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CASE REPORTS

CUTIS MARMORATA TELANGIECTATICA CONGENITA - A CASE REPORT

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ABSTRACT

Cutis marmorata telangiectatica congenita, is a rare disease, present at birth, which has a strong visual impact, especially if the form of the disease is severe.[1] The case report presents a newborn with a generalized form of the disease.

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cutis marmorata telangiectatica congenita, newborn, skin disease.

Introduction

Cutis marmorata telangiectatica congenita, is a rare disease, present at birth, which has a strong visual impact, especially if the form of the disease is severe.[1] The causes of the disease are unknown, but it is thought to be due to a genetic mutation.[1] Literature studies report less than 300 such known cases.[4] The incidence of the disease may be underestimated, as some cases are mild forms of the disease or are evolving.[5] Cutis marmorata telangiectatica congenita may improve with age, but rarely disappears completely.[5] The evolution of the disease can be with appearance of new ulcerations.[5]

Case Report

Female newborn, rank 4, delivered by caesarean section for the triple scar uterus, in cranial presentation, with Apgar score 9 to 1 minute and 9 to 5 minutes. The estimated gestational age was 35 weeks, because the pregnancy was not followed up. At birth, the weight was 1450 g, the length 38 cm and the cranian perimeter 29 cm. Lubchenco's curves was the one of a premature newborn with a 35-week-old gestational age, small for gestational age.

Heredo-collateral antecedents did not show pathological aspects. The parents are young, the mother is 23 years old and the father is 34 years old, affirmatively healthy. The other three children born: an 8-year-old boy, a 6-year-old boy and a 4-year-old girl are affirmatively healthy, but they do not have the same biological father as this newborn.

At the objective clinical examination, it was found that the newborn showed fixed patches of mottled skin on the whole body, alternating red-purple areas with pale areas, an aspect that did not change with the warming of the body (Figure 1,2). Moreover, prominent veins and telangiectasias were present. At the level of the scalp, the newborn presented blood vessels with varicose veins and left parietal ulceration area (Figure 3). All these aspects reveals the diagnosis of cutis marmorata congenital telangiectatica congenital.

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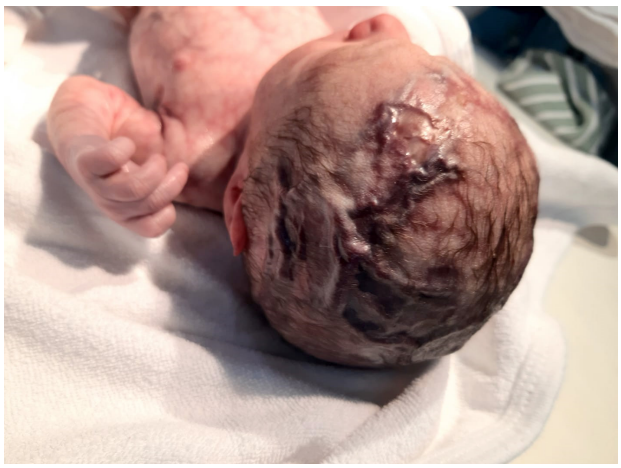
Figure 1. The newborn in the first minutes of life.

Figure 2. The newborn in the first minutes of life.

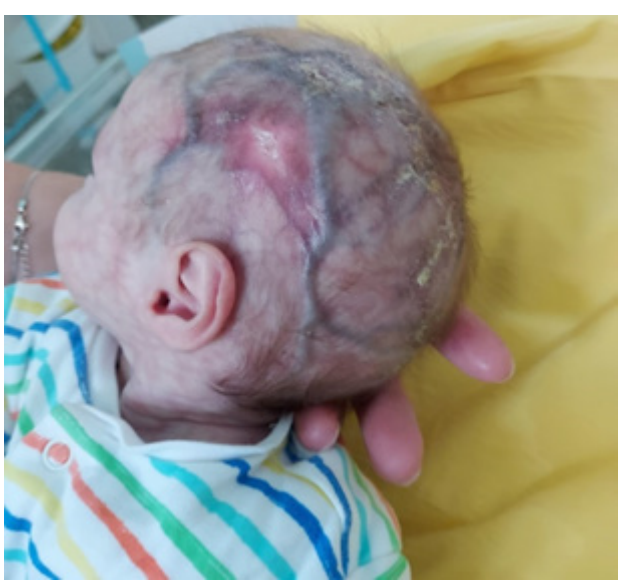
Figure 3. Appearance of the cephalic extremity with varicose vein blood vessels.



The care of the newborn immediately after birth was performed in the Neonatal Intensive Care Unit, to sustain the neonatal transition related to prematurity and related to the fact that the newborn was small for gestational age.

The evolution of the newborn was satisfactory. At the skin level, the clinical appearance remained unchanged. At the level of the scalp, the ulcerative lesions evolved towards the crustal stage, but in parallel other new ulcerative lesions appeared. (Figure 4).

Figure 4. Picture of the newborn at discharge.



At the corrected age of 39 weeks, at 4 weeks after birth, an ophthalmological consultation was performed, as the cutis marmorata telangiectatica congenita is known to be associated with ocular diseases. Ophthalmological 1,2 examination: revealed fibro-vascular band, centered on the head of the optic nerve with anterior retinocrystalline disposition in the right eye and in the temporal area in the left eye, with traction of the adjacent retina (through the macular area) and retinal pigment changes. The recommendation was a control examination after 1 month and also an examination at a department specialized in in vitro-retinal surgery.

Among other investigations the transfontanelar ultrasound at the age of 4 weeks postnatal is also relevant, as it revealed a grade 3 haemorrhage in resorption.

Discussion

Cutis marmorata telangiectatica congenita is a rare condition.[1] The causes of the disease are not known, but it is considered that it may be a genetic mutation.[3] The clinical exam of the skin reveals telangiectasias, skin atrophy, ulcerations and hyperkeratosis. The presence of atrophy, ulcers and lesions with strict localization and demarcation, allow the differential diagnosis with physiological cutis marmorata.[1]

The positive diagnosis is specified only by clinical examination. Cutaneous biopsy is not necessary because histologically there are no specific changes.[2]

Cutis marmorata telangiectatica congenita may be associated with extra-cutaneous damage.[5] Asymmetric and/or shortening limb abnormalities, retinal detachment, glaucoma, leukocoria and central nervous system abnormalities with developmental delay may be present.[1,5]

As a treatment for the disease, there is no specific treatment. Treatment for pain in areas affected by ulcers is considered.[1,2] After home discharge, there is a minimal 3 years follow-up needed, for skin and skeletal development (limb measurements) monitoring, as well as monitoring of neurological development. A multidisciplinary approach is needed.[4]

In the case of our patient, the form of the disease present at birth was severe, with generalized skin damage. The medical literature states that 24.5% of patients have a generalized form of the disease.[2,3] No limb abnormalities were identified and no skeletal / bone abnormalities such as syndactyly, congenital club foot, palatoschisis were seen. At the clinical examination only a slight ocular asymmetry was noted. Although glaucoma is mentioned in the first place in ophthalmological associations, which occurs in 4.9% of cases[3], it was not present in our patient, instead there is a risk of retinal detachment.

Conclusion

Cutis maramorata telangiectatica congenita, the generalized form that affects the entire skin is a condition with an emotional impact on parents, especially on the mother. Continuous monitoring in the first years of life is important. In our case, the greatest risk is of ophthalmic complications (retinal detachment). Furthermore, in the absence of proper care, there is a risk of infectious complications of ulcerative lesions on the scalp. Cutis marmorata telangiectatica is a condition that requires a multidisciplinary approach.

Compliance with Ethical Standards

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Conflict of Interest None

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