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"Unfortunately, we couldn't reach a definitive diagnosis": the interactional management of uncertainty in genetic counseling

"Infelizmente, não conseguimos chegar a um diagnóstico definitivo": a gestão interacional da incerteza no aconselhamento genético

"Lamentablemente no pudimos llegar a un diagnóstico definitivo": el manejo interaccional de la incertidumbre en el consejo genético

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ABSTRACT

This paper examines how participants in genetic counseling sessions interactionally manage situations where the results of tests to investigate the causes of identified fetal malformations are inconclusive or missing. The dataset consists of 54 audio-recorded interactions at a unit specialized in moderate- and high-risk pregnancies at a Brazilian public hospital. Conversation analysis was used to examine the data, revealing that the participants deployed interactional actions that exhibited highly negative valence toward diagnostic inconclusiveness, demonstrating that when there is a motivation for a medical examination, insofar as its results will serve as a basis for subsequent decision-making (in this case about future pregnancies), there is a preference for bad diagnostic news over absent or inconclusive diagnostic news. These findings are consistent with prior interactional studies.

Keywords: Uncertainty; Medical diagnosis; Doctor-patient interaction; Conversation Analysis; Genetic counseling.

RESUMO

Este artigo examina como os participantes em sessões de aconselhamento genético gerenciam interacionalmente resultados de testes genéticos inconclusivos ou ausentes – testes para investigar as causas das malformações fetais identificadas. O conjunto de dados consiste em 54 interações gravadas em áudio em uma unidade de gestação de médio e alto risco de um hospital público brasileiro. A abordagem da Análise da Conversa utilizada para examinar os dados revela que os participantes desenvolvem ações interacionais que exibem uma orientação de valência altamente negativa em relação à inconclusividade diagnóstica, demonstrando que quando há motivação para um exame médico, ou seja, usando o resultado do teste diagnóstico como base para tomada de decisão, tal como acontece com futuras gestações, haja uma preferência por más notícias diagnósticas em detrimento de notícias diagnósticas ausentes ou inconclusivas. Tais resultados são consistentes com estudos interacionais anteriores.

Palavras-chave: Incerteza; Diagnóstico médico; Interação médico-paciente; Análise de Conversa; Aconselhamento genético.

RESUMEN

Este artículo examina cómo los participantes en las sesiones de asesoramiento genético gestionan de forma interactiva los resultados de pruebas genéticas no concluyentes o faltantes, pruebas para investigar las causas de las malformaciones fetales identificadas. El conjunto de datos consta de 54 interacciones grabadas en audio en una unidad de embarazo de riesgo medio y alto de un hospital público brasileño. El enfoque de Análisis de Conversación utilizado para examinar los datos revela que los participantes desarrollan acciones interaccionales que exhiben una orientación de valencia altamente negativa hacia la falta de conclusión del diagnóstico, lo que demuestra que cuando hay motivación para un examen médico, es decir, utilizar el diagnóstico del resultado de la prueba como base para la toma de decisiones, como ocurre con futuros embarazos, se prefieran las malas noticias diagnósticas a las noticias diagnósticas ausentes o no concluyentes. Estos resultados son consistentes con estudios interaccionales previos.

Palabras clave: Incertidumbre; Diagnostico medico; Interacción médico-paciente; Análisis de conversación; Asesoramiento genetico.

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INTRODUCTION

Despite the extensive scientific, technological, and clinical advances witnessed in contemporary times, uncertainty is still an inherent part of medicine (Fox, 2000). This fact has prompted several empirical studies to examine how professionals and patients deal with uncertainty in medical interactions, especially in the context of diagnosis (Arribas-Ayllon; Sarangi, 2014; Brookes-Howell, 2006; Maynard; Frankel 2003; 2006; Pilnick; Zayts, 2014; Stivers; Timmermans, 2016; Timmermans; Buchbinder, 2010; Zayts; Sarangi, 2013; Zayts; Sarangi; Schnurr, 2016; Zuczkowski *et al.*, 2014). Despite this, studies of real-time recorded and transcribed naturalistic interactions about uncertainty in genetic counseling—the medical specialty examined in this article—remain scarce (Arribas-Ayllon; Sarangi, 2014; Stivers; Timmermans, 2016; Zayts; Sarangi, 2013; Zayts; Sarangi; Schnurr, 2016).

This study, conducted as part of a larger project that investigates the communication across different specialties of diagnostic news concerning moderate- and high-risk pregnancies at a public hospital in Brazil (Ostermann, 2013), analyzes counseling sessions scheduled to discuss fetal genetic test results, focusing on those cases where the diagnosis is inconclusive. The study shows how uncertainty is interactionally materialized in these interactions, how it is delivered and received, and what consequences it has for the unfolding consultation.

Delivery of diagnosis and treatment recommendations as an expected follow-up to screening in medical interactions

Jeffrey Robinson (2003, p. 30) proposes a schematic representation of the large-scale structure of social interaction that organizes physician–patient interactions during acute visits in which new medical problems are presented (Figure 1). The primary goals of a medical encounter of this nature are to reach a diagnosis and to recommend some treatment or conduct. Heath (1992, p. 238) also suggest that diagnosis marks the completion of a physician's practical inquiry into a patient's complaint and stands as both the reason for the consultation and the basis for the next step, which consists of recommending some treatment or other type of follow-up behavior. Doctors and patients seem to orient to the absence of such activities as a kind of "deviation" from a normative structure of activities. In other words, once a medical investigation is initiated in response to the existence of symptoms or a suspected medical problem, there is an expectation that this will lead to a diagnosis, based upon which some treatment or other type of conduct will be recommended.

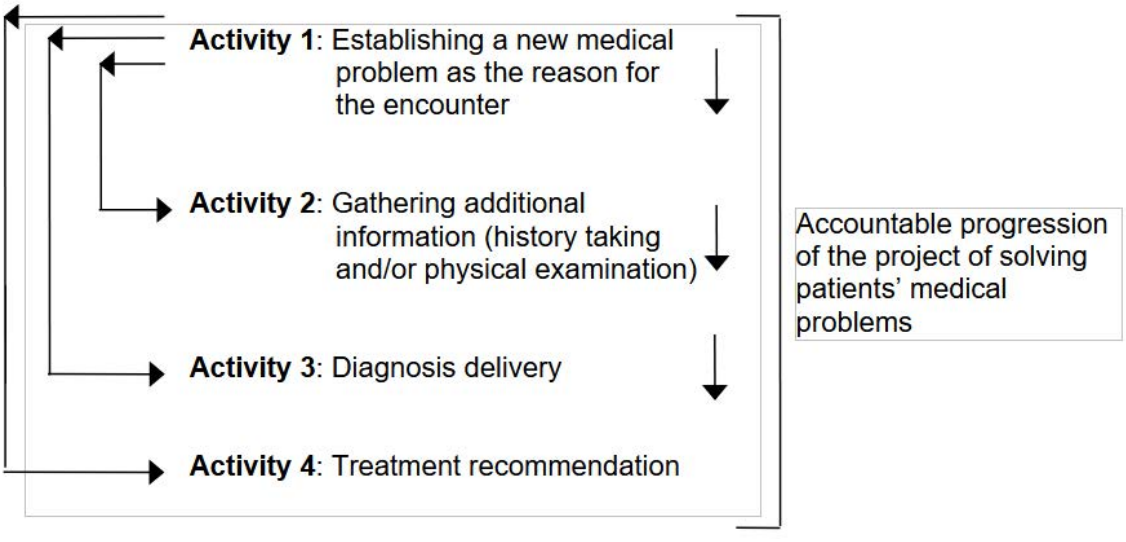

Figure 1 – Structural schema for the project of solving patients' new medical problems
Source: Robinson (2003, p. 30).

In the context of the present study, this normative expectation of a diagnosis for a presented medical problem enables such interactions to be analyzed using conversation analysis, drawing specifically on the concept of conditional relevance, in which a request for information is the first part of an adjacency pair (Sacks, 1992). In other words, once a request for information is made, some provision of information—an answer—is normatively expected. Conversation analysis also distinguishes between providing an actual "answer" to a request for information and providing a "non-answer" response, such as "I don't know." An answer effectively closes the adjacency pair by producing the action made relevant by the first-pair part, while a non-answer response fills in the interactional slot generated by the first-pair part in the form of a claim of inability to produce the requested responsive action; i.e., to provide the information (Lee, 2013; Stivers, 2010; Stivers; Robinson, 2006).

Although a search for a diagnosis cannot be seen as the first part of an adjacency pair, the expectation of a diagnosis, once a diagnostic investigation is initiated, is revealed to be pervasive and, thus, normative in the dataset from this study. As we shall demonstrate in the analysis, while health professionals engage in extensive interactional work to account for the absence of a conclusive diagnosis, patients insist on its pursuit.

Genetic counseling

Interactions in genetic counseling are known to be largely patient-centered, with a focus on patient autonomy, especially in regard to decision-making (Sarangi, 2010). A necessary precondition for achieving autonomy and thus for making an informed decision is to be sufficiently informed (Horovitz *et al.*, 2012; Kukla, 2007; Rapp, 1999). When a diagnosis cannot be obtained, the goal of autonomous, informed decision-making about future actions is rendered moot.

In genetics, the inability to obtain an accurate diagnosis is a recurring problem (Brookes-Howell, 2006), mostly because of the limitations of the state-of-the-art in the field as a whole (Sarangi; Clarke, 2002). In fetal genetics, the cause of about 50% of all malformations is unknown, and physicians are often faced with the difficult task of admitting that they "don't know" (Stevenson, 2005, p. 5).

The central role of diagnosis as a follow-up to a medical examination, as argued by Heath (1992) and Robinson (2003), is also observed in genetic counseling encounters. Brookes-Howell's (2006) analysis of 18 encounters in the United Kingdom reveals this importance. Drawing on Silverman's (1997) notions of "clinical reality" and "everyday reality," Brookes-Howell observes how the participants in genetic counseling interactions emphasize the importance of having a diagnosis (i) as a "proper explanation" to be used in a clinical sense; i.e., as a prerequisite for appropriate clinical intervention, risk management, knowledge of a prognosis, etc., which fits into the notion of clinical reality; and (ii) as a kind of "social resource" to be used in an everyday sense; i.e., as an explanation or label to be provided for others, which is related to the notion of everyday reality.

By examining the interactional management of genetic counseling consultations after genetic testing fails to provide a conclusive diagnosis, the present paper focuses on the importance of diagnosis for both clinical and everyday realities.

Diagnostic uncertainty as informed by interactional studies

Interactional studies (Maynard, 2003; Maynard; Frankel, 2006; Timmermans; Buchbinder, 2010) have observed that "the certainty of a genetic diagnosis can provide a closure to a diagnostic odyssey" (Stivers; Timmermans, 2016, p. 199–200). The metaphor of an odyssey well describes the long rounds of laboratory tests and clinical examinations that are often undertaken to arrive at a diagnosis of genetic conditions, as well as some other health conditions. In addition to the relief of completing a long investigation, the reach of a diagnosis orients patients towards reorganizing their world, knowing what to expect, and planning what to do next (Maynard, 2003).

In medicals contexts, reorganizing one's world includes discussing treatment options and follow-up conduct (Robinson, 2003). In the case of genetic disorders diagnosed in fetuses, this involves discussing

immediate therapeutic measures, planning the appropriate timing and type of delivery, understanding the child's prognosis, and estimating the risk of recurrence of the same malformation when planning future pregnancies. Thus, having a diagnosis for a particular condition makes it more possible to take further decisions or actions, drawing on the new diagnostic knowledge (Kukla, 2007; Rapp, 1999).

Previous studies of medical interactions involving diagnostic uncertainty show that patients tend to become more active in the pursuit of some kind of diagnostic "answer," while physicians tend to provide more accounts and/or even tentative diagnoses (Stivers; Timmermans, 2016). For example, in their analysis of how uncertainty is interactionally materialized in breast cancer follow-up appointments, Souza and Ostermann (2017) show that participants are oriented towards the pursuit of positive evaluations, such as "alright," at different stages of the encounters.

Analyzing the delivery and reception of diagnoses in general practice consultations in Britain, Heath (1992) suggests that a physician's authority as an expert is ratified, among other things, by how a patient receives their diagnosis. While the delivery of a diagnosis is followed by minimal acknowledgment tokens or even silence, which indicate passivity on the patient's part, the delivery of an uncertain diagnosis is followed by extended responses by the patients. Peräkylä (2006) shows similar phenomena in his analysis of Finnish primary care consultations, claiming that in situations of diagnostic uncertainty, professionals tend to provide accounts and evidence of what has prevented them reaching a conclusive diagnosis.

As Stivers and Timmermans (2016) show in their interactional analysis of genetic counseling encounters, geneticists demonstrate their expertise by actively reinterpreting ambiguous diagnoses in pursuit of a more assertive interpretation. In this process, they may invite the parents to collaborate in providing an explanation for the symptoms, even if the problem is untreatable, which can function as a comforting way of dealing with such uncertainty.

BACKGROUND TO THE CONTEXT INVESTIGATED IN THIS STUDY

The larger project that gave rise to this study (Ostermann, 2013) investigates how diagnostic news is delivered in the context of fetal medicine. The way genetic counseling is given in the context studied here reflects the structural organization at the study site, which is quite different from the kind of interactional context described by Robinson (2003) (Figure 1). In the current data, the delivery of diagnostic news is not just a stage in an encounter, but is its main purpose, as the consultations are scheduled specifically to present genetic test results and their implications for future action. As shown in the analytic session, this study demonstrates how the absence of conclusive diagnostic news becomes a burden for the participants.

Extract 1 shows two passages from our corpus in which a pregnant woman expresses her concerns about the length of the fetal femur.

Extract 1[1]: "worst thing" - HMF_ACONGEN_vanusa_JEFFERSON_08_04_14

porque a gente saindo com dúvida é a pi↑or coisa que te:m né.
because leaving with doubts is the worst thing there is right

(...)

é bom a gente sabê ↑sempre o que que tá acontecendo
it's good for us to always know what's happening
me:smo que seja o pior daí tu já vai te prepara:ndo.
even when it's the worst then you already start preparing yourself

[1] For the purposes of this article, two-line transcriptions are used, with the original interactions in Brazilian Portuguese on line 1 and their translations into English on line 2.

The patient explicitly displays what Robinson (2003) finds in his study: that doctors and patients treat the absence of a conclusive diagnosis and of treatment options as deviations from a "normal" structure of activities in medical encounters, because this absence has consequences for the patient's life. For this patient, *any* diagnostic news is better than none.

In fetal genetic testing, the absence of a definitive diagnosis for a malformation identified in a fetus creates difficulties with regards to: (i) planning treatment; (ii) planning care for a newborn with a malformation; (iii) estimating the risk of the same malformation recurring in future pregnancies; and, therefore, (iv) making decisions about future pregnancies. Given the nature of such difficulties, communicating to parents that a genetic test has failed to yield a definitive diagnosis becomes a "real-world problem in which language is a central issue" (Brumfit, 1995, p. 27), making such an event of prime investigative interest from a language-in-use and language-in-action perspectives, which are central to conversation analysis.

DATA AND PARTICIPANTS

The interactions examined in this study come from a larger dataset consisting of 54 genetic counseling encounters (Ostermann, 2013). In eight of these (3 hours 30 min of recordings) no definitive genetic diagnosis was provided. Each of these encounters was with different patients but the same geneticist.

The patients were approached individually in the waiting room on the day of their genetic counseling appointment and informed about the research. Ninety-seven percent gave their full and informed consent to participate in the study; only these patients had their consultations recorded.

The pregnant women who attend the genetic counseling consultations at the study site are referred from another institution because some type of abnormality has been identified in their fetus (or with the pregnancy itself) during regular prenatal care. These women, who receive prenatal care at a unit specializing in moderate- and high-risk pregnancies, have obstetric consultations and fetal ultrasound scans before scheduling an appointment with the geneticist. In the genetic counseling session, they are offered the opportunity to undergo further screening to investigate the possibility that the fetal problem is related to some kind of genetic syndrome.

The main genetic test offered in these consultations is the fetal karyotype test, which can reveal several genetic syndromes. If the woman decides to undergo this test, a follow-up genetic counseling visit is scheduled after the procedure to discuss the test results. This study focuses specifically on these follow-up sessions.

In order to analyze how uncertainty emerges and the interactional shape it takes in this setting, all the interactions were transcribed using the conventions proposed by Jefferson (1984) (see Table 1). Field notes were taken in addition to the recordings and transcripts. Conversation analysis (Sacks; Schegloff; Jefferson, 1974) was used to analyze the data.

Table 1 – Transcription conventions

(1.8)	Timed pause
(.)	Micropause
=	Latched speech
[text]	Overlap
,	Continuing intonation
↑text	Rising pitch
↓text	Falling pitch
.	Pitch falling at end of intonation phrase
?	Pitch rising at end of intonation phrase
-	Cut-off
:::	Prolonged sound
>text<	Increased speaking rate
<text>	Decreased speaking rate
TEXT	Louder speech
°text°	Quieter speech
°°text°°	Much quieter speech
text	Stress or emphasis
(text)	Dubious transcription
((Text.))	Comments by transcriber
.hh	Inbreath
hh	Exhalation

Source: adapted from Jefferson, 1984.

ANALYSIS

This section is divided into three main parts, which represent the central findings of this study regarding the interactional practices the participants employ to deal with a lack of definitive diagnosis in this context: the physician's orientation to the dispreference of an absent diagnosis; the physician's and patients' orientation to the expectation of a diagnosis; and the patients' agency.

Although such interactional practices typically occur in sequences involving diagnostic uncertainty, the extracts discussed in this paper are all from the same encounter, as this enables a more cohesive presentation of the analytical reasoning. In this case, the patient was a woman who had a stillbirth at 35 weeks of gestation. Several physical and neurological malformations had been identified in the fetus during the pregnancy and the woman had been informed by the geneticist about the possibility of fetal death. The interaction analyzed here was to deliver the result of the fetal autopsy.

Doctor's orientation to a dispreference for an absent definitive diagnosis: provision of accounts

Until the segment shown in Extract 2, the doctor had only informed the woman of the autopsy findings: gestational age at the time of fetal death, hydrocephaly, and a state of advanced autolysis; i.e., the fetus was already materially decomposing.

Extract 2: HMF_ACONGEN_claudia_JEFFERSON_26_11_13

GEN = Geneticist; PAT = Patient

```
223  GEN:  en[tão ]=
           so
224  PAT:    [°ãrrã°]
              uh-huh

225  GEN:          =é:, (.) .hhh (0.6) o ↑que que acontece
                    um                  what happens
226       né:, .h >até-< pela essa autólise
          you see also because of that autolysis
227       que foi vista isso acaba <atrapalhando>
          that was seen this ends up hampering
228       a adequada análise, (0.4) a avaliação dos
          an adequate analysis      an evaluation of the
229       órgãos do bebê
          baby's organs

230       (.)

231  GEN: .h porque com a- com a decomposição e a
             because with the with the decomposition and the
232       reabsorção isso vai com o tempo:: (.)
          reabsorption this ends up as time goes by
233       .h ã: (.) vai per↑dendo aquele formato da
             u:m    it ends up losing that format of the
234       estrutura usual né, e tu perde o parâmetro
          usual structure, you see, and you lose the parameter
235       do ↑que realmente, como é que tinha se
          of what really,    how that organ had
236       formado aquele órgão né,=
          formed, you see

237  PAT: =ãrrã,=
           uh-huh
```

In lines 225–29, the doctor identifies autolysis as the reason why an adequate analysis could not be made, and in lines 231–37 he explains why the decomposition of the fetus impeded the examination of the material. The patient lets the physician complete the explanation by not taking a turn at the transition-relevance place (l. 230). Up to this point, however, the doctor has not yet disclosed that there is no final diagnosis to be delivered. Instead, he has only explained that the autolysis process "hampered" the analysis, thus justifying the news he is yet to disclose, namely, that no conclusive diagnosis has been reached. This comes next in the sequence.

Extract 3: HMF_ACONGEN_claudia_JEFFERSON_26_11_13

```
249  GEN: então:, (0.6) que que acontece, (0.4) ã: a gente,
          so:           what happens here       um we
250       infelizmente não conseguiu chegá num diagnóstico
          unfortunately couldn't reach a definitive
251       definitivo né do que que realmente aconteceu.
          diagnosis you see for what really happened.
252       (.)
```

In lines 249–51, in so many words, the doctor rules out the possibility of providing a definitive diagnosis. At this point, it becomes clear that the explanations he gave in Extract 2 were in preparation for what was to come, i.e., dealing preemptively with the dispreference for the fact that there is no diagnosis to be communicated in a consultation specifically scheduled for that purpose. This is a case of an account being given to justify an inconclusive diagnosis before actually communicating this fact, serving as a "forecast" of the bad news (Maynard, 1996). In the delivery of bad news, forecasting tends to work better than bluntly delivering the news or stalling in its delivery. This is because hesitations and explanations prepare the news receivers in such a way that they may even anticipate the news.

Orientation to the expectation of a diagnosis

Doctor's provision of some speculative cause of the symptoms

Another important finding in this context relates to the fact that even in the absence of a conclusive diagnosis from the autopsy, the doctor may still provide a speculative cause for the malformations of the fetus. This is exemplified in Extract 4.

Extract 4: HMF_ACONGEN_claudia_JEFFERSON_26_11_13

253 GEN: *.hh a gente acredita que talvez o que tenha*
we believe that maybe what
254 *acontecido .h é que de repente al↑gum evento inicial*
happened is that perhaps some initial event
255 *que levô pela lesão da hidrocefalia,*
caused by the lesion of the hydrocephalus
256 *da lesã- lesão cerebral, po::de ter le-*
of the lesi- brain lesion may have
257 *leva:do secundariamente aos- os demais*
secondarily caused the the other
258 *acometimentos né: .h principalmente aquela*
symptoms you see mainly that
259 *posição dos me::mbros, né: .h é aquilo*
position of the limbs, you see, that thing
260 *dali pode ter sido secundário a uma*
that we saw may have been secondary to a
261 *lesão neurológica ↓né*
neurological lesion, you see
262 (0.9)

((Omitted lines))

293 GEN: *.h mas o- o ↑que exatamente levô: a lesão*
but wh what exactly caused the lesion
294 *a gente não conseguiu identificá: .hh*
we couldn't identify
295 *né, por[que: (.) ãrrã]*
you know, because er

Lines 253–61 show how uncertainty in genetic counseling is manifested through hedging devices, such as "maybe," "perhaps," "might," and "may" (Sarangi; Clarke, 2002). By speculating about a neurological lesion as a possible, albeit uncertain, cause of the fetus's symptoms, the clinician demonstrates an orientation to fill the void left by the search for a diagnostic answer; in other words, to fulfill the normative expectation set up by the investigation of a medical condition.

However, the doctor explains that such speculation is constrained by the limitations of the autopsy when he repeats that the underlying cause of the pregnancy outcome has not been identified (l. 293–95). This is consistent with one of the strategies used by doctors in communicating diagnostic news to patients identified in the aforementioned study of interactions in the Finnish healthcare system: they strive to make "their reasoning available to the patients, thus constructing them as understanding recipients of their reasoning" (Peräkylä, 2006, p. 222). In our study, the detailed accounts and the provision of speculative causes for the problem help bring the patient closer to the reasoning behind the professional's inconclusive diagnosis.

Patient agency in the pursuit of a diagnosis

The next extract (#5) shows the patient's agency in responding to a recommendation offered by the professional.

Extract 5: HMF_ACONGEN_claudia_JEFFERSON_26_11_13

582 PAT: *tsk a:i não sei se eu vou ter coragem [de engravi-]=*
ai I don't know if I'll dare get pregnant

583 GEN: *[sim]*
yes

584 PAT: *=de [novo.]*
again

585 GEN: *[não] a gente entende né que .hhh*
no we understand you know that

586 (0.7)

587 PAT: *é o meu mari[do:,]*
it's my husband

588 GEN: *[sim]*
yes

589 (.)

590 GEN: *ã[rrã,]*
mm-hm

591 PAT: *[ele] diz assim né [que::]=*
he says you know that

592 GEN: *[ãrrã]*
mm-hm

593 PAT: *=e:le [não]=*
he doesn't

594 GEN: *[sim]*
yes

595 PAT: *=tem mais coragem né [que::]=*
dare anymore you see it's that

596 GEN: *[sim sim]*
yes yes

597 PAT: *=eu tenho ↑muita vontade de ter*
I really want to have
598 *mais um [mais]::*
one more [more]

599 GEN: *[ãrrã,]*
mm-hm

600 (0.9)

601 PAT: *isso também não:::.* (1.1)
also this isn't

602 GEN: *sim enquanto*
yes for the moment

603 (0.8)

604 PAT: *agora não ↑é o momento pra pensá ↑ni[sso né,]*
now isn't the time to think about it, right

605 GEN: *[é: é que] é*
the thing is
606 *que eu acho também né Claudia tá >tudo<*
that I also think you see Claudia that everything is
607 *muito::=*

Extract 5: HMF_ACONGEN_claudia_JEFFERSON_26_11_13

608 PAT: *=muito re[cente é:]*
very recent it's

609 GEN: *[muito] recente então eu acho que:*
very recent so I think that

610 *às vezes a gente tem que baixá a poeira*
sometimes we have to let things settle

611 *um pouquinho, .h*
a bit

612 PAT: *nã:o >não< [↑penso agora >mas eu pen-<]=*
no I'm not thinking for now but I was thin-

613 GEN: *[né:: si:m sim]*
right yes yes

614 PAT: *=pensava em ter mais um [↓mas]=*
I was thinking of having another one but

615 GEN: *[ãrrã,]*
uh-hum

616 PAT: *=i↑sso: (.)*
that

617 GEN: *ãrrã,*
uh-hum

618 PAT: *dependendo do exame né:*
depending on the test (result) right

At the beginning of this extract, the geneticist recommends that the patient and her partner "let things settle a bit" before getting pregnant again (l. 609-611). However, the patient renders the professional's recommendation unnecessary by providing the information that she is not planning on trying for another child immediately, and that any plans for a future pregnancy depended on the test results (l. 608, 610, 612, 614, 616, 618). In so doing, she resists and rejects the doctor's recommendation (Ostermann, 2021) by orienting to the unfolding consequences of the absence of a conclusive diagnosis, namely, that it rendered any further decisions impossible. In this case, the absence of a diagnosis generates uncertainty not only about the pregnancy that has just ended, but also about what may happen to future pregnancies.

Patients' increased agency

Patients also orient to an absence of diagnosis with actions that show increased interactional agency of the kind not usually seen in consultations where a conclusive diagnosis is delivered (Brookes-Howell, 2006; Ostermann; Frezza, 2017). The following two extracts show different actions that exhibit the patient's agency: in her pursuit of diagnostic clarity (Extract 6) and in the structural organization of the encounter, specifically in initiating the closing of the interaction (Extract 7).

Extract 6: HMF_ACONGEN_claudia_JEFFERSON_26_11_13

295 GEN: *né, por[que: (.) ãrrã]*
right because er

296 PAT: *[eu só queria- eu] só queria sair*
I just wanted I just wanted to leave

297 *daqui com ↑uma certe:za,=*
here with one certainty

298 GEN: *=ãrrã*
uh-hum

299 (1.1)

300 GEN: *[sim]*
yes

301 PAT: *[que se-] eu posso ter ma:is uma gravidez,*
which is whether I can have another pregnancy

302 GEN: *ãrrã*
uh-hum

303 PAT: *porque: é assim eu tenho um sonho de ter mais um*
because it's like I dream of having another

304 *[filho né,]=*
child right

305 GEN: *[si:m si:m]*
yes yes

306 PAT: *=eu tenho só [um de]=*
I only have one who is

307 GEN: *[é:]*
yes

308 PAT: *=catorze*
fourteen

309 *>e eu queria< ver se- se isso for ge[néti]ca,=*
and I wanted to find out if- if this is genetic

310 GEN: *[é:]*
sure

311 PAT: *=eu não posso ter [mais]*
I can't have any more

312 GEN: *[sim] não, depende, né:*
yes no, it depends you see

313 *(0.5) é: >dependendo do tipo de<*
er depending on the kind of

314 *alteração genética vai ter um risco diferente*
genetic alteration there's going to be a different risk

315 *.h só que pra gente fazê esse cálculo de risco,*
but in order to make such a risk assessment

316 *a gente precisa sabê o que que o bebê tinha,*
we need to know what the baby had

317 (.)

318 GEN: *né e infelizmente mesmo com ↑todos os exames*
right and unfortunately despite all the tests

319 *que a gente fez, (.) .h a gente não conseguiu*
that we've done we weren't able

320 *chegá a uma conclusão.*
to reach a conclusion.

The patient expresses a complaint while conveying in so many words what her expectations regarding the diagnosis were (l. 296-301), and thus shows an explicit orientation to what normatively comes next after a diagnosis. In addition, the use of the verb "want" in the past and the restrictive adverb "just" makes such a request single, simple, and one that now has to be abandoned: "I just wanted to leave here with one certainty," (l. 296-97). With that, she conveys her understanding of the consequence of the lack of diagnostic clarity: the inability to estimate the risk of the same malformation occurring in future pregnancies. The design of this turn also shows: (1) the patient's under-

standing of the contingencies associated with the doctor's ability to provide some certainty; and (2) the patient's overt assessment of why a diagnosis is important to her, namely, its implications for planning future pregnancies.

There is only one point at which the patient requests the information she wants (l. 301), which she follows with three accounts that substantiate her request: (a) her "dream" of having another child (l. 303–04); (b) her only child being a teenager; and (c) her awareness that if the malformation is genetic, she will not be able to have another child (l. 309, 311).

By explaining that the recurrence of the same anomaly depends on the type of genetic alteration the baby had (l. 313–16), the doctor accounts for the impossibility of providing the requested information (l. 318–20). It is only at this point in the interaction that the doctor, prompted by the patient's complaint, directly states the implications of having no definitive diagnosis, explaining that the risk of recurrence cannot be assessed. He then goes on to provide further accounts, saying that even having exhausted all avenues, they were still unable to reach a conclusion.

The doctor's accounts in Extracts 2, 3, and 6, which are oriented to the fact that the medical staff was unable to make a definitive diagnosis, represent an important strategy identified whenever there is diagnostic uncertainty. This finding highlights the amount of interactional work uncertainty takes in medical settings. Furthermore, as the patient's complaint in Extract 6 makes evident, the lack of diagnosis generates yet another uncertainty, which is the risk of recurrence in future pregnancies. Had they reached a diagnosis, she would have been able to decide whether to plan another pregnancy.

In deploying the strategy of forecasting bad news, the doctor seems to orient to not having a diagnostic answer as the "delivery of bad news." Nevertheless, by complaining that she wants to leave "with just one certainty," the patient overtly displays what she understands bad news to be in this case: the negative consequences of a lack of diagnosis, resulting in the inability to provide a risk assessment of the same malformation recurring in future pregnancies. Thus, the bad news is constructed collaboratively by both doctor and patient, as the patient's request leads the doctor to reveal the consequences of the lack of diagnostic clarity, to which both participants orient as the worst news.

Another piece of evidence to support the argument that patients display more interactional agency in consultations without a definitive diagnosis is the fact that they are also the ones who initiate the pre-closing sequences. According to the data from the complete study corpus, when a definitive diagnosis is present, the pre-closing sequence tends to be initiated by the geneticist himself.

Extract 7: HMF_ACONGEN_claudia_JEFFERSON_26_11_13

```
1271  PAT:    °vai ficá essa dúvida [↑mas°]
                this doubt will remain but
1272  GEN:                         [vai  ] ficá °essa >coisa<
                                    this thing this gap
1273          (.) essa lacuna em aberto° °°ainda°°.
                  will still remain open
1274  PAT:    °ărrã°
                mm-hm
1275          (1.1)
1276  PAT:    então tá era isso?
              okay then is that all?
1277  GEN:    tá claudia mas a gente fica à disposição se tu
              okay claudia but we remain at your disposal if you
1278          quisé: (.) no futuro conversá de [novo:,]=
              wanna talk again in the future
1279  PAT:                                    [sim   ]
                                               yes
1280  GEN:     =tirá alguma dúvida,
                to answer any remaining questions
1281           (0.4)
1282  PAT:    tá certo en[tão ]
              okay then
```

The patient initiates the closing sequence by expressing an upshot of the missing diagnosis ("this doubt will remain") (l. 1271). This upshot reveals—as is typical for formulations—that the consultation has left an issue unresolved for the patient. By initiating the closing sequence after a one-second gap with "okay then," followed immediately by a request for information ("is that all?") (line 1276), which is common in the closing sequences of institutional encounters, the patient orients to the fact that there is nothing more to be done in this consultation. The geneticist embarks on the pre-closing sequence by offering the institution's services to discuss any questions the patient and her partner may have (l. 1277–80), which the patient minimally responds to with another closing element ("okay then").

CONCLUDING REMARKS

The findings presented in this study demonstrate the participants' orientation to the importance of genetic testing and its results in the organization of genetic counseling, as well as in the (re)organization of patients' lives. The strong orientation towards obtaining a diagnosis is related to the fact that the results would provide the knowledge that the woman and her family need to make subsequent decisions autonomously, such as assessing the risk of recurrence of the same health problem in another pregnancy. If the information the participants seeks is not available, access to knowledge is impeded, making informed decision-making impossible (Horovitz *et al*., 2012; Kukla, 2007; Rapp, 1999).

The patient's orientation to the absence of a definitive diagnostic answer as the "worst" news for her (i.e., leaving her in a state of uncertainty) seems to be consistent with findings from other studies in the field of genetics (Brookes-Howell, 2006; Decruyenaere *et al*., 2013; Stivers; Timmermans, 2016; Timmermans; Buchbinder, 2010; Zaytz; Sarangi; Schnurr, 2016). For example, Decruyenaere *et al.* (2013) show that patients who are motivated to undergo genetic testing cope with the results (even when they bring bad news) with less psychological distress than those who have unspecified test motivations. In the case of this study, the patient's expressed frustration the with the lack of diagnostic news and its impact on for further

pregnancy planning clearly demonstrate that the motivation for the genetic testing itself generated high relevance for a conclusive diagnosis.

In addition to corroborating previous studies demonstrating the various benefits of having a diagnosis, even when this means dealing with the ramifications of bad news (Maynard, 1996, 2006; Maynard; Frankel, 2006; Timmermans; Buchbinder, 2010), the analysis presented in this paper also shows how practices commonly observed in the delivery of bad diagnostic news, such as forecasting (Maynard, 1996) and presenting the medical reasoning behind the diagnosis (Peräkylä, 2006), are also used in the context of an absence of definitive diagnostic news. Thus, these findings provide evidence that the interactional orientation of conveying an inconclusive diagnosis is the same as that of conveying bad news.

Our study also contributes to the literature on the communication of inconclusive diagnosis by demonstrating the physician's resistance to providing any kind of certainty. Stivers and Timmermans (2016) find that geneticists provide some certainty when communicating variants of uncertain significance in the exome sequencing of living patients. However, in those cases, the physicians can request further testing, rely on future technological developments, or even pursue longitudinal clinical trials to confirm their suggestions. In contrast, the institutional setting of the genetic counseling sessions studied here does not provide for further testing or observation, leaving the professional with no choice but to give inconclusive answers.

It should be pointed out that in the setting studied here, if the professional had estimated the risk of recurrence (based on statistics), giving a range of possible diagnoses and the risk rate in each situation, using as a variant the fact that the patient already had a healthy child, etc., he would have exposed himself to liability if the same malformation had recurred in a future pregnancy. As noted throughout this article, the provision of an accurate diagnosis is important to patients for future decision-making. Had the doctor succumbed to the pressure to provide a tentative diagnostic answer, this answer could have misinformed the woman in her family planning decisions, which in turn could have had disastrous consequences, such as the recurrence of the same (or similar) malformations in future pregnancies. What he does, instead, is to refrain from making any sort of risk assessment. The void that is left does much more than simply violate normative expectations: to paraphrase the participant of this study, an absent diagnosis leaves a gap that remains open in the lives of all those concerned.

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