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Authors:
Olga Zayts, Srikant Sarangi and Stephanie Schnurr

Affiliation of the main author:
Olga Zayts
Room 738, 7/F, Run Run Shaw Tower (Building B, Arts)
Centennial Campus
The University of Hong Kong
Pokfulam Road
Hong Kong, China
zayts@hkucc.hku.hk

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Olga Zayts is Assistant Professor at the University of Hong Kong. She leads Health Communication Research Cluster at the Center for the Humanities and Medicine. Her research interests focus on interactional approaches to professional communication in healthcare settings, in particular in genetic counseling and prenatal screening in Hong Kong. She has published widely on the issues of information-giving vis-à-vis advice-giving and decision-making in genetic counseling; politeness; identity; and intercultural communication. Address for correspondence: School of English, The University of Hong Kong, 7/F Run Run Shaw Tower, Centennial Campus, The University of Hong Kong, Pokfulam Road, Hong Kong, China. Email: zayts@hkucc.hku.hk

Srikant Sarangi is Professor and Director of the Danish Institute of Humanities and Medicine (DIHM). His research interests include language and identity in public life; institutional and professional discourse; quality of life and risk communication in genetic counselling; HIV/AIDS; telemedicine; general practice and palliative care; intercultural pragmatics; racism and ethnicity in multicultural societies. He is the author/editor of 12 major book-length publications and more than 200 books chapters and journal articles in leading discourse and communication journals. Address for correspondence: Danish Institute of Humanities and Medicine (DIHM), Aalborg University, Kroghstræde 3, DK-9220 Aalborg, Denmark. Email: sarangi@hum.aau.dk

Stephanie Schnurr is Associate Professor at the Center for Applied Linguistics at Warwick University (UK). Her research interests are professional and medical communication. She has researched and published widely on various aspects of leadership discourse, gender, the multiple functions and strategic uses of humour, politeness and impoliteness, identity construction, the role of culture, decision making and advice giving, and other aspects of workplace discourse in a range of professional and medical contexts. Address for correspondence: Centre for Applied Linguistics, University of Warwick, Coventry CV4 7AL, UK. Email: S.Schnurr@warwick.ac.uk

The management of diagnostic uncertainty and decision-making in genetics case conferences

Abstract

In this paper we examine one type of intra-professional collaborative activity, namely case conferences in a specialist genetics clinic. Our specific focus is on how clinical geneticists manage decision making through team talk in the event of diagnostic uncertainty which is mainly attributable to limitations in the current state of genomic knowledge, 'uncertain significances' associated with genetic test results, and a lack of information/evidence pertaining to cases under discussion. The case conference then becomes a means to minimise the uncertainty and arrive at decisions that optimise the significance of the results in terms of clients' life trajectories. Adopting theme-oriented activity analysis, we examine video-recorded data from five case conferences in Hong Kong. Beginning with a prototypical structural mapping of the case conference activity type, our analysis focuses on what we call 'uncertain cases'. Our findings highlight three discourse types constitutive of team talk: pedagogic talk; diagnostic talk; and decisional talk. In paying particular attention to how uncertainty is formulated and negotiated, we suggest that access to and assessment of different kinds of evidence as well as the activity-specific expert role-positions of the participants is crucial with regard to establishing a correct diagnosis and/or striving towards a minimisation of current uncertainties.

Key words: team talk; decision-making; genetics case conferences; (diagnostic) uncertainty; activity analysis; role-relationships; evidence

Introduction

The last few decades in medicine have been marked by rapid technological and scientific advances. Fox (2002) claims that these advances have led to greater, rather than lesser, uncertainty in healthcare professionals dealing with complex diagnostic and treatment decisions. One way of managing this uncertainty and facilitating decision-making is by drawing on the expertise of colleagues both *within* and *across* various healthcare specialities. Therefore, various forms of intra- and inter-professional collaborations through the medium of talk (e.g. multidisciplinary talks, briefing sessions) and text (e.g. electronic patient records, referral letters) are becoming increasingly important activities in many healthcare sites (Lemieux-Charles and McGuire 2006).

In this paper we examine one type of intra-professional collaborative activity, namely case conferences in a specialist genetics clinical setting in Hong Kong. The cases were referred to the genetics clinic from other hospitals due to a suspected genetic condition, and typically contained limited information about clients' medical history or few confirmed clinical findings. Against this backdrop of uncertainty and inconclusive information, the team discussions are projected towards decision-making about a possible diagnosis, at times supplemented by decision-support techniques (Berg 1997). In what follows we first discuss previous research on uncertainty management and decision-making in the context of case conferences, drawing on studies in the broader health and social care settings. Next, we introduce our methodology and the discourse analytic framework through which we approach our team talk data. This is followed by the analysis of how uncertainty is

managed and decisions are made in genetics case conferences. We conclude with a discussion of our findings and their implications for professional practice.

Case conferences in health and social care settings

Previous studies of case conferences in health and social care settings have identified the multi-functional nature of this activity in fostering professional socialisation, information-sharing and decision-making vis-à-vis role-relationships. Several studies (e.g. Anspach 1988; Atkinson 1995, 1999, 2004; Fox 1957, 1959, 1980) examine the process of professional socialisation of junior medical staff (e.g. medical students, interns, residents), including their ability to manage uncertainty. In the analysis of medical students' case presentation, Erickson (1999) notes the 'face-threatening' nature of uncertainty and links this to limitations of individual expertise. In such situations, it is through inter-professional, collegial talk that 'attributions, claims or disclaimers of authority and responsibility for the knowledge and opinions are being expressed' (Atkinson 2004: 15). Lingard, Garwood, Schryer and Spafford (2002) discuss what they call the 'professional rhetoric of uncertainty' constituted in six components: limits of individual knowledge, limits of evidence, limitless possibility, limits of patient's/parent's account, limits of professional agreement and limits of scientific knowledge.

In addition to fostering professional socialisation as the 'most ostensible purpose' (Anspach 1988), case conferences are also about exchanging information among professionals with the aim of arriving at decisions. Decision-making in the institutional setting is in itself a

complex process, which is manifest at the interactional level as being 'dispersed and fragmented' (Atkinson 1999; see also Boden 1994). Bokhour (2006) identifies three communicative practices in interdisciplinary team meetings dedicated to long-term care settings: giving report, writing report and collaborative discussion. Collaborative discussion may not necessarily lead to a consensus-based future action: for example, the decision may be to defer a final decision, or not to make a decision.

Among others, Måseide (2003, 2006, 2007) specifically focuses on potential tensions in negotiating professional expertise when different medical specialists engage in decision-making. For example, in a study of thoracic ward conferences in which doctors, nurses, radiologists and other professionals participate, Måseide (2007: 204--205) observes that

> when [... different] medical experts collaborate, they have to coordinate their diverse practical conceptions into a sufficiently shared understanding of the problem at hand to be able to render their medical interventions adequate, if not effective, as well as efficient.

Also worth mentioning is the specific nature of decision-making in healthcare as discussed in another study by Måseide (2003) where most of the decisions in routine case conferences typically concern the structural organisation of the problem-solving process so as to avoid, and to a certain extent prevent, major 'life or death' decisions (see also Berg 1997; Atkinson 1999). In the genetics case conference setting where diagnostic and other kinds of uncertainty are prevalent, the process of decision-making might be more obscure

and complex. In all these, the function of professional status might be crucial for participant structure (Grosjean 2004) and consequently for decision-making, including tensions in inter-professional boundaries of expertise vis-à-vis a given institutional order.

Several studies on case conferences in other institutional/professional settings exemplify the structural influence of expert role-relations. Mehan (1986), in a study of case conferences surrounding the special education referral system, demonstrates how the role-relationships of co-present committee members are manifest at the level of language and interaction, which consequently impacts upon the authority of the claims and recommendations that are made in the meeting. Hall, Slembrouck and Sarangi (2006), in a study of a child protection case conference where professionals discuss whether a child should be put on the child protection register, show that the character work by different professionals in depicting the child's mother indexes differential significance depending on the participants' institutional and activity-specific roles in these meetings.

Following from the selective review of relevant literature, we now examine genetics case conferences, focusing specifically on two dimensions: information-sharing (in an attempt to reduce the uncertainty) and decision-making (including diagnostic reasoning) vis-à-vis participants' activity-specific role-relations.

Data and Methodology

Our data corpus includes five weekly genetics case conferences, spanning approximately 10 hours, of clinical geneticists in a Hong Kong specialist clinic. These case conferences involve discussions of clients to be seen in the clinic in the following week.

In keeping with the standard practice, at the beginning of the study the data collection methods were approved by both the University Ethics Committee and the Ethics Committee associated with the specialist clinic where the present study was conducted. The case conferences were video recorded and transcribed verbatim. Two of the case conferences were conducted in Cantonese, the participants' native language, and three case conferences were conducted in English. The Chinese data transcripts were translated into English independently by two research assistants and verified by a bilingual research team member.

All the five case conferences were attended by four geneticists, chaired by the senior medical officer (abbreviated as L in the transcripts).[1] Other members of the team (abbreviated as M, H and C in the transcripts) are all experts in clinical genetics, and share similar professional backgrounds: they were all initially trained in paediatrics and took further training in genetics. All the participants have been with the Clinic for more than 14 years. The team deals with around 3000 clients per year; as a consequence, each case conference session is rather intense in which around 30 to 40 client cases are discussed in a period of two to three hours.

For the analysis of the management of uncertainty and decision making vis-à-vis expert role-relations in these case conferences we adopt the framework of activity analysis (Sarangi 2000, 2010a), as part of theme-oriented discourse analysis (Roberts and Sarangi 2005) that focuses on specific activity types, such as case conferences. The case conference is an activity type, which is "goal-defined, socially constituted, bounded events with constraints of participants, settings and so on, but above all on the kind of allowable contributions" (Levinson 1992: 69; see also Sarangi 2000, Linell 2010).

As a first step, we map the data corpus along the structural, interactional and thematic lines. The activity analysis also involves the description of discourse types that are "specific manifestations of language form in their interactional contexts" (Sarangi 2000: 1). In addition to the discourse devices of hedging, interpretive summary, hypothetical structure etc., we then identify three discourse types constitutive of team talk: pedagogic talk, diagnostic talk and decisional talk.

Analysis of Genetics Case Conferences

A broad structural map of the case conference session reveals the following phases:

1. Introducing the client
2. Presenting the client's medical history
3. Eliciting/volunteering different kinds of information related to the client's case
4. Re-assessing and discussing the case
5. Making a decision about a case
6. Closing the case

Sequentially speaking, these phases may be dispersed depending on the specific case under discussion as well as contingent participant structure.

The case conferences follow a routine script: the discussion of each case is typically initiated by the Chair who announces the client's name (phase 1), and the team members take turns (after being nominated by the Chair) in presenting the cases (phase 2). The team members' turns usually involve (albeit to different extents) elicitation and volunteering of information, assessment and discussion of the case and decision-making (phases 3, 4 and 5). Most of the cases are then closed by the Chair (6).

The thematic map points to uncertainty management and decision making as the focal themes manifest in phases 2-5. We can distinguish between three different types of client cases in the data corpus. First, there are 'certain' client cases (around 60% of all clients) where the diagnosis has been established, and the team agrees on the management of the condition. The clients in these relatively straightforward cases are referred to by geneticists as 'inactive' as they typically come to the clinic every one to two years for follow-up appointments. The discussion of these cases takes on almost a 'ritual' character as seen below.

'Certain' client cases

In Extract 1, L chairs the meeting and initiates the discussion of the case by displaying the client's photographs on the screen:

Extract 1

1 L: Okay! The next one is (.) this one (.) ((everybody looks at the client's photographs on the screen)) this one (.) this=

2 H: =This one looks familiar.

3 M: Familiar, familiar. (.) ((looking at the photographs)) I don't quite remember.

(5.0) ((everybody studies the client's records))

4 L: Thirty. (.) Okay! Autism.

5 C: Autism, this one, we found that apart from autism, there is also MR. He is (.) he had microcephaly when we saw him. There's synophrys. Otherwise, it's normal ((inaudible)) (.) (feet). Last time we did FRAX A, and also MECP2. Both are normal. For the brain imaging, there's a thinning of corpus, corpus callosum. He is now studying in a special school, receiving speech therapy. Apart from microcephaly, otherwise I can't see any ((inaudible)). Last time we suggested doing the karyotype again. The karyotype is normal. We have counted ten cells.

6 L: Mhm.

(3.0) ((C is flipping through the client file while L is taking notes))

7 L: So there's no big problem.

8 C: Yes.

(.) ((C is flipping through the report while L is taking notes))

9 L: Is he short?

10 C: Apart from the head size, everything is normal.

((the team moves to the next client case))

This extract represents the 'ritual' character of case conferences: L announces the client's diagnosis (turn 4); C goes on to present the client's medical history (turn 5); and L and C assess the case (turns 7, 8 and 10). The language of diagnosis and physical examination in this case is characterised by the 'matter-of-factness' as autism and MR (mental retardation) become confirmed.[2] In a sense, this 'matter-of-factness' contributes to the explicit construction of 'normality' of this case (turn 7, 'So, there's no big problem'; and turn 10, 'Apart from the head size, everything is normal'). There is little uncertainty and disagreement about the diagnosis, which accounts for the absence of extended team talk.

By engaging with 'inactive' cases the team members, however, do more than just 'perform the ritual' of case presentations. In particular, cases like these are important in manifesting and reinforcing the team's consensual knowledge, and preparing the grounds for discussing more complex client cases surrounded by a higher degree of uncertainty that we will discuss in the next section.

The second type of client cases in the corpus (comprising 10% of all clients) includes those where the diagnosis is established during case discussions after the team members engage with available evidence. These cases are typically turned into a 'learning platform' for professional socialisation by the Chair who highlights 'good professional practices' concerning display of diagnostic reasoning based on available evidence, which is then

subjected to recursive assessment. In this regard, case presentations take on a pedagogic dimension.

In the remaining 30% of the cases the geneticists deal with uncertainty that is linked to limited available evidence. The presence of uncertainty generates extensive team talk (in phases 3 and 4) which is projected towards decision making regarding the clients' diagnoses. This latter cohort is our analytic focus here.

'Uncertain' Client Cases

In our data corpus, uncertainty in the client cases is manifest through a range of discourse/rhetorical strategies – ranging from explicit formulations of not being able to establish a diagnosis (e.g. *I could not find anything; we found nothing; the cause of X was not totally confirmed*); to use of hedges (e.g. *I am not sure; I suspect/think that …; this is probably/perhaps X)*; simile (e.g. *His features are similar to X*; *it is like X*); and hypothetical structures (e.g. *If this is the case, then…*). Uncertainty is also manifest prosodically through pauses, halted speech and numerous hesitation markers (e.g. *The scan, (.) um (.)…*).

In what follows we first examine how uncertainty is formulated, before considering how the team negotiates uncertainty in order to reach a decision about each case.

<u>Formulating uncertainty</u>

The uncertainty is typically formulated during the presentation of a medical history. The sources of uncertainty are attributed to the limitations of available technology; to the hospitals that have referred clients to the specialist clinic (corroborated by ambiguous referral letters); or to limitations of expert knowledge on the part of the targeted addressees of such letters. In many cases, the uncertainty is complex and lies with more than one source, as in Extract 2 where H is presenting the case:

Extract 2

1 H: This one, the previous baby passed away. The (post-mortem) said the baby has intrahepatic cholestasis. Referred to us (.) for consulting. We saw the parents last time. Oh, yes, in the last visit the mother probably=

2 L: =She was not pregnant.

3 H: Oh, yes, not pregnant, the mother should not be pregnant. Er (.) for consulting. But at that time the um cause of the death of her baby was not totally um confirmed. There was infection, other (liver problems). But anyway, after the consultation, if it was really the genetic cause of her familial intrahepatic cholestasis, er because this is relatively (uncommon) ((inaudible)). But most important was if we could test it. After seeing her last time, she is here this time to check the baby.

This is the second time the client is seen in the clinic. She was referred to the clinic previously following the death of her first infant. There is uncertainty about the reason for referral: according to a post-mortem examination the child was suspected to have a genetic condition called progressive familial intrahepatic cholestasis (PFIC). There is also uncertainty whether the team has appropriate diagnostic facilities to test for the suspected condition. Now pregnant for the second time, the client attends the clinic to check the genetic status of her second child. In the case presentation H makes an explicit reference to the diagnostic uncertainty (*the um cause of the death of her baby was not totally um confirmed*, turn 3). The uncertainty is also conveyed through the repeated use of an if-structure (*if it was really the genetic cause; if we could test it*, turn 3) and numerous hesitation markers. According to H, the uncertainty relates to the rarity of the genetic condition, and due to the fact that the first infant had other multiple problems, including an infection and a dysfunctional liver. The limitations of available diagnostic facilities are also alluded to by H (‘*But most important is if we could test it’*, turn 3).

What a case presenter reports is typically not taken prima facie by the team members. Case presentations are followed by team discussions to which each team member contributes, which we now turn to.

Negotiating uncertainty vis-à-vis activity-specific role-relations

The team members contribute to the process of negotiating uncertainty differently depending on their expert role-relations in the case conference. In particular, the Chair (L)

– both as part of his activity-specific role and genetics expert role – typically steers the discussions. For example, in Extract 3 below the team are dealing with the case where a number of tests have been performed in the clinic, and only one of them, namely MLPA, has shown the duplication within chromosome band 3p, which is a pathological abnormality. Therefore, in this case the uncertainty lies with whether this is a case of a genuine abnormality that can only be detected by newer technologies like MLPA, or whether this is an 'artefact', i.e. a false positive result. L's lead participation is evident in prompting the presenter to provide interpretation of uncertain clinical findings, eliciting further information, and issuing various assessment statements.

Extract 3

(The extract comes after the presentation of the medical history)

1 C: For the midline hand movement, it didn't- didn't mention. It is ((inaudible)) (3p duplication) ((C appears to be talking about the details of the 3p duplication while looking through the report))

2 L: It means,=

3 C: =For the karyotype, (.) it is (.) ((searching his notes)) we did (.) the normal, we have counted seventeen, seventeen cells. ((passing the report to L))

4 L: But (.) how about FISH?

5 C: For FISH, emm it's normal. FISH for 3p ((passing the report to L)) (.) (I couldn't find any=)

6 L: =3p duplication.

7. (8.0) ((L is reading the report))

((6 turns are omitted; the team examines the client's report))

14 L: (There is nothing special in the features.)

15 (3.0) ((L is reading the report))

16 M: The features are quite obvious.

17 (4.0) ((M is reading the report))

18 H: We can refer to the document ((laughs))

19 L: It's like (.) it's like, what the book mentions. There're a few similarities.

((15 turns are omitted; the team talks about the specialist literature))

35 H: See if you have the MLPA report underneath.

36 L: Do you have it? We should check. (.) (We should use another MLPA to confirm.

37 M: [Mhm.] ((nods))

38 H: [Mmm.]

39 (3.0) ((L is looking through the report))

40 L: When did the patient do the scan?

41 C: The scan, (.) umm (.) ((looking at the report)) in 2005.

((the team moves to the discussion of the next client case))

In this extract, C elaborates the results of the two previous tests, the karyotypic analysis and the FISH test, in response to L's prompt to provide the interpretation of the reported clinical findings (turns 2 and 5). Both the test results are normal, which is marked by typical case formulation. As the results do not resolve the uncertainty, this uncertainty generates extensive team talk.

The team members engage in reassessing available clinical evidence (turns 14-19). L's assessment of the client's features as 'nothing special' (turn 14) is followed by M's agreement that the features are 'quite obvious' (turn 16), i.e. there is nothing in them that may help the team resolve the diagnostic uncertainty – which comes across as a kind of neutralisation. H suggests referring to the 'document', i.e. the specialist literature as a possible source of resolving the uncertainty (turn 18). L agrees with the suggestion and recalls that the features described in the literature are 'like' the client's features (turn 19); however, the use of the rhetorical device of simile accentuates that the condition under scrutiny is similar, but *not* the same as the one in the literature. Interestingly, in examining the client's features, the team talk focuses around the client's report that is being consulted throughout the interaction and this in a sense facilitates the team talk. As reassessment of the report does not lead to resolving the uncertainty, it becomes clear that the team needs further evidence to establish a diagnosis.

The differential expert role-relations among the team members are evident in how decisions are formulated in case presentation. In the majority of case conferences, it is the Chair (L) who takes a lead in summing up the team talk (what Ferrara (1994) calls 'interpretive summary') and in formulating a decision. In Extract 3, prompted by H who asks about the previously done MLPA report (turn 35), L suggests that another MLPA test should be performed to confirm the duplication within chromosome band 3p (turn 36). The decision, however, is framed as the team's collective decision with the use of collective pronominal referencing (e.g. *we should check*; *we should use*…). The team's agreement

with this decision is conveyed through H's and M's minimal responses and nodding (turns 37-38). The limitations of available technology in this case are, therefore, dealt with by resorting to more technology.

In more complex client cases, the decisions may not be readily available like in the case of the clients in Extract 2 (continued in Extract 4) where uncertainty is attributed to two sources: the referring hospital and available diagnostic facilities. Similarly to other cases, once the uncertainty has been established in the case presentation, it generates extensive team talk.

Extract 4 (continuation of Extract 2)

3 H: Her familial intrahepatic cholestasis, because this is relatively (uncommon), ((inaudible)). But most important was if we could test it. After seeing her last time, she is here this time to check the baby.

4 L: Why do you, why are you so sure it is (intrahepatic cholestasis)?

5 H: Um? Not sure. Even for her post-mortem.

6 L: What did the post-mortem tell?

7 H: Oh, yes. Post-mortem results said progressive familial intrahepatic cholestasis. That is the referral cause.

8 M: Does she have a family history?

9 H: No.

10 M: No family history?

11 H: Last time, as I said, it was about the cause of the death. That was it.

12 L: Here it says Low gamma-GT.

13 H: Huh? Yes. Here they wrote it as the previous baby had positive ((inaudible)) for CMV. They said they initially wanted to do CMV but afterwards post-mortem said it seems like this problem. That is the familial intrahepatic cholestasis. So it is very difficult to conduct the counseling.

14 (0.9)

15 L: ((examines the reports)) Progressive familial intrahepatic cholestasis. Perhaps, usually there is some special (.)

16 M: Probably, usually for intrahepatic cholestasis. [Yes.]

17 L: [Huh?]

18 M: Yes.

19 L: Perhaps. (2) Write in this way is, this is low gamma GT, [huh?]

20 H: [Yes.] Yes. Low gamma GT.

21 L: Low gamma [GT.]

22 H: [After]wards I finished the counselling, if this is the case, still occurrence one in four.

23 L: Um. Is there ((inaudible)). But if there is not specimen for this,=

24 H: =No. no.

25 L: Did you notice?

26 H: No. that is why this time I will see how she goes.

27 L: Um.

28 M: Is there gene? What are the genes?

29 H: These.

((6 turns are omitted; L examines the report))

36 L: I think it is difficult to find.

37 M: Mhm. ((while nodding))

((the team moves to the next client case))

After the case presentation, the team members engage in reassessing the case. L again steers the discussion by eliciting further information from H (turns 4, 6, 23 and 25); drawing attention to specific clinical findings (turn 12); assessing and summarising the clinical findings in the client's report (turns 15, 19, 21, 33 and 36). L first elicits whether H is certain about the diagnosis (turn 4). H's reply that PFIC was suggested by the post-mortem examination results and that he is 'not sure' about it further accentuates the diagnostic uncertainty (turns 5 and 7). M inquires about the family history of the condition (turn 8). As clarified in the interview with the team, this additional evidence may potentially help resolve the uncertainty as in the case of PFIC the transmission of the condition is 1 in 4, and the client may be aware of the condition running in the family. While in turn 9 H asserts strongly that there is no family history, questioned by M again in turn 10, he states that the family history of PFIC was not the focus of the previous counselling session that was solely concerned with the cause of the first infant's death (turn 11). H's reply presupposes that the information is not available (rather than there is no family history); it may also be seen as an account for why an important piece of evidence was not collected in the previous counselling session that mitigates the potential threat to H's expert position.

Then L inquires about one of the test results that showed a low level of a liver enzyme, low gamma GT (turn 12). This is an important piece of evidence as well, as the low levels of this enzyme are associated with two particular types of PFIC, namely PFIC1 and PFIC 2. H notes the discrepancy between the initial diagnosis of an infection and a dysfunctional liver and the post-mortem results pointing to a genetic condition that have complicated the counselling process (turn 13). By providing this justification for why this important information has not been highlighted in the case presentation, H mitigates the threat to his expert position. This part of the interaction demonstrates the point made earlier that the team members do not take the case presentation prima facie but actively engage with and question presented case evidence. To an extent, the interaction resembles the case presentation in other medical settings (e.g. Erickson 1999; Atkinson 2004) where clinical judgements are questioned and accounted for.

Having clarified the presented evidence, the team members engage in a close re-examination of the case. The discussion between L and M about some 'special' gene defects of PFIC that follows displays an interesting example of team alignment. In particular, L's remark about the 'special' gene defects does not appear to be complete (turn 15). M, however, picks up on the meaning of that elliptical utterance and agrees with L (turn 16). Besides signalling alignment, this exchange also attests to the team's 'collective competence' (Atkinson 1995) as a resource for resolving the uncertainty that is manifest at the interactional level through several hedging devices (turns 15, 16 and 19: *perhaps*, *probably* etc).

The complexity of the case and the attendant uncertainty impact upon what happens at the decision making stage. L brings up again the results of the test that showed low gamma GT (turn 19) and suggests to H to note this information down. H downgrades the validity of this evidence through hypothetical formulation (*if this is the case...*, turn 22) and claims that the transmission of the condition is still one in four. This latter comment is ignored by L who continues examining the client reports. Having confirmed that there is no specimen for the low gamma GT test, H suggests 'seeing how she goes' (turn 26), i.e. leaving the decision open until after the consultation with the client. If accepted, this suggestion may enable the team to acquire further evidence to make a decision.

The nature of the evidence, however, is described in very vague terms, which attests to the complexity of this case. H's suggestion is not immediately accepted by the team as the assessment of the available evidence continues. M inquires about the client's genes (turn 28) and engages in discussing them with L and H. L sums up the case with 'it is difficult to find' any confirmation of the client's diagnosis by examining the genes (turn 36), and M agrees with him (turn 37).

In some cases it becomes obvious that to resolve the uncertainty the team's collective expertise and acquisition of further evidence are not sufficient. Particularly, this pertains to those conditions that require drawing on the expertise of specialists outside the domain of genetics. For example, in Extract 5, the client was referred to the clinic due to suspected Kennedy disease which has a neurodegenerative basis. H is again presenting the case, although C had seen the client in the clinic previously.

Extract 5

1 L: Kennedy.

2 H: And refer (.) this patient was referred suspected (.) Kennedy disease. And the patient healthy and until few years ago and (docu) mentions to have (.) some walking difficulty or muscle weakness and or proximal muscle wasting. And the CPK (.) checked. The result is five hundred to one thousand that is a little bit elevated. And the muscle biopsy also performed in medical unit and the result come back to be (.) chronic neurogenic atrophy. (.) And for family history unremarkable, the proband has one son. And physical examination of ((inaudible)), no facial weakness, no (noted) fasciculation, normal muscle power. And so no blood taken from the patient, maybe ask the (.) already ask the proband to bring along the MRI (.) spine (.) result for documentation.

3 (2.0)

4 L: He was referred for suspected Kennedy?

5 H: Yeah he was referred suspected (.) ((while reading the report)) he were referred with a (1.0) muscle wasting- ah yeah suspected Kennedy disease.=

6 L: =Hm hm. (2.5) hm. ((takes a report from H)) How did they suspect Kennedy?

7 H: Hm[m]

8 L: [He] has muscle fasciculation, including facial muscles. No sensory symptoms. (2.0) EMG wide spread chronic denervative changes of arms and tongue. (17.5)

((both L and H are reading the report)) Left compression. Mm hm. ((inaudible)) you you were suspecting nerve com- compression?

9 C: Mm.

10 L: ((C's first name))? ((L invites C to join the discussion))

11 C: Mm.

12. L: Why?

13 C: Ah (2.5) ((C takes the file from H)) possibly he has been commented had (.) spine has (.) disc prolapse. And also osteophytes. (.) And he has been working in ((name of hospital)) as a dark room technician. So probably I'll ask him to bring possibly the the (.) structural compression (.) mostly related to his current problem, especially the promixal muscle wasting, and leading to chronic neurogenic changes.

14 L: Mhm. [So he]

15 C: [And pa]rticular the the (.) particular with the features (they're just) asymmetric, asymmetric involvement=

16. L: =Mm [hm.

17 C: [Probably can be accounted by the (.) difference in ((inaudible)) pressure.

18 (4.0) ((C hands the report back to H))

19 L: So he gave a history of (.) spinal problem.

((10 turns are omitted in which L talks about the client's spinal problem and asks about his age))

30 L: Osteophyte, disc prolapse. (2.0) Any (.) ((inaudible)) involvement? (.) Hm?

31 C: No pos- probably not, no complaint of (.) phonation or swallowing problem.

32 L: Mm hm. (3.5) Hm. Okay so I think we need to reassess, ((hands the report to H)) okay? ((takes another report from H)) (5.0) (toe) biopsy. (.) Left (.) vastus (.) medialis (.) show chronic neurogenic atrophy. Mm hm. CP(.)K persistently elevated. Mm hm. But how- how can you explain the (.) elevated CPK if this is (.) compression.

33 C: Mm ((H hands the report to C)) (5.0) the pos-sibly he has also car-coronary heart disease the (.) to to know better (.) the (inaudible) (factor) the CK whether it is muscle enzyme or (.) cardiac enzyme.

34 L: It's cardiac enzyme?

35 C: Ya.

36 L: So you mean he is in (.) cardiac attack. (.) Right?=

37 C: =He has been ah ischemic heart with (.) balloon dialatation, just last year.

38 L: Yeh but the (.) I mean that that ((laughs)) shouldn't give a a persistently elevated CPK. Alright? It's only during the attacks. (1.5) Mhm.

39 C: The other is er he gave a history of relative static, static condition. (.) He doesn't complain any (.) deterioration, deteriorating, deterioration in his condition.=

((5 turns are omitted in which L and C talk about the onset of the condition))

45 L: Okay. Well, if I were you, I would (.) simply do the Kennedy testing for him because (.) that was a simple test. ((while C is taking notes)) And (.) and (.) I wouldn't argue (.) with (.) well probably this is a neurologist. Okay? If the neurologist (.) is suspecting Kennedy disease, maybe the geneticist is not in the

position to argue with him, right? ((C has finished taking notes and hands the report back to H)) Because that's a neurological condition.

((the team moves to the next client case))

Similarly to other cases that we have examined, L assumes a leading activity-specific, expert role in the case discussion. L questions H as to how the referring hospital established the diagnosis (turn 6). Addressing this question would require interpretation of the reported investigation, rather than their descriptive presentation. Since H's response is minimal (*hmm*, turn 7) that points to the difficulties he faces, L starts enumerating the symptoms (*muscle fasciculation*, *no sensory symptoms* etc.), thereby offering an 'interpretative summary' of the case presentation. One symptom in particular, nerve compression, that C noted when he counselled the client previously, attracts L's attention. L elicits what led C to make this diagnosis (turns 8 and 12). In response C recalls what the client told him (*he has been commented had (.) spine has (.) disc prolapse..*, turn 13), and concludes that these symptoms and the client's facial features may account for the diagnosis he made. Noteworthy here is C's very lengthy response which contains numerous hedges, pauses, hesitation markers and repetitions that attest to his uncertainty about the diagnosis. C again takes the lead in summing up these symptoms very concisely (*So he gave a history of (.) spinal problem*, turn 19).

In what follows L seeks information about the client's age (as the onset of the Kennedy disease is common in males between 20 to 40 years old) and whether the client has reported any problems with swallowing (turn 30). The absence of the latter symptom, as

reported by C (turn 31), exacerbates the uncertainty, as swallowing problems are common in clients with the Kennedy disease. As available evidence appears inconclusive, L formulates a decision about re-assessing the client (turn 32).

C continues, however, studying the client's report. One symptom in particular attracts his attention: a high level of CPK. L probes C how this symptom could be explained (turn 32). C's response that the client may have a coronary heart disease conveys his uncertainty about this symptom (which is signalled by numerous hedges and hesitation markers (turn 33). L challenges this evaluation (*So you mean he is in (.) cardiac attack. (.) Right?*, turn 36) and highlights the fact that a high level of CPK is observed at the time of the cardiac arrest, and it should not be persistently high otherwise (turn 38). The laughter indicates his sarcasm that an experienced geneticist like C could misinterpret this symptom.

To restore his expert position, C highlights another symptom in the client's history, namely that the client does not complain of any deterioration of his condition (which is common in cases of Kennedy disease, turn 39).

Once again L takes a lead in decision-making as he outlines that the team should perform the test for Kennedy disease to confirm the diagnosis (turn 45). L's decision is framed as a criticism: he stresses the simplicity and the low cost of the test, and that the team should trust the expertise of a neurologist in making a diagnosis about a neurological condition. He, however, mitigates his criticism by employing hypothetical structure (*if I were you…*), hedging (*well, may be*), and tagging (Okay?, Right?) in order to elicit the team's alignment

with the decision made. In other words, in this interaction the diagnostic decision is deferred until after a test is performed, and the clinical judgment is outsourced to a specialist external to the domain of genetics.

Discussion and Conclusion

In this paper we have examined how a team of clinical geneticists manages diagnostic uncertainty in case conferences. In addition to the information-sharing dimension of these case conferences, we have drawn specific attention to a range of discourse/rhetorical strategies that presenters and discussants of cases employ to formulate the uncertainty in client cases, ranging from explicit markers (e.g. *we could not find anything*) to hedges (e.g. *probably*, *perhaps*) to prosodic means (pauses; hesitation markers). While the responsibility of case formulation lies solely with a case presenter, we have shown that these formulations are not taken prima facie by the team. Rather, the presence of uncertainty in client cases generates extensive team talk that is projected towards decision-making. The case evidence and clinical judgments are questioned and accounted for recursively in the team discussions. In other words, the team members evoke their 'collective competence' (Atkinson 2004) as a resource for managing uncertainty in the discussed cases.

We have shown how such 'collective competence' can be mapped onto activity-specific expert role-relations (Sarangi 2010b, 2011). There are several means available to geneticists: medical representations of different artefacts (e.g. reports of investigation) and

clinical experience. When the geneticists appear to have exhausted all available case evidence, they typically refer to the specialist literature as the 'ultimate' source of evidence. We would like to stress the central role of evidence in the case discussions: it is not only an 'interactional product' (Måseide 2006: 44) that is discursively constructed by the team members as the talk progresses but it takes a central place in facilitating and enabling team talk.

We have suggested that it is not just access to evidence that structures participation in these case conferences; activity-specific expert role-positions are equally influential in how team talk is managed. While all team members are involved in team talk, the regulatory role of the Chair is evidenced in that he steers the case discussions by offering interpretative summaries, asking probing questions, challenging claims, seeking further evidence, and directing the team discussions towards diagnostic decisions. While the legitimacy of the Chair's claims and decisions are embedded in his activity-specific role in the case conferences, it is also due to the fact that he is accountable for the decisions made by the team. The contributions by the Chair may also be seen as an attempt to educate fellow team members about specific genetic conditions and good diagnostic practices. Where previous research has mainly examined case conference as a site of professional socialisation of junior medical staff (e.g. Anspach 1988; Erickson 1999; Atkinson 2004), our analysis shows that case conferences may also serve as a learning platform for continuing professional development (CPD). The associated 'threat' of implementing a pedagogic component to case conferences among professionals is that it may be seen as an attempt to question one's competency and expertise; therefore in our data these interactions

are heavily mitigated by the Chair through a range of discoursal/rhetorical strategies (e.g. if-clauses, hedges, pronominal references).

In our analysis we have also highlighted the diagnostic and decision-making component of team talk. In mainstream clinical settings outside genetics, the judgment of professional competency typically relates to one's ability to establish a correct diagnosis. The case discussions that we have analysed show that this may not be the case in genetic contexts. Genetics remains an uncertain science. The team members thus embark on making decisions about how the uncertainties in client cases can be minimised (and potentially resolved). More specifically, the decisions pertain to collecting additional, direct evidence through further testing (Extracts 3 and 5) and through clinical observation (Extract 4). Extract 5 in particular is interesting as the decision to conduct further tests implies affirming a diagnosis established by a specialist in a different area, namely a neurologist. This decision highlights the limitations of dealing with medical uncertainty within the realm of one's medical specialty, which echoes previous research on inter- and intra-professional problem-solving in healthcare (e.g. Måseide 2003, 2006, 2007)

Although our study is limited to inter-professional case-conferences in one specific context of clinical genetics, we believe that our findings about the role of team talk in the management of uncertainty and decision-making have implications beyond the analysed context of genetics case conferences. In particular, our study supports the findings of previous research about the multifaceted nature of uncertainty in healthcare and highlights the importance of intra- as well as inter-professional team collaborations in modern

healthcare. Team talk is accorded a central role in professional practice as it is essentially through talk that uncertainty is negotiated and eventually minimised or resolved.

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Appendix I. Transcription conventions

(.)	noticeable pause shorter than 0.5 second
(n)	timed pause; "n" - the interval measured in seconds
WORD	loud volume
.	falling intonation
,	level intonation
?	rising intonation
[	the beginning of overlapping
]	the end of overlapping
( )	transcriber's best guess of uttered words
(( ))	non-verbal features or transcriber's comment

A: XXX= B's utterance is latched onto A's

B: =XXX

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[1] The names of all participants have been changed to preserve their anonymity. We would like to thank the people who have participated in this research.

[2] It is worth noting that case conference team talk, from an outsider discourse analytic perspective, is full of technical jargons which are not easily decipherable. Further information about the genetic conditions and other technical aspects can be obtained from http://ghr.nlm.nih.gov/