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CASE REPORT

Newborn with cutis marmorata telangiectatica congenita: a case report and review of the literature

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ABSTRACT

Cutis marmorata telangiectatica congenita (CMTC) is a rare vascular disorder characterized by persistent marbled erythema, vascular telangiectasia, and skin atrophy, with unknown etiology and prevalence. Associated congenital abnormalities occur in up to 80% of patients. Here, we presented a case of female neonate, born at 42+1/7 weeks of gestation, who presented with bluish-red skin discoloration, marbled erythema, telangiectasias, and cutaneous atrophy. The newborn was transferred on the first day of life to our center due to suspected vascular malformations and for differential diagnosis. After admission to department, medical examination, imaging, and laboratory tests were performed. Clinical presentation was clearly suggestive of CMTC. In order to exclude other anomalies, multispecialty consultations were recommended. The newborn was discharged home on day 14th of life. Suspected CMTC requires thorough differential diagnosis. Patients with CMTC require regular follow-up visits to detect possible subsequent complications in the early stages of life.

KEY WORDS:

telangiectasis, newborn, vascular malformations, cutis marmorata telangiectatica congenita.

INTRODUCTION

Cutis marmorata telangiectatica congenita (CMTC) is a rare vascular anomaly involving capillaries and venules [1]. The etiology and frequency remain unknown, with approximately 500 cases reported [2, 3]. CMTC is mainly characterized by persistent reticulated marbled erythema, skin atrophy, and ulcerations [4]; the diagnosis is based on the clinical findings. Due to the way of disease identification, the incidence of CMTC may be underestimated [5]. Associated congenital abnormalities may occur in up to 80% of patients [6]. Due to the similarity of CMTC's clinical picture with other conditions, a thorough differential diagnosis is necessary [7].

CASE REPORT

We presented a case of a female neonate born at 42 + 1/7 weeks of gestation (gravida 2, para 2) at a secondary referral center. The girl was delivered through caesarean section due to mother's disagreement for induction of labor, and scored 10 points on the Apgar scale. The child's birth weight, length, and head circumference were 3,310 g, 55 cm, and 35 cm, respectively. The course of pregnancy was uncomplicated; the mother had a history of well-controlled hypothyroidism treated with levothyroxine, 75 µg daily dose. Also, there was no parental consanguinity or birth defects in the family. The newborn presented with bluish-red skin discoloration and lesions in the form

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FIGURE 1. Photographs of the newborn: the day of admission to our department (left, center), and the day of discharge (right)

of reticulated marbled erythema, venectasias, telangiectasias, and cutaneous atrophy, found on the trunk, limbs, and scalp. No other abnormalities were observed at birth. The baby's vital signs appeared stable, with saturation of 96–98%, heart rate (HR) of 145–167 beats per minute (BPM), and body temperature of 37.1°C. Likewise, the results of neonatal blood gas analysis and blood test were within normal limits, with no signs of infection in laboratory tests. Arterial pulses were palpable in both lower extremities. Due to suspected congenital vascular malformations and the need of differential diagnosis, the patient was transferred on the first day of life to our center (a tertiary referral hospital).

After admission to the neonatology department, CMTC was suspected. Differential diagnosis included port-wine stains, Adams-Oliver syndrome, Klippel-Trénaunay syndrome, Bockenheimer's syndrome, and neonatal lupus. Abdominal, transtemporal, and cardiac ultrasounds were performed, with no abnormalities found. A blood test revealed slightly increased level of lactate dehydrogenase (LDH) of 12 μkat/l. In the peripheral blood smear, parameters, such as hemoglobin, hematocrit, platelets, and white blood cells levels were within normal limits. The infant was breastfed, and weight gain was steady. No dysfunctions of the digestive and urinary systems were detected. Although the infant developed jaundice, there was no indication for phototherapy (transcutaneous bilirubin level of 188.1–205.2 μmol/l); heart rate of 150 BPM (occasionally, sleeping HR was 85 BPM). On day 2 of life, the newborn was examined by a geneticist, who observed symmetrical lesions affecting the whole body, except the palms, soles, and mucous membranes. Also, ulcerations or skeletal abnormalities were not found. The absence of major congenital anomalies was indicative of isolated CMTC. On the day 3 of life, dermatological examination was performed, revealing skin atrophy with significant translucency. Numerous dilated vessels and telangiectasias were observed on the skin of the trunk, head, and extremities. There was no limb asymmetry or other vascular anomalies. Conjunctivas were without petechiae, and palate and female external genital organs were normal. The clinical presentation was clearly suggestive of CMTC. In order to exclude ocular, neurological, and arteriovenous disorders, multi-specialist consultations were recommended. A reduced hip joint range of motion and left-sided motor symptom asymmetry were observed by a physiotherapist, suspecting of developmental hip dysplasia. This condition was excluded after a hip joint ultrasound, while neurological consultation confirmed no disorders. On the 7th day of life, cerebral magnetic resonance angiography and magnetic resonance imaging of the brain were performed, resulting in no pathology detected. On the 14th day of life, the girl was examined by an ophthalmologist. Unilateral iris transillumination defect (left eye) and bilateral reduced pigmentation of the iris and retina were seen, but no glaucoma features were noted.

After the exclusion of other congenital defects, the infant was discharged home on the 14th day of live in good general condition, with body weight of 3,570 g. Lesions in the form of persistent marbled erythema, telangiectasias, venectasias, and skin atrophy were present on the day of discharge, with the same degree of severity as on the day of admission to our department (Figure 1). Long-term dermatological, ophthalmological, orthopedic, and genetic follow-up appointments were recommended. Also, the baby needed physiotherapy due to left-sided motor symptom asymmetry.

Currently, the child is 4 months old and developing correctly, with parents reporting no concerns regarding its development. Also, no new anomalies were seen during medical check-ups, and the dermatological examination showed a significant clinical improvement in the skin appearance. Skin atrophy with translucency, telangiectasias, and dilated vessels are less visible.

DISCUSSION

CMTC was first described in 1922 by a Dutch pediatrician, Cato van Lohuizen. According to the International Society for the Study of Vascular Anomalies (ISSVA), the disease is classified as a capillary malformation [2, 4]. Proportions between sexes are almost identical; however, a slight predominance in females may be noticed, with female to male ratio as 1.2 : 1.0 [2, 3]. Due to the small number of published cases, the sex-related prevalence of CMTC is controversial, and differences are not statistically significant [7, 8]. The pathogenesis is not fully elucidated; though, genetic theories (autosomal mutation, mosaicism, autosomal dominance with incomplete penetrance, and teratogenic cause) were described in the literature [1, 5, 8]. In 2002, the concept of an autosomal lethal mutation surviving by mosaicism and theory of paradominant inheritance were described by Rudolf Happle [4]. Recent studies indicate that GNA11 mutations can be associated with extremity capillary malformations [9]. In Moreno-Alfonso *et al.* study [2], pathogenic gene variants were found in skin biopsies from affected areas. Lesions are usually present at birth, but can develop after a few months, even to 2 years of age [7, 8, 10]. The main symptom is the reticular erythema, which may be generalized or localized to a specific body area. When the lesions are localized, they usually remain one-sided [7, 10]. The trunk, extremities (especially lower limbs), face, and scalp are often involved in the generalized form, while mucous membranes and palms are usually lesion-free [10]. Another frequent feature is telangiectasia, and ulcerations and cutaneous atrophy may also occur [8]. The erythema pales with pressure and does not resolve with warming [4]; this information is essential to distinguish CMTC from physiological cutis marmorata (dilatation of capillaries and small venules due to low temperatures). CMTC persists despite warming, but physiological cutis marmorata resolves [10]. In the case of our patient, the lesions paled with pressure. The child's mother reported a slight improvement with warming; however, this was not observed during medical examination.

In most cases, treatment is not required as the symptoms resolve within the first few years of life, usually 2 years [1, 8]. Some vascular lesions may be treated with laser therapy, although outcomes are mixed, with reports indicating both effectiveness and ineffectiveness [1, 3]. Our patient did not require treatment, and there was no indication for laser therapy.

Kienast *et al.* [11] described criteria helpful in identifying the disorder. The major ones are congenital reticulate (marmorated) erythema, absence of venectasia in the affected area, and unresponsiveness to warming. The minor criteria include fading of erythema within 2 years of life, telangiectasia in the affected places, port-wine stains outside the affected areas, skin atrophy, and ulcerations. Although the criteria have not been validated, the presence of 3 major and 2 minor symptoms may be indicative of CMTC.

The "absence of venectasia" is controversial. There are cases of children with CMTC presenting this symptom, suggesting that the criteria should be revaluated and reconsidered [4]. In our patient's case, venectasias were also present; one of 3 major criteria (erythema) and 2 minor ones (telangiectasia and skin atrophy) were confirmed. Unresponsiveness to warming was controversial, as was reported by the mother only.

Histological examinations of skin biopsies are not relevant due to not specific findings, such as dilated capillaries and veins within the dermis [4, 5]. Molecular diagnosis is possible with identification of the pathogenic variant in *GNA11* gene in a sample of tissue from the affected area [3]. Molecular genetic tests were recommended to our patient by a geneticist; however, parents did not consent to skin biopsy and genetic testing during the child's hospitalization.

Cooccurrence of other anomalies is often described, ranging between 20 and 80%. The most frequently reported is body asymmetry defined as one-sided excessive or insufficient growth of the trunk and/or the extremities, occurring in more than 25% of patients [6, 11]. Musculoskeletal defects, especially leg length discrepancy ≥ 1 cm, if not treated correctly, might have consequences, such as scoliosis, back and lower extremity joint pain, pelvic tilt, and abnormal gait [4, 5]. For patients with mild leg length discrepancies, shoe lifts or orthotics may be used. When muscle weakness occurs, it can be treated with physical therapy [3]. In severe cases, limb lengthening or epiphysiodesis (if length discrepancy is ≥ 2 cm) might be considered [3, 4], but the procedure should be performed before maturing of the skeletal system, i.e., for girls, before the age of 14 years, and for boys, before the age of 16 years [3].

Other commonly mentioned problems are vascular anomalies (23%), and neurological (5%) or, less frequently, ophthalmologic involvement [6]. Neurological conditions include neonatal hypotonia, developmental disability, intellectual disabilities, and seizures [8]. Although involvement of the eyes is rare, it may lead to cataract, glaucoma, or retinoblastoma [12]. When glaucoma occurs, it may be treated with eye drops, trabeculotomy, and trabeculectomy with drainage implants [12]. Trabeculotomy controls the intraocular pressure without causing postoperative hypotony and suprachoroidal hemorrhage [8]. For conditions, such as exudative vitreoretinopathy with fibrovascular membrane and traction retinal detachment, vessel and optic disk dragging, congenital retinal detachment, peripheral avascular areas with vascular alterations or surgical procedure (pars plana vitrectomy) are recommended. In infants with peripheral retinal ischemia, laser photocoagulation may be indicated [12]. In contrast to most infants with CMTC, major abnormalities did not occur in our patient.

The differential diagnosis of CMTC must be conducted scrupulously. Bockenheimer's syndrome, Adams-Oliver syndrome, Klippel-Trénaunay syndrome, Sturge-Weber Syndrome, collagenosis (e.g., neonatal lupus), and capillary malformations (especially, port-wine stains), should be considered and excluded [7, 12]. Port-wine stains present more distinct borders, do not fade as children get older, and skin atrophy is not observed [7]. In neonatal lupus, cutaneous involvement includes erythematous annular lesions or macules with skin atrophy [1]. However, lesions are symmetrical, and facial involvement is more frequent than in cutis marmorata telangiectatica congenita. Moreover, anti-Ro/SSA antibodies are negative in CMTC [7]. Symptoms of Bockenheimer's syndrome include diffuse phlebectasia, which progresses to venous ectasias. Manifestation of Adams-Oliver syndrome is simplified CMTC with cardiac, extremities, and scalp abnormalities. Klippel-Trénaunay syndrome is characterized by venectasia and soft tissue hypertrophy [7], while Sturge-Weber syndrome is associated with facial port-wine stains, and ocular and neurological conditions [4, 8]. In our patient's case, differential diagnosis included disorders specified in the literature; the anti-Ro/SSA antibodies were absent.

CMTC is considered benign and the prognosis is good; however, it requires attention among healthcare professionals and long-term follow-up due to coexisting anomalies and possible subsequent complications [2, 4, 11].

Because of the small number of reported cases, clear follow-up schedules have not yet been established, though suggested in the literature. Annual medical follow-up for at least 3 years was proposed by certain authors [7, 10, 11]. Others reported that the patient's skin should be examined in the frequency determined during initial evaluation to detect forming ulceration at an early stage. Additionally, pain, muscle weakness, and sensitivity to temperature should be evaluated at each follow-up visit. In addition, length and girth of the children's limbs ought to be monitored annually until reaching skeletal maturity, especially when lower extremities are affected by the condition [3, 4]. Some researchers suggest performing a radiograph at the age of 10 years (girls) or 12 years (boys), or if the leg length discrepancy is ≥ 2 cm. Thus, follow-up by orthopedic surgeon is recommended [5]. Patients with CMTC should also have regular ophthalmologic follow-up. Intraocular pressure measurement and peripheral retinal vascular imaging are recommended every 6 months until the age of 4 years, then once a year throughout lifetime, or in case of any ocular pain or visible corneal clouding. Performing tonometry in all patients with facial CMTC is recommended; glaucoma may be associated with lesions involving the face [11]. To exclude macrocephaly, which may also be associated with CMTC, and prevent complications, the neurodevelopment of the child should be assessed by physician. Head circumference should be regularly measured [5, 7] and mental health ought to also be monitored annually, especially in school-attending children. Techniques helping patients adapting to difficult situations and self-esteem issues can be implemented. In addition, parents should be counselled on how to act when individuals not familiar with the condition perceive their child's skin lesions. Providing basic information in advance, e.g. CMTC is not contagious, may prevent unwanted curiosity and prejudice [3].

In most cases, CMTC is self-limiting and lesions show improvement as the patients grow older [13]. Erythema fades within the first two years of life in more than 50% of children [11].

CONCLUSIONS

CMTC as a rare disorder requires thorough differential diagnosis and multidisciplinary collaboration to ensure optimal management. When persistent reticular erythema is observed in a neonate, CMTC should be considered. Also, the patient ought to be referred to an experienced neonatologist, who would exclude other conditions with similar manifestation. When CMTC is diagnosed, long-term observations are necessary. Patients should be under the care of a dermatologist, ophthalmologist, and pediatrician. Imaging tests should be performed regularly to detect possible subsequent complications in the early stages. In cases without secondary complications or with minor coexisting anomalies, the prognosis is good.

DISCLOSURES

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