



APLASIA CUTIS CONGENITA

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Aplasia cutis congenita is an uncommon condition (approximately 3 in 10,000 births) where a newborn child is missing skin from certain areas of the body with a predilection for the midline of the vertex of the scalp. In about 70% of cases it is a single lesion on the scalp, but sometimes multiple lesions may appear on other parts of the body (the face, the trunk, or the limbs), sometimes symmetrically. The lesions are noninflammatory and well demarcated (circular, oval, linear, or stellate in configuration), and they range in size from 0.5-10 cm. Lesions that involve only the upper layers of skin (epidermis) are shallow and usually heal over with scarring before the child is born. A lesion involving the deeper layers of the skin (dermis, subcutaneous tissue), or rarely, the skull may be ulcerated.

Membranous aplasia cutis is the term used when there is an underlying flat, white membrane, which overlies a defect in the skull. It can be associated with a neural cranial tube defect (encephalocele or meningocele), which can be demonstrated by ultrasound scan showing misplaced brain tissue outside the skull. The "hair collar sign" refers to a ring of long, dark hair commonly encircling the lesion of membranous aplasia cutis.

Bullous aplasia cutis congenita demonstrates a fibrovascular or edematous connective tissue similar to that seen in encephaloceles and meningoceles, suggesting it may also be related to a neural tube defect. Focal preauricular dermal dysplasia is a form of aplasia cutis congenital occurring in front of the ear. It is not typically associated with any extracutaneous anomalies. The SCALP syndrome is a nevus sebaceus syndrome with CNS malformations, aplasia cutis congenita, limbal dermoid, and a giant congenital pigmented melanocytic nevus with neurocutaneous melanosis.

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, and 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 an incomplete form of a neural tube closure defect.

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. The decision to use medical, surgical, or both forms of therapy in aplasia cutis congenita depends primarily on the size, depth, and location of the cutaneous defect and therapy of associated defects as indicated.

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