Expert and lay formulation of «normality» in genetic counselling[1]

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Dans cet article, j'étudie la façon dont les notions de «normalité» et d'«anormalité» sont produites et (re)définies interactionnellement, en les traitant comme une des façons dont les acteurs gèrent les incertitudes aussi bien sociales que biomédicales dans le contexte du conseil en génétique. Puisque de nombreuses prédispositions génétiques sont présymptômatiques, une grande partie de l'échange durant le conseil porte sur les chances qu'a la personne d'être affectée par une maladie génétique dans le futur, même si au moment de l'entretien celle-ci elle est tout à fait «normale».

Sur la base de réflexions issues de la littérature philosophique et sociologique sur les différents sens que peut prendre la notion de «normalité», entendue comme «typique, ordinaire», comme «non-déviante, non-pathologique» ou encore comme «idéale, désirable», je montre comment ces sens peuvent s'imbriquer dans des explications génétiques à propos de formes d'héritage ou d'attribution de labels diagnostiques. Je suggère que les cliniciens comme les patients glissent constamment d'un sens à l'autre: alors que les clients développent un raisonnement ordinaire qui donne sens à l'absence de normalité, les cliniciens tentent d'équilibrer différents types d'évidences, basées sur les résultats cliniques, les tests de laboratoire, ou l'histoire familiale pour arriver à une décision diagnostique.

1. Introduction

In many societies and communities, the idea of seeking medical attention presupposes a dichotomy between the normal and the abnormal/pathological. A form of self-diagnosis accompanied by «lay» accounts of «not being well» is the precursor to seeking «expert» help, although one can find different degrees of «lay expertise» with which patients narrate their illnesses in and out of clinical settings (Sarangi, 2001). At a societal level, Parsons (1951) proposed the notion of «sick role» as opposed to the «social roles» to explain the patient's «exemption from normal responsibilities to society». For him (1951, p. 477), «illness is, in one of its major aspects, to be defined as a form of deviant behaviour». We have here a theoretical formulation of illness as absence of health, and by extension, a juxtaposition of deviance and norm which lies at the heart of a theorisation of the social order. In other words, the absence of health at the individual level can and does become social

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problems. The biomedical and the social thus become intertwined in the definition and management of illness. As Twaddle (1973, p. 753) puts it:

> Medical definitions of disease constitute a special type of social deviance. The norms applied are socially defined with respect to biological functioning. Judgements are made as to whether or not the organic processes exceed permissible limits... if so, it is judged that a pathology is present, the individual in question is sick (i.e., deviant) and should be subjected to the controls of a therapeutic regimen.

The social-theoretical and biomedical interest in normality and deviance can easily be mapped onto our everyday, lived experiences of health and illness. Doctors and patients alike invoke these concepts and make them locally relevant in their accounting practices. In communicative terms, the patient brings his/her abnormal/pathological body/self to the clinic, narrates the symptoms that may have caused the abnormal state and hopes for medical intervention so as to return to normality and assume social responsibilities. As far as the physician is concerned, s/he follows routine elicitation procedures to take the history of the presenting condition with a view to identifying what is non-normal. A diagnostic label to classify the condition is deemed to be essential for both doctors and patients before embarking on any treatment / intervention measures. In the biomedical paradigm, it is possible for the physician to even create a label of «nondisease» and define a state of non-illness – i.e., formulations indicating that an individual does not have such-and-so – while also accommodating incorrect diagnoses and the possibility of false positives (Meador, 1965).

In the context of genetic counselling – which is my focus of discussion here – and many other allied health and social care settings in the contemporary society, the Parsonian notion of the «sick role» – or what constitutes normality and abnormality – becomes problematic. In this article I wish to explore the extent to which normality/abnormality is formulated differently by genetic counsellors and their clients in the clinical setting. I consider genetic counselling as a critical site for the study of how normality/abnormality is locally produced and (re)defined as a way of dealing with uncertainties in both biomedical and societal spheres of activity.

The clinical activity – whether leading to disease or non-disease states – takes the form of a ceremonial order (Strong, 1979) consisting of the following phases: history-taking, physical examination, diagnosis and treatment. Genetic counselling as an activity type, however, differs from the mainstream medical encounters – both in terms of structure of interaction and content (Sarangi, 2000; Sarangi, in press). History taking in genetic clinics, for instance, follows

a significantly different course. Mainly structured around information on family trees, it pervades the entire counselling process. Patients tend to volunteer information, especially about affected and unaffected family members much more than is the case in mainstream doctor-patient encounters. Also, what constitutes physical examination in genetic counselling, which may or may not be undertaken in the clinic, is different. Similarly, diagnosis (even after physical examination) and treatment phases may be absent altogether, since for many genetic disorders there is no available treatment, and in some cases even a diagnosis is not possible. Indeed much of genetic counselling work is around prognosis, risk assessment and coping and these aspects are reflected at the interactional level. In many ways genetic counselling is a hybrid activity type (Sarangi, 2000), which has some family resemblance with other counselling discourses involving HIV/AIDS, divorce mediation, family planning, etc (Candlin and Lucas, 1986; Erickson and Shultz, 1982; Greatbatch and Dingwall, 1999; Labov and Fanshel, 1977; Perakyla, 1995; Silverman, 1997). It is worth pointing out that many of the genetic conditions are presymptomatic, i.e., one talks about the risk of being affected in the future although they may be quite normal at the time of attending the clinic. The genetic status of an individual is often characterised as being a carrier, or being affected, or where a diagnosis is possible, as having the condition. Even in the latter case, when a diagnosis is possible, the exact onset and the nature of illness is far from clear. In other words, uncertainty about future risk is a rather normal phenomenon in the activity of genetic counselling.

2.   The different meanings of «normality» in sociological and philosophical thinking

The concepts of norm and normality have a long and complex genealogy. They have engaged the minds of Foucault and Durkheim in their attempts to explain social order and change (see, for example, Foucault's accounts of sexuality and mental illness and Durkheim's accounts of suicide and anomie). Durkheim (1964) uses the analogy of health as normal and disease/morbidity as not only pathological, but also accidental and deviant. To quote Durkheim (1964, pp. 55-56):

> We shall call «normal» these social conditions that are the most generally distributed, and the others «morbid» or «pathological». If we designate as «average type» that hypothetical being that is constructed by assembling in the same individual, the most frequent forms, one may say that the normal type merges with the average type, and that every deviation from this standard of health is a morbid phenomenon.

As we can see, normality is not only to be conceived of as an either/or contrast, but also as a form of evaluation which involves scales and measurements based on averages, aggregates and patterns of distribution.

The height and weight chart used for measuring children's growth rate is a good example of how normality is constructed with reference to an aggregate. As Armstrong (1995, p. 396) puts it:

> Each line marked the «normal» experience of a child who started his or her development at the beginning of the line. Thus, every child could be assigned a place on the chart and, with successive plots, given a personal trajectory. But the individual trajectory only existed in a context of general population trajectories: one child was unique yet uniqueness could only be read from a composition which summed the unique features of all children. A test of normal growth assumed the possibility of abnormal growth...

Statements (e.g., about normal growth) are based upon observations made in the past, with the assumption that such frequencies will hold for the future. The Foucauldian notions of surveillance and governance are implicit in such institutional/professional gaze as a mode of social control. Here we can see several meanings attached to the notion of normal: normal as typical/usual; normal as distinct from abnormal/pathological; and normal as desirable or what ought to be.

It is not an overstatement to claim that normality spans all aspects of social life. The Durkheimian famous statement – «crime is normal because a society exempt from it is utterly impossible» (1964, p. 67) – implies both the typical/usual sense and the abnormal/pathological sense, because crime is logically implied in the social type, i.e., «it is bound up with the fundamental conditions of all social life» (1964, p. 70). In every society, despite the existence of normative rules, there will be crime, although it is not desirable.

Hacking (1990, 1996) is one of the major writers to have traced, historically, the notion of normality in medical and social sciences. For Hacking (1996, p. 61), normal is a «metaconcept, or a second-order concept ... [in] that it does not apply directly to individual things or living beings at all ... until we append a first-order concept, such as 'child'». He goes to suggest that «The adjective «normal» has a clear meaning only in conjunction with a noun phrase: a normal five-year-old» (1996, p. 61). According to him «normal» is both a descriptive notion and an evaluative notion. In the above example, «a normal five-year-old» is both descriptive and evaluative: the features of a normal five-year-old are identifiable as belonging to a group, while being distinguishable from what can be categorised as non-normal. Normality always involves a comparison (Smith, 1978) – a frame of reference against which professionals

and clients describe and evaluate a given state of affairs (see Sarangi and Clarke, 2002a, on the notion of contrast). The typical/usual sense of normal is perhaps more descriptive than evaluative, but the other two senses (e.g., pathological and desirable) are very much evaluative and prone to intervention measures – something which can be made normal through intervention. In what follows I focus on how these three different meanings of «normal» become intermeshed in genetic explanations concerning patterns of inheritance and (non)attribution of diagnostic labels.

3. Formulation of risk and normality in explanations for genetic inheritance

A number of studies in the healthcare setting have examined the interplay of risk and normality: how nurses recontextualise statistical risk information in lay terms for patients to make sense of their own situations (e.g. Adelsward and Sachs, 1996, 1998; Lauritzen and Sachs, 2001) and how midwives have to balance risk talk with reassurance talk when interacting with expectant mothers (Bredmar and Linell, 1999). In the context of prenatal diagnosis, Rapp (1988, pp. 148-149) draws our attention to the technical and invisible nature of the scientific vocabulary of risk, and goes on to suggest that «'statistics' implies an abstract mathematical universe that may not be shared by clients»: «counsellors are caught between the need to sound authoritative and the desire to «glide on the patien's wavelength», as one counsellor described the situation» (1988, p.151).

The notions of risk and uncertainty are intrinsic to counselling about genetic inheritance. Assessment of inheritance risk as 50:50 based on family pedigree is commonplace for many autosomal dominant conditions. In fact, genetic risk explanation centres around the notions of «normal» genes and «faulty» or «altered» genes. The following example is typical of how genetic counsellors explain the cause of genetic disorder. The case involves a parent who is concerned about the genetic status of his two sons[2]:

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2   I will use the following transcription conventions: dots or numerical between round brackets denote pause; texts within double round brackets are glosses; square brackets signal overlaps; equal sign (=) means latching; extended colons stand for lengthened sound; underlined words suggest emphasis, and untranscribable segments are signalled by [^^^^^]. The participants have been anonymised and abbreviated as: GC = genetic counsellor; AM = adult male; PM = parent male; PF = parent female; C/AM = child/adolescent male.

Example 1

01	GC:	you have a normal copy of the gene (.) and an altered copy of the
		gene (.) and the altered copy is the one that's causing the problems (.) .hhhh even
		though you've got a normal copy (.) and (.) for some reason that we not- we don't
		understand fully (.) the problem lies with the fact that you have one altered copy of a
		gene (.) that's what's causing the problem. the- but- (.) the problems basically (.5)
		.hhhhhhhhhh that means that ehm (.) because you've got one altered copy and one
		normal copy of the gene you could pass on either copy to your children
02	AM:	=right
03	GC:	=okay (.) .hhhh ehm and if they- they had the <u>altered</u> copy then
		they would- develop the condition themselves (.) .hhh so they are at eh a one-in-two
		chance (.) basically
04	AM:	fifty-fifty (.5)
05	GC:	like tossing a coin (I don't know) it's completely random. (.5) I mean
		whether which gene they- which copy of the gene they inherit (.) .hhhhh and so it
		could be that neither of them have inherited the altered copy, one of them has or
		both of them have and we- you know there's no (.) no rules to that basically
06	AM:	=right (.) (tats) is there any way of telling ^^^^^^^^^^
07	GC:	=right (.) the:::::::re (.) we <u>are</u> able to do (.) to look at the altered- to
		look at your genes and look to see if we can find any alterations in it (.) .hhhh ehm (.)
		and if we can find alterations, if we can find the alteration in <u>your</u> gene,
08	AM:	=mmh
09	GC:	= then we could check your children as well

We can notice in the above how genes are labelled as normal and altered[3]. This kind of labelling is deliberate to the extent that it does not identify the person as diseased or being ill. The carrier of a faulty gene is also a carrier of a normal gene. So, s/he is susceptible to certain genetic disorders, although there are no current symptoms to warrant this. It is the simultanous co-existence of the normal gene and the altered gene in their presymptomatic state that is given as the cause of future uncertainties (turn 5). A possible, though minimal, intervention is the offer of a test (turns 7-9). The disclaimer – «we don't understand fully» (turn 1) – prepares the ground for the counsellor to announce various possible scenarios that defy a definitive explanation of what might happen (turn 5). When it comes to inheritance, the carrier «could pass on either copy to children» or «neither of them have inherited the altered copy» or «one of them has», or «both of them have». Uncertainty is central to this prognosis («like tossing a coin») and «there's no rules to that», «it's completely random». Against this climate of uncertainty, a sense of risk is not only implied, but it is upgraded as «fifty-fifty» or «one-in-two chance». Disclaimers about expert knowledge (turns 1, 3-6) and assessment of probability go hand in hand with uncertainty about future risk (Sarangi and

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3   Other terms used to refer to faulty or altered gene are mutant, mutated, bad copy, pathological, disease-causing, etc (Clarke, personal communication).

Clarke, 2002b; Sarangi, 2002). However, GC is still able to formulate his expertise in announcing what might be regarded as a «normal» risk figure of fifty-fifty or two-in-one[4]. A relatively more definitive statement about inheritance patterns can only be made after the testing of AM and his children for exact markers and alterations.

Although the notions of risk and normality are a common thread in genetic counselling, these terms may not figure explicitly in many sessions. This can be seen in the following example concerning a mother with a history of miscarriages who wants to test her son for the faulty gene:

Example 2

PF: obviously he's totally unaware of the miscarriages or anything like that anyway (.) ehm:::::
        (.) its just to see what the risks (.) well how do I say well not really the risks what's the
        word I'm looking for (.) what the
        chances of him being a carrier are (.) you know what the odds of him being a carrier are
        really

Implicit in the mother's preference for words such as «chance» and «odds», instead of «risk», is a notion of normality. The son in question is normal now as far as his genetic status is concerned. He can even remain normal if tested positive as a carrier, although there will be a 50/50 «chance» of his transmitting the gene to his children. His current lack of awareness about his mother's miscarriages also helps to sustain a normal state of affairs in the family, which does not require a genetic explanation. Clinical expertise in these circumstances involves making decisions about (not) offering genetic tests for diagnostic, carrier or predictive purposes.

4.   (Non)attribution of normality/abnormality in genetic diagnostic reasoning

Typically, patients/clients come to genetic counselling with a referral from their GPs (General Practitioners), although there exists the possibility of self-referrals. The referral letters can be vague and unspecific, so the opening phase of many genetic counselling sessions is devoted to clarifying the clients' purpose of attendance and their main concerns. A GP's role in the referral process is routinely talked about: GPs are generally characterised as someone who have detected something «abnormal» in their patients but they

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4   This reading of «normal as typical» is based on substantial ethnographic work in the clinic, where
      both counsellors and clients display their basic understanding of genetic inheritance risk figures
      as 50:50. On many occasions, clients orient to this risk figure as given knowledge and seek more
      specific assessments of their situation in light of other intervening variables.

have held back a genetic explanation for such «abnormal» state of affairs. Let us consider some examples of how GPs» concerns are formulated in the opening phases of genetic counselling.

Example 3

GC: Dr X has asked you to come here because he was a little worried about CF's speech

GC: Dr Y has given me a description of CM's problems

GC: your family doctor sort of (.) prompted that 'coz we hadn't done it yet (1.0) and we (1.0) uh (1.0) and (.) he was (1.0) u:h (.) really asking (1.0) u:h (1.0) to see if we'd come to any (1.0) diagnosis as to the cause of ((C/AM's)) problem (1.0) and t- to see if we'd any (1.0) chromosome tests done (1.0)

In all the formulations above, GCs acknowledge the expertise of GPs as far as problem identification is concerned. In formulating certain phenomena as «problems», a notion of normality is presupposed. This is a first step towards medicalisation, which can then be followed by classification (e.g., a genetic diagnosis if available) and intervention (e.g., a predictive test)[5]. Although the GPs in question have been able to notice problems, they fall short of dealing with them (in terms of classifying and treating the problems) and so refer their patients for the specialist's attention. As we can see, GCs tend to characterise GPs» understanding of the genetic make-up of the problems as rather limited. Expressions such as «he was little worried», «he gave me a description», «he was really asking» imply a sense of uncertainty as well as a lay understanding of genetics on the GPs' part, which inevitably prompted the referral to genetic counselling.

The final episode above extends as follows and this is the consultation I shall analyse in detail in the rest of the paper[6]:

Example 4

01   GC: [it] was the fact that the GP wrote to us (.) was that because (1.0) something had happened to [trigger that]
02   PM: [no that] was because um (1.0) she knew somebody who presented with similar problems to ((C/AM)) [(1.0)] and I be:lieve she sort of=
03   GC: [right]
04   PM: =tried to put two and two together and say well I know a young lad who's got (0.6) very similar (0.5) tendencies to this other person who's got Prader-Willi (who had it) and they wanted to do a a double=

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5   More generally, de Swaan (1990) refers to the process of classifications and conceptions of troubles as problems as «the professionalisation of the client». He also draws our attention to the phenomenon of «protoprofessionalisation» whereby certain types of clients are able to appropriate the professional stance and jargon (see also Sarangi and Slembrouck [1996] for parallel notions of «professional clients» and «expert clients»).

6   I am very grateful to Lucy Howell for access to the transcribed data for this case study.

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05   GC:   [right]
06   PM:   =check I presume
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Here PM attributes to the GP some form of expertise in noticing a problem pattern – perhaps simply by association («she sort of tried to put two and two together», turn 4) – which required further medical investigation and a specialist explanation. PM's remark «they wanted to do a double check I presume» (turns 4-6) serves as an euphemism for referral to this specialist clinic. The clinic is attended by C/AM – who is a male adolescent of eighteen years of age with developmental problems – his parents (PM and PF), the GC and the Nurse (N). C/AM had undergone a test earlier which showed no sign of Prader-Willie[7], but in a separate assessment outside genetics, he has been diagnosed as being autistic. It is this diagnosis which has prompted the current visit to the genetics clinic.

Example 5

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01   GC:   there's a a test that's been developed since then (0.5) that is
            probably slightly better (0.5) not hugely better (.) but a bit better (0.5) and (0.5) so
            (0.5) if that (0.5) I (.) mean I I don't I don't think ((C/AM)) has Prader-Willi (0.5) I
            mean just from (1.0) uh [from his] from observing him and his =
02   PM:   [observing him]
03   GC:   = features and so on so (.) so I probably wouldn't have [(1.5)
            gone =
03   PF:   [no I think uh the main thing that came into it] was that small
            hands =
04   GC:   = back to that]
05   PF:   = (.) small feet which I believe is (.) one of the [symptoms (.) yes
            =
06   GC:   [that's that's yes (1.0) yeah]
07   PF:   = (1.0) you] know and uh (.) he's quite a big boy and [(1.0)] (we
            feel =
08   GC:   [yeah]
09   PF:   = it's all to do with it) [(.)] yeah so uh like I said it seemed to be
            all =
10   GC:   [yes]
11   PF:   = there [(.)] and um
12   GC:   [right] yeah I think it's worth [(1.0) it's] worth checking out (.) as
            =
13   PF:   [yeah we wanted it checked out]
14   GC:   = fully as as possible
[6 turns omitted]
20   GC:   so we can get it done very quickly anyway even if it hasn't been
            done [(0.6)] and then let you know what that shows =
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7   Prader-Willi syndrome is a condition characterised by floppiness in infancy and then delayed development, learning problems and excessive appetite leading (often) to severe obesity. Genetic testing for this condition is now highly accurate, whereas in recent past only 60-65% of cases could be confirmed on laboratory testing.

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21  PF:   [right]
22  GC:   = but (.) but I don't (.) I don't I really don't think he does have it
          (1.5) and the test we did last time would have shown most people with it (.0 (would
          have shown up) (.) mostly (0.6) um (.) and and it (.) y- know so (.) but it it's worth
          (0.6) [checking out these things further as bet- =
23  PF:   [oh yeah (.) (something on your mind get it checked out)]
24  GC:   = as better tests come]
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We have here the different senses of normality played out by the parents and
the GC and this is accomplished by drawing upon their different domains of
expertise. In turns 4-5, PF resolutely claims that the small hands and feet are
symptoms which are need of a diagnostic classification (see turns 9-13, «it's
all to do with it», «it seemed to be all there», «we wanted it checked out»).
Note that PF also goes on to characterise the small hands and feet as
abnormal in light of C/AM being quite a big boy (turn 7). So, we have here two
layers of description and evaluation: C/AM is quite a big boy in comparison to
other boys in his age range and that his hands and feet are small in light of his
big physique. In other words, the «small hands and feet» and «he's quite a big
boy» are regarded by parents as not normal in the typical/usual sense (turns
3-7). GC takes these descriptions into consideration but initially hesitates to
upgrade these as clinical evidence for a diagnosis or as a basis for inter-
vention (in the sense of offering the new test)[8].

For the parents (PM and PF), a new developed test is likely to be seen as an
integral part of an expert system and so a further test result would appear the
normal way to proceed. For the genetic counsellor, though, a laboratory test is
one of many possible ways of verifying symptoms – an important factor being
the clinical anatomy of the smallness of hands and feet which can legitimise
the administration of a test. It is worth noting GC's clinical stance here which is
formulated as «from observing him and his features and so on ... I probably
wouldn't have gone back to that» (turns 1-4). In turn 22, GC makes it clear that
«the test we did last time would have shown up most people with it» if there
was a problem. Here «mostly», «most people with it» imply the usual/typical
sense of normal. In contrast with the parental normalisation and desirability of
the new test, GC seems to be normalising the earlier test and the earlier test
results. This reluctance on GC's part is legitimated through his clinical
assessment of the presenting physical problems which do not fit the Prader-
Willi syndrome. Overall, we notice that what the parents take as usual

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8   For many genetic conditions, the clinical assessment (based on physical traits, pedigree details
     etc) may or may not be confirmed by laboratory-based genetic tests. This may explain why GC
     agrees to go along with a new test even if he has doubts about a diagnosis of Prader-Willi.

symptoms for Prader-Willi is discounted by GC in this particular instance in that they fall short of a genetic explanation in the clinical sense: «I don't think he does have it» (turn 22). Despite this absence of clinical evidence, however, GC is happy to go along with the new laboratory-based test as part of routine procedure (cf. turns 12 and 22-24).

The parents continue with their case:

Example 6

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01  GC:  when I was going back (1.0) through things (.) I think (.) I think
          it was (1.0) doctor ((name)) (.) you very first saw in ninety four who
          noted down something about club feet
02  PM:  that was virtually as soon as he was born when uh (.) to be quite
          honest people didn't know what they were on about in those days (.) as
          far as ((C/AM's)) concerned [(.)] they didn't know for the first month (.)
          =
03  GC:  [right]
04  PM:  = that his knees were (.) all wrong
05  PF:  he had plasters put on his feet as s- th- the day after he was born
          (.) to try and [(1.0) fetch them round] you now (.) and he's
06  GC:  [correct the shape of the feet] okay but the but the plastering (.)
          was all because of the (1.0) or the
07  PM:  because when he was born we'll go right back to the start [when
          he] =
08  GC:  [yeah]
09  PM:  = was born (1.0) he was born (.) and he presented with two
          deformed (1.0) legs and feet (2.0) in other words they were turned round
          backwards [(1.0)] they put him in plaster (.) to try and fetch his feet =
10  GC:  [yeah]
11  PM:  = (1.0) into a normal position [(1.0)] and then they realised that
          [(2.0)] also (.) his knees were (.) deformed as well (.) but they =
12  GC:  [yep] [it wasn't the feet]
13  PM:  = didn't realise that straight away [(1.0)] so they carried on
14  GC:  [right]
15  PM:  = correcting his feet (1.0) and after they got his feet round the
          right way (.) they then started on his knees
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GC opens this sequence with an explicit reference to the diagnostic entry in the medical records («the doctor you very first saw in ninety four who noted down something about club feet», turn 1). In the following turn PM challenges the expertise of the doctors who saw C/AM when he was a baby and then goes on to formulate an extreme case scenario (Pomerantz 1986): that C/AM's «feet and knees were all wrong» (turn 4), «he presented with two deformed legs and feet» (turn 9), «they were turned round backwards» (turn 9). Embedded here is the usual/typical sense of normal, i.e., C/AM's feet are reported as unusual/untypical. However, C/AM's abnormal knee and feet positions are also formulated in a pathological frame, which necessitated intervention so as to put them right. PM recruits the voice of the medical

profession to endorse this pathological reading. Note however that he refers to the doctors as «people [who] didn't know what they were on about in those days». In a sense, the medical expertise is undermined and the intervention is represented in unspecific terms when formulated as «try to fetch them [his feet] around» (turn 5) as compared with GC's more exact formulation:»[to] correct the shape of the feet», turn 6). This intervention is evident in the «plasters put in his feet» in order to bring them «into a normal position» (turn 11). The dichotomous pathological/normal framing is accomplished through a carefully planned premise/consequence pair (turns 7-11):

> When he was born he presented with two deformed legs and feet. In other words they were turned round backwards.

> They put him in plaster to try and fetch his feet into a normal position.

The discussion then shifts to C/AM's more recent physical problems.

Example 7

01	GC:	so about (.) ((C/AM)) physically (.) is there anything else (.0) any other problems he's had anything else that (1.0) um [(1.0) in the last few =
02	PM:	[no it's just that his main problem] his main problem is he's put on =
03	GC:	= year]
04	PM:	= so much <u>weight</u> (1.0) but when you (1.0) I'm not trying to make <u>excuses</u> for him but if you like me to already told you (.) in the last three years he's broken his femur (.) every [year] [(1.0) well if you're]
05	GC:	[mhm]
06	N:	[(^^^^^^ exercise)]
07	GC:	several months without exercise
08	PM:	exactly yeah (1.0) and there's nothing (.) else to do (.) other than (.) <u>eat</u> basically
09	GC:	yeah
10	PF:	and he can't do a lot of exercise (.) right now (.) but uh (.) he's when he's in plaster for four to six months at a a time just sitting [(0.6)] you =
11	GC:	[yeah]
12	PF:	= know it's just the weight has piled on (1.0) you know and =
13	GC:	= yeah
14	PM:	<u>that</u> is our main concern at the moment is his (.) his weight (.) coz obviously (.) he's like I say he's eighteen and a half he's eighteen stone and that (.) can't be good to (.) anybody [(1.0)] but he can't exercise
15	GC:	[no]
16	PF:	the worst part I find is that it's (.) gathered really on his chest
17	N:	does he get breathless at all
18	PF:	<u>no</u> he's pretty good (.) you know
19	PM:	he doesn't get breathless or anything because he's unable to (1.0) run he can't run (.) so he won't get out of puff like that [(.)] um he (2.0) can't =
20	N:	[no]

GC's question in turn 1 – «is there anything else any other problems he's had» – and the subsequent response by PM in the negative in turn 2 point to a restatement of what constitutes the «main problem». It is apparent that PF and PM appeal to everyday reasoning in order to explain C/AM's overweight: the cause is lack of exercise for several months (turn 7) and this lack of exercise is caused by the fact that «he's in plaster for four to six months at a time» (turn 10). This has a normalisation flavour to it which is achieved through a strategy of generalisation: gaining weight under these circumstances can happen to any one (see the use of the second person pronoun «you» in turn 4, and the formulation «that can't be good to anybody» (turn 14) when referring to C/AM's current weight). In fact, the expression «the weight has piled on» (turn 12) captures the law-like inevitability of the overweight situation. From the parental perspective, this accounting does not lend itself to a possible pattern of genetic inheritance/diagnosis, although small feet and hands were earlier offered as essential candidates for Prader-Willi (see example 6). However, C/AM's weight (eighteen stones) at his current age (eighteen and a half) is announced as a matter of concern and this prompts the question about breathlessness from the nurse (turn 17). N's query about breathlessness may be seen as a first step towards the medicalisation of C/AM's excess weight, which may or may not lead to a diagnostic label and/or treatment. As far as PM and PF are concerned, the overweight is posed as neither normal, nor pathological – in the sense that no medical intervention is necessary. The assumption is that if C/AM were to do routine exercises again, the problem will subside. What is striking though is PF's mention of the weight gathered on C/AM's chest (turn 16). The implication here is that even in comparison with other overweight children of C/AM's age there is something unusual about the distribution of fat. The deviation from the «normal» is thus accomplished through a comparative frame of reference, first implicitly in relation to other eighteen-and-a-half-year-olds, and now explicitly in relation to distribution of fat in C/AM's body. This prompts GC to undertake a physical examination.

My final example below follows immediately after the physical examination.

Example 8

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01   GC:   right (1.0) well (2.0) I think the (.) I know the tests we had
           before (1.0) failed to show any chromosomes problem or Prader-Willi
           syndrome and the (.) the newer test he's just had done (1.0) uh (.) also
           does not show the Prader-Willi [(1.5)] u:h (.) so (.) so we don't have a
           (.) proper =
02   PF:   [mhm]
03   GC:   = explanation for why ((C/AM)) has the problems he has
04   PF:   mhm
```

05 PM: are we talking about physical problems or [mental problems] (.)
            both
06 GC: [both] I suppose I'll be looking (.) what
07 PM: 'coz like I said he (1.0) th- this team as I explained to you in
            there (1.0) they have come up with that he has [(1.0)] autism (.) but he's
            in =
08 GC: [has autism]
09 PM: = the the lighter area of it he's not in the (.) apparently it works
on a scale from black to sort of (1.0) through grey into cream and then
            into (.) to [white] (1.0) and he's sort of at the top end
10 GC: [right] right I (1.5) I suppose I'd look on (.) um autism as (.) uh a
            way of describing a pattern of behaviour [(1.5)] which you could have
            for =
11 PM: [mhm]
12 GC: = lots of different reasons (1.0) so (.) uh (.) so it's a it's a
            diagnosis in a sense (.) from the point of view of people looking at his
            behaviour (1.0) but it's not an explanation for (.) why he's (.) got that as
            a pattern of behaviour (.) if that makes sense so it's [(.)] yeah
13 PM: [mhm] but it does (1.0) help [((PF))] to a certain extent that now
            when =
14 GC: [sure]
15 PM: = people when she's out shopping or something [(.)] um [(.)]
            people =
16 N: [mhm]
17 GC: [yeah]
18 PM: = tend to look and if you go (.) you know (1.0) what's the matter
            or have you got a problem they say well what's the matter with him like
            [(.)] at least now you can turn around and say [(1.0)] he [has] (.) =
19 N: [yeah] [yeah]
20 GC: [he has this]
21 PM: = so and so (.) whereas [before there's] well (.) sorry we don't
            know
22 N: [(it's a name) mhm
23 GC: yeah (1.5) yeah I s- I s- if say Prader-Willi had been found (.)
            then I suppose we'd be saying that he (.) had autism (.) in association
            with [(.) Prader-Willi]
24 PM: [yeah yeah] which makes it easily easier for ((PF))
[12 turns omitted]
36 GC: I mean I'm not in a position to add to that if I can give (1.0) a (.)
            an extra diagnosis as well [(1.0)] to uh account for it (1.0) th- there are =
37 PM: [mhm]
38 GC: = a few things (1.0) um (1.0) you know examining him then that
            (.) I suppose I h- hadn't been (1.0) aware of before (.) as as with his breasts and so
            on (1.0) so (1.0) um (1.0) I mean I'll (1.0) (1.0) think with one or two colleagues
            about (.) could some of those physical (.) problems he's had with the knees and the
            ankles and (.) um some of the other things that have developed since could they fit
            into a pattern at all (1.0) uh (2.0) and if (.) uh (1.0) hhh. If we are able to think of a
            pattern they fit into (.) then I'll get back in touch over the next couple of months

At the start of this episode, GC confirms his earlier suspicion that C/AM does
not have the Prader-Willie syndrome. His clinical judgement is now being
supported with laboratory-based evidence, i.e., results from the new test (see

example 5 above). In the rest of the interaction there is some demystification of cause and symptoms. PM recruits the voice of other professionals to characterise the exact nature and degree of autism C/AM has: «he's in the lighter area of it», «he's sort of at the top» (turn 9). Autism is conceived of in terms of a scale, and this allows for an exact specification of C/AM's condition in relation to other autistic children/adolescents. In turns 10 and 12, GC acknowledges the diagnosis of autism, but he goes on to separate autism as a recognisable «pattern of behaviour» from possible explanations «for why he's got that as a pattern of behaviour». Although C/AM's parents had come to see if the diagnosis of autism could be backed up through a genetic explanation, GC makes it clear that they are two separate things: «if say Prader-Willie had been found then I suppose we'd be saying that he had autism in association with Prader-Willie» (turn 23). It is worth noting the hypothetical passive construction «if Prader-Willie had been found» which points to the outcome of laboratory-based test results, but it also underlines his suspicion about C/AM having Prader-Willie on the basis of presenting clinical evidence.

GC's explicit mention of «his breasts» in turn 38 is particularly significant here. What the parents had referred to as excess fat on C/AM's chest (see turn 16, example 7), GC reinterprets (following the physical examination) as breast tissues at an earlier point in the interaction (not cited here). This may be seen as a pathologising stance which can lead one towards a possible diagnosis. The development of breast tissues in men, for instance, can be taken as a marker of genetic predisposition towards Prader-Willie. We see features of uncertainty and disclaimer of expertise on GC's part in turn 38. Individualised expertise gives way to collegial brainstorming – to move away from causal explanations towards possible associations and thinking about a pattern – which is somewhat different from the GP's putting «two and two together» in a kind of guesswork (see example 4 above). Normality is to be conceived of in terms of shared patterns of physical and mental conditions and such deliberations need to have the endorsement of a community of practice. Noticing a pattern and attributing it a label does not necessarily constitute expertise. Indeed the term «pattern» can be problematic. In the general medical context, a noticeable pattern of behaviour does not routinely call for a genetic causal explanation. Within genetics, however, patterns may be looked for within a syndrome and at the level of what is called «genetic expression».

A further aspect to normality – which is explicitly alluded to by the parents in this example (turns 13-22) – relates to what counts as normal in the social realm[9]. As far as shopping and other outdoor activities are concerned, abnormality of any sort becomes a normal object of public gaze. Therefore a diagnostic label for C/AM is what is required to normalise PF's interactions with strangers in these circumstances: at least when «PF's out shopping» she can «turn around and say he [C/AM] has this» (turns 18-20). A diagnostic label is being actively sought by the parents which would constitute an essential part of C/AM's social identity. So any reluctance on the part of the profession to give a diagnosis ultimately denies the patient (here C/AM) and his parents (PM and PF) «normal» participation in the sphere of social interaction. A medical diagnosis and explanation is clearly regarded by PM as desirable in preference to saying «sorry we don't know» (turn 21).

At one stage in the consultation (not shown here), the discussion also covers C/AM's academic performance in the new school. Both parents are pleased with what C/AM has been able to achieve in the new school – especially learning to read and write better than what he was doing in the previous school. According to PF, this progression is accomplished through a contrast device: «because it wasn't presented to him correctly before in a way that he could understand it». Implied here is a blaming of the old school which did not intervene correctly. In other words, it carried out its teaching «in the normal way» (in the usual/typical sense) rather than targeting it to the needs and abilities of C/AM – which perhaps required an orientation in a pathological sense. As far as the parents are concerned, the new school is more interventionist in comparison with the old school in bringing about «normal» (in the sense of «desirable») educational outcomes. We have here the use of «normal» in the sense of what the specific expectations should be for a non-normal group as opposed to the imposition of external «norms» on an already disadvantaged group.

5.   Conclusion

Categorisation of illness – with or without accompanying treatment – appears to be the hallmark of the connection between medical practice and social practice, and more generally, between the biomedical order and the social

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9   In genetic counselling, it is possible to distinguish between three different but inter-related realms – personal health, reproduction choices and social relations – where normality applies in different degrees (Clarke, personal communication).

order. The premises upon which normality and deviance are categorised can manifest themselves as potential tensions both within and across the healthcare profession and between healthcare professionals and their clients. From a professional standpoint, a proper diagnosis is desirable in the management of the normal biomedical order (so that the condition can be treated and the person can function normally). From clients' perspective, a proper diagnosis is also of vital importance for undergoing treatment, and in the absence of such treatment options at least for the maintenance of the social order (so that the parents can go shopping and respond appropriately to strangers' gaze).

Another tension in the interpretation of normality which my analysis has pointed to has to do with how professional and parental perspectives may not only differ in degree but also in kind. For clients, «normality» is mainly a first order concept (see my earlier discussion of Hacking in section 2): they interpret small feet and hands as direct evidence for absence of normality. Whereas for genetic counsellors, normality remains a second order concept in that small feet and small hands need to be assessed in relation to what is or is not categorisable as candidate features of the Prader-Willie syndrome. This clinical mentality then accounts for uncertainty in diagnosis – unlike the clients looking for a dichotomous normal/pathological explanation of the presenting «symptoms». It is evident that geneticists' explanations about inheritance and diagnosis are different from what clients often expect about genetic conditions. This tension can be extended to account for the differences between geneticists and GPs-as-lay-geneticists: while GPs can notice and identify different patterns of behavioural or anatomical phenomena, it is the geneticists who have the expertise to offer an explanation of why something is or is not genetically linked.

It seems that, in genetic counselling, unlike other medical encounters, normality is very much used in the sense of typical/usual rather than in a pathological sense. The moral sense – in terms of guilt – is a significant matter in cases of reproduction decisions – which I have not attended to here. It is the presence or absence of a genetic explanation which becomes a marker of difference in the way normal/abnormal is accounted for by genetic counsellors vis-à-vis other parties.

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