REVISTA BRASILEIRA DE MEDICINA DE FAMÍLIA E COMUNIDADE
RBMFC

www.rbmfc.org.br
ISSN 2197-7994

Quality Improvement

Training of a Family and Community Medicine resident in the context of the National Policy of Comprehensive Care for People with Rare Diseases

Formação de um residente de Medicina de Família e Comunidade no contexto da Política Nacional de Atenção Integral às Pessoas com Doenças Raras

Formación de un residente de Medicina Familiar y Comunitaria en el contexto de la Política Nacional de Atención Integral a Personas con Enfermedades Raras

Laércio Moreira Cardoso Júnior[1], Mariana Machado Aragão[1,2], Larissa Souza Mario Bueno[3]

[1]Fundação Estatal Saúde da Família, Programa de Residências Integradas em Saúde da Família – Camaçari (BA), Brazil.
[2]Prefeitura Municipal de Camaçari, Secretaria de Saúde – Camaçari (BA), Brazil.
[3]Universidade Federal da Bahia, Hospital Universitário Professor Edgard Santos, Serviço de Genética Médica – Salvador (BA), Brazil.

Abstract

Problem: Changes in the Brazilian morbidity and mortality profile have highlighted the importance of genetic diseases, but epidemiological data are still limited. Since 2014, the National Policy for Comprehensive Care of People with Rare Diseases aims to foster comprehensive care in The Brazilian Unified Health System (*Sistema Único de Saúde* – SUS). However, primary care professionals are not yet sufficiently trained to deal with genetic and rare diseases. The objective of the study was to present the experience gained by a Family and Community Medicine resident in a reference service in genetic and rare diseases. **Methods:** This is an experience report of an elective internship developed during eight weeks at the Medical Genetics Service at Hospital Universitário Professor Edgard Santos of Universidade Federal da Bahia (HUPES-UFBA). The internship consisted of rotations in outpatient clinics, laboratory, infirmary, and participation in theoretical classes. **Results:** Residents had contact with genetics knowledge and tools that are useful to their practice as a family physician, assisting in the care of people with genetic and rare diseases. They also identified how primary health care tools and principles enhance care in Medical Genetics. **Conclusion:** The experience gave the residents a better understanding of their role within the line of care for rare diseases, reinforcing the primary care professionals' responsibility for comprehensive care.

Keywords: Primary health care; Family practice; Rare diseases; Medical genetics.

Corresponding author:
Laércio Moreira Cardoso Júnior
E-mail: laerciop53@gmail.com
Funding:
No external funding.
Ethical approval:
Not applicable.
Provenance:
Not commissioned.
Peer review:
external.
Received: 07/10/2021.
Approved: 10/23/2022.
Guest editor:
Severina Alice da Costa Uchoa

How to cite: Cardoso Júnior LM, Aragão MM, Bueno LSM. Training of a Family Community Medicine resident in the context of the National Policy of Comprehensive Care for People with Rare Diseases. Rev Bras Med Fam Comunidade. 2023;18(45):3155. https://doi.org/10.5712/rbmfc18(45)3155

Resumo

Problema: Mudanças no perfil de morbimortalidade brasileiro têm evidenciado a importância das doenças genéticas, porém os dados epidemiológicos ainda são limitados. Desde 2014, a Política Nacional de Atenção Integral às Pessoas com Doenças Raras visa fomentar a assistência integral no Sistema Único de Saúde (SUS). Entretanto, os profissionais da atenção primária ainda não são suficientemente capacitados para a abordagem das doenças genéticas e raras. O objetivo do estudo é apresentar a experiência adquirida por um médico residente em Medicina de Família e Comunidade em um serviço de referência em doenças genéticas e raras. **Método:** Trata-se de um relato de experiência de estágio eletivo desenvolvido durante oito semanas no Serviço de Genética Médica do Hospital Universitário Professor Edgard Santos da Universidade Federal da Bahia (HUPES-UFBA). O estágio foi composto de rotações em ambulatórios, laboratório, enfermaria e participação em aulas teóricas. **Resultados:** O residente teve contato com conhecimentos e ferramentas de genética que são úteis à sua prática como médico de família e comunidade, auxiliando na atenção às pessoas com doenças genéticas e raras. Também identificou como ferramentas e princípios da atenção primária à saúde potencializam o cuidado em genética médica. **Conclusão:** A experiência situou o residente quanto ao seu papel na linha de cuidado em doenças raras, reforçando a responsabilidade do profissional da atenção primária na assistência integral.

Palavras-chave: Atenção primária à saúde; Medicina de família e comunidade; Doenças raras; Genética médica.

Resumen

Problema: Los cambios en el perfil de morbilidad y mortalidad brasileño han puesto en atención la importancia de las enfermedades genéticas, pero los datos epidemiológicos aún son limitados. Desde 2014, la Política Nacional de Atención Integral a Personas con Enfermedades Raras tiene como objetivo fomentar la atención integral en el Sistema Único de Salud (SUS). Sin embargo, los profesionales de atención primaria aún no están lo suficientemente capacitados para hacer frente a las enfermedades genéticas y raras. El objetivo del estudio es presentar la experiencia adquirida por un médico residente en Medicina Familiar y Comunitaria en un servicio de referencia en genética y enfermedades raras. **Método:** Se trata de un informe de experiencia de estancia electiva desarrollada durante ocho semanas en el Servicio de Genética Médica de HUPES-UFBA. La estancia consistió en rotaciones en consultas externas, laboratorio, enfermería y participación en clases teóricas. **Resultados:** El residente tuvo contacto con conocimientos y herramientas genéticas que le son útiles para su práctica como médico de familia y comunitario, ayudando a atender a personas con enfermedades genéticas y raras. También identificó cómo las herramientas y los principios de la atención primaria de salud mejoran la atención en Genética Médica. **Conclusión:** La experiencia colocó al residente en su rol dentro de la línea de atención en enfermedades raras, reforzando la responsabilidad del profesional de atención primaria en la atención integral.

Palabras clave: Atención primaria de salud; Medicina familiar y comunitaria; Enfermedades raras; Genética médica.

INTRODUCTION

In recent years, changes have been observed in the demographic and epidemiological profile of the Brazilian population, resulting in a greater importance of noncommunicable chronic diseases, combined with the persistence of infectious diseases and external causes in the morbidity and mortality profile.[1] These changes also generated a greater representation of genetic diseases in morbidity and mortality statistics.[2] In Brazil, epidemiological data on genetic diseases are limited, with information obtained from regional studies or data from the information systems of the Unified Health System (*Sistema Único de Saúde* – SUS), largely underreported.[3] So far, there is no national census on the representation of these diseases in the country, but there is a multicenter project on the subject in progress.[4]

In general, it is estimated that genetic diseases affect 3 to 7% of the world's population (31.5 to 73.0 per thousand individuals), which signals genetic diseases as a public health problem.[5,6] Regarding birth defects, which are totally or partially determined by genetic factors, have an estimated prevalence of 5% among live births. It was observed that, between the years 1980 and 2000, birth defects went from the fifth to the second cause of infant mortality in the first year of life.[7] Birth defects also accounted for more than a third of all pediatric hospitalizations in university hospitals and are responsible for an increase in hospitalization costs, reinforcing the importance of policies for rare diseases in Brazil.[3]

Among the policies to deal with the problem of genetic diseases instituted by the Brazilian government, the National Policy for Comprehensive Care for People with Rare Diseases (*Política Nacional de Atenção Integral às Pessoas com Doenças Raras* – PNAIDR) was regulated in 2014.[8] It aims to guarantee comprehensive care in SUS for people with rare diseases, including diseases of genetic origin and also rare non-genetic diseases. The Ministry of Health defines a rare disease as one that affects up to 65 people per 100,000 individuals (or 1.3 people per 2,000 individuals). There are from 6 to 8 thousand different types of rare diseases, 80% of which are of genetic etiology; the other 20% comprises infrequent immunological and infectious diseases. Although rare from an individual point of view, when these diseases are combined, they represent a significant epidemiological impact. Many of them are chronic, progressive, and degenerative.[6,9,10]

The role of Primary Care or Primary Health Care (PHC) is recognized as the first contact and preferred route for users to the health system.[2] The essential elements of PHC have the potential to maximize the recognition of genetic problems at this level of care, as well as how to advise on environmental risk factors and family predispositions.[2,11] In addition, as it is the main gateway to the Health Care Networks (*Redes de Atenção à Saúde* – RAS), it must have qualified professionals to deal with the comprehensive care of people with genetically determined conditions.[2,11]

The PNAIDR also recognizes PHC as preponderant in the integral care of people with genetic and/or rare diseases, defining that the line of care for people with rare diseases permeates both basic and specialized care. It is primarily incumbent upon PHC to: promote actions aimed at health promotion, health education, early diagnosis, timely follow-up and referral, coordination and maintenance of care for users with rare diseases, among others.[6,8,9]

Despite this recognized importance, there are still difficulties in the implementation of care for people with rare diseases at the PHC level, mainly due to the lack of information on the part of professionals and the insufficient approach, which results in obstacles in the diagnosis and referral.[2,11] Some experiences of articulation of genetics with PHC were developed in a timely manner, mainly through educational activities.[11-15]

The training of professionals to care for people with genetic diseases has been advocated since 2014 by the national guidelines for the medical course.[9] Specifically in the training of medical residency in Family and Community Medicine (FCM), the approach to genetic diseases is a required competence, beyond the importance shown in PHC as a whole.[16] This is not the reality of most undergraduate courses in Medicine in the country, nor the training of other health professionals working in PHC, who report insufficient knowledge of genetics and rare diseases.[17-19]

Motivated by this problem, the authors developed the experience of inserting a resident doctor in FCM in a reference service in genetic and rare diseases. The objective of this article is to present the experience acquired during the internship developed by the resident physician, one of the authors of this work. It aims to explain how the performance in the internship could contribute to the formation of the intern as a family physician and to the expansion of the care provided to people with genetic and rare diseases at the PHC level.

METHODS

This is a descriptive, quality improvement article of an elective internship developed by a resident physician in a reference service in genetic and rare diseases. The elective internship comprises residency

training in FP of the Integrated Residency Program in Family Health of *Fundação Estatal Saúde da Família* (FESF-SUS) and *Fundação Oswaldo Cruz* (Fiocruz Bahia). It recommends the insertion of the resident in an area of interest, as long as it is articulated with the objectives of PHC.

The internship was carried out for eight weeks, between September and October 2020, with a weekly workload of 40 hours (totaling 320 hours), between practical, theoretical, and theoretical-practical activities. The development site was the Medical Genetics Service of the University Hospital Professor Edgard Santos of Universidade Federal da Bahia (*Serviço de Genética Médica do Hospital Universitário Professor Edgard Santos da Universidade Federal da Bahia* – SGM/HUPES/UFBA).

The internship was structured in shifts in outpatient clinics and in the laboratory of the SGM, participation in theoretical classes, clinical meetings, visits to wards, and shifts dedicated to case studies. The proposed activities aimed to provide the resident with general knowledge about the main areas of activity in Medical Genetics and rare diseases, focusing on knowledge that could be useful to family and community physicians and in their work process. The activities developed and perceptions were recorded in an internship report developed at the end of the period and served as support for the elaboration of this article. The main references of theoretical-methodological support to the experience are cited throughout the text.

RESULTS AND DISCUSSION

The SGM/HUPES was founded in 1969 and is a pioneer in teaching, research, and assistance in Medical Genetics in the state of Bahia.[20] Since then, the service has become a reference in assisting people with genetic and rare diseases, providing an important service to the community of the state and SUS.[21] In 2019, HUPES was accredited by the Ministry of Health as a reference service in rare diseases (*serviço de referência em doenças raras* – SRDR), with the SGM responsible for assistance in the axis of rare diseases of genetic origin.[22] The HUPES Complex (COM-HUPES/UFBA) is integrated by HUPES, Ambulatório Magalhães Neto (AMN), and *Centro Pediátrico Professor Hosannah de Oliveira* (CPPHO), currently administered by *Empresa Brasileira de Serviços Hospitalares* (EBSERH). The HUPES Complex is a reference in medium and high complexity, with assistance fully provided by SUS.[23]

The SGM is made up of a multidisciplinary team consisted of medical geneticists, doctors from other specialties, and professionals in the biomedical area. It has outpatient clinics in various fields of genetics, a Medical Genetics laboratory, and the Teratogenic Agents Information Service.[21] Additionally, resident physicians, academics and UFBA graduate students work in the service. Several surveys are carried out at the service, with funding from the main development agencies and in collaboration with other centers in the country.

External patients access the service through *Sistema Vida* and the State Regulation Center, where patients are referred for specialized evaluation by a geneticist. This is the service's main line of communication with PHC. These patients are evaluated at the screening outpatient clinic and subsequently referred to specific outpatient clinics. The service also performs genetic tests through SUS and partnerships with other centers and research projects.[21] Thus, in line with PNAIDR, the SGM acts as a point of interest in the line of care for rare diseases in Bahia, exercising the role of specialized care and genetic counseling.

The internship developed by the resident physician was carried out on the SGM premises and units linked to COM-HUPES. The rotations and activities described in Table 1 were proposed during the first

Table 1. Practical and theoretical-practical activities developed during the elective internship.

Elective Internship		
Program	**Family and Community Medicine — FESF-SUS/FIOCRUZ-BA**	
Internship unit	**Medical Genetics Service — COM-HUPES/UFBA/EBSERH**	
Month	**Proposed activities**	**Acting areas**
September	Cytogenetics Laboratory	Medical Genetics Service Laboratório de Genética Médica Ambulatório Magalhães Neto
	Consultation Outpatient Clinic	
	Oncogenetics Outpatient Clinic	
	General Genetics Outpatient Clinic	
	Sexual Development Anomalies Outpatient Clinic	
	Skeletal Dysplasias Outpatient Clinic	
	Screening Outpatient Clinic	
	Clinical Meeting	
	Case studies	
	Theoretical classes of the MRP of Genetics	
October	Cytogenetics Laboratory	Medical Genetics Service Laboratório de Genética Médica Ambulatório Magalhães Neto Centro Pediátrico Professor Hosannah de Oliveira
	Consultation Outpatient Clinic	
	Osteogenesis Imperfecta Outpatient Clinic	
	General Genetics Outpatient Clinic	
	Inborn Errors of Metabolism / Neurometabolic Outpatient Clinic	
	Neurogenetics Outpatient Clinic	
	Consultations in Pediatric Ward	
	Clinical Meeting	
	Case studies	
	Theoretical classes of the MRP of Genetics	

FESF-SUS/FIOCRUZ-BA: Fundação Estatal Saúde da Família/Sistema Único de Saúde/Fundação Oswaldo Cruz – Bahia; COM-HUPES/UFBA/EBSERH: Complexo do Hospital Universitário Professor Edgard Santos/Universidade Federal da Bahia/ Empresa Brasileira de Serviços Hospitalares; MRP: medical residency programs.

and second months. Some of them took place in collaboration with the resident physicians of the medical residency programs (MRP) in Medical Genetics and Pediatrics at the institution.

Among the theoretical activities developed, there was participation in classes at the MRP in Medical Genetics, held virtually due to the context of the pandemic. Some topics addressed were: neonatal screening and general approach to inborn errors of metabolism; assessment elements in dysmorphology and physical examination; characteristics of genetic tests; screening and counseling in cancer genetics. These classes had an expository character and were presented by preceptors or invited professionals.

The other theoretical activities developed by the resident corresponded to participation in weekly clinical meetings, case study shifts, in addition to discussions held before and after the outpatient clinics. These activities, as they are based on more active methodologies, were evaluated by the resident as a more significant learning tool. In the weekly clinical meeting, residents presented and discussed with the preceptorship the

cases that would be seen in the following week, trying to elaborate diagnostic hypotheses and develop clinical reasoning in genetics. The study shifts were dedicated to preparing and deepening the cases, as well as complementing the theoretical support based on reference materials in genetics and scientific literature.

The laboratory activity was developed in the cytogenetics sector of the SGM and presented the resident with laboratory tests in genetics, focusing on the karyotype. The resident had contact with the stages of performing the technique, the overview of chromosome analysis, and the main indications of the exam. The space was important for a better understanding of the logistics of a laboratory unit and the importance of dialogue between the clinic and the laboratory for greater investigative effectiveness. Visits to the CPPHO pediatric ward were aimed at evaluating hospitalized patients in need of specialized evaluation in Medical Genetics. In some of these consultation requests, residents had the opportunity to perform the clinical history and physical examination of these patients, with subsequent discussion of the case with the responsible preceptor. At other times in the ward, they accompanied the evaluation of patients with *osteogenesis imperfecta*.

During the development of outpatient activities, residents received support from preceptors and also help from residents of the genetics MRP. In this process, residents actively participated in the consultations, being responsible for carrying out consultations, prescribing, requesting exams, registering in medical records, and preparing reports, always discussing them with the preceptor physicians of the service and under their supervision. In this outpatient experience, residents had contact with genetic knowledge and tools that would be useful to their practice as future family physicians, helping to care for people with genetic and rare diseases.

At the screening clinic, they had contact with outpatients referred to the hospital for genetic evaluation. Most of them were referred by the PHC or by other focal specialists due to suspected genetic alteration, such as patients with a history of hypotonia, congenital anomalies, recurrent pregnancy loss, exposure to teratogens, counseling of consanguineous couples, among others. In this way, residents developed the clinical reasoning for the initial evaluation and investigation of a genetic suspicion, as well as for knowing when to discard this suspicion.

At the Interconsultation Outpatient Clinic, they participated in the evaluation of patients with suspected genetic alterations who were referred by other sectors of the hospital, also developing clinical reasoning for the investigation of genetic and rare conditions. At the General Genetics Outpatient Clinic, they participated in the care of adult and pediatric patients with different genetic conditions and already followed up at the service, including congenital anomalies, autism spectrum disorder, and intellectual disability. Skills were mainly developed in morphological physical examination and the differential diagnosis of genetic syndromes based on pattern recognition (*gestalt*) and the investigation of occult anomalies.

The internship included other outpatient clinics of subspecialties of Medical Genetics. At the Oncogenetics Outpatient Clinic, residents evaluated patients with a history of neoplasms to identify hereditary characteristics and provide genetic counseling. In this sense, it was important to approach family history based on lineage, in order to recognize family patterns of susceptibilities. At the Sexual Development Anomalies Outpatient Clinic, residents had contact with genetic conditions related to changes in biological sexual development that made a broader approach to the individual essential, in which psychological and cultural conditions weighed.

At the Skeletal Dysplasia Outpatient Clinic, they participated in the care of patients with genetic alterations in the formation of the skeleton. These are conditions that also bring important aesthetic and systemic repercussions, so that the residents' knowledge in interpreting imaging tests and osteoarticular clinical

evaluation was strengthened. At the Ambulatory of Osteogenesis Imperfecta, they had contact with patients suspected and diagnosed with this condition, addressing the initial investigation and follow-up evaluations.

At the Outpatient Clinic for Innate Metabolism/Neurometabolic Errors, residents had contact with patients with hereditary metabolic diseases, in which chronic and multisystemic manifestations weigh, representing a challenge to longitudinal care and rehabilitation. At the Neurogenetics Outpatient Clinic, the focus was on approaching children with neurological disorders for whom there was a suspicion of genetic etiology. The exercise of the syndromic diagnosis and the skills of the neurological physical examination were especially encouraged.

In this sense, the residents' perception of in-service learning can be especially defined along two converging lines: contributions from Medical Genetics to FP and contributions from FP to Medical Genetics — both strengthening the professionals' role in the network. In this first line, it can be justified that the knowledge and skills of Medical Genetics strengthen the look of the PHC professional, especially family physicians, for the suspicion and initial approach of genetic and rare conditions. In the second line, it is possible to identify how specific PHC and FP tools and principles enhance care in Medical Genetics and can positively change the work process in the service specialized in rare diseases. These two views are detailed below.

In the first aspect, knowledge and skills in genetics were deepened during the period, such as notions of genetic semiology, morphological physical examination, recognition of inheritance patterns, main diagnostic tests, indications for genetic counseling, teratogenic agents, and prevention of birth defects, among others. This knowledge expands the scope of the family doctor to act in the integral care of people with genetic and rare diseases, also being in accordance with the recommendations of the PNAIDR.[2,8]

This knowledge is useful for an adequate referral, in a timely and qualified manner. It was observed by residents that part of the primary care referrals for genetic evaluation had a report that was not very descriptive and without a prior basic investigation being carried out by the general practitioner in order to rule out non-genetic causes.

In a previous experience in the city of São Carlos, a physician residing in FP completed an internship at a Medical Genetics outpatient clinic. In this training, the resident was trained to deal with common genetic problems in PHC, developing skills in the construction of lineage, recognition of genetic risk, and primary prevention measures.[12] These skills were also achieved in the experience of the present report, which has the differential of having been produced in an SRDR, for a longer period of time and already in the context of the publication of the PNAIDR. The residents' home program did not include specific training to care for genetic and rare diseases in its pedagogical project, and the experience was innovative in this regard.[24]

In 2019, the Brazilian Society of Medical Genetics and Genomics proposed a competency profile for Brazilian physicians, detailing knowledge, skills, and attitudes in genetics needed by all physicians and guiding their adoption in medical schools and continuing education actions.[25] The actions developed in this elective internship are close to this profile and can serve as a model for continuing education actions in health, as well as fostering other internship opportunities in other SRDR. They can also guide the adequacy of medical residency programs in FP, aiming to achieve both the expected profile for the family physician and the specific objectives of the PNAIDR.[8,9,16,25]

In the second aspect, it is reinforced that the principles and practices of FP and PHC can qualify the work of the specialized professional and the attention in Medical Genetics, insofar as they broaden the look on the individual, their family and community and their needs beyond genetic pathology, as well as providing contact with care coordination technologies.

The essential and derived attributes of primary care refer to the operational characteristics of the services provided at this level. Namely: first contact, longitudinality, integrality, coordination, family and community orientation, and cultural competence.[26-28] When these attributes are understood and/or applied, as far as possible, to the reality of specialized services, they have the potential to boost care for genetic and rare diseases.

Thus, an SRDR can be recognized as one of the entry points for people with a rare disease in the RAS, even though PHC remains the priority entry point, but which often does not cope with their demands and complexities. Iriart and collaborators[10] showed a long therapeutic journey for patients with rare diseases until the proper diagnosis, in part associated with the lack of knowledge in genetics of PHC professionals. In this way, the services are complementary and must act together, respecting the responsibilities in the network and what is recommended in the PNAIDR.[8]

Genetic and rare diseases require lifelong care, building bonds with the team and a comprehensive approach to all the needs of affected individuals, both for PHC professionals and for genetic caregivers, in their different perspectives. Directing the investigation according to the evidence, the use of clinical epidemiology for probabilistic reasoning, the rational use of tests, and the application of quaternary prevention, the evaluation of diagnostic and therapeutic limits, the need for communication with other focal specialists — fundamental aspects in the action of the family doctor — are also necessary in genetic care and require the geneticist, in a certain way, to participate in the coordination of care for people with genetic and rare diseases.

Family and community orientation should also be part of the geneticists' work, insofar as their gaze should not only be focused on the patients, but also involve the family or community context, based on risk estimates, predictive and preventive attitudes, and genetic counseling. These last tools must also contemplate a systemic view of the health-disease process, considering the socio-cultural, economic, and environmental aspects involved. Cultural competence must be valued in the approach to genetic counseling for patients, family, and community.

Thus, all these tools that are traditionally part of FP professional practice can contribute to the caregivers' work in genetics, as long as they incorporate part of their knowledge into their care practice and establish more fruitful dialogues with professionals at the PHC level.[26,28,29] Clinical management in FP and PHC, in addition to the aspects already mentioned, also involves the organization of access and demand, professional agenda models, consultation time and medical records, among other aspects, which to a certain extent can be useful elements in the supply management of services specialized in rare diseases.[29]

Also, the person-centered clinical method, widely used in the practice of family physicians, can be an approach used in the care of people with genetic and rare diseases and contribute to the integrality of care.[30] The resident applied the method whenever possible during consultations, in order to value the experience and subjectivity of the person, respect their autonomy and meet their needs and expectations, according to the components of the method.[28,29] This view was extended to the service so that these aspects were taken to the discussion of cases and clinical meetings, but it has the potential to offer a more effective and lasting contribution to the SGM if it starts to receive more FP residents in its routine and the theme becomes a part of educational actions in the service.

That is, what is suggested in this report is not that specialized services in genetics replace or assume the attributes of PHC, nor the opposite, but that both can incorporate mutual elements in order to enhance the integrality of care for people with rare diseases in all levels of care mentioned.

Thus, the experience in the service was evaluated by the resident as very useful in all aspects already mentioned. Regarding the evaluation of the service, the structure of the SGM is adequate to host a reference service in the care of genetic and rare diseases, which contributed to the richness and success of the experience. The clinical and laboratory team is highly qualified and actively participates in research projects and academic activities. The greatest difficulties were observed in the unavailability of some biochemical and molecular tests due to material limitations and funding restrictions. The expectation is that, with the consolidation of the PNAIDR and adequate financial support, these shortcomings will be remedied in the service.

The internship allowed the resident to understand the general approach of the main genetic conditions assisted in the service, acquiring basic skills for comprehensive care for people with genetic and rare diseases at the PHC level. This training was in line with the PNAIDR, which recognizes primary care as the gateway for patients with genetic and rare diseases, being responsible for longitudinal care in families and communities. Thus, the experience places residents in terms of their role in the line of care for rare diseases and RAS.

The internship also allowed residents to recognize themselves as a family physician, appropriating the main FP tools to reflect on their own work process and to encourage changes in the local reality of the service. The articulation between genetics and FP developed in the internship reinforced, both for the resident and for the service, that care for rare diseases is complex and challenging precisely because it involves people, their families and communities, deserving emphasis not only on its biological determinants, but also on their care specificities and social vulnerabilities.

The SGM was given another field of articulation with PHC. In view of the experience gained from the residents' internship, the service can open space for the training of other residents and PHC professionals. Receiving an FP resident in its various spaces contributed to reflections on the role of the service in the RAS, its place as a reference center and for closer articulation with the PHC, through permanent education and matrix support actions.

ACKNOWLEDGMENTS

We would like to thank Professor Angelina Xavier Acosta for reviewing the work.

CONFLICT OF INTERESTS

Nothing to declare.

AUTHORS' CONTRIBUTIONS

LMCJ: Project administration, Formal analysis, Conceptualization, Data curation, Writing – original draft, Writing – review & editing, Investigation, Methodology, Supervision, Validation. MMA: Project administration, Formal analysis, Conceptualization, Data curation, Writing – original draft, Writing – review & editing, Investigation, Methodology, Supervision, Validation. LSMB: Project administration, Formal analysis, Conceptualization, Data curation, Writing – original draft, Writing – review & editing, Investigation, Methodology, Supervision, Validation.

REFERENCES

1. Peixoto SV. A tripla carga de agravos e os desafios para o Sistema Único de Saúde. Ciênc Saúde Coletiva 2000;25(8):2912. https://doi.org/10.1590/1413-81232020258.14672020
2. Vieira T, Giugliani R. Manual de genética médica para atenção primária à saúde, Porto Alegre: Artmed; 2013.
3. Passos-Bueno MR, Bertola D, Horovitz DDG, Ferraz VEF, Brito LA. Genetics and genomics in Brazil: a promising future. Mol Genet Genomic Med 2014;2(4):280-91. https://doi.org/10.1002/mgg3.95
4. Félix TM, Oliveira BM, Artifon M, Carvalho I, Bernardi FA, Schwartz IVD, et al. Epidemiology of rare diseases in Brazil: protocol of the Brazilian Rare Diseases Network (RARAS-BRDN). Orphanet J Rare Dis 2022;17(1):84. https://doi.org/10.1186/s13023-022-02254-4
5. Melo DG, Lessa ACO, Teixeira Filho JL, Nhoncanse GC, Drizlionoks E, Klein C, et al. Perfil clínico-epidemiológico da genética médica no Sistema Único de Saúde: análise do município de São Carlos, SP. BEPA Boletim Epidemiológico Paulista 2010;7(75):4-15.
6. Santos CS, Kishi RGB, Costa DLG, Silva DSD, Narciso TRF, Avó LRS, et al. Identificação de doenças genéticas na atenção primária à saúde: experiência de um município de porte médio no brasil. Rev Bras Med Fam Comunidade 2020;15(42):2347. https://doi.org/10.5712/rbmfc15(42)2347
7. Horovitz DDG, Llerena Jr. JC, Mattos RA. Atenção aos defeitos congênitos no Brasil: panorama atual. Cad Saúde Pública 2005;21(4):1055-64. https://doi.org/10.1590/S0102-311X2005000400008
8. Brasil. Ministério da Saúde. Gabinete do Ministro. Portaria nº 199, de 30 de janeiro de 2014. Institui a política nacional de atenção integral às pessoas com doenças raras, aprova as diretrizes para atenção integral às pessoas com doenças raras no âmbito do sistema único de saúde (SUS) e institui incentivos financeiros de custeio [Internet]. Diário Oficial da União; 2014 [cited on Feb. 1, 2021]. Available at: https://bvsms.saude.gov.br/bvs/saudelegis/gm/2014/prt0199_30_01_2014.html
9. Melo DG, Germano CMR, Porciúncula CGG, Paiva IS, Neri JICF, Avó LRS, et al. Qualificação e provimento de médicos no contexto da política nacional de atenção integral às pessoas com doenças raras no sistema único de saúde (SUS). Interface 2017;21(Supl. 1):1205-16. https://doi.org/10.1590/1807-57622016.0211
10. Iriart JAB, Nucci MF, Muniz TP, Viana GB, Aureliano WA, Gibbon S. Da busca pelo diagnóstico às incertezas do tratamento: desafios do cuidado para as doenças genéticas raras no Brasil. Ciênc Saúde Coletiva 2019;24(10):3637-50. https://doi.org/10.1590/1413-812320182410.01612019
11. Vieira TA. Genética comunitária: a inserção da genética médica na atenção primária à saúde em Porto Alegre [tese de doutorado]. Porto Alegre: Programa de Pós-Graduação em Ciências Médicas da Faculdade de Medicina da Universidade Federal do Rio Grande do Sul; 2012.
12. Melo DG, Gomes TLCS. Capacitação em genética médica para residentes em medicina de família e comunidade: relato de experiência. Rev APS 2009;12(1):83-7.
13. Vieira DKR, Attianezi M, Horovitz DD, Llerena Jr JC. Atenção em genética médica no SUS: a experiência de um município de médio porte. Physis 2013;23(1):243-61. https://doi.org/10.1590/S0103-73312013000100014
14. Acosta AX, Abé-Sandes K, Giugliani R, Bittles AH. Delivering genetic education and genetic counseling for rare diseases in rural Brazil. J Genet Couns 2013;22(6):830-4. https://doi.org/10.1007/s10897-013-9570-x
15. Silva LP. Integração da genética médica com a atenção primária à saúde: uma estratégia de suporte remoto [dissertação de mestrado]. Porto Alegre: Programa de Pós-Graduação em Ciências Médicas da Faculdade de Medicina da Universidade Federal do Rio Grande do Sul; 2017.
16. Sociedade Brasileira de Medicina de Família e Comunidade. Currículo baseado em competências para medicina de família e comunidade [Internet]. 2015 [cited on Feb. 01, 2021] Available at: https://www.ufrgs.br/telessauders/documentos/biblioteca_em_saude/006_material_saude_curriculo_competencias_mfc.pdf
17. Tavares DF, Teixeira TRG, Cardoso Júnior LM, Acosta AX. Radiografia do ensino de genética humana e médica nos cursos de Medicina das Universidades Federais do Brasil. Rev Bra Edu Saúde 2020;10(2):71-7. https://doi.org/10.18378/rebes.v10i2.7465
18. Vieira TA, Giugliani C, Silva LP, Faccini LS, Leite JCL, Artigalás OAP, et al. Inclusion of medical genetics in primary health care: report of a pilot project in Brazil. J Community Genet 2013;4(1):137-45. https://doi.org/10.1007/s12687-012-0110-4
19. Melo DG, Paula PK, Rodrigues SA, Avó LRS, Germano CMR, Demarzo MMP. Genetics in primary health care and the national policy on comprehensive care for people with rare diseases in Brazil: opportunities and challenges for professional education. J Community Genet 2015;6(3):231-40. https://doi.org/10.1007/s12687-015-0224-6
20. Ferrari N, Azevedo ES. Relatos sobre a história da genética na Bahia. Gazeta Médica da Bahia 2007;77(2):237-40.
21. Brasil. Ministério da Educação. Empresa Brasileira de Serviços Hospitalares. Carta de serviços ao cidadão. Complexo Hospitalar Universitário Professor Edgard Santos. Home [Internet]. [cited on Nov. 15, 2022]. Available at: https://www.gov.br/ebserh/pt-br/hospitais-universitarios/regiao-nordeste/hupes-ufba/acesso-a-informacao/acoes-e-programas/carta-de-servicos
22. Brasil. Ministério da Saúde. Gabinete do Ministro. Portaria nº 1.237, de 06 de junho de 2019. Habilita o Hospital Universitário Professor Edgard Santos como Serviço de Referência em Doenças Raras e estabelece recurso do Bloco de Custeio das Ações e Serviços Públicos de Saúde da Atenção de Média e Alta Complexidade Ambulatorial e Hospitalar – Grupo de Atenção de Média e Alta Complexidade – MAC, a serem disponibilizados por meio do Fundo de Ações Estratégicas e Compensação – FAEC do Estado da Bahia [Internet]. Diário Oficial da União; 2019 [cited on Feb. 1, 2021]. Available at: https://www.in.gov.br/web/dou/-/portaria-n-1.237-de-6-de-junho-de-2019-157606612

23. Brasil. Ministério da Educação. Empresa Brasileira de Serviços Hospitalares. Universidade Federal da Bahia. Complexo Hupes. Home [Internet]. [acessado em 10 nov. 2021]. Disponível em: https://www.gov.br/ebserh/pt-br/hospitais-universitarios/regiao-nordeste/hupes-ufba
24. Costa AJ, Valadares AMA, Silva BHDA, Barreto GSA, Campos HFS, Souza ICS, et al. Projeto Político Pedagógico. Programa de Residência de Medicina de Família e Comunidade e Multiprofissional em Saúde da Família. Fundação Estatal Saúde da Família; 2022.
25. Melo DG, Silva AA, El Husny AS, Ferraz VEF. Perfil de competência em genética para médicos do Brasil: uma proposta da Sociedade Brasileira de Genética Médica e Genômica. Rev Bras Educ Med 2019;43(1 Supl. 1):451-61. https://doi.org/10.1590/1981-5271v43suplemento1-20180257
26. Duncan BB, Schmidt MI, Giugliani ERJ, Duncan MS, Giugliani C. Medicina ambulatorial: condutas de atenção primária baseadas em evidências. 4ª ed. Porto Alegre: Artmed; 2013.
27. Oliveira MAC, Pereira IC. Atributos essenciais da atenção primária e a estratégia saúde da família. Rev Bras Enferm 2013;66(spe):158-64. https://doi.org/10.1590/S0034-71672013000700020
28. Gusso G, Lopes JMC, Dias LC. Tratado de medicina de família e comunidade: princípios, formação e prática. Porto Alegre: ARTMED; 2019.
29. Valladão Júnior JBR, Gusso G, Olmos RD. Medicina de família e comunidade. Série manual do Médico-Residente do Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo. São Paulo: Atheneu; 2017.
30. Stewart M, Brown JB, Weston WW, McWhinney IR, McWilliam CL, Freeman TR. Medicina centrada na pessoa: transformando o método clínico. 3ª ed. Porto Alegre: Artmed; 2017.