

The first case of Béguez César syndrome in Iraq

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Abstract

Beguez Cesar syndrome was first reported by Antonio Beguez Cesar, and he called it chronic malignant familial neutropenia with atypical leukocyte granulations. The most common manifestations of Beguez Cesar syndrome are neutropenia, oculocutaneous albinism, and the susceptibility to infections, particularly *Staphylococcus aureus*, and *Streptococci*.

Beguez Cesar syndrome is a rare disorder that has not been described or documented in Iraq. The aim of this chapter is to describe the first case of the syndrome in Iraq.

Keyword: béguez césar syndrome; antonio beguez cesar; chronic malignant; leukocyte granulations

Introduction

Beguez Cesar syndrome was first reported by Antonio Beguez Cesar, and he called it chronic malignant familial neutropenia with atypical leukocyte granulations. The most common manifestations of Beguez Cesar syndrome are neutropenia, oculocutaneous albinism, and the susceptibility to infections, particularly *Staphylococcus aureus*, and *Streptococci*. The susceptibility to infections in Beguez Cesar syndrome is mainly attributed to impaired bacteriolysis which is caused by failure of phagolysosome formation. The resulting disordered intracellular trafficking and impaired lysosome degranulation within phagosomes make phagocytosed bacteria remain undestroyed by the lysosomal enzymes [1,2].

Patients with Beguez Cesar syndrome have light skin, and silvery hair (partial albinism) and photophobia. Recurrent infections are commonly associated with the syndrome.

Infections in Beguez Cesar syndrome usually involve mucous membranes, skin, and the respiratory tract. During childhood, patients with this syndrome are susceptible to infection by Gram-positive, gram-negative bacteria, and also fungi. However, infections with *Staphylococcus aureus* are probably the most common infection associated with this syndrome[1,2].

Beguez Cesar syndrome is a rare disorder that has not been described or documented in Iraq. The aim of this chapter is to describe the first case of the syndrome in Iraq.

Case report

A five year old girl was seen during December, 2016 because of recurrent infections and delayed development with poor speech. She had fair light skin and blond hair (Figure-1), and she didn't say any word.

She also had refractive error and needed corrective eye glasses, photophobia, and also nystagmus. Her gross and fine motor developed

was normal; she could hold a pencil, and was unable to draw a circle which was not very perfect.

However, she could play games on smart phones. Her parents were cousins, and she had three normal sisters. The older sisters both had normal development, one at third primary class, and the second at first primary class. The two older sisters were doing well at school. The younger sister was normal, and aged three years. None of the family had fair or light skin, and all were considered to be normal.

The girl had recurrent skin, oral, and respiratory infections. She didn't have organomegaly. She was previously hospitalized because of serious lower respiratory infection.



Figure-1: The girl had fair light skin and blond hair .She also had refractive error and needed corrective eye glasses

Full blood examination was performed during November, 2016 and showed hypochromic microcytic anemia, anisocytosis, few target cells, and marked rouleaux. While blood count showed absolute neutropenia with relative lymphocytosis. There were atypical lymphocytes (15%), and few of them had cytoplasmic vacuulations. Reticulocyte count was 0.5%. Platelet count was $286 \times 10^9/L$, and large and giant platelets were observed.

The hemoglobin was 9.76 g/dl (hematocrit 34.2%) during November, and was 9.76 g/dl (hematocrit 30.0%). Table-1 shows the while blood counts, the differential counts, red blood cell counts, red blood cell indices, platelet counts, and mean platelet volume performed during November,

and December, 2016.

Bone marrow examination showed cellular marrow with mixed cellularity. Erythropoiesis was normoblastic and micro-normoblastic and the myeloid to erythroid ratio was 1.72 to 1. Granulopoiesis had different maturation series to the end stages. Large myelocytes and metamyelocytes were present. Ring bands were also detected.

Late during December, 2016, she developed fever and cough with clinical and radiological evidence of lower respiratory tract infection (Figure-2). The girl also had fungal infection of the nail and slight clubbing (Figure-3).

	November	December
WBC	$2.74 \times 10^9/L$	$3.45 \times 10^9/L$
Neutrophil	9.37%	8.12%
Lymphocyte	74.9%	77%
Monocyte	14.1%	14.1%
Eosinophil	0.2%	0.27%
Basophil	1.49%	1.22%
RBC	$5.18 \times 10^{12}/L$	$4.62 \times 10^{12}/L$
Mean corpuscular volume (MCV)	65.9 FL	64 FL
Mean corpuscular hemoglobin (MCH)	18.8 pg	19.1 pg
Mean corpuscular hemoglobin concentration (MCHC)	28.5g/dL	29.8g/dL
Red blood cell distribution width (RDW)	18 %	18.4 %
Platelet count	$286 \times 10^9/L$	$226 \times 10^9/L$
Mean platelet volume (MPV)	6.76 FL	7.70 FL

Table-1: The while blood counts, the differential counts, red blood cell counts, red blood cell indices, platelet counts, and mean platelet volume performed during November and December, 2016.



Figure-2: She developed fever and cough with radiological evidence of lower respiratory tract infection



Figure -3: The girl also had fungal infection of the nail and slight clubbing

Discussion

Management of Beguez Cesar syndrome includes treatment of manifestations and genetic counseling.

Beguez Cesar syndrome is transmitted in an autosomal recessive pattern. When both parents are heterozygous, each siblings of an affected individual has a 25% chance of having the disease, and a 50% chance of being carrier, and a 25% chance of being unaffected and not a carrier.

Early aggressive use of antibiotics and antiviral agents for bacterial and viral illnesses is an integral component of the management of Beguez Cesar syndrome.

Boxer et al (1976) found that ascorbic acid could correct certain functional abnormalities of the cells in Beguez Cesar syndrome. Therefore, the decision was made to put the patient in this report on long term vitamin C supplementation[3].

Syndromes in medicine including genetic syndromes are generally named after the physician or group of physicians that discovered them or initially provided the full clinical picture or the best description of the syndrome. However, many of the rare genetic syndromes have been described by doctors in many areas of the world before the era of the internet which has been associated with easy access to clinical reports throughout the world. Unfortunately, some syndromes have been attributed unfairly and inappropriately to doctors other than those first described them. Beguez Cesar syndrome has not been named appropriately in many or most medical literature[2].

The condition was first reported by Antonio Maria Beguez Cesar, and he called it chronic malignant familial neutropenia with atypical leukocyte granulations. Dr. Antonio Beguez Cesar published the first report about the disorder in the Bulletin of the Cuban Society of Pediatrics in January, 1943.

Dr. Antonio Maria Beguez Cesar reported the cases of three children of a family in Santiago de Cuba. He observed them during the period from 1933 to 1940. The three children had a disease characterized by albinism, nystagmus, undetermined febrile states, leukopenia, neutropenia, lymphomonocytosis, as well as the presence of atypical granulocytes in the leukocytes.

The three patients of Dr. Beguez died despite treatment. The abnormal leukocytes with granulations were found in the father and all the children. Dr. Antonio Beguez Cesar concluded that the children had a new disease or a new clinical entity called chronic familial malignant neutropenia with atypical granulocytes of the leukocytes. Dr. Beguez thought that the disorder was possibly transmitted by direct inheritance or similar inheritance from father to offspring[1,2].

A German doctor, Dr. William Steinbrinck published a paper on this condition in 1948 [4].

In 1952, a Cuban physician and serologist Dr. Alejandro Moises Chediak (1903 1993) published in a French magazine a paper entitled "Nouvelle anomalie leucocytaire de caractere constitutionnel et familial", which means "New leukocyte anomaly of constitutional and family character"[5].

Chediak mentioned that the patient was first seen during the later part of the year 1940, and during more than a decade, he presented the leukocyte anomalies to a number of American and European hematologists, but none of them recognized the condition[5].

During August, 1953, Boturao and colleagues presented a case of Beguez Cesar syndrome at the Third Regional Congress of the Associaoca Paulista Medicina Santos.

The patient of Boturao and colleagues had the leukocyte anomaly of Beguez Cesar syndrome, photophobia, albinism of fundus oculi, and whitish-gray hair. The patient remained healthy at the age 13 years old [6].

In 1953, a Japanese pediatrician, Dr. Otokata Higashi (1883 1981) published a case similar to that of Beguez, but the Japanese physician was also unmindful to the observations of Dr Antonio Beguez Cesar [7].

The syndrome was called Chediak and Higashi's disease by Sato in 1955. Sato also missed the report of Antonio Beguez Cesar, and suggested the emergence of a new clinical entity Chediak and Higashi's disease or Chediak-Higashi's Leuco-anomaly. Sato cited the paper of Chediak, published in 1952 entitled Nouvelle anomalie leucocytaire de caractere constitutionnel et familial, and also the paper of Higashi entitled Congenital gigantism of peroxidase granules which was published in 1953. Sato stated I had not seen any case of such an abnormality during my own 30 years experience with the peroxidase reaction [8].

Sato also missed the cases of Steinbrinck and Boturao and colleagues. Sato considered the disease reported by Chediak in 1952, and the disease described by Higashi in 1953 to be two different manifestations of one and the same clinical entity, despite the fact that Chediak didn't describe a peroxidase or oxidase reaction [2,8].

During the 1960s and 1970s some authors including Mauri and Silingardi (1964); Santino and Scialfa (1966); Balsamo and colleagues (1967); Pachioli and colleagues (1970) called the syndrome Beguez Cesar-Steinbrinck- Chediak-Higashi anomaly or syndrome [2].

However, at the closing session of the First Latin American Hematology Conference, which was held in Havana during February, 1973, the Ministry of Public Health of Cuba, and all the representatives of the countries of Latin America who attended this conference, and Scientists officially recognized that the discoverer of the so-called "Chediak-Higashi disease" had been Dr. Antonio Maria Beguez Cesar.

It was according to suggestions of doctors Sanchez Medal and Tulio Arends, a special recognition was given to Dr. Antonio Beguez Cesar, for being the first who described a new hematological disease that he published in the Bulletin of the Cuban Society of Pediatrics, number 12 of 1943 with the Title 'Familial Malignant Chronic Neutropenia with Atypical Leukocyte Granulations[2]'.

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References

1. Beguez Cesar A. Neutropenia Cronica Maligna Familiarcon Granulaciones Atipicas de los Leucocitos. Bol Soc Cubana Pediatr 1943;15(12): 900-22.
2. Al-Mosawi AJ. Bequez Cesar syndrome. 1st ed., Saarbrücken; LAP Lambert Academic Publishing: 2017(ISBN: 978-3-330-03284-2).
3. Boxer L A, Watanabe AM, Rister M, Besch HR Jr, Allen J, Baehner RL. Correction of leukocyte function in Chediak-Higashi syndrome by ascorbate. New Eng J Med 1976; 295: 1041-1045.PubMed: 184391.
4. Steinbrinck W. Uber eine neue Granulatsanomalie der Leukocyten. Deutsches Archiv fur klinische Medizin (Leipzig) 1948;193: 577-81.
5. Chediak MM. New leukocyte anomaly of constitutional and familial character. Rev Hematol. 1952; 7(3):362-7.PMID:13004553.
6. Boturao E, Barbosa JU, Bonna L De .Anomalia de Chediak. Third Reg Congr of the Associaoca Paulista Medicina Santos 1953; August.
7. Higashi O. Congenital abnormality of peroxidase granules; a case of congenital gigantism of peroxidase granules, preliminary report. Tohoku J Exp Med 1953 Oct 25; 58(3-4):246.PMID:13136163.
8. Sato A. Chediak and Higashi's disease: probable identity of a new leucocytal anomaly (Chediak) and congenital gigantism of peroxidase granules (Higashi). Tohoku J Exp Med 1955 Feb 25; 61(2-3):201-10. PMID: 14396888.



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