ECRONICON OPEN ACCESS

EC PAEDIATRICS
Research Article

Blackfan-Diamond Syndrome After Bone Marrow Transplant

Ariadne Godinho*, Emannuel Luther Valentin Leal, Joanna Cardoso's Hemilly and Julia Maria Pinho Costa

Advanced Institute of Higher Education of Barriers - IAESB, San Francisco University Center of Barriers - UNIFASB, Brazil

***Corresponding Author:** Ariadne Godinho, Advanced Institute of Higher Education of Barriers - IAESB, San Francisco University Center of Barriers - UNIFASB, Brazil.

Received: February 27, 2023; **Published:** March 06, 2023

DOI: 10.31080/ecpe.2023.12.01282

Abstract

Blackfan-diamond syndrome is a rare congenital anemia characterized by the reduction of erythroid precursors in the bone marrow, severe macrocytic anemia and reticulocytopenia, as well as the presence of congenital anomalies and predisposition to cancer. It has hereditary character in 40 - 45% of cases by autosomal dominant inheritance and the remaining 55 - 60% are sporadic. Diagnosis is made in the first months of life in 90% of cases. It is evaluated by complete blood count, reticulocyte count and bone marrow aspirate. The therapy of the syndrome is initially based on corticosteroid therapy and close follow-up of blood count for periodic blood transfusion, adhering to this some patients require iron quelantes. The cure for the disease comes from bone marrow transplantation from a 100% compatible allogeneic donor. Transplantation is not always achieved quickly, and the patient is then submitted to corticosteroid and blood transfusion cycles and presenting certain corresponding adverse effects, such as drug intolerance, iron hemoconcentration and problems related to growth retardation, the latter as well as being seen in other types of chronic anemias. Thus, the aspects of microscopic diagnosis, histocompatibility test characteristics (HLA-Human Leukocyte Antigen) and bone marrow transplantation and its positive consequences for the history of the disease will be addressed.

Keywords: *Blackfan-Diamond; Aplasia; Anaemia; Transfusion; Transplantation; Bone Marrow*

Introduction

Blackfan-diamond syndrome is characterized by reduction of erythroid precursors in the bone marrow, severe macrocytic anemia, and reticulocytopenia. It is a rare congenital anemia, with an estimated incidence of 5 - $10/10^6$ live births [1] and heterogeneous when observing the pattern of transmission, clinical picture, laboratory tests and treatment results. The syndrome is hereditary in 40 - 45% of cases by autosomal dominant inheritance and the remaining 55 - 60% are sporadic. Autosomal recessive inheritance has been reported less frequently [2].

The clinical picture manifests itself early in life, usually before 6 months of age (mean between 2 - 4 months) [3]. Its clinical manifestation is a progressive and slow anemia, in which the decrease in hemoglobin is more pronounced and the lower limit varies with the degree of prematurity, may present hemoglobin values of 7 to 8 g/dl, still in the 2^{nd} week of life, and even after this period, when the baby is already about 10 weeks [4]. In addition, congenital malformations are present in 25% of cases, such as craniofacial and upper limbs deformities [5].

Diagnosis is made in the first months of life in 90% of cases [6]. Laboratory analysis begins with the request for a complete blood count, reticulocyte count and, if possible, fetal hemoglobin level (HbF) and adenosine deaminase (ADA) activity in red blood cells. The large por-

tion of patients present macrocytosis, with increased HbF. The activity of ADA is high, for reasons not fully understood, in 80% to 85% of patients with Blackfan-Diamond Syndrome [7].

The essential treatment is linked to corticosteroid therapy, red cell transfusions and Hematopoietic Stem Cell Transplantation (HSCT). HSCT is the only definitive treatment, in the presence of an HLA-compatible donor, for the hematological manifestations of BDS [8]. However, all sibling donors should be carefully indicated, including genotyping when known, even when the donor has no evidence of any hematological or physical manifestation of Blackfan-Diamond Syndrome [9,10].

Justification

Bone marrow transplantation is the crucial therapy for the cure of Blackfan-Diamond Syndrome. Some patients soon after diagnosis are submitted to corticosteroid therapy and blood transfusion, in which they are exposed consequently to undesirable effects, and end up suffering for a longer period with the consequences of erythrocytic failure, presenting growth retardation among other complications. The search for a compatible donor is usually harsh, but when it finds success, the patient cures himself of the disease and will be free of corticosteroid therapy or blood transfusion and its negative consequences.

This work raises the steps of accurate diagnosis, histocompatibility test (HLA-Human Leukocyte Antigen) and bone marrow transplantation by washing into account the too positive step towards the cure of these patients.

Objectives of the Study

General

- Evaluate the characteristics of Blackfan-Diamond Syndrome and the diagnostic and therapeutic steps in the search for compatible donor and bone marrow transplantation, as well as its benefit.

Specific objectives

- Characterize Blackfan-Diamond Syndrome;
- Evaluate the criteria for accurate diagnosis;
- Describe the initial therapies of the Syndrome;
- List the criteria of the histocompatibility test (HLA- Human Leukocyte Antigen);
- Explain bone marrow transplantation as a cure for the disease;
- Elucidate the picture and positive consequences after bone marrow transplantation.

Methods

Type of search

The ongoing research has a primary and observational descriptive character in the qualitative scientific literature.

Description of the search location

The patient in question undertakes medical care at the Jardim Ouro Branco Family Health Unit, located in the municipality of Barreiras, Bahia. The research was carried out through the unit's medical records and previous examinations presented by the patient. In addition, research was done in literary banks.

Study population

The population of this study is equivalent to 01 participant, having the diagnosis for Blackfan Diamond Syndrome and literature review, by examining the written material and published in books.

Sample

This sample was 01 case report compatible with Blackfan Diamond Syndrome, and in order to elaborate the research, medical records, reports and examinations of the patient from diagnosis to discharge from the MOT outpatient clinic were analyzed. It also presents a survey and analysis with the theme of this research, filtering those that are especially interested in Blackfan Diamond Syndrome.

Data collection instrument

As a mechanism for data collection, it was elected through the selection of the relevant material, interpretation of data and law. The insertion criterion was articles that constituted information about the research syndrome presented.

Description of data collection

As a technique for data collection, we chose to use systemic analysis in search of articles that presented complete textual structures available, such as scientific studies, articles published in journals, symposia, conferences and dissertations published between 2000 and 2018.

Descriptions of data analysis

In the search for selection of the collected data, an analysis of the selected content was made, which responded statically to the descriptive methods to evaluate the precise diagnostic criteria of Blackfan Diamond Syndrome.

Inclusion criteria

In the election of the articles, we used the criteria of: articles that contained information about the presented syndrome, the interest for the theme and scientific articles that presented relevant content and sample data. Throughout the analysis, the following descriptors were used: Blackfan-diamond, aplasia, anemia, transfusion, transplantation, bone marrow.

Exclusion criteria

The following were discarded from the research: theses, dissertations, repeated articles, monographs and articles that did not respond to the theme presented, as well as those works that only related to the proposed subject.

Risk analysis to research subjects (Sample)

In this work, no methodology applies to which to perform actions that affect physiological, social and psychic variables. Data collection was found in medical records that would not be necessary for this research, exams and reports, in which it will include only the age and initials of the participant.

Analysis of benefits to research subjects (Sample)

In this study, it will not address to patients any intervention or intentional alteration in social, physiological or psychological aspects, a view that the intention will be only the analysis of the medical records in the sample.

Return of benefits to the population

The present study includes the dissemination of final results of the investigation of data collected with the population, which includes research participants, and not exclusively to students and professionals in the area, therefore, it is evident the intention in methods of publication in public dissemination media, in order to add the information obtained on the subject.

It is important to note that the dissemination of this study collaborates with intervention protocols and postponement to the treatment of Blackfan-Diamond Syndrome, so that it improves therapy, ensures greater effectiveness when diagnosing the disease.

There is no privilege to the participants, because it is a direct contactless analysis. It then relates to a general privilege, to improve knowledge.

Criteria to terminate or suspend search

In case there is mediation snares for the acquisition of information for the progress and ratification of the study, as impediments to the collection of data is realized and the participant's withdrawal in disclosing the case. In this situation, any data that makes it impossible to access the information provided will be considered as an example.

Ethics in research with human beings (Resolution 466/12)

The study is part of a project of greater expression in the area of Blackfan Diamond Syndrome submitted to the admiration of the Research Ethics Committee (CEP). Given that the study subjects are human beings, we will comply with the provisions of Resolution 466/12 of the Ministry of Health in Brazil by submitting it to the analysis and judgment of the Ethics Committee on Research with Human Beings of the São Francisco University Center of Barriers - UNIFASB/UNINASSAU.

Expected Results, Discussion and Conclusion

It doesn't apply.

Commitment to make the results public

The publication of the results obtained in the research will take place in order to propagate the information obtained through the analysis and studies of this research on websites, scientific initiation congress, seminars, journals, scientific studies, articles published in journals, symposia, conferences and dissertations.

Schedule

The schedule is the graphical disposition of the time that will be spent in carrying out a work or project, according to the activities to be fulfilled. It serves to assist in the management and control of this work, allowing quickly the visualization of its progress.

Activity	March	April	may
Project development	X		
Referral to zip code		X	X
Data Analysis			X
Results and Discussion			X
Conclusion/Final Considerations			X
Final Version			X

Budget

Permanent material	Quantity	Unit price R$	Total Price R$
Notebook	03	0,0	0,0
		Subtotal	0,0
Third Party Services Individuals	Quantity	Unit price R$	Total Price R$
Fuel	60Liters	7,80	468,00
		Subtotal	468,00
Third Party Services Individuals	Quantity	Unit price R$	Total Price R$
Impression	5,00	0,25	1,25
		Subtotal	1,25
Overall Total			R$ 469,25

Table 1: *Forecast of expenses with the execution of the work.*

Note: The burden of this research is the responsibility of the researchers, and there are, therefore, no costs for the participants, nor for the institutions involved.

Bibliography

1. Vlachos A., *et al*. "Clinical utility gene card for: Diamond - Blackfan Anemia - update 2013". *European Journal of Human Genetics* 21.10 (2013): 1187.

2. Engidaye G., *et al*. "Diamond Blackfan Anemia: genetics pathogenesis, diagnosis and treatment". *eJIFCC* 30.1 (2019): 67-81.

3. Bravo LM and Rodríguez ZN. "Diamond-Blackfan anemia: Clinical experience in 20 patients (1968-1998)". *Revista Chilena de Pediatria* 71.3 (2000).

4. Martino-Röth., *et al*. "Frequency of multiple malformations in newborns in the city of Pelotas, Rio Grande do Sul, Brazil, and associated sociodemographic factors". *Caderno de Saúde Pública* 22.5 (2006): 1009-1015.

5. Pasquini Ricardo. "SSEA bone marrow transplantation in aplastic anemias". *Medicine, Ribeirão Preto, Symposium: SSEA Bone Marrow Transplantation* 33 (2000): 219-231."

6. Da Costa L., *et al*. "An update on the pathogenesis and diagnosis of Diamond–Blackfan anemia". *F1000 Research* 7 (2018): 1350.

7. Vlachos Adriana and Muir Ellen. "How I treat Diamond-Blackfan anemia". *Blood* 116 (2010): 3715-3723.

8. Vlachos A., *et al*. "Diagnosing and treating Diamond Blackfan anaemia: results of an international clinical consensus conference". *British Journal of Haematology* 142.6 (2008): 859-876.

9. Hernández Marino A. "Anemias in childhood and adolescence. Classification and diagnosis". *Comprehensive Pediatrics* 5.10-10 (2020): 5.

10. Willig T., *et al*. "Diamond-Blackfan anemia". *Current Opinion in Hematology* 7.2 (2000): 85-94.

Volume 12 Issue 4 April 2023