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Short Communication

Bazex-Dupré-Christol syndrome (BDCS)

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Identity

Other names

Bazex syndrome
Follicular atrophoderma and basal cell carcinomas

Note

Bazex-Dupré-Christol syndrome (Bazex et al., 1964; Bazex et al., 1966) should not be confused with acrokeratosis paraneoplastica/Bazex syndrome, a paraneoplastic syndrome characterized by psoriasisform cutaneous lesions with acral distribution (fingers, toes, nose and ear), most often associated with a squamous cell carcinoma of the upper aerodigestive tract.

Inheritance

Bazex-Dupré-Christol syndrome is an X-linked dominant genodermatosis, with a prevalence below 1/1 000 000.
The female to male ratio reaches the expected 2:1, and females appear to be less affected than males.
There is intrafamilial and interfamilial phenotypic variability (Lacombe and Taïeb, 1995; Kidd et al., 1996).
Most cases are European patients. Oley syndrome (Oley et al., 1992) may be a variant of Bazex-Dupré-Christol syndrome (Vabres and de Prost, 1993; Kidd et al., 1996).
It has been suggested that Bazex-Dupré-Christol syndrome is a disorder of the hair follicle; it can be considered an ectodermal dysplasia, an heterogeneous group of inherited disorders resulting from abnormal organogenesis of the skin and its appendages (Castori et al., 2009).

Clinics

Phenotype and clinics

The phenotype, reviewed by Kidd et al., 1996 on about 120 cases, associates:
- Follicular atrophoderma (ice pick depressions of the skin) of the dorsa of hands and feet, extensor areas of the elbows and knees, and face, presenting at birth or developing early in life.
They are discrete areas of dilated hair follicules. It is found in 100% of the cases.
- Congenital hypotrichosis, found in 90% of the cases, with hair shaft abnormalities such as pili torti and trichorrhexis nodosa with spontaneous breakage, jagged or absent cuticular scales.
- Hypohidrosis, either generalized or confined to the face, found in 60% of the cases.
- Multiple milia, predominating on the face, reported at birth in some cases, and developing during childhood in other cases; found in 90% of the cases.
- 'Pinched' nose with hypoplastic alae and prominent columella (Kidd et al., 1996).
- And early onset of basal cell carcinomas, mainly confined to the face.
- Trichoepitheliomas can also be found (Castori et al., 2009).
Trichoepitheliomas derive from the trichoblast (i.e. the folliculo-sebaceous-apocrine germ).
They are small skin-colored papules or nodules, with nests of basaloid cells forming cysts containing horn cells (with keratin) (Lee et al., 2005).

- Sweet glands have been reported to be absent in 9 cases in a study of 11 patients (Kidd et al., 1996). Comedones and hidradenitis suppurativa, flexural hyperpigmentation can also be found (Castori et al., 2009; Castori et al., 2012).

Differential Diagnosis

The main differential diagnoses are :
- Gorlin syndrome, an autosomal dominant disease associating multiple basal cell carcinomas, jaw keratocysts, dyskeratotic palmar/plantar pits and skeletal malformations;
- Rombo syndrome, a probably autosomal dominant disease that presents with cyanosis, vermiculate atrophoderma of the face and sun-exposed areas, telangiectasia, milia-like papules, and basal cell carcinomas;
- Generalized basaloid follicular hamartoma, an autosomal-dominantly inherited disorder associating disseminated milia, palmoplantar pits, hypotrichosis and basaloid follicular hamartomas (Wheeler et al., 2000);
- Schöpf-Schulz-Passarge syndrome, an autosomal recessive disease in most cases (with a genetic heterogeneity with a possible dominant variant), characterized by multiple eyelid apocrine hidrocystomas, palmo-plantar keratoderma, hypodontia, hypotrichosis and nail dystrophy (Castori et al., 2008).

Neoplastic risk

Basal cell carcinomas (BCC) develop in the second or third decade of life in patients with Bazex-Dupré-Christol syndrome (BDCS), mostly in the sun-exposed areas of the head and neck. Age at onset varies from 3 years (Abuzahra et al., 2012) to 50 years. 3 out of 4 BDCS patients will experience BCCs. Predisposing inherited diseases with an increased risk of BCCs are the following: Bazex-Dupré-Christol, Gorlin, Rombo syndromes, Xeroderma Pigmentosum, Bloom, Werner, Rothmund-Thomson and Muir-Torre syndromes, Brooke-Spiegler, Schöpf-Schulz-Passarge and Cowden syndromes, cartilage-hair hypoplasia and epidermodysplasia verruciformis, oculocutaneous albinism and Hermansky-Pudlak syndrome, and some epidermal nevus syndromes (review in Parren and Frank, 2011; Castori et al., 2012).

Genes involved and proteins

Note

The gene involved in BDCS is still unknown. It has been mapped to Xq25-27.1 (Vabres et al., 1995; Parren et al., 2011).

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