

Anna Giulia Mello Paiva

**Significados produzidos pelos cuidadores de crianças com
Doenças Raras sobre a trajetória de cuidado em saúde: um
estudo qualitativo**

*Meanings produced by caregivers of children with Rare Diseases
about healthcare trajectory: a qualitative study*

Dissertação apresentada à
Faculdade de Odontologia da
Universidade Federal de Uberlândia,
para a obtenção do Título de Mestre
em Odontologia, Área de Clínica
Odontológica Integrada

Uberlândia, 2023

Anna Giulia Mello Paiva

Significados produzidos pelos cuidadores de crianças com Doenças Raras sobre a trajetória de cuidado em saúde: um estudo qualitativo

Meanings produced by caregivers of children with Rare Diseases about healthcare trajectory: a qualitative study

Dissertação apresentada à Faculdade de Odontologia da Universidade Federal de Uberlândia, para a obtenção do Título de Mestre em Odontologia, Área de Clínica Odontológica Integrada

Orientador: Prof. Dr. Álex Moreira Herval

Banca Examinadora:

Prof. Dr. Álex Moreira Herval (UFU)

Profa. Dra. Jaqueline Vilela Bulgareli (UFU)

Profa. Dra. Viviane Elisângela Gomes (UFMG)

Profa. Dra. Alessandra Maia de C. Prado (UFU)

Prod. Dr. Túlio Eduardo Nogueira (UFG)

Uberlândia, 2023



UNIVERSIDADE FEDERAL DE UBERLÂNDIA
Coordenação do Programa de Pós-Graduação em Odontologia
Av. Pará, 1720, Bloco 4L, Anexo B, Sala 35 - Bairro Umuarama, Uberlândia-MG, CEP 38400-902
Telefone: (34) 3225-8115/8108 - www.ppgoufu.com - copod@umuarama.ufu.br



ATA DE DEFESA - PÓS-GRADUAÇÃO

Programa de Pós-Graduação em:	Odontologia				
Defesa de:	Dissertação de Mestrado Acadêmico, nº 435 , PPGODONTO				
Data:	Vinte e Cinco de Outubro de Dois Mil e Vinte e Três	Hora de início:	08:00	Hora de encerramento:	10:45
Matrícula do Discente:	12122ODO005				
Nome do Discente:	Anna Giulia Mello Paiva				
Título do Trabalho:	Significado produzido pelos cuidadores de crianças com Doenças Raras sobre a trajetória de cuidado em saúde: um estudo qualitativo				
Área de concentração:	Anna Giulia Mello Paiva				
Linha de pesquisa:	Patologia e Diagnóstico Bucal				
Projeto de Pesquisa de vinculação:	Patologia e Diagnóstico Bucal				

Reuniu-se em Web Conferência pela plataforma Zoom, em conformidade com a PORTARIA Nº 36, DE 19 DE MARÇO DE 2020 da COORDENAÇÃO DE APERFEIÇOAMENTO DE PESSOAL DE NÍVEL SUPERIOR - CAPES, pela Universidade Federal de Uberlândia, a Banca Examinadora, designada pelo Colegiado do Programa de Pós-graduação em Odontologia, assim composta: Professores Doutores: Viviane Elisângela Gomes (UFMG); Jaqueline Vilela Bulgareli (UFU); Álex Moreira Herval (UFU) orientador da candidata.

Iniciando os trabalhos o(a) presidente da mesa, Dr. Álex Moreira Herval, apresentou a Comissão Examinadora e o candidata, agradeceu a presença do público, e concedeu ao Discente a palavra para a exposição do seu trabalho. A duração da apresentação do Discente e o tempo de arguição e resposta foram conforme as normas do Programa.

A seguir o senhor(a) presidente concedeu a palavra, pela ordem sucessivamente, às examinadoras, que passaram a arguir a candidata. Ultimada a arguição, que se desenvolveu dentro dos termos regimentais, a Banca, em sessão secreta, atribuiu o resultado final, considerando a candidata:

Aprovada.

Esta defesa faz parte dos requisitos necessários à obtenção do título de Mestre.

O competente diploma será expedido após cumprimento dos demais requisitos, conforme as normas do Programa, a legislação pertinente e a regulamentação interna da UFU.

Nada mais havendo a tratar foram encerrados os trabalhos. Foi lavrada a presente ata que após lida e achada conforme foi assinada pela Banca Examinadora.



Documento assinado eletronicamente por **Alex Moreira Herval, Professor(a) do Magistério Superior**, em 25/10/2023, às 09:35, conforme horário oficial de Brasília, com fundamento no art. 6º, § 1º, do [Decreto nº 8.539, de 8 de outubro de 2015](#).



Documento assinado eletronicamente por **Jaqueline Vilela Bulgareli, Professor(a) do Magistério Superior**, em 25/10/2023, às 09:36, conforme horário oficial de Brasília, com fundamento no art. 6º, § 1º, do [Decreto nº 8.539, de 8 de outubro de 2015](#).



Documento assinado eletronicamente por **Viviane Elisângela Gomes, Usuário Externo**, em 25/10/2023, às 09:44, conforme horário oficial de Brasília, com fundamento no art. 6º, § 1º, do [Decreto nº 8.539, de 8 de outubro de 2015](#).



A autenticidade deste documento pode ser conferida no site https://www.sei.ufu.br/sei/controlador_externo.php?acao=documento_conferir&id_orgao_acesso_externo=0, informando o código verificador **4910380** e o código CRC **D15CDBDD**.

Ficha Catalográfica Online do Sistema de Bibliotecas da UFU
com dados informados pelo(a) próprio(a) autor(a).

P149 2023	Paiva, Anna Giulia Mello, 1994- Significados produzidos pelos cuidadores de crianças com Doenças Raras sobre a trajetória de cuidado em saúde: um estudo qualitativo [recurso eletrônico] / Anna Giulia Mello Paiva. - 2023. Orientador: Álex Moreira Herval. Coorientador: Fabiana Sodr� de Oliveira. Disserta�o (Mestrado) - Universidade Federal de Uberl�ndia, P�s-gradua�o em Odontologia. Modo de acesso: Internet. Dispon�vel em: http://doi.org/10.14393/ufu.di.2023.566 Inclui bibliografia. 1. Odontologia. I. Herval, �lex Moreira, 1989-, (Orient.). II. Oliveira, Fabiana Sodr� de , 1967-, (Coorient.). III. Universidade Federal de Uberl�ndia. P�s-gradua�o em Odontologia. IV. T�tulo. CDU: 616.314
--------------	---

Bibliotec rios respons veis pela estrutura de acordo com o AACR2:
Gizele Cristine Nunes do Couto - CRB6/2091
Nelson Marcos Ferreira - CRB6/3074

DEDICATÓRIA

À todas as mães de crianças com Doenças Raras
por toda dedicação, amor e força.

AGRADECIMENTOS

À Deus por todo amor e todos os cuidados como Pai.

Ao meu marido, *Vinicius Henrique Batista Paiva*, que em todos os momentos sempre acreditou em mim, sempre me motivou e nunca soltou minha mão, mesmo com tantas dificuldades pelo caminho.

À minha querida avó, *Dagmar Borges Gonçalves*, que hoje está nos braços do Pai, mas sempre foi um alicerce na minha vida e sei que de onde ela estiver, continua emanando muitas bençãos e luz.

Ao meu orientador *Prof. Dr. Álex Moreira Herval* que além de um orientador incrível, foi uma verdadeira fortaleza em todos os percalços nessa trajetória.

À *Profa. Dra. Fabiana Sodr  de Oliveira* que me abriu portas para esse estudo maravilhoso e t o necess rio, al m de ser uma grande amiga e conselheira.

A todos os professores do PPGO-UFU que participaram e contrib ram compartilhando seus conhecimentos para a minha forma  o como mestre em odontologia e t m m   toda equipe do PPGO e em especial aos coordenadores e secret rias que sempre se dispuseram prontamente a orientar e auxiliar na condu  o desta jornada.

  minha fam lia e amigos que me deram for a, me incentivaram o tempo todo e sempre refor aram a confian a que depositam em mim.

Finalmente, agrade o   Funda  o de Amparo   Pesquisa do Estado de Minas Gerais (FAPEMIG) pela bolsa de estudos e   Coordena  o de Aperfei oamento do Pessoal de N vel Superior (CAPES) pelo apoio financeiro.

SUMÁRIO

LISTA DE ABREVIATURAS	10
RESUMO/ PALAVRAS-CHAVE	11
ABSTRACT/ KEYWORDS	12
1. INTRODUÇÃO E REFERENCIAL TEÓRICO	13
2.CAPÍTULO ÚNICO	16
REFERÊNCIAS	39

LISTA DE ABREVIATURAS E SIGLAS

OMS	Organização Mundial de Saúde
DR	Doenças Raras
PNAIPDR	Política Nacional de Atenção Integral às Pessoas com Doenças Raras
SUS	Sistema Único de Saúde

RESUMO

Introdução: Cuidadores de crianças com Doenças Raras enfrentam dificuldades para efetivarem uma adequada atenção às suas crianças, apesar de existirem políticas públicas voltadas para esses pacientes. *Objetivo:* Construir uma teorização sobre os significados produzidos por cuidadores de crianças com Doenças Raras na trajetória de cuidado em saúde. *Metodologia:* Foi realizado um estudo qualitativo com abordagem da Teoria Fundamentada nos Dados e referencial teórico do Interacionismo Simbólico. Os dados foram coletados no Hospital Odontológico da Universidade Federal de Uberlândia por meio de entrevistas semiestruturadas, audiogravadas e analisados por meio da Teoria Fundamentada dos Dados. *Resultados:* Foram realizadas oito entrevistas com mães de crianças com Doenças Raras. A análise do material qualitativo produzido possibilitou identificar os significados produzidos nas trajetórias de cuidados e a organização em cinco categorias: 1) Rede de saúde insatisfatória; 2) Contexto de dúvidas e medos; 3) Angústia no tempo de espera por atendimento; 4) Alívio e desespero diante do diagnóstico; 5) Atendimento odontológico integrado. O significado de uma rede de saúde insatisfatória foi definido como a categoria central. *Conclusão:* O significado central é uma rede pública de saúde insatisfatória para lidar com as Doenças Raras, seja no aspecto estrutural da rede de serviços ou nas posturas profissionais. No contexto desse estudo, o atendimento odontológico foi dado como eficiente.

Palavras-Chave: Doenças Raras; Políticas de Saúde; Acesso aos Serviços de Saúde; Assistência Odontológica; Pesquisa Qualitativa.

ABSTRACT

Introduction: Caregivers of children with Rare Diseases face difficulties in providing adequate care for their children, despite the existence of public policies aimed at these patients. *Objective:* Build a theorization about the meanings produced by caregivers of children with Rare Diseases in the health care trajectory. *Methodology:* A qualitative study was carried out using a Grounded Theory approach and the theoretical framework of Symbolic Interactionism. Data were collected at the Dental Hospital of the Federal University of Uberlândia through semi-structured, audio-recorded interviews and analyzed using Grounded Data Theory. *Results:* Eight interviews were carried out with mothers of children with Rare Diseases. The analysis of the qualitative material produced made it possible to identify the meanings produced in the care trajectories and the organization into five categories: 1) Unsatisfactory health network; 2) Context of doubts and fears; 3) Distress in the waiting time for care; 4) Relief and despair at the diagnosis; 5) Integrated dental care. The meaning of an unsatisfactory healthcare network was defined as the central category. *Conclusion:* The central meaning is an unsatisfactory public health network for dealing with Rare Diseases, whether in the structural aspect of the service network or in professional attitudes. In the context of this study, dental care was considered efficient.

KEYWORDS: Rare Diseases; Health policies; Access to Health Services; Dental care; Qualitative research.

1. INTRODUÇÃO E REFERENCIAL TEÓRICO

A Organização Mundial de Saúde (OMS) classifica como rara a doença com prevalência baixa. Entretanto, os parâmetros para determinar essa prevalência pode variar de um país para outro, especialmente em função da inexistência de um limite de prevalência para essa classificação. No Brasil, as Doenças Raras (DR) são aquelas que afetam menos de 65 pessoas a cada 100 mil indivíduos (BRASIL, 2014), alcançando, em média, 13 a 15 milhões de pessoas acometidas (AURELIANO, 2018; BRASIL, 2019; IRIART et al., 2019). Apesar da baixa prevalência de cada DR, há cerca de 6 a 8 mil doenças diferentes classificadas como raras (LOPES et al, 2018).

As DR são difíceis de serem diagnosticadas e aglutinam um grupo heterogêneo de doenças, como as anomalias congênitas, os erros inatos do metabolismo, os erros inatos da imunidade, as deficiências intelectuais (CHUNG et al, 2022). A maioria dessas doenças possui algum tipo de componente genético, mas outras etiologias podem estar associadas, como os fatores nutricionais, os fatores ambientais, os medicamentos e os agentes teratogênicos (BRASIL, 2022).

Para organização do cuidado às pessoas com DR no Brasil, o Ministério da Saúde propôs, no ano de 2014, a Política Nacional de Atenção Integral às Pessoas com Doenças Raras (PNAIPDR), cujo objetivo era reduzir a morbimortalidade e contribuir para a redução das manifestações secundárias dessas doenças (BRASIL, 2014). Essa política aponta que a atenção às condições genéticas deve fazer parte tanto da Atenção Primária à Saúde, quanto dos Centros de Referência Médica Regionais, sendo que estes últimos deveriam receber financiamento para realização de testes e aconselhamento genéticos (AURELIANO, 2018).

Embora as diretrizes do Sistema Único de Saúde (SUS) para atenção integral dos indivíduos com DR prevejam o atendimento multiprofissional destes pacientes, o acesso a esses profissionais ainda é precário, seja pela escassez de recursos materiais e humano, seja pelos desafios organizacionais e regulatórios da Rede de Atenção à Saúde do SUS (IRIART et al, 2019).

Nos últimos anos, as DR obtiveram uma maior visibilidade mundial, embora ainda exista uma falta de conhecimento por parte de muitos profissionais, limitação de investimentos em pesquisas e uma considerável carência de medicamentos disponíveis no SUS (AMARAL, REGO, 2020). Esse cenário contrasta com as necessidades assistenciais das pessoas ou famílias acometidas por estas doenças, que são complexas em termos de diagnóstico, tratamento e acompanhamento (IRIART et al, 2019). As constantes mudanças nas dinâmicas sociais e familiares, gerada pelos longos períodos de internação ou de cuidado domiciliar intensivo, trazem elevado sofrimento físico e psicossocial às famílias e pessoas com DR (BRASIL, 2022).

Os cuidadores de crianças com DR desempenham um papel crucial no bem-estar físico e emocional da criança afetada (WETTERAUER, SCHUSTER, 2008). No entanto, as dificuldades enfrentadas por esses cuidadores ao longo de suas vidas podem gerar impactos substanciais em suas vidas nos âmbitos profissional, social e familiar (BOETTCHER et al, 2021), promovendo prejuízos ao seu próprio bem-estar (ZURYNSKI et al, 2008). O processo de enfrentamento do adoecimento de uma criança é uma dificuldade encontrada nos relatos dos pais e, muitas vezes, a origem genética das DR faz com que os pais tendem a se culpar pela doença dos seus filhos (WITT et al, 2021). O longo processo de aceitação do estado de saúde da criança é muitas vezes acompanhado pela falta de aceitação de terapias específicas e de ajuda psicossocial, pois muitas vezes as famílias não reconhecem que precisam de apoio pois apresentam preconceitos contra os cuidados psicossociais e receiam ser estigmatizados ou considerados fracos devido à sua necessidade de ajuda (WITT et al, 2021).

Vale destacar o papel culturalmente ocupado pelas mulheres na dinâmica familiar é o de responsável pelo cuidado, ou seja, diante de um adoecimento quem assume esta posição de cuidador, normalmente, é a mãe, que podem acabar por negligenciar suas próprias necessidades e iniciar sintomas depressivos (GIMENES, ZANETTI, HAAS, 2009; LOPES et al., 2018; OLIVEIRA et al., 2016). Nesse sentido, o acompanhamento psicológico por parte dos cuidadores de crianças com DR é relevante para melhor lidar com sentimento de frustração, vergonha, culpa e tristeza (BROTTO, ROSANELI, 2021).

Muitas vezes os cuidadores das crianças com DR buscam formas de reduzir os desconfortos em relação ao “ser diferente”, minimizando, assim, sofrimentos relacionados a uma situação de não enquadramento do filho aos padrões impostos como normalidade, tanto pelos profissionais de saúde quanto pela sociedade em geral (SOARES, ARAUJO, BELLATO, 2016). O diagnóstico de uma DR pode desencadear a presença de vulnerabilidades morais, seja na discriminação e/ou estigmatização, ou até mesmo devido privações de atividades diárias geradas pelo capacitismo das pessoas (FELIPE et al, 2020). Algumas das experiências negativas assistidas pelos cuidadores de crianças com DR incluem a exclusão da criança, o isolamento e o bullying e, muitas das vezes, a aceitação é influenciada através da forma como os pais são aceitos, valorizados e incluídos pela escola e pela comunidade ao qual vivem, além do diagnóstico e da gravidade da DR da criança (FOSTER et al, 2022).

Muitas das vezes, os cuidados odontológicos às crianças com DR podem ser dificultados (BALDOVINO et al, 2016), seja pela falta de profissionais e estruturas especializadas que oferecem condições de diagnosticar, acompanhar e tratar todas as doenças da forma multidisciplinar necessária (DENIS et al, 2010); seja porque as DR com comprometimento cognitivo impõem dificuldade para o cuidado em saúde bucal (FRIEDLANDER et al, 2023).

Por se tratar de uma temática ainda recente, viu-se a necessidade de aprofundamento sobre os desafios enfrentados por pessoas com DR em suas trajetórias de cuidado. Por isso, o objetivo desse estudo foi compreender os significados produzidos pelos cuidadores das crianças com DR, considerando todo o trajeto do cuidado em saúde, buscando, inclusive compreender o acesso ao atendimento odontológico.

2. CAPÍTULO ÚNICO

ARTIGO - Meanings produced by caregivers of children with Rare Diseases about healthcare trajectory: a qualitative study

Artigo a ser enviado para publicação no periódico *Special Care in Dentistry*

Anna Giulia Mello Paiva, Thamyres Isabel Borges de Andrade, Fabiana Sodré de Oliveira, Álex Moreira Herval

Meanings produced by caregivers of children with Rare Diseases about healthcare trajectory: a qualitative study

Anna Giulia Mello Paiva¹, Thamyres Isabel Borges de Andrade², Fabiana Sodré de Oliveira³, Álex Moreira Herval^{4*}

¹Post-Graduation Program in Dentistry, Universidade Federal de Uberlândia (UFU), Uberlândia, Minas Gerais, Brazil

²School of Dentistry, Universidade Federal de Uberlândia (UFU), Uberlândia, Minas Gerais, Brazil

³Area of Pediatric Dentistry, School of Dentistry, Universidade Federal de Uberlândia (UFU), Uberlândia, Minas Gerais, Brazil

⁴Area of Preventive and Community Dentistry, School of Dentistry, Universidade Federal de Uberlândia (UFU), Uberlândia, Minas Gerais, Brazil

*Corresponding author

Álex Moreira Herval

E-mail: alexmherval@ufu.br

Federal University of Uberlândia

School of Dentistry – Community and Preventive Health Area

Av. Pará, 1720, Bloco 2G

Uberlândia, MG, Brazil - ZIP Code: 38405-320

ACKNOWLEDGMENTS

The authors acknowledge to FAPEMIG and CAPES for partially funding this study.

Abstract

Introduction: Caregivers of children with Rare Diseases face difficulties in providing adequate care to their children, despite the existence of public policies aimed at these patients. **Objective:** To produce a theorization about the meanings produced by caregivers of children with Rare Diseases. **Methodology:** A qualitative study was carried out with a Grounded Theory and the theoretical framework of Symbolic Interactionism. Data were collected at the Dental Hospital of the Federal University of Uberlandia through semi-structured interviews, audio-recorded and categorized according to GT. **Results:** Eight interviews were conducted with mothers of children with Rare Diseases. The analysis of the qualitative material produced made it possible to identify the meanings produced in the care trajectories and the organization in five categories: 1) Unsatisfactory health network; 2) Context of doubts and fears; 3) Anguish in the waiting time for care; 4) Relief and despair in the face of the diagnosis; 5) Integrated dental care. It was identified that the unsatisfactory health network was defined as the central category. **Conclusion:** The central meaning is an unsatisfactory public health network to deal with Rare Diseases, either in the structural aspect of the service network or in professional postures. In the context of this study, dental care was considered efficient.

Keywords: Rare Diseases; Health policies; Access to Health Services; Dental care; Qualitative research.

INTRODUCTION

Rare Diseases (RD) generally have a chronic, progressive and disabling nature, and can also be degenerative, which influences the quality of life of affected people and their families¹. Considering the impact on quality of life, a support network is essential to help family members, and especially the mother, in facing the difficulties that emerge day after day². It is noted that caregivers of children with RD have become increasingly lonely, with insufficient support from institutions and absence of empathy from some professionals^{3,4}.

As a result of low prevalence, there is a known deficit of specific treatments for the majority of DR⁵, leaving some patients with only palliative treatments to alleviate the manifestations and symptoms of these diseases^{6,7}. In Brazil, the “*Política Nacional de Atenção às Pessoas com Doenças Raras*” (National Policy for Comprehensive Care for People with Rare Diseases) was proposed to organize care for people with RD, however there is a considerable delay in its implementation, with only 10 institutions reporting consultations and procedures to the Ministry of Health in 2020, resulting in delays in diagnosis and limited access to resources and treatment^{8,9}.

Among the challenges faced by family members and patients with RD, obtaining a diagnosis can be difficult because many signs and symptoms are similar to other common diseases¹⁰. The absence of information about the disease, stigma and prejudice impacts the quality of life of people with RD¹¹. The treatment of RD requires multidisciplinary monitoring by multidisciplinary team, including geneticist, physiotherapist, speech therapist, nutritionist and psychologist, among others, aiming to alleviate symptoms or delay their onset¹². Therefore, as a result to high demand for care, one of parents to give up work in order to dedicate themselves exclusively to child with RD¹¹.

Dental care for children with RD can often be hampered¹¹, either due to the lack of professionals and specialized services that offer conditions to diagnose, monitor and treat all diseases in the necessary multidisciplinary way¹²; or because some diseases with cognitive impairment do not have adequate services to manage their oral health in these patients¹⁰.

It is important to highlight that access to dental treatments must be discussed within the scope of multidisciplinary care for patients with RD. In this context, this study aims to produce a theorization about the meanings produced by caregivers of children with RD from diagnosis to treatment, also seeking the meanings attributed to access to dental treatment.

MATERIALS & METHODS

Study Design and Ethical Aspects

Qualitative study was developed, guided by Grounded Theory and theoretical framework of Symbolic Interactionism, to theorize about the meanings produced by caregivers of children with RD in healthcare trajectory (from diagnosis to treatment). This theoretical basis understands human behavior as result of interaction between individual and society, which results in construction of symbols and meanings that guide social interactions between individuals¹³. In this context, this research was based on identification of symbols produced, identification of self (self-awareness) and mind (the understanding between subjects), understanding of human action (dynamic process of taking decision), and interactions (communication and interpretation) of the interviewees¹⁴. The research protocol was assessed by the Research Ethics Committee of the Federal University of Uberlândia and approved by CAAE: 52311221.6.0000.5152.

Sampling and Study Participants

A convenience sample was used, which included caregivers of children with RD up to 10 years of age treated at the Dental Hospital of the Federal University of Uberlândia. In total, eight caregivers of children with RD were included. The sample size was determined by theoretical data saturation^{15,16}. The interviews were interrupted when the questioning about how to access dental care reached saturation.

Data Collections

Data collection was carried out remotely, which was previously scheduled and lasted an average of 12 minutes. The interviews took place from June/2022 to July/2023, when theoretical saturation occurred. The data collection method was based on semi-structured audio-recorded interviews, conducted by a single female interviewer (author: AGMP), a dental surgeon, with no previous contact with the interviewees, trained by the author AMH. The interviewer followed an interview guide with these main questions: 1) How was the diagnosis for you and your family?; 2) Tell me about the challenges you faced in getting the necessary treatments; 3) What strategies, in your opinion, facilitated access and healthcare for RD?; 4) What are the challenges in accessing dental care?

After each interview, the audio-recorded content was transcribed, when names were removed to preserve confidentiality of the participants. The caregivers interviewed were numbered sequentially as M1, M2, M3, and successively. After each interview, memos were created pointing out the first impressions found by the interviewer, which facilitated the identification of the categories elaborated.

Data analysis

The analysis of qualitative data produced in the interviews was executed using Grounded Theory from the Straussian post-positivist perspective¹⁶. All transcribed material resulting from the interviews was analyzed in three coding phases. In first stage

of coding (Open Coding), the transcribed qualitative material was exhaustively read and received line-by-line coding. Through initial coding, codes were created and analyzed in the next stage. In second stage of coding (Axial Coding), the codes were compared to understand the frequent and/or significant speeches produced by mothers of children with RD from diagnosis to treatment. Thus, the codes formed in previous stage were grouped and linked into explanatory categories of studied phenomenon. For this grouping, the Paradigmatic Model was used (context and intervening conditions, causes, strategies, consequences and phenomenon). In short, this Paradigmatic Model organizes the different categories previously identified in the first codification and establishes the relationships created between them, following the conditions presented. After theoretical saturation of the explanatory categories of phenomena, the last stage of coding began (Selective Coding). In this stage, categories created were compared and analyzed exhaustively to identify the central category (or main theme of the study). At the end of three coding stages, a theory was generated based on the elements that form the Paradigmatic Model.

RESULTS

Eight mothers of children aged 1-8 years old with the following RD were interviewed: Fanconi syndrome; Turner syndrome; Goldenhar syndrome; DiGeorge syndrome; Osteogenesis Imperfecta; and Interstitial Deletion in the Long Arm of Chromosomes 6 (Table 1). After open and axial coding, five categories emerged: unsatisfactory healthcare network; relief and desperation in face of diagnosis; context of doubts and fears; distress over the waiting time for care; and integrated dental care. They represent the meanings produced by mothers in their care trajectories. Parts of speech are presented in Table 2.

Relief and desperation in face of diagnosis: The healthcare trajectory begins with search for the diagnosis and the resulting reactions. There is a dichotomy of meanings attributed to diagnosis: relief and desperation. On the one hand, there is a relief attributed to end of initial journey (search for diagnosis), which helps to explain health condition of child. On the other hand, the diagnosis of an RD is frightening, brings desperation and helplessness in the face of the unknown, sometimes leading to persistent denial of the diagnosis.

Context of doubts and fears: The diagnosis of RD results in a context of fears and insecurities regarding health and future of child. This context emerges from lack of information about RD, which raises doubts about health child prognosis and social development.

Distress over the waiting time for care: Mothers attribute the meaning of insufficient health services to the waiting time for medical care and exams. However, it was evident that the time was not, in fact, too long. The notion of delay comes from anguish, the feeling of impotence as a result of to not being able to get answers about the disease and the perspective that the time to start treatment is crucial for the prognosis. Given this, there may be a loss of hope regarding the treatment of their children.

Unsatisfactory healthcare network: In the healthcare trajectory, mothers interviewed attributed to professionals and health services a meaning of unpreparedness, which comes from the experiences of professionals who are unaware of RD or even from insensitivity in dialogue with families. Faced with these experiences and meanings produced, mothers demonstrate an expectation of creating specialized RD services as a possible solution to these unwanted experiences, but also to know where to find support.

Integrated dental care: Mothers of children with RD consider inclusion in dental care as a surprise. It was not expected by mothers who received referral or scheduling call

with surprise, since it was not part of care previously sought or thought of by them, but whose possibility of happening arises during consultations at the clinical hospital.

The care trajectory of children with RD develops in a context of doubts and fears, since there is little information about these diseases, leaving families with few perspectives regarding healthcare and future of child. This context is full of uncertainty, diagnosis of an RD initially generates relief, having overcome the first stage of healthcare trajectory. However, the diagnosis of RD also generates desperation, since absence of knowledge about RD does not allow them to have any idea about future of child, nor which services to seek. Therefore, meanings attributed to health network are unprepared and slow, as it does not overcome urgency of treating RD. The search for treatment of clinical needs brought about by RD does not cover oral healthcare and as caregivers receive the scheduled appointments, it is treated as a surprise. A scheme was constructed to show the theorization of this study, which includes the categories formed, highlights the central category of the theory and is presented in Figure 1.

The central category, which corresponds to the point of correlation of all created categories, is meaning of the insufficiency of public health network to deal with RD. This category assumes centrality in the meanings since public health network was not able to overcome fear and anxiety of mothers, either due to the lack of clear existence and integration of services, or lack of preparation of professionals to deal with children with DR. In an inverse logic, the trajectory of oral health care seems to be a surprise and result of an organized functioning of services, as it appears independently of mothers' search for this care.

DISCUSSION

To theorize about meanings produced by caregivers of children with RD in the trajectory of healthcare, the main results of this study was the meaning of insufficiency attributed by caregivers to public healthcare network, because it does not have neither organization for care, nor trained professionals. This meaning is mainly attributed to sense that healthcare services do not overcome the fear and sense of urgency that mothers feel for treatment of their children. For oral healthcare is given a different meaning, as it appears as a surprise.

The desperation, fear and anguish observed in this study by mothers is lined up to scientific literature^{18,19,20}. After diagnosis, family takes responsibility for disease, generating feelings of guilt, worthlessness, depression and anxiety, especially regarding the idealization of “perfect child”¹⁸. Furthermore, the fact of dealing with a RD child generates emotional vulnerability in the family members²¹. Cultural characteristics can influence way of family relates to RD, and suffering of child can cause significant suffering to other family members¹⁹. Caring for a child with RD can often be demanding, stressful and anxiety-provoking^{22,23}, as more intense care is often required due to rarity and complexity of conditions brought about by disease, thus resulting in an increase of physical and emotional anxiety in these caregivers^{24,25}.

The negative feelings generated after diagnosis can be accentuated due to insufficient guidance and lack of reception from health professionals²⁰. When health professionals are able to welcome individuals efficiently, result is considerable emotional stability and better understanding of disease²⁶. In addition to attitude of health professionals, the organization of healthcare network also proved to be relevant in the context of this study. Late diagnoses, insufficient responses and unmet needs bring insecurity to patients and families²¹. Patients with RD will probably go through different health services until they reach a specialized service²⁷. The lack of clear flows between

points of healthcare networks and absence of knowledge of professionals makes patients feel lost in the health network services¹², which can aggravate the feelings of anguish and suffering of family and patients²⁸. The central point of this study highlights to health professionals adequately welcome patients with RD and to organize necessities of healthcare, whether primary, specialized or even hospital. In this sense, in addition to reception, Primary Healthcare must to develop and monitor therapeutic plan and make appropriate referrals to specialized care⁴. When necessary, homecare should be promoted, aiming to reduce hospital stays, increase autonomy, humanize care, and support caregivers aiming for comprehensive and qualified healthcare⁴.

Some countries have defined strategies to overcome the difficulty in accessing specialized health services that people with RD encounter. Since 2009, European Union members have sought to implement plans and strategies to ensure adequate medical care for patients affected by RD²⁹. In France, territorial networks of university hospitals were created to provide care for RD, made up of health professionals, researchers and patient associations³⁰. In addition to improving the healthcare of patients with RD, these networks contribute to support, training, information, research and innovation³⁰. In Germany, the National Action Plan for People with Rare Diseases, was launched in 2013 to improve the medical treatment situation for patients with RD and support research into health services aimed at these patients³¹. In China, a network of 324 hospitals was set up covering the entire territory of country, forming a national system responsible for resolving problems caused by DR³². These international experiences can contribute to reflection on organization of Brazilian DR healthcare network and overcome meaning of inefficiency currently attributed to health services. In Brazil, national policy to RD was established in 2014 aiming of guaranteeing access to diagnostic means and treatments for RD, in addition to qualifying care for these people¹. However, there are still difficulties

in guaranteeing access to treatment through the Unified Health System, leaving patient associations with access to healthcare and medicines, to detriment of effective implementation of the national policy⁴.

The diagnosis of RD brings urgency for care aimed at overcoming negative feelings that have emerged. However, urgency of healthcare contrasts with the long waiting time, worsening the insecurities of those involved²⁰. The inadequate responses, unresolved care demands and requirements are added with the long waiting time²¹. Although these studies point to the slowness of health services in meeting care demands, the results of this study open the question of whether there is a changed notion of waiting time due to negative feelings and the insufficiency of professionals in welcoming patients and families with RD. In this sense, the proposal disseminated by the International Rare Diseases Research Consortium is that health systems enable the accurate diagnosis of RD, the initiation of healthcare with available therapy within a year, from contact with the healthcare service³⁰. Despite this deadline, health services in Brazil still need to move forward with the feasibility of comprehensive multi-professional and multidisciplinary care in regional reference centers focused on RD, optimizing the time and resources available, in addition to minimizing sequelae and suffering of people with RD and their family members and caregivers²⁶. Although dentist is not part of multidisciplinary team to DR care, it is known that around 15.0% of RD may present oral and maxillofacial manifestations³³. Furthermore, due to clinical and systemic conditions, patients with RD tend to have a higher prevalence of oral diseases when compared to patients without RD³⁴. Physical, mental, behavioral, and sensory changes result in compromising the quality and maintenance of oral health³⁵. Some patients with RD may also present craniofacial and dental changes that interfere skeletal development and can cause chewing and speech disorders, pain, infections and respiratory complications³⁶. Patients with RD may also

have their microbial defenses impaired and reduced motor capacity to perform oral hygiene³⁷. Given this situation, it is known oral healthcare could reduce impact of nutritional disorders, occurrence of microbial infections and inflammations, often related to RD³⁸.

Despite clinical scenario reinforce importance of dental treatment in these patients, children and adolescents with disabilities have less access to dental care compared to general population³⁹. Reduced access of patients with RD to dental care is related to lack of professionals and specialized structures capable of diagnosing, monitoring and treating these patients in a multidisciplinary way²⁸. This data differs from the reality observed in this study, as referral to dental care was executed even before mothers identified their need. However, this difference was a limitation of this study, once the regional hospital service has resident dental surgeons who identify and refer the dental needs of children with special needs to the university service linked to the hospital. Despite being a limitation, the observed scenario reinforces the importance of incorporating the dentist into specialized care teams for people with RD.

This study was done remotely, which means that data collection was guided by information and communication technologies. In this sense, it is possible that some empirical data important for the interpretation of the data were not captured in full, especially those revealing the self of the interviewed caregivers. It is recognized, however, that this adversity did not harm the construction of the theory.

CONCLUSION

The meanings identified in the interviews allowed us to understand that the diagnosis of an unknown disease generates a sense of urgency, which is incompatible with the current organization of health services for RD care and with the attitude of health

professionals. This is because mothers do not find support and clear guidance for the diagnosis and care of their children, which generates more and more anguish and despair.

As implications for practice, the results highlight the importance of preparing health professionals to welcome and coordinate the care of patients with RD, with a view to overcoming mother's suffering. With this same objective, care flows from the thematic RD care network should be more clearly organized and disseminated among health professionals.

REFERENCES

1. Brasil. Ministério da Saúde. Coordenação Geral de Média e Alta Complexidade, Departamento de Atenção Especializada e Temática, Secretaria de Atenção à Saúde, Ministério da Saúde. *Diretrizes para atenção integral às pessoas com DR no Sistema Único de Saúde*. Brasília: Ministério da Saúde; 2014.
2. Aymé S, Kole A, Groft S. Empowerment of patients: lessons from the rare diseases Community. *Lancet*. 2008; 371(9629):2048-51. doi.org/10.1016/S0140-6736(08)60875-2
3. Čagalj D, Buljevac M, Leutar Z. Being a Mother of a Child with Prader-Willi Syndrome: Experiences of Accessing and Using Formal Support in Croatia. *Scand J Disabil Res*. 2018; 20(1):228-237. doi.org/10.16993/sjdr.13
4. Lima MAFD, Gilbert ACB, Horovitz DDG. Redes de tratamento e as associações de pacientes com doenças raras. *Cien Saude Colet* 2018; 23:(32):47-56. doi.org/10.1590/1413-812320182310.14762018
5. Šimić G. Rare diseases and omics-driven personalized medicine. *Croat Med J*. 2019; 60(6):485-487. doi.org/10.3325/cmj.2019.60.485
6. Tabor HK, Goldenberg A. What precision medicine can learn from rare genetic disease research and translation. *AMA J Ethics*. 2018; 20(9):e834-440. doi.org/10.1001/amajethics.2018.834

7. Dharssi S, Wong-Rieger D, Harold M, Terry S. Review of 11 national policies for rare diseases in the context of key patient needs. *Orphanet J Rare Dis*. 2017;12(1):63. doi.org/10.1186/s13023-017-0618-0
8. Abdon Barbosa A, de Oliveira MR, Martins R, Grumach AS. Assessment on hereditary angioedema burden of illness in Brazil: a patient perspective. *Allergy Asthma Proc*. 2019; 40(3):193–197. doi.org/10.2500/aap.2019.40.4207
9. Blumer H. *Symbolic Interactionism: Perspectives and Method*. University of California Press, Berkeley; 1969.
10. Luboya E, Tshilonda JCB, Ekila MB, Aloni MN. Psychosocial impact of sickle cell disease in the parents of children living in Kinshasa, Democratic Republic of Congo: a qualitative study. *Pan Afr Med J*. 2014; 19(5):28-30.
11. Santos Luz G dos, Silva MRS da, DeMontigny F. Priority needs referred by families of rare disease patients. *Texto contexto - enferm*. 2016;25(4):e0590015. doi.org/10.1590/0104-07072016000590015
12. Aureliano WA. Trajetórias terapêuticas familiares: Doenças raras hereditárias como sofrimento de longa duração. *Cien Saude Colet*. 2018; 23(2):369-79. doi.org/10.1590/1413-81232018232.21832017
13. Vicente JB, Sanguino GZ, Riccioppo MRPL, Santos MR, Furtado MCC. Syphilis in pregnancy and congenital syphilis: women's experiences from the perspective of symbolic interactionism. *Rev Bras Enferm*. 2023;76(1):e20220210. doi.org/10.1590/0034-7167-2022-0210p
14. Fontanella BJB, Luchesi BM, Saidel MGB, Ricas J, Turato ER, Melo DG. Amostragem em pesquisas qualitativas: proposta de procedimentos para constatar saturação teórica. *Cad Saude Publica*. 2011; 27(2):389-394.
15. Charmaz K. *Constructing Grounded Theory*. 2.ed. Los Angeles: Sage, 2014.
16. Santos JLG, Cunha KS, Adamy EK, Backes MTS, Leite JL, Sousa FGM. Data analysis: comparison between the different methodological perspectives of the Grounded Theory. *Rev Esc Enferm USP*. 2018;52:e03303. doi.org/10.1590/S1980-220X2017021803303
17. Oliveira APB, Schimidt BD, Santos CJ, Amatneeks TM, Cavallet LHR, Michel RB. Quality of life patients on hemodialysis and the relationship with mortality, hospitalizations and poor treatment adherence. *J Bras Nefrol*. 2016; 38(4):411-420. doi.org/10.5935/0101-2800.20160066

18. Xavier DM, Gomes GC, Cesar-Vaz MR. Meanings assigned by families about children's chronic disease diagnosis. *Rev Bras Enferm.* 2020; 73(2):e20180742.
19. Felipe RNR, Rosaneli CF, Cunha TR, Munhoz CJM. Fatores de vulnerabilidades em itinerários terapêuticos de doenças raras: uma revisão integrativa. *Revista Saúde e Desenvolvimento Humano.* 2020; 8(3):1-13.
20. Brotto A, Rosaneli C. Vulnerabilidades de cuidadores familiares de pacientes com doenças raras: uma revisão integrativa. *Psicologia, Saúde & Doenças.* 2021; 22(2):659-673.
21. Barbosa RL, Portugal S. O Associativismo faz bem à saúde? O caso das doenças raras. *Cien Saude Colet* 2018; 23(2):417-430. doi.org/q0.1590/1413-81232018232.24032017
22. Cousino MK, Hazen RA. Parenting stress among caregivers of children with chronic illness: a systematic review. *J Pediatr Psychol.* 2013;38(8):809–28. doi.org/10.1093/jpepsy/jst049
23. Pinquart M. Parenting stress in caregivers of children with chronic physical condition—a meta-analysis. *Stress Health J Int Soc Investig Stress.* 2017; 34(2):197-207. doi.org/ 10.1002/smi. 2780
24. Pelentsov LJ, Fielder AL, Laws TA, Esterman AJ. The supportive care needs of parents with a child with a rare disease: results of an online survey. *BMC Fam Pract.* 2016;17:88. doi.org/10.1186/s12875-016-0488-x
25. Zurynski Y, Deverell M, Dalkeith T, Johnson S, Christodoulou J, Leonard H, Elliott EJ; APSU Rare Diseases Impacts on Families Study group. Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. *Orphanet J Rare Dis.* 2017;12(1):68. doi.org/10.1186/s13023-017-0622-4
26. 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. *Cien Saude Colet* 2019; 24(10):3637–50. doi.org/10.1590/1413-812320182410.01612019
27. Silva SF. The organization of regional and integrated healthcare delivery systems: challenges facing Brazil's Unified Health System. *Cien Saude Colet,* 2011; 16(6):2753-2762. doi.org/10.1590/S1413-81232011000600014

28. Baldovino S, Moliner AM, Taruscio D, Daina E, Roccatello D. Rare Diseases in Europe: from a Wide to a Local Perspective. *Isr Med Assoc J.* 2016;18(6):359-63.
29. Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on Orphan Medical Products.
30. Austin CP, Cutillo CM, Lau LPL, Jonker AH, Rath A, Julkowska D, Thomson D, Terry SF, de Montleau B, Ardigò D, Hivert V, Boycott KM, Baynam G, Kaufmann P, Taruscio D, Lochmüller H, Suematsu M, Incerti C, Draghia-Akli R, Norstedt I, Wang L, Dawkins HJS; International Rare Diseases Research Consortium (IRDIRC). Future of Rare Diseases Research 2017-2027: An IRDiRC Perspective. *Clin Transl Sci.* 2018; 11(1):21-27. doi.org/10.1111/cts.12500
31. National Action League for People with Rare Diseases. National Plan of Action for People with Rare Diseases: Action Fields, Recommendations, Proposed Actions. German Federal Ministry of Health, German Federal Ministry of Education and Research, Alliance for Chronic Rare Diseases, 2013.
32. Ren Q, Wang J. Network established to collaborate on diagnosis and treatment of rare diseases in China: a strategic alliance backed by tiered healthcare is the key to the future. *Intractable Rare Dis Res.* 2019; 8(1):78–79. doi: 10.5582/irdr.2019.01024
33. Kühne A, Kleinheinz J, Jackowski J, Köppe J, Hanisch M. Study to investigate the knowledge of rare diseases among dentists, orthodontists, periodontists, oral surgeons and craniomaxillofacial surgeons. *Int J Environ Res Public Health.* 2020;18(1):139-53. doi.org/10.3390/ijerph18010139
34. Prado HV, Carneiro NCR, Perazzo MF, de Abreu MHNG, Martins CC, Borges-Oliveira AC. Assessing a possible vulnerability to dental caries in individuals with rare genetic diseases that affect the skeletal development. *Orphanet J Rare Dis.* 2019; 14(1):145-152. doi.org/10.1186/s13023-019-1114-5
35. Hanisch M, Hanisch L, Benz K, Kleinheinz J, Jackowski J. Development of a database to record orofacial manifestations in patients with rare diseases: a status report from the ROMSE (recording of orofacial manifestations in people with rare diseases) database. *Br J Oral Maxillofac Surg.* 2017; 55(5):500-503. doi.org/10.1016/j.bjoms.2017.02.003
36. Debossan SAT, Deps TD, Prado HV, de Abreu MHNG, Borges-Oliveira AC. Access to oral health care services for individuals with rare genetic diseases

- affecting skeletal development. *Spec Care Dentist*. 2022; 42(1):32-40. doi.org.10.1111/scd.12639
37. Aslan BI, Gülşen A, Tirank ŞB, Findikçioğlu K, Uzuner FD, Tutar H, Üçüncü N. Family Functions and Life Quality of Parents of Children With Cleft Lip and Palate. *J Craniofac Surg*. 2018; 29(6):1614-1618. doi.org/10.1097/SCS.00000000000004611
38. Friedlander L, Berdal A, Boizeau P, Licht BA, Manière MC, Picard A, et al. Oral health related quality of life of children and adolescents affected by rare orofacial diseases: a questionnaire-based cohort study. *Orphanet J Rare Dis*. 2019; 14(1):124–4. doi.org10.1186/s13023-019-1109-2
39. Ruy Carneiro NC, Duda Dets T, Campos França E, Ribeiro Valadares E, Almeida Pordeus I, Borges-Oliveira AC. Oral health of children and adolescents with mucopolysaccharidosis and mother's Sense of Coherence. *Spec Care Dentist*. 2017; 37(5):223-229. doi.org/10.1111/scd.12238

TABLES

Table 1. Information about the caregiver and the child with a RD

Caregiver Code	Degree of kinship	Child's age	Child's gender	Diagnosis of the RD
M1	mother	8	M	Fanconi syndrome
M2	mother	5	F	Turner syndrome
M3	mother	1	F	Goldenhar syndrome
M4	mother	8	M	Osteogenesis Imperfecta
M5	mother	6	F	DiGeorge syndrome
M6	mother	4	F	Turner syndrome
M7	mother	7	F	Interstitial deletion in the long arm of chromosomes 6
M8	mother	6	F	Osteogenesis imperfecta

Table 2. Exemplary excerpts and categories formed from the categorization of interviews carried out with mothers of children with RD.

Categories	Codes	Exemplary excerpts
<i>Relief and despair at the diagnosis</i>	Despair offered by the diagnosis	Everyone dreams of having a healthy child and if the doctor says it won't happen, our ground collapses. (M6) So, for me, when we found out it was like that, you know, I lost my ground. (M1)
	Relief at finding the answers	Despair of not knowing the syndrome, of not knowing what the consequences will be in her life. And, I think there was also a bit of relief knowing that those episodes of pneumonia, of other episodes that we had in the hospital, had a reason. (M5)
		When the doctor made the diagnosis we were aware, he said: 'she has little malformation and she doesn't have the most extreme malformation, hers can be corrected'. So her [disease] is calmer, thank God! (M3)
<i>Context of doubts and fears</i>	Fear of prejudice	[...] of course I'm worried, because when she's older she has to go to school, socialize with other children. I'm afraid of prejudice and things like that (M3)
	Uncertainties about the future	So I think it's because of the unknown. Because she didn't know what would happen in her life in the future, right? What problems could she have, right? I think all of this makes us very desperate. (M5)

		We are afraid of not being able to handle it. I thought, I think a lot about her future. I think when there is no me, nor my mother, who will take care of her? Because it's just us. So I'm afraid of it. (M8)
	Anguish about the prognosis	Then you were in that anguish, you know? I was distressed thinking: "What if my daughter gets worse in a month?"; "Can't she get better in a month?" (M4). [...] So it seemed like I was alone, I was lost. I was afraid of not knowing how to deal with the syndrome, afraid of losing my daughter. I was afraid of everything. (M2)
<i>Distress in waiting time for care</i>	Distress altering the notion of time for care	And if care is delayed, we lose hope [...] because I feel distressed. I know there are a lot of people to help, but it's hard to stay at home waiting for days without an answer (M2) I say it depends a little on your point of view, but as a mother, I think it took a while because it took an average of six months to start the treatment. [...] So, I say that it took a long time, right, but by telling you the step-by-step process we realize that it didn't take that long, right? (M1)
	Facing delays in service	Medical appointments take time. Because we looked for assistance, we went after it, we took the bus, we spent on driving, we spent whatever, but then we got there and there was no timetable to call you, or it would take 15-20 days. Or a month. (M2) So, the most complicated thing is the public system. Because everything takes time. [...] Waiting is slower, even if you run, and sometimes there's no way you can run any further because there's no way to keep help. [...] If I were a still, calm mother, I wouldn't close her diagnosis to this day. (M3)
<i>Unsatisfactory healthcare network</i>	Absence of professional support	For me, I found their preparation very lacking, you know? [...] There [in the specialized care unit] the information he gave me was what made me afraid. I found it to be a very difficult moment at that time [...], but after I went to the clinical hospital, where they guided me, they explained everything to me clearly [...] then it hit me, it hit me. (M6)
	Ignorance of diseases	I don't think even the geneticist knew this syndrome well, she had to do some research to tell me better about the syndrome. (M5)

		[At the hospital] there was a nurse who had never seen the disease, there was a doctor who had never seen it. It could be a teaching hospital for those who are interested in learning about this RD, but it has to be a hospital that already directs the mother (M3)
<i>Integrated dental care</i>	Surprise with dental care	At one of the appointments, they made an appointment and told me that she would receive dental care in a specialized department. [...] I was scared because I wasn't expecting to get specialized care, especially for free. (M5)
		So, he actually scheduled it for her without me knowing anything! I think they took her data when she went to a consultation (M3).
	Integration of services	Because if we hadn't been providing care there [at the clinical hospital] she wouldn't have gotten the position. I wouldn't even know about the dentist who treats special needs children, I would continue taking them privately and spending a lot of money. (M2)
		The doctors at the hospital who referred her. And so, I didn't even know. (M6)

FIGURE

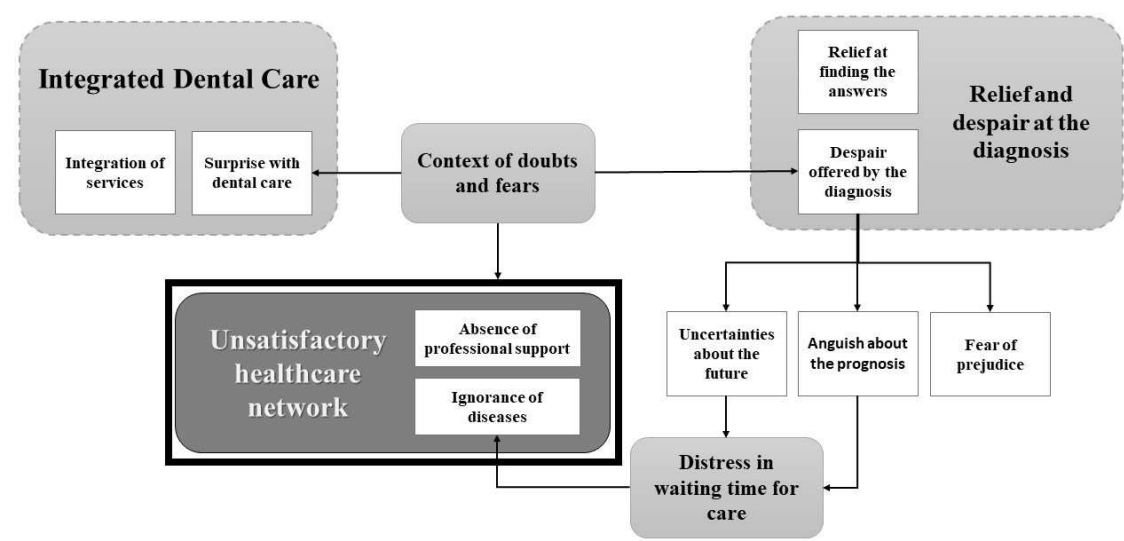


Figure 1. The dissatisfactions experienced by mothers of children with RD regarding public health system - Schematic model of the relationship between the categories identified in the study and which bring the meanings produced by mothers in the trajectory of caring for children with rare diseases.

REFERÊNCIAS*

AMARAL, Marise Basso; REGO, Sergio. Doenças raras na agenda da inovação em saúde: avanços e desafios na fibrose cística. **Cadernos de Saúde Pública**, v. 36, n. 12, dez. 2020.

AURELIANO, Waleska de Araújo. Trajetórias Terapêuticas Familiares: doenças raras hereditárias como sofrimento de longa duração. **Ciência & Saúde Coletiva**, v. 23, n. 2, p. 369–380, fev. 2018.

BARBOSA, Rogério Lima; PORTUGAL, Silvia. O Associativismo faz bem à saúde? O caso das doenças raras. **Ciência & Saúde Coletiva**, v. 23, n. 2, p. 417–430, fev. 2018.

BOETTCHER, Johannes *et al.* Being the Pillar for Children with Rare Diseases—A Systematic Review on Parental Quality of Life. **Internacional Journal of Environmental Research and Public Health**, v. 18. N. 9, p. 4993, mai. 2021.

BRASIL. Diretrizes para atenção integral às pessoas com Doenças Raras no Sistema Único de Saúde. Ministério da Saúde. Coordenação Geral de Média e Alta Complexidade, Departamento de Atenção Especializada e Temática, Secretaria de Atenção à Saúde, Ministério da Saúde. Brasília: Ministério da Saúde; 2014.

BROTTO, Aline; ROSANELI, Caroline. Vulnerabilidades de cuidadores familiares de pacientes com doenças raras: uma revisão integrativa. **Psicologia, Saúde & Doenças**, v. 22, n. 2, p. 659-673, mai. 2021.

CHUNG, Claudia Ching Yan *et al.* Rare disease emerging as a global public health priority. **Frontiers in Public Health**, v. 10, out. 2022.

FELIPE, Raquiel Naiele Ramos *et al.* Fatores de vulnerabilidades em itinerários terapêuticos de doenças raras: uma revisão integrativa. **Revista Saúde e Desenvolvimento Humano**, v. 8, n. 3, p. 1-13, jul. 2020.

FOSTER, Mandie *et al.* Parents' experiences of children with a rare disease attending a mainstream school: Australia. **Journal of Pediatric Nursing**, v. 63, p. e50-e57, mar. 2022.

GIMENES, Heloisa Turcatto; ZANETTI, Maria Lúcia; HAAS, Vanderlei José. Fatores relacionados à adesão do paciente diabético à terapêutica medicamentosa. **Revista Latino-americana de Enfermagem**, v. 17, n. 1, jan. 2009.

GOPAL-SRIVASTAVA, Rashmi; KAUFMANN, Petra. Facilitating clinical studies in rare diseases. **Advances in Experimental Medicine and Biology**, v. 1031, p. 125–40, 2017.

IRIART, Jorge Alberto Bernstein *et al.* Da busca pelo diagnóstico às incertezas do tratamento: desafios do cuidado para as doenças genéticas raras no Brasil. **Ciência & Saúde Coletiva**, v. 24, n. 10, p. 3637–3650, out. 2019.

LOPES, Marcos Thomazin *et al.* Difficulties in the diagnosis and treatment of rare diseases according to the perceptions of patients, relatives and health care professionals. **Clinics**, v. 73, abr. 2018.

OLIVEIRA, Araiê Prado Berger *et al.* Quality of life in hemodialysis patients and the relationship with mortality, hospitalizations and poor treatment adherence. **Brazilian Journal of Nephrology**, v. 38, n. 4, p. 411–420, out. 2016.

SOARES, Juliana de Lima; ARAÚJO, Laura Filomena Santos de; BELLATO, Roseney. Cuidar na situação de adoecimento raro: vivência da família e sua busca por amparo dos serviços de saúde. **Saúde e Sociedade**, v. 25, n. 4, p. 1017–1030, out. 2016.

WETTERAUER, Birgit; SCHUSTER, R. Rare diseases. Funding programs in Germany and Europe. **Bundesgesundheitsblatt Gesundheitsforschung Gesundheitsschutz**, v. 51, n. 5, p. 519-528, mai. 2008.

WITT, Stefanie *et al.* Rare pediatric diseases and pathways to psychosocial care: a qualitative interview study with professional experts working with affected families in Germany. **Orphanet journal of rare diseases**, v. 16, n. 1, p. 497, nov. 2021.

ZURYNSKI, Yvonne *et al.* Rare childhood diseases: How should we respond? **Archives of Disease in Childhood**, v. 93, n. 12, p. 1071-1074, dez. 2008.