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Aplasia cutis congenita and associated disorders: an update

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We present an update of disorders in which aplasia cutis congenita is a feature. Localization of the lesion, important other features, and possible etiology are tabulated. Disorders are classified as chromosomal, monogenic, teratological/exogenous, and unknown. Points of particular interest in history taking and examination of patients with aplasia cutis congenita are presented.

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Aplasia cutis congenita (ACC) is defined as congenital localised absence of skin. Cordon described the first case in 1767. More than 500 cases have been reported (Gorlin et al. 1990).

Several etiological theories have been postulated. The amniogenic theory proposed that ACC is caused by adhesion of the amniotic membrane to the fetal skin. However, in most cases examination of the placenta did not show any abnormality (Demmel 1975). The vascular theory assumed thromboplastic material from a fetus papyraceus (Lemke et al. 1993) or placental abnormalities to be the main cause (Levin et al. 1980). Biomechanical forces on the vertex during the embryogenesis (Stephan et al. 1982), varicella (Jones 1988) and herpes simplex (Harris et al. 1986) infections, intrauterine involution of a hemangioma (Pozzati et al. 1983) have all been suggested as causes of ACC. It may also be the result of the teratological action of medicaments, like methimazole (Frieden 1986) and misoprostol (Fonseca et al. 1993). High doses of vitamin A in rats (Kjaer 1968) and triamcinolone acetonide in non-human primates (Hendrickx et al. 1980) resulted in ACC in offspring. Genetic factors (Aknin et al. 1992) play an important role in the development of ACC.

Clinically, ACC presents as an ulcer or a transparent membrane through which the underlying structures are visible. The location is usually on the scalp; less frequently the trunk and limbs are involved (Demmel 1975).

ACC occurs as an isolated defect, or with one or more other congenital anomalies as part of a syndrome, sequence or association.

Several classifications of this heterogeneous disorder have been proposed (Demmel 1975, Sybert 1985, Frieden 1986, Küster & Traupe 1988, Gorlin et al. 1990).

In this article we give an up-to-date list of disorders with ACC according to etiology (Table 1).

Localisation of the lesion and important features are mentioned. Finally, we present points of particular interest in history taking and examination of patients with ACC (Table 2).

For the research into the literature, the following resources have been used: CD-Rom on-line, CD-Rom Silver Platters, POSSUM (Pictures of Standard Syndromes and Undiagnosed Malformations) and Oxford Medical Databases (Neurogenetics, Dysmorphology and Cytogenetics).

Discussion

ACC features in a heterogeneous group of disorders which we classified into four groups. The monogenic group is composed of autosomal dominant,

Table 1. Disorders associated with aplasia cutis congenita

	References	Localisation ACC		Important other features	Remarks
		Scalp	Body		
I. CHROMOSOMAL					
Del (4p) syndrome Wolf-Hirschhorn syndrome	Schinzel (1984)	midline		ocular hypertelorism and broad beaked nose, mental retardation, microcephaly and/or cranial asymmetry, low-set ears with preauricular dimple	±35% ACC
Tetrasomy 12p Pallister-Killian syndrome	Zakowski (1992)		axilla, lateral neck	coarse facies, mental retardation, cardiovascular anomalies, pigmentary skin anomalies, alopecia areata, supernumerary nipples	demonstrated in fibroblasts, seldom ACC
Trisomy 13 Patau syndrome	Jones (1988)	vertex		microphthalmia, cleft lip and palate, holoprosencephaly, polydactyly, narrow hyperconvex nails	±50% ACC
II MONOGENIC					
IIa *AUTOSOMAL DOMINANT*					
Adams-Oliver syndrome, ACC with terminal transverse limb defects	Koiffmann (1988) Whitley (1991)	vertex	one case also on knee	asymmetric terminal limb malformations, cutis marmorata telangiectatica, occasionally supernumerary nipples, tortuous scalp veins	Mc Kusick 100300 Koiffmann 1988: autosomal recessive high% met ACC, expression highly variable
Autosomal dominant ACC	Guillen (1985) Rauschkolb (1962)	+	+		Mc Kusick 107600 reduced penetrance
Ectrodactyly-ectodermal dysplasia-clefting syndrome, EEC-syndrome	Johnson (1976) Jones (1988)	+		ectrodactyly, ectodermal dysplasia, cleft lip and palate, absent breasts, renal anomalies	Mc Kusick 129900 seldom ACC
Ectodermal dysplasia (TOOD)	Tuffli (1983) Salinas (1988)	vertex		TOOD: Tricho-Odonto-Onychodysplasia and Dyshydrosis, nipple/breast hypoplasia	Mc Kusick 129550
Scalp-Ear-Nipple syndrome (SEN syndrome)	Finlay (1978)	+		ear malformations (rudimentary tragus and antitragus) rudimentary nipples	Mc Kusick 181270
Ear malformations, unilateral n. VII paresis and dermal sinus	Anderson (1979) Singman (1990)	+		ear malformations: 'lop ears', unilateral n. VII paresis, dermal sinus	Mc Kusick 134100
Postaxial polydactyly	Buttiëns (1985)	vertex		postaxial polydactyly, encephalocele (one case)	Mc Kusick 181250
IIb AUTOSOMAL RECESSIVE					
Autosomal recessive ACC	Stephan (1982)	+	+	abnormal low set ears, prematurity, small nose, micrognathia, arthrogryposis, renal-, gastrointestinal-, and brain anomalies	Mc Kusick 207700
Intestinal lymphangiectasia	Bronspiegel (1985)	vertex		intestinal lymphangiectasia, simian creases	Mc Kusick 207731
Johanson-Blizzard syndrome	Rüdnik-Schöneborn (1991)	+		aplasia of the alae nasi, dental anomalies, pancreatic insufficiency, deafness, mental retardation	Mc Kusick 243800
Setleis syndrome "bitemporal forceps marks", focal facial dermal dysplasia	Clark (1989) Artlich (1992)	temporal		double upper eyelashes, absent lower eyelashes, "coarse" face, hypoplastic lateral eyebrows, prominent thick lips	Mc Kusick 227260 high% ACC Artlich 1992: autosomal dominant
Ectodermal dysplasia-clefting syndrome	Bowen (1976)	+	+	genital hypoplasia, hyperpigmentation, mental retardation, skeletal anomalies	Mc Kusick 225000
Familial 46 XY-gonadal dysgenesis	Brosnan (1980)	+		'streak gonads", renal anomalies, VSD, mild mental retardation, short stature, sturdy neck, facial anomalies	Mc Kusick 233430
IIc X-LINKED					
Goltz-Gorlin syndrome focal dermal hypoplasia (FDH)	Wechsler (1988)	vertex parietal	thoraco-lumbal	linear poikiloderma, localized lipomatous herniations, angiofibromas of mucous membranes, nail dystrophy, hypoplasia of teeth, ophthalmologic anomalies: coloboma, ear malformations	Mc Kusick 305600 XL dominant ±15% ACC lethal in males

Cont.

Table 1. Continued

MIDAS syndrome	Happle (1993)	face, scalp, neck	upper part of thorax	Microphthalmia, Dermal Aplasia and Sclerocornea sometimes agenesis of corpus callosum or congenital heart defect	the lesions follow Blaschko-lines, X-functional mosaicism, XL dominant, lethal in males
III TERATOLOGICAL/EXOGENOUS					
Alcohol and heroin		+		see Spear-Mickle syndrome (IVa)	
Alcohol and cocaine		+		see Singman (IVa)	
Marijuana		+		see thrombosis (IVb 2)	
Methimazole/Carbimazole	Dutertre (1991)	+		sometimes urachal malformations	antithyroid drugs
Misoprostol	Fonseca (1993)	+			PGE_1 analogue as abortifacient
Congenital herpes simplex infection	Harris (1986)	various	+	signs of congenital herpes simplex infection	maternal primary HSV^1-infection
Congenital varicella infection	Baillie (1983), Jones (1988)	various	+	mental retardation, chorioretinitis, limb hypoplasia	maternal primary VZV^2-infection
Foetus papyraceus	Lemke (1993)	+	symmetric +	placenta with foetus papyraceus	dragged thromboplastic material from the dead foetus
Placental anomalies	Levin (1980) Munkvad (1981)	+	+	infarction of placenta, single umbilical artery (one case), macerated and bleeding umbilical cord (one case)	
Amniotic band disruption complex	Higginbottom (1979)	various	various	depending upon timing of amniotic rupture various malformations (limb amputation, cleft lip-palate, omphalocele)	cause of amniotic rupture unknown, low% ACC
IV UNKNOWN					
IVa "MULTIPLE'					
Delleman syndrome oculo-cerebro-cutaneous syndrome	Giorgi (1989) Clericuzio (1990)	above the ears		focal dermal hypoplasia, skin appendages, orbital cyst, microphthalmia, coloboma of eyes, cerebral malformations (e.g. multiple intracranial cysts)	autosomal dominant mutations?
Epidermal naevus syndrome and ACC, linear sebaceous naevus syndrome	Fryburg (1993)	+		epidermal naevus syndrome (=epidermal naevus associated with extracutaneous anomalies)	low% ACC
Greig	Greig (1931)	+		cleft lip-palate, polydactyly, renal anomalies	
Halper	Halper (1991)	+		syndactyly, supernumerary nipples	autosomal dominant? Possibly Adams-Oliver syndrome
Kosnik	Kosnik (1975)	midline		choanal atresia, imperforate anus, laryngomalacia, ambiguous genitalia, congenital hip dislocation	
Leichtman	Leichtman (1990)	vertex		generalised ectodermal dysplasia, thin breakable hair, hypohidrosis, prefrontal lipoma, epicanthus, hypoplastic midface, broad nasal bridge	dominant, autosomal or X-linked, possibly form of TOOD.
Mimouni	Mimouni (1986)	+		epidermal naevus syndrome, giant pigmented naevocellular naevus, chondroma, conjunctival lipodermoid, naevus flammeus, brain anomalies	
Sakati syndrome	Cohen (1986)	above the ears		acrocephalopolysyndactyly, short limbs, heart defects, ear anomalies	low% ACC
Singh	Singh (1987)	+		dextrocardia, cleft lip and palate, low set ears, depressed nasal bridge, short neck, upward slanting of eyes, ridged metopic sutures, hydrocele, talipes equinovarus	
Singman	Singman (1990)	+		infratentorial arteriovenous fistula, prominent scalp veins, depressed nasal bridge, flattened philtrum, high-arched palate	cocaine and alcohol abuse during pregnancy
Spear-Mickle syndrome	Spear (1983)	+		cranial stenosis, scalp asymmetry, several soft-tissue tumors in the frontal area of the scalp, bifid nose, small lumbar meningocele	heroin and alcohol abuse during pregnancy

Cont.

Table 1. Continued

Stevenson	Stevenson (1993)	+		short stature, myopia, delayed dentition, facial dysplasia	
Toriello	Toriello (1993)	+		epibulbar dermoids, macrocephaly, hyperpigmentation	
IVb "SINGLE'					
1 Central nervous system					
ACC and Morbus Charcot-Marie-Tooth (HMSN I)	Castle (1992)	+		HMSN type 1	XL recessive? seldom ACC
Holoprosencephaly	Kurokawa (1990)	midline		Dandy-Walker cyst	
Hydranencephaly	Fowler (1973)	vertex	seldom		
Hydrocephalus	Ingalls (1933)	+			
Leptomeningeal angiomatosis and cerebral aneurysm	Pozzati (1983)	in the region of the 1st division of n.V		ipsilateral parieto-occipital leptomeningeal capillary venous angioma, giant aneurysm of the ipsilateral distal posterior cerebral artery	possibly atypical Sturge-Weber syndrome
Spastic paralysis and mental retardation	Ruiz-Maldonado (1974)	midline	symmetric back, belly, limbs	spastic paralysis, especially of lower limbs	
Neural tube defects:					
Craniospinal rachischisis	Greig (1931)	+		congenital brevicollis	
Meningocele	Ingalls (1933)	+			see also Spear-Mickle syndrome
Occult spinal dysraphism	Higginbottom (1980)		lumbosacral		
Tethered cord	Pauli (1987)	+	lumbosacral		
2 Cardiovascular system					
ACC and coarctatio-aortae (CA) ACCCA-syndrome	Dallapiccola (1992)	vertex			CA in 4 generations (autosomal dominant), ACCCA: mother and son
Arteriovenous malformation	Vasconez (1973)	midparietal		short, stubby distal phalanges	proband and her mother: chromosome 16–18 defect
Patent ductus arteriosus	Deeken (1970)	temporal			
Ventricle septal defect/pulmonary stenosis	Paltzik (1985) Dubosson (1978)	vertex			
Thrombosis of the sagittal sinus	Lavine (1978)	+			marijuana abuse during prenancy
3 Gastrointestinal system					
Cleft lip-palate	Kosnik (1975)	+			
Omphalocele	Argenta (1986)	+			
Tracheoesophageal fistula	Deeken (1970)	midline			
4 Miscellaneous					
Cutis marmorata	Voirin (1992)	vertex			no limb anomalies
Closure defects of ventral body wall and/or neural tube	Hoyme (1989)	vertex	sternal	partial sternal agenesis/long agenesis	

[1] Herpes Simplex Virus.
[2] Varicella Zoster Virus.
+=localisation not further specified.

autosomal recessive, and X-chromosomal inherited syndromes, respectively. The unknown group is subdivided into "multiple" and "single". "Multiple" implies, in addition to ACC, two or more other congenital defects, and "single" one other congenital defect.

The lesions in epidermolysis bullosa (Kanzler & Smoller 1992), in Knobloch syndrome (Seaver et al.

Table 2. Important issues in history taking and examination in patients with ACC

History
- ▶family history: cutaneous defects, limb defects
- ▶pregnancy:
  - medication (antithyroids, misoprostol)
  - infections (varicella, herpes simplex)
  - intra-uterine trauma
  - placenta: amniotic bands, infarctions, color (pale), single umbilical artery
  - presence of fetus papyraceus
  - birth trauma

Physical examination
- ▶description and localisation of the lesion
- ▶other ectodermal anomalies, like
  - teeth: hypoplasia, delayed dentition
  - nails: hypoplasia
  - skin: blisters, hyperpigmentation, cutis marmorata
- ▶central nervous system: mental retardation, neural tube defects, paresis
- ▶ophthalmologic anomalies: coloboma, myopia, epibulbar dermoid
- ▶anomalies of limbs, ears, nipples, heart

Other investigations on indication
- ▶imaging:
  - roentgenograms of hands and feet
  - roentgenograms of scalp
  - cerebral CT-scan or MRI
- ▶skin biopsy: light and/or electron microscopy
- ▶karyotyping
- ▶culture/serology: varicella zoster and herpes simplex virus
- ▶examination of family members

1993), in atrophic alopecia of Leung (Leung et al. 1988), in the branchio-oculo-facial syndrome (Fujimoto et al. 1987), and in perinatal trauma are not included in Table 1 because we do not consider these as ACC.

This updated list of disorders with ACC, and the points for special attention in history taking and examination, may serve as a tool in the diagnostic process and genetic counseling of patients with ACC.

Note added in proof

Jewell et al. (1994) have recently reported a possible new autosomal dominant genetic syndrome consisting of cutis aplasia, alopecia areata, ear anomolies and perianal skin tags. Hubert et al. (1994) have reported an infant with ACC of the scalp who was exposed to valproic acid *in utero*.

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