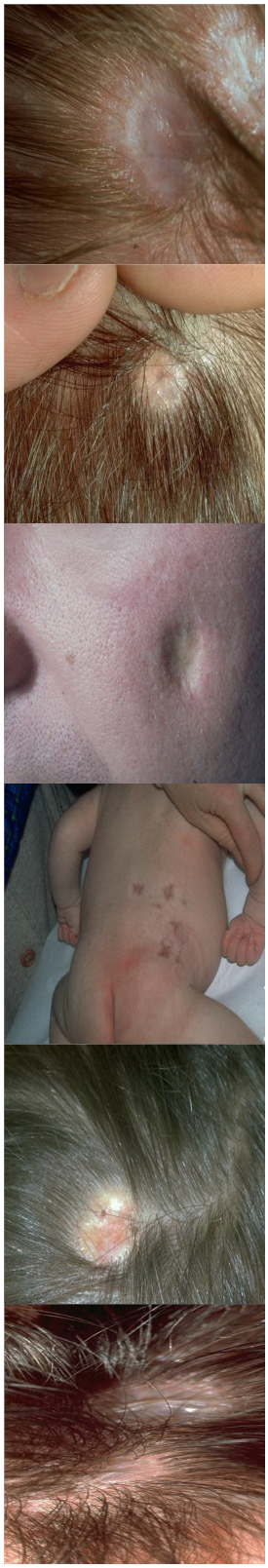

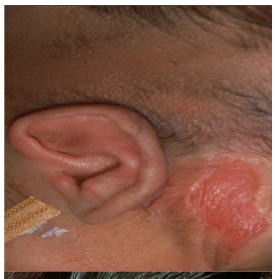
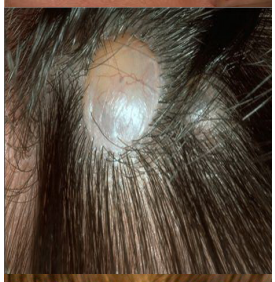

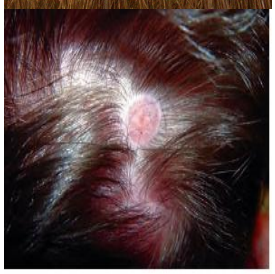
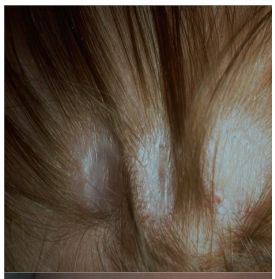
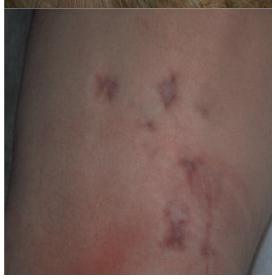

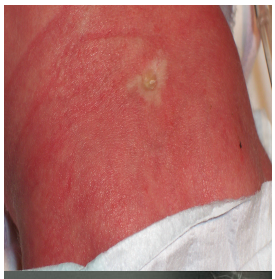
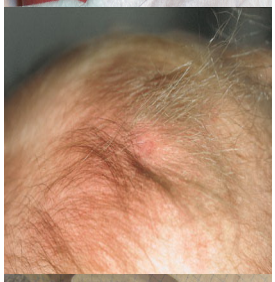
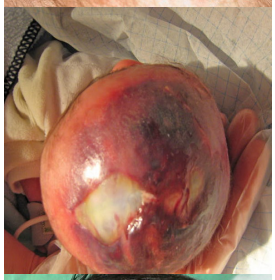
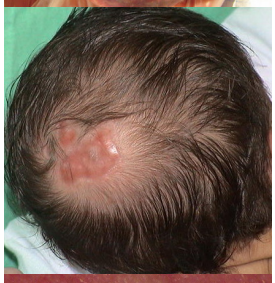
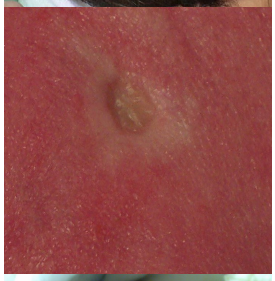
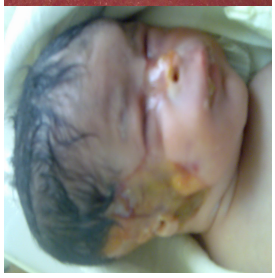

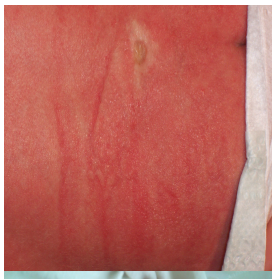
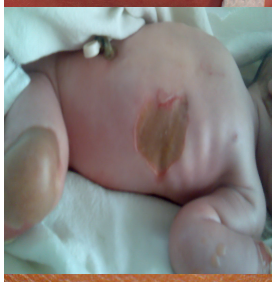
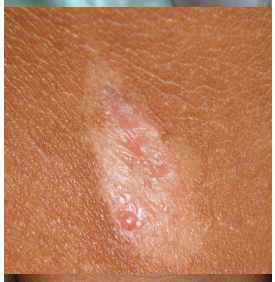
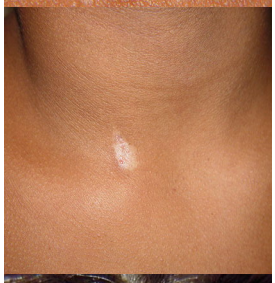
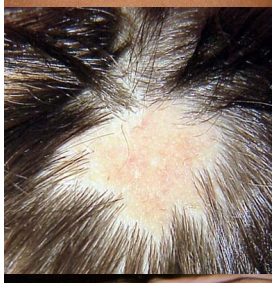
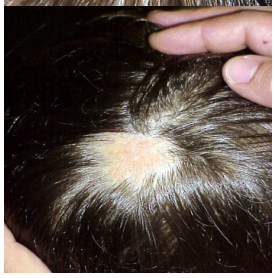


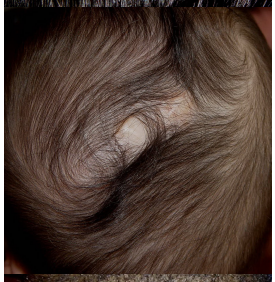
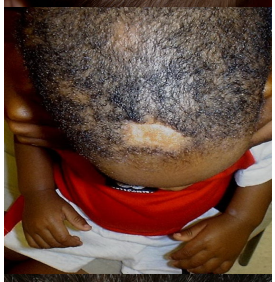
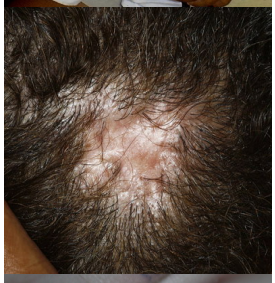
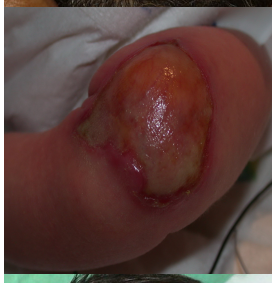
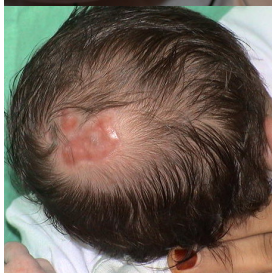

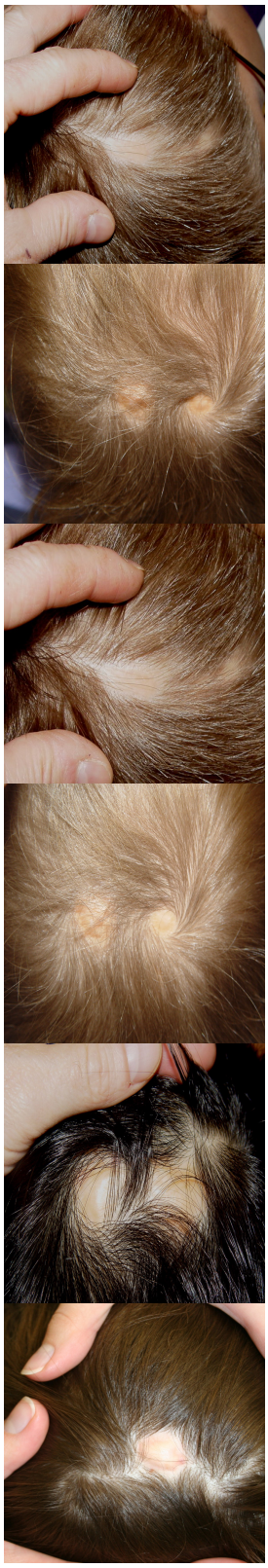

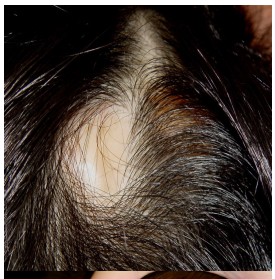
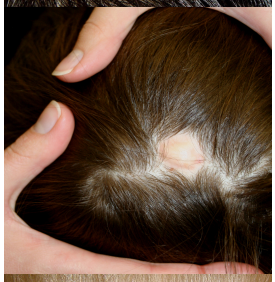
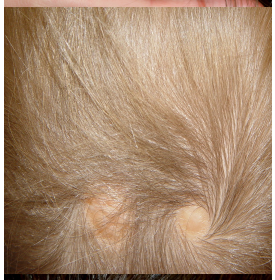
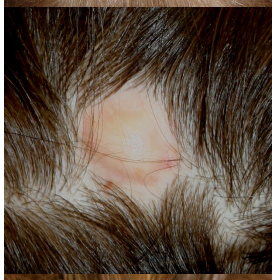
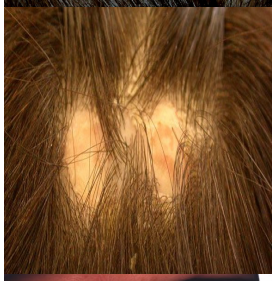
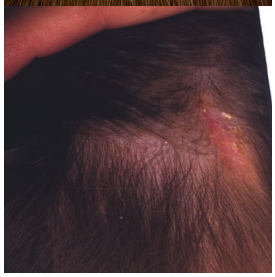

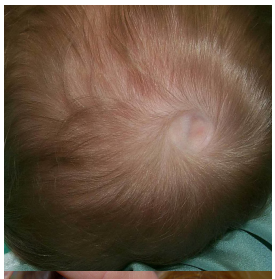
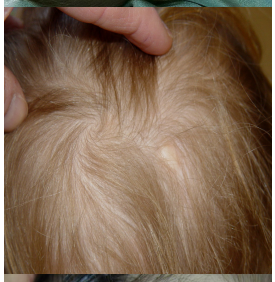
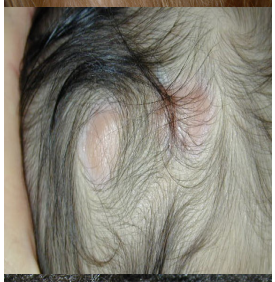
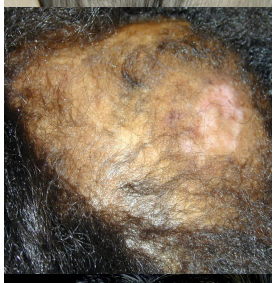
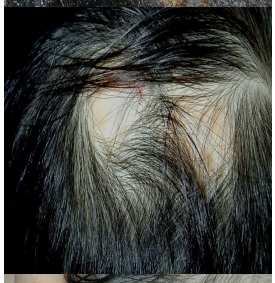
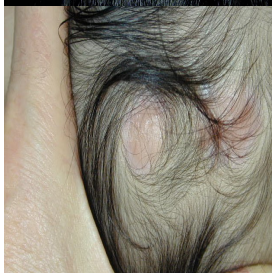

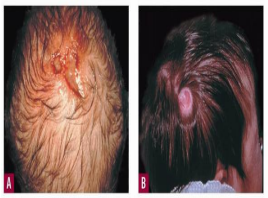

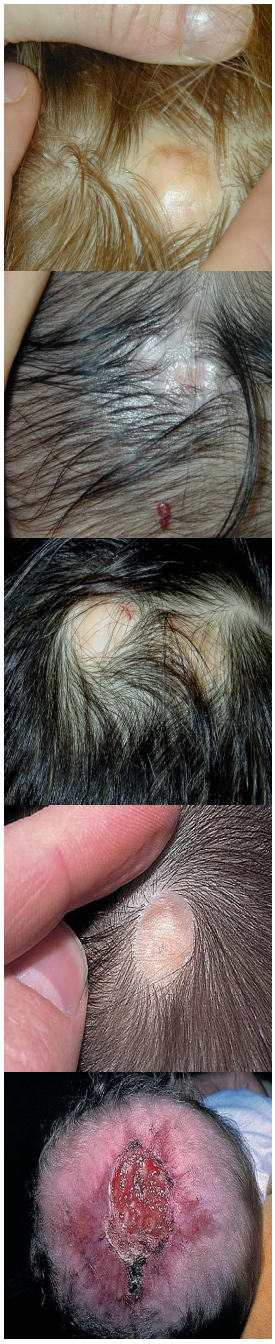

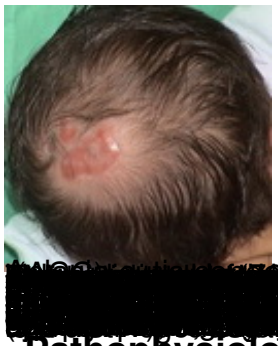

[redacted]

Pathophysiology

The skin and occasionally the underlying structures are affected in aplasia cutis congenita. Lesions are clean, sharply demarcated, and noninflammatory in appearance.

Frequency

United States

Aplasia cutis congenita is an uncommon anomaly of newborns. More than 500 cases have been reported since it was first described, but because of significant underreporting of this generally benign disorder, the precise frequency is unknown. One estimate of incidence is approximately 3 in 10,000 births.

International

The international frequency of aplasia cutis congenita is expected to be similar to that in the United States.

Mortality/Morbidity

If the defect is small, recovery is uneventful, with gradual epithelialization and formation of a hairless, atrophic scar over several weeks. Small underlying bony defects usually close spontaneously during the first year of life. Surgical repair of large or multiple scalp defects with excision and primary closure, if feasible, or with the use of tissue expanders and rotation of a flap, may be considered. Truncal and limb defects, despite their large size, usually epithelialize and form atrophic scars, which can later be revised if necessary.

Underlying or associated defects may also significantly affect mortality and morbidity. Full-thickness defects of the scalp, skull, and dura are associated with a mortality rate of greater than 50%. Even large defects on areas other than the scalp usually heal well with conservative skin care using silver sulfadiazine ointment. The rare larger scalp defects are prone to complications of hemorrhage and infection; subsequently, patients are at risk for death. Extensive aplasia cutis congenita of the scalp may be associated with an increased risk of

sagittal sinus thrombosis. For these reasons, surgical intervention may be required for large, full-thickness scalp defects.

Race

No racial predilection is reported for aplasia cutis congenita.

Sex

Unless associated with an X-linked malformation syndrome, no sexual predilection exists in aplasia cutis congenita.

Age

Aplasia cutis congenita lesions are congenital.

Clinical

History

- History should include a review of maternal medications taken during the pregnancy and evidence of infections such with varicella or herpes viruses. Because many forms of aplasia cutis congenita are inherited, a thorough family history is important.

Physical

- Aplasia cutis congenita diagnosis is made on the basis of physical findings indicative of an in utero disruption of skin development. Most lesions occur on the scalp lateral to the midline, but they may also occur on the face, the trunk, or the limbs, sometimes symmetrically.
- The lesions are noninflammatory and well demarcated. The appearance of the lesions varies, depending on when they occur during intrauterine development. Lesions that form early in gestation may heal before delivery and appear as an atrophic, membranous, parchmentlike or fibrotic alopecic scar, whereas less mature defects may present as an ulceration of variable depth. With only the epidermis and the upper dermis involved, minimal alopecic scarring may result, but deeper defects may extend through the dermis, the subcutaneous tissue, and rarely the periosteum, the skull, or the dura. Distorted hair growth around a scalp lesion, known as the hair collar sign, is a marker for underlying defects.

- A bullous variant of aplasia cutis congenita manifesting as a tense yellow vesicle on the scalp has been reported.
- A complete physical examination should be performed to search for associated physical anomalies or recognizable malformation syndromes

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Causes

- No unifying theory can account for all lesions of aplasia cutis congenita. Because this condition is the phenotypic result of more than one disease process, it is likely that more than one mechanism is involved. Mechanisms include genetic factors, teratogens (eg, methimazole, carbimazole, misoprostol, valproic acid), compromised vasculature to the skin, and trauma. Of particular note is the association of fetus papyraceous with bilaterally symmetric aplasia cutis congenita.
- The proximity of scalp aplasia cutis congenita to the scalp hair whorl, which is thought to be the point of maximum tensile force during rapid brain growth, has led to the hypothesis that tension-induced disruption of the overlying skin occurs at 10-15 weeks of gestation when hair direction, patterning, and rapid brain growth occur.
- Early rupture of the amniotic membranes, forming amniotic bands, has appeared to be the cause of aplasia cutis congenita in several cases.
- The bullous variant of aplasia cutis congenita reveals a distinct histologic pattern identical to those in encephaloceles and meningoceles. This supports a hypothesis that this variant of aplasia cutis may represent the form fruste of a neural tube closure defect.

Treatment

Medical Care

The decision to use medical, surgical, or both forms of therapy in aplasia cutis congenita (ACC) depends primarily on the size, depth, and location of the cutaneous defect and therapy of associated defects as indicated.[43,44,45,46,47,48,49,50,51]

- Local therapy includes gentle cleansing and the application of bland ointment or silver sulfadiazine ointment to prevent desiccation of the defect.
- Antibiotics are only indicated if overt signs of infection are noted.

- Other treatment is rarely necessary because the erosions and the ulcerations almost always heal spontaneously.
- A variety of specialized dressing materials have been used.[52,53]

Surgical Care

- Surgical repair is not usually indicated if the defect is small. Recovery is uneventful, with gradual epithelialization and formation of a hairless, atrophic scar over several weeks. Small underlying bony defects usually close spontaneously during the first year of life.
- Surgical repair of large or multiple scalp defects may require excision with primary closure, if feasible, or the use of tissue expanders and rotation of a flap to fill the defect. Skin and bone grafts may also be required. Occasionally, skin grafting may be required.
- Patients with large, full-thickness scalp defects may face a treatment dilemma: conservative versus surgical therapy of the defect. Complete osseous regeneration of a large skull defect associated with aplasia cutis congenita has been obtained with a conservative approach of skin care and silver sulfadiazine dressings. No surgical treatment of bone or soft tissue reconstruction was necessary. On the other hand, aplasia cutis congenita of the scalp may be complicated by sagittal sinus hemorrhage or thrombosis, and primary closure with scalp flaps may prevent a potentially fatal outcome.
- Truncal and limb defects, despite their large size, usually epithelialize and form atrophic scars, which can later be revised if necessary.
- Patients should be evaluated for evidence of epidermolysis bullosa (EB) before a surgical endeavor is undertaken.