Journey into Genes: Cultural Values and the (Near) Future of Genetic Counselling in Mental Health

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28.1 Introduction

Our understanding of the role of genetics in individual vulnerability to mental ill-health and response to treatment is increasing every day. Correlatively, the cost of genetic testing[1] is decreasing and will soon reach the point where an individual's genetic profile will become a normal part of doctor patient consultations. Not only that but it is now possible to buy for around £100 a range of kits that purport to offer DNA[2]-based personalised health solutions, thus theoretically empowering people to take more control of their health-related decisions.

Although not yet routinely part of everyday health consultations, these developments in the science of genetic medicine are sufficiently advanced as to have become the subject of anticipatory health planning in many parts of the world. In the UK, they are the subject of a recent (2019) Government Green Paper,[3] 'Advancing our health: prevention in the 2020s' [1]. This sets out a clear agenda for genetics and

[1] The term genetic testing covers a range of techniques designed to identify individuals' unique genetic make-up and how this might relate to their risk of particular conditions and their likely responses to treatments.

[2] DNA is an abbreviation for deoxyribonucleic acid. This is the molecule that contains the genetic code of organisms.

[3] A Green Paper is a consultation paper circulated for comment before moving to specific proposals in a White Paper and from there to implementation (in some cases including parliamentary processes of producing enabling legislation).

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D. Stoyanov et al. (eds.), *International Perspectives in Values-Based Mental Health Practice*, https://doi.org/10.1007/978-3-030-47852-0_28

Table 28.1 Extract from the Government Green Paper (2019) 'Advancing our health: prevention in the 2020s'

In the 2020s, people will not be passive recipients of care. They will be co-creators of their own health. The challenge is to equip them with the skills, knowledge and confidence they need to help themselves
We are: • Embedding genomics in routine healthcare and making the UK the home of the genomic revolution • Reviewing the NHS health check and setting out a bold future vision for NHS screening • Launching phase 1 of a predictive prevention work programme from Public Health England (PHE)

Table 28.2 Patient and public engagement in psychiatric genetics: a summary of the Mental Health Foundation Research

Patient and public engagement in psychiatric genetics—Mental Health Foundation research
Aim: Explore thoughts, feelings and opinions of psychiatric genetic testing and any personal insights into this subject area. Psychiatric genetic testing has the potential to develop more personalised treatment, yet there are many ethical considerations
Ethical considerations: Access to information (e.g. police, employers, family), how much information individual receives, length information is stored for and potential mental health impact of testing (e.g. if person receives result of higher mental health vulnerability will information enable them developing said mental health difficulty, also potential impact on family and family choices)
Participants: Individuals ranging in age, ethnicity and gender with lived experience
Method: Qualitative, interactive discussion groups discussing three subject areas: Psychiatric genetic testing; mental health impact of non-psychiatric genetic testing; and pharmacogenomics
Results: Indicates how personal the decision-making process is for genetic testing and was mixed in views of for and again such testing. Many factors were considered, however there was unanimous decision that the choice for whether you get genetic testing should remain a free choice and not something imposed upon you. Research like this is important as it puts the voice of lived experience at the forefront, which is crucial for developing future mental health treatment options

genomic medicine to become integral to public health within the coming few years. An extract from the Green Paper illustrating the level of commitment already being made to this agenda is given in Table 28.1.

The following case narrative illustrates the impact of these anticipated developments on mental health through an imaginary consultation set in the near future when the aspirations of the 2019 Green Paper have become incorporated into everyday practice, it is also informed by the Mental Health Foundation's on-going research summarised in Table 28.2.[4]

The consultation is of course fictional, but the scientific developments assumed by those concerned are all within the reach of current technologies—so the case

[4] Further details of this research are given in the Guide to Further Information at the end of this chapter.

narrative is fiction but not *science* fiction. The science is indeed real and (almost) upon us. Note however that the actual risk and probability scores used are illustrative only and should not be read as actual scores for the conditions or treatments mentioned.

28.2 Case Narrative: *'Come In and Sit Down'*: A Doctor/Patient Consultation

The patient, Jim Smith, is in his early twenties. He has been experiencing particular forms of visions and voices consistent with a diagnosis of schizophrenia. There is a family history of mental ill-health: this has not been discussed in detail among family members but a grandmother is talked about in a way that suggests she spent a number of spells in psychiatric hospitals. Jim Smith had been referred to a psychiatrist who gave a provisional diagnosis of schizophrenia. This is the follow up consultation that will by then be required under the new personalised medicine policy introduced (so the story line assumes) following the 2019 Green Paper and its subsequent development and implementation in UK health policy and law.

The doctor, Helen Jones, is well versed in psychiatric genetics (as a medical student she did a genetics research project and had been encouraged to follow a career in this field). After inviting the patient to 'come in and sit down', she starts by reminding Jim Smith of his diagnosis of schizophrenia based on reported and observed symptoms. She then outlines the established intervention recommended by NICE Guidelines[5] *for schizophrenia and reminds him of the informed consent process. She explains that the recommended first treatment achieves the desired therapeutic effect for approximately one third of people with this diagnosis; has a marginal therapeutic effect with some adverse reactions for another third; and has no therapeutic benefit and significant adverse effects for the remaining third. Only through trial and error can the best therapeutic outcome with the minimal adverse impact be established; the process could take many months; and there is no way to tell in advance which of the various treatment options will do good or harm until the good or harm has been done.*

Under the personalised medicine policy (now newly introduced), she then offers Jim Smith the option of his genome being sequenced. If he accepts this, Dr Jones explains, it will entitle him to an enhanced personal treatment plan (EPTP) based on his genotype. Although the EPTP is free it is contingent on enrolling into the UK National Biobank Programme (see Guide to Further Information), a project that

[5] NICE is an independent body, the National Institutes for Health and Care Excellence, set up by the UK government to commission regular reviews of treatment options for different conditions and, on the basis of these, to publish evidence-based treatment guidelines.

aims to improve the prevention, diagnosis and treatment of a wide range of serious and life-threatening conditions with a view to improving personalised medicine.[6]

Dr Jones goes on to explain the role of the Biobank to Jim Smith, spelling out that the more complete the Biobank becomes, the more of the population it is able to include, the more effective it will be in meeting its aims. She explains that on current best evidence an EPTP for schizophrenia improves the likelihood of therapeutic benefit to 80% and reduces the risk of significant adverse effects to about 5% (as noted above, these figures are at the time of writing fictitious and used for narrative purposes only).

There are two further considerations to bear in mind, she continues, before reaching a decision about genomic screening: first, how much more health information (i.e. *over and above likely response to treatment) does he (Jim Smith) want to be reported and shared, and with whom; and, second, bearing in mind the fast moving nature of the field, how far would he be happy with possible developments in the use of Biobank data currently being considered, for example by academic researchers, the police, credit agencies, insurance companies, DVLA, employers, and benefits agencies.*

28.3 Discussion

In this discussion, we review the above consultation from the perspective of the values issues arising (individual and cultural) and how these interact with the scientific and medical advances assumed to have taken place. The discussion is in part informed by our experience with a small series of discussion groups (see Acknowledgements) exploring patients' perspectives emerging practice in psychiatric genetics. Although the details are still being analysed, one clear message from these discussions is the individuality of personal values. For example, some people in the discussion group were very clear that they would not want to know anything about their genetic make-up or any risks, whereas others would want to know everything.

28.3.1 Developments in Psychiatric Genetics

Other than for the specifics of the risks mentioned, the above consultation draws on established findings of the growing field of psychiatric genetics. To the best of

[6] Terminology is sometimes confusing here. The term 'personalised medicine' is generally used to mean medicine that is geared to the particular biological make up of the individual – their genetic profile is particularly important in this respect, we have coined the term "Enhanced Personal Treatment Plan" to reflect this significant increase in how individual treatments may become. 'Person-centred medicine' is different in that it focuses on the particular needs, wishes and expectations of the patient. The 'person-values-centred care' of values-based practice focuses particularly on the values of the patient as an aspect of person-centred care (see chapter "Surprised by Values: an Introduction to Values-based Practice and the Use of Personal Narratives in this Book").

current knowledge, our genetic make-up really does contribute to our risk of being affected by a range of health conditions, including, within mental health, many of those widely described by contemporary psychiatric diagnostic categories. These include, as in the above narrative, Jim Smith's diagnosis of 'schizophrenia' based on the presence of a number of specific forms of experiences (specific forms of 'visions and voices').

Psychiatric diagnostic categories of this kind are not universally accepted not least within the contemporary neuroscience research community (see also Sect. 4, below). Yet notwithstanding the likely scientific limitations of these diagnostic concepts, there is growing evidence of a genetic contribution to individual differences in how people respond to established treatments for conditions so defined. This is why 'pharmacogenomics' as it is called (basing drug treatments on an individual's genetic profile) is becoming an ever more important aspect of personalised medicine.

28.3.2 Scientific Solutions to Values Issues?

On first inspection, the above advances in psychiatric genetics might seem if not to resolve at least to ameliorate the values issues raised by treatment choice in Jim Smith's story. In the first part of his consultation, he is presented with three possible outcomes for the NICE recommended first line treatment for schizophrenia. In deciding whether or not to consent to this recommended treatment, he is in effect asked to take a gamble on the balance of risks and benefits that he will experience (depending on which of the three risk groups described by Dr. Jones he turns out to fall into). Then, in the second part of the consultation, he is offered access to genetic testing that will greatly reduce his gamble. It will give him (in the fictitious numbers given in the story) a large degree of certainty about his likely response to treatment.

Not only that but, to extend the story a little (though again not beyond the immediate future of genetic testing), Jim Smith's test results may well resolve further questions he may have had about specific risks. For example, one recognised risk of the treatments in question is weight gain. This is an outcome of treatment that (carrying as it does a range of aesthetic and health implications) is valued differently by different people. In coming to a shared decision, therefore, as the basis of contemporary best practice and legal rules on consent [2, 3], the risk of weight gain would have to be explored by Dr. Jones with Jim Smith. Such discussion would be better informed with more precise genetics-profile-based information about his likelihood of weight gain to hand.

So, job done? Well, no, because on further inspection, we see that the values issues have been increased rather than reduced by the addition of the option of genetic testing. Certainly, within the terms of reference of the story, genetic testing makes available to Jim Smith better information about the likely balance of harms and benefits of treatment in his case. This is nothing if not extremely helpful both to Jim Smith and to Dr. Jones in coming to a shared decision about how to proceed.

But this information comes at a price. It comes at the price of having to make a whole series of further choices about whether or not to proceed with genetic testing and if so to what extent and for what purposes. For each of these contingent choices, there will be a range of risks and benefits to consider. Thus, in the first place, as Dr. Jones points out, the test is free, but only if Jim Smith agrees to 'sign up' to the Biobank Programme. There are benefits (mainly to others) if he agrees to this but also risks (around the confidentiality of his genetic information now and in respect of future possible uses of the Biobank data by other agencies). In respect of these issues, he, Jim Smith, will have as it were a unique 'values profile' sitting alongside his unique genetic make-up. He will, that is to say, have a unique personal take on what matters or is important to him about these issues. He will have a unique values profile, similarly, about all the further issues concerning what additional genetic information he, Jim Smith, will want reported to him (and/or others) about his own test results over and above his likely response to the proposed treatment for his schizophrenia.

28.3.3 The 'Science Driven' Principle of Values-Based Practice

Jim Smith's story thus illustrates the 'Science Driven' principle of values-based practice, namely, that the impact of advances in medical science and technology is not to diminish but rather to enhance the need for values-based practice as a partner to evidence-based practice in the shared decision-making that underpins contemporary person-centred clinical care (see chapter "Surprised by Values: An Introduction to Values-Based Practice and the Use of Personal Narratives in This Book"). His story shows too why this should be so. For as in Jim Smith's story, the impact of advances in medical science and technology is to widen the range of choices that we (patients and clinicians) have available to us—and with choices go values.

28.3.4 How to Do It

Helen Jones, the doctor in the above narrative, is well aware of the advances in genetic medicine relevant to Jim Smith's condition, and given her research as a medical student, she is better versed in them than many of her colleagues [4]: this is perhaps why Jim Smith's doctor made the referral. Certainly, she makes a good showing of explaining the technical issues to her patient. This as we have seen is not enough for purposes of shared decision-making as the basis of consent to treatment. Nor is it enough, so the evidence suggests [5], for purposes of psychiatric genetic counselling[7] if it is to empower rather than disempower patients.

[7] Genetic counselling is the process of "helping people to understand and adapt, to the medical, psychological and familial implications of genetic contributions to disease" [6]. In the psychiatric context, it involves helping people make personal meaning from what is known about how genetic and environmental factors contribute together to the development of mental health difficulties and using this to frame an enhanced understanding of how to protect mental health. The counsellor will help develop strategies for coping with the risk in family, the related uncertainty of testing and help people living with a mental health difficulty.

What more, then, is needed and how is it to be delivered? We do not have space to consider the challenges of delivery in detail here. But it is important at least to be aware of the resources available. There is a generic resource of training and other materials available to support values-based practice in different areas of health care including mental health (see Guide to Further Information below). There is also, specifically in relation to psychiatric genetic counselling, a growing resource, first, of evidence-based information about what is important to patients about the counselling they receive and the counsellor from whom they receive it, and, second, of practical aids to support delivery. Again, we do not have space to describe these resources in detail but will offer two examples with the above consultation particularly in mind (Table 28.3).

First, then, as to the evidence about what works, Table 28.3 is adapted from *The Empowering Encounter* developed using Grounded Theory to study psychiatric genetic counselling [7]. The factors shown are a small subset of those that make up the full *Empowering Encounter*. But they illustrate the range of factors that are

Table 28.3 What matters to patients in psychiatric genetic counselling

Factor	Example patient quote	Dr Helen Jones
Receiving support and information		
• Being heard	*[The genetic counsellor] let me talk, which is something that I think a lot of people don't do... to be able to talk about what I do [to manage my MI], to be heard and be validated was helpful*	**NO**
• Feeling validated	*I liked that [GC] affirmed a lot of the things that I've been trying to do to manage my mental illness... so its good to know that, like it validates my efforts*	**NO**
• Knowledge	*Until genetic counselling, no one ever coherently explained to me why I have a mental illness. And I think that's a conversation that needs to be had because most people just think they're having a bad time of it or they just think that they just need to try harder and that's [because] they don't understand that it's an illness*	**YES**
• Tool for understanding	*I think that the simplified jar analogy* (see text and Fig. 28.1) *was good, it was easy to understand and was presented in a really good way. I thought it was a useful tool*	**NO**
Characteristics of the genetic counsellor		
• Empathetic	*I just felt that [the genetic counsellor] really understood what I was going through and was really open to connecting*	**NO**
• Non-judgemental	*I didn't feel like [the genetic counsellor] was judging me, I felt like she genuinely wanted to help me*	**YES**
• Knowledgeable	*With [the genetic counsellor] there was a lot more knowledge on her part and [it was] evident that she's heard stories and worked with people who have mental health issues that are the same as I have*	**YES**

important if a consultation in psychiatric genetics counselling is to be empowering for the patient concerned (and thereby effective in motivating health-supporting behaviours). Note that among these factors, giving information and technical competence is indeed highly valued by patients ('knowledgeable' comes up strongly in the *Encounter*—see Table 28.3). But to be effective in empowering patients, giving information competently has to be twinned with a range of what the authors call 'emotional factors', such as (in Table 28.3) the patient feeling that they are 'being heard' and end up 'feeling validated' and that the genetic counsellor is 'empathetic' and 'non-judgemental'.

Understood through the model of values-based practice, the factors that make up the *Empowering Encounter* represent patients' values in the consultation. There will be, as we have seen, further individually unique values that figure crucially in shared decision-making between the clinician and patient. But the factors identified in the *Empowering Encounter* study, representing as they do what matters or is important to patients in the context of psychiatric genetic counselling, are essential as enabling values—they are essential if the counselling is to be effective in engaging with the particular values of the individual patient concerned and hence empower that individual in processes of shared decision-making.

The right hand column of Table 28.3 represents our 'score' for Dr. Jones as represented in the consultation with Jim Smith at the start of the chapter. Her success with the information side of the consultation contrasts with her relative failure in its emotional aspects. And yet as we have seen, both are essential to an effective consultation.

All this of course, as Dr. Jones would no doubt be the first to point out, carries costs—training costs—and, in clinical practice, time costs. These costs we believe should not be overstated. First, there are, at the very least, quick wins to be had. The consultation at the start of this chapter, for example, would have gone very differently if Dr. Jones had introduced herself. Instead of 'Come in and sit down,' she might have said 'Come in and please have a seat, Mr Smith. My name is Dr Jones' (See Guide to Further Information). The time cost of this minimal courtesy would have been negligible and such costs, as there were, would have been amply repaid in terms of enhanced patient engagement and shared decision-making.

Then again, to come to the second of our examples of the resources already available to aid implementation, there are well-validated tools available to support the consultation. The 'metal illness jar' analogy, illustrated in the Fig. 28.1 is a case in point. Derived from practical experience of genetic counselling and developed with input from people with mental health conditions and their family members, the use of this analogy emerged from the *Empowering Encounter* study as a valued resource for meeting the 'understanding' component of an effective counselling experience (see [7]; also Table 28.3).

In offering these brief comments on the resources available to support effective implementation in genetic counselling, we do not wish to be taken to be underestimating the entailed costs. Our point is rather that at least in the case of *psychiatric*

Fig. 28.1 The Mental Illness Jar Analogy (see text)

genetic counselling, there are resources available that make the costs in principle affordable. Which is good news. For as the story of Jim Smith and his doctor Helen Jones illustrates, the further our journey into genes takes us over the next few years, the more will it become important to attend equally to the values-base as to the evidence-base of this promising but increasingly challenging area of mental health care.

28.4 Conclusions

Our aim in setting the consultation between Dr. Helen Jones and Jim Smith at the start of this chapter in the near future was to anticipate our bottom line, namely, that, as an instance of the wider 'Science Driven' principle of values-based practice, advances in psychiatric genetics, far from resolving the values issues involved in shared decision-making, actually make them ever more acute.

We have focussed here on the implications of the Science Driven principle for practice. But it has in, as it were reverse engineered form, implications for scientific and medical research or at any rate for the translation of such research into improvements in clinical care. Translation of research was the subject of a (2014) blog by Thomas Insel [8], at the time Director of the world's largest neuroscience research funder, the USA's National Institute for Mental Health. Responding to widespread frustration at the failure of the neurosciences to translate into tangible improvements in patient care, Insel launched the RDoC (Research Domain Criteria, [9]) as an alternative to the long-dominant American Psychiatric Associations' DSM (Diagnostic and Statistical Manual). Insel was surely right that the DSM is not the last word in psychiatric diagnostic classification. But the message of the Science Driven principle illustrated by this chapter is that if genetics is any guide, the reasons for the translational failures (with which Insel and others are rightly) concerned have less to do with the deficiencies (real or imagined) of neuroscientific research and more to do with a failure to attend to the factors involved in—to

borrow a further phrase from values-based practice (see chapter "Surprised by Values: An Introduction to Values-Based Practice and the Use of Personal Narratives in This Book")—linking science with people.

Acknowledgements The 'mental illness jar analogy in the Fig. 28.1 is based on an illustration by Cindy Campbell-Lashley found in [10]. We are grateful to the Laces Trust for support for the Mental Health Foundation programme exploring patients' perspectives on emerging practice in psychiatric genetics. We would also like to thank our colleagues from the Dragon Cafe for their contribution to this research.

28.5 Guide to Further Information

For more on values-based practice including training resources, please see the website for the Collaborating Centre for Values-based Practice at St Catherine's College, Oxford: valuesbasedpractice.org

Details of the UK Biobank can be found at https://www.ukbiobank.ac.uk/

The 'Hello my name is' campaign website is at: https://www.hellomynameis.org.uk

For further details of the Mental Health Foundation programme exploring patients' perspectives on emerging practice in psychiatric genetics, please see www.mentalhealth.org.uk/our-work/research.

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