to be removed and replaced by the pedicled tissue. Also, this case report is the first in which reparative surgery has been employed.

Case 4. S. S. (New York Hospital Case #37 06 75). This female infant was born November 8, 1943, by low forceps delivery. The pregnancy and delivery had been uneventful in all other respects. The birth weight was 2890 Gm.

At birth a defect measuring 6 by 13 cm., and involving both the parietal and the posterior portion of the temporal bones, was noted. The area was covered by a fine membrane through which the convolutions of the brain could be seen. A systolic heart murmur was heard over the precordium leading to the presumptive diagnosis of congenital heart disease.

Treatment of the defective area of the scalp and skull was aimed at keeping the area clean and dry. Gentian violet was applied locally and sulfadiazine was given systemically in an attempt to prevent infection. On this regimen a thick eschar formed over the defect and the wound of the scalp became smaller. The child was discharged after a month with plans to readmit her at a later date for repair of the defect.

The skull and scalp defect continued to become smaller and by the age of 3 it measured about 4 cms. in diameter. The parents refused operation at this time, and further follow-up has been unobtainable.

Although the above patient survived with conservative therapy in the presence of a large defect, it is felt that this is a dangerous course to follow. The application of thick-split grafts to cover the defect during the first 24 to 48 hours of the child's life would seem to be a safer plan to follow.

SUMMARY

1. Congenital absence of the tissues of the vertex is a rare finding. A defect of the scalp alone is more frequent than that of the skull and scalp. Only 22 cases of combined osteocutaneous defect of the head have been found in the literature.

2. Treatment should be aimed toward immediate coverage of the granulomatous defect with a skin graft, unless it is small enough to close spontaneously.

3. Two additional cases of congenital absence of the scalp and two cases of total osteocutaneous defect of a portion of the head are reported.

BIBLIOGRAPHY

1. Billard, C. M.: Traité des Maladies des Enfans Nouveaux-nés et à la Mamelle. Paris, 1828.
2. Bonnaire, E. and G. Durante: Arrêt de Développement Limité des Enveloppes Cutanée et Osseuse du Crane. La Presse Médicale, **21**: 185, 1913.
3. Carreño, C. and M. Seoane: Defecto Congénito, Limitado, de Cráneo y Epicráneo en un Recién Nacido. Archivos Argentinos de Pediatria, **5**: 271, 1934.
4. Campbell, W.: Case of Congenite Ulcer on the Cranium of a Fetus. Edinburgh Journal of Medical Science, **2**: 82, 1826. Quoted by Greig Edinburgh Medical Journal, **38**: 341, 1931.
5. Franke, K.: Beitrag zur Genese der Kongenitalen Haut und Knochendefekete am Schädeldach. Monatsschrift Geburtshülfe und Gynäkologie, **5**: 107, 1937.
6. Gare, A. L. Ch.: Een merkwaardig Geval van een Hangeboren Hâid—En Been-Defect bij een Passgeborene. Nederlandsch Tijdschrift Voor Geneeskunde, **68**: 1391, 1924.
7. Heidler, H.: Kongenitaler Hautdefekt am Kopfe. Wiener Klinische Wochenschrift, **37**: 114, 1924.
8. Heinrichsbauer, F.: Über angeborene Hautdefekte beim neugeborenen mit besonderer Berücksichtigung ihrer Ätiologie. Monatsschrift für Geburtshülfe und Gynäkologie, **75**: 267, 1927.
9. Ingalls, N. W.: Congenital Defects of the Scalp, Studies in The Pathology of Development III. American Journal of Obstetrics and Gynecology, **25**: 861, 1933.

10. Ingraham, F. D. and D. D. Matson: Neurosurgery of Infancy and Childhood. C. C. Thomas, Springfield, Ill., 1954.
11. Kehrer, E.: Über kongenitale Defekte am Schädel infolge amniotischer Verwachsungen. Monatsschrift für Geburtshülfe und Gynäkologie, 31: 183, 1910.
12. Lequeux: Zentralblatt für Gynaekologie, 34: 636, 1910.
13. Mace: L'Obstétrique, 13: 97, 1908.
14. Matthes, V. W.: Ueber angeborene, noch nicht verheilte, umschriebene Hautdefekte bei Neugeborenen. Inaugural-Dissertation, Marburg, 1894.
15. Möller, K. O.: Congenital Defects of the Cranium Caused by Amnionic Adhesion. Acta Gynecologica Scandinavica, 2: 144, 1923.
16. Møller, K. O.: Un Cas d'Acranie et d'Hydromicrocéphalie avec une Bride Amniotique Persistante. Acta Gynecologica Scandinavica, 2: 380, 1923.
17. Robyn, G.: Absence Congénitale de la Voûte Cranienne Chez un Nouveau-Né. Survie et Processus de Réparation. Bruxelles—Medical, 17: 1035, 1937.
18. Schrader, V.: Zentralblatt für Gynäkologie, 17: 374, 1893.
19. Scheyer, von H. E.: Zur Kasuistik der kongenitalen Haut- und Knochendefekte am Schädeldach. Monatsschrift für Geburtshülfe und Gynäkologie, 97: 65, 1934.
20. Terruhn, E.: Über die Entstehung amniogener Hautdefekte während der Schwangerschaft mit besonderer Berücksichtigung des Schädels beim Neugeborenen. Archiv für Gynäkologie, 140: 428, 1930.
21. Turrettini, A.: Un Cas de Malformation Congénitale Rare: Le Défaut Osteo-cutané Total du Crâne. Gynäecologia, 128: 455, 1949.
22. Vignocchi, R.: Contributo allo Studio delle Aplasie Congenite cutanee ed Ossee delle volta cranica. La Clinica Ostetricae Ginecologica, 42: 58, 1940.
23. Weinberg, A. and S. Gittelson: Congenital Scalp Defect. New York State Journal of Medicine, 54: 809, 1954.