Thinking About Dementia

Annette Leibing, Lawrence Cohen

Published by Rutgers University Press


For additional information about this book
https://muse.jhu.edu/book/17686

For content related to this chapter
https://muse.jhu.edu/related_content?type=book&id=619776
I think it’s safe to say we will have individualized, preventive medical care based on our own predicted risk of disease as assessed by looking at our DNA. By then each of us will have had our genomes sequenced because it will cost less than $100 to do that. And this information will be part of our medical record. Because we will still get sick, we’ll still need drugs, but these will be tailored to our individual needs. They’ll be based on a new breed of designer drugs with very high efficacy and very low toxicity, many of them predicted by computer models.

—Francis Collins, Director of the National Human Genome Research Institute, Time

Comments and claims, such as the preceding by Francis Collins, as well as reports about newly located genes, appear with increasing frequency in the media these days. The sociologist Alan Peterson argues that such stories are deemed newsworthy “precisely because they offer people the promise of being able to re-make themselves anew—to ‘play God’—so that they can better deal with, if not overcome, the reality of disease, disability and death” (2001, 267). Because of the possibility that such stories may bring about changes in individual behaviors, Peterson insists that it is important to investigate how “gene stories” selectively present “facts, themes, and claims, and thereby help limit what can be known about health, disease, and embodiment” (267). Adding a sense of urgency to such an investigation is the claim commonly made by clinical
geneticists and many other medical specialists today, that all diseases must be recognized as having a genetic basis (Lock, forthcoming).

While social scientists such as Peterson have contributed to an understanding of the way in which media coverage of knowledge claims in science are biased, albeit often inadvertently, additional factors are clearly at work in connection with knowledge uptake (Conrad 1999, 2001; Peterson 2001). Authors often assume that a relatively simple trickle-down process takes place as knowledge is passed from one domain to another, for example, on the one hand, from basic scientists to health-care professionals and then to patients and, on the other hand, from basic scientists to the media and then to the public at large. Embedded in this assumption lies another: that a uniform body of scientific knowledge exists and experts are in effect united in their understanding about what types of action should be taken on the basis of this knowledge. Peterson is critical of such a position, as are we, and goes on to note that research shows that laypeople have a more sophisticated and critical view of genetics than is generally acknowledged by medical experts (2001, 1256; see also Kerr, Cunningham-Burley, and Amos 1998). However, observing that the public holds a critical or skeptical view of genetic research tells us little about the way in which genetic science is understood or used by this same public.

The expansion in knowledge about molecular genetics that has occurred during the past two decades, particularly since 2001, when the "completed" map of the human genome was heralded, have indeed led to Manichaean prophecies about the implications of this new knowledge. Genetic advancements are thought to have unlimited promise for medical developments that will benefit all humans, but at the same time, fears have run rampant about the unleashing of unprecedented eugenic powers and the potential engineering of social inequality at the molecular level. Often scapegoated, the media is but one culprit in the authorship and perpetuation of these parallel discourses: the scientific community, politicians, advocacy groups, ethicists, and social scientists have contributed to both. In this chapter we set out to counter both some of the more hyperbolic of these claims that genetics is revolutionizing medicine and the way health and illness are understood by both professionals and the public.

Much of what has been written to date by social scientists about the new genetics has focused on life-threatening conditions in which the involved gene or genes follow a Mendelian pattern of transmission, and therefore inheritance patterns and calculations of risk (but rarely severity) can be made with great accuracy. However, as yet, rather little has been written by social scientists about complex, multifactorial diseases such as Alzheimer’s disease (AD). Being alerted to susceptibility, particularly for a disease such as AD that is largely restricted to the elderly, does not confer the same order of knowledge as that of being told that one carries a mutation for a disease that is lethal much earlier in life and that may already have been transmitted to one’s children. Before gener-
alizations are made about what effect genetic “truths” are likely to have on individuals, the scientific “facts” and material effects of specific diseases must be taken into account. Social scientists can be as guilty of reductionism as are certain geneticists when theorizing about abstracted futures, if such theorizing is not grounded in the materiality of the body, or if it conflates the lived experiences associated with various diverse illnesses.

To counter this type of reductionism, we focus here on a particular disease, late-onset AD, which provides an interesting example precisely because of the way in which genetic knowledge has recently been implicated in etiological theories about this condition. Unlike other diseases that exhibit a Mendelian pattern of inheritance, including early-onset AD, genetics is implicated in an as yet poorly understood, complex fashion in late-onset AD, as it is in numerous other common diseases. When making estimates about the susceptibility of individuals to a particular disease on the basis of their genotype, which is inevitably compounded by the varying “penetrance” of virtually all disease-producing genes, resulting in a wide range of phenotypic effects in individuals, uncertainty is compounded enormously, and the prediction of who exactly is at increased risk is fraught with difficulty. Under these circumstances, conveying genetic information to people who are affected by a specific disorder or to those who are “carriers” of a gene for the disorder, and to their family members, becomes problematic. In this chapter we consider the state of current genetic knowledge surrounding late-onset AD, specifically, the way in which key parts of it are packaged and disseminated in clinical settings, by the media, and for the public at large. Emphasis is given to the types of information to which people in each of these domains have access, the information they communicate to others, and the information they use personally. We have found that in the course of translation of information across groups, there is an uneven penetrance of ever changing genetic knowledge into both popular and professional discourse about AD causation.

Enlightened Geneticization?

In 1992 Abby Lippman coined the term geneticization to capture what she perceives as an ever growing tendency to distinguish people one from another on the basis of genetics. She argues that geneticization is both “a way of thinking and a way of doing, with genetic technologies applied to diagnose, treat, and categorize conditions previously identified in other ways” (1998, 64). Lippman is concerned above all with a possible reinforcement of racism, inequalities, and discrimination of various kinds that already exist in abundance, as a result of a renewed conflation of social realities and biological difference grounded in genetics. She suggests that we may well be witnessing an incipient neo-eugenics, as do many other contemporary writers (see, for example, Kitcher 1997).
Adam Hedgecoe, a social scientist, understands the use of genetic knowledge and technologies as just the latest in a long line of attempts to advance our understanding of the body at the molecular level and is less inclined than he believes Lippman to be to see geneticization as “an opportunistic tactic employed by doctors to gain power over patients” (2001, 877). Hedgecoe argues for recognition of a concept of “enlightened geneticization,” by which he means that even though the contribution of environmental and other factors are today widely accepted in scientific discourse about disease causation, genetic explanations are nevertheless prioritized and subtly divert attention away from nongenetic factors (see also Spallone 1998). Hedgecoe (2001) has shown how this discourse of enlightened geneticism is reproduced in psychiatric medical literature. He agrees with Lippman that genetic determinism, although more subtle than was formerly the case, is at work, but he points out that geneticization, as does medicalization more generally (Lock and Kaufert 1993; Lock, forthcoming), has some positive attributes. For example, it is abundantly clear that once a disease is medically recognized, particularly when behavioral changes are involved, then social stigma and allocation of individual and family responsibility for the occurrence of such conditions are reduced (McGuffin, Riley, and Plomin 2001). What is more, many families appear to take comfort in being told that a disabling condition is the result of faulty genetics and therefore, by implication, has nothing to do with moral shortcomings (Turney and Turner 2000). It remains to be established if this finding applies to AD, as it does to psychiatric illness. Preliminary inquiries indicate that in many circles, stigma is no longer associated with AD, now regarded as the result of unavoidable neurobiological changes and, moreover, that a good number of people believe that these conditions are part of the unavoidable process of aging. However, stigma clearly remains attached to mental illness, even in contemporary psychiatric practice (Marc Miresco, personal communication), so that the “urge” to geneticize may be greater in psychiatric circles and among the families of psychiatric patients.

The arguments of Lippman and Hedgecoe are insightful, but in the case of AD, and no doubt other multifactorial conditions, even when a susceptibility gene is incontrovertibly associated with a disease, it does not follow that geneticization, even in an “enlightened” form, will become the dominant mode of conceptualizing the condition at all sites where it is investigated, treated, or managed. Although an enlightened geneticism is apparent in certain types of professional discourse in connection with AD, dissent and disagreements, and above all differences in emphasis, are evident among various groups of specialists—basic scientists, neurologists, clinical geneticists, clinical psychologists, psychiatrists, and others (Lock, forthcoming). Nor is a focus on genetics steadfastly embraced by the media, which usually acknowledges that, almost without exception, multiple risk factors are at play. Advocacy-group literature mini-
mizes the contribution of genetics, as do the majority of patients and their families as they face the massive burden of dealing with a disease as devastating as is AD. Our research shows that what is common when involved patients and families discuss the possible causes of AD is that, in addition to behavioral, social, and environmental factors, references to hereditable tendencies are made at times. However, other than among geneticists, highlighting the contribution of specific genes is left at the margins of most discussions.

At a broad level of generalization, the concept of geneticization alerts us to the persistence of a particularly insidious form of reductionism that may well be exacerbated by the current widespread fascination with emerging knowledge in molecular and population biology. As technologies of molecularization and knowledge in connection with the new genetics change, so too will the social meanings associated with specific conditions, and it will be of great interest to track such changes. In the idiom used by researchers engaged in mapping out the sociology of science and technology, late-onset AD is a boundary marker that serves as an “interface” between multiple social worlds that draw upon and emphasize different sources and types of information and interventions (Fujimura and Clarke 1992).

**Living with the New Genetics: Biosociality and Genetic Citizenship**

Social scientists have introduced several new concepts in order to convey the responses of the public and, more specifically, of individuals and families who are directly affected by the new technologies of genetic testing and screening. Rayna Rapp and colleagues have posited the concept of “genetic citizenship” as one response to new information yielded from genomics research. Their research has documented how certain families and networks of families increasingly coalesce around lethal and highly disabling single-gene diseases that afflict their children. Such groups provide mutual social support and lobby the United States Congress for improved research funding (similar activities happen in many other countries). These activists are painfully aware that only rarely will drug companies invest in research for the kinds of diseases that affect their families. Because of their relative rarity, there is no profit to be had in researching the “orphan diseases,” and lobbying for public funding is deemed essential. This is genetic citizenship in action, and it involves not only mobilization of affected people but also new ways of envisioning the future (Rapp 2003).

Two decades ago, Edward Yoxen (1982) suggested that our newfound abilities to detect “pre-symptomatically ill” individuals would ensure that virtually all of us would shortly be subject to increased medical surveillance. More recently, Carlos Novas and Nikolas Rose have raised important questions about what it means to be designated as “genetically at risk” (2000). On the basis of their perusal of Huntington’s disease Web site exchanges, Novas and Rose argue...
that genetic testing does not generate a sense of fatalism, as many have pre-
dicted, but rather brings about “genetic responsibility,” a bonding that is
grounded in a molecular optic and that transforms relationships between ex-
pert and patient and within and between affected individuals, families, and
communities. However, it has also been shown that among those families in
which Huntington’s disease is prevalent, the majority of individuals choose not
to undergo genetic testing.

In his 1996 essay “Artificiality and Enlightenment: From Sociobiology to
Biosociality,” Paul Rabinow cites the geneticist Neil Holtzman, who envisions a
time when early detection of genetic susceptibility and predispositions will be-
come routine. Rabinow chooses to underline only one of the many issues that
he believes arise from this prediction. First, he notes “the likely formation of
new group and individual identities and practices arising out of these new
truths.” He then points out that such groups already exist, formed on the basis
of the firsthand experience of diseases such as neurofibromatosis, groups
whose members meet to share their experiences, educate their children about
the condition and its genetic transmission, finance changes in the home envi-
ronment to facilitate home care, lobby for funding for further research in con-
nection with the particular disease associated with their family, and so on. “This
is what I mean by biosociality,” writes Rabinow. In rhetorical mode he adds: “I
am not discussing some hypothetical gene for aggression or altruism. Rather it
is not hard to imagine groups formed around chromosome 17, locus 16,256, site
54,376 allele variant, with a guanine substitution.” Rabinow is quick to add
that these newer groupings will not overtake older categories and, contrary to
the claims of certain population geneticists, may even enhance the spread of
racism because of a heightened sensitivity to biological difference (1996, 91).

Ironically, Holtzman has since gone on to forcibly argue against claims that
genetics will revolutionize the way in which disease and illness are understood.
“Statements like these clothe medicine in a genetic mantle. The result of efforts
to identify genes that have a role in common disease suggests a different pic-
ture: the genetic mantle may prove to be like the emperor’s new clothes”
(Holtzman and Marteau 2000, 141). He argues that, ultimately, with the excep-
tion of Mendelian disorders, the mapping and sequencing of the human ge-
nome will have little impact on the understanding, treatment, or prevention of
disease, in large part because of the incomplete penetrance of genotypes for
common diseases.

Of course, in contrast to the “orphan” diseases, no genetic citizenship is
required in order to place heart disease, cancer, or AD in the spotlight; gov-
ernments and the pharmaceutical industry are both deeply invested in trying
to reduce the financial and social burden associated with these conditions. The
existence of support groups for such diseases precedes the consolidation of
molecular genetics by many years, and in the case of late-onset AD, their ac-
tivities continue largely unchallenged and unchanged by the new genetic knowledge.

For the remainder of this chapter, we will focus on AD, providing a brief review of current medical understanding about the role of genetics in AD causation, followed by an overview of how this knowledge is usually interpreted among experts. The opinions of several clinicians concerning the state of genetic knowledge of AD will be presented. The position taken by AD societies and the media with respect to current theories about the genetics of AD will be discussed. Ethnographic data based on interviews in progress with relatives of people who have been diagnosed with AD will then be presented. These findings show that even though AD is molecularized by basic scientists and many clinicians conceptualize the disease very differently from the way they did so a few years ago, thus far almost nothing has changed in actual clinical practice as a result of insights gained from genetics. This is also the case for the literature produced by advocacy groups and for families, as they confront the daily reality of caring for someone with AD. Any thought of being genetically at risk, or of shouldering a genetic “responsibility” is generally outweighed by numerous other exigencies for the majority of people whom we interviewed.

**Disease of the Century**

It is estimated by many experts that AD—a “living death” as some describe it—will strike between a quarter and a half of us by the time we are in our eighties. The range of these estimates demonstrates the fallibility of epidemiological data, but hyperbole is useful for maintaining funding. Even if the lower rate is a better prediction, very many of us are implicated as future patients, caregivers, or both, suggesting a remarkable challenge for social imaginaries of the future.

AD is ranked as the fourth-biggest killer in the United States, and the massive sum of $100 billion a year is currently spent on the care of 4 million AD patients in the United States. It is assumed that these costs will double as the baby boomers grow older, and by 2030 the number of cases will have tripled unless a “cure” is found. It has been reported that AD costs U.S. businesses more than $33 million in “lost productivity and absenteeism” and that these costs will soar as the baby boomers take time off to care for their elderly relatives ("Alzheimer’s Disease" 1998). Clearly, no lobbying is needed to encourage a search for a cure for this disease—very well funded research is under way to pin down the genes implicated in AD.

**Genetics and the Clinician**

*An Abundance of Genes*

Until recently, late-onset AD had been described as sporadic, because familial inheritance patterns did not appear to be at issue. However, ten years ago, an
event radically disrupted this perception: one particular allelic variation of the
gene known as apolipoproteinE (ApoE) located on chromosome 19 was shown
by linkage studies carried out in the laboratory of Alan Roses to be associated
with late-onset AD (Strittmatter et al. 1993). This finding has since been verified
in more than one hundred laboratories and has been described as the best dem-
"onstrated of all scientific findings in recent years. ApoE, already implicated in
heart disease before its association with Alzheimer’s was noted, is a polymor-
phic protein with three alleles, 2, 3, and 4, that appear to be universally but
unequally (clinically) distributed around the world. It is the ApoE ε4 variation that
places individuals at increased risk not only for contracting AD, but also for an
earlier age of onset of the disease by as much as seven to nine years. Despite
consensus on these findings, it is at the same time agreed that the allele deter-
"mines nothing with respect to the incidence of AD. All that can be inferred is
that the ApoE ε4 genotype confers a greater degree of susceptibility but that it is
neither necessary nor sufficient to cause the disease. It is assumed that combi-
nations of gene-gene and gene-protein interactions as well as interactions with
environmental variables must also be implicated in the onset of AD, and a great
deal of time and energy is currently being invested in establishing what contri-
bution these other variables make to the disease occurrence and its age of onset
(Tilley, Morgan, and Kalsheker 1998).

In 2001 a newly located susceptibility locus was reported in Science on chro-
mosome 10 (Myers et al. 2000). Other sites are currently under investigation to
locate yet more genes that may contribute in some way to AD incidence. These
sites are located on chromosomes 9, 12, 13, 15, and 19. In carrying out these in-
vestigations, researchers are attempting to do much more than pinpoint what is
happening at the molecular level once AD has become established; they are
attempting to demonstrate how risk of AD is increased by the presence of spe-
cific alleles, which are activated under as yet poorly understood conditions at
various points along biological pathways that are essential to normal brain
functioning. The excitement around these investigations is palpable, and the
molecularization of professional understanding of AD is undeniable.

John Hardy, the chief of the genetics laboratory of the United States Na-
tional Institute of Aging, stated during a presentation at the 2002 biannual AD
conference held in Stockholm that “genetics underpins our understanding of
this disease AD,” adding that “findings from genetics are the baseline for re-
search into AD.” But Hardy’s presentation, and others like it that focused on
genetics, while they received a great deal of attention at Stockholm, caused only
a limited stir. The reason is that, including the ApoE ε4 discovery ten years ear-
lier, no findings that derive from knowledge about the genetics of AD have as
yet resulted in clear advances of any kind in the prevention or treatment of the
disease. Similarly, there is limited predictive value of knowing one’s ApoE sta-
tus because of the difficulty of converting this knowledge into useful informa-
tion for clinicians and individuals attempting to relate individual status to future risk of developing the disease.

Estimating Risk: Epidemiological Shuffling of “Knowledge in Flux”

Research in connection with late-onset AD is amply demonstrating that genes are unequaled shape-shifters, the products of both evolutionary and recent human history and, at times, of toxic environments or of serendipitous mutations as a result of faulty replication.

Population genetics must be relied upon in attempts to establish what characteristics place certain populations at an increased risk for AD. The problem with assessing individual risk from these types of studies is no different from the difficulties inherent in the application of large-scale epidemiological information to any specific case; epidemiology is not “about” individual cases, but “about” populations and probabilities. In addition to this basic problem, there are several other sources of potential misunderstanding.

Since 1993, a large number of population studies have been published on the ApoE gene and its relationship to AD in which the focus has been on so-called Caucasians (Growdon 1998; Korovaitseva et al. 2001; Roses 1998; Saunders 2000; Silverman et al. 2003). Inconsistencies about the effects of the ApoE gene within this literature have the potential to cause confusion. For example, estimates of the number of individuals with AD who carry the ε4 allele range from 30 to 90 percent (Liddell, Lovestone, and Owen 2001; Ritchie and DuPuy 1999) and many studies do not specify whether these numbers refer to those who are hetero- or homozygous, further confounding the matter. In addition, researchers report that between 23 and 68 percent of AD patients do not have the ApoE ε4 allele, serving to highlight the complex and elusive nature of the association between susceptibility genes and the pathology of AD (Farlow 1997).

In addition to retrospective studies of individuals who already have AD, many studies attempt to estimate the number of people with ApoE ε4 alleles who will eventually develop AD. There is considerable variation between the estimates presented in these prospective studies. Depending on the study consulted, the number of individuals who are heterozygous for the ApoE ε4 allele and who are expected to develop AD range from 7.6 to 47 percent. The range for homozygous individuals is between 21.4 to 91 percent (Holmes 2002; Farlow 1997). For a healthy person to be given a 91 percent chance of getting AD in the future no doubt creates a significantly different anxiety level from that of a 21 percent chance.

In contrast, there is better agreement that individuals with ApoE ε4 alleles have an increased relative risk of developing AD. The literature suggests that a person with one ε4 allele has three times the chance, and a person with two ε4 alleles has between eight and thirty times the chance of developing AD compared with someone with no ε4 alleles (Holmes 2002; Swartz, Black, and St.
George-Hyslop 1999). However, the baseline on which this probability is estimated is rarely provided, and without this information, relative-risk estimates are highly misleading, although they still have the power to create anxiety.

Other studies report that individuals with first-degree relatives suffering from AD have between a 10 and 50 percent chance of developing AD by the time they are ninety (St. George-Hyslop 2000). In this case, the broadness of the risk estimate seems to be so wide as to be of little value for individuals, particularly given that the majority will have died of some other cause before age ninety.

For those individuals with two ApoE ε3 alleles (about 60 percent of the population in Europe and North America), risk for AD is estimated as “average,” and it is calculated that about a quarter will develop AD once over the age of eighty. Relatively few people carry ApoE ε2, although it is proportionally more frequent among several populations other than “Caucasians.” Those who inherit two copies of this gene are thought to be at very low risk of contracting AD, and this allele appears to be protective, in contrast to the ApoE ε4 allele.

One of the principle causes of confusion about genetic risk for AD is inherent to the research design. Holmes (2002) and Ritchie and Dupuy (1999) suggest that in many studies, present numbers do not represent the population at large, because they are based on clinical samples.

The emphasis on ApoE ε4 in research literature obscures the fact that many other factors have been associated with late-onset AD (including other candidate genes, head trauma, environment, diet, and lifestyle). Further, ApoE ε4 has been shown to work in unexpected ways in certain populations. For instance, among Pygmies, and other populations whose subsistence economy was relatively recently predominantly that of hunting and gathering, ApoE ε4 apparently protects against AD. This finding holds when controlled for age (Corbo and Scacchi 1999). Low rates of AD have been reported for parts of Nigeria, and the presence of an ApoE ε4 allele does not appear to be implicated when it does occur. However, ApoE ε4 is significantly associated with AD among African Americans, although less so than in populations of “whites” (Farrer 2000). It is argued that other risk-reducing factors (in Africa) and risk-enhancing factors (in North America) must therefore be implicated, including no doubt diet and environment and other genes and their protein products, but researchers also acknowledge limitations to the research methodologies used thus far. Clearly the specific role of ApoE ε4 in AD is far from perfectly understood.

The bottom line is that individual risk assessments for late-onset AD that make use of genetics are at present so vague as to be deemed by the majority of clinicians and researchers to be of little or no use in clinical care (Farlow 1997; Liddell, Lovestone, and Owen 2001; McConnell et al. 1998; St. George-Hyslop 2000; Tilley, Morgan, and Kalshaker 1998). Although it is acknowledged that this situation may change in the future, currently, official guidelines put out by professional and health-policy-making institutions and organizations, and by
advocacy groups in the United States, Canada, and the United Kingdom, state that genetic testing for ApoE status should not be carried out routinely. This is justified especially because there is no known prevention or treatment for AD that is more than minimally effective.

**Clinicians’ Practice and Knowledge in Flux**

AD is frequently used in departments of genetics today as a model for teaching genetic complexity and for illustrating gene-environment interaction. The general consensus among medical professionals about the contribution that an ApoE ε4 allele makes to increased risk for late-onset AD has brought about a fundamental shift in thinking that is not as yet reflected in the media or in advocacy literature (see below). As noted earlier, what was until very recently understood as a sporadic disease—as happenstance—is now considered by the majority of medical experts to be genetically transmitted, if at an uncertain rate. The assumption is that a great deal more remains to be learned about the genetics of AD and that when these advances are made, we may then be in a position to make some major breakthroughs with respect to medications, perhaps in the form of pharmacogenetics (Hedgecoe n.d.). However, this shift in theoretical perspective does not as yet in any way affect patient management. The following are excerpts selected from interviews with twenty-eight clinicians who practice in Canada, the United Kingdom, and the United States, carried out between 2000 and 2002.

One of the interviewed clinicians described his position:

It’s very hard now to talk about sporadic—nongenetic—Alzheimer’s. Sporadic would be rare—and even then genes are involved—the result of a sudden isolated mutation but not passed on in families. Until recently we thought that no genes were involved in this disease, that it was just aging—but now we know there are: genes are involved with aging and with the pathologies associated with aging. But genes are not causative—it’s just that genes increase the risk, or accelerate a process that is already under way. This process would not get expressed as pathology unless genes like ApoE ε4 are present, so that the “normal” changes of aging are transformed, resulting, for example, in an excess of plaques.

An eminent molecular geneticist who is also a clinician stated:

Genes for Alzheimer’s are not for anything at all, they simply contribute to one’s risk for getting late-onset Alzheimer’s. We’re talking about common polymorphisms, not mutations, and there’s going to be at least four to six of them involved with Alzheimer’s—maybe more. They could have arisen simply by chance, or it could be a bit like cystic fibrosis or sickle cell. In
the case of Alzheimer’s, the gene protects you from something (we don’t know what, of course) during most of your life, but you pay the price later on because of the long-term negative effects it has on the body.

A third clinician, although he does not dispute the importance of genetics, sounds a cautionary note:

All we can do is make a judgment of likely cause. Genetics are one influence among many leading to dementia. I’m very skeptical about the whole genetic frenzy—there’s so much hype and no money left over for improved nursing homes and home care. We simply don’t have a coherent story for why dementia happens—we have fashions in theories about it, that’s all. The more genes they find, the more reasonable and sensible the story will become because of course genes, or their products, are involved with just about everything that happens to the body.

A geriatric epidemiologist openly challenges the idea of AD as a disease.

As far as I’m concerned, genes are just other variables. They are markers, inherited markers, that have to be put into the pot for analysis along with other variables to see how they look. The great mistake with AD is to assume that because there is one end point there is one cause and one treatment. I don’t think AD is a disease. I think we should be talking about the brain and about dementia because now we know that virtually all dementias are mixed.

One clinician only, a psychogeriatrician, when focusing on incidence, chose to emphasize the effects of lifestyle in clear preference to genes:

You can have Alzheimer’s without predisposing genes—the genetics are overblown. Alzheimer’s is clearly linked to lifestyle. It all depends on where you work professionally as to what you think about cause, prevention, and best treatment. I think that lifestyle changes are crucial—there is evidence that exercise lowers incidence, and diet is crucial because cholesterol is involved in plaque production. People with diabetes are vulnerable.

In none of the clinicians’ accounts is a naked language of genetic determinism made use of in discussion of late-onset AD. Although all involved researchers and clinicians recognize the contribution of the ApoE ε4 allele, this knowledge has no effect on the clinical care they offer. Given these conditions of uncertainty, a consensus does not exist among health care professionals dealing with late-onset AD about the utility of current genetic knowledge in connection with this condition and not one believes that genetic testing for ApoE ε4 should be used routinely in the clinic.14
The Transfer and Translation of Knowledge in Flux

In this section we focus on how information about risk factors, assumed mechanisms of the disease process, theories of causation, and therapeutic options are portrayed in various forums in order to understand what types of information reach AD sufferers and their families and caregivers.

A common assumption held on the part of health care professionals interviewed in this study is that most, though not all, media reports create unwarranted hype about the possibility of curing life-threatening diseases once the involved genes are located. This claim has also been made by social scientists who have argued that there has been an increase in genetic determinism and reductionism by the general public, whose ideas have been largely shaped by media representations (Nelkin and Lindee 1995). However, a number of social scientists critical of Nelkin and Lindee’s statements have pointed out the anecdotal nature of many of their claims and emphasized the need for thoroughly grounded empirical studies on this issue. In fact, contextualized studies by such critics demonstrate that genetic determinism is perhaps not as strong as has been suggested and that recent discourse about genetics in the media is more nuanced than was previously assumed (see, for example, Miller 1995; Condit 1999; Condit, Ofulue, and Sheedy 1998; Hedgecoe n.d.). Our research tends to support this latter perspective.

The sensationalism of the claims made at times in media headlines cannot be denied, but in order to judge media reports, careful reading of their content is required. For example, the manner in which genetic knowledge is presented varies in complexity and in the weight granted to it as compared to other variables implicated in disease causation. Moreover, the popular media is not the only source of information about genetic knowledge directed at the public. Systematic empirical examination of multiple sources of knowledge about genetics is needed for full evaluation of the way in which scientific knowledge is translated and transmitted to the public. To this end, we begin our study by examining discourse about genetic risk and AD that has appeared over the past decade within three bodies of literature: popular media (newspapers and magazines), educational materials produced by pharmaceutical companies that is distributed through clinics and by general practitioners, and the literature produced and distributed by advocacy and support groups.15

Media

It has been shown that as scientific data are translated for use in the media, a process of simplification and homogenization frequently takes place (Conrad 1999) and that “user-friendly” vocabularies are made use of. Conrad (2001) has further analyzed the manner in which media coverage of genetic discoveries tends to fall into a “frame” of genetic optimism, in no small part because of the
style of journalistic writing and editorial concerns. Good news is always in short supply, and coverage of genetic discoveries and their associated promise, no matter how remote, are easily deemed newsworthy and make it past the editorial cutting-room floor. By contrast, stories that tell of retractions or discredited scientific studies stand little chance of making it to print, except in specialty science magazines.

To a certain extent, Conrad’s observation that media coverage of genetic knowledge tends to optimistically herald the promise of new discoveries and to oversimplify, or even misrepresent, the studies on which their stories are based, holds true for some of the media coverage of the science of AD. However, while “AD-gene stories” may gloss the scientific knowledge surrounding AD and make bold headlines, they do not necessarily fall into the trap of presenting simple stories of genetic determinism or pipe-dream cures, as described by Nelkin and Lindee (1995). In fact, rather than focusing on genetics, the majority of news coverage of AD concerns itself with caregiving responsibilities, celebrity victims, and studies that explore “nongenetic” theories of causation. Genetics is but one small part of the picture of AD that has been portrayed over the past decade. While the media may be guilty of promoting genetic optimism in certain headlines and in a limited number of stories, especially older ones, it is not at all evident that this is reflective of media coverage of the disease in general.

One reason that the genetics of AD may no longer have a central place in newspapers and magazines is that the media tends to have peaks of interest in studies presenting novel findings at the time of their initial “discovery.” As Conrad (2001) predicts, there is a saturation point at which time something is just no longer news, and unless a new research angle is presented regarding a disease, its coverage in the media will dwindle. Within the media coverage, the promise of genetics has been displaced by the most recent findings from scientific studies that do not relate directly to genetics. Recent media stories have focused upon possible prevention of AD through cognitive and dietary strategies, experimental vaccines, and the promise of early detection through monitoring of homocysteine blood levels or by means of brain scans.

When the genetics of AD are mentioned today in newspaper articles, such reports are more often than not cautious and nonspecific: “It is unclear what causes AD, though it is likely a combination of genetic predisposition and environmental factors” (Picard 2002). Others clearly state that while researchers have identified genetic links to a number of diseases—of which AD is listed as but one—the inheritance of a genetic mutation does not guarantee that a disease will develop, only that there is an increased susceptibility as additional genes and environmental factors also appear to be implicated (Andrews 2002).

In recent media coverage, the ApoE ε4 gene is usually mentioned in connec-
tion with findings from other studies. For example, the New York Times reports that the protective effects of Vitamin E are not observed in those “with a gene variation apolipoprotein ε4, which has been linked to Alzheimer’s” (Kolata 2002; emphasis added). The involvement of genetics, regarding the same finding about Vitamin E, is reported in another newspaper as “the ApoE ε4 gene, which indicates an inherited risk of developing Alzheimer’s” (Immen 2002; emphasis added). Other articles report on studies showing that high-fat diets increase sevenfold the risk for those who are genetically predisposed to develop AD (“Les graisses” 2000). It appears that in newspaper coverage the linkage of ApoE ε4 to AD is understood as a given, but its role is portrayed in subtly different ways and in association with other variables.

The most problematic element observed in media coverage of AD and genetics was figures and estimates of incidence and risk that were uncontextualized. For example: “Those who carry a certain genetic characteristic have 16 times the risk of developing Alzheimer’s disease” (“La génétique” 2001). Or, as claimed in Time: there is a 91 percent chance that someone with two copies of an “Alzheimer’s-related gene will develop Alzheimer’s” (“Numbers” 2000). This failure to contextualize figures and the misrepresentation of estimates of risk are not confined to media coverage, as the source of figures quoted in the media are most often taken directly from involved scientists, or from spokespersons for or media releases supplied by advocacy groups such as the Alzheimer’s Society and the Alzheimer’s Organization.

Therefore, while these stories tend to reflect a certain simplification of details, as predicted by Conrad (2001), upon careful reading, the media cannot be accused of creating unwarranted hype about an imminent discovery of “cures” for or ultimate causes of AD.

Pharmaceutical Educational Materials

Educational material about AD produced by pharmaceutical companies is heavily relied upon by physicians and nurses when explaining AD to newly diagnosed patients and their families, but these information sources adroitly sidestep theorizing about causation: “In some cases, Alzheimer’s disease may run in the family. In other cases, no other family members are affected . . . researchers generally believe that the cause may be a combination of several factors. They are working very hard to find out more about the causes of Alzheimer’s disease” (Pfizer Canada 2001, 2). While theories of causation in general, and more particularly of genetics and heredity, are given scant attention in the pharmaceutical educational materials, when they do appear, they are usually glossed over as “family history,” and there is a failure to clearly distinguish between early- and late-onset Alzheimer’s disease.
The few allusions to genetics that appear in pharmaceutical-company educational handouts are usually drawn from other sources, such as the Canadian Alzheimer’s Society’s *Alzheimer’s Disease and Heredity Fact Sheet* (2002). Pharmaceutical handouts also direct patients and caregivers to “educational” Web sites that are more detailed in their discussion of causality and risk. Here ApoE ε4 makes a brief appearance: “Recent medical research suggests that people who carry a specific gene variant—apolipoprotein (ApoE), ApoE ε4—may be at increased risk of AD after age 60. Research is being undertaken to further explore this possibility.”

**Advocacy Organizations**

In addition to drawing on the material distributed by pharmaceutical companies, physicians routinely refer patients to local support and advocacy groups such as the Canadian Alzheimer’s Society or the American Alzheimer’s Association. These organizations offer a broader range of information than can be found in the general media or through the pharmaceutical-industry-sponsored Web sites, in large part because such organizations have mandates to both support and make available “state of the art” research and to convey this information in a manner that is accessible to patients and caregivers, as well as to the research and clinical communities.

Despite their mandates to pass along the newest information, the Alzheimer’s Society statements with regard ApoE ε4 are cautious and qualified: “[S]cientists now believe, *in some cases*, the pattern of inheritance of the ApoE ε4 allele can be used to estimate subsequent risk, and age of onset.” The Alzheimer’s Society specifies that while ApoE ε4 has been linked to various forms of the disease, it is clear that the allele does not *cause* the disease but rather is a potential risk factor. Risk estimates provided by the Alzheimer’s Society are also vague: “[T]hose with 2 copies of ApoE ε4 will have a higher risk of developing the disease in their early sixties, while those with 1 copy of the allele will more probably develop AD after the age of seventy” (emphases added). No concrete figures are provided. Given that relatives of AD patients are not routinely tested for their ApoE genotype, it would be interesting to know if such vague statements aroused an interest in genetic testing among relatives of AD patients. An unspecified “higher risk” may be cause for concern, as might be the prospect of “more probably” developing AD, even if only in one’s eighth decade of life. But such statements could equally have the opposite effect.

The Alzheimer’s Association’s Web site offers general estimates of average worldwide lifetime risk for developing any type of Alzheimer’s: (5 percent by age sixty-five, 10–15 percent by seventy-five, and 20–40 percent by eighty-five; having one parent or a sibling doubles risk to 10 percent by sixty-five, 20–35 per-
The authors state that risk increases with the number of affected relatives, and having more than one affected sibling results in the greatest increase in risk. “This is explained through shared genes, whose influence may be large or small—deterministic genes, in the case of FAD (familial AD). In the case of sporadic AD ‘susceptibility’ or ‘risk’ genes—they raise the likelihood of a particular outcome but do not ensure it” (Alzheimer’s Association 2002).

The association cautions, “Analyses of the exact amount that ApoE ε4 raises risk are inconsistent, and the precise mechanism by which the ApoE genotype affects risk is not yet completely understood” (Alzheimer’s Association 2002). What the society fails to point out is that the general-risk figures provided by lifetime estimated risk are not well defined in the scientific literature. For example, included in lifetime estimated risks of succumbing to AD are not only those diagnosed with late-onset AD, but also those with early-onset AD. Further, as with the materials produced by pharmaceutical companies, these pamphlets at times conflate early- and late-onset forms of AD: “For a few families, there is a definite connection between family history and AD. While for others, a family history of Alzheimer’s Disease puts them at a greater risk than someone with no family history” (idem.). Such a statement does not clearly differentiate between the two forms of AD, which have very different meanings and outcomes for afflicted individuals and their family members.16

MUCH OF THE information and commentary that makes its way into pharmaceutical educational materials and media coverage is based upon the information provided by organizations such as the Alzheimer’s Association and the Alzheimer’s Society. The groups are often quoted in media accounts to bolster and confirm the importance of new scientific studies and findings concerning AD. These sources of knowledge are also primary channels for information about the nature and causes of AD. We suggest that the import of these sources is probably as significant as that of the clinician in providing information about AD to patients, family members, and the public at large.

Participation in an eight-week support group for families of patients with AD confirmed the impression gained from the advocacy literature: the care of patients is given top priority for discussion in these organizations, and the presumed causes of AD, including the possible contribution of genetics, is granted only a minimal amount of attention, if more than is available through other sources. Our interviews with family members of AD sufferers make it clear that a very discriminating reading of what is presented in the media and as the latest “breaking news” is common; readers critically assess the importance of “discoveries” and most often recall only those articles that are directly related to their preexisting interests.
Disparate Uptake of Knowledge in Flux among Families of Alzheimer’s Patients

We began our study of the circulating information on AD causation and genetics with the assumption that this knowledge flowed from basic science and epidemiology through the clinical setting then through pharmaceutical, advocacy, and media representations, finally “trickling down” to the public at large. However, what was discovered was characterized less by “flow” than by disjunction, translation, and a disparate uptake of this fluctuating knowledge. Interviews carried out in Montreal with the children of AD patients indicate that the children’s concerns and beliefs about AD are no more a simple reflection of the information available in the media than they are a result of current epidemiological or clinical knowledge.17

While many were quick to state that they kept abreast of media coverage of AD, it was unusual for respondents to recall in any detail the stories that they had read or watched on television. Furthermore, their statements did not indicate that they necessarily remembered or put their faith in information given to them in clinical settings. Pharmaceutical and advocacy information pamphlets were more likely to be sources of information on daily coping and care than on AD causation. For the most part, respondents’ concerns were focused on the day-to-day management of their parents. Learning to cope with a sick and dying family member was much more pressing for them than was thinking about one’s own risk of developing AD.

These findings contradict the writings of those social scientists who have indicated, in relation to “genetic” disorders, that people tend to worry about genetic risk, that this risk will be central to their life choices, and that knowledge about genetic risk will play a role in increasing self-surveillance. Clearly, the specific pathologies and prognostic effects of the disease under consideration limits what generalizations can be made. In what follows, we present several of the most common themes that have appeared in interviews in connection with attitudes about AD and beliefs about its cause(s).

Clinical Encounters

Many respondents indicated that their parents’ clinicians did not add substantially to their knowledge about the causes of AD, for the simple reason that doctors rarely took time to discuss potential causes of the disease. Instead, most of the discussion tended to center around diagnosis, therapy, and the immediacy of caregiving. Respondents said that they spent time with the doctor inquiring about the stage of their parent’s disease, prognosis, and daily care.

Replies to the question “What did the doctor tell you about AD?” usually took the following form: “That it is progressive, that it would simply continue and there will be a point in time that my mother will need another type of
care . . . that it is not reversible; there are some medications that might help stabilize it to some degree, but that none of these are . . . there is no cure. So it is a progressive, degenerative disease” (Ari Rosen, age fifty-three, salesperson).

While sitting in on doctor-patient appointments, we observed that the monitoring of patients’ complicated drug regimens, often including multiple medications to slow disease progression, in addition to others prescribed for concurrent conditions, often consumed virtually all the available time, making it difficult to engage in in-depth discussions about disease causation or other matters.

However, in most cases respondents reported that their parents’ clinicians had at some point suggested several potential causes of AD, none of which was granted much more weight than the others. Clinicians reported that they were particularly uncomfortable discussing potential genetic causes of AD, since so little was known about their influence on risk assessments and because this information might result in unfounded concerns among patients. Answers such as the following were commonly given to the question “Did your doctor tell you anything about cause or prevention?”

I don’t inquire very far into the doctor’s knowledge base. They don’t know, they don’t know exactly, the doctors, do they? I don’t think so. They research and research, but they don’t know where it comes from” (Françoise Boisvert, age fifty-eight, hairdresser).

I can’t say that we were overwhelmed by medical evidence. It’s not like we get the feeling anybody really knows. With there being so many questions out there, as to what is AD. It’s a catch-all term for many illnesses. What is it? Unless you can even grasp why it happens, I guess to an extent, then you aren’t doing really more than just giving it a name.” (Hélène Talbot, age thirty-six, lawyer)

Diseases Run in Families

Although most respondents were not aware of any specific genetic risk factor involved in the development of AD (ApoE was not once introduced spontaneously by respondents into the discussion), those who did speak of a genetic component to AD usually thought of it in one of two ways. They either stated that they probably had a fifty-fifty chance of developing AD (as a result of inheriting the gene from their affected parent) or that this parental gene makes an unknown contribution (the increased risk is uncertain and possibly quite small). The vast majority of respondents usually presented their understanding of intergenerational transmission of AD in terms of familial tendencies; in other words, anything one’s parents “had” had a chance of being passed along. For instance, it was commonly explained that one could inherit from one’s parent
personality, eye color, or a tendency toward a particular sickness, but when respondents were asked to describe their understanding of hereditary transmission, they resorted to statements such as “Certain traits are in the family”:

I have always said to myself that it’s hereditary. Because there is so much of it in the family, it makes me think that it’s hereditary. (Françoise Boisvert, age fifty-eight, hairdresser)

What I understand about this disease is that . . . I think . . . that it is inevitable. It transmits from parents . . . that is what I think. . . . My fears are important. My mother often said to me, “Look at me, I am seventy-seven and I am in good shape, you can hold on to that.” And that’s right, except that I have a 50 percent chance of getting the disease because my father suffered from the disease. So I have worries about the fact that I could potentially have the disease. This worry doesn’t prevent me from functioning or anything. I just think about it. It is just something that could happen because my father had it and because other members of the family had this disease . . .

I wouldn’t say that inheritance of AD was automatic, but the percentage is larger. But I wouldn’t say that it was automatic. It is sure that if you look at the percentages, if there is someone close to you, like a mother or father, you have more chances of having it, but I wouldn’t say 100 percent. The chances are probably just higher. (Nicole Boucher, age fifty-two, office worker)

It’s possibly not transmissible but probably I have more chances to have it than my neighbor who doesn’t have it in his family, yes. I try not to think about it though. This topic isn’t on my mind each day. (Jean-Pierre Côté, age thirty-nine, lawyer)

Some respondents reasoned that if they shared other traits (especially personality) with their affected parent, then it was more likely that they would have inherited the AD trait from that same parent:18 “My mother worries more about my brother than about me. She thinks his personality is more likely to be similar to hers than mine. My brother and I have talked a bit about that” (Celine Goulet, age sixty-two, college professor). Or they suggested that they had had enough bad luck in the form of other diseases that they presumed were inherited from their parents to ensure that it was unlikely that they would also inherit AD: “My father died of a sudden heart attack, and that would be my choice. And there’s Crohn’s in my father’s family and I somehow think: OK, if I picked up that gene, how likely is it that I have the AD gene, too. Can I have them both? I hope not” (Celine Goulet, age sixty-two, college professor).

Several respondents had an understanding of the genetics of AD that mir-
rors that of the geneticists who were interviewed: “Do I think that there is a genetic component . . . yes! I think everything has something genetically related, but I think there could be a propensity towards whatever it may be, could be asthma, heart disease, AD, cancer, and I think there’s others factors that may make it surface or not” (Celine Goulet, age sixty-two, college professor).

Many respondents suggested that they made an effort not to research or think about AD, its causes, or their chances of getting the disease. Their explanation for this response was that because no cure existed, they were not interested in estimating their risk for AD, since they would not be able to do anything with the information: “Dr. Jefferson once said to me, ‘There is a test.’ I said, ‘Do you have a cure?’ He said, ‘No.’ I said, ‘Then why should I take the test? Because if I took the test I could go straight, if the results were not to my liking, I would go straight to the Jacques Cartier Bridge and jump!’ I know that my greatest fear is I don’t ever want to put my husband or my children through that. But I don’t see the point of knowing” (Esther Blumberg, age fifty-eight, homemaker).

Others stated that they had concerns about other diseases that might either threaten their future health or with which they or a family member were already struggling that were of more immediate import in their lives: “We have other medical histories that I have to deal with in my family, my father as I said had colon cancer, my mother had lymphoma, my mother’s late brother had lymphoma, so there is clearly some other risk profile in the family that I’m more concerned about than I am about the AD actually” (Ari Rosen, age fifty-three, salesperson).

A Good and Active Life

The importance of “good living” is a common theme that comes out in many of the interviews. Virtually all respondents suggested that they tried to live a healthy life by reducing stress, by exercising, and by eating well. Some, responding at times to medical recommendations, took vitamins, and others used or considered use of hormone replacement therapy (though most were aware of the recent stopping of clinical trials in connection with this treatment). The detrimental effects of stress were acknowledged and the importance of an active lifestyle, social and cognitive, was frequently mentioned as likely to be protective factors against AD. But people were well aware that good living was unlikely to stave off AD forever: “Well, prevention . . . I mean, I’ve gone to lectures and everything on Alzheimer’s and they say, you know, keep your brain working, keep your brain active, try and do things. But you know, my mum worked her whole life, she played bridge, she played mah-jongg, you know, she did everything. Her mind was working all the time, so you know, how do you figure that one out? There is no answer, you know? She was very active her whole life” (Ethel Goldman, age fifty-six, homemaker).
Aluminum . . . and Other Theories of Causation

When asked directly what they believed may have caused their parents’ AD, respondents were likely to postulate causes that did not originate from clinical encounters; and in most cases, these theories had not been “run by” their clinicians. These speculative theories apparently arose from efforts to reconcile many pieces of information on risk factors that respondents had accumulated from various sources. Head injuries, aluminum, stress, depression, diet, alcohol consumption, and lack of exercise and cognitive stimulation were liberally evoked as causative. On the whole, respondents held to complex explanations about who gets AD, and why, which they kept separate from unfocused ideas about the hereditary aspect of the disease. In other words, many people intuitively held theories of multiple causation and implicitly recognized a complexity, over which they may have some limited control. Secondarily, inherited traits, over which there can be no control, may also make a contribution. No doubt for this reason, their significance was not given a great deal of prominence:

You know, my mother never exercised. She never dieted. She never had to diet, thank goodness. She ate everything, and she always prided herself on eating fatty foods and ice cream and fried things, and she never had to lose weight. And my mother’s brother, who was also brought up in the same household, same thing, he never did anything to watch his weight or watch his diet, and they never exercised. So now I’m thinking, maybe that was one of the reasons, who knows? (Ethel Goldman, age fifty-six, homemaker)

My mum got divorced about twenty-five years ago . . . and then her world kind of crumbled, and at that time too her job she was doing—she had only worked in the workforce a couple years, but her job was eliminated, the company moved and so she babysat for me full time. But she became more and more reclusive in terms of less social activities, less outings, and I think really, lack of stimulation, lack of outside interests, beyond the family not a lot of friends, so possibly lack of stimulation and social activity. (Nancy Robertson, age forty-two, nurse)

My mother had an accident. We have a country place up north, and she was cutting wood one day—this is all before I even brought her to the doctor—and she got hit by an ax. She hit the wood and the wood hit right here [on the head]. She became blue, black, her eyes, everything. So I always thought that this . . . maybe this was the cause of it. (Rosa Bellini, age fifty-four, factory worker, retired)

I think of environmental factors, OK. I think of the pollutants in the air and I am very neurotic about things like that in general. So I would just
write that [AD] off as another example of living in a highly industrialized
city, where there’s a lot of pollutants. My mother didn’t live near high-
tension wires, but close to the highway for most of her life, so I . . . I mean
that’s also what my focus is, and I don’t know about her early childhood,
what may or may not have been a factor, on her own personal level. Until
she remarried it was high stress. I don’t know if that contributes or not, for
me it’s guessing, really. (Celine Goulet, age sixty-two, college professor)

Hearsay or Heresy

The sources that contributed to these “lay” theories of causation were difficult
to pin down. When pressed for detail, respondents claimed to have read some-
thing somewhere or seen something on TV, to have been just guessing, or to
have often made extrapolations from other illnesses and widespread discourse
about health and disease prevention. It was rare that respondents would be able
to specifically recall whether the source of information had been the media, an
advocacy organization such as the Alzheimer’s Society, their doctor, or commu-
ication with others with some intimate experience of AD. And yet these di-
verse, vaguely recalled pieces of information seemed to have had as much or
greater influence than the actual clinical encounter in shaping respondents’
perceptions of AD and the role that they believed genetics might play in the
development of the disease.

While only a few respondents could identify the sources of their theories of
disease causation or of particular knowledge about AD, circulating speculations
about risk factors and possible protective strategies had clearly penetrated the
minds of family members. Although none mentioned folic acid or the reported
benefits of Vitamin E, many had heard that staying “cognitively active” might
help, as might regular ingestion of anti-inflammatories, and many are familiar
with the debated benefits of hormone replacement therapy in women. There is
little doubt that hearsay was at work. Does this selective uptake and use of
knowledge about AD, with its subjugation of genetics factors, amount to heresy
against current scientific knowledge?

Knowledge in Flux

Interview responses clearly indicate that first-degree relatives of patients diag-
nosed with AD were generally not very interested in learning about the causes
of the disease. They were unlikely to push their clinicians for detailed informa-
tion about the science of AD. Nor were they searching for ultimate causes and
exhibited a more than healthy skepticism of the scientific and genetic discover-
ies they stumbled across. They were well aware that the discoveries announced
in the media did not always prove to be accurate and that promised advances
were slow to materialize. Most respondents said that they were not interested in learning about their risk of developing AD until there was something they could do about it, something more than maintaining a healthy lifestyle. They trusted that the clinician following their parent, with whom they had regular contact, would keep them abreast of what they needed to know.

It might seem surprising that so few relatives of AD patients were interested in understanding the cause, or causes, of the disease or in estimates of their own risk. But perhaps this is not so surprising in light of the fact that all those people interviewed in Montreal were in the throes of caring for a sick parent. These people suggested that the possibility of their developing AD was far away and not nearly as immediate as were their concerns about the illnesses with which they were currently dealing—be it their own Crohn’s disease, their spouse’s cancer or heart disease, or their parent’s AD. While stating that they saw their risk of developing AD as something far off in the future, most also intimated that they did not want to think in terms of future risk. This might be a situation in which pragmatic disregard has a place, since circumspection does not lead to circumvention.

These interviews have helped to identify some of the factors implicated in the uptake, or lack thereof, of genetic knowledge. There is no simple “trickle-down effect” at work here. Instead, we observe an extreme form of incomplete penetrance of the current genetic knowledge surrounding AD. Despite the fact that genetic researchers have uncovered one gene that is indisputably associated with an increased risk for AD, respondents, when they spoke about heredity, did so in a simplified fashion with no reference to ApoE. They recalled having read about genes in newspapers, hearing about them in public lectures, and being told certain things by their physician. However, the knowledge was recollected in general terms (quite possibly as it was given to them) and was positioned alongside other speculative theories. Interestingly, respondents more often introduced aluminum exposure as a possible risk factor than they did genetics, and if they read about aluminum, they likely read about ApoE ε₄, since these two items hit the general media at roughly the same time, ten years ago. This suggests a selectivity in uptake and use of knowledge that has not been emphasized in previous social science studies of the public understanding of science. It is possible that the apparent immediacy of poisoning by toxic metals, along with the fact that one can relatively easily choose to avoid this purported source of AD risk, creates more concern than does the possibility of risk estimates based on genotypes, which one can do nothing directly to modify.

Even though media headlines have announced a genetic component to AD, the content of the articles tends to stress the complex interaction of genes, the environment, and yet-to-be-determined cofactors. However, the majority of current popular media coverage of AD is concerned with the shortage of home
care for sufferers, or with the latest studies concerning anti-inflammatories or folic acid. Genetics is but one small part of the picture portrayed in the popular media, and these articles are often less striking to afflicted families than other articles, of which there are many, that emphasize the lived reality of the disease.

Clinicians are located at a major point of disjunction in the transfer of knowledge concerning genetics and AD. Certain clinicians, such as those in specialty memory clinics, are conversant with cutting-edge genetic science and are active researchers, but they do not transmit this information to their patients, because in their opinion it has no effect on clinical care or recommendation about family care of patients.20

The goals and purposes of the clinical encounters at which family members are present need to be held in mind: the interview respondents were not patients suffering from AD and did not necessarily perceive themselves as potential patients even in the future; rather, they were caregivers. It seemed to be the general opinion of our informants that to engage in speculation about susceptibility and possible risk for themselves would be inappropriate in the clinical setting, where their parents’ needs took precedence.21

**Conclusion**

As a result of developments in molecular genetics, most health-care professionals now think that genetics is implicated in all disease processes, including those of complex diseases, such as AD, but they also recognize that multiple genes, proteins, and the environment are inevitably implicated. Our data show that there is a disjunction in the uptake of this knowledge, at least in connection with AD, by patients, families, and the public. Advocacy groups, authors of pharmaceutical literature, and clinicians themselves do not emphasize the implications of genetic findings to patients, families, and the public; on the contrary, they often withhold their professional position with respect to these findings, or else make clear their ambivalence about the clinical usefulness of genetic information. In the popular media, the “discovery” of new genes associated with AD are reported, but these are far outnumbered by those articles about the benefits of vitamins and lifestyle that may help to prevent the occurrence of this disease. Further, at least as often as prevention, caregiving is the subject of these reports. Similarly, support groups for AD will, for the foreseeable future, continue to focus on patient care rather than highlighting the numerous genes and proteins that have been targeted as contributing to the disease, whose predictive value is little or none.

Reasons for deemphasizing new genetic research within a clinical setting appear to be twofold. First, to date, the complex mechanisms whereby genetics
has an impact on AD disease causation are by no means understood. The effects of ApoE, which is the closest thing to a genetic “smoking gun” scientists have found, are anything but obvious. Knowledge of an individual’s ApoE status does not affect diagnosis, prognosis, or treatment and therefore does not make a difference in a clinician’s ability to care for that individual. Second, aside from withholding information about genetics and AD because of the limited predictive value of ApoE status, a further reason for the deemphasis of genetic knowledge is that caregivers in the clinical setting are most concerned and worried about the details of caregiving and what the future has in store for their parent. Concerns and speculation about their own future risk is actively avoided.

Professional and popular understandings of the role of genetics in illness vary from disease to disease, and so too do the form and degree of geneticization, genetic citizenship, genetic responsibility, and biosociality. In order to predict the effects that genetic knowledge may have, it is necessary first to understand and contextualize the state of professional and popular knowledge surrounding a particular disorder. Most important, what is relevant for Mendelian diseases cannot be extended to complex diseases in which susceptibility, rather than causative genes, are implicated. Further, it is necessary to recognize that different diseases will have very different effects on individuals, families, and communities. Some of the reasons for this include the varying age of onset, the form that the disease takes, and its particular pathological effects, along with differing perceptions among individuals about the disease. For instance, some respondents in this study indicated that their fears of developing AD were less than their fears of developing other disorders, because they assumed that AD would strike only very late in life. Nonetheless, others had grave fears of putting their children through the difficulty of dealing with a parent with AD in the future. Without detailed knowledge of these factors, it is not possible to understand what form geneticization or genetic responsibility may take.

Not only scientists, but also social scientists, risk participating in hyperbole about the new genetics when they focus attention on geneticization at the expense of considering the impact of the lived experience of disease on patients and families. Our research demonstrates that the transfer and uptake of knowledge and its penetrance into lay understanding of disease causation is in part dependent on its pragmatic value for the listener and its consistency with individuals’ broader beliefs about health and illness. Rarely did respondents suggest that their understanding of AD was significantly different from their understanding about other late-onset diseases; they posited that heredity and lifestyle were probably both implicated in AD, cancer, and heart disease. Beyond the understanding of some respondents that the disease might “run in the family,” genetics played a small part in perceptions about AD, taking a backseat to the pragmatics of living with, and caring for, an afflicted family member.
As the mapping of genes becomes increasingly sophisticated, it has become clear that risk estimates are very often lower than was previously believed to be the case, in some instances, considerably lower. What was formerly assumed to be certain is no longer the case (Almquist et al. 1997).

As a result of genetic research, AD is now conventionally divided into early- and late-onset forms. Early onset, “familial” AD, is associated thus far with approximately 170 extended families worldwide accounting for perhaps 2 percent of all cases of AD. Genetic markers for this form of the disease have been found on chromosomes 1, 14, and 21, one variation of which is inevitably present in vulnerable families. These genes are autosomal dominant mutations and are understood by most specialists as genetic “determinants,” although twin studies have shown that age of onset for identical twins can differ by as much as ten years, strongly suggesting that the environment also plays a role (Tilley, Morgan, and Kalsheker 1998). Onset of the early forms of the disease is almost without exception between the ages of thirty-five and sixty, with another form starting a little later in life and occasionally not making an appearance until age seventy. In all cases of early onset, the condition progresses rapidly to florid dementia and death.

Many genes are polymorphic and have a number of variations that are widespread in the human population. Those allelic variations that have been associated with an increased risk of developing a disorder are known as susceptibility genes. Such gene variants are neither necessary nor sufficient to cause specific diseases, however, as, compared with the population at large, an individual who carries one and especially two copies of such alleles is believed to have an increased risk of protracting the relevant disease. Even so, people with two copies of a susceptibility gene may not get the disease, indicating that other, as-yet-unidentified factors are involved.

Penetrance (a term coined by Vogt [1926]) refers to the frequency with which a gene manifests itself in the phenotype of carriers. Phenotypic expression is a function of both genotype and environmental factors. Penetrance is reduced or incomplete when carriers fail to demonstrate the associated phenotype.

A shift toward a molecular approach in biology began in the 1930s (Kay 1993). This shift was associated with a search for what constitutes “life,” and was made possible by the development of several new technologies. For two decades, molecular biology focused on protein structure and function. After 1953, when the significance of the discovery of DNA was recognized, the emphasis switched dramatically to genes, culminating in the Human Genome Project. In recent years proteomics has again become a major focus in molecular biology and is the subdiscipline that currently holds out the most hope for the development of new, individualized medications and for elucidating numerous complex biological pathways, including those associated with AD.

This Internet research is open to critique in that it represents only those people who have been motivated to turn to the Web sites. Others, even when they have “genetics” in the family, avoid such sites. There is no evidence that they exhibit a “genetic responsibility” or that they believe that it is necessary. It is also worth noting that only between 10 and 15 percent of people in Huntington families take up offers for testing.

Concepts of genetic citizenship, genetic risk, and genetic responsibility are likely to be useful when researching early-onset AD, which has a Mendelian inheritance pattern.

Epidemiological figures for other countries, such as Canada, are often derived from
estimates of the figures in the United States. With Canada having one-tenth the population, Canadian estimates are commonly assumed to be one-tenth of those in the United States. What is lost in mathematical gerrymandering are potential differences in disease incidences that might result from different social and economic circumstances such as very different medical and social service systems.

9. When a disease has a very late onset, as does AD, it is difficult to reconstruct family genealogies effectively to document disease transmission, with the result that disease occurrence appears sporadic.

10. At this same conference, an epidemiologist clarified the way in which genes are believed to be implicated from before birth in connection with what will happen to one in old age. Genes influence the building of “cognitive capacity,” this epidemiologist argued, starting in the intrauterine environment and playing a large role throughout infancy and childhood and into early adult life. During interviews, AD experts frequently claimed that people with high IQs and with extensive education are at considerably less risk for AD than are others. The epidemiologist’s presentation made it clear that what is assumed in such statements is that genetic predisposition influences the laying down of the neurological networks required for brain functioning. Certain biologically predisposed individuals will end up as adults with fewer synapses and, as a result, are likely to have lower IQs and therefore will complete less schooling. It is assumed that the plaques, tangles, and cell death associated with AD will do proportionally more damage in a short space of time to such people.

11. A recent New York Times article reported debate among specialists with regard to the limited efficacy of existing drugs, stating that it may be decades before real progress is made (Grady 2004).

12. The term heterozygous refers to the case in which a person carries only one ApoE ε4 allele (along with an ApoE ε2 or 3, for example). Someone who is homozygous for ApoE ε4 has two of these alleles.

13. In the examples, the involved gene has been shown to protect against a specific disease, but if individuals are homozygous for the gene then they will develop cystic fibrosis or sickle cell disease.

14. In the two clinics located in teaching hospitals with which we are most familiar, patients and their families are routinely asked to provide blood for the various research projects that are under way. Several of these projects involve testing for genes and proteins believed to be associated with AD in the hope of finding clues that could lead to the development of an effective medication. If the family agrees to participate in one or more of these studies, blood is drawn and the sample is then anonymized before it is sent for analysis. The results of such testing are not made available to patients, families, or clinicians, and therefore participation has no effect on clinical encounters. Should an effective medication be developed, only then would samples be rematched with specific patients.

15. This analysis is based upon database searches (Canada Newsdisc and Factiva [Dow Jones Index]) covering the above-mentioned time period, which cover major Canadian newspapers in French and English, such as the Montreal Gazette, La Presse, Le Devoir, Le Soleil, the Globe and Mail, the National Post, and the Toronto Star, in addition to popular magazines such as Canadian Living, Chatelaine, and Macleans and CBC news coverage. The Washington Post, the New York Times, Report on Business, Time, and Scientific American were also examined. (It was not within the scope of this analysis to examine British and European media sources, but a comparison of the manner in which media representation of AD and genetics might differ from the North American con-
text might prove an interesting cross-cultural comparison). Because of the multina-
tional makeup of the Canadian population, it is to be expected that many Canadians
will be exposed to foreign media sources. However, as the intent of this analysis is to
explore the media coverage that our “typical” respondent would have most likely
been “exposed” to, we have limited ourselves to these representative sources.

16. Familial Autosomal Dominant AD (FAD) is identified as rare (5–10 percent of all
cases), and “Sporadic AD” is described in the pamphlet as the common form, ac-
counting for 90–95 percent of all cases. It is noted, “The role of heredity in Sporadic
AD is unclear and continues to be the subject of intense research” (Alzheimer’s Soci-
ety 2002).

17. This study was based on interviews of 40 first-degree relatives whose parent was suf-
f ering, or had suffered, from late-onset AD. The sample includes English- and French-
speaking Montrealers and represents three different clinical sites. Recent immigrants
were excluded. No detectable differences between responses of men and women or
between people of different ethnic backgrounds were observed. All names that ap-
pear are pseudonyms, to ensure anonymity.

18. Richards (1996) suggests that Mendelian genetic explanations may contradict lay un-
derstandings of inheritance grounded in kinship concepts. He finds that disorders
that “run in the family” are often linked to other associated groups of characteristics,
which one may or may not inherit from the affected parent. This anti-Mendelian phe-
nomenon, which Richards calls blended inheritance, is taken up in another paper by
the authors that looks at the manner in which adult children of Alzheimer’s disease
sufferers grapple with the complexity of risk estimates for AD based on ApoE status
(Prest, Lloyd and Lock, in progress, “When It ‘Runs in the Family’: The Interplay of
Heredity, Inheritance, and Genetics in Understanding Risk for Late Onset AD). In
spite of the fact that these respondents have undergone education and genetic-coun-
seling sessions, in addition to being followed for one year and completing numerous
qualitative scales that have reviewed the genetic information about ApoE and for AD,
we found continued references to blended inheritance and extremely simplified and
abstracted genetic explanations. These findings echo the data reported here, but are
of great interest in light of the unique nature of the sample population. This paper in
progress more fully engages the social science literature on lay understandings of ge-
netics and explores the significance of the lack of penetrance of genetic knowledge
surrounding susceptibility and risk for developing AD.

19. For a rich account of how family members grapple with theories of causation, see
Annette Leibing’s research on AD in Brazil (2002), in which she depicts “the struggle
for meaning situated at the interface of biomedical explanations and intuitive in-
sights.”

20. One future objective of this ongoing study is to characterize the exchange of knowl-
edge concerning AD that takes place between general practitioners (or nonspecialist
clinicians) and patients and their caregivers. Such interactions are likely to constitute
a majority of the clinical encounters concerning memory loss. Although the theory is
speculative, we anticipate that even less genetic knowledge is transferred in these
encounters, in light of findings from our research conducted in the specialist clinics.

21. It is possible that the family members raise their concerns about their risk for getting
AD with their own general practitioners or family doctors. This part of a research
project is yet to be carried out.
REFERENCES


