

The intertwining of discourses in the diagnostic construction of cystic fibrosis: a perspective on access and barriers

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FREE THEMES

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Abstract *The complexities referred to in the search for “accuracy” in the diagnosis of cystic fibrosis (CF) point to reflections around “what is needed” in the current situation of “precision medicine”. We analyzed the discourses of 19 social actors belonging to the community of specialists in cystic fibrosis, exploring the semantic meanings of the word “precision”, and the barriers to diagnosis and innovations in therapeutics. We adopted the critical discourse analysis (CDA) of Norman Fairclough in order to achieve the discursive constructions around the integrality of care, the guarantee and equitable supply of basic social needs. Access was identified as an emic category when in the social arenas of dispute are health needs and the right to life.*

Key words *Cystic fibrosis, Precision medicine, Health needs*

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Introduction

The word *precision*, present in *precision medicine*, entices an analytical look into the (im)precisions that cystic fibrosis evokes as a health condition that is rare, chronic, and complex. The current article explores the meanings of “what is needed” in the process of the construction of a diagnosis, and “what are the needs” for the individuals with this health condition. The innovations in therapeutics of cystic fibrosis with Cystic Fibrosis Transmembrane Regulator (CFTR) modulating medication can therefore be placed somewhere between precision/exactness and health needs. Such modulators, according to Amaral and Rego¹, are significant in achieving quality of life and strengthening hope.

Cystic fibrosis (CF) is a rare genetic, autosomal, and recessive disease connected to mutations in the CFTR gene. It is progressive and requires continuous care. Mutations located in chromosome 7q31.2 alter the codification of the CFTR gene, leading to an imbalance in the concentration of chlorine and sodium in the cells responsible for bodily secretions, which are quite dense and provoke changes in different systems of the human body, especially in the functioning of the lungs, pancreas, liver, intestine, and reproductive system².

With this description, CF resends to us its genetic complexity, in the field of precision medicine and in terms of hope for medical care that can provide a quality of life despite the chronicity of the illness. The word precision is placed among the synonyms of exactness, perfection, rigor, care, and need, establishing a dialogue between “*exactness*”, by one definition, and “*what is needed*” in the universe of needs. Its meaning as a verb – “*what is needed*” – is aligned with health needs; whereas its meaning as an adjective, as a synonym of exactness, relates to the location of the diagnosis and its implications in therapy.

These senses permeate the scenario of contemporary biomedicine and are updated by the discourse of social actors, here understood as a universe in which health professionals, researchers, patients associations, patient specialists, professionals of the pharmaceutical industry, legislators, and decision-makers join together.

These social actors, considered here to be a community of CF specialists, seize social reality, providing meaning and significance to it. This occurs from the standpoint of the position they occupy in the social arenas in relationships of race, generation, cultural values, and the possi-

bilities of satisfactory access to basic health needs in the social arenas. It is in this socially constructed place that the dialectics of influences, alliances, recognitions, and power struggles empower the discourses and redefine them^{3,4}.

In the social arenas, demands are considered and answered by a political system, given that that society and institutions negotiate the decisions. This analytical model indicates a prolific field for public policies, a *locus* for negotiations of power and interests present in a *policy community* (community of specialists)³.

In the community of specialists, there are the *alike* and the *informed*⁵, and it is possible to establish connections and relationships, influences and identities in dispute and negotiation. It is a complex process of the elaboration of needs and a definition of demands for the public agenda of decision-makers and producers of public policies.

The understanding of the discourse as a practice by social actors, inscribed in social arenas, requires one to consider the combination present between structural perspectives and actions, since social practice is, “on the one hand a relatively permanent manner of acting in society, determined by a position within the network of structured practices, and on the other hand, a domain of social action and interaction that reproduces structures and is able to transform them”⁶ (p.308). Considering the field of integrality in medical care provided to people with cystic fibrosis, we consider it a daily experience that occurs in the body and in life as a rare health condition that is chronic and complex.

The current article is supported by findings from Marins⁷, who articulates a dialogue between the needs and the demands for healthcare, using the scenario of contemporary biomedicine within its limits and possibilities. Our aim, therefore, is to analyze – by means of definition/precision x undefinition/imprecision in terms of cystic fibrosis – the discursive scenario of the search for the exact treatment, for which the ultimate expression is “*what is needed*”.

Methodology

The cystic fibrosis experience is far from being something homogeneous among those who are part of the community of specialists, in terms of appropriation, meaning their acting and identity with disputes, as well as strategies to act in and upon reality. Therefore, the research which su-

ports this article is defined by the presumptions of Critical Discourse Analysis (CDA) in the perspective of Norman Fairclough^{4,6}.

The theoretical-methodological perspective of CDA assumes language as a social practice. And, in the dialectic function of social relationships, the conditions for discourse production represent the social-historical and cultural standpoints of the social actors, here understood as a community of specialists in cystic fibrosis. Language is something appropriated as a form of social practice, transforming discourse into a form of action; it is the way people act upon the world, upon other individuals, and in representations⁷.

For Fairclough⁴, discourse is something with the capacity to adjust and restrict itself to social structure. Social structure is understood in a broad sense and in the placement of classes, as well as in and through social and institutional relationships, norms, conventions, and classifications, be they of a discursive nature or not. Discursive events, in a given social structure, provide variability according to the social and institutional domain in which they are produced. Moreover, they are also socially constructed by the very dimensions of the social structure that both adjusts and restricts them. Hence, discourse is a representation practice and a process of constitution and construction of a meaning for reality^{4,6}.

We assume that there is a dialectic relationship between discourse and social structure, since the discursive constitution of society is produced by social practices supported by objective social structures and directed in a contradictory, partial, and flexible manner. On the other hand, social practice is structured by a diversity of semiotic elements: productive activities, means of production, social relationships, cultural values, and consciousness. And this discourse is accomplished considering all of these elements^{4,6}.

The discourse presents itself as a manner of political and ideological practice, to the point that it is able to act in the maintenance or transformation of power relationships, as well as to constitute, naturalize, maintain, or transform the attributed meanings, depending on the diverse social positions in the power relationships. The manner by which the power relationships, the ideologies and the conventions interact in discursive practice are therefore transformed into arenas of dispute^{4,8}.

Social actors, as social-historical subjects, act and are not restricted to being “passive subjects”

but rather are defined by the ambivalence of those two states. Through these dynamics, “discourse is determined by the construction of identities and the modeling of social relationships and the ways of conceiving/reading the world”⁹ (p.08).

Based on these theoretical assumptions, we employed a network which indicates familiar universes, in which each interviewee indicated other members of the community of specialists to become a participant in the study. This network of universes based on familiarity is not random, it is not like a snowball, occurring in a supposedly natural manner. It rather assumes familiarity as the basis for indication^{10,11}, reference, and knowledge, hence reinforcing the intentionality in the indication.

With the dynamics we employed, we reached 42 indications, with 11 people indicated more than once. Thirty-one individuals were contacted – one, in person and 20 through Whatsapp, with text messages inviting them to participate in the research.

Of the contacted individuals, 20 responded, allowing for 19 individual interviews to be conducted from January to April 2021. One interview could not be performed, as the participant was not available until after the interview stage. The open interviews lasted an average of 1 hour and 30 minutes, and were guided by a script, which included: identification and three triggering statements related to the connection with CF, the diagnosis dimension, and the healthcare needs in the current scenario of precision medicine.

The interviews were conducted in an in-person format, using audio recording (one interview) and remote format (18 interviews) through online digital platforms (Zoom: 11 interviews; Google Meet: seven interviews). The method allowed us to reach more participants, when distance and/or time zones did not favor in-person dynamics.

The collection of discourses produced in the interviews was transcribed, subjected to exhaustive reading, in dialogue with CDA's theoretical-methodological design, and conducted separately by each of the authors in this study. Afterwards, the descriptive, interpretative, and explanatory aspects present in the discourses of the social actors were compared.

All of the participants accepted to participate through a Free and Informed Consent Form, with their names kept anonymous, identified only by a capital letter, followed by numbers (Indo-Arabic numbers), including: six healthcare professionals (S), three members of associations

of CF patients (A); five professionals connected to the pharmaceutical laboratory industry (I); and one researcher (P). Also included were social actors, considered to have a hybrid identity: one person with CF and a researcher (PCF); one person with CF and a member of the association of CF patients (ACF); one family member of a person with CF and a researcher (PF); one family member of person with CF, a member of an association, and a researcher (APF).

It is important to highlight that, considering the geographic location of the participants, the study reached a good level of territorial distribution and of different states/regions of the country, as well as reached beyond the Brazilian territory. In other words, there were participants from the Brazilian states of Rio de Janeiro, São Paulo, Minas Gerais, Paraná, and Mato Grosso do Sul, along with Brazilian nationals residing in the United States and Portugal. The present study was approved under CAAE: 40374320.0.0000.526 and decision no. 4,472,657.

Results and discussion

Beginning with the expression *“what is needed”*, our study reflected upon needs as social-historical products present in the discourses of the social actors, taking place in social arenas. In these are disputes with diverse revindication agendas, which often make use of the argument of the recognition of rights. In the discourses, diagnosis and medication were interpreted analytically as steps in the line of care for cystic fibrosis and as points of care in the perspective of integrality. The possibility of a “precise” diagnosis is linked to the construction of a process of search for care, which is made concrete by naming what “one has” according to the symptoms. In this construction, the medication’s importance is to provide quality and years of life: *“This is an issue in which I understand perfectly the need for having efficient medication, allowing us to provide to the patient, let’s say, a horizon for that patient”* (S-6).

Considering the manner in which the social actors consider representations, as inserted in the network of social practices and through the relationship with other particular practices, diagnosis is seen as both an identity and a field in which imprecisions are obstacles for the needs of life. In diagnosis, the disease transcends the clinical manifestations and becomes increasingly more related to other dimensions of life. Therefore, we understand the concept of diagnosis, not as con-

finied to the disease itself, but in the sphere of the health condition and in its (re)arrangements in terms of daily practices¹².

Moreover, diagnosis incorporates brands, the stigma, and the symptoms to clarify the common points and the singularities among those who embody it¹³. It can also be defined as a mechanism of analysis of a wider society system, in which the practices of the social actors and their relationship with science, technology, and social rights are located.

[...] diagnosis still is é... I think it is a key element... (I-3).

So, if I could put diagnosis on a scale of importance, it is indeed the most important thing, no question about it. All of the policies, the guidelines, are based on the patient with cystic fibrosis, and not on the patient without cystic fibrosis. So, diagnosis is highly important, on a scale of 0 to 10, it is 10 (I-2).

In the discourses, diagnosis is considered to be very important. It is constructed in the negotiated expressions in daily comments and in the scientific arguments of social actors, such as “key element” (I-3) and/or as the apex in a graduated scale. Furthermore, it relates, almost instantaneously, to the means necessary for the articulation of its (in)definition.

Many mothers did not have a set diagnosis of the disease. So, they could do nothing, they could not take legal measures, could not get the support from the State. They live for the purpose of taking care of their children, with no legal support, no social support, nothing (APF).

A typical report, including two pathogenic variants is easy; however, the reports that are kind of on the fence, the variant with uncertain meaning, if it is cis, if it is trans, if one single variant is detected, then things start to get complicated (S-3).

The use of metaphors like *“on the fence”* (S-3) and the antithesis *“set”* (APF) to show the definitions related to diagnosis and the consequences in life when it is absent, by the use of the negative adverb *“no”*, the preposition *“without”* and the term *“nothing”*, when evoking the legislative and interventionist character of the State in the guarantee of effectiveness and in relation to the other “points” of care that having a diagnosis might mean for someone’s life.

The centrality of the diagnosis is noticeable in the very textual construction of the discourses, in the use of expressions such as *“diagnosis still is”*, as well as in the use of the present tense *“it is”*, *“it has”*, *“it achieves”*, and in the sequences described in the past, demonstrating a process of the events

until their definition and an enumeration in order to define the degrees of importance.

The presence of diagnosis confirming identity and belonging is intertwined, in terms of discourse, with other social processes: as a guide for actions, in the search for access (made into a pilgrimage), in terms of participation in the understanding about the market of diagnosis technologies and/or research and development, and the struggles to achieve one's rights, which may include legal measures.

Still regarding diagnosis, it is important to include a Goffmanian analysis, when in the discourses of the social actors, what is at stake is not just the physical space, but also the social situations, the interactions, and the reflective and recontextualized representations¹⁴. Therefore, diagnosis as a semiotic element is materialized in the representations, articulates what is general and particular, and is not limited to the doctor-patient binome, since it is influenced by beliefs, values, and disputes¹².

Diagnosis becomes a mediator – in the necessary interaction of a codification for the disease – and to become such, it makes use of the contemporary methods and technologies, which are increasingly more precise. It also becomes a structuring element of social practice, since it relates to needs and demands in health. Diagnosis becomes an “efficient lenses through which social aspects in health can be seen”¹³ (p. 845).

[...] my opinion is: cystic fibrosis is a disease that does not affect one specific organ, right? It affects the body as a whole [...] Diagnosis is essential; however, after it, there is an entire universe, only those who work with it or live through it can really tell (A-2).

Diagnosis is only the first step, and it is the dividing line, because once you know what you have, you can try to adapt – I wouldn't say, like, having no symptoms – but trying to minimize them (PF).

As a complex health condition, cystic fibrosis transcends the physiopathological aspect that defines it. Diagnosis is something present in the metaphorical statement by A-2, wondering “It is a path, right?” or in PF's metonymy “first step”. In this path, which begins with the essential criteria of a diagnosis, as a definition of what is happening to the body, also builds itself in the discourse as a “dividing line” (PF) or something that “affects the body as a whole” (A-2), by bringing changes which impose themselves and materialize in terms of life and needs.

The figures of language operate as an environment for discourse production as images that

facilitate interlocution with the audience, building and maintaining discourses in a political environment. As we can see in the hyperbole: *world that is opened, a huge window, that ranges from the emotional level to the financial structure level (A-2)*, or in the metonymy highlighted in the statement by S-1: *Few families are able to face this disease in a positive manner. In general, everyone says: ‘you pulled the rug from under our feet with this diagnosis’.*

The social practices are translated from the intensity of the experience, unrestricted to the name itself, and to what it represents in terms of symptoms. It appears in the discourses (even in the most biomedical ones in terms of language) as limitations and possibilities of access to integral care. It is noticeable the presence of the disruptive experience of the announced diagnosis, which is unknown and rare.

In this sense, light is shed upon the demands and needs in terms of the health care present in the semiotics of the (im)precisions in the diagnosis, and how this diagnosis is built in terms of meaning and sense. It constitutes and is constituted in the discourses of the social actors faced with the socioeconomic and cultural aspects of language and informed knowledge, of diagnosis and treatment, and of time spent with the daily routine of treatment; the construction of autonomy and identity; the transition into adult care; and the construction of sexual rights, reproductive rights, and a productive life represented by possibilities and potentialities.

[...] the diagnosis is the first barrier [...] Some still have the difficulty of the lack of a neonatal triage in stage III, which was universalized in 2011, so diagnosis is still a barrier, and we still have many late diagnoses in Brazil; access to confirmatory exams is also difficult... Few health facilities do the sweat test, and even fewer do genetic testing. So, this is still a barrier (A-1).

As far as exams go, there is the issue of the newborn blood spot test, which stopped being offered for a while because we ran out of the marker. We ended up with a big six-month gap in terms of tests for patients (A-3).

By adopting the words “barriers”, “limitations”, “gap” in the discursive textual constructs, occasionally presented together with ordinal or cardinal numbering, the social actors characterize the lack of healthcare for cystic fibrosis and/or what can be identified as health needs to be addressed.

However, when reflecting in terms of the regency of the words presented in the previous pa-

ragraph, there was the mention of “barriers to”, “limitations to”, “gaps of”. There is a demand for a complement which is capable of providing more sense, or in other words, when looking dialectically into the discursive structures used by the social actors in the discourses, we wonder: what nominal complement could make them more intelligible? Our conclusion is that the sense of the words directs us to the concept of *access*. Access, in the discourse by the social actors, is recognized as a health need, which has an impact on the quality of health care offered – *I believe that access is another ‘gap’ which must be looked into with care, with great care* (I-1).

Access goes beyond the textual constructs and the discourse of diagnosis, of treatment, of the existence and the structure of reference facilities, and of information, and has a position in current concerns regarding CFTR medication. In this sense, more than being a concept in the perspective of integral care for cystic fibrosis, access is something which appears as an emic category in the discourses, as it acts upon the representation of social practices, resulting in more attention (to and from) the value of that access.

As semantic production, access may have a meaning that is close to accessibility, with the existence of theoretical models which relate to both concepts (access and accessibility) and refer to healthcare. It is also evident in the variability which results from the relationship between individuals and the providing of services, or as objectives/results by healthcare systems in relation to what is established with the elements included within it¹⁵.

In general terms, the concept of access involves the entry into the healthcare system and the results in terms of the providing of health care. There is a current trend to use the concept to refer to the results of the care received, with the presence of explanatory models focused on “access and use of healthcare services; access and continuity of healthcare; and access to effectiveness in terms of the care provided”¹⁵ (p. 197).

As regards accessibility, the use defined by Donabedian (1973) relates to the quality of being accessible as one of the aspects of the offer of services and responses to the needs of life. In other words, the characteristics which facilitate or limit use by a given population are relevant in the dimension of equity of healthcare systems¹⁵.

In the conceptual review of access to healthcare, there is also the possibility of having access to use of healthcare services, together with multiple other factors (individual, contextual, quality,

and effectiveness of care). In general terms, it is defined by determining factors of the use of healthcare services, whose related factors include: a) health needs; b) demographic, geographic, cultural, psychic and socioeconomic aspects of the users; c) service providers; d) characteristics of the resources and availability to provide services; e) aspects related to politics (the kind and the manner of financing of the healthcare systems; the quantity, distribution, and regulation regarding the services; as well as the professionals and the systems themselves)¹⁵.

Considering the complexity that involves the concept of access to health, and the possibility of this concept being understood in terms of indicators of processes and results, four dimensions can be considered: availability, acceptability, ability to pay, and information. These dimensions of access establish relationships with less tangible aspects, such as culture, education, and socioeconomic status. Moreover, improvements in access to health care also require intersectoral actions, along with other social and economic policies¹⁶.

Based on this dialogue about the concept of access, we discussed the discursive excerpts of the social actors regarding cystic fibrosis, shedding light on some inferences. First, one needs to perceive how senses constitute and are constituted by the equivalence of the appropriate use of healthcare services, and to what extent they correspond to an efficient answer to healthcare needs.

Cystic fibrosis is a complex disease. It is basically multidisciplinary. Therefore, there are questions which are not just medical, but also psychological, social... many times children, from the time they are little, need to use medication continuous, they often have to be hospitalized, which results in issues like missing school or repeating a school year. So, I would say, it is not restricted to the disease in medical terms, the surrounding reality of the child becomes more difficult. And how does that affect the family? This is a view that we often have (S-1).

The complexity that defines the conceptualizing of access to health is also an integral part of cystic fibrosis. Our study made reference to this conceptual complexity, which permeates (and is permeated by) senses and meanings (in terms of definition), and to what it represents in daily life. For instance, if we consider the dimension of acceptability¹⁶ in terms of information and connection, we can see how centrally and intervening it constitutes a “therapeutic pilgrimage”, in terms of rare diseases in general and in terms of cystic fibrosis, which is the core theme of this study. This

dimension has an interface with stigma, which tends to repel both common people and non-specialized professionals, educators, and other elements of sociability.

Such complexity lies in the fact that it “*is not just medical*”, or psychological, or family related, but it is present integrally in life and in the social practices of those who live with it, advocate for it, work on it, produce science and technology for it. Therefore, in terms of integral care, it constitutes itself in the availability, acceptability, financing, and information.

[...] *she noticed that the newborn blood spot test could not be performed by a lack of inputs, by a lack of professionals, so there were blood spot tests that took a year to be done, even though it should be done in two weeks at most. And it is part of the SUS system, so diagnosis is still a challenge* (APF)

To go beyond the limits of diagnosis primarily means to access the necessary, as it can be defined. The discourses of the social actors have textual constructs that are related to absences, disparities, interruptions of exams, and a lack of inputs and services that can ensure diagnosis, such as neonatal triage (“*newborn blood spot test*” – APF) and the Sweat test at SUS. That is parallel to the unequal realities that occur in Brazil, requiring more efforts to be better addressed.

We need some political attitude or action from the Ministry of Health for homogenizing this in the entire national territory [of Brazil]. This is still a problem. Another difficulty that I see is that the confirmation exam, which is the Sweat Test, in some states, we can see that it works quite smoothly, while in some others, there are enormous difficulties to conduct the Sweat Test (I-3).

The National Program of Neonatal Triage, universalized in 2011, includes the triage for cystic fibrosis in stage III. The Clinical Protocol and Therapeutic Directives for cystic fibrosis (in its last revision, 2021) establishes that two altered results for Immunoreactive trypsin (IRT), conducted with newborns, in the interval of three to four weeks of age, indicates the need to investigate by means of electrolytes present in the baby’s sweat – the Sweat Test. This consequently requires follow up by a specialized reference center.

We understand that, although present positively in terms of decisions and protocols, it does not result in equal access in terms of availability at SUS. It does, however, create the possibility of including it for discussion in agendas, in a much needed interaction, aimed at *homogenizing this in the entire national territory [of Brazil]* (I-3). In other words, in the synonymy of the verb “to ho-

mogenize”, it is defined as something that appropriates similarity in terms of structure, function, and distribution, guaranteeing thus unity and effectiveness throughout Brazil.

The discourses of the social actors regarding access to diagnosis and the representation of senses are structured in two main points: a) a bumpy trajectory due to diverse healthcare services, the carrying out of tests and consultations with specialists in search of a definition; b) reach, quality, quantity, and functioning of the healthcare services provided, which report to the availability (or not) of inputs, equipment, and appropriate means for the very diagnosis.

However, we noticed that, be it due to the bumpy road in search of a diagnostic definition, or be it due to the limitations in effective coverage of healthcare services, diagnosis is built discursively in terms of a “*process*” (PCF) or by a “*set of actions*” (PCF) which are adjectively expressed as “*very difficult*” (A-3). This is then personified in the direct object of the phrase “*you waste a lot of time*” (A-3).

Meanwhile, the strategies to face the problem appear in the field of semantics as “*partnership*” (A-3) and “*the help*” (A-3), understood here as a (re)joining of social actors and strategies mobilized in the search for qualified health care.

That “*process*” (PCF) or “*sets of actions*” (PCF) which are, by nature, difficult, may also appear as late diagnoses of cystic fibrosis, regardless of the onset of considerable symptoms, enabling an unwanted (un)predictability of what is to come – *therefore we should have a wave of patients that will soon appear with respiratory symptoms, gastric ones, with a diagnosis or not* (A-3).

The Brazilian cystic fibrosis registration system (*Registro Brasileiro de Fibrose Cística – RE-BRACF*)¹⁷ indicates that the median age of diagnosis over the past six years remained below six months. However, in terms of conditions for the diagnosis, availability of access to an effective coverage of neonatal triage is below what is found for persistent respiratory manifestations and for deficit in growth/undernourishment, respectively.

As Brazil is a country of continental dimension, the presence of several “*Brazils*” (as mentioned by the anthropologist Darci Ribeiro) is quite notorious, considering the sociocultural and economic diversity of the country, which in turn determines inferences in access to health care and the satisfaction of patient needs. That, in the representation of order in the discourse on cystic fibrosis, appears as the “*without*” and the “*no-*

thing” (APF) which live side by side with the country of the “well” and the “has” (S-6), and appears as well in the dimensions of access to diagnosis.

Access to diagnosis proves to be a paradox in the discursive and social practices of the social actors. Though it appears in the social-territorial Brazilian context as collective experiences of “very difficult”, “erroneously”, “not”, “without”, “takes very long”, there is also the “well cared for”, “have”, “fast and efficient”, “early”, “correct”. This means, in discourse production, that there is a paradoxical dimension adjectivating the (im) precision of access to health.

By contrast, the statement – *It is something golden and goes faster than the time we have, so we cannot wait for our time, we have to move in the time that our patients have* (S-6) – has as its semantic nucleus, the word “time”, characterized as being indefinite, precious, and swift, perceived differently by the social actors whose pace must be established by the person that suffers from cystic fibrosis.

“Time” (S-6) translates representations of sense, considered cross-sectional in the entire rationale of seeking medical care, since this concept of time is expressed in the objectivity of the diagnosis and/or as the access to and relative length of the treatment, as well as in terms of the moment that is being lived and in which life is happening, in terms of opportunities, considering that (im) precision is a part of life.

The representation of “time” in terms of treatment appears as a semiotic element of the discourse. It is present in the dimensions of access, in terms of place of reference for treatment and in terms of medication. It is also defined as time dedicated to taking medication and undergoing therapy. Moreover, it acquires a dimension, amid access to and satisfaction of basic survival needs, related to aspects which are inherent to the centers of reference, such as location, physical structure, and composition of multidisciplinary teams.

We know that the life expectancy of these patients has been increasing with the passing of years, with the development of therapies, with access to medication, for example, tobramycin. Therefore, there is an improvement in the quality of life for these patients, and that, consequently, has increased life expectancy. Consequently, these patients are living longer. They will have, for instance, a transition, from treatment in a pediatric facility to an adult facility; consequently, it is not simply a matter of the diagnosis (I-4).

The discursive elements around the treatment may appear in terms of the expected outcome,

such as in the textual constructs: “increase in life expectancy” (I-4), “improvement in quality of life” (I-4), “access to medication” (I-4), and even more specifically when mentioning care at the time of the transition to adult age. However, treatment is not represented discursively in a linear manner, following diagnosis, it rather relates to disputes in the social arenas and to the very experience of the health condition.

This treatment, the PCDT, today includes only the digestive enzymes, dornase alfa and tobramycin, which is one of the antibiotics used for cystic fibrosis, and there are states which only have those medications available, and states where, by lawsuits or by state law, or state protocols, managed to include more therapeutic options (A-1)

From the standpoint of availability of access to treatment, we see in the discourse the reference to the existence of PCDT in terms of medication already incorporated by SUS (such as the recent CFTR modulator: active principle, *ivacaftor*), also present in the struggles for its ensured provision and equanimous access, through lawsuits, agreements, and pacts at federated instances, according to the dynamics of the correlation of forces that may be present in each occasion.

We can present access to treatment in relation to the dispute between what is needed versus what is offered, as in “only includes” (A-1) and “only those available” (A-1), from a financial aspect related to the costs of the healthcare system. However, the struggle to “include more” (A-1) implies considering the indirect costs of care for the families.

Therefore, in the discourses, access to what is basic care still produces realities defined by “do not have”, “lacks”, “deserted” (A-3) in the correspondence established with “what is needed”, although already predicted. And the use of legal instruments – “injunctions” (A-3) “public civil lawsuit” (A-2) “resource lock” (A-3) – as the most accessible means to make the transition to the semantic field of having, of what is due – “obligation” (A-2) – thereby enabling, in the Brazilian scenario, access to diagnosis and integral care for cystic fibrosis patients.

Conclusion

Cystic fibrosis produces impacts of a socioeconomic, cultural, subjective, and identity nature, related to the daily, shared experience of caring for the disease. It is an issue that is present, from the trajectories in search of diagnostic definitions, to

the very process of caring for the disease, often performed by the woman as a duty implicit in the concept of maternity and managing care in the family realm.

There are also impacts in the process of the socialization of children and teenagers with their peers, often related to the differentiation that can be seen in the physical impression of their bodies and in the symptoms, such as constant coughing and secretion, frequent hospitalization interrupting one's school routine, the wearing of masks for respiratory protection, among others.

Therefore, cystic fibrosis, understood as a rare health condition, has a perspective of care which incorporates the social determinants of sickness, which address life, the sphere of rights, the guarantee of care, social iniquity, and the organization of the public health system, bringing demands and needs in terms of health. It places health in a central position and interconnects many other needs, for which groups, families and associations are mobilized in the search for quality of life and the guarantee of rights.

Hence, in the discourses of the social actors who are part of the specialist community, diagnosis takes on a key role; it is a sort of passport

for access to and definition of the health conditions and their treatments. It is pressing that diagnosis is interpreted as part of an identity, collective construct, essential in the search for rights and the consolidation of means through which to access those rights.

However, once the diagnosis is obtained, a journey begins, with the tensioning of social arenas. In the struggle to receive the diagnosis, it is required/necessary to have access to medical services, such as Neonatal Triage, the Sweat Test, and Genetic Exams, available and provided by SUS for the entire territory of Brazil. We reiterate the need to ensure the existence of means through which to satisfy the health needs resulting from a rare health condition that is chronic and complex.

In the discourse constructions, integrality of care describes in *precision*, what is necessary, from an equitable providing of the basic need for diagnosis and treatment. Access itself, understood as an emic category in this type of discourse, provides clear evidence of barriers, even in the consolidation of the bare minimum of the health needs and the right to life.

Collaborations

KAC Marins: created the study, analyzed the data collection, and drafted the article. MCN Moreira: guided the methodological design, reviewed the data collection, and drafted the article.

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