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CASE REPORT

Congenital Dyskeratosis - case report

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Abstract

Congenital dyskeratosis is a rare, recessive, X-linked, autosomal dominant and recessive disease. The most common manifestations are the triad of mucosal leukoplakia, reticulated pigmentation of the skin and nail dystrophy. However, the patients may present other systemic alterations, such as hematologic, ophthalmologic, gastrointestinal, genitourinary, dental, neurological, pulmonary and skeletal alterations. We report the case of a 5-year old patient, who was followed up for 3 years, who developed pancytopenia, recurrent transfusions and hospitalizations due to recurrent infections.

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INTRODUCTION

Congenital Dyskeratosis is a serious, rare disease with just a few cases reported in the literature - estimated to be 1/1,000,000 in Europe - and that can lead to a spinal failure. Initially, the diagnosis can be suspected through classic mucocutaneous alterations, such as nail dystrophy; reticular pigmentation of the face, neck and shoulders; and mucosal leucodysplasia. The definitive diagnosis is made through the determining the telomeres and the treatment is palliative until bone marrow transplantation is achieved.

GOAL

To report the case of an individual with Congenital Dyskeratosis that evolved to bone marrow aplasia, frequently requiring blood transfusions and frequent hospitalizations due to infections.

METHODOLOGY

This is a descriptive study of the Case Report type. The information contained in this study was obtained through review of the medical records, interview with the person in charge of the patient and review of the literature.

CASE REPORT

A 5-year-old male patient, from Uberaba/MG, started follow-up at the Pediatric Hematology and Hemotherapy outpatient clinic in February 2013 due to mild thrombocytopenia (125,000/mm^3), evidenced in routine exams. Due to the persistence of thrombocytopenia, a myelogram was performed with erythrocytic, granulocytic and lymphoplasmocytic series normoplasia, with hypoplasia of the megakaryocytic series. Since the patient was asymptomatic, we opted for regular follow-up with clinical and laboratory reassessment. In subsequent consultations, thrombocytopenia progressed to pancytopenia. On January 16, 2014, a sensitivity test was performed with Diepoxybutane with a negative result, and bone marrow biopsy revealed hypocellular bone marrow with 15% hematopoietic tissue and 85% adipose tissue, with rare granulocytes and a large predominance of lymphocytic lineage, hemosiderin present grade 2 in 4; suggestive of aplastic anemia.

In 2014, the patient started monthly transfusions of packed red blood cells and platelets. Physical examination revealed nail dystrophies (Figure 1 and 2) and reticular skin pigmentation.

In 2016, telomere length measurement was performed, revealing short telomeres for the patient's age, closing on the diagnosis of Dyskeratosis Congenita.

During the three years of medical follow-up, the patient had worsening pancytopenia. Dermatology indicated skin barrier repairers for dyskeratosis (without treatment success) and Ophthalmology diagnosed fungal retinitis. He



Figure 1. Nail dystrophies in the left hand.

is currently under platelet transfusion twice a week and red blood cell counts every fifteen days. Due to severe persistent neutropenia (neutrophils less than 500), the number of treatable admissions with a broad-spectrum antibiotic increased.

DISCUSSION

Congenital Dyskeratosis is a rare disease with chromosomal instability. The classic manifestation is the triad: reticulated pigmentation of the skin, nail dystrophy and mucosal leukoplakia[1]. Although the classical triad characterizes the disease, only ¾ (three quarters) have at least one clinical characteristic and just under half have the three. Other changes, in addition to mucocutaneous ones may be present, such as progressive bone marrow failure, increased chance



Figure 2. Nail dystrophies in the left foot.

of developing myelodysplasia and leukemia, liver changes, pulmonary fibrosis and central nervous system calcifications[2].

There are two major types of congenital diagnosis: (1) Hoyeraal-Hreidarsson syndrome, characterized by cerebellar hypoplasia, microcephaly, ataxia, developmental delay, immunodeficiency, and early aplastic anemia; and (2) Revez's syndrome, which presents bilateral exudative retinopathy, intrauterine retardation, severe aplastic anemia and central nervous system calcifications. The prognoses of these two forms are very bleak.

Congenital Dyskeratosis is a disorder that can be inherited autosomal recessive, autosomal dominant or linked to the chromosome. Approximately 70% of patients will mutate into one of the following genes: ACD, CTC1, DKC1-DC, NAF1, NHP2, NOP10, PARN, RTEL1, STN1, TERC, TERT, TINF2, and WRAP53[4].

The diagnosis is made by determining the sizes of telomeres (less than 1% for age) in various leukocyte subtypes and the clinical findings of the disease[5]. Treatment consists of attenuating symptoms with blood transfusions, Danazol and treatment of infections[6]. Already the definitive treatment happens with the transplant of bone marrow. Even after transplantation, there is a chance of complications such as pulmonary or hepatic fibrosis.

CONCLUSION

Congenital Dyskeratosis is a serious disease with an increased chance of survival when diagnosed early. The treatment consists in attenuating the symptoms and the definitive cure is obtained through bone marrow transplant.

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