CASE REPORT

Anhidrotic ectodermal dysplasia syndrome in the neonatal period - case report

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Abstract

Objective: to describe a rare syndrome affecting children, and to urge pediatricians to consider this diagnosis when investigating idiopathic fever in neonates.

Methods: we report a case of anhidrotic ectodermal dysplasia syndrome in a 10 day old newborn in the neonatal intensive care unit at Hospital Geral de Caxias do Sul. The child presented recurrent episodes of fever since the first days of life.

Results: the patient presented dry mucus, dry skin, and fever. Skin biopsy was performed in the child’s back. Absence of sweat and sebaceous glands as well as hypoplasia of follicular structures were identified. The patient is receiving adequate treatment, and is being followed as an outpatient.

Conclusion: anhidrotic ectodermal dysplasia syndrome is a rare disorder that must be considered when investigating newborns with recurrent episodes of fever. During the neonatal period, the clinical manifestations of the disease are subtle and unspecific. Such findings become more visible after a few months of life. There is no definitive treatment for this syndrome.


Introduction

Ectodermal dysplasias are a heterogeneous group of hereditary diseases characterized by the presence of abnormalities in tissues and organs of ectodermal origin including skin, hair, nails, teeth, and eccrine glands.1,2 The hypohidrotic and anhidrotic forms are classical examples of this type of dysplasia.1

The first cases of ectodermal dysplasia involving skin, hair, and teeth were described in 1848 by Thurman.3-5 Moreover, Weech was the first to employ the term anhidrotic ectodermal dysplasia for patients with absence of sweat glands.4

Anhidrotic or hypohidrotic ectodermal dysplasia, or Christ-Siemens-Touraine syndrome, is a recessive, X-linked disease; it is also rare and nonprogressive and presents a triad of partial or total absence of sweat glands, hypotrichosis, and hypodontia.5 In addition, there are other signs and symptoms that can be found depending on the involvement of the ectodermal tissue.4 Most cases of total Christ-Siemens-Touraine syndrome involve male patients (more than 90%); female patients usually present the asymptomatic form of the disease.5
It is our objective to report a case of anhidrotic ectodermal dysplasia and to underscore the importance of considering this pathology in differential diagnosis of fever of unknown origin in infants.

Case report

A male newborn with 10 days of life from the city of Caxias do Sul, state of Rio Grande do Sul, southern Brazil, was admitted to the Neonatal ICU of the Teaching Hospital at the Universidade de Caxias do Sul (UCS) on August 20th, 1998. Patient presented with history of episodes of persistent hyperthermia since the first days of life. At physical examination, ES presented with dry mucous membranes, dry and desquamative skin, hyperthermia (39ºC), and umbilical granuloma.

In order to investigate the origin of the fever we carried out chest X-ray, hemogram, hemoculture, urinalysis, urine culture, and liquor examination. The exams presented no alterations. Next, an umbilical stump culture was collected for suspected diagnosis of omphalitis, which also presented negative. We started empiric therapy with oxacillin and gentamicin despite patient presenting normal for the examinations carried out.

On August 25th, 1998 patient peak temperatures persisted and the above examinations were carried out for a second time; once more, results were negative. We replaced oxacillin and gentamicin with vancomycin, amicacin, and cefotaxime.

On the 14th day of hospital stay the patient still did not present any improvement of clinical status. Consequently, we carried out exams to rule out HIV and congenital neonatal infection (STORCH). Again, patient presented negative for both exams. He also presented normal for ultrasonography examination of the abdomen.

Only on September 9th of that same year we suspected that the patient had anhidrotic ectodermal dysplasia. Biopsy of a specimen taken from the dorsum of the patient and histopathological report confirmed the diagnosis. Specimen submitted to anatomopathological exam indicated absence of eccrine and sebaceous gland structures and hypoplasia of follicular structures (Figure 1).

Patient was discharged from the hospital on September 12th, 1998. The mother was instructed regarding procedures for control of temperature and use of emollient for dry skin.

One year later, the clinical signs of anhidrotic ectodermal dysplasia were more evident. At physical examination, patient presented typical facies with frontal boss, small nose, lip protrusion, erythematous malar region rash, and rhinorrhea. Patient also presented hypotrichosis; thin and sparse hair; depigmentation of the hair; and sparse eyebrow hair. The skin was dry, pale, thin, and with protruding vessels (figures 2 and 3).
Anhidrotic ectodermal dysplasia is a disease with genetic heterogeneity. Many types of inheritance have been described, including those of dominant and recessive autosomal forms of the disease and of X-linked inheritance. In general, the mechanism of recessive inheritance is linked to the X chromosome; as a result, male patients are more seriously affected by the disease, presenting significant morbidity and mortality. Heterozygote women can be affected differently, ranging from being asymptomatic carriers to presenting clinical manifestation identical, or similar to, that of men.

The characteristic clinical manifestations of anhidrotic ectodermal dysplasia are absent or reduced sweat, hypotrichosis, total or partial anodontia, and typical facies.

In anhidrotic ectodermal dysplasia, the hypotrichosis is generalized and the alopecia is not complete. The hair presents thin, sparse, dry, and with depigmentation. In addition to the scalp, other areas of the body can also be affected, such as the axillae, pubic region, face, and thorax. The eyebrow can be completely absent or sparse; the eyelashes, in turn, remain normal. Anhidrotic ectodermal dysplasia patients tend to have pale, dry, and translucent skin with thin wrinkle formation and characteristics of early aging.

Dental abnormalities include anodontia or hypodontia and odontodystrophy (disorder of the format and size of teeth). Usually, the incisor and/or canine teeth are small, conoid, and pointed. Moreover, onychodystrophy occurs in half of the cases of anhidrotic ectodermal dysplasia.

A classification for congenital ectodermal dysplasia was suggested by Freire-Maia and colleagues of the Center of Studies on Ectodermal Dysplasia from the Department of Genetics at Universidade Federal do Paraná. The classification is based on the phenotype of each group of the disease. The hair, tooth, nail, and sweat gland structures are designated as 1, 2, 3, and 4, respectively. According to the abnormalities found in each structures, it is possible to establish subgroups: 1-2; 1-3; 1-4; 2-3; 2-4; 3-4; 1-2-3; 1-2-4; 1-3-4; 2-3-4; 1-2-3-4.

The deficiency or absence of mucosal glands, especially in the upper respiratory tract, oral cavity, and nasopharynx usually develops to rhinitis, pharyngitis, laryngitis, bronchitis, and otitis media. Rhinorrhea occurs conversely to absence of mucus.

Less frequently, it is also possible to find cases of palmar-plantar hyperkeratosis; cleft palate; susceptibility to atopic dermatitis; abnormalities of the eye; abnormalities of the skeleton and gonad; absence or hypoplasia of mammary glands; and mental retardation.

Children with anhidrotic ectodermal dysplasia present persistent hyperthermia that manifests itself especially following the practice of physical exercise, the appearance of infections, or the use of inappropriately warm clothing. This occurs due to absence of eccrine glands, which, in turn, results in anhidrosis.

Early diagnosis of the disease is rare since its clinical alterations are not manifest at the moment of birth. The final diagnosis is obtained with histopathological exam of the skin. Currently, it is also possible to carry out prenatal diagnosis with studies of DNA.

One of the main obstacles that doctors have to face in relation to this pathology is the lack of a specific treatment. Therapeutic measures focus basically on the control of the temperature of the patient, use of air-conditioning, use of light-clothing, cold showers, practice of exercise in cold environments, and limitation of physical activities. The use of emollient for dry skin is also recommended. The only treatment available is the dental treatment with use of prostheses, which can help improve patient nutrition and physical appearance. Moreover, plastic surgery can be carried out in adolescent and adult patients in order to correct deformities.

Despite the fact that the congenital anhidrotic ectodermal dysplasia is a rare disease and with late typical clinical alterations, we conclude that it is necessary to include it in the differential diagnosis of newborns with persistent fever of unknown origin. In this sense, it would be possible to establish early diagnosis of the disease and adequate treatment, thus avoiding unnecessary empirical use of antibiotics and other pharmaceuticals in cases of persistent fever of unknown origin in newborn patients.
References


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