Strategic analysis of a medical genetics center at a national health research institute in Brazil: challenges for the country's public health system

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Abstract Objective: to discuss strategic challenges in complex health organizations sustained by an analysis in a Medical Genetics Center at a National Health Research Institute in Brazil. Method: Démarche Stratégique adapted for use with research institutes, including the policy formulation function. The study used institution's databases and workshops with key actors between Dec/2014 and Jun/2015. Results: Were defined the segments: clinical diagnosis, lysosomal diseases, osteogenesis imperfecta, genetic counseling, cytogenetics, biochemistry, and genomics. The findings reveal that the segments are aligned with the Institute's mission when it comes to referral care, although investment is required in strategic areas. The clinical diagnosis, lysosomal diseases, and genomics segments stood out in the scientific output portfolio. The findings show that progress needs to be made with policy implementation and planning to improve access to new technologies, together with investment in training and development, and the strengthening of coordination and collaboration between research institutes and referral services and between the wider healthcare network and research, care, and teaching. The inclusion of the policy formulation dimension was essential to highlight the unique role medical genetics plays in Brazil's public health system and the importance of developing action plans in the field of public health.

**Key words** Medical genetics, Health planning, Prospective analysis, Hospital management, Health policy

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### Introduction

According to the World Health Organization, genetic disorders affect between 3% and 10% of the world population. These disorders are defined as diseases caused by an alteration in the genetic material (DNA) and are mostly present from conception. They manifest themselves in different ways and at different stages of life, from gestation, as in the case of birth defects, through to adulthood, as with hereditary cancer for example<sup>1-3</sup>.

In Brazil, epidemiological data show that birth defects, more specifically congenital malformations, are the second leading cause of early child mortality, following the global trend4. Cancer is the second leading cause of death among the general population. It is estimated that between 5% and 10% of all cases of cancer are hereditary and that most cancer predisposition syndromes are transmitted in an autosomal dominant manner<sup>5,6,7</sup>. The social impact of these diseases is considerable and can reach beyond the patient and immediate family. Genetic disorders must not be neglected, especially given the burden they represent in terms of disability adjusted life years (DALY). Important advances in molecular genetics have provided a deeper understanding of these diseases, enhancing the potential contribution this area can make to healthcare<sup>2,3</sup>.

The recognition of the relevance of this theme led to the inclusion of medical genetics services in Brazil's public health system, while discussions about the diagnosis and treatment of genetic disorders and the organization of genetic healthcare resulted in the creation of the National Policy for Comprehensive Care in Clinical Genetics<sup>8</sup> and National Policy for the Comprehensive Care of People with Rare Diseases<sup>9</sup>.

However, various challenges remain in the field of genetic healthcare in Brazil, including a shortfall of available specialist consultations, centralization of services in large urban centers, difficulties in accessing specialized services, and poor coordination and collaboration between different levels of attention. These factors hamper access and contribute to a shortfall in care for people with genetic disorders and their families<sup>2,5</sup>.

To address these and other challenges in this area, the *Instituto Fernandes Figueira* (IFF) created the Medical Genetics Center (MGC) in 1989. The Center's mission is to promote referral care, research, and teaching in the municipality and state of Rio de Janeiro and support the formu-

lation of policies that address issues concerning medical genetics. In 2016, the MGC was accredited as a rare disease referral center (Rare Diseases Center) after meeting the criteria established by the Ministry of Health<sup>10</sup>.

The IFF is part of the Fundação Oswaldo Cruz (Fiocruz) and was renamed the National Institute of Women's, Children's, and Adolescent's Health (hereafter called the Institute) by Ministerial Order MS 4.159/2014. Since this change, the IFF has intensified its strategic advisory role in the following areas: proposition, implementation, and evaluation of policies and programs designed to improve access to healthcare; development of care models; research and technological innovation; and support for the coordination of the Women's, Children's, and Adolescent's Healthcare Network<sup>11</sup>.

This reconfiguration has demanded strategic changes in the IFF's departments, including the MGC, to address the need to strengthen competencies and reconcile the organization to its new role. To this end, the IFF has promoted debates to discuss the redefinition of its care service profile and management model, sustained by the principles and guidelines set out in the National Humanization Policy (NHP)<sup>12-14</sup> and the hospital's strategic management plan, the latter of which uses démarche stratégique (a method of strategic analysis that literally means "Strategic Approach") as its theoretical and methodological framework<sup>15,16</sup>.

Using the opportunity criterion, this framework can be used to redefine the mission of hospitals and complex care organizations with the aim of promoting the integration of the facility into an effectively and efficiently structured care network through a process of progressive cultural transformation<sup>15</sup>.

This approach draws on elements of microeconomics, strategy and policy, and organizational and cultural development, and also introduces aspects from the area of health and epidemiology, taking into account the unique nature of the public health sector. The *démarche stratégique*, developed originally by French authors, has been tailored to the Brazilian reality<sup>17,18</sup> and applied to hospitals and research institutes<sup>19</sup>.

The approach also focuses on the accountability of actors in the development of communicative and internal and external negotiation processes that result in agreements on types of care provided within a coordinated and interdependent health service network and in the creation of collegiate spaces that bring together workers,

managers, and users around more participatory and democratic decision-making<sup>16</sup>.

In this sense, the provisions of the NHP and premises of the démarche stratégique have synergies, since they are opposed to the normative and technocratic definition of guidelines and seek to create the conditions for communicative processes, developing a link between strategic management and care centers and enabling actors to define strategies and build a common project.

In light of the reconfiguration of the IFF and the strategic challenges faced by complex care organizations<sup>20</sup>, this work discusses the strategic position of the MGC and actions that enhance its referral care, teaching, and research activities. This study is unique in so far as it conducted the strategic analysis using the démarche stratégique and was the first one to include the support for policy formulation function in the method.

# Methodology

Medical genetics was chosen as the area of study because of its strategic position in the care network, the relevance of genetic disorders in the national context, and the potential for technological innovation in clinical research and healthcare<sup>21</sup>.

The MGC provides several referral services, including: clinical cytogenetics laboratories; applied molecular genetics services; a center for intravenous infusion and enzyme replacement therapy for metabolic disorders; diagnosis of inborn errors of metabolism; general medical genetics outpatient clinics (birth defects, genetic syndromes, developmental disorders); and osteogenesis imperfecta, inborn errors of metabolism, high-risk pregnancy, and genetic counseling outpatient clinics. The Center also provides post-graduate training for medical geneticists, conducts research on genomics, and develops and participates in national clinical trials.

With regard to support for health policy formulation, the MGC participates in the development of clinical guidelines, provides consultancy services to the thematic areas of the Ministry of Health, coordinates the Osteogenesis Imperfecta Referral Center (OIRC), provides training and guidance to health professionals in the management of genetic disorders, partners with the Latin American Collaborative Study of Congenital Malformations (ECLAMC, acronym in Portuguese), and is a managing member and collaborator of the National Institute of Population Medical Genetics (INAGEMP).

The adaptation of the *Démarche Stratégique* used an extended interview guideline to encompass research institutes with an emphasis in teaching and research actions and mixed production indicators and includes the applied model for assessing scientific output<sup>19</sup>. This study included an analysis of support for health policy formulation, which has not been previously addressed, thereby constituting an innovative adaptation of the original method.

The work involved meetings and workshops with key actors and the collection of data and institutional documents between December 2014 and June 2015. It is important to stress that the interview guideline – designed based on the assumptions of the démarche stratégique and adapted to the Brazilian context to encompass research institutes<sup>19</sup> – served as the basis for the strategic analysis presented here.

The key actors were the manager of the MGC, health professionals, managers or staff responsible for health care processes/sectors, professionals who develop teaching and research activities, and staff involved in service coordination and planning, resulting in a total of 25 participants with a frequency of participation of over 90%. The study was approved by the IFF's Research Ethics Committee (application number: 1,740,977) and conducted in accordance with the ethical, legal, and regulatory norms and standards for research involving human subjects. All participants signed an informed consent form.

The following stages of the d*émarche stratégique* were followed<sup>16-18</sup>:

1) Initial situation analysis – this stage consists of an administrative and medical appraisal to provide the elements necessary for defining the activity segments (stage 2). We used data from the hospital's statistics departments, MGC, nursing service, sub-directories of research and teaching, and Lattes Platform curriculum vitae from each participant.

The administrative appraisal focused on the following information: number of patients and patient characteristics by category, number of tests performed, and scientific publications. The medical appraisal concentrated on the pathologies treated, type of population, type of care according to patient specificities, technologies used, synergies and partnerships, collaborating services inside and outside the Institute, and types of cohort studies and clinical research underway. The study periods for data analysis were: 2014 for clinical consultations; 2013 and 2014 for laboratory tests and diagnoses; and 2010 to 2014

for fundraising projects, published scientific articles, and other scientific outputs.

- 2) Segmentation in this stage a multi-criteria analysis is performed to define homogeneous activity segments and therefore ensure that the strategic analysis is representative. Segmentation takes into account the complexity of the institution's mission, whereby all activity groups do not necessarily have the same level of development or strategic value. The criteria used for the purposes of this study were: pathologies and health status of service users, degree of complexity, links between pathologies, main features of the consultation process and core care areas, technologies used, infrastructure needs, and necessary competencies.
- 3) Strategic and competitive position analysis – this analysis is performed to determine the prospective value of each segment. The results are used to hierarchize/prioritize each segment according to its strategic value, specific problems, and competitive position in terms of excellence. Competitive position is determined based on the scores assigned by the study participants for a set of key success factors (national and international benchmarks in areas such as technologies, specialized human resources, expertise, relational capacities, and financial capacities) and on the level of control the institution has over these factors to be able to ensure excellence. The higher the level of control over key success factors the higher the level of competitiveness.

The following evaluation criteria were used to determine the prospective value of each segment: growth possibilities, competitive intensity, investment in equipment, investment in human resources, synergies, external partnership opportunities, teaching and research potential, contribution to the Institute's general political project, degree of internal staff motivation, and local/state potential and regional/national potential. It is important to note that these criteria were analyzed considering the four core functions of the Institute: referral care, teaching, research, and support for health policy formulation.

4) Activity portfolios – these are represented by graphs showing the segments (medical specialty) in question, allowing for set and comparative value analysis. The X-axis represents competitive position and the Y-axis represents comparative value. Each segment is represented by a bubble whose size corresponds to the relative output volume of the activity and whose position depends on the score obtained from the evaluation of the value of and degree of control over

the key success factor (KSFs), thus defining the segment's competitive position. The graph is divided into four quadrants, where the two upper quadrants correspond to high value segments, the right quadrants correspond to segments with a "high" competitive position, and the left quadrants correspond to segments with a low level of competitiveness.

Bearing in mind the complexity of the Institution's mission<sup>18</sup>, which involves not only teaching and research activities, but also healthcare activities in institutions where research has a high value, two portfolios were created (healthcare output and scientific output) to provide a broad picture of the set of segments, strategic risk assessment, and output volume<sup>19</sup>.

5) Action plan – this consists of a set of actions aimed at revaluing segments that are strategic both for the organization and healthcare network, focusing on workable low-scoring value analysis criteria (when the political decision is to seek to increase value). These actions – including social and intersectoral actions – may translate into a reduction in activities, when it is more appropriate to provide them in other facilities, and into modifications of the care model, the types of consultation that characterize a given segment, and the types of technology used.

## Results and discussion

The following segments were identified in stage 2: osteogenesis imperfecta; clinical diagnosis (congenital anomalies, adult degenerative diseases, inborn errors of metabolism, developmental disorders, hereditary diseases and genetic syndromes); lysosomal diseases; cytogenetics; biochemistry; genomics; and genetic counseling (which encompasses criteria related to pathology and technology).

The prospective value of each segment was determined using multi-criteria analysis (growth possibilities, competitive intensity, investment in equipment, investment in human resources, synergies, external partnership opportunities, teaching and research potential, contribution to the Institute's general political project, internal staff motivation, and local/state potential and regional/national potential), according to the guidelines of the interview adapted for the application of the *Demarche Strategique* in Brazil, providing an assessment of the attraction capacity of each segment. Table 1 shows segment weighting and the average scores assigned to each segment

by the study participants in the strategic and competitive position analysis.

Table 1 shows a relatively small difference between the values assigned to the segments, with scores ranging between 14.23 and 16.21, demonstrating their strategic importance in relation to the Institution's mission.

The leading segment was genomics. This is due to recent technological advances regarding new generation DNA sequencing that have improved access to precision diagnosis<sup>21</sup> and the need to deepen medical and clinical knowledge in this area, combined with prioritization of this segment by research funding agencies. Despite the decline of childhood disease incidence and stabilization of epidemiological indicators, the clinical diagnosis segment also shows a growing trend due to advances in antenatal diagnosis – leading to improved control of newborn illnesses - and to the potential the Institution has to provide support and advice to other facilities in the neonatal care network with a view to promoting the creation of new healthcare centers.

The forecast of strong growth for the biochemistry segment can be explained by the expansion of the Newborn Screening Program<sup>22</sup>, identification of new treatments for inborn errors of metabolism, and development of more accurate diagnostic methods and biomarkers. The growing trend in cytogenetics is explained by the fact that the IFF's laboratory is the only public laboratory that performs cytogenetic diagnosis in the State of Rio de Janeiro.

Clinical diagnosis, genetic counseling, and lysosomal diseases were the segments that show greatest growth potential. This is because the MGC/IFF was recently accredited as a rare diseases center, leading to an increase in demand for diagnostic and prevention services and treatment<sup>9,10</sup>.

All segments were well-positioned when it comes to competitive intensity, notably clinical diagnosis and genomics. The low level of competition could be explained by the scarce provision of public health services in this area<sup>17</sup>, primarily due to the lack of effective policy, planning, and investment<sup>5,7</sup>.

The segments where investment in technology and equipment are needed most were clinical diagnosis and laboratories. This is due to the rapid advances in technology in these areas, requiring the adoption and update of these new technology and reform of the current structure of the Genetics Center. All segments pointed to major investment on human resources, given the

 Table 1. Prospective values of the segments of medical genetics services.

	Weight		IO	Clin	Clinical diagnosis	Cli	Clinical diagnosis	Bioche	Biochemistry	Cytogenetics	netics	Genomics	mics	Genetic counselin	Genetic counseling
Evaluation criteria	(%)	•	Score (p x	Score	Score (p.x	Score	Score (p.x	Score	Score (p.x	Score	Score (p.x	Score	Score (p.x	Score	Score (p.x
		(20)	n)/100	(20)	n)/100	(20)	n)/100	(20)	n)/100	(20)	n)/100	(20)	n)/100	(20)	n)/100
1. Growth possibilities	12	13	1.56	13.5	1.62	17	2.04	17	2.04	16	1.92	18	2.16	17	2.04
2. Competitive intensity	7	∞	0.56	16	1.12	13	0.91	13	0.91	13	0.91	15	1.05	12	0.84
3. Investment in equipment	9	10	9.0	17	1.02	13	0.78	16	96.0	15	6.0	17	1.02	7	0.42
4. Investment in human resources	6	13	1.17	15	1.35	17	1.53	15	1.35	15	1.35	16	1.44	13	1.17
5. Synergies (competences and shared facilities)	10	17	1.7	17	1.7	12	1.2	16	1.6	17	1.7	16	1.6	18	1.8
6. External partnership opportunities	10	17	1.7	15.5	1.55	17	1.7	18	1.8	18	1.8	18	1.8	18	1.8
7. Teaching and research potential	12	17	2.04	17	2.04	17	2.04	17	2.04	16	1.92	18	2.16	12	1.44
8. Contribution to the jnstitute's general political project	12	17	2.04	14	1.68	17	2.04	16	1.92	16	1.92	16	1.92	15	1.8
9. Internal staff motivation	^	13	0.91	18	1.26	17	1.19	18	1.26	18	1.26	18	1.26	16	1.12
10.Local/municipal potential	9	17	1.02	17	1.02	16	96.0	13	0.78	15	6.0	15	6.0	15	6.0
11. Regional/national potential	6	15.5	1.39	16	1.44	12	1.08	10	0.9	10	6.0	10	6.0	10	6.0
	100		14.60		0		17		70 01		15 40		16.51		60.7
	100		14.09		15.8		15.47		00.01		15.48		10.21		14.25
Source: Results from the workshops with key actors															

current shortness of staff, present growth trends, and low level of competition.

All segments obtained a high score for synergies due to the interdependence between specialties within the Institute. The segment that obtained the highest score was lysosomal diseases. This segment has the potential to enhance coordination with the pediatric outpatient clinic, nursing and rehabilitation services, laboratory, pharmacy, and cohort studies.

All segments have the potential to develop partnerships with specialist referral centers, family clinics, and research institutes (ECLAMC and INAGEMP) and healthcare networks (OIRC and rare disease centers), and need to improve aspects concerning the formalization of these partnerships. In this respect, the study participants suggested that the lysosomal diseases segment needs to strengthen links with primary care services to enable home enzyme infusion, avoiding the need for hospital visits and optimizing the use of hospital resources.

All segments except for genetic counseling show major potential for both teaching and research and participation in research networks<sup>23</sup>. Factors that drive research include: technological advancements within these segments and their contribution to diagnostic and treatment research (next-generation DNA sequencing and replacement therapy for example)21; the potential for developing partnerships with national and international research networks; participation in drug trials conducted by large companies; and the need to develop national clinical guidelines in this area. With regard to teaching, the findings show that there is major potential for medical residency (medical genetics and osteogenesis imperfecta), undergraduate and postgraduate courses (biochemistry and lysosomal diseases), and expansion of training and development for professionals working in primary healthcare and referral centers (particularly in the genetic counseling and osteogenesis imperfecta segments), which is essential to improving the quality of care.

Since the MGC provides specialist services and develops actions aligned with the functions of the Institute, all segments obtained a high score for contribution to the hospital's political project. Scores were also high for internal staff motivation and for local/state potential and regional/national potential across the majority of segments, due to the concentration of specialized services in state referral centers<sup>5</sup>.

The competitive position of each segment was determined (stage 3) using between six and

nine KSFs. Degree of control over these KSFs was scored using the ideal degree of control (high-quality performance) as the parameter.

The main strengths are represented by the following KSF's: accreditation of the MGC as a rare diseases center and osteogenesis imperfecta referral center; availability of retrospective modality of genetic counseling technology, osteogenesis imperfecta treatment technology, and enzyme replacement therapy technology; expertise and availability of basic equipment; synergies with hospital care areas and IFF specialties; partnerships with other local and national centers; development of cohort studies and clinical trials. Most of the segments showed good control of the resources professional expertise and basic equipment necessary for the operation of specialist laboratories, which are decisive for the work of the MGC/IFF.

The general analysis of the segments showed that the osteogenesis imperfecta and lysosomal diseases segments had the highest competitive position, while the biochemistry, genomics, and clinical diagnosis segments had the lowest positions. The remaining segments had an average position.

The segments clinical diagnosis, osteogenesis imperfecta, lysosomal diseases and genetic counseling showed a high level of synergy with other sectors and specialist services provided by the IFF. The partnership between the osteogenesis imperfecta segment and National Institute of Traumatology and Orthopedics (INTO) and State Institute of Diabetes and Endocrinology (IEDE) also showed a high level of synergy. This partnership is necessary to complement the treatment of patients in orthopedic and endocrinology specialist services. Since the MGC does not provide biochemistry services, the partnership with the Federal University of Rio de Janeiro's Inborn Errors of Metabolism Laboratory (LABEIM) is essential to provide specific tests required for diagnosis. The possibility of promoting research through the creation of research cohorts and clinical trials represents a strong KSF, involving mainly the osteogenesis imperfecta and lysosomal diseases segments.

KSFs with a score below 12 indicate weaknesses or critical resources, such interdisciplinary teams in the clinical diagnosis and genetic counseling segments. Given its level of complexity and to fulfill its function with excellence, the clinical diagnosis segment will require specialist skills in several areas, including neurology, psychology, orthopedics, physical therapy, social services, and nursing. On the other hand, genetic counseling will need professionals from areas such as psychology and social services and experts in clinical genetics in order to enhance its service capacity and expand teaching and research <sup>23</sup>.

With regard to facilities, the findings show that there is a need to improve information technology, including databases, specific software, specialized technical support, and equipment to support the growth of healthcare and research functions in the osteogenesis imperfecta, cytogenetics, and genomics segments. High resolution equipment was identified as a critical resource for the cytogenetics and lysosomal diseases segments. With regard to operational matters, it will be necessary to reinforce resources such as laboratory technicians, supplies, and physical structure and maintenance, especially in the genomics segment.

Data related to the healthcare output portfolio (stage 4) was obtained from internal reports of consultations performed by the clinical diagnosis, lysosomal diseases, genetic counseling, and osteogenesis imperfecta segments and a report of tests performed by the cytogenetics (internal), biochemistry (external), and genomics (internal) segments. The scientific output portfolio was built using data on fund raising for research projects obtained from the hospital's internal sectors. Data on published articles, participation in congresses and courses, and supervision of MSc and PhD students was obtained from the curriculum vitae CNPq Lattes Platform. The portfolios were presented at a workshop to analyze and define strategies for addressing weaknesses.

The portfolios represent the value and competitive position of the strategic segments. Their analysis provides an idea of the whole picture and an appraisal of the strategic risk that the segment is exposed to. This risk results from the adaptation of service provision to greater or lesser degrees (activity value) and the effort required to achieve greater control over the KSFs, which in turn increases competitive position and, consequently, the capacity to meet the demand relative to the activity segments<sup>17-19</sup>. Portfolio analysis also allows key focus areas to be defined according to the prospective strategic position.

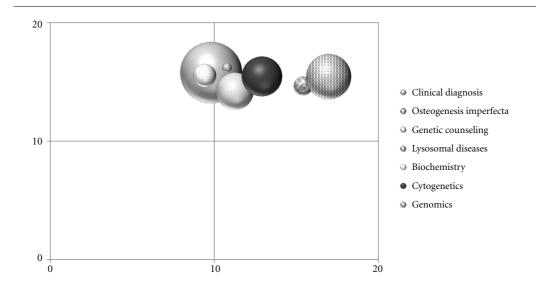
The healthcare output portfolio (Graph 1) shows that all segments have a high value, indicating that medical genetics is well tailored to the IFI's mission, represented by the provision of referral care activities and high-density technology. The segment with the highest level of competitiveness is genomics (16.21), followed by clinical diagnosis (15.8), biochemistry (15.56),

cytogenetics (15.48), lysosomal diseases (15.47), osteogenesis imperfecta (14.69), and genetic counseling (14.23). The segments show different competitive positions. Clinical diagnosis, biochemistry, and genomics are positioned in a situation of strategic risk with scores of 9.79, 9.4, and 10.75, respectively. It is important to note that the challenges that will need to be faced to overcome this situation are related to difficulties in regulating technology incorporation in Brazil<sup>24</sup> and issues concerning political and economic feasibility in the area of health. The formulation of policies that speed up the technology incorporation process and support cutting-edge research involving actors and institutions across all levels of the public and private sector<sup>25</sup> is essential. The right to health enshrined in our constitution needs to be anchored in policies that strengthen the democratization of healthcare provision across all levels, including specialist services, without overlooking the need for effective coordination and collaboration within the care network to ensure the effective delivery of genetic health care.

The scientific output portfolio (teaching and research) shows that the clinical diagnosis segment has the largest volume (817.75 points), followed by the lysosomal diseases (385 points), genomics (236 points), cytogenetics (199 points), osteogenesis imperfecta (75 points), genetic counseling (23.5 points), and biochemistry (16 points) segments. Scientific output related to teaching and research was calculated using parameters with different weightings corresponding to published articles, fundraising for research, participation in congresses, supervision of MSc and PhD students, participation in academic panels, and coordination of courses<sup>19</sup>.

The portfolios were analyzed in a workshop and served as a basis for defining strategies for the various segments and developing an action plan (stage 5) to implement the agreed strategies.

The strategies were based on the KSFs and weaknesses identified by the analysis, notably: strengthen the interdisciplinary team (neurologist, psychologist, orthopedist, nurse, physical therapist, social worker), which is essential because genetic health care requires an interdisciplinary approach<sup>1</sup>; negotiate with area coordinators to increase the availability and support of these professionals for the MGC; increase staff numbers using resources from the Rare Diseases Center's budget; and develop partnerships with Fiocruz's Genomic Diagnostic Network to complement diagnosis services.



Graph 1. Healthcare output portfolio.

Source: Workshops with key actors, databases, record books and institutional documents.

With regard to facilities, the main needs identified were support of information technology resources, key laboratory supplies and equipment, and research resources. It will also be necessary to: incorporate the development of operating systems and acquisition of specific software into the Institute's Investment Plan; plan the incorporation of supplies and equipment; modernize the laboratory's physical structure; and provide maintenance services via Fiocruz's shared technology platform.

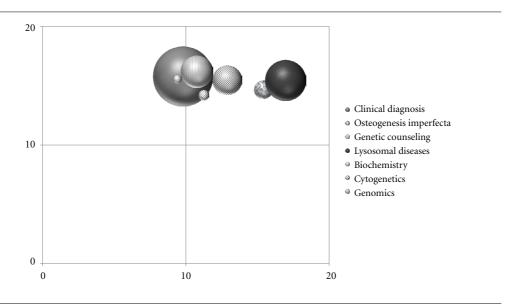
Specific strategies identified for each segment include: streamlining test processing to better meet the demands of the clinical diagnosis segment; promotion of clinical trials in the osteogenesis imperfecta segment; expansion of fund raising capacity in the segments that use laboratory technology; and formalization of partnerships with the IEML/UFRJ in the lysosomal diseases segment.

It is important to problematize the difference between various activities. To this end, we used different portfolios for healthcare and research activities in the discussions with the key actors in the workshops. Research and healthcare output should not be looked at in isolation. The genetic counseling segment, for example, has a high output, but is situated at the interface between the quadrants that represent strategic risk. This segment obtained average scores because this activi-

ty should be better shared with the network. This was the strategy suggested for the action plan in this case. The discussion between the group of professionals and director of strategy contributed to decision making.

The research and knowledge production potential of each segment should be reflected in scientific output. The cytogenetics segment showed the highest output volume in the healthcare portfolio and lowest output volume in the scientific portfolio. This is explained by the fact that cytogenetics is a diagnostic activity. It is expected that the incorporation of new molecular diagnostic methods<sup>21</sup> (such as array CGH and whole exome sequencing) proposed in the Activity Plan will lead to an increase in scientific output. The analysis proposed by the method is always prospective<sup>16</sup>.

Some segments are better positioned in the healthcare portfolio, whereas others are better positioned in the scientific portfolio. The factors that led to these scores were examined to define repositioning strategies for each segment. In this respect, low healthcare output in a segment with high value and a good strategic position may indicate a need for more professionals with this expertise, as in the example of the genomics segment, while high scientific output and low healthcare output may indicate that professionals are more involved in research and teaching than



Graph 2. Scientific output portfolio.

Source: Workshops with key actors, institutional documents and Lattes Platform.

in healthcare activities. Only strategic analysis such as the one presented here is able to show whether it is appropriate to invest in healthcare output within the hospital or share activities with the healthcare network<sup>17</sup>. In institutions with complex missions, coordination and collaboration between healthcare, teaching, and research should be improved since, apart from providing health services to those who need them, the results of clinical research linked to healthcare activities produce knowledge that can help advance diagnostic and healthcare technologies, improve protocols, and inform policy-making, positively impacting public health and quality of life.

## Final considerations

This study analyzed segments aligned with the new configuration of the Institute, which is sustained by the principles and guidelines set out in the National Humanization Policy and the hospital's strategic management plan. The analysis considered internal synergies and partnerships, external partnership opportunities, and coordination and collaboration with the wider healthcare network. Sector analysis does not contribute to the development of bolder and more wide-ranging proposals. We therefore adopted a

prospective approach focusing on specialist services thereby avoiding reductionism.

The findings show that the segments have a good competitive position and underscore the need for investment in strategic areas, including: multidisciplinary teams, incorporation of medical equipment and information technology, and coordination and collaboration with the primary care network and other referral centers that provide genetic healthcare services.

The MGC's alignment to the IFF's new mission brings new challenges and the need to develop actions that this area has the capacity to enhance, including: greater coordination and collaboration with the network, particularly primary care services, with a view to expanding prevention activities via genetic counseling<sup>23</sup>; identification of coordination strategies and care technologies that allow dehospitalization, homecare, and clinical follow-up; expansion of service provision with the proposal of new core care areas and national diagnosis and treatment referral centers; training courses and actions, including a strategy to promote nationwide distance learning courses; development of clinical guidelines; and development of research projects focusing on the assessment of technology incorporation.

These strategies are essential given that certain genetic conditions require early hospital

admission, readmission, surgery, and chronicity, increasing the burden on health services and care costs<sup>5</sup>.

The discussions also showed that technological advances alone are not enough to solve genetic healthcare problems. Progress needs to be made with policy implementation and planning in order to improve access to new technologies, together with investment in training and development and the strengthening of coordination and collaboration between research institutes and referral services and between the wider healthcare network and research, care, and teaching. It is also necessary to consider the macro scenario and to strengthen the role of the State through health policies that articulate the economic, social and regulatory dimension and to promote the participation of actors with expertise in various types of knowledge that contribute to increase the value chain in innovation health<sup>25</sup>. However, the inclusive ethics represented in the principles of the SUS should predominate, considering the common good and values of solidarity.

The methodology adopted helped improve communication and collaboration, providing the opportunity to discuss the strategy of the MGC with the participation of the directors and all staff, embracing the features of autonomy of professional organizations based on Minstzberg<sup>20</sup>, who suggests that the participation of frontline and operational staff is essential to ensure effective decision-making. It is important to highlight that this study conducted an in-depth analysis of research activities, building a specific portfolio for scientific output19. We included the policy formulation dimension given that the MGC is a point of reference at national level and has made an important contribution towards the development of policies in the area of genetic health care, thus highlighting the unique role medical genetics plays in Brazil's public health system and the importance of building more inclusive public health policies.

### Collaborators

Artmann E, Llerena Junior JC and Pereira LT worked on the conception, survey and analysis of information, in the preparation and revision of the manuscript. Binsfeld L and Rivera FJU worked on the survey and analysis of information, in the preparation and revision of the manuscript.

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