





Pathophysiology

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

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

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.

Other Problems to Be Considered

Few other conditions present with ulcerations or scars in the newborn period. Aplasia cutis congenita (ACC) must be differentiated from the erosions of epidermolysis bullosa (EB); this differentiation is especially important because aplasia cutis congenita may be diagnosed alone before new lesions from EB become apparent.

Neonatal herpes may present with an erosion, often on the scalp, but rarely at delivery. Some lesions have been attributed to fetal varicella.

Lesions are sometimes mistakenly attributed to birth trauma secondary to vacuum extraction, forceps, or fetal scalp monitor electrodes.

Setleis syndrome includes bitemporal aplasia cutis congenita with other facial cutaneous abnormalities and has been termed congenital ectodermal dysplasia of the face.

Transient bullous dermolysis of the newborn is a rare, benign, blistering disease evident at birth or shortly thereafter; this condition resolves during the first months of life leaving no dystrophic scars.

Laboratory Studies

- No specific laboratory abnormalities are consistently found in this condition. Abnormalities due to associated conditions may be present. Chromosome analysis may be indicated if a pattern of abnormalities suggests a genetic disorder.

Imaging Studies

- Imaging studies are seldom required for small circular or oval aplasia cutis congenita of the scalp with no apparent associated anomalies.
- Atypical or very large scalp defects should be imaged for possible underlying bone or soft tissue defects. In one neonate, a ventricular diverticulum of the posterior horn of the lateral ventricle was associated with aplasia cutis congenita of the scalp. It manifested as an expanding bulge of the subdural space.
- Distal radial epiphyseal dysplasia associated with aplasia cutis congenita over the dorsum of the distal forearm has been reported. The cutis aplasia was diagnosed at birth but the radial dysplasia was not recognized until presentation to an orthopedist at age 5-10 years. Radiographs of the involved extremity should be performed in cases of aplasia cutis congenita involving the distal forearm.

Other Tests

- Other tests may be indicated for evaluation of associated conditions.

Histologic Findings

The histologic features of the skin vary according to the depth of the aplasia and its duration. At birth, ulcerated lesions may show a complete absence of skin. After healing, the epidermis may appear flattened with a proliferation of fibroblasts within the connective-tissue stroma and an absence of adnexal structures. Bullous aplasia cutis congenita is a rare clinical subtype of aplasia cutis congenita with distinctive histologic findings. Histologic evaluation of such lesions reveals a distinct pattern containing fibrovascular stromas, edematous stroma, or both. Identical histologic findings are found in encephaloceles and meningoceles.

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.

Consultations

- Genetic counseling should be considered if associated anomalies are noted.
- Consultations based on other findings are appropriate to evaluate these conditions.