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ORIGINAL ARTICLE

Working Up Policy: The Use of Specific Disease Exemplars in Formulating General Principles Governing Childhood Genetic Testing

Paula Boddington · Susan Hogben

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Abstract Non-therapeutic genetic testing in childhood presents a "myriad of ethical questions"; questions which are discussed and resolved in professional policy and position statements. In this paper we consider an underdiscussed but strongly influential feature of policy-making, the role of selective case and exemplar in the production of general recommendations. Our analysis, in the tradition of rhetoric and argumentation, examines the predominate use of three particular disease exemplar (Huntington's disease, Tay-Sachs disease and sickle cell disease) to argue for or against particular genetic tests (predictive testing and testing for carrier status). We discuss the influence these choices have on the type and strength of subsequent recommendations. We argue that there are lessons to be drawn about how genetic diseases are conceptualised and we caution against the geneticisation of medical policy making.

Keywords Genetic testing · Geneticisation · Huntington's disease · Policy-making · Tay Sachs' disease · Sickle cell disease · Stigma

P. Boddington
School of Medical Genetics, Cardiff University,
Cardiff, Wales, UK

S. Hogben
Cardiff School of Journalism, Media and Cultural Studies,
Cardiff University, Wales, UK

P. Boddington (✉)
CESAGen, 6 Museum Place,
Cardiff, CF10 3BG Wales, UK
e-mail: BoddingtonP@cardiff.ac.uk

Introduction

Genetic testing[1] which can help provide a diagnosis identifying the cause of individual's medical condition is generally considered uncontroversial. However, the nature of the genome means that genetic testing can also generate information about an individual's and possibly his/her family's future disease status. Genetic testing can identify people who are currently asymptomatic but for whom there is a likelihood (so-called susceptibility testing) or certainty (so-called predictive testing) that they will develop a condition in the future. Testing may also identify individuals with a gene mutation who will not themselves develop a condition but who can be identified as carriers of a condition and who could have affected children (so-called carrier testing). The benefits of these sorts of genetic tests are much less clear. Whether an adult wants to acquire such information is generally left to that adult but generating such prophetic information during childhood raises, as Ross and Moon ([37], p. 873) (cf. [10, 21], and many others) suggest "a myriad of ethical questions"; questions which a range of policy documents and position statements which form the data for our analysis in this paper attempt to resolve.

Policy (as both a process and a product) has recently become an increasingly important topic for analysis. Particularly, though not exclusively (see [15, 20, 22, 38]) in health care ([24, 39] amongst others), in the area of genomics[2] and to genetic testing in particular [23, 31].

Our paper concerns itself specifically with important methodological points about the formulation of policy recommendations. Our primary focus is how arguments for and against childhood genetic testing are centred on specific disease conditions which are used (we argue) to work up particular recommendations which differ in modality.

The choice of example is significant in itself but crucial too is just how examples are described. Chambers [8] examines the ways in which the description and use of cases in bioethics impacts upon the ethical conclusions generated (cf. Lippmann, 1994). His warnings are of relevance here. In its use and in the particular way an example is described, an example can be seen as a type of case. We show how the ways in which examples are both selected and described impacts upon the formation of policy. By taking such cautions on board, we hope to identify shortcomings and specific biases in the formulation of genetic policy and suggest steps to avoid unnecessary limitations.

Policy as a Topic for Analysis

Policy-making, like many other socio-political practices, is now widely recognised to be "made of language" ([30], p. 1), to be a "thoroughly rhetorical activity" ([40], p. 1) and, as Fischer and Forester ([15], pp. 1–2) appreciate, to be a "constant discursive struggle." A struggle which is as much about defining and creating the issue as it is about resolving it ([35], p. 148); in both cases it is one that depends fundamentally on choice, contrast [15] and framing [34]; each of which selectively organise, interpret and "distinguish some aspects of a

[1] Genetic testing refers to the analysis of a specific gene, its product or function, or other DNA and chromosome analysis, to detect or exclude an alteration likely to be associated with a genetic disorder (Harper, 1997). Testing can be specifically distinguished from screening which is the same practice across populations.

[2] The Human Genetics Commission (The UK Government's advisory body on new developments in human genetics) has recently commissioned an analysis of its own consultative process.

situation rather than others" ([20], p. 45) to provide "a perspective from which an amorphous, ill defined problematic situation can be made sense of and acted upon" ([34], p. 263).

Policy, then, is as much about what is said as it is about what is not said is. Accordingly policy analysis which is centred on the discursive construction of matters can focus on how arguments are structured, how particular factors that writers[3] consider relevant are selectively included or excluded, how these features are downplayed or upgraded, given precedence or silenced, how and what evidence is presented in terms of generalisation and/or exemplar, how potential actions are prescribed and how conclusions or recommendations are modulated and, perhaps most importantly, how these are or are not implemented in practice. In this latter respect we can also see policy-making as interactional, irrefutably dependent on how a reader interprets policy statements or how those texts are left open for interpretation.

It is also important to appreciate that policy-making is not a naïve or neutral activity; it "always takes place within a nested context" ([35], p. 154). That is to say, policy issues emerge in connection with (non)governmental programmes, which exist within a wider policy environment, itself part of a broader political, social and economic setting, which is simultaneously situated within, and a result of, historical practices (cf. [16]). Policy is thus influenced by, reacts to and is constitutive of prevailing sets of assumptions and values (cf. [30]); each of which is accomplished rhetorically.

Examples *in* Argument - Examples *as* Argument

> A main cause of philosophical disease: a one-sided diet: one nourishes one's thinking with only one kind of example. ([42], *Philosophical Investigations*, 593)

One underdiscussed area of policy making and, indeed policy analysis, and one which is of particular interest in ethically troubled arenas, is the use of the exemplar or the case in the generation or working up of recommendations (cf. [8]). The use of examples in the development of arguments and the production of conclusions has long been appreciated to be both key and problematic. Not least because examples are supposed to relate to generalisations in terms of typicality – in terms that is of representativeness ([13], p. 45). But there are problems with matters of both typicality and representativeness.

Examples may be more or less good examples of a category or class. Rosch [36] demonstrates how robins, for many English speakers, are treated as a more typical example of a bird than are penguins; an effect attributed to robins sharing more prototypical properties of "birdiness." The issue of typicality matters greatly when we examine how some disease conditions are worked up to be more representative of particular classifications of disease conditions as the use of examples are not arbitrary . An example will not only "guide the readers" interpretations of situations they judge comparable with those the author investigates: it also directs reactions and evaluations of them" ([13], p. 45).

The use of the example relies on the selective presentation of particular features from which generalisations may be built. An example also can be granted the status as an argument itself, particularly where particular features are presumed to be shared with other types in what Gillam ([18], p. 400) argues are "all morally relevant respects." The selective attribution of what is deemed to be morally relevant is crucial too. In this respect examples, we will argue, need to be understood within the context that gives them salience as cases.

[3] We appreciate that the discursive nature of policy making is multi-modal [20]. We use the term writers here as we are dealing directly with printed documents.

In the context of genetic-testing much research is carried out in disease-specific contexts or considering individual, family or community responses to testing and we might anticipate how in the policy data the illustrative warrants might not unsurprisingly make mention of the impact of testing for specific conditions. With childhood genetic testing it matters whether attempts are being made to generalise from one condition to (ethical) principles or to populations of diseases or how cases may utilise contextual information such as the unique circumstances of each family, the form of social or medical interventions that are available and the particular features of a condition and the values that it may be deemed to share with other conditions factors relating to the inheritance, onset, and morbidity of specific conditions.

Let us now flesh out the policy issues by examining the data in which examples are used.

The Data: Five Position Papers and Their Terms of Reference

Our data consist of five extensive statements produced by medical professionals and patient interest groups concerned with each of these types of genetic testing. These documents, although not regulatory, present policy, principles and guidelines for preferred practice with respect to genetic testing in childhood. The documents under examination are produced by: The UK Clinical Genetics Society (CGS) in 1994 [11]; the UK Genetic Interest Group (GIG) in 1995 [17]; the American Society of Human Genetics and the American College of Medical Genetics (ASHG/ACMG) in 1995 [3]; the American Academy of Paediatrics (AAP) in 2001 [1] and the Canadian Paediatric Society (CPS) in 2003 [7].

A number of other documents provide guidance on practice in this area including papers produced by the American Medical Association [2], Belgian Society of Human Genetics [5] and the National Society of Genetic Counsellors [32] Resolutions Ballot. However, these documents do not report the arguments that informed the conclusions and recommendations that they prefer. Because our analytic focus is on how the recommendations are worked up, on what factors inform the process of argumentation as well as the product of final recommendations we have not been able to include such documents in our analysis.

The issues addressed in the five papers primarily concern predictive testing for child and adult onset conditions and testing for carrier status. Three of the papers reflect on practice with regard to adoption and two papers also pay attention to newborn screening and pre-natal testing.

Each of the papers locate arguments amongst the four cornerstones of biomedical ethical principles [19]: respect for autonomy (the obligation to respect the decision making capacities of autonomous persons); non-maleficence (the obligation to avoid causing harm); beneficence (obligations to provide benefits and to balance benefits against risks), and to a far lesser extent, justice (obligations of fairness in the distribution of benefits and risks). The main ethical principles under consideration in these policy papers are the first three of these; the significance of this emphasis is discussed later. Where these principles might conflict, one question to be addressed is which has ascendancy in a particular context.

Most of the papers reach broadly similar conclusions regarding the appropriateness of testing for carrier status or for late onset conditions, generally positioning themselves against such testing. Yet, the degree to which they are opposed to testing varies. Only GIG [17] produced by a patient and family interest group, positions itself in favour of carrier testing at parental request. Importantly, the papers differ significantly in how these conclusions are reached and what cases or exemplars of diseases are deployed, and, (even when the same

cases or exemplars are used), *how* the ones selected are used with respect to the four ethical principles.

In the following sections we will consider the use of the three most prominent conditions cited in the data: Huntington's disease; Tay-Sachs disease and sickle cell disease (see Appendix 1) as well as exploring just how when and why other examples are deployed in support of particular positions/conclusions.

Arguments Concerning Predictive Testing: The Use of Huntington's Disease as an Exemplar for Late Onset Disorders

Huntington's disease (HD) has a particular scientifico-historical role as the first single gene disorder to be identified for which testing is available (Gusella *et al*., 1983). Perhaps because of this, it is referred to by each of the datum and this plays an instrumental role in the formulation of the resulting recommendations for both predictive testing and, as we shall show, through the use of contrast, for testing for carrier status. It thus sets a precedent [12, 41]. However, the use of findings referring to the uptake of HD testing, and its description as an especially onerous condition, give it a particular complexion which threatens the safety of any conclusions drawn. Indeed, the earliest paper [11] in our data recognises but does not resolve this problem when the following question is posed: "(T)o what extent is Huntington's disease (HD) unique, and to what extent has the discussion about HD served to awaken us to similar issues raised by the testing of children for other disorders" ([11], p. 786).

It seems that choosing Huntington's disease as the exemplar for adult onset disorders *and* the exemplar condition for predictive testing inevitably acts to construct the particular information generated by predictive testing for HD as highly undesirable categorically (cf. [6]). Huntington's disease is described for example as "burdensome" ([11], p. 786) or with an assumption of a strongly negative impact where "a positive result in a well-prepared individual may not be as devastating as one might predict" ([7], p. 43).

Such quantitative differences in the construal of the degree of severity of testing ultimately translate into qualitative differences in argumentation. The presentation of the disease as onerous works with the selective recruitment of ethical principles under discussion to modulate the generation of policy conclusions.

In those policy papers that consider whether the genetic testing of children is in accord with the principle of autonomy, selecting HD as an onerous, especially serious disease, that strikes in the prime of life, is neurological, untreatable, burdensome and fatal means that information generated from testing is seen as having a serious impact upon individual autonomy. There would be a significant breach of autonomy to test in childhood: the future autonomous choice of the adult to test or not would be denied.

Reference is made in all of these policy papers to the relatively low uptake of HD testing in adults ([1], p. 1454; [11], p. 791; [7], p. 42; [17], p. 5). Note that this low uptake is seen as a given: the right of the individual to refuse this "burdensome" information is taken as read. This contrasts with the testing of other diseases and notably with testing for carrier status, in which low uptake of genetic testing is seen as a problem to be overcome. It seems that the papers assume that carrier testing *will* be done at some time in life. For example, GIG [17] conducts the discussion as if the question is only about when testing should occur, not whether it will ever occur. This normalising of testing reduces the extent to which testing is

seen as a deliberate choice; hence downplaying its intrusive effects upon autonomy. These points will be taken up further in the final section of this paper.

Hence, it is argued, for adult onset predictive testing the principle of autonomy would be breached when testing a child and this breach of autonomy is presented as sufficiently serious that autonomy then becomes the master principle to be protected. This conclusion becomes generalised in these policy papers to the childhood testing of all adult onset disorders.

The use of HD as an exemplar also assists in sustaining a distinction between predictive and carrier testing. All but one of the position statements [3] organise their arguments around a distinction forged between predictive and carrier testing: a distinction grounded in the putative difference in degrees of gravity of the genetic information generated through testing. "Testing healthy individuals for carrier status for X-linked or autosomal recessive conditions is often considered to be of minimal risk when comparing with testing those who are at risk of adult onset disorders" ([7], p. 43). Using HD as the exemplar condition for predictive testing is one device that is used to construe carrier testing as "less serious" through implied contrast. For example, GIG ([17], p. 5) says of carrier testing that "we believe the seriousness of this information has been exaggerated because it is still relatively new," then swiftly moves to claim that, for adult-onset conditions, "the argument that testing of the child takes away their right to make an informed decision as an adult overrides all other considerations," citing as almost sole reason for this the low uptake of testing for HD. This low uptake, premised upon the reluctance of adults to expose themselves to this "serious" information, then is pivotal in constructing seriousness of predictive test information as "overriding" in contrast to the seriousness of carrier test information as "exaggerated."

The question of whether or not testing children for carrier status violates the principle of autonomy is considered in the policy papers. However, any encroachment of autonomy is construed as less serious, therefore possibly counterweighed by the competing principles of beneficence and non-maleficence (as accrual of sufficient benefit in the absence of harm). Although most of the papers argue against carrier testing for children, this conclusion is therefore held less strongly than the conclusion against predictive testing and is more open therefore to individual exceptions, and GIG [17] argues, perhaps not unsurprisingly, in favour of childhood carrier testing at parental request. Where the main ethical consideration is a weighing of harms and benefits, principles will be held less absolutely than when dealing with a notion such as that of autonomy, as unfolding evidence and difference in cases changes assessments of overall harm and benefit. A difference of *degree* of seriousness attributed to knowledge of a disease state hence translates into a substantive difference of *kind* in justifying ethical argument.

What one is left to wonder is how arguments might have gone, had a different genetic disease of delayed onset been used as an exemplar. Other possible candidate conditions include haemochromatosis, familial Alzheimer disease, adult onset blindness, adult polycystic kidney disease and many inherited cancers including autosomal dominant breast cancer. These differ in relevant respects such as inheritance patterns, severity, and the availability of treatment. Many of these differences would act to mark these conditions as less onerous than HD, and hence different policy conclusions may have been drawn. Grouping these conditions together as genetic diseases may act to foreground their commonalities and encourage a generalisation that would not be warranted were other aspects of the conditions to be highlighted. This can be seen as an example of geneticisation, [28] the reductionist phenomenon of foregrounding the genetic over other aspects of a disease or condition.

Arguments Concerning Carrier Testing (I) the Use of Sickle Cell Disease as an Exemplar

Sickle cell disease is a disorder of haemoglobin that causes anaemia, tissue death, multiple infections and may lead to premature death. Some treatment is available. It is present amongst certain populations, such as the African population, African Americans, and also in Mediterranean and Middle Eastern populations ([25], p. 33). Diagnostic testing and testing for carrier status are possible; antenatal and newborn screening programmes exist and are being introduced. In the policy papers in which sickle cell disease is used as an exemplar of carrier status testing, the apparent onerousness or degree of severity of the resulting genetic information is presented in discussions of stigma and discrimination. Here we note problems in argumentation created by the use of this example in the absence of consideration of the particular social, ethnic and community context.

Three papers [1, 7, 11] refer to the same screening programme for sickle cell disease in African American populations carried out in the USA in the 1970s. "The wider issues of social stigma have also caused concern even in the recent past, particularly in relation to sickle cell carrier screening in the USA" ([11], p. 792) Interestingly, despite the context of stigma and discrimination, the racial elements evident in this practice are consistently downplayed; indeed only one paper [7] even mentions that African Americans were targeted, leading the reader to infer that race is not held pertinent to issues of stigma and carrier testing. Selective highlighting of aspects of this case modulates the conclusions generated as the following two extracts show:

> Indeed, there are sufficient historical data to substantiate the concern when African Americans were compelled in some American states to undergo testing for sickle cell disease . . . and were subsequently discriminated against on the basis of carrier status. Therefore . . . an individual . . . should . . . have the right to control [carrier status] information. ([7], p. 43)

> An historical example is provided by the carrier screening programmes for sickle cell disease in the 1970s in the United States that were not preceded by adequate broad-based education. The subsequent misunderstanding of the benign nature of being a sickle cell carrier . . . led to many cases of discrimination and stigmatisation. ([1], p. 5)

It is interesting to note how different ways in which this same screening programme is described also modulate policy conclusions drawn. CPS [7] draws attention to the element of compulsion apparently leads to a corollary of the individual's right to control the information produced through testing and to resist such compulsion. It is important that this control is extended here only to the information produced by the screening, rather than perhaps more appropriately to the screening itself, but nonetheless it is a solution which gives back power to the individual from third parties. Quite distinctly, in the AAP paper, attention to inadequate provision of education is identified as leading to misunderstandings, which are in turn identified as the apparent source of stigma and discrimination. Hence the problem of stigma and discrimination is presented as one that can potentially be remedied by the obvious route of improving education about screening. This differs markedly from the CPS treatment of this case in that it retains the matter out of the hands of individuals by identifying something provided socially, i.e. education, as the root cause of the problem. Note that initial descriptions of the same programme as being "compelled" ([7], p. 43) or as "not preceded by adequate broad based education" ([1], p. 1453) suggest these quite different solutions. This selective highlighting of descriptive factors has weighty normative implications.

Even more centrally for our argument, conclusions are drawn about stigma and discrimination with scant or absent reference to other socially salient issues of race and ethnicity.

But, having already seen the normative effects of selective description, how valid is such generalisation? It remains entirely possible, indeed likely, that this particular case of sickle cell screening cannot be fully understood *without* reference to the pre-existing stigma and discrimination experienced by African Americans. Indeed, it has been argued specifically in relation to sickle cell disease that "particular diseases associated with already low status or discriminated against population groups may themselves take on the discreditable attributes of those population groups, or come to be seen as a further reason for discrimination" ([29], p. 1097). Hence, the stigma occasioned by carrier testing in this case can *only* be understood as mediated through previously existing stigma. Stigma added to an already stigmatised group may not generalise well to stigma laid on a non-stigmatised group. For example, some Tay Sachs screening programmes have been instigated from within the affected communities and have taken steps to avoid stigmatisation of carriers [26].

In conclusion, the way the example of sickle cell disease is used in these contexts incorporates the notion that stigma attaches to certain genetic characteristics, but the choice of genetic condition associated with an already stigmatised group works to makes this conclusion unsafe. Additionally the focus on the example as a genetic disease encourages us to overlook its social and community context, which factors are arguably of greater import in understanding the issues of stigma and discrimination. We are encouraged to generalise perhaps inadvisably from this example to other genetic conditions. This then is a further example of geneticisation.

Arguments Concerning Carrier Testing (II) the Use of Tay Sachs Disease as an Exemplar

Tay Sachs disease is an untreatable, progressive neurological disorder with onset in infancy which causes mental impairment, blindness and deafness and leads to death by the age of five. It is a disorder prevalent amongst, although not exclusive to, the Ashkenazi Jewish population, but references to it similarly lack explicit reference to ethnicity although it is widely thought of as a "Jewish disease" [9]. Unusually in these policy papers, which tend to draw on existent longitudinal studies and qualitative research (cf. [33]) there is a "bioethics style" case [8] as part of an argument that childhood testing for carrier status may not be in the best interests of the child. Examining this case gives insight into how ethnicity in particular, and social context in general, may operate below the surface of examples and cases despite, or because of, the fact that explicit attention is not drawn to it.

> The practice of medical genetics provides some examples of tests that may not be in the best interests of the child. For example, parents may request a determination of their young daughter's Tay Sachs carrier status, for the purpose of encouraging her to be sexually responsible when she is older. The possibility of stigmatisation without any clear immediate benefit is a serious concern. (ASGH, 1995, p. 1239)

The case is told, as in the tradition of medical case telling, from the point of view of the physician involved. The views of the parents and of the daughter are absent. The medical story rather than a social or more contextualised story is told and this again reinforces the way that the disease exemplar is understood and generalised from: as a medical, specifically genetic phenomenon. The framing of this case implies that it derives from real life ("the practice of medical genetics provides"). As Chambers notes, this "contract of truthfulness" is a hallmark of bioethics and is used to demonstrate that any conclusions have been subjected to a legitimate test of validity ([8], p. 8). Yet it remains a thinly articulated account into which much can be inferred by the reader.

The choice of Tay Sachs as an exemplar disease, rather than using a hypothetical example which talks of testing for some unspecified carrier status, invites unstated and so unchallenged, speculation on pressures within a family with a certain ethnic identity. Culture is present in this case by implication only. This case is an extremely clear example of how stories may be read in different ways, with different implied meanings. The unclear notion of "sexual responsibility" muddies the waters. It may allude to "responsible" sexual restraint; to "responsible" use of contraception; to "responsible" choice of partner; to "responsible" use of pre-natal diagnosis. It is of interest that there are also implied gender issues in attitudes to sexual responsibility in the choice of "daughter" rather than "son" or "child" in this case. The example leaves us unclear whether the parents want to encourage a specific sexual responsibility related to possible carrier status, (which may or may not be legitimate) or are using this as a device for smuggling in their concerns to encourage a more general sexual responsibility in their daughter (for which read: "this looks disingenuous and is hence dubiously legitimate"). However, by simply talking of "sexual responsibility," as opposed to the more specific "reproductive responsibility," the reader is arguably guided towards the latter interpretation. The reader is left with a negative impression of carrier testing in childhood inviting generalisation to other cases when much of any negative attitude stems from what this case suggests but does not state, much in this instance deriving from stereotyped notions of ethnicity. Without clear articulation, positions become less easy to rebut [14] and again we are seeing the operation of geneticisation in the generation of policy.

Explicit reference to ethnicity in the context of these genetic diseases could have assisted in making useful discriminations both between and within ethnic groupings. Ethnicity intersects with salient features of any carrier screening in at least one significant way; the 1970s USA sickle cell screening programme was mandated on African Americans in some states, a practice consistent with actions targeted at groups of low social status. Conversely, many of the screening programmes for Tay Sachs disease are either instigated by the affected communities themselves, or offered to them as an option, consistent with actions deriving from higher status, more highly resourced groups. The imposition of screening from without a community is of course ethically problematic compared with instigation from within; conclusions, in our data, generated from examples of diseases that make no reference to such highly salient differences are to that extent problematic.

Moreover, within the Ashkenazi Jewish population there are different groups who may have quite diverse attitudes towards carrier status; significantly there exist screening programmes with divergent rationales. Some aim to alert people to carrier status so that the resources of genetic technology, here pre-natal diagnosis followed by selective termination, can be utilised. In contrast the Dor Yeshorim programme, specifically motivated by Hasidic beliefs against abortion and in favour of formal marriage arranging, aims to influence choice of marriage partner. To avoid stigma, carrier status is not disclosed directly; pin numbers are used to identify cases where two potential marriage partners are both carriers.

Leaving out reference to the social and community context of this disease, as of others, hence risks impoverishing any resulting discussion. "Stigma" is seen only as attaching to a disease insofar as it is characterised as "genetic," rather than in any more fully significant characterisation. Attempts at generalising from a specific disease exemplar that do not take account of community values that help form how a disease is experienced, understood and reacted to, will be to that extent flawed.

Like sickle cell disease, Tay Sachs is also a disease associated with a particular ethnic grouping, yet the position papers do not usually cite Tay Sachs as an example in the context of stigma and discrimination, rather in terms of psychological harms and benefits. This

is intriguing especially given that it is well known in the literature that many Tay Sachs screening programmes have gone to great lengths to avoid stigmatisation. Reference to the distinct social status of Ashkenazi Jews as distinguished from that of African Americans is textually absent, yet rhetorically present below the surface of the papers in the use of particular diseases to illustrate different points.

Community Context and Disease Testing: Lessons for Policy Making and Practice

Any attempt to draw up general principles from particular human experience is going to have limits, and any use of cases is necessarily selective. The points we make may be seen as having relevance to any attempt to produce general policy from particular cases, but we focus on lessons for genetic policy making, in particular looking at problems about how diseases are delineated and generalised from within an exclusively genetic context.

We have located our discussion in the reality that policy making "always takes place within a nested context" ([35], p. 154). In this concluding section we examine the form and effect of some of the nesting that occurs in these policy papers. Here, as elsewhere within genetics, the construal of genetic testing issues is nested within a context of positive assessment of fast progress in genomic research and technological developments. Opening statements for the policy papers make such statements as that "rapid developments" ([3], p. 1233) have "greatly enhanced" ([7], p. 42, also [11], p. 786) our ability to test and "promise great strides" ([1], p. 1451). As Lemke ([27], p. 1) also appreciates, this "will alter the way individuals experience pregnancy and birth as much as it will change social institutions (such as the health system, the legal and insurance systems) and influence the way we collectively treat disability, illness and death." This nesting helps to set the scene as a technological and medical one. We have already pointed out how the disease exemplars used display the phenomenon of geneticisation, which can be seen as a sub-set of the technological and medical realm.

Despite the "rapid progress" of this new technology it already of course has a history. The use of Huntington's disease as a precedent setting disease is a prominent part of that history, and our examination of the shortcomings of using it as an exemplar highlighted for historical reasons, or because it represents an "extreme" or "clear" case, should act as a warning. The very newness of the field gives few historical examples from which to choose. Greater awareness of this very problem could have acted as a warning against excessive reliance upon this exemplar.

Community context is present in the use of some of the exemplars but consistently downplayed. Because the wish in these policy papers is to generalise out to other genetic conditions, the diseases are presented as "genetic diseases" standing in for all genetic diseases, or for all late onset genetic diseases, or for all genetic diseases where one can be an unaffected carrier. As a result, significant features of testing and screening in particular contexts may be lost to view in this tendency to understand the diseases only or chiefly as genetic. This then can be seen as a concrete manifestation of the phenomenon of geneticisation. This can have a direct bearing on the formation of policy as genetic or medical features of a disease are highlighted at the expense of examining the impact and meaning of a disease within a community or individual context.

This nesting under the category of the genetic as we have seen acts to downgrade considerations of the social. Further lessons can be learned from considering how the different disease exemplars are differentially nested with respect to the social. An intriguing feature of the disease exemplars studied is that HD, the chief exemplar for a late onset disease

used in the context of predictive testing for individuals, is a disease presented free of any community context. The prevalence rate of HD is about 1/20,000 in Caucasians, and less in other populations such as Africans. It is a disease of the dominant population covered by the policy papers, and perhaps because of this does not carry any markers of particular identity or of community. In contrast, sickle cell and Tay Sachs disease are associated with particular community groups, and even if this feature is downplayed in the policy papers, as we have seen it nonetheless has a presence: the question of the community is immediately raised. In the case of African Americans, past sickle cell screening has had implications for the community as a whole; in the case of Tay Sachs disease, particular affected communities have themselves mobilised screening programmes. In these very different ways, testing for carrier status for these diseases is hence easily and naturally seen in a community context where responsibilities to others, including reproductive responsibilities but expanding to include responsibilities to avoid stigma and discrimination, are foregrounded. In contrast, testing for HD is presented simply as an individual decision, and this is highlighted by the frequent use of individual autonomy as the most important ethical principle.

The significance of this is that it acts to underline a striking difference between predictive and carrier testing. The information generated by predictive testing is seen as knowledge of the self, private, individualised and protected by the dominant principle of autonomy. The information generated by carrier testing is presented as additionally having wider implications for others, including reproductive implications. This is despite the fact that predictive testing also has reproductive implications. This contrast between predictive and carrier testing may be present for independent reasons, but the choice of the specific disease exemplars used in these policy papers helps to reinforce this distinction. More careful use of exemplars, or use of different exemplars, may therefore have modulated resultant policy.

Foregrounding the genetic basis (e.g. inheritance) of disease in order to classify conditions ignores crucially relevant differences within sub-populations. Focussing on social salience and cultural practices would produce quite different groupings and in this data perhaps influence the final recommendations with respect to testing. In particular, reasoning about the reproductive implications of genetic carrier status, without reference to the social contexts which modulate reproduction, is unwise.

The nesting of the genetic disease exemplars provides for a further limitation. Many commentators have noted how frequently much discussion within medical ethics gives a very attenuated role to considerations of justice. Chambers identifies one mechanism by which this lesser role is ensured, by pointing out how the setting of the bioethics case is within the context of an unspecified clinic. This then acts to shield out considerations of justice, which can only be raised within a known socio-economic context. Likewise, the focus on the genetic and the medical in the use of these disease exemplars and the eclipsing of the social also acts to obscure considerations of justice; as noted above, and in common with much other literature in bioethics, these policy papers make reference to principles of autonomy, beneficence and non-maleficence but not to justice. This is an unfortunate omission in discussions of genetic policy.

Any attempt to draw up general principles from human experience is going to have its limits, and any use of cases is necessarily selective. Lessons for any policy making must include warnings about generalising from exemplar and cases where certain highly salient features are disregarded. In particular we have seen many different respects in which the foregrounding of the genetic aspects of a disease at the expense of consideration of other aspects, in particular the social and community context, will result in impoverished thinking at the policy level.

Appendix

Genetic Conditions Used in Argumentation in the Data	Number of Times	
Adult Onset Blindness/Retinoblastoma	2	
Alpha-1 Antitrypsin Deficiency	1	
Alzheimer's Disease	2	
Balanced Chromosomal Translocations	2	
Coronary Heart Disease	2	
Cystic Fibrosis	4	
Diabetes		2
Duchenne's Muscular Dystrophy	2	
Familial Adenomatous Polyposis Coli	2	
Familial Hyperlipidema	1	
Fragile X		3
Friedreich's Ataxia		1
Haemochromotosis		1
Haemoglobin Disorders	2	
Hereditary Cancers (incl BRCA1)*	6	
Hypertension	1	
Huntington's disease	**16**	
Multiple Endocrine Neoplasia	1	
Myotonic Dystrophy		2
Neurofibromatosis		1
Phenylketonuria	1	
Polycystic Kidney Disease		2
Prion Dementia	2	
Sickle Cell		**8**
Tay Sachs		**6**
Von Hippel Landau Disease	1	

*The hereditary cancers are primarily susceptibility tests and are not within the scope of this paper.

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