

## Prevalence of Genetic Diseases in Four Basic Health Units Located in Bahia State

### Prevalência de Doenças Genéticas em Quatro Unidades Básicas de Saúde Localizadas no Interior da Bahia, Brasil

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#### Abstract

Genetic diseases affect about 3 to 7% of the world population and represent an important cause of infant morbidity and mortality in Brazil. Thinking of this reality, more in-depth studies on the clinical and epidemiological characteristics of these diseases are necessary for the implementation of a more effective and comprehensive health system. The objective of this study was to analyze the prevalence of genetic diseases in patients treated at four Basic Health Units (BHU), located in the city of Paulo Afonso, Bahia. This is a quantitative, exploratory and documentary study, based on secondary data obtained by consulting the users' medical records assisted by four BHU, during the period from 2015 to 2018. The data were presented as both absolute and relative values, considering an estimated prevalence of 10% in the general population, a sampling error relative of 5% and a confidence level of 95%. It was noticed for the prevalence of three noted disturbances in primary health clinics: Down's syndrome, cleft palate and epilepsy. The disease with the highest prevalence was Down's syndrome (27.78%), followed by cleft palate (16.67%), then Crohn's Disease, sickle cell trait, and epilepsy (11.10% each). The other genetic diseases presented only one case each (5.56%). It was evidenced that there is a difference in the proportion of genetic diseases among the studied communities ( $\chi^2 = 13.32$ ;  $p = 0.003$ ).

**Keywords:** Community Genetics. Primary Attention. Genetic Disorders. Medical Secondary Data. Hereditary Diseases.

#### Resumo

*As doenças genéticas afetam cerca de 3 a 10% da população mundial e representam um importante causa de morbimortalidade infantil no Brasil. Diante dessa realidade, tornam-se necessários estudos mais aprofundados sobre as características clínicas e epidemiológicas dessas doenças para um sistema de saúde mais efetivo e abrangente. O objetivo desse estudo foi analisar a prevalência de doenças genéticas nos pacientes atendidos em quatro Unidades Básicas de Saúde (UBS), localizadas no município de Paulo Afonso, Bahia. Trata-se de um estudo quantitativo, exploratório e documental, baseado em dados secundários obtidos por meio de consulta aos prontuários dos usuários assistidos por quatro UBS, durante o período de 2015 a 2018. Os dados encontrados foram apresentados na forma de números absolutos e relativos, considerando uma prevalência estimada de 10% na população em geral, um erro de amostragem relativo a 5% e um nível de confiança de 95%. As doenças com maiores prevalências foram a síndrome de Down (27,78%), fenda palatina (16,67%), doença de Crohn, traço falciforme e epilepsia (11,10%, cada). As demais doenças genéticas apresentaram apenas um caso (5,56%). Evidenciou-se uma diferença significativa na proporção de doenças genéticas entre as comunidades estudadas ( $\chi^2 = 13,32$ ;  $p = 0,003$ ).*

**Palavras-chave:** Genética Comunitária. Atenção Primária. Desordens Genéticas. Dados Secundários Médicos. Doenças Hereditárias.

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#### 1 Introduction

According to the World Health Organization (WHO), diseases of genetic character affect approximately from 3% to 10% of the population and bring important economic and psycho-social impacts to the affected individuals (VIEIRA *et al.*, 2013). In addition, information from the Department of Informatics of *Sistema Único de Saúde* (SUS) or DATASUS reveals that in 2017, congenital diseases, which include genetic disorders, were the second largest proportional cause of infant mortality (FRANCA *et al.*, 2017).

Considering the expressiveness of these data, greater involvement of present health care sectors with medical genetics becomes imperative, most notably in the Primary Health Care system. Thus, at this level of care that direct

contact was found between the population and Family Health Strategy (FHS) representatives. Greater involvement in medical genetics would allow for more effective prevention and control actions, monitoring, assistance, and treatment for both the individuals and families who suffer because of some genetic disorder (MEIRA; ACOSTA, 2009; SILVA; CASOTTI; CHAVEZ, 2013).

While using a regional context as a basis for the actions and interventions of its professionals, the Brazilian health system provides integral health care. Considering the reported incidence of genetic diseases, objective knowledge concerning regional populations through mapping of health conditions, and emphasis on the importance of genetic diseases is necessary. Since the FHS has intimate contact with

the community and provides continuing assistance in order to achieve positive results, current localized mapping of these diseases and qualification of its working professionals through specific knowledge concerning medical genetics are essential (MELO *et al.*, 2015).

Considering the importance to the community of genetics and the mapping of genetic diseases for knowledge of the health conditions in a certain population, and for realization of a more comprehensive qualified health care, the present study was performed with the objective of identifying the prevalence of genetic diseases as diagnosed at certain Primary Health Clinics in the city of Paulo Afonso/BA.

## 2 Material and Methods

### 2.1 Sample design

The city of Paulo Afonso-BA has a population of approximately 118 thousand inhabitants. Considering an estimated prevalence of 10% in the general population, a sampling error relative of 5% and a confidence level of 95%, a necessary minimum sample of 12,408 observations to build reliable parameters for the occurrence of genetic diseases was obtained.

From March 2015 to February 2018, data were collected at the Center for Applied Studies in genetics (NEAGen), involving the São Francisco Valley Federal University – UNIVASF, Paulo Afonso campus medical school. The research was performed at four primary health clinics: São João, Perpétuo Socorro, Jardim Bahia, and Santa Quitéria, all in the Paulo Afonso, Bahia municipality. Health services located in central and peripheral areas of the city were chosen in order to increase the socioeconomic spectrum of the population studied. The study was based in quantitative,

exploratory and documentary methods.

Data collection was carried out through visits to the primary health clinics, in which medical records were analyzed with the aim of finding genetic etiology pathologies. 14,257 records were surveyed corresponding to 3,459 families. When a genetic disorder was identified, this was recorded and tabulated under a general genetic disease framework and then genetically analyzed by the project director.

After detection and accompanied by Community Health Agents (CHA), patients were actively sought out and interviewed using a structured questionnaire, containing information such as ethnicity, age, types of abnormalities, family history of genetic and/or congenital disease, and if female, the number of previous pregnancies. As each cycle of visits to the primary health clinics ended, a questionnaire was applied as well to the unit's health care professionals. Topics related to basic genetics, as well as themes related to the most prevalent disorders discovered during the medical records analyses were also addressed. The questionnaire was applied with the aim of identifying possible gaps in the professional health care giver's knowledge concerning medical genetics.

### 2.2 Data analysis

Inferential analysis was performed to test the hypothesis of difference among the health units. For that, a Pearson's chi-square test ( $\chi^2$ ) was performed, adopting a significance level of 5%.

## 3 Results and Discussion

Primary health units' records were analyzed from São João, Perpétuo Socorro, Jardim Bahia, and Santa Quitéria unit, as shown in Table 1.

**Table 1** - Data concerning genetic diseases; by primary health clinic in Paulo Afonso/BA - 2018

Primary Health Clinic	Medical Records Analyzed	Number of Families	Genetic Disease Cases	Genetic Disease Total
São João	5299	1121	Albinism (1); Friedreich's ataxia (1); Cleft palate (2); Down's Syndrome (3).	7
Perpétuo Socorro	1040	338	Multiple hereditary osteochondromatosis (1); Down's Syndrome (1); Sickle cell trait (1).	3
Jardim Bahia	3348	916	Crohn's Disease (2); Cleft palate (1).	3
Santa Quitéria	4570	1804	Sickle cell anemia (1); Epilepsy (2); Down's Syndrome (1); Sickle cell trait (1).	5
<b>Total</b>	14257	3459	-	18

Source: Research data.

Of the total number of records analyzed, 18 presented genetic diseases with confirmed diagnoses; this provided a percentage of approximately 0.13%, (of the city) for

individuals with genetic disease. It is important to highlight the prevalence of three specific diseases: Down's syndrome, cleft palate and epilepsy, which among all the genetic

disorders presented a higher prevalence in the primary health clinics visited.

Upon analysis of São João primary health unit records seven patients were found with some type of genetic disorder, being a patient with albinism, a patient with Friedreich's ataxia, two patients with cleft palate, and three patients with Down's syndrome.

For Perpétuo Socorro and Jardim Bahia primary health units three cases were found each of genetic diseases per unit. Santa Quitéria health unit presented the third highest prevalence of genetic disorders, with a total of five recorded cases in 4570 records, presenting one case of sickle cell anemia, two cases of epilepsy, one case of Down's syndrome and one case of sickle cell trait.

Modern society is undergoing a major epidemiological transition, in which contagious diseases are becoming less frequent in relation to chronic and hereditary diseases. Within this new reality, genetic diseases play an important role in this relative increase in morbidity and mortality rates. Such statistics can be observed in the Brazilian scenario, in which congenital basis pathologies, including those of genetic etiology, represented the 5th cause of infant mortality in 1980, yet in 2015 occupied the second position (FRANCA *et al.*, 2015).

During the period of research, three data collections were carried out, examining 14,257 medical records belonging to 3,459 families, being on average 4.12 people per household in the region. According to data from the Brazilian Institute of Geography and Statistics (IBGE) for the year 2017, the estimated population of the city of Paulo Afonso-BA, being 1,580 km<sup>2</sup>, was 120,706 inhabitants, an average of 76.39 inhabitants per km<sup>2</sup>.

Correlating the data already mentioned herein, it was verified that the population percentage (of the city) analyzed was 11.81%.

According to WHO, rare diseases are disorders that affect up to 65 per 100,000 individuals. Among the various pathologies encountered by the study, certain rare diseases were found, among them two cases of Crohn's Disease (North American incidence of 20.2:100,000 inhabitants/year; similar to other continents), one case of Friedreich's ataxia, and one case of multiple hereditary osteochondromatosis (1:50,000 for both) (RUIZ JR *et al.*, 2008; CASTRO; DUARTE, 2013; ROSA; SILVA JÚNIOR; ROSA, 2014).

However, considering the number of records analyzed from each clinic, Perpétuo Socorro health clinic presented a smaller number of patients, yet with a three times greater prevalence in relation to the Jardim Bahia health clinic. Of all the records analyzed, Perpétuo Socorro clinic presented the highest prevalence of genetic disorders, with one case of multiple hereditary osteochondromatosis, one case of Down's syndrome, and one case of sickle cell trait. Jardim Bahia clinic presented two cases of Crohn's disease and one case of cleft palate.

Among other reasons, the increase in genetic diseases rates in Brazil results from greater scientific knowledge concerning such diseases among medical professionals (MELO *et al.*, 2015), and confirms the reality here with the one found in the health clinics, where health professionals achieved relatively satisfactory performances in the questionnaires.

Analysis of the results revealed that Perpétuo Socorro health clinic presented the highest rate of genetic disease, considering that for every 1000 individuals (Table 2), three patients were carriers of genetic disease (0.28% of health clinic users). Jardim Bahia neighborhood presented fewer cases of genetic diseases, and for every 1000 individuals approximately 1 person carried a genetic disorder (0.09% of users).

**Table 2** - Prevalence of genetic diseases found in the primary health clinics of Paulo Afonso/BA, 2018

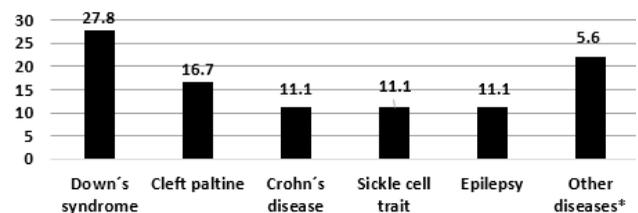
Primary Health Clinic / Neighborhood	Prevalence of Genetic Diseases
São João	0.13%
Perpétuo Socorro	0.28%
Jardim Bahia	0.09%
Santa Quitéria	0.10%

Source: Research data.

It was evidenced that there is a difference in the proportion of genetic diseases among the studied communities ( $x^2 = 13.32$ ;  $p = 0.003$ ), with Perpétuo Socorro unit being more prevalent than other neighborhoods beyond chance. (This can be explained by two facets: there are actually more diseases or there is a better diagnosis).

The disease with the highest prevalence was Down's syndrome (27.78%), followed by cleft palate (16.67%), then Crohn's Disease, sickle cell trait, and epilepsy (11.10% each). The other genetic diseases presented only one case each (5.56%) (Figure 1).

**Figure 1** - Prevalence of genetic diseases found in Paulo Afonso/BA primary health clinics - 2018.



\*Albinism, Friedreich's ataxia, multiple hereditary osteochondromatosis and sickle cell anemia.

Source: Research data.

The prevalence of down syndrome was more significant, corroborating with the high rate in Brazil (1:700 births) (FIGUEIREDO *et al.*, 2008). This syndrome is a genetic disorder in which trisomy of chromosome 21 occurs. Individuals thus affected commonly follow a clinical description, with characteristic physical, mental and cognitive changes (SIMÕES *et al.*, 2016).

By prevalence ranking for the survey findings, cleft

palate was 0.021%. When compared to the same prevalence in the state of Rio Grande do Norte, which was 0.006%, the frequency identified in Paulo Afonso municipality was much higher, however, lower than the overall Brazilian incidence of 0.15% (FIGUEIREDO et al., 2011, CTMC, 2018). Cleft palate is a congenital malformation identified by the presence of a cleft in the bone region or mucosa of the palatal vault. Currently, treatments have been developed and through corrective surgeries are capable of improving the quality of life, aesthetics, and social interactions of the patient (FERREIRA, 2012; WALKER; PODDA, 2020).

However, the prevalence of epilepsy, being one of the most prevalent mental disorders was 0.00014%, well below the average for Brazil, which is around (CUNHA, 2011). This may have occurred because only cases indicating related genetic factors were included in the study. For example, the two cases found in Santa Quitéria primary health clinic were brothers, and there remains a third brother with undiagnosed psychiatric disorders.

In accordance with the International League against Epilepsy (ILAE), epilepsy is a brain disorder caused by a persistent predisposition in which there are manifestations of epileptic seizure, with cognitive, psychosocial, social and neurobiological consequences; and in which there must have already occurred at least one epileptic crisis in life. It presents a complex etiology; it is a multifactor disease, or one which can be triggered by several factors. Yet the association of specific genes has already been confirmed for this disorder, such as mutations in the gene *GRIN1* that cause polymicrogyria (FRY et al., 2018).

From analysis of the results it was seen that the amount of genetic disease found was relatively proportional to the number of records analyzed, and the number of families serviced at each clinic, thus, demonstrating a certain pattern regarding genetic diseases in Paulo Afonso-BA population. The primary health clinic is the first point of public health access to the population, As decentralized as possible with the aim of meeting the needs of all regions, and also being responsible to identify potential risks and demands (BRASIL, 2012). Thus, it is seen that the importance of seeking the project data from clinics that (theoretically) are the points of first contact for patients with the public health network.

In the questionnaires applied to the health care professionals, higher scores were achieved by the doctors who obtained on average: 95% correct answers. The other professionals who made up the multidisciplinary framework of the health clinics presented a more basic knowledge about genetics, such as the type and the number of chromosomes and genetic changes found in the most prevalent diseases, such as Down's syndrome. However, they presented little knowledge when asked about very specific genetic disorder topics, such as physiopathology and conducts suggested for specific diseases; among them Down's syndrome, Crohn's disease, and other genetic disorders diagnosed even within the

health clinics themselves.

The methodologies used in the research, such as reading of the records and active searches for patients in the chosen city health clinics, allowed an overview of the diseases that affect the communities, and of the users who attend the clinics. The information first analyzed was from records presenting confirmed diagnoses. Students associated to the research could examine the charts as a way of learning, aiding both their technical knowledge and acquisition of common medical terms and abbreviations in current practice. However, some charts were analyzed with difficulty due to either poor calligraphy or missing patient information.

As for the questionnaire applied to health professionals on knowledge of basic medical genetics, greater scores were observed for the physicians, involving specific knowledge about specific diseases. This can be justified by their more in-depth multidisciplinary training in genetics (NOGUEIRA, 2014). According to the Ministry of Health (BRASIL, 2000), the lack of health knowledge in care professionals represents a major obstacle to both the quality and effectiveness of assistance provided at health clinics. Facing this reality and seeking to optimize and qualify the assistance provided to the end users who suffer from genetic disorders, constant training is essential for all health professionals who work in this area.

#### 4 Conclusion

Facing the increased incidence of genetically based diseases, there is a need for greater training of health professionals towards diagnosis and elaboration of effective therapies, which would ensure significant improvements in the quality of life for patients affected by these types of sickness.

For having greater contact with the population and a deeper more realistic knowledge of each individual's health conditions, the Primary Health Attention constitutes an extremely important locale for developing actions geared towards medical genetics. They naturally possess greater sensitivity towards recognition and diagnosis of genetic diseases, to promote continued and qualified therapy.

During the project, it became apparent that the health professionals were familiar with more common diseases, such as Down's syndrome. Yet they displayed little knowledge in relation to less frequent genetic disorders. The importance of interventional training became clear afterwards, as it was observed that more qualified health professionals deal more appropriately with these diseases, minimizing (in relation to genetic disorders) the professional deficit currently present in primary health clinics.

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