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Ten Fingers, Ten Toes: Newborn Screening for Untreatable Disorders

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TEN FINGERS, TEN TOES: NEWBORN SCREENING FOR UNTREATABLE DISORDERS

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The topic of newborn screening for untreatable disorders evokes the movie GATTACA in which all newborns have extensive genetic testing at birth. The following lines by the protagonist, Jerome, recounting his own birth appear in the original script:

JEROME (VO)

Of course, there was nothing wrong with me. Not so long ago I would have been considered a perfectly healthy, normal baby. Ten fingers, ten toes. That was all that used to matter. But now my immediate well-being was not the sole concern.

Antonio [Jerome's father] turns his attention from his baby to the data appearing on the monitor. We see individual items highlighted amongst the data - *"NERVE CONDITION - PROBABILITY 60%", "MANIC DEPRESSION - 42%", "OBESITY - 66%", "ATTENTION DEFICIT DISORDER - 89%"--*

JEROME (VO)

*My destiny was mapped out before me--**all my flaws, predispositions and susceptibilities - most untreatable to this day.** Only minutes old, the date and cause of my death was already known.*

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> Antonio focuses on a final highlighted item on the monitor's screen, *"HEART DISORDER - 99% - EARLY FATAL POTENTIAL." "LIFE EXPECTANCY - 33 YEARS."*[1]

Note the bolded text in Jerome's second voice over, which did not appear in the film. In a nightmare of genetic discrimination, Jerome suffers numerous indignities as a result of his genetic heritage, from being denied admission to a nursery school for fear of liability were he to fall to being employed as a janitor. But this is a story of redemption, and by using the genetic identity of Eugene (literally, good gene), Jerome is able to fulfill his lifelong dream of becoming an astronaut. He also wins the heart of Irene (Uma Thurman). And it turns out that the awful predictions made at his birth were not borne out. Far from dying of heart disease at the age of 33, Jerome passes a cardiac stress test with flying colors.

This movie makes two important points despite its admitted unreality. The first, which the screen writer probably did not fully appreciate at the time, is that genetic testing cannot now[2] and probably will never be able to predict with complete certainty the occurrence and course of complex diseases. It is not true that "Genes-R-Us." Rather, we are the products of complex interactions of our genes, the genomes of other organisms (many of which we live in relation with),[3] and the environment, broadly understood to include the air we breathe, the water we drink, the food we eat, the drugs we take, our social structures, and our cultural practices.[4] Moreover, the effects of these interactions can vary depending on when they occur in the life cycle. For example, it appears that caloric deprivation during pregnancy may predispose children to a host of health problems later on, whereas the same deprivation during adulthood might be life prolonging. As a result, even the most sophisticated genetic analysis at birth is not going to reveal everything about a person's future health.

The other lesson of GATTACA, the ultimate of a genetic essentialist dystopia, is the importance of characteristics of the society in which the information is being used. Why might *we* want to screen

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[1] Andrew M. Niccol, UNTITLED, http://www.imsdb.com/scripts/Gattaca.html (last visited Nov. 2, 2008) (emphasis added).

[2] Olivia Judson, Op-Ed., *Testing Genes, Solving Little*, N.Y. TIMES, Aug. 17, 2008, at WK10.

[3] Colin Nickerson, *Of Microbes and Men: Bacteria Disappearing from Our Bodies May Harm Human Health*, BOSTON GLOBE, Feb. 25, 2008, at C1.

[4] COMM. ON ASSESSING INTERACTIONS AMONG SOC., BEHAVIORAL, & GENETIC FACTORS IN HEALTH, INST. OF MED., GENES, BEHAVIOR, AND THE SOCIAL ENVIRONMENT: MOVING BEYOND THE NATURE/NURTURE DEBATE *passim* (Lyla M. Hernandez & Dan G. Blazer, eds., 2006).

newborns for untreatable disorders? Here the excellent paper by Bailey et al. in Pediatrics is particularly thoughtful.[5] In this article, the authors discuss a number of arguments for newborn screening for these problems.[6] These include avoiding delays in diagnosis often after numerous consultations, providing parents with information about their children's health, allowing the earlier initiation of interventions, learning more about the natural history of these disorders, providing reproductive information to the family, and creating more opportunities to support these families.[7]

The authors also point out some of the problems that might arise. They point, for example, to potential changes in childrearing.[8] For instance, in GATTACA, Jerome's parents kept telling him not to dream about opportunities that were not open to him because of his "broken ladder." Other problems include the difficulty of sharing what could be unwanted news with family members and the gendering of responsibility for the health of children. Regarding the latter, women not only tend to deliver more hands on care but also are more likely to be seen to be at "fault" for their child's health problems even for autosomal recessive disorders, which require receiving a mutant gene from both parents.

Finally, the authors make recommendations. One of these, which is already coming to pass, is to set up a research network to learn what works and what does not.[9] They proposed that parents be asked to give informed consent prior to newborn screening for untreatable disorders and that new models be developed to do so.[10] They also oppose newborn screening for adult onset disorders.[11]

Despite this very thoughtful analysis, some questions and issues remain. One question is why this screening should be done 1) by the state and 2) in the newborn period. Children get most of their health care from clinicians in either public clinics or the private sector. These clinicians screen children all the time for growth and development[12] and for a myriad of health problems. Indeed, Medicaid re-

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[5] Donald B. Bailey, Jr. et al., *Ethical, Legal, and Social Concerns About Expanded Newborn Screening: Fragile X Syndrome as a Prototype for Emerging Issues*, 121 PEDIATRICS, e693 (2008).

[6] *Id.* at e694-701.

[7] *Id.*

[8] *Id.* at e694-95. *See also* Rachel Grob, *Is My Sick Child Healthy? Is My Healthy Child Sick? Changing Parental Experiences of Cystic Fibrosis in the Age of Expanded Newborn Screening*, 67 SOC. SCI. & MED. 1056 (2008).

[9] *Id.* at e699-700.

[10] *Id.*

[11] *Id.*

[12] The types of screening already in use or capable of being developed may

quires this screening for the children it covers. One can ask, then, why not screen for particular disorders when it is time to intervene? Admittedly, not all children receive optimal health care, and it may be more efficient to do genetic testing all at once, but given the risks to both parent and child that can attend newborn screening particularly for untreatable disorders, we should at least consider the possibility of a staged approach.

Another reason to include clinicians in genetic screening for untreatable disorders is that they are more likely than state officials to talk with parents about what is at stake. As a clinician myself, I am under no illusions that these conversations will automatically rise to the level of informed consent. Clinical care is often subject to routinization and the pressures of limited time, but the fact that the clinician who orders the test then has to deal with the consequences of bad news is a powerful incentive to say something about what the test is for and what it might reveal before it is done.[13] By contrast, having state run programs obtain consent would require a huge sea change since the vast majority of these programs have never sought parental permission.

The next two issues broaden out from the newborn screening process itself. Deciding to focus on genetic variations can make them seem more important than their actual contribution to health and disease warrants, possibly diverting attention from environmental and social factors. In addition, people worry that information about their genetic makeup will be used to their disadvantage. Only time will tell whether anti-discrimination laws, such as the Genetic Information Nondiscrimination Act,[14] will allay those fears. The good news is that the social implications of genetic information can be shaped by public will, for which there is powerful precedent in laws against discrimination on the basis of race, sex, and disability.[15] Ongoing vigilance, however, is essential.

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prove cheaper and more thorough than genetic testing. The M-CHAT is almost certainly going to be better at picking up a child with autism than a complex genetic test because the former is able to look at the results of the interactions of environment and genetics rather than just the genetics. It might also be less expensive.

[13] It also increases the likelihood that the clinician will know what to do with the results.

[14] Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-233, 122 Stat 881 (2008).

[15] I am one of the most optimistic people I know about the possibility of creating a more inclusive society. I grew up in Texas in the early 1950's. I went to segregated schools. There was no accommodation for people with disabilities although one of my classmates in high school was blind. Also, as a girl, all kinds of opportunities were foreclosed to me, including among other things, participating in

The final issue goes to the question of who decides what in the health care system. There is strong pressure to adopt evidence-based medicine, using only those interventions of proven efficacy in an effort to exercise stewardship over limited resources and to reduce the incidence of adverse events from unnecessary tests and treatments. Moreover, clinicians for decades have had a legal monopoly over a host of medical interventions, having sole authority to decide when a particular test or drug is "clinically indicated." Professional organizations promulgate guidelines for care. For example, there is a general consensus that children should not receive genetic tests for adult onset disorders unless screening or treatment must begin prior to the age of majority.

Patients and their families, however, have resisted these limits in a number of ways. The ethical doctrine of informed consent teaches that patients' values matter. [16] Political action by patients and parents is also having a profound impact on medical care. This is clearly evident in newborn screening. Parents played a critical role in the initial adoption of state newborn screening laws in the late 1960s. Their role has only grown since, especially with the expansion of the internet, which facilitated advocacy for the adoption of tandem mass spectrometry and other tests. Over and over, they have expressed their impatience with the medical system. Parents urge that that there is no time to wait for evidence from clinical trials. They repeatedly say that they want to know what is going on with their children and argue that they, and not clinicians, are best positioned to decide what is best for their children and their families. These sentiments underlie the current assault on the "consensus" that children should not be tested for adult onset disorders, even though many adults continue to avoid genetic tests themselves, especially for untreatable disorders.

In the end, the fate of newborn screening for untreatable disorders will probably rest not upon the results of science or ethical discourse, but with the outcome of the struggle for control between health care providers and parents in the political sphere. Whether children and their families benefit will depend on the shape of the health care system and the society into which these children are born.

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sports unless I wanted to play tennis or volleyball. The world has changed a lot, even if not as much as we would like. So I am a person who is hopeful about the possibility of creating a world that is more accepting of difference.

[16] *See, e.g.*, JAY KATZ, THE SILENT WORLD OF DOCTOR AND PATIENT (1984). Legal rules that protect the reasonable patient or the reasonable physician are not so deferential.