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Original Article

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From emergence to extinction: the trajectory of an Outpatient Medical Genetics Service in Brazil

Do surgimento à extinção: a trajetória de um Serviço Ambulatorial de Genética Médica no Brasil

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Abstract

Introduction: The number of health professionals involved in specialized care in genetics is considered insufficient in Brazil and it is assumed that most patients and families do not receive adequate care. In addition, the availability of human and material resources for care in genetics is unequal in various regions of the country. **Objectives**: To describe the trajectory of an outpatient medical genetics service in Brazil and to highlight the importance of organizing the medical genetic care network in Brazil. **Methodology**: A quantitative, cross-sectional, and retrospective study, in which care maps of all 792 patients evaluated at the Medical Genetics Outpatient Clinic of São Carlos, São Paulo, from June 2006 to December 2018 were analyzed. Results: There was a great diversity of genetic diseases that affected people in different age groups. A genetic diagnosis was defined in 290 patients (36.6%) and ruled out in 153 patients (19.3%); 23 patients (2.9%) received genetic counseling for future offspring and in 326 patients (41.2%) a genetic diagnosis was neither defined nor ruled out. The frequency of unresolved cases explains the difficulties inherent in a clinical genetic diagnosis and barriers to accessing molecular genetic tests. **Conclusion**: Experiences such as the Medical Genetics Outpatient Clinic of São Carlos show obstacles in terms of providing adequate health services to the population in genetics, reinforcing the need to organize the genetic care network in the Brazilian Unified Health System, based on the National Policy for Comprehensive Care for People with Rare Diseases.

Keywords: medical genetics; genetic counseling; rare diseases; referral and consultation; health services research; Brazilian Unified National Health System.

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Resumo

Introdução: O número de profissionais de saúde envolvidos na assistência especializada em genética é considerado insuficiente no Brasil, e supõe-se que a maior parte dos pacientes e famílias não recebe cuidado adequado. Além disso, a disponibilidade de recursos humanos e material para atendimento em genética é muito desigual nas várias regiões do país. **Objetivos**: Descrever a trajetória de um serviço ambulatorial de genética médica no Brasil e ressaltar a importância de organizar a rede de atenção em genética médica no país. **Metodologia**: Estudo quantitativo, transversal e retrospectivo, no qual foram analisados os mapas de atendimento de todos os 792 pacientes avaliados no Ambulatório de Genética Médica de São Carlos, São Paulo, entre junho de 2006 e dezembro de 2018. Resultados: Houve grande diversidade de doenças genéticas, que acometeram pessoas de diferentes faixas etárias. Um diagnóstico genético foi definido em 290 pacientes (36,6%) e afastado em 153 pacientes (19,3%); 23 pacientes (2,9%) receberam aconselhamento genético para futura descendência e em 326 pacientes (41,2%) um diagnóstico genético não foi definido nem afastado. A frequência de casos não resolvidos explicita dificuldades inerentes ao diagnóstico clínico genético e barreiras para acesso aos testes genéticos moleculares. **Conclusão**: Experiências como a do Ambulatório de Genética Médica de São Carlos mostram os obstáculos de se prestar um serviço de saúde adequado à população, reforçando a necessidade de organizar a rede de atenção em genética no Sistema Único de Saúde, com base na Política Nacional de Atenção Integral às Pessoas com Doenças Raras.

Palavras-chave: genética médica; aconselhamento genético; doenças raras; encaminhamento e consulta; pesquisa sobre serviços de Saúde; Sistema Único de Saúde.

Introduction

Genetic diseases are numerous and diverse. It is estimated that there are between 6,000 and 7,000 different genetic diseases, which together affect about 31.5 to 73 per 1,000 individuals in the general population[1]. They can be congenital or develop throughout life and can occur in all age groups. Many of them are hereditary, affecting several individuals in the same family, and others occur by a new mutation without a family history. They are usually chronic, severe, progressive, and in addition to any personal and family impacts, they cause socioeconomic loss for the country as they increase health expenses and decrease productivity[2].

The number of health professionals involved in specialized assistance in genetics is considered insufficient in Brazil and it is assumed that most patients and families do not receive suitable care[3]. In 2020, there were 332 physicians specializing in genetics registered in the Federal Medicine Council[4]. In addition to being few, doctors, as well as genetics services, are concentrated unequally, predominantly in the Southeast and South regions[4 -7]. Undoubtedly, the lack of human and specialized service resources is an obstacle for the inclusion of assistance in medical genetics in the Brazilian Unified National Health System, called Sistema Único de Saúde (SUS)[3,6].

A first attempt to include genetics in the SUS was made in 2009 when the National Policy for Comprehensive Care in Clinical Genetics[8] was published, although it was not effectively implemented. The National Policy for Comprehensive Care for People with Rare Diseases was implanted in 2014[9] and, as approximately 70% of rare diseases have a genetic etiology, a new opportunity arose to organize a care network in medical genetics in SUS[3,10]. The National Policy for Comprehensive Care for People with Rare Diseases establishes the actions that must be developed by primary healthcare and those that must be developed by specialized healthcare specifically. For this purpose, this Policy recognizes and qualifies

Specialized Care Services for Rare Diseases and Reference Services for Rare Diseases[9].

A survey conducted on the Brazilian Society of Medical Genetics and Genomics website[11] identified 108 public medical genetics services, including highly complex hospital services and outpatient services for specific diseases, which were distributed unequally: 56 services in the Southeast, 23 in the Northeast, 15 in the South, 8 in the North and 5 in the Center-West. Most of these centers were integrated with hospitals and/or public university institutions.

Despite the repressed demand from people who need specialized care in the area, by December 2020, the Ministry of Health had enabled only one "Specialized Care Service for Rare Diseases" and 16 "Reference Services for Rare Diseases", in line with the National Policy for Comprehensive Care for People with Rare Diseases, nationwide[11]. There are many difficulties in accessing genetic tests (especially in services that are not enabled), which are usually offered in a research context and are carried out only during a certain period with no continuous offer over time[6,7].

In 2006, in São Carlos, a city in São Paulo State, an outpatient medical genetics service was set up, as an academic extension project, which was part of the health-school network[12]. This service worked regularly until December 2018 when attendance was discontinued. The aim of this study was to describe the trajectory of the Medical Genetics Outpatient Clinic of São Carlos, São Paulo, and to perform a retrospective analysis of the work conducted during the period of operation, characterizing the population served, to highlight and discuss the importance of organizing the genetic care network in Brazil.

Methodology

This is a quantitative, cross-sectional, and retrospective study approved by the Ethics Committee on Human Research at the Federal University of São Carlos (CAAE 66080517.2.0000.5504). The city of São Carlos is located in the geographic center of São Paulo State, Brazil and has an estimated population of approximately 250,000 inhabitants. In 2010, it had a human development index of 0.805 and was considered the 28th most developed municipality in the country[13].

The Medical Genetics Outpatient Clinic of São Carlos operated from June 2006 to December 2018 at the Municipal Center of Health Specialties (CEME in Portuguese). Two doctors, who were both university professors, attended patients on a weekly basis and were often accompanied by medical students. Officially, two medical appointments were offered to new patients and three appointments to patients who were already in outpatient follow-up. Sometimes, other arrangements regarding these vacancies were made over the years.

The Outpatient Clinic was the only public genetics service in the city, also meeting the demands of eight other cities in the micro-region[12] and totalizing a coverage area of about 530,000 people[13].

The only genetic test routinely offered at the Outpatient Clinic during the whole period of operation was the conventional karyotype, performed by contracting outsourced laboratory services by the Municipal Health Department as this complementary test is a diagnostic procedure that SUS covers. The test was performed using a culture of peripheral blood lymphocytes with G banding staining and resolution between 400-550 bands.

The cytogenetic study was carried out in patients treated at the Outpatient Clinic using the following clinical indications: suspicion of classic chromosomal syndromes; multiple congenital defects, with or without facial dysmorphism; intellectual disability or global developmental delay of undefined etiology, with or without facial dysmorphism; pathological short stature in

girls; genital ambiguity; infertility; primary amenorrhea and recurrent miscarriage.

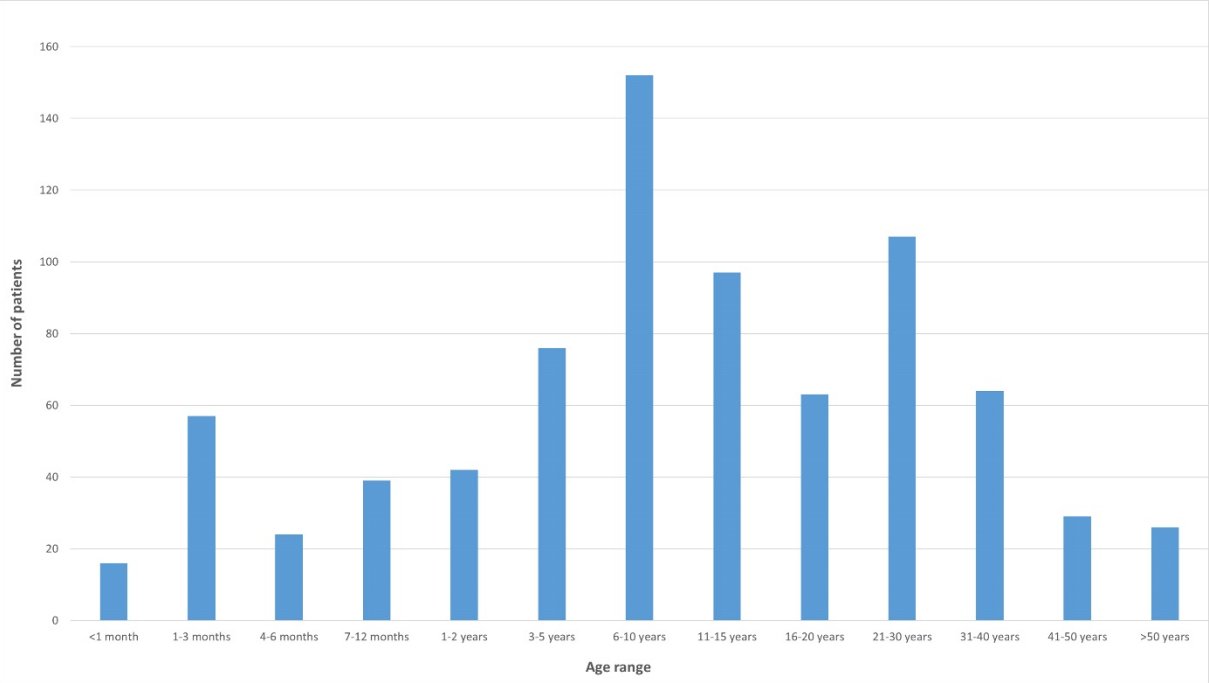
For retrospective analysis of attendance, data collection was performed by analyzing the "Daily Medical Care Maps" of patients who attended the Outpatient Clinic between June 2006 and December 2018. There was no exclusion criterion, and all the "Maps" of the patients seen in the mentioned period were included in the review. The "Daily Medical Care Maps" are files filled out daily, after outpatient clinical care, containing basic information about these consultations. From the revised "Maps", information was extracted on the year of service; the patient's age and sex; reason for referral; and genetic diagnosis, when defined.

The collected data were analyzed and categorized using descriptive statistics. The results were discussed and examined using the literature and the clinical experience of the authors, adopting an interpretive and comprehensive approach.

Results

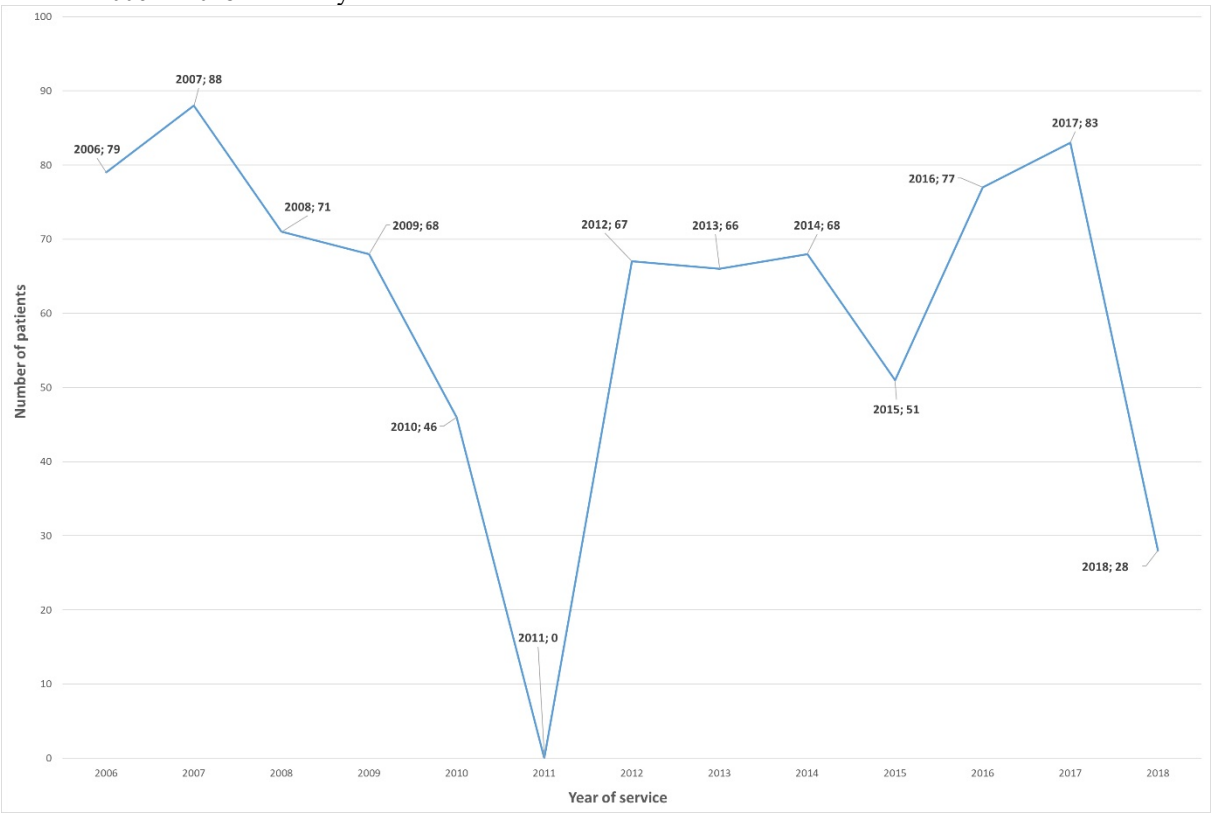
During the study period, 792 different patients were included, of which 48 (6%) were related. Regarding gender, 397 (50.1%) were female, 385 (48.6%) were male and in 10 situations (1.3%) the patient's gender was not determined by incomplete information. The age varied between 5 days of life and 79 years (Figure 1), but predominantly the young population: 71.5% of the patients were under 20 years old.

Figure 1: Distribution of 792 patients seen at the Medical Genetics Outpatient Clinic of São Carlos, SP from 2006 to 2018 by age group.



The distribution of attendance over the years (Figure 2) showed an interruption of new cases during 2011 and a decrease in this number in 2018.

Figure 2: Distribution of the number of new patients seen at the Medical Genetics Outpatient Clinic of São Carlos, SP from 2006 to 2018 over the years.



The reasons for referral of different patients were categorized and are shown in Table 1. Tables 2, 3 and 4 present the genetic diagnoses in 290 situations (36.6%), in which this one was defined.

Table 1: Reasons for initial referral to the Medical Genetics Outpatient Clinic of São Carlos, SP from 2006 to 2018 (N=792).

REASON FOR REFERRAL	N	%
Isolated intellectual disability or with minor facial dysmorphia	207	26.1
Mild	55	6.9
Moderate	93	11.7
Severe to profound	59	7.4
Multiple malformations, including or not intellectual disabilities	144	18.2
Recurrent miscarriage	46	5.8
Isolated congenital malformation	43	5.4
Short stature	43	5.4
Genodermatosis/hamartomatosis suspected	40	5.0
Isolated Global developmental delay from children under the age of 5 years old.	40	5.0
Clinical suspected Down syndrome	37	4.7
Genetic counseling for offspring	28	3.5
History of perinatal death of fetus with multiple birth defects	14	1.8
Consanguineous couple	6	0.7
Positive family history of previously diagnosed genetic disease	6	0.7
Advanced maternal age	2	0.2
Suspected connective tissue disease and/or marfanoid habitus	26	3.3
Child with facial dysmorphia, without major dysfunctions or malformations	25	3.1
Suspected neurogenic disease	21	2.6

REASON FOR REFERRAL	N	%
Autism spectrum disorder without intellectual disability	17	2.1
Visual impairment	17	2.1
Suspected hereditary cancer syndrome	15	1.9
Male infertility	13	1.6
Deafness or significant hearing loss	10	1.3
Osteochondrodysplasia	7	0.9
Disturbance of sexual differentiation and/or determination	5	0.6
Female pubertal delay and / or primary amenorrhea	4	0.5
Male pubertal delay	3	0.4
Child assessment resulting from incestuous consanguineous	1	0.1

Table 2: Distribution of patients with chromosomal alterations, that were attended at the Medical Genetics Outpatient Clinic of São Carlos, SP from 2006 to 2018, in which a genetic diagnosis was defined (N = 77).

GENETIC DIAGNOSIS	N	%
Chromosomal abnormalities identified by the conventional karyotype		
Numerical chromosomal disorders	58	20.0
Down syndrome	33	11.4
Edwards syndrome	5	1.7
Patau syndrome	2	0.7
Turner syndrome	6	2.1
Klinefelter syndrome	6	2.1
Double Y syndrome	3	1.0
Triple X syndrome	1	0.3
Abnormalities of undefined chromosomes (chromosomal markers)	2	0.7
Chromosomal polymorphisms	7	2.4
Imbalanced structural chromosomal disorders	6	2.1
Balanced structural chromosomal disorders	6	2.1

Table 3: Distribution of patients with non-chromosomal genetic diseases, seen at the Medical Genetics Outpatient Clinic of São Carlos, SP from 2006 to 2018, in which a genetic diagnosis was defined (N = 165).

GENETIC DIAGNOSIS	N	%
Non-chromosomal genetic disorders		
Multisystemic syndromes	58	20.0
X-fragile syndrome	8	2.7
Crouzon syndrome	7	2.4
Prader-Willi syndrome	5	1.7
Cornelia de Lange syndrome	4	1.4
Silver-Russel syndrome	3	1.0
Robinow syndrome	3	1.0
Williams syndrome	3	1.0
Stickler syndrome	2	0.7
22q11.2 microdeletion syndrome	2	0.7
Joubert syndrome	2	0.7
Sotos syndrome	2	0.7
Goldenhar syndrome	2	0.7
Kartagener syndrome	1	0.3
Waardenburg syndrome	1	0.3
Seckel syndrome	1	0.3
Beckwith-Wiedemann syndrome	1	0.3
Lowe syndrome	1	0.3
Peters Plus syndrome	1	0.3
Mucopolysaccharidosis type IV-A	1	0.3
Alport syndrome	1	0.3
Pfeiffer type 2 syndrome	1	0.3
Cohen syndrome	1	0.3

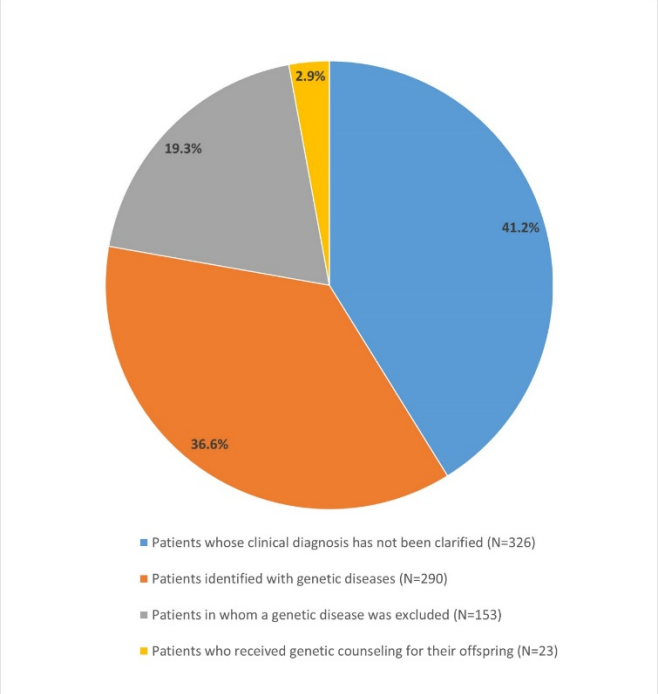
GENETIC DIAGNOSIS	N	%
Treacher Collins syndrome	1	0.3
Noonan syndrome	1	0.3
Frontonasal dysplasia syndrome	1	0.3
1p32.3 microdeletion and15p11.2 microduplication syndrome	1	0.3
2q36.1q36.3 microdeletion syndrome	1	0.3
Genodermatosis/hamartomatosis	35	12.1
Neurofibromatosis type 1	25	8.6
Pigmentary incontinence	3	1.0
Unspecified ectodermal dysplasia	2	0.7
Klippel Trenaunay Weber syndrome	2	0.7
Autosomal recessive bullous epidermolysis	1	0.3
Gorlin syndrome	1	0.3
Hypomelanosis of Ito	1	0.3
Predominantly neurogenic diseases	23	7.9
Duchenne muscular dystrophy	8	2.7
Unspecified autosomal dominant hereditary motor sensitive neuropathy	3	1.0
Unspecified Congenital myopathy	4	1.4
Unspecified autosomal dominant familial spinocerebellar ataxia	2	0.7
Steinert myotonic dystrophy	2	0.7
Progressive spinal amyotrophy type 1	1	0.3
Friedreich's ataxia	1	0.3
Huntington Disease	1	0.3
Metachromatic leukodystrophy	1	0.3
Predominantly connective tissue diseases	15	5.2
Marfan syndrome	12	4.1
Ehlers-Danlos syndrome	3	1.0
Hereditary cancer syndromes	11	3.8
Hereditary breast and ovarian cancer	6	2.1
Intestinal adenomatous polyposis	3	1.0
Familial retinoblastoma	1	0.3
Li Fraumeni syndrome	1	0.3
Osteochondrodysplasias	9	3.1
Achondroplasia	2	0.6
Osteogenesis imperfecta	2	0.6
Pycnodysostosis	1	0.3
X-linked hypophosphatemic rickets	1	0.3
X-linked spondyloepiphyseal dysplasia	1	0.3
Unspecified congenital spondylomethaepiphyseal dysplasia	1	0.3
Thanatophoric dwarfism	1	0.3
Sensory defects	8	2.7
Usher syndrome	3	1.0
Deafness with unspecified autosomal recessive inheritance	2	0.6
Deafness with unspecified autosomal dominant inheritance	1	0.3
Pigmentary retinosis with autosomal recessive inheritance	1	0.3
Leber's hereditary optic neuropathy	1	0.3
Defects of differentiation and sexual determination	3	1.0
True hermaphroditism 46, XX	1	0.3
Morris syndrome	1	0.3
Rokitansky syndrome	1	0.3
Sequence and Association	3	1.0
Septo-optic dysplasia	1	0.3
VACTERL association	1	0.3
Pierre Robin sequence	1	0.3

Table 4: Distribution of patients with isolated congenital malformations, seen at the Medical Genetics Outpatient Clinic of São Carlos, SP from 2006 to 2018, in which a genetic diagnosis was defined (N = 48).

GENETIC DIAGNOSIS	N	%
Isolated congenital malformations		
Defect of central nervous system	16	5.5
Face defect	9	3.1
Heart defect	9	3.1
Limb defect	9	3.1
Defect of the chest, abdomen or digestive system	3	1.0
Genitourinary defect	2	0.6

Figure 3 demonstrates outpatient resolvability. After clinical evaluation, genetic disease was ruled out in 153 cases (153/792; 19.3%). Most of these patients were in a situation of recurrent miscarriage (43/153; 28.1%); cases in which children had familial and/or constitutional short stature or even pathological short stature of non-genetic cause (37/153; 24.2%); children with facial dysmorphisms, with no definition or without major malformation and considered eutrophic (25/153; 16.3%); cases of intellectual disability and/or congenital defects due to teratogenesis (13/153; 8.5%); and people with intellectual disabilities secondary to neonatal anoxia (10/153; 6.5%).

Figure 3: Resolubility of the Medical Genetics Outpatient Clinic of São Carlos, SP from 2006 to 2018.



The diagnosis was not defined or ruled out in 326 patients. Most of them were people with isolated intellectual disabilities or with minor facial dysmorphisms (174/326; 53.4%); cases of multiple malformations, including or not an intellectual disability, without syndromic diagnosis established (77/326; 23.6%), children with isolated global developmental delay (37/326; 11.3%); and children with autism spectrum disorder without associated intellectual disability (17/326; 5.2%).

Twenty-three patients received genetic counseling for their future offspring. Among these, nine patients had a

history of perinatal death of a fetus diagnosed with a genetic disease, six patients had a positive family history of a previously diagnosed genetic disease, six people came to the Outpatient Clinic for preconception genetic counseling because of inbreeding and two people because of advanced maternal age.

Discussion

The Medical Genetics Outpatient Clinic of São Carlos started operating in June 2006. At the time, the Department of Medicine at UFSCar had just been opened and the professors maintained a close relationship with the managers and attended health facilities in the municipality. The invitation to open the Outpatient Clinic, which functioned since the beginning as a teaching-assistance integration activity, came from the coordinator of the Municipal Center of Specialties in Health (CEME). At that time, working conditions were agreed related to physical space, scheduling appointments, the number of vacancies made available, the collaboration of nursing, social service, and administrative support teams from CEME, as well as access to the karyotype by G banding for patients with clinical indications.

Considering the population coverage and the prevalence of genetic diseases, it is estimated that approximately 15,900 to 37,100 citizens, living in the cities comprised by Considering the population covered and the prevalence of genetic diseases, it is estimated that approximately 15,900 to 37,100 citizens, residing in the cities comprised by the Medical Genetics Clinic of São Carlos, will need health care in genetics at some point of their lives. The Medical Genetics Outpatient Clinic of São Carlos met part of the needs of this population during the period of its operation. However, many patients were not identified as people who would benefit from assistance in the field of genetics or did not have access to the Outpatient Clinic.

The suspicion of genetic diseases or situations of risk for their development is an expected competence for health professionals working in primary healthcare, which is not always sufficiently developed[3,14,15]. The systematic use of instruments, such as questionnaires and forms, which help to recognize risk factors and suspicious situations, has been identified as a useful strategy to be incorporated by the primary healthcare network to facilitate the identification of people with genetic diseases, which allows for regulated referral to specialized services[16,17].

Access to specialized genetic care is another problem. The number of medical genetic services in the country is insufficient, and although most of these services are linked to public education institutions, they are not always located within a region and state regulatory systems of the SUS network. Situations in which patients are informally referred to services through direct communication with genetic physicians, who are responsible for these services, are not unusual. Alternatives described in the literature to circumvent the lack of genetics specialists in the country include the organization of itinerant genetics services[16] and the use of telemedicine[18].

Concerning the flow of care at the Medical Genetics Outpatient Clinic of São Carlos over the years, the absence of new patients attended in 2011 calls attention. This happened because in that year, one of the professors responsible for attending was on leave. On this occasion, there was an attempt to replace this doctor offering a public examination to a geneticist physician, but there were no interested candidates. Genetics is the medical specialty with the fewest specialists in Brazil[4]. Although the Ministry of Health considers priority specialty, there are only 11 genetic residence programs in the country, which offer a total of 27 new resident physicians opening per year and,

except for one program linked to the Health Department of the Federal District and another program linked to the Federal University of Bahia, all the others are located in the South and Southeast regions[15]. Therefore, training specialists in the field has been slow and unequal.

The reasons for referring patients to the Medical Genetics Outpatient Clinic of São Carlos showed the great diversity of genetic diseases, which affect people in very different age groups and affect different organs and systems. The frequency of unsolved cases indicates the difficulties inherent in the genetic clinical diagnosis, in addition to the limitation of accessing molecular genetic tests. More common situations of undefined diagnoses, which are related to intellectual disability, global developmental delay in children, and the presence of multiple congenital defects, the patients would benefit from a systematic investigation of X-fragile syndrome including chromosomal analysis by microarray tests and by complete exome sequencing, as recommended in the literature[20]. In 12 years of operation at the Outpatient Clinic, some patients had access to molecular genetic tests, through research projects[21-23] or because they were able to negotiate the performance of the tests directly with the Municipal Health Department through administrative processes, or even through judicialization of SUS. However, the offer and access to molecular genetic tests occurred irregularly over time. In particular, patients who did not live in the city of São Carlos found it more difficult to carry out complementary exams as they should be authorized by the Health Departments of their municipalities of origin.

Additional diagnostic difficulties were perceived in situations of neurogenetic diseases, in which the clinical diagnosis was often established, but specific etiological genetic diagnosis remained inconclusive, largely because of inaccessibility to molecular genetic testing[24] and muscle biopsy with adequate pathological study[25]. There were also five perinatal death situations of fetuses with multiple birth defects, that is, polymalformed fetuses, in which it was not possible to perform a necropsy, making it impossible to define syndromic diagnoses and impairing family genetic counseling[26].

Since the release of the National Policy for Comprehensive Care for People with Rare Diseases, there have been several negotiations between the Outpatient Clinic coordination and the Municipal Health Department of São Carlos to enable the qualification of the Outpatient Clinic with the Ministry of Health, as a “Specialized Care Service for Rare Diseases” in line with the National Policy for Comprehensive Care for People with Rare Diseases[9]. The negotiations were hampered by changes in municipal management and the lack of a stable, long-term governance plan for the health area in the municipality. Municipal management plays a key role in the health care of the population; this level of public management makes citizens' demands for health care explicit, and at this level of management state and federal governments who offer support must guarantee the offer and access to services, according to the required health needs. This is a challenge that municipal management has in the field of public policies[27].

All contingencies considered, the Medical Genetics Outpatient Clinic of São Carlos operated regularly until June 2018. As of July 2018, the municipal health administration made it difficult for the Outpatient Clinic to operate as they imposed limitations regarding the physical space where the visits took place in the CEME, and bureaucratizing the request of the conventional karyotype, a complementary test that until then was performed routinely. It can be hypothesized that these obstacles were related to the presence of medical students in the CEME, which changed the dynamics of the environment, as well as to the increased

demand for molecular genetic testing, which led to more administrative and judicial processes. Faced with this scenario, and perceiving a loss in the health care offered to patients, the Outpatient Clinic coordination opted to suspend the admission of new patients as of August 2018 and interrupt the appointments in December 2018. The municipal administration was officially communicated about the suspension of consultations three months in advance so that it could restructure the line of care for patients in longitudinal follow-up.

In 2019, a new municipal contest was held for a geneticist doctor in the municipality and, this time, a specialist doctor was approved, although he has not been asked to take up the position so far. It is expected that when this professional is called, the Outpatient Clinic will be reopened and the partnership with the University may be resumed.

This is not the first report of stopping a medical genetics service in Brazil. Between 1969 and 1989, the medical genetics service at the Federal University of Bahia had a major impact on local public health. However, important administrative changes reduced it for many years to a small genetics practice[29], until the resumption of its structure in the early 2000s. Currently, Professor Edgard Santos University Hospital in Salvador is one of 16 "Reference Services for Rare Diseases" enabled by the National Policy for Comprehensive Care for People with Rare Diseases (SBGM, 2020). In 1996, the genetics department of the Menino Jesus Children's Hospital, which operated for four years in the city of São Paulo, was closed down. This service stopped considering changes in the municipal health system of São Paulo on the occasion of the Health Care Plan[30]. In both situations, there were no public policies related to genetic care in SUS, as they currently exist.

Even today, it can be observed that the existence of public policies in the area does not guarantee the organization of a care network and a line of health care for patients with genetic diseases in SUS. People with rare genetic diseases continue to face long therapeutic trajectories until diagnosis; the unfamiliarity of non-geneticist physicians about rare diseases; difficulties in accessing specialists, diagnostic and complementary tests; and difficulties in accessing medicines and high-cost food supplies[10]. Additionally, late diagnosis often prevents the patient from receiving appropriate and timely treatment, impacting their quality of life[2,10].

Conclusions

There is a common misconception that genetic diseases are rare and that healthcare in the area would be costly. However, genetic diseases in their entirety affect a large number of people of different age groups. Although the availability of human and material resources for genetic care is very unequal in various regions of Brazil, adequate health care is the right of these patients and can be achieved with the effective implementation of the National Policy for Comprehensive Care for People with Rare Diseases.

Health regulation in the area of genetics in the country is a subject that needs to be better studied and understood. Experiences such as that of the Medical Genetics Outpatient Clinic of São Carlos, São Paulo, show the obstacles of providing an adequate health service to the population, reinforcing the need to organize the genetic care network in SUS, respecting the attributions of primary healthcare and enabling specialized care services to reduce health inequalities among people with rare genetic diseases in the country.

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Conflict of interest

The authors declare that there is no conflict of interest.

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