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The genetic testing of children raises many ethical concerns. This paper examines how five position statements from Canada, UK and USA, which present guidelines for good practice in this area produce different recommendations for carrier testing and predictive testing. We find that the genetic information generated through carrier testing is routinely presented as less serious than that generated from predictive testing. Additionally, the reproductive implications of predictive testing are also routinely erased. Consequently, the papers argue strongly against predictive testing but advise caution against carrier testing in somewhat weaker terms. We argue that these differences rest on assumptions about the status of reproduction in people’s lives and on an ethical stance that foregrounds the self over others. We propose that questioning the crude and sharp distinction between carrier and predictive testing in principle may enable practitioners and parents/families to make more nuanced decisions in practice.

KEY WORDS: genetic testing; children; policy; carrier testing; predictive testing; discourse analysis; ethics.

INTRODUCTION

The issue of the genetic testing of children, especially in circumstances where there is no immediate medical benefit to the child such as testing for carrier status and for adult onset disorders, is one that has received a great deal of ethical and policy attention. In this article we examine how five influential policy documents deal with one area that has received less attention as an ethically problematic issue: the distinction between testing children for carrier status, and predictive testing of children for later-onset disorders. By carrier testing we refer to testing used to identify usually asymptomatic individuals who have a gene mutation for an autosomal recessive, X-linked recessive or chromosomal disorder. Predictive testing refers to testing offered to asymptomatic individuals with a family history of a genetic disorder and a potential risk of eventually developing the disorder (cf. NIH, 2004). To make this distinction in an absolute way is problematic since “carriers” of some conditions may not be entirely asymptomatic (Leehey et al., 2003). Furthermore, one of our main interests is the way that, in the published guidelines we examine, the distinction is made in that carrier testing is seen as having implications for reproduction, but the potential...
implications for the reproductive self, which result from predictive testing, are routinely overlooked.

Through our analysis of five formative policy/position statements which present guidelines for preferred practice with respect to genetic testing in childhood, we hope to illuminate assumptions pervading much of the childhood testing literature and, possibly, much thinking elsewhere within genetic testing protocols and practice. We will argue that the dichotomy of carrier testing and predictive testing is drawn too sharply, and although it may appear to be value neutral, it operates with, and is partially constructed by, other value distinctions, including the sometimes-competing ethical principles of respect for autonomy, avoiding harm and achieving benefit.

We will introduce our analytic perspectives and summarize the recommendations in the published guidelines. Using illustrative excerpts from the data we will demonstrate just how a rhetorical distinction is wrought between carrier and predictive testing, indicating how this influences the hierarchical deployment of ethical principles and the inevitable effects this has on the final forcefulness of the resulting recommendations.

ARGUMENTATION AND POLICY-MAKING

Policy-making, Fischer and Forester argue, is “a constant discursive struggle” (1993, p. 1–2). The struggle occurs in how arguments are structured, between the ways certain “facts” are represented, the selective inclusion and exclusion of particular factors that the writers consider relevant and how conclusions are generated particularly when their incorporation into practice relies on how the reader interprets the text. Policy-making and policy-implementation is thus a linguistic, rhetorical, discursive, and interactional activity (Majone, 1989) which depends fundamentally on choice. This can be illustrated simply at a linguistic level where Fairclough (1992, p. 77) suggests that “people make choices about the design and structure of their clauses which amount to choices about how to signify (and construct) knowledge and belief.” That is to say, a particular discursive construction of a state of affairs is implicitly or explicitly designed for, and has, particular (sociopolitical) purposes.

Analyzing how this is accomplished is at the core of the complementary disciplinary approaches of applied philosophy and discourse analysis we employ here. These disciplines examine how versions of reality are construed, how arguments are constructed, what features of social and moral life are recruited, downplayed or ignored and how underlying ideological influences are utilized in reasoning. Both these analytic perspectives aim to show that distinctions that may appear neutral or simply factual may in fact “smuggle” in further value assumptions (cf. van Dijk, 1993; Fairclough, 1989). The relevance of such analysis is deemed essential if policy is to be considered critically, as Fischer and Forester (1993, p. 1) recognize when they state:

If . . . ways of representing policy and planning issues must make assumptions about causality and responsibility, about legitimacy and authority, and about interests, needs, values, preferences and obligations, then the language of policy and planning analyses not only depicts but also constructs the issues at hand.

Consequently, analyzing the role of language in the policy-making process can, as Hastings (1998, p. 193) argues “help to reveal how social policy is implicated in constructing and sustaining a ‘system of belief’ or ‘ideational knowledge’ about the nature of social reality” (cf. Hajer, 2002).

The distinction between carrier testing and predictive testing can be treated as one such “system of belief” which, we argue, may contribute to a way of thinking which Hajer (1993, p. 48) describes as, “discourse institutionalization” where professionals or “central actors are persuaded by, or forced to accept the rhetorical power of a new discourse” and “this is reflected in [how] the institutional practices [and] actual policy processes are conducted according to the ideas of a given discourse.” In this respect we are concerned with the way that a distinction wrought at policy level, premised on particular arguments, may innocently be applied in practice. A related concern is how, conversely, published policy documents may fail to capture or influence actual practice. Here we simply raise the question of how exactly in practice, the distinction between carrier and predictive testing is managed.

ETHICS AND RECOMMENDATIONS FOR GENETIC TESTING IN CHILDHOOD

The position statements we examine provide guidelines or advice related to appropriate professional practice with respect to genetic testing in childhood, and in doing so draw upon recognized ethical principles, notably here the principles of autonomy, beneficence (the aim to produce benefit) and non-maleficence (the aim to avoid harm).